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The effect of psychosocial information resources on the psychological impact of genetic testing for patients

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The effect of psychosocial information
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genetic testing for patients

LEWIS C.

PhD

2011

The effect of psychosocial information
resources on the psychological impact of
genetic testing for patients

by

Celine Lewis

A thesis submitted to the University of Plymouth in partial
fulfilment for the degree of

DOCTOR OF PHILOSOPHY

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Abstract:

The effect of psychosocial information resources on the psychological impact of genetic testing for patients

Celine Lewis

Background: The genetic testing process has been shown to have a profound psychosocial impact on patients and families, yet research suggests that there is a lack of practical and helpful psychosocial information written to support decision-making. Ideally, this should be available for use both before and after genetic testing and should be easily accessed through genetic clinics. The development of pre-written leaflets or on-line resources which draw on the experiences and advice of families who have been through similar experiences, and are readily available through genetic clinics, might be one way of helping families make necessary adjustments.

Aim: The aim of this study was to develop information resources for a) people undergoing carrier testing, and b) parents of children with undiagnosed conditions, and to pilot the use of these resources with service users.

Methods: A systematic literature review was conducted to identify key themes to inform the content of the resources. To build on these findings, in-depth interviews were conducted with 11 people who had undergone carrier testing and 14 parents of children without a diagnosis. Interview data were analysed using the grounded theory method. A grey literature search of existing patient information was also conducted. These three phases informed the content of information resources. The development process also included input from genetic specialists, patient group representatives and interviewees. Finally, a pilot study was conducted through three genetic centres to assess the feasibility of a study testing the use of the resources.

Findings: The participants in this study were striving for empowerment: carriers sought reproductive empowerment; parents developed empowerment strategies in order to advocate for their child. Moreover, a theory named 'reconstructing the meaning of being a parent' was constructed to describe the experience of parenting a child for whom no clear care pathway existed. The importance of providing timely information was identified as being a key factor in supporting parents during their search for a diagnosis. A new model was built to summarise the overarching experience of participants in this study.

Conclusions: Empowerment was identified as a dynamic and multi-faceted construct. Health professionals and support groups can help facilitate the empowerment process through the provision of timely psychosocial information. This is particularly important in an age when patients are expected to take greater control than ever before over decisions affecting their healthcare.

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List of Abbreviations

AGNC	Association of Genetic Nurses and Counsellors
BDI	Beck Depression Inventory
BMD	Becker muscular dystrophy
CF	Cystic fibrosis
CGD	Chronic granulomatous disease
CPHVA	Health Visiting Union
CRI	Coping Responses Inventory
DMD	Duchenne muscular dystrophy
FX	Fragile X
GHQ-12	General Health Questionnaire (12 item)
GP	General practitioner
H	Haemophilia
HADS	Hospital Anxiety and Depression Scale
HbPs	Haemoglobinopathies
HOS	Health Orientation Scale
IES	Impact of Event Scale
IPA	Interpretative phenomenological analysis
IVE	In vitro fertilisation
NFCS	Need for Closure Scale
NHS	National Health Service
PANAS	Positive and Negative Affect Scale
PPC	Perceived Personal Control Scale
PKU	Phenylketonuria
PSW	Patient Satisfaction with Information Scale
PGD	Preimplantation genetic diagnosis
R&D	Research and Development
RCT	Randomised controlled trial

SD	Standard deviation
SMA	Spinal muscular atrophy
STAI	Spielberger State-Trait Anxiety Inventory
SWAN	Syndromes without a Name
TSCS	Tennessee Self Concept Scale
VAS	Fragile X Visual Analog Scale

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Author's Declaration

At no time during the registration for the Doctor of Philosophy has the author been registered for any other University award.

During the course of the study a number of relevant postgraduate courses were attended to gain transferable and research skills. Relevant seminars and conferences were also regularly attended, at which work was sometimes presented in the form of a poster or presentation. These are recorded in the Graduate School logbook.

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Chapter One:

Introduction

1.1 Introduction

Individuals and families affected by a genetic condition have a range of healthcare needs. These may include psychological support to enable them to deal effectively with the challenges presented by being affected by or at risk of the condition. The focus of this doctoral study was the exploration of psychosocial needs of two specific groups of patients accessing genetic services, development of evidence-based psychosocial information resources for those individuals and assessment of the resources. In this first chapter, I will set the scene for the study. I will begin by providing a brief historical overview of genetic testing, followed by an introduction to genetic services currently offered within the United Kingdom (UK). I will then give a description of genetic counselling and genetic testing. Previous research focusing on the psychological impact of genetic testing will then be presented, followed by a discussion of the rationale behind this body of research. Finally, I will outline the four phases of this study, including a brief description of each chapter.

1.2 An Historical Overview

The principles of genetic inheritance were discovered by Gregor Mendel in the mid 19th century (Miko, 2008). However, the significance of his work was not recognized until the turn of the 20th century when scientists began explaining the *inheritance of genetic diseases through Mendelian patterns* (Sapp, 1990). An important step in the development of human genetics and its application to

medicine came with Garrod's demonstration of a Mendelian mode of inheritance in alkaptonuria disease and other inborn errors of metabolism in 1902 (Motulsky, 2010). Further milestones were Pauling's elucidation of sickle cell anemia as a "molecular disease" in 1949, and the discovery of genetic enzyme defects as the causes of metabolic disease in the 1950s and 1960s (Motulsky, 2010).

New understanding of metabolic processes paved the way for advances in biochemistry and genetics. In the late 1960's phenylketonuria (PKU) became the first genetic disease for which neonatal screening was offered, when scientists' pioneered mass biochemical testing for newborn babies through dried blood spot samples (Wilcken and Wiley, 2008). During the 1970's, the development of enzyme assay testing enabled the medical community to start testing for two new diseases, Tay Sachs (Schneider et al., 2009) and sickle cell anemia (Ashley-Koch et al., 2000). Both these diseases are associated with certain ethnic groups; Tay Sachs being a disease predominantly affecting children of Jewish ancestry and sickle cell a disease that mainly affects people of African descent. Mass screening to identify carriers of sickle cell in the United States led to misunderstanding and discrimination, and many African-Americans were stigmatized as a result of their carrier status (Markel, 1992).

The discovery that normal human cells contain 46 chromosomes occurred in 1956 (Trask, 2002), paving the way for the development of prenatal diagnosis by amniocentesis for the detection of chromosomal conditions. The first use of amniotic fluid examination in the diagnosis of genetic disease was reported in 1956 (Fuchs and Riis, 1956). Fuchs and Riis determined fetal sex from cells found in amniotic fluid, based on the presence or absence of the Barr body (Moore et al., 1953). In the same year, John Edwards discussed for the first time the possibility of

the antenatal detection of hereditary disorders (McKusick, 2007). Prenatal diagnosis through amniocentesis was first carried out in 1960 (Cowan, 1994) and was conducted to ascertain the sex of a fetus whose mother was a carrier of haemophilia (Riis and Fuchs, 1960). The suitability of amniotic fluid for karyotyping was demonstrated in 1966 (Steele and Breg, 1966) and in 1968 Nadler reported one of the first diagnoses of Trisomy 21 (Nadler, 1968) using cultured cells for full chromosomal analysis. In the following years, amniocentesis and genetic diagnosis became more prevalent, with genetic laboratories analysing amniotic fluid for detection of chromosomal abnormalities, X linked conditions, inborn errors of metabolism and neural tube defects. Significant improvements in prenatal screening were made in the 1980's, in particular for detecting Down syndrome in the fetus, when it was demonstrated that combining maternal age data with maternal serum alpha fetal protein level could increase the detection rate substantially (Cuckle et al., 1987). Invasive testing was offered to those women whose fetus was found to be at a specific level of risk or higher.

Prior to the identification of specific gene-causing mutations, linkage was the technique used for genetic testing in families (Skirton and Patch, 2002). Linkage disequilibrium (also known as allelic association) was originally predicted back in 1909 by Weinberg, who documented that alleles of two adjacent loci asymptotically approach random association in a population (Xiong and Guo, 1997). Yet it wasn't until 1947 when the technique was first used to identify human genetic disorders, in this case for the blood antigen rhesus factor (Fisher, 1947). However, initially the linked markers were too few and not close enough to be practical in prediction. The key development was the discovery that deoxyribonucleic acid (DNA) polymorphisms are so abundant that they could

provide close markers for any disease gene, even if the gene itself was still unknown. In the 1970's and early 1980's, the availability of DNA-based polymorphisms led to the application of linkage disequilibrium to search for causative genes in diseases such as cystic fibrosis and Huntington disease (Cardon and Bell, 2001); and in 1989 a gene mutation causing cystic fibrosis (CF) was identified (Kerem et al., 1989). The ability to detect mutations at the DNA sequence level had important implications for genetic diagnosis. Previously, genetic testing for CF had only been available to families with affected children and to their close relatives (Beaudet et al., 1988). Knowledge of the CF mutation at the DNA sequence level enabled testing of any random individual. Since then, our understanding of the impact of gene mutation on common and complex conditions has advanced rapidly, as have genetic testing techniques (Ali-Khan et al., 2009). As the range of conditions and susceptibilities for which genetic testing is available has broadened, so too have the numbers of requests by patients and families for counselling and genetic testing (Ibarreta et al., 2004).

1.3 Genetic Services in the United Kingdom

Genetic services have grown out of a need for professionals who can provide genetic information, education and support to patients and families with current and future genetic health concerns (Lea, 2000). The first genetic clinic in Europe was set up by Dr Fraser Roberts at The Hospital for Sick Children, Great Ormond Street in 1946, and fairly soon after he was joined by Cedric Carter (Harper et al., 2010 p.26). This in turn led to the decision in 1957, by the Medical Research Council, to create a formal research unit in clinical genetics at the Institute of Child Health, London, with Roberts as the director (Pembrey, 1987). Roberts had a particular interest in multifactorial causation and genetic influences

in common diseases (Pembrey, 1987). He was also interested in genetic counselling issues, for example, how far couples understood the information they were given on risks, and the decisions the couples took about planning further pregnancies (Carter et al., 1971). In 1960, Paul Polani became director of the Paediatric Research Unit at Guy's Hospital, London, a unit funded by what was then called the Spastics Society (Scope since 1994), with the specific remit of developing medical genetics research on a broad front in relation to the prevention of developmental disorders. The unit provided an unparalleled model for combined research and service in medical genetics across a wide range of laboratory areas and helped to establish medical genetics as a specific discipline (Harper, 2007). Moreover, links with clinicians were strengthened by providing clinical genetic services outside of central London, which were linked administratively with Guy's Hospital. This regional approach was to become one of the hallmarks and strengths of medical genetics in the UK, and still features in existing services today.

Currently within the UK, twenty five genetic services exist which are located regionally as part of the National Health Service (NHS). Broadly, the aim of genetic services is to respond to the needs of individuals and families, particularly their wish to know whether they are at risk of developing a genetic disorder or bearing an affected child (Godard et al., 2003). A more detailed definition is provided by The Clinical Genetics Committee of the Royal College of Physicians of London (Royal College of Physicians, 1996) who define a clinical genetics service as having three main objectives:

1. making a genetic diagnosis, pedigree analysis and estimate of transition risk for people who are affected or referred because of a genetic risk;

2. supporting the identification and surveillance of relatives who are at risk of a serious genetic disorder but who may not have been directly referred; and
3. providing support to family members, both affected and unaffected.

As well as personal communication between the genetic specialist and client, there are a number of other ways in which this support process may take place including telephone support, the provision of written summary letters and patient information leaflets. Specialists are also encouraged to maintain good, ongoing relationships with support groups and direct patients and families to relevant organisations if further support and information is desired (Genetic Alliance UK, 1998).

Information provision within the clinical genetics setting is part of the process referred to as genetic counselling, which also includes helping people understand and adapt to medical, psychological and familial implications of genetic contributions to disease (Resta et al., 2006). It is understood to be a dynamic psycho-educational process between the provider and the client, the goal being to facilitate the client's ability to use genetic information in a personally meaningful way that minimises distress and increases personal control (Biesecker and Peters, 2001).

Clinical Genetics is a recognised medical specialty in the UK and referral to the service is usually made through a consultant physician or general practitioner. Patients and families may be referred for a number of reasons, including:

- there is a known or suspected genetic condition in the family;

- a child has learning difficulties, developmental delay or congenital abnormalities and a genetic link is suspected;
- an abnormality of the fetus has been diagnosed during pregnancy;
- a woman has experienced recurrent miscarriage or stillbirth; and
- there is a strong family history of cancer.

The clinical team consists of specialist physicians working with genetic counsellors (who may have master's degrees in genetic counselling) or specialist nurses. Some genetic counselling which does not involve clinical diagnosis is provided directly by genetic counsellors (Donnai, 2002). Contact with the genetic service may be for a single visit; in other cases it may be recurrent. Complex conditions, multi-disciplinary and long-term management of patients may be co-ordinated by the genetics service.

1.4 Genetic Counselling

The introduction of nurses and social workers within genetic services has been relatively recent, with the formation of an association in the UK - The Genetic Nurses and Social Workers Association, now The Association of Genetic Nurses and Counsellors (AGNC) - only occurring in 1980 (Farnish, 1988). An early definition of genetic counselling was given by Fraser (1974), which was the result of a workshop sponsored by the National Genetics Foundation in America to evaluate and make recommendations about the status of the profession. Genetic counselling was defined as a communication process to:

- help patients and families understand the medical facts;
- understand the prognosis and management of a condition;
- understand the inheritance and recurrence risk;

- be aware of options for dealing with the recurrence risk;
- help families come to an informed decision regarding the course of action in accordance with their beliefs and goals; and
- make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

This definition was adopted by the American Society of Human Genetics in 1975, and is still used to this day by the AGNC.

1.5 Genetic Testing

The term 'genetic testing' is used in a variety of different settings, including medical care, forensic services and the insurance industry, often with very different meanings (Pinto-Basto et al., 2010). In the healthcare context, genetic testing has been defined by a UK Advisory Committee on Genetic Testing as "testing to detect the presence or absence of, or alteration in, a particular gene, chromosome or gene product" (Advisory Committee on Genetic Testing, 1998, p.8). Zimmern's (1999) definition focuses more on outcomes, defining genetic testing as "any clinical, haematological, radiological or biochemical test, from which information about the gene or the inheritability of a disorder to be inferred," (p153).

In the context of clinical genetics, genetic tests are used for a number of reasons. These include:

Diagnostic testing - a genetic test performed in a symptomatic individual to confirm or exclude a genetic condition;

Predictive testing - a genetic test performed in a healthy high-risk relative for a specific later-onset monogenic disorder. The mutation in the family leads to the

disease or a considerably high risk for the disease (like in high risk familial cancers). Sometimes the term 'presymptomatic test' is used when referring to mutations with full penetrance;

Carrier testing - a genetic test that detects a gene mutation that will generally have limited or no consequences to the health of the individual. However, it may confer a high risk of disease in the offspring, if inherited from one parent (in the case of X linked inheritance, autosomal dominant premutation or chromosomal translocation) or in combination with the same or another mutation in the same gene from the other parent (in case of autosomal recessive inheritance);

Prenatal testing - a genetic test performed during pregnancy where there is an increased risk for a certain condition in the fetus;

Genetic screening - testing where the target is not high risk individuals or families, but where the test is systematically offered to the general population or a part of it (for example newborns or a particular ethnic group). (Kaariainen et al., 2008).

1.6 Psychological Impact of Genetic Testing

The genetic testing process can have a profound impact on patients, both positive and negative, and there is an abundance of literature on the subject, particularly in the area of predictive and presymptomatic testing for conditions such as hereditary breast and ovarian cancer, and Huntington disease. Lim et al. (2004) found that emotional turmoil was experienced by patients when first receiving test results for hereditary breast/ovarian predisposition genes. Carriers also had difficulty divulging results to family members. Watson et al. (2004) found that women who were younger than 50 years of age had a significant rise in cancer-related worry one month after testing. Women undergoing BRCA1 and

BRCA2 testing who had been previously diagnosed with breast and ovarian cancer, found the information beneficial as it ended the uncertainty, in a study conducted by Hallowell et al. (2002). However, the women had difficulties in disclosing information to kin and increased anxiety about their own or others' cancer risk. D'Agincourt-Canning (2006) also identified mixed experiences of women identified as having an increased risk for cancer. Genetic information was generally found to be enabling and enabled women to take measures such as surveillance and prophylactic surgery to confront the disease. However, for a small number of women, the experience was negative and had a profound and limiting effect on their sense of control over their lives. Gargiulo et al. (2009) conducted a cross-sectional study which was conducted with 351 persons who underwent presymptomatic testing for Huntington disease. They found that 58% of carriers were found to be experiencing depression. Furthermore, 27% of non-carriers did not cope well with a favourable result, and 24% were depressed during follow-up. Tibben et al. (1997) also looked at the psychological impact of receiving carrier test results for Huntington disease. They found that one week after disclosure of test results, carriers had increased levels of hopelessness. Carrier partners also followed the same course of distress as carriers, and those with children were significantly more distressed than those with no offspring.

1.7 Psychosocial Impact of Living with a Genetic Condition

The psychosocial needs of patients and families living with a genetic condition are also well documented in the literature, as the following examples highlight. Skirton found that parents whose child had been referred for possible genetic diagnosis reported feeling stress, fear, anxiety and guilt (Skirton, 2006b). The impact of the child's condition on the family was powerful, altering the way in

which the entire family functioned. In a study (Packman et al., 2010) in which the emotional health, psychosocial needs and concerns of individuals with Gaucher disease were assessed, 64% of participants said the condition had an effect on their ability to do their job, and 22% said that the condition had an effect on their ability to make plans, travel, or do physical activities. Furthermore, participants described an array of emotions including sadness and depression (25%), and anxiety, worry or increased stress (35%). Szyndler et al. (2005) found that young people with a delayed diagnosis of cystic fibrosis who are alienated from their families may be in need of additional psychosocial support. Family cohesiveness, expressiveness and organisation were also associated with better psychological functioning. Lewis et al. (2006) identified that mothers of sons with fragile X syndrome were pessimistic about their son's future, experienced conflict within the family and had lower levels of reciprocated closeness. In a study conducted by Read et al. (2010), siblings of children with Duchenne muscular dystrophy were more likely to score at high-risk for psychological morbidity compared to the general population norm. Parents reported high levels of overall impact of the condition on the family, in particular concerning mental health, finances and leisure time.

1.8 Written Information – An Important Aspect of the Counselling Process

Whilst the experiences cited above may be natural responses to stressful situations, psychosocial support has been shown to have a positive impact on patient outcomes. Cognitive-behavioural therapy was found to be an effective pain management treatment for sickle cell disease, in a study conducted by Edwards and Edwards (2010). Arnaboldi et al. (2010) established that a psychosocial cancer phone centre staffed by professional psychologists was an

effective support mechanism for a broad range of cancer patients. In a study conducted by Barilla et al. (2010), providing comprehensive psychosocial follow-up support for new mothers in the postpartum period reduced the rate of normal newborn readmissions. Authors of a systematic review on risk communication in genetics highlighted that it was the supportive or emotional elements of counselling that provided benefits to users, rather than the informational or educational elements (Edwards et al., 2008).

Psychosocial support through the provision of written information has also been shown to help improve a number of outcomes including patient satisfaction (Austoker and Ong, 1994, Iconomou et al., 2006), information retention and understanding (Beaver et al., 2009, Bucker et al., 2010, Entwistle and Watt, 1998) and anxiety reduction (Kutluturkan et al., 2010). Moreover, there is overwhelming evidence that patients want written information about medical conditions, treatments and their outcome (Nachtigall et al., 2010, Paul et al., 2004, Soltner et al., 2009).

Within the context of clinical genetics, a number of studies have been conducted to assess the impact of providing written information to patients and families. A booklet provided to women considering genetic testing for a BRCA1/2 mutation was found to have a positive impact on satisfaction with the information provided, to decrease the decisional conflict due to lack of information, and have a marginal positive impact on knowledge (Mancini et al., 2006). Roshanai et al. (2009) found that receiving written information was one of the factors that improved satisfaction with the content of the information provided by counsellors in a cancer genetics service. Increased knowledge was a main outcome of providing parents of children with beta-thalassaemia with an information booklet

(Dehkordi and Heydarnejad, 2008) and decisional conflict was found to be reduced through the use of leaflets in women considering prenatal screening (Dahl et al., 2006).

As genetics is an emerging field within healthcare, genetic literacy amongst the general population is relatively low (Skirton and Eiser, 2003). Furthermore, the *complex nature of genetic information and the fact that it is often provided when a patient is in distress*, means that understanding and retention of information is often poor for many people (Gordon et al., 2003, Henneman et al., 2002a, Jedlicka-Kohler et al., 1996, Lakeman et al., 2008, Rona et al., 1994). For these reasons, it is imperative that oral information is supported by written material during the counselling process. As well as enhancing the communication process between clinician and patient, written information can be kept and re-read at a later date, and shown to other family members who may themselves be at increased risk.

There has been some concern recently regarding the quality of the written information patients and families receive. In a study conducted by Dahl et al. (2006), written materials discussing prenatal testing were found to have important information missing, and were often insufficient in detail. Alouini et al. (2007) reported that written prenatal information could have been improved by using simple and accessible language. Members of the public who were provided with a seemingly 'neutral' leaflet about genetic testing, were found to have more positive attitudes towards testing than those who did not receive it, in a study of the effects of an information leaflet on public attitudes (Sanderson et al., 2005). In another example, Hargreaves et al. (2005) reported that leaflets on newborn bloodspot screening supported the public health agenda by informing parents of

the benefits of screening, but few supported the informed choice agenda by mentioning either the limitations of screening, or choice, or by being easy to read. In addition to this body of evidence, I conducted my own study to evaluate the quality of written patient information about genetic testing provided by clinicians from across Europe (Lewis et al., 2007). I identified that written material was far more likely to include hard, factual information related to the condition and the test than the more qualitative, experience based information related to the psychological and social implications of genetic testing. This is a concern since studies have shown that patients and families do have psychosocial needs.

1.9 Rationale for the Doctoral Study

Research suggests that there is a lack of practical and helpful psychosocial information, written for patients and families to support decision-making both before and after genetic testing, which can be easily accessed through genetic clinics (Lewis et al., 2007). The shortage of resources and time experienced by staff at genetic services may be one reason for this (Donnai, 2002). Another reason may be that the acceptability and expectation of writing about psychosocial issues may vary across conditions, as well as across countries and more specifically departments. Either way, patients and families do have psychosocial needs, and hence written information exploring this subject may be of benefit to patients during the genetic testing process. The development of pre-written leaflets or on-line resources which draw on the experiences and advice of families who have been through similar experiences, and are readily available through genetic clinics, might be one way of helping families make the necessary adjustments during these difficult periods.

My previous study assessing the quality of written information about genetic testing (Lewis et al., 2007) as well as informal investigation at the beginning of this doctoral study (looking specifically at available information through genetic clinics) identified two areas where very little psychosocial information was available for patients and families.

1. Whilst the information relating to being a carrier of an autosomal recessive and X linked condition that is available through genetic clinics provides factual, scientific information, there is little discussion of the qualitative patient experience. This is a concern because people who have undergone carrier testing for a variety of conditions have been shown to have very real psychosocial needs. Women found to be carriers of fragile X syndrome perceived this as a barrier for having healthy biological children (McConkie-Rosell et al., 2001). Carriers of CF and haemoglobinopathies intended to use information derived from test results to inform reproductive decisions (Lakeman et al., 2008). Some carriers of CF felt less healthy as a result of their carrier status (Henneman et al., 2002a). Guilt, shock and grief were some of the experiences cited by mothers of children affected by haemophilia (Dunn et al., 2008).
2. Another area where patient information appeared to be severely lacking is in supporting parents whose children have been referred for genetic investigation but no diagnosis has been given. Formal research into this area also appears to be sparse; a literature search identified only two papers on the subject (Graungaard and Skov, 2006, Rosenthal et al., 2001), both of which highlighted the psychosocial difficulties of

coping with an uncertain future. However neither of these studies was conducted in the UK. At the time of beginning this doctoral study, and since the closure of the support group Syndromes Without A Name (SWAN) in 2009, there was no support group in the UK specifically for parents of children without a diagnosis. Yet my experience of working at Genetic Alliance UK, a national charity supporting people affected by genetic conditions, has highlighted that there are many parents in this situation who want help, support and contact with other families. The Genetic Alliance UK has produced a 'route map' for parents of undiagnosed conditions, which signposts them to sources of information and appropriate services (Genetic Alliance UK, 2008). The organisation Contact a Family has also developed information for parents (Contact a Family, 2009), however neither look in detail at social and psychological outcomes. Furthermore, through my own informal investigation, it did not appear that any of the genetic centres in the UK have developed psychosocial written information materials supporting parents in this position.

I therefore felt there was a need to look at these two areas more closely.

1.10 Description of the Doctoral Study

1.10.1 Aims

There were four broad aims associated with this doctoral study:

1. explore the impact of carrier testing;
2. explore the experience of parenting a child with an undiagnosed condition;

3. develop evidence-based psychosocial information resources related to these two areas; and
4. assess the utility of providing this psychosocial information to service users.

1.10.2 Objectives

In order to fulfil these aims, the following five objectives were set:

1. undertake a systematic review of the literature in order to establish a baseline of information for these two areas of research;
2. corroborate the findings from the systematic review through conducting *qualitative research with service users who have taken a carrier test or are the parent of an undiagnosed child*, to inform the content of the information resources;
3. conduct initial piloting of resources with the help of patients and professionals to ensure accuracy, readability and relevance of content to service users;
4. develop two versions of the resources, the first with scientific content only, the second with both scientific and psychosocial content; and
5. pilot these two versions with service users to determine whether the resources with additional psychosocial information had a greater impact on patient satisfaction and reduction of unwanted psychosocial responses than those receiving scientific information only.

1.10.3 Mixed methods study design

This was an exploratory study using a sequential, mixed methods approach in which the second phase elaborated and built on the first. In order to gather in-

depth data that would give a rich understanding of the two areas of interest and inform the content of the information resources, qualitative methods were considered most appropriate. This was especially the case for the resource for parents of undiagnosed children (from now on referred to as the 'non-diagnosis resource') for which there was little information currently available which could feed into it. However, in order to test the resources for effectiveness, a quantitative method using validated psychology measures was considered most suitable as it makes it possible to do statistical analyses that cannot be done using qualitative research methods. For these reasons, a mixed methods approach was considered most suitable. Furthermore, by combining a variety of methods, triangulation, whereby results from different methods can be compared to corroborate the findings, could be achieved (Mays and Pope, 2006).

The concept of mixing different methods probably originated in 1959 when multiple methods were used to study validity of psychological traits (Creswell, 2003). The method gained popularity as researchers recognised that by mixing methods, biases inherent in any single method could be overcome (Creswell, 2003). Through this emerged the method of triangulation, a means of verifying qualitative and quantitative methods (Jick, 1979). Moreover, further benefits of mixing data emerged as a result of the increased popularity of this method. For example, Greene et al. (1989) identified that the results from one method could help develop or inform other methods.

A common reason for conducting a mixed methods study is to expand the scope of the enquiry by accessing a wider range of data (O'Cathain and Thomas, 2006). According to O'Cathain and Thomas, there is increasing recognition of the numerous factors that affect health and healthcare. This awareness, coupled with

a desire to answer a wider range of questions, has increased the popularity of the mixed methods approach, particularly because qualitative and quantitative methods are suited to different types of enquiry and have their own strengths and weaknesses (further discussion on qualitative and quantitative research methods is presented in Chapter Three).

A particular benefit of using a mixed methods approach is that the researcher may use the theoretical and methodological differences within a study to generate more insights than they might have done if they had only used a quantitative or qualitative method (Greene and Caracelli, 1997). Furthermore, mixed methods studies can produce a whole greater than the sum of the parts (Barbour, 1999). One potential drawback if the research is being conducted by more than one researcher is that each researcher may have their own theoretical and methodological perspectives which may create problems within the team (Sandelowski, 2000). However, as there was only one main researcher working on this study, this issue would not be relevant in this particular case.

There are a number of recent examples of where mixed methods approaches have been used in exploratory studies. Freer et al. (2010) examined trauma narratives from survivors of interpersonal violence using qualitative and quantitative methods. This enabled the authors to assess coherence and explore narrative characteristics among differentially exposed groups. In a study conducted by Buckley (2010), descriptive statistics and qualitative methods were used to explore nurses' experiences of nursing young people with mental health problems. A mixed methods approach was also used by Phelan (2010) to *determine the feasibility of an innovative approach to targeting pain in older*

people in the acute care setting. The pilot study design included pre and post evaluation surveys and interviews.

This doctoral study comprised four distinct phases:

Phase 1: I undertook a systematic review of the current literature relating to the psychosocial impact of carrier testing, and key themes, similarities and differences across the studies were identified. As a literature search only identified two studies in which the parental impact of living without a diagnosis was assessed, a synthesis of the findings was incorporated into the Discussion section of Chapter Five.

Phase 2: I expanded the findings from the systematic review by conducting in-depth interviews with (a) people who had undergone carrier testing and (b) parents of undiagnosed children. I used grounded theory method to analyse the data. I then compared the key themes with those identified in the systematic review. A theory that described the experience of service users accessing the genetic clinic began to emerge.

Phase 3: I used the findings from the systematic review and the qualitative interviews to inform the content of the information resources. Furthermore, I conducted a grey literature search of patient information materials from support groups and genetic centres to build on the key issues identified during Phase 1 and Phase 2. Draft versions of the resources were piloted with interviewees to ensure respondent validation as well as key stakeholders including patient group representatives, genetic health professionals and interview participants.

Phase 4: In this final phase, I conducted a pilot study through three regional genetic centres. The objectives were to: (1) check the acceptability of the study procedure; (2) check the acceptability of the control and intervention; (3) assess

recruitment and attrition rates; and (4) carry out a trial-run of the types of analysis that would be used in a definitive trial. The pilot study also provided an opportunity to corroborate the findings from the previous three phases of the study by validating the 'key issues' identified through the systematic review and qualitative interviews with service users, as well as gather further feedback about the resources. In addition, the findings from the pilot study were incorporated into a final overarching theory.

1.10.4 Outline of chapters

Chapter One: I provide a brief overview of genetic services, and present the rationale for the study and description of the study process.

Chapter Two: I present the findings from a systematic review of the carrier testing literature. I discuss the key themes in relation to theoretical concepts.

Chapter Three: I describe the method chosen to investigate the psychosocial experiences of participants. I discuss and justify the study procedure and the method chosen to analyse the interview data.

Chapter Four: I present and discuss the findings from the qualitative interviews with participants who underwent carrier testing. I then propose and examine theoretical explanations that can be used to interpret the data. Finally, I introduce a theoretical model summarising the experience of participants.

Chapter Five: I present and discuss the findings from the qualitative interviews with parents of undiagnosed children. I propose a new theoretical model to help interpret the data as well as examine existing theoretical constructs.

Chapter Six: I describe the method used to develop the information resources, and discuss the procedure used to test and revise the resources.

Chapter Seven: I introduce the method used to pilot the educational resources. I outline the study design, recruitment procedure, measures chosen and method for data analysis.

Chapter Eight: I present and discuss the findings from the pilot study. I also discuss the implications of the results and make suggestions for a subsequent study.

Chapter Nine: In the final chapter, I begin by presenting an overarching theoretical framework that draws together the findings from the four phases of this research. I then discuss the relevance of the study findings within the context of genetic counselling and beyond. I provide a reflective assessment of the study process, and finally make recommendations for clinical practice and future research.

1.11 Summary

In this chapter, I have given a brief overview of genetic services in the UK as a background to the study. I have described the findings from previous research in the field and presented the rationale for conducting this study. In the following chapter, I will present a systematic review of the literature on the psychosocial impact of carrier testing.

Chapter Two:

A Systematic Review of the Carrier Testing Literature

2.1 Introduction

The psychosocial impact of carrier testing and living as a carrier has been the focus of numerous studies. In order to identify key themes associated with this experience, I conducted a systematic review to synthesise this body of knowledge. In this chapter I will describe the method used to conduct this review, and present the main findings. I will also discuss the findings in relation to established theoretical concepts.

2.2 Aim and Objectives

The aim of the systematic review was to investigate the psychological and social impact of genetic carrier testing on the individual being tested. The main objectives were to:

- identify the key factors affecting the impact of carrier testing results on individuals;
- establish commonalities and differences across different condition types and inheritance patterns; and
- assess the methodological quality of the literature; and
- identify any gaps in the knowledge.

Furthermore, the systematic review gave me an opportunity to familiarise myself with the existing body of research in the field and ascertain the methods used by other researchers to inform my own research method in Phase 2.

2.3 Design

This was a systematic review including qualitative, quantitative and mixed methods studies. A systematic review is a reliable method of collating empirical evidence, synthesising and interpreting the findings of the included studies in a transparent and nonbiased way (The Cochrane Collaboration, 2009). The processes used for the systematic review were based on those described by Pope, Mays and Popay (2007). These include: the development of the review question and boundaries; a comprehensive search; application of inclusion criteria to studies identified; quality assessment of included studies; data extraction; synthesis of findings and reporting of findings and their implications. Methods of data analysis and display as described by Miles and Huberman (1994), including presenting key aspects of each study in a matrix, were also used.

2.4 Search Methods

Advice regarding relevant databases was sought from an experienced genetic counsellor and the University of Plymouth librarian for health. The following databases were recommended and used: CINAHL, Embase, Ovid, Medline, PsychINFO, Pubmed, Web of Science. Grey literature, such as newsletters and reports, and book chapters were not searched as they do not have the same level of robustness as peer reviewed academic studies.

Initially, my supervisor and I conducted a 'brainstorming' session to identify relevant keywords related to the subject of the review. These keywords along with their relevant truncations were used as search terms. The inclusion and exclusion criteria were also decided at the outset. Where possible, the inclusion criteria were included as search terms to try to limit the retrieval of non-relevant articles.

In the first instance, the following search terms and limits were used:

carrier test or carrier test*

AND

genetic or DNA or chromosome or autosomal recessive or recessive or X
linked

AND

depression or emotion or guilt or anxiety or worry or stress or blame or
psychological or psychosocial or social or effect or impact or psychological
impact or social impact or personal or carrier status or distress or relief or burden or
coping or coping strategy or communication.

NOT children NOT cancer NOT prenatal NOT predictive.

Limits set: 1998-2008, English

Initially the limit was set to publication dates between January 1998 and October 2008 to limit the search to studies written within the last 10 years. Twelve potentially relevant studies were found. However there were a number of relevant studies that were written between 1990 and 1998 that I became aware of during the search. It is likely that there were relevant papers written during this period because from about 1990 onwards DNA carrier testing became feasible clinically and tests started to become routinely performed on patients with a family history of recessive and X linked conditions such as CF, fragile X and Tay-Sachs disease (Broide et al., 1993, Kerem et al., 1989). At the same time, studies that assessed the impact of the test on the patient began to appear in the literature. It was

therefore decided to extend the search to include those papers written between 1990 and 2008 so as not to omit any relevant studies. In addition, a number of new keywords were added to the search. These were identified by looking through the 'keywords' given on the relevant studies.

In the second database search, the following search terms and limits were used:

carrier test or carrier test or carrier screening or genetic screening or population screening or cascade testing or heterozygote testing*

AND

genetic or DNA or chromosome or autosomal recessive or recessive or X linked

AND

depression or emotion or guilt or anxiety or worry or stress or blame or psychological or psychosocial or social or effect or impact or psychological impact or social impact or personal or carrier status or distress or relief or burden or coping or coping strategy or communication or coping behaviour or emotion* or stigma or self concept or attitude* or genetic counseling or genetic counselling*

NOT children NOT cancer NOT prenatal NOT predictive.

Limits set: dates (1990-2008), language (English) and population (Human; Age: Adult.)

Note: italicised words denote keywords/limits added for second database search.

2.4.1 Inclusion and exclusion criteria

Studies were included if they were:

- systematic reviews, literature reviews, randomised controlled trials (RCT's), quasi-experiments, observational studies, surveys or qualitative studies;
- published between January 1990 to 2008;
- focused on the psychological and social impact of the test result on the patient; and
- focused on either autosomal recessive and X linked conditions, or carriers of chromosomal changes such as translocations.

Studies were excluded if they were:

- about cancer, adult onset conditions or other dominantly inherited conditions;
- ones in which there was potential for participants to find out that they were homozygotes for a particular gene mutation where the age of onset of the disease was in adulthood (e.g. haemochromatosis);
- focused on pregnant women;
- ones that included children or adolescents;
- focused only on recall of information about risk; or
- focused only on motivation for taking/not taking the test.

2.4.2 Comments on inclusion and exclusion criteria

Female fragile X carriers can sometimes be affected by the condition (although in general more mildly) and for that reason fragile X does differ from autosomal recessive and some other X linked conditions. However, carriers of

fragile X were included in the review because a body of knowledge exists relating to the psychosocial effect of carrier testing on this group (Anido et al., 2007, Anido et al., 2005, McConkie-Rosell et al., 2000, McConkie-Rosell et al., 2001). Furthermore, because the aim was to ascertain similarities and differences across a range of different conditions, I felt it was relevant to include this group in the systematic review.

Studies relating to cancer, adult onset conditions or other dominantly inherited conditions were excluded because the aim of this doctoral study was to develop information for patients that informs them of the possible psychosocial consequences of carrier testing for recessive, X linked or chromosomal conditions. Furthermore, the nature of the information derived from predictive and presymptomatic tests will be different from receiving carrier information for recessive, X linked or chromosomal conditions, as conditions identified through carrier testing will not go on to affect the carriers' health. Studies focusing on pregnant women were excluded because their feelings might be highly influenced by worry for their offspring, and also because their decision to seek testing would be influenced by the immediate needs of a current pregnancy (Cheuvront et al., 1998). Studies including children were also excluded because the focus of this doctoral study is to develop information for adults (who are able to give informed consent). It is also likely that children will have very different psychosocial reactions and information needs to adults. Only studies that assessed the psychological and social impact of the test result on the patient were included. Therefore, studies that addressed recall of information about risk and reasons for taking/not taking the test but did not address psychosocial impact were excluded.

2.5 Search Outcome

Eight databases were searched and 1537 articles were identified overall. After reading through the titles and abstracts, 1516 studies were excluded. This left 21 studies which were retrieved in full.

Table 2.1 Results of database searches

Database	Retrieved	Potentially relevant articles after reading abstract
CINAHL	27	4
Embase	44	10
Medline	54	10
Ovid	64	4
PsychINFO	7	4
Pubmed	1095	13
Web of Science	246	11

A hand search of my Endnote archive was also carried out. This archive has been compiled over the last four years, during which time I have been looking at the information needs of patients considering genetic testing. Three further potentially relevant studies were identified in this way. Twenty four studies were retrieved and read. Eleven were then excluded for reasons that were not obvious when reading the abstract alone. Reasons for their exclusion included: the inclusion of pregnant women in the cohort (in two cases this was not explicit in the papers and the author was contacted for clarification); inclusion of children or adolescents; the focus was on family communication towards carrier testing or recall/knowledge of test results and not on psychosocial impact; the paper was an opinion letter. This left thirteen relevant studies.

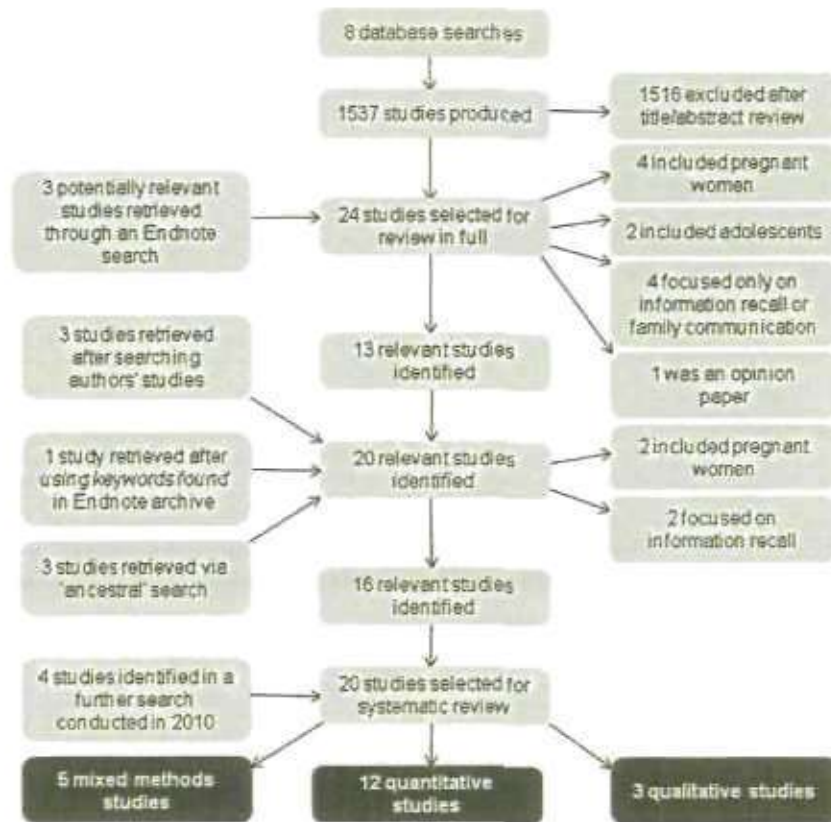
A number of other methods were then used to identify studies. First, an author search was conducted on Pubmed, using the names of the authors

identified through relevant studies. This was considered a useful method because the identified authors were likely to have a special interest in the subject and thus may have published more than one study in the area. Three potentially relevant articles were retrieved through this method. Secondly, the database search was repeated using new keywords found in studies identified through Endnote. The additional keywords included at this stage were: **psychology, social adaptation, reproductive uncertainty, risk perception, carrier couples, family planning, and prospective risk**. One potentially relevant study was found in this way. Thirdly, an ancestral search (reference search) was carried out. Three potentially relevant studies were retrieved in this way. The full text to these seven papers was retrieved, of which three were relevant, leaving sixteen relevant studies. A further search was conducted in May 2010 to identify whether any new and relevant papers had been published since the previous search, or if any papers had been overlooked. Four relevant papers were identified at this stage and these were then included in this systematic review. Therefore, twenty relevant papers were found in total (Table 2.5).

There are a number of reasons why some of the potentially relevant articles retrieved through the Endnote, author and ancestral searches were not identified through the database searches. First, qualitative studies can be poorly indexed and keyworded (Pope et al., 2007) which means that identifying relevant studies can sometimes be quite 'hit or miss'. In some cases, abstracts were not published, making it harder to assess the relevance of the study. In others, the psychosocial findings were not mentioned in the abstract, even though this was a main aspect of the study. Some of the older papers were not always uploaded onto all the

relevant databases. In other papers, the keywords did not appear to be highly relevant or appropriate.

Figure 2.1 Flow diagram of the study selection process



2.5.1 Quality appraisal

Before analysing the findings from the studies, each study was assessed using a quality appraisal tool (Kmet et al., 2004) developed to evaluate primary research papers from a variety of fields. The tool comprises two 'checklists'; one for assessing the quality of quantitative studies; the other for assessing the quality of qualitative studies. Each checklist comprises a number of questions regarding the quality of the paper, for example, "Is the design evident and appropriate to answer the study question?", "Is the analysis described and appropriate?" and "Is there a connection to a theoretical framework/wider body of knowledge?"

Papers are scored depending on the degree to which specific criteria were met ("yes" = 2, "partial" = 1, "no" = 0). A score was calculated for each paper by summing the total score obtained across relevant items and dividing by the total possible score.

This tool has proven internal validity and provides a systematic, reproducible and quantitative means of simultaneously assessing the quality of research encompassing a broad range of study designs. Using this tool, I scored each paper based on quality criteria including the description of the research question, appropriateness of design, justification of sampling strategy, appropriate data collection and analysis and estimates of variance (for quantitative studies) to produce a score phrased as a percentage. Five papers across the range of scores were selected and a blind appraisal was made by my supervisor to verify the results. The papers were ranked in the same order by both appraisers.

Although there remains controversy over the utility of such tools (Garratt and Hodgkinson, 1998, Pope et al., 2007) it is agreed that research should be clear and transparent. Quality appraisal tools do provide a means to assess rigorously the methodological quality of each study, to ascertain whether it is of a good enough standard to be included in the review, and give an indication of the appropriate weight that should be given to the evidence in the different papers. Thus conducting a quality appraisal of the papers was considered worthwhile.

Table 2.2 Quality appraisal scores

Author and year of publication	Quantitative score (out of 100)	Qualitative score (out of 100)
Anido et al. 2007		85
Anido et al. 2005		85
Bekker et al. 1994	63	55
Callanan et al. 1999	75	
Chevront et al. 1998	88	
Dunn et al. 2008	83	
Fanos & Johnson 1995	72	65
Gordon et al. 2003	90	
Henneman et al. 2002	81	
Honnor et al. 2000	90	
James et al. 2006	90	
Lakeman et al. 2008	90	
Marteau et al 1992	72	
McConkie-Rosell et al. 2001	95	75
McConkie-Rosell et al. 2000	95	75
McConkie-Rosell et al.1997	81	
Newman et al. 2002	79	
Pastore et al. 2008	72	
Watson et al. 1992	68	
Williams et al. 1997		85

All 20 papers scored greater than 50% on the Kmet scale and therefore none were excluded on the basis of quality (Kmet et al. do not provide a 'cut-point' at which studies should be discarded on the grounds of poor quality but I assigned a score of 50% or more as acceptable as this indicated that the paper had scored the majority of questions either "partial" or "yes" on the three scaled scoring system). The studies included in the review had notable strengths. Validated questionnaires were used in many of the studies [e.g., Honnor et al. (2000)]. The use of a longitudinal study design enabled changes in psychosocial wellbeing to be measured over time [e.g., Lakeman et al. (2008)], and the use of in-depth qualitative interviews enabled rich and complex data to be acquired by the researchers [e.g., Anido et al. (2007)]. There were a number of study limitations

that should be considered when interpreting the findings from these research studies. First, potential confounding variables, such as whether participants had completed their families at the time of testing [e.g., Bekker et al., (1994)] or whether they knew anyone with the condition for which they were a carrier [e.g., Watson et al., (1992)] were not addressed by some of the authors. In addition, authors of many studies did not justify their sample size [e.g., Pastore et al., (2008)] while some studies lacked clear conceptual definitions and were not based on particular theoretical models [e.g., Henneman et al., (2002)].

2.5.2 Data extraction

To enable comparison across the studies, a matrix (Miles and Huberman, 1994) of studies was drawn up (Table 2.5, p.72) including aspects of each relevant study considered to be most important (study design, sample and size, methods, quality issues and findings related to the psychosocial impact of carrier testing). The matrix method is an effective way of displaying key variables, to facilitate the reviewer to compare and contrast variables and findings.

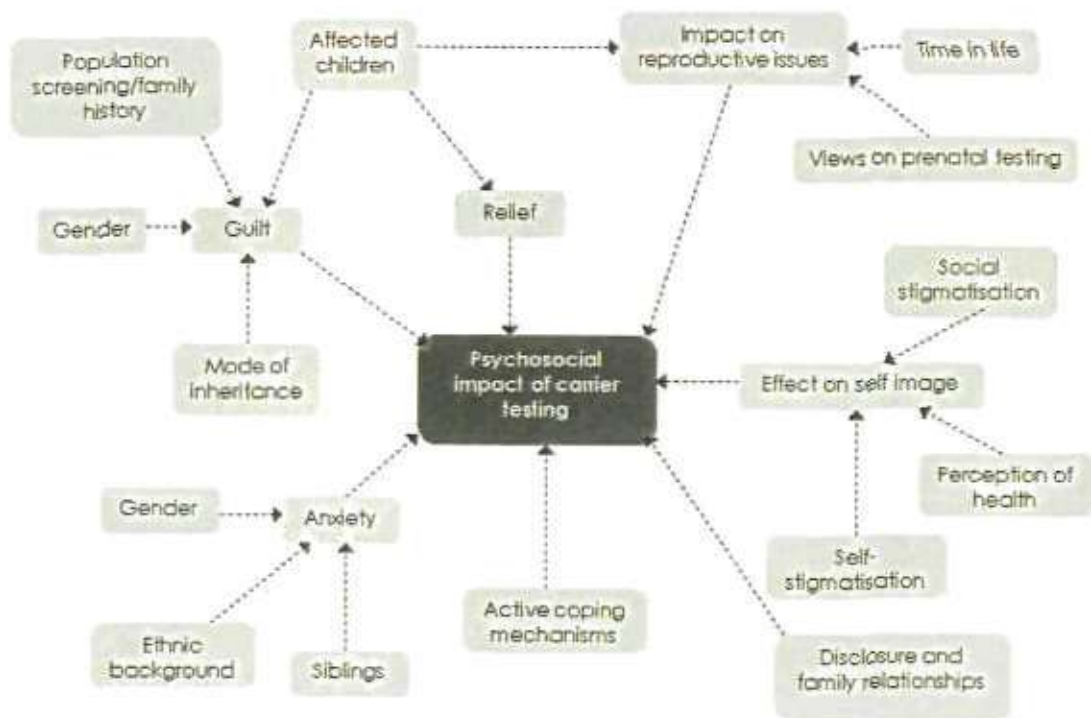
2.6 Analysis of Findings

Due to the range of different quantitative measures and quantitative philosophical approaches used, a meta-analysis was not performed. A thematic analysis was undertaken to elicit general overarching themes from the papers studied; this type of analysis has also been undertaken by other authors such as Legare et al. (2008), Edwards et al. (2008) and Hoey et al. (2008). This method involves 'the identification of prominent or recurrent themes in the literature, and summarising the findings of different studies under thematic headings' (Dixon-Woods et al., 2005). It is a useful method for summarising the data retrieved in a

systematic review as it has the ability to deal with studies with a diverse methodology and allows organised and structured ways of categorising key findings in the literature in terms of themes. Dixon-Woods et al. (2005) highlight that there is sometimes a lack of clarity about exactly what thematic analysis involves and the processes by which it can be achieved. However, stating explicitly the processes involved in analysing and synthesising the data, and where possible using terms that were taken directly from the studies included in the review, should negate the criticism that the method lacks transparency.

Themes were identified by reading and re-reading the studies. The findings were discussed by myself and my supervisor, and compared to identify areas of agreement and disagreement across studies. This resulted in a set of overarching themes. These themes were labelled using the terminology commonly contained in many of the studies, such as guilt, anxiety and stigmatisation. Once this process was complete, a 'spider diagram' or 'mindmap' (Burgess-Allen and Owen-Smith, 2010) was drawn so that the key themes could be visualised and the relationships between them identified (Figure 2.2). Displaying data visually as a spider diagram is an effective and user-friendly method of amalgamating and interpreting data (Pope et al., 2007). It also enables relationships between themes to be seen more clearly than by using the matrix only. Finally, after reviewing the studies, I compared the results with well established psychological models relating to self concept (Shavelson et al., 1976) and coping (Lazarus and Folkman, 1984). These two models have been used in other health-related studies looking at psychosocial adaptation (McConkie-Rosell et al., 2000, Street et al., 2010).

Figure 2.2 Mindmap showing relationships between key themes



2.7 Results

The studies identified had been published between 1992 and 2008. Twelve studies were quantitative, three were qualitative and in five mixed methods were used. Table 2.3 gives an overview of the condition, population group, sample size and country of origin of each paper included in the review. Cystic fibrosis was the most commonly studied condition. No studies that assessed the impact of carrier testing on carriers of chromosomal abnormalities, were identified from the literature review.

There were substantial differences in construct, design, measures, population and outcomes across the studies. Study designs comprised longitudinal studies [e.g., Bekker et al. (1994)], RCT's [e.g., Callanan et al. (1999)],

and cross sectional studies [e.g., Dunn et al., (2008)]. Samples from different populations including the general population [e.g., Henneman et al. (2002a), high risk groups [e.g., McConkie-Rosell et al. (2001) of Jewish decent (Marteau et al., 1992) and women only [e.g., Anido et al. (2005)] were included. Sample size varied from eight participants (Anido et al., 2007) to 2220 participants (Honnor et al., 2000). Data collection methods varied from questionnaires [e.g., Bekker et al. (1994)], to focus groups (Anido et al., 2005) and in-depth interviews [e.g., Williams and Schutte (1997)]. Various measures were used, including the Spielberger State-Trait Anxiety Inventory (STAI), developed by Spielberger (1970) [e.g., Watson et al. (1992)], the Health Orientation Scale (HOS), developed by Wooldridge and Murray (1988) [e.g., James et al. (2006)] and the Tennessee Self Concept Scale (TSCS), developed by Fills and Warren (1996) (McConkie-Rosell et al., 2000). In two cases, the same cohort of participants was involved in two studies, however because different findings were presented in the different papers, both studies were included in the review [e.g., Newman et al. (2002); Chevront et al. (1998)].

Table 2.3 Table comparing condition, population group, sample size and country of origin of each study included in the systematic review

Authors and year of publication	Condition	Family history/ general population	Sample size	Country in which study conducted
Anido et al. 2007	FX	population	8	USA
Anido et al. 2005	FX	both	40	USA
Bekker et al. 1994	CF	population	441	UK
Callanan et al. 1999	CF	family history	372	USA
Chevront et al. 1998	CF	family history	299	USA
Dunn et al. 2008	H	family history	66	UK
Fanos et al. 1995	CF	family history	84	USA
Gordon et al. 2003	CF	population	353	Australia
Henneman et al. 2002	CF	population	826	Netherlands
Honor et al. 2000	CF	population	2220	Australia
James et al. 2006	CGD, DMD, BMD, SMA	family history	112	USA
Lakeman et al. 2008	CF and HbPs	population	87	Netherlands
Marteau et al 1992	TS	both	82	UK
McConkie-Rosell et al. 2001	FX	family history	42	USA
McConkie-Rosell et al. 2000	FX	family history	42	USA
McConkie-Rosell et al. 1997	FX	family history	28	USA
Newman et al. 2002	CF	family history	299	USA
Pastore et al. 2008	FX	family history	28	USA
Watson et al. 1992	CF	population	285	UK
Williams et al. 1997	CF, DMD, FX	family history	34	USA

Note: Family history = Participants with a family history of the condition; Population = Participants identified from the general population; FX = fragile X; CF = cystic fibrosis; H = haemophilia; CGD = chronic granulomatous disease; DMD = Duchenne muscular dystrophy; BMD = Becker muscular dystrophy, SMA = spinal muscular atrophy; HbPs = haemoglobinopathies.

2.7.1 Major themes

A number of overarching themes were identified. The most prominent were anxiety, guilt, relief, effect on self image, active coping mechanisms, impact on reproductive issues and disclosure of test results (Table 2.4).

Table 2.4 Themes by mode of inheritance

Themes	Autosomal recessive		X linked	
	Family history	Population	Family history	Population
Anxiety dissipated by 6 months	Callanan 1999 Cheuvront 1998 Newman 2002	Bekker 1994 Henneman 2002 Honnor 2000 Lakeman 2008 Watson 1992	Anido 2005* McConkie-Rosell 2001	Anido 2007
Guilt	Williams 1997*		Anido 2005* Dunn 2008* James 2006* McConkie-Rosell 1997* Williams 1997*	
Altered perception of health	Fanos 1995 Marteau 1992*	Bekker 1994 Henneman 2002 Marteau 1992	McConkie-Rosell 2000	
Evidence of self-stigmatisation		Gordon 2003	James 2006* McConkie-Rosell 2000 McConkie-Rosell 1997*	
Evidence of social stigmatisation		Gordon 2003	McConkie-Rosell 2001	
Use of active coping mechanisms			Anido 2005* McConkie-Rosell 2001	Anido 2007
Impact on reproductive plans			Anido 2005* Dunn 2008* McConkie-Rosell 1997*	Dunn 2008*
No impact on reproductive plans	Callanan 1999	Henneman 2002 Watson 1992 Lakeman 2008		Anido 2007
Disclosure of test results to family	Williams 1997*	Henneman 2002 Watson 1992	McConkie-Rosell 1997* Williams 1997*	Anido 2007

*Note: Family history = Participants with a family history of the condition; Population = Participants identified from the general population; * = cohort included participants with affected children*

2.7.2 Anxiety

Two categories of anxiety emerged; one related to testing and the other related to child health. In relation to testing, all longitudinal studies investigating patient anxiety over time either found no significant difference in anxiety between carriers and non-carriers (Honor et al., 2000), or found that any anxiety experienced by carriers upon first receiving their test result had, for the vast majority, dissipated by six months as assessed by the STAI (Bekker et al., 1994, Callanan et al., 1999, Cheuvront et al., 1998, Lakeman et al., 2008, Watson et al., 1992), the Fragile X Visual Analog Scale (VAS) (McConkie-Rosell et al., 2001), or qualitative interviews (Anido et al., 2007, Anido et al., 2005).

Carrier anxiety dissipated for a number of reasons. Watson et al. (1992) found that the provision of written information and genetic counselling was helpful for most participants (92% and 97%, respectively). Bekker et al. (1994) found that the passage of time appeared to dissipate anxiety. Gender was also an issue discussed in relation to anxiety, in a number of studies. Newman et al. (2002) and Henneman et al. (2002a) found that women reported higher anxiety than men while waiting for their test results (mean = 16.5 and 14.6, respectively on the STAI in the Newman study, $p < 0.001$; and 24% versus 13%, $p < 0.001$ measured on a five-point Likert scale in the Henneman study); however there was no significant difference between the genders once the test results had been received. Lakeman et al. (2008) found that Western participants generally reported lower levels of anxiety compared with non-Western (defined as those who do not have their ancestry in Europe, e.g. people from Turkey, Surinam, Morocco and the Netherlands Antilles, who form the four largest immigrant groups in the

Netherlands) participants (General Linear Model analysis at 4 time points, $p < 0.001$).

Anxiety did however appear to be an issue for both carrier and non-carrier siblings of people with cystic fibrosis, in the interview-based study conducted by Fanos et al. (1995b). Identified carriers and non-carriers were equally likely to have moderate or severe anxiety around their child's health. Forty-one percent had had their children sweat-tested to rule out the condition, nine percent had had their child tested for carrier status, and 55% planned to do so before their child reached 18 years of age. Siblings who had had their children sweat-tested or tested for carrier status were equally divided between those who knew their own carrier status and those who did not.

2.7.3 Guilt

Guilt was a prominent theme in the data. Feelings of guilt associated with carrier status were cited as findings in five studies. These results were identified through interviews (Anido et al., 2005, McConkie-Rosell et al., 1997, Williams and Schutte, 1997), an open-ended questionnaire (Dunn et al., 2008), a VAS (James et al., 2006, McConkie-Rosell et al., 1997) and the guilt subscale of the Multiscore Depression Inventory (sMDI) (James et al., 2006). Guilt is also an issue found to be closely associated with gender, mode of inheritance, and whether the participants had affected children. In the study conducted by Dunn et al. in which 81% of respondents had a son with haemophilia, 18 of 48 (38%) female carriers reported the timing of testing as negative. Reasons cited for the timing being negative included feeling blamed by their partner and a prolonged sense of guilt. James et al. (2006) found that mothers who were carriers of X linked

conditions felt substantial guilt and self-blame related to their child's condition. When measured on the Visual Analog Scale, mothers of children with X linked conditions had significantly higher levels of guilt than mothers of children with recessive conditions ($p < 0.01$) and were more likely to blame themselves ($p < 0.001$). A similar finding was identified in the Williams and Schutte study (1997), in which it was found that many of the participants who expressed feelings of grief and guilt were women who were carriers of fragile X or Duchenne muscular dystrophy. Anido et al. (2005) also found that in families affected by fragile X, even those women without affected children experienced guilt to some extent, by virtue of the condition being in the family.

In one study (Gordon et al., 2003) there was no significant difference evident between carriers and non-carriers on the 'guilt' scale (as measured by the HOS). The participants were from the general population, were screened for cystic fibrosis, and did not have a family history of the condition.

2.7.4 Relief

Anido et al. (2005), McConkie-Rosell et al. (1997) and Lakeman et al. (2008) all identified that relief was an emotion experienced by carriers. In the study conducted by Anido et al. reactions of relief were expressed equally as strongly as reactions of guilt, with nearly all carriers expressing this emotion during interviews. For these individuals, finding out their carrier status was an inevitable result of finding a diagnosis for their child. Similarly, in the study by McConkie-Rosell et al., participants' responses indicated that while they felt angry or depressed about their carrier status, there was an *"emotional relief in finding out the cause of the mental retardation in the family"* (p. 65). Lakeman et al. found that 68% of

participants, including seven out of ten carriers, felt relief one week after receiving their test results, as measured on a structured questionnaire assessing emotional outcomes.

2.7.5 **Effect on self-image**

Three main issues arose within this theme: perception of health, self-stigmatisation and social stigmatisation.

Perception of health

Of the seven studies in which perception of health was measured, findings from three studies indicate that some carriers believed their current or future health to be significantly poorer after learning their carrier status (Fanos and Johnson, 1995b, Henneman et al., 2002a, Marteau et al., 1992). Seven out of 17 carriers (41%) in the study conducted by Henneman et al. felt less healthy (measured on a multiple-choice questionnaire) due to their test results, despite being informed both verbally and by letter that their carrier status would have no effect on their own health. Marteau et al., also using a multiple-choice questionnaire that measured perceived health from three time perspectives, identified that carriers of Tay-Sachs held the least optimistic view of future health compared with non-carriers and the control group ($p < 0.01$) and Fanos and Johnson reported that during interviews, sibling carriers retrospectively redefined health problems as related to cystic fibrosis, although the authors do not report how many.

Authors of four studies found that perception of health did not alter after learning one's carrier status, using measures such as the TSCS (McConkie-Rosell et al., 2000), a multiple choice questionnaire (Bekker et al., 1994), the HOS (Gordon et al., 2003) and a five point Likert-scale (Lakeman et al., 2008). However, both

McConkie-Rosell et al. and Bekker et al. do provide some anecdotal evidence to suggest that carriers might attribute previous health problems to their carrier status. McConkie-Rosell et al. found that 12% of participants at Time 1 and 20% of carriers at Time 2 reported feeling they had mild clinical features of fragile X. They felt that perhaps if they were carriers it would explain why they had to "study hard in school" (p. 340). A participant in the Bekker et al. cohort wondered whether her allergies and chest colds were in some way linked to her carrier status. Therefore, even though perception of health did not alter when measured quantitatively, during qualitative interviews there were some indications that it did in fact occur in a small number of cases. In fact, in the case of fragile X, it is possible that carriers did experience a mild manifestation of the disease due to skewed X-inactivation (Skirton et al., 2005). Furthermore, this finding may also be attributable to the repeat length itself which appears to be associated with toxicity due to elevated mRNA levels (Koldewyn et al., 2008).

Self-stigmatisation

There is evidence from four studies to indicate that self-stigmatisation occurred in carriers to some extent (Gordon et al., 2003, James et al., 2006, McConkie-Rosell et al., 1997, McConkie-Rosell et al., 2000). Gordon et al. identified that carriers experienced less positive feelings; more afraid, worse, weaker, less relieved, less happy, more marked (although the authors do not explain what is meant by this) and angrier, compared to those who tested negative, on the HOS. Similarly, James et al. found that carrier status is associated with stigma and is significantly associated with mode of inheritance using the same scale. The only other study (Pastore et al., 2008) specifically looking at stigma using the HOS consisted of just one carrier, and therefore findings were not significant. Stigma

was also evident in two of the qualitative studies. Just under half (9/19) of the fragile X carriers in one study (McConkie-Rosell et al., 1997) indicated that there had been a negative change in the way that they viewed themselves. One reason cited for this change was a "*feeling of being abnormal or inferior*" (p. 64), a statement indicative of self-stigmatisation.

Social stigmatisation

Evidence of social stigmatisation was evident in two studies, one quantitative (Gordon et al., 2003) and one mixed methods (McConkie-Rosell et al., 2001). Gordon et al. found that carriers and non-carriers attributed significantly more negative feelings to CF carrier status than non-carrier status. This finding was significant for all emotional scales on the HOS ($p < 0.001$).

2.7.6 Active coping mechanisms

Use of active coping mechanisms was identified in five out of a possible seven studies in which qualitative research techniques were employed. These studies included participants from the general population without affected children (Anido et al., 2007) and participants with a family history (Anido et al., 2005, McConkie-Rosell et al., 1997, McConkie-Rosell et al., 2000, McConkie-Rosell et al., 2001). McConkie-Rosell et al. (2001) found no change in the level of distress or perceived seriousness of fragile X when women were at risk of being a carrier as when they were found to be carriers. The increase in perception of seriousness only occurred in the non-carriers when the threat was no longer present. This possibly indicates that 'threat minimisation' was used by the participants as an active coping mechanism in both situations. McConkie-Rosell et al. (2001) also found, during in-depth interviews, that 11 out of 20 (55%) carrier women used

spontaneous coping statements such as "life goes on" (p.41) and "if I am, I am... I'll deal with it" (p.41). Coping behaviour statements were also evident during interviews in the study conducted by Anido et al. (2007).

For carriers identified in the study by Anido et al. (2007), most appeared to be considering their carrier status over the course of the interview, having not given the subject much thought previously. This attitude is consistent with the coping mechanism known as 'just-in-time' learning, as described in Adult Learning Theory (Wlodkowski, 1999), wherein adult learners process information which is relevant and applicable to them at the time they need it.

2.7.7 Impact on reproductive issues

The impact of carrier status on participants' views on reproductive issues varied depending on their life stage, their views on prenatal testing and abortion, whether their partners were also carriers, and whether they were carriers of an X linked or recessive condition. Authors of four studies (Callanan et al., 1999, Henneman et al., 2002a, Lakeman et al., 2008, Watson et al., 1992) of CF carriers identified from both high risk groups and the general population who did not have affected children all reported that the majority of carriers showed no change in reproductive plans after testing, as measured on questionnaires which included multiple-choice options (Callanan et al., 1999, Watson et al., 1992) or a five point Likert-scale (Henneman et al., 2002a, Lakeman et al., 2008). Reasons given included the availability of prenatal diagnosis (Henneman et al., 2002a, Lakeman et al., 2008, Watson et al., 1992) and having completed their families (Watson et al., 1992). Furthermore, in two of the studies (Cheuvront et al., 1998, Henneman et al., 2002a), only couples in which only one was a carrier were included. If one

partner tests positive and the other negative, the risk of having a child with CF is approximately 1 in 640 (Watson et al., 1992).

However, in two interview-based studies, females carrying X linked mutations, many of whom were mothers of affected children, were more likely to indicate their carrier status had caused a change to their reproductive plans (Anido et al., 2005, McConkie-Rosell et al., 1997). In the study conducted by McConkie-Rosell et al. (1997) 19 out of 28 (67%) fragile X carriers stated that they would not have any more children because of their carrier status, and 25 out of 28 (89%) would have either reduced the size of their families or not had any biological children, if they had known earlier. Anido et al. also found through in-depth interviews that many women with fragile X children stopped planning to have more children after receiving their test results. Furthermore, those without affected children expressed a strong desire "*to figure out a way to end it with me*" (p. 301). Dunn et al. (2008) also reported findings from open-ended questions that revealed some respondents felt they might not have had as many children if they had known their carrier status earlier.

Findings differed however, in the study conducted on fragile X carriers identified from the general population (Anido et al., 2007). Many carriers expressed that although the information could be relevant in the future, it was not relevant at this stage of their lives in terms of family planning. Some had not really considered the implications for family planning and their thoughts about prenatal testing, but for those that had, carrier status did not have an apparent effect on their attitudes about termination. The issue of premature ovarian failure appeared to be more prominent than the risk of having children affected with fragile X.

2.7.8 Disclosure of test results and family relationships

In six studies in which disclosure of test results was assessed, the researchers found that participants did share their test results with others, although this disclosure was selective (Anido et al., 2007, Dunn et al., 2008, Henneman et al., 2002a, McConkie-Rosell et al., 1997, Watson et al., 1992, Williams and Schutte, 1997). Anido et al. found that providing information to partners primarily depended on the seriousness of the relationship. Watson et al. found that 89% (47/53) of CF carriers informed their partners of their test results, 83% told their parents, 82% their siblings and 48% told other relatives. Henneman et al. reported that most CF carriers shared the information with parents and siblings. All but one of the carriers whose parents were still alive had told them about their test results. Ten carriers had shared the information with their brothers and sisters, but two had not. With respect to participants who did not disclose carrier information to other family members, their reasons included not wanting to disclose results to relatives who had affected siblings, and not wanting to cause feelings of guilt (Williams and Schutte, 1997).

The effects of sharing information about one's carrier status with a partner and/or family members varied across the studies. Positive experiences related to disclosure of test results were documented by Dunn et al. (2008) and McConkie-Rosell et al. (1997). Of the 18 carriers who indicated in a change in their relationship with their husband in the McConkie-Rosell et al. study, 13 carriers (72%) indicated this change had been positive. Seventeen (61%) felt that there had been an improvement in their relationship with their siblings. Difficult or distressing experiences were highlighted in three studies (Dunn et al., 2008, McConkie-Rosell et al., 1997, Williams and Schutte, 1997). Dunn et al. and McConkie-Rosell et al.

identified a negative effect on the relationship with the partner in 13/31 (42%) and 5/18 (27%) of cases, respectively. Reasons cited included anxiety and anger from the male partner (Dunn et al.) and feeling blamed by their spouse (McConkie-Rosell et al.). In cases where the experience had a positive effect on the relationship (in 4/31 and 13/18 of cases respectively), the carrier felt completely accepted by her partner (Dunn et al., 2008) and there was an increase in understanding and communication. Henneman et al. (2002a) found the majority of participants (98%) perceived no impact of carrier testing results on the relationship with their partner. For the majority of participants in the Anido et al. (2007) study, providing information about fragile X carrier status to family members was not problematic. However, providing the information to partners depended on the seriousness of the current relationship.

2.8 Discussion

This review is useful in that it identifies a number of factors that appear to influence the emotional consequences of carrier testing. These include population group, whether the carrier has an affected child, stage of life, psychological coping mechanisms, and mode of inheritance. In this respect the results of this systematic review provide some interesting insights into how genetic testing for different conditions may have a varying psychological impact that is dependent on the context in which testing occurs.

Anxiety, an emotion frequently measured in studies investigating the impact of carrier testing on individuals, dissipated in the long term for the majority of participants in all studies. In addition to the reasons suggested by authors which include the passage of time and the provision of written information, another

reason may have been because none of the participants were pregnant at the time of receiving their carrier test results and were therefore not anxious about the possibility that the fetus was affected. For carriers, knowledge that reproductive options were available to them if there was a risk of having an affected child may also have overridden any initial anxiety. Furthermore, good quality genetic counselling services may have lessened the impact of the test results.

Variables including mode of inheritance, gender and whether the carrier already had a child affected by the condition appear to be strongly linked to the issue of guilt. The finding that guilt was more dominant in women than men, indicates that it may be strongly connected with what Peters and Jackson (2009) describe as a unique emotion concerning a mother's relationship with her affected child. Guilt also appeared to be more commonly reported by mothers of children with X linked conditions. One possible explanation lies in the close association of guilt and blame. In the case of X linked conditions, it only takes a carrier mother to pass along an X linked condition rather than having both parents contribute the "faulty" gene. Therefore, the burden of having passed on a faulty gene cannot be shared with a partner. In these cases men may externalise their emotional response to devastating news and blame, while women are likely to internalise their responses and to accept this blame (Lewis, 1998). Mothers are also more likely to self blame (Peters and Jackson, 2009).

Guilt may also be an emotion linked to family history. Anido et al. (2005) found that women who did not have affected children but had the condition in the family, expressed feelings of guilt, which may indicate a form of 'survivor guilt'. Survivor guilt has also been identified in CF families. In a study (which was excluded from this review as it contained women who were pregnant at the time

of testing) in which barriers to carrier testing for adult CF siblings were identified, carrier status served an important function in binding guilt, with 15% of siblings either hoping they were carriers or feeling guilty they were not (Fanos and Johnson, 1995a).

The issue of guilt has also been identified in other areas of healthcare, *highlighting that it is not an emotion uniquely linked to conditions that are inherited*. For example, guilt has been identified as a factor in studies of children with diabetes. Bowes et al. (2009) found that guilt, as well as grief and anger, were expressed by both mothers and fathers of children with type 1 diabetes. Similarly, in a study conducted in Finland, guilt was identified as key emotion of parents of diabetic children in the immediate period after the diagnosis (Seppanen et al., 1999).

There are conflicting results in the literature regarding the issue of perceptions of health. Yet even those studies in which carriers did not indicate feeling less healthy on surveys or questionnaires, during in-depth interviews, some participants reported clinical features of the disorder for which they were being tested or for which they were found to be a carrier. In the case of fragile X, it is possible that carriers did experience a mild manifestation of the disease due to skewed X inactivation (Skirton et al., 2005). Furthermore, this finding may also be attributable to the repeat length itself which appears to be associated with toxicity due to elevated mRNA levels (Koldewyn et al., 2008). However this finding also suggests that participants may have been seeking support for beliefs they held about themselves.

In interpreting this finding, McConkie-Rosell et al. (2000) refer to the theory of self-concept as described by Shavelson et al. (1976). Shavelson et al. hypothesize that self-concept is hierarchical, with perception of personal behaviour in specific situations at the base of the hierarchy, inferences about the self in broader domains (e.g., social, physical) at the middle, and a global, general self-concept at the apex. Global self-concept is stable, but as one descends the hierarchy self-concept becomes increasingly situation specific and less stable. Seeking clinical features related to actual or possible carrier status might be indicative of situation-specific changes in feelings about self. Additionally, it may be the case that scales such as the HOS (used by Gordon et al., 2003) and TSCS (used by McConkie-Rosell et al., 2000) are not sensitive enough to detect the subtleties concerning how carriers perceive their own health, which are more likely to be expressed during in-depth interviews.

Reproductive intent also appeared to be closely linked to mode of inheritance, stage of life and whether the participant already had an affected child, with the greatest impact being identified for carriers of X linked conditions with affected children. This group was most likely to refrain from having more children. One possible reason involves the documented psychological difficulties of raising a child with fragile X (Abbeduto et al., 2004, Lewis et al., 2006). When Anido et al. (2007) interviewed fragile X carriers who did not have affected children and were from the general population, the information did not appear to have an impact on family planning with many not having considered the issue. This is likely to be because they did not have any experience, either themselves or through other family members, of raising a child with the condition. It may be that these carriers would experience increased distress as they consider reproduction

more seriously. Similarly, carriers of CF in the general population did not change their reproductive plans as a result of their carrier status. Participants in these studies did not have affected children, and even as a carrier, there would only be a risk to future children if the partner was also a carrier.

McConkie-Rosell (2000) suggests that knowledge that one is a carrier causes a barrier to having biological children or grandchildren and hence is a threat to the parental role. In the case of X linked conditions, this threat may be more accentuated for carriers than it is for carriers of autosomal recessive conditions because there is a more immediate risk (due to the fact that only one partner needs be a carrier), particularly if the carrier has not yet begun or completed her family. It could also be the case that women in X linked families are more likely to know affected relatives, because there are likely to have been family members affected across several generations. This may in turn impact on their perception of the severity of the condition and hence the threat may be more immediate. For carriers of recessive conditions the threat is less acute because the risk only exists if the partner is also a carrier of the same condition.

Active coping mechanisms, such as "threat minimisation," significant changes to reproductive intentions and the use of active coping statements, were identified in those participants at an increased risk of carrying the fragile X gene. Lazarus and Folkman (1984) describe coping as consisting of two different strategies, problem-focused coping and emotional-focused coping. The findings from this systematic review suggest that women at high risk of being a carrier of fragile X engaged in problem-focused coping by managing their health threat through genetic testing, and if found to be carriers, by changing their reproductive intentions. They engaged in emotional-focused coping through threat

minimization and active coping statements. Similar coping mechanisms have been found in other studies looking at the psychosocial impact of waiting for or receiving genetic test results. Men and women interviewed in a qualitative study about the impact of genetic risk information for hereditary breast cancer (d'Agincourt-Canning, 2006) revealed that genetic information was 'enabling' and allowed participants to take proactive measures to confront the disease. In a similar study, Lim et al. (Lim et al., 2004) identified that some participants reported discovering their hereditary breast cancer mutation status as a positive life-changing experience. Participants who took part in a study (Gooding et al., 2006) in which susceptibility testing for Alzheimer disease was conducted associated their motivation to partake in genetic testing with a preference to seek information about health threats in order to feel in control of their health. These studies highlight that coping strategies are used to reduce distress by people undertaking genetic testing, across the genetic testing spectrum.

In addition to these coping strategies aimed at lessening distress, Lazarus and Folkman (1984) describe a smaller group of cognitive strategies directed at increasing distress. For some individuals, there is a need to feel worse before they can feel better. Self-blame, a coping mechanism found to be used by carriers of X linked conditions, is one such form of self-punishment individuals may use. This deliberate emotional distress may mobilise individuals into action. Evidence that women use self-blame as a coping strategy has been identified in other studies; for example, self-blame was found to be significantly correlated with both problem-focused and emotional-focused coping strategies in a study of patients with diabetes (Tuncay et al., 2008). Self-blame was also used as a strategy to cope

with depression in a study of how primary care patients manage their illness (Brown et al., 2007).

Other studies, in which participants became aware of their carrier status through family history or newborn screening, have identified similar psychosocial issues to those in this review. Fanos et al. (1999) recognized a number of coping mechanisms used by parents of children with *ataxia-telangiectasia*, including rationalising their child's condition as a "statistical quirk" (p.417), and imbuing the occurrence with meaning and significance through connecting it with the wider sphere of human suffering or to the spiritual world. Guilt was not however a common finding in their study, and surprisingly when it was mentioned, it was in reference to fathers. Undue concerns about the health of carriers was also identified in a minority of parents in a study assessing the impact of carrier status information following newborn screening (Kai et al., 2009), as was a sense of responsibility to share carrier status information with extended families. Stigmatisation was also evident in a study which included participants from high risk CF families who did not want to learn their carrier status (Fanos and Johnson, 1995a). For example, one untested woman was worried that she would be "less desirable" (p. 88) to men if they knew she was a carrier.

While this review provides an overview of the psychosocial experience of living as a carrier, it is important to keep in mind the limitations of making comparisons across different conditions, in particular CF and fragile X (the conditions included most often in this review). These two conditions vary greatly in terms of their effects on the affected individual, the implications for the health of the carrier, and risk of the carrier having an affected child. Furthermore, variations in study design, the different population subsets compared, and the obvious

complexities of comparing qualitative and quantitative data, mean that the findings should be interpreted with some degree of caution. For example, there were indications from some studies that validated scales detected no changes in perception of health. However, when the authors used in-depth interviews, changes in health perception were evident (Bekker et al., 1994, McConkie-Rosell et al., 2000). Some authors used the STAI to measure anxiety, whereas others using qualitative methods relied on participants' own terminology. Studies using the HOS were much more likely to identify evidence of stigmatisation than those that did not use this scale, as this scale specifically measures aspects of self image. Future systematic reviews may therefore benefit from the inclusion of samples involving population groups which are more similar in kind in terms of risk to offspring, severity of the condition or family history. Future research studies may be better synthesised if the studies focus on using similar groups of patients and validated tools.

Yet this does not necessarily mean the findings of the present review fail to provide valuable insight into the psychosocial experience of living as a carrier. In particular, the review provides an overview of the commonality of experiences across conditions with different inheritance patterns. Furthermore the overview identifies a number of issues that collectively apply to carriers as a group, because of the familial nature of genetics.

2.9 Strengths and Limitations of the Review

As stated previously, findings from the review should be considered in light of the difficulties and limitations of combining studies undertaken with different study designs, subsets of the population, measures and outcomes. These factors

may have diluted the strength of the comparisons. Furthermore, many of the studies lacked theoretical models or presentation of a conceptual model to help place the variables and their possible interactions in context (Henneman et al., 2002a, Pastore et al., 2008, Watson et al., 1992). Such omissions possibly weaken the validity of the results. Nevertheless, in the present systematic thematic analysis, the findings were able to be explained within established theoretical models of coping and self-concept (Lazarus and Folkman, 1984, Shavelson et al., 1976).

The systematic review does have notable strengths. Seven databases were used to retrieve studies to maximize the chance of finding all relevant research. In addition, several iterations of the search were conducted using different combinations of keywords, to ensure the search was rigorous. At the present time there does not appear to be another systematic review in the literature that compares the psychosocial experience of carrier testing for autosomal recessive and X linked conditions; thus, this review provides unique and useful information.

2.10 Conclusion

The findings from this systematic review provide insight into the variety of psychosocial emotions experienced by individuals undergoing carrier testing and the commonality and differences of experience across different condition types and inheritance patterns. Moreover, the findings contribute a general overview of the psychosocial impact of living as a carrier. Prominent themes that occur in the literature include anxiety, guilt, relief, effect on self image, active coping mechanisms, impact on reproductive issues and disclosure of test results. Variables that influence the psychosocial effects of carrier testing include whether the carrier has an affected child, mode of inheritance, genetic counselling and life

stage. In the next chapter, I will describe the method used to conduct my own research looking at the psychosocial impact of genetic testing.

Table 2.5 Summary of papers

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Anido et al. 2007	To explore the attitudes towards FX mutation carrier testing from women identified from the general population.	Qualitative in-depth interviews.	8 women from the general population, of reproductive-age, who had been identified as FX carriers. Aged 21-44.	Semi-structured qualitative in-depth interviews. Topic guide consisted of 28 questions covering topics including testing experience, premature ovarian failure, affect of information on relationships and family planning.	interpretative phenomenological analysis. Recurring primary patterns in data noted. After independent analysis, team members compared their assessment to discuss clarification and development of themes.	Women were wholly unprepared for positive carrier results. For many carriers the information was not relevant at this stage of their lives in terms of family planning and personal relationships. Many expressed the information could be relevant in the future. For majority, providing information to family was not problematic. Providing information to partners depended on seriousness of relationship. Resulting information came as a surprise but for most women was put quickly out of mind.	

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Anido et al. 2005	To identify issues related to carrier testing and population screening for premutation carrier women.	Qualitative focus groups.	General population and women from families with FX from Atlanta, aged 28-50. 40 focus group participants. 10 women per focus group.	Focus groups were audio and video recorded. Questions included: How would you describe your first reaction to getting results? Did your test result cause you to take any action or make decisions about your life plans? Did your result make you feel anything new about yourself?	Thematic analysis. Primary patterns noted and classified into themes. Data analysed independently then findings compared. Themes then compared to existing literature to determine novel findings.	Nearly all carriers from FX families reported some sort of guilt experience. Reactions of relief expressed equally strongly – relief for carriers in terms of finding diagnosis, and relief for non-carriers. Anxiety dissipated either immediately or over a few months. Carrier status led to reconsideration of life plans. 'Grandmother guilt' experienced by several carriers. Timing of carrier testing with respect to a woman's life stage and views on abortion dictates whether the information on carrier status will be seen as beneficial or detrimental.	

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Bekker et al. 1994	To evaluate the short term effects of population based screening for carriers of CF.	Mixed methods longitudinal study. Self completed postal questionnaire completed at 3 time points.	Adults between 18-45 years registered with a general practitioner in Inner London. 5,529 adults approached (5529). 957 patients tested (637 females 320 males) Full data received from 427 with negative results and 14 carriers.	Questionnaires at 3 time points: before testing, upon receiving results, three months later. STAI used to assess anxiety. 6 interviews with carriers 6 months after receiving test results.	MANOVA univariate analysis. No information on how interview data were analysed.	Receipt of results had no effect on perceptions of health. Those who received a positive result were significantly more anxious upon receipt but by 3 months this anxiety had dissipated. Main problem of population carrier screening would be false reassurance as opposed to anxiety.	Does not address potential confounding variables. No information about method used to analyse data. No justification of sample size.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Callanan et al, 1999	To identify risk perceptions, psychological status and reproductive plans of carrier by non carrier couples.	Randomised controlled trial comparing home education and testing with clinic education and testing.	CF carrier testing offered to 120 partners of previously identified carriers of individuals with CF. 57 partners were tested. Participants: 18+ years, not pregnant, living in study inclusion area (N. Carolina). Participants randomly assigned to either clinic or home screening.	Survey during interval between partners' testing and receiving results six months later. STAI used to measure anxiety and personality trait. Positive and negative affect measured using PANAS.	Paired comparison t test. Descriptive statistics used to summarise data.	Both relatives and their partners showed slightly higher anxiety scores while waiting for partners test results, although within 'normal' anxiety range. No significant differences between the relative and partner at either time point. Both showed decrease in anxiety score six months after completion of testing. 89% of relatives reported no change in reproductive plans.	Does not address potential confounding variables.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Cheuvront et al., 1998 (same cohort as Newman 2002)	To look at psychosocial and knowledge outcomes of two different approaches to CF gene pre-test education and carrier testing.	Randomised controlled trial comparing home education and testing with clinic education and testing. Secondary analysis comparing carriers and Non-carriers.	299 accepted offer of free education and testing, aged 18+ years (majority between 26-45 years). Participants were relatives of people with CF living in N. Carolina. Participants randomly assigned into 2 groups before being contacted: those assigned to the clinic (91) and those assigned to education and testing at home (208).	Participants completed a baseline telephone interview, and completed a questionnaire at 2 time points: while waiting for their test results and immediately after learning their test results. STAI used to measure anxiety.	Data analysed using SUDAAN software for clustered samples. Value of $p < 0.05$ was considered significant.	No significant differences in positive or negative affect or anxiety while waiting for results or after results were known as a result of where person had been educated and tested. No statistically significant differences on any of the outcome measures based on carrier status (anxiety, positive and negative affect and satisfaction with education and testing arrangements)	Mainly focuses on comparing knowledge, anxiety and patient satisfaction with education and testing at 2 different testing settings (home or clinic). Not much information on the psycho-social consequences of genetic testing itself. Does not address potential confounding variables. No justification of sample size.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Dunn et al, 2008	A descriptive study which aims to report adult carriers' and their husbands/partners' experiences of carrier diagnosis for haemophilia A and B.	Cross sectional study. Self completed postal questionnaire	Carriers registered on the Katharine Dormandy Haemophilia Centre database at the Royal Free, London. 235 carriers identified aged 18-65 and sent introductory letter. 66 carriers responded. Median age 42. 76% were in a relationship and 78% of their husbands/partners completed the questionnaire.	Questionnaire influenced by Systematic Theory. Included yes/no, multiple choice, example scenario, ranking and open ended questions.	Most of the data analysis was descriptive; where appropriate a Student's t-test or a Mantel-Haenszel chi-squared test was used and a value of $p < 0.05$ was considered statistically significant.	Carriership was considered an issue by both groups at 2 key points: 34% once their relationship had become serious and 39% once their son had been diagnosed with haemophilia. 38% of carriers said the timing of the test had a negative effect on them. Reasons cited were: sense of shock and grief, concern about having children, feeling blamed by their partner and a prolonged sense of guilt. Where timing was seen as positive (22%) it was because of feeling completely accepted despite the diagnosis. 42% reported a negative effect on the husband/partner, 13% reported a positive effect. Self-assessments of both carriers and husbands/partner showed the effects had mostly been negative or neutral.	No justification of sample size.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Fanos and Johnson 1995	To explore levels of understanding and feelings about carrier status and genetics of CF in affected families.	Mixed methods study. Qualitative interviews and questionnaire to measure anxiety and depression and knowledge of CF.	84 individuals interviewed including 54 adult CF siblings and 30 spouses. Of the 54 interviewed siblings, 45 had been tested and 9 had not at the time of the interview. Participants ranged in age from 18-55 years, 26 were male siblings and 28 female siblings. They were recruited through the Genetics Division at Children's Hospital, Oakland, US.	Semi-structured, face to face interviews including: What was your reaction to the results? Were they what you had expected or different? Scales were developed for various categories capturing important aspects of family functioning and psychosocial adaptation. Questionnaires included anxiety and depression scales from Hopkins checklist.	Inter-rater reliability obtained for interview codes. Hypotheses analysed using X ² procedure. Relationship between guilt and sibling resentment explored through a Pearson's correlation.	53% of CF siblings assumed they were carriers before testing and were neither upset nor surprised by positive results. Those with a negative result were relieved. For those that did not assume they were carriers, a positive result did not come as a shock as they knew there was a strong possibility they were. Identified carriers and non-carriers were equally likely to have moderate or severe anxiety around their child's health.	Much of the data related to siblings as a whole, some of whom had not been tested and therefore we could not use this data. There was only a small amount of data that specifically addressed the psychosocial impact on tested individuals.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Gordon et al, 2003	An assessment of CF knowledge and emotional consequences of CF population testing 18 months after screening was offered. (results of initial screening and 3-6 month follow-up reported in Honnor et al. 2000)	Cross sectional study. Self completed postal questionnaire survey.	English speaking individuals aged 18-50 years who presented (for visits unrelated to pregnancy) to either Family Planning Clinic or one of 20 GP practices in Perth, Western Australia. 353 (59.5%) responded to questionnaire.	18 months after testing follow-up questionnaires were sent to a consecutive sample. Questionnaire contained the HOS.	Significance level for t-test set at 0.001	Carriers felt more afraid, worse, weaker, less relieved, less happy, more marked, and angrier than test-negative individuals. However there was no difference on the guilty, ashamed, ability, activity and health scales. No difference was found between carriers and non-carriers relating to feeling guilty, ashamed, ability, activity and health scales. Carriers did not have a poorer perception of their current health than non-carriers. Carriers described more positive feelings about themselves when they considered their own result compared to feelings they attributed to most carriers - 'unrealistic optimism'. Non-carriers tended to be slightly more negative about 'most carriers' than carriers.	No justification of sample size.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Henneman et al. 2002	Impact, understanding of test results and satisfaction among participating couples in a preconception CF carrier screening project.	Longitudinal survey. Self completed postal questionnaire at 3 time points.	Recruitment of individuals aged 20-35 years, through GP or Municipal Health Service 1997-2000. 559 couples gave written consent to CF testing after education. Testing was step-wise. 18 carriers identified. Partners tested negative. Response rate for completion of all three questionnaires was 17 carriers, 15 partners with negative test results and 794 other participants.	3 self-administered questionnaires at 3 Time points. Time 1 = before pretest education and counselling; Time 2 = before receiving test results, Time 3 = 6 months after receiving test results.	χ^2 or Fisher exact tests. P Values of $p < 0.05$ considered significant. Multiple logistical regression.	Carriers and partners reported no impact of the test results on their reproductive plans. 154 (19%) felt worried when waiting for results; women worried more than men. 6 months after results only 8 felt worried, 4 of whom were carriers. 7 out of 17 carriers reported feeling less healthy due to test results. 98% perceived no impact on relationship with partner. The other 2% perceived a positive influence (improvement in communication, increased certainty in having children) All carriers and 95% of the other participants would have the test again. 10 carriers shared info with their brothers and sisters, all but one had told parents. Only 2 shared info with more distant relatives.	

