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PRESYMPTOMATIC TESTING FOR FAMILIAL CANCER SYNDROMES IN YOUNG ADULTS: CONSIDERATIONS, DECISION MAKING AND IMPACT

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PRESYMPTOMATIC TESTING FOR FAMILIAL CANCER SYNDROMES IN YOUNG ADULTS: CONSIDERATIONS, DECISION MAKING AND IMPACT

by

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ABSTRACT

PRESYMPTOMATIC TESTING FOR FAMILIAL CANCER SYNDROMES IN YOUNG ADULTS: CONSIDERATIONS, DECISION MAKING AND IMPACT

Lea Godino

Background: Presymptomatic genetic testing should always involve a considered choice. Young adults are at a key life stage as they may be developing a career, forming partnerships and potentially becoming parents. Presymptomatic testing may therefore affect the future lives of consultands significantly when testing is undertaken in early adulthood.

Aim: To explore presymptomatic testing for hereditary cancer in consultands aged 18-30 years with particular reference to psychosocial impact, the decision-making process and the consequent counselling needs.

Methods: A mixed-methods sequential exploratory design was used, comprising a systematic review, a qualitative study and a quantitative study. Results of all phases were used to build a theoretical model regarding the process of presymptomatic testing in young adults.

Findings: The systematic review indicated that many participants grew-up with little or no information concerning their genetic risk. The experience of genetic counselling was either reported as an opportunity for discussing problems or associated with feelings of disempowerment. Parents appeared to have exerted pressure on their children during the decision-making process. However, as a result of the qualitative study, the influence of other people and the decision-making process prior to counselling were identified as key factors. Further results from the quantitative phase underlined that parents felt they had control over the decisions their children made, while the majority of the young adults reported the request for the genetic test as their own decision. A new theoretical model of decision making and impact on young adults was built to synthesise the overarching experience of participants in this research project.

Conclusion: Counselling approaches to this population may require modification both for young adults and their parents. Young adults may benefit from a multi-step approach to presymptomatic testing. Parents need to be more informed that genetic counselling is a forum where information can be obtained and young adults can talk about the testing decision, regardless of whether they want to be tested or not. The traditional 'wait until they come to us' approach by health services may be failing to meet the educational and emotional needs of this population.

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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 without prior agreement of the Graduate Committee.

Work submitted for this research degree at Plymouth University has not formed part of any other degree either at Plymouth University or at another establishment.

During the course of the doctoral study a number of relevant postgraduate courses were attended to gain transferable and research skills including a Joanna Briggs Institute course on systematic reviews. Several courses facilitating the use of NVivo software were attended. A summer course in Manchester was attended to better understand the philosophy and processes of qualitative research. Three university courses in statistics (statistical inference, sampling methods and data analysis) by University of Bologna were attended. These are recorded in the Graduate School logbook.

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CHAPTER ONE

INTRODUCTION

1.1 INTRODUCTION

In the past, medical genetics was a small specialty, providing services to a limited number of families with rare conditions (Neri and Genuardi, 2010). However, the study of genetics in the health care context is important to a wide range of health-care providers in Europe as an estimated 30 million people now suffer from genetic diseases (Cassiman, 2005). As a result of the expansion of genetic knowledge and application of genetics in mainstream health care settings, genetic counselling activities have to be offered by a wider group of health professionals in order to provide genetic information, education and support to patients and their families at risk of genetic problems (Frazier et al., 2004; Lashley, 2005; Godino and Skirton, 2012). An important patient group for whom genetic services should be provided are those families at risk of being affected by an inherited cancer syndrome (Evans, 2006).

The focus of this doctoral study was the exploration of psychosocial implications of a specific group of clients (young adults) accessing presymptomatic testing for cancer. Specifically, I will address considerations

made by young people undergoing a test, the decision making process and the impact of testing. In this doctoral dissertation, the term *client* will be used instead of *patient* to refer to all individuals undergoing genetic counselling, as suggested by the American Nurses Association and the International Society of Nursing in Genetics (2016). *Patient* may have connotations of illness, while by using the term *client* I include individuals, and families with or at risk for a genetic condition.

In this first chapter, I will set the scenario for the doctoral study. I will begin by providing a brief overview of the definition of a young adult, followed by an introduction to genetic counselling and genetic testing. Genetic counselling in the Italian scenario, in terms of who provides genetic counselling, will be described. Previous research focusing on the psychological impact of presymptomatic genetic testing will then be presented, followed by a discussion of the rationale behind this body of research. Finally, I will outline the three phases of this doctoral study and provide a brief description of each chapter in the thesis.

1.2 DEFINITION OF YOUNG ADULT

The focus of this doctoral study, as described above, was the way in which young adults made testing decisions, what they considered before making the decision and the impact of presymptomatic testing for inherited cancer syndromes on those young adults. It is therefore important to define the term 'young adult'. The definition can be extremely broad and is not clearly defined in terms of one specific age group. For example, young adults have been defined as persons in their late teens, twenties and thirties who

represent diverse cultural, racial, ethnic, educational, vocational, social, political, and spiritual backgrounds. They could be college students, workers and professionals; they could be persons in military service, they could be single, married, divorced, or widowed; they could be with or without children; they could be newcomers in search of a better life (United States Conference of Catholic Bishops, 2016).

Although, the definition of a young adult is not often clear, Rindfuss (1991) defined young adults as aged between 18 and 30 years old, because 18 years is an age that is often recognized in law (i.e. individuals start to vote legally) and 30 years often represents the time for taking stock in life. However, Zebrack et al. (2010) considered 39 years age as the upper age limit to be considered an individual as a young adult. It is the period of human development during which a young person who has been dependent on his or her parents throughout childhood starts to take definitive steps towards independence (financial, residential, and emotional) and take on adult roles (i.e. citizen, spouse, parent, and worker) (Modena and Rondinelli, 2010). Figure 1.1 shows the features of young adults as described by Arnett and Tanner (Arnett and Tanner, 2006): these include self-focus, creating an adult identity and awareness of new possibilities. However, for the purposes of this doctoral study, where independence may be considered an issue in decision making, it is important also to consider that the current generation of young adults have higher levels of student debt and are more likely to experience poverty and unemployment compared with the two prior generations. Fry (2013) showed that 53% of young adults (aged 18-24

years) either currently lived with parents or had moved back temporarily after a period of living independently.

FIGURE 1. 1 FEATURES OF YOUNG ADULTS (ARNETT AND TANNER, 2006)

AGE OF IDENTITY EXPLORATION

Young adults are deciding who they are and what they want out of work, school and love.

AGE OF INSTABILITY

The post-high school years are marked by repeated residence changes, as young adults either go to college or live with friends or a partner. For most, frequent moves end as families and careers are established in the 30s.

AGE OF SELF-FOCUS

Freed of the parent- and society-directed routine of school, young adults try to decide what they want to do, where they want to go and who they want to be with before those choices get limited by the constraints of marriage, children and a career.

AGE OF FEELING IN BETWEEN

Many young adults are taking responsibility for themselves, but still do not completely feel like adults.

AGE OF POSSIBILITIES

Most young adults believe they have good chances of living "better than their parents did".

Moreover, many young people experience obstacles to their development as adults due to factors such as becoming parents too soon, dropping out of school, failing to find work, or getting into trouble with the legal system (Children's Defense Fund, 1999). These experiences make the transition to adulthood more difficult (Malekoff, 2014). In order to help young adults effectively, it is important to understand the relevant lifestage and the developmental processes that are involved (Geldard and Geldard, 2009).

For the purposes of this doctoral study, where, as I have already reported, independence may be considered an issue in decision making, it is important also to consider the Italian context of young adults' lives. First of all, it is important to consider that Italy is a country that has been characterized by the age until which young adults remain in the parental home, which is very high when compared to other countries (Ferrari et al., 2014). This could affect young adults' development into mature adulthood, as one of the key developmental tasks of emerging adulthood is living independently, as declared by Shanahan (2000). In this scenario, the transition to adulthood occurs within the family contex (Scabini et al., 2006). Parents support their children, who are expected to leave the parental home only after completing their education, finding a stable job, and marrying (Albertini and Kohli, 2013). This takes place in a context currently characterized by high levels of job insecurity and unemployment. The Italian Institute of Statistics (Istat, 2016) reported that in 2015, seven million young adults (62.5%) aged 18-34 years lived with their nuclear families. Of those 35.5% were students, 29.7% were unemployed, and 31.8% chose to live in their parental home. These data show that economic difficulties only partly explain this phenomenon: there is a group of young adults who, although they have the financial security necessary to live independently and form a new family, still postpone the choice to leave the parental home and continue to live with their parents (Buzzi et al., 2007).

In this Italian context, it is clear that the age at which young adults were forming partnership and becoming parents has changed. The Italian Institute of Statistics (Istat, 2016) reported that in 2015 the mean age of the

bridegroom was 34.7 years, compared to a mean of 31.7 years for the bride. Similar data were reported by the Italian Institute of Statistics (Istat, 2016) considering the mean age for becoming parents: young women become mothers at 32.3 years and young men became fathers at 35.3 years of age.

Specifically for my doctoral study aims, there is some inconsistency in the definition of young adults correlated with presymptomatic genetic testing as definitions used in the literature may be people under 30 years of age (Patenaude et al., 2006) or under 25 years (Bradbury et al., 2008), while in other relevant work authors use only the terms "children", "adolescent" or "adulthood" (Borry et al., 2006, 2009; Van der Meer et al., 2012) without specifying any age. In addition, while some authors write about young people and presymptomatic testing, they are usually referring to minors (age <18). For the purpose of this doctoral study, I will use the definition suggested by Patenaude et al. (2006) (30 years of age and under).

1.3 GENETIC COUNSELLING AND GENETIC TESTING

In this section, I will provide relevant background on the practice and processes of genetic counselling and testing. The Italian scenario will be compared with clinical practice in other countries.

1.3.1 GENETIC COUNSELLING

The American Society of Human Genetics (ASHG) (American Society of Human Genetics Ad Hoc Committee on Genetic Counseling, 1975) initially

defined genetic counselling as a process of communicating genetic risks to families, but updated this definition in 2006 to

> *'the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease*' (Resta et al., 2006, p. 77).

This changed definition emphasized the need for genetic counsellors to also provide psychological support to clients, which means practitioners need to have a range of counselling skills (Skirton, Lewis, et al., 2010). Each client arrives for the first consultation with a set of expectations, which may or may be not congruent with those of the counsellor (Skirton, 2001). The counsellor therefore needs to open the counselling session with a concise summary of what is known about the client and his/her family, and to ask what he/she hopes to get out of the counselling session (Uhlmann et al., 2009; Harper, 2010). A genetic counsellor aims to create an ambience allowing the client to talk about his/her feelings and emotions (Evans, 2006). When the process of information gathering is completed, the counsellor should use that information to determine the risk of recurrence of a disorder for the client or his/her family members. After the initial consultation, there may be a lapse of time during which all of the information is assimilated (Uhlmann et al., 2009; Harper, 2010). However, in order to provide psychological support, the counsellor has to provide the mechanisms in order to enable the client to express their emotions and feelings and resolve their situation in the best possible way (Evans, 2006; Harper, 2010). Although, genetic counsellors have many roles, it is suggested that the main role of genetic counselling is helping clients make appropriate decisions on the basis of the information that they receive (Evans, 2006). Decisions made in the

context of genetic counselling may lead to significant actions such as testing for hereditary cancer predisposition (Lea et al., 1998). It is known that individuals at risk of a genetic condition, even members of the same family, may make entirely different decisions regarding, for example, use of protective or risk-reducing strategies (Paalosalo-Harris and Skirton, 2016). Theoretical models of health behaviour underline factors that are relevant for understanding health-related actions, and explain why some people take protective actions and others do not (Steptoe and Matthews, 1984). For example, in the Health Belief Model, Becker et al. (1977) state that the principal predictors of the likelihood of engaging in a specific health behaviour associated with taking the action are the perceived susceptibility to a health threat, its perceived severity, and the perceived benefits and barriers. These four major constructs of perception are modified by other variables, such as culture, education level, past experiences, skills, and motivation. Although the European Board of Medical Genetics guidelines considers the Master's degree level education as essential to prepare health professionals for practice as genetic counsellor or genetic nurses (European Board of Medical Genetics, 2010), in Italy apart from one example, nurses' training in genetics is provided "on the job" and through conferences and seminars, because in Italy there is no Master programme or any other specialized training available to become a genetic nurse (Godino et al., 2013).

A proportion of clients seeking genetic counselling do so because of their concerns about familial cancer (Lea et al., 1998; Harper, 2010). Although the majority of common cancers are sporadic, due to somatic changes in the genes of particular cells, hereditary cancer accounts for 5-10%

of estimated cancer occurrences (Riley et al., 2012). It is possible in many cases to test an affected person to detect the gene mutation that has contributed to the development of the cancer and that may be passed on to biological relatives. The inherited gene mutation makes the person more prone to develop cancer and to be affected by cancer at a younger age than in the normal population, therefore clinical surveillance and possibly risk reducing treatment may be offered to those with a possible inherited predisposition (Neri and Genuardi, 2010). Development of neoplasia is associated with at least 200 of the known single gene disorders (Lashley, 2005). Table 1.1 provides data on some of the hereditary cancer syndromes with the clinically available tests in detail (Schneider, 2011).

The general focus of genetic counselling has been described earlier in the chapter. More specifically, onco-genetic counselling is aimed at identifying individuals and families who may benefit from further cancer risk assessment and testing, providing support and follow-up throughout the counselling and testing process and monitoring clients' responses to diagnostic, preventive and intervention methods (Skirton and Patch, 2009; Uhlmann et al., 2009; Harper, 2010). The probability that an individual has inherited a mutation and/or that he will develop cancer is generally estimated based on the analysis of the pedigree and accompanying family medical history, according to validated criteria specific for each syndrome (Neri and Genuardi, 2010).

TABLE 1. 1 HEREDITARY CANCER SYNDROMES WITH CLINICALLY AVAILABLE TESTS (SCHNEIDER, 2011)

SYNDROME	**GENES**	**MODE**
Ataxia Telangiectasia	ATM	Autosomal Recessive (AR)
Autoimmune Lymphoproliferative syndrome (Canale-Smith syndrome)	CASP10	Autosomal Dominant (AD)
Beckwith-Wiedemann Syndrome (also Exomphalos Macroglossia Gigantism syndrome)	BWS	AD 85% of cases are sporadic
Birt-Hogg-Dubé syndrome	FLCN	AD
Bloom syndrome	BLM^{ASH}	AR
Hereditary breast-ovarian cancer syndrome (HBOC)	BRCA1/2	AD
Carney complex	PRKAR1A	AD
Diamond-Blackfan Anaemia	RPL5/11/35A RPS7/10/17/19/24/26	AD 10-25% of cases are familial A few cases AR
Familial adenomatous polyposis (also attenuated FAP, Gardner's syndrome, Turcot syndrome, Hereditary desmoid disease, and FAP)	APC	AD
Fanconi Anaemia	FANCA-FANCN	AR X-Linked recessive only for FANCB
Hereditary diffuse gastric cancer	CDH1	AD
Juvenile polyposis syndrome	BMPR1A SMAD4	AD
Hereditary leiomyomatosis renal cell cancer	FH	AD
Li-Fraumeni syndrome	TP53	AD
Lynch syndrome (also termed HNPCC)	MLSH1/2/6 PMS2 TACSTD1	AD
Melanoma, cutaneous malignant (includes familial atypical mole-malignant melanoma syndrome, dysplastic nevus syndrome, and melanoma-astrocytoma syndrome)	CDK4 CDKN2A	AD

Multiple endocrine neoplasia, type 1 (also Wermer syndrome or MEN1)	MEN1	AD
Multiple endocrine neoplasia, type 2 (also Sipple syndrome, familial medullary thyroid carcinoma syndrome or MEN2)	RET	AD
MYH-associated polyposis	MYH	AR
Familial neuroblastoma	ALK PHOX2B	AD
Neurofibromatosis, type 1 (also Von Recklinghausen disease)	NF1	AD
Neurofibromatosis, type 2	NF2	AD
Nevoid basal cell carcinoma syndrome (also Gorlin syndrome, basal cell nevus syndrome)	PTCH	AD
Hereditary paraganglioma-pheochromocytoma (including Carney-Stratakis syndrome)	SDHB/C/D	AD : SDHD AR : SDHB/C
Peutz-Jeghers syndrome (PJS)	STK11	AD
PTEN hamartoma syndrome (also Cowden syndrome, includes Bannayan-Riley-Ruvalcaba syndrome and Proteus syndrome)	PTEN	AD
Hereditary papillary renal cell carcinoma	MET	AD
Hereditary retinoblastoma	RB1	AD
Rothmund-Thomson syndrome	RECQL4	AR
Tuberous Sclerosis complex	TSC1/2	AD
Von Hippel Lindau syndrome	VHL	AD
Familial Wilms' tumor (includes Denys-Drash syndrome, Frasier syndrome, WAGR syndrome)	WT1	AD
Xeroderma pigmentosum (includes XP/CS complex, XP variant)	XPA XPC	AR

In addition, counsellors may use statistical models to predict risk. Examples are the Gail model (Gail et al., 1989), the Claus model (Claus et al.,

1993, 1994) and the Tyrer-Cuzick model (Tyrer et al., 2004) to determine breast cancer risk. If appropriate, genetic testing is performed in order to further clarify the genetic risk for the individual (Neri and Genuardi, 2010).

1.3.2 GENETIC TESTING

The term "genetic testing" is used in a variety of different settings (Pinto-Basto et al., 2010). In the health care context, genetic testing was defined by a UK Advisory Committee on Genetic Testing as

> '*testing to detect the presence of absence or an alteration in a particular gene, chromosome or a gene product*' (Advisory Committee on Genetic Testing, 1998, p. 8).

In the context of clinical genetics, genetic tests are used in a number of scenarios, and may be used for diagnostic, presymptomatic, predictive, carrier or prenatal testing. In more detail:

- diagnostic testing is a genetic test performed in a symptomatic individual to confirm or exclude a genetic condition
- presymptomatic/ predictive testing is available for a number of heritable genetic disorders including hereditary cancer syndromes, inherited cardiac conditions and neurodegenerative genetic disorders. The terms 'presymptomatic' and 'predictive' genetic testing refer to the possibility of detecting a genetic mutation that causes a particular condition before the presentation of symptoms
- carrier testing is a genetic test that detects a gene mutation that will generally have limited or no consequences to the health of the individual. However, if inherited from one parent or in combination

with the same or another mutation in the same gene from the other parent, it may confer a high risk of disease in the offspring

- prenatal testing is a genetic test performed during pregnancy because of an increased risk for a certain condition in the fetus (Neri and Genuardi, 2010).