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Honnar et al. 2000	Acceptance of carrier testing for CF in the community when offered in a primary care setting.	Longitudinal, self completed postal questionnaire survey at 2 time points (pre and post test)	5102 individuals age 18-50 years, recruited through 20 general practices and a family planning clinic in Western Australia completed questionnaire at Time 1. 2220 (43.5%) took test, 69 carriers identified. Response rate at 3 to 6 month follow up was 58.4%.	2 questionnaires administered at 2 time points. Time 1 = before testing and both before and after receiving information about CF; Time 2 = 3 to 6 months after receiving test results. STAI used to measure anxiety at both time points.	χ^2 test used to compare proportions and odds ratios (OR) and 95% confidence intervals (CI) were calculated using SPSS.	There were no significant differences between counselled and uncounselled carriers; carriers and test-negative individuals; or test-negative and untested individuals for State Anxiety Inventory scores.	

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
James et al. 2006	A survey of family members with chronic granulomatous disease and Duchenne/Becker muscular dystrophy and spinal muscular atrophy types II/III	Cross-sectional study. Self-completed postal questionnaire survey.	Recruitment of 'adults' (no age range given) was through US registries, participants in other studies and patient organisations. Consisted of 112 members of 51 families (59% response) with granulomatous disease and 96 members of 51 families with Duchenne/Becker muscular dystrophy and spinal muscular atrophy types II/III.	Cross-sectional mail survey of adults with the conditions mentioned. Included Multiscore Depression Inventory and the HOS.	X ² tests, Two-tailed <i>t</i> test Mann-Whitney Logistic and linear regressions	Mothers carrying X linked conditions were more worried about risks to future generations than mothers carrying recessive conditions. X linked mothers were more likely to feel guilty (both currently and in the past) and blame themselves. X linked fathers blamed their child's mother and X linked mothers felt more blamed by fathers. There were no differences in level of guilt or self-blame between autosomal recessive mothers and fathers. X linked family-members were more likely to consider being a carrier stigmatising.	No justification of sample size.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Lakeman et al. 2008	To study outcomes, knowledge, recall and understanding of test results, satisfaction, and reproductive intentions among 97 Western and 46 non-Western participants undergoing pre-conceptual ancestry-based carrier couple screening for CF and HbP's in the Netherlands.	Longitudinal, self completed postal questionnaire survey at 4 time points (before and after pre-test consultation, 1 week after and 3 months after receiving test results).	Screening offered to 9453 individuals (20-35 years) in Amsterdam from January-December 2005. Invitees who had a partner with whom they were planning a pregnancy were eligible for participation. It was estimated that 33% (3120) belonged to the 'target population' based on reply forms and telephone survey, 87 participated in testing, providing a total of 72 couples. 47 couples were eligible for CF screening only, six for HbP's only.	Questionnaire at 4 time points (before and after pre-test consultation, 1 week after and 3 months after receiving test results). Questionnaire included 6 item STAI, Reproductive intentions, and satisfaction.	Independent sample T-tests or ANOVA analysis were performed to compare the mean scores for the variables between groups at the same measurement moment. T-test(s) and General Linear Model-analysis for repeated measurements for longitudinal comparison. χ^2 test for statistical comparison of proportions.	Participants reported low levels of anxiety at the start, which decreased further during the study. Carriers felt anxious one week after receiving test results and only one was still anxious at the three month follow-up. Seven out of ten carriers felt relieved one week after receiving test results. None of the participants, including carriers, perceived themselves as being less healthy after receiving the test-results. 68% felt relieved at Time 3, and 62% at Time 4. 4 participants, including 2 carriers, were disappointed 1 week after receiving results at Time 3, but none were disappointed at Time 4 (3 month follow up). 4 other respondents reported feelings of disappointment at Time 4, including two non carrier partners of CF carriers. 27% of	

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
			and 19 for both disorders. CF carrier testing was carried out step-wise. For HbP's carrier testing, both partners were tested. 3 CF carriers and seven HbP carriers were identified, but no carrier couples. 110 participants completed all 4 questionnaires.			participants stated they would have considered not having more children if found to be in a carrier couple, although majority reported no change. At 3 month follow-up, 93% of all participants including all carriers, states that the test results had not changed their ideas about having children. Western compared with non-Western participants generally reported lower levels of anxiety.	
Marteau et al. 1992	A study to determine how carriers of a recessive gene for Tay-Sachs perceived their health related to non-carriers.	Cross sectional study. Self completed postal questionnaire survey.	3 groups screened for Tay-Sachs. 1 group recruited from cultural exhibition for Jewish community, another through the local synagogue, the 3 group	Questionnaire including three questions from SF-36 Health Status Questionnaire and 2 questions developed for the study. Multiple choice questions.	χ^2 test for statistical comparison of proportions.	No differences between the 3 groups in how they perceived their current health. Contrasting current health with health status 2 years previously showed no difference between the groups. Groups altered in their expectations about future health. Carriers of Tay-Sachs held the least	Does not address potential confounding variables. No justification of sample size.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
			included relatives (Jewish and non-Jewish) of previously affected children, and previously identified carriers as well as persons identified by routine screening, 27 carriers and 55 non-carriers responded to the questionnaire. Mean age = 29 years (non-carriers) and 39.5 years (carriers).			optimistic view of their future health compared to the other 2 groups. Carriers were less optimistic about their risk of developing something wrong in the future.	
McConkie-Rosell et al. 2001 (same cohort as McConkie-	A study of women at-risk to inherit the FX mutation.	Mixed methods, longitudinal study at 2 time points. Interview included	Women all had 50% a priori risk of being a carrier. Study sample = 42 Caucasian women (20	Questionnaire including Fragile X VAS developed by principle investigator and her colleagues, and	Analysis of variance, Mann Whitney U procedure, Two-tailed t test, Open ended	Being at risk was upsetting, frightening and scary. At Time 2 non-carriers reported feeling happy, relieved, grateful. Relief related to no longer having to worry about	Small sample size. No justification of sample size. Author does not assess likely impact

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Rosell 2000)		open and closed questions.	carriers and 22 non-carriers) from 17 families. Women aged 18+ years, mean age 42.3 years. 81% married and 71% had at least one child. All women had a family member diagnosed with the condition.	structured interview with open and closed ended questions including: How do you feel about your carrier status?	interview data analysed using thematic analysis.	their children or grandchildren's risk. Carriers said they were upset and concerned, mainly for their children, grandchildren or own reproduction. 55% used active coping statements in discussing feelings about being a carrier. Possible 'survivor guilt' reported by 1 non-carrier. Significant difference between carrier and non-carrier at Time 2 was result of changed perceptions of the non-carrier women. Women who were carriers were equally as upset learning their at-risk status as learning they were in fact carriers. Carriers said initially learning carrier status was upsetting. Carriers reported an improvement in level of upset at Time 2. At Time 2 non-carriers viewed condition as more serious than at Time 1.	of their own characteristics on data.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
McConkie-Rosell et al. 2000 (same cohort as McConkie-Rosell 2001)	A study to explore self-concept of women at risk for inheriting the FX mutation.	Mixed methods, longitudinal study at 2 time points (before testing and 6 months after). Interview included open and closed questions.	Women all had 50% a priori risk of being a carrier. Study sample = 42. Caucasian women (20 carriers and 22 non-carriers) from 17 families. Women 18 years +, mean age 42.3 yrs, 81% married and 71% had at least one child. All women had a family member diagnosed with the condition.	Questionnaire including Tennessee Self-Concept Scale, Fragile X VAS. Structured interview consisting of 50 questions including: Has finding out your carrier status changed the way you view yourself?	Analysis of variance. Mann Whitney U procedure. Two-tailed t test. Open ended interview data analysed using thematic analysis.	No evidence of diminished social self related to at-risk status or to actual carrier status at Times 1 and 2 on Tennessee Self-Concept Scale: 59% of the non-carriers reported feeling better after 5 months using the Fragile X Visual Analog Scale. There was no evidence that being at risk or being a carrier altered perception of health. Some anecdotal evidence to suggest that carriers at Time 2 believed they had mild clinical features of FX. Carriers did not report feeling worse about themselves than they had reported at Time 1. The difference between carriers and non-carriers at Time 2 occurred because non-carriers felt better about themselves and not because the carriers felt worse.	Small sample size: Author does not assess likely impact of their own characteristics on data.

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
McConkie-Rosell et al, 1997	A study of obligate carriers of the FX syndrome to ascertain opinions and attitudes regarding carrier testing.	Mixed methods study using 48 question structured interview.	28 female carriers were recruited through the Fragile X Clinic at Duke University Medical Centre. All women had undergone genetic counseling and all knew they were carriers. 25 premutation carriers, 3 full mutation carriers, mean age 41.8 years, mean number of children 2.28, mean number of years since diagnosis = 5.9 years.	Structured interview with open and closed ended (multiple choice) questions at 1 time point. Questions were asked regarding family planning issues, how relatives should be told of genetic risk, and marital and family relationships. Also included an 11 item VAS.	Descriptive univariate analysis, Fisher's Exact test in the case of 2x2 frequency tables or Student's t-test for differences between means.	67% felt knowing about the condition had changed their plans about having more children. 89% felt that if they had known earlier they would have either reduced the size of their family or not have had any biological children. 82% said they would have used prenatal diagnosis. 76% said learning they were carriers had changed the way they viewed themselves: 53% in a positive way, 47% in a negative way. Subjects reported that over time there was a lessening in the intensity of the negative feelings associated with first learning carrier status. 87% indicated they did not feel guilty, however were sometimes angry or depressed and would change their carrier status if they could. 64% noted a change in relationship	

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Newman et al. 2002 (same cohort as Chevront 1998)	To assess how gender as well as carrier testing arrangements (home and clinic based) affected testing experience.	Randomised controlled trial comparing home education and testing with clinic education and testing.	299 accepted offer of free education and testing, aged 18+ (majority between 26-45 yrs). Participants were relatives of people with CF living in N. Carolina. Participants randomly assigned into 2 groups; those assigned to the clinic (91) and those assigned to education and testing at home (208).	Participants completed a baseline telephone interview, and completed a questionnaire at 2 time points: while waiting for their test results and immediately after learning their test results.	All reported regression analysis were conducted using SUDAAN software for clustered samples.	with the partner; 72% a positive change, 27% a negative change. 60% reported improvement in relationships with siblings. Women reported higher anxiety than men on the STAI administered at baseline but not at follow-up. Men tended to describe themselves with more positive adjectives than women while waiting for test results.	

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Pastore et al. 2008	A study assessing female FX premutation carriers.	Longitudinal, self completed postal questionnaire survey at 2 time points.	Female participants who had been diagnosed with diminished ovarian reserve aged 18-42 years and infertile. Women with a family history of FX were excluded, 20 women completed questionnaire at baseline and 17 who were non-carriers and one carrier completed the questionnaire at 3 months.	Self administered questionnaires at baseline and 3 months after learning test results.	Exact Mann-Whitney U, Health Orientation Scale, McNemar tests.	Perception of the seriousness of FX premutations increased 3 months after learning they were not a carrier but this did not reach statistical significance. Non-carriers thought carriers would be more angry and regretful than they felt themselves 3 months after testing. Self esteem of non-carriers was essentially unchanged 3 months after learning they were not carriers.	

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Watson et al. 1992	A study assessing the effect of screening for CF carrier status on anxiety levels, attitudes, knowledge and actions of participants.	Longitudinal, self completed postal questionnaire at 3 time points.	Participants (men and women age 16-44) recruited through primary health care services. 3176 individuals were screened identifying 100 carriers with no known history of CF. 88 carriers responded to questionnaire at Time 2. 60 carriers responded to questionnaire at Time 3. 51 carriers (85%) have responded at Time 2 and 3.	Self administered questionnaires. STAI used to measure anxiety. Time 1 = pre-test. Time 2 = 2 weeks after results. Time 3 = non-carriers after 3 months, carriers after 6 months.	Descriptive statistics were calculated.	81% of carriers said they were glad they had been tested. Carriers expressed being surprised (67%) at first. 25% said they were slightly anxious. Provision of written information and genetic counselling helpful (92% and 97% respectively). 22% were worried (some still waiting for results of partner). At 6 months, 70% reported no anxiety or depression. Those not planning further children were either 'not worried' or 'indifferent' about their results. 89% of carriers told result to partner, 83% to parents, 82% to siblings, 48% to other relatives and 63% to friends. If found to be in an 'at risk' partnership 33% would consider not having children, 42% were unsure. 78% would request prenatal diagnosis. 36% would consider termination.	

Reference	Purpose of study	Study design	Sample and size	Data collection	Methods of analysis	Results	Limitations
Williams et al. 1997	A study examining experiences (emotional and social consequences) of adults requesting carrier testing for 4 autosomal recessive and X linked disorders.	Qualitative study. Interviews conducted at 1 time point.	34 adults recruited from a genetic counselling clinic. People requesting carrier testing for CF, TS, DMD, or FX from 1993 to 1996 were informed of study by genetic nurses and counselors. Ages of participants ranged from 18-71 years, the mean age was 32. 76% were women.	Semi-structured interviews (17 face to face, 17 telephone) one month after learning test results. Questions included: Tell me about how your carrier testing turned out, positive and negative aspects of knowing their carrier status and to whom they disclosed test results.	Interviews transcribed, reviewed and coded. Each rater's codes were compared, and analysis continued until there was agreement on data codes. Codes then collapsed into more general categories.	Non-carriers experienced the freedom to look ahead regarding their own reproduction and relief from fear regarding future generations. Carriers experienced loss of hope to have children or grandchildren who would be free of the condition. Most participants shared results with some members of their families, although members of both groups experienced some difficulties regarding disclosure of results to family members and expressed uncertainty regarding disclosure to insurance providers. Many participants who expressed feelings of grief and guilt were women who were carriers of FX or DMD.	

Note. Cf = cystic fibrosis; DMD = Duchenne muscular dystrophy; FX = fragile X; HbP = hemoglobinopathies; TS = Tay-Sachs; Fragile X VAS = Fragile X Visual Analog Scale; HOS = Health Orientation Scale; PANAS = Positive and Negative Affect Scale; STAI = Spielberger State-Trait Anxiety Inventory.

Chapter Three:

Methodology

3.1 Introduction

In this chapter, I will present the method chosen to gather data for the content of the information resources. I will begin by providing a brief background to qualitative research methods, looking at their use within the healthcare setting, and justifying their use in this phase of the doctoral study. I will then present the grounded theory method, and discuss why I felt it was the most suitable method for data analysis. Finally, I will describe the study design, focusing on the recruitment process, ethical issues, and the procedure used to analyse the data.

3.2 Background

To investigate the psychosocial issues and experiences identified in the systematic review further and gather data that would help inform the content of the psychosocial information resources, qualitative research methods were considered most suitable as they enable in-depth exploration of the subject matter. Qualitative research is concerned with the meanings people attach to their experiences of the social world and how they make sense of that world (Pope and Mays, 2006). The paradigm has a number of key features that distinguish it from quantitative methods. Instead of asking questions such as 'how many' or 'how much', measurements frequently used in quantitative research, qualitative research is concerned with the 'how' and 'why' questions. These are questions which are fundamentally concerned with the nature of social phenomena, a person's lived experiences, behaviours, emotions and

feelings (Strauss and Corbin, 1998). Compared to quantitative methods, use of qualitative methods can provide a 'deeper' understanding of social phenomena (Silverman, 2001). The methods used in qualitative research are very different to those of quantitative research. Methods such as interviews, focus groups and observation are commonly used in order to obtain data as opposed to using techniques such as multiple choice questions or scales commonly used in quantitative questionnaires. Another distinguishing feature is that this method enables the researcher to study people in their natural settings as opposed to artificial or experimental ones (Mays and Pope, 1995a). People may be observed in their own homes for example, or interviewed in a way in which they are encouraged to talk freely. For these reasons, qualitative research methods were considered most useful and relevant for gaining an in-depth understanding of the psychosocial impact of carrier testing and parenting a child with an undiagnosed condition, the two areas focused on for this doctoral study.

There has been considerable debate concerning the trustworthiness of qualitative research, and how it should be judged (Meyrick, 2006). Some of the criticisms levied at qualitative research have been that it lacks scientific rigour, consists of anecdotal and personal impressions rather than valid and reliable findings and that it lacks reproducibility. It has also been said that the results of qualitative research lack generalisability, in that they only represent the experiences of a small group or within a particular setting (Mays and Pope, 1995a).

Yet these criticisms rest on the assumption that there is some sort of underlying "truth" that we are trying to attain through conducting social research, a position rejected by the anti-realists. Supporters of this viewpoint believe that people construct their own realities in different ways, and maintain

that the impossibility of a context-free reality precludes categorising some versions of reality as "trustworthy" (Smith, 1984). Others maintain that all research involves subjective perception, but that there is an underlying reality that does exist and can be judged using the same quality criteria (Mays and Pope, 2000). Finally, a more moderate viewpoint exists which maintains that some quality criteria can be applied to both qualitative and quantitative research, but that other criteria may have to be modified to account for particular features of qualitative research (Popay et al., 1998). This is a position held by Meyrick (2006) who stresses that qualitative enquiry is not the same sort of enquiry as quantitative research, which utilises different methodologies and has distinctive goals. Hence we should not necessarily judge these two methods using the same quality criteria, nor be biased towards quantitative aspects of rigour. A similar position is held by Mays and Pope (1995b), who assert that all research is selective and "depends on collecting particular sorts of evidence through the prism of particular methods, each of which has its strength and weaknesses" (p.109).

Whilst this conceptual debate is important, various strategies are available within qualitative research to ensure systematic and self-conscious research design and data interpretation, to protect against bias and to enhance the reliability of the findings. For example, the researcher can demonstrate that appropriate methods have been used through referencing their use in similar studies (Meyrick, 2006). Reliability of data analysis can be achieved by having transcripts coded by more than one researcher and comparing agreement between the coders (Mays and Pope, 1995b). Furthermore, triangulation can be used as a method to address the issue of internal validity by using more than one technique or participant group to corroborate findings (Barbour, 2001).

Qualitative research methods have gained popularity in the healthcare setting over the past fifteen years: in 1995 the British Medical Journal ran a series of articles advocating their utility in health research (Britten, 1995, Mays and Pope, 1995b, Pope and Mays, 1995). More recently, a further series of articles focusing on a variety of qualitative research methods was published (Hodges et al., 2008, Kuper et al., 2008, Reeves et al., 2008). Qualitative methods have been widely used in studies looking into the psychosocial effects of genetic testing on patients. For example, Anido et al. (2005) conducted a qualitative analysis of responses from women concerning their attitudes towards carrier testing for fragile X syndrome. D'Agincourt-Canning explored the impact of genetic risk information from BRCA1/2 testing through in-depth qualitative interviews (d'Agincourt-Canning, 2006). Personal theories of inheritance, coping strategies and risk perception in hereditary non-polyposis colon cancer were analysed using qualitative methods in a study conducted by McAllister (2003).

3.3 Study Design

Semi-structured interviews were chosen as the primary method for collecting data as this method would enable me to explore the subject's thoughts and beliefs in great detail, whilst covering a number of relevant issues. Furthermore, it is a method likely to produce rich and complex data. Semi-structured interviews are widely used in the field of genetics as highlighted in studies included in the systematic review (Bekker et al., 1994, McConkie-Rosell et al., 1997, McConkie-Rosell et al., 2000, McConkie-Rosell et al., 2001, Williams and Schutte, 1997).

There are a number of methods that have been extensively used to analyse qualitative data and that might have been suitable for this particular

study. These include discourse analysis (Potter and Wetherell, 1987), interpretative phenomenological analysis (Smith, 1996) and grounded theory (Glaser and Strauss, 1967). Interpretative phenomenological analysis (IPA) aims to explore in detail the individual, personal and lived experience and examine how participants make sense of their personal and social world, as opposed to attempting to produce an objective record of the event itself. This method of analysis also emphasises that research is a dynamic process in which the researcher is an active participant. The participant's experience is seen through the lens of the researcher, who will carry his or her own conceptions. In this way the process is interpretative as the researcher is engaged in making sense of someone's experience (Smith, 1996). The methodology of IPA entails firstly a close examination of the data followed by identification of initial themes, clustering themes and finally identifying connections between them. The researcher then produces a narrative account of their interpretation of the participants' experience (Smith and Osborn, 2003). Even though IPA has its roots in psychology, it is now widely used in health and clinical psychology (Donnison et al., 2009, Kay et al., 2009). Whilst this method would have helped me to understand how participants made sense of their experience, it would not necessarily further my understanding of why these experiences take place or why there may be differences between individuals (Willig, 2008). Furthermore, the aim of this method is not to develop a theory that can guide action, such as informing recommendations for health services or health policy. For these reasons it was not considered the most appropriate method.

Similarly, discourse analysis was also considered as a possible method to use in this study as it is a method that has been widely used in the field of health psychology and in particular in the field of genetics, in recent years (Arribas-Ayllon et al., 2008, Babul-Hirji et al., 2009, Williams and Noyes, 2009). Discourse

analysis holds that language is represented not as reflecting psychological and social reality but as constructing it. Thus there are no objective truths; instead language is the building blocks of 'social reality'. The main topic of interest is the underlying social structures, which may be assumed or played out within the conversation or text. The way we can gain a better understanding of social life and social interaction, is through the study of social texts and interviews, looking at the tools and strategies people use when they communicate, such as their choice of words, use of metaphors, and so on (Potter and Wetherell, 1987). Potter and Wetherell describe a number of steps to perform when doing a discourse analysis. First, there is a close examination and coding of the text, looking for themes, ideas, views, roles, and so on. The researcher looks for recurrent patterns shared throughout the data. Wider discourses are then identified, developed and revised e.g. a biomedical discourse or a psychological discourse. The function of the text is also considered. Although using discourse analysis in this study would have provided an understanding of how the interview participants constructed their own version of events, and identified commonly shared patterns of talking, the aim of this method is not to guide action which was one of the main desired outcomes of the study. For this reason it was not considered the most appropriate method to use.

Grounded theory (Glaser and Strauss, 1967) was developed by two sociologists, Barney Glaser and Anselm Strauss, who were in part protesting against the fact that so much sociological research was based on pre-existing theories. In response, they developed an inductive method of data analysis that was 'grounded' in the data, and that enabled a theory to emerge or be discovered (Willig, 2008). The method was devised while researching the experiences of chronically ill patients, with data collection and interpretation

following a systematic, clear and precise method which offered guidelines for the verification and validation of findings (Payne, 2007).

Even though Glaser and Strauss published *The Discovery of Grounded Theory* in 1967 (Glaser and Strauss, 1967) it was not until the late 1970's that the method began to gain popularity in studies relating to healthcare and health psychology (Atwood, 1977, Mullen, 1978). The theory itself has also developed in a number of ways since its inception. The original text (Glaser and Strauss, 1967), is regarded as the 'classic' account of the method. Since then however, Glaser and Strauss have differed in their views concerning what constitutes the 'correct' approach to the method. Glaser upholds the original methodology which states that theoretical insights emerge directly from the data if the researcher frees him or herself from any previous theoretical knowledge (Glaser and Strauss, 1967). However for Strauss (Strauss and Corbin, 1990) the idea of the researcher freeing himself from any previous theoretical knowledge is simply not feasible. Strauss and Corbin therefore proposed the utilisation of a specified theoretical framework based on knowledge of human action and certain theoretical concepts when explaining the phenomenon researched. Glaser was however critical of this method, claiming that the method was too prescriptive and "forced conceptual description" (Glaser, 1992 p.5).

Another fundamental issue which Glaser and Strauss disagreed over was the timing of consultation of the literature. Strauss and Corbin (1990) advocate reviewing the literature early on in the study as it stimulates theoretical sensitivity, provides a secondary source of data, stimulates questions, directs theoretical sampling, and provides supplementary validity. However, Glaser (1992) strongly disagreed with this stand and believed that professional literature should not be examined until codes and categories had begun to emerge from the data.

Despite these conflicting opinions, there remain a set of fundamental features associated with the grounded theory method. The process begins with the identification of an area of interest which may have been relatively ignored in the literature or given little attention (Payne, 2007). The aim is for the researcher to build his or her own theory from the ground (Strauss and Corbin, 1998). Samples are selected purposively in order that the sample can contribute to the topic under investigation (Payne, 2007). During data collection, interviews or focus groups (any source of textual data may be used but these are the most common) are transcribed and categories are developed from the data by 'open coding' of the transcripts (Payne, 2007). This means identifying and labelling meaningful units of text. These are clustered into categories which are concepts that stand for a particular phenomenon (Strauss and Corbin, 1998). Categories are usually descriptive to begin with and become more analytic as the process goes on.

As coding continues, the number of new categories emerging from the data will generally progressively reduce, until no new examples occur. This is described as saturation (Glaser and Strauss, 1967). At this point, the specific properties and dimensions of the categories can be further developed through constant comparison. This refers to examining segments of the text against previous categories for similarities and differences (Strauss and Corbin, 1998). At this stage, theoretical sampling of participants who are specifically selected to help test and develop categories further and test emerging theories occurs. Theoretical sampling, which is both guided by and contributes to the emerging model, is a key feature of grounded theory (Payne, 2007).

Axial coding is used to relate categories to their subcategories. Axial refers to coding around the axis of a category, linking categories at the level of properties and dimensions (Strauss and Corbin, 1998). Throughout the whole

process, researchers are encouraged to write memos to record any thoughts, interpretations, questions and directions for further data collection (Strauss and Corbin, 1998). In the final stages, a core category which has major explanatory power is identified which may build into an emergent theory. To validate the theory, the researcher returns to the data to check it against segments of the text (Payne, 2007).

Grounded theory provides a systematic and rigorous method of data analysis, with specific procedures such as coding and memo-writing, to ensure that the analysis is transparent and any evolving theory remains closely tied to the data. However, as is the case with any research method, it does have a number of limitations. Theoretical sampling is a key component of conducting grounded theory. However, as Backman and Kyngas point out (1999), using this method, the researcher may make conclusions that are based on his or her preliminary analyses, which may influence too strongly the subsequent data collection and the emerging theory. The method also requires the researcher to commit to a time-consuming and long process. Where time or budget constraints exist, the researcher may have to compromise between the demands of the approach and the resources available. (Backman and Kyngas, 1999)

Conrad (1990) and Riessman (1990) suggest that fracturing the data in grounded theory might limit the researcher's understanding, as the aim is analysis of the data rather than the portrayal of the subjects' experience in full. Another criticism of grounded theory is that it does not address questions of reflexivity satisfactorily. Dey suggests that even if we do 'discover' categories, what we discover will depend to a certain extent on what we are looking for (Dey, 1999). Hence, whatever emerges from the data is in some way shaped by us as researchers, our particular agendas and our preconceptions. One

way to ensure as far as possible that emerging concepts are grounded in the data is to document in detail each stage of the research process, for example, through memo-writing. This increases reflexivity and demonstrates the ways in which the researcher's assumptions, views and beliefs have shaped the research (Pidgeon and Henwood, 2004). Using independent coders also helps to ensure that the findings have an appropriate degree of trustworthiness (Barbour, 2001).

Despite these limitations, grounded theory was considered to be the most suitable method of analysis for a number of reasons:

1. it is a method which is 'likely to offer insight, enhance understanding and provide a meaningful guide to action' (Strauss and Corbin, 1998 p.12). By using this method, insightful accounts that would inform genetic counsellors, health policy makers and academics were likely to emerge. Furthermore, the findings could be used to underpin the content of the information resources;
2. it is suitable for exploratory and explanatory research (Payne, 2007). Thus it was a method considered particularly useful for exploring the impact of parenting a child with no clear diagnosis, an area of genetics in which very little research currently exists;
3. the method aims to enable the researcher to elicit participants' understandings, perceptions and experiences of the world (Payne, 2007). Therefore, the findings should represent the reality of the phenomena under investigation and not a subjective interpretation of reality. This aspect of grounded theory was important as the aim was that the information resources would reflect 'real world' experiences;

4. grounded theory method provides an explanatory framework with which to understand the phenomenon under investigation (Willig, 2008), and so guides the research process; and
5. it was hoped that by using grounded theory, major themes and possible theories relating to the two issues under examination, would emerge from the data (Strauss and Corbin, 1998).

In addition, I decided to use the methods described by Strauss and Corbin (Strauss and Corbin, 1998). This was firstly because I felt that my existing knowledge of some of the key psychosocial concepts related to genetic testing would prohibit me from approaching the subject without any previous knowledge, as Glaser advises, and secondly, because I felt a literature review was useful in order to clarify concepts, and gain a deeper understanding of the key questions that needed to be asked during interviews.

3.4 Ethical Approval

Full ethical approval was obtained through the National Research Ethics Service (Appendix 1) and Research and Development (R&D) approval was given from Guy's and St Thomas NHS Foundation Trust (Appendix 2). Approval for the study was also granted by the University of Plymouth ethics committee.

3.5 Participants

Participants were invited to participate in either the carrier testing sub-study or the 'non diagnosis' sub-study.

3.5.1 Carrier testing sub-study

In order to achieve maximum variation in the purposive sample, I aimed to recruit:

- both males and females

- participants who were identified as carriers of recessive conditions, X linked conditions and chromosome translocations, and participants identified as non-carriers, after carrier testing;
- carriers of manageable and non-manageable conditions;
- participants with children (affected and non-affected) and without; and
- participants who varied in respect of how long ago they had received their test results.

Participants were excluded if they were:

- mutation positive for adult onset conditions or other dominantly inherited autosomal conditions;
- children or adolescents (under 18 years of age);
- carriers without sufficient grasp of the English language; or
- carriers who were not competent to consent to the research.

3.5.2 'Non-diagnosis' study

In order to achieve maximum variation in the purposive sample, I aimed to recruit:

- both males and females
- parents who were still actively searching for a diagnosis for their child and those for whom it was not a priority;
- non-diagnosed children who varied in age;
- parents of children where the child without a diagnosis was an only child and parents of children where there was more than one child in the family;
- parents who had a 'working diagnosis' for their child and parents without; and

- parents who found out there was an undiagnosed condition when the woman was pregnant, and parents who found out after the child was born.

Participants were excluded if they:

- did not have sufficient grasp of the English language;
- were under 18 years of age; or
- were not competent to consent to the research.

3.6 Recruitment Process

All participants were recruited through the South East Thames Regional Genetics Service (Guy's genetics department). Guy's genetics department were informed of the study before I applied for ethics approval. They agreed to act as the lead for the study once ethics approval was granted. Once ethics was granted, I met with one of the genetic counsellors from the service who agreed to take responsibility for sending out invitations. The lead counsellor was informed of the inclusion and exclusion criteria and the ideal purposive sample for the study. Other members of staff from the department were also informed of the study and asked to identify and suggest potential participants. Purposive sampling was used to ensure participants with a variety of experiences were recruited to the two sub-studies. The lead member of staff from Guy's genetics department identified participants that would be most relevant to the study and sent out the invitations. This consisted of a letter from the department (on letter headed paper) stating their involvement in the study (Appendix 3) and a patient information sheet from myself on letter headed paper from the University of Plymouth (Appendix 4 and Appendix 5). The patient information sheet provided information concerning:

- what the study was about;

- why it was being done;
- what would be involved if the participant agreed to take part;
- what would happen if the participant changed their mind about being involved in the study once it had started; and
- my contact details if the potential participant wanted to ask further questions or be involved.

The last page of the information sheet was a reply slip which could be returned to me in a pre-paid envelope. The potential participant was asked to return the reply slip whether they wanted to participate or not. If the potential participant did not want to be involved in the study it was stated that this was 'absolutely fine' but they were asked to briefly explain why. This was considered useful information because knowledge of the reasons why participants were unwilling to participate could help inform recruitment for future studies. Potential participants were only invited to participate in the study once. If they did not respond to the initial invitation they were not contacted again.

3.7 Ethical Issues

A number of procedures were put in place to ensure that any ethical issues that might arise as a result of the interviews were anticipated and able to be addressed appropriately.

3.7.1 Consent

A patient information sheet (Appendix 4 and Appendix 5) was sent to potential participants explaining the study. The patient information sheet explained that if any difficulties arose as a result of the interview the participant would be put in contact with an appropriate person (such as a genetic counsellor) if the participant felt it was necessary. It was also explained that the

participant could change their mind about participating in the study at any time, withdraw without giving a reason and request that their details, audio-tapes and transcripts be deleted. My contact details were included on the patient information sheet if the potential participant wanted further information.

Before the interview began the reason for the study was explained once again and any questions were answered. Permission was sought to record the interview and participants were asked to sign a consent form (Appendix 6). Any partners present during the interview were also asked to sign a consent form. The consent form stated that the participant had read and understood the patient information sheet, understood that their participation was voluntary and agreed to take part in the study.

3.7.2 Freedom from coercion

The patient information sheet stated that if the potential participant was not interested in being interviewed, this was 'absolutely fine'. Terms such as 'willing to help' were also used to ensure potential participants did not feel pressurised into participating. Participants were informed that neither their own medical care nor that of their child would be affected by either accepting or declining to be involved in the study.

3.7.3 Confidentiality

In the patient information sheet it was explained that the interviews would be audio-taped and transcribed, but that all transcripts would be anonymised by changing names and other personal details. This would ensure that any comments made by the participants were anonymised when the findings were presented. It was also clarified that no one other than those researchers involved in the study would hear the interview or read the interview

transcripts, and all personal details would be kept securely and not passed on to anyone else. This was reiterated before the interview began.

All interview data including audio-files and transcripts were kept in a secure office, on an encrypted memory stick which only I had access to. To safeguard confidentiality, all participants were assigned an identification number so that their identity was not known by anyone other than me. No names or distinguishable characteristics were used when writing up the study or when using quotes from the interviews. Personal data will only be kept for three years, after which time it will be destroyed.

3.8 Data Collection

Face to face interviews were organised with participants who responded to a joint invitation from Guy's genetics department and from me to be involved in the study. Face to face interviews were considered to be the most appropriate method as it enables the subject matter to be pursued in-depth and provides the opportunity to probe and ask follow up question if necessary. The advantage of using face to face interviews rather than telephone interviews is that non-verbal cues from the interviewee are more likely to be picked up, and any discomfort or stress can be more easily detected. The questions asked in the interviews (Appendix 7 and Appendix 8) were informed by the literature reviews and were chosen in order to elicit as much understanding as possible about participants' psychological and social experiences.

The topic guide for the carrier testing sub-study was designed to examine issues including:

- why the participants had decided to undergo carrier testing;
- the emotional impact of receiving the test result;

- whether there had been any long term psychological impact;
- what decisions had been made based on the test result; and
- whether they had passed the information on to other family members.

The topic guide for the non-diagnosis sub-study was designed to examine issues including:

- when and why parents started searching for a diagnosis;
- whether they were still searching;
- what information they hoped a diagnosis would provide;
- what had been the main issues, both practical and emotional, that had arisen as a result of not having a diagnosis; and
- what advice they had for other parents in a similar position.

Questions which addressed less sensitive topics, such as when the participant first started looking for a diagnosis, were asked first and more difficult or sensitive topics were addressed further on during the interview. Draft questions were sent to relevant patient groups in order to ensure they were relevant and clear, and amendments were made in light of their suggestions. In addition, questions were refined and amended over the course of the interviews to take into account possible theories emerging from the data. In this way, potential theories could be challenged or elaborated on during interviews. Where both partners had undergone carrier testing, the participants stated a preference to be interviewed together. In these cases the same questions were asked to both participants and any differences in responses were explored further. Each interview was recorded, with the subject's permission, so that it could be reproduced verbatim. This was so that a detailed analysis of the transcript could be performed at a later stage. It also ensured

that there was no bias by the interviewer. Interviews were conducted between December 2008 and August 2009. Interviews were then transcribed by myself, given a code number and the names changed in order to ensure anonymity.

3.9 Recruitment and Interview Procedure

1. Staff from Guy's genetics department were approached to be involved in recruiting for the study and agreed, subject to ethical approval.
2. Ethics Approval was sought from The Joint UCL/UCLH Committees on the Ethics of Human Research (Appendix 1) and R & D approval from Guy's and St Thomas NHS Foundation Trust (Appendix 2).
3. Participants fitting the selection criteria were approached by a letter from Guy's genetics department (Appendix 3) highlighting the staff involvement in the study and including a patient information sheet (Appendix 4 and Appendix 5) from myself. The patient information sheet detailed the reason for the study, why they were being asked to participate and what they would be expected to do. Ethical issues such as confidentiality and the right to withdraw at any point were also addressed. Those who were interested in being involved in the study were asked to either complete the section at the bottom of the information sheet and return it, e mail or telephone me.
4. Invitations were staggered in order to maximise variation in participants.
5. Participants who responded in favour of being in the study were contacted and an appointment made to visit them in a place of their choosing at a convenient time. All chose to be interviewed at their home.

6. Before the interview began, the reason for and aims of the study were explained in person to each participant. Each participant was then asked to read and sign a consent form (Appendix 6). Participants were asked if they had any questions before the interview began and permission was sought to record the interview on a Dictaphone.
7. The participants were interviewed using semi-structured interview techniques. Draft questions (Appendix 7 and Appendix 8) had been approved by the Ethics Committee and had been tested out with patient groups.
8. I transcribed the taped interviews (an example of a transcribed interview can be found in Appendix 9). Names and identifying features were changed to protect the anonymity of the participants. Transcripts were kept on an encrypted memory stick in a locked office.

3.10 Data Analysis

The grounded theory method (Strauss and Corbin, 1998) was used to analyse the interview data. Data collection and analysis was done concurrently (Strauss and Corbin, 1998). After the first few interviews had been conducted, transcripts were coded to establish themes which were clustered to form categories. Any major themes and possible theories from the data were then worked back into the interview questions so that the issues could be probed more closely. Data analysis also progressively focused the sampling. The process of recruitment, interviews and data analysis was ongoing until saturation was reached and no new categories were emerging. This was achieved after nine months. At this point the lead counsellor at Guy's genetics

department was informed and no more invitations were sent out. The procedure used to analyse the data will now be described below.

1. To facilitate the grounded theory process, the software package NVivo, version 8, was used (QSR International, Pty Ltd). This programme facilitates the indexing and retrieval of data, and all coding and categorising was done using NVivo (an example of coding can be found in Appendix 10).
2. As transcripts were imported into NVivo, attribute information, (e.g. sex, carrier type, age of undiagnosed child etc) was created and stored for each case.
3. The first two transcripts were read repeatedly. During these readings, any words or phrases that were considered important or meaningful were coded through the creation of 'nodes', a storage area in NVivo for referencing coded text. This process is known as 'open coding' and the coding was descriptive in nature. Where possible names of codes were taken directly from the text ('in vivo').
4. As more interviews were transcribed and coded, similarities between the codes started to appear, and general categories were developed into which codes could be appropriately placed. This was facilitated using NVivo by creating 'tree nodes'. Each category had a number of branches beneath it into which codes that related to that category were placed. Sorting codes in this way enabled common properties and early comparisons to be made. During this process any overall thoughts, interpretations or questions relating to the data were noted separately as 'memos'.
5. During the coding process, one of the transcripts was sent to a supervisor to code. The codes and emerging categories were then

compared to ensure inter-rater reliability of the findings. Any disagreements were discussed until consensus was reached.

6. The process of interviewing, coding and categorising was fluid and continuous. As the analysis of the interviews continued, data were coded either into pre-existing codes or new codes were developed, and these codes were clustered to form categories. The interview schedule was also continuously amended in order to probe further any interesting observations or theories emerging from the data (theoretical sampling). For example, during the first few interviews in the carrier testing study, it became apparent that there had been a change in self-concept for some of the carriers. I therefore amended the topic guide so that I could explore this area further during interviews. Furthermore, during these interviews it became apparent that the experience of carrier testing varied greatly between those participants who were tested as a result of having an affected child, and those who were tested because of a family history or ethnic background. This was fed back to the lead genetic counsellor, who then tried to recruit participants from these two groups. Conducting the interviews and analysis process in this way ensured that it was iterative and early data collection and analysis was informing subsequent sampling.
7. During the process of theoretical sampling, where possible 'outliers' (negative cases) were recruited to test the emerging theory. For example, for the study assessing the impact of parenting a child without a diagnosis, difficulties accessing services was a prominent theme in the findings. To test whether this was also the case when there was a diagnosis, participants who had a 'working diagnosis'

(even if genetic testing was still ongoing) such as autism, were recruited into the study. This then enabled me to compare the experiences accessing services across these two groups.

8. Categories were checked and compared against segments of the text (constant comparison) to identify any similarities among and differences between them. During this process categories were revised, relabelled, merged or split. For example, the category 'coping mechanisms' was divided into subcategories of 'emotional coping mechanisms' and 'behavioural coping mechanisms' to account for the difference between activities such as information seeking which is an action, and 'remaining positive' which is more of an emotional state. Previously coded transcripts were revised and recoded.
9. To further develop emergent concepts into higher-level analytic concepts, categories were examined in order to identify links and relationships between them and their subcategories (axial coding). The coding stripes function of NVivo was particularly useful during this process as it provided a visual overview of how categories related to one another. For example, frustration was one of the categories that had arisen from the data derived from parents. To gain insight into how this might contribute to the psychosocial effect of parenting a child without a diagnosis, I examined the coding stripes attached to this node to identify any potential relationships with other nodes. This revealed that communication was an important subcategory of frustration. Analysing the data in this way generated new lines of enquiry and subsequently helped the development of a theory. Concurrently, annotations and memos were being continuously

developed to reflect on the process and capture any thoughts, questions or explanations relating to the data. This ensured that they were not forgotten.

10. Interviews and subsequent coding continued until no new categories were emerging from the data. At this point it was felt that saturation had been reached.
11. Category development continued until no new properties, dimensions or relationships were emerging (theoretical saturation). At this point, more theoretical, higher-level analytic accounts were developed (selective coding) by integrating and refining the major categories. The aim was also to identify a core category which related to the other sub-categories.
12. Possible core categories were then 'bounced' between the supervisor and me. This proved to be a very productive way of testing hypotheses and possible theories. Diagrammatic representations (Figure 4.1) were used to represent the connections between the categories, and helped provide a visual 'storyboard'. This helped in devising an emergent theory. Furthermore, a theoretical model based on grounded theory method and developed by Morrow and Smith (1995) (Figure 5.2) was adapted to help conceptualise the emerging theories.
13. The emergent theory was then grounded by returning to the data and validating it against segments of the text. Furthermore, as many of the findings were used to inform the content of the information resources, respondent validation occurred during the drafting stage (Chapter Six). The questionnaire sent to patients during the pilot

study was also another method of validating the key findings. The results from this final validation are discussed in Chapter Eight.

3.11 Conclusion

In this chapter I have described the methods used to recruit participants to the study, investigate the psychosocial experiences of patients and parents involved in genetic testing, and analyse the data derived from the interviews. The results from the data analysis for both sets of interviews will now be presented in the following two chapters.

Chapter Four:

Interviews with Participant who Have Undergone Carrier Testing

4.1 Introduction

The methods used to analyse the carrier testing interview data were described in the previous chapter. In this chapter, I will begin by introducing the key findings from the interviews. I will then present and describe a theory that encapsulates the overarching experience of participants undergoing carrier testing. I will present research evidence to support this theory, including the findings from the systematic review. Additionally, I will discuss the secondary themes identified during data analysis and link these to established theoretical concepts.

Quotations (in italics) are used to illustrate the points made in the analysis. All names have been changed to protect the identities of the participants. Comments made by myself will be marked in bold italics. Codes such as CT1 (Carrier Testing 1) are included at the end of each quote to identify the interview, and line numbers have also been included.

4.2 Sample Characteristics

Thirty invitations were sent out to invite participation, of which eight responses (27%) were received. In two cases, both partners had taken a carrier test at Guy's genetics department and both agreed to participate in the interview. In one case, one of the participants had taken a carrier test previously but not at Guy's genetics department, but agreed to be interviewed. In total, eight interviews were conducted with 11 participants. Interviews lasted between 30 to 50 minutes.

One hundred and twenty-two codes were derived from the interview data; these were organised into 15 categories. The number of new codes that resulted from each interview is shown in Table 4.1. A decision was made to stop conducting interviews after interview number eight. Very few new codes were being generated after the fifth interview and no new categories had emerged since the fourth interview. I felt therefore that saturation had been reached.

Table 4.1 Number of new codes and categories derived from each interview

Interview	New codes	New categories
CT1	53	14
CT2	22	0
CT3	7	0
CT4	14	1
CT5	14	0
CT6	5	0
CT7	3	0
CT8	4	0

4.2.1 Demographic characteristics of the participants

The characteristics of the participants are described in Table 4.2 and discussed in more detail below. In keeping with the grounded theory approach, maximum variation across the sample was sought.

Table 4.2 Pseudonyms and characteristics of participants

Interview	Pseudonym	Gender	Condition	Family history	Affected child	Unaffected children	How long test results known	Carrier
CT1	Diane Cot	F	FX	no	yes	no	3 months	yes
CT2	Jo Green	F	H	yes	no	1	always suspected but confirmed 2 years ago	yes
CT3	Gail Goss	F	CF	no	yes	no	1.4 years	yes
CT4a	Claire Hives	F	TS	no	no	1	15 years but confirmed 8 years ago	yes
CT4b	Sam Hives	M	TS	yes	no	1	20 years but confirmed 8 years ago	yes
CT5	Sally Night	F	balanced translocation	no	no	2 carry balanced translocation, 1 unsure	19 years ago	yes
CT6a	Leah Brown	F	SMA	yes	no	no	2 weeks	yes
CT6b	Ian Brown	M	SMA	no	no	no	2 weeks	no
CT7	Kim Bale	F	DMD	no	yes	1	3 months	no
CT8a	Cindy Cocks	F	CF	yes	no	no	2 months	no
CT8b	John Karr	M	CF	no	no	no	4 years	yes

Note: F = female; M = male; FX = fragile X; H = haemophilia; CF = cystic fibrosis; TS = Tay-Sachs; SMA = spinal muscular atrophy; DMD = Duchenne muscular dystrophy

Manageable and non-manageable conditions

There was a range in life expectancy and severity for the conditions for which carrier testing was carried out. Spinal muscular atrophy (type 1) and Tay-Sachs disease are non-treatable conditions and both result in short life expectancies, usually around two years and five years of age respectively (Bach et al., 2002, Gason et al., 2005). Cystic fibrosis is now considered to be a manageable condition and according to UK CF data, life expectancy is estimated to be greater than 50 years of age for those born in 2000 (Dodge et al., 2007). The life expectancy of a person with haemophilia (if treated adequately) and fragile X is approaching that of the general population (Partington et al., 1992, Tagliaferri et al., 2010).

In the case of someone with a balanced chromosome translocation, there are a number of possibilities in terms of the status of the offspring in any pregnancy. The pregnancy may end in miscarriage; the child may inherit an unbalanced translocation and may be born with some degree of developmental delay or learning difficulties; the child may inherit the same balanced translocation as the mother and not have any health or developmental problems as a result of the translocation; or the child may inherit entirely normal chromosomes (Bache et al., 2007, Pazarbasi et al., 2008, Stephenson and Sierra, 2006).

Previous knowledge of condition in the family

In four cases (CT2, CT4b, CT6a, CT8a), participants had a family history of the condition for which they were being tested. In seven cases there was no family history of the condition (CT1, CT3, CT4a, CT5, CT6b, CT7, CT8b), however, in two cases (CT4a, CT4b), Tay-Sachs screening had been conducted as a result of the participant's ethnic background. In two cases the test had been

taken because the partner was either a carrier (CT8a), or at risk of being a carrier (CT6b).

Length of time since participant received test result

The length of time since the participant received their test result varied from two weeks to 20 years. Five participants had been aware of their results for three months or less, and six participants had been aware for a year or longer.

Ethnic background

Although information about the ethnic background of participants was not requested at the time of the interview, it was acknowledged during the interview that the two participants being tested for Tay-Sachs were both Jewish. Tay-Sachs is a condition that is more prevalent in members of the Ashkenazi Jewish population than in other individuals.

4.3 Findings

The core concept that encapsulated the overriding experience of carrier testing for participants was 'reproductive empowerment', as illustrated by the following comment:

"Well once you know you are a carrier and if your partner is a carrier too, then you can make decisions. It means you have the power to make decisions that are right for you, like whether you would abort if you found out the baby had the condition. So it gives you that control which is a positive thing." CT8b L262

During the carrier testing process, participants gathered information which enabled them to take control of and make informed decisions regarding reproduction. They were also enabled to empower their (future) children as well as other family members.

A number of secondary outcomes emerged. Guilt was an issue that appeared to be prominent for women with children affected by both X linked and recessive conditions. The issue of stigma was also prevalent, and there was evidence to suggest that some participants experienced an altered sense of identity as a result of their carrier status. Lastly, there appeared to be differences in the language men and women used to describe their experience of carrier testing.

4.3.1 **Empowerment**

Reproductive empowerment emerged as the overriding concept that described both the participants' motivation for carrier testing, and the outcome of the counselling process. Participants' primary aim when accessing genetic services was to manage their risk, or their families' risk, of having a child affected by a genetic condition. In order to do this they gathered information around the condition, the risk of having an affected child, and the prenatal testing options available. This information was passed on primarily by health professionals, but support groups and the Internet were also utilised. Armed with this information, participants were then able to partake in autonomous and informed decision-making regarding the most appropriate way forward. Furthermore, they passed on information to family members, empowering them to take control over their reproductive lives.

4.3.2 **Managing risk – the main motivator for carrier testing**

The most commonly cited motivator for carrier testing was to manage one's reproductive risk. Two participants already had a child affected by a genetic condition, three had relatives affected by a genetic condition, and two were partners of carriers. These participants were therefore aware that there was a risk to any future children they might have and hence wanted to

know for certain before having any (more) children, as this comment from John highlights.

"We are thinking of starting a family and there is a suspected CF gene in my family through my nephew having CF, so we thought it was a responsible thing to do." CT8b L6

Similarly Jo, whose father had haemophilia and it was therefore very likely that she was a carrier, had a test to confirm her carrier status when she wanted to have children. Jo's experience of haemophilia had been very negative as it had affected her father quite severely, and therefore she had put off having a carrier test until it became important for her to know for reproductive purposes.

"And I suppose I'd been avoiding having the test for years until I had to for reproductive purposes as you say because I don't think I really wanted to face up to it. I think I was avoiding it for quite a long time until I really had to deal with it." CT2 L26

Kim had been informed by her general practitioner that she might be a carrier of spinal muscular atrophy, after her niece had been diagnosed with the condition. As Kim and her partner Ian wanted to start a family, they were referred to the genetic clinic in order to identify and manage their risk of having an affected child.

"So the doctor recommended that we got tested because we want to have children and so we decided to both get tested didn't we." CT6a L7

Increased risk of being a carrier due to ethnic background was another motivator for carrier testing. Claire and Sam, who were both Ashkenazi Jews and therefore at increased risk of being carriers of Tay-Sachs disease, had both been tested at a young age at their Chedar (religious) school. They had had their test results reconfirmed at the genetic clinic because they wanted to have children.

"Well it is quite prevalent in the Jewish community and I used to go to Chedar which is like Sunday school and it was just something that was offered to all the children I think when I was about 15 or 16, we were offered to be tested, and so I found out then...So when our relationship got more serious and we started thinking about children we got retested, so it was confirmed that we were both carriers." CT4a L7

4.3.3 Information gathering

Information gathering was a central part of the empowerment process. Participants gathered information about the condition, the risk of having an affected child and the various prenatal testing options available to them both before and after testing. This information was primarily provided by genetic professionals, however patient support groups and the Internet were also cited as sources for further information. The process of information gathering was ongoing throughout the carrier testing process.

The genetic counsellor was a central figure who helped facilitate the empowerment process. All the participants in this study had developed good trusting relationships with their genetic professionals, which ensured that they felt supported and informed throughout the carrier testing journey.

"The doctor we saw was very calming and reassuring. She made us feel very at ease. She provided us with all the information we needed at the right time. She was really good." CT6a L266

After reconfirming their carrier status, Claire and Sam had visited the genetic counsellor to discuss the various prenatal testing options available to them during pregnancy.

"Seeing Emma [the genetic counsellor] was very reassuring because before then I hadn't really understood that there was this thing called a CVS test or amniocentesis and so it was just good to have a professional explain all the different options. It was very helpful." CT4a L238

Discussion had also focused around the timings of invasive testing. Knowing they were both carriers of Tay-Sachs and could undergo prenatal testing

around 12 weeks of pregnancy enabled Claire and Sam to take control of their pregnancy by identifying whether the fetus was affected at the earliest possible stage. This was important as if the fetus was affected, they would be able to have a termination of pregnancy as early as possible.

"What it did do, the information around the test helped us to understand how the timings would work which was really important" CT4b L116

The genetic counsellor had provided information about the condition as well as the prenatal testing options available. However, Sam and Claire had also contacted a Tay-Sachs support group, who put them in touch with a couple who had had an affected child to help them more fully understand the implications of the disease.

"Yeah that was really interesting meeting somebody, they were both carriers and they had unfortunately had children who had the disease, so that was actually quite upsetting." CT4a L91

This had confirmed their decision to have a termination, if the fetus was found to have the condition.

"Tay-Sachs is such a serious illness it was just then kind of obvious to us that if I had had a pregnancy with Tay-Sachs I would have an abortion" CT4a L112

For those that were opposed to termination of pregnancy, alternative options could be investigated. For Diane who already had a child affected by fragile X, knowing she was a carrier gave her the opportunity to seek out a prenatal testing procedure that was more suited to her particular beliefs. Preimplantation genetic diagnosis (PGD) was an option that she had first heard about through the Fragile X Society.

"We went to the Fragile X conference in Birmingham and it was dealing with future children and testing and all the various options open to carriers who want to have fragile X free children." CT1 L297

The support group had also provided her with information about her son's condition, and been a good source of psychosocial support.

"We've found that we've got quite a lot of information and [they have] passed a lot of information on and so it's been helpful that way. And knowing that because some nights they work late so if you want to ring them up and have a chat to them they will chat to you over the phone, send things in the post, work with the schools." CT1 L230

Sally, who was a carrier of a balanced chromosome translocation, had looked for information in medical textbooks to understand the implications for a child born with her particular chromosome imbalance. This was primarily because genetic specialists had been unable to provide her with this information.

"This was long before the Internet, but I went to the post-graduate centre at [local] Hospital and looked up a lot of medical genetic books and scared myself silly looking at all the various genetic birth defects there were. Partly because at [the genetic clinic] they were unable to tell me if I delivered a baby that had an unbalanced translocation whether it would survive, or what abnormalities it would have, and so I was trying to look to see if there was anything in the textbooks and research papers about a 9 and 10 balanced or unbalanced translocation." CT5 L68

This information had given her a broader understanding of the possible implications of her chromosome imbalance.

As well as gathering information about the condition and prenatal testing options, knowledge and understanding surrounding inheritance and risk was another key factor that informed decision-making. Cindy, whose partner John was a carrier of CF, had spoken to a genetic counsellor about her risk of also being a carrier and if she was, having an affected child.

"She said there is a 1 in 25 chance that you are a carrier, and if you are, statistically on average if we had 4 children 1 of them would have CF, 2 would be carriers and 1 would be affected." CT8a L68

This information had been the catalyst for Cindy's decision to go ahead with carrier testing. Similarly, the information Kim had been given by the genetic counsellor had helped her understand the implications for her daughter and nieces, if she was found to be a carrier. This had been the primary reason for her decision to go ahead with carrier testing. Furthermore, they had recommended that her daughter should still be tested whatever her own results.

"The thing they were explaining to me at the appointment is that they do now realise that there is a chance that Susan is still potentially at risk, so they would still recommend that she be checked." CT7 L87

The information gathered through specialists, the Internet and support groups facilitated informed and shared decision-making. Additionally, it enabled participants to pass on information to and empower other family members.

4.3.4 Informed decision-making

For those participants who were identified as non-carriers, the knowledge was empowering because they were able to begin their reproductive lives knowing they were not at increased risk of having an affected pregnancy. This proved to be a huge relief for participants, both for themselves as well as their family.

"It's a huge relief not having to feel like you had inflicted pain on other people, even if it was negative for them. I would have had to ring my sister up and say 'you're going to have to get yourself tested', and that would have been really hard. So that was a huge relief." CT7 L34

Yet for those participants identified as carriers, whilst the information was not necessarily what they wanted to hear, it was reframed in a positive way that enabled them to make fully informed and autonomous decisions around reproduction. One of the benefits of knowing one's carrier status in advance of a pregnancy was that participants could find out about and think through

what options were most suitable for them based on their own particular views and beliefs. Cindy and John, for example, had engaged in shared decision-making through discussing their feelings about raising a child with CF.

"We discussed whether or not we could cope with a disabled child, and to be very honest about it we said we can't, we are not geared up mentally. And I know if you have a baby who is disabled you bond with it, but I don't know if I would, so we knew that if we did have an [affected] child at 12 weeks we would have seriously considered termination." CT8b L190

The information Claire and Sam had been given by the genetic counsellor and the support group, had helped inform their decision that they would go through with invasive testing and termination of pregnancy if necessary. Diane had also made an informed decision that if she were to have more children she would try to pursue the option of PGD.

"We still want to have children but if we do we will do it through IVF." CT3 L108

4.3.5 Empowering other family members

Information derived through the genetic clinic was useful to participants themselves, but it was also important for other family members. Providing information to children and relatives was a motivational reason for carrier testing cited by four participants. For Kim, whose son had Duchenne muscular dystrophy, it was important for her to find out whether she was a carrier because her daughter and nieces were getting to an age where they themselves would soon be thinking about having children.

"It's not going to change anything for Toby, its more Susan [daughter] that I was concerned about. And by that stage my sister's two girls were getting older too. The oldest one was now 17, going on 18, so it's beginning to be the point at which they will need to know one way or the other." CT7 L71

The testing process was a particularly emotional and difficult experience for Kim because it made her "wade through all the history of what had happened" (CT7 L95) when her son was first diagnosed. In addition, she felt particularly anxious about what her parents-in-law would think if the results confirmed she was a carrier.

"I found it quite hard at the beginning thinking, 'are John's family going to think I'm a carrier and have brought this in?'" CT7 L174

Gail also discussed how her carrier test results would provide useful information for other family members, not just in the immediate future, but in the longer term.

"You have to think about the long term and not just the short term. In the end it's going to help other people, other family members." CT3 L130

After having received their test results, participants were able to pass on carrier information to family members, thus empowering the wider family. All participants had informed family members of their test results, in particular those who might be at an increased risk themselves. Kim had informed her daughter, her sister and her two nieces. Diane had informed her brother. Other interviewees spoke of informing their 'extended families'. As a result, a number of family members had accessed the genetic clinic and had undergone carrier testing.

"My mum had a brother and it's on her side of the family, because they tested my parents just so that everybody in the family could all be traced and everyone could work out whether they were carriers or not." CT4 L14

The results of other family members' test results had an emotional impact on some participants. For Sally, finding out her mother was also a carrier had resulted in her feeling less isolated and her carrier status less significant.

"My mother's sister, my aunt, carries this as well so it has obviously gone back three generations and every time I found more people had it I think

I felt better because at the time that it was just me I felt very singled out and then once I found a lot of other people had got it in the family and had children and no problems whatsoever really or minimal problems I felt this was just a nuisance, and nothing else." CT5 L123

Most participants had found the process of passing on carrier information to family members a positive one. Gail's mother-in-law was pleased that she had been informed that she might be a carrier of CF, and had gone on to have testing herself.

"But she was happy I told her, and I gave her the details for Guys and she went and did the test and we found out that she's a carrier." CT3 L70

Leah's sister, who had had a child affected by spinal muscular atrophy, was pleased to find out that only Leah and not Ian was a carrier of the condition.

"But telling them the results was not a problem. I mean I think they were just pleased for us at the end of the day, that we were ok." CT6a L190

There were, however, examples of where there had been difficulties communicating test results to family members. Diane had found it difficult to speak to her relatives because her family was, as she described it, quite 'broken'.

"It's just difficult to tell people in my family, my cousins who we don't talk to very much, we haven't been able to tell yet. I just find it very difficult to tell them because there has been a lot of hardship, my mum and dad have split up, and my dad passed away a couple of years ago so it's all a bit broken families." CT1 L29

Jo found it difficult telling her parents-in-law that she was a haemophilia carrier because she felt that she was "bringing a blight into someone else's family" (CT2 L76). There were also a number of examples of parents reacting badly to the news that they may be carriers themselves and therefore may have unknowingly passed on the gene mutation. John commented that his mother was "adamant" (CT8b L231) the mutation was not from her side of the family.

Sally remarked that her mother *"felt at first that she could not possibly have it"* (CT5 L133). Diane's grandmother reacted by denying that it could have been passed on from her.

"My mum spoke to my nan and she said 'it's not from me full stop'. So she blocked it off, she said 'I didn't bring anything into the family'." CT1 L109

Nevertheless, all the participants understood the importance of passing on this information.

"I think that that is difficult knowledge to have, but obviously it's important for other people" CT5 L345

Whilst the dimensions of reproductive empowerment discussed above were the most prominent and overarching findings that emerged from the data, a number of secondary themes were also identified. I will now go on to look at these.

4.3.6 **Guilt**

The issue of guilt was a prominent theme in those cases where the participant already had an affected child. Three mothers, two of whom were (or were likely to be) carriers of recessive conditions and the other who was a carrier of an X linked condition, cited this as their initial reaction when they first received their test results.

"I felt very guilty about what had happened and wasn't sure where I went and what happened next." CT1 L33

Kim also expressed feelings of guilt after her son was diagnosed, even though this had not been confirmed through a genetic test.

"So when it was explained to you about Toby's diagnosis, were you aware that you could be a carrier?"

"Yes, right from the beginning. And to begin with that is a huge guilt, and it's horrible." CT7 L111

For Kim and Gail, this feeling appeared to have subsided over time. For Kim, finding out she was not in fact a carrier, was likely to have been the key factor. For Diane, however, the feeling still appeared to be prominent.

"I do feel guilty about having made Simon the way he is." CT1 L70.

The issue of 'grandmother guilt' was also prevalent for Gail and Kim. Gail described her mother, who had also found out she was a carrier, as feeling *"really cut up about it. She felt guilty"* (CT3 L74). Similarly Kim, who knew that she was likely to be a carrier if her daughter was, commented that her mother, *"found it very, very hard, and she felt responsible. She thought 'what have I done without realising it, what have I caused'."* (CT7 L51)

4.3.7 Altered sense of identity

The issue of genetic identity and the effect that carrier knowledge had on the participants' sense of self was an area explored during interviews. For the most part, there was no evidence that carriers felt 'less healthy' as a result of their carrier status. In fact, four participants made comments in which they made explicit their awareness that being a carrier did not affect their health in any way. The one exception was Diane who was aware that because she was a premutation carrier of fragile X (55-200 CGG repeats), she might experience an early menopause. Although she was aware that this would not impact her general health, the prospect of an early menopause had made her feel like a *"ticking time bomb"* (CT1 L41).

For a couple of participants, knowledge of their carrier status had caused a change in their sense of self (or genetic identity). For Sam, finding out he was a carrier of Tay-Sachs had made him feel *"more Jewish"* (CT4b L132).

"It just kind of felt like some of the genetic burden I've been given along with very short sight and a tendency to get fat and things like that." CT4b L153

Similarly, Leah felt differently about herself after finding out she was a carrier of spinal muscular atrophy.

"For 31 years you think there is nothing wrong and then all of sudden you find out there is this genetic thing. It does feel a bit odd" CT6a L216

4.3.8 **Stigma**

The issue of 'stigma' was raised by three participants. Sam felt that there was a certain stigma or "shame" associated with being a carrier of Tay-Sachs, particularly with the older generation.

"There was this real kind of stigma thing that our parents' generation have, which is a bit sad really and very unhelpful." CT4b L148

His partner Claire, who was also a carrier, felt that her parents viewed it as a "black mark" against her (CT4a L41). Similarly, John found that when discussing his carrier status with friends, they reacted as if it was "a sexual disease that's about to be passed on to them through me" (CT8b L98), suggesting that they viewed it as something negative.

4.3.9 **Language variation between the sexes**

There were prominent differences in the ways in which males and females described their experience of carrier testing. Male participants were more likely to talk about 'chance' and 'risk' than female participants were, suggesting that they viewed it primarily as a mathematical phenomenon rather than an emotional one. Sam, for example, spoke of "understanding the odds", and that "the odds were in [their] favour" (CT4b L50) when asked how he felt about being a carrier. Likewise John spoke of having a "mathematical mindset" and "thinking in a very logical way" (CT8b L68) in comparison to his partner Cindy

who was *"fixated on the worst case scenario"* (CT8a L71). Ian said that he was *"concentrating on the percentages"* (CT6b L370). In comparison, female participants described the experience of carrier testing using emotive and descriptive language. Describing the experience of waiting for their test results, female participants used words such as *"scared"* (CT5 L110) and *"nervous"* (CT6a L22). Words such as *"guilty"* (CT1 L33) *"upset"* (CT4a L36) and *"gutted"* (CT4a L158) were used to describe the impact of being confirmed as a carrier. Similarly non-carriers used emotional expressions such as *"absolutely delighted"* (CT8a L167) and *"huge relief"* (CT7 L86) to describe their experience. For Jo and Kim, the process of undergoing carrier testing brought difficult past emotions to the surface again. *"It did bring up a lot of emotions, and that was hard"* (CT7 L243).

4.4 Discussion

The number of participants in this cohort was small and therefore the findings do need to be treated with some degree of caution. Nevertheless, the finding that empowerment summarises the main benefit patients derive from using clinical genetics services, identified by McAllister et al. (2008), is further confirmed through this study. Furthermore, I have extended the work of McAllister et al. by specifying 'reproductive empowerment' as the key outcome of carrier testing. A number of prominent secondary themes including guilt, stigma and altered sense of identity have also been identified. I will now look at these themes in more detail, beginning with a discussion of the emergence of the core category.

4.4.1 Reproductive empowerment - the core category

A number of stages occurred during the identification of 'reproductive empowerment' as the core category. These included: the identification of a

concept that integrated many of the themes established during the initial coding stage; the discovery of an existing theory that fitted with the concepts emerging from my data (emergent fit); and finally, modification of the existing theory to describe more specifically the findings as they related to carrier testing.

Identification of a concept

During the initial stages of theory development, a prominent theme from the data was 'reproductive control'. The term 'control' had been taken directly from the data (it was a word used by John), and during the analysis stage, it became evident that reproductive control was an important motivator and outcome of carrier testing. During axial coding, the major categories (reproductive control, guilt, sense of self etc.) were compared to see which held the most conceptual interest. Reproductive control was clearly the central phenomenon because it was most frequently discussed by participants and 'pulled together' the other emerging themes.

Using emergent fit to describe the core concept

During the course of my study, I became aware of Marion McAllister's work looking at the patient benefit from using clinical genetics services (McAllister et al., 2008), and realised it had congruence with my own findings. McAllister et al. had identified that empowerment, a key outcome of clinical genetics services, enabled service users to take control of their lives and have responsibility and autonomy over important life decisions and choices. Similarly, control and responsibility over decisions and choices concerning reproduction were the central findings from this study. My findings supported the findings of McAllister et al. in a specific area of clinical genetics services, namely carrier testing. McAllister et al. had, however, named this concept 'empowerment'

whereas I had named it 'control'. Yet empowerment was a more suitable term because it encapsulates both a process, incorporating actions, activities, or structures, and an outcome (Zimmerman, 1995).

Furthermore, McAllister et al. identified four dimensions of empowerment that were all evident in my own findings. These were: (1) informing decisions; (2) knowledge and understanding about the condition, risk to children and family members and prevention; (3) enabling effective use of the healthcare system; and (4) enabling hope for a fulfilling family life for oneself, one's family and/or one's future descendants (p898). In this study, participants were also making important life decisions, in this case as a result of knowledge derived from carrier testing (Dimension 1), they were gathering knowledge and understanding from health professionals and other sources such as the Internet and support groups (Dimension 2), family members were benefiting from the information and many were accessing the genetic clinic themselves (Dimension 3), and finally, the participants and their family members could hope for a future family life without the particular disease they were concerned about (Dimension 4). For these reasons it seemed more fitting to adopt the term 'empowerment' in my own description of the central phenomenon, as it better described the phenomenon under investigation, and built on the work that had already been conducted in the area.

To ensure this was in keeping with the grounded theory method, I went back to the literature to see what Strauss and Corbin (1998) said about the use of existing theories. They acknowledge that as professionals, "we cannot completely divorce ourselves from who we are or from what we know...it is by using what we bring to the data in a systematic and aware way that we become sensitive to meaning without forcing our explanations on the data" (p.47). Further, they go on to say:

"All assumptions of pre-existing theories are subject to potential scepticism and, therefore, must be scrutinized in light of one's own data. The latter allow the researcher to question and qualify as well as to give assent to his or her received theories. Concepts must 'earn their way' into a study rather than be blindly accepted and imposed on data. Received theories might work brilliantly for some data but not so well on other data." (p292).

Thus, as long as the pre-existing theory is rigorously tested through constant comparison, (i.e. going back and checking that the concepts I have identified fit, without being forced, with the existing theory), then there does not appear to be any problem using an existing theory to explain or interpret what is occurring in my own findings.

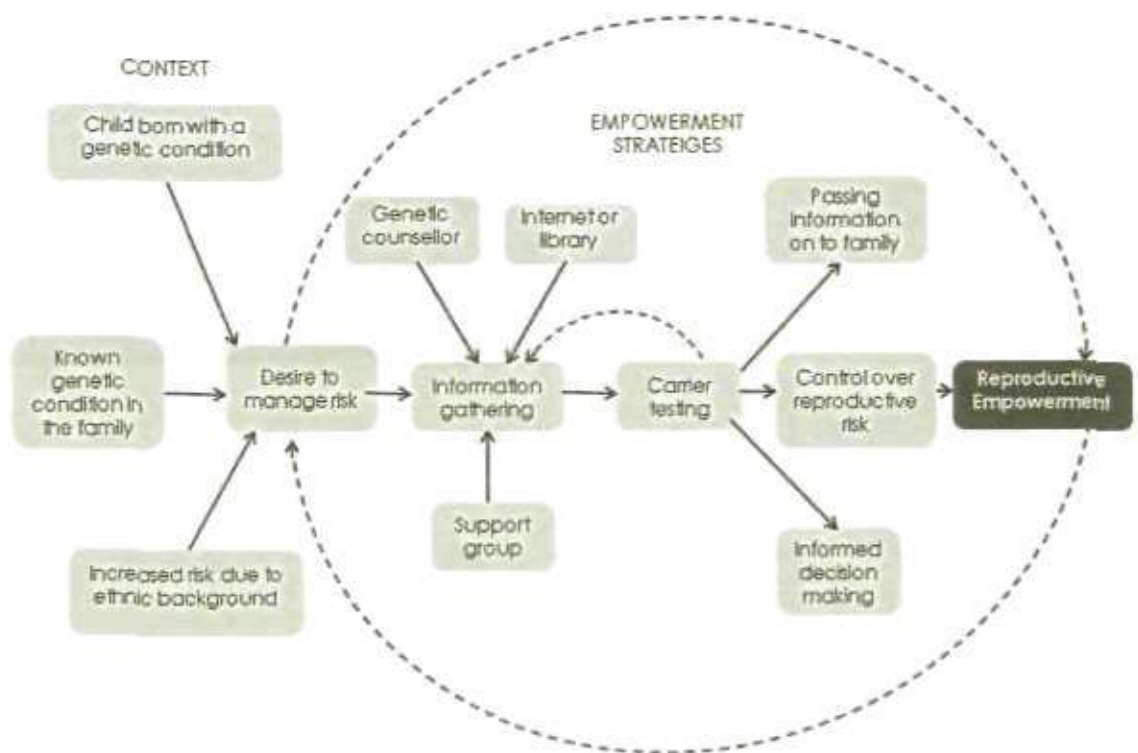
Modifying the existing theory

During the constant comparative process, I modified and built on the existing theory so that it fitted more accurately with my own findings. In the study conducted by McAllister et al. (2008), the scope of the study had encompassed all aspects of genetic services. However in this study I was focusing on carrier testing. The main motivator and outcome for the participants in this study had been to inform reproduction, either their own, their children's, or other family members'. I therefore named the central phenomenon 'reproductive empowerment'.

Reproductive empowerment was identified as a multi-dimensional process, with the various dimensions feeding into each other. A desire to manage reproductive risk was the first stage of reproductive empowerment, and the main motivation for carrier testing. The context behind this motivation was either the birth of a child affected by a genetic condition, a known genetic condition in the family, or increased risk due to ethnic background. In order to manage this risk, participants gathered information from genetic

professionals, the Internet and libraries, and patient support groups. Information gathering was a central facilitator of empowerment and was conducted in stages, as and when it was needed, both before and after testing. With this information, participants were able to make informed decisions, regain control over their reproductive risk, and pass on information to family members. These dimensions were all part of the empowerment process.

Figure 4.1 A model of reproductive empowerment



4.4.2 Reproductive empowerment: research evidence

A number of the key themes including reproductive decision making, the use of prenatal testing and passing on information to family members were identified in the systematic review and were also apparent during this phase of the study, confirming their importance within the carrier testing process. Moreover, the term 'actively coping', a term used by McConkie-Rosell et al. (2000) was used to describe the way in which participants were dealing with

their test results in the systematic review. However, in this phase of the study I have identified that reproductive empowerment is a more accurate description.

Lakeman et al. (2008) found that participants intended to draw reproductive decisions from test results, with 27% stating they would have considered not having (more) children if they had been identified as a carrier couple, 89% saying they would have opted for prenatal diagnosis, and 68% saying they would have considered termination of pregnancy. Similarly, Henneman et al. (2002b) found that all viable pregnancies of carrier couples were monitored by prenatal diagnosis and all affected pregnancies were terminated. In a qualitative study in which the reproductive decisions of CF carrier couples was explored. Where disclosure of test results was assessed, authors found that participants did share their test results with family members. Watson et al. (1992) found that 83% of CF carriers told their parents, 82% their siblings and 48% told other relatives. Likewise, Henneman (2002a) reported that most CF carriers shared the information with parents and siblings. These findings support the identification of reproductive empowerment as a central phenomenon of carrier testing.

Reproductive empowerment has also been discussed widely in the healthcare setting more generally. In a study conducted by van Peperstraten et al. (van Peperstraten et al., 2010), couples going through in vitro fertilisation (IVF) were given a decision aid, the support of a nurse specialist, and the offer of reimbursement by way of an extra treatment cycle, to enable empowered decision-making about the number of embryos transferred during IVF. Similarly, women reported using PGD as empowering and led them to feel in control of their reproductive futures, in a qualitative study conducted by Karatas et al. (2010). Empowerment was a key theme for women who had home births. The

confidence arising from their intense preparation and partnership with their midwives permitted them to choreograph their birth experience to a degree that they felt would not be possible in a formal setting (Janssen et al., 2009). Furthermore, in a study conducted in Nigeria, it was found that providing reproductive health information alongside training in basic business skills and micro-credit facilities, empowered women especially in terms of their reproductive behaviour (Odutalu et al., 2003).

4.4.3 Secondary Themes

Guilt

The issue of guilt was one that was prominent in the findings, particularly for women who were mothers of affected children. Similar findings were identified in studies included in the systematic review (Anido et al., 2005, Dunn et al., 2008, James et al., 2006, McConkie-Rosell et al., 1997, Watson et al., 1992, Williams and Schutte, 1997). Yet whilst James et al. (2006) found that mothers of children with X linked conditions were more likely to experience feelings of guilt and self-blame than mothers of children with recessive conditions, this finding was not evident in this study, with all mothers of children with X linked and recessive conditions discussing feelings of guilt. Unfortunately, there were no male carriers who had affected children interviewed during this study, and therefore it is not possible to comment on whether this experience affected both males and females. As well as parental guilt, grandmother guilt was also evident from the data. This finding has been identified in other carrier testing studies (Anido et al., 2005).

Guilt, according to Weil (2000), is a normal response to adverse events over which one has little or no control. It is also believed to serve as a psychological defence against feelings of helplessness, implies a sense of

responsibility, and provides a sense of control over recurrence (Weil, 2000). Guilt, in this respect, may have been an active coping mechanism employed by carriers to retain a sense of control over their situation. Knowledge that if carrier status had been known sooner, procedures such as prenatal testing could have been put in place to avoid the birth of an affected child, may also have aggravated this feeling.

Altered sense of identity

The issue of identity was one that featured in the data. A small number of participants made comments about feeling genetically different, implying that on a psychological level finding out they were a carrier of a gene mutation had affected their sense of self. These findings are consistent with the findings of McConkie-Rosell et al. (2000), where some of the women described feeling 'genetically different' after receiving positive carrier test results for fragile X. Similar findings have been identified in studies outside the carrier testing spectrum. In a study in which women's and men's responses to genetic risk information about BRCA1 and BRCA2 were assessed, participants linked their positive test results to becoming more aware of their physical selves (d'Agincourt-Canning, 2006).

One possible reason why carrier status may affect feelings about identity has been suggested by McConkie-Rosell (2000). McConkie-Rosell posits that "carrier testing may challenge a 'wished-for' parental role" and may "alter how a person defines himself or herself in relation to reproductive expectations and future roles as a parent, grandparent, or great-grandparent." (p.297). Therefore, when participants were found to be carriers, this may have created a perceived barrier to having children. This in turn may have caused them to feel differently about their (future) identity as a parent. Findings from a number

of studies support this theory. Callanan et al. (1999) found that carriers and their partners showed slightly higher anxiety scores whilst waiting for their partners' test results, but that this decreased significantly when the test results came back negative, indicating that carriers were no longer worried about their carrier status once there was no risk of having an affected child. Anido et al. (2007) identified that the psychological implications of being a carrier were linked to a woman's life stage. For those who were not at a reproductive stage of their life, the information had little relevance. However, for those who were, the information had more impact and was actively processed. Furthermore, the information appeared to affect self-concept, as highlighted by one woman who said "it's something that you can't change and something you didn't know before now" (p101). Outside the sphere of carrier testing, women who had had unsuccessful fertility treatment described the existential challenges to their sense of self as a major consequence of their infertility (McCarthy, 2008).

Another possible reason that carrier status affected participants' sense of identity may be related to how people define themselves within their ethnic and religious world. For Sam, being a carrier of Tay-Sachs had made him feel "more Jewish". Due to the fact that the Tay-Sachs mutation is common and well known within the Ashkenazi Jewish population, finding out he was a carrier of this condition acted to reconfirm, possibly even strengthen his identity, within this social context. A similar finding was reported in a study in which women's and men's responses to risk information for cancer was assessed (d'Agincourt-Canning, 2006). D'Agincourt-Canning found that being a carrier of a BRCA2 mutation specific to Ashkenazi Jewish ancestry, made one woman, who had never considered herself Jewish, reconsider her religious identity and where she fitted into a larger social structure. It would be interesting to explore the

relationship between ethnic identity and carrier status further, as so far the evidence is limited to a small number of cases.

Identification as a carrier also had repercussions within a familial context. Empowering other family members was identified as a main motivator for testing. Furthermore, all the participants in this study had informed their family members of their test results. For Diane, the knowledge that she was a carrier of fragile X had imbued within her a sense of duty to re-contact family members who may themselves have been at risk. Being a carrier might therefore have the effect of changing one's value or role as a family member. As Finkler (2005) argues, genetic information has led to the 'biologisation' of kinship. Furthermore, it generates new forms of 'genetic responsibility', locating actually and potentially affected individuals within new communities of obligation and identification (Novas and Rose, 2000). Knowing one was a carrier may have created a sense of responsibility or 'moral duty' that participants felt towards other family members.

Familial responsibility has been identified in a number of studies assessing the impact of genetic testing. Hallowell et al. (2006) found that all men undergoing genetic testing for the BRCA1 and 2 mutation, did so because they felt it was a familial duty, and d'Agincourt-Canning (2006) found that almost all participants viewed genetic testing for inherited cancer as a way to 'do right' by their families. In addition, Anido et al. (2005) identified that there was a desire among carrier women from fragile X families to know their status in order that family members could make more informed choices. Disclosure of test results to other family members was also evident in a number of other studies included in the systematic review (Anido et al., 2005, Watson et al., 1992, Williams and Schutte, 1997).

Stigma

A number of participants in the study discussed how they had encountered stigma as a result of their carrier status, either from friends or family. These findings are consistent with the findings identified in the systematic review (Gordon et al., 2003, McConkie-Rosell et al., 2001). For Sam, stigma was something he had only experienced from his parents' generation. One reason for this may be that genetics would have only been introduced relatively recently to the school science curriculum and therefore may not have been a subject his parents' generation had much understanding of. Furthermore, older relatives may have lived through or soon after the Second World War, and therefore associate genetics with eugenics and abuses conducted during Nazi Germany and in other countries (Garver and Garver, 1991, Harper, 1992). In contrast, the younger generation are more likely to have learned about genetics at school and be more in touch with advances in genetics through multiple media sources including documentaries and the internet. Molster et al. (2009) found that participants aged 18 to 44 years had the highest level of genetic knowledge in an Australian study on public knowledge of human genetics and health.

Information about a condition may lessen the stigma associated with it. This finding has been identified in a number of health-related studies. Higher education was associated with better knowledge and more positive attitudes towards people with schizophrenia, in a study conducted in Greece (Economou et al., 2009). Similarly, it was found that participants who were more stigmatising of people with HIV/AIDS were less educated and less knowledgeable about the condition, in a study conducted in a South African community (Visser et al., 2009). It may also be the case that with the rise of the disability rights movement, and the passing of the Disability Discrimination Act

(DDA) in 1995, modern society is less tolerant of discrimination based on disability.

Language and gender

Men in this study were more likely than women to express their carrier status using terms related to chance and risk, which expresses a certain rational, mathematical way of thinking. Carrier females were also more likely to express negative emotions as a result of their test results than males were. These findings are consistent with those reported by Marteau et al. (1997) in a study assessing the emotional impact of carrier testing for cystic fibrosis. Gender differences in genetic testing were also reported by Newman et al. (2002) who found that women were more anxious than men prior to testing, and were less likely to view themselves positively after testing, but before learning test results.

There have been a number of possible theories as to why the men and women in this study responded differently to their carrier status. First, there may be differences in the way that men and women perceive and evaluate health threats, with women perceiving greater risks than men do. In a study of smoking risk perceptions in Swedish adolescents, it was found that girls perceived the mortality risk of smoking as significantly greater than boys did (Lundborg and Andersson, 2008). Similarly, in a study conducted in Zambia, men were more likely than women to have multiple sex partners, however were less likely to consider themselves at risk of HIV, compared to women (Do and Meekers, 2009).

If women perceive them to pose greater health threats than men do, it would seem implicit that in the context of genetics, women would be more likely to seek genetic counselling than men would, as a means of managing that health threat. However the literature only underpins this theory to some

degree. In studies in which attendance rates for BRCA1/2 counselling were reported, men were less likely to seek counselling than women were. For example, in a study in which uptake of BRCA1/2 testing for at risk family members in two UK centres was assessed, Brooks et al. (2004) identified that uptake for genetic counselling using a non-proactive approach was 53% and 29% for women, but only 11% and 12% for men. Cody et al. (2008) noted that only 19% of males compared to 80% of females who were at risk of carrying the BRCA1/2 mutation attended genetic counselling. However, these results are not surprising considering that the health implications for men carrying breast cancer mutations are not as severe as they are for women. Christiaans et al. (2008) identified no significant difference between uptake of counselling for males and females in a study in which uptake of genetic counselling for hypertrophic cardiomyopathy was assessed. Similarly, men and women were equally interested in genetic testing for hereditary nonpolyposis colorectal cancer (HNPCC) and undertook genetic counselling (Hadley et al. (2003).

Another possible theory to explain the finding that women expressed their carrier status in more negative terms than men did, may be that in the case of carrier testing, the threat is related to reproduction, an area of life in which women could be perceived to take most responsibility. Findings from studies have shown that women take a more prominent role in decisions related to reproduction and child health than men do, including communicating genetic health information to relatives. An analysis of posts about the MMR vaccine on an online chat forum for parents, found that most participants who contributed were female (Skea et al., 2008). In families at risk of hereditary cancer, it was found that women were more likely than men to be gatherers and disseminators of health information within families (Koehly et al., 2009) and in a study conducted by d'Agincourt-Canning, women took primary

responsibility for disclosing genetic risk information about BRCA1/2 mutations to other kin, even in cases where the mutation originated on the paternal side of the family (d'Agincourt-Canning, 2001).

4.5 Conclusion

The findings from the carrier testing interviews support the finding identified by McAllister et al. (2008), that the concept of empowerment summarises the patient benefits from using clinical genetics services. Furthermore, in the context of carrier testing, 'reproductive empowerment' more accurately describes the patient benefit, and was identified as a process involving information gathering, informed decision-making and information sharing. Guilt, altered identity and stigma were secondary themes that emerged from the data, confirming findings identified during the systematic review. I will now go on to present the findings from the second set of interviews, focusing on the parental experience of raising a child without a diagnosis.

Chapter Five:

Interviews with Parents of Undiagnosed Children

5.1 Introduction

In this chapter I will present the findings from the interviews conducted with parents of undiagnosed children which I refer to as the non-diagnosis study. As in the previous chapter, I will present a theoretical interpretation of the data which emerged using grounded theory method. I will then compare the theory to the findings of Rosenthal et al. (2001) and Graungaard and Skov (2006) who conducted similar qualitative studies based on in-depth interviews with parents. Finally, I will use existing theories to help interpret the data.

5.2 Sample Characteristics

The parents of 20 children were invited to participate in the non-diagnosis study. Nine interviews consisting of 14 participants were conducted (45% response rate). Where both parents were present and willing to be interviewed, they were interviewed together as a couple. Interviews lasted between 30 – 80 minutes.

In total there were 248 open codes derived from the interview data; these were organised into 29 categories during the initial coding process (Table 5.1). The number of new codes and categories that resulted from each interview was as follows:

Table 5.1 Number of new codes and categories derived from each interview

Interview	New codes	New categories
ND1	93	24
ND2	43	1
ND3	28	1
ND4	24	2
ND5	16	0
ND6	19	0
ND7	10	1
ND8	8	0
ND9	7	0

I decided to stop conducting interviews after interview number nine (ND9) because whilst there were still new codes being generated over the last few interviews, they were very specific to those particular cases. In addition, no new categories had been generated since interview number seven (ND7) and previous to that, interview number four (ND4). I therefore felt that saturation had been reached and conducting more interviews was unlikely to generate any further significant findings.

5.3 Demographic characteristics of participants

In keeping with the grounded theory approach, maximum variation between participants was sought. The characteristics of the participants and their children are presented in Table 5.2. and discussed further below.

Table 5.2 Pseudonyms of the parents, and other characteristics of parents and undiagnosed children

Interview	Pseudonym	Gender	Marital status	No. of children	Position of undiagnosed child	Age when problem first detected	Age of undiagnosed child	Gender of child
ND1	Sally Black	female	separated	2	oldest	12 week prenatal scan	9 years	female
ND2	Donna Franks	female	separated	2	twins	22 week prenatal scan	8 years	male
ND3a	Laura Dent	female	married	2	twins	3 years	6 years (twins both affected)	female
ND3b	Abe Dent	male						
ND4a	Mary Williams	female	married	2	youngest	birth	3 years	male
ND4b	Tony Williams	male						
ND5a	Rachel Brown	female	married	1	only child	2 years	5 years	female
ND5b	Alan Brown	male						
ND6	Alison Pearson	female	married	1	only child	20 week prenatal scan	3 months	female
ND7a	Sophie Kent	female	married	1	only child	9 months	15 months	male
ND7b	Collin Kent	male						
ND8a	Julia Bard	female	married	2	youngest	6 months	20 months	male
ND8b	David Bard	male						
ND9	Samantha Clark	female	separated	3	youngest	20 week prenatal scan	4 years	male

Number of children in the family

In three families the child without the diagnosis was an only child. In two families there were twins; in one case both twins were affected with an undiagnosed condition, in the other case only one of the twins was affected. In the remaining four families, in one case the undiagnosed child was the oldest, in the other three cases they were the youngest.

Point at which parent(s) identified medical problem

Four participants were informed there was a medical problem with the expected child during the pregnancy; one was in the first trimester at 12 weeks, the other three were in the second trimester. All found out during a routine scan at around four months into the pregnancy. In all other cases the parent(s) found out after the child was born, but the actual age of the child when the medical issue became apparent ranged from six weeks to three years.

Activity level in terms of searching for a diagnosis

There was a range of activity level concerning the search for a diagnosis. This ranged from "still persevering with all the different channels" (ND5a) to making a decision not to continue actively searching; "I felt I was putting her through unnecessary pain. So I said no more" (ND1).

Having a 'working diagnosis'

Even though many of the parent(s) described their children as having 'developmental delay', two specifically used a 'working diagnosis' to describe their child's condition. These included autism and a bilateral cataract. Autism had been given as a 'possible' diagnosis by a paediatrician, but the geneticist was still investigating a genetic cause. Similarly Mary and Tony had been given

a diagnosis of a bilateral cataract for their child's condition, but the cause of this was still unknown and thought to possibly be genetic.

Although Alison (ND6) had been given a diagnosis of a chromosome translocation, the prognosis in this case was unclear because the particular translocation had never been seen before. For this reason the family regarded it as a non-diagnosis because they had no information about what they could expect for the child in the future. I decided to include this participant in the study as a possible 'outlier' to test whether having an unclear prognosis generated the same difficulties as not having a diagnosis.

Employment

Three mothers had given up work. For two mothers, this was because it did not make sense financially (these participants were both single mothers) and in the other case, the reason given was that it was too difficult to work and look after a child with disabilities. Of the six married women participating in the study, two had given up working for the first five years of their child's life, but had recently returned to work, one mother was working part time, one mother was still on maternity leave, one mother chose not to work and one worked full time. Of the five fathers involved in the study, all worked full time.

Range of problems experienced by undiagnosed children

Parents described a vast range of problems affecting their children during interviews. These are detailed in Table 5.3.

Table 5.3 Range of problems affecting undiagnosed children as described by parents

Interview	Range of problems
ND1:	Growth deficiency, severe speech problems, bowel incontinence, learning difficulties, hearing problems, poor eyesight
ND2:	Hydrocephalus, problems with mobility, large head size, large body weight
ND3:	Not reaching milestones, no speech, 'episodes', defects on hands, nanocephaly, microcephaly, prominent defect on spine
ND4:	Pinprick pupils, abnormalities in eyes, bilateral cataracts
ND5:	Low muscle tone, unable to walk, discoloration in eyes, dysmorphic features
ND6:	Chromosome translocation, 6 weeks premature, low amniotic fluid, ears 'a bit low down'
ND7:	Dysmorphic features, wide set eyes, low weight, small holes in the heart
ND8:	Low muscle tone, not reaching milestones, torticollis, wobbly
ND9:	Developmental delay, fits, unable to walk

5.4 Findings

The central phenomenon, or core category, that emerged from the data and describes the overarching experience of parenting a child without a diagnosis, was named 'reconstructing the meaning of being a parent'. This concept denotes the process parents went through in order to effectively manage and control the day-to-day challenges of parenting a child with no diagnosis and thus no clear care pathway. As part of this ongoing and dynamic process, parents developed empowerment strategies in order to ensure necessary and appropriate care, treatment and services were accessed. I will begin by setting the scene and describing the context behind the parental experience.

5.4.1 Setting the scene – the beginning of the journey

The parents in this study were unable to find a certain diagnosis or aetiological explanation for their child's condition. Thus began a journey, ongoing at the time of the interview, in which parents searched for a diagnosis and simultaneously parented a child with a disabling condition. Parents first became aware that there was a medical problem either during pregnancy, or after the child had been born. Where a problem was identified during pregnancy, usually during a routine scan, the news came as a shock and was devastating.

"It was like being hit with a truck. I was sobbing, sitting outside sobbing in the garden and it was awful. It's a real shock because you don't anticipate it. It was horrible, it was really, really horrible." ND6 L78

"Well it was devastating to begin with. Absolutely devastating. It was like, well what do you do, how far do you take this. Every mother's worst fear isn't it. You're not going to have a normal child. Simple as that." ND9 L34

After this initial shock came the realisation for some women that difficult decisions would have to be made regarding termination or continuation of the pregnancy, as the following comment from Alison highlights.

"But when they finally found it and when we finally got the diagnosis which is still unclear and we still don't really know what it is, it was quite late on then, maybe 30 weeks. So we were quite far into the pregnancy so had we had to make a difficult decision." ND6 L39

When problems became apparent after birth, the realisation that there was a problem was more gradual. It often began when parents started to notice that the child was not reaching certain milestones.

"They were not doing things at the stages or around the stages they should have been. Like they didn't sit up very early, they didn't walk until they were past two. And obviously there was no speech, and then they started having little flip outs – little episodes" ND3a L17

Others described their child as "floppy" (ND8a L11), "had a problem with his eyes" (ND4a L7) or had "failure to thrive" (ND7 L8). Statements such as "there has been a lot of anxiety" (ND4a L163) and "it was very stressful" (ND3a L107) were used to describe the experience.

Thus parents found themselves plunged into a world of emotional turmoil, having to deal with experiences including frustration and loss, whilst simultaneously negotiating the broader intervening 'real world' issues including the health and education systems. Before looking at these broader social issues parents faced, I will begin by focusing on the emotional, psychological issues parents encountered.

5.4.2 The psychological experience

Frustration

Frustration was a common experience cited by participants that was encountered at many stages during the parental journey. In fact, the word frustration (or derivatives of) occurred 61 times during interviews, and was mentioned in all but one of the interviews. Parents were frustrated for a number of reasons. First, parents felt that without a diagnosis they did not know how their child would progress over time.

"Because there is no diagnosis, there is no prognosis, so we don't know what to expect, what he is going to do, we don't know whether he is going to walk, going to talk, to any extent at all, which is really frustrating, I mean really frustrating." ND9 L44

Without a diagnosis they were also unable to access appropriate treatments for their child.

"I'd just like to know how we treat it really. If it's Russell Silver syndrome it can be treated by growth hormones which is a positive thing." ND7a L68

It was also frustrating for parents not to have a 'term' to help summarise what was 'wrong' with their child, particularly when friends or other parents asked.

"...it would be nice to sort of say, oh yes, Laura has got rather than we don't know." ND5a L339

Not having a reason why their child had learning difficulties or developmental delay was another cause of frustration, and this difficulty was alluded to during three interviews. For Samantha, the 'not knowing' was frustrating on what she called a "personal emotional level, not a clinical level" (ND9 L347). This desire for a reason highlights that having a diagnosis would, for some parents, provide psychological relief, even if it would not change their child's condition. For Alison who did have a diagnosis of a chromosome translocation, there was some psychological relief in being given a genetic explanation, even if it failed to provide any information regarding her child's prognosis.

"I just feel better knowing what it is, even though it doesn't really mean anything." ND6 L161

Communication issues, both with the undiagnosed child as well as with health professionals, were a source of frustration for some parents. Two parents specifically discussed the frustration that arose with being unable to communicate with their child. Sally described the frustration that came with being unable to communicate with her daughter around the issue of toileting.

"...frustrating because me and Claire we couldn't communicate with each other which has been the biggest thing. Her frustration in wanting to do like, the walking, erm, even now the toileting, it's frustrating because I try and stick to a routine that will stop her having accidents, but when she does she gets disappointed, I get frustrated, it's always been frustrating." ND1 L170

Three parents highlighted the frustration of constantly having to repeat their child's 'life story' because of the numerous different specialists being seen.

"..you are always telling everything again and you just think, 'why can't you just open your notes and read what is in front of you because I have told at least a 100 people this, please I do not want to tell anybody again', and this is quite frustrating." ND2 L177

"If I go to a doctor or specialist and I've found it over so many years, that when I go in, I have to go over and over and over the same problem." NDT L56

Lack of information was another factor that contributed to feelings of frustration, in particular because health professionals were unable to provide more information about the condition, including issues such as quality of life, the genetic cause of the problem, chances of having another child with the same condition, and whether children would develop more problems later on in life. For Alison, even though she had a diagnosis of a chromosome translocation, it was almost meaningless as the genetic specialists could not give her any information regarding what impact this would have on her daughter.

"Nobody can give you that information no matter how many times you ask them, which we did, all the time. They won't give you information. Some will hazard a guess but others will go they don't know, and that's very frustrating." ND6 L216

Yet participants were also aware of how frustrating the situation was for the health professionals. There was an awareness that professionals wanted to give parents more information but couldn't.

"And as a parent you want to ask all these questions and you are repeating yourself all the time, always saying 'what do you think, what do you think?' It's frustrating for people, the specialists, the physiotherapist, the occupational [therapist], for them, but it's just because I want to know, that need to know for how to plan." ND9 L172

Lack of control

Parents felt out of control as a result of not having a diagnosis. This occurred for a number of reasons. First, parents were unsure what would

happen in the future. Three parents specifically discussed the issues of whether they would have to care for their child for the rest of their lives, and if they were unable to, were unsure who would, as the following comment from Donna highlights.

"I do not know what is going to happen to him in the future and that is the worst thing I think, it is just not knowing because as far as I am concerned I now have to be his carer for the rest of his life because I do not even know if he will be able to at some point go into, like an assisted living home and I think that is hardest thing." ND2 L97

Sally spoke of the difficulties of not being able to discuss puberty with her daughter, and this created a feeling of helplessness.

"I'm petrified about her going to school and hitting puberty, getting her periods. It petrifies me. I can't sit down with her and say darling, this is what you're going to go through." ND1 L265

For Samantha, not having a diagnosis was akin to "being blind" (ND9 L330), another term highlighting the lack of control parents felt regarding the future. Not knowing about recurrence risk was another factor within this theme. This was the case for parents' future children, as well as for their (unaffected) children's children.

"If we do not know what the syndrome is, if it is a syndrome, how do you know how the next child will be affected?" ND4b L290

"My concern is if it might be genetic...if it is something that's carried through the female line, when Helen [daughter] has children, she may pass it on." ND4 L150

The term 'rollercoaster', used by four parents, again emphasising the lack of control parents felt, as well as the 'ups' and 'downs' that made up the parental journey. Julia used this term to describe the two months of intense genetic testing her son had had whilst they were waiting for his test results. Sally used the word to summarise the highs and lows she had experienced during

her daughter's life. Laura used it to describe the different emotional states she found herself in day to day.

"Some days you feel angry, some days you are emotional, some days you are just normal, but at the end of the day that's what we are used to so that is normal to us." (ND3 L606)

Similarly, Tony described the sense of helplessness he felt during the search for a diagnosis. Finding a diagnosis was *"just a case of waiting to see if they do the things that a child with those syndromes would do."* (ND4b L192).

Loss of expectation

Loss of expectation was a recurring theme that emerged during the interviews. Where parents had once expected that they would have a 'normal' family life with a healthy child who would grow up, go to school, and eventually leave home, the reality that this was not necessarily going to be the case quickly dawned. For many, there were associated difficulties in accepting this new situation that they found themselves in. Samantha described how she would *"set goals"* (ND9 L73) for her son, however these hopes were inevitably dashed and she found herself having to come to terms with this disappointment. During one account, she described the difficulty in accepting the fact that her son would never go to a mainstream school.

"He's coming up to starting school this year and my aim was for him to go to mainstream. And you hope for that and you hope he is progressing but then it comes down to the fact that you have to face facts, he's not going to go to mainstream and that brings you back down again, that he hasn't reached the potential you'd hoped he would reach." ND9 L66

There was also the difficulty of accepting that her child would probably never walk. This loss of expectation was aggravated when she went from having to push a buggy to a wheelchair.

"I used to be a little bit ashamed. That walk of shame. Like when I went from a normal buggy into his bigger one, pushing it along, but you get over it, and I don't know why you feel like that, because it's almost like you are embarrassed of your own child, but I'm not, but it's coming to terms with it, its again something to come to terms with. It's that transition again, it's all the little transition periods, and it's never going to stop. I'm going to get more and more emotional as time goes on probably. But that's just something you live with and you accept." ND9 L469

A similar sentiment was echoed by David who described the difficulty he had accepting the way his child looked.

"...you almost just rush over to stick his tongue back in because you are conscious of what others might think of him. It's not because you are embarrassed but you are aware of the fact that he is unusual, you know, and you don't want to.....you almost feel sorry for him as if you want to say 'you don't have to go through this'. Because I know how I would feel if everyone was staring at me." ND8b L610

Parents often compared their child's progress with other children, either their own or those of friends, an act which inevitably compounded this loss of expectation.

"I mean my friend had a baby the same week as Elise and I saw her the other week and her baby is twice the size of Elise, he's smiling, etc, and she isn't..." ND6 L303

Additionally, there was the realisation for some parents that they might never be able to return to work. Sally had tried to return to work but found the pressures of looking after a disabled child and holding down a job too much.

"I've tried two or three times to go back to work but it's beaten me in the end and it's like, OK, I can't do this". ND1 L215

Sophie found that her son's constant appointments meant it was difficult for her to commit to an employer, so had only gone back to work part time. For Samantha, who was a single mother, it did not make any sense financially for her to return to work. This sentiment was also echoed by Donna.

"...in the long run, and it's a horrible thing to say, but we would be a lot worse off. If I had a part-time job I would lose my house and benefit and I would lose the council tax benefit..." ND2a L351

Other parents talked about coming to the realisation that other aspects of their life had to change. Donna spoke of the fact that she was unable to do the 'normal' things that children liked to do, like go to the cinema. Mary and Tony spoke of how they could not take walks on the beach they lived near because the noise of the waves and the seagulls frightened their son. Similarly Laura and Abe could not go on 'normal' family holidays.

"Things that we do in every-day life, things that you take for granted, we weren't able to do. We weren't able to just jet off here, take a holiday here, do this or that because our children couldn't handle it." ND3a L95

For Tony and Mary, their child's disability had meant that they had to put their life on hold.

"I would sum it up by saying your life kind of goes on hold as it probably would do with a young child anyway but to a much greater magnification." ND4b.L623

These factors contributed to the loss of expectations parents faced as a result of their child's condition.

Guilt, anxiety and stress

Guilt was a common emotional experience which existed for a number of reasons. Three parents were worried that their child's condition was something that they caused when they first found out there was a medical problem. Alison and Samantha were both worried it was a result of something they had done during their pregnancy, such as drinking alcohol, and Julia wondered whether her son's condition was linked to her epilepsy.

"I hoped I had not passed anything on to him, I felt responsible.... When you realise something is wrong with Nick then you think oh god, is it because of the tablets?" ND8 L59

Two parents felt guilty because they could not be as attentive to their other children as they would otherwise have been.

"I have his twin sister who I have so much guilt towards because we can't go out and do the things that a normal eight year old girl would love to be doing." ND2 L376

Samantha had tried to rectify this feeling by taking her two daughters out shopping when her son was being looked after by a friend. Accessing respite was another cause of guilt, in particular for Laura and Abe, who were very reluctant to leave their children with a carer.

"I think one of the biggest things with a lot of it is guilt, like with respite, Accessing respite we felt so guilty." ND3 L554

Anxiety and stress were also experienced by parents as a result of their child's condition. Parents commonly experienced anxiety when they first found out that there was a medical problem, especially if this occurred early in the child's life, before certain possible syndromes could be ruled out.

"There has been a lot of anxiety because when he was very small there was no indication that he didn't have severe learning difficulties which were one of the cluster of conditions that may have been associated with the other symptoms." ND4 L163

Three participants described the experience of looking after a child with special needs as being particularly stressful. Reasons included the lack of sleep coupled with a full time job, the constant visits to the hospital and looking after a child with disabilities as a single mother. For Laura and Abe, who had twins who were both affected, the experience was doubly stressful because of having to access two of everything.

"It is very very difficult because people see it again as it's not one it's two. So you have to get two of everything, two of all the services, so we find that we are up against a lot with that." ND3a L234

Parenting a child with a disability also caused a great deal of stress on relationships. One couple found that because they were constantly tired, they rarely went out and spent time together. Two couples acknowledged that they argued more as a result of the stress and worry of looking after a child with special needs. For Laura, a major source of arguments was her husband's reluctance to help access support and care.

"He doesn't step in and say anything which obviously then causes more problems because then I end up having a go at him because he hasn't said anything. Because he should be fighting as well as me." ND3a L446

There was awareness that many couples that have children with disabilities ended up separating. Sally, Donna and Samantha were already separated and Rachel and Alan knew a number of parents who had separated as a result of the stress of raising a disabled child. For those mothers that were separated from their child's father, there were examples given of the father's unwillingness to offer help and support.

"I don't have a great relationship with him and he does not really have an awful lot to do with them either." ND2 L514

Not all relationships were described in negative terms. Two interviewees commented that they were lucky to be in supportive relationships. Sally felt particularly lucky that her new husband was so accepting of her daughter as well as being supportive of her own needs.

"....but I'm so lucky cause I've found that in my husband, and he's so supportive, he accepted Claire instantly and has done nothing but try and forward her" ND1 L330

Distrust of health professionals

Although this was not a common finding, a number of participants did describe cumulative disappointments or mistakes made by health professionals which led to feelings of distrust. There was one example of a patient getting 'lost in the system' because the general practitioner (GP) had not followed the proper referral procedure. In another case, the clinician had not made the necessary speedy referral. A number of errors had also been made in which blood samples were lost, or DNA samples contaminated.

"The last set of blood I took her for got mucked up three times, the blood clotted, then they tested her for the wrong things..." ND1 L240

Other examples suggesting distrust between the parent and the health professional were also provided. Sally was not believed by the medical team when she said that her amniotic waters had broken, even though it later turned out that they had. Samantha did not believe the original diagnosis made by a specialist, and had had another scan which showed that the situation was not as bad as originally thought. Furthermore, there were a number of examples whereby health professionals appeared not to have allowed informed decision-making to take place, with three women implying that there had been pressure to terminate their pregnancies.

"Well, I was told when I was 22 weeks' pregnant at my scan that there was a problem and I was told to terminate, so I asked for a second opinion." ND2 L12

5.4.3 The sociological experience

In addition to the numerous psychological experiences parents were dealing with, they were simultaneously navigating their way through a maze of care pathways including health and support services, schools and the benefits system. These issues will now be discussed in more detail.

Accessing health, education and benefit services

Numerous parents described the difficulties they had experienced accessing certain services, in particular speech and language therapy, and occupational health.

"We actually want a speech and language [therapist] to come here, or give us an appointment at the hospital so we can take the girls there so they can actually have therapy which is the big thing that is lacking. It's really lacking. And no matter how hard we push we don't seem to get anywhere with it." ND3 L420

Another issue raised by a number of parents concerned the difficulty and length of time it took to complete forms, particularly when there were other children to look after.

"And when you're a single mum and trying to run a family, I've got two teens, a disabled child, you haven't got time to sit down and fill out forms and even take it in....and I'm not unintelligent by any stretch of the imagination, but its physically sitting down and having the time to process it." ND9 L140

One issue raised by parents that was particularly troublesome was the difficulty of completing forms when your child had no 'label', and thus no 'box to tick'. Parents found themselves having to explain that their child had no diagnosis, and list their symptoms. However, for Mary and Tony, having the label 'visually impaired' for their child's condition, was hugely beneficial as it enabled them to easily complete forms as well as explain to others what was wrong with their son.

"Because he has a physical disability that cannot be missed, so in this respect we are lucky if you like, because it means that we do get help. And when you write in forms he is visually impaired nobody says to you 'are you sure?'. It is a medical diagnosis that, you can try and query it but he has no lenses, he has no ability to focus." ND4 L454

The difficulties associated with getting a Statement (of educational need) were mentioned during two interviews. Sally had to take her local education authority to a tribunal because they would not give her a Statement, primarily because she did not have a diagnosis. Laura explained that getting a Statement was such a long and laborious process that by the end of it she had "hit rock bottom" (ND3a L191). However, for Rachel and Samantha who were supported by social and educational services during this period, getting a Statement appeared to be a much smoother journey. Rachel explained that "early years, that's county council based from when [her daughter] was at pre-school, they came into the home and they are the ones that drove for the statement." (ND5a L189).

Parents had mixed reports concerning how willing schools and nurseries were to take on their children. Laura explained that "as soon as we told people about the problems it was like 'oh sorry we haven't got any spaces'" (ND3a L138). Yet for Rachel and Alan, the process was much easier.

"When we went there we said, you know, we have got no diagnosis she is just Lisa, she is just unique. They said fine, that's fine, this is what we have, this is what we can do for her, so they were absolutely brilliant and they still are brilliant." ND5a L177

Housing and employment

For those parents living in council housing, the issue of appropriate housing was very pressing. Donna wanted the council to move her into a bungalow because her son was unable to get up the stairs, but she had so far been unsuccessful in this pursuit. However, she had received some assistance from social services as a result of her child's condition.

"Social Services came out they assessed Rob, they assessed our needs and then they moved everything around. So the toilet went into the

cupboard under the stairs and the downstairs toilet was made into a walk-in shower." ND2 L248

For Samantha, the lack of a diagnosis meant that the council were unwilling to spend money building a downstairs bedroom for her son.

"He is coming up to four now and he is still not walking or anything. The bedroom is upstairs and he is sharing a bedroom with his sister, and they are saying that they won't grant any funds because they say 'well if we do that and spend £50,000 then in six months he might be walking and it will be a waste of money'." ND9 L89

As previously discussed, having a child with a disability had an impact on parents' ability to work, with five having given up working as a direct result of their child's condition. However, for those parents who were still working, it appeared that in most cases employers understood parents' needs when it came to taking time off. Some employers were happy for parents to work from home if need be, and others offered flexible shifts. Mary, who worked for the civil service, was able to take school holidays off under the Disability Discrimination Act. All the fathers interviewed worked full time. Alan commented that his boss was not very understanding or accommodating of his daughter's situation, and would not allow him to take time off for hospital visits. Collin commented that he did not frequently attend hospital appointments because he was self employed.

Two parents commented that they were concerned, particularly when they first found out about their child's condition, of the impact the condition would have on them financially. For Mary and Tony, there had been a concern when the condition was first identified that they might have to sell their house and buy somewhere cheaper so that one of them would be able to give up work.

Coping Strategies Employed by Parents

The contextual and intervening conditions described above highlight the climate in which parents without a diagnosis found themselves in. These factors contributed to the emergence of a new parental dynamic whereby parents first and foremost saw themselves as carers and advocates, rather than parents. Parents became proactively engaged in the search for a diagnosis, advocated for their child to ensure they received appropriate care and treatment, and at the same time continued to care for their child with a disabling condition. A number of emotional and behavioural coping strategies were employed by parents, both intuitively and consciously, empowering parents to manage this new role. It is these empowerment strategies which I will now go on to discuss.

Parents as experts

Over the years, parents had developed expertise regarding their child's particular condition and were knowledgeable about a variety of issues including their child's specific symptoms as well as issues such as feeding, behaviour and development.

"The girls have certain defects on their hands, the lines, and their heads are both small, and they have microcephaly and obviously that has to be monitored to make sure it grows in line with their body. They also have a prominent defect in their spine which protrudes out." ND3 L51

By developing expertise regarding their child's condition, parents were able to engage in more egalitarian-type, empowering relationships with their health professionals. Professionals treated them as partners by respecting and seeking their input, keeping them well informed and ensuring they were involved in decisions about testing and treatment. Parents wanted to be active participants in the diagnostic process and felt the medical team

acknowledged them as experts on their child's condition, as highlighted by Sophie who felt she was free to "ring up [the geneticist] and say 'can you check this on the database'" (ND7a L43). Furthermore, a number of parents were instrumental in developing a 'working diagnosis' for their child's condition, in collaboration with health professionals. This 'label' was a useful tool which enabled them to complete forms and provide responses when asked by people what was wrong with their child. Donna, for example, used the term 'global developmental delay' to describe her son. Laura and Abe used the term 'autism' which had been suggested as a possible diagnosis by their paediatrician. Samantha and Donna both used the term 'developmental delay'. In Samantha's case, this had been decided in collaboration with other health professionals.

"I just say developmental delay, I just say he's two years behind. I sat there in a key worker meeting and I said 'what do I say? If I'm asked what is Charlie's disability, how do I define it?' And we came up with developmental delay." ND9 L187

As well as being acknowledged as experts, parents also wanted to be acknowledged as being good parents. This idea was articulated by Sally who wanted to be seen as a "good mum" (ND1 L515). This provided a sense of achievement and made the experience rewarding.

"When you get someone that says 'well done, you've done fantastic' that is something that not a lot of people get and It makes it all worthwhile." ND1 L509

Developing relationships: gathering information

As well as developing expertise and relationships with health professionals, parents developed good relationships with other service providers and parents. Through this, parents were able to access useful information and ensure their child received appropriate care and support. The health visitor

seemed to be a particularly important person along the parental journey, and was instrumental in providing information about local special needs nurseries and disability benefits, as well as organising meeting between health and service professionals. For two mothers, the health visitor was seen as a "rock", providing practical and psychological support not just to them as mothers, but to the whole family. Samantha had developed a good relationship with her health visitor, who in turn had helped "organise all the meetings" (ND9 L123) with health professionals. She described her health visitor as a "substitute mum. She looks after us and makes sure that everything is tickity-boo and running like clockwork." (ND9 L12)

Samantha had also developed a close relationship with her housing officer who had played an important role in helping her access housing benefit.

"...she just came round with every single form and benefit that I was entitled to. Every single thing, things that I hadn't even thought or known about. And she sat here for hours and hours, days on end writing them all for me, making sure I got exactly what I needed." ND9 L128

Similarly, Julia had developed a good relationship with her health visitor as well as with her paediatrician, who had applied for speech and language sessions for their son.

Both Donna and Laura had developed close ties with the staff at their children's schools and nurseries. Donna was always "happy to volunteer to help out" at her son's nursery because they were "very short staffed" (ND2 L325). For Laura, the head of the nursery her children went to "made such a difference" and "was just brilliant from the day we met him" (ND3 L155). One outcome of these close networks was that parents were given good advice regarding services and schools in the area. Furthermore, they received help and support accessing services. Laura and Abe, for example, were able to

access a block of six speech and language sessions for their twins because of a referral from the nursery. For Donna, the close collaboration she had with the nursery facilitated a smooth transition from nursery to school.

"It was very easy for me to have him in the system and all the people who worked at the nursery knew me and knew Rob...so straightaway he was in nursery at three and then got 'statemented' while he was in the nursery so that he definitely went to [name] school." ND2 L229

Parents also developed strong networks with other parents, in particular with those whose children had similar disabilities. For Laura, *"you find when you have special needs children most of your friends have got special needs children"* (ND3a L287), primarily because they *"are more understanding"* (ND3a L302). Other parents were also a good source of information. For Julia, developing relationships with other parents had been helpful because they were in a good position to judge whether you were being an *"overly worried mother"* (ND8a L503). Alison, who was told her expected baby was at high risk of having Down syndrome after a 20 week scan, described how she *"spent that weekend talking to colleagues that worked with kids with Down's, talking to my friend who is a social worker, reading about Down's syndrome, getting as informed as I could"* (ND6 L129).

The Internet was a valuable tool for developing networks with other parents and experts, in particular with people that were in other parts of the country or overseas. As one father commented, the Internet enabled you *"to take the geography out of it"* (ND4b L944). Mary and Tony had found useful information about coping with their son's visual impairment by accessing chat forums. For them, they were able to access useful information relating to day-to-day issues such as eating, by speaking with other parents.

"Online they say 'well I always feed my child on a white plate because you do not get any white food so the child can always see the food'.

Just the little things that you would not dream of going to the doctors for but people don't tell you that. But if you go to these chat rooms all the trivial things that are actually quite big things as a parent you can resolve quite happily and quite easily because other people have been through it or are in the same boat as you." ND4 L949

Similarly Donna had been informed by her social worker of a number of different charities that she could access to help fund equipment.

"I have had help with getting things like the washing machine and a fridge and a holiday every year and if I hadn't already got a computer they would have provided me with a computer." ND2 L273

For Donna, a key factor that made a huge difference to the experience of caring for a child with a disability was "how much access you have to information" (ND2 L272), highlighting the importance of information within the empowerment process.

Becoming the 'gatekeeper'

Parents played an active role in navigating the system to ensure that appropriate care, treatment and services were met by service providers. In this way, parents became 'gatekeepers' for their child's care. Donna, for example, began looking into appropriate nurseries as soon as she moved into the area.

"The day after I came down here which was on a Sunday, on Monday I went to my old doctors to register with them and asked if I could speak to a health worker because I had the children and they were ten months old at the time. And asked if there is anywhere for children with disabilities because although they're only ten months old, I had to put things in place..." ND2 L224

Rachel ensured her daughter received optimal care at school by identifying where services were lacking, and reporting this back to the school.

"I go in there and I liaise with the SENKO lady who's in charge of the whole school and if I have got a problem I just say to her look and she goes and sorts it out... So I just go in there and I say to them look, this isn't

happening, this isn't happening and she says right I will look into that."
NDS L218

Parents also proactively ensured that health professionals were kept up-to-date with any appointments and tests that had been conducted. Julia, Sophie and Sally all discussed how they kept a file with all correspondence they received from health professionals, and ensured that all the professionals involved in their child's care were kept informed.

"I carry all the letters I've received from the paediatric consultant and genetics and sometimes those letters haven't yet been put on file and sometimes I have to pull out the letter and show it to them" ND7a L165

Julia ensured that her paediatrician, health visitor and GP were copied in to all correspondence to ensure there was a "team of people pulling the same way" (ND8 L493). These strategies ensured parents took control, where possible, over their child's care and treatment.

Fighting for services

Pushing or fighting in order to access services was identified as a key strategy employed by parents. In fact, the words 'push', 'pushing', 'fight' or 'fighter' were mentioned 48 times during interviews. Sally, for example, had to fight her local council in order to get her daughter into a special needs school because the local council wanted to keep her in mainstream schooling as she did not have a diagnosis.

"I was willing to take them to tribunal and fight them because I know Claire deserves more. And it took a lot of time and effort to do that."
ND1 L438

Laura and Abe had been fighting the council to get access to respite care, and had only recently succeeded after months of persisting.

"Well we've just managed to get some respite which again we've had to push for" ND3 L389

Likewise, Rachel and Alan reported being constantly on the phone to try to organise occupational therapy, and Samantha was currently fighting her housing association in order to try and get a downstairs bedroom fitted.

"I've got another appointment coming up and we're just pushing and pushing." ND9 L88

Whilst most parents discussed examples of where they had had to push or fight for services, those parents that had a good support network around them managed to share the burden, resulting in what appeared to be a less traumatic experience overall.

"My support group that I've got, I don't actually go to a support group, but the support group that I have, the occupational therapist, my health visitor that is still very much involved... They are all really pushing hard for me, they are doing all they can." ND9 L100

Developing explanatory theories and diagnoses

Developing theories about cause and diagnosis of the child's condition was evident during five interviews. Mary, for example, wondered whether she had contracted chicken pox from her brother during her pregnancy and this had somehow affected the fetus. Alan attributed his daughter's shaking as a symptom of a particular kind of autism. Similarly Laura believed that her two children had autism through *"research and the Internet and other people we spoke to"* (ND3a L39). These explanatory theories and diagnoses may have been psychological coping mechanisms which imbued parents with a greater sense of control over the situation.

Readjusting 'the norm'

A number of parents talked about comparing their child to others whom they considered to be 'worse off'. By doing this, parents readjusted the spectrum of what was 'normal', possibly in order to accommodate their own

child in a position that was not at the worst end of the scale. Donna, for example, felt "lucky" (ND2 L587) that her son was happy and was not constantly in and out of hospital like some of the other children she had seen. After reading stories of other children on the website of a patient organisation, Sophie felt that her and her husband weren't "too badly off" (ND7 L263). Similarly Julia and David felt their own situation was not as bad as it could be after seeing other children at a hydrotherapy centre.

Julia: "...you realise there are children there that are far worse off than Nick. Children with severe cerebral palsy, spina bifida, all sorts of problems and you just think..."

David: "It puts it in perspective"

Julia: "Puts it in perspective and you kind of think what am I complaining about...." (ND8 L265)

These may have been coping mechanisms parents employed in order to remain positive.

Internally readjusting –accepting and remaining positive

There were numerous comments related to 'accepting the situation', 'getting on with life' and 'just enjoying' your children. These highlight the positive outlook parents tried to maintain during what was frequently a difficult and complex journey. For Julia, it was important to accept her son for who he was, and not compare his progress to that of his sister.

"What he'll be he'll be, what he is he is, we don't mind" (ND8a L64)

Laura had found that she had accepted her child's condition, and instead of worrying about a diagnosis was keen to "just get on with day to day life" (ND3a L592). Similarly Samantha, who was not as focused on finding a diagnosis as she had once been, acknowledged that "even if we had a diagnosis he's still

Charlie to me, it's not going to change him, not going to change him in the slightest." ND9 L345

Parents were keen to enjoy their children regardless of their medical problems. Comments such as *"Let's enjoy him"* (ND7a L302) were echoed in four interviews. Parents were also aware of the need to focus their energy on being optimistic, as highlighted by comments such as *"you have to keep positive thinking"* (ND5a L498), and *"when you focus on the positive your life becomes so much easier"* (ND8a L229). Furthermore, they were anxious to express how rewarding and loved their child was. During seven interviews, parents made positive comments about their child such as *"I love her to death"* (ND1 L205), *"we just love her to bits"* (ND5a L506) and *"he's inspiring sometimes as to how loving and gorgeous he is"* (ND9 L368). For Alison, even though the experience had been difficult, it had been *"positive overall"* (ND6 L328).

5.4.4 **Consequences of strategies used for coping**

By using these strategies, parents empowered themselves to effectively care for their child, and they were able to keep from becoming overwhelmed with the day to day practical and emotional difficulties of raising a child with a disability. The consequence of this was that the nature of being a parent evolved, and the main role shifted to focus on advocating for the child. Furthermore, as the parental situation became increasingly manageable, parents were more able to cope with the difficulties of living with uncertainty. When probed, five parents said that a diagnosis was no longer a priority.

"There is nothing I can do is there. And I will chase it, but not to the point where it's like I wake up every day and worry about it." ND9 L332

5.4.5 Reconstructing the meaning of being a parent: the central phenomenon

The journey experienced by parents in their search for a diagnosis, had a profound impact on how parents viewed themselves. In order to cope with the challenges and demands they encountered, parents moved from being reactive to proactive and empowered themselves through the development of a number of management strategies. Parents had to go above and beyond what in other circumstances they would be expected to do. Furthermore, they had to adapt in order to effectively manage the situation. During this process, the meaning of what it was to be a parent adjusted and the primary focus shifted to caring and advocating for the child's condition. The following comment by Laura perfectly encapsulated this idea.

"You have to push, you have to say what's happening and you have to chase people. I've had people where I've been waiting for appointments and I've had to ring them every day, and you end up getting to a point where you're like 'I don't care if I upset you now, my children matter and you don't'. And I'm not that sort of person but I've had to learn to be that sort of person. And I think that's the biggest thing." ND3a L437

This phenomenon I have described as 'reconstructing the meaning of being a parent', and it forms the basis of the theory I will discuss in the next section.

5.5 Discussion

In this section, I will elucidate the theory presented at the beginning of the Findings section and discuss how it emerged inductively from the data. I will compare the theory with the findings of Rosenthal et al. (2001) and Graungaard and Skov (2006) who conducted similar studies assessing the parental impact of raising a child without a diagnosis. I will also apply pre-existing theories to help interpret the findings. To begin with, I will look at how

the coping strategies I identified relate to the wider body of literature within healthcare.

5.5.1 Previous research

Previous research into the parental experience of raising a child with a disability has identified similar contextual and intervening conditions as the ones identified in this study. Frustration, for example, is an experience frequently discussed in the literature. In a qualitative study of parents' experience of hearing loss, Russ et al. (2004) found that parents were generally frustrated with appointment delays, resource limitations, and the accessibility of information given to them by the medical team. In a study conducted in Canada in which parents' experiences seeking respite care for children with special needs was reviewed, Doig et al. (2009) established that caregivers were frustrated with the process of finding and obtaining respite care. Families of children diagnosed with type 1 diabetes (Wennick and Hallstrom, 2007) felt acceptance yet frustration with the care-giving experience, and parents of children with Kawasaki disease (Chahal et al., 2010) were frustrated by the lack of information available in lay language and the limited scientific knowledge regarding the long-term consequences.

Frustration was also a prominent theme in the study conducted by Rosenthal et al. (2001). Parents of children with undiagnosed conditions reported feelings of frustration because people did not believe that their child had a syndrome. In addition, they were thwarted in their attempts to locate appropriate support groups and meet parents of children with similar diagnoses in order to find a tangible vision of their child's future. Similarly, in Graungaard and Skov's qualitative study (2006), parents of undiagnosed children were frustrated at the lack of information specialists were able to give them.

Other experiences, including distrust of health professionals, loss of control, loss of expectation and loss of employability have also been identified. Huang et al. (2009) explored the experience of mothers after learning their child's diagnosis of cerebral palsy and found that parents mistrusted healthcare professionals when appropriate referrals were not made or when there was a mistaken diagnosis. Furthermore, mothers felt out of control and powerless, with the diagnosis feeling like the 'end of the world', and physicians giving no hope for their child's future. Godress et al. (2005) found that grief due to loss of expectation was a common experience for parents of children with mental illness in a study conducted in Australia. In a study of children with conditions including cerebral palsy, Down syndrome, stroke and spina bifida, Green (2007) found that the process of looking after a disabled child was 'physically exhausting' (p155), and had serious consequences for maternal ability to meet employment expectations with more than 50% of participants out of work. Mothers also required intensive, time consuming contact with the medical team.

Empowerment strategies used by parents to manage raising a child with a disability are also frequently discussed in the literature and give weight to the findings from this study. Green (2007) found that mothers took responsibility and often had to fight to ensure children received the medical treatments prescribed to them, despite opposition from health and social service providers. In addition, they engaged in advocacy activities, negotiations and paper work that were necessary to provide their children with the services experts had recommended, and became experts in navigating the healthcare and social service systems. Similarly, Doig et al. (2009) identified that navigating the system was one of the main strategies used by parents of children with special needs to find respite care. In a study conducted by Nuutila and Salanterä (2006) ,

parents were found to become experts in the skills and knowledge needed to care for children with conditions such as asthma, epilepsy, muscular dystrophy and leukaemia, and developed the ability and willingness to participate in decision-making. Graungaard and Skov (2006) also found that parents wanted to be acknowledged by clinicians as experts on their child. Networking was a key strategy adopted by parents in a study conducted by Weust (2000). Parents actively networked with health professionals, support groups, school staff and friends in order to increase knowledge and understanding of system structures that potentially influenced caring. Furthermore, information seeking was identified as a strategy used by some parents in order to cope with the diagnosis, management plan and prognosis of children with chronic illnesses, in a study conducted by Hummelinck and Pollock (2006).

5.5.2 Findings unique within the experience of having no diagnosis

Whilst the experiences and strategies cited above are encountered by parents of children with diagnosed and undiagnosed conditions, there do appear to be a number of findings particularly pertinent to the experience of having no diagnosis. First, the difficulties described in studies where parents did have a diagnosis are primarily sociological constraints, i.e. they encompass the difficulties meeting the personal, medical, educational and social needs of the child within a system that is often non-responsive, poorly organised and underfunded. Whilst the parents in this study did experience these burdens, a key difference is that they also experienced ongoing, long-term psychological issues, specifically the difficulty of not having a reason for their child's condition, or 'a tangible vision of their child's future' (Rosenthal et al., 2001), perpetuating a feeling of being out of control. Similar findings were identified by Graungaard and Skov (2006). In their study, in which half the children had a diagnosis and

the other half did not, it was found that parents without a diagnosis were found to fluctuate much more between emotional and cognitive dimensions such as frustration/anger and confidence, and powerlessness and possibilities for action, than parents with a diagnosis.

Another key difference concerns the issue of future risk. Some of the parents in this study and the Rosenthal et al. (2001) study were concerned about whether future children might be at risk of the condition. However, this finding was not apparent in studies where children did have a diagnosis. The reason for this is likely to be that for diagnosed conditions, health professionals are able to provide parents with a recurrence risk. Where the etiological explanation for the child's condition is unknown, it is not possible to provide such a risk.

Parents of children with very rare conditions for which very little information is available are also likely to experience some of the psychological issues experienced by parents without a diagnosis. Alison, for example, had been given a diagnosis of a chromosome translocation yet experienced frustration at having no clear prognosis for her child, just as the other parents in this study did, as the translocation was so rare. Feelings of powerlessness and frustration at not having a clear prognosis may also be experienced by parents of children with highly variable conditions. It would be interesting to explore this further to identify whether there are similarities of experience across parents with very rare, variable and undiagnosed conditions. If so, psychosocial information developed for parents of undiagnosed children may also be relevant for parents of very rare and variable conditions.

5.5.3 Development of a grounded theory

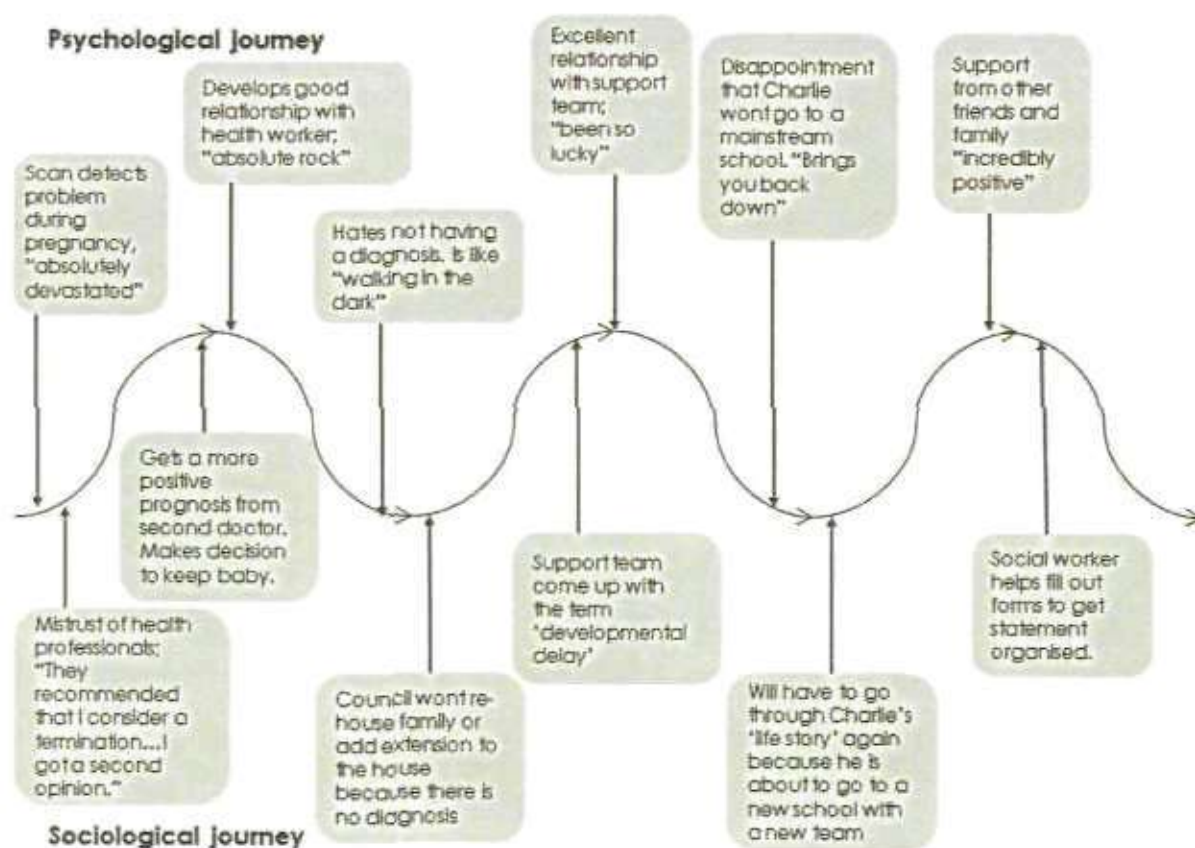
The development of a grounded theory was the culmination of numerous stages of analysis of the data. During these stages, different relationships between the categories were identified. Towards the final stages of analysis, a central category was chosen which linked together the other major categories and logically explained what was occurring in the data.

I will now provide an overview of these stages to account for how the grounded theory was developed inductively from the data. A number of integrative diagrams were developed during these stages which helped visualise the parental story and link categories.

The parental journey – a series of waves

During the initial stages of theory development, it became clear from the data that parents were describing a journey that began when a medical problem was identified (either during pregnancy or during the early stages of the child's development), and was still ongoing at the time of the interview. This journey can be viewed as a series of waves or transition periods made up of both positive and negative experiences. These 'ups' and 'downs' include both social as well as psychological encounters, and highlight the 'rollercoaster' that parents described, a unifying feature of all the experiences documented.

Figure 5.1 Wave diagram representing the psychosocial experience of Samantha Clark (ND9)



Identifying empowerment strategies

It became clear during axial coding that whilst parents were describing a journey filled with psychological and social complexities, there was another process occurring. Parents were consciously as well as unconsciously developing numerous strategies to cope with the difficulties they were encountering. These strategies were being used as a tool to tackle the real world issues, such as accessing benefits, or finding suitable nurseries or schools, as well as inner world issues, such as dealing with the loss of the child you thought you had and re-evaluating feelings about disability.

Table 5.4 Coping strategies as a part of the parental journey

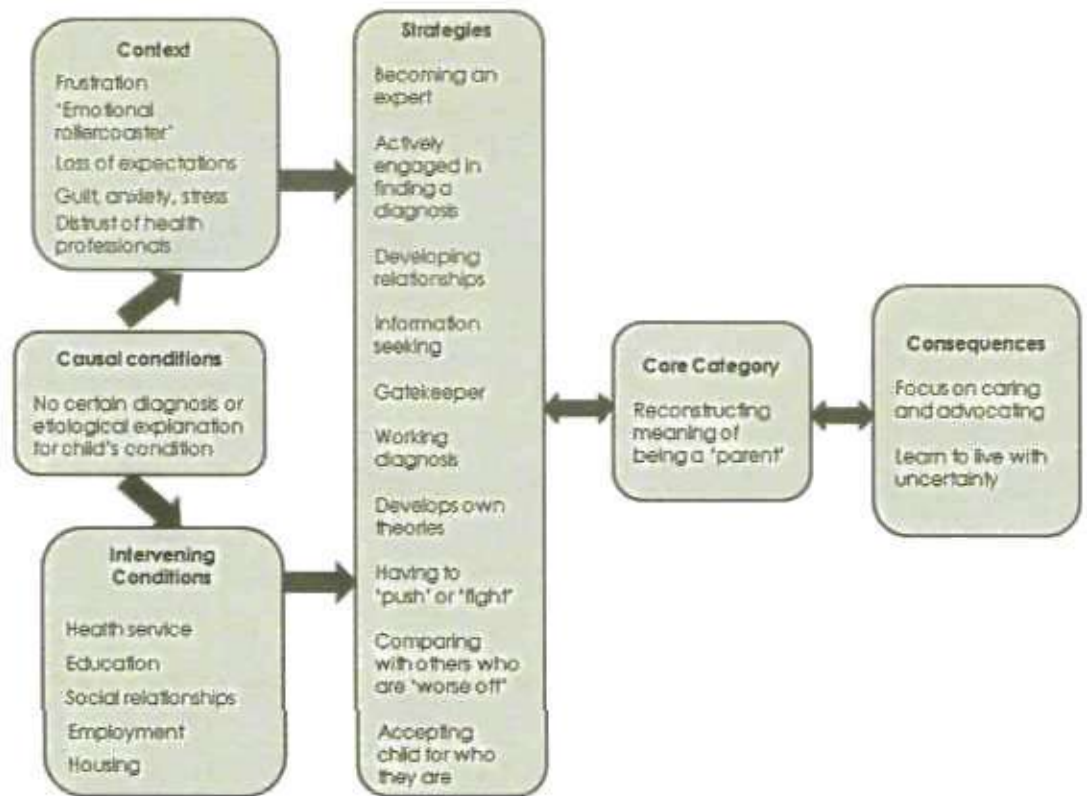
Problem experienced	Empowerment strategy
Problem with fetus identified during routine scan. Advised to terminate pregnancy	Parent requests second medical opinion
Child not reaching certain milestones. Concern about being an "overly worried mother"	Parent networks with other parents through Internet and support groups to discuss concerns
No speech and language therapy available at local hospital	Parent lobbies to get service funded
Parents notice new aspect of their child's condition	Parent contacts consultant to discuss the observation. Engage in mutually active decision-making
Not having a diagnosis leads to problems accessing services or benefits	Parent develops a 'working diagnosis' with the professional support team
Poor communication between health professionals	Parent carries folder with all correspondence to appointments. Keep professionals 'in the loop' regarding tests and consultations

Identification of a central phenomenon

There were a number of key categories considered important to the interpretation of the parental experience. These included 'frustration', 'empowerment strategies', 'learning to live with uncertainty' and 'parent engaged in caring'. Empowerment was a central and unifying experience for parents in this sub-study, as it had been for the participants interviewed in the carrier sub-study. This finding therefore confirmed empowerment as a key outcome of clinical genetics in a specific area. Yet whilst empowerment was a central finding, there was, I believed, a more profound parental experience occurring. In order to achieve a more overarching explanation, I went back to the data to establish further links among categories, using the paradigm model

(Strauss and Corbin, 1998) to identify the conditions, actions, interactions, and consequences in the data. Visualising the data in this way helped me to recognise that there was a dynamic relationship between the empowerment strategies parents employed and their experience of being a parent, and that this was central to the story. With this in mind I went back to the data to look for clues to help decipher what was happening as parents empowered themselves to advocate for their children. Laura's comment about having to 'learn' to be a certain type of person (ND3a L437) struck me as central. There was a shift or adaptation occurring for these parents who were having to "re-learn" what it meant to be a parent. This was either because the experience was so different from a previous one (if they had more than one child), or it was different from the experiences of parenting other friends and/or family had had. I conceptualised this central idea as 'reconstructing the meaning of being a parent'. I then went back to the data to ensure that the other categories logically fitted with this major category.

Figure 5.2 The central phenomenon



In order to verify the core concept I used the flip-flop technique (Strauss and Corbin, 1998) to obtain a different perspective on the key finding. I asked myself how this experience was different from the 'normal' parenting role in which parents care for their children. To help answer this question, I went back to the data to search for answers. I was particularly interested in re-reading the interviews from parents who had previous parental experience before the birth of the undiagnosed child, to find out how their perceptions of parenting had changed. If reconstructing the meaning of being a parent was the phenomenon that was occurring here, to what extent did they have to reconstruct this meaning as a result of caring for an undiagnosed child? Comments made during the interview with Julia and David helped answered this question. Julia had discussed how her first child Zoe, had gone through life "problem free" and that because of this, "every time Nick did something

unusual we didn't know if it was something medical" (ND8 L572). She then goes on to say:

"I think this whole process is a very steep learning curve because you learn about your child, about the development of your child and that it's not going the way it's supposed to be going in terms of milestones and you think 'oh it's not going the way it's supposed to be going', especially when you've had a child who has done it so easily." (ND8 L627)

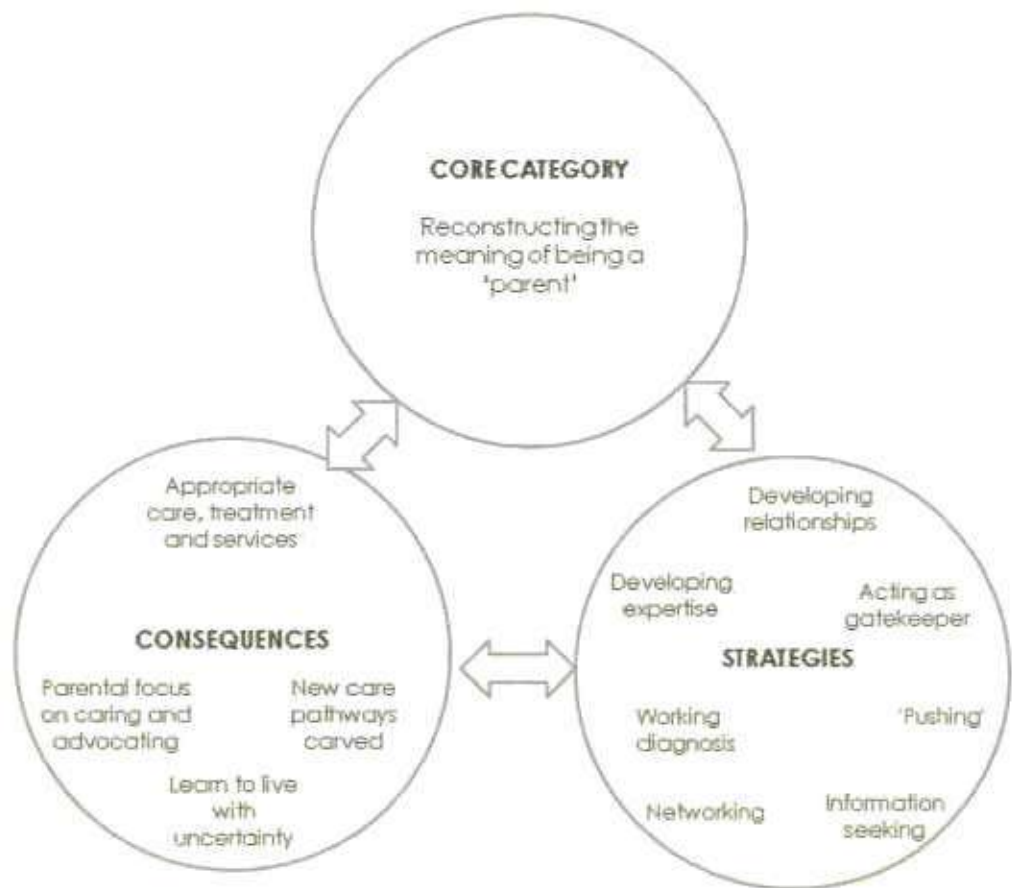
This comment clearly articulated how different the experience of parenting was for Julia the second time around. The experience was 'problem free' the first time because for 'normal' children, clear pathways in terms of child development, health, education etc. exist. However, children with rare or undiagnosed conditions don't fit into this regular care pathway because they are not progressing 'the way it's supposed to be going'. They require different pathways. However, for these conditions, such pathways don't exist. Parents are therefore left to carve these pathways for themselves, a process which is a 'steep learning curve'. In this sense, these parents are pioneers. They have no model, from their peers or their own parents, to copy or reject. This is one of the reasons many of them joined support groups. Support groups provided the opportunity for parents to gain knowledge and skills from other parents (Mary's comment about being told by another parent to put food on a white plate is a good example of this). Furthermore, parents learnt to fight and be advocates for their children in order to ensure that these new pathways were developed and that their children received the services, care and treatments they deserved.

Reconstructing the meaning of being a parent; a dynamic process

In the final stage of theory development, I decided that the consequences were not in fact an 'end point' in the parent journey, but were part of an ongoing dynamic process. The core category, strategies and

consequences were interrelated, continuously feeding back into each other; parents became empowered as a result of creating new pathways, such as developing relationships. This, in turn, enabled them to push for additional services, develop further expertise and become further empowered. I therefore developed a new and final model which highlighted this dynamic process.

Figure 5.3 Final model showing the parental experience of raising a child with no diagnosis



5.5.4 Comparing theory to findings of Rosenthal et al. and Graungaard and Skov (2006)

Many of the strategies identified in this study were also evident in the findings of Rosenthal et al. (2001) and Graungaard and Skov (2006). Graungaard and Skov highlighted that parents became advocates for their child's care through information seeking, learning new skills, becoming active

participants in their child's care and treatment, adopting the role of the gatekeeper and facilitating the diagnostic process. Parents were also forced to 'fight for the child' (p304) in order to ensure investigations were carried out quickly. Emotional coping strategies included perceiving the child in a positive light and retaining hope. Rosenthal et al. identified similar coping strategies. Parents were active in ensuring their child received appropriate services, wanted as much information as possible, and developed networks with other parents in order to learn strategies for procuring services and obtain emotional support. Fighting for services from school districts and therapeutic services as well as for assistive equipment was also an issue. In addition, parents expressed an evolving acceptance of their child's limitations, a finding similar to the consequence I identified in which parents learnt to live with uncertainty. Whilst none of the authors discuss their findings explicitly in relation to parents reframing their role, both studies do give examples of parents developing strategies in order to care effectively for their child.

5.5.5 Interpretation of the findings through existing theories

Well established theories of coping can also be used to help explain the findings from this study. Miller (1987) describes two styles of coping when confronted with a threatening situation. There are the monitors, who seek to reduce threat by seeking as much information as possible, and there are the blunters, who avoid information. In this study, information seeking was an important strategy adopted by parents. In this respect, the participants in this study could be described as 'monitors' because their method of coping was to actively seek information. In addition to information seeking, other behavioural coping strategies employed by parents included developing relationships and networking with professionals and other parents and becoming the 'expert' in

the child's condition. From an emotional perspective parents learnt to accept and enjoy their children for who they were, and remain in a positive mindset. These coping strategies are very similar to those described by Lazarus and Folkman (Lazarus and Folkman, 1984). They describe coping as consisting of two different strategies. There is problem-focused coping, which is 'coping that is directed at managing or altering the problem causing the distress', and emotional-focused coping, which is 'coping that is directed at regulating the emotional response to the problem'. Parents used both these strategies in this study suggesting they were successfully coping with their situation.

The term 'coping' however, describes a parent that is just managing or surviving. Empowerment is a more accurate description of the parents in this study because it denotes more than just coping. Funnell et al. describe an empowered patient as someone who has "the knowledge, skills, attitudes, and self-awareness necessary to influence their own behaviour and that of others...to improve the quality of their lives" (Funnell et al., 1991), attributes that parents in this study had acquired.

The theory of the need for cognitive closure (NFCC) is another theory that can be used to interpret the findings. The theory was developed as an explanation for the cognitive-motivational factor that underlies the way people interpret and respond to their social environments (Webster and Kruglanski, 1994). People at the high end of the continuum are motivated to search for certainty and have a low tolerance for ambiguity, and those at the low end of the continuum prefer uncertainty and have a high tolerance for ambiguity. Whilst some parents indicated that a diagnosis was no longer their main priority, participants in this study were still actively searching, indicating a desire for certainty. A diagnosis was seen as a means of having a prognosis, would provide psychological relief, and for some parents would resolve issues of

personal responsibility. Further investigations using the Need for Certainty Scale would help to confirm how strong this need was for parents of undiagnosed children, however the findings from both Graungaard and Skov (2006) and Rosenthal et al. (2001) also imply that parents without a diagnosis are searching for certainty.

The need for certainty has been identified as a motivator for individuals seeking genetic counselling in other studies. In a study by Skirton (Skirton, 2001), in which the need for certainty was assessed in users of a clinical genetic health service, it was found that clients preferred predictability to unpredictability, and were uncomfortable with ambiguity. Discomfort with ambiguity was significantly higher when the participants' main concerns centred on their children and need for closure was also verbally expressed by participants during interviews. Whilst it is the case that the participants in this study were found to have developed empowerment strategies, were searching for certainty and appeared to conform to the monitoring style of coping described by Miller (1987), it may not be the case that all parents of undiagnosed children are coping as effectively. Those who do not seek certainty and are 'blunters' may be less likely to participate in this kind of research.

Finally, the principles of information seeking described by Harris and Dewdney (1994) help to explain the manner in which participants sought information. Harris and Dewdney describe several principles of information seeking, including that people seek information in familiar and comfortable patterns, prefer their information to be accompanied by emotional support, often follow an informal to formal continuum, and seek information in an opportunistic way. These principles might explain some of the ways in which participants in this study gathered information. Alison, for example, who was

told her expected baby was at high risk of Down syndrome, sought information from colleagues and talked to a friend who was a social worker instead of first speaking with health professionals or consulting interpersonal sources such as the Internet. Julia frequently discussed the importance of speaking with other parents to find out about how other children developed, and whether she was being an 'overtly worried mum'. Similarly, Tony and Mary found chat forums useful sources of information because they could speak with other parents around the world. By gathering information in the first instance through informal sources such as friends, other parents or trusted professionals (such as the health worker), parents were able to gather information in an informal yet emotionally supportive setting.

5.6 Conclusion

Parents with undiagnosed children undergo a complex and frequently difficult journey. In order to manage this situation effectively, parents develop numerous strategies to empower themselves. These include actively participating in the search for a diagnosis, information gathering, and establishing mutually active relationships. During this process, parents carve new care pathways for their child, and are more able to access appropriate care, treatment and services. Furthermore, during this process, the previous anticipation of what it means to be a parent is lost. So begins a new trajectory, whereby parents begin to reconstruct the meaning of what it is to be a parent, a main theoretical finding from this study. In the next chapter I will discuss how the findings from the interviews were used to inform the development of the patient information resources.

Chapter Six:

Development of the Educational Resources

6.1 Introduction

In this chapter I will present the method used to develop the information resources for patients and families. I will discuss the reason why a booklet format was considered most suitable, describe the aims and objectives of the booklets, and present the method by which the content was developed. Furthermore, I will set out the procedure used to test and revise the booklets.

6.2 Setting the Scene

Patients are taking a greater interest than ever before in managing their health (Coulter, 1997). Printed information, such as leaflets and booklets, is an important part of the information giving process, and has been shown to improve information retention by up to 50% (Entwistle and Watt, 1998, Macfarlane et al., 2002), enhance the communication process between patient and health professional (Bucker et al., Cheung et al., 2007), improve patient knowledge (Arhan et al., 2009, Bucker et al., 2010, Scala et al., 2008), and reduce anxiety (Kutlurkan et al., 2010). In the context of genetics, used alongside verbal communication, written information has been shown to improve patient satisfaction (Austoker and Ong, 1994, Mancini et al., 2006, Roshanai et al., 2009) and support the decision-making process (Mancini et al., 2006). With genetic literacy among the general population being relatively low (Skirton and Eiser, 2003), written information is essential to support verbal communication with a genetic specialist. Furthermore, genetic mutations identified in individuals have implications across extended families. Written

information can provide important information about risk and inheritance to those who may not themselves have seen a genetic specialist.

As outlined in Chapter One, this study consisted of four phases. In Phase 1, a systematic review was conducted to identify key themes which could be included in the information resources. During Phase 2, further themes were identified through in-depth interviews with service users. Phase 3 involved further development of the content of the resources through a review of existing patient information and through consultation with key stakeholders including patient group representatives, health professionals and interviewees. In Phase 4, the resources were piloted with service users through genetic clinics. The remainder of this chapter details Phase 3 of this study.

6.3 Aims and Objectives

The aim of Phase 3 of this study was to develop evidence-based resources for: (1) people who have been identified as carriers through genetic testing; and (2) parents of undiagnosed children; which provide both scientific and psychosocial information to the reader. Moreover, they should:

- enhance the communication between service user and the genetic specialist through reiterating some of the important genetic concepts and through the use of diagrams;
- ensure that scientific information, such as the function of genes and chromosomes or genetic inheritance patterns, are presented in a way that is clear and understandable;
- provide psychological and social information, and highlight these through the use of personal quotes;
- reduce any anxiety that might exist as a result of either receiving carrier test results or being the parent of a child without a diagnosis,

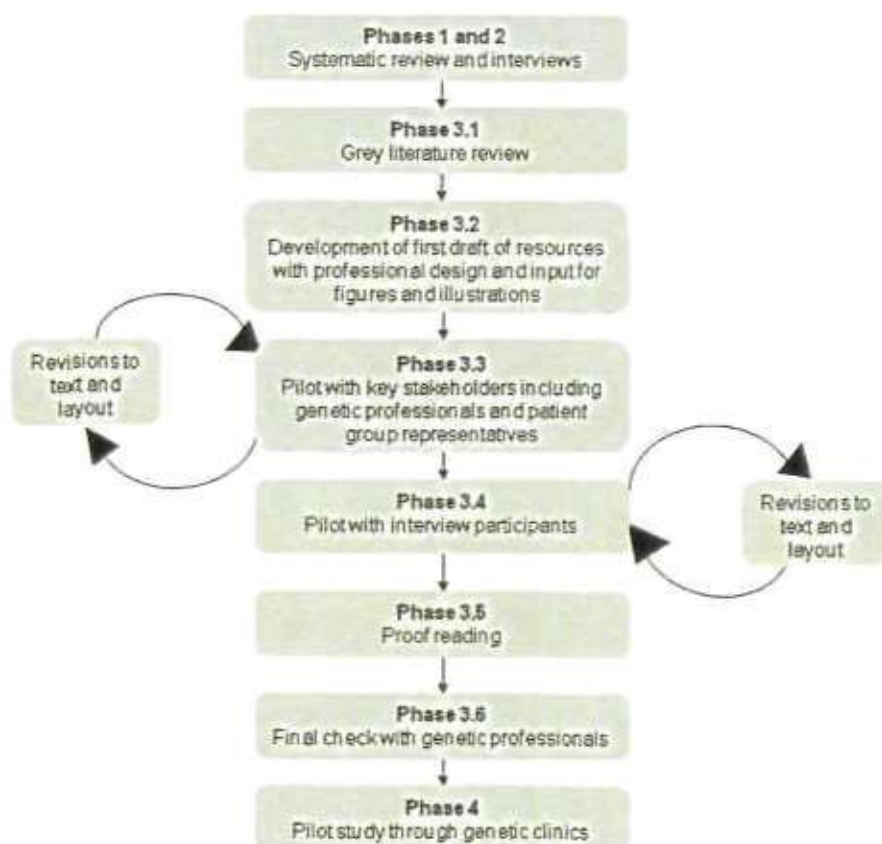
by providing service users with information that is clear, accurate and useful, and by directing them to further information sources that may be of interest, such as patient organisations, charities and government bodies;

- ensure that the information is relevant, effective and satisfies service users' information needs;
- ensure the language and design of the content is suitable, clear and logical; and
- ensure the resources are available in a medium that is easily accessible to health professionals and patients, and keep development and reproduction costs to a minimum.

6.4 Methods

Details of the four phases of this study are provided in Figure 6.1.

Figure 6.1 Overview of the development process



6.4.1 Grey literature review (Phase 3.1)

A number of key themes and issues to inform the psychosocial content of the resources had already been identified through a systematic review of the literature and through in-depth interviews. In order to further establish the content of the resources, a review of existing information resources for patients and families was conducted via a web-based search in April 2009 and through a catalogue search of patient information available at the Genetic Alliance UK.

Carrier resources

Due to the large amount of information on the Internet that related to the impact of carrier testing and on being a carrier, only patient information that had been developed either by a patient organisations or by a clinical genetics centre was reviewed. Information relating to a variety of condition types was included. The resources included in the review are presented in Table 6.1.

Whilst reading through each resource, the key issues that related to the impact of carrier test results and living as a carrier were recorded (Appendix 12). Issues relating to making a decision about carrier testing and the testing procedure were not included because the booklet was aimed at people who had already been identified as carriers through testing. This was because the majority of themes derived through the systematic review and the interviews related to the experience of being identified as a carrier, and not the decision-making process beforehand. Once this process was complete, the issues were grouped together into main themes. A comparison was then made between the issues identified across all three phases. These are presented in Table 6.2. There was significant overlap, highlighting the reliability of the findings. This can

be seen by the repeated themes occurring across all three columns in the table.

Table 6.1 List of organisations and resources identified for grey literature search on being a carrier

Organisation	Location
CF Trust – 'Genetics'	www.cftrust.org.uk/aboutcf/publications/booklets/Genetics-0107.pdf
Cystic Fibrosis Foundation – 'Genetic Carrier Testing'	www.cff.org/AboutCF/Testing/GeneticCarrierTest/
Women's & Children's Hospital, N Adelaide – 'Carrier Testing for Cystic Fibrosis'	www.wch.sa.gov.au/services/az/divisions/paedm/metabolic/documents/cf_pamphlet.pdf
Haemophilia Foundation Australia – 'On Being a Carrier'	http://www.haemophilia.org.au/documents/item/54
The Haemophilia Society UK – 'Haemophilia Gene Carrier'	www.haemophilia.org.uk/index.php?content_id=63&parent=279
New York University Medical Centre – Carrier Testing for Ashkenazi Jews (video)	www.med.nyu.edu/pediatrics/genetics/clinical/carrier.html (no longer available)
New York University Medical Centre – 'About Carrier Testing'	http://pediatrics.med.nyu.edu/genetics/clinical-services/carrier-testing/about-carrier-testing
The National Fragile X Foundation USA – 'What Does It Mean To Be A Carrier?'	http://www.fragilex.org/html/carriers.htm
Unique: The Rare Chromosome Disorder Support Group – 'Balanced Translocations'	http://www.rarechromo.org/information/Other/Balanced%20translocations%20FTNW.pdf
Guy's and St Thomas' Hospital – 'Translocations'	Appendix 11
Muscular Dystrophy Campaign – 'Carrier Tests and Prenatal Diagnosis' and 'Genetic Issues'	www.muscular-dystrophy.org/how_we_help_you/publications/

Table 6.2 Main themes and issues identified across the 3 phases relating to being a carrier

Main Themes	Systematic Review	Interviews	Grey Literature
Scientific background		<ul style="list-style-type: none"> ▪ Risk and inheritance ▪ Impact on health 	<ul style="list-style-type: none"> ▪ Risk and inheritance ▪ Impact on health ▪ Genes and chromosomes, gene mutations ▪ Inheritance patterns ▪ How common it is to be a carrier
Emotional impact	<ul style="list-style-type: none"> ▪ Guilt ▪ Anxiety ▪ Relief 	<ul style="list-style-type: none"> ▪ Guilt ▪ Relief ▪ Difficulty adapting to information 	<ul style="list-style-type: none"> ▪ Guilt ▪ Difficulty adapting to information ▪ No one's fault
Reproductive options	<ul style="list-style-type: none"> ▪ Prenatal testing ▪ Termination of pregnancy 	<ul style="list-style-type: none"> ▪ Prenatal testing ▪ Termination of pregnancy 	<ul style="list-style-type: none"> ▪ Prenatal testing ▪ Termination of pregnancy ▪ Other choices – adoption, donor egg/sperm, not having children ▪ Increased risk of miscarriage for translocation carriers
Existing children		<ul style="list-style-type: none"> ▪ Could be carriers ▪ When to discuss issue with children 	<ul style="list-style-type: none"> ▪ Could be carriers ▪ When to discuss issue with children
Partner		<ul style="list-style-type: none"> ▪ Discussing issue with partner ▪ Testing partner 	<ul style="list-style-type: none"> ▪ Discussing issue with partner ▪ Testing partner
Other family members	<ul style="list-style-type: none"> ▪ Disclosure to family 	<ul style="list-style-type: none"> ▪ Disclosure to family 	<ul style="list-style-type: none"> ▪ Disclosure to family ▪ How to inform family members ▪ Confidentiality of test results
Further information		<ul style="list-style-type: none"> ▪ Genetic clinic ▪ Patient support groups 	<ul style="list-style-type: none"> ▪ Genetic clinic ▪ Patient support groups
Other issues	<ul style="list-style-type: none"> ▪ Stigma ▪ Altered sense of identity ▪ Perception of health 	<ul style="list-style-type: none"> ▪ Stigma ▪ Altered sense of identity ▪ Perception of health 	

'Non-diagnosis' resources

Only three resources related to living without a diagnosis were identified through a review of existing patient literature, highlighting the lack of information available to parents in this position. The resources identified are presented in Table 6.3.

Table 6.3 List of organisations and resources identified for grey literature search on living without a diagnosis

Organisation	Location
Contact a family – 'Living Without a Diagnosis'	Appendix 13
Early Support – 'Information for Parents when your Child has no Diagnosis'	Appendix 14
Syndromes Without A Name USA – short information pamphlet	Appendix 15

The main issues from these resources were recorded (Appendix 16) and then compared to the findings from the parent interviews and the Graungaard and Skov (2006) and Rosenthal et al. (2001) studies. A comparison was then made between the findings. Once again, there was significant overlap, particularly between the literature review and the interviews, verifying the findings (Table 6.4).

Table 6.4 Main themes and issues identified across the three phases relating to living without a diagnosis

Main Themes	Systematic Review	Interviews	Grey Literature
Desire for a diagnosis	<ul style="list-style-type: none"> ▪ Recurrence risk ▪ Know what will happen in the future ▪ Improve care and treatment ▪ A diagnosis wouldn't change the situation 	<ul style="list-style-type: none"> ▪ Recurrence risk ▪ Know what will happen in the future ▪ Improve care and treatment ▪ A diagnosis wouldn't change the situation 	<ul style="list-style-type: none"> ▪ Know what will happen in the future ▪ Improve care and treatment ▪ A diagnosis wouldn't change the situation
Emotional impact	<ul style="list-style-type: none"> ▪ Frustration ▪ Lack of control ▪ Psychological impact ▪ Retaining hope ▪ Identifying positive aspects ▪ Acceptance ▪ Guilt/self-blame ▪ Isolation 	<ul style="list-style-type: none"> ▪ Frustration ▪ Lack of control ▪ Psychological impact ▪ Retaining hope ▪ Identifying positive aspects ▪ Acceptance ▪ Guilt/self-blame ▪ Isolation 	<ul style="list-style-type: none"> ▪ Frustration ▪ Lack of control ▪ Psychological impact ▪ Retaining hope ▪ Acceptance ▪ Guilt ▪ Isolation
Doctors and specialists	<ul style="list-style-type: none"> ▪ Difficulties accessing services ▪ Mistrust of health professionals ▪ Desire to be active partners in diagnostic process 	<ul style="list-style-type: none"> ▪ Loss of expectation ▪ Difficulties accessing services ▪ Mistrust of health professionals ▪ Desire to be active partners in diagnostic process ▪ Lack of information from professionals 	<ul style="list-style-type: none"> ▪ Difficulties accessing services ▪ Lack of information from professionals ▪ Who's who specialities ▪ Investigations
Education	<ul style="list-style-type: none"> ▪ Difficulty accessing school services 	<ul style="list-style-type: none"> ▪ Difficulty accessing school services ▪ Getting a 'Statement' 	<ul style="list-style-type: none"> ▪ Getting a 'Statement'
Family & relationships		<ul style="list-style-type: none"> ▪ Impact on relationship 	

Main Themes	Systematic Review	Interviews	Grey Literature
Empowerment strategies / advice from parents	<ul style="list-style-type: none"> ▪ Acknowledged as 'experts' ▪ Information seeking ▪ 'Fighting' ▪ Developing networks ▪ Active participant in care and treatment ▪ Negotiating services 	<ul style="list-style-type: none"> ▪ Acknowledged as 'experts' ▪ Information seeking ▪ 'Fighting' ▪ Developing networks ▪ Active participant in care and treatment ▪ Negotiating services ▪ Developing theories ▪ 'Working diagnosis' ▪ Keep a diary ▪ Make a list of questions ▪ Be persistent ▪ Have a support letter from clinician confirming symptoms 	<ul style="list-style-type: none"> ▪ Developing networks ▪ Negotiating services ▪ Keep a diary ▪ Make a list of questions ▪ Be persistent ▪ Have a support letter from clinician confirming symptoms ▪ Take a friend to appointment
Information and support	<ul style="list-style-type: none"> ▪ Lack of information ▪ Unable to join specific support group 	<ul style="list-style-type: none"> ▪ Lack of information ▪ Unable to join support specific support group ▪ Other support groups contacted and useful ▪ Support from health visitor ▪ Importance of support and advice from other parents 	<ul style="list-style-type: none"> ▪ Importance of support and advice from other parents ▪ Signposting to further services and support groups

6.4.2 Development of first draft of resources (Phase 3.2)

After Phase 3.1 had been completed, the first draft of each resource was developed using, where appropriate, the themes identified through the

systematic review, interviews and grey literature search. It was decided that an A5 booklet format was the most appropriate size to use, the reasons being:

- I had previous experience developing information booklets using Microsoft Office Publisher 2003;
- a booklet can be kept for future reference and shared with family and friends which is important in the context of genetics;
- it does not require access to a computer or the internet;
- it is relatively cheap to publish as it only requires A4 plain paper and a printer;
- an A5 format is easy to carry.

The content of the resources was loosely based around the key themes and issues identified during Phases 1, 2 and 3.1. Scientific information was provided at the beginning of the resource, followed by psychosocial information. Quotes from the interviews were embedded in the text to enhance understanding and give a personal account of the psychosocial themes discussed. Sentences were kept as short as possible and the text was presented in short paragraphs to promote ease of reading (The Plain English Campaign, 2002). Recommendations from the National Health Service (NHS) for producing patient information were also followed (NHS, 2003). These recommendations include using clear communication, patient-friendly text, bulleted or numbered points, heading, diagrams and pictures. White paper and black ink were used for the booklet to produce a clear contrast, and a font size of 12 was used to ensure the information was legible and suitable for general readers (NHS, 2003). Permission was provided by the designer to use a diagram of genes, chromosome and DNA that had been identified in a Cancer BACUP publication. Additional figures, which included visual representations of

inheritance patterns and chromosome translocations, were developed with an illustrator.

After the first draft of each booklet had been completed, the text was subjected to the Flesch-Kincaid Readability Test (Flesch, 1948), in which the rating is based upon the average number of syllables per word and words per sentence. The carrier booklet scored 70 on the Flesch Reading Ease Score, indicating it was easily understandable by 13-15 year old students. The non-diagnosis booklet scored 67, also indicating it was understandable by 13-15 year old students. The booklets were therefore considered to be pitched at an acceptable level for most readers.

6.4.3 Pilot with key stakeholders (Phase 3.3)

The next phase involved piloting the booklets with key stakeholders. These included genetic specialists, a clinical psychologist, a consultant paediatrician, the Health Visiting Union (CPHVA) and a number of patient group representatives. The patient group representatives were from the following organisations: Mencap UK, Syndromes Without A Name (SWAN), the Fragile X Society and the CF Trust. Draft copies of the leaflets were sent out and stakeholders responded either via email or telephone. Respondents were asked to comment on content, layout and design and encouraged to use track changes and comments boxes. Booklets were then revised in accordance with the suggestions made. Minor changes to the figures were also made with the illustrator.

Most of the suggestions made referred to the content of the booklets, a number of which have been included below by way of example.

Carrier booklet

- *"Not sure this level of detail is needed."* CF Trust. Referring to a paragraph discussing uncertainty of test results.
- *"It is good to show different reactions, but maybe this could be shorter – scenarios also seem a bit black and white."* Fragile X Society. Referring to the section 'Relationship with Partner'.
- *"I would also include here the sentence 'It is much better to do this before you are pregnant if possible, to give the medical team and the laboratory time to prepare for a possible test.'"* Genetic specialist. Referring to information about discussing prenatal testing with your doctor, in the section 'Future Children'.

'Non-diagnosis' booklet

- *"It can be a good idea to have a letter from a consultant explaining what is meant by 'undiagnosed' which you can send with the applications."* Paraphrased from telephone conversation with the founder of SWAN. Referring to the 'Advice for Parents' section.
- *"It would be useful to have an uplifting quote at the beginning of the booklet."* Mencap.
- *"Health visitors would not ordinarily refer children to consultant paediatricians, they are more likely to refer them to community paediatricians."* CPHVA. Referring to the section on the role of the health visitor.
- *"I think it is important to say, in response to the fact that parents find it frustrating having to repeat their child's life story, that in most cases it is necessary because there will be very specific things the specialist is interested in finding out that relate to his or her particular*

speciality." Paraphrased from a conversation with a genetic specialist when discussing the 'Doctors and Specialists' section.

6.4.4 **Piloting booklets with interview participants (Phase 3.4)**

After amendments had been made following consideration of responses from key stakeholders, a second pilot was conducted with interviewees who expressed an interest in reviewing the information resource. This was to obtain feedback concerning content and layout, and also to ensure that the content was relevant to interviewees' experiences (respondent validation). This is a method recommended by Strauss and Corbin (1998) to ensure the findings mirror the experience of the interviewee. A short survey (Appendix 17 and Appendix 18) was developed for each booklet incorporating some of the questions from the DISCERN tool (Charnock et al., 1999) and the Patient Satisfaction With Information tool (PSWI) (Jones et al., 1999). Text boxes were included for comments. Surveys were tested out with a number of researchers at the Genetic Alliance UK to ensure they were clear and answerable. Surveys were then posted to interviewees with a letter inviting them to participate in the survey (Appendix 19), a copy of the booklet, and a freepost envelope. Participants were asked to respond within two weeks. Those that had not responded were sent a reminder letter after three weeks. Five out of 11 (45%) participants responded to the carrier testing survey. Eight out of 14 participants (57%) responded to the Non-Diagnosis survey. Even though both members of a couple were sent a survey each and asked to respond independently, some may have completed their survey jointly, which may explain why not everyone responded. Responses to the surveys can be found in the Table 6.5 and Table 6.6.

Table 6.5 Responses to the survey concerning the carrier booklet

Description	Definitely no	Not entirely	To some degree	Definitely yes
Are the aims of the leaflet clear?		2	2	1
Does the leaflet achieve its aims?	1		1	3
Did you understand the scientific information (Section 1)?	1	1	2	1
Was the Genes, Chromosomes and DNA diagram clear?			1	4
Was the discussion about the emotional and social consequences (Section 2) of the test results useful?		1	4	
Does the leaflet provide sufficient information about where you can find additional information and support?			3	2
Is the leaflet balanced and unbiased?		1	2	2
Is the information relevant to your own experience?		1	4	
Is it clear who wrote the leaflet and when it was produced?			2	3
Do you like the overall design of the leaflet?	1	1	2	1
Based on the answers to all of the above questions, rate the overall quality of the leaflet as a source of information about carrier testing.		1	3	1

Feedback regarding carrier booklet

One person commented that the format of the text was too dense, another felt uncomfortable that she was the only spinal muscular atrophy (SMA) carrier that had been quoted. In response to these comments as well as the scores from the survey, two major amendments were made to the booklet. These were:

1. the booklet was divided into three separate booklets, each one specific to the three types of inheritance patterns discussed

(recessive inheritance, X linked inheritance and chromosome translocations) to make the scientific information clearer and less text heavy; and

2. the name of the condition was removed after the quotes from carriers so that there was no possibility that carriers could be identified (after discussion with the SMA carrier).

One concern I had related to the fact that the booklet on chromosome translocations had only been informed through one interview. In addition, many of the quotes included in the booklet had been taken from carriers of recessive or X linked conditions. To ensure that the quotes were relevant, and that the content of the booklet was appropriate for translocation carriers, the booklet was piloted with members of Unique, the patient organisation for people with rare chromosome disorders. Overall, the response to the booklet was very positive, however a number of helpful comments were suggested by members. These included some minor amendments to the text to improve readability, and the inclusion of a quote, provided by a woman who had a balanced translocation, about her experience of terminating a pregnancy.

Table 6.6 Responses to the survey concerning the 'non-diagnosis' booklet

Description	Definitely no	Not entirely	To some degree	Definitely yes
Are the aims of the leaflet clear?				8
Does the leaflet achieve its aims?			1	7
Did it tell you anything new?		1	7	
Did you feel overwhelmed with information?	4	3	1	
Did you find the information too limited?	8			
Is the information relevant to your own experience?			4	4
Is the leaflet balanced and unbiased?			1	7
Does the leaflet provide sufficient information about where you can find additional information and support?			1	7
Is it clear who wrote the leaflet and when it was produced?	1			7
Do you like the overall design of the leaflet?		1	1	6
Overall, do you think the leaflet is a useful source of information about parenting a child without a diagnosis?			1	7

Feedback regarding 'non-diagnosis' booklet

There were numerous positive comments made about the 'non-diagnosis' booklet. These included:

- *"Overall I was very impressed with the leaflet and was interested to learn of other people's opinions."*
- *"It is very clear and helpful especially to new parents with special needs, well done."*
- *"Really liked the 'advice from parents' section".*
- *"I liked the quotes from parents all the way through...the information on support groups and other useful resources at the end is a really good reference guide."*

- *"Clear print, easy to read, non-threatening."*

There were also a number of helpful suggestions on how to improve the booklet. These included: making it clearer that the booklet was produced by the Genetic Alliance UK; moving the section 'How important is a diagnosis?' nearer to the beginning; including a contents page; and including a physiotherapist in the description of health professionals parents may come into contact with. One person also felt that the booklet was rather text heavy. In response to these comments and the scores from the survey, a number of changes were made to the booklet at this point. These were:

1. adding a further four pages to the booklet and spacing out the text so that it was less dense and easier to read. Some of the larger paragraphs were split in two. A number of new illustrations were added so that the pages were not so text heavy;
2. adding a contents page at the beginning of the booklet to make it easier to navigate;
3. including the role of the physiotherapist to the list and description of professionals;
4. moving the section 'How Important is a Diagnosis?' to the beginning of the psychosocial section; and
5. providing a more detailed description of Genetic Alliance UK.

6.4.5 Proof reading (Phase 3.5)

After amendments had been made, the booklets were sent to a proof reader. Formatting and grammatical errors identified by the proof reader were then corrected.

6.4.6 Final check with genetic professionals (Phase 3.6)

A final check with genetic professionals was conducted to ensure that the final versions of the booklets were scientifically accurate. A number of small amendments to the text were made at this stage. The final version of the booklets can be found in Appendices 20 - 26.

6.5 Discussion

The development of written information materials for patients has been discussed widely in the literature. Moreover, many of the methods frequently used are evident in this study. For example, Paul et al. (2004) conducted a literature review and semi-structured interviews to elicit key themes to incorporate into an information booklet about transfer from an intensive care unit. The authors also drafted the booklet with a multidisciplinary team to ensure the content was comprehensive and reflected local services. A literature review was undertaken to investigate what information was available to patients about gastrointestinal cancer, in a study in which the development of a patient information booklet was detailed (Nicklin, 2009). Baty et al. (2003) conducted qualitative analysis of focus groups with African Americans in order to inform the content and layout of a culturally sensitive cancer genetics communication aid. The effectiveness of the materials has been evaluated through follow-up questionnaires with service users. Patient and professionals reviewed a draft booklet for patients with colorectal cancer considering therapy in addition to surgery (Jefford et al., 2005). The Flesch-Kincaid Readability Test was also applied to the booklet.