In the context of genetic counselling, genetic testing should be offered with adequate and appropriate education provided at the level of the client's understanding. The understanding of the client should be taken into consideration while showing respect for their ethnic and cultural values and beliefs (Lashley, 2005). Clients should have the right to choose testing without coercion. These points are consistent with the recommendations on genetic testing made by both the Council of Europe (2008) and the Organisation of Economic Co-operation and Development (2007). Both sets of recommendations require that genetic testing must be accompanied by relevant information provided by a genetic counsellor with adequate educational and training preparation. In the systematic review conducted by Skirton et al. (2015), it was reported that genetic counsellors were mostly educated to at least postgraduate level. This is in line with the European Board of Medical Genetics guidelines that considers Master's degree level education as essential to prepare health professional for practice as genetic counsellors or genetic nurses (European Board of Medical Genetics, 2010).

1.4 PSYCHOLOGICAL IMPACT OF PREDICTIVE AND PRESYMPTOMATIC GENETIC TESTING

Presymptomatic and predictive genetic testing are available for a number of heritable genetic disorders including hereditary cancer syndromes, inherited cardiac conditions and neurodegenerative genetic disorders (Evans et al., 2001). The terms 'presymptomatic' and 'predictive' genetic testing refer to the possibility of detecting a genetic mutation that causes a particular condition before the presentation of symptoms. The first term generally refers to those diseases in which a positive test result will inevitably lead to the development of the disease later in life (i.e. Huntington disease); the second term refers to a broader range of diseases in which the risk for a disorder is increased but without necessarily implying any degree of certainty (i.e. hereditary breast and ovarian cancer). However, these terms are often used in a broadly interchangeable manner (Skirton et al., 2013).

A substantial difference in types of disorder for which presymptomatic or predictive testing can be offered exists in terms of preventive measures and or early detection. Those with predisposition to inherited cancer can often be monitored through a surveillance protocol or take preventive measures via surgical intervention, while no such measures are currently available for diseases such as Huntington disease or cerebellar ataxia (American Society of Clinical Oncology, 2003). Therefore, the choice to undergo a presymptomatic test for disorders with incomplete penetrance (a form of penetrance in which not all individuals who have a mutation manifest the disease (Strachan and Read, 2011), for example Hereditary Breast and Ovarian Cancer syndrome, and where there are preventive measures could

have a highly different psychological and social impact when compared with testing for disorders with complete penetrance (a form of penetrance in which all individuals carrying a mutation will develop signs and symptoms (Strachan and Read, 2011)), for example Huntington disease and no preventive options, particularly in young adults. For brevity, in this dissertation, the term 'presymptomatic' will be used to indicate both predictive and presymptomatic genetic tests, but the different impact will be considered whenever appropriate.

Overall, the presence in the family of a disorder such as Huntington disease, can influence the relationship between parents and their children for different reasons. These include becoming preoccupied with the diagnosis, changes in the family social system and concern about the fact that children are at risk for developing the disease (Tibben, 2007). In the context of impact of testing on the individual and the family, presymptomatic testing has been extensively studied in cohorts of adults of all ages. Collins et al. (2007) studied a sample (age range: 21-75 years) of asymptomatic individuals with a hereditary non-polyposis colon cancer (HNPCC) mutation identified in their family. Those authors described an increase in mean cancer-specific distress in those with a mutation after two weeks, with a return to baseline levels by 12 months that was maintained until three years post-test. However, in those without the mutation there was a decrease in distress after testing, with a significantly reduced level at three years compared with the baseline. Conversely, members of the two groups (mutation-positive and mutation-negative) did not differ in relation to mean depression and anxiety scores (Collins et al., 2007). The authors reported a

lower breast cancer risk perception in those who were mutation-negative, compared with those who were mutation-positive, 12 months after BRCA1/2 genetic testing. Julian-Reynier et al. (2011) did not specify the age range of the sample, but declared the mean age of those with a BRCA1/2 mutation at the time of disclosure was 37.2±10.2 years, while mean age of those without a mutation was 41.7±11.8 years. The authors described the risk-prevention decisions made by healthy women up to 5 years after disclosure of their BRCA1/2 test result, based on behaviour of women in various age groups. Breast surveillance alone was opted for by 50% of healthy women who were mutation-positive: 31% of those underwent either magnetic resonance imaging and other imaging (22 out of 31 women were under 40 years of age) and 19% chose mammography alone (nine out of 19 women were under 40 years of age). Risk reducing salpingo-oophorectomy and breast surveillance (based on magnetic resonance imaging and other imaging or mammography alone) was used by 38% of mutation positive women, risk reducing mastectomy and risk reducing salpingo-oophorectomy by 5% (four out of seven women were under 40 years of age), and risk reducing mastectomy alone by 2% (two women that were up to 40 years of age). Six percent of these women decided not to undergo either risk reducing mastectomy and/or risk reducing salpingo-oophorectomy or surveillance. Of those who would rather not undertake any preventive strategies, two women were under 30 years of age. However, Watson et al. (2004) showed that the risk perceptions of those with a BRCA1/2 mutation were higher at six months post-test than they were before testing, but it is suggested that risk perception does eventually decrease over time (Heshka et al., 2008). However, none of these studies focussed on the experiences of young adults.

Various guidelines and position papers have been produced on presymptomatic and predictive genetic testing in minors (Borry et al., 2006). It is clearly suggested that undergoing presymptomatic testing too early in life may increase the risk of unfavourable impact, and, therefore, the appropriate age to undergo presymptomatic testing is still a matter of debate (Borry et al., 2006; Richards, 2008; Duncan et al., 2008). Presymptomatic and predictive genetic testing offers the possibility of defining the individual risk for a genetic disorder (Neri and Genuardi, 2010). A variety of psychosocial responses have been observed in those who have chosen testing (Meissen et al., 1991; Williams et al., 1999; Baig et al., 2016). For these reasons, presymptomatic genetic testing for adult-onset disorders is not generally recommended for those aged less than 18 years, unless it is in a child's best interests either in terms of immediate relevance for their health or of psychological or social benefits.(Borry et al., 2009). Conversely, according to UK guidelines, people aged 16 or 17 years are presumed to be capable of consenting to their own medical treatment, and, in specific cases, children under 16 years who have sufficient understanding and intelligence to enable them to fully understand what is involved in a proposed intervention will also have the capacity to consent to that intervention (Department of Health, 2009). In addition, it has been argued that young persons who are considered as adults on the age-based criterion of 18 years are not all necessarily truly autonomous (Richards, 2008). There is no specific age when a person is able to give autonomous consent, but it is important to consider psychological maturity (Richards, 2008) that is cumulative with age, life experience and cognitive development (Steinberg and Cauffman, 1996).

Prior to testing, young adults need to be aware of the potential risk to them of hereditary cancer, and this is usually disclosed by their parents. Patenaude et al. (2006), Bradbury et al. (2007a) and Van der Meer et al. (2012) all reported that 50% or more of parents who have a BRCA mutation inform their minor children of their mutation status, despite recommendations against genetic testing for BRCA mutation or other presymptomatic genetic testing in minors (Borry et al., 2009). Prevalence and experiences of parental communication of BRCA results to children under the age of 25 years old was described by Bradbury et al. (2007). Specifically, in their study, the majority of parents (55%, n= 23/42) reported sharing family history and/or genetic risk with at least one child: 91% (n=21/23) shared genetic test results and 9% (n=2/23) shared only family history. Their results indicate that the 43% (n=18/42) of children in these families were learning of their potential genetic risk of cancer before the age of 18 and 57% (n=24/42) between 18 and 24 years of age. It came to light in that study that children of those with a BRCA mutation learnt of their parents' genetic test results many years before preventive interventions were indicated. In fact, in a study of 273 women tested for hereditary breast and ovarian cancer mutation, Patenaude et al. (2006) noted that although most children were told by their mother, the child's age influenced the communication with offspring in same age groups. However, they showed there was no significant difference between numbers of minors (14 to 17 years, 85%) or young adults (18 to 30 years, 92%) and children age 30 or older informed of the risk by their parents. Borry et al. (2009), in their paper on genetic testing in asymptomatic minors, concluded that minors, considering their age and degree of maturity, are able to participate in the

decision making and their opinions regarding genetic testing should be taken in consideration.

Bradbury et al. (2007) described offspring reactions to disclosure of information about BRCA mutations and they reported that almost half of the children of mutation-positive parents did not appear to understand the significance of the information. In that study, some parents believed that sharing their genetic status did not add significant information because their children were already conscious of the hereditary risk in the family. Thirty percent of those who disclosed this information reported that they did this between several months to six years after receiving their test result: waiting for the child to get older was the main reason for delaying the communication. There does seem to be evidence to support this strategy, as when comparing the mean age of offspring at disclosure with their understanding (based on the parents' perception) older children (mean age 20.1) understood the significance of the information better than younger offspring (mean age 16.8). Parents also described a variety of emotional reactions in their offspring, varying from concern or anxiety to crying or fear.

In the context of adverse responses to disclosure of genetic risk, genetic counselling may assume an important role. It is recommended, for example, in the Huntington disease scenario, that there is a screening interview between the genetic counsellor and the client at risk by means of an initial telephone interview (Lea et al., 1998). A minimum of three pre-test genetic counselling sessions to ensure informed decision making and follow-up sessions to discuss the test results over a two year period in the context of a genetic counselling was initially suggested when pre-symptomatic testing

for neurodegenerative conditions was first introduced (Went. et al., 1994), although these requirements have since been modified (MacLeod et al., 2013). Although the guidelines have provided data on how genetic counsellors can help clients in their presymptomatic genetic testing decision making process, clinical experience has shown the importance of a case by case approach (Tibben, 2007). In the most recent guidance, while there is still an emphasis on both pre- and post-test counselling to prepare the patient and to support him or her to deal with the result (MacLeod et al., 2013), this should be adjusted to suit the individual.

1.5 GENETIC COUNSELLING IN ITALY: WHO PROVIDES IT?

In Italy, genetic counselling is provided by medical geneticists, specialists specifically trained in medical genetics. In 2009, the Italian Society of Human Genetics stated that the presence of a genetic nurse on the team in clinical genetic units was an obligatory requirement. However, despite this recommendation, the role of the genetic nurse has not yet been well defined (Società Italiana di Genetica Umana, 2009). There are very few genetic centres in Italy employing genetic nurses (i.e. Trento, Bolzano, Milano, Bologna) (personal communication presented at the Italian Society of Human Genetics, Rimini, Italy, October 2015), but in these centres nurses with genetics knowledge work in collaboration with medical geneticists.

In countries such as the United States, Canada, Japan, the United Kingdom, Belgium, Australia, New Zealand, and the Netherlands, the contribution of nurses to specialist genetic health care is well established (American Nurse Association and International Society of Nurses in Genetics,

2016). Basic and advanced levels are acknowledged in the scope of genetic speciality nursing practice. Application of genetic knowledge, in risk assessment, identification of possible outcomes, intervention and evaluation are included in both levels of the genetic nurse's daily work. The genetic nurse working at the basic level is considered able to conduct a risk assessment based on fundamental genetics knowledge. It is part of her/ his daily work to collect and record a family pedigree, to identify components of the family history that may benefit genetic counselling, to explain the client's potential genetic risk, to develop a referral plan with the client, to facilitate a referral to a genetic nurse in advanced practice, to provide psychological support, to evaluate the interventions, and to assess the client's understanding and ability to implement a plan (i.e. of surveillance or treatment in a oncological context) following the referral. The genetic nurse working at the advanced level is expected to conduct a more thorough risk assessment based on family and other risk factors and to provide understandable information about genetic testing to enable the client to make an informed decision about whether to be tested or not. It is also part of her/ his work to discuss interpretation of genetic test results with the client, to determine the client's need for assistance in communicating test results within the family, to discuss surveillance and any risk reduction options (if available for the disorder in the client's family), and to monitor outcomes of the interventions. In summary, expanded practice skills and knowledge in nursing and genetics, and an increased complexity of decision making processes are the main characteristics that distinguish advanced from basic level in genetics nursing.

In general, information giving and exploration of the client's circumstances and needs are included within the role of the genetic counsellor (Skirton, Patch, et al., 2010). In more detail, in the systematic review conducted by Skirton et al. (2015) to assess the role of the genetic counsellor, the authors found that taking family history, drawing the pedigree, risk assessment, discussion of the genetic disease, psychosocial impact of the disease, providing client education, discussion of options, addressing ethical issues, making, providing psychosocial assessment and support, and delivering professional and public education were encompassed in the role. Because of the genetic counsellor's contribution, for example in the process of decision making (it has been detailed in this chapter, Section 1.3.2), an empathic client-centred approach is a fundamental requirement (Skirton, Patch, et al., 2010).

1.6 RESEARCH PROBLEMS

Based on the potential adverse impact of genetic counselling for hereditary cancer on young adults and the scarcity of evidence regarding presymptomatic testing in this group, I wished to investigate how young adults make testing decisions, the emotional impact of the process and how genetic services can provide optimal support to them in this process.

1.7 AIMS AND OBJECTIVES OF THE STUDY

The purpose of this programme of doctoral study was to explore the implications of presymptomatic testing for hereditary cancer in consultands aged 18-30 years. The specific objectives were:

- to explore how young adults interpret cancer presymptomatic testing
- to explore the basis for young adults' decisions to undergo testing or not
- to explore the influence that parents have in the choice, with reference to family dynamics and lifestage theory
- to analyse the psychosocial impact of test disclosure, according to mutation status
- to develop a theoretical model regarding the decision making process in young adults considering presymptomatic testing for hereditary cancer
- to inform the process of cancer genetic counselling for young consultands.

1.8 STUDY DESIGN: MIXED METHODS

The overall design chosen for this doctoral research project was a mixed methods approach (Creswell and Plano Clark, 2011). The programme of study comprised a systematic review, qualitative and quantitative phases. I chose a mixed methods design because both qualitative and quantitative methods, in combination, provide a better understanding of a research

problem or issue than either method alone (Creswell, 2002; Tashakkori and Teddlie, 2003). More detail about the methods used will be provided in the next Chapter.

This doctoral study comprised three distinct phases:

- Phase 1: I undertook a systematic review of the current literature relating to my thesis. The purpose of the review was to answer the following questions: which factors influence young adults' or adolescents' choices to have a presymptomatic or predictive test? Eleven studies were identified using inclusion/exclusion criteria. This systematic review appears, to the best of my knowledge, to be the first published review on factors that influence young adults' or adolescents' choices to have a presymptomatic or predictive test. Because the systematic review was the first phase of this doctoral study, in order to understand what was published later, I have updated the search, using the same keywords on the eight databases that I previously searched. The systematic review is discussed in Chapter Three and the update of the literature has been reported in a section of Chapter Six.
- Phase 2: I expanded the findings from the systematic review by conducting in-depth interviews in this qualitative phase. The psychosocial implications of presymptomatic testing for hereditary cancer in young consultands (aged 18-30 years) referred for cancer genetic counselling were assessed. As part of this phase, I evaluated the cancer perception and psychological status of young adults and explored the extent to which the parents' influence was important. In addition, the major issues emerging from the systematic review were

explored. In this phase, I interviewed young consultands (18-30) without personal history of cancer who were members of families with a hereditary cancer predisposition. I interviewed participants on three occasions: one month before genetic counselling, and two weeks and six months respectively after genetic counselling. The interviews were designed to explore the participants' journey through testing, including emotions, experiences and the psychosocial implications of predictive testing for hereditary cancer. Interview data were analysed using grounded theory. A theory named "Finding yourself in front of the mirror" was constructed to describe the experience of young adults who decided to undergo presymptomatic testing for hereditary cancer.

- Phase 3: I used the findings from the systematic review and the qualitative interviews to design the third phase of my doctoral study. I performed a quantitative study to systematically assess the most relevant findings emerging from the qualitative study in a wider population. To dermine how young individuals interpret presymptomatic cancer testing, the basis for the young individual's decision to undergo testing or not, the experiences of the counselling process of both young adults and parents, the influence that parents have on the choice to be tested or not, the influence that parents have on the young adult's decisions after the disclosure of the positive test result and how the experiences of young adults being tested in Italy and their parents compared with those in other countries were the specific objectives. To achieve these objectives, based on the results of the literature (Phase 1) and the qualitative study (Phase 2), some

specific variables were identified, and the most appropriate tools were chosen to measure those variables. For some topics, new questionnaires were designed. The data obtained were entered into a dedicated database, arranged by variables and finally analysed in order to assess the relationships between variables. Both the influence of other people and the decision making process were identified as being key factors in the process.

1.9 STRUCTURE OF THE THESIS

I have organised the dissertation into six chapters.

In this first chapter I introduced the doctoral study and described the organization of the overall doctoral dissertation.

In Chapter Two I describe, discuss and justify the method chosen to address the aims and objectives of this doctoral dissertation. The methods used for each phase are detailed.

Chapter Three is focussed on the systematic review of the literature. I present the findings from the systematic review on which factors influence young adults or adolescents' choices to have a presymptomatic or predictive test. I discuss the key themes in relation to theoretical concepts.

In Chapter Four I report the qualitative phase of the doctoral study concerning the psychosocial implications of presymptomatic testing for hereditary cancer in young consultands (aged 18-30 years) referred for cancer genetic counselling. I present and discuss the findings from the qualitative interviews with participants who underwent presymptomatic

genetic testing. I then propose and examine a theoretical model summarising the experience of participants.

Chapter Five includes the results from the quantitative phase of the study. The results from the questionnaires designed to explore the psychosocial impact, the decision making process and the consequent counselling needs of young adults and parents are presented and discussed.

In Chapter Six I summarise, synthesise and discuss the findings of the three phases of the doctoral study and critique the study. I then present an update of the literature based on the systematic review performed (Phase 1). A new theoretical model is presented in this chapter. The new model was built to summarise the overarching all three phases of this doctoral project. This new theoretical model of decision making and impact on young adults who underwent presymptomatic genetic testing showed it as a dynamic process. It emphasises the interaction between sensitive experience and temporal dimensions, according to the definition of dynamic provided by Valsiner et al. (2009). In the conclusion, recommendations for practice and further research are provided, as well as a reflexive account and a statement on the novel aspects of the doctoral study.

1.10 IN SUMMARY

In this chapter I have given a brief overview of the thesis, a definition of young adults and described the findings from previous research focusing on the psychological impact of presymptomatic genetic testing. I have also presented the rationale for conducting this doctoral study. In the following

chapter, I will describe, discuss and justify the mixed methods research approach chosen to conduct the doctoral study and detail each method used.

CHAPTER TWO

MATERIALS AND METHODS

2.1 INTRODUCTION

In this chapter, I will present the methods chosen to conduct the research. Initially I will discuss the choice and use of a mixed methods approach for the research in this doctoral study. A brief account of the process carried out in order to complete the systematic review for Phase 1 of the study will be described: this will be expanded upon in Chapter Three, along with the results and discussion of the review. I will provide a brief background to qualitative research methods, looking at their use within the health care setting, and justifying their use in this phase of the doctoral project. I will then present the grounded theory method and discuss why I considered it was the most suitable method for this phase of the doctoral study. Finally, I will describe the quantitative research method with a critique and justification of the use of the method. I will also describe the doctoral study design, focusing on the recruitment process, ethical issues, and the procedures used to analyse the data.

2.2 MIXED METHODS DESIGN

In this section the definition and development of mixed methods research will be examined. The specific research design will then be presented, followed by the key decisions made relating to this research design. Finally, limitations of the research design will be discussed.

Mixed methods research is often used in health care to address research questions relevant to improving quality of patient care (Bryman, 2016). Some authors believe that mixed methods studies involve use of a combination of qualitative and quantitative methods to achieve the research aims, while other researchers formally adopt mixed methods as a paradigmatic underpinning of the research process (Creswell and Plano Clark, 2011; Coolican, 2014).

Mixed methods research is used because it has the potential to intensify the findings in a manner that is not always possible with one type of data (Tashakkori and Teddlie, 2003). The inclusion of both qualitative and quantitative data allow the researcher to achieve two objectives simultaneously: a) to generalise findings from a sample to a population and b) to facilitate a deeper understanding about the topic being studied (Creswell and Plano Clark, 2011).

Although the mixed methods approach was introduced in the 1960s (Leech and Onwuegbuzie, 2009), pragmatism, the paradigm that provides the philosophical underpinnings of mixed methods research (Doyle et al., 2009), did not emerge until the 1990s (Denzin, 2010). Methodological choice does not exist within a philosophical void and Brannen (2005) shows that the choice of method used is driven by philosophical assumptions. Ontology and

epistemology are two facets of the philosophical assumptions that researchers should consider. Questions such as '*what is the nature of reality?*' (ontology) (Creswell and Plano Clark, 2011, p. 42) and '*what is the relationship between the researcher and that being researched?*' (epistemology) (Creswell and Plano Clark, 2011, p. 42) are considered by the researcher before starting the study. Within their worldview, pragmatists believe that the consequences are more important than the process (Doyle et al., 2009) and that determining the quality of a study should be based on the planned purpose, allocated resources, procedures followed and findings generated (Patton, 2002). Because mixed methods research involves using both qualitative and quantitative methods, it is important that the researcher reflects on both approaches. Traditionally, qualitative researchers work within the constructivist worldview with an ontologic stance based on the belief that there are multiple realities based on different perspectives (Creswell and Plano Clark, 2011). Qualitative researchers increase knowledge by observing and or collecting narrative from participants and analysing the data while acknowledging that, as reareachers, they are immersed in the study, leading to value laden, subjective interpretations (Tashakkori and Teddlie, 2003; Doyle et al., 2009). This contrasts with the quantitative method, as the epistemological view of quantitative researchers is that they remain objective when collecting numerical data with structured data collection methods, and in analysing the data using statistical procedures to make impartial interpretations (Tashakkori and Teddlie, 2003; Doyle et al., 2009; Creswell and Plano Clark, 2011). Creswell and Plano Clark (2011) suggest that usually quantitative researchers work within the postpositivist worldview with an ontologic stance that there is a singular

reality centred on rejecting or failing to reject the null hypothesis (Creswell and Plano Clark, 2011).

A mixed methods approach can therefore be utilised to maximise the rigour of the findings in complex situations. As such, the mixed methods approach to social, behavioural and health care research has become progressively more common (Tashakkori and Teddlie, 2003; Doyle et al., 2009; Leech and Onwuegbuzie, 2009; Chow et al., 2010; Creswell and Plano Clark, 2011).

Jick (1979) pioneered the mixed methods approach in 1979, advocating combining both qualitative and quantitative methods rather than simply collecting both types of data without deliberately planning integration of the two in the research design (Jick, 1979). He claimed that, by using this approach, researchers could have more confidence in their findings. Triangulation and sequential design are the two types of mixed methods research designs developed by Morse (1991), who further developed the thinking about mixed methods. Morse (1991) also stated that the use of mixed methods research was not just to obtain complementary findings using two approaches, but could lead to the development of new knowledge and theory.

The key issue when choosing a mixed methods approach are priority, timing, and mixing (Creswell and Plano Clark, 2011; Coolican, 2014). Priority refers to the relative importance of the quantitative and qualitative strands within the design for answering the research questions. There are three possible weighting options: equal priority, quantitative priority and qualitative priority. Equal priority occurs when both methods play an

equally important role in addressing the research aim; the quantitative priority occurs when a greater emphasis is placed on the quantitative methods, while a greater emphasis on the qualitative strand equates to qualitative priority (Creswell and Plano Clark, 2011; Coolican, 2014). Timing refers to the temporal relationship between the quantitative and qualitative strands within a study. The selection of timing is pragmatically based on the objectives of the researcher: (a) concurrent timing occurs when both qualitative and quantitative methods are implemented during a single phase of the research; (b) sequential timing occurs when both methods are implemented in two distinct phases; (c) multiphase combination timing occurs when multiple phases are implemented that include sequential and /or concurrent timing over the programme of study (Creswell and Plano Clark, 2011; Coolican, 2014). The concept of mixing relates to the stage when qualitative and quantitative findings are integrated. This decision is again based on the purpose of the research (Creswell and Plano Clark, 2011; Coolican, 2014). Cresswell and Plano Clark (2011) described four possible strategies: mixing data during interpretation, mixing during data analysis, mixing during data collection, mixing at the level of design.

Based on these key decisions, Cresswell and Plano Clark (2011) distinguished six possible designs. These are:

- convergent parallel design, when qualitative and quantitative data are collected simultaneously with equal priority
- exploratory sequential design, when qualitative data are collected and analysed prior to the collection of quantitative data

- explanatory sequential design, entailing the collection and analysis of quantitative data followed by the collection and analysis of qualitative data
- embedded design, when either quantitative or qualitative research is the priority approach
- transformative design, when the researcher works within a transformative theoretical framework investigating the needs of a specific population and calling for change
- multiphase design, combining both sequential and concurrent design over a period of time within a programme of study addressing an overall programme objective.

After considering all the possible approaches, a sequential exploratory mixed method design (Creswell and Plano Clark, 2011; Coolican, 2014) was adopted to address the research aim and objectives. I chose this design because it enabled initial deep exploration of the topic, followed by wider investigation of the results. To provide a comprehensive background to the empirical study phases of the doctoral research, a systematic review was conducted, focussing on studies assessing the factors that influence young adults' or adolescents' choices to have a presymptomatic test for a genetic condition. Qualitative interviews were then conducted with young clients (18-30) undergoing genetic counselling and, possibly, presymptomatic testing for cancer. Building from the qualitative findings, the quantitative phase was conducted collecting data using two questionnaires: one for young adults and one for parents of young adults.

The strengths and limitations of a mixed methods design have been widely discussed in the literature (Creswell et al., 1996; Green and Caracelli, 1997; Creswell, 2003, 2005; Moghaddam et al., 2003). Strengths include the opportunities for the researcher to answer a broader and more complete range of questions because he or she is not confined to a single method. It is also an opportunity for the exploration of results arising from one type of method (qualitative or quantitative) in more detail, especially useful when unexpected results arise (Morse, 1991). Also, researchers can provide stronger evidence for a conclusion through convergence and corroboration of findings. Generally, qualitative and quantitative methods used together produce the more complete knowledge necessary to inform theory and practice (Johnson and Onwuegbuzie, 2004). The limitations of this design are the extensive time and resources required to collect and analyse multiple types of data. Moreover, the researcher needs to have skills in both qualitative and quantitative research. The researcher is required to learn multiple methods and be able to know how to mix each method effectively (Johnson and Onwuegbuzie, 2004).

FIGURE 2. 1 VISUAL MODEL FOR MIXED METHODS: SEQUENTIAL EXPLORATORY DESIGN PROCEDURES

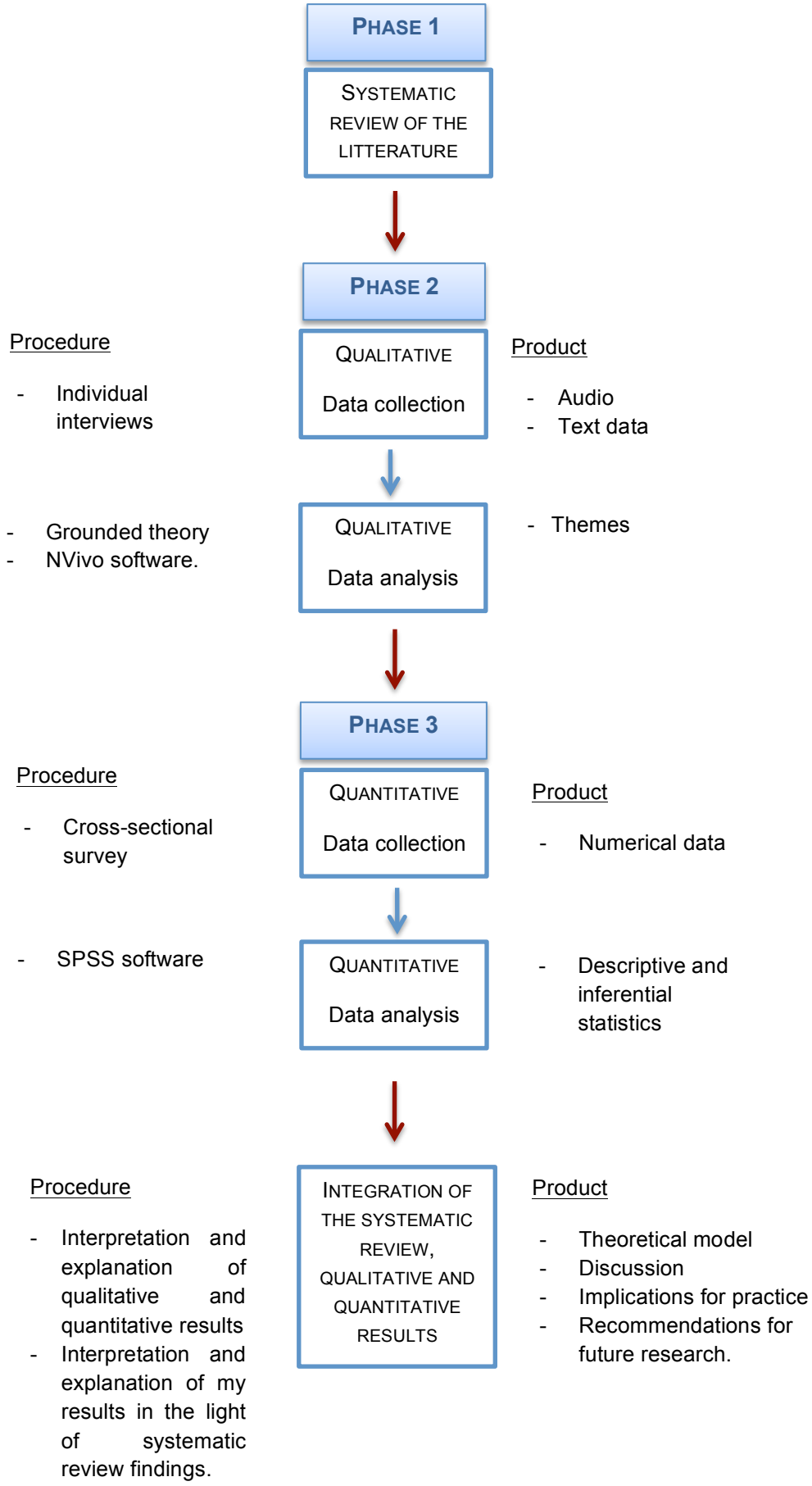


Figure 2.1, on the previous page, shows a graphical representation of the mixed methods sequential exploratory design procedures (Creswell and Plano Clark, 2011; Coolican, 2014) used for this research study. The first phase, the systematic review, informed the qualitative research (Phase 2), which in turn informed the questions for the quantitative study (Phase 3). All the findings were used to construct the final theoretical model described in the last chapter of this dissertation.

2.3 SYSTEMATIC REVIEW (PHASE 1)

2.3.1 DESIGN

In this section I will outline the method used for the systematic review, to demonstrate the entire process of the mixed methods study. Further details on the methods used in the systematic review are presented in Chapter Three.

A systematic review is a method of amassing, assessing and synthesizing a body of evidence on a particular topic (CRD, 2009). It is used when there is an important clinical question and it seeks to provide an overview of the findings of the individual research, highlighting possible answers, as well as any remaining gaps in knowledge (Clarke, 2011). The strengths and limitations of a systematic review are presented in Figure 2.2. This systematic review was conducted in accordance with the Centre for Reviews and Dissemination methods for undertaking reviews in health care (CRD, 2009).

FIGURE 2. 2 THE STRENGTHS AND LIMITATIONS OF A SYSTEMATIC REVIEW (CRD, 2009)

STRENGTHS	LIMITATIONS
CAN REDUCE BIAS IN REACHING CONCLUSIONS IS REPLICABLE MAY RESOLVE CONTROVERSY BETWEEN CONFLICTING STUDIES CAN IDENTIFY GAPS IN CURRENT RESEARCH PROVIDES RELIABLE BASIS FOR DECISION MAKING.	RESULTS MAY STILL BE INCONCLUSIVE THERE MAY BE NO TRIALS/EVIDENCE THE TRIALS/STUDIES MAY BE OF POOR QUALITY THE INTERVENTION MAY BE TOO COMPLEX TO BE TESTED BY A TRIAL PRACTICE DOES NOT NECESSARILY CHANGE BECAUSE OF EVIDENCE OF EFFECT/EFFECTIVENESS.

In this methods chapter, I will outline the systematic review method for completeness but additional details on the systematic review will be provided in Chapter Three.

The research questions for the systematic review were:

- which are factors influencing young adults' or adolescents' choices to have a presymptomatic test (or not)?
- what is the emotional impact of young adults' or adolescents' choice to have a presymptomatic test (or not)**?**

The literature on parent passing information about their condition or genetic status to their children was not included in this systematic review. The decision was taken not to include those studies unless it was possible to link the findings directly to the young adults' decision making process. If a

study included data in less directly relevant contexts, it was not included in the systematic review.

However, the topic of parental disclosure of information about the genetic condition is recognised as being of some relevance to this thesis and has been discussed in Section 3.5 in the context of the findings of the review related to young adults' decision making.

Furthermore, the systematic review gave me an opportunity to familiarise myself with the existing body of research in this context and ascertain the methods used by other researchers to inform my own research methods in Phase 2 and then in Phase 3.

2.3.2 SEARCH STRATEGY

A search of the published peer-reviewed literature on presymptomatic testing in young adults was conducted in December 2014. I chose to start the search period at 1993 because presymptomatic testing based on mutation analysis (i.e. not based on linkage) became available for Huntington disease that year (Harper, 1993) and this was a landmark in presymptomatic testing for adult onset conditions. As I am bilingual, papers published in either English or Italian were eligible (there were no papers identified that were written in Italian). The literature search employed variations and Boolean connectors of the key terms.

2.3.2.1 DATABASES

The databases used in this research were as follows:

- Embase
- The Cochrane Library
- Cumulative Index of Nursing and Allied Health Literature (CINAHL)
- Medline
- PsychInfo
- PubMed
- SocIndex
- Web of Science

Figure 2.3 provides a complete description of the databases used in the search (http://ovidsp.tx.ovid.com, accessed on 3th October 2014; http://onlinelibrary.wiley.com/cochranelibrary/search/quick, accessed on 3th October 2014, http://web.a.ebscohost.com, accessed on 3th October 2014; http://search.proquest.com/psycinfo, accessed on 3th October 2014; https://www.ncbi.nlm.nih.gov/pubmed?db=PubMed, accessed on 3th October 2014; https://apps.webofknowledge.com, accessed on 3th October 2014).

FIGURE 2. 3 DESCRIPTION OF DATABASES USED IN SEARCH

EMBASE

- Biomedical and pharmaceutical database
- 1947 - present

THE COCHRANE LIBRARY

- Provides access to high-quality, independent evidence to inform health care decision making.
- 1972 - present

CUMULATIVE INDEX OF NURSING & ALLIED HEALTH LITERATURE (CINAHL)

- Provides access to nursing and allied health journals
- 1937 - present

MEDLINE

- Provides access to literature in medical information on medicine, nursing,

dentistry, veterinary medicine, heath care system, pre-clinical sciences
- 1949 - present

PSYCHINFO

- Provides access to literature in the psychological, social and behavioural, and health sciences
- 1806 - present

PUBMED

- Provides access to literature in medical research
- 1966 - present

SOCINDEX

- Provides access to literature in sociology and related subjects
- 1895 - present

WEB OF SCIENCE

- Provides access to literature in science and technology, social sciences, arts and humanities
- 1900 - present

2.3.2.2 OTHER RESOURCES

Targeted Internet searching using Google Scholar was also used and reference lists of relevant papers were examined for any additional studies of interest.

2.3.3 KEYWORDS

An exploratory search with the terms ["genet*" or "predict* test*" or "presymptom* test*"] and ["young*" or "adult*" or "adolescent*"] and ["decision*" or "choic*" or "communicat*" or "psycho*"] resulted in 976 studies. However, this search failed to identify some papers on this theme already known to me, therefore the main search was conducted with some general key terms. The whole new search was: ["young*" or "adult*" or

"adolescent*"] and ["BRCA" or "APC" or "Lynch" or "Huntington"] and ["genetic* test*"].

2.3.4 INCLUSION AND EXCLUSION CRITERIA

The criteria for inclusion in this systematic review were papers:

- published in English or Italian
- published in peer-reviewed journals between 1993-2014 and reporting original research (using any methods)
- where the study sample explicitly included young adults or adolescents (14-30 years)
- focussed on presymptomatic or predictive testing in young people
- focused on the factors influencing young adults or adolescents' choices to have a presymptomatic or predictive test and the emotional impact of those choices.

Papers were excluded from the review if they were:

- published in languages other than English and Italian
- guidelines for testing
- educational or opinion papers
- focused on perceptions and attitudes of college students/young adults who were not at known risk of a specific adult-onset genetic condition.

2.3.5 SELECTION OF THE STUDIES

I and two supervisors (LJ and DT) independently screened the titles and abstracts of articles identified in the first search against the inclusion criteria and decided which papers should be retrieved. Articles were rejected at this stage if the title or abstract did not focus on the topic, was not in English or Italian, or was not original research. We reviewed selection decisions and resolved disagreements by consultation with a third reviewer (HS).