The methods employed in this study have incorporated many of those promoted as 'good practice' by other information providers. For example, the booklets comprise the three key attributes of quality healthcare information

materials, as identified by the Centre for Health Information Quality (1997): clear communication; evidence based; and the involvement of patients in the development stage. To help to ensure clear communication the guidance provided by The Plain English Campaign was used (The Plain English Campaign), this entailed presenting the text in short paragraphs with one main idea in each sentence. The booklets also signposted the reader to where they could find further information (Coulter et al., 1998).

Finally, the value of this study is that the process of information development has been systematic and transparent. Therefore, the method can be adopted as a guide to develop patient information in other areas. The next phase of this doctoral study, Phase 4, was a pilot study with patients recruited through genetic clinics. This final stage further enhanced the readability and content of the resources.

6.6 Conclusion

In this chapter I have detailed the systematic steps taken to develop a set of evidence-based information resources. It was important that the resources were developed in a way which took into account the views of stakeholders in order to ensure that they were both scientifically accurate, mirrored that patient experience and presented in a way that was clear, informative and useful. For these reasons, the resources were piloted with health professionals, support group representatives and interviewees. A crucial stage in the development of information resources is rigorous evaluation of their effectiveness and utility with service users. In the next chapter, I will outline the methodology for the pilot study used to evaluate the resources further.

Chapter Seven:

Methodology for Resource Evaluation

7.1 Introduction

In this chapter I will discuss the method used to evaluate the educational resources, Phase 4 of this doctoral study. I will look at study design, the recruitment process, study procedure for the two sub-studies (including how and why they differed) and how the measures used in the questionnaire were selected. I will then go on to discuss data collection and data analysis. The results of the pilot study will be presented in Chapter Eight.

7.2 Study Design

7.2.1 Experimental design

For this phase of the study I used an experimental design, meaning that I used a method whereby the independent variable was manipulated to see its effect on the dependent variable (Field, 2009). More specifically, I used an RCT design which is the classic experimental method and the most rigorous way of determining whether a cause-effect relationship exists between an intervention and an outcome (Sibbald and Roland, 1998). Although the 'standard' RCT design is one intervention and one control group, many have more than one intervention group (Bains et al., Jones et al., 2006).

Experimental designs have a number of key features. First, study participants are randomly allocated into either the control or the experimental (intervention) group. This means that each participant has an equal chance of being allocated into either group (Bowling, 2002). Often this is done through the use of random number tables (Pocock, 1983). Ideally, the investigators and

participants are 'blind' to which group they have been allocated (known as 'double-blinding'). If the researcher but not the participant is aware then this is known as single blind. Sometimes it is impossible to 'blind' the study, for example, if during the consent process it becomes obvious which group they are in. In this case the study is described as 'open'. The disadvantage of this type of study is that it can sometimes lead to what is known as the Hawthorne effect, whereby people alter their behaviour as a result of knowing they are part of an experimental study (Campbell et al., 1995). Even if the study is 'open', it is important that the investigator, not the health professionals involved in the care of the patient, conducts the randomisation (Bowling, 2002).

Sample size is another key feature of experimental designs. The size of the sample influences the ability of the experiment to distinguish a real phenomenon from an effect of sampling fluctuation. If the sample is too small then the phenomenon being tested is unlikely to be distinguishable from the effect of sampling fluctuation. Thus, experiments need to have large enough samples to ensure sufficient power and reliability (Boniface, 1995). The analysis is focused on estimating the size of the difference in predefined outcomes between the groups (Sibbald and Roland, 1998). If the researcher can validly infer that the results obtained were owing to the influence of the experimental variable then the researcher is said to have achieved internal validity. When it is possible to generalise the results to a wider setting then the experiment is said to have external validity (Bowling, 2002). Randomised controlled trials should also be reliable in that the procedures and measures employed should be clearly laid out so that the experiment can be replicated (Bryman, 2008). Furthermore, they should be piloted in order to 'test' the consent and randomisation process as well as the intervention and measurement of outcomes (Feeley et al., 2009).

Experimental methods possess several advantages over other types of research methods:

- as participants are randomly assigned to either a control or intervention group, the risk of extraneous variables confounding the results is minimised (Bowling, 2002);
- the experimental design is the only research design which can, in principle, show a causal link (Bowling, 2002); and
- it allows for precise control of variables (Bowling, 2002).

Yet the method also poses several difficulties:

- exposing patients to an intervention believed to be inferior to current treatment is often thought unethical (Sibbald and Roland, 1998);
- randomised controlled trials are generally more costly and time consuming than other studies (Sibbald and Roland, 1998); and
- experiments cannot capture the diversity of goals, objectives and service inputs which may contribute to healthcare outcomes in natural settings (Nolan and Grant, 1993).

Despite these limitations, I decided to use a RCT design for the pilot study as it is considered the 'gold standard' of research design in healthcare (Bryman, 2008), and has been used in similar studies assessing the impact of information resources on patients (Iconomou et al., 2006, Jones et al., 1999, Raynes-Greenow et al., 2009). In reporting the method, results and discussion, CONSORT guidelines (CONSORT, 2010) were followed.

7.2.2 Aims of the pilot study

This was a pilot study which comprised two independent sub-studies; the carrier study and the non-diagnosis study. The aim of the pilot study was to

determine the feasibility of a larger RCT of the two educational resources. The primary objectives were to:

- validate procedures and data collection;
- assess likely recruitment and dropout rates;
- check the acceptability of the intervention and the control;
- test the outcome measures to see if they are reliable and likely to be sufficiently sensitive to enable a difference to be detected between groups in the context of an RCT; and
- estimate the likely effect size and therefore the sample size required in an RCT.

The secondary objective was to:

- assess patient satisfaction with the information resources in a limited sample.

7.2.3 Participants - carrier sub-study

Participants were eligible for inclusion if they:

- were over 18 years of age; and
- had just been confirmed as a carrier of an autosomal recessive condition, an X linked condition or a chromosome translocation.

Individuals were excluded if they were:

- under 18 years of age;
- pregnant at the time of receiving their test results (as their primary concern at this point would have been the health of the fetus and this may have skewed their questionnaire responses);
- were unable to understand the information resource, study description and/or questionnaire due to insufficient fluency in the English language;

- lacking capacity to consent to participating in the study (as decided by the genetic professional); or
- believed by the clinician to be vulnerable to undue distress by being invited to participate in the study.

7.2.4 Participants - non-diagnosis sub-study

Participants were included if they:

- were parents of children who currently did not have a certain diagnosis or aetiological explanation for their condition, and whom the genetics team consider 'undiagnosed'; and
- were parents of children who had only recently been referred to the genetics clinic, or who had been attending the genetics clinic for a prolonged period.

Individuals were excluded if they were:

- under 18 years of age;
- unable to understand the information resource, study description and/or questionnaire due to insufficient fluency in the English language; or
- parents whom the clinician believed would be caused undue distress by being invited to participate in the study.

7.3 Ethical Approval

The study was reviewed and approved by the Camden and Islington Community Research Ethics Committee of the National Health Service in March 2010 (Appendix 27). To ensure that any ethical issues that might arise were addressed appropriately, a number of procedures were put in place.

7.3.1 **Informed consent**

The patient information sheet (Appendices 28 - 30) was sent to potential participants explaining the study. It was made clear that participants could withdraw from the study at any time without giving a reason. My contact details were also included at the end of the information sheet, and potential participants were encouraged to contact me if they had any further questions about the study.

7.3.2 **Freedom from coercion**

To ensure potential participants did not feel under any pressure to take part, terms such as 'if you would like to participate' and 'willing to help' were used in the information sheet and invitation. It was also made clear that whether or not they agreed to participate, neither they nor their child's health care would be affected in any way.

7.3.3 **Confidentiality**

It was explained in the patient information sheet that questionnaire responses would be treated as confidential and that the only person that would have access to responses would be myself. Furthermore, it was clarified that personal details would be kept secure and not passed on to anyone else.

7.3.4 **Recruitment Process**

Before I applied for ethics approval, the clinical staff at the South East Thames Regional Genetics Service were informed of the study. They agreed to participate and act as the lead genetics department. The study procedure was discussed with a staff member prior to submission for ethical approval, to ensure that it was logistically viable and all ethical issues had been addressed. After ethics approval was granted, seven other regional genetic centres across

the United Kingdom were contacted to see if they would be willing to participate in the study. Meetings were arranged with two regional genetic centres that were interested in participating: North East Thames Regional Genetics Service and Yorkshire Regional Genetics Service. Staff of both agreed to participate and the study was approved by the Research and Development (R&D) departments of all three genetic services (Appendices 31-33).

Recruitment began as soon as R&D approval was given (South East Thames Regional Genetics Service - May 2010; North East Thames Regional Genetics Service - August 2010; Yorkshire Regional Genetics Service - September 2010). Potential participants were invited to take part in the study by letter. The lead genetic counsellor who had taken responsibility for the study at each genetic centre accepted responsibility for ensuring the department was aware of the study and that invitation letters were being sent out, as per the inclusion criteria. Potential participants were sent an invitation letter detailing the clinic's involvement in the study (Appendix 34 and Appendix 35) by their genetic counsellor or medical consultant and a patient information sheet. The patient information sheet provided information concerning:

- what the study was about and why it was being conducted;
- the study procedure if the participant agreed to take part;
- issues concerning confidentiality;
- withdrawing from the study;
- how to participate; and
- my contact details if the potential participant wanted to ask further questions.

In both sub-studies, the last page of the information sheet was a reply slip. The invitee was asked to return the reply slip in the pre-paid envelope if

they wished to participate in the study, or alternatively contact me by email, telephone or text. Participants were asked to provide their name, address and/or email so that a booklet and/or questionnaire could be sent to them.

The aim was to recruit at least 50 participants into each sub-study; each sub-study having 25 in the control group and 25 in the intervention group, which would provide enough data from the two pilot sub-studies to inform subsequent RCTs. Thirty to 50 patients is a typical size for a pilot study (Gilbert et al., 2010, Pelsser et al., 2010) and would have provided sufficient numbers to allow testing of methods and calculation of sample size for the main RCT.

7.4 Study Procedure

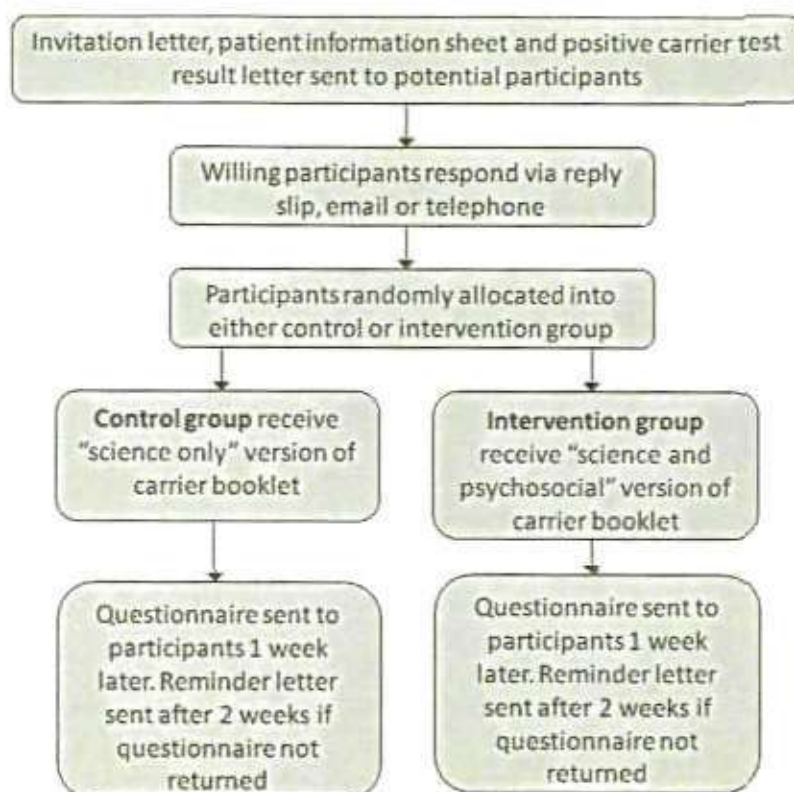
The study procedure varied for the two sub-studies due the differing nature of the two resources. The booklet for the carrier sub-study contained both scientific as well as psychosocial information, therefore I was able to make a comparison between those who received a 'science only' version of the booklet (Appendix 20, Appendix 22, Appendix 24) and those who received the complete 'science and psychosocial' version (Appendix 21, Appendix 23, Appendix 25). However, due to the fact that there was very little scientific information suitable to include in the booklet for the non-diagnosis sub-study (as without a diagnosis, one cannot describe the genetic cause of the condition), only one version of the booklet was produced and this contained mainly psychosocial information. Therefore, for the non-diagnosis sub-study I compared those who did not receive a booklet and received the standard care from the clinic (summary letter) to those who received standard care plus the new booklet.

7.4.1 Carrier sub-study procedure

1. Potential participants were invited to participate at the same time as receiving a positive carrier test result from the genetic clinic. Those who wished to participate were asked to return the reply slip or alternatively respond by email, telephone or text. Participants were asked to indicate whether they were a carrier of a recessive condition, an X linked condition or a chromosome translocation so I knew which version of the carrier booklet to send them.
2. Participants were randomly allocated into either the control group or the intervention group. Randomisation was performed through the use of random number tables, generated in blocks of ten. The generation of the number tables had been performed prior to the start of the study. Odd numbers represented the control group, even numbers the intervention group. Hence, if the numbers generated were 07, 09, 08, 06, 05, 02, 04, 01, 03, 10, the first participant would be assigned to the control group, the second to the control group, the third to the intervention group, etc.
3. Participants allocated to the control group were sent the science only version of the carrier booklet (Appendix 20, Appendix 22, Appendix 24); participants allocated to the intervention group were sent the complete (science + psychosocial) booklet (Appendix 21, Appendix 23, Appendix 25).
4. Participants were sent a questionnaire (Appendix 36) either by post or email, depending on the preference indicated on the reply slip, one week after receiving the booklet. Participants in the control group received a pink questionnaire and those in the intervention group a yellow questionnaire, to help identify to which group the

participant belonged to when the questionnaire was returned. In addition, an identifying number was added to the back of the questionnaire to help identify those participants that had responded. Those that had not responded after two weeks were sent a reminder letter.

Figure 7.1 Flow diagram showing carrier sub-study procedure



7.4.2 Discussion of study procedure

For the carrier sub-study, people were invited to participate when they received their results letter. It is well documented that anxiety is an issue that is prominent for some carriers in the first three months after receiving positive test results (Anido et al., 2005, Bekker et al., 1994), and one reason for this may be the lack of psychosocial information available. One of the aims of the pilot study was therefore to explore this hypothesis further. Participants were given one week between receiving the booklet and receiving the questionnaire, as

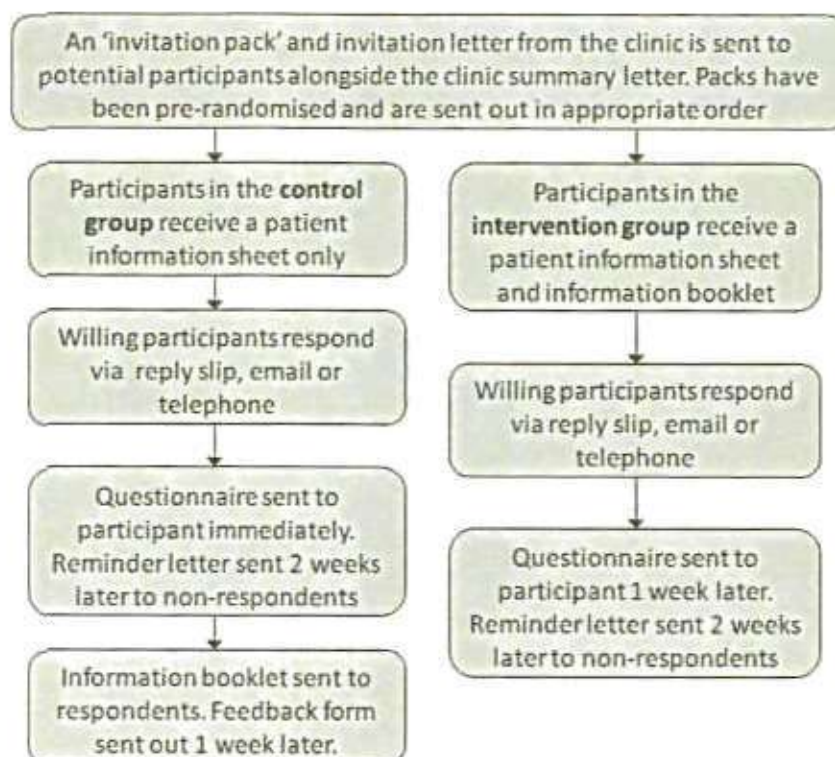
this was considered a sufficient amount of time to read and absorb the contents of the booklet.

7.4.3 **'Non-diagnosis' sub-study procedure**

1. The potential participant was sent an invitation letter from the clinic and an 'invitation pack' inside the envelope that contained the routine summary letter sent by the clinician after an appointment. The invitation packs for the control group included a patient information sheet and a pre-paid envelope only. The invitation packs for the intervention group included a patient information sheet, pre-paid envelope and the non-diagnosis booklet. Invitation packs were randomised and sequenced in the appropriate order before being given to the genetic clinic. The clinic was informed that the packs were to be sent in the correct order to ensure randomisation.
2. Willing participants were asked to return the reply slip in the pre-paid envelope, or alternatively email, telephone or text their responses. The reply slips for the control group were pink and for the intervention group yellow, to indicate to which group the participant had been allocated. Those that responded by email, telephone or text were asked what colour their reply slip was.
3. Questionnaires (Appendix 37 and Appendix 38) were sent to participants one week after agreeing to participate, to give those in the intervention group enough time to read and absorb the contents of the booklet. An identifying number was added to the back of the questionnaire so that respondents could be identified. Those that had not responded after two weeks were sent a reminder letter.

4. Those participants in the control group who had not received an information booklet were sent one after returning their questionnaire. One week later they were sent a feedback form (Appendix 39).

Figure 7.2 Flow diagram showing 'non-diagnosis' sub-study procedure



7.4.4 Discussion of study procedure

There were a number of reasons for including the information resource at the same time as the summary letter.

1. For this study, only one version of the resource existed. I therefore wanted to compare those who received the traditional summary letter only (control) with those who received a summary letter and additional information booklet (intervention) to see what impact this additional psychosocial information had.
2. Certain parents, in particular those whose children were being monitored heavily, were likely to be receiving letters from a variety of

sources in addition to the genetic clinic (such as from paediatricians, social services, etc). Sending the booklet at the same time as the standard letter, and then clarifying that parents should think about the written information they received from the genetic clinic only when completing the questionnaire, would, I felt, ensure the questionnaire results were not unduly influenced by respondents' feelings towards any other information they might have received around the time of the study. It also meant there was a shorter time frame in which they might receive written information from other sources.

3. Findings from the interviews (Phase 2) indicated that having a consultation with a geneticist and receiving a summary letter was not a particularly stressful or anxious event for parents. By the time parents were seen at the genetic clinic, they would have already been seen by a number of other health professionals along their journey, such as a paediatrician. Knowledge that their child did not have a diagnosis would have been evident for some time. Therefore, I felt that inviting them to participate in the study at the same time as receiving the summary letter would not cause unnecessary distress or anxiety. On the contrary, all the parents interviewed during Phase 2 of this study desperately wanted more information. Many resorted to searching the Internet which can give misleading or incorrect advice. The information resource had been developed with the help of genetic professionals and had been validated by healthcare professionals and patient organisations to ensure it was accurate, accessible and informative.

7.5 Questionnaire

A questionnaire, which included demographic questions and validated psychological measures, with mainly pre-coded response choices, was used to measure the impact of the interventions. This method was considered most appropriate because it ensures that all questions are presented to respondents in the same way, with no variation in question wording, and facilitates the collection of unambiguous and easily analyzable responses. Furthermore, compared to conducting face to face interviews, it is relatively economical, therefore enabling large samples of participants to be included in the study (Bowling, 2002). One of the disadvantages of using a structured questionnaire, however, is that pre-coded responses may not be sufficiently comprehensive to fully represent respondents' views. Therefore, free text boxes were provided so that respondents had the opportunity to add comments relating to the accessibility and comprehensibility of the resources.

7.5.1 Measures used

This was an exploratory pilot study in preparation for a subsequent RCT. A key outcome being explored was the impact that receiving the information resource had on participants. However, due to the exploratory nature of the study, it was not entirely clear what impact the psychosocial interventions would have on participants and therefore what the most suitable measures to use were. Initially, a number of measures were selected that I felt assessed the prominent psychosocial experiences identified in the three previous phases of the study, such as anxiety for the carrier sub-study. I also selected measures that had been used in similar studies in genetics. The measures selected were then assessed for their suitability for each of the sub-studies and those considered most suitable were chosen (Table 7.1).

Table 7.1 Identified and selected measures for the two studies

Name of measure	Description of measure	Carrier sub-study	Non-diagnosis sub-study	Reason for decision
Hospital Anxiety and Depression Scale (HADS) (Zigmond and Snaith, 1983)	<p>This is a 14-item measure of self-reported emotional distress comprising two subscales, each containing seven items assessing current levels of anxiety (HADS-A) and depression (HADS-D). A review of 71 studies using HADS found that it performed well in assessing both anxiety and depression in non-psychiatric hospital patients and in the general population.</p> <p>Cronbach's alpha for HADS-A varied from 0.68 to 0.93 and for HADS-D from 0.67 to 0.90 (Bjelland et al., 2002). This measure has also been used in studies assessing the psychological impact of genetic counselling, for example for cystic fibrosis (Chapman and Bilton, 2004).</p>	√	x	<p>Whilst anxiety and depression were not emotional experiences explicitly evident from the carrier interviews, expressions including shock, cross and upset were commonly used to explain the initial reaction of receiving positive test results, which may suggest some form of depression or anxiety. Furthermore, anxiety did feature as a key issue in the systematic review, even though it was found to dissipate in the vast majority of carriers by six months. I felt therefore that it would be interesting to measure anxiety and depression in this phase of the study, first to see if there was any evidence that it did exist for carriers in this study, and secondly to see if the provision of psychosocial information had any effect in dissipating this psychological reaction.</p> <p>Due to the similarities with the GHQ-12 scale, and because I wanted to look at other aspects such as coping and impact of event, I decided not to include this measure in the non-diagnosis sub-study.</p>

Name of measure	Description of measure	Carrier sub-study	Non-diagnosis sub-study	Reason for decision
Patient Satisfaction with Information (PSWI) Scale (Jones et al., 1999)	<p>Scale that measures patient satisfaction and understanding of information. Whilst the validity of this scale has yet to be assessed, versions of it have been used successfully in a number of studies (Jones et al., 1999, Jones et al., 2001, Jones et al., 2006).</p> <p>After discussion with the author it was decided to omit a question related to information accessibility, as this question applied mainly to web-based information, and add one further question to identify whether participants had shown the booklet to anyone else. This question was included because of the importance that genetic information has for other family members. Text boxes were also added so that participants could record any further thoughts or comments they had about the booklet.</p>	√	√	<p>Other identified scales that are used to assess the quality of written patient information relate specifically to treatment choice (Charnock et al., 1999) or a genetic test (Shepperd et al., 2006). The PSWI scale is more general in its remit and suitable to assess patient satisfaction with information about being a carrier and living without a diagnosis. This scale was included in the questionnaire sent to the intervention group (who received the booklet) but not in the questionnaire sent to the control group (who did not receive the booklet).</p>
The Beck Depression Inventory (BDI) (Beck et al., 1961)	<p>The BDI consists of 21 multiple-choice questions which measure the severity of depressive symptoms and has been used in the context of genetics (Almqvist et al., 2003, Padua et al., 2009)</p>	x	x	<p>This tool only measured depression. Therefore the HADS tool was preferable because it measured both depression and anxiety.</p>

Name of measure	Description of measure	Carrier sub-study	Non-diagnosis sub-study	Reason for decision
State Trait Anxiety Inventory (STAI) (Spielberger, 1970)	The STAI consists of two 20-item scales measuring two forms of human anxiety: state anxiety (emotional responses to a specific stressor which are likely to fluctuate over time and may vary in intensity) and trait anxiety (stable and enduring individual differences in vulnerability to anxiety). It has been used widely in the context of genetics (Bekker et al., 1994, Decruyenaere et al., 2003), and in the context of cancer genetic testing has excellent internal consistency (Vadaparampil et al., 2005).	x	x	Although this would have been a suitable tool to use, the HADS tool was preferable because it measured depression as well as anxiety.
Positive and Negative Affect Schedule (PANAS) (Watson et al., 1988)	This is a 20-item mood scale, with high negative affect epitomised by subjective distress and low negative affect by the absence of these feelings. By contrast, positive affect represents the extent to which an individual experiences pleasurable engagement with the environment. Reliability and validity reported by Watson (1988) was moderately good. For the Positive Affect Scale, the Cronbach alpha coefficient was 0.86 to 0.90; for the Negative Affect Scale it was 0.84 to 0.87.	x	x	The scale was rejected because a large number of emotional states used in the scale, e.g. interested, excited or proud, did not relate very strongly to the emotional states identified through semi-structured interviews, and therefore I was unsure how useful the findings from the scale would be.

Name of measure	Description of measure	Carrier sub-study	Non-diagnosis sub-study	Reason for decision
<p>The General Health Questionnaire (GHQ-12) (Goldberg, 1992)</p>	<p>This questionnaire was designed to detect non-psychotic psychiatric disorder in people in community and medical settings. It is constructed to identify cases, but is also used to measure degree of disorder in users. The GHQ has been used extensively with adults assessing psychological distress in relation to a variety of stressors, such as traumatic events (Dyregrov et al., 1996).</p> <p>The 12-item GHQ (GHQ-12) is easy to administer, simple, short and requires only two minutes to be completed by the respondent (Goldberg, 1985). Furthermore, it has good internal consistency ranging from 0.82 to 0.90 according to Goldberg and Williams (1988).</p>	<p>✓</p>	<p>✓</p>	<p>For the carrier study, the questionnaire was considered a useful tool to measure whether the provision of psychosocial information reduced the psychological impact of being identified as a carrier. Furthermore, the responses could be cross-checked with the responses from the Hospital Anxiety and Depression Scale (HADS) as they contain similar questions regarding depression. Similarly, for the non-diagnosis sub-study, the measure could help identify whether parents experienced any psychiatric disorder as a result of not having a diagnosis, and also identify whether the intervention had an impact on the psychological well being of the user.</p>
<p>Coping Responses Inventory (CRI) (Moos, 1988)</p>	<p>This inventory identifies cognitive and behavioural responses that individuals use to cope with a recent problem or stressful situation. The CRI measures eight types of coping responses which can be classified according to approach vs. avoidance and cognitive vs. behavioural coping strategies. Reliability and validity for most of the subscales are good (Moos, 1988).</p>	<p>x</p>	<p>x</p>	<p>The CRI was rejected because I felt it was too strategy focused, i.e. It focused on specific strategies people use to cope with a particular problem. I therefore felt it would not productively measure the impact of the booklets.</p>

Name of measure	Description of measure	Carrier sub-study	Non-diagnosis sub-study	Reason for decision
Need For Closure Scale (NFC5) (Kruglanski et al., 1993)	This 42 item scale measures a person's need for closure: a person with a high need for closure prefers order and predictability, a person with a low need will be more comfortable with unpredictability and ambiguity. It has been used in clinical genetics settings (Skirton, 2006a).	x	x	This scale was rejected on the grounds that the questions were very focused on the personality type of the respondent. It was felt that this would not sufficiently measure the impact of the intervention.
Perceived Personal Control Scale (PPC) (Berkenstadt et al., 1999)	This scale measures how genetic counselling impacts perceived personal control over health threats. The scale has been validated and the internal consistency for the total PPC was good (Cronbach's alpha = 0.79-0.81) (Smets et al., 2006).	√	x	The issues of control and empowerment were key findings from the carrier interviews conducted during Phase Two. To validate these findings the scale was considered an appropriate tool to measure counsees' understanding and perception of control over their carrier status, and, in addition, whether the provision of psychosocial information had an impact on perception of control. The questions were not appropriate for the non-diagnosis sub-study because many of the statements related to actions participants would be unable to respond to without a diagnosis.

Name of measure	Description of measure	Carrier sub-study	Non-diagnosis sub-study	Reason for decision
Impact of Event Scale (IES) (Horowitz et al., 1979)	This scale measures distress related to a specific life event by asking participants to respond to commonly reported experiences of intrusion and avoidance. It has been used numerous times in genetic counselling research for a variety of conditions (Decruyenaere et al., 2003, Keller et al., 2002) and has a good internal consistency, with a Cronbach alpha coefficient ranging from 0.89 for the intrusion subscale, and 0.79 for the avoidance subscale with a reliability of 0.87 for the total stress scores (Horowitz et al., 1979).	x	√	Analysis of the findings from the interviews identified that living without a diagnosis was a stressful life event for many parents, and therefore the scale was considered a useful method of measuring the impact of the intervention (receiving the psychosocial booklet). If participants who received the booklet were found to have less intrusive thoughts than those who did not, it would suggest that the resource reduced the impact of the stressful event. If no intrusion was shown using the scale for either group, it would indicate that not having a diagnosis did not have a major impact on parents. The scale would also have been appropriate for the carrier study, however we were already using the PPC scale to measure the impact of the carrier test result.

7.5.2 Demographic questions

In addition to these measures, a number of demographic questions were incorporated at the beginning of the questionnaire to help interpret the findings. Questions relating to sex, age, ethnic background and level of education were included. For the carrier sub-study, additional questions relating to the condition for which they were a carrier were included, e.g. whether they had any affected children. For the non-diagnosis sub-study additional questions that related to the undiagnosed child were incorporated, e.g. what statement best describes your child's health or developmental problems.

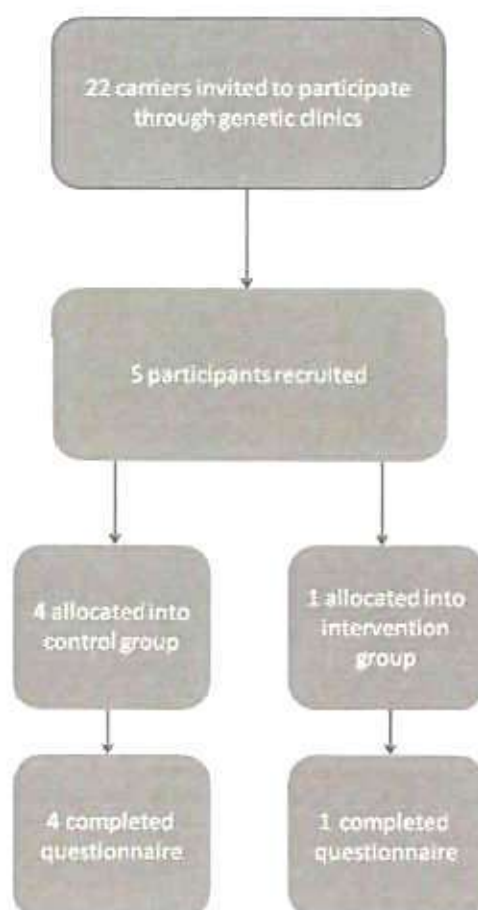
7.6 Data Collection

Data collection began as soon as R&D permission had been obtained from the respective hospital trusts, with the aim of recruiting 50 participants into each sub-study by the end of December 2010.

7.6.1 Carrier sub-study

Uptake into the carrier sub-study was slow. By the end of December 2010, 22 carriers had been invited to participate but only five had agreed to participate and had completed the questionnaire. Due to the small number of responses, not enough data were available to conduct any meaningful statistical analysis.

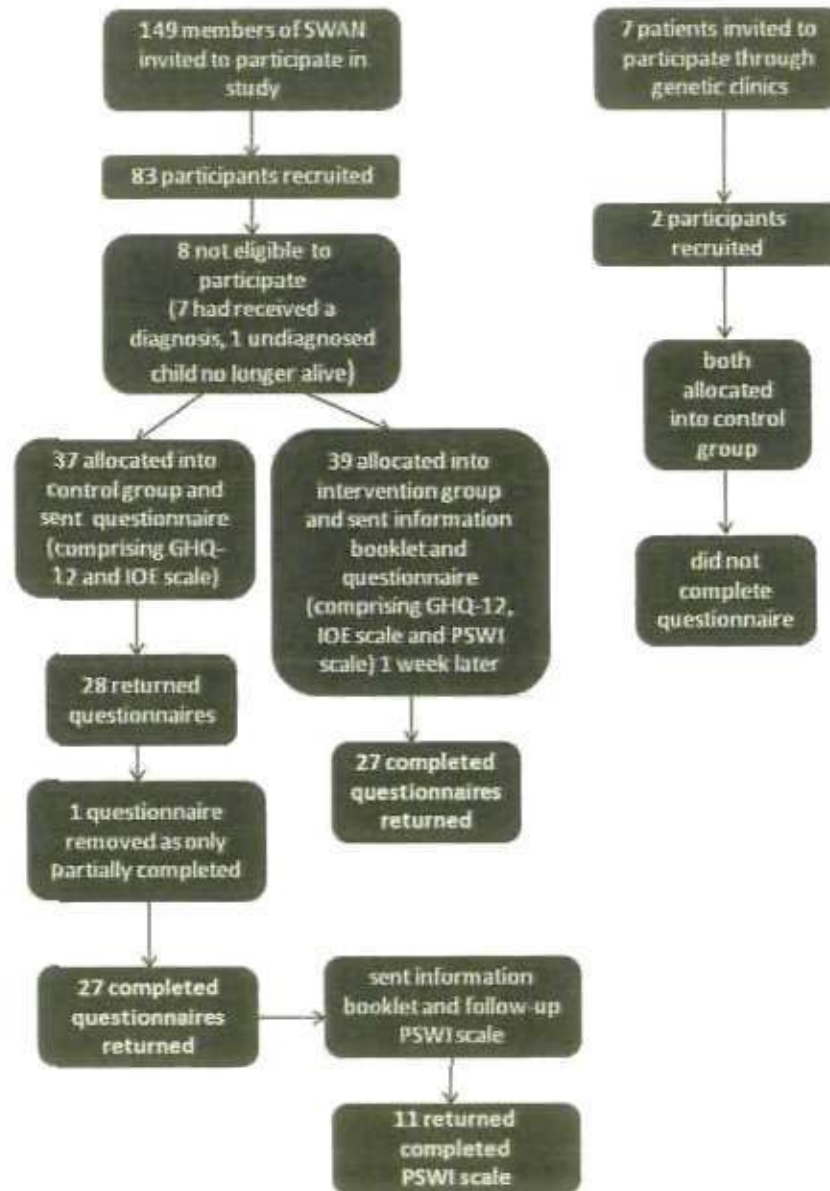
Figure 7.3 Patient flow diagram for carrier sub-study



7.6.2 'Non-diagnosis' sub-study

By November 2010, no responses to the non-diagnosis sub-study had been received, despite numerous reminder emails and telephone calls with the lead genetic counsellors at the three participating centres. I therefore decided to widen the recruitment net by inviting members of the patient group SWAN, an organisation for parents of children without a diagnosis, who had previously agreed to participate in future studies conducted by the Genetic Alliance UK. SWAN is not currently active as an organisation since the founder retired in 2008. Hence no new members have joined for the past few years.

Figure 7.4 Patient flow diagram for 'non-diagnosis' sub-study



Ethical approval was given by Camden and Islington Community Research Ethics Committee for this amendment to the study procedure (Appendix 4Q). Invitations were sent to 147 potential participants in September 2010, either via email or letter, depending on what they had previously indicated to be their preferred method of contact. Whilst the information resource and questionnaire

remained exactly the same, the study procedure was amended, with randomisation into either the control or intervention group being conducted as and when a participant was recruited (as I would not be comparing the resource with information received from the clinic). Participants were also given the option of completing the questionnaire via the online survey tool Survey Monkey (www.surveymonkey.com). The reason behind this was that in a previous survey conducted through Genetic Alliance UK, a large number of participants had chosen to complete the survey online. This option was therefore offered to ensure the highest possible rate of participation.

Questionnaire responses from SWAN members were kept separately from genetic clinic responses. By December 2010, seven participants had been invited to participate in the pilot study through genetic clinics. Two had agreed to participate but neither had completed the questionnaire. Eighty-three participants had been recruited into the pilot study through the SWAN membership. Eight were not eligible to participate, and the remaining 76 had been allocated into either the control or intervention group. Fifty-four questionnaire responses were received in total (27 in the control group, 27 in the intervention group). With no more responses expected from the SWAN members, I decided to stop recruiting for this sub-study on 2nd December 2010.

7.7 Preparation of the Data

The data gathered from the non-diagnosis sub-study was entered into the statistical software package PASW Version 18 (PASW Statistics 18, 2009). Before this could be done, the data had to be coded. This comprised assigning numerical codes to responses that were not already in numerical form, e.g. 1=males, 2 =

females. Numerical responses, such as age, remained the same. During this process, codes and abbreviated variable names were documented in a codebook. Once this had been completed, the data were checked for errors. Frequencies, minimum and maximum values, mean scores and valid and missing cases were checked and any unusual values examined and corrected if an error had been made.

7.7.1 Scoring

Scores were assigned to each of the responses on the three scales chosen for the 'non-diagnosis' sub-study outlined in Table 7.1. The scoring systems are further described below.

Impact of Event Scale (IES): For the IES, a scoring system of 0-1-3-5 (Horowitz et al., 1979) was used. The total maximum score is 75, with a maximum score of 40 for the avoidance scale and 35 for intrusion scale. The total score can be interpreted according to the following dimensions of post-traumatic stress symptoms: 0 to 8 (subclinical range); 9 – 25 (mild range); 26 to 43 (moderate range); 44+ (severe range) (Cornell et al., 1999). It is suggested that the cut-off point is 26, above which a moderate or severe impact is indicated (Cornell et al., 1999).

General Health Questionnaire (GHQ): Each response to the questions in this instrument has four possible responses, typically 'not at all', 'no more than usual', 'rather more than usual' and 'much more than usual'. There are two methods of scoring: (1) GHQ-case scoring (0-0-1-1) which gives a possible score range of 0-12; and (2) Likert scoring (0-1-2-3) which gives a possible score range of 0-36, with a higher score indicating greater levels of psychiatric distress. Likert scoring is more

useful for assessing severity than the GHQ-case scoring system, which is more appropriate for detecting cases. As the aim was to identify whether those receiving the information booklet were less anxious and depressed than those who did not receive it, the Likert scoring system was considered more appropriate. However, as most studies were found to use GHQ-case scoring, scores using this method were also calculated. Cut-offs have not been validated for Likert scoring (Johnston et al., 1995), however a cut-off score of ≥ 18 has been recommended by Hawley et al. (2003) as it signifies a high proportion of reporting at the 'rather more than usual' and 'much more than usual' level. A GHQ-case cut-off score of 3/4 was used as it was the recommended cut-off in a study of parents of children with developmental disabilities (Wong and Poon, 2010)

Patient Satisfaction with Information scale (PSWI): The PSWI was scored using a scoring system agreed with the author of the scale. A scoring system of either 1-2-3 or 1-2-3-4 was used depending on the number of responses available to choose from. In scoring negatively worded questions, scores were inverted so that the highest score always went to the most satisfactory outcome. The minimum possible score was 8, the maximum 29. Higher scores indicated greater satisfaction with the information booklet.

7.7.2 Reliability analysis

IES: According to Horowitz et al. (1979), the IES has a good internal consistency, with a Cronbach alpha coefficient ranging from 0.89 for the intrusion subscale and 0.79 for the avoidance subscale, with a reliability of 0.87 for the total scores. In this study, the Cronbach alpha coefficient showed excellent internal

consistency for the three measures ($\alpha = 0.91$ for the intrusion subscale, $\alpha = 0.86$ for the avoidance subscale and $\alpha = 0.93$ for the total scores).

GHQ: According to Goldberg and Williams (1988), the GHQ has a good internal consistency ranging from 0.82 to 0.90. In the current study, Cronbach's alpha coefficient was 0.91 using GHQ-Likert scoring, and 0.89 using GHQ-symptom scoring. This indicated excellent internal consistency for both the IES and GHQ scales (DeVellis, 2003).

PSWI: The scale has not yet been validated for internal consistency by the author. On the study sample, Cronbach's alpha coefficient was low ($\alpha = 0.57$) suggesting poor internal consistency reliability for the scale with this sample. Question seven ("Did you find the information too technical?") appeared to be problematic as it scored -0.058 on the corrected item-total correlation. Scoring was checked to ensure this question had been correctly reverse scored which it had, so it may be that this question is measuring something different to the others. Without this question, Cronbach's alpha coefficient would have been 0.626. Whilst still low, it is a more acceptable reliability statistic. This low score may also have been due to the small number of items on the scale.

7.8 Data Analysis – 'Non-Diagnosis' Sub-Study

A combination of descriptive statistics and statistical techniques were used to fulfil the objectives of the pilot study. These are summarised in Table 7.2.

Table 7.2 Statistical techniques used to analyse the data

Technique/Test	Reasons for selection
Descriptive statistics (mean scores, standard deviation, Kolmogorov-Smirnov test, histograms, boxplots)	To describe the characteristics of the sample, check recruitment and attrition rates, assess the normality of the distribution of scores and check for outliers.
Cronbach's alpha coefficient	To measure the reliability of the scales with the sample group.
Independent sample T-test	To compare the mean scores of the two groups (control and intervention) to see if they were likely to be sufficiently sensitive to notice a difference in an RCT.
Power analysis	To estimate the likely effect size and therefore the sample size that will be required in an RCT.

7.9 Conclusion

In this chapter I have discussed the design, recruitment process, and study procedure for the two sub-studies, including reasons why the study procedure for one of the sub-studies was changed midway through this phase. I have also looked at the measures selected to be used in the questionnaires for the two sub-studies and given a brief overview of the statistical techniques used to analyse the data. In Chapter Eight I will discuss the results of the pilot study.

Chapter Eight:

Results of the Pilot Study

8.1 Introduction

In this chapter, I will report the results of the two pilot sub-studies. The assumed model (to be tested in a subsequent RCT) is that by giving patients and/or parents additional psychosocial written information, their satisfaction with information received will increase, they will experience lower levels of psychiatric distress and the impact of the event will be lessened. For the carrier sub-study, this 'event' is the positive carrier test result; for the 'non-diagnosis' sub-study, this 'event' is the overall experience of parenting a child with an unknown condition. The objectives of the pilot study were therefore to prepare for the RCT by: assessing study procedure, recruitment and attrition; checking the acceptability of the control and intervention; testing the outcome measures for reliability and acceptability; estimating the likely effect size and sample size needed for an RCT; and carrying out a trial-run of the types of analysis that would be used in a definitive trial.

8.2 Recruitment and Attrition

8.2.1 Carrier sub-study

As presented in Chapter Six, twenty-two carriers were invited to participate in the carrier sub-study. Of these, five (23%) agreed to participate and completed the questionnaire. Three were in the control group; two were in the intervention group. Due to the small number of responses, not enough data were available to

conduct any meaningful statistical analysis. However, suggestions as to why the study procedure was unsuccessful will be examined in the Discussion section (8.8).

8.2.2 'Non-diagnosis' sub-study

A total of seven people were invited to participate in the study through three genetic clinics. Two participants were recruited, however neither of them completed the questionnaire.

A total of 149 members of the patient organisation SWAN were invited to take part in the study. Of these, 83 (56%) agreed to take part. Eight were not eligible to participate, either because they had received a diagnosis (but had remained members of the organisation) or their undiagnosed child was no longer alive (which was not known at the time of recruitment). One questionnaire was partially completed and therefore removed. Of the 74 eligible participants, 37 were recruited into the control group and 39 were recruited into the intervention group. Fifty-four (73%) completed and returned the questionnaire (32 [59%] online; 22 [41%] by post). There was no significant difference in rates of attrition between control group (27/37) and intervention group (27/39). Overall, 36% of the original 149 members invited to participate completed the questionnaire.

The questionnaire sent to participants in the control group did not include the PSWI scale (as those participants had not received the information booklet yet). Therefore, those who completed the first questionnaire were sent a follow-up questionnaire, which consisted solely of the PSWI scale, one week after being sent the information booklet. Eleven (41%) participants completed and returned this follow-up questionnaire, giving a total of 38 responses (70%) for the PSWI scale.

8.3 Characteristics of Participants

The demographic characteristics of the participants appear in Table 8.1. Chi-square test for relatedness found no difference between the intervention and control groups in their demographics, therefore Table 8.1 shows the whole sample. Participants ranged in age from 30 years old to 61 years old (mean = 44; median = 44).

Table 8.1 Demographic and health characteristics of participants in the 'non-diagnosis' study (N=54)

Variables	Frequency	Percentage
Relation to child (N=54)		
Mother	53	98%
Father	1	2%
Parental age (N=54)		
≤ 30 years	1	2%
31-40 years	18	33%
41-50 years	27	50%
51 + years	8	15%
Marital status (N=54)		
Married	39	72%
Living with Partner	6	11%
Single	4	7%
Divorced	3	6%
Separated	1	2%
Widowed	1	2%
Region (N=54)		
South East England	14	26%
West Midlands	6	11%
North West England	5	9%
East of England	5	9%
Scotland	5	9%
Greater London	4	7%
Wales	4	7%
North East England	3	6%
Northern Ireland	3	6%
Yorkshire and the Humber	2	4%
South West England	2	4%
East Midlands	1	2%

Variables	Frequency	Percentage
Education (N=54)		
Secondary School (GCSE/O Level)	13	24%
Secondary School/College (A Level, HND, etc)	20	37%
University First Degree (BSc, BA)	12	22%
University Higher Degree (MA, MSc, PhD etc)	6	11%
Prefer not to answer	3	6%
Total annual household income (N=54)		
≤ £20,000	10	19%
£21,000 - £40,000	22	41%
£41,000 - £60,000	8	15%
£61,000 - £80,000	5	9%
£81,000 +	4	7%
Prefer not to answer	5	9%
Ethnic background (N=54)		
White	50	93%
Mixed	3	6%
Other	1	2%
Asian	0	0%
African or Caribbean	0	0%
Prefer not to answer	0	0%
Number of children without diagnosis (N=54)		
1	48	89%
2	5	9%
3	1	2%
Age of 1st child without diagnosis in years (N=54)		
1-5 (infant/preschool)	6	11%
6-11 (child)	23	43%
12-18+ (adolescent)	25	46%
Years without a diagnosis (N=54)		
1-6	16	30%
7-12	21	39%
13-18+	17	31%
Description of (most severely affected) child's health (N=54)		
No real effect	0	0%
Some effect	8	15%
Major effect	46	85%
Do you have any unaffected children? (N=54)		
Yes	41	76%
No	13	24%

Note: Percentages may not total 100 because of rounding

8.4 Assessing Normality

A pilot study offers the opportunity to have a 'trial-run' of the analysis that would be used in a definitive study. This allows the identification of problems such failure to collect data, incorrect assumptions about the type of analysis, or identification of sample sizes needed. Analysis began with tests to assess normality, as the assumption of normality is a prerequisite for many inferential statistical techniques and it is a key assumption for parametric tests (Pallant, 2007). This assumption was first explored graphically using a visual examination of all histograms, boxplots, and normal probability QQ plots. Furthermore, a number of statistics including a Kolmogorov Smirnov statistic, skewness and kurtosis were obtained.

The histograms in Figures 8.1 and 8.3 suggest the scores for the IES and GHQ-Likert variables are reasonably consistent with a normal distribution, with most scores occurring in the centre and tapering out towards the extremes. This was confirmed by a Kolmogorov Smirnov test, which found that all tests were not significant (IES: $p = .987$; GHQ-Likert: $p = .519$). Distribution of scores using GHQ-case scoring (Figure 8.2) had a high number of scores skewed to the left indicating a high number of low scores, however, using the Kolmogorov Smirnov test this was not found to be significant (GHQ-case: $p = .138$). In contrast, the distribution of scores for the PSWI scale (Figure 8.4) appear skewed to the right indicating that most participants scored the booklets highly. However, the Kolmogorov Smirnov test found the data were consistent with a normal distribution of scores ($p = .125$). Therefore all distributions are not significantly different from a normal distribution (i.e., they are *normal*) (Field, 2009).

Figure 8.1 to 8.4 Histograms showing distribution of scores for GHQ, IES and PSWI scales

Figure 8.1

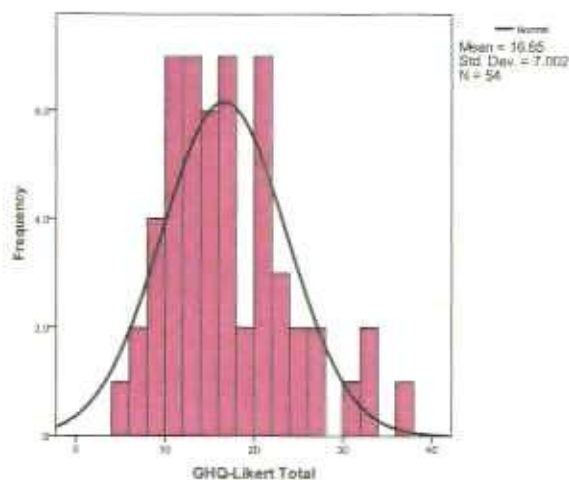


Figure 8.2

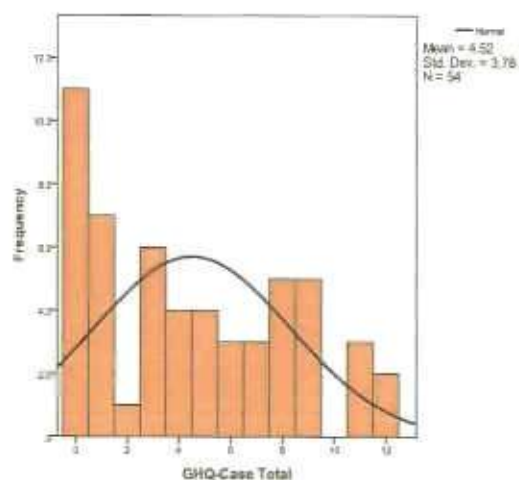


Figure 8.3

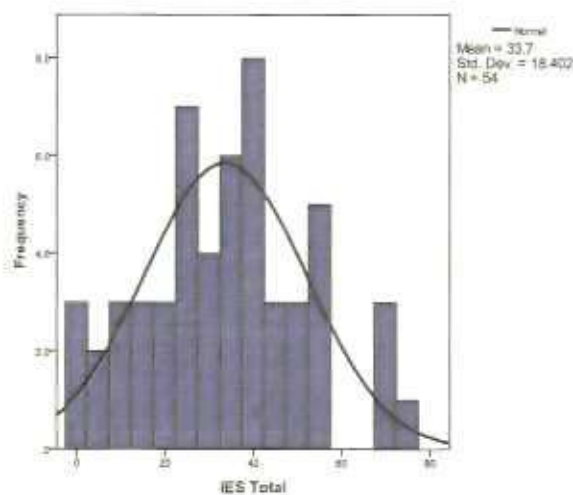
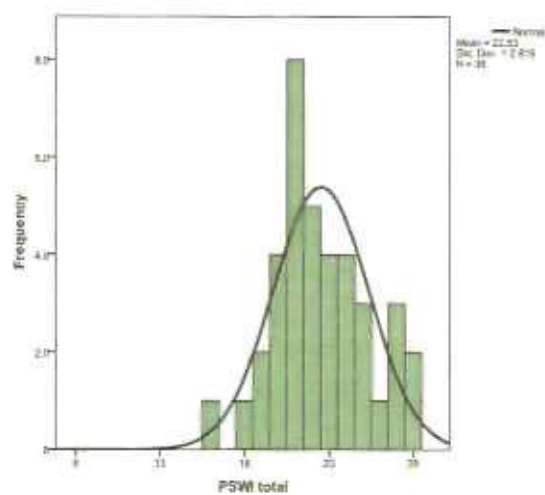


Figure 8.4



8.5 Descriptive Statistics

In this study, IES intrusion scores ranged from 0 – 35 (mean = 19.2, SD = 10), IES avoidance scores ranged from 0 – 40 (mean = 14.5, SD = 9.8), and total scores ranged from 0 – 75 (mean = 33.7, SD = 18.4) indicating a large variability in scores. Scores for the GHQ-Likert ranged from 5 – 36 (mean = 16.7, SD = 7.0), GHQ-case 0 –

12 (mean = 4.5, SD = 3.8) again, showing a large variability in scores. Scores were less variable for the PSWI scale, ranging from 16 - 28 (mean = 22.5, SD = 2.82).

Table 8.2 Descriptive statistics for IES, GHQ and PSWI scale

Variables	Mean (S.D)	Median	Min	Max	95% CI-	95% CI+	α
IES Total	33.7 (18.4)	34.5	0	75	28.7	38.7	0.93
IES Intrusion	19.2 (10)	20.5	0	35	16.5	19.4	0.91
IES Avoidance	14.5 (9.8)	13	0	40	11.8	17.2	0.86
GHQ-Likert	16.7 (7)	15.5	5	36	14.7	18.6	0.91
GHQ-Case	4.5 (3.8)	4	0	12	3.5	5.7	0.89
PSWI	22.5 (2.8)	22	16	28	21.6	23.5	0.57

Note: SD = standard deviation; α = Cronbach's alpha; CI- = lower confidence interval; CI+ = upper confidence interval;

IES = Impact of Event Scale. Fifteen item scale. Scores range from 0-75, with a maximum score of 40 for the avoidance scale (8 items) and 35 for intrusion scale (7 items) with higher scores indicating higher levels of distress

GHQ-Likert: General Health Questionnaire 12 item version, Likert scoring system. Scores range from 0-36 with higher scores indicating a greater possibility of psychiatric disorder

GHQ-Case: General Health Questionnaire 12 item version case-scale. Scores range from 0-12 with higher scores indicating a greater possibility of psychiatric disorder

PSWI - Patient Satisfaction with Information Scale. Scores range from 8-29, with a higher score indicating higher satisfaction with the information.

Min and max refers to the minimum and maximum scores in this study.

8.6 Data Analysis

8.6.1 Analysis of IES data

Scores from the IES were analysed in two ways. First, the distribution of scores was assessed according to the clinical significance of the scores (subclinical, mild, moderate or severe (Cornell et al., 1999)) to get a broader understanding of the impact of raising a child without a diagnosis and to look for

differences between the control and intervention groups. Second, the mean scores on the intrusion and avoidance scales were calculated to gain a deeper understanding of the results across the two components of the scale.

Table 8.3 Clinical significance of IES scores

Clinical significance	IES score	Control		Intervention		Total	
		%	n	%	n	%	n
Subclinical	0-8	19	5	4	1	11	6
Mild	9-25	22	6	19	5	20	11
Moderate	26-43	33	9	48	13	41	22
Severe	≥44	26	7	30	8	28	15

Note: IES = Impact of Event Scale

Percentages may not total 100 because of rounding

Table 8.4 Mean scores for IES including intrusion and avoidance sub-scales

	Control M (SD)	Intervention M (SD)	P value	Total M (SD)
IES Total (out of 70)	31.9 (21.9)	33.5 (14.1)	0.484	33.7 (18.4)
IES Intrusion (out of 35)	18.6 (11.5)	19.9 (8.5)	0.640	19.2 (10.0)
IES Avoidance (out of 40)	13.4 (11.4)	15.6 (7.9)	0.401	14.5 (9.8)

Note: IES = Impact of Event Scale; M = mean score; SD = standard deviation

Twenty-two participants (41%) scored in the moderate range of clinical significance; fifteen (30%) scored in the severe range. Thirty-seven participants (64%) scored above the cut-off point (≥ 26) for post traumatic stress disorder.

Twenty-one participants (78%) in the intervention group scored in the moderate and severe category, compared to sixteen participants (59%) in the control group.

For the IES total score, the mean score was 33.7. The mean values for the intervention group (mean = 33.5, SD = 14.1) were higher than for the control group (mean = 18.6, SD = 21.9). Similarly for the intrusion subscale (intervention: mean = 19.9, SD = 8.5; control: mean = 18.6, SD = 11.5) and avoidance subscale (intervention: mean = 15.6, SD = 7.9; control: mean = 13.4, SD = 11.4). None of these findings were statistically significant ($p < .05$). Participants scored higher on the intrusion sub-scale than on the avoidance sub-scale.

8.6.2 Analysis of GHQ data

Similar to the IES analysis, GHQ scores were analysed in two ways. First, distribution of scores was assessed according to the clinical significance (Hawley et al., 2003, Wong and Poon, 2010). Secondly, the total mean score, and mean scores for the control and intervention groups were calculated.

Table 8.5 Clinical significance of GHQ scores

Clinical significance	GHQ-L score	Control		Intervention		Total	
		%	n	%	n	%	n
Low	0-17	52	14	74	20	63	34
Significant	18-36	48	13	26	7	37	20
Clinical significance	GHQ-C score	Control		Intervention		Total	
		%	n	%	n	%	n
Low	≤3	41	11	52	14	46	25
Significant	≥4	59	16	48	13	54	29

Note: GHQ-L = General Health Questionnaire, Likert scoring. GHQ -C = General Health Questionnaire, case scoring. Percentages may not total 100 because of rounding.

Table 8.6 Mean scores for GHQ

	Control M (SD)	Intervention M (SD)	P value	Total M (SD)
GHQ-Likert score (out of 36)	17.7 (8.2)	15.6 (5.6)	0.273	16.7 (7)
GHQ-case score (out of 12)	5.0 (4.3)	4.0 (3.1)	0.355	4.5 (3.8)

Note: GHQ = General Health Questionnaire; M = mean score; SD = standard deviation

8.6.3 GHQ-Likert scale

Using GHQ-Likert scoring, the majority of participants ($n = 34$, 63%) scored below the cut-off for clinical significance, with 20 (37%) scoring above. Fourteen participants (52%) in the control group scored below the cut-off for clinical significance, 13 (48%) scored above. In the intervention group, 20 participants (74%) scored below the cut-off compared to seven (26%) who scored above.

Although not statistically significant, the mean values on the GHQ-Likert scale show that the intervention group (mean = 15.6, SD = 5.5) had lower scores when compared with the control group (mean = 17.7, SD = 8.2).

8.6.4 GHQ-Case scale

Using the GHQ-case scoring method, more participants scored above the cut-off for clinical significance than they did using GHQ-Likert scoring. Twenty-five participants (46%) scored less than four, and 29 (54%) scored four or more. In the control group, 11 (41%) scored below the cut-off for clinical significance compared to 16 (59%) who scored above. In the intervention group, 14 (52%) scored below the cut-off compared to 13 (48%) who scored above.

The mean values on the GHQ-case show that the intervention group (mean = 4.0, SD = 3.1) had lower scores than the control group (mean = 5.0, SD = 4.3), although these findings were not statistically significant.

8.6.5 Effect size

The effect size was calculated (Cohen's d) using means and standard deviations, using a free online effect size calculator (Becker, 2000). Cohen (Cohen, 1988) defined d as the difference between the means (M), $M_1 - M_2$, divided by standard deviation, σ , of either group.

$$\text{Cohen's } d = M_1 - M_2 / \sigma_{\text{pooled}}$$

$$\text{where } \sigma_{\text{pooled}} = \sqrt{[(\sigma_1^2 + \sigma_2^2) / 2]}$$

$$r_{YX} = d / \sqrt{(d^2 + 4)}$$

To interpret the strength of the effect sizes, Cohen proposes the guidelines as presented in Table 8.7.

Table 8.7 Cohen's effect sizes

Size	Cohen's d (standard deviation units)
Small	0.2
Medium	0.5
Large	0.8

IES: The effect size for the intrusion subscale ($d = -.13$), the avoidance subscale ($d = -.22$), and the total scale ($d = -.09$) were all small. Cohen's d scores for the IES were all negative because the mean difference was not in the predicted direction (the mean for the intervention groups was higher than the control groups).

GHQ-Likert: The effect size using GHQ-Likert scoring was small ($d = .29$).

GHQ-Case: The effect size using GHQ-case scoring was small ($d = .27$).

8.6.6 Sample size calculation

GPower version 3.1 software was used to calculate the sample sizes needed if this study was to be replicated using a larger sample size. The estimates in the table below are based on using an independent samples t-test (one-tailed) to test if the control group will score higher on the measures than the intervention group, using a margin of error of .05 (5%), a significance level set at .05 (5%), and a power level of .95 (95%).

Table 8.8 Table showing sample size needed to generate each effect size

Effect size	Sample size
Small (0.2)	1,084 (N1 = 542, N2 = 542)
Medium (0.5)	176 (N1 = 88, N2 = 88)
Large (0.8)	70 (N1 = 35, N2 = 35)

8.6.7 Analysis of PSWI Data

Table 8.9 Table showing significance of scores on the PSWI scale

Significance	PSWI score	Total	
		%	n
Low	8-15	0	0
Medium	16-22	55	21
High	23-29	45	17

All participants scored the booklet either in the medium or high scoring range, with the majority (n = 21, 55%) scoring the booklet in the medium range. Scoring for the individual questions on the PSWI scale can be seen in the bar charts below.

Figures 8.5 to 8.12 Bar charts showing scores for individual questions on the PSWI scale

Figure 8.5

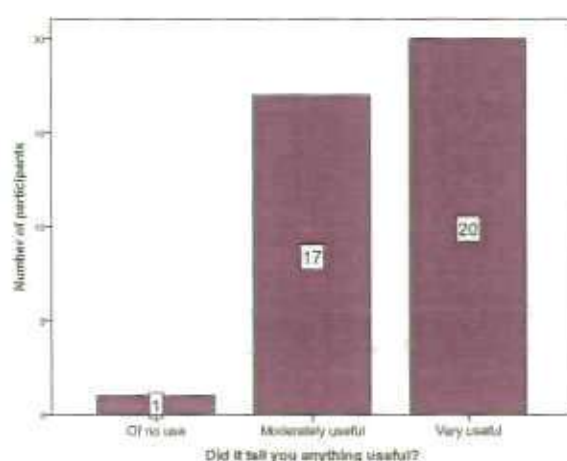


Figure 8.6

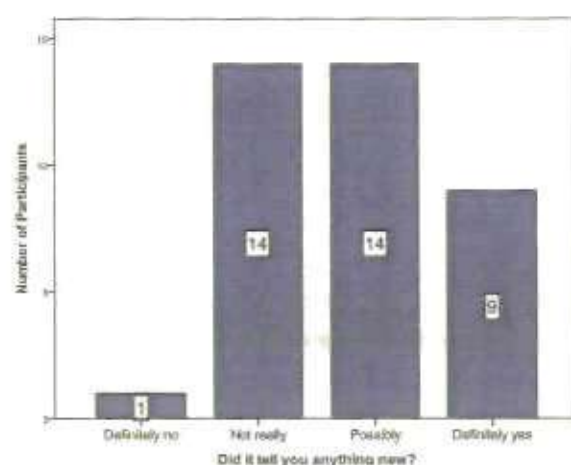


Figure 8.7

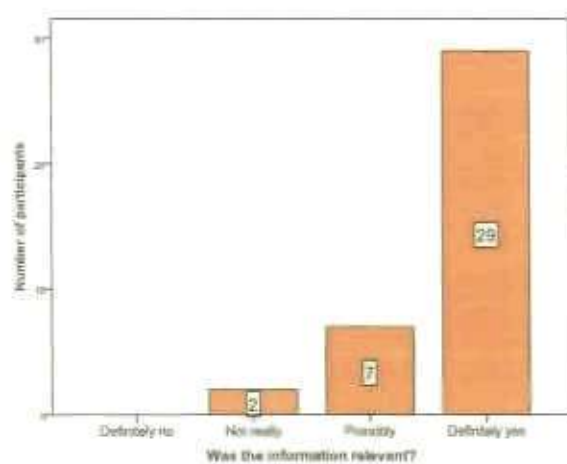


Figure 8.8

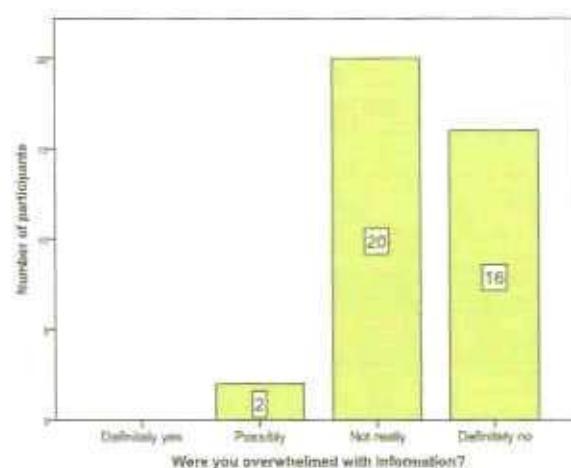


Figure 8.9

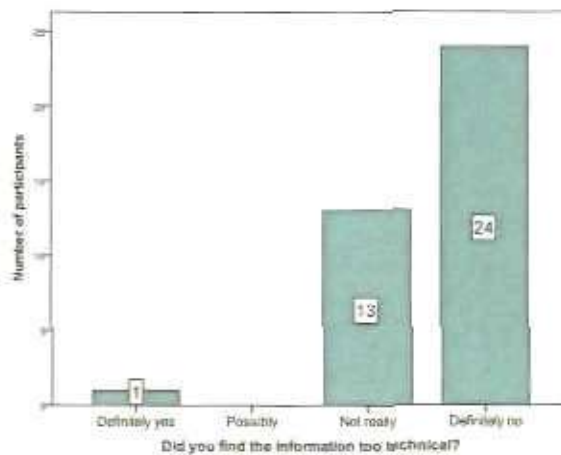


Figure 8.10

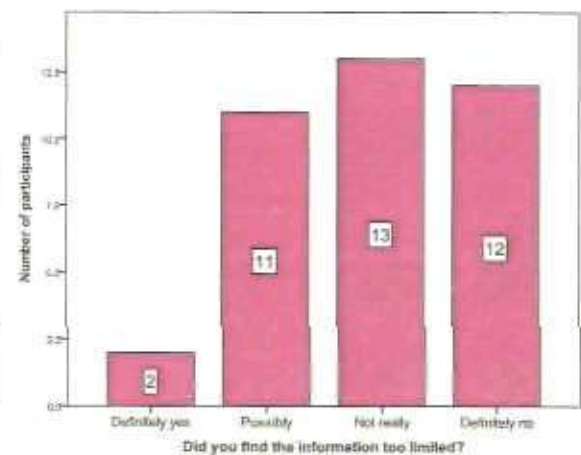


Figure 8.11

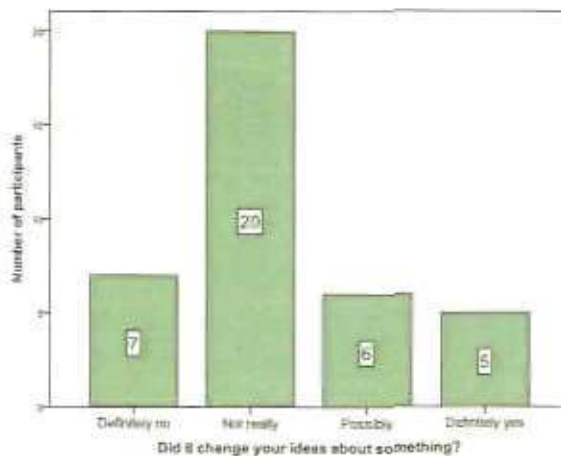
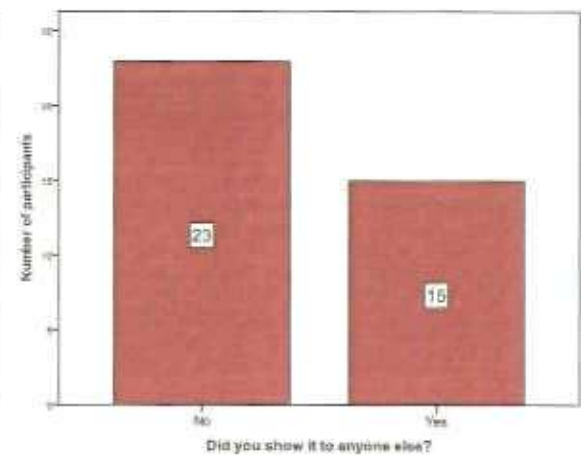


Figure 8.12



8.7 Qualitative Results

Thirty-seven out of forty participants (93%) provided qualitative feedback about the information booklet. Of those, the vast majority (n=31, 84%) left positive feedback about the booklet, such as "I found it extremely helpful" (P17), "It is brilliant to have this information in one booklet" (P47) and "Very good booklet, very impressed, well done. A must for new parents of undiagnosed children" (P43).

Six categories of findings emerged from this analysis:

1. timing of the booklet;
2. ease of reading;
3. reassurance from reading about similar experiences;
4. useful advice;
5. *suggestions about the booklet*;
6. aspects of the booklet that were disliked.

8.7.1 **Timing of the booklet**

The most common theme that emerged, cited by 15 participants (41%), related to timing. Parents felt that the booklet would be particularly useful for parents at the beginning of the non-diagnosis journey. One participant commented *"This would have been so helpful 17 years ago. We had little help or advice and it has taken years to find out what a book like yours would have told us in a few moments."* (P40) Similarly, another parent said *"I thought that as an introduction to someone coming to terms with an unknown diagnosis it was an excellent body of information - not too technical but with clear, detailed and realistic advice."*(P12)

8.7.2 **Ease of reading**

Ten participants (27%) commented that the booklet was easy to read, for example, one participant said that the booklet was *"very accessible"* (P6). Another said she *"found the layout good and it was easy to read."* (P32). There were no negative comments made regarding the readability of the booklets.

8.7.3 Reassurance from reading about similar experiences

Nine participants (24%) discussed that it was reassuring to know that there were other parents going through a similar experience to their own. *"I also liked the quotes from other parents; it made me feel like I wasn't alone and other parents are having the same fears and thoughts about their children as me."* (P28)

8.7.4 Useful advice

For seven participants (19%), the advice from other parents was particularly useful, as highlighted by one participant who said she particularly liked *"the focus on just enjoying time with your child, rather than pursuing an endless battle with the professionals. In my case I look back and wish that's what we'd focused more on and now it's too late."*(P4) Another participant commented that she *"liked that there was advice from parents, rather than lots of professionals"* (P28). The contact list at the end of the booklet was also considered very useful by seven (19%) participants; *"The information websites of the support groups, charities etc is very good to have."* (P9)

8.7.5 Suggestions about the booklet

Twelve participants (32%) gave suggestions about the booklet. These included adding the website details for Mumsnet (a website which provides information and advice for parents), and including information about the Early Support Programme, a new government programme which aims to help parents and carers be more actively involved in decisions about their child. Other suggestions included: information about transferring to adult services; alternative therapies; and healthy eating. In addition, one mother suggested including information about what to do when your children want children of their own. *"I*

think the one thing I noticed that was missing is...my eldest son who is unaffected is now asking if he has children will they be affected." (P29)

8.7.6 Aspects of the booklet that were disliked

There were a small number of 'dislikes' raised about the booklet. Two participants (5%) felt that the quotes were overly positive and should be balanced with more quotes looking at the negative impact of raising a child without a diagnosis.

"The parents' quotes should reflect the diversity of the experience of parenting a disabled child. Having a child with a disability can be very, very isolating, difficult and stressful. Marriages end, and it can be financially disastrous for a family too. Parents need to be told it's OK to feel resentful, angry and negative." (P2)

One parent felt that the information was too general but acknowledged that it was hard to make it more specific when it had to cover a wide range of symptoms. Two parents said that they disliked the front cover. Lastly, two participants commented that some of the information included in the booklet (such as around the issue of education) was only relevant in England, and that differences in the Scottish system needed to be addressed.

8.8 Discussion

8.8.1 Recruitment and participant demographics

Carrier sub-study

One reason for the low numbers of invitations sent out for this sub-study may be related to the small number of carriers identified each month through genetic clinics. One genetic counsellor estimated that between five to ten carriers were

identified through the clinic each month, this may have been lower in the other two clinics. A second reason for the low recruitment rate (23%) may be that due to good quality genetic counselling, carriers did not feel they needed any further information and therefore declined to participate in the study. Further, there is good quality scientific and psychosocial information available to carriers through patient organisations, hence carriers may not have felt it was necessary to take part in a study in which they would receive additional information.

'Non-diagnosis' sub-study

Recruitment of participants to the 'non-diagnosis' sub-study through genetic clinics was similarly low with only seven invitation packs sent out by clinics. There are a number of reasons as to why this method may have been unsuitable. A number of these were suggested by the lead genetic counsellors helping to run the study.

1. At each of the genetic clinics it was a genetic counsellor who was leading the study. Therefore they had to remind genetic consultants (who see undiagnosed patients) about the study. Furthermore, those genetic consultants then had to remember to remind the departmental secretaries to send out the information packs alongside the summary letters. The process was therefore fairly complicated with numerous steps that had to occur to ensure an invitation pack was sent out.
2. I was informed that secretaries are already under a lot of pressure due to staff shortages and therefore may not have engaged in the study (acknowledging when they were sending out a letter to a non-diagnosis

patient and querying the clinician as to whether an invitation pack might be appropriate).

3. Often testing, such as comparative genomic hybridization (CGH) array, is conducted on a sample taken during the appointment. The possibility of a result and therefore a diagnosis may therefore have been a hindrance to inviting parents to participate.
4. The clinicians may not like to admit to parents that they cannot find a diagnosis.
5. Even if clinicians remembered when seeing a patient that they were suitable for the study, they may have overlooked ensuring an invitation pack was sent out with the summary letter.

To remove some of these apparent obstacles, for a future study, it might be more appropriate to ask clinicians to give out the invitation packs during the consultation. Alternatively, given that the issue of timing was identified as important through the qualitative feedback, participants could be recruited retrospectively through patients records, and only those parents who have been without a diagnosis for one or two years could be invited to participate.

Recruitment rates for the SWAN members (56%) were comparable to other questionnaire based studies of parental coping (Korenromp et al., 2007). Attrition was fairly low (27%), and all participants except one completed the questionnaire fully. Other studies assessing the impact of information resources have reported attrition rates of 22% (Raynes-Greenow et al., 2009) and 33% (Wilson et al., 2010). This suggests the attrition rates for this study were within an acceptable range for this selected population. However, attrition rates might have been higher had the sample been recruited through the genetic clinic. Hence we cannot necessarily

assume that participants recruited through the genetic clinic would be similar in a subsequent RCT.

There are a number of reasons why attrition rates may have been low for this particular sample. As members of a patient organisation, these participants are likely to have an interest in helping with the development of an information booklet that would benefit future members. In addition, with very little information currently available on the subject of having no diagnosis, participants may have been keen to read and offer feedback on any information available to them. Participants may also have found the questions engaging and the length of the questionnaire to be acceptable. Finally, offering participants the option to complete the questionnaire either on paper or online may have facilitated more people to complete the questionnaire in a way that was preferable to them.

Attrition was higher (59%) for those participants in the control group who were asked to complete a follow up questionnaire, which consisted solely of the PSWI scale, after receiving the information booklet. One reason for this may have been because it was not clear on the original patient information sheet that some participants would receive a second questionnaire asking for feedback about the booklet. It would therefore be important to clarify this point in the patient information sheet in a subsequent RCT. Additionally, after having already completed one questionnaire, participants may not have wanted to complete another.

With 46% of the undiagnosed children in this study between the ages of 12-18+, the age was fairly high in comparison to other studies of children without a diagnosis (Graungaard and Skov, 2006, Rosenthal et al., 2001). However, this is

likely to be because the organisation has not been recruiting members recently as it is currently not functioning, and those parents that are members will have been so for a number of years. Furthermore, all but one of the participants involved in the study were mothers. This may be because mothers are generally more likely to be responsible for the day-to-day aspects of their child's health than fathers are (Bailey, 1994, Pelchat et al., 2003, Wood and Repetti, 2004)

8.8.2 Scoring methods

As this was a pilot study, the numbers in the sample were not sufficient to find significant differences in the outcome variables. Comparisons between intervention and control groups were therefore made partly as a 'practice run' for a definitive RCT and partly to see if differences were in the 'right direction' and might be found by a larger study. Two scoring methods were used for the GHQ. Using Likert scoring, the majority of participants (63%) scored under the cut-off for clinical significance. Using GHQ-case scoring, the majority (54%) scored over the cut-off, but for this small sample these differences are not significant. In the literature, the GHQ-case scoring method appears to be more commonly used. Therefore, in a future RCT it might be preferable to use GHQ-case scoring as it allows one to compare the scores with cross sectional studies. On the other hand, GHQ-Likert scoring appears more normally distributed and may therefore be preferable in a comparison of groups within one RCT. For these reasons, one might choose to use both methods of analysis in a future RCT.

8.8.3 IES and GHQ results

Using the GHQ-Likert scoring method, the mean values for the intervention group (mean = 15.6) were lower than for the control group (mean = 17.7).

Similarly, using the GHQ-case scoring method, the mean values for the intervention group (mean = 4.0) were lower than for the control (mean = 5.0). On a larger sample these differences might be statistically significant. Surprisingly, for the IES, the mean value for the intervention group (mean = 33.5) was higher than for the control group (mean = 18.6). This could simply be random variation in a small sample. If the study was to be replicated in a larger sample it would be unexpected, as one would have thought that receiving the information booklet would lessen the impact of not having a diagnosis. One reason for the intervention group scoring higher on the IES may have been that most of the participants included in the study had lived without a diagnosis for a long time (71% of parents had lived without a diagnosis for over seven years, and 32% of those had lived without a diagnosis for over 13 years). These parents may therefore have come to terms with the fact that their child did not have a diagnosis, and the booklet may have acted as a reminder, bringing back a lot of the negative feelings associated with not having a diagnosis. The problem therefore may not necessarily be the measure itself but rather the sample which participated in this study. The GHQ may not have picked this up as it was measuring something different – anxiety and depression. The IES results may have been different if it had been piloted with newly undiagnosed parents. It might be worthwhile retesting the measure in a further pilot study, targeting parents of younger children.

8.8.4 PSWI results

The high scores on the PSWI scale (mean = 22.5) and the positive qualitative feedback suggest that the information booklet was well accepted by participants. Those questions where the booklet did not score so well related to the scope of the

information in the booklet (Did it tell you anything new? 'Did you find the information too limited?' and 'Did it change your ideas about something?') As the qualitative feedback suggests, this may be because the majority of participants in the study have lived without a diagnosis for many years, and much of the information is already known to them. However, a significant number of people (41%) commented in the qualitative feedback that the booklet would be particularly useful for parents just beginning to search for a diagnosis. Hence, in a subsequent RCT, the assumed model needs to be adapted. It would be important to try and recruit parents as early as possible into their non-diagnosis journey as this is where it appears the booklet would have the most impact.

Cronbach's alpha coefficient for the PSWI scale was low ($\alpha = 0.57$), indicating that the individual questions do not combine to measure the concept of 'satisfaction'. To make the scale more reliable in a subsequent trial, removing any items with an alpha level below 0.6 would be recommended. As the alpha coefficient was low in this study, it is important to give more weight to the individual questions, in particular the qualitative feedback, than to the total score.

8.8.5 Results compared with similar studies

Mean scores for the IES were high (Total mean = 33.7, Intrusion mean = 19.2, Avoidance mean = 14.5) with thirteen (64%) of participants scoring over the cut-off point (≥ 26) for post traumatic stress disorder. This suggests that living without a diagnosis has a huge impact on parents, especially considering that these participants have been living without a diagnosis for many years. Compared with other studies of care giving and parental coping, participants in this study scored highly on the IES. For example, family caregivers of psychogeriatric in-patients had

a mean score of 27.4 in a study conducted by Aakhus et al. (2009). Parents of children diagnosed with malformations postnatally, had a mean total score of 18.6 (13.5 on the intrusion sub-scale and 5.1 on the avoidance sub-scale), nine years after the birth of the child. Authors of a prospective study looking at parental coping four months after the termination of a pregnancy for fetal anomalies, found that women's total mean score on the IES was 25.1 (Korenromp et al., 2007).

Similarly high scores were found on both versions of the GHQ, with participants in this study scoring 16.7 (GHQ-Likert scoring) and 4.5 (GHQ-case scoring). As with the IES scores, these were high in comparison to other health-related studies. In a study of mothers of children with cancer (Dockerty et al., 2000), the GHQ-Likert mean score was 13.7 (SD = 6.3). The mean score for mothers whose children had died of cancer was more comparable, yet still slightly lower at 16.3 (SD = 6.6). Parents of children who had suffered a severe traumatic brain injury (Hawley et al., 2003) also scored slightly lower than the parents in this study (M = 14.6, SD = 5.7). The control group (children of the same age, sex, social background and school class) had a mean score of 9.1 (SD = 2.7).

Using the GHQ-case scale, Nosarti et al. (2002) assessed the psychological well-being of women referred for breast cancer diagnosis, and found that pre-diagnosis, women's mean score was 4.5 (SD = 3.8), the same as this study. In addition, 34% of patients in the Nosarti et al. study were considered clinically significant cases (>4) compared to 54% of participants in this study. Ong et al. (2011) reported a mean score of 1.3 (SD = 2.0) for mothers of children with spina bifida aged 1–18 compared with mothers of able-bodied controls (mean = 0.5, SD = 1.3), (these figures were provided by the author as they are not presented in the paper). However, the scores in this study of undiagnosed children were not as high

as they were for women clinically diagnosed as suffering from postnatal depression six weeks after delivery (Navarro et al., 2007). Clinically depressed women had a mean score of 8.1 (SD = 2.8) compared to the control group who had a mean score of 2.3 (SD = 2.4).

The high scores on the IES and GHQ in this study, compared with other *health and parental coping studies using the same measures, highlight the major impact that living without a diagnosis has on parents' psychological wellbeing, even after many years.* One reason for this may be because for these parents, there is very little long-term psychosocial support available outside of the support group. Compared to more common conditions such as cystic fibrosis or fragile X for which clear medical and educational pathways are in place, parents may also have to navigate their way through services that are not clearly signposted, and which may change as their child gets older. Furthermore, without a prognosis parents are unsure how their child's condition will progress. These factors are all likely to contribute to parental anxiety, depression and psychiatric distress.

8.8.6 Future studies

A sample size calculation identified that in a subsequent RCT, a sample size of 1,084 would be needed in order to identify a small effect size (0.2), 176 for a medium effect size (0.5), and 70 for a large effect size (0.8). In theory, as the booklet intervention is relatively cheap and had good qualitative feedback, it would be worth knowing about a small effect size (as even if the effect was small, it would still be worthwhile making available to parents). Yet given the difficulties experienced recruiting participants, this option does not appear to be viable and it would be more realistic if a subsequent RCT were to be conducted, to aim for a

large effect size as it would require a smaller sample. Perhaps with a more suitable study sample, it would be possible to find a larger effect size. Other studies in which the same measures were used have concluded that a medium effect size is an effect size 'worth having'. Bech et al. (2009) concluded that an effect size of 0.47 was of value in a study in which the GHQ-12 Likert scoring was used to assess the effect of an antidepressant compared to a placebo. Similarly, using the IES, Monti et al. (2007) interpreted an effect size of 0.05 as significant, in a study in which cancer survivors with traumatic stress symptoms were enrolled to receive three sessions of Neuro-Emotional Technique.

As a result of this pilot study it is clear that receiving timely information is vital. This finding has been identified in a number of other studies in which the impact of information provision has been assessed (Nanton et al., 2009, Satterlund et al., 2003, Trask et al., 2009). Therefore, the assumed model needs to be changed to acknowledge the importance of receiving information early on. To test the assumed model in an RCT, it would be necessary to conduct another pilot study using the same measures but with a more appropriate sample. However, given the difficulties experienced recruiting, an RCT does not look like a viable option. Given the positive qualitative feedback with no evidence to suggest that the booklet caused any harm, and the low cost of printing it, a qualitative 'action research/service improvement' approach seems most suitable. In this case the clinic could give the booklet to parents who are likely to benefit from it, and ask recipients to provide feedback about it. This might be through the PSWI scale or alternatively a combination of PSWI scale plus qualitative telephone interviews with some of the recipients.

8.9 Conclusion

In this chapter I have presented the results from the pilot study. I have discussed the reasons why recruitment to the pilot study was unsuccessful, and looked specifically at the study procedure and appropriateness of the sample. I have also made suggestions for future studies. I have identified that the assumed model needs to be adapted to take into account the importance of timing in relation to receiving the information resource. In the next and final chapter, I will summarise the main findings from this body of research and present an overarching theory.

Chapter Nine:

Overview of the Study Including an Overarching Theory, a Reflective Assessment and Recommendations for Practice and Research

9.1 Introduction

In this final chapter, I will present an overarching theoretical framework that draws together the findings from all four phases of this doctoral study. I will discuss the relevance of the study findings within the context of genetic services and healthcare in general, and provide a reflective assessment of the study process. Finally, I will make recommendations for practice and future research.

9.2 An Overarching Theory

A number of themes were identified during each phase of this study that fed into the final, overarching theory. During the systematic review, a variety of emotional experiences and active coping mechanisms were identified that were commonly cited within the carrier testing literature. These included guilt, anxiety, stigmatisation, information gathering and reproductive decision-making. In the next phase, analysis of the carrier testing interviews confirmed a number of these themes. An active coping strategy named 'reproductive empowerment' was found to be the central phenomenon that unified the carrier testing experience. Carriers were found to empower themselves to make important life decisions and manage their risk of having an affected child, as a result of the carrier testing process. Information gathering around both scientific and psychosocial issues was a central part of this process, and enabled carriers to take control over their

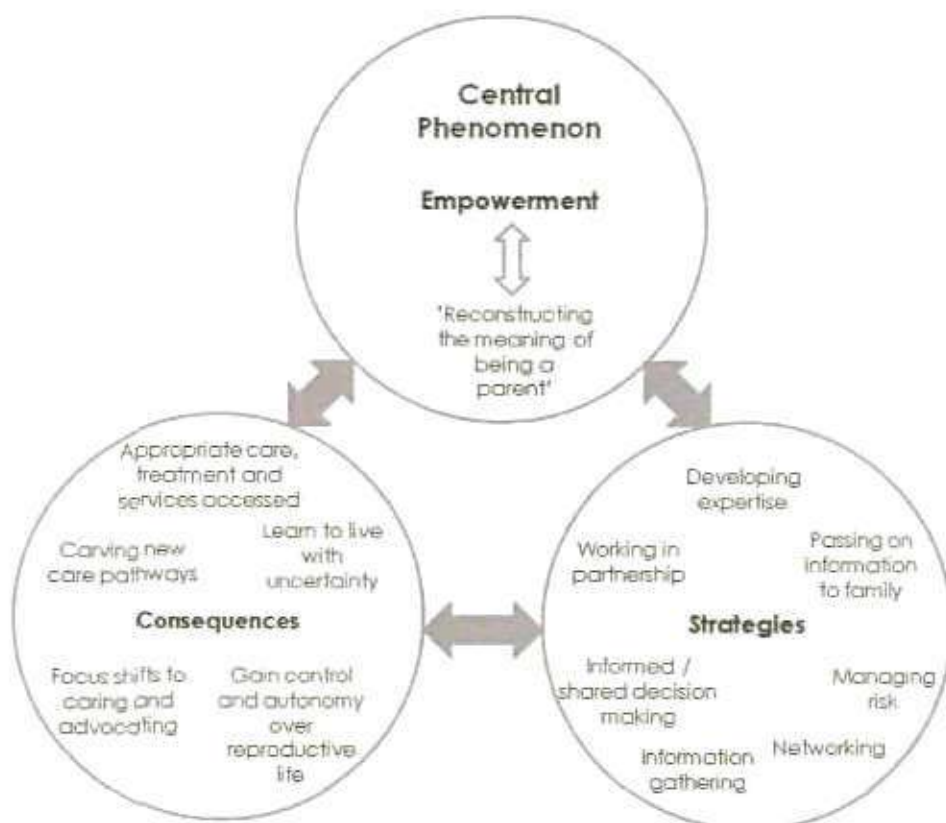
reproductive lives. This finding built on the work previously conducted by McAllister et al. (2008) looking at outcomes of clinical genetic services, and confirmed reproductive empowerment as a key motivator and outcome of carrier testing.