2.3.6 SEARCH OUTCOME

The search of eight databases produced 3373 citations. There were 755 duplicates, leaving 2618 for examination. Following review of the title and abstract, 29 papers were assessed as potentially relevant. These papers were read in detail by my supervisors and me. The eleven remaining papers were included in the review.

2.3.7 QUALITY APPRAISAL

All papers considered for inclusion criteria in the review were then subjected to independent analysis by the researcher and one supervisor (HS) using standard quality assessment criteria for evaluating original research papers from a variety of fields (Kmet et al., 2004).

2.3.8 DATA ABSTRACTION AND ANALYSIS

It was not possible to undertake a meta-analysis or meta-synthesis of the data due to the heterogeneity of the methods and samples. We therefore prepared a narrative description of the findings, as suggested by the Centre for Reviews and Dissemination (CRD, 2009), using the thematic analysis method described by Braun and Clarke (Braun and Clarke, 2008), in order to employ a clear, replicable, and transparent methodology.

2.4 QUALITATIVE PHASE (PHASE 2)

2.4.1 RESEARCH DESIGN

Qualitative research is a systematic approach to understanding and exploring qualities of a phenomenon within a particular context (Hansen, 2006): this approach was therefore appropriate for a study focussed on health-related decision making.

There are a number of approaches that have been used within the qualitative paradigm and that might have been suitable for this particular study. These include interpretative phenomenological analysis (Smith, 1996), discourse analysis (Potter and Wetherell, 1987), thematic analysis (Braun and Clarke, 2008, 2014) and grounded theory (Glaser and Strauss, 1967; Corbin and Strauss, 2014).

The aim of interpretative phenomenological analysis is to explore the way in which people experience specific and important events in their lives (Smith, 1996; Coolican, 2014) and how participants make sense of their personal and social world (Coolican, 2014). 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. An important aspect is the reflexive role of the researcher in the analysis of the participants' experience (Smith, 1996; Coolican, 2014).

Users of another qualitative approach, discourse analysis, hold that language is represented not as reflecting psychological and social reality, but as constructing it (Potter and Wetherell, 1987; Gee, 2014). The main topic of interest is the underlying social structures, which may be assumed or played out within the conversation or text. One 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 and use of metaphors (Potter and Wetherell, 1987; Coolican, 2014; Gee, 2014).

The aim of thematic analysis is to identify and describe both implicit and explicit ideas within the data, through extraction of themes. The latter are a set of aptly named and derived constructs that tell a compelling story when described by the researcher and are evidenced from the data. However, unlike grounded theory, thematic analysis does not require that saturation has been achieved nor that the themes are linked or integrated into an overall theoretical model (Coolican, 2014).

After consideration of a range of approaches, including those described above, I chose to use grounded theory. Grounded theory (Glaser and Strauss, 1967) was developed by two sociologists, Barney Glaser and Anselm Strauss, as an inductive method of data analysis that was grounded in

the data and that enabled a theory to emerge from those data (Coolican, 2014). However, after their initial development of this approach, Glaser and Strauss subsequently had differing views concerning the methods. Glaser supported the 'pure' form which states that theoretical insights emerge directly from the data if the researcher avoids being influenced by any previous theory knowledge (Glaser and Strauss, 1967). In this way, the data are analysed with no preconceptions held by the researcher (Coolican, 2014). However for Strauss the idea of the researcher avoiding influence from any previous theory or knowledge was simply not feasible (Corbin and Strauss, 2014). The timing of consultation of the literature was another fundamental issue that Glaser and Strauss disagreed upon. While Glaser (Glaser, 1992) believed that literature should not be examined until codes and categories had begun to emerge from the data, Strauss strongly disagreed with this stance. Strauss and Corbin (2014) believed that reviewing the literature early stimulates questions, directs theoretical sampling and provides supplementary validity. Moreover, literature can be used as 'data' and constantly compared with the emerging categories to be integrated in the theory (Corbin and Strauss, 2014).

Despite these differing opinions, there are sets of fundamental features associated with grounded theory methods. Once the area of interest, which may have been given little previous attention, is identified, the researcher's aim is to build his or her own theory (Corbin and Strauss, 2014). Many types of data collection techniques are compatible with grounded theory, but interviews or focus groups are the most commonly used (Payne, 2007). During data collection, the raw data are transcribed and categories

are developed from the data by 'open coding' of the transcripts (Payne, 2007). This is performed by identifying important words, or group of words, in the data and then labelling them accordingly. These are grouped into categories, which are concepts that label phenomena (Corbin and Strauss, 2014). The process of data collection and analysis continues until theoretical saturation has been achieved. This means that the researcher continues to sample and code data until no new categories can be identified, categories are well developed and the relationships between them are well established and validated (Corbin and Strauss, 2014). At this stage, a key feature of grounded theory is theoretical sampling (Payne, 2007). While the earlier stages require openness and flexibility to identify a wide range of categories, theoretical sampling is important when exploring new or uncharted areas (Corbin and Strauss, 2014). The researcher makes a decision about who provides the most rich information sources of data to meet her or his needs. Another step, not necessarily sequential, is the process of putting back together data that were split during open coding (Corbin and Strauss, 2014). This process is called axial coding and it is used to relate categories to their subcategories to outline more precise and complete elucidations of explored scenarios (Corbin and Strauss, 2014). Throughout the process of data collection and analysis, the researcher is encouraged to write memos of theory development. This means writing any thoughts, interpretations, questions and directions for further data collection (Corbin and Strauss, 2014). An overview of grounded theory is shown in Figure 2.4 provided by Birks and Mills (2011, p. 37), who illustrated the grounded theory method as three cogs that can drive a researcher to generate new theory.

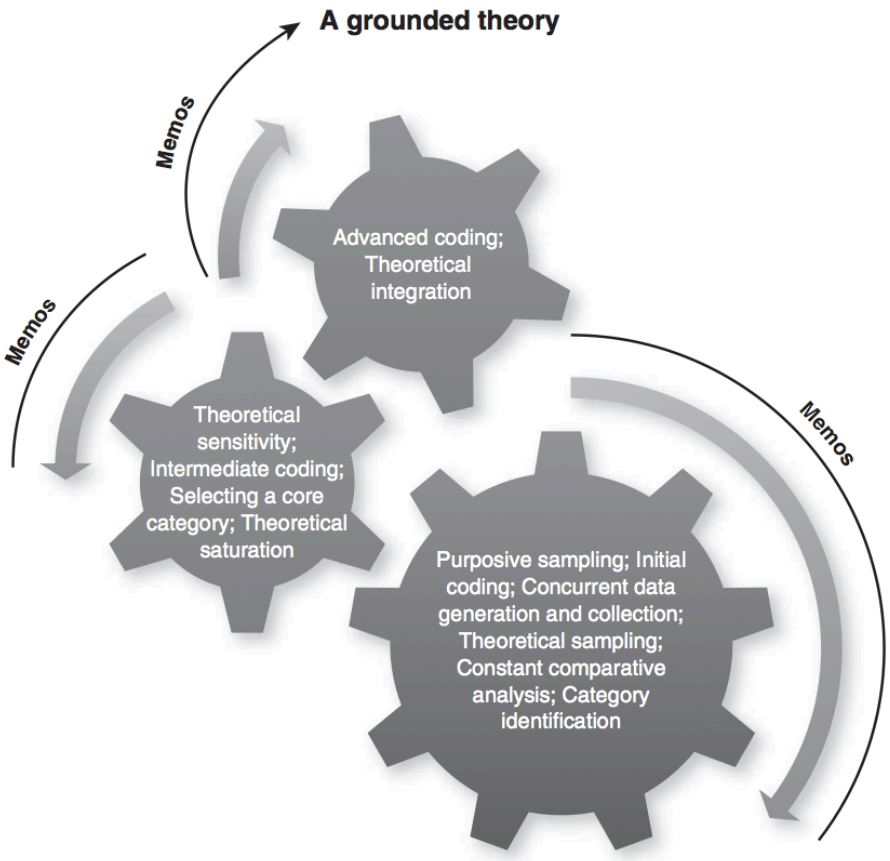
Grounded theory provides a systematic and rigorous method of data collection and analysis. However, it does have a number of limitations, as is the case with all research methods. One of the problems associated with allowing new theories emerging from data is that the role of the researcher is not analysed with sufficient attention, because data are seen as self-explanatory (Birks and Mills, 2011; Coolican, 2014; Corbin and Strauss, 2014).

Another facet of grounded theory which has been criticised is that questions of reflexivity are not addressed satisfactorily. In this regard, Dey (1999, p. 104) affirms that

> '*even if we accept the (doubtful) proposition that categories are discovered, what we discover will depend in some degree on what we are looking for – just as Columbus could hardly have 'discovered' America if he had not been looking for the 'Indies' in the first place*'.

Therefore, whatever emerges from the data is influenced by the researcher. To enhance rigour and reduce the impact of this on the eventual findings, Pidgeon and Henwood (2004) recommend that the researcher documents each phase of the research process, for example through memo-writing. This increases reflexivity throughout the research process and demonstrates the ways in which the researcher's assumptions, views and beliefs have shaped the research (Pidgeon and Henwood, 2004).

FIGURE 2. 4 ESSENTIAL GROUNDED THEORY METHODS



Despite these limitations, grounded theory is considered to be a suitable method of analysis for many reasons. While other theoretical perspectives emphasise theory developed by consistent deduction from an *a priori* theory (Patton, 2002), grounded theory focuses on inductive strategies of generating theory.

I decided to use grounded theory because of the desire to step beyond the known and enter into the world of young adults, to see the world from their perspective. I resolved to use the methods described by Strauss and Corbin (2014) because my previous knowledge of some psychosocial concepts related to genetic testing would prohibit me from approaching the

subject without any previous knowledge, as Glaser suggested (Glaser, 1992). I also believed that a literature review was useful in order to make comparisons, enhancing sensitivity, providing descriptive materials, supplying questions for initial interviews, stimulating analytic questions and confirming findings or the reverse. Finally, it is important to attend to Becker, who said

> '*use the literature, do not let it use you*' (Becker, 1986, p. 149).

2.4.2 ETHICS APPROVAL

Ethics approval for the study was sought and obtained both from Plymouth University Faculty Research Ethics Committee, reference number 14/15-324 (Appendix 1), and St. Orsola-Malpighi Hospital Ethical Board, reference number 132/2014/O/Oss (Appendix 2). Specific ethical issues arising in this doctoral study are discussed in Section 2.4.5.

2.4.3 PARTICIPANTS

Participants were young consultands, who were invited to take part in the study if they were:

- aged 18-30 years
- without personal history of cancer
- members of families with a hereditary cancer predisposition
- able to give informed consent, and
- able to speak Italian or English fluently.

Young adults were excluded from the study if they were:

- clients counselled by the principal researcher.

2.4.4 RECRUITMENT PROCESS

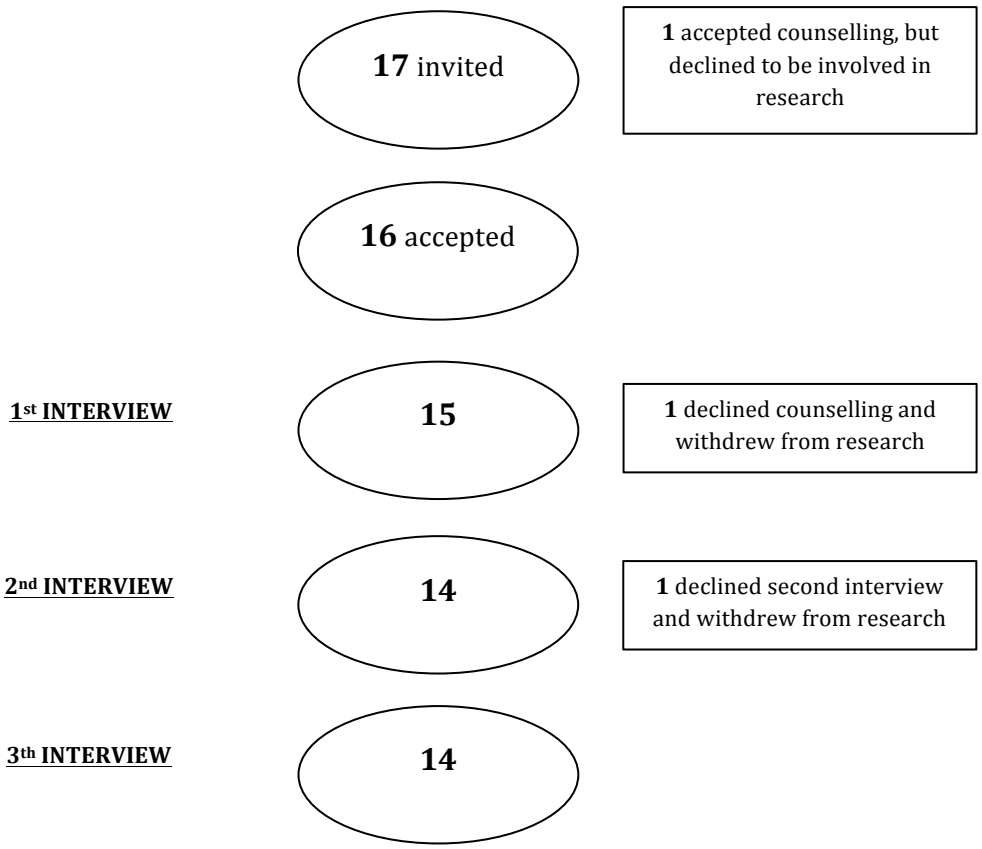
All participants were recruited at Genetics Unit of Bologna University Hospital Authority St. Orsola Malpighi Polyclinic (Italy). The Medical Genetics Unit staff were aware of the project before I applied for ethics approval because one of my supervisors is the geneticist who performs onco-genetic counselling and because I worked in that genetic clinic. After ethics approval was granted, I met my colleagues so that they were informed of the inclusion and exclusion criteria. I also asked them to identify and suggest potential participants. Every new young consultand making an appointment to the cancer genetics clinic in Bologna University Hospital Authority St. Orsola Malpighi Polyclinic, Italy, was contacted before the consultation via telephone and invited to take part in the study. I informed them of the study and I asked for their email address to send the invitation. The invitation consisted of a letter from the Medical Genetics Unit describing patient involvement in the study and a patient information sheet. The young adult consultand information sheet (Appendix 3) 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
- my contact details so that the potential participant could ask me any further questions they had about the study or inform me that they would like to be involved.

The potential participant was asked to reply via email whether they wanted to participate or not. If the potential participant did not want to be

involved in the study they were invited to say why, because knowledge of the reasons why participants were unlikely to participate could help inform recruitment for future research. However, due to sensitivity to their wishes, potential participants were only invited to participate in this study once. If they did not respond to the initial invitation they were not contacted again. Figure 2.5 shows the recruitment process.

FIGURE 2. 5 THE RECRUITMENT PROCESS



2.4.5 ETHICAL ISSUES

When we speak about psychological research we need to follow strict ethical principles, such as those devised by the British Psychological Society (2014) and the American Psychological Association (2003, amended 2010). Issues applying to research participants are described in Figure 2.6 (Coolican, 2014).

FIGURE 2. 6 DESCRIPTION OF ISSUES APPLYING TO RESEARCH PARTICIPANTS (COOLICAN, 2014)

ANONYMITY

- Keeping participant's identity concealed in any publication and avoiding any possible inadvertent disclosure.

CONFIDENTIALITY

- Keeping any data that could identify participants as confidential.

DEBRIEFING

- Informing participants about the full nature and rationale of the study they have taken part in and attempting to reverse any negative influence on them that has occurred as a result of their participation.

DECEPTION

- Leading participants to believe that something other than the actual research question is being investigated, or withholding information such that the nature of their involvement.

INFORMED CONSENT

- Ensuring participants agree to be involved in the study in the full knowledge of the research context and participant rights.

RIGHT TO PRIVACY

- Right that upholds people's expectation that their personal lives will not be intruded upon by voluntary or involuntary research participation.

In my study, a patient information sheet was sent to potential participants explaining the research. The patient information sheet explained that, if any harm arose as a result of the interview and the participant or I felt it was appropriate, the participant would be put in contact with a psychologist for further support. The information also included the fact that the participant could change his or her mind about participating in the study at any time without giving a reason and could request that their interview data (digital recordings and transcripts) were removed from the study at any time up to four weeks after the interview. My contact details were included on the patient information sheet in case the potential participant wanted further information.

Before the interviews began I explained once again the reason for the study and I answered any questions. I also sought permission to record the interview and I asked the participant to sign a form to record their consent. The consent form stated that they had read and understood the patient information sheet, understood that their participation was voluntary and agreed to take part in the study. In the information sheet, I explained that the interviews were digitally recorded so I could ensure participants' views were recorded accurately but I would change all names or other details so participants could not be identified in the final report. In this way, participants' personal details were kept completely confidential. Participants were informed also that their health care would not be in any way affected if they decided not to be involved in the study.

All interview data including digital audio-files and transcripts were kept in a secure office and on an encrypted memory stick to which only I had access. To safeguard confidentiality, all participants were assigned an

identification number and a pseudonym, so that their identity was not known to anyone other than myself.

2.4.6 DATA COLLECTION

Face to face interviews were organised with participants who responded to an invitation to be involved in the study. The data collection method of one-to-one semi-structured interviews (Hansen, 2006) was chosen to provide young adults with a means by which the researcher could direct the conversation towards areas that were important to the study. However, data collection using focus groups was also considered. Unlike one-to-one interviews, focus groups allow researchers to capture group interaction between participants and to make use of interaction as a way of prompting discussion (Kevern and Webb, 2001). I chose to use one-to-one interviews because focus groups could have posed additional ethical challenges related to confidentiality and anonymity (Hansen, 2006). In addition, interviews are more likely to enable exploration of individual experiences (Kevern and Webb, 2001). In fact, throughout the interviews, young adults were invited to describe their emotions, experiences and the psychosocial implications of presymptomatic testing for hereditary cancer. Each interview began with questions regarding demographic information. Later sections were designed to understand the attitudes of young clients, to evaluate their cancer perception and psychological status and to explore the extent to which the parents' influence had been important. In addition, questions were refined and amended over the course of the interviews to take into account possible theories emerging from the data. The interviews

were written in Italian (English version in Appendix 4) and each lasted between 10 and 45 minutes. Data were collected using a digital recording device and interviews were transcribed verbatim, with names and other identifying material altered to ensure confidentiality.

2.4.7 DATA ANALYSIS

Data were analysed using the grounded theory method described by Strauss and Corbin (2014). In accordance with grounded theory analysis, each interview transcript was analysed as soon after transcription as possible. To facilitate the analytical process, the software package NVivo, version 10, was used (QRS international, Pty, Ltd). This programme facilitates the indexing and retrieval of data. All coding and categorising was done using NVivo: an example of coding can be found in Appendix 5.