Through the analysis of interviews conducted with parents, a grounded theory emerged inductively from the data, which I named 'reconstructing the meaning of being a parent'. This theory described the process parents went through in order to effectively manage and control the day-to-day challenges of caring for a child with no diagnosis and no clear care pathway. Furthermore, throughout this process, parents developed empowerment strategies such as networking, developing expertise and information gathering. These strategies enabled parents to advocate effectively and carve new care pathways in order to ensure their child received appropriate care and services. This finding built on and verified the concept of empowerment that I had already identified through the carrier interviews, but here I identified it in another area of genetic services. In the same way as carriers were empowering themselves to regain control and autonomy over their reproductive lives, so parents with no diagnosis for their child were striving to empower themselves and their children in a system which did not necessarily facilitate this. Obtaining psychosocial information was a key part of the empowerment process.

In the fourth and final phase of this doctoral study, I further corroborated the findings from the first three phases of the study (triangulation) through the piloting of the information resources. Parents of undiagnosed children were found to be experiencing high levels of psychological morbidity, even after many years of having no diagnosis. This finding supported the experiences reported by parents during interviews. Furthermore, the value of psychosocial information was

confirmed through participant feedback. The importance of providing timely information was identified as being a key factor in supporting parents during their journey. Findings from the qualitative and quantitative methods were then integrated in the final interpretation phase of the study which resulted in a model of the overarching theory.

Figure 9.1 A model of the overarching theory



In summation, empowerment has been identified as a multi-faceted and dynamic construct. The participants in this study were striving to achieve a greater sense of control over their life. In order to do this, they employed a number of coping strategies. Participants undergoing carrier testing sought information through genetic professionals, support groups and the Internet in order to make informed and autonomous decisions. Furthermore, they empowered other family

members through information sharing, and engaged in shared decision-making with their partner. Parents of undiagnosed children also developed empowerment strategies. For example, they developed relationships with health professionals that were mutually active and shared information and knowledge with other parents in order to develop skills and expertise. Through this, they learnt to be advocates for their child, carving new pathways that did not previously exist. This process was ongoing with each influencing and enhancing the other. During this process, the meaning and role of what it meant to be a parent shifted significantly.

Whilst these findings are highly relevant within the field of genetic healthcare, they can also be applied more widely. The importance of empowerment, and the role that psychosocial information plays in that process, is relevant in any area where health professionals provide a supportive as well as an educational role. Many of the empowerment strategies identified through the interviews with parents have also been identified by authors focusing on the parental impact of raising a child with a range of known conditions. These include diabetes (Wennick and Hallstrom, 2007), hearing loss (Russ et al., 2004) and cerebral palsy (Huang et al., 2009). Therefore, the findings from this study may be relevant across an array of paediatric specialisations. The importance of acknowledging that many parents will have developed knowledge and expertise regarding their child's condition, and may desire a doctor-patient relationship in which both parties are working together to achieve desired outcomes through shared decision-making and information transfer, is also an important and relevant finding across the healthcare spectrum.

The rapid advancements that are currently taking place in genomic medicine and diagnostic testing (Miller et al., 2010) offer hope to those patients

who are currently living without a diagnosis. Advances in diagnostic capacity through new technologies such as chromosomal microarrays (which are increasingly being used for genetic testing of individuals with unexplained developmental delay or multiple congenital anomalies), offer much higher diagnostic yields for various types of chromosomal aberrations, with testing techniques more efficient at finding submicroscopic deletions and duplications (Miller et al). This, in theory, is promising for those currently living without a diagnosis. Yet the clinical significance of much of the genomic information derived from these tests is not yet certain. The challenge will lie in ensuring that this technology has clinical utility and patient benefit (Ali-Khan et al., 2009). Furthermore, genetic practitioners will need to determine best practices for effective communication of this genetic information, in particular with respect to incomplete knowledge and diagnostic uncertainty if they are to ensure a service that is useful, informative and appropriate for patients and families.

9.3 Context

9.3.1 Patient empowerment – research evidence

Evidence of genetic counselling outcomes encompassing aspects of empowerment (such as control, knowledge gain, informed decision-making etc), are apparent throughout the genetics literature. Information derived through carrier testing has been shown to empower carriers in reproductive decision-making (Henneman et al., 2002a, Henneman et al., 2002b, Lakeman et al., 2008). Empowerment strategies employed by parents of children with disabilities include navigating the system (Dolg et al., 2009), developing expertise (Nuutila and Salanterä, 2006) and networking (Wuest, 2000). Empowerment is also a frequent

outcome of presymptomatic and predictive testing. Crotser and Dickerson (2010) identified that family members given information about a BRCA1/2 mutation felt empowered as a result of the information and turned to genetic counselling in order to adapt to their potential hereditary risk. Women at increased risk of hereditary breast and ovarian cancer were found to have empowered themselves by actively engaging in risk management strategies such as mammograms, bilateral risk-reducing mastectomies and oophorectomies, in a study conducted by Watson et al. (2004). Similarly, Lim et al. (2004) reported that for some BRCA1/2 carriers, discovering their mutation status was a positive life-changing experience, as it was enabling both practically (through surveillance programs and prophylactic surgery) as well as emotionally (reducing uncertainty and increasing awareness of options and knowledge about risk). Smith et al. (2004) identified that information for future planning was a key reason for genetic testing, in a study in which presymptomatic testing for hereditary ataxia and neuromuscular disorders was conducted. In a study conducted by Decruyenaere et al. (2007), family planning was found to be one of the motivations for predictive testing for carriers of Huntington disease. Information derived through genetic counselling enabled carriers to make reproductive decisions regarding prenatal diagnosis, preimplantation genetic diagnosis and not having further children.

Furthermore, a number of outcome measures related to the concept of empowerment have been developed for use in genetic counselling. Berkenstadt et al. (1999) identified control as central to coping with health threats and to adapting to a broad spectrum of health problems. As a result, they developed the Perceived Personal Control scale (PPC) as a means of measuring outcomes of genetic counselling. The scale has been used widely in the field of genetic

healthcare (Aalfs et al., 2007, Boormans et al., 2010, Davey et al., 2005). More recently, McAllister et al. (2011) have been developing a patient outcome measure based on four dimensions of empowerment. These include knowledge and understanding, decision-making, instrumentality and future-orientation. The scale is still being tested, but it is likely to be a useful measure for assessing genetic counselling outcomes.

Research evidence more broadly highlights patient empowerment as an important outcome of healthcare services. Authors of a systematic review (Crawford et al., 2002) found that higher levels of patient involvement in the planning and development of care across a range of settings resulted in better quality of care, increased satisfaction and improved self-esteem for patients. Shared decision-making and receipt of mental healthcare were both positively associated with patient satisfaction, in a study in which satisfaction in a community sample of patients with depression was explored (Swanson et al., 2007). Further, in an RCT of facilitating information giving, patients with chronic medical conditions who were given copies of their medical record progress notes and completed question lists for physician review, were more satisfied with their physician's care and reported better physical functioning (Maly et al., 1999).

The importance of psychosocial information has been identified as central to the facilitation of patient empowerment. For example, France et al. (2008) found that a new brochure addressing common blood donor concerns and suggesting specific coping strategies, correlated positively with improvements in attitude, anxiety, self-efficacy and donation intention. The value of written information to support verbal 'one-to-one' information was identified in a qualitative study of the information needs of parents supporting children with

disabilities (Jackson et al., 2008). The provision of contact details for a named professional so that further information could be obtained when required, was also identified as important. Mancini et al. (2006) found that a patient information booklet supporting decision-making around genetic testing had a positive impact on satisfaction with information provided, decreased decisional conflict and had a marginal impact on knowledge. Psychosocial interventions including information, goal setting, homework assignments, exercise, discussion forums and multidisciplinary team support were found to help individuals manage the psychosocial, social and emotional challenges of living with multiple sclerosis, in a review conducted by Malcomson et al. (2007).

While the authors of these studies indicate that forms of patient empowerment such as information provision and shared decision-making have a positive impact on patients, it is important to bear in mind that not all patients will want to take such an active role over their healthcare. In a study of patient desire for information and decision-making, Nease and Brooks (1995) found that whilst most patients wanted to be given relevant information about their condition, not all wanted to be involved in the decision-making process. Those that did tended to be women, younger, highly educated and affluent. Similarly, Fujimori et al. (2007) reported that 30% of patients with cancer preferred not to receive information about their life expectancy. These preferences were related to level of education and mental adjustment to cancer. Ethnicity may also play a part in influencing patient desire for information and involvement in decision-making. Authors of a study looking at differences in end-of-life communication between Korean American and Non-Hispanic White older adults, found that Non-Hispanic Whites were more likely to engage in end-of-life communication than Korean

Americans (Ko and Lee, 2009). Other factors such as religion may also have an impact. Mobeireek et al. (2008) identified that decisions regarding life prolonging therapy and assisted suicide were influenced by religious beliefs. These findings highlight that the empowerment model will not be relevant to all patients. Finding out from patients themselves what role they want to play in the decision-making process and how much information they want to be given, is therefore vital.

9.3.2 Empowerment in policy and practice

Whilst the studies cited above highlight the current importance placed on empowerment within the context of healthcare, the concept of patient empowerment in fact emerged during the later part of the twentieth century. Previous to this, medicine had been recognised as a scientific profession, primarily as a result of scientific discoveries such as X-rays, vaccines and medications (Steele et al., 1987). By the 1960's however, broad social changes were occurring in the United States. The civil rights movement, the women's movement and the growing consumer self-help movement all emphasised a growing mistrust of the authority and an emphasis on self-determination (Roberts, 1999). The Roe v Wade case in 1973, in which the Supreme Court decided to legalise the right of women to chose to have an abortion, is a good example of this (Reiser, 1993). There was also a growing awareness, following the Nuremberg trials, on the notion of patients' rights (Brazell, 1997). Public attention had also been drawn to another vulnerable medical group - the subjects of human studies - following reports in the press in the 1960's of subjects inducted into experiments without being informed of their risks (Reiser, 1993).

A key change that emerged from these events was the modern medical ethics movement in which a new standing was given to the rights of individuals to learn about their illness and decide on therapies (Reiser, 1993). In the 1970's, the American Hospital Association issued a 'Patient Bill of Rights' laying out the rights patients had if they were hospitalised. The 'living will' was also invented during this time, which was a legal document enabling individuals to make decisions about which medical interventions they did and did not want (Roberts, 1999).

The concept of patient empowerment continued to gain recognition during the 1980's. New approaches to health care assessment began to develop, including a focus on the outcomes of healthcare, such as quality of life and functional capacity, further enhancing the importance of the patient perspective (McKenna and Stern, 1996). In addition, numerous court cases occurred during this period, particularly in the United States, highlighting the importance of the patient or family to decide their own care and treatment (Reiser, 1993). These themes were reflected in the internationally endorsed Ottawa Charter for Health Promotion (Canadian Public Health Association, 1986), which advocated health promotion as a means of "enabling people to increase control over and to improve their health" (p.1). The Charter also emphasised the importance of "a supportive environment, access to information, life skills and opportunities for making healthy choices" (p2).

More recently, professional and government guidelines in the United Kingdom have promoted patient empowerment as a vital component in the provision of quality health services. The General Medical Council, the body that registers and regulates doctors, promotes patient empowerment as a core duty of the doctor towards their patient. Their Good Medical Practice guidelines state

that doctors have a responsibility to "give patients the information they want or need in a way they can understand" and "support patients in caring for themselves to improve and maintain their health" (General Medical Council, 2009 p.2). Similarly, the Department of Health's 2001 publication, 'Your Guide to the National Health Service', states that "The NHS will shape its services around the needs and preferences of individual patients, their families and their carers" and "provide open access to information about services, treatment and performance" (Department of Health, 2001 b p.2-3).

The notion of the 'expert patient' has also been widely promoted by the Government (Department of Health, 1999, Department of Health, 2001a) linking patient expertise to ideas of empowerment and a user-led NHS. It is believed that expert patients will be "empowered to take some responsibility for [the condition's] management and work in partnership with their health and social care providers" in order to have "greater control over their lives" (Department of Health, 2001a p.5). The availability of accurate, informative and accessible information is viewed as central to this initiative, and websites such as the National Electronic Library for Health (NELH) and NHS Direct Online have been established in order to encourage this process (Price and Leaver, 2002). Furthermore, there has been an emphasis by the Government on patients taking care of themselves through the promotion of good practice such as healthy eating, regular exercise and breast self examination (Department of Health, 2000, Department of Health, 2009). These measures have created an environment whereby people are encouraged to take a more active role in their own healthcare.

Another key movement which is also relevant to this study is the Disability Rights Movement. This movement gained significant momentum during the 1960's.

during which time there was a growing desire of people with disabilities to participate more fully in society (Pfeiffer, 1993). There was also a realisation within the disabled community that they could not fully participate in society, not because of their disability, but because society as a whole did not cater for them. For example, the facilities were not in place for them to attend university, hold down a job or find a place to live outside of an institution (Driedger, 1989). A struggle for the recognition and right to live as independently as possible began, and in 1970, the civil rights organisation Disabled in Action (DIA) was founded with the aim of ending discrimination through litigation and demonstrations (Fleischer and Zames, 2001). The Centre for Independent Living was also established in Berkeley, California around this time in order that people with disabilities could live independently on university campus. It grew later into an advocacy program for the disabled community, providing impetus for the founding of the Disability Law Resource Centre (now called the Disability Rights Education and Defense Fund) (Switzer, 2003 p.74). In 1973, The Rehabilitation Act came into being and was the first national law to address employment protections for disabled individuals, and prohibited employment discrimination against otherwise qualified disabled workers by federal, state, and local governments (Richards, 1985). In 1975, the Education for All Handicapped Children Act was passed, following lobbying from parents' groups, for the right of disabled children to a free and appropriate public education that would emulate as closely as possible, the educational experience of non-disabled students (Abeson and Zettel, 1977).

Similar changes were occurring in the United Kingdom. The Chronically Sick and Disabled Persons Act was introduced in 1970, and stipulated that it was a duty of local authorities to provide certain social services for disabled persons and to

make their availability known (Elian and Dean, 1983). More recently, the Disability Discrimination Act 1995 was passed. This made it unlawful to discriminate against people with disabilities in relation to employment, the provision of goods and services, education and transport (Bell and Heitmueller, 2009).

This historical and political overview provides a useful framework in which to view the findings from this study. We live in an age in which the notion of empowerment is promoted in all areas of our lives. Within healthcare, patients are expected to take some control over their condition, and practitioners are supposed to facilitate this. People living with disabilities are also more empowered to be active participants in society than ever before. This is partly as a result of rights legislations that have occurred over the past 50 years, but is also likely to be due to shifts in societal attitudes towards disability. Thus, in some respects, the findings from this study are very much in tune with the wider political and cultural climate.

9.4 A Reflective Assessment and Identification of Strengths and Limitations of the Study Process

9.4.1 Qualitative Interviews

During this study, I experienced conducting face to face qualitative interviews for the first time. During interviews, I felt that it was important to reiterate that I was a researcher doing a PhD study at the University of Plymouth but that I also worked at a patient organisation for people affected by genetic conditions. Talking about my own background and interest in the subject helped me to build a good rapport with the interviewees. My background may have had an impact on my interaction with the participants, but I feel that this impact was positive in

that it made participants feel comfortable talking to me, and made me feel more confident talking to them.

One aspect of conducting interviews that I had not foreseen was that interviewees found the opportunity to talk about their own personal experience therapeutic. This was particularly the case for mothers of children with undiagnosed conditions, who were used to being asked by professionals about issues concerning their child. For many, this was the first time that anyone had specifically expressed interest in their well-being, in particular their personal feelings and experiences. The opportunity to talk about these issues was liberating for some mothers, as indicated by the length of many of the interviews. Most carrier interviews lasted around half an hour. In the case of the 'non-diagnosis' interviews, the average length was around 50 minutes with one interview lasting one hour 20 minutes. Furthermore, a number of interviewees commented at the end of the interview how much they had enjoyed taking part.

One of the difficulties I experienced as a researcher was keeping the agenda flexible, allowing scope for new issues to come to the surface and be explored, and at the same time ensuring that the conversation did not stray too far off the topic guide. Possibly, my inexperience as an interviewer meant that I was not as forthright as I could have been in ensuring the conversation remained focused. During one particular interview, when I was interviewing a woman who had undergone carrier testing, the participant began talking about the experience of being given her son's diagnosis of Duchenne muscular dystrophy. Although this was not strictly in line with the topic guide, I felt that because the subject was clearly an emotional one, it would be inappropriate to try and interrupt. I therefore listened attentively until the interviewee had said all she

wanted to about the issue, and then brought the conversation back in line with the topic guide. On another occasion, a father of an undiagnosed child spent a considerable amount of time describing his daughter's symptoms and I felt that he had perhaps mistaken me for a medical doctor who might have been able to offer advice. After this, I made sure at the beginning of interviews, that it was clear I was a researcher and not a doctor, and was therefore unable to offer any medical advice regarding their child's condition. In future studies it would be worthwhile clarifying this in the Patient Information Sheet.

In those cases where both partners were eligible to participate in the interview, they all agreed to be interviewed together. I felt that participants might find it useful to hear each other's experience as it might provoke an interesting discussion. I was careful to ensure that each question was asked to both participants. During the carrier interviews, both participants contributed equally to discussion. However, during the 'non-diagnosis' interviews, women were far more dominant. One reason for this may have been that women generally take a more active role in the area of child health and development (Skea et al., 2008). In general, it was the mothers who took children to appointments, or had given up work in order to look after their child, and therefore the day-to-day experience of caring for a child without a diagnosis appeared to have a greater impact on mothers than fathers, giving them more experiences to discuss during the interview. It may also be that from a psychological perspective, mothers may have had more emotional issues and experiences to discuss, or were happy to discuss, during the interview. In hindsight, I may have identified more salient findings relating to the emotional impact on fathers, if I had interviewed parents separately. One further limitation to interviewing partners together may have

been that discussion around topics such as the impact having an undiagnosed child had on the couples' relationship, and how the issue of carrier testing was first raised with the partner, may have been influenced by the fact that the partner was present.

One of the limitations of this study is that participants were self selecting and were keen to be involved in research. Thus, the findings from this research are likely to reflect the experiences of participants who are a certain 'type' of person: active, informed, engaged and more likely to be coping. This is reflected in the fact that the participants in this study were striving to empower themselves. Those that were not striving for empowerment are not represented in this study, and their experiences are likely to have been very different. Furthermore, by recruiting participants through the same regional genetics service, many of the parents were under the supervision of the same medical geneticist. This may have limited the range of experiences. More diverse experiences may have been identified if participants had been recruited from different regional genetics centres in different geographical locations across the United Kingdom. Lastly, participants had to have sufficient grasp of the English language to be eligible to participate, which may have excluded people from ethnic minorities for whom English was not a first language. This group may have had different experiences to the ones which I identified in this study.

Nevertheless, a wide range of themes and issues were uncovered during the two sets of interviews, and in the case of the undiagnosed children, even though parents accessed the same regional genetics centre, all the children had very different symptoms, were under the care of different medical specialists, attended different schools or nurseries and lived in different areas ensuring the

experiences of parents varied greatly. Furthermore, pertinent and profound themes that help us to understand the service user experience have been identified, which can be used to inform policy and practice.

9.4.2 Analysis of interview data

In reflecting on the method chosen to analyse interview data, I believe that the grounded theory approach worked well for the two studies. Even with small samples, the method provided a good framework for key themes to surface from the data and be explored in-depth. In addition, the findings and theories derived from this approach have contributed to the literature, provided useful content for the information resources, enabled the development of a set of useful recommendations for genetic healthcare specialists and policy makers, and supported a successful application to the Big Lottery Fund. However, as with all qualitative research, it is important to acknowledge that my own values, interests and experiences will have shaped my interpretation of the findings. Nevertheless, I hope that by using the validation techniques described in grounded theory, my impact on the research findings has been minimal.

One of the main challenges I faced in analysing the data lay in identifying themes and categories that were broad enough to reflect the range of experiences, but specific enough to be relevant and meaningful. The majority of qualitative studies previously conducted, in which the psychosocial impact of carrier testing was assessed, focused on particular genetic conditions (Anido et al., 2007, Fanos and Johnson, 1995b). However, in this study I chose to look at a broad spectrum of conditions to try and identify whether there were overarching themes and a possible theory that reflected the experience of being a carrier more

generally. Furthermore, because a systematic review of the carrier testing literature had already been conducted, a number of key themes had been identified before qualitative research began. Whilst this information gave me a good understanding of the key issues, it was important to ensure that this knowledge did not exert too much influence over my coding. I therefore tried to remain as open as possible to new categories and themes, identifying them inductively from the data in keeping with the grounded theory approach. One method I used to achieve this was to create hundreds of codes (free nodes) before attempting to group them together into higher-level categories (tree nodes). This ensured all thoughts about the data were given value and were not discarded on the grounds that they did not fit in with preconceived ideas. To further ensure the emerging theory was grounded in the data, I labelled codes and categories 'in-vivo' where possible, using words or phrases directly taken from participants in the study.

9.4.3 **Development of educational resources**

Once I began writing the first draft of the text for the resources, a number of issues became apparent. First, it became clear that there was a lack of scientific information suitable for inclusion into the non-diagnosis resource. This was primarily because without a diagnosis, one cannot talk specifically about a particular gene mutation, chromosome rearrangement, or inheritance pattern. For this reason, the resource would have to be heavily biased towards psychosocial information. This in itself was not a problem. However, it did mean that the study procedure originally planned had to be revised. Discussion with my supervisors as to the best way forward was a useful way of thinking through an alternative plan.

As is the case when developing written information with key stakeholders, each will have their own agenda that will influence their thoughts and beliefs regarding the content. The comments received from the various stakeholders who reviewed the texts were incredibly helpful, and a number of points and suggestions were raised which I had not considered. However, occasionally thoughts differed and I had to make difficult decisions which ultimately relied on my own professional opinion and previous experience developing patient information. I also had to accept the impossibility of pleasing everyone. One major revision which was made towards the end of the piloting stage came about as a result of interviewee feedback. A number of comments were made concerning the readability of the carrier testing resource. Furthermore, I had my own doubts about whether it was sensible to discuss recessive inheritance, X linked inheritance and chromosome translocations in the same resource. The feedback was therefore useful as it confirmed that the best way forward would be to divide the information up into three separate resources.

9.4.4 Piloting of resources

Discussion of the study procedure with staff at the regional genetics centres was extremely valuable as a number of practical issues were raised which I had not considered. For example, it was flagged up by one genetic counsellor that parents of children without a diagnosis are likely to receive numerous letters from a range of clinicians and other service providers. Therefore, the letter from the clinician was likely to be only one of a number of pieces of written information received at that time. This was important because it might have affected the impact and effectiveness of the information resource. For this reason, it was decided to design the study so that parents were given the resource at the same

time as the clinic letter to reduce the possibility of other information being received between acquiring the resource and completing the questionnaire. This example demonstrates that it is good practice to talk through study design with those people who will be implementing the study, before beginning.

The difficulties of recruiting sufficient participants to conduct a successful RCT became apparent during the final phase of this research. In hindsight, one of the reasons for the failure of the non-diagnosis sub-study was that the recruitment procedure was probably too complicated as it relied on the involvement of numerous people. This should be kept in mind for future studies and where possible, a study procedure in which potential participants are recruited at one point in time and which requires the input of one staff member, should be used. In addition, the difficulties experienced during this pilot study have highlighted that where there is only a small sample of potential participants available, quantitative research methods are not necessarily the most appropriate to use. In the case of the 'non-diagnosis' resource, it was the qualitative data that was most valuable, and provided a convincing argument for the effectiveness of the intervention. Therefore, quantitative research methods should not always be presumed to be most appropriate, and considerations such as sample size, cost and potential harm caused by the intervention should be taken into account when formulating a study procedure.

9.4.5 **Mixed methods design**

By combining qualitative and quantitative methods in this study, I was able to use triangulation in order to cross-check the findings throughout the study. As similar findings were identified using the different methods, the comprehensiveness

and validity of the findings was verified. In addition, each method was used as a basis for following phase. For example, the systematic review helped inform the questions asked during the patient interviews and the key themes derived from the patient interviews informed the content of the information resources. By using a mixed methods design, I was also able to develop a richer and more complex picture of what was occurring. Respondent validation, i.e. cross checking interim research findings with respondents, was also carried out by sending the draft versions of the information resources to the participants who had helped inform them, to ensure the resources reflected the parent experience.

One challenge that using this method posed was that extensive data collection using a variety of methods was necessary, which was in itself time consuming. Furthermore, it entailed becoming familiar with both qualitative and quantitative methods (although this could also be considered a benefit as I now have an understanding of both methods).

9.5 Recommendations for Practice

There are a number of recommendations that can be made to service providers in light of the findings from this study.

9.5.1 Recommendations aimed at improving clinical outcomes of carrier testing

The evidence of this study indicates that having an affected child alters the individual's response to the carrier test results. For those carriers who already have an affected child, the impact of receiving the test results in these cases may reinforce feelings of guilt, self-blame and maternal blame in the case of X linked conditions. Counsellors therefore need to be aware of these issues when testing

parents of affected children as these psychological issues may need to be addressed both before and after testing.

Counsellors should look to address misconceptions related to health and carrier status; some individuals may seek support for beliefs they have about their health by identifying clinical features of the disorder for which they are being tested or are found to be a carrier. Furthermore, while some clients will effectively manage anxiety and their carrier status through threat minimisation and other active coping mechanisms, professionals should ensure that those who appear to be managing well do not minimise their threat to the extent that they disengage from protective health actions, particularly when it comes to reproductive issues.

To ensure anxiety regarding carrier status is not unduly prolonged, genetic practitioners should provide understandable and comprehensive information regarding the reproductive options available to carriers, as this was the issue found to be most relevant to participants. In addition, there is a need to explore with patients when they would like this information (pre or post test results), in order that it is provided at a time when it will reduce any undue anxiety.

Ensuring that other health professionals, such as midwives and GP's, understand the implications of carrier status with regard to the risk to the fetus, and are aware of the various prenatal testing options that are currently available, is also imperative. Ideally, couples should be referred to genetic services before conception, so that the risk can be clarified and prenatal testing options discussed with a trained genetic counsellor.

Whilst genetic specialists may routinely provide patients with written information that can be passed on to family members, it is important that discussion focuses on how the subject might be broached and handled as

guidance and support may not necessarily be given in this area. Any written information provided to family members should include psychosocial information as identified in this study. The psychosocial booklets developed for this study could provide the basis for this communication.

9.5.2 Recommendations aimed at improving clinical outcomes when a child has no clear diagnosis

Whilst the majority of parents that attend genetic services because their child has learning needs and developmental delay will be searching for a diagnosis, between 30-50% do not receive one (Daily et al., 2000). The need for, and lack of certainty, is therefore an issue for many parents seen by genetic specialists. Specialists may therefore want to highlight to parents at the beginning of the diagnostic process that a diagnosis is not always achievable in order that parents do not have unrealistic expectations. As Skirton suggests (2006a), it may be helpful to discuss other areas where increased certainty may be possible in order to give parents an expectation that is more realistic and more likely to be satisfied. Furthermore, parents should be reassured that even without a diagnosis, their child should receive care, treatment and services that are tailored to their needs.

'Frustration' was an issue frequently experienced by parents. For example, participants repeatedly talked about their frustration in having to repeat themselves to different healthcare professionals. One way of reducing this would be to inform health professionals of this issue and recommend that they take more responsibility for reading back through their notes. At the same time, parents should be informed that there will be very specific things a specialist will be interested in finding out that relate to their particular speciality, so sometimes repeating information is necessary. Another source of frustration concerned the

time needed to complete all the necessary forms to access services, and the difficulties of completing forms when there was 'no box to tick'. Health visitors should be encouraged to help with form-filling. Furthermore, a letter from a clinical specialist, clarifying in writing the child's symptoms and that there is no confirmed diagnosis, might be useful for parents when submitting forms as well as when seeing other health specialists.

The findings from this study provide further evidence of the need for families to have one person or 'key worker' who acts as their main point of contact and who provides psychological and social support. This recommendation has been made by authors of other studies in which parental needs have been assessed (Allford and Hillier, 2008, Liabo et al., 2001, Rahi et al., 2004). In most cases, the health visitor was identified as the person who provided this service and was identified as key to the wellbeing of the whole family. If health visitors are the most appropriate professionals to provide this service, it is important that they receive appropriate training to enable them to understand service users' needs, provide psychological and informational support, and access relevant services in a timely manner.

A number of parents felt that there was no appropriate support group for them to contact because their child did not have a diagnosis. This may have further added to feelings of isolation and stigmatisation. Nevertheless, support groups do exist that are nonspecific and can provide general information and support to parents without a diagnosis, such as Unique and Contact a Family. Furthermore, even without a diagnosis, parents can benefit from the information provided by organisations for particular conditions, where similar symptoms exist. There was also a perception by some participants that joining a support group

meant that you had to physically attend group meetings. However, many support groups have chat forums which people can access online from anywhere around the world. This point should be stressed in the literature produced by patient support groups. Furthermore, patient groups may want to invest in setting up these forums if they have not already done so.

9.5.3 **General recommendations**

Not all service users will want the same level of input into their or their child's healthcare. Some, like many of the participants recruited into this study, strive for empowerment through information seeking and shared decision-making. Others, however, may want the clinician to take a more active role in this area and may not want to pursue further sources of information. This issue is relevant to all healthcare disciplines. Clinicians should therefore address this issue during the consultation in order to identify the patients' needs.

Participants valued being kept informed and up-to-date with information regarding their or their child's healthcare. For those service users who are keen to take control in this area, health professionals across the board should facilitate this by copying patients in to letters, signposting them to relevant organisations and keeping them updated about any scientific developments that might be of interest.

9.5.4 **Recommendation for policy**

The importance of the health visitor in providing support to parents and acting as a key worker for the family has been identified in this study. Unfortunately, due to the steady erosion of services which health visitors once offered, it will be very difficult for them to undertake this kind of role in the future (Craig and Adams, 2007).

Therefore, it is vitally important to promote this finding through organisations such as the Genetic Alliance UK, which informs and influences policy makers.

9.6 Contribution of Study Findings to Theory, Method and Practice

9.6.1 Theory

Whilst empowerment has been suggested by other researchers to be a central outcome of genetic services, what is novel in this study is that I have identified a multi-faceted and dynamic process of empowerment in two specific areas of genetic services. Moreover, I have developed a new construct, 'reconstructing the meaning of being a parent', to describe the experience of parenting a child with no clear diagnosis.

9.6.2 Methods

Whilst the methods employed in this study have been used widely in health research, the mixed methods design was novel and formed a robust method to develop patient information. By combining qualitative and quantitative methods, I was able to verify the findings through triangulation. Moreover, each phase built on and added to the findings from the previous phase which enabled me to gain a richer theoretical insight than if I had only used one particular method. The mixed method approach outlined in this study can be adopted by other organisations as a guide to develop validated patient information in other areas.

9.6.3 Practice

One of the main findings from this study has been the long term impact of parenting a child without a diagnosis, and the lack of psychosocial information and support available. The need for and importance of timely, comprehensive

psychosocial information has therefore been identified as key. Furthermore, reproduction has been confirmed as a central motivator and outcome of carrier testing. This underlines the need for timely information provision around this topic through genetic clinics and other healthcare providers.

9.6.4 Outcomes of Research

One positive outcome so far of this research has been that the findings and recommendations were used to support an application to the Big Lottery Fund to provide a new service for families without a diagnosis. This application has been successful and Genetic Alliance UK has been awarded £300,000 to undertake this work. Findings from this study highlight the need for the support group to provide the following services:

1. opportunities for families to share knowledge, experiences, difficulties and solutions through a variety of mediums such as a chat forum, a website, a regular newsletter, an annual meeting and by linking families for mutual support;
2. a website containing relevant information and signposting to useful government, health, educational and service organisations; and
3. links with health and social care professionals with expertise in undiagnosed conditions.

Furthermore, the booklets were entered into the British Medical Association Patient Information Awards 2010. The booklet 'Living without a Diagnosis – Information for Parents' won a Commended prize (Appendix 41).

9.7 Recommendations for Research

This study was primarily concerned with assessing the psychosocial impact of genetic testing from the service users' perspective. However, in order to achieve a more holistic overview of empowerment, it would be important to look at the concept from a variety of perspectives. A discussion of the concept with service providers such as clinicians, health visitors, and support workers would provide a more complete theoretical framework. Furthermore, it would be interesting to see if the desire for and definitions of empowerment varies across different ethnic or religious groups.

Findings from the carrier study suggest that there are differences in the way that men and women experience carrier testing. Men focused more closely on the mathematical aspects of carrier testing; women on emotional issues. As the number of men in this sub-study was small, the findings are not necessarily significant. However, they do add weight to the small number of studies that have also identified differences between the way men and women interpret and experience carrier testing (Marteau et al., 1997, Newman et al., 2002). Research is needed to explore this area further as there might be a need to develop carrier testing information that is specifically targeted at men. Furthermore, it was identified that carrier status appeared to impact on participants' sense of identity within their ethnic and religious world. This finding has been identified by d'Agincourt-Canning (2006) who was looking at the impact of predisposition testing for hereditary breast cancer. It would be worthwhile looking more closely at this issue, as so far the evidence is limited to a small number of studies.

It was difficult to recruit men into this study, both at the interview phase and during the pilot study. In addition, fathers of children without a diagnosis contributed less during the interviews than mothers did. Although this might reflect the fact that mothers generally take a more 'hands on' role with the day-to-day aspects of child care, the work conducted by the Foundation for People with Learning Disabilities (Towers, 2009) highlights that raising a child with a disability does have a substantial impact on fathers. Fathers often struggle to combine paid employment with caring responsibilities and looking after their own health. Efforts should be made to reach fathers in future work looking at the impact of parenting an undiagnosed child. This is something that the new service for families without a diagnosis could focus on.

The results of the pilot study highlight that in some circumstances the logistics of doing a quantitative study is more challenging than a qualitative one, particularly when it is difficult to recruit sufficient numbers to ensure significant findings. In order to test the effectiveness and acceptability of the information resources further, it might be preferable to forgo the RCT approach and conduct a 'service evaluation' study instead. Using this approach, patients who might benefit could be provided with a copy of the booklet and asked to complete a feedback form to assess its acceptability. For the 'non-diagnosis' booklet, it would be important for clinicians to give the booklet to parents who have only just begun searching for a diagnosis, rather than those who have lived without a diagnosis for many years. The qualitative feedback from the pilot study could then be compared with the qualitative feedback from the evaluation study to see whether giving it to parents at the beginning of their journey had a greater impact.

9.7.1 Suggested studies

Patient Empowerment in Genetic Services – The Professionals' Perspective

This is a mixed methods study exploring the concept of empowerment from the professionals' perspective. In-depth interviews with genetic service professionals would be conducted. Areas for exploration include: professionals' definition and views regarding patient empowerment, what professionals like and/or dislike about empowered patients, how they facilitate empowerment, their views on whether all patients strive for empowerment and how empowerment affects their role. Written information materials provided by genetic specialists would also be analysed to see if and how they promote patient empowerment. Empowerment from the perspective of support groups could also be explored to understand the concept from outside of the healthcare setting.

The Psychosocial Experience of Carrier Testing – Do Differences Exist Across Genders?

This is a qualitative study looking at the experience of carrier testing from both the male and female perspective. The aim would be to explore whether males and females experience the 'same' event in different ways. Particular attention would be paid to the terminology used by participants, to explore similarities and differences. Discourse analysis might be a suitable method to analyse the data as this method is concerned specifically with language and its role in the constitution of social and psychological life (Willig, 2008). If profound differences exist, further work developing and piloting 'gender specific' carrier information, could be conducted.

Living Without a Diagnosis – The Fathers' Perspective

This is a qualitative study looking at the impact of parenting a child without a diagnosis from the fathers' perspective. In-depth interviews would be conducted with fathers to explore the psychosocial impact of the condition from their position. Particular areas to focus on would include: whether it impacted on their feelings about being a 'father', the impact on their relationship with the child, issues related to employment, impact on the relationship with the mother. Furthermore, the need for and content of information in this area aimed specifically at fathers could be explored.

A 'Service Evaluation' Study Assessing the Acceptability of Written Psychosocial Information Resources

The psychosocial resources developed in this doctoral study are piloted through genetic clinics. Genetic professionals would give them to anyone who it was felt might benefit from the resource. Participants would be asked to complete a feedback form (either online or paper) in order to ascertain the acceptability, readability and comprehensibility of the resources. Questions from the PSWI scale would be included. A number of participants would be invited to take part in a short telephone interview. Interviews would enable participants' thoughts regarding the resources to be probed in more detail.

9.8 Conclusion

In this study I have used a range of methods to explore the impact of psychosocial information resources for patients accessing genetic services. As a result of the findings, I have been able to develop a novel theoretical model of empowerment, showing it as a dynamic and engaging process, and have

demonstrated that it is a desired outcome for many people accessing genetic services. This finding is in line with current trends in healthcare policy, which promote patient empowerment as a vital component in the provision of high quality health services. Furthermore, the resources produced during this doctoral study are the result of a systematic and innovative development process, which can be adopted across all areas of healthcare. They will now be promoted widely to genetic centres and support groups, helping empower patients and their families across the UK and beyond.

Appendices

Appendix

- 1 NHS Research Ethics Committee approval letter for patient interviews; 12 September 2008
- 2 Guys and St Thomas' NHS Foundation Trust R&D approval email; 23 September 2008
- 3 Letter from Guy's and St Thomas' to potential participant stating their involvement in the study
- 4 Patient information sheet for Carrier Testing study
- 5 Patient information sheet for Non-Diagnosis study
- 6 Consent form for interview
- 7 Topic guide for Carrier Testing interviews
- 8 Topic guide for Non-Diagnosis interviews
- 9 A sample transcript (ND9)
- 10 Example of coding using NVivo
- 11 Guy's and St Thomas' Hospital – 'Translocations' leaflet
- 12 Grey literature search - key issues relating to living as a carrier
- 13 Contact a Family – 'Living Without a Diagnosis' leaflet
- 14 Early Support – 'Information for Parents. When your Child has no Diagnosis'
- 15 Syndromes Without A Name USA – short information pamphlet
- 16 Grey literature search – key issues relating to living without a diagnosis
- 17 Survey to interviewees assessing carrier testing booklet

- 18 Survey to interviewees assessing non-diagnosis booklet
- 19 Letter inviting interviewees to complete survey
- 20 Recessive booklet (control version)
- 21 Recessive booklet (intervention version)
- 22 X linked booklet (control version)
- 23 X linked booklet (intervention version)
- 24 Translocation booklet (control version)
- 25 Translocation booklet (intervention version)
- 26 Booklet for parents of undiagnosed children
- 27 NHS Research Ethics Committee approval letter for pilot study; 26 March 2010
- 28 Patient Information Sheet for Carrier Testing pilot study
- 29 Patient Information Sheet for Non-Diagnosis pilot study (control)
- 30 Patient Information Sheet for Non-Diagnosis pilot study (intervention)
- 31 Guys and St Thomas' NHS Foundation Trust R&D approval letter; 30 April 2010
- 32 Great Ormond Street Hospital for Children NHS Trust R&D approval letter; 27 July 2010
- 33 Leeds Teaching Hospitals NHS Trust R&D approval letter; 24 August 2010
- 34 Invitation letter from genetic clinic to Carrier Testing pilot study
- 35 Invitation letter from genetic clinic for to Non-Diagnosis pilot study
- 36 Questionnaire for Carrier Testing pilot study
- 37 Questionnaire for Non-Diagnosis pilot study (control)
- 38 Questionnaire for Non-Diagnosis pilot study (intervention)
- 39 Feedback form for participants allocated into control group

- 40 NHS Research Ethics Committee approval for amendment to study procedure; 21 September 2010
- 41 Patient Information Award from the BMA; July 2010
- 42 Published paper: Lewis C., Skirton H., Jones, R. (2010) '*Living without a diagnosis; the parental experience*' *Genetic Testing and Molecular Biomarkers*, 14 (6) 1-9.
- 43 Published paper: Lewis C., Skirton H., Jones R. (2011) '*Can we make assumptions about the psychosocial impact of living as a carrier, based on studies assessing the effects of carrier testing?*' *Journal of Genetic Counseling*, 20, 80-97.



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Our Ref: 08AL 289
12 September 2008

Ms. Celine Lewis
Project Officer at GIG/ PhD student at University of Plymouth
Genetic Interest Group (patient organisation)
Unit 4D Leroy House
438 Essex Road
London
N1 3QP

Dear Ms. Lewis

Full title of study: A qualitative study of the psychosocial impact of genetic testing to inform development of information resources for patients.
REC reference number: 08/H0715/71

Thank you for your letter of 15 August 2008, responding to the Committee's request for further information on the above research and submitting revised documentation.

The further information has been considered on behalf of the Committee by the Chair.
Confirmation of ethical opinion

On behalf of the Committee, I am pleased to confirm a favourable ethical opinion for the above research on the basis described in the application form, protocol and supporting documentation as revised, subject to the conditions specified below.

Ethical review of research sites

The Committee has designated this study as exempt from site-specific assessment (SSA). The favourable opinion for the study applies to all sites involved in the research. There is no requirement for other Local Research Ethics Committees to be informed or SSA to be carried out at each site.

Conditions of the favourable opinion

The favourable opinion is subject to the following conditions being met prior to the start of the study.

Management permission or approval must be obtained from each host organisation prior to the start of the study at the site concerned.

Management permission at NHS sites ("R&D approval") should be obtained from the relevant care organisation(s) in accordance with NHS research governance arrangements.

This Research Ethics Committee is an advisory committee to London Strategic Health Authority
The National Research Ethics Service (NRES) represents the NRES Directorate within
the National Patient Safety Agency and Research Ethics Committees in England

Guidance on applying for NHS permission is available in the Integrated Research Application System or at <http://www.rdforum.nhs.uk>.

Approved documents

The final list of documents reviewed and approved by the Committee is as follows:

Document	Version	Date
Application		
Investigator CV		
Protocol	1	04 June 2008
Covering Letter		
Interview Schedules/Topic Guides	1	04 June 2008
Participant Information Sheet: Carrier testing study	1	04 June 2008
Participant Information Sheet: Non-diagnosis after genetic testing study	1	04 June 2008
Participant Information Sheet: Carrier testing study	2	22 July 2008
Participant Information Sheet: Non-diagnosis after genetic testing study	2	22 July 2008
Participant Consent Form	1	04 June 2008
Participant Consent Form	2	22 July 2008
Response to Request for Further Information		23 July 2008
Response to Request for Further Information	2	15 August 2008
CV Heather Skirton		
Guys and St Thomas Clinical Genetics letter of support		05 June 2008
Insurance policy		17 July 2008
Sponsor letter		09 June 2008
Draft questions for interview: Carrier testing	2	23 July 2008
Draft questions for interview: Non-diagnosis	3	15 August 2008
Reply slip	1	23 July 2008

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

After ethical review

Now that you have completed the application process please visit the National Research Ethics Website > After Review

You are invited to give your view of the service that you have received from the National Research Ethics Service and the application procedure. If you wish to make your views known please use the feedback form available on the website.

The attached document "After ethical review – guidance for researchers" gives detailed guidance on reporting requirements for studies with a favourable opinion, including:

- Notifying substantial amendments
- Progress and safety reports
- Notifying the end of the study

Appendix 2

Dear Ms Watts

<ul style="list-style-type: none">• A qualitative study of the psychosocial impact of genetic testing to inform development of information resources for patients.	08/H 0715/71	Ms Sally Watts
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Thank you for submitting your research project to the R&D Department. The project has now been approved by the Trust and has been allocated the Trust R&D registration number **RJ1 08/0246**. The project has been registered on the Trust's research database.

Please quote the R&D registration number in any communications with the R&D Department regarding your project.

Conditions of Approval:

- The principal investigator must notify R&D of the actual start and end date of the project.
- The Principal Investigator is responsible for ensuring that Data Protection procedures are observed throughout the course of the project.
- The agreed protocol must be followed. R&D must be notified of any changes to the protocol prior to implementation.
- The Principal Investigator and research team must have appropriate substantive or honorary contracts with the Trust. The Principal Investigator is responsible for ensuring that the team is covered.
- All members of the research team must have completed GCP training - please contact me if training or annual updates are required.
- Please submit a copy of the progress report on the anniversary of the Ethics favourable opinion (12th September)

Trust approval for the research is subject to the research being undertaken in line with the Department of Health's Research Governance Framework, and Trust policies relating to Research Governance. The Research Governance Framework and details of you and your researchers responsibilities within this framework can be found on the Department of Health's website at:

http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4108962

If appropriate it is recommended that you register with the Current Controlled Trials website: <http://isrctn.org/>

There is a fee per trial registered - if this fee will impose a considerable burden upon your trial resources please contact myself or one of my colleagues in the R&D Department.

In line with the Research Governance Framework, your project may be randomly selected for monitoring for compliance against the standards set out in the Framework. For information, the Trust's process for the monitoring of projects and the associated guidance is available from the Trust's intranet or on request from the R&D Department. You will be notified by the R&D Department if and when your project has been selected as part of the monitoring process. No action is needed until that time.

Many thanks for registering your research project

Best regards

Karen

Karen Ignatian BSc Hons

Research Governance Specialist

Guy's & St Thomas' Foundation NHS Trust

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Appendix 3

CLINICAL GENETICS
7th Floor Borough Wing
GUY'S HOSPITAL
Great Maze Pond
London SE1 9RT

Tel: 020 7188 1364

Fax: 020 7188 1369

Dear...

Re: Developing Resources for Patients

We are writing to inform you about a research study that we think might be of interest to you. The study is about people's experiences of genetic testing. The researcher would like to interview patients to hear about their experiences of genetic testing. This is to help inform a new information booklet that will be given to future patients. The booklet will provide information and support. More information about the study can be found on the Information Sheet we are sending with this letter.

Please read the Information Sheet, and if you are interested in participating please **contact the lead researcher, Celine Lewis**. You will find her contact details at the end of the Information Sheet.

Many thanks,

Sally Watts

REGISTERED PRINCIPAL GENETIC COUNSELLOR



Research Study Information Sheet

Developing Resources for Patients

Carrier Testing study

My name is Celine Lewis. I work for a patient group called the Genetic Interest Group where I develop information about genetics for patients and families. I am also doing a PHD at the University of Plymouth. I am currently doing some research into the effects of genetic testing on patients for a new information resource I am going to be developing. This information sheet explains a bit more about this research, and why I need your help.

What is the study about? The study is being done to find out how patients feel after receiving the results of a carrier test. This information will help me to develop a new booklet for patients about carrier testing. This booklet will be available to patients and families who visit a genetic clinic, and will be translated so that it can be used by patients in other countries. In order to write this booklet I need to hear the views and experiences of people who have taken a carrier test, whether they are found to be a carrier or not. This study is important because at the moment there is not very much information for people when they receive their test results. An information booklet, that provides helpful and practical information, can be a good source of support.

Does this study concern me? If you are 18 years or over and you have taken a carrier test, I would value your help with this study. For the purposes of this study it is not relevant what the results of your carrier test were, or whether you are male or female.

What will happen if I decide to help? If you would be willing to help with this study, I would like to interview you at your home or another convenient place. The interview will take between 30 and 60 minutes.

If you would like to ask me more questions about the study, or you have decided you would be willing to help, you can either fill in the return slip at the bottom of this letter, phone or write to me. We will then be able to arrange a date and time to meet, which is convenient for you. If you need to travel to the interview, I would be able to reimburse your travel costs. The discussion will be recorded on audio-tape but when it is typed out your name will not be on it. I will be writing a report but any comments you make will be anonymous. Your personal details will be kept securely and not passed on to anyone else.

If any difficult issues arise for you as a result of our discussion and you would like to talk with someone about them, I would be able to put you in contact with an appropriate person. I would be very happy to answer any further questions you might have about the study before you decide about being involved.

Can I change my mind about being involved? Even if you tell me you would like to help, you can change your mind about being in the study at any time, without giving a reason. Your health care will not be affected in any way, whether or not you decide to be involved.

How do I get involved?

If you are willing to help or want to ask a question, please contact me, in the next 2 weeks if possible, by:

Telephone: (daytime) 020 7704 3141 OR

OR Email: celine.lewis@plymouth.ac.uk

OR Mail: Fill out the form at the bottom of this information sheet and send it back to me in the pre-paid self addressed envelope.

Finallythank you for reading this information sheet.

Celine Lewis

I am interested in participating in this research and am happy for you to contact me.

Name.....

Phone Number.....

E mail.....

If you are not interested in participating this is absolutely fine, but it would be helpful for me if you could explain the reason why you have decided not to participate. Many thanks.

.....

.....

.....



Research Study Information Sheet
Developing Resources for Patients
Genetic Testing study

My name is Celine Lewis. I work for a patient group called the Genetic Interest Group where I develop information about genetics for patients and families. I am also doing a PHD at the University of Plymouth. I am currently doing some research about what it is like for families when doctors cannot find a name for their child's condition. This is for a new information booklet I am going to be developing for families. This information sheet explains a bit more about this research, and why I need your help.

What is the study about? The study is being done to find out what it is like for parents and families when doctors cannot provide a specific name for a child's condition. I would like to speak to people who have been in this situation. This is so that I can find out information that will be useful for other families that go through this difficult and sometimes distressing experience. The information will help me to develop an information booklet to support parents through this time, that will be available in genetic clinics. It will also be translated so that it can be used by patients in other countries. This study is important because at the moment there is not very much information for people who are unable to find a diagnosis for their child.

Does this study concern me? If you are 18 years or over and you are the parent of a child or young person (up to 19 years) with a possible genetic condition but no clear diagnosis, I would value your help with this study. I am interested in interviewing mothers and fathers as well as couples together. I will ask you questions such as what it was like waiting for a diagnosis and then not receiving one, and whether not having a diagnosis has caused any problems in practical terms (schools, healthcare etc) as well as emotionally. I will ask you whether you have any advice for other families going through a similar situation.

What will happen if I decide to help? If you would be willing to help with this study, I would like to interview you at your home or another convenient place such as Guy's genetics department or my office in north London. The interview will take between 30 and 60 minutes.

If you would like to ask me more questions about the study, or you have decided you would be willing to help, you can either fill in the return slip at the bottom of this letter, phone or write to me. We will then be able to arrange a date and time to meet, which is convenient for you. If you need to travel to the interview, I would be able to reimburse your travel costs (up to £50). The discussion will be recorded on audio-tape but when it is typed out your name will not be on it. I will be writing a report but any comments you make will be anonymous. Your personal details will be kept securely and not passed on to anyone else.

If any difficult issues arise for you as a result of our discussion and you would like to talk with someone about them, I would be able to put you in contact with an appropriate person. I would be very happy to answer any further questions you might have about the study before you decide about being involved.

Can I change my mind about being involved? Even if you tell me you would like to help, you can change your mind and withdraw from the study at any time, without giving a reason. Your health care or your child's health care will not be affected in any way, whether or not you decide to be involved.

How do I get involved?

If you are willing to help or want to ask a question, please contact me, in the next 2 weeks if possible, by:

Telephone: (daytime) 020 7704 3141

OR Email: celine@gig.org.uk

OR Mail: Fill out the form on the next page of this information sheet and send it back to me in the pre-paid self addressed envelope.

Finallythank you for reading this information sheet.

Celine Lewis

Reply Slip - Non Diagnosis

Please post this back to me in the stamped addressed envelope provided.

I am / I am not interested in participating in this research and am happy for you to contact me.

Name.....

Phone Number.....

E mail.....

If you are not interested in participating this is absolutely fine, but it would be helpful for me if you could explain the reason why you have decided not to participate. Many thanks.

.....

.....

.....



CONSENT FORM

Title of Project: A qualitative study of the psychosocial impact of genetic testing to inform development of information resources for patients.

Name of Researcher: Celine Lewis

Please initial

1. I confirm that I have read and understand the information sheet dated 22 July 2008 (version 2) for the above study and have had the opportunity to ask questions.
2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.....
3. I agree to take part in the above study.
4. I would like to receive a copy of the patient information booklet once it has been developed.

Name of Participant

Date

Signature

Researcher

Date

Signature

Patient Identification Number for this study:

Appendix 7

Topic Guide – Carrier Testing

Introduce why you are doing this study – to write an information booklet for people who have taken a carrier test. Opportunity to hear the experiences of people who have taken the test.

1. Can you tell me the reasons why you decided to take a carrier test? Did you have any worries or fears? If so what were they?

2. Can you describe what that 'waiting period' was like whilst you were waiting for the results?

3. Can you describe how you felt, when you first received your results?
(Prompt: talk about emotions e.g. relief, anxiety, shock, guilt etc)

4. Was anyone with you at the time e.g. your partner, a parent, a friend? Was this useful?

5. Do you think that having this information about yourself makes you feel differently about yourself in any way?
(Prompt: Do you feel any differently about your health, your identity, as a mother, partner)

6. In terms of family planning, did the results affect your plans to have children, or have more children, in any way? Did you make any other decisions based on your test results?
(Prompt: did you consider options such as prenatal testing, not having children, didn't matter as already had children)

7. Did you discuss your test results with other family members? How did they react? Did you experience any kind of stigma from other family members?

8. Did you/do you still have contact with any support groups or other professional services? If so, were these helpful?

9. Thinking about your carrier status now, how do you feel?

10. If you could go back in time, would you still take the carrier test?

11. Thinking about all the issues and emotions that you have described to me today, what advice would you give to other people thinking about carrier testing?

Appendix 8

Topic Guide: Non-diagnosis

Introduce why you are doing this study – to write an information booklet for parents of children without a diagnosis. Opportunity to hear the experiences of parents.

1. When and why did you start searching for a diagnosis for your child's condition? What information did you hope a diagnosis would provide?
2. What have been the main issues that have arisen due to not having a diagnosis (problems accessing services, schools, explaining to other people/health professionals etc,).
3. How have you attempted/managed to get round these problems?
4. How has the situation affected you on an emotional level?
(Prompt: depressing, frustrating)?
5. What about on a practical level?
(Prompt: relationship with partner/other children, taking time off work, expense, etc)
6. Do you think there have been any positive aspects about the experience?
7. Is a genetic diagnosis something you are still pursuing? If so why? If not, why not?
8. Did you/do you still have contact with any support groups or other professional services? If so, were these helpful?
9. What advice –practical and emotional, would you give to other families in this situation?

1
2 CL. OK, so I wonder if you could just start by telling me when you began searching
3 for a diagnosis?

4 SC. Well basically we knew there was a problem during the pregnancy. They didn't
5 know what it was at all.

6 CL. How did you find out?

7 SC. That was with the nuchal fold to begin with. They do the nuchal fold, the fluid
8 at the back of the neck, that was actually enlarged. So it went on from there really.
9 And I had numerous scans, 3D scans, at about 30 weeks they said that he was
10 going to be really badly disabled, almost vegetative sort of thing, and they
11 recommended that I consider a termination, waited 2 weeks, and I got a second
12 opinion and they said he would have learning difficulties, but they didn't know to
13 what extent but it wasn't going to be anywhere near as severe as what they
14 thought. So he was born, there were difficulties with the birth and he had to have
15 an emergency caesarean etc, and then he started fitting at about 3 weeks, his
16 head was very deformed and he was onand then he went in to have his
17 head operated on to make it more round, and they just started straight away at
18 looking at what was wrong with him basically.

19 CL. So doing lots of tests

20 SC. Lots of tests, blood tests, MRI's.

21 CL. And so what did they say?

22 SC. Nothing. Couldn't come up with anything. Absolutely nothing.

23 CL. When you were pregnant, they must of done a blood test then, did they find
24 anything then?

25 SC. Nothing. They tested his heart everything.

26 CL. Over what period of time did these tests occur?

27 SC. From about 4 months. They did numerous tests. They never really stopped
28 testing with him.

29 CL. How old is he now?

30 SC. He's 4. He recently had an MRI last year. I took him to see the geneticist a
31 few weeks ago, and she said there is still nothing, absolutely nothing.

32 CL. Can you tell me what it was like when you found out there was something
33 wrong.

34 SC. Well it was devastating to begin with. Absolutely devastating. It was like well
35 what do you do, how far do you take this. Every mother's worst fear isn't it. Your'e
36 not going to have a normal child. Simple as that. And obviously I was in a bit of a
37 state. My dad was with me at the time. I said I wanted another scan and they did a
38 3D scan and they said it wasn't going to be as bad. And I think the relief of it not
39 going to be as bad as what I expected it to be, and for them to say that he will have
40 learning difficulties but not as bad really made the decision for me. And also I was
41 so late into the pregnancy anyway and I didn't really want to have to go through
42 giving birth and a funeral basically. So that kind of made the decision for me. That's
43 it. Its just...its difficult to explain. Because there is no diagnosis, there is no
44 prognosis, so we don't know what to expect, what he is going to do, we don't know
45 whether he is going to walk, going to talk, to any extent at all, which is really

46 frustrating, I mean really frustrating. I mean Dad you came with me to see the
47 geneticist the first time didn't you?

48 They just talk a load of mumbo jumbo really don't they. They said it was probably
49 this syndrome or whatever, and then we got a letter saying it wasn't what he said.

50 *CL. So they came up with a possible diagnosis then?*

51 SC. Possible yeh.

52 *CL. What did they say that was?*

53 SC. Something like Dodo syndrome, something like that. And then it was like no, it
54 isn't a syndrome and then there is no prognosis at all.

55 *CL. So when they say it might be something*

56 They confirmed that it wasn't

57 *CL. Right and what was that like?*

58 SC. Its frustrating. The actual diagnosis is like a personal thing for me because it
59 would be nice to turn around and say to somebody 'he's got such and such', but
60 not only that, when it comes down to what he will achieve in later life, there is
61 nothing to compare him to. There is no guidance, no guidelines or anything. Will he
62 ever walk? I don't know, no one knows. Will he ever talk? Well I don't know, no one
63 knows. So you have to make decisions based on what he is doing today, not what
64 he can possibly do. It's very much an emotional rollercoaster in the sense that
65 obviously you want him to progress, like he's coming up to starting school this year
66 and my aim was for him to go to mainstream. And you hope for that and you hope
67 you is progressing but then it comes down to the fact that you have to face facts,
68 he's not going to go to mainstream and that brings you back down again, that he

69 hasn't reached the potential you'd hoped he would reach. So in that sense it is, it is
70 so up and down. It's like he starts school in September and my goal, because I
71 personally set goals for him, is for him to have a dual placement so he does part
72 time in special needs school and part time in mainstream. But he may not make
73 that, and then obviously I'm back down again, and then I'm disappointed, not
74 disappointed in him but just generally emotional of the fact that he's not making
75 that. So you know its yeah.....it's just as each thing he doesn't achieve...its
76 upsetting, just upsetting because like I say if we knew what to expect we'd be
77 prepared. It's being prepared. Like Dad and I have spoken about it and got
78 emotional when we've spoken about him, about what he's achieved and what he
79 hasn't achieved etc. So that side of it is really difficult. The personal housing side of
80 it, they won't re-house me or do any extension to this property because there is no
81 diagnosis for him.

82 *CL. That's something I was going to ask you about. Because you haven't got a*
83 *diagnosis what are the practical problems that you've encountered been? So*
84 *housing, I mean did they send anyone out to assess him?*

85 SC. Oh yes he's been assessed, I've got another appointment coming up and
86 we're just pushing and pushing because he is coming up to 4 now and he is still
87 not walking or anything. The bedroom is upstairs and he is sharing a bedroom with
88 his sister, and they are saying that they wont grant any funds because they say
89 'well if we do that and spend £50,000 then in 6 months he might be walking and it
90 will be a waste of money'. So ok I can see it from the business logistics side of it,
91 but, also that doesn't help me because he might not be walking in 6 years. So what
92 am I supposed to be doing with a 10 year old up and down stairs etc. But luckily,

93 my support group that I've got, I don't actually go to a support group, but the
94 support group that I have, the OT, my health visitor that is still very much involved,
95 I'm on the key worker system which is fantastic, they are all really pushing hard for
96 me, they are doing all they can so I can turn around and go 'look this is ridiculous,
97 you cannot go on a non-diagnosis, you cannot. It is not just affecting Charlie and
98 his mum, its affecting his sisters, its affecting the whole family'. But I am very lucky
99 in the medical network that I've got that they are fantastic but we work very closely
100 together. Very closely together.

101 *CL. So who are the medical team that you are generally in contact with a lot? You*
102 *said your health visitor..*

103 SC. My health visitor, not the geneticist. She gets in touch every few months and
104 says we still haven't found anything. Physio, occupational therapist, speech and
105 language therapist, all my basic ones. Fantastic nursery. He has a one to one at
106 nursery. They're fantastic too.

107 *CL. Was it easy to get him into nursery?*

108 SC. Very. With [name of nursery], Pam my health visitor has been an absolute rock
109 because I mean I've had other problems not just with Charlie but with Gemma, his
110 elder sister, eating disorders and things like that. So she has been really involved
111 in my whole family network.

112 *CL. Did you have her from the very beginning?*

113 SC. No not from the very beginning. I had one that I didn't really gel with and I met
114 Pam on an off chance when I took him for a weigh in and I complained about my

115 last one and she said well I'll be your health visitor if you're happy with that. And I
116 couldn't sing her praises enough. I really couldn't.

117 *CL. And how long has she been with you?*

118 Four years. She's absolutely brilliant. And she's also my key worker. She organises
119 all the meetings.

120 *CL. So she is like your gatekeeper in a way*

121 SC. Yes she is. She is my substitute mum. She look after us and makes sure that
122 everything is tickiti-boo and running like clockwork. I mean even my housing officer
123 in fact. Because this is run by [name of housing association] and they have a
124 support worked at Orbit housing and when he was about 18 months, 2 years old,
125 she just came round with every single form and benefit that I was entitled to. Every
126 single thing, things that I hadn't even thought or known about. And she sat here for
127 hours and hours, days on end writing them all for me, making sure I got exactly
128 what I needed. And I know so many people that wouldn't have a clue on what they
129 would be entitled to, and I think Fiona was a godsend to me. She was my housing
130 support worker. And it was only just by off chance. I think a housing officer came
131 for a visit and he said 'we've got a support worker. I'll send her round'. I mean I
132 would not have what I have got now if it hadn't been for her, which is where I think
133 that it is not clear to anybody who has a disabled child. How to do these forms and
134 what is available to you. It's really unfair in that sense because she completely did
135 everything. And when you're a single mum and trying to run a family, I've got 2
136 teens, a disabled child, you haven't got time to sit down and fill out forms and even
137 take in....and I'm not unintelligent by any stretch of the imagination, but its
138 physically sitting down and having the time to process it. And I think that that needs

139 to so be made readily available and so much easier to other people who are in my
140 situation.

141 *CL. So she was the one that was in charge of telling you about the benefits?*

142 SC. My health worker, she did touch on it with me but obviously her job is
143 not to sit down and do the forms whereas Fiona completely did everything. Pam
144 had mentioned it to me, I'd sort of taken it in, but its one of those things, 'I'll do it
145 next week', but next week never comes. With Fiona it was I'm coming now, we are
146 doing it now.

147 *CL. So did you have to contact her and ask her to come or did she contact you?*

148 SC. No, my housing officer went back to [name of housing association] and
149 contacted the support worker and said 'I have a lady on my books who I think will
150 totally benefit from you'. She contacted me within a week. And it took about 2 or 3
151 months of getting forms, filling out forms, copying, everything. Even down to filling
152 out the disabled form. She did everything for me, all I had to do was put my paw
153 print on basically. So that was fantastic. And I don't think people benefit from that,
154 they really do need that and it's just not made clear. You just haven't got time to do
155 it as a single mum. Even as a working parent you are so tied up with what's going
156 on with your child's disabled life, especially if you've got other children as well, you
157 just don't have time for it. I don't know what I would have done without Fiona
158 (support worker). But I hate not having a diagnosis, it's awful, it's been called
159 'Charlie syndrome' for the time being, because there is not one like him in the
160 whole world. And it's the not knowing. The not knowing is the most awful thing.

161 *CL. Is that because of the implications for the future?*

162 SC. Yeah implications for the future. I mean it's, I mean a child like my daughters
163 can sit there and go 'right this is what I'm going to do in the future'. They can plan. I
164 can't even plan for Charlie. I cannot make any plans for Charlie because I do not
165 know what he is going to be doing. I have no clue. It is like walking in the dark.
166 Completely and utterly. And as a parent you want to ask all these questions and
167 you are repeating yourself all the time, always saying 'what do you think, what do
168 you think?' It's frustrating for people, the specialists, the physio, the occupational,
169 for them, but it's just because I want to know, that need to know for how to plan.
170 How do you do it. Silly things like, I'm a single mum, and say for instance I meet
171 somebody, and obviously I have to say I have a special needs child, they conjure
172 up the worst possible scenario, someone with severe cerebral palsy. And that is a
173 stereotypical how humans perceive a disabled child. They don't see it on a certain
174 scale and then when they turn around and go 'well what's wrong with him?' 'Well
175 he can't walk or talk'. That's still really massive and huge. Its so difficult to relay to
176 people the extent to which his disabilities are because they have no understanding.

177 *CL. One of the things people have said is that when you don't have a name like*
178 *cystic fibrosis you have to list a whole long list of things...*

179 SC. I just say developmental delay, I just say he's two years behind. I can't be
180 bothered, I sat there in a key worker meeting and I said 'what do I say?' We are all
181 sat here, if I'm asked what is Charlie's disability, how do I define it?' And we came
182 up with developmental delay.

183 *CL. And do you use that with other people as well as other health professionals?*

184 SC. Yeah I'll say developmental delay in other words he is about 2 years behind.
185 Physically and mentally, in a nutshell. Which is basically what he is. He is like a 2

186 year old. He's doing things that my girls were doing at 2. That is where I do find it a
187 slight benefit is that I do have 2 older children. So I can look back and see where
188 my girls were at a certain age and how far Charlie is behind. But that's only
189 reference, a personal reference for me for how I deal with my son, not how other
190 people perceive him. I mean the geneticist said to me that mine and my exes
191 genes just didn't fit, that's it. Whether its genetic, a condition, there are so many
192 different possible reasons why, but they do think its genetic.

193 *CL. Have you come up with your own reason as to why it might have been?*

194 SC. I haven't got a clue. I guess its genetic

195 *CL. So you think its something to do with you and your partners genes?*

196 SC. That's the only thing I can put it down to. He hasn't got any other children. So
197 for him it was very hard in the beginning obviously because I gave birth to normal
198 children, able bodied children, but of course with him we ended up with a disabled
199 child, which he took quite personally, which I can understand. I can tell why he
200 would take on the guilt. You know it's, why are people born with blonde hair and
201 blue eyes, its difficult isn't it. There is no medical reason at all, so where do you go,
202 I don't know.

203 *CL. Have you found that you've stuck with the same medical team?*

204 SC. Yes but that's going to change now which I am absolutely dreading. Because I
205 don't do new people very well at all.

206 *CL. Why is that changing?*

207 SC. Because he's going to a new school, a special needs school so they have in
208 house physio, in house speech therapist, he'll go to the government occupational

209 therapist not the under 5's. The health visitor I will lose her because she only goes
210 up to 5 years old, and that to me I find really daunting. I've spoken to Pam about it
211 and she says she will always be here to support me and do it as a private thing just
212 because she is very close to us, which I have to say I am very lucky. But yeh I will I
213 have to start all over again which will be horrible, because I will have to explain the
214 whole thing over and over again repeating myself. And also when he starts school
215 they can have it in black and white, what Charlie is able to do, what he's achieved,
216 but they are not going to know him whereas Catherine and Claire and Claire and
217 Diane have known him 4 years. They are his physio, his speech, his OT, the ones
218 he has had for the last 4 years, but they are going to disappear off the scene now.
219 But talking to them, they know Charlie, they know he's very quiet to start off with
220 but then he comes out of himself, they know what he's capable of. So the transition
221 to the old speech and language therapist to the new one is only going to be in
222 black and white, which is completely different to having 4 years experience with
223 him. It is very difficult. Special needs school..

224 *CL. Was it hard to get him into the special needs school?*

225 SC. No, I have, round here, I don't know what anywhere else is like, but round here
226 I have been so lucky, so so lucky, but it is also you have to get off your backside,
227 and you do have to, not go to them because they did come to me, but certain
228 people would be like, 'whatever, I'll wait a year for them to come to me', they won't
229 push it kind of thing.

230 *CL. So you find that you have to be a fighter?*

231 SC. I don't think I have been, I don't think I have had to but then I think that's in my
232 nature anyway. And I've built up a relationship with his therapists. They know what

233 I'm like and I know what they're like. So no I haven't had to push. Maybe Pam had
234 to, my health visitor, but they were sent to me. But because Pam was my go-
235 between, I probably told her how passionate I was about it so she has been
236 passionate about it too. If I had been complacent then maybe she would have been
237 complacent about it too. I think it depends on what the parents are like too, and I
238 think that's with any child anyway. It doesn't matter if its special needs or not.
239 Some parents can be bothered some parents ca'nt. It depends how passionate you
240 are about it.

241 *CL. So did you have to get a statement to get him into special needs school?*

242 *SC. Yeah a statutory assessment.*

243 *CL. And was that difficult to get hold of or not?*

244 *SC. Absolutely not. We all went to the key worker meeting. And I received all my*
245 *copies, signed it, Pam (health visitor) put it all together for me, sent it to me, I*
246 *signed it and it was done. That's what I'm saying – I was so lucky, I am one of the*
247 *luckiest ones. I don't know whether that's the area as well. It might be where we*
248 *live. But then like I said, Pam (health visitor) is like a family friend now, and like I*
249 *said I'm quite not pushy, but passionate and outgoing and I've built up relationships*
250 *with his therapists. I would never say my relationship with his therapist is clinical. I*
251 *mean obviously its professional, but I'm talking on a official professional level. Its*
252 *professional but its not official. That guard has gone down and we can chat.*

253 *CL. Have you worked at all during that time?*

254 *SC. No no.*

255 *CL. And is that because of Charlie?*

256 SC. I couldn't commit to Charlie. I probably can do when he starts school. But him
257 not being at school, when he has appointments he has to go. Because
258 appointments are so few and far between so there is no getting out of it. And when
259 you have a child like that you cannot commit to an employer, personally, I don't
260 think so anyway.

261 *CL. So you spend a lot of time going to appointments?*

262 SC. Not quite so much now, but in the early days yeah. You have to see a special
263 dentist as well. Lots of different things that crop up and OT and we are trying to get
264 them to put us down for housing. But that will all settle down. Once he starts
265 school, because they have in house therapists, it will all be in his school time, so
266 my life is going to completely change as well. I'll be at a bit of a loose end, so yeah
267 it's the first 5 years I think....i mean when I went to look round schools and Grove
268 Park is his special needs school, it was so emotional. It was such a horrid time. It
269 was just all the way through, you reach these emotional hurdles with Charlie, total
270 emotional hurdles because you are so up and down you know, and you have to
271 come to terms with the fact, ok he's now 3 and he hasn't walked, he's now 3 he
272 hasn't talked, and its emotional, and having to come to terms with the fact that my
273 son is going to special needs was an emotional hurdle. It was heart wrenching,
274 absolutely heart wrenching, but I'm not an unrealistic mother and just talking to the
275 educational needs people. Some mums, some parents will chuck them into
276 mainstream and hope for the best. But that's not good for the child. So that is really
277 difficult, that side of it is really really difficult. When I went to visit Grove Park I went
278 to speak to Nina who is going to be his teacher about his needs, and obviously in
279 his classroom there are children that are severely, severely disabled. Nothing like

280 Charlie and of course I am trying to explain to her about Charlie and she's not
281 getting it, just seriously not getting it in the slightest. So I took him in, I said can I
282 bring him in for a morning so you can see what he's capable of etc, and he..to use
283 an analogy, he's like Einstein compared to the rest and he's a lot further ahead
284 than the rest of him. So now my concern is that they are going to bring him down,
285 and I have got to trust the system, I've got to try and sit down and say these are my
286 concerns that he is going to fall back. There is a boy there who is a year older than
287 Charlie who is sitting next to Charlie around the classroom table and he can't talk
288 and he is seriously disabled. And children learn by copying so of course that is a
289 concern to me.

290 *CL. You said about doing the dual placement. Is that something you suggested or*
291 *they suggested?*

292 SC. They suggested because they knew really that I was quite upset about coming
293 to terms with it. And eventually he may be able to go to dual placement but of
294 course he's got to go to special needs because he's too far behind and he's be lost
295 in mainstream and of course I wouldn't do that to my son. He may come on in
296 leaps and bounds.

297 *CL. You've got 2 other daughters. What's it been like for them?*

298 SC. The girls have been my rocks. They are incredibly positive. They are totally
299 positive. They are like 'Charlie's Charlie'. He's not disabled, he's Charlie, he will
300 talk, he will walk. There is no grey area with my girls. He will talk he will walk, ok
301 he's a bit behind mum but he's a normal little boy. And whether they are doing that
302 to be positive to me, but I genuinely do feel that that is how they do think. They are
303 genuinely positive. I mean 16 and 13, they are old enough to be left with Charlie so

304 I can go out. They have friends over, they all adore him, their friends, they all help
305 to look after him. They have all been amazing. Absolutely. The girls do now see
306 Charlie as anything but a normal little boy. I don't think anyone does really.
307 Anyone close to us, everyone loves him, he's just little special Charlie isn't he.

308 *CL. Have your feelings changed over the years, from initially the frustration of not*
309 *having a diagnosis, do you see things differently now?*

310 SC. In what way?

311 *CL. For example some mothers have said to me that you spend the first few years*
312 *desperately trying to find a diagnosis and going to all these appointments that you*
313 *tend to forget that this is your son and just to enjoy him...*

314 SC. No, I mean I am still desperate for a diagnosis but it's not been first and
315 foremost.....that is a lie.....its been very important to me but more on an emotional
316 level, not on a clinical level, more on an emotional level, a personal level, just
317 because I hate being blind and that's how I feel.

318 *CL. So you are still actively searching?*

319 SC. Yes, well the geneticists are. There is nothing I can do is there. And I will
320 chase it, but not to the point where its like I wake up every day and worry about it.
321 The only times when I worry about it is if it comes up in every day life. If someone
322 asks me, or schooling and that is the only time it really worries me. To begin with it
323 was very very hard because it was guilt as well, maybe I'd done something wrong,
324 maybe you know, I'd done something wrong when I was pregnant, so you tend to
325 blame yourself a lot, but having been reassured over the years, I know it has
326 nothing to do with me, because I used to beat myself up over it and everything, but

327 I know it's nothing to do with me, it is the chicken and the egg syndrome, what
328 came first you just don't know, you just do not know, so what can you do, you just
329 get on with it and hope there is a diagnosis. But it is frustrating, I could bang my
330 head against a brick wall without a doubt, but then that's because of a personal
331 emotional level, not a clinical level, because even if we had a diagnosis he's still
332 Charlie to me, it's not going to change him, not going to change him in the
333 slightest, it's just on a personal emotional level and I hate not knowing, I hate not
334 knowing.

335 *CL. I don't know if you were thinking of having more children anyway but did it*
336 *change your attitude about having more children?*

337 SC. Not more on my own but it definitely would have done if I'd stayed with Steve.
338 If I'd stayed with Steve absolutely no way would I have thought about having
339 another child. I love babies, I'd have 12 of them. It's just when they get to 5 and are
340 able bodied, because he's still lovely. I would have more if the situation was right.
341 That hasn't put me off at all.

342 *CL. So they didn't say anything about risk of having another?*

343 SC. If I did with Steve?

344 *CL. Right, and did they give you a figure?*

345 SC. No, just a probability. Because the only thing they could put it down to was our
346 genes. But no, the whole thing about having a special needs child has completely
347 changed my outlook on having a special needs child as well. Before when I was
348 carrying the 2 girls it was your worst nightmare them not having 10 fingers 10 toes,
349 you want the perfect baby. But now having not a 'perfect' baby so to speak, it's

350 nothing. People say 'how do you cope'. You just do because Charlie is really easy.
351 We are a very loving family and we just cope, we just do. I have a bad day but
352 everyone has bad days don't they? But how do you cope with a special needs
353 child, well I don't really because he's all I know. Although I've had 2 girls who were
354 normal he's all I know because he's just Charlie. He's inspiring sometimes as to
355 how loving and gorgeous he is to other children his own age, he's so affectionate,
356 just things to do with his syndrome that make him special, just a gorgeous little
357 boy.

358 *CL. You said you have lots of people supporting you. Do you have a patient*
359 *support group?*

360 SC. No my support group is my key worker etc.

361 *CL. Right and have you accessed any other kind of support group?*

362 SC. No because I'm not a people person at all and I would rather be left alone to it.
363 One of the early years ladies gave me a leaflet about a support group for
364 undiagnosed children but personally I don't feel I need it, personally. I'm coping, its
365 not as if I'm not coping and its taking over my life or anything like that. I've got too
366 much to worry about than go to groups. I've got too much going on. And I don't do
367 well with strangers. You know I've never been a coffee morning mum.

368 *CL. I suppose also because you have people around you that are so supportive*
369 *you perhaps don't need a support group.*

370 SC. Yeah but I wouldn't anyway because that is just the person I am . I'm quite
371 cocooned, I like my own space and I'm ever so independent. I have Jess and my
372 dad and Pam (health worker) and those people are fantastic and I wouldn't be

373 where I am without them but generally as a rule going to groups just isn't my thing.
374 I know a lot of parents would find that incredibly useful and get a lot out of it, but
375 I'm a bit of a nobbin know-all, I'd rather do it my way than no way. So I know what
376 I'm like, unless somebody sits down and tells me I'm doing it wrong....and I've
377 seemed to cope ok so far. If I do come across a problem then I will phone Pam,
378 and she'll come round and see me and we'll chat. And that's very rarely but she is
379 there if I need her.

380 *CL. So I wondered what advice you would give to other parents. Practical advice.*
381 *One of the things you mentioned is that you had a health worked but you didn't*
382 *really like her so you asked for another one. So obviously that would be something*
383 *you would advise.*

384 SC. Absolutely

385 *CL. So if you don't get on with one of the people you are working with, don't be*
386 *scared to ask for somebody else.*

387 SC. Absolutely, because that will put you off going for what you can get. That will
388 limit you straight away.

389 *CL. Is there anything else? Any other practical information...people talk about*
390 *keeping all your appointment letters...*

391 SC. I'm rubbish, I'm so disorganised so so badly disorganised. I don't have a filing
392 system, I don't do photos, I get a letter and I try and memorise it or stick it on the
393 fridge I'm not organised. But I would love to be like that but that's not my nature.

394 *CL. Are there any things that you have done that you think have been really helpful*
395 *that you would advise to other people?*

396 SC. Useful advice would be to be positive. Be positive and don't, I mean in the
397 early years I compared Charlie to other children as well. That's very very upsetting.
398 That's something that I would say do not do. People used to say to me don't do it
399 but you still do it, it's a natural thing. That's something I would say do not because
400 every child is different. Looking back I used to compare my kids to my friends kids
401 and think mine were fabulous and theirs were rubbish, so badly behaved, so in that
402 sense definitely don't. With the health visitor, with the professional side of things, I
403 think that a new parent with an undiagnosed child they need to pick out one person
404 on that professional side and cling on to them and build a rapport and relationship
405 with them. From my experience, what I have gained is phenomenal from having
406 Pam (health visitor). Totally. I mean it doesn't have to be your health visitor. It
407 could be your doctor, the midwife that put you on to somebody, and make sure first
408 and foremost that you have got an honest relationship with them because that is
409 just so important, so important. In the first 6 months I was near a breakdown with
410 Charlie, not with Charlie but personally, because my daughter was going through
411 eating disorders and getting bullied at school and then I had to deal with a special
412 needs child who was having fits, and I just phoned Pam up in an absolute state,
413 drunk as well, and she was on my door step and we just worked through it, and
414 you need that, someone who is not emotionally involved to be able to sit there and
415 rationalise with you. And I would say anybody in this sort of situation you need that
416 one person. Its great having your friends but they are emotionally involved. They
417 are going to be biased, they are going to be this, they are going to be that, but a
418 professional, they will be honest and straight with you. And I was honest and
419 straight with Pam, I told her I had been drinking and we were able to sit down and
420 rationalise and that was amazing, totally amazing. And I would say that that is first

421 and foremost with anyone who has a special needs child, get that one person on
422 who you can rely and trust, that is definitely, because you need them, because you
423 are going to have really crap days, and there will be days when your mum, your
424 dad, your best friend are not going to understand why you feel so shitty, but a
425 professional will. That borderline friendship professional, and it built up from there,
426 because she was purely professional at that time and you know its become a
427 professional friendship. So that is something. This is all in hindsight but try not to
428 beat yourself up over it, the why's and the wherefores. I think you have to just try
429 and stay positive. I mean I'm so rubbish. I've missed appointments in the past, I
430 cant organise anything, I loose my purse, my keys, I'm not off that nature, but try
431 and be organised, but I think if there are older children that are 'normal', I think they
432 have to be so involved in it, give them responsibilities and make them feel part of it.
433 Because I was always so worried because although the girls were a lot older, Sally
434 was 9 and Gina was 11 when Charlie was born, a funny age anyway, I always
435 worried they would feel pushed out. I think Gina did to begin with because it had
436 always been just the 2 of them, the 3 of us, but you know, I just got them involved
437 from the off. Don't mollycoddle. I was 'right, you feed your brother, I'm going to sit
438 down'. Although you are putting responsibility on them, you are shoving it in their
439 face so to speak, you are still making them feel part of the family, that they exist to,
440 that we do this together, we live together we do it together. My relationship with my
441 two girls is excellent. Absolutely excellent, I have no trouble with them whatsoever,
442 they are grade A students, because we work so hard at keeping the family
443 together. I make sure we have days out together. I say 'right I haven't seen you,
444 we're going to stay in and watch a movie together'. Or I make time, Jess will take
445 him and I'll take the girls out on their own. That's hard, sharing yourself, that is

446 really hard, but I will say always make time for your other children, because that
447 can have a bad knock on effect. Even having a normal new baby can have a bad
448 knock on effect, let alone a special needs one who can be incredibly demanding.
449 So that is definitely a work of advice I would give. That's really all I can say...I don't
450 know....also I used to be a little bit ashamed. That walk of shame. Like when I went
451 from a normal buggy into his bigger one, pushing it along, but you get over it, and I
452 don't know why you feel like that, because it's almost like you are embarrassed of
453 your own child, but I'm not, but it's coming to terms with it, its again something to
454 come to terms with. It's that transition again, it's all the little transition periods, and
455 it's never going to stop. I'm going to get more and more emotional as time goes on
456 probably. But that's just something you live with and you except.

457

458 End of interview.

Appendix 10

Links Code Tools Window Help

Code Kit

Nodes

- Free nodes
- Tree nodes
- Cases
- Relationships
- Memories
- Search Folders
- All Nodes

Sources

Nodes

Sets

Queries

Models

Links

Classifications

Folders

CL: 296 items

Look for: Search In: Tree nodes Find Now Clear Options

Tree Nodes

- Name
- [-] A Repositioning as parent
 - causal conditions
 - context
 - Emotional difficulties experienced
 - Lack of control
 - Loss of expectations
 - Frustration**
 - Role of parenting has changed
 - intervening conditions
 - Social relationships (partner, family, friends)
 - Support organisations
 - Housing
 - Health service
 - Housing issues
 - Insurance issues
 - Education
 - Employment
 - Disability in Society
 - strategies
 - Parent engaged in finding a diagnosis
 - Parent becomes expert in child's condition
 - 'Gatekeeper'
 - Having to 'push' or 'fight'

Parent becomes expert in child's c Parent engaged in finding a diagn Information seeking Frustration

Parent becomes expert in child's condition

Reference 1 - 0.18% Coverage

It's very frustrating and a drain but saying that she is very special

Parent engaged in finding a diagnosis

Reference 1 - 0.31% Coverage

but it was very worrying just not to know, very frustrating to be given a diagnosis of 'we don't know what it is'

Information seeking

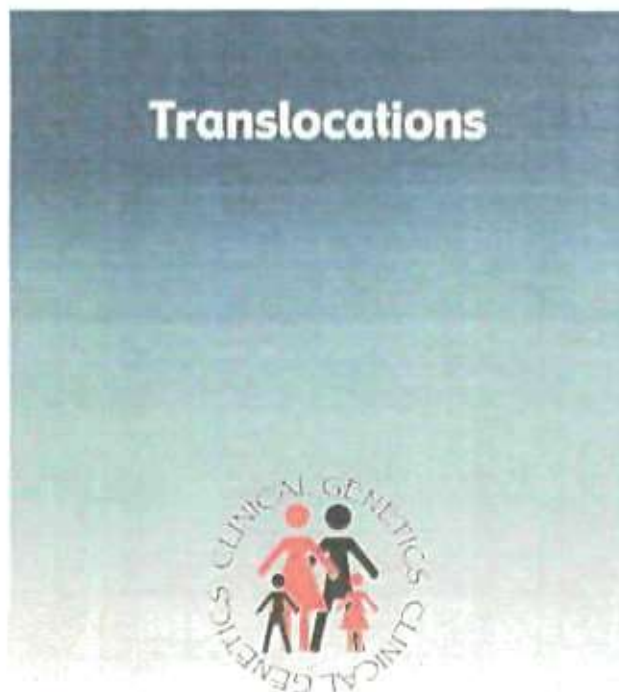
Reference 2 - 0.52% Coverage

So its just frustrating really. Its horrible. It must be the same if you're told your child's got a heart problem or a physical problem. Its just horrible being told there is something wrong

Frustration

Reference 3 - 0.82% Coverage

Appendix 11



Information booklet

Guy's and St Thomas' Hospital **NHS**
NHS Trust

Introduction

You may have been told that you, or a member of your family, has what is known as a **translocation** of the chromosomes. A translocation is an unusual arrangement of the chromosomes (or genetic material) in the cells of the body.

Most people have never heard of a translocation, and can feel worried if told that they have (or may have) something like this which is rather unusual. Understanding a little more about the translocation can often help to lessen such anxiety. We hope that this booklet will help to explain what this means, and answer some of your questions.

Genes and chromosomes: What are they and why are they important?

To understand what a translocation is, it is helpful to understand something about genes and chromosomes.

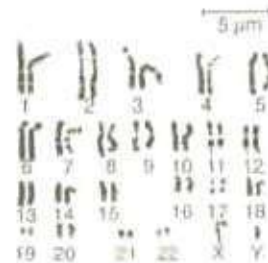
Genes are tiny 'packages' of vital information which influence our growth and development. Each person probably has about 30,000 genes, which all do something different. They work rather like special computer programmes, and determine such things as the colour of our eyes, how many fingers and toes we have, how tall and short we are, and so on. Genes are so small that they cannot be seen, even under a powerful microscope. They are arranged along little threadlike strands, rather like the way that beads are arranged along a string. These strands are called **chromosomes**, and are inside most of the cells in our body. Unlike genes, chromosomes can be seen under a microscope.

How many chromosomes does a person usually have?

Our bodies are made up of billions of cells. A person usually has a total of 46 chromosomes in each cell. They come in pairs. We inherit one of each pair of chromosomes from our mother and the other from our father. This is how we inherit characteristics from our parents.

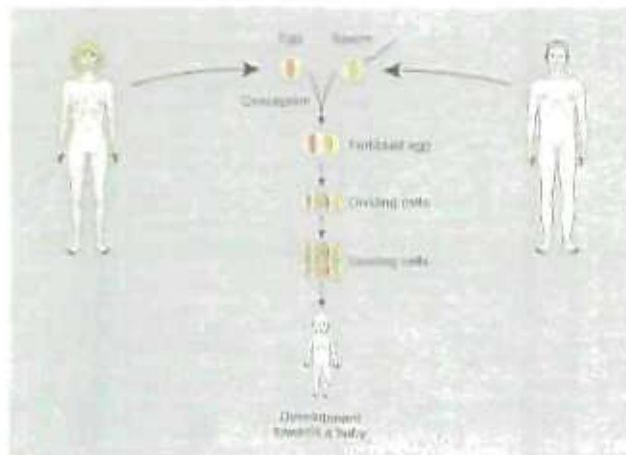
The following picture shows what chromosomes would look like if we arranged them in pairs in order of their size. Each pair is numbered to help us identify them more easily.