First I listened to the digital recordings and I transcribed the interviews, so that ideas about the newly collected data were fresh in my mind. Also, I made choices about the words I used to label the ideas or themes that I saw occur repeatedly in the study data, because coding is a central and important process in qualitative analysis (Richards and Morse, 2002). At this stage of the process, data were coded either into pre-existing codes or new codes. The next step was integrating these codes into something more compact and coherent so that I was able to make sense of them, before grouping them into categories. This was facilitated using NVivo by creating tree nodes. An example of axial coding can be found in Appendix 6. During this process any overall thoughts, interpretations or questions relating to the data were noted as memos. I translated 21 interviews into

English and I sent them to one of my supervisors and my Director of Studies to code independently. Translations were checked by my Italian supervisor. All three interviews conducted with one young woman and one young man both in Italian and English language can be found in Appendices (English version in Appendix 7 and Appendix 8; Italian version in Appendix 9 and Appendix 10). The codes and emerging categories derived by the supervisors were then compared with mine to ensure trustworthiness of the findings. Any disagreements were discussed until consensus was reached. Finally, I interpreted the data further by grouping categories into major themes in order to synthesise and understand the relationship between data from all participants. After the first 21 transcripts were analysed, I started to apply theoretical sampling (Corbin and Strauss, 2014) by recruiting more young adults aged under 25 years to better understand the experience of this subgroup of participants. This then enabled me to consider the experiences across these two groups of participants (under 25 years and over). Theoretical sampling allowed me to explore issues and problems from different points of view. The process of recruitment, interviews and data analysis was ongoing until saturation (Corbin and Strauss, 2014) was reached and no new categories were emerging. This was achieved after 16 months and 28 interviews. At this point all my colleagues at the Medical Genetics Unit were informed and no more invitations were sent out.

The results from the qualitative phase and discussion of the findings are presented in Chapter Four.

2.4.8 ENSURING RIGOUR

It is felt that qualitative research is subjected to researcher bias, to lack reproducibility and to lack generalizability: in fact, generally, it is criticised for lacking scientific rigour (Mays and Pope, 1995). I conducted all of the individual interviews to ensure that the participants were subject to a constant interviewer effect. All participants received the same information via email prior to the meeting. No leading questions were asked and when participants were interviewed this was done using open-ended questions such as "Please, tell me more about …". I transcribed all the data, which I considered was the best way to represent the data as consistently and accurately as possible. For analysis, recorded data were combined with notes written at the time.

The use of contemporaneous notes kept by the researcher increases reflexivity throughout the research process and demonstrates the ways in which the researcher's assumptions, views and beliefs have shaped the research (Pidgeon and Henwood, 2004). Many formal approaches have been described for keeping research notes. For example, Burgess (1981) suggests including autobiographical details about the research, Pope and Mays (2006) discuss the use of a personal research diary to record the researcher's reactions during the research, together with personal and intellectual biases, and Gibbons et al. (1986) suggest the recording of reflexive data next to notes. Although being guided by these approaches, I used an informal approach to the reflexive notes and made *ad hoc* notes thoughout the data collection and analysis.

The analysis of the data, as mentioned previously, was by grounded theory (Corbin and Strauss, 2014). Independent coding of several transcripts was carried out by two of my supervisors, who are experienced researchers, to maximise the validity of my analysis. Furthermore, the grounded theory approach had the advantage of permitting me to go back and refine questions, develop hypotheses, and enable me to look for negative or deviant cases as suggested by Pope et al. (2000). Karl Popper, the philosopher of science who developed the empirical falsification theory regarding scientific method, declared that even if any negative cases were not found, the researcher could not assume they did not exist (Miles and Huberman, 1984). Being able to seek negative or deviant cases helps the researcher to refine the hypothesis being developed. Thoughout the analysis process findings may emerge that require the purposeful selection of participants to reinforce or amend any theory being developed: this is called theoretical sampling by Strauss and Corbin (2014). Moreover, the grounded theory approach takes this even further, saying that the researcher should actually look for negative cases (Corbin and Strauss, 2014).

2.5 QUANTITATIVE PHASE (PHASE 3)

2.5.1 RESEARCH DESIGN

For this phase of the study I used a quantitative design. Quantitative research is a deductivist and objectivist approach and incorporates a natural science model of the research process (Bryman, 2016).

There are a number of possible quantitative designs within a hierarchy in which a randomised controlled trial is considered the most robust level of data collection (Akobeng, 2005). As a randomised controlled trial was not feasible in this situation. I chose cross-sectional design in order to collect a body of quantitative data in relationship with two or more variables (Bryman, 2016). Specifically, the study design was a cross-sectional self-completion survey (Mann, 2003) to investigate the experiences of young adults and their parents undergoing genetic counselling and presymptomatic testing for cancer. I chose to write questionnaires both in the Italian and English languages. This enabled me to explore the experiences of Italian participants because, to the best of my knowledge, there is no published literature on an Italian sample and it was also then possible to compare the Italian findings with the experiences of other young adults.

2.5.2 PARTICIPANTS

The participants were young adults and their parents. Those who were eligible to take part in the study fitted either of the two groups below.

1. Young adults who were:

- aged 18-30 years when they underwent the presymptomatic genetic test for a familial cancer syndrome
- without personal history of cancer when they underwent a presymptomatic genetic test and
- members of families with a hereditary cancer predisposition.

2. Parents of young adults who were tested between 18-30 years of age.

2.5.3 ETHICS APPROVAL

Ethics approval was sought and obtained both from St. Orsola-Malpighi Hospital Ethical Board, reference number 198/2015/O/Oss (Appendix 11), and Plymouth University by Faculty Research Ethics Committee, reference number 15/16-519 (Appendix 12). Further details on the way in which ethical issues were addressed are included in Section 2.5.6.

2.5.4 RECRUITMENT PROCESS

Data were collected using i) online questionnaires uploaded to the Survey Monkey® website and ii) paper versions of the same questionnaire. The surveys were open to respondents between 23 December 2015– 30 June 2016.

As shown by Jones et al. (2008) online questionnaires have similar strengths to their hard copy equivalents, such as the low cost of implementation and the ability to reach a larger number of the target population than would be possible via interviews. However, hard copy or email responses subsequently have to be manually entered into the

dedicated database, which is considered to be quite an arduous and time-consuming task, with a potential for transcription error (Jones et al., 2008). Online questionnaires have features that minimise such disadvantages; specifically, online surveys can streamline the data process by the responses being directly submitted into a dedicated database. Nonetheless, web-based questionnaires are noted for the initial expertise required to configure the survey (Jones et al., 2008), for historically lower response rates (Solomon, 2001) and the alienation of some respondents who are reluctant to use the Internet (Denscombe, 2014). One potential limitation of online questionnaires has been recognised as coverage due to accessibility of the Internet (Fricker and Schonlau, 2002).

I decided to use both online and traditional methods of recruitment and data collection because, although online surveys are a convenient way of collecting data from a wide range of people (Dillman et al., 2014), there is evidence that many members of the Italian population do not use the Internet regularly. The Istat and Ugo Bordoni Foundation (Gruppo di Lavoro congiunto Istat-FUB, 2014) discovered in research aimed at understanding which Italians use the Internet that the Italian population is divided into three main categories: 1) "strong" users of the Internet (people who connect to the Internet every day), approximately 19 million people, 33.1% of the total; 2) non-users, over 23 million people, about 40% of the total; 3) 'weak' users, who connect to the Internet at least once a week, comprising about 20%, of the population. Those authors also found there were some differences in socio-demographic variables between the groups. In particular, there was a strong difference in respect to age between users and non-users: the use of the Internet was very high in the age groups 14-18

years (85%) and 19-34 years (78%), lower in the age group 35-54 years (62%), and even lower after 55 years of age with 39% in the age group 55-64 years and 10% in the group aged over 65 years. Another variable they considered was educational qualifications: 84% of people with graduate level education were likely to use the Internet, compared with only 18% of people with elementary education or less. As my study sample comprised young adults and parents of young adults it was unlikely, in the light of my study sample age, that they would have not been reached. However I decided that it was essential to use online and traditional approaches to data collection to ensure that participants who wanted to take part were not excluded because of lack of access to the Internet. The figure below (Figure 2.7) indicates the overall Internet use in other European countries, which is in general higher than in Italy. Of note, in 2013 85% of UK citizens are reported to use the Internet compared to 32% of Italians (Eurostat Statistics Explained, 2013).

FIGURE 2. 7 INTERNATIONAL COMPARISONS OF INTERNET USERS IN EUROPE

	Internet users and non-users			Frequency of use (on average)	
	Used internet within the last 3 months	Used internet within the last 12 months	Never used internet	Every day or almost every day	At least once a week (including daily use)
EU-28	75	77	21	62	72
BE	82	83	15	68	80
BG	53	56	41	43	51
CZ	74	76	17	54	70
DK	95	95	4	84	91
DE	84	86	13	68	80
EE	80	82	16	63	77
IE	78	80	18	61	75
EL	60	61	36	47	56
ES	72	74	24	54	66
FR	82	84	14	66	78
HR	67	68	29	53	63
IT	58	61	34	54	56
CY	65	66	32	53	62
LV	75	76	22	60	71
LT	68	69	29	53	65
LU	94	95	5	82	93
HU	73	74	24	62	71
MT	69	70	28	59	66
NL	94	94	5	83	92
AT	81	82	16	63	77
PL	63	65	32	47	60
PT	62	65	33	48	58
RO	50	55	42	32	45
SI	73	74	23	58	69
SK	78	81	15	61	74
FI	92	92	6	80	89
SE	95	95	4	81	92
UK	90	91	8	78	87
IS	97	97	(3)	91	95
NO	95	96	(3)	85	93
TR	43	46	51	30	40

( ) Data with reduced reliability due to small number of respondents.

2.5.4.1 SPECIFIC METHODS OF RECRUITMENT USED

In this section, I will discuss the various methods of recruitment that were used involving social media and more traditional methods.

Facebook description

Facebook was launched to facilitate online communication for Harvard students in 2004, when its founder Mark Zuckerberg was a Harvard sophomore student (Dobinick, 2013). It is now estimated that Facebook has 1550 million active users (Statista, 2016). Messages can be posted on an individual Facebook profile, Facebook pages and/or Facebook groups. Specifically, Facebook pages are created by an individual or institution about a specific topic, and messages can be posted by the administrator, while

Facebook groups can also be created by an individual or institution about a specific topic, and consist of members who share the group interest. Membership in open groups is available to the Facebook public. Conversely, in closed groups, membership is only available to users who met the group administrator's specified criteria (https://www.facebook.com, accessed on 4th January 2016).

Facebook recruitment

Recruitment was conducted by posting recruitment messages, in both Italian and English, to open Facebook groups that I felt would have members who were interested in the study.

Example of English message posted:

"Hello, I'm a PhD student at Plymouth University. I'm carrying out a survey on motivations and the impact of genetic testing for hereditary cancer in young adults. Please read the flyer below: if you satisfy the requirements and are willing to help me, please follow the link https://www.surveymonkey.com/r/PhDENG and complete the survey. Thank you!".

The English version of the flyer advertising study is showed in Appendix 13.

Twitter description

Twitter was launched as a social communications platform in 2006 by Jack Dorsey, Evan Williams, Biz Stone, and Noah Glass (https://twitter.com, accessed on 4th January 2016). It is now estimated that Twitter has 316 million active users (Statista, 2016), generating approximately 500 million tweets per day (Internet Live Stats, 2016). As a form of micro-blogging, Twitter uses messages, called tweets, of a maximum of 140 characters. Twitter users can send short messages (tweets) to share online material. In the tweet there are often words or collections of word preceded by a hash symbol (#) called a hashtag, that indicated topics of interest that can be searched for in other tweets (https://twitter.com, accessed on 4 January 2016). Also, tweets can be tagged to specific Twitter user or Twitter organizations using the '@' symbol before their Twitter name. Twitter users can then 'retweet' the messages, that is to say share these tweets with their own followers (https://twitter.com, accessed on 4th January 2016).

Twitter recruitment

Recruitment consisted of tweeting recruitment text in both Italian and English to other Twitter users that I felt might be interested in the study. In a recent study, it has been shown that Twitter could be used in health research (O'Connor et al., 2014). I asked Twitter users to retweet my original tweet containing the link to the online survey to their own followers and this activity was continued throughout the duration of the study.

Examples of English tweets posted:

"Have you undergone #cancer #GeneticTest? Seen this? Pls help and do our online survey goo.gl/gkB3iT #BRCA #LynchSyndrome

What is your experience about #GeneticTest? Pls help and do our online survey goo.gl/gkB3iT Can you RT pls?

Are you a #parent? Did you undergo #GeneticTest? Can you help with our survey and RT? goo.gl/gkB3iT

Are you a #YoungAdult? Did you undergo #GeneticTest? Can you help with our survey and RT? goo.gl/gkB3iT ".

Google+ description

Google+ was launched as an invitation-only social network site in 2011 by Google (Gonzalez et al., 2013). It is now estimated that Google+ has 2.2 billion active profiles, but only 6% of those have any post activity in 2015 (Barrie, 2015). Google+ offers a combination of Facebook- and Twitter-like services (Gonzalez et al., 2013). Google+ uses messages, called posts, that may have attached files. Other Google+ users can react to post with '+1 button' (similar to the 'like button' in Facebook) with which other Google+ users can indicate their interest in a post, with comments on a post and/or a 'reshare' (similar to a retweet in Twitter), with which other Google+ users can share a post to their followers (Gonzalez et al., 2013).

Google+ recruitment

Recruitment consisted of posting recruitment text in both Italian and English to other Google+ users who may have been interested in the study. I asked Google+ users to reshare my original post containing the link to the online survey to their own followers and this activity was continued throughout the duration of the study.

Examples of English posts posted:

"Hello, I'm a PhD student at Plymouth University. I'm carrying out a survey on motivations and the impact of genetic testing for hereditary cancer in young adults. Can you help me with a reshare please? Pls, read the flyer below: if you satisfy the requirements and are willing to help me, please follow the link https://www.surveymonkey.com/r/GPhDENG and complete the survey. Thank you!"

Traditional recruitment

Traditional recruitment was used only at the Medical Genetics Unit of Bologna University Hospital Authority St. Orsola Malpighi Polyclinic (Italy). The Medical Genetics Unit was informed of the study before I applied for ethics approval because one of the supervisors is the geneticist who provides onco-genetic counselling and because I worked in that genetic clinic. After ethics approval was granted, I informed my colleagues of the inclusion and exclusion criteria and asked them to identify potential participants. Every young adult (or parent of a young adult who had been tested) who met the inclusion criteria and was being contacted by the cancer genetics clinic in the

Bologna University Hospital Authority St. Orsola-Malpighi Polyclinic, Italy, for a follow-up session, was invited to take part in the study. I informed them of the study and, if they were interested, I asked them for their email address or home address so I could send them an invitation.

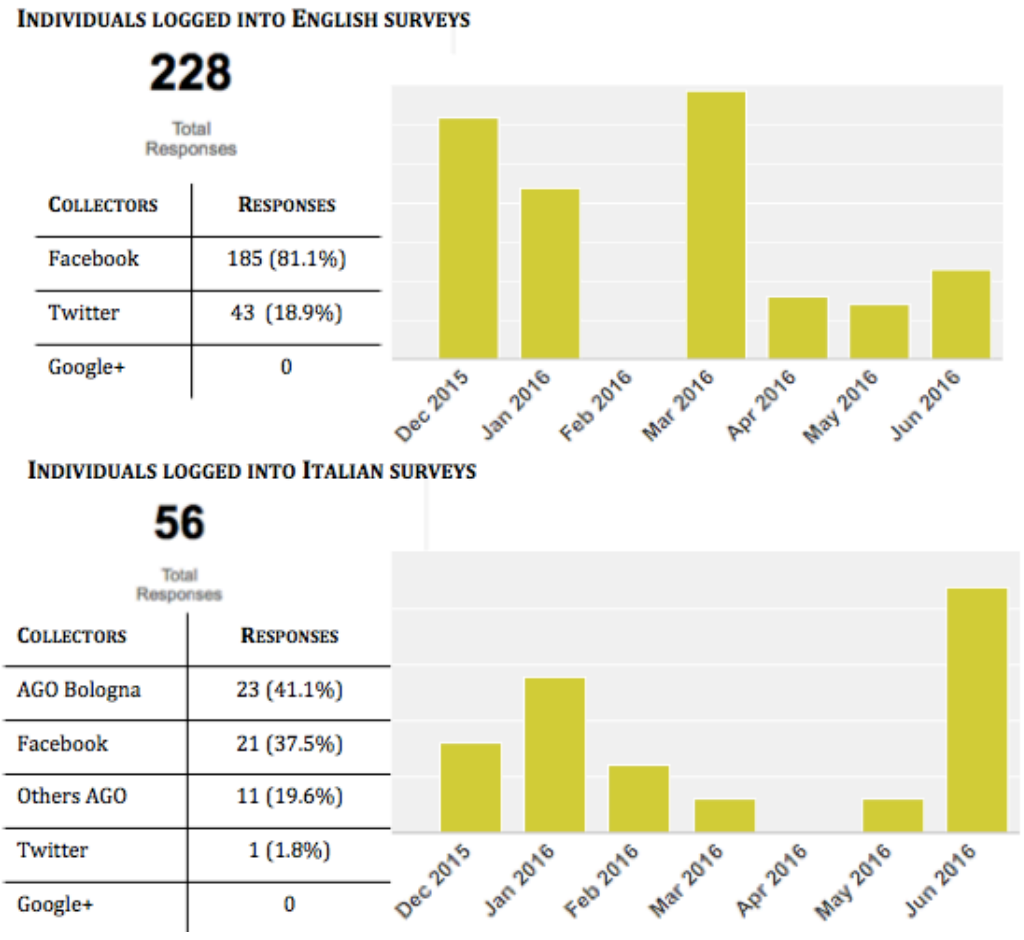
The email invitation consisted of a letter from the Medical Genetics Unit stating their involvement in the study and a patient information sheet. The potential participants were asked to follow the link https://www.surveymonkey.com/r/APhDITA (in the email text) and complete the survey. I also asked them to inform me via email whether or not they wanted to participate. If the potential participant did not want to be involved in the study, it was stated that this was considered useful information because knowledge of the reasons why participants were unlikely to participate could help inform recruitment for future research. Potential participants were only invited once to participate in this study: if they did not respond to the initial invitation they were not contacted again.

In addition to this approach, paper questionnaires were sent to the home address of each eligible young adult and their parents within a package including an invitation letter, the participant information sheet, the consent form and a prepaid envelope to return the filled forms to me. In order to obtain more Italian responses, I also asked Italian colleagues in other clinics via the "Gruppo di Genetica Oncologica Clinica" (Genetic Oncology Clinical Group) to share my research flyer with clients in their clinics.

Using different collectors created in Survey Monkey® I recorded the number of individuals who had accessed the surveys based on the different routes of access. Of those who visited the English survey site, 185 came via a

link from Facebook, 43 from Twitter, and 0 from Google+. Of those who visited the Italian survey site, 23 came via a link from the flyer emailed to clients of the clinic in Bologna, 21 from Facebook, 11 from emails sent by colleagues in different Italian genetic centres, one from Twitter, and 0 from Google+ (Figure 2.8). Of the six young adult questionnaires and four parent questionnaires mailed, none were returned.

FIGURE 2. 8 INDIVIDUALS LOGGED INTO SURVEYS



AGO is abbreviation of "ambulatorio di genetica oncologica" that means cancer genetics clinic.

On the next page the recruitment flow-chart is presented (Figure 2.9).

FIGURE 2. 9 RECRUITMENT FLOW-CHART

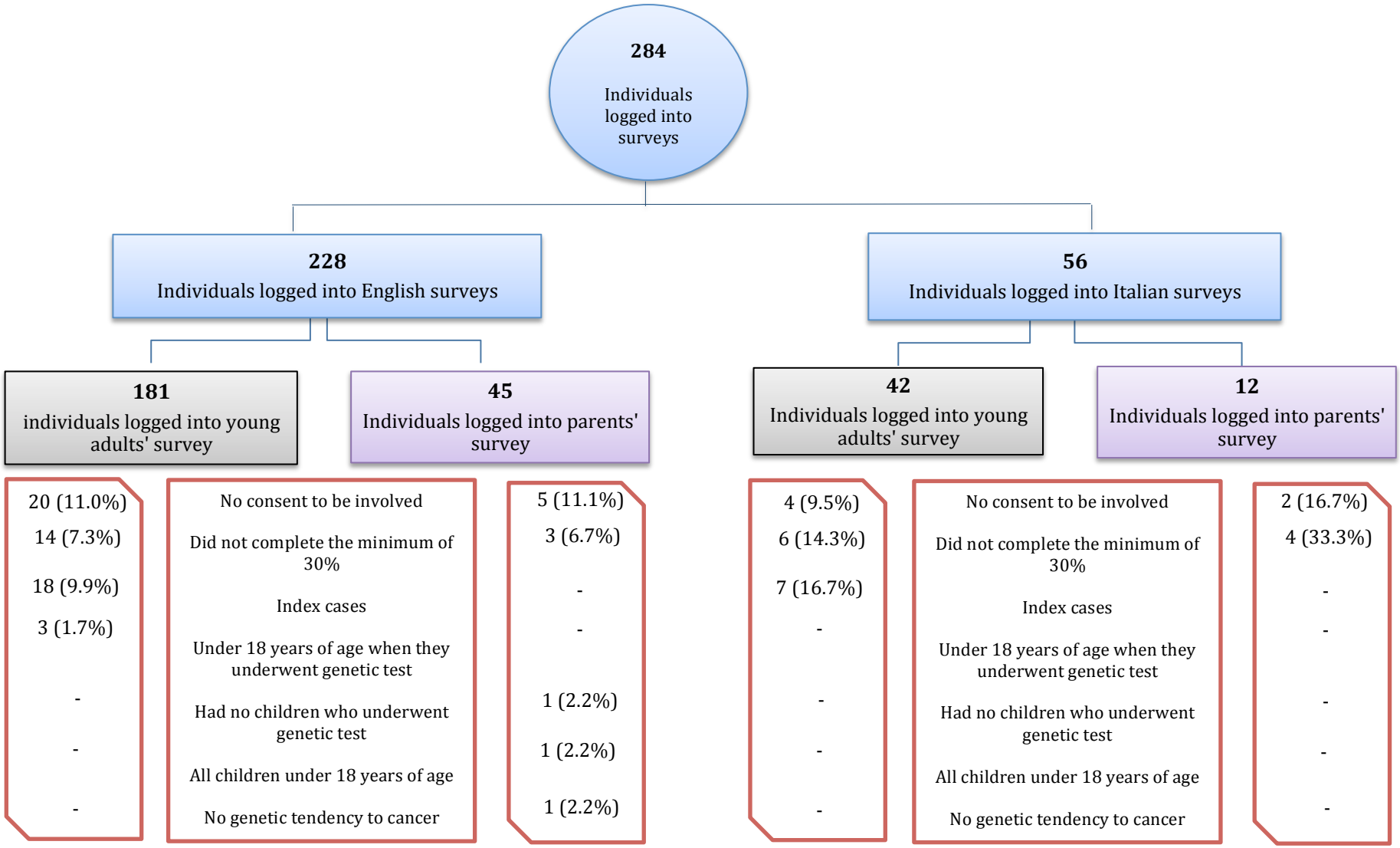
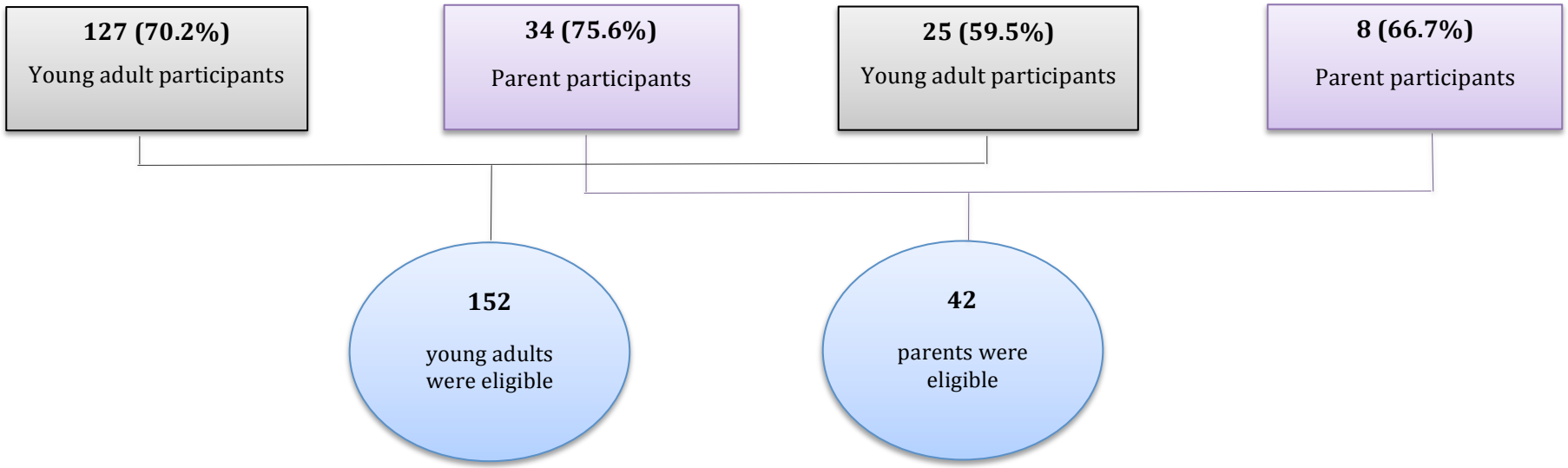


FIGURE 2.9: RECRUITMENT FLOW-CHART (continue)



2.5.5 QUESTIONNAIRES

In the systematic review of this topic (Chapter Three), I was unable to identify a validated survey tool. I utilised the findings of other phases of this mixed methods study to derive questions. The surveys are therefore based on the results of the systematic review (Godino et al., 2016) and a qualitative study of young people's experiences of presymptomatic testing (Phase 2 of this doctoral project) as well as questions in other similar surveys (Shiloh et al., 1990; Bruno et al., 2004). Because it was important to also investigate the parents' point of view, two questionnaires were designed. The questionnaires were written both in Italian and English (English version in Appendix 14 and Appendix 15). My Italian supervisor (who is bilingual) and I checked cross-translations.

The young adult questionnaire began with questions regarding the demographic characteristics of respondents and their parents and their genetic status. Later sections were designed to examine the respondent's experiences in the period prior to genetic testing, the experience of genetic counselling, involvement of parents in decision making, impact of personal test results and finally living with genetic risk (English version in Appendix 14).

The parent questionnaire began with questions regarding demographic information of respondents, their genetic status and their partner's genetic status. Later sections were designed to test how and why they told their young adult children about the genetic risk, their perception of their adult child's experience of the genetic test and their feelings about genetic testing for their children (English version in Appendix 15).

When draft questionnaires had been developed, I used a Think-Aloud technique (Ericsson and Simon, 1993), to test the understanding and relevance of the questions with a small sample of young adults and their parents before the main study began. Those data were used to ensure validity of the tool and did not contribute to the overall findings of the study.

For each participant I noted down any information they gave, such as when they asked for clarification of the question. After completing the questionnaire, each participant was asked for their opinion about the survey, including:

- what they thought about the questions in general
- whether any of the questions seemed to be strange or unusual
- their opinion on the order of questions
- whether any questions should not be asked in the survey
- to point out any questions they do not want to answer, or think they should not be asked, and
- whether two or more questions appeared to be asking the same thing.

Based on feedback during using a Think-Aloud technique (Ericsson and Simon, 1993), minor revisions were made to the questionnaires.

The English version of the first webpage on Survey Monkey® is included in Appendix 16.

2.5.6 ETHICAL ISSUES

As I described in the previous section (see 2.4.5 Ethical issues section), I followed the ethical principles for research (American Psychological Society, 2003; The British Psychological Society, 2014).

Below I detail the ethical considerations for my recruitment approach, both online and traditional. For the online recruitment, any individual entering the survey site was able to leave the site without entering data or to leave when the survey was only partly completed. Respondents were asked to signify their consent to be involved at the start of the survey and were unable to progress to the questions unless they had recorded their consent. Data were only submitted to the survey if the respondent gave consent and also specifically pressed the 'submit' button.

To ensure anonymity and confidentiality, the participants did not need to provide any identifying details linked to their questionnaire. Using this system, personal data were completely disconnected from the data collection process. Survey Monkey® data can only be accessed by the registered account holder and is protected using a username and password.

For the 'traditional' recruitment (via clinics), a patient information sheet was sent to potential participants explaining the study. The information sheet explained that if any harm arose as a result of the questionnaire, the participant would be put in contact with a psychologist for further support, with the participant's consent. It was also explained that the participant could change their mind about participating in the study at any time and request that their questionnaire data were removed from the study up to four weeks after the questionnaire was returned. My contact details

were included on the patient information sheet for use if the potential participant wanted further information.

All questionnaire data were kept in a secure office and on an encrypted USB memory stick, to which only I had access. To safeguard confidentiality, all participants were assigned an identification number, so that their identity was not known by anyone other than me.

2.5.7 DATA ANALYSIS

The analysis was conducted using the data derived from the young adult questionnaire and the parent questionnaire. The data were entered into two respective dedicated databases. Before analysis could be performed, the data were coded: this comprised assigning numerical codes to responses that were not already in numerical form (e.g. 1=Male, 2=Female, 3=I prefer not to say). During this process, codes and abbreviated variable names were documented. When this had been completed, the data were checked for errors. Frequencies, minimum and maximum values, means scores and valid and missing cases were checked and any unusual values examined and corrected if an error had been made. The data were then analysed using the Statistical Package for the Social Sciences (SPSS) Ver. 21.0 for Windows (IBM Corporation, Armonk, NY, USA).

Descriptive statistics were used to determine the means, standard deviations, percentages and frequencies of variables. The Chi-squared test for independence, also called Pearson's chi-square test, was used to discover if there was a relationship between two categorical variables (Azzalini, 2001). The Fisher's exact test for independence was used to discover whether the

proportions of one categorical variable were different, depending on the value of the other categorical variable (Azzalini, 2001). The Independent Samples t-test (or Independent t-test) was used to compare the means between two unrelated groups on the same continuous, dependent variable (Azzalini, 2001). The Independent t-test assumes equal variances of the two groups. The variance indicated how widely members in a group vary. The variance will be large if the member observations vary greatly from the group mean, and *vice versa* (Cicchitelli, 2014). If variances are unequal, this can affect the Type I error rate. This type of error occurs when the researcher rejects a null hypothesis when it is correct. In other words, the researcher accepts an alternative hypothesis (the real hypothesis of interest) when the results can be attributed to chance. The probability of committing a Type I error is called the significance level, and it is often denoted by α (Cicchitelli, 2014). The assumption of homogeneity of variance was tested using Levene's Test of Equality of Variances. If the Levene's Test for Equality of Variances was statistically significant, indicating unequal variances, the Independent t-test was not performed. To assess whether there were any significant differences between the means of two or more independent groups, a one-way analysis of variance, also called one-way ANOVA, was performed (Azzalini, 2001). If the dependent variable was normally distributed in each group being compared, there was a homogeneity of variance (this means that the variances in each group were equal), and the independence of observations were the three main assumptions for using the one-way analysis of variance. Because the one-way analysis of variance could not tell me which specific groups were significantly different from each other, but only indicate an overall difference between the groups considered,

post hoc tests were also performed. However, post hoc tests were usually performed to confirm where the differences occurred between groups. These tests should only be run when the one-way analysis of variance has been shown an overall statistically significant difference in group means. There are a great number of different post hoc tests suitable. Although the Bonferroni post hoc test was a conservative measure, it was chosen and performed to enhance reliability of the results.

Exploratory factor analysis was then carried out. Reducing the number of variables (from large to small), establishing underlying dimensions between measured variables and constructs, and providing construct validity evidence were the three main reasons for using this multivariate statistical technique (Mignani and Montanari, 1997). Exploratory factor analysis was performed using the Kaiser-Meyer-Olkin (KMO) index of sampling, the analysis of the principal components (Kaiser, 1974), and Bartlett's test of sphericity to determine appropriateness for factor analysis (Bartlett, 1954). The KMO was used to assess the sampling adequacy: the results of the KMO analysis lie between 0 and 1. Sampling was considered adequate if KMO was higher than 0.6. Additionally, Bartlett's test of sphericity was related to the significance of the sample and by using that test it was possible to demonstrate the validity and suitability of the responses collected for the purpose being addressed through the study. The result of the Bartlett's test of sphericity must be less than 0.05 to indicate validity.

According to Cattell (1978), a minimum of three cases per item is considered necessary to perform factor analysis. The Scree Plot and

eigenvalue methods were used to calculate the number of factors (Zwick and Velicer, 1986). The criterion used for the classification of the factors was an inflection point of 0.4 as the minimum factor loading required to keep the item in the factors extracted through factor analysis, and eigenvalues greater than one were accepted. The orthogonal (varimax) and oblique (promax) rotation was used for the simplification and interpretability of the factor constructs (Mignani and Montanari, 1997). Throughout the study, P values were considered statistically significant when they were less than 0.05.

The results from the quantitative phase are presented in Chapter Five. All statistical test results are reported in the tables, but to aid fluency of the account those results that indicate a significant difference will be reported in the narrative alongside any highly relevant results where a significant difference was not found.

2.5.8 ENSURING RIGOUR

A pilot of the survey was conducted with five collegues, permitting an opportunity to test the online surveys and transference of data. The data were entered into two respective dedicated databases and were coded before performing the analysis. During this process, codes and abbreviated variable names were documented. When this had been completed, the data were checked for errors.

The SPSS syntax was used to ensure replicability. Furthermore, before performing any statistical test, the assumptions were considered as mentioned in Section 2.5.7. The statistical tests were perfomed only if the

assumptions were satisfied. The choice of statistical tests and the SPSS outcomes were assessed by my supervisors, who are experienced researchers, to maximise the validity of my analysis.

2.6 PREPARING A THEORETICAL MODEL

At the end of each phase I prepared a model to explain the findings. The theoretical model was built on consecutively during the doctoral research, as each phase within the sequential mixed methods design was completed. After the last phase, the final model was constructed.

2.7 IN SUMMARY

In this chapter I have described the mixed methods sequential exploratory design used in this doctoral project. It included a systematic review, a qualitative study, and a quantitative study. Initially I have discussed the choice of a mixed methods approach to the research in this doctoral study. I have described the process carried out in order to complete the systematic review. I have also provided a brief background to qualitative research methods, looking at their use within the health care setting, and justifying their use in this phase of the doctoral project. I then have presented the grounded theory method and discussed why I felt it was the most suitable method for this phase of the study. The quantitative research method with a critique and justification of the use of the method, as well as a description of the various methods of recruitment used involving social media and more traditional methods have been discussed. The ethical issues related to

quantitative design, and the procedures used to analyse the data have been also presented. Finally I have explained the process of building a theoretical model consecutively during the doctoral research.

The results from the data analysis for the systematic review, the qualitative phase and the quantitative phase will be presented in the following three chapters. In the final chapter, the overall theoretical model will be presented and discussed.

CHAPTER THREE

PHASE 1: SYSTEMATIC REVIEW

3.1 INTRODUCTION

To provide a comprehensive background to the empirical study phases of the doctoral research, a systematic review was conducted, focussing on studies assessing which factors influence young adults' or adolescents' choices to have a presymptomatic test for a genetic condition. The following sections provide a detailed account of the process carried out in order to complete the review for Phase 1 of the study.

3.2 AIM AND OBJECTIVES

The aim of the review was to systematically identify and analyse factors influencing young adults' or adolescents' choices to have a presymptomatic test (or not) and the emotional impact of those choices. The specific objectives were:

- to explore how young individuals interpret cancer presymptomatic testing

- to explore the basis for the young individual's decision to undergo testing or not
- to explore the influence that parents have in the choice young adults make about testing
- to analyse the psychosocial impact of test result disclosure, according to mutation status.

3.3 METHODS

The main justification for use of a systematic review in this study and the main methods used were included in the previous chapter, in Section 2.3. Here I will present in more detail the selection of studies, the search outcome, the quality appraisal, and the data abstraction and analysis.

3.3.1 SELECTION OF STUDIES

A complete list of reasons for excluding articles screened through reading the title and abstract is included in Figure 3.1.

Moreover, the principal reasons for exclusion of each study that seemed initially to meet our inclusion criteria but on closer inspection (i.e. reading the full text) did not do are documented in Appendix 17.