Pairs number "1" to "22" look the same in boys and girls. Pair number "23" are the sex chromosomes. They determine our sex. Girls have two "X" chromosomes, one inherited from their mother and one from their father. Boys inherit an "X" chromosome from their mother, and a "Y" chromosome from their father. The picture to the right shows chromosomes from a male.



At conception, the egg (with 23 chromosomes) joins with the sperm (also with 23 chromosomes) to make a fertilised egg. This divides and divides millions of times, and the baby develops.

It is important that we have the correct amount of chromosome material, as the genes (which control the way we grow and develop right from when we are conceived) are found on the chromosomes. Having some part of a chromosome missing, or having an extra part of a chromosome, can therefore result in a problem with normal development and lead to physical disabilities and learning difficulties in a child.



What happens in a translocation?

Sometimes, when cells are dividing during the formation of the egg or the sperm, or in the very early development of the baby, one or more of the chromosomes can break. This can cause a "translocation": an unusual arrangement of the chromosomes. There are two main types of translocation: a **RECIPROCAL** translocation and a **ROBERTSONIAN** translocation.

A **Reciprocal translocation** occurs when two fragments break off from two different chromosomes and "swap places", as shown in the diagram below.



A **Robertsonian translocation** occurs when two whole chromosomes become "stuck together". This diagram shows a Robertsonian translocation.



Why do translocations happen?

Although about 1 person in 500 has a translocation, we still do not really understand why they happen. We know that chromosomes seem to break and *join* quite often, and it is only sometimes that this leads to problems. Translocations happen in either the egg or the sperm cell before they join together, or shortly afterwards. These changes are totally out of our control and are unlikely to be caused by anything that happens during a pregnancy.

To understand why translocations can be important, it is helpful to understand the difference between **BALANCED** and **UNBALANCED** translocations.

Balanced translocations

In both examples already shown in the pictures, the chromosome material has been rearranged in such a way that **no chromosome material has been lost or gained**. This is known as a **balanced translocation**.

The health of a person who "carries" such a balanced rearrangement of their chromosomes is **NOT** affected by it. The only time it is important to them is when they come to have children, when a baby can inherit what is called an unbalanced form of the translocation.

Unbalanced translocations

If one or other parent carries a balanced translocation, it is possible for a child of theirs to inherit a rearrangement of the chromosomes in which there is an **extra piece of one chromosome and/or a missing piece of another chromosome**. This is what is known as an **unbalanced translocation**.

As was explained earlier, having too little or too much chromosome material can result in disability. An **unbalanced** translocation can cause serious problems in the development of a child who inherits this kind of rearrangement. There are different sorts of disability, some more serious than others. The seriousness of the disability in individual cases may depend on exactly which chromosomes are involved, and how much of the chromosome material is missing or extra. Some parts of the chromosome do seem to be more important than others. However, if a baby does have an **unbalanced** translocation, there is nearly always some degree of disability.

If a parent has an unbalanced translocation, will he/she always pass it on?

Not necessarily. Each time a "carrier" of a balanced translocation has a pregnancy, there are several possibilities:

- (1) A baby may have an entirely normal set of chromosomes.
- (2) A baby may inherit the same balanced translocation as the parent. In this case the child would be expected to be a healthy carrier, and not be in any way disabled because of this.
- (3) A baby may have an unbalanced translocation, and be mentally and physically disabled because of this.
- (4) The pregnancy may end in a miscarriage.

Therefore, it is quite possible for a person who carries a balanced translocation to have healthy children, and many do. However, the risk that a "carrier" of a balanced translocation may have a disabled child is higher than average, although the precise risk depends on the exact type of the translocation.

Can a carrier of a balanced translocation have a special test in a pregnancy?

Yes. Some people who carry a balanced translocation choose to have a special test in pregnancy by which the chromosomes of a developing baby can be checked.

One of these is called an **AMNIOCENTESIS** test (there is a special leaflet available which explains this test in more detail). A sample of the amniotic fluid ("water") which surrounds the baby in the womb is withdrawn through a fine needle. This is usually performed from 16 weeks of pregnancy onwards. The result takes about 2-3 weeks to come through.

Another test is called **CHORIONIC VILLUS SAMPLING (CVS)** (there is a special leaflet available which explains this test in more detail). A sample of the developing placenta (afterbirth) is taken, either through the neck of the womb (rather like a cervical smear test) or through a needle in a similar way to an amniocentesis. The CVS test is usually performed from the 11th week of pregnancy until the 13th week of pregnancy, with the result taking 2-3 weeks.

If you would like more information about either of these tests, we would be pleased to send you some, or talk to you about the tests, preferably before a pregnancy. If, after one of these tests, the baby was found to have either the "normal" set of chromosomes or the same **balanced** translocation as the carrier parent, then nothing further would need to be done. However, if the baby was found to have an **unbalanced** translocation, the parents would then need to consider whether or not they wanted to continue with the pregnancy, or have a termination (abortion). This is because, sadly, there is no way of "correcting" the baby's chromosomes.

How can someone find out if they carry a balanced translocation?

A simple blood test is all that is needed. A small amount of blood is taken, and some of the blood cells examined in a special laboratory.

Should other family members be told about the translocation?

If anyone in the family already has children, and/or is likely to have children in the future, it is usually sensible for them to be told. This gives them the opportunity to have a blood test to see if they also carry the translocation. If they do not carry the translocation, they cannot pass it on to any of their children, and the matter can be forgotten about from their point of view. If it is found that they do carry the translocation, they too could be offered a special test in pregnancy to check the baby's chromosomes, and any existing children can be offered genetic counselling and testing when they are older.

Children who carry (or may carry) a balanced translocation: why, and when, should they be told about this?

Why? Because a child who inherits the same balanced translocation as their carrier parent will have the same increased risk of having a disabled child when he or she comes to have a family of their own.

When? Each child is different, so there is really no one time when they 'should' be told that they do (or may) carry a translocation. It is probably sensible to leave this until they are old enough to be able to understand without being too worried by it. However, it may be best not to leave it until adolescence, when suddenly learning that you are 'different' from their friends can be very upsetting. Also, it is usually best for people to learn about the translocation well before having a family of their own. If any parent would like to talk more about this with us, we would be very happy to do so.

Can someone who 'carries' a translocation still be a blood donor?

Yes. A translocation is NOT a disease, and is not something that can be 'caught' from other people. It can only be passed from parent to child. Having a translocation should not stop you from giving blood if you want to do so.

And finally..... some important points to remember

(1) A balanced translocation of the chromosomes does NOT affect the health of someone who carries it. The only time it is important is when there is a pregnancy.

(2) It is important that children who carry, or may carry, a translocation know about it before they plan to have children of their own.

(3) People often feel guilty about something like a balanced translocation that runs in the family. It is important to remember that it is **no-one's fault**, and that no-one has done anything to cause it to happen. These things come 'out of the blue' and are out of **anyone's control**.

(4) Sometimes, people find it difficult to tell other family members about translocations. Often they feel it is their duty to tell their relatives about the translocation, as they may be at risk of having a child with a serious disability but they don't know quite how to go about this without causing undue worry. Some families are closer to each other than other families. Sometimes people may have lost touch with relatives and feel awkward about contacting them after so long. In such situations, it may be helpful to talk to someone from the genetic counselling clinic about how to approach family members, and which relatives in particular may need to know.

Therefore, if you, or any of your relatives, would like to talk about anything further, please feel free to get in touch with:

at the address or telephone number below. We can always arrange an appointment for you here or at your local genetic counselling clinic, or if that's be possible to visit you at home.

We do hope that this leaflet has answered some of your questions. Thank you for reading it. If you do have any comments about the leaflet, we would be very grateful if you could let us know.

Genetic Counselling Unit

If you need more advice about any aspect of Translocations, you are welcome to contact:

Clinical Genetics Department
7th Floor, New Guy's House
Guy's Hospital, St. Thomas Street
London SE1 8RT
Telephone: 020 7188 1364
Facsimile: 020 7188 1369

The production of this leaflet was funded by
the Guy's & St. Thomas' Charitable Foundation.

**Key issues relating to being a carrier as discussed in
patient information resources**

Appendix 12

<p>CF Trust – 'Genetics'</p>	<p>How is CF inherited How carriers of the CF gene can be identified Who should be offered carrier testing When should you see a genetic counsellor How you might avoid having a child with CF – amnio, cvs, PGD Gene Therapy Further Information</p>
<p>Cystic Fibrosis Foundation – 'Genetic Carrier Testing'</p>	<p>How does someone inherit a mutation of the CF gene? What does a positive genetic carrier test for CF mean? If I have one copy of the defective CF gene, can my children have CF? How does CF 'appear' when no one in my family has ever had the disease? Do certain types of the defective CF gene make the disease more mild or severe? Can genetic testing diagnose CF? Can a fetus be tested for CF?</p>
<p>Women's & Children's Hospital, N Adelaide – 'Carrier Testing for Cystic Fibrosis'</p>	<p>What does it mean to be a carrier? If I am a carrier, can my children be carriers? Can this test always detect carriers? If I am a carrier should my partner be tested? What does it mean if I am a carrier and my partner's carrier test is -ve? What happens if I plan to have children with a different partner? If my partner and I are carriers, can we have a test during pregnancy to see if our baby has CF?</p>
<p>Haemophilia Foundation Australia – 'On Being a Carrier'</p>	<p>-different possible reactions when receiving results (guilt, difficulty adapting to unexpected info) -dealing with reality of carrier status, - how it relates to family members, partner, community Genetic Counselling (best before pregnancy) Prenatal testing (CVS and amnio) Reproduction choices (IVF, infant adoption, permanent care, foster care)</p>
<p>The Haemophilia Society UK – 'Haemophilia Gene Carrier'</p>	<p>Personal Issues (experience of other members of family, severity of condition) Personal feelings about being a carrier – confusion, impact on relationship with others How do I tell my partner? Be well informed and up to date in treatment How do I tell my daughter that she is a carrier? Different opinions about when is the right age Different kinds of carriers - Obligate Carriers, potential carriers Further Information</p>
<p>NYU Medical Centre – 'About Carrier Testing'</p>	<p>What is a carrier? How do carriers use genetic information? Having children – prenatal diagnosis. Sometimes carriers decide not to have children with other carriers. Is genetic testing accurate? Test cannot detect all carriers/mutations. Confidentiality of test results.</p>

	Further information
NYU Carrier Testing for Ashkenazi Jews	<p>What a carrier is.</p> <p>Askenazi Jews – high risk of certain diseases. Some are less well known.</p> <p>Genetic testing minimises risk of having a child with genetic condition.</p> <p>Genetic screening can be carrier testing or prenatal screening.</p> <p>Prenatal diagnosis if both carriers (explanation of amnio, CVS, procedure, risk)</p> <p>Genetic counsellors provide information about test results and reproductive options. Do not make recommendations or judgements</p> <p>Knowing about problems before your baby is born may help you and your partner make important decisions about healthcare for your unborn child.</p> <p>In some cases may have to make difficult decision about terminating pregnancy.</p> <p>Information is confidential. Intended to help you make decisions appropriate to your own cultural background and individual experience.</p> <p>Should volunteer info about your ethnicity or race if obstetrician does not ask you.</p> <p>Genetic screening is one more technique used to make sure you have a healthy baby.</p>
The National Fragile X Foundation USA – What Does it Mean To Be A Carrier	<p>Definition of a carrier</p> <p>Females with a permutation</p> <p>Females with a full mutation</p> <p>Informing family members</p> <p>Prenatal diagnosis for fragile X – amnio, CVS, PGD, donated eggs</p>
Unique – ‘Balanced Translocations’	<p>What is a balanced translocation?</p> <p>Why did it happen?</p> <p>Will it affect my health?</p> <p>Can it be cured? Is it my fault?</p> <p>How common are they?</p> <p>Can we have children? Different possibilities for inheritance – balanced, unbalanced, miscarriage</p> <p>Tests during pregnancy – amnio, cvs, PGD, ICSI, egg and sperm donation, adoption</p>
Guy’s and St Thomas’ Hospital – ‘Translocations’	<p>Genes and chromosomes</p> <p>What happens in a translocation?</p> <p>Why do translocations happen?</p> <p>Balanced / unbalanced translocations</p> <p>If a parent has a balanced translocation, will he/she always pass it on?</p> <p>Tests in pregnancy – amnio, CVS</p> <p>Should other family members be told?</p> <p>Why and when should children be told?</p> <p>Can someone who carries a translocation be a blood donor?</p> <p>No one’s fault</p> <p>For more information...</p>
Muscular Dystrophy Campaign – Carrier Tests and Prenatal Diagnosis; Genetic Issues	<p>What is a carrier?</p> <p>Prenatal diagnosis – CVS, amnio, termination, PGD, egg/sperm donation,</p> <p>How is MD inherited?</p> <p>Support groups</p>

Appendix 13

contact a family
for families with disabled children



www.cafamily.org.uk

Helpline 0808 808 3555 Interpreters in over 100 languages via

 **Länguāģe Līnē**

Contact a Family - 200-211 City Road, London EC1Y 1JH, Tel: 020 7608 8200 Fax: 020 7608 8202

Telephone 0808 808 3555 e-mail: info@cafamily.org.uk Web: www.cafamily.org.uk

Helpline 0808 808 3555. Free phone for parents and families (11am-6pm, Mon-Fri). Chief Executive Françoise Bates

Registered Office: 200-211 City Road, London EC1Y 1JH. Registered Charity Number: 294413

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This Pamphlet is one of a series produced for parents and groups concerned with the care of disabled children. A full list of Contact a Family publications is available on request.

Incorporating
The Lady Hoare Trust

November 2005

www.cafamily.org.uk



Introduction

This fact sheet is aimed at parents who are caring for a child who does not currently have a diagnosis. It may help to answer some of the questions you may have, more especially it will highlight that you are not alone. There are many other families in a similar situation, all searching for information that will help them understand their child's particular symptoms and needs and ensure that they receive the help, support and services they need.

"I felt that getting a name for our daughter's condition would help us when explaining to others what was wrong, but in the end it didn't really matter. At the GP consultation involved with Helen only needed to know what her immediate difficulties were so that the right care could be arranged."

When you discover your child has a disability, your emotions can be very varied and intense. Parents commonly describe feelings of isolation, anxiety, sadness, despair, guilt and anger mixed up with an overwhelming love and protectiveness.

It is natural to want to know the cause of your child's disability. Without a clear diagnosis you can feel in limbo, trying to come to terms with what is happening and unsure what the future holds for your child and the rest of the family. Many parents describe their feelings of frustration as they are referred from one specialist to another, desperately seeking a diagnosis. Finding ways around the system to access information, help and support can be rewarding and parents often worry that their child will be deprived of appropriate services because they have no 'label'.

There are no easy solutions and although some families receive a diagnosis at a later stage, for others, sadly, this does not happen.

Why did it happen?

There are many reasons why children are born with disabilities and/or health needs. These include:

- Difficulties during the pregnancy including infections in the mother
- prematurity
- environmental problems
- genetic conditions
- asphyxia or trauma during or shortly after the birth

Although in many cases it is almost impossible for paediatricians to single out a specific cause, children will be left with a number of problems that do not fit into any one condition. Sometimes parents who have a child with an apparently obvious condition can discover that the cause is quite different from what they believed it to be.

"I'm tired of trying to explain to people that my child isn't naughty but has an abnormal condition."



Helpline 0808 808 3555



Why are some conditions so hard to diagnose?

There are a number of reasons why making a diagnosis is not easy:

- Doctors are now seeing a large number of children with very rare conditions, which are sometimes difficult to identify.
- Many conditions have similar features and symptoms, which make it difficult to be accurate about a diagnosis.
- Some children will have a number of problems that do not fit into one specific condition.
- There are substantial variations in the degree to which a child may be affected by a disorder or syndrome, which might also be the problem of making a diagnosis.
- Certain identifying features may not appear until later in the child's development, resulting in a late diagnosis or even a change of diagnosis.
- Identifying a medical professional who specialises in particular conditions can take time.

Another factor is the multitude of new syndromes that are being identified. Parents who may once have been told simply that their child was "developmentally delayed", might now have different syndromes suggested as possibilities. This can raise hopes of a precise diagnosis – if only you could find the right expert.

The problem with statistics

It has been estimated that there are 500,000 disabled children under the age of sixteen years. However, it is difficult to know how many of these children do not have a diagnosis. Problems with identifying and diagnosing very rare conditions. Misdiagnosis and late diagnosis have added to the difficulties of knowing how many children come into this category. Attempts have been made to estimate numbers but lack of research has made it impossible to be accurate about figures.

Is it genetic?

Genetics is a branch of biology concerned with heredity and individual characteristics. Some specific conditions and rare syndromes have a genetic basis. There are a variety of reasons why genetic conditions arise, including structural or numerical changes in chromosomes, change or mutation in a single gene or change (or mutation) in a number of genes. Some conditions tend to run in families while others may occur for the first time in a family (sporadic).

The field of clinical genetics is rapidly advancing and it is now possible to diagnose a large number of conditions using a variety of testing methods available. If there is evidence of a specific condition amongst family members, the option of genetic testing may be discussed. This may help to confirm or rule out whether your child's difficulties are the result of a genetic condition. Your GP or paediatrician are in a position to refer you to your nearest Regional Genetics Centre so you can self-ref.

Contact a Family doctor "A genetic condition in the family" is factored, which gives a brief introduction to genetic medicine, what genetic counselling involves and how to address them in the workplace and someone that surround it.



www.cafamily.org.uk



Exploring all avenues

Some families feel it is important to explore all avenues when seeking a diagnosis for their child. One of the greatest difficulties for parents in this situation is identifying the medical professional who may be able to advise them on the options available. Some children will be under the care of the local hospital while others will be under a specialist centre. Children with a number of differing problems may be under the care of more than one professional worker. Dealing with the medical professionals can be frustrating due to limited appointment times, crowded outpatient appointments, lack of information and not always having access to a specialist, if you are prepared:

- Make a list of the questions you want to ask.
- Request a longer appointment time.
- Take a friend or family member with you so that they can take notes.
- Ask for any written information that may help you to understand your child's specific problems.
- If possible request an appointment with the consultant without your child being present so that you can have a discussion without being distracted.
- If your child is under the care of the local paediatrician or ask if there are any other options available to you. This could include a referral to a specialist centre or specialist paediatrician but you may have to travel long distances for appointments.

If you feel strongly that all avenues to getting a diagnosis have not been explored, do discuss this with your child's paediatrician. Parents have no legal right to a second opinion but they can request one. It is not, however, guaranteed to provide you with further information re a firm diagnosis. Families in England can ask their local MALS (Patient Advocacy and Liaison Service) for more advice about this or any other health issue. Community health councils in Wales, health and social services councils in Northern Ireland and Health Councils in Scotland provide similar support.

Living without a diagnosis

"A sense of helplessness and frustration may be caused if parents can be encouraged to keep a diary to an ongoing extent of their child's development. This may not allow her to progress, but it may give a perspective to health professionals and others who monitor the child. Parents may also need to record their own thoughts and feelings."

A diagnosis can give a good indication of how a child may or may not develop. Without this information parents feel that it will be difficult to obtain suitable services. It might be helpful to bear the following points in mind:

- In any assessment, focus on the child's current symptoms.
- It may help to have a supporting letter from your child's paediatrician confirming the current symptoms.
- Consider keeping a diary of appointments, side effects and attention needed by the child and all the demands on your time and finances. Record details of the child's milestones and current stage of physical and mental development in relation to able-bodied children.
- It helps if an annual review can be provided in the light of the child's developing condition. Use any such reviews as an opportunity to express your views of how your child is developing and his or her current needs, and to re-state any earlier concerns if necessary.
- It is helpful if you can build confidence with service providers and find ways of working in co-operation with them. At the same time you may have to be persistent – parents often say that they received help only because they have been compared to sweet up and battle for services.
- Under various pieces of legislation throughout the UK, you are entitled to ask for an assessment of your needs as a carer and your child's needs.

For further information about the assessment process Contact a Family produces 'Assessments and Services in England and Wales' and 'Assessments and Services in Scotland'. Contact our Helpline (0808 808 3555) for a diary.

Influencing the services your child receives

You may find that arranging services for your child without medical diagnosis takes issues which parents of children with a diagnosis do not come across. There is an increasing shift in government policy and legislation towards parents of disabled children being able to be involved and influence how services are delivered. If you would like to get involved in influencing your local service providers and make them more aware of the needs of you and your child, make contact with other parents of disabled children in your area to see if you can work together. For more information about parent participation in service projects please get in touch with the Contact a Family helpline.

My unique child

Some parents, after several years, do not have a diagnosis for their child only to find that the label does not matter anymore. With or without a diagnosis, all parents of disabled children face the challenges of accessing adequate services and the same heartache and joys of having a special child.

For many parents the best support comes from other parents in local multi-disability groups that typically represent a range of physical and learning disabilities, some with labels and some without. Contact a Family can provide information about joining or starting a local group. Call the Contact a Family local groups officer, tel: 020 7608 8700 for more information. SWAN (Syndromes Without A Name), the UK support group (see page 7) can also provide information and support.

In any gathering of parents of children with mixed disabilities it can be guaranteed that a few will say:

"I did not get a diagnosis until Amy was 12" or "I have never had a diagnosis for Sammy". However, they will all talk of the same needs for information and support and inevitably add that their greatest help has come from other parents of disabled children.

When the word 'diagnosis' comes back to me, I think that it was time to accept that Christa is unique. I have support from her school and from other parents who give us morale. We have to deal with each new stage in the best way we can. Now I am not pre-occupied on the day of a diagnosis or a solution, I can get on with my own life. When Christa gave through one of her 'hobbies' projects I know I will not feel this way.¹⁴



Investigations

There are a number of tests and investigations that can be used to help determine the way in which your child is affected. These tests may help in obtaining a diagnosis. The most common tests and investigations are:

Blood Tests – These can be used to help identify minor things such as the number of red and white blood cells, infections, the amount of oxygen in the bloodstream and the child's blood group. Chemical imbalances indicating a possible metabolic disorder may be identified by blood tests.

Chromosomal Studies – These can give detailed information of the chromosomal structure. They are only available in specialist centres.

Computerised Tomography (CT) Scan – This scan gives detailed images of the inside of the skull and the body.

Developmental Tests – During early childhood all children have developmental tests. These start from birth and are then followed by regular routine assessments of height, weight, head circumference, reflexes, co-ordination, speech, hearing, sight and physical development. These can be the first indication that a child is not reaching the expected milestones.

Electroencephalogram (EEG) – Records the electrical activity in the brain.

Gene tests – These may give detailed information about changes (or mutations) in the genes or genes. Such tests are usually carried out at regional genetics centres.

Magnetic Resonance Imaging (MRI) Scan – Gives a detailed picture of the internal organs of the body using a computer.

Ultrasound – Produces a detailed image of organs such as the kidneys, gallbladder and liver onto a screen, which is then recorded on film.

X-rays – Used to give a detailed image of the bone structure of the body.

Who's who

For parents one of the most confusing areas is that of understanding the roles of the various professionals involved in the care of their child. These are some you may have contact with:

Radiologist – Works with children who have hearing difficulties and can advise on aids to improve hearing.

Care Manager – Assesses, organises and reviews the total care required for an individual. Can work for the local Social Services or the Health Authority. Please note that this is a generic term and that decisions are made by more than one individual.

Children's Community Nurse – Not available in all areas. Responsible for carrying out certain nursing procedures and treatments at home. Can also advise families on caring for their child at home.

Clinical Psychologist – Works with the child and family to help with any behavioural and developmental problems. **Community Paediatrician** – Co-ordinates community health services and makes referral referrals to other agencies. May be involved with educational assessments.

Consultant – The most senior member heading a team of doctors in overall charge of a child's care.

Continence Adviser – Provides information, advice and support to promote continence and the management of incontinence. May also supply continence aids. Some areas have Enuresis Clinics to help with daytime and night-time wetting.

Dietician/Nutritionist – Will offer advice and help on special diets and feeding.

Educational Psychologist – Works with parents and teachers in assessing the psychological and educational needs of children with any learning difficulties.

Geneticists – Doctors and genetic counsellors who may have a nursing background. Provide information about the likelihood of a genetic condition reappearing in a family and about the medical management of a condition.

Key Worker – Maintains regular contact with your family. Ensures that information about your child is shared effectively with everyone who is working with your family. However, they are not available in all areas.

Occupational Therapist – Will help and advise on special aids and equipment, which will help in all areas of mobility and daily living.

Ophthalmologist – Works with children who have vision problems, abnormal eye movements and can check for squints.

Paediatrician – Doctor who specialises in the care and treatment of children whilst in hospital or Out-Patients Department.

Physiotherapist – Using exercise and movement will help the child to gain as much independence as possible.

Social Worker – Provides help and advice as well as emotional support with social and financial problems. Will also help to obtain local services such as respite care.

Speech and Language Therapist – Works with children who are experiencing speech or language problems to enable them to communicate. Also helps with other difficulties such as feeding caused by oral problems.

Specialities

Hospitals will vary in the range of conditions they are able to treat. There is a lot of speciality in the specialist departments and the functions they cover:

Cardiology – heart and circulation

Dermatology – skin

ENT – ears, nose and throat

Endocrinology – any internal organs which produce hormones, such as the thyroid gland, stomach and liver.

Gastroenterology – oesophagus, stomach and intestines

Haematology – blood

Immunology – the body's reaction to disease with bacteria and viruses

Nephrology – kidneys and the urinary tract

Neurology – brain and nervous system

Ophthalmology – eyes

Orthopaedics – bones and joints

Psychology – study of behaviour

Respiratory – lungs and associated organs which involve breathing

Rheumatology – joints and muscles

Helpline 0808 808 3555

References and Acknowledgements

- (1) Family Resource Survey (FRS), UNICEF/ICEI
- (2) Helen Wright, Consultant in Child & Family Support, United Royal Hospital for A&E, Oxford
- (3) The last date used here is based on Family, Children, and Families 1997

And the parents who kindly allowed us to quote their experiences.



Support groups

SWAN (Syndromes Without A Name)

8 Acorn Close

Great Witley

Worthing

W Sussex

Tel: 01902 701234

e-mail: info@undisgnosed.org.uk

Web Site: <http://www.undisgnosed.org.uk>

The group offers a 24-hour helpline (info@undisgnosed.org.uk) where you can discuss your child's condition. It publishes a newsletter free once a year. It offers a 24-hour telephone helpline and a web site. It also offers an information pack on various issues such as benefits, mobility and funding to all its members. The group has a members' area.

Other Support

Children's Centres may be able to offer you support. Sure Start is a government programme which aims to achieve better outcomes for children, parents and communities. Some Sure Start Children's Centres are places where children under five years old, their carers, families, can receive integrated services and information. To find a Children's Centre in your area visit the search the Children's Centre Online database at <http://www.beyon.org.uk>

The Department for Education and Skills (DES) has produced the Early Support Family Pack. As a part of this pack, one of a series of booklets, When your child has an Disability, can be downloaded from the following website or ordered online.

Web: <http://www.gov.uk/25/>

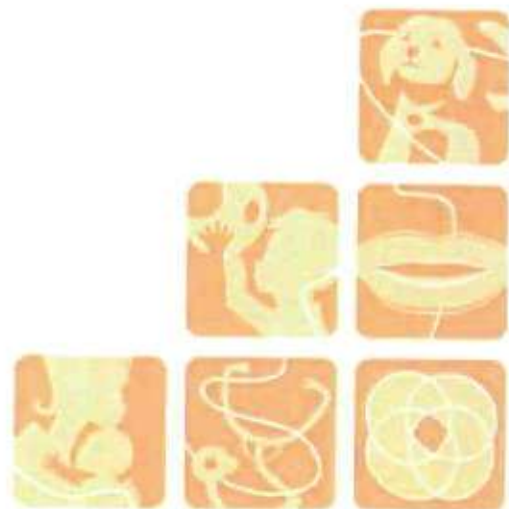
If you do not have internet access, please get in touch with our Helpline (0808 808 3555) as we can post you the information.

A leaflet entitled 'Making Medical Advances in the Home' has been produced by Contact a Family and approved by the Royal College of Paediatrics and Child Health. For a copy please visit our website (www.contacta.org.uk) or phone the Helpline (0808 808 3555).

MakingContact.org

Parents with children who do not have a diagnosis can make a request on <http://www.makingcontact.org>. It finds others with similar undiagnosed conditions or a diagnosed rare condition or undiagnosed rare condition.

Information for parents When your child has no diagnosis



About this publication

This booklet is for parents with young children with additional support needs where no diagnosis has been made and there is no obvious cause for the difficulties that a child experiences.

It was developed by the Early Support programme in partnership with *Common Ground*, in response to requests from families, professional agencies and voluntary organisations for better, standard information and reflects what parents who have 'been there before' say they would have liked to have known in the early days of finding out about their child's situation.

To find out more about Early Support visit www.earlysupport.org.uk

Information for parents
When your child has no diagnosis



It's natural to want to know the cause of a disability in your child or an identified need for support. Without a clear diagnosis, it can feel like you're in limbo, not knowing what's happening to your child or what the future holds. It can also feel frightening if you don't know how a condition might progress.

It's natural that parents want to know the cause of a disability in their child or an identified need for support. Without a clear diagnosis, it can feel like you're in limbo, not knowing what's happening to your child or what the future holds. It can also feel frightening if you don't know how a condition might progress.

It can be very frustrating when a child is referred back and forth between different specialists for more tests and there is still no clear answer to the question, 'What's happening and why?'

Information for parents
When your child has no diagnosis

Why is it so hard to get a diagnosis?

There are many things that can cause disability in a child. Difficulties during a pregnancy, personality, genetic conditions or trauma during birth itself can all be relevant. In some cases it's impossible to single out a specific cause - especially where children have a range of health problems that do not fit easily into any known **syndrome**. A **syndrome** is a characteristic pattern, or group of symptoms, which often appear in combination with one another.

Some children have a **rare condition**, which may only affect a handful of other children across the country, or perhaps none at all. It's harder for doctors to diagnose a condition they've never seen before, and where there are very few studies which would make it possible to compare the features of your child's difficulties with other cases. Many conditions have some similar features and symptoms, which can also make it hard to be specific about your child's particular condition.

Some features may not appear until your child is older, when it will become more obvious that they are affected by a particular syndrome or disorder. More and more syndromes are being discovered each year, so it may be that a diagnosis will be achieved for your child in the future, even if it does not seem to be possible now.

If you feel strongly that all avenues to getting a diagnosis have not been explored, you should certainly discuss this with your child's doctor and request a second opinion. But sometimes everything that can be done has been done and you are still left with no diagnosis.

Information for parents
When your child has no diagnosis



Does it matter whether you get a diagnosis or not?

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(Mother)

Not having a diagnosis may matter very much to you as a parent and it may matter to your child as they get older, so they can understand why they can't walk or see or have an impairment. However, for many practical purposes, it doesn't make any difference whether you have a diagnosis or not. This is because:

- Treatment, therapy or teaching should be tailored to your child's needs, not to the name of their condition.
- You're entitled to have a social services assessment of your child's needs and of your needs as a parent or carer, whether your child has a named diagnosis or not.
- You're entitled to receive benefits such as Disability Living Allowance on the basis of the difficulties that your child has and the support they need. Entitlement does not depend on being able to name the disorder your child has.
- Your child is entitled to have extra or different support to help them at school, if they need it. This does not depend on knowing the cause of their learning difficulties.

Some families are never able to achieve a diagnosis for their child, but as the years go on, some begin to feel that it's not as important to them as it once was.

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(Mother)

Information for parents
When your child has no diagnosis

Where to get support, if you don't know why your child needs help

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(Father)

Many areas in the UK have a local parents' support group where families of children with all kinds of disabilities come together for mutual support and contact. It might be helpful to find out if there's one near you. Even if you had a diagnosis, it's unlikely that another child in the group would have the same condition. However, a lot of the issues you face on a day-to-day basis will be familiar to other parents and they often have practical advice to share. Local groups have the advantage of meeting regularly and locally, which can be important in providing a support network, if you need one. Another advantage is that other members can pass on information about support and services that are available in your area and that they have already used.

Information for parents
When your child has no diagnosis



Voluntary organisations

Voluntary organisations are usually charities that provide help and advice. Find out if there are any that specialise locally and what they can offer. Or contact the following national organisations if you are looking for information and help:

Coventry Family, the UK charity for families with disabled children, can tell you whether there is a local support group in your area. You can also talk things through with a parent adviser, order free factbooks and find out about the support and help that is available. The service is free and confidential.

Contact a Family

209-211 City Road
London EC1Y 1JH

National Freephone Helpline: 0800 808 3555

Telephone Helpline: 0800 808 3556

Email Helpline: helpme@cofamily.org.uk

Web: www.cofamily.org.uk

Syndrome Without a Name (SWAN), the support group for families who have a child with an undiagnosed condition. SWAN has information resources, a newsletter and a helpline, and they can put you in touch with other families where possible.

Syndromes Without a Name (SWAN)

Tel: 01922 701 234

Email: info@undiagnosed.org.uk

Web: www.undiagnosed.org.uk

Information for parents
When your child has no diagnosis

Early Support

Early Support is the central government mechanism for achieving better co-ordinated, family-focused services for young disabled children and their families across England. It is developing at a time of significant change, as part of the restructuring of children's services in response to Every Child Matters and alongside new integrated assessment, information and inspection frameworks for children's services.

Early Support builds on existing good practice. It facilitates the achievement of objectives set by broader initiatives to integrate services, in partnership with families who use services and the many agencies that provide services for young children.

To find out more about Early Support programmes, specialist training opportunities and to view other materials produced by the programme, visit www.earlysupport.org.uk

This booklet is one in a series produced in response to requests from families, professional agencies and voluntary organisations for better standard information about particular conditions or disabilities. The other titles in the series are:

Autistic spectrum disorders (ASD)	(ES12)
Cerebral palsy	(ES10)
Deafness	(ES11)
Down syndrome	(ES13)
If your child has a rare condition	(ES18)
Learning disabilities	(ES15)
Multi-sensory impairment	(ES9)
Speech and language difficulties	(ES14)
Visual impairment	(ES8)

Information for parents
When your child has no diagnosis



This is the second edition of the booklet, which updates information and incorporates comments from those who used the material in 2004-2006.

Other Early Support information about services is available separately, or as part of the Early Support Family pack. The Family pack (ES1) helps families who come into contact with many different professionals to co-ordinate activity and share information about their child through the first few years of life, using a Family file (ES5).



To obtain copies of any of the Early Support materials mentioned here, ring 0845 602 2260 quoting the reference number for the publication.

Early Support would like to thank the many families and professionals that have been involved in development of these resources and to thank **Contact a Family** for their help in writing and more recently revising this booklet.

Contact a Family provides advice, information and support to families with a disabled child. **Contact a Family** provides advice about financial and practical help as well as information on medical conditions and disabilities. They also put families in touch with others through support groups and one to one linking, assist parents to develop their own support groups and provide a voice to raise awareness and campaign for families.

The **Contact a Family** website contains all publications, also available in paper format, including the **Contact a Family Directory**.

Contact a Family

209-211 City Road
London
EC1Y 1PJ

Tel: 020 7608 8700
National Freephone Helpline: 0800 808 3555
Textphone Helpline: 0800 808 3556

Email: info@cafamity.org.uk
Web: www.cafamity.org.uk

Contact a family
for families and disabled children



RESOURCES:

- NIH Office of Rare Diseases (ORD)
http://rarediseases.info.nih.gov/asp/resource/swardia_info.asp
- National Organization for Rare Disorders (NORD) <http://www.rarediseases.org>
- National Human Genome Research Institute: "Undiagnosed Condition in a Child"
<http://www.genome.gov/17515951>
- Genetics Education Center, University of Kansas Medical Ctr., "Unknown Conditions"
<http://www.kumc.edu/gec/prof/genecentr.html>
- National Library of Medicine/Medline Plus
<http://www.nlm.nih.gov/medlineplus>
- National Society of Genetic Counselors
<http://www.nsgc.org/resource/link.cfm>
- Syndromes Without a Name (UK)
<http://www.undiagnosed.org.uk>
- S.W.A.N. in Australia
<http://www.swanusa.bigpondhosting.com>



SWAN-USA
1742 Lorna Lane
Oshego, NY 13828
Amy Chaghtin,



Syndromes

Without

A

Name

USA

SWAN-USA

providing support, information,
and connections to families of
children with undiagnosed
medical conditions



What is a Syndrome?

- A **Syndrome** is a group of signs, symptoms, or features that occur together and create a picture which may suggest the presence of a particular medical condition or disorder.
- **Symptoms** are subjective evidence of a medical condition that are experienced by a patient and reported in the medical history such as pain, nausea, or dizziness.
- **Signs** are objective evidence of a disorder that a physician, or nurse, can recognize during a physical examination such as wheezing, visual impairment, or a seizure.
- Signs and symptoms are important tools that can't help a physician make a diagnosis.
- Some children have only a single symptom, while other children may have a number of symptoms affecting multiple body systems.
- Medical researchers have identified over 6000 rare medical conditions.
- Many disorders have similar signs and symptoms, making diagnosis difficult.
- Specific symptoms of some disorders may not appear until a child is older.
- Some children have signs and symptoms that are so unique, physicians are unable to make a diagnosis.



Why is a Diagnosis Important?

- Information
- Support
- Services
- Treatment Options
- Planning

Without a name or "label" for your child's condition, you may have difficulty obtaining reliable information to help you understand your child's medical condition, finding qualified medical professionals, gaining access to healthcare supports, or even making plans for the school programs your child may need. Finding ways around the "system" to obtain services, or insurance coverage can be confusing, overwhelming, or provoke feelings of anger. You may feel isolated or alone as your child is referred from one specialist to another in the search for a diagnosis. Dealing with physicians or medical professionals can be frustrating due to the long waits, limited appointment time, and difficulty finding a specialist who may be able to offer helpful advice on treatment options, testing, or clinical trials. You may feel like you are living in "limbo," unsure what the future holds for your child and the rest of the family.

SWAN-USA is here to help by offering your family a place to talk, and gain information, advice, ideas, and even encouragement for managing your child's unique medical condition. Although our members children may not have the same exact symptoms as your child, there are many families in a similar situation—living with a child who has an undiagnosed rare disorder. Medical researchers estimate that about 33% of children with special healthcare needs lack a specific diagnosis.

Who is SWAN-USA?

SWAN-USA is a non-profit organization dedicated to providing support to the families of children living with an undiagnosed medical condition. Our goal is to be a resource for patients, their families and the medical community. We try to help families with similar conditions connect for mutual support, help understanding medical information, and exchange ideas, advice, or information through our website and our online support group. Our mission is to:

- Promote policies that will improve the quality of life, health, and welfare of those lacking a diagnosis.
- Provide educational information, programs and support through our newsletter, emails, online support group, and website.
- Help patients and their families obtain benefits, support services, insurance coverage, and medical programs.
- Create a database of undiagnosed syndromes that can be used by physicians as a tool for identifying "unique" conditions.
- Connect with international groups to raise public awareness and understanding of children living with undiagnosed disorders.
- Create a network of medical professionals who can offer advice, support, or help.

For more information, or to join SWAN-USA, please visit our website: www.undiagnosed-usa.org

Or contact:

SWAN-USA
1745 Lorna Lane
Oshtemo, MI 48878



Amy Clugston, President
amyclugston@undiagnosed-usa.org
Phone: (269) 692-2090
Toll Free: (888) 850-7926
www.undiagnosed-usa.org

Appendix 16

Key issues relating to living without a diagnosis as discussed in patient information resources

<p>Contact a family – 'Living Without a Diagnosis'</p>	<p>Possible reasons why it happened Why some conditions are hard to diagnose Whether it's genetic Emotional impact: frustration, feel alone, guilt, anger, despair, acceptance Lack of information Difficulties accessing services, getting a Statement Advice: make a list of questions; take friend or family member with; keep a diary; ask for written information; have a support letter from clinician confirming symptoms; be persistent Influencing services Investigations Who's who Specialities Support groups</p>
<p>Early Support – 'Information for Parents when your Child has no Diagnosis'</p>	<p>Emotional impact – frustration, feel like you are in limbo, lack of control, no prognosis, frightening Why is can be hard to get a diagnosis Does it matter whether you get a diagnosis? Acceptance Still entitled to treatment therapy, social services assessment, benefits, support at school. Support groups</p>
<p>Syndromes Without A Name USA – short information pamphlet</p>	<p>What is a syndrome? Why a diagnosis is important – difficulties obtaining information, gaining access to healthcare support, improve care and treatment, prognosis. Difficulties accessing services and finding way around the 'system' Emotional impact - confusing, overwhelming, anger, frustrating. Difficulty finding a specialist Role of support group Connect families for mutual support</p>

Carrier Testing Leaflet Survey

Below are a number of questions each concerning a different aspect of the carrier testing leaflet. After having read the leaflet, please indicate which you feel is the most appropriate response to each of the questions by circling one of the numbers on the right (or if emailing this form back please use the highlight tool). The scale is explained below.

Scale	
1	Definitely no
2	Not entirely
3	To some degree
4	Definitely yes

Question	Scale			
	1	2	3	4
1. Are the aims of the leaflet clear?	1	2	3	4
2. Does the leaflet achieve its aims?	1	2	3	4
3. Did you understand the scientific information (Section 1) ?	1	2	3	4
4. Was the Genes, Chromosomes and DNA diagram clear?	1	2	3	4
5. Was the discussion about the emotional and social consequences (Section 2) of the test results useful?	1	2	3	4
6. Does the leaflet provide sufficient information about where you can find additional information and support?	1	2	3	4
7. Is the leaflet balanced and unbiased?	1	2	3	4

8. Is the information relevant to your own experience?	1	2	3	4
9. Is it clear who wrote the leaflet and when it was produced?	1	2	3	4
10. Do you like the overall design of the leaflet?	1	2	3	4
11. Based on the answers to all of the above questions, rate the overall quality of the leaflet as a source of information about carrier testing.	1	2	3	4

12. Please use this space to tell me about anything you thought was missing from the leaflet, any sentences you thought were unclear, or any aspect of the leaflet that you disliked.

13. Please use this space to tell me about anything you particularly liked about the leaflet, or any other general comments that you have

Appendix 18

'Living Without A Diagnosis' Leaflet Survey

Below are a number of questions each concerning a different aspect of the leaflet. After having read the leaflet, please indicate which you feel is the most appropriate response to each of the questions by circling one of the numbers on the right (or if emailing this form back please use the highlight tool). The scale is explained below.

Scale	
1	Definitely no
2	Not entirely
3	To some degree
4	Definitely yes

Question	Scale			
1. Are the aims of the leaflet clear?	1	2	3	4
2. Does the leaflet achieve its aims?	1	2	3	4
3. Did it tell you anything new?	1	2	3	4
4. Did you feel overwhelmed with information?	1	2	3	4
5. Did you find the information too limited?	1	2	3	4
6. Is the information relevant to your own experience?	1	2	3	4
7. Is the leaflet balanced and unbiased?	1	2	3	4

8. Does the leaflet provide sufficient information about where you can find additional information and support?	1	2	3	4
9. Is it clear who wrote the leaflet and when it was produced?	1	2	3	4
10. Do you like the overall design of the leaflet?	1	2	3	4
11. Overall, do you think the leaflet is a useful source of information about parenting a child without a diagnosis?	1	2	3	4

12. Please use this space to tell me about anything you thought was missing from the leaflet, any sentences you thought were unclear, or any aspect of the leaflet that you disliked.

13. Please use this space to tell me about anything you particularly liked about the leaflet, or any other general comments that you have



Developing Resources for Patients

Dear

At the beginning of this year (or the end of last year), I interviewed you about your experience of genetic testing. One of the aims of this interview was to help me develop a new leaflet for patients and families. At the time of the interview, you said that you would be interested in receiving a copy of this leaflet once it had been developed.

I have now completed the first main draft of this leaflet, and I am sending it to you to see what you think. It would be really helpful for me if you could read through the leaflet which I have enclosed, and then respond to the questions attached to this letter. You can then send your responses back to me in the Freepost envelope. If I interviewed both you and your partner, I have enclosed two copies of the questions (and two Freepost envelopes). This is because I would like to hear from both of you separately.

Once again, many thanks for letting me interview you, and for taking the time to answer these questions.

Celine.

What Does It Mean To Be A Carrier Of A Recessive Condition?



Information for Patients and Families

What Does It Mean To Be A Carrier Of A Recessive Condition?

What is a carrier?

A carrier is someone who carries a changed copy of a gene, which can lead to future children having a genetic condition.

- Being a carrier does not mean you have a genetic condition.
- Being a carrier usually does not affect your health in any way.
- The only time when being a carrier can cause problems is if it can lead to your children having a genetic condition.

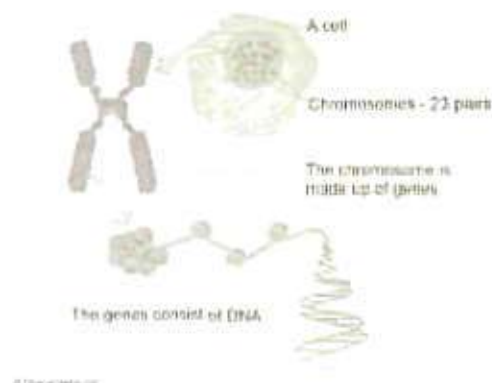
To understand better what it means to be a carrier, it is helpful to understand what genes and chromosomes are.

Genes and chromosomes

Our bodies are made up of millions of cells. Most cells contain a complete set of genes. We have thousands of genes. Genes act like a set of instructions, controlling our growth and how our bodies work. They are responsible for many of our characteristics, such as our eye colour, blood type or height.

Genes are carried on thread-like structures called chromosomes. Usually, we have 46 chromosomes in most cells. We inherit our chromosomes from our parents, 23 from our mother and 23 from our father, so we have two sets of 23 chromosomes, or 23 pairs. Because the chromosomes are made up of genes, we therefore inherit two copies of most genes, one copy from each parent. This is the reason why we often have similar characteristics to our parents. The chromosomes, and therefore the genes, are made up of a chemical substance called DNA.

Figure 1: Genes, chromosomes and DNA



Sometimes, there is a change (mutation) in one copy of a gene which stops it from working properly. However, because we have two copies of most genes, the normal copy compensates for the copy with the change. Being a carrier therefore means that you do not have the condition, but carry a changed copy of the gene on one of a pair of chromosomes.

Being a carrier usually does not affect your health in any way. In fact, we are all carriers of a number of gene mutations. The only time when being a carrier can cause problems is if it can lead to your children having a genetic condition.

When can being a carrier lead to our children being affected by a genetic condition?

If both partners are carriers of the same changed gene, they may pass on either their normal gene or their changed gene to their child. This occurs randomly.

Each child therefore has a 25% (1 in 4) chance of inheriting a changed gene from both parents and being affected by the condition.

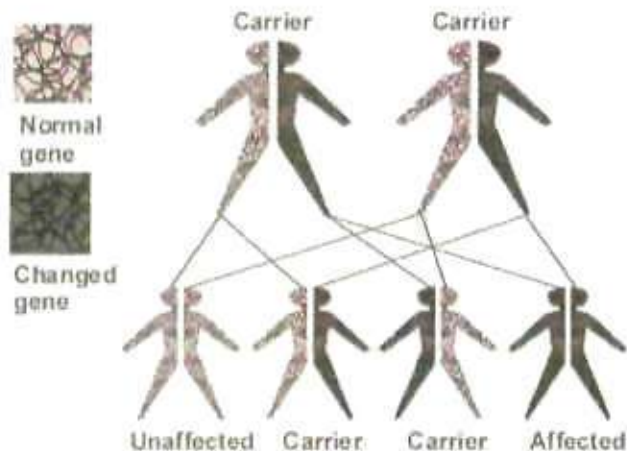
This also means that there is a 75% (3 in 4) chance that a child will not be affected by the condition.

There is also a 50% (2 in 4) chance that the child will inherit just one copy of the changed gene from a parent. If this happens, then they will be healthy carriers like their parents.

Lastly, there is a 25% (1 in 4) chance that the child will inherit both normal copies of the gene. In this case the child will not have the condition, and will not be a carrier.

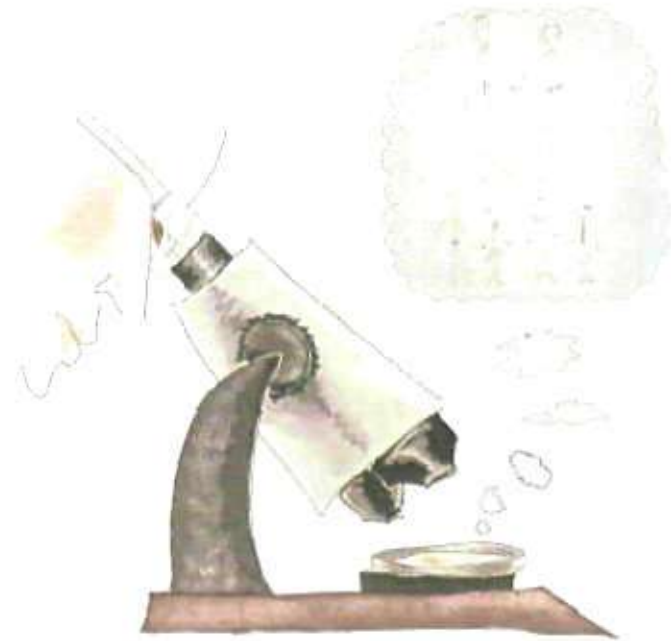
These possible outcomes occur randomly. The chance remains the same in every pregnancy and is the same for boys and girls.

Common conditions inherited in this way include: cystic fibrosis, sickle cell, beta-thalassaemia, Tay Sachs disease, inherited haemochromatosis and spinal muscular atrophy.



Publication date: January 2010

What Does It Mean To Be A Carrier Of A Recessive Condition?



Information for Patients and Families

What Does It Mean To Be A Carrier Of A Recessive Condition?

This information is for people who have found out they are a carrier of a recessive condition. It provides information about what it means to be a 'carrier', and practical and emotional information about living with your test results. Much of this information has been gathered by speaking to people who are themselves carriers. We hope you will find it helpful.

Section 1: What is a carrier?

A carrier is someone who 'carries' a changed copy of a gene which can lead to future children having a genetic condition.

- Being a carrier does not mean you have a genetic condition.
- Being a carrier usually does not effect your health in any way.
- The only time when being a carrier can cause problems is if it can lead to your children having a genetic condition.

To understand better what it means to be a carrier, it is helpful to understand what genes and chromosomes are.

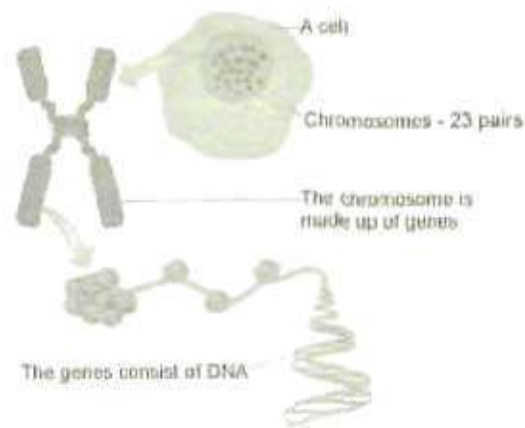
Genes and chromosomes

Our bodies are made up of millions of cells. Most cells contain a complete set of genes. We have thousands of genes. Genes act like a set of instructions controlling our growth and how our bodies work. They are responsible for many of our characteristics, such as our eye colour, blood type or height.

Genes are carried on thread-like structures called chromosomes. Usually, we have 46 chromosomes in most cells. We inherit our

chromosomes from our parents, 23 from our mother and 23 from our father, so we have two sets of 23 chromosomes, or 23 'pairs'. Because the chromosomes are made up of genes, we therefore inherit two copies of most genes - one copy from each parent. This is the reason why we often have similar characteristics to our parents. The chromosomes, and therefore the genes, are made up of a chemical substance called DNA.

Figure 1: Genes, chromosomes and DNA



© Unicef/Debi Ltd

Sometimes, there is a change (mutation) in one copy of a gene which stops it from working properly. However, because we have two copies of most genes, the normal copy compensates for the copy with the change. Being a carrier therefore means that you do not have the condition, but carry a changed copy of the gene on one of a pair of chromosomes.

Being a carrier usually does not affect your health in any way. In fact, we are all carriers of a number of gene mutations. The only time when being a carrier can cause problems is if it can lead to your children having a genetic condition.

When can being a carrier lead to our children being affected by a genetic condition?

If both partners are carriers of the same changed gene, they may pass on either their normal gene or their changed gene to their child. This occurs randomly.

Each child therefore has a 25% (1 in 4) chance of inheriting a changed gene from both parents and being affected by the condition.

This also means that there is a 75% (3 in 4) chance that a child will not be affected by the condition.

There is also a 50% (2 in 4) chance that the child will inherit just one copy of the changed gene from a parent. If this happens, then they will be healthy carriers like their parents.

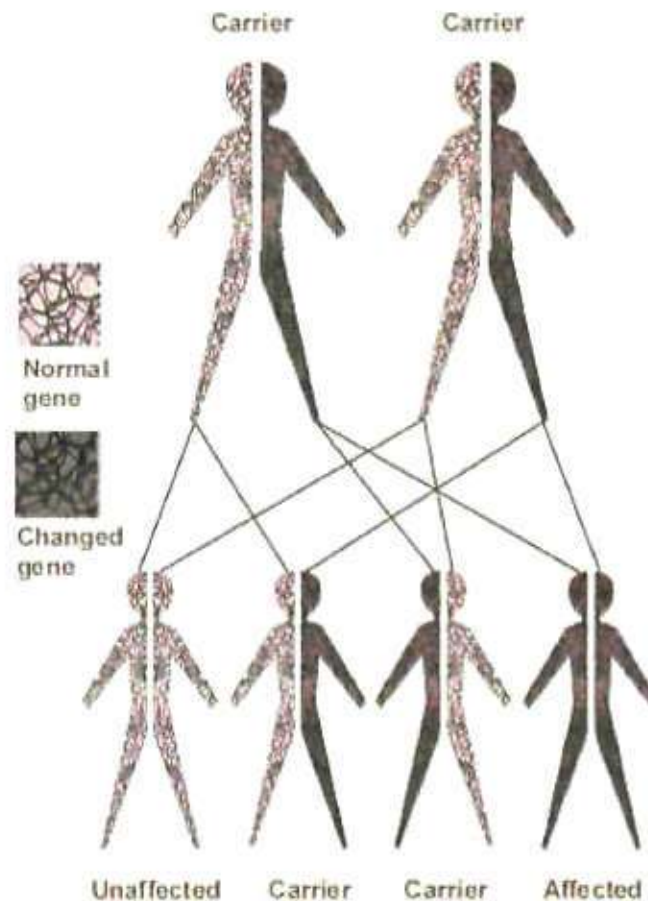
Lastly, there is a 25% (1 in 4) chance that the child will inherit both normal copies of the gene. In this case the child will not have the condition, and will not be a carrier.

These possible outcomes occur randomly. The chance remains the same in every pregnancy and is the same for boys and girls.

Common conditions inherited in this way include:

- cystic fibrosis
- sickle cell
- beta-thalassaemia
- Tay-Sachs disease
- inherited haemochromatosis
- spinal muscular atrophy

Figure 2: How recessive conditions are passed on from parent to child



Section 2: Living with your carrier status

The following information discusses the various experiences of people who have found out they are carriers. We have tried to highlight a range of issues and emotions that finding out you are a carrier can cause, however they may not all be relevant to you.

How might I react?

People react in all sorts of ways when they receive their test results. Many people say that they feel angry or worried when they first find out that they are a carrier. Some people report that they feel sad, surprised or shocked. All these reactions are normal. For most people these feelings subside after a few months.

"To find out I was a carrier and my partner was also a carrier, I just felt gutted. Also, when you look at the statistics, to be one and then your partner to be one is so rare, I just felt really cross about it, it was just such awful bad luck."

"I was actually quite surprised, I don't know why because my sister is a carrier so I guess I should have expected it in a way, but I was quite shocked that I was a carrier."

It is quite normal to feel somehow different about yourself when you find out you are a carrier. Some people say that it is strange to find out something new about yourself when you think you know everything. It can take time to adapt to this new information.

Some people say that finding out they are a carrier makes them feel 'less healthy'. Other carriers say that they are concerned that, in the future, they will be more likely to develop health problems. These reactions are very normal but it is important to remember that your carrier status has no effect on your health. We are all carriers of a number of gene mutations.

Studies have shown that carriers cope better with their results if they recognise that being a carrier is something that you cannot change, and that the information can be used positively.

"I've just accepted it now. I've moved on from that. Life throws things at you and you just get on with it really. It's just another one of life's twists."

"My mother said to me, 'well how do you feel about it?', and I said 'fine, at least I know'."

Future children

For some people, finding out they are a carrier is a source of worry because it affects their plans to have children. For other people, it is a relief to know so that they can plan ahead. Knowing that there is an increased risk that your future children might have a genetic condition means that you can be prepared and take the time to make important decisions. There are a number of options you may wish to consider.

For some genetic conditions, it is possible to perform a test during pregnancy (prenatal test), such as an amniocentesis or CVS test, to see if the baby has inherited the genetic condition. If you think this might be an option for you, speak to your doctor about whether these tests are available for the condition you are concerned about. If possible do this before the pregnancy as the laboratory may have to make preparations that can take several months.

If you are considering prenatal testing you should think about what you would do if the baby was found to have a genetic condition, and how you might feel about a termination of pregnancy.



It may be possible to perform a technique called Preimplantation Genetic Diagnosis (PGD) as an alternative to testing the baby during pregnancy. This involves the couple undergoing medically assisted reproduction, after which the fertilised eggs are tested to see if they have the changed gene. Only those eggs without the changed gene are implanted into the woman's womb. This is a demanding process and is not suitable for everyone. For more information about PGD, and whether it is available to you, you should speak to your doctor.

Other options you might want to consider include adoption, conceiving using donor eggs or sperm, or the possibility of not having children.

"The good thing is when I went for the genetic counselling I found out there were choices. If you want to have more children, there are ways you can go about it."

What if you already have children?

Some parents who already have a child with a genetic condition, say that one of the emotions that they experience when they find out they are a carrier is feeling 'guilty' for having 'passed on' the child's condition. It is entirely natural to feel this way. If you are having these kinds of feelings you should talk them through with your genetic specialist. It is important to remember that genes are distributed by chance and having a changed gene is not your fault. Over time it has been shown that these feelings usually lessen in intensity.

If you find out that you are a carrier and you have children, even if they are not affected by the condition, there is still a chance that they could be carriers. It is important that you



discuss this risk with your children at an age which is suitable. For some parents this might be when they think the child is old enough to understand. For others it might be when their children begin to have serious relationships. The age at which a young person can have a carrier test varies, but the person having the test has to make their own decision about it.

Relationship with partner

Knowing about your carrier status can have an effect on your relationship with your partner. For some people it can bring them closer together and they can help support one another. However, knowing your carrier status can also cause tension and strain within the relationship. Sometimes couples will have to discuss issues that are very difficult and upsetting.

"Knowing that we were both carriers, I knew that was going to add a big complication to our lives, but it never made me not want to be with him. It just made me realise, ok so we're both carriers, so this is something we're going to have to think about and discuss in terms of our future together."

Other family members

If you find out that you are a carrier, you may wish to discuss this with other family members. This gives other family members the opportunity to have a blood test to see if they are also carriers, if they wish. This information may also be useful in helping diagnose other family members. It might also be particularly important to family members who are likely to have children in the future. Discussing your carrier test results is your choice and your test results will never be shared with other family members without your permission.

"They now know that it's in the family and they can be tested if they want to, they have that choice. And it's useful for them when they want to have children."

For some people, sharing information with other family members is a positive experience. It can bring families closer together, and family members can provide a good source of support. Others find it difficult because they are worried about causing anxiety in the family. In some families, people have lost touch with relatives and may feel it is difficult to contact them.

Discussing test results with grandparents can be particularly difficult. They may not want to accept that the changed gene is something that could have been passed down from them. It is also not uncommon for grandparents to feel guilty because they feel it is their fault. These are reactions you should be aware of.

"My mum spoke to my nan and she said 'it's not from me full stop'. So she blocked it off, she said 'I didn't bring anything into the family.'"

"She was really cut up about it. She felt guilty. And I said look, it's just one of these things, it's not your fault."

Genetic specialists often have a lot of experience with families in these situations and may be able to offer you help in discussing the situation with other family members. It can also be helpful to have information provided by your genetic specialist to show to other family members to help explain what it means to be a carrier, and that being a carrier is something that occurs by chance.

Other sources of support

Genetic specialists and other health professionals (such as counsellors and psychologists) are experienced in helping people talk through the emotions that relate to receiving carrier test results. They can be a good source of information and support.

"Seeing Emma [the genetic counsellor] was very reassuring because before then I hadn't really understood that there was this thing called a CVS or an amniocentesis. So it was just good to have a professional explain all the different options. It was very helpful."

Some people also find it helpful to contact a patient support group. Patient support groups can provide information about the practical and emotional aspects of being a carrier of a condition. Many have a website and helpline giving information and advice. They can often put people and families in touch with others who are in a similar situation. They may have a chat forum on which members can email other members.

"We've got quite a lot of information from the patient support group and so it's been helpful. Also, knowing that some nights they work late, if you want to ring them up and have a chat to them, they will chat to you over the phone, send things in the post, work with the schools and so on."

Further information

For further information, contact your local regional genetics service. Their contact details can be found at www.gig.org.uk/services.htm

Additional information can also be found at:

Contact a Family

UK charity for families with disabled children.
Offers information on specific conditions and rare disorders.
Helpline 0808 808 3555
(Mon-Fri, 10am-4pm and Mon, 5.30pm-7.30pm)
Email helpline@cafamily.org.uk
Web: www.cafamily.org.uk



Climb

The National Information
Centre for Metabolic Diseases.
Have information about Tay-
Sachs disease.

Tel: 0800 652 3181
Email: info@svcs@climb.org.uk
Web: www.climb.org.uk

Cystic Fibrosis Trust

Tel: 0845 859 1000
Email: enquiries@cftrust.org.uk
Web: www.cftrust.org.uk

Genetic Alliance UK

Provides information about
specific genetic conditions and
contact details of support
organisations.

Tel: 020 7704 3141
E-mail:
mail@geneticalliance.org.uk
Web: www.geneticalliance.org.uk

Haemochromatosis Society

Tel: 02084491363
Email:
info@haemochromatosis.org.uk
Web:
www.haemochromatosis.org.uk

**The Jennifer Trust (For Spinal
Muscular Atrophy)**

Tel: 0800 975 3100 (9am-5pm)
Email: jennifer@jtsma.org.uk
Web: www.jtsma.org.uk

The Sickle Cell Society

Tel: 020 8961 7755
Tel: 0800 0015666 (24-hour
helpline)
Email: info@sicklecellsociety.org
Web: www.sicklecellsociety.org

UK Thalassaemia Society

Tel: 020 8882 0011
Email: office@ukts.org.uk
Web: www.ukts.org

We are extremely grateful to all the people that allowed us to interview them during the making of this leaflet.

This information was developed by Genetic Alliance UK, a national alliance of patient organisations which supports children, families and individuals affected by genetic disorders.

Publication date: January 2010

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What Does It Mean To Be A Carrier Of An X Linked Condition?



Information for Patients and Families

What Does It Mean To Be A Carrier Of An X Linked Condition?

What is a carrier?

A carrier is someone who 'carries' a changed copy of a gene, which can lead to future children having a genetic condition.

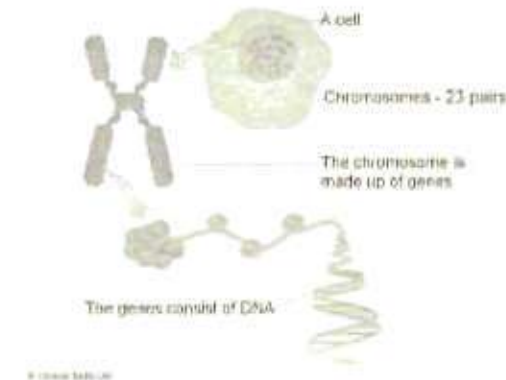
- Being a carrier does not mean you have a genetic condition.
- Being a carrier usually does not effect your health in any way.
- The only time when being a carrier can cause problems is if it can lead to your children having a genetic condition.

To understand better what it means to be a carrier, it is helpful to understand what genes and chromosomes are.

Genes and chromosomes

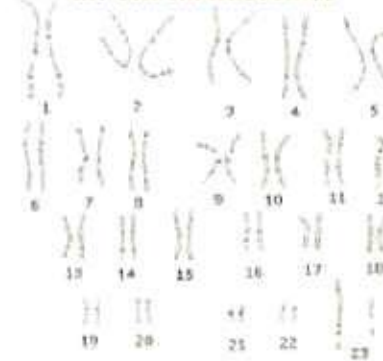
Our bodies are made up of millions of cells. Most cells contain a complete set of genes. We have thousands of genes. Genes act like a set of instructions, controlling our growth and how our bodies work. They are responsible for many of our characteristics, such as our eye colour, blood type or height.

Figure 1: Genes, chromosomes and DNA



Genes are carried on thread-like structures called chromosomes. Usually, we have 46 chromosomes in most cells. We inherit our chromosomes from our parents, 23 from our mother and 23 from our father, so we have two sets of 23 chromosomes, or 23 'pairs'.

Figure 2: 23 pairs of chromosomes. The last pair are the sex chromosomes



One of the pair of chromosomes are called the 'sex chromosomes', and control whether we are male or female. Females normally have two X chromosomes (XX). Males normally have an X and a Y chromosome (XY).

Figure 2 therefore shows the chromosomes of a male as the last pair of chromosomes are XY.

Sometimes, there is a change (mutation) in a gene which stops it from working properly. In fact, we all carry of a number of gene mutations. However, X linked conditions occur when there is a changed gene (mutation) on the X chromosome.

Females have two X chromosomes (XX). Therefore, if a female has a changed gene on an X chromosome, the normal gene on the other chromosome can compensate for the changed copy. If this happens the female is usually a healthy carrier of the X linked condition. Being a carrier means that you do not have the condition, but carry a changed copy of the gene. In some cases, females show mild signs of the condition.

Males have an X and a Y chromosome (XY). Therefore, if a male has a changed gene on an X chromosome, he does not have another copy of that gene to compensate for the changed copy. This means that he will be affected by the condition.

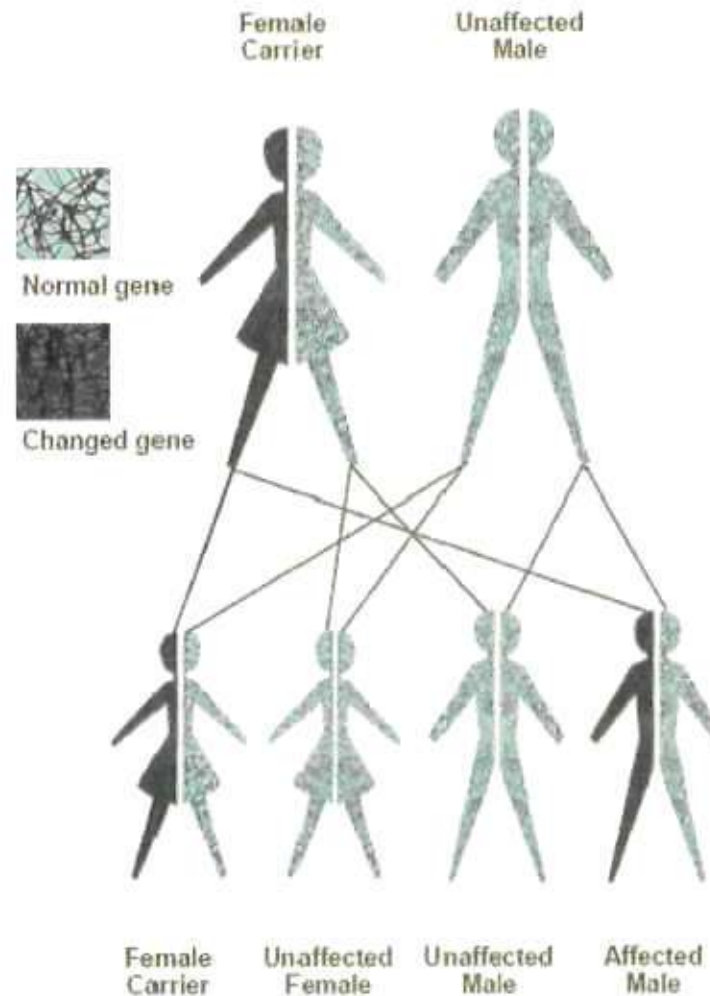
Some examples of X linked conditions include haemophilia, Duchenne muscular dystrophy and fragile X.

When can being a carrier lead to our children being affected by a genetic condition?

If a female carrier has a son, there is a 50% chance (1 in 2) of inheriting the changed gene and being affected by the condition. There is also a 50% chance (1 in 2) that the son will inherit the normal gene. If this happens he will not be affected by the condition. This chance remains the same for every son.

If a female carrier has a daughter, there is a 50% chance (1 in 2) of inheriting the changed gene and being a carrier, like her mother. There is also a 50% chance (1 in 2) that the daughter will inherit the normal gene. If this happens she will not be a carrier, and will be totally unaffected by the condition. This chance remains the same for every daughter.

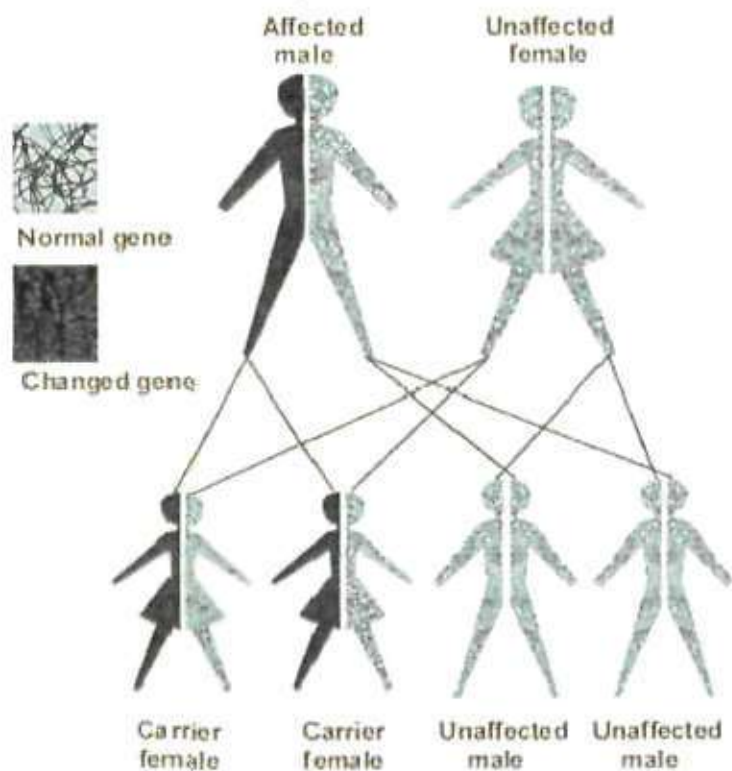
Figure 3: How X linked recessive conditions are passed on by female carriers



If a male who has an X linked condition has a daughter, he will **always** pass on the changed gene to her. All his daughters will therefore be carriers. The daughters will usually not have the condition, but they are at risk of having affected sons.

If a male who has an X linked condition has a son, his son will **never** inherit the changed gene. All his sons will therefore be unaffected by the condition, and cannot pass the condition on.

Figure 4: How X linked recessive conditions are passed on by affected males



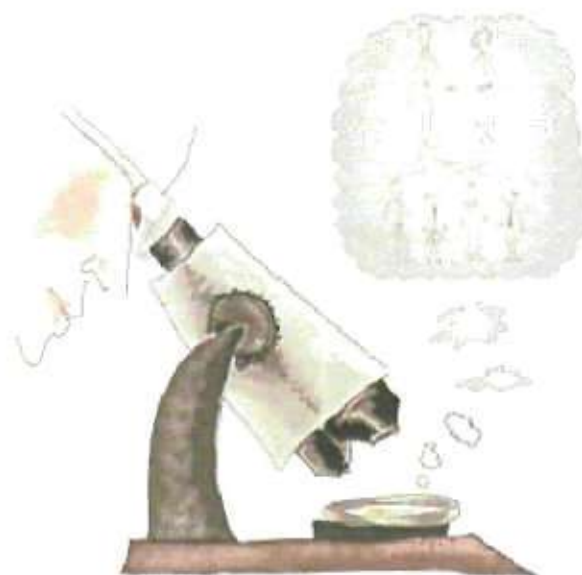
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What Does It Mean To Be A Carrier Of An X Linked Condition?



Information for Patients and Families

What Does It Mean To Be A Carrier Of An X Linked Condition?

This information is for people who have found out they are a carrier of an X linked condition. It provides information about what it means to be a 'carrier', and practical and emotional information about living with your test results. Much of this information has been gathered by speaking to people are themselves carriers. We hope you will find it helpful.

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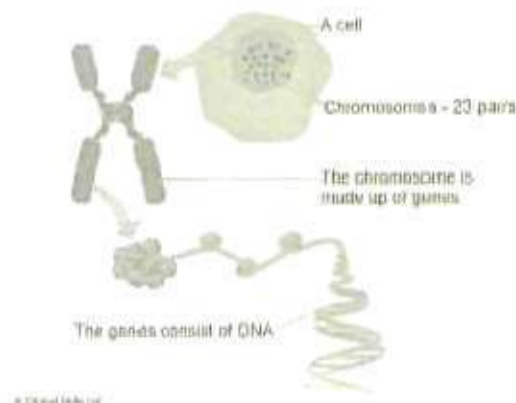
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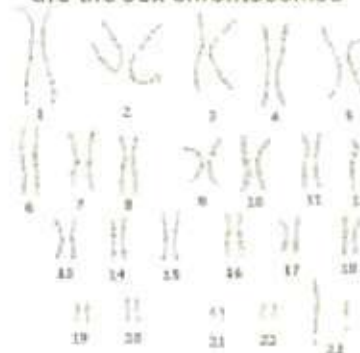
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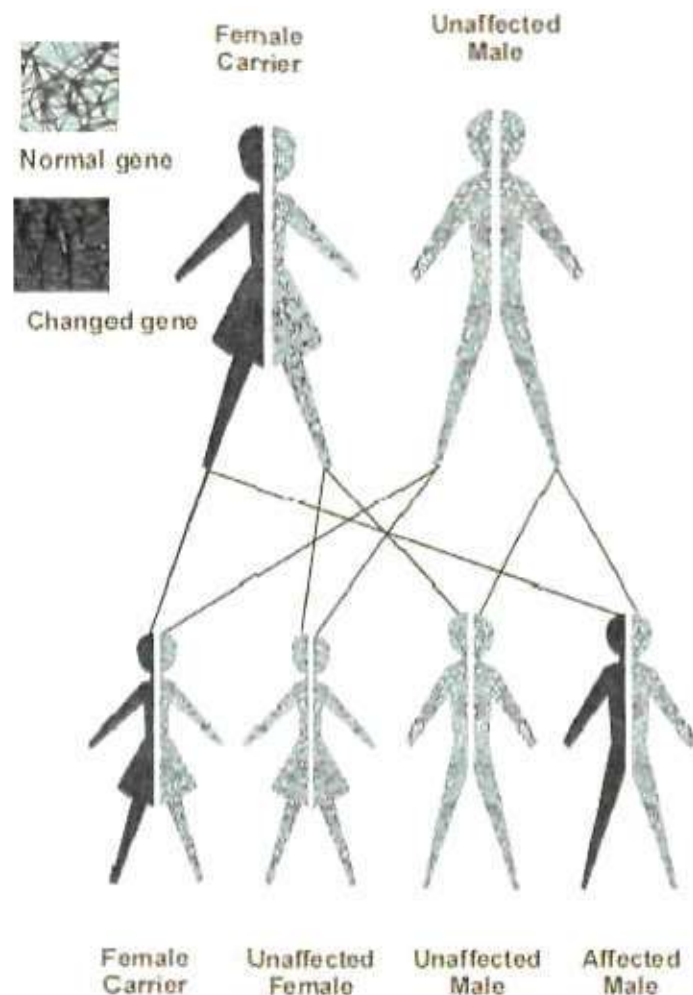
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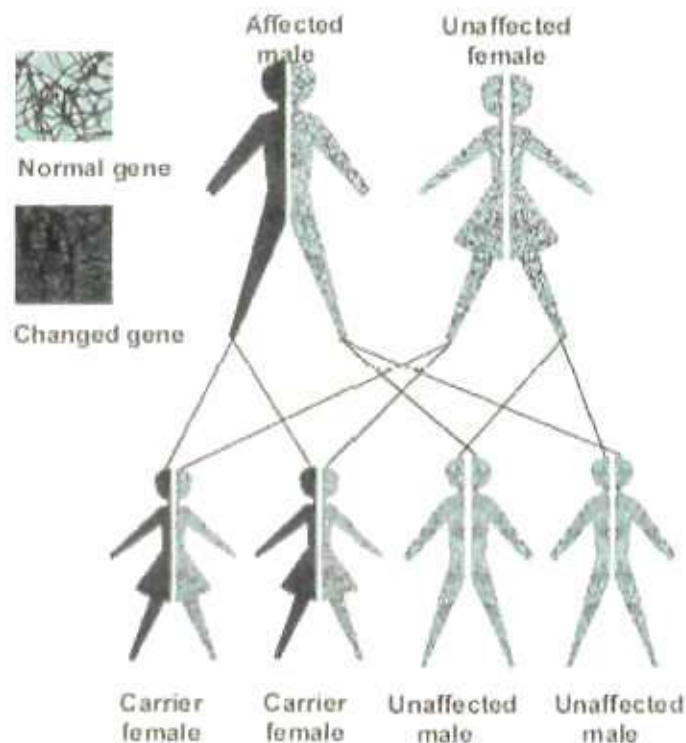
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Section 2: Living with your carrier status

The following information discusses the various experiences of people who have found out they are carriers. We have tried to highlight a range of issues and emotions that finding out you are a carrier can cause, however they may not all be relevant to you.

How might I react?

People react in all sorts of ways when they receive their test results. Many people say that they feel angry or worried when they first find out that they are a carrier. Some people report that they feel sad, surprised or shocked. All these reactions are normal. For most people these feelings subside after a few months.

"I just felt really cross about it."

It is quite normal to feel somehow different about yourself when you find out you are a carrier. Some people say that it is strange to find out something new about yourself when you think you know everything. It can take time to adapt to this new information.

"For thirty-one years you think there is nothing wrong and then all of sudden you find out there is this genetic thing..."

Some people say that finding out they are a carrier makes them feel 'less healthy'. Other carriers say that they are concerned that, in the future, they will be more likely to develop health problems. These reactions are very normal but it is important to remember that your carrier status should have no effect on your health. Your doctor will tell you if there are any health implications relating to your carrier status.

Studies have shown that carriers cope better with their results if they recognise that being a carrier is something that you cannot change, and that the information can be used positively.

"I've just accepted it now. I've moved on from that. Life throws things at you and you just get on with it really. It's just another one of life's twists."

"My mother said to me, 'well how do you feel about it?' and I said 'fine, at least I know'."

Future children

For some people, finding out they are a carrier is a source of worry because it affects their plans to have children. For other people, it is a relief to know so that they can plan ahead. Knowing that there is an increased risk that your future children might have a genetic condition means that you can be prepared and take the time to make important decisions. There are a number of options you may wish to consider.

For some genetic conditions, it is possible to perform a test during pregnancy (prenatal test), such as an amniocentesis or CVS test, to see if the baby has inherited the condition. If you think this might be an option for you, speak to your doctor about whether these tests are available for the condition you are concerned about. If possible do this before the pregnancy as the laboratory may have to make preparations that can take several months.

If you are considering prenatal testing you should think about what you would do if the baby was found to have a genetic condition, and how you might feel about a termination of pregnancy.

It may be possible to perform a technique called Preimplantation Genetic Diagnosis (PGD) as an alternative to testing the baby during pregnancy. This involves the couple undergoing medically assisted reproduction, after which the fertilised eggs are tested to see if they have the changed



gene. Only those eggs without the changed gene are implanted into the woman's womb. This is a demanding process and is not suitable for everyone. For more information about PGD, and whether it is available to you, you should speak to your doctor.

Other options you might want to consider include adoption, conceiving using donor eggs or sperm, or the possibility of not having children.

"The good thing is when I went for the genetic counselling I found out there were choices. If you want to have more children, there are ways you can go about it."

What if you already have children?

Some parents who already have a child with a genetic condition, say that one of the emotions that they experience when they find out they are a carrier is feeling 'guilty' for having 'passed on' the child's condition. It is entirely natural to feel this way. Mothers of boys who have X linked conditions sometimes say that they feel 'blamed' by their male partners for having passed on the changed gene, or blame themselves. If you are having these kinds of feelings you should talk them through with your genetic specialist. It is important to remember that genes are distributed by chance and having a changed gene or chromosome rearrangement is not your fault. Over time it has been shown that these feelings usually lessen in intensity.

"I do feel sort of that I've let my family down or let my husband down, and obviously let my son down because obviously I gave him something that he didn't really need to deal with for the rest of his life."

If you find out that you are a carrier and you have children, even if they are not affected by the condition, there is still a chance that your daughters could be carriers. It is important that you discuss this risk with your children at an age which is suitable.

For some parents this might be when they think the child is old enough to understand. For others it might be when their children begin to have serious relationships. The age at which a young person can have a carrier test varies, but the person having the test has to make their own decision about it.

Relationship with partner

Knowing about your carrier status can have an effect on your relationship with your partner. For some people it can bring them closer together and they can help support one another.

"I just had to try and be there. She was very upset. You have to just listen and talk about it, just really be there and be supportive." (partner of a carrier)

However, knowing your carrier status can also cause tension and strain within the relationship. Sometimes couples will have to discuss issues that are very difficult and upsetting.

"I feel like I can cope with a lot, but to have to pull in someone you really care about, that felt horrible. That felt worse than dealing with it myself."

Other family members

If you find out that you are a carrier, you may wish to discuss this with other family members. This gives other family members the opportunity to have a blood test to see if they are also carriers, if they wish. This information may also be useful in helping diagnose other family members. It might also be particularly important to family members who are likely to have children in the future. Discussing your carrier test results is your choice and your test results will never be shared with other family members without your permission.

"They now know that it's in the family and they can be tested if they want to, they have that choice. And it's useful for them when they want to have children."

For some people, sharing information with other family members is a positive experience. It can bring families closer together, and family members can provide a good source of support. Others find it difficult because they are worried about causing anxiety in the family. In some families, people have lost touch with relatives and may feel it is difficult to contact them.

"You kind of feel like you're bringing a blight into someone else's family and that really did affect me."

For grandparents, it can be particularly difficult. They may not want to accept that the changed gene is something that could have been passed down from them. It is also not uncommon for grandparents to feel guilty because they feel it is their fault. These are reactions you should be aware of.

"My mum spoke to my nan and she said it's not from me full stop. So she blocked it off, she said I didn't bring anything into the family."

"She was really cut up about it. She felt guilty. And I said look, it's just one of those things, it's not your fault."



Genetic specialists often have a lot of experience with families in these situations and may be able to offer you help in discussing the situation with other family members. It can also be helpful to have information provided by your genetic specialist to show to other family members to help explain what it means to be a carrier, and that being a carrier is something that occurs by chance.

Other sources of support

Genetic specialists and other health professionals (such as counsellors and psychologists) are experienced in helping people talk through the emotions that relate to receiving carrier test results. They can be a good source of information and support.

"Seeing Emma [the genetic counsellor] was very reassuring because before then I hadn't really understood that there was this thing called a CVS or an amniocentesis. So it was just good to have a professional explain all the different options. It was very helpful."

Some people also find it helpful to contact a patient support group. Patient support groups can provide information about the practical and emotional aspects of being a carrier of a condition. Many have a website and helpline giving information and advice. They can often put people and families in touch with others who are in a similar situation. They may have a chat forum on which members can email other members.

"We've got quite a lot of information from the patient support group and so it's been helpful. Also, knowing that some nights they work late, if you want to ring them up and have a chat to them, they will chat to you over the phone, send things in the post, work with the schools and so on."



Further information

For further information, contact your local regional genetics service. Their contact details can be found at: www.gig.org.uk/services.htm

Additional information can also be found at:

Contact a Family

UK charity for families with disabled children. Offers information on specific conditions and rare disorders.
Helpline 0808 808 3555
(Mon-Fri, 10am-4pm and Mon, 5.30pm-7.30pm)
Email ihelpline@cafamily.org.uk
Web: www.cafamily.org.uk

Fragile X Society

Tel: 01371 875100
Email: info@fragilex.org.uk
Web: www.fragilex.org.uk

Genetic Alliance UK

Provides information about specific genetic conditions and contact details of support organisations
Tel: 020 7704 3141
E-mail: mail@geneticalliance.org.uk
Web: www.geneticalliance.org.uk

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Haemophilia Society

Tel: 020 7831 1020

Email: info@haemophilia.org.uk

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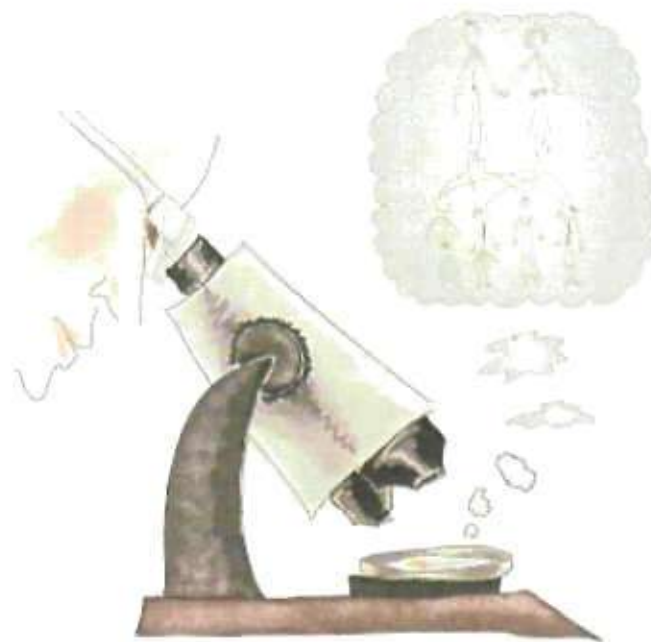
Muscular Dystrophy Campaign

Tel: 0800 652 6352 (freephone information line)

Email: info@muscular-dystrophy.org

Web: www.muscular-dystrophy.org

What Does It Mean To Be A Carrier Of A Balanced Translocation?



Information for Patients and Families

What Does It Mean To Carry A Balanced Translocation?

What is a carrier?

A translocation carrier is someone who 'carries' an unusual arrangement of chromosomes.

- Being a carrier does not mean you have a genetic condition.
- Being a carrier usually does not effect your health in any way.
- Usually, the only time when being a carrier can cause problems is when it comes to having children.

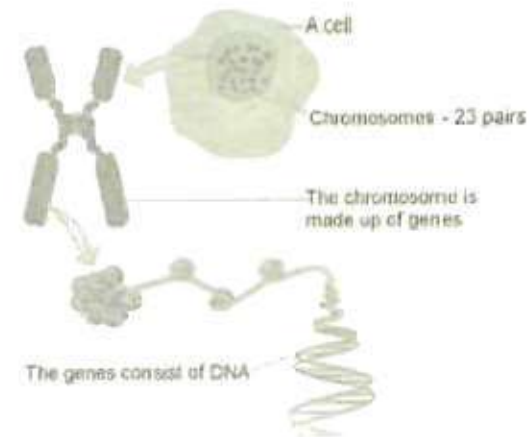
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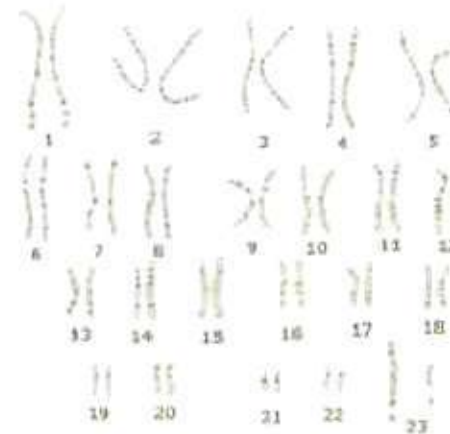
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Figure 1: Genes, chromosomes and DNA



© 1999 OF BAHU, Ltd

Figure 2: 23 pairs of chromosomes arranged according to size



It is important that we have the correct amount of chromosome material, as the genes (that instruct the cells in our body) are found on the chromosomes. Having some part of a chromosome missing, or having an extra part of a chromosome, can result in a person being born with learning difficulties, developmental delay and health problems.

What is a translocation?

A translocation means that there is an unusual arrangement of the chromosomes. This can happen because:

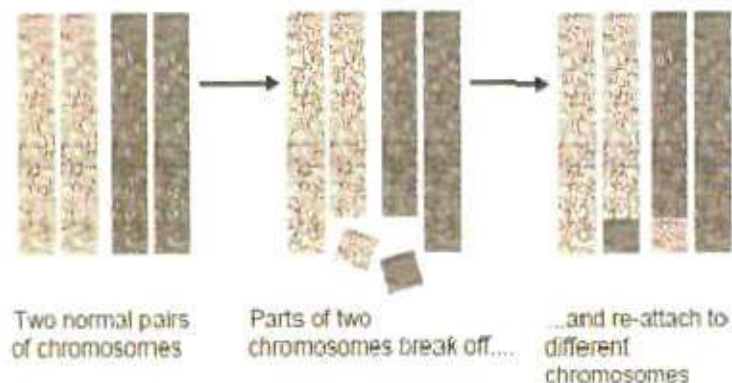
- a change has arisen during the making of the egg or the sperm or around the time of conception
- an altered chromosome arrangement has been inherited from either the mother or the father

There are two main types of translocations: a **RECIPROCAL** translocation and a **ROBERTSONIAN** translocation.

Reciprocal translocations

A reciprocal translocation occurs when two fragments break off from two different chromosomes and swap places.

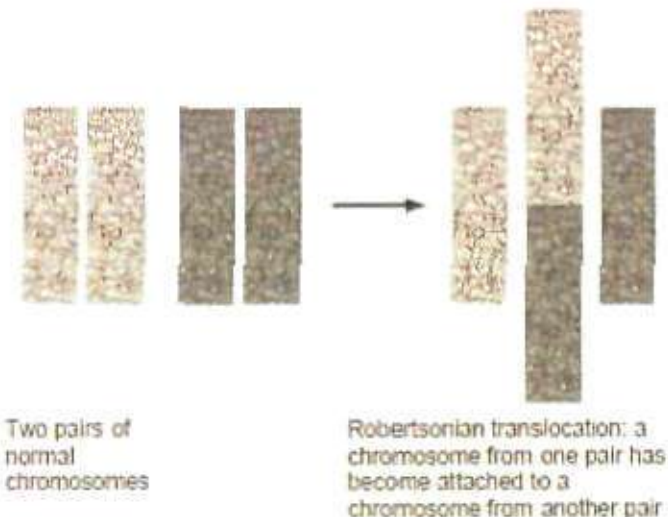
Figure 3: How a reciprocal translocation arises



Robertsonian translocations

A Robertsonian translocation occurs when one chromosome becomes attached to another. Picture 4 shows a Robertsonian translocation involving two chromosomes.

Picture 4: How a Robertsonian translocation arises



Why do translocations happen?

Although about 1 person in 500 has a translocation, we still do not really understand why they happen. We know that chromosomes seem to break and rejoin quite often during the making of sperm and eggs or around the time of conception, and it is only sometimes that this leads to problems. These changes occur without us being able to control them.

When might this lead to problems?

In both the examples we have looked at, the chromosomes have been rearranged so that no chromosome material has been lost or gained. This is called a **balanced translocation**. A person who carries a **balanced translocation** is not usually affected by it, and is often unaware of having it. The only time it may become important is when he or she comes to have children. This is because the child may inherit what we call an **unbalanced translocation**.

Unbalanced translocations

If either parent carries a balanced translocation, it is possible that their child may inherit an **unbalanced translocation** in which there is an **extra piece of one chromosome and/or a missing piece of another chromosome**.

A person who has an unbalanced translocation may have learning disability, developmental delay and health problems. The seriousness of the disability depends on exactly which parts of which chromosomes are involved and how much missing or extra chromosome material there is. This is because some parts of the chromosome are more important than other parts.



If a parent has a balanced translocation will he or she always pass it on?

Not necessarily, there are several possibilities for each pregnancy:

- The child may inherit entirely normal chromosomes.
- The child may inherit the same balanced translocation as the parent and not have any problems.
- The child may inherit an unbalanced translocation, and may be born with some degree of developmental delay, learning disability or health problems.
- The pregnancy ends in miscarriage.

Therefore it is quite possible for a person who carries a balanced translocation to have healthy children, and many do. However, the risk that a carrier of a balanced translocation will have a child with some degree of disability is higher than average, although the severity of the disability depends on the amount of material affected by the translocation.

Frequently a child can be born with a translocation although both parents' chromosomes are normal. This is called a "*de novo*" (from Latin) or new rearrangement. In this case the parents are unlikely to have another child with a translocation in the future.

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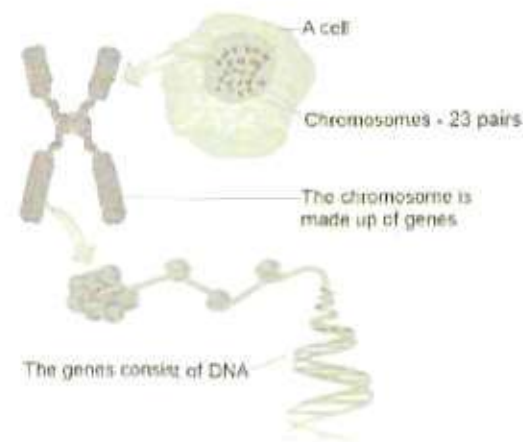
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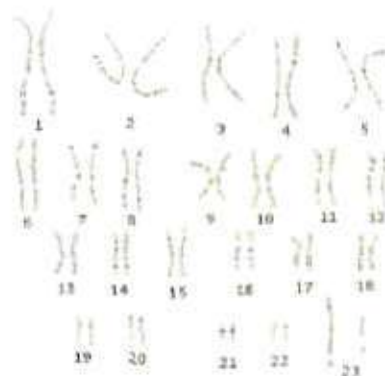
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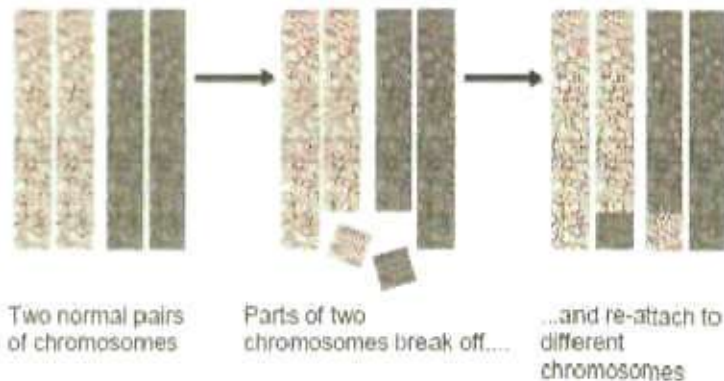
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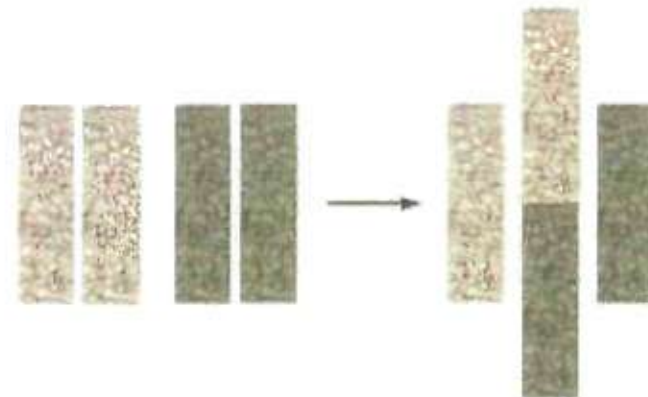
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Two pairs of normal chromosomes

Robertsonian translocation: a chromosome from one pair has become attached to a chromosome from another pair

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If either parent carries a balanced translocation, it is possible that their child may inherit an unbalanced translocation in which there is an extra piece of one chromosome and/or a missing piece of another chromosome.

A person who has an unbalanced translocation may be born with learning disability, developmental delay and health problems. The seriousness of the disability depends on exactly which parts of which chromosomes are involved and how much missing or extra chromosome material there is. This is because some parts of the chromosome are more important than other parts.



If a parent has a balanced translocation will he or she always pass it on?

Not necessarily, there are several possibilities for each pregnancy.

- The child may inherit entirely normal chromosomes.
- The child may inherit the same balanced translocation as the parent and not have any problems.
- The child may inherit an unbalanced translocation, and may be born with some degree of developmental delay, learning disability or health problems.
- The pregnancy ends in miscarriage.

Therefore it is quite possible for a person who carries a balanced translocation to have healthy children, and many do. However, the risk that a carrier of a balanced translocation will have a child with some degree of disability is higher than average, although the severity of the disability depends on the amount of material affected by the translocation.

Frequently a child can be born with a translocation although both parents' chromosomes are normal. This is called a "de novo" (from Latin) or new rearrangement. In this case the parents are unlikely to have another child with a translocation.

Section 2: Living with your carrier status

The following information discusses the various experiences of people who have found out they are carriers. We have tried to highlight a range of issues and emotions that finding out you are a carrier can cause, however they may not all be relevant to you.

How might I react?

People react in all sorts of ways when they receive their test results. Many people say that they feel angry or worried when they first find out that they are a carrier. Some people report that they feel sad, surprised or shocked. All these reactions are normal. For most people these feelings subside after a few months.

"At the time it seemed of enormous importance that this translocation had been found. But then as time went on it seemed like just a quirk."

It is quite normal to feel somehow different about yourself when you find out you are a carrier. Some people say that it is strange to find out something new about yourself when you think you know everything. It can take time to adapt to this new information.

"For thirty-one years you think there is nothing wrong and then all of sudden you find out there is this genetic thing..."

Some people say that finding out they are a carrier makes them feel 'less healthy'. Other carriers say that they are concerned that, in the future, they will be more likely to develop health problems. These reactions are very normal but it is important to remember that your carrier status has no effect on your health. We are all carriers of a number of gene mutations.

Studies have shown that carriers cope better with their results if they recognise that being a carrier is something that you cannot change, and that the information can be used positively.

"I've just accepted it now. I've moved on from that. Life throws things at you and you just get on with it really. It's just another one of life's twists."

Future children

For some people, finding out they are a carrier is a source of worry. For other people, such as those who have had multiple miscarriages, it is a relief to know so that they can plan ahead. Knowing that there is a risk that your future children might be born with an unbalanced translocation means that you can be prepared and take the time to make important decisions. There are a number of options you may wish to consider.

It is possible to perform a test during pregnancy (prenatal test), such as an amniocentesis or CVS test, to see if the baby has an unbalanced translocation. If you think this might be an option for you, speak to your doctor about whether these tests are available for the condition you are concerned about. If possible do this before the pregnancy as the laboratory may have to make preparations that can take several months.

If you are considering prenatal testing you should think about what you would do if the baby was found to have an unbalanced translocation, and how you might feel about a termination of pregnancy.

"Terminating the pregnancy was an agonising decision for my husband and me, but it was easier knowing that other couples have made the same decision."



It may be possible to perform a technique called Preimplantation Genetic Diagnosis (PGD) as an alternative to testing the baby during pregnancy. This involves the couple undergoing medically assisted reproduction, after which the fertilised eggs are tested. Only those eggs without the unbalanced translocation are implanted into the woman's womb. This is a demanding process and is not suitable for everyone. For more information about PGD, and whether it is available to you, you should speak to your doctor.

Other options you might want to consider include adoption, conceiving using donor eggs or sperm, or the possibility of not having children.

"The good thing is when I went for the genetic counselling I found out there were choices. If you want to have children, there are ways you can go about it."

What if you already have children?

Some parents who already have a child with an unbalanced translocation, say that one of the emotions that they experience when they find out they are a carrier is feeling 'guilty' for having 'passed on' the child's condition. It is entirely natural to feel this way. If you are having these kinds of feelings you should talk them through with your genetic specialist. It is important to remember that genes and chromosomes are distributed by chance and having a translocation is not your fault. Over time it has been shown that these feelings usually lessen in intensity.

"I do feel sort of that I've let my family down or let my husband down, and obviously let my son down because obviously I gave him something that he didn't really need to deal with for the rest of his life."



If you find out that you are a carrier and you have children, even if they do not carry an unbalanced translocation, there is still a chance that they could be carriers. It is important that you discuss this with your children at an age which is suitable. For some parents this might be when they think the child is old enough to understand. For others it might be when their children begin to have serious relationships. The age at which a young person can have a carrier test varies, but the person having the test has to make their own decision about it.

"The two girls have been up to the genetics department and had it explained to them."

Relationship with partner

Knowing about your carrier status can have an effect on your relationship with your partner. For some people it can bring them closer together and they can help support one another. However, knowing your carrier status can also cause tension and strain within the relationship. Sometimes couples will have to discuss issues that are very difficult and upsetting.

"I feel like I can cope with a lot, but to have to pull in someone you really care about, that felt horrible. That felt worse than dealing with it myself."

Other family members

If you find out that you are a carrier, you may wish to discuss this with other family members. This gives other family members the opportunity to have a blood test to see if they are also carriers, if they wish. This information may also be useful in helping diagnose other family members. It might also be particularly important to family members who are likely to have children in the future. Discussing your carrier test results is your choice and your test results will never be shared with other family members without your permission.

"They now know that it's in the family and they can be tested if they want to, they have that choice. And it's useful for them when they want to have children."

"Once I had discovered that my mother was a carrier and had not had any problems having babies, I felt much more comfortable about it."

For some people, sharing information with other family members is a positive experience. It can bring families closer together, and family members can provide a good source of support. Others find it difficult because they are worried about causing anxiety in the family. In some families, people have lost touch with relatives and may feel it is difficult to contact them.

For grandparents, it can be particularly difficult. They may not want to accept that the translocation is something that could have been passed down from them. It is also not uncommon for grandparents to feel guilty because they feel it is their fault. These are reactions you should be aware of.

"My mum spoke to my nan and she said 'it's not from me full stop'. So she blocked it off, she said 'I didn't bring anything into the family.'"



Genetic specialists often have a lot of experience with families in these situations and may be able to offer you help in discussing the situation with other family members. It can also be helpful to have information provided by your genetic specialist to show to other family members to help explain what it means to carry a translocation, and that being a carrier is something that occurs by chance.

Other sources of support

Genetic specialists and other health professionals (such as counsellors and psychologists) are experienced in helping people talk through the emotions that relate to receiving carrier test results. They can be a good source of information and support.

"Seeing Emma [the genetic counsellor] was very reassuring because before then I hadn't really understood that there was this thing called a CVS or an amniocentesis. So it was just good to have a professional explain all the different options. It was very helpful."

Some people also find it helpful to contact a patient support group. Patient support groups can provide information about the practical and emotional aspects of being a carrier of a condition. Many have a website and helpline giving information and advice. They can often put people and families in touch with others who are in a similar situation. They may have a chat forum on which members can email other members.

"We've got quite a lot of information from the patient support group and so it's been helpful. Also, knowing that some nights they work late, if you want to ring them up and have a chat to them, they will chat to you over the phone, send things in the post, work with the schools and so on."

Further information

For further information, contact your local regional genetics service. Their contact details can be found at:

www.gig.org.uk/services.htm

Additional information can also be found at:

Contact a Family

UK charity for families with disabled children. Offers information on specific conditions and rare disorders.

Helpline 0800 808 3555

(Mon-Fri, 10am-4pm and Mon, 5.30pm-7.30pm)

Email: ihelpline@cafamily.org.uk

Web: www.cafamily.org.uk

Genetic Alliance UK

Provides information about specific genetic conditions and contact details of support organisations.

Tel: 020 7704 3141

E mail: mail@geneticalliance.org.uk

Web: www.geneticalliance.org.uk

Unique- The Rare Chromosome Disorder Support Group

Telephone: 01883 330766

Email: info@rarechromo.org

Web: www.rarechromo.org



We are extremely grateful to all the people that allowed us to interview them during the making of this leaflet.

This information was developed by Genetic Alliance UK, a national alliance of patient organisations which supports children, families and individuals affected by genetic disorders.

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Living Without A Diagnosis Information For Parents



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Living Without A Diagnosis Information For Parents

What is this booklet about?

There are some children who have learning difficulties or health problems, and no-one can give a diagnosis to explain the cause of these problems. If you are the parent of a child without a diagnosis, this information booklet has been written for you. It is designed to help answer some of the questions you may have. We also give you some ideas of helpful things you might do as a parent and talk about some of the experiences other parents have had. Unfortunately, it won't be able to give you a diagnosis.

"We have got no diagnosis, she is just Lisa, she is just unique. But to be honest with you, we get so much enjoyment out of her it totally outweighs any problems."

How common is it not to have a diagnosis?

Even though most people do not realise it, having an 'undiagnosed condition' is actually quite common. For example, it is thought that about half (50%) of children with learning difficulties have no definite diagnosis. Even without a diagnosis, children are still entitled to receive healthcare, education, benefits and services that are appropriate to their particular needs. Not having a diagnosis won't stop a child from reaching their full potential.

Why are some conditions difficult to diagnose?

There are a number of reasons why making a diagnosis is not easy:

- Sometimes children have a number of different problems that do not all fit into one specific condition,
- Many conditions have similar features and health problems, which makes it difficult to be accurate about a diagnosis

- Certain conditions are so rare that it can be difficult for doctors to identify the condition.
- The same condition may affect children in different ways, which can add to the problem of making a diagnosis.
- Some features of the condition may not appear until later on in the child's life. This may cause a delay in getting a diagnosis, or in some cases a change of diagnosis.

(taken from Contact a Family, *About Diagnosis*)

Doctors are now able to diagnose more conditions than ever before due to advances in our knowledge, and improvements in testing techniques and medical equipment. Some of these conditions will be genetic, that means they are caused by changes in our genetic material. Others will be the result of problems at birth or infection during pregnancy or in the first few years of life. However, because of improvements in science, there may be an expectation that doctors can always find a diagnosis. If they can't, it can be extremely disappointing and confusing for parents.

It is worth remembering though that, even if you do not have a diagnosis now, advances in medical knowledge and testing techniques mean that you may be able to get one in the future.



Living without a diagnosis: the experience of other parents

"If you have a diagnosis you're on some sort of track. Without a diagnosis it feels like you're lost in a swamp."

To help us write this booklet, we spoke to a number of parents of children who have no diagnosis. Hearing other people's stories can be interesting and helpful. They also show you that you are not alone! We have included many of their experiences, some of which may be of interest to you. Parents also gave some suggestions of useful things you can do as a parent, and these have also been included.

The emotional impact

Finding out a child was not developing as expected was a very worrying and anxious time for some parents. Parents described feeling alone, angry and sad, particularly because their child was not the 'problem-free' child they had hoped for.

Waiting for the results of tests was also a stressful time, and it was disappointing and frustrating when the results did not give any definite answers. Some women said that when they first found out there was a problem they were worried that it was somehow their fault, or something they had caused whilst they were pregnant. However, the reality is that these things are rarely anyone's fault. In most cases these things just happen, and there is no way that the parents could have changed the outcome.

Some parents said that not having a diagnosis made them feel out of control of the situation. They were unsure whether they would ever get a diagnosis and they didn't know what the future would hold for either them or their child. Without a 'name', they were unable to search for information. This was very frustrating.

"I don't know what is going to happen to him in the future and that is the worst thing I think, the 'not knowing'."

Parents often used the term 'emotional rollercoaster' to describe the day-to-day experience of looking after a child with a disability. There were difficulties and frustrations, but there was also an overwhelming sense of love and admiration that parents had for their child.

"She's so lovable. She makes me smile, she makes me glow. I just love her to bits."

Advice from parents

- Try to enjoy your child. It is easy to focus on all their problems and forget to enjoy watching them grow up.
- Once you accept your child for who they are, your life becomes easier.
- Don't compare your child to other children of the same age, or how your other children were at that age, as this can be upsetting. Every child is different.
- Try not to focus on the milestones your child has not reached. Focus on those that they have.
- Try not to worry too much about the future. Try to focus instead on dealing with each day at a time.
- Raising a child with a disability can be hard work, both physically and emotionally. Don't be afraid to ask for help, and take help when it is offered.
- Having someone you can really talk to and who is not emotionally involved (such as a counsellor or health visitor) can be a great help, even if they don't have all the answers.
- For peace of mind, try to be as organised as you can. You might want to put certain procedures in place, for example organising your will.

- It's easy to forget to look after yourself when you are looking after a child with a disability. Take time out every once in a while; go out with friends; look after your health.

"People kept saying to me 'just enjoy him because they grow up so quickly', and I do feel that we didn't enjoy him as much because of all the anxieties that we had about what was going to happen. If it was to happen over again I'd definitely say try and put that to the back of your mind and just enjoy who he is."

"As much as you think you are coping, take a step back and say, well, perhaps I need help with this or that. Don't be afraid to ask for help. It doesn't make you any less of a parent."

How important is a diagnosis?

Whilst all the parents that we spoke to said that they would like a diagnosis, over time parents had found that getting a 'name' was not as important as it had been at first. For most parents, the priority was ensuring that the day-to-day issues related to their child's condition were met. Many said that, even with a diagnosis, they would still have to explain what the diagnosis meant, particularly to people who did not work in genetics or medicine. For many parents, getting a diagnosis was just something that 'would be nice to know'.

"Even if we did have a diagnosis, I'm sure it would be some really long word and people would look at me and say, 'what's that then?' and then I'd have to explain it anyway."

"Even if we had a diagnosis, he's still Charlie to me. It's not going to change him, not going to change him in the slightest."

At Genetic Alliance UK, (a patient organisation supporting individuals and families affected by genetic disorders), we speak to patients and families every day who are living with both diagnosed and undiagnosed conditions. We find that they all

face similar challenges accessing services and raising a child with a disability. Sometimes, even with a diagnosis, it can still be difficult to know what the future holds.

"He's inspiring sometimes as to how loving and gorgeous he is to other children his own age. He's so affectionate. There are just things to do with his syndrome that make him special. He's just a gorgeous little boy."

Doctors and specialists

Going to lots of hospital appointments and seeing different specialists was a frustrating and time-consuming experience for some parents. Often, a number of different specialists were involved in their child's care, and children were often referred from one specialist to another as the search for a diagnosis continued.

One issue that parents found particularly frustrating was the frequency with which they have to repeat their child's 'life story', including a history of all the symptoms, tests and the specialists they had seen. It can be even more frustrating when you don't have a diagnosis because you do not have a 'label' that summarises all your child's problems. Whilst repeating this information every time you see a new specialist is understandably frustrating, in most cases it is necessary because there will be very specific things the specialist is interested in finding out that relate to his or her particular speciality.

"If you are able to go in and say, 'He's got so and so disease' then they go 'Okay that is fine', but you can't. You have to go back from birth when you were first told, and explain the whole thing."



Parents also found that sometimes communication between different specialists was not as up to date as it should be. Many therefore had taken an active role in keeping their GP and other specialists such as their paediatrician and geneticist up to date with any tests done, results given or specialists seen.

"I carry all the letters I've received from the paediatric consultant and geneticist to every appointment, and sometimes those letters haven't yet been put on file. Sometimes I have to pull out the letter and show it to them."

Some parents found that the lack of information, particularly about their child's future development, was very frustrating. However, it's worth remembering that it is also frustrating for your specialist. They too will want to find a diagnosis for your child's condition. Yet even without a diagnosis, specialists will still be able to offer treatment and support that is tailored to your child's needs.

Remember that you are ultimately the expert in your child's condition. If there are developments in your child's condition, or if new milestones are reached, you should keep a record of these to discuss with the specialist at your next appointment.

Advice from parents

- Make sure you receive copies of all the letters sent by different specialists, to ensure you are kept up to date.
- Take responsibility for ensuring that all the different specialists, including your GP and healthcare worker, are kept up to date of any appointments and developments.
- Keep a paper trail. Keep copies of all letters, appointments, test results, etc. in a folder.
- Keep a record of your child's progress. Keep a diary; take

photos and videos if possible. As well as being a useful record to show to doctors, it can also be comforting to look back and see the progress your child has made.

- Create a 'passport' for your child. This can be a series of flashcards that explain things such as what your child can and cannot do, what they like/dislike, any health problems they have etc. These can be taken to appointments and are a quick way of passing on information about your child.
- If you feel that you need to see a specialist, or if you think you may have somehow got 'lost in the system', don't be afraid to phone and find out. Ask to speak to the doctor's secretary to find out what is happening with your child's appointment.
- Whenever you think of a question that you want to ask the specialist, write it down immediately so you don't forget it. A number of parents, for example, kept a note-pad handy on their fridge, to jot down their questions.
- Don't be afraid to ask your specialist questions that are concerning you, however silly or insignificant you think they may seem.
- Take someone else with you to your appointments as a 'second pair of ears'. This person can take notes of the discussion you have with the specialist. These can be a great help later on when you try to remember what was said.
- If possible, try to stick with the same health professionals so you don't have to keep retelling your child's life story.

Other services and professionals

Many parents found that their health visitor was a good source of information and support. A health visitor is a qualified and registered nurse or midwife with a specialist qualification in Community Health who visits families with children up to the age of five, at their home. Parents found that their health visitor:



- Provided useful information about their child's growth and development.
- Organised referrals, if necessary, to specialists such as community paediatricians and speech and language therapists.
- Provided information about local services such as special needs nurseries, and had knowledge about issues such as disability benefit.
- Provided emotional support, particularly to parents who were concerned or anxious about their child, or were finding it difficult to cope.

"In my case the health visitor has been a very good 'rock' in this whole process. I think also that is part of her job, to support us as a family and me as a mother."

There were a number of other professionals that families had come into contact with, and found helpful. These included:

- The social worker – can provide an assessment of your child's needs, which can lead to support and services for you and your child, and advise you about financial benefits you may be entitled to.

- The **housing officer** – works for a housing association or local council. Your housing officer may be able to provide you with information about housing benefits and entitlements, including home adaptations and facilities.
- **Children's centre / nursery / play group staff** – helps with activities to encourage your child's development and has useful information about local schools in the area.
- The **key worker** – a health professional or social worker who supports families and co-ordinates services, often for children with complex needs. These services can be involved from birth.
- The **occupational therapist** – can advise on equipment for daily living and managing more easily within the home.
- The **speech and language therapist** – assesses and treats speech, language and communication problems in people to enable them to communicate to the best of their ability. They may also work with people who have eating and swallowing problems.
- The **clinical or educational psychologist** – can assess whether a child has learning difficulties.
- The **physiotherapist** – advises and helps with exercises to improve mobility and coordination.

Certain professionals, such as the speech and language therapist or physiotherapist, will offer you advice and put into place programmes that will help with your child's development. Attending appointments and seeing professionals is only one aspect of your child's care. As your child's main carer, it will be up to you to make sure you follow their advice on a day-to-day basis.

Some of the parents we spoke to found they had to be persistent

in order to access services or get appointments. Other parents said that it was a 'battle' just to find out about the help and services that were available. Some parents said that they had to learn to be 'pushy'. Unfortunately, this is often the reality of caring for a child with a disability, particularly when they have a number of health-related or developmental problems. However, the professional team around you (such as your health or social worker, paediatrician or geneticist) should help you to navigate your way through the system and ensure you receive an effective service.

Parent support groups and charities may also be able to provide you with information and support. In addition, all councils now have a Family Information Service that provides information, advice and guidance to parents and carers on childcare and early years services in the local area. Their contact details are listed in the Contacts section at the end of this leaflet.

Another issue parents frequently raised was the amount of paperwork that needed to be completed to access certain services or benefits, and the amount of time this took. Some parents said that their housing officer or social worker helped them to complete these forms. Others had found voluntary organisations (such as the Citizens Advice Bureau) who were willing to help with this work. If you are having trouble filling out these forms, try to find someone who can take the time to help you.

Advice from parents

- Don't be afraid to chase things up and be persistent.
- Be aware of all the services and benefits that are available to you. Ask the professional team around you to provide you with this information.
- Find out about the local services in your area such as

special needs nurseries, respite services or local parent support groups.

- Other organisations such as patient support groups, charities, the Citizens Advice Bureau (CAB) and other parents are also a good source of information.
- If you feel you can't cope with the number of appointments you have to attend, you can *always* speak to one of your specialists or your key worker about pushing back some of your appointment dates. It's important for you and your child not to feel overwhelmed.
- Not having a 'name' for your child's condition can make it difficult when filling out forms. You find you end up writing 'undiagnosed condition'. Having a letter from your consultant that explains what this means can be helpful.

"My health visitor came round with all the forms and she said, 'this is disability benefit, I'm sure you can claim this'. And she gave me these claim packs and I thought, 'well I'll just fill them out and see'. And then all of a sudden I got this letter through saying, 'you're entitled to full disability benefit', so that wasn't a problem. I mean I didn't even know about disability benefit until the health visitor brought the form round."

Education

Getting the right school for their child was one of the most important issues for parents. There were a number of things that parents had to think about when deciding which school was best for their child. These included the educational needs of the child, and whether mainstream schools in the area were able to cater for their child's educational or physical needs.



Some children were in local mainstream schools, some were in schools for children with special needs, and some were in a 'dual placement' system which meant that for some of the week the child was educated at a mainstream school and for some of the week in a school specifically for children with special needs.

A number of parents commented that choosing a school was an anxious time. For some parents, the reality that their child needed extra assistance in class, or was best placed in a school for children with special needs, was hard to come to terms with. However, many parents whose children had started school made positive comments about the attention and support their child received.

"When we went to the school we said, 'we have got no diagnosis, she is just Leah, she is just unique'. They said, 'fine, this is what we have, this is what we can do for her', so they were absolutely brilliant and they still are brilliant."

Your health or social worker should be able to offer you advice about the options in your area, or the staff at your child's pre-school or nursery. In addition, you may want to go and look at different local schools and speak to the staff to find out which would be most appropriate for your child's needs. Most schools nowadays have a special educational needs co-ordinator (SENCO) who should also be able to offer you advice.

Special educational needs assessments

A number of the parents we spoke to had gone through the process of applying for an assessment of special educational needs from their local education authority (LEA). This was because it was felt that the local nursery or mainstream school was unable to meet their child's educational needs.

An assessment is carried out by the LEA to find out about a child's difficulties and agree upon the extra help they need. Those children with the greatest learning difficulties receive a

statement of special educational needs (usually just called a Statement).

A Statement describes the special educational help a child must be given by law. Applying for an assessment can be a long and difficult process, but the professionals around you will be able to help you.

If you feel that your child's educational needs are not being met, the first thing to do is to speak to your child's teacher or special educational needs co-ordinator (SENCO). You may also find it helpful to talk to local and national voluntary organisations (see the Education contact details provided at the end of this booklet) or your local Parent Partnership Service.

Parent Partnership Services

Parent Partnership Services are local organisations that offer free information, advice and support to parents and carers of children and young people with special educational needs. They can also offer support in preparing for and attending meetings, help in filling in forms and writing letters, and provide support in resolving disagreements with your child's school and the local authority.

For further information about your local parent partnership service (National Parent Partnership Network), see the Education contact details provided at the end of this booklet.

Advice from parents

- Visit the nurseries and schools in your area and speak to the teachers, before making a decision about which is best for your child.
- Applying for a statutory assessment can be a long and complicated process. If you are going to apply, it's a good idea to know as much as possible about the process beforehand.



Family

Raising a child with a disability can cause a strain on relationships. Some of the parents we spoke to said they argued more as a result of the stress and worry of looking after a child with a disability.

"I think you have to stop and say, 'I need to take time out for myself and my relationship'. I think in that we've suffered because we didn't have time for each other."

Parents can have different ideas about what's best for their child. Often it is the mother who attends all of the appointments, and the father can feel uninvolved. Many couples emphasised the importance of being supportive of each other and communicating about issues such as what happened at an appointment, so that both parents feel involved in their child's care. It's also important to accept help (either from friends, family, or short break (respite) services) so that you can have time alone together, even though it can be quite daunting at first to let someone else look after your child.

"We felt really guilty sending the girls somewhere so we could have a break. But you need to accept all the help and support you can get in order to keep on top of things."

For single parents the experience can be very lonely. Many parents turned to family members and friends for support.

"I think my dad took on the dominant, father role. He used to come round a lot and help me cope."

Parents who had more than one child often described feeling guilty for being unable to provide the same amount of attention to all their children. They worried that siblings would feel they were not as loved as their brother or sister. Growing up with a brother or sister who has some kind of disability and who needs more attention, can be very hard for children. However, some parents were surprised at how affectionate and supportive siblings could be.

"She just likes to be involved, she wants to help him. She is perfectly aware of the fact his eyes don't work properly and that he needs extra help, and all she is concerned with is, you know, has he got his glasses, does he need his contact lenses, is he okay, making sure nobody is mean to him."

Advice from parents

- Explain the situation to siblings, taking into account their age and maturity level.
- Try to make siblings feel involved in caring for their disabled brother or sister.
- It is important to try to take time out once in a while to spend with other siblings.
- It's a good idea to find a term such as 'developmental delay', or 'SWAN child' (syndrome without a name) that describes your child's condition. This can be helpful when explaining your child's disability to other people (such as friends and other parents). Ask your healthcare specialists to help you come up with a term that best describes your child's disability.

"That's hard, shaming yourself, that is really hard. But I will say

always make time for your other children, because otherwise that can have a bad knock on effect."

Information and support

It's natural to want to find as much information as possible about your child's disability. Parents used a number of different methods to find information. Some looked on the internet, others used chat forums to speak to other parents, and some contacted patient support groups.

Even without a confirmed diagnosis, you may still be able to find a lot of useful information from patient support groups and other parents who have a child with similar symptoms, particularly about the day-to-day issues of caring for your child. There are a number of parent groups that can help put you in touch with other families. Their contact details are listed at the end of this booklet. There may also be other charities and support services in your area that can provide respite care (a short break from caring), help you fill out forms, and provide other forms of support and information for you and your child.

"If you go to these parent chat rooms, all the trivial things that are actually quite big things as a parent, you can resolve quite happily and quite easily because other people have been through it, or are in the same boat as you. It is nice to know that you are not the only one, it is not a stupid question and you don't actually have to go to the doctor to ask."

Whilst the internet can be a great source of advice and information, some parents found that it was easy to scare themselves by looking online. In addition, not all the 'medical' information found on the internet has been verified with medical professionals and may not be correct. This is something you should be aware of.



"There is a certain level of 'ignorance is bliss' I do regret trawling the internet to find out about the things that she had listed down as possible syndromes. It just meant eight to ten months of complete worry."

Advice from parents

- Be careful of the information you read on the internet. It can be very easy to scare yourself.
- Parent support groups and chat forums can be good sources of information, particularly about the 'day-to-day' issues of caring for your child, that your specialist may not be able to help you with.
- Find out about local services in your area. Your council's Family Information Service can be a valuable source of information about local services.
- Having a computer and being able to access the internet can be very helpful. Being able to do your shopping on-line, for example, can make life much easier.

Charities, support groups and other useful information services

General

Contact a Family

UK-wide charity that provides support, advice and information for families with disabled children, no matter what their condition or disability. They also put families in contact with other families who are affected by the same or similar disabilities or medical conditions.

Free helpline: 0808 808 3555

Email: helpline@cafamily.org.uk Web: www.cafamily.org.uk

Citizens Advice Bureau

Gives free, confidential information and advice to help people with their money, legal, consumer and other problems across the UK. Web: www.adviceguide.org.uk

Carers UK

Offers information and advice to carers of children and adults with disabilities.

Tel: 020 7378 4999

Email: info@carersuk.org Web: www.carersuk.org

Directgov

Government website providing easy access to government information and services. Has information about education, carer advice, benefits and financial support. Also has a link to local councils throughout the UK. Web: www.direct.gov.uk

Every Child Matters

Programme set up by the Department for Children, School and Families which has a useful website providing information on early years and childcare, health and education for children. Particularly useful information can be found on the following pages. Web: www.dcsf.gov.uk

www.dcsf.gov.uk/everychildmatters/earlyyears/

www.dcsf.gov.uk/everychildmatters/healthandwellbeing/ahdc/AHDC/

www.dcsf.gov.uk/everychildmatters/healthandwellbeing/ahdc/earlysupport/home/

www.dcsf.gov.uk/everychildmatters/healthandwellbeing/ahdc/earlysupport/resources/esresources/

Family Information Service

Provides information, advice and guidance to parents and carers on childcare and early years services in the local area.

Tel: 0207 953 4085 Web: www.nafis.co.uk

NHS Direct

Provides health information and advice.
Helpline: 0845 4647
Web: www.nhsdirect.nhs.uk

Support groups**Ableize**

UK and Ireland information resource run by people with disabilities. Offers information on mobility, and provides information, support and advice.
Web: www.ableize.com

Afasic – speech, language & communication

UK charity helping children affected by speech, language and communication impairments, and their families.
Helpline: 0845 355 5577 Web: www.afasic.org.uk

Changing Faces

Charity supporting people who have disfigurement of the face or body from any cause.
Tel: 0845 4500 275 Web: www.changingfaces.org.uk

Genetic Alliance UK

Provides information about specific genetic conditions and contact details of support organisations across the UK.
Tel: 020 7704 3141
Email: mail@geneticalliance.org.uk
Web: www.geneticalliance.org.uk

Kith and Kids

Kith and Kids' overall aim is to empower families living with disability to overcome their social isolation and access the services they need.
Tel: 020 8801 7432
Email: projects@kithandkids.org.uk
Web: www.kithandkids.org.uk

Mencap

UK charity supporting people with learning disabilities.
Helpline: 0808 808 1111
Email: help@mencap.org.uk Web: www.mencap.org.uk

National Autistic Society

Information and support for people with autism and their families.
Helpline: 0845 070 4004 Web: www.nas.org.uk

SIBS

UK charity for people who grow up with a disabled brother or sister.
Tel: 01535 645453 Web: www.sibs.org.uk

SWAN (Syndromes Without A Name) USA

American organisation offering support, advice and information to families with children who have undiagnosed medical conditions. They also enable families to contact other families through an e-support group, MySpace, Facebook and Twitter page.
Tel: (001) 269 962 2090
Email: swanusa@undiagnosed-usa.org
Web: www.undiagnosed-usa.org

United Response

Supports people with learning disabilities, mental health needs and physical disabilities to live in the community, across England and in Wales.
Tel: 020 8246 5200
Email: info@unitedresponse.org.uk
Web: www.unitedresponse.org.uk

Unique

The Rare Chromosome Disorder Support Group.
Tel: 01883 330 766
Email: info@rarechromo.org
Web: www.rarechromo.org

Education**Advisory Centre for Education (ACE)**

A national charity that provides advice and information to parents and carers on a wide range of school-based issues including special education needs.

General helpline: 0808 800 5793

Exclusion helpline: 0808 800 0327

Web: www.ace-ed.org.uk

Independent Panel of Special Education Advice (IPSEA)

Provides free education advice and representation for parents of children with special educational needs and disabilities in England and Wales.

Tel: 0800 0184016 Web: www.ipsea.org.uk

National Parent Partnership Network (NPPN)

Your local authority will have a parent partnership service. They offer free, confidential and impartial information, advice and support to parents and carers of children and young people with special educational needs (SEN). They can give you advice about statutory assessments, statements, SEN Code of Practice, your rights and responsibilities and who you can talk to about any concerns you may have about SEN.

Tel: 020 7843 6058

Email: nppn@ncb.org.uk Web: www.parentpartnership.org.uk

Portage

A home-visiting educational service for pre-school children with additional support needs and their families.

Tel: 0121 244 1807

Email: info@portage.ork.uk Web: www.portage.org.uk

Special Educational Needs and Disability Tribunal (SEND)

Parents whose children have special educational needs can appeal against decisions made by local education authorities in England about their children's education.

Tel: 0870 241 2555 Web: www.sendist.gov.uk

Special Education Needs Tribunal for Wales

The equivalent body in Wales. Tel: 01597 829 800

Web: www.wales.gov.uk/sentws/sub/home/?lang=en

Additional Support Needs Tribunal for Scotland

The equivalent body in Scotland. Tel: 0845 120 2906

Web: www.asntscotland.gov.uk/asnts/141.html

Education Support for Northern Ireland

The equivalent body in Northern Ireland.

Tel: 028 3751 2383

Web: www.education-support.org.uk/parents/special-education/sendist

The Children's Legal Centre

Provides legal advice, information and representation for children and young people who are involved in a dispute with a school or local authority.

Free Child Law helpline: 08088 020 008

National Education helpline: 0845 345 4345

Email: clc@essex.ac.uk Web: www.childrenslegalcentre.com

Finance, benefits, equipment and holidays**3H Fund**

A charity that makes it possible for disabled people and their carer or family to have memorable and fulfilling holidays.

Tel: 01892 860 219

Email: info@3hfund.org.uk Web: www.3hfund.org.uk

Caudwell Children

Provides direct donations for treatment, therapy and specialised equipment.

Tel: 0845 300 1348

Email: charity@caudwellchildren.com

Web: www.caudwellchildren.com

Children Today

Raise funds to provide special equipment for children and young

people throughout the UK.

Tel: 01244 335622

Email: info@childrentoday.org.uk

Web: www.children-today.org.uk

Dream Makers

A national children's charity sending sick and disabled children on dream holidays as well as providing equipment.

Tel: 0121 711 8982

Web: www.dreammakerschildrenscharity.com

Disability Alliance UK

Provides information on social security benefits and tax credits to disabled people, their families and carers.

Tel: 020 7247 8776 (not an advice line)

Email: office.da@dial.pipex.com Web: www.disabilityalliance.org

Disability Benefits Helpline

Offers advice on Disability Living Allowance and Attendance Allowance.

Helpline: 08457 123 456

E mail: DCPU.Customer-Services@dwp.gsi.gov.uk

Web: www.dwp.gov.uk/about-dwp/customer-delivery/disability-and-carers-service/

Family Fund

Helps families with severely disabled children by providing grants for things that make life easier and more enjoyable for the disabled child and their family, such as washing machines, computers and holidays.

Tel: 0845 130 4542

Email: info@familyfund.org.uk Web: www.familyfund.org.uk

Newlife Foundation for Disabled Children

Charity providing equipment to help individual children. Also provides a nurse service that offers support and advice.

Tel: 01543 468 999 Nurse service: 0800 902 0095

Email: info@newlifecharity.co.uk

Web: www.newlifecharity.co.uk

Pearson's Holiday Fund

Provides grants that financially assist disadvantaged children and young people (between 4 and 16 inclusive) in the UK to have holidays or take part in respite activities.

Email: general.secretary@pearsonsholidayfund.org

Web: www.pearsonsholidayfund.org

The Aidis Trust

Helps people with disabilities make the best use of information and communication technology by giving information, help and support on all aspects of disability computing.

Tel: 0207 426 2130

Email: info@aidis.org Web: www.aidis.org

Whizz Kidz

Charity giving disabled children the chance to lead more independent lives. Provides mobility equipment, advice and training.

Tel: 020 7233 6600

Web: www.whizz-kidz.org.uk

We are extremely grateful to all the parents who allowed us to interview them during the making of this leaflet.

This information was developed by Genetic Alliance UK, a national alliance of patient organisations which supports children, families and individuals affected by genetic disorders.

Scientific information had been checked by Dr Simon Holden, Consultant in Clinical Genetics, Guy's Hospital, London.

Publication date: January 2010

Review date: January 2012

Illustrations by Rebecca J Kent

www.rebeccajkent.com

rebecca@rebeccajkent.com





National Research Ethics Service

Camden & Islington Community Research Ethics Committee

REC Offices
South House, Royal Free Hospital
Pond Street, London
NW3 2QG

Telephone: 020 7764 0500 extn 30906
Facsimile: 020 7764 1004

28 March 2010

Ms Celine Lewis
Project officer at GIG
Genetic Interest Group
Unit 4D Leroy House
436 Essex Rd
London, N1 3QP

Dear Ms Lewis

Study Title: A pilot study, in preparation for a randomised controlled trial, exploring the effectiveness of two information resources to provide psychosocial information about genetic testing to patients

REC reference number: 10/H0722/25

Protocol number: Version 1

The Research Ethics Committee reviewed the above application at the meeting held on 22 March 2010. Thank you for attending to discuss the study.

Ethical opinion

The members of the Committee present gave a favourable ethical opinion of the above research on the basis described in the application form, protocol and supporting documentation, subject to the conditions specified below.

Ethical review of research sites

The favourable opinion applies to all NHS sites taking part in the study subject to management permission being obtained from the NHS/HSC R&D office prior to the start of the study (see "Conditions of the favourable opinion" below).

Conditions of the favourable opinion

The favourable opinion is subject to the following conditions being met prior to the start of the study.

Management permission or approval must be obtained from each host organisation prior to the start of the study at the site concerned.

For NHS research sites only, management permission for research ("R&D approval") should be obtained from the relevant care organisation(s) in accordance with NHS research governance arrangements. Guidance on applying for NHS permission for research is available in the Integrated Research Application System or at <http://www.r4forum.nhs.uk>

This Research Ethics Committee is an advisory committee to London Strategic Health Authority
The National Research Ethics Service (NRES) represents the NRES Directorate within
the National Patient Safety Agency and Research Ethics Committees in England

Where the only involvement of the NHS organisation is as a Participant Identification Centre, management permission for research is not required but the R&D office should be notified of the study. Guidance should be sought from the R&D office where necessary.

Sponsors are not required to notify the Committee of approvals from host organisations.

It is responsibility of the sponsor to ensure that all the conditions are complied with before the start of the study or its initiation at a particular site (as applicable).

Approved documents

The documents reviewed and approved at the meeting were:

Document	Version	Date
REC application		16 February 2010
Protocol	Version 1	16 February 2010
Investigator CV	CI's CV - Celine Lewis	
Investigator CV	Supervisor's CV - Heather Skilton	16 February 2010
Letter from Sponsor	Prof Richard Stephenson, Dean of Faculty of Health, University of Plymouth	16 February 2010
Letter of invitation to participant	Version 1, from Clinical Genetics Service	16 February 2010
Evidence of insurance or indemnity	Zenith Municipal for University of Plymouth	05 August 2009
Letter of invitation to participant	Non-diagnosis Intervention, Version 1, from CI with reply slip	16 February 2010
Letter of invitation to participant	Non-diagnosis Controls, Version 1, from CI with reply slip	16 February 2010
Letter of invitation to participant	Carrier Testing, Version 1, from CI with reply slip	16 February 2010
Questionnaire: Carrier Testing	Version 1	16 February 2010
Questionnaire: Non-diagnosis Controls	Version 1	16 February 2010
Questionnaire: Non-diagnosis Intervention	Version 1	16 February 2010
Participant Information Sheet: Balanced Translocation: Intervention Group	Version 2	
Participant Information Sheet: Balanced Translocation: Control Group	Version 2	
Participant Information Sheet: X-Linked Condition: Intervention Group	Version 2	
Participant Information Sheet: X-Linked Condition: Control Group	Version 2	
Participant Information Sheet: Recessive Condition: Control Group	Version 2	
Participant Information Sheet: Recessive Condition: Intervention Group	Version 2	

Membership of the Committee

The members of the Ethics Committee who were present at the meeting are listed on the attached sheet.

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

After ethical review

Now that you have completed the application process please visit the National Research Ethics Service website > After Review

You are invited to give your view of the service that you have received from the National Research Ethics Service and the application procedure. If you wish to make your views known please use the feedback form available on the website.

The attached document "After ethical review – guidance for researchers" gives detailed guidance on reporting requirements for studies with a favourable opinion, including:

- Notifying substantial amendments
- Adding new sites and investigators
- Progress and safety reports
- Notifying the end of the study

The NRES website also provides guidance on these topics, which is updated in the light of changes in reporting requirements or procedures.

We would also like to inform you that we consult regularly with stakeholders to improve our service. If you would like to join our Reference Group please email referencegroup@nres.npsa.nhs.uk.

10/H0722/25

Please quote this number on all correspondence

With the Committee's best wishes for the success of this project

Yours sincerely


Ms Stephanie Ellis
Chair

Email: katherine.cuseley@royalfree.nhs.uk

Enclosures List of names and professions of members who were present at the meeting and those who submitted written comments

"After ethical review – guidance for researchers"

Copy to: Sponsor's contact - Dr Richard Stephenson, University of Plymouth

R&D contact - Mrs Karen Ignation, Guy's & St. Thomas' NHS Foundation Trust

Educational Supervisor – Prof. Heather Skirton, University of Plymouth

An advisory committee to London Strategic Health Authority

Camden & Islington Community Research Ethics Committee

Attendance at Committee meeting on 22 March 2010

Committee Members:

Name	Profession	Present	Notes
Dr Adedotun Adenuga	Staff Doctor	No	
Professor David Caplin	Emeritus Professor of Physics	Yes	
Ms Heidi Chandler	PA/Administrator	No	
Dr Claudia Cooper	Senior Lecturer in Old Age Psychiatry	No	
Ms Stephanie Ellis	Former Civil Servant	Yes	
Ms Victoria Fox	Lawyer	No	
Dr Angela Hassiotis	Senior Lecturer in Learning Disabilities	Yes	
Mr Matthew Lewin	Journalist and Author	No	
Dr Roshan McClenahan	Retired Consultant Speech & Language Therapist	Yes	
Ms Peggy Papada	Clinical Research Officer	Yes	
Dr Frederic Shaw	Sessional GP/GP Appraiser	No	
Dr Charlotte Warren-Gash	SpR Public Health/Academic Clinical Fellow	Yes	
Ms Eleni Yerolaki	Specialist Counsellor	Yes	
Ms Biddy Youell	Head of Child Psychotherapy	No	

Also in attendance:

Name	Position (or reason for attending)
Miss Katherine Ouseley	REC Coordinator

Written comments received from:

Name	Position
Ms Victoria Fox	Lawyer
Ms Biddy Youell	Head of Child Psychotherapy



Would you like to look at a new booklet for people who have recently found out they are a carrier of a genetic condition?

My name is Celine Lewis and I work for a charity called Genetic Alliance UK where we provide information to individuals and families affected by genetic conditions. I am also doing a PhD at the University of Plymouth. As part of my PhD I have developed a new information booklet for people who have recently found out they are a carrier of a genetic condition. I hope that the booklet provides clear and supportive information about being a carrier but this needs to be tested to see if people really do find it useful. This is where I am hoping you may be able to help me.

What will happen if you decide to help? If you agree to participate in the study, I will send you a copy of my new booklet in the post. One week later I will send you a questionnaire (either by post or e mail, whichever you prefer). The questionnaire will ask questions about what you thought of the booklet, and also about how you feel about being a carrier. The questionnaire should take no more than 15 minutes to complete. You will then be able to return the questionnaire to me either in the freepost envelope I will have sent you, or by emailing it back to me.

If you agree to participate in the study, your questionnaire responses will be treated as confidential. I am the only person who will have access to your responses. I will keep your personal details secure on a password controlled computer and not pass them on to anyone else. Even if you tell me that you would like to help, you can change your mind and withdraw from the study at any time, without giving a reason. Your health care will not be affected in any way, whether or not you decide to be involved. I would be very happy to answer any further questions you might have about the study before you decide about being involved.

How do you get involved?

If you are 18 years of age or over, have recently found out you are a carrier, and willing to help or want to ask a question, please contact me, in the next 2 weeks if possible, in one of the following ways:

Mail: Fill out the form on the next page of this information sheet and send it back to me in the freepost envelope

Telephone: (daytime) 020 7704 3141

Text: (anytime) 07974 867 967

Email: celine@gig.org.uk or celine.lewis@plymouth.ac.uk

Finallythank you for reading this.

Celine Lewis

Reply Slip – Carrier Testing study

Please post this back to me in the freepost envelope provided.

[] I am interested in participating in this research and am happy for you to send me the booklet and questionnaire

[] I am not interested in participating in this research. Please do not contact me again

Name.....

Address.....

.....

.....

.....

Email.....

The carrier testing booklet will be sent to you by post. However, I am happy to email you the questionnaire if you would prefer. **If you would prefer me to email you the questionnaire, please tick (✓) the box next to your email address.**

Please indicate which condition type you are a carrier of. This is so that I know which booklet to send you (if you are unsure please contact me).

[] **Recessive** (e.g. cystic fibrosis, sickle cell, thalassaemia, Tay Sachs, spinal muscular atrophy)

[] **X linked** (e.g. fragile X, Duchenne muscular dystrophy, haemophilia)

[] **Chromosome Translocation**



Would you like to be involved in a study in which you will receive a new booklet for parents of children without a diagnosis?

My name is Celine Lewis and I work for a charity called Genetic Alliance UK, where we provide information and support to individuals and families affected by genetic conditions. I am also doing a PhD at the University of Plymouth. As part of my PhD, I am developing a new information booklet for parents of children without a diagnosis. As part of this process, I have developed a short questionnaire. This is where I am hoping that you may be able to help me.

What will happen if you decide to help? If you agree to participate in the study, I will send you a questionnaire (either by post or e mail, whichever you prefer). The questionnaire will include questions about you how you feel as the parent of a child without a diagnosis. The questionnaire should take no more than 10 minutes to complete. You will then be able to return the questionnaire to me either in the freepost envelope I will have sent you, or by emailing it back to me. **I will then send you a copy of the booklet which will be yours to keep.**

If you agree to participate in the study, your questionnaire responses will be treated as confidential. I am the only person who will have access to your responses. I will keep your personal details secure on a password controlled computer and not pass them on to anyone else. Even if you tell me that you would like to help, you can change your mind and withdraw from the study at any time, without giving a reason. Your health care will not be affected in any way, whether or not you decide to be involved. I would be very happy to answer any further questions you might have about the study before you decide about being involved.

How do you get involved?

If you are 18 years of age or over, are the parent of a child without a diagnosis, and are willing to help or want to ask a question, please contact me, in the next 2 weeks if possible, in one of the following ways:

Mail: Fill out the form on the next page of this information sheet and send it back to me in the freepost envelope

Telephone: (daytime) 020 7704 3141

Text: (anytime) 07974 867 967

Email: celine@gig.org.uk or celine.lewis@plymouth.ac.uk

Finallythank you for reading this.

Celine Lewis

Please post this back to me in the freepost envelope provided.

[] I am interested in participating in this research and am happy for you to send me the questionnaire

[] I am not interested in participating in this research please do not contact me again

Name.....

Address.....

.....

.....

.....

Email.....

If you would prefer me to e mail you the questionnaire instead of sending it by post, please tick (✓) the box next to your e mail address.

Appendix 30



Would you like to be involved in a study looking at a new booklet for parents of children without a diagnosis?

My name is Celine Lewis and I work for a charity called Genetic Alliance UK, where we provide information and support to individuals and families affected by genetic conditions. I am also doing a PhD at the University of Plymouth. As part of my PhD, I have developed a new information booklet for parents of children without a diagnosis. The booklet has been developed with the help of parents who, like yourself, do not have a diagnosis for their child's condition. You will find a copy of this booklet enclosed. I hope that the booklet provides information and support but this needs to be tested to see if parents really do find it useful. This is where I am hoping that you may be able to help me.

What will happen if you decide to help? If you would like to participate in the study, I will send you a questionnaire (either by post or e mail, whichever you prefer). The questionnaire will include questions about what you thought of the booklet, and also questions about you how you feel as the parent of a child without a diagnosis. The questionnaire should take no more than 10 minutes to complete. You will then be able to return the questionnaire to me either in the freepost envelope I will have sent you, or by e mail.

If you agree to participate in the study, your questionnaire responses will be treated as confidential. I am the only person who will have access to your responses. I will keep your personal details secure on a password controlled computer and not pass them on to anyone else. Even if you tell me that you would like to help, you can change your mind and withdraw from the study at any time, without giving a reason. Your health care will not be affected in any way, whether or not you decide to be involved. I would be very happy to answer any further questions you might have about the study before you decide about being involved.

How do I get involved?

If you are 18 years of age or over, and you are the parent of a child who currently does not have a diagnosis, and are willing to help or want to ask a question, please contact me, in the next 2 weeks if possible, in one of the following ways:

Mail: Fill out the form on the next page of this information sheet and send it back to me in the freepost envelope

Telephone: (daytime) 020 7704 3141

Text: (anytime) 07974 867 967

Email: celine@gig.org.uk or celine.lewis@plymouth.ac.uk

If you would like to participate, please keep hold of the information booklet provided in this envelope, as well as the letter sent to you by the genetic clinic.

Finallythank you for reading this.

Celine Lewis

Reply Slip – Non-Diagnosis Study

Please post this back to me in the freepost envelope provided.

[] **I am interested** in participating in this research and am happy for you to send me the questionnaire

[] **I am not interested** in participating in this research please do not contact me again

Name.....

Address.....

.....

.....

.....

Email.....

If you would prefer me to e mail you the questionnaire instead of sending it by post, please tick (✓) the box next to your e mail address.

Research & Development
Guy's & St Thomas' Foundation NHS Trust
18th Tower Wing
Guy's Hospital
St Thomas Street
London SE1 9RT
Tel: 02071895731

Sally Watts
Clinical Genetics
F07 New Guy's House
Guy's Hospital
St Thomas Street
UK
SE1 9RT

30 April 2010

Dear Sally Watts,

Title: A pilot study, in preparation for a randomised controlled trial, exploring the effectiveness of two information resources to provide psychosocial information about genetic testing to patients.

In accordance with the Department of Health's Research Governance Framework for Health and Social Care, all research projects taking place within the Trust must receive a favourable opinion from an ethics committee and approval from the Department of Research and Development (R&D) prior to commencement.

- Ethics number: 10/H0722/25
- Sponsor: Faculty of Health, University of Plymouth
- Funder: No external funder
- End date: 30/12/2010
- Protocol: Version 1
- Site: GSTFT
- R&D approval Date: 30/04/2010

R&D have reviewed the documentation submitted for this project and I am pleased to inform you that we are approving the work to proceed within Guy's and St Thomas' NHS Foundation Trust and has been allocated the Trust R&D registration number **RJ110/N091** Please quote the R&D registration number in any communications with the R&D Department regarding your project.

Conditions of Approval:

- The principal investigator must notify R&D of the actual end date of the project.
- The Principal Investigator is responsible for ensuring that Data Protection procedures are observed throughout the course of the project.
- The project must follow the agreed protocol and be conducted in accordance with all Trust Policies and Procedures especially those relating to research and data management.
- R&D must be notified of any changes to the protocol prior to implementation.
- Please submit a copy of the progress report on the anniversary of the Ethics favourable opinion **28th March 2011**

If appropriate it is recommended that you register with the Current Controlled Trials website:
<http://www.isrctn.org/>

Please ensure that you are aware of your responsibilities in relation to The Data Protection Act 1998, NHS Confidentiality Code of Practice, NHS Caldicott Report and Caldicott Guardians, the Human Tissue Act 2004, Good Clinical Practice, the NHS Research Governance Framework for

Health and Social Care, Second Edition April 2005 and any further legislation released during the time of this study.

Members of the research team must have appropriate substantive or honorary contracts with the Trust prior to the study commencing. Any additional researchers who join the study at a later stage must also hold a suitable contract.

If the project is a clinical trial under the European Union Clinical Trials Directive the following must also be complied with:

1. The EU Directive on Clinical Trials (Directive 2001/20/EC) and UK's implementation of the Directive: The Medicines for Human Use (Clinical Trials) Regulations 2004;
2. The EU Directive on Principles and Guidelines for Good Clinical Practice (EU Commission Directive 2005/28/EC); and UK's implementation of the Directive: The Medicines for Human Use (Clinical Trials) Amendment Regulations 2006;

Amendments

Please ensure that you submit a copy of any amendments made to this study to the R&D Department.

Annual Report

It is obligatory that an annual report is submitted by the Chief Investigator to the research ethics committee, and we ask that a copy is sent to the R&D Department. The yearly period commences from the date of receiving a favourable opinion from the ethics committee.

Should you require any further information please do not hesitate to contact us.

In line with the Research Governance Framework, your project may be randomly selected for monitoring for compliance against the standards set out in the Framework. For information, the Trust's process for the monitoring of projects and the associated guidance is available from the Trust's intranet or on request from the R&D Department. You will be notified by the R&D Department if and when your project has been selected as part of the monitoring process. No action is needed until that time.

Many thanks for registering your research project

Yours Sincerely,



Rachel Fay

R&D Coordinator

cc. Celine Lewis
cc. Sponsor, University of Plymouth



Ms Sally Taffinder
Great Ormond Street Hospital
Clinical Genetics
Great Ormond Street
London
WC1N 3JH

27/07/2010

Dear Ms Sally Taffinder

TITLE: A pilot study in preparation for a randomised controlled trial, exploring the effectiveness of two information resources to provide psychosocial information about genetic testing to patients

R&D Number: 10CM27

REC reference number: 10/H0722/25

Sponsor: Plymouth University

This project has been granted Management Approval by the R&D Office.

Approval Conditions:

Your research study must adhere to the Department of Health's Research Governance Framework. For more information please see the attached leaflet, further information can be found on the Department of Health website at www.doh.gov.uk.

You must submit an ICH/GOSH annual report which will be sent to you by the R&D office when it is due.

The PI must inform the R&D office of any changes to the start and end dates of the project, or if there are any changes to the protocol, personnel or ethical status. At the end of the study the PI will be sent a final report form to complete and return to the R&D Office.

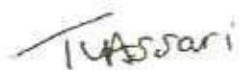
During your study you should remember to send in Annual Progress Reports to your Funder and the Research Ethics Committee that provided ethics approval for your study.

If you need statistical support you can contact the Statistical Support Service
<http://www.ich.ucl.ac.uk/ich/html/education/sss/intro.html>.

Attached is a checklist of documentation which you should keep on file. These documents should be

retained for 25 years. All patients entered into this study must be registered on the hospital PIMS system.

Yours Sincerely

A handwritten signature in black ink that reads "T. Assari". The signature is written in a cursive style with a long horizontal stroke at the beginning.

Dr Tracy Assari
Research Governance Co-ordinator

Enc.

Reference: 10/08/2010

24/08/2010

Ms Celine Lewis
Genetic Interest Group
Unit 4 D Leroy House
346 Essex Road
London
N1 3QP

Research & Development

Leeds Teaching Hospitals NHS Trust

34 Hyde Terrace
Leeds
LS2 9LN

Tel: 0113 392 2678
Fax: 0113 392 6897

r&d@leedsth.nhs.uk
www.leedsth.nhs.uk

Dear Ms Celine Lewis:

Re: LTHT R&D Approval of: A pilot study, in preparation for a randomised controlled trial, exploring the effectiveness of two information resources to provide psychosocial information about genetic testing to patients.
LTHT R&D Number: CG10/9426
REC: 10/H0722/25

I confirm that this study has R&D approval and the study may proceed at The Leeds Teaching Hospitals NHS Trust (LTHT). This organisational level approval is given based on the information provided in the documents listed below.

In undertaking this research you must comply with the requirements of the *Research Governance Framework for Health and Social Care* which is mandatory for all NHS employees. This document may be accessed on the R&D website http://www.leedsth.nhs.uk/sites/research_and_development/

R&D approval is given on the understanding that you comply with the requirements of the *Framework* as listed in the attached sheet "Conditions of Approval".

If you have any queries about this approval please do not hesitate to contact the R&D Department on telephone 0113 392 2678.

Indemnity Arrangements

The Leeds Teaching Hospitals NHS Trust participates in the NHS risk pooling scheme administered by the NHS Litigation Authority 'Clinical Negligence Scheme for NHS Trusts' for: (i) medical professional and/or medical malpractice liability; and

Chairman Mike Collier or Chief Executive Maggie Boyle

The Leeds Teaching Hospitals incorporating:

Chapel Allerton Hospital, Leeds Dental Institute, Beckett Hospital,
St James's University Hospital, The General Infirmary at Leeds, Wharfedale Hospital



(ii) general liability. NHS Indemnity for negligent harm is extended to researchers with an employment contract (substantive or honorary) with the Trust. The Trust only accepts liability for research activity that has been managerially approved by the R&D Department.

The Trust therefore accepts liability for the above research project and extends indemnity for negligent harm to cover you as principal investigator and the researchers listed on the Site Specific Information form. Should there be any changes to the research team please ensure that you inform the R&D Department and that s/he obtains an employment contract with the Trust if required.

Yours sincerely


Dr D R Norfolk
Associate Director of R&D

Approved documents

The documents reviewed and approved are listed as follows

Document	Version	Date of document
NHS R&D Form	3.0	16.02.2010
SSI Form	3.0	24.06.2010
Protocol	1.0	16.02.2010
REC Letter confirming favourable opinion		26.03.2010
Information Sheet (REC Approved) <i>Balanced Translocation Intervention Group</i>	2.0	<i>Not Dated</i>
Information Sheet (REC Approved) <i>Balanced Translocation Control Group</i>	2.0	<i>Not Dated</i>
Information Sheet (REC Approved) <i>X-Linked Condition Intervention Group</i>	2.0	<i>Not Dated</i>
Information Sheet (REC Approved) <i>X-Linked Condition Control Group</i>	2.0	<i>Not Dated</i>
Information Sheet (REC Approved) <i>Recessive Condition Intervention Group</i>	2.0	<i>Not Dated</i>
Information Sheet (REC Approved) <i>Recessive Condition Control Group</i>	2.0	<i>Not Dated</i>
Invitation to Participant (REC Approved) <i>Clinical Genetics Service</i>	1.0	16.02.2010
Invitation to Participant (REC Approved) <i>Non-diagnosis Intervention</i>	1.0	16.02.2010
Invitation to Participant (REC Approved) <i>Non-diagnosis Control</i>	1.0	16.02.2010
Questionnaire (REC Approved) <i>Carrier Testing</i>	1.0	16.02.2010
Questionnaire (REC Approved) <i>Non-Diagnosis Intervention</i>	1.0	16.02.2010
Questionnaire (REC Approved) <i>Non-diagnosis Control</i>	1.0	16.02.2010
Letter from Sponsor		16.02.2010
Evidence of Insurance		09.08.2009

Conditions of R&D Approval

- Approval from your Directorate must be obtained before starting the study
- Approval of the appropriate Research Ethics Committee, where necessary, must be obtained before starting the study. Any changes made to the project during ethical review must be reviewed and approved by the R&D Department to maintain R&D Approval status
- Arrangements must be made to ensure that all members of the research team, where applicable, have employment contracts with the Trust (either full or honorary).
- Agreements must be in place with appropriate support departments regarding the services required to undertake the project and arrangements must be in place to recompense them for the costs of their services.
- Arrangements must be in place for the management of financial and other resources provided for the study, including intellectual property arising from the work.
- Priority should be given at all times to the dignity, rights, safety and well being of participants in the study
- Healthcare staff should be suitably informed about the research their patients are taking part in and information specifically relevant to their care arising from the study should be communicated promptly.
- Each member of the research team must be qualified by education, training and experience to discharge his/her role in the study. Students and new researchers must have adequate supervision, support and training
- The research must follow the protocol approved by the relevant research ethics committee. Any proposed amendments to or deviations from the protocol must be submitted for approval to the Research Ethics Committee, the research sponsor, regulatory authority and any other appropriate body. The R&D Department should be informed where the amendment has resource implications within the Directorate and the Directorate research lead/clinical director notified.
- Adverse Events in clinical trials of investigational medicinal products must be reported in accordance with the Medicines for Human Use (Clinical Trials) Regulations 2004.
- Complete and return 6 monthly Study Status Reports to the R&D Department within 28 days of receipt as requested. (NB Failure to comply to such request with the requirement will lead to suspension of R&D Approval.)

- Procedures should be in place to ensure collection of high quality, accurate data and the integrity and confidentiality of data during processing and storage.
- Arrangements must be made for the appropriate archiving of data when the research has finished. Records must normally be kept for 15 years.
- All data and documentation associated with the study must be available for audit at the request of the appropriate auditing authority. Projects are randomly selected for audit by the R&D Department. You will be informed by letter if your study is selected.
- Findings from the study should be disseminated promptly and fed back as agreed to research participants.
- Findings from the study should be exposed to critical review through accepted scientific and professional channels.
- All members of the research team must ensure that the process of informed consent adheres to the standards GCP outlined in the UK Clinical Trials Regulations. Investigators are directed to the R&D website for further information and training availability.
- Where applicable, this managerial approval includes aspects of the study previously covered by the NRES Site Specific Assessment (SSA) process.

Commercially Sponsored Trials

If the study is commercially sponsored approval is given subject to provision of the following documents.

- Clinical Trials Agreement - agreed and signed off by the R&D Department (on behalf of the Leeds Teaching Hospitals NHS Trust) and the Sponsor. Investigators do not have the authority to sign contract on behalf of the Trust.
- Indemnity agreement, if not included in the Clinical Trials Agreement- (standard ABPI no fault arrangements apply) signed by the R&D Department and the Sponsor.

It is essential that all the responsibilities set out in the Research Governance Framework, including those outlined above are fulfilled. The Trust reserves the right to withdraw R&D approval where the above criteria are not being met. The Trust will not accept liability for any activity that has not been fully approved.

Appendix 34

Dear ...

We are writing to let you know about a study *Guy's /Great Ormond Street /Leeds genetic department* are involved in, which might be of interest to you. It concerns a new information booklet for carriers that we think you may find helpful. Please find more information about this study on the Information Sheet sent with this letter. If you would like any further information about the study, please contact the lead researcher, Celine Lewis. Her contact details can be found at the end of the information sheet.

Many thanks,

Name of referring genetic counsellor

Dear ...

We are writing to let you know about a study *Guy's / Great Ormond Street / Leeds genetic department* are involved in, which might be of interest to you. It concerns the development of a new information booklet for parents of children without a diagnosis. Please find more information about this study in the Information Pack sent with this letter. If you would like any further information about the study, please contact the lead researcher, Celine Lewis. Her contact details can be found at the end of the information sheet inside the pack.

Many thanks,

Name of referring genetic counsellor



CARRIER STUDY
QUESTIONNAIRE



Dear ...

Thank you for agreeing to help me with this study. About a week ago you should have received an information booklet in the post, giving you some additional information about being a carrier. I have enclosed another copy of the booklet in this envelope. Please make sure you have read this information before completing the questionnaire. The questions are designed to help me assess how useful the booklet is, and get a better understanding of what it is like for people when they find out they are a carrier of a particular genetic condition. There are no right or wrong answers and don't worry if some of the questions seem a bit unusual. They have been used in other studies and have been shown to be good indicators of how people deal with certain situations.

Once you have completed the questionnaire, please send it back to me in the enclosed freepost envelope or at the address below. I would be grateful if you could try and return the questionnaire to me within a week of receiving it.

Thank you very much for your help with this project.

With best wishes,

Celine Lewis

Celine Lewis
Carrier Testing Study
Genetic Alliance UK
Unit 4D Leroy House
436 Essex Road
London
N1 3QP
Tel: 0207 704 3141
Email: celine@geneticalliance.org.uk

SECTION 1: GENERAL QUESTIONS

Form ref:

The following questions are designed to give me some background information.

Please tick (✓) one box only for each question, or fill in the blanks.

1. Your sex:

- Male
 Female

2. Your age: _____ years

3. Your ethnic background:

- White
 African or Caribbean
 Asian
 Mixed
 Other
 Prefer not to say

4. Your marital status:

- Single
 Married
 Living with Partner
 Separated
 Divorced
 Widowed

5. Your level of Education:

- Secondary School (GCSE, O Level)
 Secondary School/College (A Level, HND, Diploma etc.)
 University First degree (BSc, BA)
 University Higher Degree (MA, MSc, PhD etc.)
 Prefer not to say

6. Condition for which you are a carrier:

- Chromosome translocation/deletion
 Cystic fibrosis (CF)
 Duchenne muscular dystrophy (DMD)
 Fragile X
 Haemophilia
 Sickle cell
 Spinal muscular atrophy (SMA)
 Tay Sachs
 Thalassaemia
 Other (please specify).....

7. Is your partner also a carrier of the same condition?

- Yes
 No
 Don't know
 No partner

8. Do you have any children?

- Yes
 No

9. Do you or have you had any children affected by the condition?

- Yes
 No

10. Are you planning on having any (more) children in the future?

- Yes
 Possibly
 No

SECTION 2: QUESTIONS ABOUT THE BOOKLET

The following questions are about the information booklet about carrier testing that you received in the post (a copy was also enclosed with this questionnaire). Please ensure you have read the booklet before answering the questions below.

Please tick (✓) one box only for each question.

1. Do you have a copy of the booklet?

- Yes
 No
 Don't know

2. Did you read it?

- Yes
 No

3. Did you find the information:

- Very useful
- Moderately useful
- Of no use

5. Was the information relevant?

- Definitely yes
- Possibly
- Not really
- Definitely no

7. Did you find the information too technical?

- Definitely yes
- Possibly
- Not really
- Definitely no

9. Did the information change your ideas about something?

- Definitely yes
- Possibly
- Not really
- Definitely no

4. Did it tell you anything new?

- Definitely yes
- Possibly
- Not really
- Definitely no

6. Did you feel overwhelmed with information?

- Definitely yes
- Possibly
- Not really
- Definitely no

8. Did you find the information too limited?

- Definitely yes
- Possibly
- Not really
- Definitely no

10. Have you shown the booklet to anyone else? (Tick all that apply)

- Partner
- Parent(s)
- Children
- Sibling(s)
- Other family members
- Friends
- Other (please specify).....
- No one

If you have any comments on things that you particularly **liked** about the booklet, write them here.

If you have any comments on things that you particularly **disliked** about the booklet, write them here.

If you have any general comments about the booklet, please write them here.

SECTION 3: QUESTIONS ABOUT YOUR UNDERSTANDING OF YOUR CARRIER STATUS

Thinking about your understanding of what it means to be a 'carrier', please answer the following questions.

Please tick (✓) one box only for each question.

1. I think I understand why I went to genetic counselling.

- Do not agree
 Somewhat agree
 Completely agree

2. I feel I know the meaning of the problem for me and my family's future.

- Do not agree
 Somewhat agree
 Completely agree

3. I think I know what caused the problem.

- Do not agree
 Somewhat agree
 Completely agree

4. I feel I have the tools to make decisions that will influence my future.

- Do not agree
 Somewhat agree
 Completely agree

5. I feel I can make a logical evaluation of the various options available to me in order to choose one of them.

- Do not agree
 Somewhat agree
 Completely agree

6. I feel I can make decisions that will change my family's future.

- Do not agree
 Somewhat agree
 Completely agree

7. I feel there are certain things I can do to prevent the problem from reoccurring.

- Do not agree
 Somewhat agree
 Completely agree

8. I feel I know what to do to ease the situation.

- Do not agree
 Somewhat agree
 Completely agree

9. I think I know what should be my next steps.

- Do not agree
 Somewhat agree
 Completely agree

SECTION 4: QUESTIONS ABOUT HOW YOU FEEL

Below are a list of comments made by people about stressful life events or situations. Thinking about your carrier test results, please indicate how frequently these statements were true for you in the last week. If any item is not relevant to you, please tick the 'not at all' option.

Please tick (✓) one box only for each question.

1. I thought about it when I didn't mean to.

- Not at all
 Rarely
 Sometimes
 Often

2. I avoided letting myself get upset when I thought about it or was reminded of it.

- Not at all
 Rarely
 Sometimes
 Often

3. I tried to remove it from my memory.

- Not at all
 Rarely
 Sometimes
 Often

4. I had trouble falling asleep or staying asleep, because of pictures or thoughts about it that came into my mind.

- Not at all
 Rarely
 Sometimes
 Often

5. I had waves of strong feelings about it.

- Not at all
- Rarely
- Sometimes
- Often

7. I stayed away from reminders of it.

- Not at all
- Rarely
- Sometimes
- Often

9. I tried not to talk about it.

- Not at all
- Rarely
- Sometimes
- Often

11. Other things kept making me think about it.

- Not at all
- Rarely
- Sometimes
- Often

13. I tried not to think about it.

- Not at all
- Rarely
- Sometimes
- Often

15. My feelings about it were kind of numb.

- Not at all
- Rarely
- Sometimes
- Often

6. I had dreams about it.

- Not at all
- Rarely
- Sometimes
- Often

8. I felt as if it hadn't happened or wasn't real.

- Not at all
- Rarely
- Sometimes
- Often

10. Pictures about it popped into my mind.

- Not at all
- Rarely
- Sometimes
- Often

12. I was aware that I still had a lot of feelings about it, but I didn't deal with them.

- Not at all
- Rarely
- Sometimes
- Often

14. Any reminder brought back feelings about it.

- Not at all
- Rarely
- Sometimes
- Often

SECTION 5: MORE QUESTIONS ABOUT HOW YOU FEEL

Please answer the following questions according to how you have felt since receiving your carrier test results. Don't take too long over your replies: your immediate reaction to each question will probably be more accurate than a long thought-out response.

Please tick (✓) one box only for each question

1. I feel tense or 'wound up':

- Most of the time
- A lot of the time
- From time to time, occasionally
- Not at all

3. I get a sort of frightened feeling as if something awful is about to happen:

- Very definitely and quite badly
- Yes, but not too badly
- A little, but it doesn't worry me
- Not at all

2. I still enjoy the things I used to enjoy:

- Definitely as much
- Not quite so much
- Only a little
- Hardly at all

4. I can laugh and see the funny side of things:

- As much as I always could
- Not quite so much now
- Definitely not so much now
- Not at all

5. Worrying thoughts go through my mind:

- A great deal of the time
- A lot of the time
- From time to time
- Only occasionally

7. I can sit at ease and feel relaxed:

- Definitely
- Usually
- Not often
- Not at all

9. I get a sort of frightened feeling like 'butterflies in the stomach':

- Not at all
- Occasionally
- Quite often
- Very often

11. I feel restless as if I have to be on the move:

- Very much indeed
- Quite a lot
- Not very much
- Not at all

13. I get sudden feelings of panic:

- Very often indeed
- Quite often
- Not very often
- Not at all

6. I feel cheerful:

- Not at all
- Not often
- Sometimes
- Most of the time

8. I feel as if I am slowed down:

- Nearly all the time
- Very often
- Sometimes
- Not at all

10. I have lost interest in my appearance:

- Definitely
- I don't take as much care as I should
- I may not take quite as much care
- I take just as much care as ever

12. I look forward with enjoyment to things:

- As much as ever I did
- Rather less than I used to
- Definitely less than I used to
- Hardly at all

14. I can enjoy a good book or radio or TV programme:

- Often
- Sometimes
- Not often
- Very seldom

Thank you very much for completing this questionnaire and for your help with this study. I hope that you have found it interesting.

Please return it in the freepost envelope or to the address on the cover.

With best wishes,

Celine Lewis



LIVING WITHOUT A DIAGNOSIS STUDY
QUESTIONNAIRE

Dear ...

Thank you for agreeing to help me with this study. These questions are designed to give me a better understanding of what it is like for parents who currently do not have a diagnosis for their child's condition. There are no right or wrong answers and don't worry if some of the questions seem a bit unusual. They have been used in other studies and have been shown to be good indicators of how people deal with certain situations. Only one parent (or guardian) need complete the questionnaire.

Once you have completed the questionnaire, please send it back to me in the enclosed freepost envelope or at the address below. I will then post you a copy of the information booklet. I would be grateful if you could try and return the questionnaire to me within a week of receiving it.

Thank you very much for your help with this project.

With best wishes,

Celine Lewis

Celine Lewis
Genetic Alliance UK
Unit 4D Leroy House
436 Essex Road
London
N1 3QP
Tel: 0207 704 3141
Email: celine@geneticalliance.org.uk

SECTION 1: GENERAL QUESTIONS

The following questions are designed to give me some background information.

Please tick (✓) one box only for each question, or fill in the blanks.

1. Your relation to the child:

- Mother
 Father
 Other (please specify) _____

2. Your age: _____ years

3. Your marital status:

- Single
 Married
 Living with Partner
 Separated
 Divorced
 Widowed

4. In which region of the UK do you live?

- North East England
 North West England
 Yorkshire and the Humber
 East of England
 East Midlands
 West Midlands
 Greater London
 South East England
 South West England
 Wales
 Scotland
 Northern Ireland

5. Your level of education:

- Secondary School (GCSE, O Level)
 Secondary School/College (A Level, HND, Diploma, NVQ etc)
 University First degree (BSc, BA)
 University Higher Degree (MA, MSc, PhD etc)
 Prefer not to answer

6. Total annual household income:

- less than £20,000
 £21,000 - £40,000
 £41,000 - £60,000
 £61,000 - £80,000
 £80,000 +
 Prefer not to answer

7. Your ethnic background:

- White
 African or Caribbean
 Asian
 Mixed
 Other
 Prefer not to answer

8. How many of your children are affected by an undiagnosed condition?

- One
 Two
 Three

9. How old is your child (children) without a diagnosis?

First child.....
Second child.....
Third child.....

10. At what age did their symptoms first become apparent?

First child.....
Second child.....
Third child.....

11. Do you have any children not affected by the condition?

- Yes
 No

12. Which of the following statements do you think best describes your child's (or children's) health or development problems?

	First child	Second child	Third child
Has no real effect on the child's daily life (the child can enjoy all the usual activities and needs no additional health care or support with schooling).	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Has some effect on the child's daily life (the child requires some support regarding their health care or learning).	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Has a major effect on the child's daily life (the child requires constant support with regard to activities, health and/or learning).	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

SECTION 2: QUESTIONS ABOUT HOW YOU FEEL

Below are a list of comments made by people about stressful life events or situations. Thinking about the fact that you do not have a diagnosis for your child's condition, please indicate how frequently these statements were true for you in the last week. If any item is not relevant to you, please tick the 'not at all' option.

Please tick (✓) one box only for each question.

- | | |
|--|---|
| <p>1. I thought about it when I didn't mean to.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> | <p>2. I avoided letting myself get upset when I thought about it or was reminded of it.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> |
| <p>3. I tried to remove it from my memory.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> | <p>4. I had trouble falling asleep or staying asleep, because of pictures or thoughts about it that came into my mind.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> |
| <p>5. I had waves of strong feelings about it.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> | <p>6. I had dreams about it.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> |
| <p>7. I stayed away from reminders of it.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> | <p>8. I felt as if it hadn't happened or wasn't real.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> |
| <p>9. I tried not to talk about it.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> | <p>10. Pictures about it popped into my mind.</p> <p><input type="checkbox"/> Not at all
 <input type="checkbox"/> Rarely
 <input type="checkbox"/> Sometimes
 <input type="checkbox"/> Often</p> |

11. Other things kept making me think about it.

- Not at all
- Rarely
- Sometimes
- Often

13. I tried not to think about it.

- Not at all
- Rarely
- Sometimes
- Often

15. My feelings about it were kind of numb.

- Not at all
- Rarely
- Sometimes
- Often

12. I was aware that I still had a lot of feelings about it, but I didn't deal with them.

- Not at all
- Rarely
- Sometimes
- Often

14. Any reminder brought back feelings about it.

- Not at all
- Rarely
- Sometimes
- Often

SECTION 3: MORE QUESTIONS ABOUT HOW YOU FEEL

Please answer the following questions according to how you feel about not having a diagnosis for your child. **Don't take too long over your replies: your immediate reaction to each question will probably be more accurate than a long thought-out response.**

Please tick (✓) one box only for each question.

Have you recently.....

1. Been able to concentrate on whatever you're doing?

- Better than usual
- Same as usual
- Less than usual
- Much less than usual

3. Felt that you are playing a useful part in things?

- More so than usual
- Same as usual
- Less useful than usual
- Much less useful

5. Felt constantly under strain?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

7. Been able to enjoy your normal day-to-day activities?

- More so than usual
- Same as usual
- Less so than usual
- Much less than usual

2. Lost much sleep over worry?

- Not at all
- No more than usual
- Rather more than usual
- Much less than usual

4. Felt capable of making decisions about things?

- More so than usual
- Same as usual
- Less so than usual
- Much less so than usual

6. Felt you couldn't overcome your difficulties?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

8. Been able to face up to your problems?

- More so than usual
- Same as usual
- Less so than usual
- Much less than usual

9. Been feeling unhappy and depressed?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

10. Been losing confidence in yourself?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

11. Been thinking of yourself as a worthless person?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

12. Been feeling reasonably happy, all things considered?

- More so than usual
- About same as usual
- Less so than usual
- Much less than usual

Thank you very much for completing this questionnaire and for your help with this study. I hope that you have found it interesting. Please return it in the freepost envelope or to the address on the cover.

Celine.



LIVING WITHOUT A DIAGNOSIS STUDY
QUESTIONNAIRE

Dear...

Thank you for agreeing to help me with this study. You should have received the information booklet 'Living without a Diagnosis; Information for Parents' in the post a week ago. Please make sure you have read the booklet before you answer this questionnaire. The questions are designed to help me assess how useful the information you received is, and to get a better understanding of what it is like for parents who currently do not have a diagnosis for their child's condition. There are no right or wrong answers and don't worry if some of the questions seem a bit unusual. They have been used in other studies and have been shown to be good indicators of how people deal with certain situations. Only one parent (or guardian) need complete the questionnaire.

Once you have completed the questionnaire, please send it back to me in the enclosed freepost envelope or at the address below. I would be grateful if you could try and return the questionnaire to me within a week of receiving it.

Thank you very much for your help with this project.

With best wishes,
Celine Lewis

Celine Lewis
Non Diagnosis Study
Genetic Alliance UK
Unit 4D Leroy House
436 Essex Road
London
N1 3QP

Tel: 0207 704 3141
Email: celine@geneticalliance.org.uk

SECTION 1: GENERAL QUESTIONS

The following questions are designed to give me some background information.

Please tick (✓) one box only for each question, or fill in the blanks.

1. Your relation to the child:

- Mother
 Father
 Other (please specify)

2. Your age: years

3. Your marital status:

- Single
 Married
 Living with Partner
 Separated
 Divorced
 Widowed

4. In which region of the UK do you live?

- North East England
 North West England
 Yorkshire and the Humber
 East of England
 East Midlands
 West Midlands
 Greater London
 South East England
 South West England
 Wales
 Scotland
 Northern Ireland

5. Your level of education:

- Secondary School (GCSE, O Level)
 Secondary School/College (A Level, HND, Diploma, NVQ etc)
 University First degree (BSc, BA)
 University Higher Degree (MA, MSc, PhD etc)
 Prefer not to answer

6. Total annual household income:

- less than £20,000
 £21,000 - £40,000
 £41,000 - £60,000
 £61,000 - £80,000
 £80,000 +
 Prefer not to answer

7. Your ethnic background:

- White
 African or Caribbean
 Asian
 Mixed
 Other
 Prefer not to answer

8. How many of your children are affected by an undiagnosed condition?

- One
 Two
 Three

9. How old is your child (children) without a diagnosis?

First child.....
 Second child.....
 Third child.....

10. At what age did their symptoms first become apparent?

First child.....
 Second child.....
 Third child.....

11. Do you have any children not affected by the condition?

- Yes
 No

12. Which of the following statements do you think best describes your child's (or children's) health or development problems?

	First child	Second child	Third child
Has no real effect on the child's daily life (the child can enjoy all the usual activities and needs no additional health care or support with schooling).	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Has some effect on the child's daily life (the child requires some support regarding their health care or learning).	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Has a major effect on the child's daily life (the child requires constant support with regard to activities, health and/or learning).	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

SECTION 2: QUESTIONS ABOUT THE BOOKLET

The following questions are about the information booklet.