3.3.2 SEARCH OUTCOME

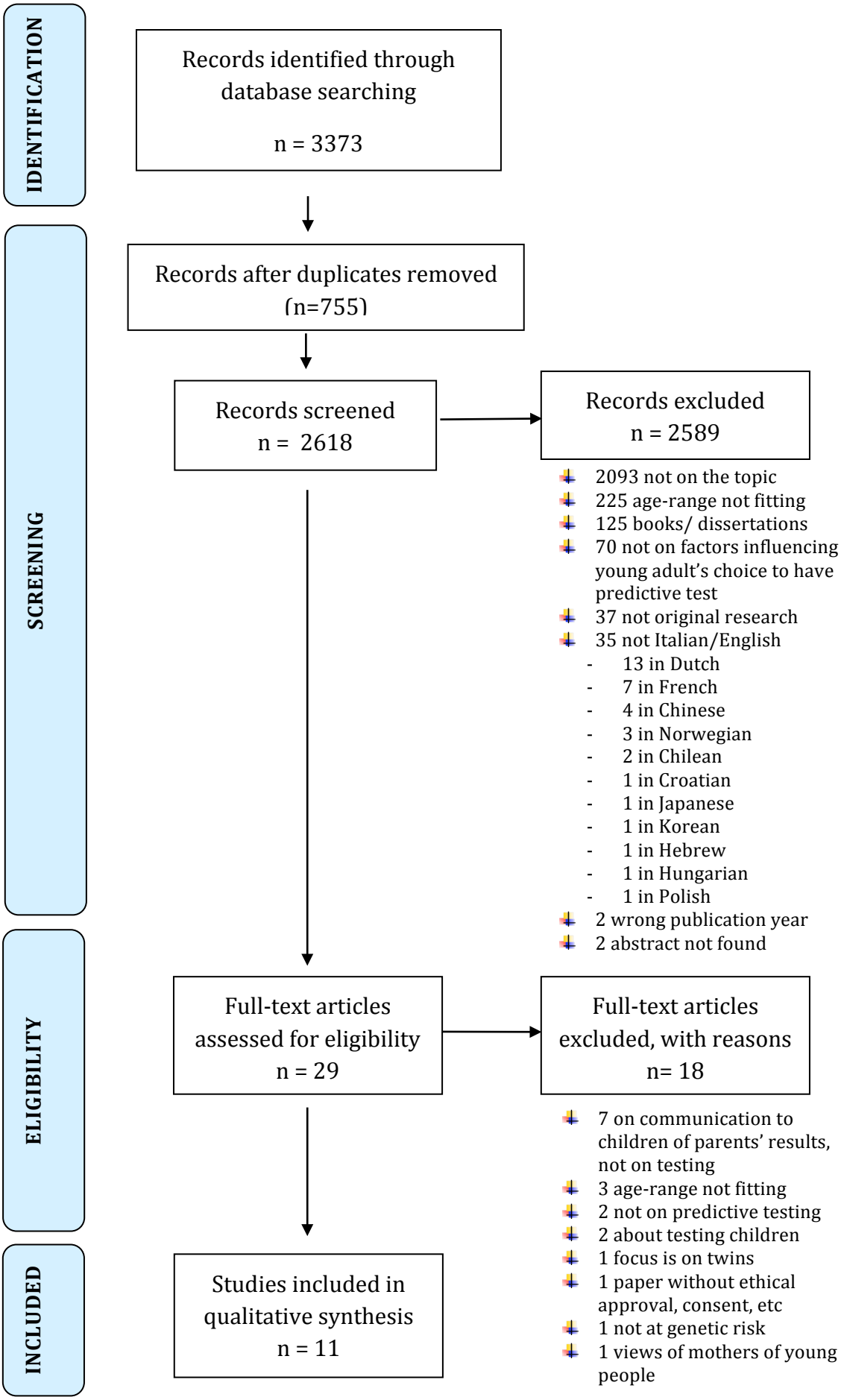
The Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) flowchart showing the consecutive methodological steps in this systematic review is displayed in Figure 3.1 (PRISMA, 2014). The search of

eight databases initially produced 976 potential papers. With the second search, 2397 papers were found. From the total of 3373 papers, 755 were duplicates, leaving 2618 for examination. Following review of the title and abstract, 29 papers were assessed as potentially relevant. These papers were read in detail by me and my supervisors. The eleven remaining papers were included in the review. The main characteristics of the included studies are presented in Table 3.1.

3.3.3 QUALITY APPRAISAL

All papers considered for inclusion criteria in the review were then subjected to independent analysis by the researcher and one supervisor (HS) using standard quality assessment criteria for evaluating original research papers from a variety of fields (Kmet et al., 2004). This evaluation method allows the systematic evaluation of both quantitative and qualitative original research and across a broad range of study designs. Specific aspects of the paper, relating to methodology and reporting of results are assessed and assigned 0 points (not addressed), 1 point (partially addressed) or 2 points (satisfactorily addressed) as detailed in Table 2.2.

FIGURE 3. 1 PRISMA flowchart of study selection



Any disagreements about scoring of papers were discussed. Although Kmet et al. (2004) do not enforce a minimum score for inclusion in a review, they suggest 60% as a reasonable cut-off point. However, according to current guidance from The Centre for Reviews and Dissemination (Centre for Reviews and Dissemination, 2008) studies should not be excluded on the basis of quality, I therefore included all papers in the review. Thus, all papers contributed to the synthesis and development of themes. To enable judgements to be made about the robustness of the evidence from each paper, the strengths and weakness of the methods used have been reported in Table 3.1, which shows the checklist for assessing the quality of qualitative papers included. Comments on the quality of each paper are also reported in the final column of Table 3. 2.

3.3.4 DATA ABSTRACTION AND ANALYSIS

Because it was not possible to undertake a meta-analysis or meta-synthesis of the data, a narrative description of the findings was prepared using thematic analysis with the methods described by Braun and Clarke (2008). First of all, codes pertinent to the research purpose were identified in each paper. Furthermore, different codes were combined, whenever similar across papers or because considering the same aspect, to define themes that explained larger sections of the data and were given specific names. Finally, direct quotes from each of the papers were chosen to illustrate examples of themes. The thematic analysis was confirmed by the researcher and two of the supervisors (CH, HS).

TABLE 3. 1 CHECKLIST FOR ASSESSING THE QUALITY OF QUALITATIVE PAPERS

	Duncan *et al.* (2010)	Duncan *et al.* (2007)	Duncan *et al.* (2008)	Hamilton *et al.* (2009)	Hamilton (2012)	Hoskins *et al.* (2014)	MacLeod *et al.* (2014)	Macrae *et al.* (2013)	Mand *et al.* (2013)	Patenaude *et al.* (2013)	Werner-Lin *et al.* (2012)
1. Question/objective clearly described?	Partial	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
2. Design evident and appropriate to answer study question?	Partial	Partial	Partial	Yes	Yes	Partial	Partial	Yes	No	Partial	Yes
3. Context for the study is clear?	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Partial	Partial	Yes
4. Connection to a theoretical framework/ wider body of knowledge?	Partial	Partial	Partial	Yes	Yes	Yes	Yes	Yes	No	Partial	Yes
5. Sampling strategy described, relevant and justified?	Partial	Yes	Yes	Yes	Partial	Yes	Yes	Yes	Partial	Yes	Yes
6. Data collection methods clearly described and systematic?	Partial	Partial	Partial	Yes	Partial	Yes	Yes	Yes	Partial	Yes	Yes

7. Data analysis clearly described, complete and systematic?	Partial	Yes	Partial	Yes	No	Yes	Yes	Yes	Partial	Partial	Yes
8. Use of verification procedure(s) to establish credibility of the study?	No	Yes	Yes	Partial	No	Yes	Yes	Yes	Partial	Partial	Partial
9. Conclusions supported by the results?	Yes	Yes	Partial	Yes	Partial	Yes	Yes	Partial	Yes	Yes	Partial
10. Reflexivity of the account?	No	Partial	Partial	Partial	Yes	Partial	Partial	Partial	Partial	Partial	No

3.4 RESULTS

3.4.1 CHARACTERISTICS OF INCLUDED STUDIES

In total, the systematic review included 11 qualitative studies. The key characteristics of the individual studies are presented in Table 3.2. Methods adopted by the authors were: interpretative phenomenological analysis (MacLeod et al., 2014), thematic analysis (Duncan et al., 2007; Duncan et al., 2010; Macrae et al., 2013; Mand et al., 2013; Patenaude et al., 2013), a combination of interpretative content analysis and thematic analysis (Duncan et al., 2008), or grounded theory (Duncan et al., 2008; Hamilton et al., 2009; Hamilton, 2012; Werner-Lin et al., 2012; Hoskins et al., 2014). Patenaude et al. (2013) also included a quantitative analysis of their data.

All the included studies were published between 2007-2014 and were focused on few specific heritable disorders, namely autosomal dominant cerebellar ataxia (Mand et al., 2013), familial adenomatous polyposis (FAP) (Duncan et al., 2008; Duncan et al., 2010), familial cardiomyopathy (MacLeod et al., 2014), hereditary breast and ovarian cancer (HBOC) (Hamilton et al., 2009; Hamilton, 2012; Werner-Lin et al., 2012; Macrae et al., 2013; Mand et al., 2013; Patenaude et al., 2013; Hoskins et al., 2014; MacLeod et al., 2014), hereditary diffuse gastric cancer (Mand et al., 2013), Huntington disease (HD) (Duncan et al., 2007; R E Duncan et al., 2008; Mand et al., 2013; MacLeod et al., 2014) and Lynch syndrome (Mand et al., 2013). Samples included participants within an age range of 12-39 years, thus including, but not limited to, the age range of 14-30 years identified as the focus of this study. Cross-cultural comparisons were hampered by the fact that all eligible studies had been conducted in the United Kingdom (MacLeod et al., 2014) or

countries that were originally colonies of the British Isles, namely Australia (Duncan et al., 2007; Duncan et al., 2008; Duncan et al., 2010; Mand et al., 2013), Canada (Hamilton et al., 2009; Hamilton, 2012; Macrae et al., 2013), and the United States (Hamilton et al., 2009; Hamilton, 2012; Werner-Lin et al., 2012; Patenaude et al., 2013).

TABLE 3. 2 MAIN CHARACTERISTICS OF INCLUDED STUDIES

STUDY	COUNTRY	NUMBER OF PARTICIPANTS	AGE	CONDITION	AIM	ANALYSIS	MAIN FINDINGS	QUALITY ISSUES
Duncan et al. (2007)	Australia	8	17-25 years	Huntington disease	To explore the experience of presymptomatic genetic testing for Huntington disease from young persons' perspectives and document the impact that testing has upon various aspects of young peoples' lives	Thematic analysis	Some of young people interviewed, uncertainty about their genetic status constituted a barrier in their lives and prevented them from moving forward. Testing in similar circumstances may therefore allow other young people to move forward with their lives.	Although research question is clear, study design is not clearly identified. The sampling strategy and the data analysis are clearly described and justified, while data collection process is not clearly described.
Duncan et al. (2008)	Australia	18	14-26 years	Huntington disease and familial adenomatous polyposis	To evaluate the potential effects associated with presymptomatic genetic tests in young people	Grounded theory	The results were analysed in two categories: harms and benefits. These categories have been separated into three sub-categories: a) experiences relating to a gene-positive test result; b) experiences relating to a gene-negative test result; c) experiences relating to the testing process in general.	The objective is clear, but study design is not clearly identified. The sampling strategy is clearly described and justified, while data analysis and data collection are not clearly described. Moreover, the

								conclusion are only partly supported by the findings.
Duncan et al. (2010)	Australia	10	12-25 years	Familial adenomatous polyposis	To evaluate some of the key ethical challenges associated with presymptomatic genetic testing for familial adenomatous polyposis in young people	Combination of interpretative content analysis and thematic analysis	Five themes emerged: 1) the significance of the test; 2) young people's lack of involvement in the decision to be tested; 3) young people's limited understanding; 4) provision of the blood test at the first visit; 5) group testing of family members. These themes highlighted key ethical challenges. From these themes, authors draw eight recommendations for future practice to provide developmentally appropriate care to young adults undergoing presymptomatic genetic testing.	Research question and study design are not clearly identified. Sampling strategy verification procedures to establish credibility of the study were not evident.
Hamilton et al. (2009)	United States of America and Canada	44	18-39 years	Hereditary breast and ovarian cancer	To describe the decisional process of young women with increased risk for hereditary breast and ovarian cancer	Grounded theory	Four life trajectories that influenced the decision in young women to have genetic testing and subsequent risk reduction decisions after receiving a positive mutation result: 1) long-standing awareness of	Overall a good quality paper. Sampling strategy verification procedures to establish credibility of the study were not well described.

							breast cancer in the family; 2) loss of one's mother to breast cancer at a young age; 3) expression of concern by a health care professional; 4) personal diagnosis of breast cancer. Understanding possible influences behind decision making for genetic testing and risk reduction in young women may assist health care providers in offering age appropriate guidance and support.	
Hamilton (2012)	United States of America and Canada	44	18-39 years	Hereditary breast and ovarian cancer	To explore how young women live with a BRCA mutation	Grounded theory	Among 13 unmarried women, issues of when to disclose information about their genetic risk with their partners were discussed. 24 women who had children reported "staying alive" for their children as a primary goal; on the other hand women without children reported an urgency to have children. Several of the 21 who had a breast cancer diagnosis said knowledge of their genetic risk influenced their decision to undergo prophylactic mastectomy.	Research question and study design are clearly identified. The sampling strategy, data collection methods and analytic methods are not well described.

Hoskins et al. (2014)	United States of America	32	21-25 years	Hereditary breast and ovarian cancer	To explore patient-centred perspective on the dilemma faced by 18 to 24 year olds as they considered BRCA1/2 genetic testing and risk management		Young adults expressed needs for greater clarity in recommendations for screening and prevention before age 25, and ongoing contact with providers to discuss risk management protocols.	Overall a good quality paper. Study design is not clearly identified.
Macrae et al. (2013)	Canada	8	22-37 years	Hereditary breast and ovarian cancer	To assess the experiences of those mutation-negative young women	Thematic analysis	To investigate the experience of BRCA mutation-negative young women, eight themes were analysed: 1) timing; 2) disclosure; 3) risk perceptions; 4) cancer worry; 5) cancer burden; 6) hope; 7) plans for the future; 8) explanatory models for mutation status. These young women were likely still affected by the degree of cancer history in their family, even with their understanding of the genetic contribution to disease.	Overall a good quality paper. The conclusion are only partly supported by the findings.
MacLeod et al. (2014)	United Kingdom	36	15-31 years	Huntington disease,	To evaluate the motivation of	Interpreta tive	Young adults saw the value of pre-test counselling not in	Although the research question is

				Hereditary breast and ovarian cancer, Hypertrophic Cardiomyopathy or Dilated Cardiomyopathy,	young adults to be tested when young, their experiences of the counselling process and the advice they would offer to health professionals and other young adults considering testing	Phenomenological Analysis	facilitating a decision, but rather as a source of information and support. Differences emerged between the disease groups in terms of parental attitudes to testing. Parents in familial cardiomyopathies families were a strong influence in favour of testing, in hereditary breast and ovarian cancer the decision was autonomous but congruent with the parent's view, and in Huntington disease the decision was autonomous and sometimes went against the opinions of relatives.	clear, study design is not clearly identified. The sampling strategy, data collection and the data analysis are clearly described and justified.
Mand et al. (2013)	Australia	9	17-21 years	Huntington disease, Autosomal Dominant Cerebellar Ataxia, Lynch Syndrome, Hereditary breast and ovarian cancer , Hereditary	To assess the experiences of young people who request a presymptomatic genetic testing	Thematic analysis	Three themes emerged: 1) life before the test; 2) the battle to be tested; 3) living with the knowledge. The results convey young adults, from families affected by genetic conditions, might possess task-specific competence relating to decision making about presymptomatic testing.	The design and connection to a theoretical framework is not completely described, and the verification procedures to establish credibility of the study not described.

				Diffuse Gastric Cancer				
Patenaude et al. (2013)	United States of America	40	18-24 years	Hereditary breast and ovarian cancer	To evaluate what daughters understand about their 50% chance of carrying BRCA mutation and about risk reduction or management options for mutation carriers. To assess the extent and nature of daughters' cancer-related distress and the effects of knowing mother's mutation status on daughters' future plans.	Thematic analysis	Daughters of mothers who tested positive for a mutation in BRCA genes showed scarce genetic knowledge. Also, the genetic information was raised by young women regarding their future plans, such as childbearing.	Design, the setting, and data analysis are partially described. However, the sampling strategy is clearly described and justified.
Werner-Lin et al. (2012)	United States of America	32	18-24 years	Hereditary breast and ovarian cancer	To build on and increase findings of experiences of BRCA 1/2 mutation carriers in their reproductive years	Grounded theory	Feeling vulnerable to a cancer diagnosis were described by participant. Also, they described a quandary regarding their care, a wide range of genetic	Overall a good quality paper. Sampling strategy verification procedures to establish credibility of the study were not

	by focusing on those challenges specific to 18-24 year-olds	and health literacy. Several young women contemplated risk-reducing mastectomy before age 25. Parents were a primary source of emotional and financial support for young adults.	clearly identified.

3.4.2 FINDINGS OF THE ANALYSIS

The systematic review found that issues emerging from the studies included family and partner relationships, plans for the future, emotional state and general approach to life.

Themes identified were:

- the period prior to testing
- the experience of genetic counselling
- involvement of parents in decision making
- impact of personal test result communication
- living with genetic risk.

2.4.2.1 THE PERIOD PRIOR TO TESTING

Many participants reported having grown up without awareness or with misinformation about the genetic disease running in their family or the inheritance mode (Duncan et al., 2007; Patenaude et al., 2013; Hoskins et al., 2014). They also lacked information about the appropriate age for testing (Duncan et al., 2007; Patenaude et al., 2013).

Two sets of authors reported that the first communication about the genetic risk was made by parents (Macrae et al., 2013; Patenaude et al., 2013). None of the participants was younger than 12 years of age when informed, about half experienced disclosure before they were 18 years old and half between 18 and 21 years old. Many participants stated that the disclosure was made during an occasional encounter and in a casual moment (i.e. while driving) or by telephone (Patenaude et al., 2013). Almost all

daughters were informed of their mother's test result in a private conversation with their mother, and it was rare for both parents to participate in the disclosure (Macrae et al., 2013; Patenaude et al., 2013). In some studies, participants expressed a preference toward being informed by both parents, although they knew that the information given by parents was limited and sought genetic counselling almost immediately after disclosure (Macrae et al., 2013). In other cases, once aware of the family genetic disorder, those who did not understand what it really meant sought information online or in professional journals (Patenaude et al., 2013), while those who were more conscious of their own risk (or potential risk) arranged the first counselling session to have their blood test (Hamilton et al., 2009).

However, interviewees described the disclosure of a positive parental test result as the most important information of their lives (Mand et al., 2013), reporting concerns about their mother's health (Mand et al., 2013; Patenaude et al., 2013) and, only secondarily, their own (Mand et al., 2013). In the quantitative sub-study by Patenaude et al. (2013), one third of the daughters of BRCA1/2 mutation carriers reported normal levels of general distress but high cancer-related distress, which was not significantly different from distress levels of women with known BRCA1/2 mutations.