Please tick (✓) one box only for each question

1. Do you still have the booklet?

- Yes
 No
 Don't know

2. Did you read it?

- Yes
 No

3. Did you find the information:

- Very useful
 Moderately useful
 Of no use

4. Did it tell you anything new?

- Definitely yes
 Possibly
 Not really
 Definitely no

5. Was the information relevant?

- Definitely yes
 Possibly
 Not really
 Definitely no

6. Did you feel overwhelmed with information?

- Definitely yes
 Possibly
 Not really
 Definitely no

7. Did you find the information too technical?

- Definitely yes
 Possibly
 Not really
 Definitely no

8. Did you find the information too limited?

- Definitely yes
 Possibly
 Not really
 Definitely no

9. Did the information change your ideas about something?

- Definitely yes
 Possibly
 Not really
 Definitely no

10. Have you shown the information to anyone else? (Tick all that apply)

- Partner
 Parent(s)
 Children
 Sibling
 Other family members
 Friends
 Others (please specify).....
 No one

If you have any comments on things that you particularly **liked** about the booklet, write them here.

If you have any comments on things that you particularly **disliked** about the booklet, write them here.

If you have any general comments about the booklet, please write them here.

SECTION 3: QUESTIONS ABOUT HOW YOU FEEL

Below are a list of comments made by people about stressful life events or situations. Thinking about the fact that you do not currently have a diagnosis for your child's condition, please indicate how frequently these statements were true for you in the last week. If any item is not relevant to you, please tick the 'not at all' option.

Please tick (✓) one box only for each question.

1. I thought about it when I didn't mean to.

- Not at all
- Rarely
- Sometimes
- Often

2. I avoided letting myself get upset when I thought about it or was reminded of it.

- Not at all
- Rarely
- Sometimes
- Often

3. I tried to remove it from my memory.

- Not at all
- Rarely
- Sometimes
- Often

4. I had trouble falling asleep or staying asleep, because of pictures or thoughts about it that came into my mind.

- Not at all
- Rarely
- Sometimes
- Often

5. I had waves of strong feelings about it.

- Not at all
 Rarely
 Sometimes
 Often

7. I stayed away from reminders of it.

- Not at all
 Rarely
 Sometimes
 Often

9. I tried not to talk about it.

- Not at all
 Rarely
 Sometimes
 Often

11. Other things kept making me think about it.

- Not at all
 Rarely
 Sometimes
 Often

13. I tried not to think about it.

- Not at all
 Rarely
 Sometimes
 Often

15. My feelings about it were kind of numb.

- Not at all
 Rarely
 Sometimes
 Often

6. I had dreams about it.

- Not at all
 Rarely
 Sometimes
 Often

8. I felt as if it hadn't happened or wasn't real.

- Not at all
 Rarely
 Sometimes
 Often

10. Pictures about it popped into my mind.

- Not at all
 Rarely
 Sometimes
 Often

12. I was aware that I still had a lot of feelings about it, but I didn't deal with them.

- Not at all
 Rarely
 Sometimes
 Often

14. Any reminder brought back feelings about it.

- Not at all
 Rarely
 Sometimes
 Often

SECTION 4: MORE QUESTIONS ABOUT HOW YOU FEEL

Please answer the following questions according to how you feel about not having a diagnosis for your child. **Don't take too long over your replies; your immediate reaction to each question will probably be more accurate than a long thought-out response.**

Please tick (✓) one box only for each question.

Have you recently.....

1. Been able to concentrate on whatever you're doing?

- Better than usual
 Same as usual
 Less than usual
 Much less than usual

2. Lost much sleep over worry?

- Not at all
 No more than usual
 Rather more than usual
 Much less than usual

3. Felt that you are playing a useful part in things?

- More so than usual
- Same as usual
- Less useful than usual
- Much less useful

4. Felt capable of making decisions about things?

- More so than usual
- Same as usual
- Less so than usual
- Much less so than usual

5. Felt constantly under strain?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

6. Felt you couldn't overcome your difficulties?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

7. Been able to enjoy your normal day-to-day activities?

- More so than usual
- Same as usual
- Less so than usual
- Much less than usual

8. Been able to face up to your problems?

- More so than usual
- Same as usual
- Less so than usual
- Much less than usual

9. Been feeling unhappy and depressed?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

10. Been losing confidence in yourself?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

11. Been thinking of yourself as a worthless person?

- Not at all
- No more than usual
- Rather more than usual
- Much more than usual

12. Been feeling reasonably happy, all things considered?

- More so than usual
- About same as usual
- Less so than usual
- Much less than usual

Thank you very much for completing this questionnaire and for your help with this study. I hope that you have found it interesting.

Please return it in the freepost envelope or to the address on the cover.

With best wishes,

Celine Lewis



Dear

Many thanks for completing the questionnaire. Please find enclosed the information booklet. I hope you find it useful but it would be really helpful for me if you could give me some feedback, letting me know what you think and ways that I could improve it. If you would like to do this, I have enclosed a short questionnaire.

Many thanks again for your support,

Celine

Celine Lewis
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N1 3QP

Tel: 0207 704 3141

Email: celine@geneticalliance.org.uk

QUESTIONS ABOUT THE BOOKLET

The following questions are about the information booklet.

Please tick (✓) one box only for each question

1. Do you still have the booklet?

- Yes
- No
- Don't know

2. Did you read it?

- Yes
- No

3. Did you find the information:

- Very useful
- Moderately useful
- Of no use

4. Did it tell you anything new?

- Definitely yes
- Possibly
- Not really
- Definitely no

5. Was the information relevant?

- Definitely yes
- Possibly
- Not really
- Definitely no

6. Did you feel overwhelmed with information?

- Definitely yes
- Possibly
- Not really
- Definitely no

7. Did you find the information too technical?

- Definitely yes
- Possibly
- Not really
- Definitely no

8. Did you find the information too limited?

- Definitely yes
- Possibly
- Not really
- Definitely no

9. Did the information change your ideas about something?

- Definitely yes
- Possibly
- Not really
- Definitely no

10. Have you shown the information to anyone else? (Tick all that apply)

- Partner
- Parent(s)
- Children
- Sibling
- Other family members
- Friends
- Others (please specify).....
- No one

If you have any comments on things that you particularly **liked** about the booklet, write them here.

If you have any comments on things that you particularly **disliked** about the booklet, write them here.

If you have any general comments about the booklet, please write them here.

REC Offices
South House, Royal Free Hospital
Ford Street, London
NW3 2QG

Tel: 020 7794 0500 - Extn: 38906
Fax: 020 7794 0714

21 September 2010

Ms Celine Lewis
Project officer at GIG/PHD student at University of Plymouth
Genetic Interest Group (patient organisation)
Project officer at GIG/PHD student at University of Plymouth
Unit 4D Laroy House
436 Essex Rd
London N1 3QP

Dear Ms Lewis

Study title: A pilot study, in preparation for a randomised controlled trial, exploring the effectiveness of two information resources to provide psychosocial information about genetic testing to patients.

REC reference: 10/H0722/25

Amendment number: 2

Amendment date: 14 September 2010

The above amendment was reviewed on 20 September 2010 by the Sub-Committee in correspondence.

Ethical opinion

Favourable Opinion

The members of the Committee taking part in the review gave a favourable ethical opinion of the amendment on the basis described in the notice of amendment form and supporting documentation.

Approved documents

The documents reviewed and approved at the meeting were:

Document	Version	Date
Notice of Substantial Amendment (non-CTIMPs)	2	14 September 2010

Membership of the Committee

The members of the Committee who took part in the review are listed on the attached sheet.

R&D approval

All investigators and research collaborators in the NHS should notify the R&D office for the relevant NHS care organisation of this amendment and check whether it affects R&D approval of the research.

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

10/H0722/25:

Please quote this number on all correspondence

Yours sincerely



Veronica Oke
Committee Co-ordinator

E-mail: veronica.oke@nhs.net

Enclosures: List of names and professions of members who took part in the review

Copy to: Dr Richard Stephenson, R&D office

Appendix 41

British Medical Association
0300 123 123 3 bma.org.uk



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Library

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Fax: +44(0) 20 7386 2544
Email: bma-library@bma.org.uk

Thursday, 15 July 2010

Dear Celine Lewis

BMA Patient Information Awards 2010

I am pleased to be able to inform you that your resource, **Living Without A Diagnosis - Information For Parents**, has won a **Commended** prize for the 2010 Patient Information Awards. The results were approved by the BMA's Board of Science last month.

Please note that the results are confidential until immediately after the awards ceremony.

All Highly Commended titles are shortlisted for the Patient Information Award 2010. The winning title has been chosen by our final judging panel but we will not disclose it in advance. The winner will be announced at a ceremony and reception on the evening of **Tuesday 14th September** at BMA House.

Unfortunately, because of the number of prizes to be awarded this year, we are unable to invite winners of commended leaflets to the award ceremony. I will email these certificates to you before the ceremony.

I enclose a form which details the inscription that will appear on the certificate. Please post this back as soon as possible, indicating either that the wording is correct or detailing any amendments. We do not make a charge for the presentation certificate which will be emailed to you immediately after the awards. Additional certificates made out to different recipients or personalised in some other way, can be ordered from our on-line Additional Certificate order form, <http://web2.bma.org.uk/bmalibrary.nsf/bcac>

Additional certificates cost £40 plus VAT and are invoiced after dispatched after the awards.

All forms should be returned by Tuesday 27th July so that the certificates can be prepared. As our awards work to a very tight schedule we regret that we will be unable to edit any forms received after this date.

There will be an announcement of the awards in the *BMJ* after the ceremony and they will also be listed on our BMA library website: <http://www.bma.org.uk/librarybookcomp>

Any queries regarding press and publicity should be addressed in the first instance to Mr Paul Gashby in

Chief Executive/Secretary: Tonybourne

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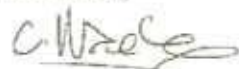


**STANDING UP
FOR DOCTORS**

the BMA press office on 020 7383 6165.

Congratulations on your award. If you have any queries, please do not hesitate to contact me

Yours sincerely

A handwritten signature in black ink, appearing to read 'C. Waide', with a stylized flourish at the end.

Manager,
BMA Patient Information Award

Living Without a Diagnosis: The Parental Experience

Celine Lewis¹, Heather Skilton,² and Ray Jones²

Aim: The aim of this study was to explore the parental experiences of raising a child without a diagnosis. **Method:** Qualitative semistructured interviews were conducted with 14 parents recruited through a large Regional Genetics Centre in the United Kingdom. The interview guide was designed to examine issues such as when and why parents started searching for a diagnosis, whether they were still searching, and what psychosocial issues had arisen as a result of not having a diagnosis. Data were analyzed using the Grounded Theory method. **Results:** The parental experience can be viewed as a journey, which comprises of two distinct components: the inner, emotional experience, and the outer, sociological experience. Issues that comprise the emotional journey include the realization that there is a problem, the experience of testing, reasons for wanting a diagnosis, the emotional impact, and active coping mechanisms. Social issues include the experience with professionals, the various support networks accessed by parents, and issues such as education and housing. The issue of frustration was one that occurred throughout the journey. **Conclusion:** Although some of the experiences cited by parents are common to families raising a child with a diagnosed condition, lack of diagnosis adds a layer of complexity.

Introduction

IT IS ESTIMATED THAT IN 30%–38% of cases of children with severe learning disability (Hodgson, 1998; Dally *et al.*, 2000) and 60% of cases of children with multiple congenital anomalies (Cohen, 1997). It is not possible to provide parents with a certain diagnosis or etiological explanation. The psychological and social implications of raising a child with a known disability are well documented (Carmichael *et al.*, 1999; Fares and Mackintosh, 1999; Saei *et al.*, 2006; Wiedelbruch *et al.*, 2008; Wong and Heriot, 2008), but there are only few studies detailing the psychological and social experience of parenting a child without a diagnosis.

Studies investigating the psychosocial experience of raising a child with a known condition have highlighted a variety of experiences encountered by parents. In a qualitative study investigating the experience of parents of children with Down syndrome, parents expressed anxiety about the future, including whether their children would be able to look after themselves independently in later years (Saei *et al.*, 2006). Parents of children with ataxia telangiectasia were found to experience multiple mourning processes including mourning the loss of the child they thought they had and the child they used to have (Fares and Mackintosh, 1999). In a study focusing on parents of children with fragile X syndrome, parents felt there was a lack of support from clinicians after they had received a diagnosis (Carmichael *et al.*, 1999), and self-

blame, behavioral disengagement, and information seeking were found to be some of the coping mechanisms used by parents of children with cystic fibrosis (Wong and Heriot, 2008) and Turner syndrome (Stark and Muller, 2002).

A review of the literature identified two studies in which the parental experience of diagnostic uncertainty was investigated. In one study (Kozathal *et al.*, 2001) conducted in the United States, researchers identified six areas in which parents claimed a diagnosis would have an impact. These included providing a label, informing treatment, and establishing social support. Results from a study conducted in Denmark (Grongaard and Skov, 2006) showed that parental satisfaction with the diagnostic process was strongly related to the certainty of the diagnosis and that parents found it difficult coping with an uncertain future. Our review of the literature, however, did not identify any studies conducted in the United Kingdom investigating the experience of parenting a child without a diagnosis.

It is important to investigate this issue from a United Kingdom perspective as differences in healthcare systems across these countries may have an impact on psychosocial outcome for parents. In 1997, the provision of genetic services in the European countries was evaluated by the Concerted Action on Genetic Services in Europe (CAGSE). This evaluation revealed that practices, resources, as well as traditions in the different countries varied considerably. Access to genetic services will also vary. Denmark, like the United Kingdom,

¹Genetic Alliance UK, London, United Kingdom.

²Faculty of Health and Social Work, University of Plymouth, Plymouth, United Kingdom.

regulates genetic testing through the national healthcare system, whereas in the United States, it is provided by state and private sector organisations (Godard *et al.*, 2003). Further, across Europe and the United States there exists a diversity of cultures and opinions about a number of issues relevant to genetics, including human reproduction issues and community and individual approaches to the significance of disabilities (Godard *et al.*, 2003).

The aims of this qualitative study were therefore to (1) assess the psychosocial parental impact of having a non-diagnosed child in the United Kingdom, (2) identify whether the psychosocial implications were similar to findings from studies in which parents did have a diagnosis, and (3) identify whether there were any issues that were unique to this situation.

Methods

This was an exploratory study of parents' feelings and experiences, and therefore, we chose a qualitative method (Pope and Mays, 2006). A Grounded Theory approach enables the researcher to draw appropriate themes from the data and to develop explanatory theories in connection with the topic being studied (Strauss and Corbin, 1998). Ethical approval for the study was obtained from the NHS Ethics Committee.

Participants were recruited through staff at a large Regional Genetics Centre in the United Kingdom. Eligible parents had a child who had been seen at the genetic clinic, but who did not, at the time of recruitment, have a diagnosis of an identified syndrome. To achieve maximum variation in the sample, the inclusion criteria consisted of (1) parents who were still actively searching for a diagnosis and those for whom it was not a priority, (2) children who varied in age, (3) parents of children where the non-diagnosed child was an only child and parents of children where there was more than one child in the family, (4) parents who had a "working diagnosis" (a diagnosis given by the clinician that was socially enabling, but did not provide an etiological explanation) such as autism or bilateral cataract, and parents without; and (5) parents who found out there was an undiagnosed condition during pregnancy and parents who found out after the child was born.

Parents of children considered suitable were invited to participate in the study via a letter that included a participant information sheet. They were asked to return the reply slip if they were interested in participating, and face-to-face interviews were then arranged. All participants stated a preference to be interviewed in their own homes. The interviews were conducted by the lead researcher (C.L.) and permission was given by all interviewees for the interview to be audiotaped. Interviews took place between December 2008 and August 2009 and lasted from 30 min to 1 h. Recruitment occurred in stages to enable the researcher to identify specific characteristics of potential participants, to recruit purposively.

Consent was recorded in writing before each semi-structured interview. Where both parents were available, they were asked whether they would prefer to be interviewed together or separately. All couples preferred to be interviewed together. The interview guide was designed to examine issues including when and why parents started searching for a diagnosis, whether they were still searching for a diagnosis and what information they hoped a diagnosis would provide, and

what had been the main issue, both practical and emotional, that had arisen as a result of not having a diagnosis.

Interviews were transcribed by the researcher and data were analyzed using the Grounded Theory method (Strauss and Corbin, 1998). The process of interviewing, coding, and categorizing was fluid and continuous. As analysis of the interviews continued, data were coded into either pre-existing codes or new codes. Coding and sorting was done using the qualitative research software NVIVO (QSR International Pty Ltd.) and interview questions were amended to take into account the emerging concepts. Categories were continuously checked and compared against segments of the text (constant comparison) to determine their properties and dimensions. During this process, codes and categories were revised, re-organized, split, or merged. A second researcher also coded and categorized parts of the interviews to ensure trustworthiness of the findings. Any disagreements were discussed and amended. At the point when no new codes or categories were emerging, examination of categories was carried out to identify links and relationships between them (axial coding). Finally, as greater insight was achieved, more theoretical and abstract accounts were developed (selective coding). The results of this analysis, including proposed theoretical explanations about the data and their implications for health service provision, are discussed below. All identifying details have been changed to protect confidentiality.

Results

Sample characteristics

The parents of 20 children were invited to participate and parents of 9 children agreed to be interviewed (45% response rate). During five interviews, both parents were present. In the other four interviews, only the mother was present (in three of these cases the parents had separated), resulting in a total of 14 participants. All participants were living in Southern England. Three mothers had given up work either for financial reasons or because it was too difficult to work and look after their children. Two mothers had recently returned to work because their child had turned five, one mother was working part time, one mother was still on maternity leave, one mother chose not to work, and one worked full time. Of the five fathers involved in the study, all worked full time. The ages of undiagnosed children ranged from 3 months to 9 years. Table 1 shows further characteristics of the parents and affected children, including a range of problems as described by the parents. Many of the children had some form of learning difficulty, but the range of physical manifestations was broad. There was a range of activity level concerning the search for a diagnosis. This ranged from "still persevering with all the different channels" to "no longer actively searching". Even though many of the parents described their children as having "developmental delay," two had been given a working diagnosis to describe their child's condition, in one case this was autism and in the other, bilateral cataracts. Another woman had been told her child had a chromosome translocation; however, because the diagnosis was so rare, she regarded it as a non-diagnosis as there was no prognosis.

The overriding theme that emerged was that the experience of having an undiagnosed child was a "journey" encountered by parents. The journey began with the identification of a medical problem and was still ongoing at the time of the

TABLE 1. PSYCHOSIS OF THE PARENTS, AND OTHER CHARACTERISTICS OF PARENTS AND NONDIAGNOSED CHILDREN

Interview	Pseudonym	Parents marital status	No. of children	Position of nondiagnosed child	Age when problem was detected	Age of nondiagnosed child	Gender	Problems experienced by nondiagnosed child as described by parents
1	Sally Black	Separated	2	Oldest	3-month scan	9 years	F	Growth deficiency, severe speech problems, bowel incontinent, severe learning difficulties, hearing problems, poor eyesight
2	Deena Franka	Separated	2	Twins	22-week scan	8 years	M	Hydrocephalus, problems with mobility, large head size, large body weight
3	Abe & Laura Dent	Married	2	Twins	3 years	6 years 6 years (twins both affected)	F	Not reaching milestones, no speech, epiglottis, defects on hands, small head, microcephaly, prominent defect on spine
4	Macy & Anthony Williams	Married	2	Youngest	Birth	3 years	M	Pupils, abnormalities in eyes, bilateral cataracts
5	Rachel & Alan Bateman	Married	1	N/A	2 years	5 years	F	Low muscle tone, unable to walk, disclination in eyes, dysmorphic features
6	Alison Foxcross	Married	1	N/A	20-week scan	3 months	F	Chromosome translocation
7	Sophie & Colin Keef	Married	1	N/A	9 months	15 months	M	Dysmorphic features, wide set eyes, low weight, small holes in the heart
8	Julia & David Bird	Married	2	Youngest	6 months	20 months	M	Low muscle tone, not reaching milestones, teokville, wobbly
9	Samantha Clark	Separated	3	Youngest	20-week scan	4 years	M	Developmental delay, fits, unable to walk

N/A, not applicable.

interview. This journey comprises two distinct components: the "inner world" experience, comprising emotions and cognitions, and the "outer world" sociological experience.

The "inner" journey

The initial emergence of the problem: There was a noticeable difference in the experiences described by parents depending on whether the problem was identified during pregnancy or not. For those who found out during pregnancy, the information came as a shock and was devastating.

"It was like being hit with a truck. I was sobbing, sitting outside sobbing in the garden and it was awful. It's a real shock because you don't anticipate it. It was horrible, it was really, really horrible." (Alison)

When problems became apparent after birth, the realization that there was a problem was more gradual. It often began when parents started to notice that the child was not reaching certain milestones.

"We realized after six or seven months he wasn't reaching his milestones. He wasn't sitting up, his head in particular, he was always quite floppy." (Julia)

Statements such as "there has been a lot of anxiety" (Macy) and "it was very stressful" (Laura) were used to describe the experience.

Experience of testing: An integral part of the journey was the testing that children underwent during the search for a diagnosis. Most participants named a variety of procedures including blood tests and MRI scans, but other tests such as X-rays were mentioned. Many of the children had undergone testing over prolonged periods. The experience of testing varied for parents. Some were happy for their child to continue testing if it might lead to a diagnosis, whereas for others the experience was traumatic and upsetting. Comments from three parents suggested a certain level of trust with their health professionals.

"If there is an issue and they want to carry on and dig a little further then ok lets just go with it." (Julia)

For other parents, however, the experience was more negative. One had a particularly bad experience because of a number of mistakes being made with blood samples.

"And I wish that last time it upset me so much that I said to everyone 'take the bloods you need, store them, and there are no more bloods being taken because I can't put Clire through this.'" (Sally)

For those women who were pregnant at the time of testing, the experience was particularly traumatic because it was at a late stage in the pregnancy.

Reasons for wanting a diagnosis: "Help with care and treatment" and "to know about the future" were the two most common reasons given for wanting a diagnosis. Parents felt that if they had a diagnosis it would enable them to make more accurate decisions regarding their child's care and treatment and provide information about the prognosis.

"I'd just like to know how we treat it really. If it's Russell Silver syndrome it can be treated by growth hormones which is a positive thing." (Sophie)

Without a diagnosis, parents were unsure how their child would progress over time, and this was a great source of frustration. One parent described it as being "blind." During three interviews, participants also commented that they

wanted a diagnosis just to have "a reason." This desire for a reason implies that having a diagnosis would, for some parents, provide psychological relief, even if it would not change the situation.

"... because even if we had a diagnosis he's still Charlie to me, its not going to change him... its just on a personal emotional level I hate not knowing..." (Sandra)

A diagnosis was seen as a social tool by parents of affected children. Three parents raised the point that it would be helpful to have a term that could help summarize what was wrong with their child when asked by friends or other parents.

"... it would be nice to sort of say oh yes, Laura has got rather than we don't know." (Rachel)

Two mothers explained how they had decided to use the terms "severe special needs" and "developmental delay" to describe their child's condition. This "social diagnosis" made it much easier when asked (by nonmedical people) as it meant they did not have to go into the specific details. One mother had decided upon the term "developmental delay" in a key worker meeting attended by members of her support team (including her health visitor and occupational therapist amongst others). This had been a very useful exercise that she recommended to other parents. Other reasons cited for wanting a diagnosis included concern about recurrent risk in future pregnancies, to help with schooling, and to have an explanation to give to their child.

Feelings about getting a diagnosis: Although parents all said that they would like a diagnosis, when probed further many said that getting a diagnosis was no longer a priority. This was either because they were more concerned with dealing with the day-to-day issues related to the condition, they felt that their child was happy, and therefore, a diagnosis was not vital, or they had come to accept their child's condition and wanted to move on.

"If you worry about having a name for something, if your child is happy and your child isn't in and out of hospital then does it really matter?" (Donna)

Two parents commented that a diagnosis was just a name and would not "change" their child. For one mother, the actual problems, including a hole in the heart, were more important than a name. Three parents mentioned that they would still have to explain what the diagnosis meant to others.

For one woman who had been told her child had a translocation, she still felt that this "diagnosis" did not provide any useful information about the condition or the future.

"So when they told you it was a translocation was that a relief?" (interviewer)

"Well it's like being told something in a foreign language really. It wasn't a relief because I didn't understand it. I'd never heard of a translocation... It was still a meaningless diagnosis. You need to know what it means. I don't care what it's called. I just want to know what it means, or what I can do, or what I can do to help." (Alison)

However, for one couple who had been given a working diagnosis of autism while testing was ongoing, the diagnosis was a relief as it proved to others that there was something wrong. Many of their emotional difficulties had been connected with the fact that no one believed there was anything wrong with their children, other than bad behavior.

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"We got a lot of relief out of our diagnosis because at least somebody believed and somebody could see we were not imagining these problems, they were actually there. The girls were actually having difficulties and up until our diagnosis nobody seemed to accept that." (Abe)

Emotional impact: Parents commonly experienced anxiety when they first found out that there was a medical problem, especially if this occurred early in the child's life, before certain possible syndromes could be ruled out. However, most parents did not report feeling anxious over the long term. The emotional issues with a long-term impact related to the lack of control parents felt, particularly regarding their child's future. Issues raised included whether they would have to care for their child for the rest of their lives, whether their child might eventually die from their condition, recurrence risk in future pregnancies, the emotional difficulties of being unable to make "life-plans" for their child, and not knowing how to deal with aspects of their child's development such as the onset of puberty.

"I do not know what is going to happen to him in the future and that is the worst thing I think, it is just not knowing because as far as I am concerned I now have to be his care for the rest of his life." (Dorina)

These participants described their experience as being stressful. Reasons cited included the lack of sleep coupled with a full-time job, the constant visits to hospital, and looking after a special needs child as a single mother. Two parents described the sense of helplessness they felt in trying to find a diagnosis, one father felt that it was "just a case of waiting to see if they do the things that a child with these syndromes would do" (Anthony). A number of parents expressed a sense of guilt relating to their nonaffected children. One woman explained that she felt guilty toward her other child "because we can't go out and do the things that a normal eight-year-old girl would love to be doing" (Dorina). Other emotions expressed by parents included the strain on relationships, the lack of psychological help available, and the difficulty in coming to terms with the fact that their child did not look "normal."

However, not all the emotions described by parents were negative in nature. A number of parents commented that the experience was an "emotional rollercoaster." There were "lows," but there were also "highs." For one mother, even though the experience had at times been difficult, it had been "positive overall" (Sophie). Many of the parents were also keen to express how rewarding and loving their child was. Comments including "I love her to death" (Sally) and "he's surprising sometimes as to how loving and gorgeous he is" (Samantha) were made during the interviews.

Coping mechanisms: The majority of participants used terminology during interviews that implied they were actively coping with their situation and trying to remain positive. Many of these coping statements related to accepting the situation, getting on with life, and just enjoying their children. Parents were also aware of the need to focus their energy on being optimistic, as highlighted by comments such as "you have to keep positive thinking" and "when you focus on the positive your life becomes so much easier." One particular coping mechanism that four parents engaged in was to compare their own situation to others:

"... you realize there are children there that are far worse off than Nick. Children with severe cerebral palsy, spina bifida, all sorts of problems..." (Julia)

Father: "It puts it in perspective"

Mother: "...and you kind of think, what am I complaining about..." (Julia)

Looking for information, either online or by speaking with other parents, was another coping strategy parents employed. One mother who was given a high risk of Down syndrome after a 20-week scan described how she "spent that weekend talking to colleagues that worked with kids with Down's, talking to my friend who is a social worker, reading about Down syndrome, getting as informed as I could" (Aileen). Another means by which parents coped was by playing an active role in ensuring appropriate care, treatment, and services were met by service providers. Many parents were keen to take control of the situation and, in essence, act as the "gatekeeper" for their child's care. However, there were many examples of parents having to push or fight for services and appointments:

"You have to push, you have to say what's happening and you have to chase people... and you end up getting to a point where you're like 'I don't care if I upset you now, my children matter and you don't'... I've had to learn to be that sort of person." (Laura)

The sociological journey.

Experience with professionals: Communication and support between medical professionals and parents was on the whole positive, with most parents feeling that there was a good line of communication maintained and support provided. In addition, parents wanted to be active participants in the diagnostic process and felt the medical team acknowledged them as experts on their child's condition, as highlighted by one woman who felt she was free to "ring up and say 'can you check this on the database?'" The experience was less positive when it came to communication between health professionals. Two parents found that there was a lack of communication between specialists, and often it was up to them to keep the medical team informed of test results and any specialists seen since the last appointment. A further issue that was particularly frustrating for parents was the need to constantly repeat their child's "life story" because of the numerous different specialists involved in their child's care, and the lack of communication between them:

"... you are always telling everything again and you just think why can't you just open your notes and read what is in front of you because I have told at least a 100 people this, please I do not want to tell anybody again and this is quite frustrating." (Dorina)

The importance of support and advice from professionals such as health visitors, social support workers, and housing officers was evident from interviewees' comments. The health visitor seemed to be a particularly important person along the parental journey, who provided information and support about issues including local nurseries and playgroups, and disability benefits. In some cases the health visitor had adopted a "key worker" role by ensuring all health professionals were kept up-to-date with tests and appointments and by helping facilitate access to services. They also provided psychological support to parents, particularly mothers.

"Pam my health visitor has been an absolute rock because I mean I've had other problems not just with Charlie but with Leah, his elder sister, eating disorders and things like that. So she has been really involved in my whole family network." (Samantha)

Other specialists who were seen to play an important role included the support worker, the housing officer, and staff at local nurseries and playgroups.

One negative aspect concerning professionals that was mentioned was the high turnover of staff. One woman commented that "You hardly ever see the same person" (Sally). Another parent was also very fearful of the transition to a new team of professionals that would inevitably happen soon, as her son was about to turn 5 years old.

Other support networks: Parents found that having a child with a disability placed a strain on their relationship. One couple found that because they were constantly tired, they rarely went out and spent time together. Two couples acknowledged that they argued more as a result of the stress and worry of looking after a child with special needs. On the whole, parents found that their family members were very supportive; however, there were examples of family members being unsupportive and insensitive. One mother described her sister as treating her son "as a second class citizen."

Two mothers commented that over the years they had built up a network of friends who also had children with special needs. One mother commented that "you find when you have special needs children most of your friends have got special needs children because they are more understanding" (Laura). She had also found that there was a certain stigma attached to having a child with special needs, and that most of her other friends did not "want to know. You don't get invited to parties or things like that." Parents had made friends through nurseries, schools, and patient support groups.

Support groups, chat forums, and the Internet were further places where parents had found information and support. One father commented that chat forums were good places to retrieve information concerning the important day-to-day issues one might not receive from specialists, and support groups and other charitable organizations helped in some cases by providing respite care and funding equipment and travel. A number of parents, however, commented on the lack of support groups specifically for families without a diagnosis.

Although it was natural for parents to want as much information as possible, many parents spoke of how easy it was to scare yourself while searching for information online. Comments such as "you end up petrified," "it freaks you out," and "I went on line which is a dangerous thing to do" were made about searching on the Internet.

Education, employment, housing, and insurance: Schooling was an important and, sometimes, problematic issue for parents, with some parents unsure whether their child would be better off in a special needs or mainstream school. For two parents, the concept of "dual placement" was particularly appealing, whereby the child spends part of the time in a mainstream school and part of the time in a special needs school. Getting access to appropriate schooling was another hurdle for some parents. The difficulties associated with getting a statement were mentioned during two interviews. The process was so

long and laborious for one mother that by the end of it she had "hit rock bottom."

The issue of employment was frequently raised during interviews. For mothers in particular, having a child with a disability had an impact on their ability to work. Five mothers had to give up work as a result of their child's condition, although two had gone back to work as their children had entered full-time education.

"I've tried 2 or 3 times to go back to work but its beaten me in the end and it's like OK I can't do this." (Sally)

One mother found that her son's constant appointments meant it was very difficult for her to commit to an employer. Another had decided to go part time so that she would have the free time needed to take her son to hospital appointments.

A number of participants discussed issues related to housing, particularly the lack of suitable housing made available by the council, which was particularly frustrating for parents. One mother remarked that her council was unwilling to spend money adapting the house because she had no diagnosis.

"He is coming up to 4 now and he is still not walking or anything. The bedroom is upstairs and he is sharing a bedroom with his sister, and they are saying that they won't grant any funds because they say 'well if we do that and spend £30,000 then in 6 months he might be walking and it will be a waste of money.'" (Samantha)

Discussion

The findings from this analysis highlight the main emotional and social issues that occur throughout the journey undertaken by parents on their search for a diagnosis. This journey can be viewed as a series of waves, or "highs" and "lows." There were the associated difficulties that raising a child with a nondiagnosed condition posed, such as the lack of information about the future and problems in acquiring appropriate housing and education, but there were also positive experiences, such as the supportiveness of professionals and the joy their child had brought to them. This journey, or "emotional rollercoaster" as some parents described it, was a unifying feature of all the experiences documented.

One of the main issues that weaves its way throughout the findings is the issue of frustration. In fact, the issue of frustration was also found to be an issue experienced by parents with undiagnosed syndromes in a similar study conducted in the United States (Resenthal et al., 2001). Yet, frustration is not an issue experienced only by those without a diagnosis. Other studies have shown that frustration also occurs when parents do have a diagnosis (Ruin et al., 2004; Wennick and Hallstrom, 2007; Doig et al., 2009; Henderson et al., 2009). Further, a number of other findings from this study, including the reasons given for wanting a diagnosis, the active role that parents play in managing care and treatment for their child, and the maternal difficulties in meeting employment expectations, were experienced in other studies in which the child did have a diagnosis (Young et al., 2002; Hummelink and Pollock, 2006; Green, 2007). Yet, there does appear to be one key difference. The difficulties described in studies where parents did have a diagnosis are primarily sociological constraints. However, many of the difficulties in this study are emotional stresses, or as Green (2007) refers to them, "subjective burdens." As well as experiencing sociological burdens, parents in this study ex-

experienced ongoing psychological issues, particularly the difficulty of coming to terms with not having a reason why, which perpetuated a feeling of being out of control. This emotional distress in particular may make the experience of raising a child without a diagnosis uniquely different.

Parents used a number of different coping methods during their journey. One commonly used was "information seeking," a proactive engagement that is consistent with the "monitoring" style of coping described by Miller (1987). Further, many of the parents in this study took an active role in ensuring their child received appropriate care and support, tried to remain in a positive mindset, and accept the situation (coping behaviors echoed in the findings of Rosenthal *et al.* (2001)). These strategies are similar to the "problem-focused coping" and "emotional-focused coping" strategies used by people to cope with a particular situation, as described by Lazarus and Folkman (1984). Parents appeared to use both these strategies, suggesting they were successfully coping with their situation, yet further quantitative research would be needed to test this theory.

Although all parents were still searching for a diagnosis, the importance placed on finding a diagnosis varied, with five parents indicating that for them it was no longer their main priority. Yet, for these parents, the psychological burden of living in a state of uncertainty and the lack of control they had over their situation was very much apparent. These parents appeared to be more motivated to search for certainty, and they had a lower tolerance for ambiguity, a cognitive-motivational factor described in the theory of the need for cognitive closure (Webster and Kruglanski, 1994). However, although the majority of parents who attend genetic services, because their child has learning needs and developmental delay, will be searching for a diagnosis, between 30% and 50% do not receive one (Daly *et al.*, 2000). The need for, and lack of certainty, is therefore an issue for many parents seen by genetic specialists. Specialists may, therefore, want to highlight to parents at the beginning of the diagnostic process that a diagnosis is not always achievable if parents do not have unrealistic expectations. It may be helpful, therefore, to discuss other areas where increased certainty may be possible, to give parents an expectation that is more realistic and more likely to be satisfied (Skirton, 2006). Parents should also be reassured that even without a diagnosis, their child should receive care, treatment, and services that are tailored to their needs, and not having a diagnosis should not stop their child from reaching their full potential.

Parents who had found a professional (such as a health visitor) who coordinated care, facilitated access to specific services, and provided emotional and social support found this assistance to be extremely valuable. In addition, their experience was noticeably less traumatic, with fewer instances in which they had to "push" or "fight" for services or appointments. A number of other United Kingdom studies and reports assessing the impact of a key worker have reported similar findings (Lalor *et al.*, 2001; Rabi *et al.*, 2004; Greco *et al.*, 2006; Allford and Hillier, 2008), highlighting the need for and importance of a key worker program for all families of children with disabilities, particularly those with multiple syndromes. In the United Kingdom, studies evaluating the impact of a key worker program found that parents benefited from the important information, especially about education and social services, and emotional

support provided by the community link team. Coordinating care and supporting families with care planning and review, speaking on behalf of the family when dealing with services, mediating between schools and families to tackle problems and to resolve sensitive or contentious issues, and providing help and support in a crisis are further areas where the role of the key worker could be expanded. If health visitors are the most appropriate professionals to provide this service, it is important that they receive the appropriate training to enable them to understand service users' needs, provide psychological and informational support, and access relevant services in a timely manner. Further, community health workers exist in many settings, but the background and actual role may differ from country to country or even state to state. The key worker in each case should be the professional who is best placed to be effective and most acceptable to the family, regardless of actual title. A further finding from this study was that without a diagnosis many parents felt there was no appropriate support group to contact. However, a number of organizations in the United Kingdom, such as Unique and Contact a Family, do cater for parents without a diagnosis. Clinicians and health workers need to be aware of these organizations so that they can effectively signpost parents toward them.

Numerous similarities in methods and findings were identified between this study and the Danish (Grungaard and Skov, 2006) and American (Rosenthal *et al.*, 2001) studies. All three were qualitative studies based on semistructured in-depth interviews with parents. However, in the Danish study, half the parents did have a diagnosis for their child's condition. Similar reasons for wanting a diagnosis were found in this study and that conducted in the United States. These included wanting information about their child's prognosis and wanting a "label" as a way to help other people understand their child's condition. However, parents in the Rosenthal *et al.* study seemed more concerned with the issue of reproductive risk than was evident in our findings. This may be attributed to the fact that some of the siblings of the affected children in the American study were at or reaching reproductive age. Parents in all three studies found it emotionally difficult coping with an uncertain future; however, like this study, some of the participants in the study by Rosenthal *et al.* felt that their interest in a diagnosis had diminished over time. There was an acknowledgment that even with a diagnosis, the situation would not substantially change. Similar to our findings, Rosenthal *et al.* found that parents had to fight to access services, including services from school districts, therapeutic services, or assistive equipment. This issue was not evident in the Danish study, possibly because this area was not explored or perhaps because services and equipment are more easily accessible and available in Denmark. This is an area that would benefit from further research. Use of problem-focused and emotional-focused coping strategies by parents was evident across all three studies. Like our study, Grungaard and Skov highlighted that parents sought information, learnt new skills, sought social support, and tried to focus on the positive. Similarly, Rosenthal *et al.* found that parents were active in ensuring their child received appropriate services, wanted as much information as possible, had come to accept the way their child was, and wanted to meet other parents whose children were similar to their own. Interestingly, the term "frustration," a common theme in our findings,

was also used on a number of occasions by Czerniack and Skow and Rowental *et al.* to describe the experience of parenting a child without a diagnosis. These findings highlight that even across countries with very different healthcare systems and cultures, many parents living without a diagnosis experience very similar psychological and day-to-day difficulties and challenges.

Parents suggested a number of useful measures that other parents could employ, to minimize the difficulties frequently encountered in accessing services, communicating with health professionals, and remaining positive.

- Be aware of all the services and benefits that are available to you. Ask the professional team around you to provide you with this information.
- Other organizations such as patient support groups, charities, your council's Family Information Service, the Citizens Advice Bureau (CAB), and other parents are also a good source of information.
- Not having a "name" for your child's condition can make it difficult when filling out forms. You find you end up writing "undiagnosed condition." Having a letter from your consultant that explains what this means can be helpful.
- It is a good idea to find a term such as "developmental delay" or "SWAN child" (syndrome without a name) that describes your child's condition. This can be helpful when explaining your child's disability to other people (such as friends and other parents). Ask your healthcare specialists to help you come up with a term that best describes your child's disability.
- Keep a record of your child's progress. Keep a diary; take photos and videos if possible. As well as being a useful record to show to doctors, it can also be comforting to look back and see the progress your child has made.
- Create a "passport" for your child. This can be a series of flashcards that explain things such as what your child can and cannot do, what they like/dislike, any health problems they have, etc. These can be taken to appointments and are a quick way of passing on information about your child.
- Do not compare your child to other children of the same age, or how your other children were at that age, as this can be upsetting. Every child is different.
- Try to enjoy your child. It is easy to focus on all their problems and to forget to enjoy watching them grow up.

The rapid advancements that are currently taking place in genomic medicine and diagnostic testing (Miller *et al.*, 2010) offer hope to those patients who are currently living without a diagnosis. Advances in diagnostic capacity through new technologies such as chromosomal microarrays (which are increasingly being used for genetic testing of individuals with unexplained developmental delay or multiple congenital anomalies) offer much higher diagnostic yields for various types of chromosomal aberrations, with testing techniques more efficient at finding submicroscopic deletions and duplications (Miller *et al.*, 2010). This, in theory, is promising for those currently living without a diagnosis. However, the clinical significance of much of the genomic information derived from these tests is not yet certain. The challenge will lie in ensuring that this technology has clinical utility and patient

benefit (Ali-Khan *et al.*, 2009). Further, genetic practitioners will need to determine best practices for effective communication of this genetic information, particularly with respect to incomplete knowledge and diagnostic uncertainty if they are to ensure a service that is useful, informative, and appropriate for patients and families.

Limitations

The participants in this study were self-selecting, and therefore, these results will not necessarily be a true reflection for all parents of children without a diagnosis. In addition, all the participants were living in Southern England and had been seen by staff at one regional genetics service. Further research is needed to see if the psychosocial experience of parenting a child without a diagnosis differs for parents in different geographical locations across the United Kingdom who access different regional genetic centers, primary care trusts, and local educational authorities. In addition, it would be interesting to explore how ethnic background, socioeconomic factors, and personal coping styles affect the parental experience.

Conclusion

The findings from this study highlight some of the main psychosocial issues experienced by parents living without a diagnosis. Although a lack of diagnosis may add a further layer of complexity to an already difficult situation, a number of experiences cited are common to families raising a child with a known disability. Genetic professionals may find it useful to highlight that with or without a diagnosis, families still face similar challenges, uncertainties, and joys.

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Can We Make Assumptions About the Psychosocial Impact of Living as a Carrier, Based on Studies Assessing the Effects of Carrier Testing?

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Abstract Receiving the results of genetic carrier testing may have an impact on the psychosocial health of the individual. Numerous studies have been conducted to assess the psychosocial effects of carrier status for a range of conditions. To systematically review research focused on the psychological and social impact of carrier testing on individuals in order to identify factors affecting the impact of carrier testing results, and discern areas where further research is needed. Twenty relevant papers meeting criteria for inclusion in this review were found. The main themes identified across these studies included: anxiety, guilt and stigmatization, effect on family relationships, effect on self image, active coping mechanisms and reproductive issues. Variables related to the psychosocial effect of carrier testing included whether the carrier has an affected child, mode of inheritance, genetic counseling, and life stage. A key finding concerns carriers who already have an affected child, they are more likely to experience guilt and self-blame, and change their reproductive plans compared to carriers without affected children. Additionally, some participants reported clinical features of the disorder for which they were being tested. Genetic counselors may erroneously

assume that parents with affected children are aware of their own carrier status in the absence of testing, and they may offer inadequate support. Additionally, counselors should attempt to address patient misconceptions related to their health and carrier status.

Keywords Systematic review · Carrier testing · Genetic · Psychosocial · Genetic counseling

Introduction

Variations in genetic material are inherent in all humans. While many of these variations do not change the protein product for which the gene codes, others may have a more deleterious effect (Addison and Brown 2007). In autosomal or X linked recessive conditions, one normal copy of the gene is usually sufficient to ensure the protein product is not adversely affected, however if individuals are heterozygous, having one normal and one mutated copy of the gene, they are said to be a “carrier” of the condition. The offspring of carriers could be at risk of inheriting the disease. A woman carrying an X linked recessive condition, such as fragile X or Duchenne muscular dystrophy, could pass the condition to her children (usually her sons) but in the case of autosomal recessive conditions, such as cystic fibrosis or thalassaemia, both parents need to be carriers of the same disease for their children to be at risk. Fragile X does differ from most other X linked conditions because premutation carriers (individuals with 55 to 200 CGG repeats (Kunquist et al. 2008)) can be mildly affected by the condition, can present with late-onset

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fragile X-associated tremor/ataxia syndrome (FXTAS) (Reis et al. 2008), and have an increased risk of premature ovarian failure (Hunter et al. 2008).

The term "carrier" is also used to describe individuals who have a balanced chromosome translocation. The translocation does not affect their own health, but their children could inherit an unbalanced form of the translocation that could have an adverse phenotypic effect (Maza-Espinosa et al. 2008).

Because of the potential reproductive implications of carrier status, individuals who are aware they could be carriers may wish to be certain of their own status. Carrier testing for a range of genetic diseases has been offered to individuals with a family history of the condition, or through community based screening programs in particular ethnic groups, for example community based screening for Tay Sachs disease in the Ashkenazi Jewish population conducted since the 1970's (Kaback et al. 1993). Population carrier testing has also been offered either regionally or nationally, for conditions including cystic fibrosis, thalassaemia and fragile X (Hartley et al. 1997; Kaufman et al. 2008; Metcalfe et al. 2008; Saugkijaporn et al. 2004; Zlotogora et al. 2009).

Numerous studies have assessed the psychosocial impact of carrier testing and living as a carrier (Anido et al. 2005; Childs et al. 1976; Kenen and Schmidt 1978; McConkie-Rosell et al. 2001). As early as 1976, Childs et al. highlighted a number of emotional issues experienced by carriers including anxiety, self-stigmatization, and concern for offspring. Since then, other studies have assessed the impact of carrier testing in relation to a number of variables such as the particular condition (Anido et al. 2005), the mode of inheritance (James et al. 2006), the population being screened (Axworthy et al. 1996) and the impact of gender (Martean et al. 1997). We conducted a systematic review to synthesize this body of knowledge. Carriers of fragile X were included in the review because a body of knowledge exists relating to the psychosocial effect of carrier testing on this group (Anido et al. 2007; Anido et al. 2005; McConkie-Rosell et al. 2000; McConkie-Rosell et al. 2001). Because female fragile X carriers can sometimes be affected by the condition, although in general more mildly, fragile X does differ from autosomal recessive and some other X linked conditions. However, because we were trying to ascertain similarities and differences across a range of different conditions, we felt it was relevant to include this group in the systematic review.

Specifically, the aim of this systematic review was to answer the following questions: (1) What are the factors affecting the impact of carrier testing results on individuals?

(2) What is the methodological quality of the body of literature examining the psychosocial effects of carrier testing? (3) Can we make any assumptions about the psychosocial impact of living as a carrier, based on studies assessing the impact of carrier testing?

Methods

In conducting this systematic review the methods described by Pope et al. (2007) which involve using specific search parameters, defining inclusion and exclusion criteria, and undertaking quality appraisal of the studies that are included, were used as a guide. Due to the wide range of methods, conditions and samples in the studies reviewed, we did not conduct a meta-analysis of the data.

Search Methods

The following databases were searched: CINAHL, Embase, Ovid, Medline, PsychINFO, Pubmed and Web of Science, using the following search terms:

carrier testing or carrier test* or carrier screening or genetic screening or population screening or cascade testing or heterozygote testing AND genetic or DNA or chromosome or autosomal recessive or recessive or X-linked AND depression or emotion or guilt or anxiety or worry or stress or blame or psychological or psychosocial or social or effect or impact or psychological impact or social impact or personal or carrier status or distress or relief or burden or coping or coping strategy or communication or coping behavior* or emotion* or stigma or self concept or attitude* or psychology or social adaptation or reproductive uncertainty or risk perception or genetic counselling or genetic counseling or carrier couples or family planning or prospective risk AND NOT children NOT zygote NOT prenatal NOT predictive

Limits were set on publication dates (January 1990 to May 2010), language (English), and population (Human; Age: Adult).

An author search and an ancestral search (reference search) were also carried out after relevant studies were identified. Keywords from relevant studies found this way were fed back into the search terms to ensure the search was thorough.

Inclusion and exclusion factors

Studies were included if they were:

- systematic reviews, literature reviews randomized controlled trials, quasi-experiments, observational studies, surveys or qualitative studies
- published between January 1990 to May 2010. We included studies published from 1990 onwards as around this time DNA carrier testing became feasible clinically for patients with a family history of recessive and X-linked conditions (Broide et al. 1993; Kerem et al. 1989). At the same time, studies that assessed the impact of the test on the patient began to appear in the literature
- focused on the psychological and social impact of the test result on the patient
- focused on either autosomal recessive and X linked conditions, or carriers of chromosomal changes such as translocations.

Studies were excluded if they were:

- about cancer, adult onset conditions or other dominantly inherited conditions, because the nature of the information derived from these tests will be different from receiving carrier information for recessive, X-linked or chromosomal conditions
- ones in which there was potential for participants to find out that they were homozygotes for a particular gene mutation where the age of onset of the disease was in adulthood (e.g., hemochromatosis)
- focused on pregnant women because their feelings may be influenced by worry for their offspring, and also because their decision to seek testing would be influenced by the immediate needs of a current pregnancy (Cheuvront et al. 1998)
- ones that included children or adolescents because it is likely that they will have very different psychosocial reactions and information needs than adults
- focused only on recall of information about risk
- focused only on motivation for taking not taking the test.

Search Outcome

The literature search generated 1694 articles for consideration. Following exclusion based on title and abstract, the full text of 41 articles was retrieved. An ancestral and an author search identified 10 further studies. After reading the papers in full, 31 studies were excluded because they did not meet the inclusion criteria, leaving 20 relevant

studies to be included in the systematic review. There were substantial differences in construct, design, measures, population and outcomes across the studies. In this section we cite one example of each particular design, measure or outcome studied. Table 1 contains a more detailed report of the characteristics of each study. Thirteen studies were quantitative, three were qualitative and four were mixed methods. Study designs comprised longitudinal studies (Becker et al. 1994), randomised controlled trials (Callanan et al. 1999), and cross sectional studies (Dunn et al. 2008). Samples from different populations including the general population (Henneman et al. 2002), high risk groups (McConkie-Rosell et al. 2003) of Jewish decent (Marceau et al. 1992) and women only (Anido et al. 2005) were included. Sample size varied from eight participants (Anido et al. 2007) to 2220 participants (Honnor et al. 2000). Data collection methods varied from questionnaires (Becker et al. 1994), to focus groups (Anido et al. 2005) and in-depth interviews (Williams and Schute 1997). Various measures were used, including the Spielberger State-Trait Anxiety Inventory (STAI) (Spielberger et al. 1970), the Health Orientation Scale (HOS) (Woodridge and Murray 1988) and the Tennessee Self Concept Scale (TSCS) (Fitts and Warren 1996). In two cases the same cohort of participants was involved in two studies, however because different findings were presented in the different papers, both studies were included in the review (Cheuvront et al. 1998; Newman et al. 2002).

Quality Appraisal

Each study was assessed using a quality appraisal tool developed by Kmet et al. (2004). The Kmet scale enables assessment of both qualitative and quantitative studies. This tool has proven internal validity and provides a systematic, reproducible and quantitative means of simultaneously assessing the quality of research encompassing a broad range of study designs. Using this tool, the first author scored each paper based on quality criteria including the description of the research question, appropriateness of design, justification of sampling strategy, appropriate data collection and analysis and estimates of variance (for quantitative studies) to produce a score phrased as a percentage. Five papers across the range of scores were selected and a blind appraisal was made by the second author to verify the results. The papers were ranked in the same order by both appraisers.

All 20 papers scored greater than 60% on the Kmet scale (range between 62% to 95%; median: 81%) and therefore none were excluded, on the basis of quality (Kmet et al. do not provide a 'cut-score' at which studies

Table 1 Summary of studies

Reference	Purpose of study	Study design	Sample and size	Data collection	Method of analysis	Results
Smith et al. (2007)	To explore the attitudes towards CE lessons learnt during their lifetime education from the general population	Qualitative, in-depth interviews	Eight women from the general population, of non-graduate age, who had been identified as CE users. April 21–01	Focus group discussions to explore their views on education in general, and to discuss their perceptions of their CE experience (including writing, experiential, practical or other skills). Effect of education on self-identity and health planning.	Interpretive phenomenological analysis. Topics identified. Exploring primary purposes for CE uptake. A list of individual activities, based on their responses, identified the categories to discuss (definition and characteristics of themes).	Women more actively engaged in their lifetime education. The more active the education was at the advanced CE level, the more active the women were in their planning and health activities. More advanced the education, the more active the women. For actively pursuing education to benefit a career progression. Pursuing education is positive, especially in anticipation of retirement. Health planning activities were also reported but by more women than in the early and middle
Smith et al. (2007)	To identify issues related to career seeking and education amongst the general population	Qualitative, Focus groups	General population and a sample from families with CE users. March 2007 (9–11 focus). Focus group participants less than 100	Focus groups were audio and video recorded (duration included). How would you describe your first reaction to getting started? How long had been since you started any other or more education than your life time? How can you make your life fulfilling and more enjoyable?	Thematic analysis. Topics identified. Primary purposes, social and individual issues (health, time, financial independence, etc). Healthier comparison. Themes were compared to existing literature in education and health.	Early life uptake from CE benefits increased with career path progression. Education of self reported as more complete – could be positive in terms of health planning, and could be more active. Healthier compared other respondents of more active health plans. Educational path experienced to avoid career. Taking of education with reported careers. No clear link between education uptake and education or career plans will be seen in health planning.
Walker et al. (1994)	To evaluate the short term effects of progressive health screening for coronary CE.	Longitudinal, study, 400 completed postal questionnaires completed at three time points.	Adults between 40–60 years registered with a general practitioner in Dorset, London. First followed the standard anti-smoking and health screening (5,526). This included an ECG screen (407), patients aged 40–70, female 125, male 1788. Second screen from 417 with complete responses 14 patients.	Questionnaires at three time points before taking, upon receiving results, three months later (T1), and at three years. Six activities with completion monthly after receiving test results.	MANOVA. Dichotomous analysis. No adjustment on how activities data were analysed.	Results of results had no effect on completion of tests. There is a reported a positive short term improvement seen in the first three months. This activity has improved. High proportion of participants taking screening which has been maintained or improved in action.
Collins et al. (1997)	To identify the psychosocial, psychological issues and reproductive plans of women by age related groups.	Experimental research that examining how education and working with a life insurance and savings.	CE groups using either in CE systems of previously identified samples of 1000 adults with CE CE systems. Income were varied (high/low). CE users, not complete living in stable relationships (N = 2000). Participants randomly assigned to either active or health screening.	Focus group sessions were audio taped, lasting and lasting each 45 minutes (N = 11). Used to examine the use and perceived value. Positive and negative effect (movement) using PSYSA.	Factor comparison t-test. Descriptive statistics used to summarise data.	Both education and their activities showed higher higher income women were working for education and savings, although women reported activity might be explained differences between the religion and culture in some time points. There were no differences in working status or results after completion of working. Significant positive of education appeared to change in reproductive plans.
Chewman et al. (1999) and Norman et al. (2002)	To look at perceptions and knowledge acquisition of low-ability population in relation to CE post primary education and career planning.	Two focused research that comparing basic education and learning with other education and working. Secondary analysis comparing current and N/A variables.	Two hundred adult women were given an ability of low education and working. April 1999–2000 completed between 10–45 years. Participants were women of people with CE living in N. Carolina. Participants randomly assigned into two groups before being recruited. First assigned to the class CE and then assigned to education and learning activities. 2000.	Focus groups completed a baseline telephone interview, and completed a questionnaire at two time points while waiting for their test results and immediately after learning their test results. T1,11 used to measure anxiety.	Data analysed using SPSS 15.5. Value of p < 0.01 was considered significant.	No significant differences in position or negative effect in anxiety with waiting for results after results were known as a result of their career had been education and health. Significantly significant differences in any of the outcomes. Education health outcomes were not significantly different between education and learning interventions.

Table 1 (continued)

Author(s) Year	Focus of study	Study design	Sample and size	Data collection	Measures of impact	Results
Boonick & Tansik 1998	Assessments of career readiness in 1,100 in the community college workforce	1,100 students, self-administered survey, survey of 100 faculty members	For students who completed and had not completed the survey, the mean scores were 3.14 and 3.08, respectively. For faculty members, the mean score was 3.14.	For a questionnaire, students rated on a 5-point scale (1 = not at all, 5 = very much) their agreement with the statement: "I am confident about my career future."	F ² score for comparison of responses between students who completed and did not complete the survey was 10.14, $p < .001$.	From a survey of 1,100 students in the community college workforce, 100 faculty members completed and analyzed the survey. The mean score for students who completed the survey was 3.14, and the mean score for faculty members was 3.14.
Carroll & Jones 2001	A review of faculty members' perceptions of the impact of leaving as a career choice	Focus on faculty members who had completed the survey	Interviews of 10 faculty members who had completed the survey	A review of faculty members' perceptions of the impact of leaving as a career choice	F ² score for comparison of responses between students who completed and did not complete the survey was 10.14, $p < .001$.	Faculty members who had completed the survey were interviewed. The mean score for students who completed the survey was 3.14, and the mean score for faculty members was 3.14.
Carroll & Jones 2001	Faculty members' knowledge of the impact of leaving as a career choice	Faculty members' knowledge of the impact of leaving as a career choice	Faculty members' knowledge of the impact of leaving as a career choice	Faculty members' knowledge of the impact of leaving as a career choice	F ² score for comparison of responses between students who completed and did not complete the survey was 10.14, $p < .001$.	Faculty members who had completed the survey were interviewed. The mean score for students who completed the survey was 3.14, and the mean score for faculty members was 3.14.

Table 1 (continued)

Reference	Purpose of study	Study design	Sample and size	Data collection	Method of analysis	Results
McIntyre-Roth et al. (1997)	A study of child gatekeepers of the EE system to evaluate caregivers and parents regarding carrier usage.	Mixed methods study using 49 parents' personal narratives.	Twenty-eight single carriers was recruited through the Eagle XC line at Duke University Medical Center. All carriers had undergone genetic counseling and all their first wives passed. Twelve first partners were carriers. There left residual carriers must age 17.8 years, most number of children 2.9, mean number of years since diagnosis 7.8 years.	Structured interview with open and closed-ended multiple choice questions at end (one week). Interviews were audio-taped, lasted 30-45 min, covering carrier planning issues. Two interviews should be held at parents' request and family characteristics. After included as 11 carrier XAs.	Thematic and open analysis. Fisher's F test due to the size of 315 frequency tables or tables used for chi-square, however, none.	As a carrier parents felt a meaning about the condition and changed their plans after being diagnosed. They were nervous but that it was not known whether they could have a child before the age of their partner or not knew that any biological children. Eighteen percent said they would have used genetic diagnosis because of nervous and during this time carriers had changed the way they lived of themselves. 17% of a partner was 47% in a negative way. Partners reported that carriers were a new beginning in the intensity of the support feelings associated with the learning about carrier. Eighty-seven percent indicated they did not feel guilty because of their partner's illness or diagnosis and would change their carrier plans. It was stated that their partner would a change in relationship with the partner. 27% a partner change. 17% a negative change. They percent reported satisfaction in relationship with siblings.
Neuman et al. (2002)	To assess how genetic at will as carrier usage, reproductive choice and their family affected carrier usage.	Randomized controlled trial comparing those who used and those who did not undergo and testing.	In a randomized trial, 100 carriers were recruited. 50 carriers received 10-40 day. Participants were subjects of genetic with CE living in N. Carolina. Participants randomly selected into two groups: those assigned to receive CE and those assigned to education and testing of carrier (CE).	Participants completed a baseline questionnaire, and completed a questionnaire at two-time points, while waiting for their results, and immediately after learning their test results.	All reported symptoms reported were conducted using SF-36V2 and SF-36V2 (not shown) analysis.	Women reported higher scores than men on the SF-36 subscales of function, vitality and well-being. After learning to carrier, differences with their partner adjustment was lower with waiting for test results.
Patterson et al. (2001)	A study examining family CE psychological distress.	Longitudinal, self-completed mailed questionnaire survey at six time points.	Family participants who had been diagnosed with sickle cell disease. Between 16-47 years old, white. Between 16-47 years of CE was included. Twelve women completed questionnaire at baseline and 17 women completed questionnaire at baseline and one month completed the questionnaire at three months.	All questionnaire questionnaires at baseline and three months plus testing for results.	Tests: Mann-Whitney U, Health Transition Scale (McNamee and	Diagnosis of the wife increased CE psychological distress over the year following that time. The carrier had the the most psychological difficulties. Non-carrier family members might be more angry and hospitalized than the CE. Women in three months after learning that carrier of sickle cell was essentially equivalent three months after learning the wife was carrier.
Waters et al. (2001)	A study assessing the effect of learning the CE carrier status on parents health, attitudes, knowledge and access of healthcare.	Longitudinal self-completed postal questionnaire of New York parents.	Participants (non-carrier and carrier age 18-40) completed baseline survey health and attitude. Data showed that carrier and carrier was individuals were without identifying CE carriers at 10 carrier times at 17. Eighty-eight percent completed questionnaire at Time 1. Carrier's current satisfaction in questionnaire at Time 1 was 1.81 (on sample 0-10) which indicated at Time 1 was not poor.	All questionnaire questionnaires. SF-36 used to measure anxiety. Time 1 was one year. Time 2 was two weeks after results. Time 3 was two weeks after two months after the CE results.	Stepwise statistics were conducted.	Eighty-one percent of parents said they were satisfied but their third carrier was diagnosed. 70% of the CE was 17% and the non-carrier 10% percent. Psychological effects of diagnosis and genetic counseling helped 17% and 17% respectively. Twenty-one percent were married (some still waiting for results of partner). At 17 months, 70% reported to parents of diagnosis. There was planning further children was possible. An increase in satisfaction after two months. Eighteen percent of carriers did not use a w-

Fig. 1 Mindmap showing relationship between key themes



Results

The impact of carrier testing for cystic fibrosis was the condition most commonly investigated, with ten studies focusing on this condition, followed by carrier testing for fragile X (five studies), Tay Sachs (one study) and hemophilia A and B (one study). The focus of the remaining three studies was the effects of carrier testing for a number of conditions. No studies which assessed the impact of carrier testing on carriers of chromosomal abnormalities were identified from the literature review. Carrier testing for people who had a family history of a genetic condition (and were therefore at an increased risk) was assessed in 11 studies, risk in the general population was assessed in seven studies, and in two studies people in both groups were assessed. Only three of the studies included in the systematic review were intervention studies (Callanan et al. 1999; Chevroux et al. 1998; Newman et al. 2002). All three compared levels of anxiety related to home education and testing with clinic education and testing. A number of overarching themes were identified. The most prominent were anxiety, guilt, relief, effect on self image, active coping mechanisms, impact on reproductive issues and disclosure of test results (Table 2).

Anxiety

Two categories of anxiety emerged: one related to testing and the other related to child health. In relation to testing, all longitudinal studies investigating parent anxiety over time either found no significant difference in anxiety between carriers and non-carriers (Hinnor et al. 2000), or

found that any anxiety experienced by carriers upon first receiving their test result had, for the vast majority, dissipated by six months as assessed by the state STAI (Bekker et al. 1994; Callanan et al. 1999; Chevroux et al. 1998; Lakeman et al. 2008; Watson et al. 1992), the Fragile X Visual Analog Scale (VAS) (McConkie-Rosell et al. 2001), or qualitative interviews (Anido et al. 2007; Anido et al. 2005).

Carrier anxiety dissipated for a number of reasons. Watson et al. (1992) found that the provision of written information and genetic counseling was helpful for most participants (92% and 97%, respectively). Bekker et al. (1994) found that the passage of time appeared to dissipate anxiety. Gender was also an issue discussed in relation to anxiety, in a number of studies. Newman et al. (2002) and Henneman et al. (2002) found that women reported higher anxiety than men while waiting for their test results (mean = 16.5 and 14.6, respectively on the STAI in the Newman study, $p < 0.001$, and 24% versus 13%, $p < 0.001$ measured on a five-point Likert scale in the Henneman study); however there was no significant difference between the genders once the test results had been received. Lakeman et al. (2008) found that Western participants generally reported lower levels of anxiety compared with non-Western participants (General Linear Model analysis at 4 time points, $p < 0.001$).

Anxiety did however appear to be an issue for both carrier and non-carrier siblings of people with cystic fibrosis, in the interview-based study conducted by Fanso and Johnson (1995b). Identified carriers and non-carriers were equally likely to have moderate or severe anxiety around their child's health. Forty-one percent had had their

Table 2 Themes by mode of inheritance

	Autosomal recessive		X-linked	
	Family history	Population	Family history	Population
Anxiety/disappointment by 6 months	Callanan 1999 Chatterton 1998 Newman 2002	Bekker 1994, Henneman 2002, Honor 2000, Lakeman 2008, Watson 1992	Anido 2005*, McConkie-Rosell 2001	Anido 2007
Guilt	Williams 1997*		Anido 2005*, Dunn 2008*, James 2006*, McConkie-Rosell 1997*, Williams 1997*	
Altered perception of health	Fanos 1995, Martens 1992*	Bekker 1994, Henneman 2002, Martens 1992	McConkie-Rosell 2000	
Evidence of self-stigmatisation		Gordon 2003	James 2006*, McConkie-Rosell 2000, McConkie-Rosell 1997*	
Evidence of social-stigmatisation		Gordon 2003	McConkie-Rosell 2001	
Use of active coping mechanisms			Anido 2005*, McConkie-Rosell 2001	Anido 2007
Impact on reproductive plans			Anido 2005*, Dunn 2008*, McConkie-Rosell 1997*	Dunn 2008*
No impact on reproductive plans	Callanan 1999	Henneman 2002, Watson 1992, Lakeman 2008		Anido 2007
Disclosure of test results to family	Williams 1997*	Henneman 2002, Watson 1992	McConkie-Rosell 1997*, Williams 1997*	Anido 2007

Family History – Participants with a family history of the condition

Population – Participants identified from the general population

* Cohort included participants with affected children

children sweat-tested to rule out the condition, nine percent had had their child tested for carrier status, and 55% planned to do so before their child reached 18 years of age. Siblings who had had their children sweat-tested or tested for carrier status were equally divided between those who knew their own carrier status and those who did not.

Guilt

Guilt was a prominent theme in the data. Feelings of guilt associated with carrier status were cited as findings in five studies. These results were identified through interviews (Anido et al. 2005, McConkie-Rosell et al. 1997, Williams and Schutte 1997), an open-ended questionnaire (Dunn et al. 2008), a VAS (James et al. 2006, McConkie-Rosell et al. 1997) and the guilt subscale of the Multiscore Depression Inventory (sMDI) (James et al. 2006). Guilt is also an issue found to be closely associated with gender, mode of inheritance, and whether the participants had affected children. In the study conducted by Dunn et al. (2008) in which 81% of respondents had a son with hemophilia, 18 of

48 (38%) female carriers reported the timing of testing as negative. Reasons cited for the timing being negative included feeling blamed by their partner and a prolonged sense of guilt. James et al. (2006) found that mothers who were carriers of X-linked conditions felt substantial guilt and self-blame related to their child's condition. When measured on the VAS, mothers of children with X-linked conditions had significantly higher levels of guilt than mothers of children with recessive conditions ($p < 0.01$) and were more likely to blame themselves ($p < 0.001$). A similar finding was identified in the Williams and Schutte study, in which it was found that many of the participants who expressed feelings of grief and guilt were women who were carriers of fragile X or Duchenne muscular dystrophy. Anido et al. (2005) also found that in families affected by fragile X, even those women without affected children experienced guilt to some extent, by virtue of the condition being in the family.

In one study (Gordon et al. 2003) there was no significant difference evident between carriers and non-carriers on the "guilt" scale (as measured by the HHS). The participants were from the general population, were

screened for cystic fibrosis, and did not have a family history of the condition.

Relief

Anido et al. (2005), McConkie-Rosell et al. (1997) and Lakeman et al. (2008) all identified that relief was an emotion experienced by carriers. In the study conducted by Anido et al. reactions of relief were expressed equally as strongly as reactions of guilt, with nearly all carriers expressing this emotion during interviews. For these individuals, finding out their carrier status was an inevitable result of finding a diagnosis for their child. Similarly, in the study by McConkie-Rosell et al. (1997), participants' responses indicated that while they felt angry or depressed about their carrier status, there was an "emotional relief in finding out the cause of the mental retardation in the family" (p. 65). Lakeman et al. (2008) found that 68% of participants, including seven out of ten carriers, felt relief one week after receiving their test results, as measured on a structured questionnaire assessing emotional outcomes.

Effect on Self-image

Three main issues arose within this theme: perception of health, self-stigmatization and social-stigmatization.

Perception of Health

Of the seven studies in which perception of health was measured, findings from three studies indicate that some carriers believed their current or future health to be significantly poorer after learning their carrier status (Fanos and Johnson 1995b; Henneman et al. 2002; Marteau et al. 1992). Seven out of 17 carriers (41%) in the study conducted by Henneman et al. (2002) felt less healthy (measured on a multiple-choice questionnaire) due to their test results, despite being informed both verbally and by letter that their carrier status would have no effect on their own health. Marteau et al. (1992), also using a multiple-choice questionnaire which measured perceived health from three time perspectives, identified that carriers of Tay Sachs held the least optimistic view of future health compared with non-carriers and the control group ($p < 0.01$) and Fanos and Johnson (1995b) reported that during interviews, sibling carriers retrospectively redefined health problems as related to cystic fibrosis; although the authors do not report how many.

Authors of four studies found that perception of health did not alter after learning one's carrier status, using measures such as the Tennessee Self Concept Scale (McConkie-Rosell et al. 2000), a multiple choice questionnaire (Bekker et al. 1994), the HOS (Gordon et al. 2003)

and a five point Likert-scale (Lakeman et al. 2008). However, both McConkie-Rosell et al. and Bekker et al. do provide some anecdotal evidence to suggest that carriers might attribute previous health problems to their carrier status. McConkie-Rosell et al. found that 12% of participants at Time 1 and 20% of carriers at Time 2 reported feeling they had mild clinical features of fragile X. They felt that perhaps if they were carriers it would explain why they had to "study hard in school" (p. 340). A participant in the Bekker et al. cohort wondered whether her allergies and chest colds were in some way linked to her carrier status. Therefore, even though perception of health did not alter when measured quantitatively, during qualitative interviews there were some indications that it did in fact occur in a small number of cases. In fact, in the case of fragile X, it is possible that carriers did experience a mild manifestation of the disease due to skewed X-inactivation (Skirton et al. 2005). Furthermore, this finding may also be attributable to the repeat length itself which appears to be associated with toxicity due to elevated mRNA levels (Koldewyn et al. 2008).

Self-stigmatization

There is evidence from four studies to indicate that self-stigmatization occurred in carriers to some extent (Gordon et al. 2003; James et al. 2006; McConkie-Rosell et al. 1997, 2000). Gordon et al. identified that carriers experienced less positive feelings; more afraid, worse, weaker, less relieved, less happy, more marked (although the authors do not explain what is meant by this) and angrier, compared to those who tested negative, on the HOS. Similarly, James et al. found that carrier status is associated with stigma and is significantly associated with mode of inheritance using the same scale. The only other study (Pastore et al. 2008) specifically looking at stigma using the HOS consisted of just one carrier, and therefore findings were not significant. Stigma was also evident in two of the qualitative studies. Just under half (9/19) of the fragile X carriers in one study (McConkie-Rosell et al. 1997) indicated that there had been a negative change in the way that they viewed themselves. One reason cited for this change was a "feeling of being abnormal or inferior" (p. 64), a statement indicative of self-stigmatization.

Social Stigmatization

Evidence of social stigmatization was evident in two studies, one quantitative (Gordon et al. 2003) and one mixed methods (McConkie-Rosell et al. 2001). Gordon et al. found that carriers and non-carriers attributed significantly more negative feelings to cystic fibrosis carrier status than non-carrier status. This finding was significant for all emotional scales on the HOS ($p < 0.001$).

Active Coping Mechanisms

Use of active coping mechanisms was identified in five studies, out of a possible seven studies in which qualitative research techniques were employed. These studies included participants from the general population without affected children (Anido et al. 2007) and participants with a family history (Anido et al. 2005; McConkie-Rosell et al. 1997, 2000, 2001). McConkie-Rosell et al. (2001) found no change in the level of distress or perceived seriousness of fragile X when women were "at risk" of being a carrier as when they were found to be carriers. The increase in perception of seriousness only occurred in the non-carriers when the threat was no longer present. This possibly indicates that "threat minimization" was used by the participants as an active coping mechanism in both situations. McConkie-Rosell et al. (2001) also found, during in-depth interviews, that 11 out of 20 (55%) carrier women used spontaneous coping statements such as "life goes on" (p.41) and "If I am, I am, I'll deal with it" (p.41). Coping behavior statements were also evident during interviews in the study conducted by Anido et al. (2007).

For carriers identified in the study by Anido et al. (2007), most appeared to be considering their carrier status over the course of the interview, having not given the subject much thought previously. The authors postulated that this attitude is consistent with the coping mechanism known as "just-in-time" learning, as described in Adult Learning Theory (Wlodkowski 1999), wherein adult learners process information which is relevant and applicable to them at the time they need it.

Impact on Reproductive Issues

The impact of carrier status on participants' views on reproductive issues varied depending on their life stage, their views on prenatal testing and abortion, whether their partners were also carriers, and whether they were carriers of an X-linked or recessive condition. Authors of four studies (Callanan et al. 1999; Henneman et al. 2002; Lakeman et al. 2008; Watson et al. 1992) of cystic fibrosis carriers identified from both high risk groups and the general population who did not have affected children, all reported that the majority of carriers showed no change in reproductive plans after testing, as measured on questionnaires which included multiple-choice options (Callanan et al. 1999; Watson et al. 1992) or a five point Likert-scale (Henneman et al. 2002; Lakeman et al. 2008). Reasons given included the availability of prenatal diagnosis (Henneman et al. 2002; Lakeman et al. 2008; Watson et al. 1992) and having completed their families (Watson et al. 1992). Furthermore, in two of the studies (Chevrant et al. 1998; Henneman et al. 2002), only carrier by non-carrier

couples were included. If one partner tests positive and the other negative, the risk of having a child with CF is about 1 in 640 (Watson et al. 1992).

However, in two interview-based studies, females carrying X-linked mutations, many of whom were mothers of affected children, were more likely to indicate their carrier status had caused a change to their reproductive plans (Anido et al. 2005; McConkie-Rosell et al. 1997). In the study conducted by McConkie-Rosell et al. (1997) 19 out of 28 (67%) fragile X carriers stated that they would not have any more children because of their carrier status, and 25 out of 28 (89%) would have either reduced the size of their families or not had any biological children, if they had known earlier. Anido et al. (2005) also found through in-depth interviews that many women with fragile X children stopped planning to have more children after receiving their test results. Furthermore, those without affected children expressed a strong desire "to figure out a way to end it with me" (p. 501). Dunn et al. (2008) also reported findings from open-ended questions that revealed some respondents felt they might not have had as many children if they had known their carrier status earlier.

Findings differed however, in the study conducted on fragile X carriers identified from the general population (Anido et al. 2007). Many carriers expressed that although the information could be relevant in the future, it was not relevant at this stage of their lives in terms of family planning. Some had not really considered the implications for family planning and their thoughts about prenatal testing, but for those that had, carrier status did not have an apparent effect on their attitudes about termination. The issue of premature ovarian failure appeared to be more prominent than the risk of having children affected with fragile X.

Disclosure of Test Results and Family Relationships

In six studies in which disclosure of test results was assessed, the researchers found that participants did share their test results with others, although this disclosure was selective (Anido et al. 2007; Dunn et al. 2008; Henneman et al. 2002; McConkie-Rosell et al. 1997; Watson et al. 1992; Williams and Schutte 1997). Anido et al. (2007) found that providing information to partners primarily depended on the seriousness of the relationship. Watson et al. (1992) found that 89% (47/53) of CF carriers informed their partners of their test results, 83% told their parents, 82% their siblings and 48% told other relatives. Henneman et al. (2002) reported that most CF carriers shared the information with parents and siblings. All but one of the carriers whose parents were still alive had told them about their test results. Ten carriers had shared the information

with their brothers and sisters, but two had not. With respect to participants who did not disclose carrier information to other family members, their reasons included not wanting to disclose results to relatives who had affected siblings, and not wanting to cause feelings of guilt (Williams and Schutte 1997).

The effects of sharing information about one's carrier status with a partner and/or family members varied across the studies. Positive experiences related to disclosure of test results were documented by Dunn et al. (2008) and McConkie-Rosell et al. (1997). Of the 18 carriers who indicated in a change in their relationship with their husband in the McConkie-Rosell et al. (1997) study, 13 carriers (72%) indicated this change had been positive. Seventeen (61%) felt that there had been an improvement in their relationship with their siblings. Difficult or distressing experiences were highlighted in three studies (Dunn et al. 2008; McConkie-Rosell et al. 1997; Williams and Schutte 1997). Dunn et al. and McConkie-Rosell et al. identified a negative effect on the relationship with the partner in 13/31 (42%) and 5/18 (27%) of cases, respectively. Reasons cited included anxiety and anger from the male partner (Dunn et al.) and feeling blamed by their spouse (McConkie-Rosell et al.). In cases where the experience had a positive effect on the relationship (in 4/31 and 13/18 of cases respectively), the carrier felt completely accepted by her partner (Dunn et al. 2008) and there was an increase in understanding and communication. Henneman et al. (2002) found the majority of participants (98%) perceived no impact of carrier testing results on the relationship with their partner. For the majority of participants in the Anido et al. (2007) study, providing information about fragile X carrier status to family members was not problematic. However, providing the information to partners depended on the seriousness of the current relationship.

Discussion and Conclusion

Discussion

This review is useful in that it identifies a number of factors that seem to influence the emotional consequences of carrier testing. These include population group, whether the carrier has an affected child, stage of life, psychological coping mechanisms, and mode of inheritance. In this respect the results of this systematic review provide some interesting insights into how genetic testing for different conditions may have a varying psychological impact that is dependent on the context in which testing occurs.

Anxiety, an emotion frequently measured in studies investigating the impact of carrier testing on individuals, dissipated in the long term for the majority of participants

in all studies. In addition, the reasons suggested by authors, another reason may have been because none of the participants were pregnant at the time of receiving their carrier test results and were therefore not anxious about the possibility that the fetus was affected. For carriers, knowledge that reproductive options were available to them if there was a risk of having an affected child may also have overridden any initial anxiety. Furthermore, good quality genetic counseling services may have lessened the impact of the test results.

Variables including mode of inheritance, gender and whether the carrier already had a child affected by the condition appear to be strongly linked to the issue of guilt. The finding that guilt was more dominant in women than men, indicates that it may be strongly connected with what Peters and Jackson (2009) describe as a unique emotion concerning a mother's relationship with her affected child. Guilt also appeared to be more commonly reported by mothers of children with X-linked conditions. One possible explanation lies in the close association of guilt and blame. In the case of X-linked conditions, it only takes a carrier mother to pass along an X-linked condition rather than having both parents contribute the "faulty" gene. Therefore the burden of having passed on a faulty gene cannot be shared with a partner. In these cases men may "externalize their emotional response to devastating news and blame, while women are likely to internalize their responses and to accept this blame" (James et al. 2006). Mothers are also more likely to self-blame (Peters and Jackson 2009).

Guilt may also be an emotion linked to family history. Anido et al. (2005) found that women who did not have affected children but had the condition in the family, expressed feelings of guilt, which may indicate a form of "survivor guilt." Survivor guilt has also been identified in CF families. In a study (which was excluded from this review as it contained women who were pregnant at the time of testing) in which barriers to carrier testing for adult cystic fibrosis siblings were identified, carrier status served an important function in binding guilt, with 15% of siblings either hoping they were carriers or feeling guilty they were not (Fanos and Johnson 1995a).

There are conflicting results in the literature regarding the issue of perceptions of health. Yet even those studies in which carriers did not indicate feeling less healthy on surveys or questionnaires, during in-depth interviews, some participants reported clinical features of the disorder for which they were being tested or for which they were found to be a carrier. In the case of fragile X, it is possible that carriers did experience a mild manifestation of the disease due to skewed X-inactivation (Skirton et al. 2005). Furthermore, this finding may also be attributable to the repeat length itself which appears to be associated with

nervosity due to elevated mRNA levels (Koldewyn et al. 2008). However this finding also suggests that participants may have been seeking support for beliefs they held about themselves.

In interpreting this finding, McConkie-Rosell et al. (2000) refer to the theory of self-concept as described by Shavelson et al. (1976). Shavelson et al. hypothesize that self-concept is hierarchical, with perception of personal behavior in specific situations at the base of the hierarchy, inferences about the self in broader domains (e.g., social, physical) at the middle, and a global, general self-concept at the apex. Global self-concept is stable, but as one descends the hierarchy self-concept becomes increasingly situation specific and less stable. Seeking clinical features related to actual or possible carrier status might be indicative of situation-specific changes in feelings about self. Additionally, it may be the case that scales such as the IOS (used by Gordon et al. 2003) and TSCS (used by McConkie-Rosell et al. 2000) are not sensitive enough to detect the subtleties concerning how carriers perceive their own health, which are more likely to be expressed during in-depth interviews.

Reproductive intent also appeared to be closely linked to mode of inheritance, stage of life and whether the participant already had an affected child, with the greatest impact being identified for carriers of X-linked conditions with affected children. This group was most likely to refrain from having more children. One possible reason involves the documented psychological difficulties of raising a child with fragile X (Abbeduto et al. 2004; Lewis et al. 2006). When Anido et al. (2007) interviewed fragile X carriers who did not have affected children and were from the general population, the information did not appear to have an impact on family planning with many not having considered the issue. This is likely to be because they did not have any experience, either themselves or through other family members, of raising a child with the condition. It may be that these carriers would experience increased distress as they consider reproduction more seriously. Similarly, carriers of cystic fibrosis in the general population did not change their reproductive plans as a result of their carrier status. Participants in these studies did not have affected children, and even as a carrier, there would only be a risk to future children if the partner was also a carrier.

Active coping mechanisms, such as "threat minimization," significant changes to reproductive intentions and the use of active coping statements, were identified in those participants at an increased risk of carrying the fragile X gene. Lazarus and Folkman (1984) describe coping as consisting of two different strategies, problem-focused coping and emotional-focused coping. The findings from this systematic review suggest that women at high risk of being a carrier of fragile X engaged in problem-focused

coping by managing their health threat through genetic testing, and if found to be carriers, by changing their reproductive intentions. They engaged in emotional-focused coping through threat minimization and active coping statements.

In addition to these coping strategies aimed at lessening distress, Lazarus and Folkman describe a smaller group of cognitive strategies directed at increasing distress. For some individuals, there is a need to feel worse before they can feel better. Self-blame, a coping mechanism found to be used by carriers of X-linked conditions, is one such form of self-punishment individuals may use. This deliberate emotional distress may mobilize individuals into action. Evidence that women use self-blame as a coping strategy has been identified in other studies; for example, self-blame was found to be significantly correlated with both problem-focused and emotional-focused coping strategies in a study of patients with diabetes (Tuncay et al. 2008). Self-blame was also used as a strategy to cope with depression in a study of how primary care patients manage their illness (Broseth et al. 2007).

Other studies, in which participants became aware of their carrier status through family history or newborn screening, have identified similar psychosocial issues to those in this review. Famos and Mackintosh (1999) recognized a number of coping mechanisms used by parents of children with ataxia-telangiectasia, including rationalizing their child's condition as a "statistical quirk" (p.417), and imbuing the occurrence with meaning and significance through connecting it with the wider sphere of human suffering or to the spiritual world. Guilt was not however a common finding in their study, and surprisingly when it was mentioned, it was in reference to fathers. Uptake concerns about the health of carriers was also identified in a minority of parents in a study assessing the impact of carrier status information following newborn screening (Kai et al. 2009), as was a sense of responsibility to share carrier status information with extended families. Stigmatization was also evident in a study which included participants from high risk CF families who did not want to learn their carrier status (Famos and Johnson 1995a). For example, one untested woman was worried that she would be "less desirable" (p. 88) to men if they knew she was a carrier.

While this review provides an overview of the psychosocial experience of living as a carrier, it is important to keep in mind the limitations of making comparisons across different conditions, in particular cystic fibrosis and fragile X (the major conditions included in this review). These two conditions vary greatly in terms of their effects on the affected individual, the implications for the health of the carrier, and risk of the carrier having an affected child. Furthermore, variations in study design, the different population subsets compared, and the obvious complexities

of comparing qualitative and quantitative data, mean that the findings should be interpreted with some degree of caution. For example, there were indications from some studies using validated scales of no changes in perception of health. However, when the authors used in-depth interviews, changes in health perception were evident (Bekker et al. 1993; McConkie-Rosell et al. 2000). Some authors used the STAI to measure anxiety, whereas others using qualitative methods relied on participants' own terminology. Studies using the HOS were much more likely to identify evidence of stigmatization than those that did not use this scale, as this scale specifically measures aspects of self image. Future systematic reviews may therefore benefit from the inclusion of samples involving population groups which are more similar in kind in terms of risk to offspring, severity of the condition or family history. Future research studies may be better summarized if the studies focus on using similar groups of parents and validated tools.

Yet this does not necessarily mean the findings of the present review fail to provide valuable insight into the psychosocial experience of living as a carrier. In particular, the review provides an overview of the commonality of experiences across conditions with different inheritance patterns. Furthermore the overview identifies a number of issues that collectively apply to carriers as a group, because of the familial nature of genetics.

Strengths and Limitations

As stated previously, findings from the review should be considered in light of the difficulties and limitations of combining studies undertaken with different study designs, subsets of the population, measures and outcomes. These factors may have diluted the strength of the comparisons. Furthermore, many of the studies lacked theoretical models or presentation of a conceptual model to help place the variables and their possible interactions in context (Hermeman et al. 2002; Pastore et al. 2008; Watson et al. 1992). Such omissions possibly weaken the validity of the results. Nevertheless, in the present systematic thematic analysis, the findings were able to be explained within established theoretical models of coping and self-concept (Lazarus and Folkman 1984; Shavelson et al. 1976).

The systematic review does have notable strengths. Seven databases were used to retrieve studies to maximize the chance of finding all relevant research. In addition, several iterations of the search were conducted using different combinations of keywords, to ensure the search was rigorous. At the present time there does not appear to be another systematic review in the literature that compares the psychosocial experience of carrier testing for autosomal recessive and X linked conditions, thus, this review provides unique and useful information.

Conclusion

The findings from this systematic review provide insight into the variety of psychosocial emotions experienced by individuals undergoing carrier testing and a general overview of the psychosocial impact of living as a carrier. Prominent themes that occur in the literature include anxiety, guilt, relief, effect on self image, active coping mechanisms, impact on reproductive issues and disclosure of test results. Variables that influence the psychosocial effects of carrier testing include whether the carrier has an affected child, mode of inheritance, genetic counseling and life stage. A key finding concerns the different emotions experienced by carriers who already had an affected child compared with carriers who did not. Studies indicated that carriers with affected children were more likely to experience guilt and self-blame. Furthermore, fragile X carriers with affected children were more likely to indicate that carrier status had affected their reproductive plans. In contrast, carriers identified from the general population did not change their reproductive plans as a result of their carrier status. Due to the commonality of experiences identified through this systematic review, it would appear that we can make certain assumptions about the psychosocial impact of living as a carrier. Yet at the same time it is important to bear in mind the limitations of making generalizations across different population groups and condition types.

Practical implications

Genetic counselors and other health professionals offering genetic testing should pay attention to the variety and complexity of psychosocial experiences that may be encountered by individuals undergoing carrier testing. One key finding from this systematic review is that carriers who already have an affected child often react differently when receiving their test results than carriers who do not. For those carriers who already have an affected child, the impact of receiving the test results in these cases may reinforce feelings of guilt, self-blame and maternal blame in the case of X linked conditions. Counselors therefore need to be aware of these issues when testing parents of affected children as these psychological issues may need to be addressed both before and after testing. In addition, counselors should look to address misconceptions related to health and carrier status; some individuals may seek support for beliefs they have about their health by identifying clinical features of the disorder for which they are being tested or are found to be a carrier. Furthermore, while some clients will effectively manage anxiety and their carrier status through threat minimization and other active coping mechanisms, professionals should ensure that those who appear to be managing well do not

minimize their threat to the extent that they discourage from protective health actions, particularly when it comes to reproductive issues.

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