Some participants reported that at the time they were told of their risk, the implications for themselves seemed distant, but now, as young adults, the fear of developing the adult-onset disease recurring in their family had increased (Patenaude et al., 2013). Conversely, others felt that early disclosure of the family disease gave them the time to digest the information (Macrae et al., 2013). However, the knowledge of being at risk of a disorder such as Huntington disease for some participants involved engagement in

risk behaviours such as drugs use, trouble with the police or difficulty at school (Duncan et al., 2007).

When approaching the decision about testing, in the study by MacLeod et al. (2014) most of the participants did not understand that having a presymptomatic test was a choice, but rather something they felt obliged to undergo in order to obtain information about themselves and to remove uncertainty. For example, a young woman said '*I knew, I had to'* (MacLeod et al., 2014, p. 397). By contrast, those who perceived there was a choice prepared themselves for the result; some prepared themselves for the worst possible outcome because then they would not be surprised by bad news (Duncan et al., 2007; Mand et al., 2013; MacLeod et al., 2014), while other participants were scared that receiving the test result would be devastating (Patenaude et al., 2013). Some study participants expected to test positive because of identification with a gene-positive family member (Macrae et al., 2013; Mand et al., 2013; MacLeod et al., 2014).

Choosing to undergo genetic testing constituted a major life event (Duncan et al., 2010), so important that participants reported it had a significant impact on their outlook and sense of self (Duncan et al., 2008; Mand et al., 2013). For example, a young woman said '*that was the day the clock stopped; that was the day the uncertainty began'* (Mand et al., 2013, p644). Nevertheless, while Mand et al. (2013) and MacLeod et al. (2014) report that no interviewees expressed regret regarding the decision to undertake testing, the timing for testing emerged as important because of potential interference with schooling: one young woman tested during her final year, said that looking back, she wished she had been tested at a different time of her life (Duncan et al., 2008). Another factor relevant to the

timing of testing was childbearing planning (Macrae et al., 2013; MacLeod et al., 2014), with participants split on the issue of undergoing presymptomatic testing prior to versus after having children (Macrae et al., 2013).

3.4.2.2 THE EXPERIENCE OF GENETIC COUNSELLING

Undergoing genetic counselling for the young adults studied by Duncan et al. (2008) was reported to have helped discussion of problems, for example a young woman who was mutation positive for Huntington disease said that the counsellor helped her with every type of problem in her life. Even when the genetic counselling was a source of information and support, it did not appear to facilitate the decision to be tested (MacLeod et al., 2014). Moreover, Duncan et al. (2010) showed that when counselled and tested at the same time as their siblings, participants felt this had limited the individual attention and support during the counselling process.

Some negative feelings were reported about counsellors (Mand et al., 2013), such as the perception of not being understood and the feeling that the counsellor was the person with the power over the testing decision. Some participants were disappointed to hear that the counsellor believed they were not ready to deal with the psychological consequences of the genetic test and that they needed to take time to reach an autonomous decision. The need to wait for genetic testing increased the feeling of disempowerment raised by uncertainty: a girl said '*I wanted the maybe to become yes or no; I was over maybe*' (Mand et al., 2013, p. 645). Others focused their attention on the procedure, instead of the meaning of testing,

with the fear of the needle overshadowing the purpose of the counselling (Duncan et al., 2008; Duncan et al., 2010).

A consequence of discussion during pre-test counselling was that most young adults had shared their test result with only close friends and family. This was because they felt that other people would not understand the complexities of the process from decision making to the result (MacLeod et al., 2014).

3.4.2.3 INVOLVEMENT OF PARENTS IN DECISION MAKING

Although theoretically an autonomous choice to undergo presymptomatic testing is a fundamental requirement of the process of genetic counselling, some parents were reported to exert pressure on their young adult children (Duncan et al., 2007; Duncan et al., 2008; Duncan et al., 2010; Hamilton et al., 2009; Werner-Lin et al., 2012; MacLeod et al., 2014). In addition, Hoskins et al. (2014) reported that a young woman underwent genetic testing because of her gynaecologist's suggestion. As a consequence of parental pressure, interviewees conveyed feelings of disempowerment and lack of control and declared that they underwent genetic testing because of pressure from family members or "for" a parent, which also raised the ethical problem of respecting young adults' developing autonomy (Duncan et al., 2007; Duncan et al., 2008; Duncan et al., 2010; Hamilton et al., 2009; Werner-Lin et al., 2012). A young man said "*I was 12 when I was told that I had to have the test, I didn't want to have it, but then I sort of had to*" (Duncan et al., 2010, p. 30); a female participant said that her parents did not ask her what she wanted to do, they just said "*you know, you have to go get a blood*

test" (Duncan et al., 2010, p. 30). Older participants were more likely than younger ones to decide autonomously to have genetic testing, so much so that some described it as '*a way to take control and not to be like their mothers'* (Hamilton, 2009, p. 152).

Even when the decision making was autonomous and pragmatic, the family experience was still important, especially when parents had developed cancer (Hamilton et al., 2009). Some of these young adults had lost a parent in their adolescence or earlier, so they grew up without a parent and with the knowledge that their parent's death was due to "the gene" and that they may carry the same risk. In this way, the study conducted by Hamilton et al. (2009) showed that participants both desired and feared genetic testing. Another key motivation was the perception that they were doing something to alter the course of a disease that had led to the death of affected relatives (MacLeod et al., 2014). A young woman said '*I just thought that you know if she (mum) would have had the opportunity to have the test, then things could have been a lot different*' (MacLeod et al., 2014, p. 397).

However, differences emerged in parental involvement in the decision making process, based on the specific disease (MacLeod et al., 2014). Parents in families with familial cardiomyopathy had a strong influence in favour of testing; in hereditary breast and ovarian cancer the decision was autonomous but usually congruent with the parents' point of view, while in Huntington disease the decision was autonomous and sometimes went against the parents' opinion (MacLeod et al., 2014).

3.4.2.4 IMPACT OF PERSONAL TEST RESULT COMMUNICATION

Once the test was undertaken, waiting for the test result was associated with anxiety (Duncan et al., 2008). At the time of the communication of the genetic test result, participants generally had to face the idea of disease (Macrae et al., 2013). Usually at their age young adults do not think about disease; their attention is focused on plans for their future such as university and/or job plans (Macrae et al., 2013). Nevertheless, none of the participants of the study by Mand et al. (2013) reported a catastrophic emotional response to their test result, but conflicting emotions of relief, happiness, guilt, fear and anger were generally reported (Macrae et al., 2013). In more detail, authors described the emotional impact of both gene-negative and gene-positive test results. Surprisingly, positive and negative emotional outcomes were not correlated with test results: in any case interviewees thought that the best thing was to find out the test result (Duncan et al., 2008). Accordingly, participants described themselves as happy just to know their genetic status or as willing to begin enjoying life and to make behavioural changes (Duncan et al., 2007; Mand et al., 2013). Specifically, a positive result led participants to feel able to move forward and to understand what was important (or not) in their lives (Duncan et al., 2008). Although some participants stated their positive result induced a change of lifestyle, others showed no reaction to testing positive; this lack of reaction sometimes created uneasiness because the counsellors failed to understand the underlying feelings, which are well explained by one woman: "*I kept on the same direction I was already going*" (Mand et al., 2013, p. 646). In others, a gene-positive result created some negative emotions such as depression and anxiety, either in general or related to potential gossip by

other people (Duncan et al., 2008; Mand et al., 2013), connected with employment (Duncan et al., 2008), related to the possibility of passing on the mutation to their future children (Mand et al., 2013) or because of a different test result in other family members (MacLeod et al., 2014). A young man said " *when I first found out I didn't want to be too happy around them because it's still not the best of situations because my mum's still poorly with it [...], I'm still upset about my mum*" (MacLeod et al., 2014, p. 399); while a young woman said that she had been only thinking of herself during the decision making process, but now, receiving a negative test result, she wondered "*what does she (sister) feel about me now because I haven't got it and she has*" (MacLeod et al., 2014, p. 399). In addition, some interviewees described their shock at finding out that they had not inherited the family mutation (Mand et al., 2013; MacLeod et al., 2014): they had prepared themselves for something and then it just did not happen (Duncan et al., 2007; MacLeod et al., 2014). This was particularly true for young adults receiving a Huntington's disease test result (MacLeod et al., 2014). One young man, in the study by Macrae et al. (2013), received a negative test result but clearly expressed the desire to have been mutation-positive. Also, negative test results generated unexpected negative emotions in some participants, such as guilt and feeling distanced from family members. Moreover, some interviewees expressed the desire to receive additional screening regardless of their results, because of their familial cancer experience and residual cancer worry (Macrae et al., 2013). Nevertheless, in other participants the negative test result was associated with feeling able to plan for the future (Duncan et al., 2008).

3.3.2.5 LIVING WITH A GENETIC RISK

Authors of seven papers analysed risk management in terms of behaviour and attitudes. However, six of those papers were focused on BRCA1/2 carriers or daughters of BRCA1/2 carriers (Hamilton et al., 2009; Hamilton, 2012; Werner-Lin et al., 2012; Macrae et al., 2013; Patenaude et al., 2013; Hoskins et al., 2014; MacLeod et al., 2014) and only one on familial cardiomyopathy and Huntington disease (MacLeod et al., 2014).

Even though interviewees stated that having time before surveillance began gave them the opportunity to think about surveillance protocols or prophylactic surgery (Macrae et al., 2013), younger participants were more likely to feel out of place in the health care system and frustrated at their inability to access screening. This was emphasised so much so that some described themselves as '*paralyzed'*: one young woman underwent bilateral mastectomy at age 22, believing it was the only way to manage her cancer risk (Hoskins et al., 2014). Others expressed frustration at receiving inconsistent, inaccurate, ambiguous or incomplete recommendations by genetic counsellors or doctors, during the initial phase of their mutation-positive experience (Hoskins et al., 2014). They complained that each doctor explained only their own discipline-specific perspective and knowledge base. Others wondered about when to share with a new partner their genetic risk or how early in a relationship to discuss having children (Hamilton, 2012; Werner-Lin et al., 2012; Patenaude et al., 2013; Hoskins et al., 2014; MacLeod et al., 2014) or plans for prophylactic surgery (Hamilton, 2012; Werner-Lin et al., 2012; Patenaude et al., 2013).

Some participants with children described the impact of knowing they may have passed on the mutation to their children in terms of feeling guilty,

worried and, in some cases, leading to a decision to limit the number of children. However none regretted the choice to have children (Hamilton, 2012); they declared that they first considered the possibility of avoiding having children, but then realized that there were many options and over time there will hopefully be more (Hamilton, 2012). However, participants with children thought also about '*staying alive*' related to their children (Hamilton, 2012, p. 28) or of not being the next in the family to die (Hamilton et al., 2009). As a consequence of the wish to stay alive, young women were making the choice to have prophylactic surgery sooner rather than later (Hamilton et al., 2009; Hamilton, 2012). On the other hand, those opting for surveillance did not feel confident in surveillance protocols and reported being anxious waiting for the next screening (Hamilton et al., 2009). A 35 year old woman said '*I admire women who can live with surveillance, but that was not for me*' and she felt herself as being a '*ticking time bomb*' (Hamilton et al., 2009, p. 153). Werner-Lin et al. (2012) also showed that some parents exerted pressure on their children to pursue risk reduction surgery, while other young adults erected a barrier, because of their young age, to addressing aspects of cancer risk, for example in terms of being too young for surveillance.

3.5 DISCUSSION

Although this systematic review focused on presymptomatic testing, one major issue emerging from the papers reviewed is when and how at-risk individuals are informed of their genetic risk. Although many participants grew up with no or scarce information concerning their potential genetic

risk, communication generally occurred due to the parents' initiative and in a casual manner, several years before testing or clinical actions could be undertaken (Duncan et al., 2007; Patenaude et al., 2013; Hoskins et al., 2014). This is in line with findings by Rew et al. (2009), that showed that the majority of children of BRCA mutation carriers learnt of their potential genetic risk of cancer many years before preventive interventions were recommended. Indeed, intra-familial communication is a highly complex process, especially when an inherited genetic condition is involved, thus it is understandable that parents face the dilemma of when, how and what to tell their children about it (Sobel and Cowan, 2000; Sobel and Cowan, 2003). On the other hand, appropriate communication of genetic risk information by parents to their children is highly desirable, since it has been shown to have long-term consequences in terms of informed reproductive decision making and better family cohesion (Metcalfe et al., 2008). To achieve this, health professionals may have a role in both supporting parents and young people, but their involvement in parents' decisions to communicate genetic risk to young family members was found to be limited in both our search and previous reports (Bradbury et al., 2007; Rew et al., 2009; Werner-Lin et al., 2015). Although this may be partly due to the parents' wish to undertake this task alone, it is reported that some parents desired health professionals to be available in a supporting role, but found that this support was limited (Gaff et al., 2006; Metcalfe et al., 2008). This evidence highlights the need for a comprehensive, longitudinal counselling process with appropriate timing and setting, which supports 'parent-to-offspring' risk communication first and young people's decision making about presymptomatic testing and risk management afterwards. Accordingly, participants perceived that their lack

of emotional experience at the time of testing had made it difficult for them to envisage the possible psychological impact of a test result (MacLeod et al., 2014). Furthermore, establishing a deeper and long-standing relationship with the counsellor may reduce the feelings of disempowerment reported by some study participants about the experience of genetic counselling. Such an approach would also help limit parents' pressure towards testing or risk-reducing surgery, which was a relevant issue in the studies reviewed (Duncan et al., 2007; Duncan et al., 2008; Duncan et al., 2010; Hamilton et al., 2009; Werner-Lin et al., 2012; MacLeod et al., 2014).

Concerning the impact of test results, overall, our findings do not support any substantial risk of adverse emotional outcome in mutation carriers, which is in agreement with previous findings (Broadstock et al., 2000). However, possible reactions to being tested *per se* should be explored before undertaking testing, instead of focusing only on the potential effects of specific test results.

Nevertheless, there is general concern that undergoing presymptomatic testing too early in life may increase the risk of unfavourable impact, and, therefore, the right age to undergo presymptomatic testing is still a matter of debate (Borry et al., 2006; Richards, 2008; Duncan et al., 2008). In most of the papers analysed, the age at which participants had undergone genetic testing was not specified (Hamilton et al., 2009; Duncan et al., 2010; Hamilton, 2012; Werner-Lin et al., 2012; Macrae et al., 2013; Patenaude et al., 2013). However, Duncan et al. (2008), who included in their analysis 10 individuals who were aged 10-17 years at the time of their genetic test for FAP, concluded that harms observed in younger persons were no different in nature from those described in

adults. According to UK guidelines, people aged 16 or 17 are presumed to be capable of consenting to their own medical treatment, and, in specific cases, children under 16 years who have sufficient understanding and intelligence to enable them to fully understand what is involved in a proposed intervention will also have the capacity to consent to that intervention (Department of Health, 2009). Conversely, according to international guidelines (Borry et al., 2009), presymptomatic testing for adult-onset disorders are recommended to be made available to those aged 18 years and older, unless there it is in a child's best interest either in terms of immediate relevance for their health or of psychological or social benefits. Nevertheless, Richards argued that young persons who are considered as adults on the age-based criterion of 18 years are not all necessarily truly autonomous (Richards, 2008). She pinpointed that the most important aspect of the decision making process is the recognition that the knowledge obtained from the test result is irreversible. There is no specific age when a person is able to give autonomous consent, but it is important to consider psychological maturity (Richards, 2008) that is cumulative with age, life experience and cognitive development (Steinberg and Cauffman, 1996). Therefore, future studies should aim at defining the optimal moment when to undergo presymptomatic genetic tests, not only on the basis of age, but also considering psychosocial maturity (Richards, 2006). In any case, genetic health care professionals, in the context of presymptomatic counselling, should support young adults to become aware of their own individual needs and capacities and of the fact that, sometimes, waiting to be tested may be helpful to better understand potential harms and benefits of testing.

In addition, although it is reasonable to hypothesize that undergoing testing at the right time reduces the risk of negative effects, it is important to consider the influence of the specific disease considered: the perception and experience of harms and benefits from the test result. These may differ for a potentially treatable condition (such as BRCA, FAP, etc.) when compared with conditions for which there are no preventive treatment or cure (such as Huntington disease).

A potential limitation of this systematic review is that all the papers analysed are based on studies conducted in only four countries with similar British historical and cultural legacies, thus the findings may not generalize to other countries with different sociocultural backgrounds, supporting the need for further studies in other contexts. On the other hand, the papers analysed spanned across several diseases, while considering similar age ranges, thus providing a comprehensive overview of how young adults deal with genetic testing overall and according to the specific disease.

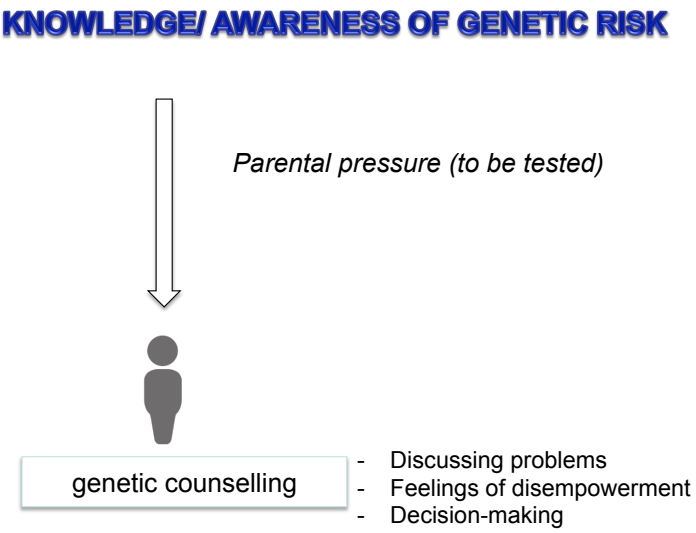
3.6 DEVELOPMENT OF THE MODEL

Figure 3.2 shows the model developed after the Phase 1 of this doctoral project. From the findings of the systematic review it came to light that young adults received the information on their potential genetic risk from parents. Parents, on the other hand, appeared to have exerted pressure on their children during the decision making process about testing.

It appeared that young adults underwent genetic counselling just to have the presymptomatic genetic test: there was no evidence of them

declining a test after being seen in the genetics clinic. This may indicate that they were not aware that one of the roles of the genetic counsellor is to support clients to make an appropriate decision regarding whether to be tested or not (Evans, 2006). Although the experience of genetic counselling was reported as an opportunity for discussing problems by young adults, it was also associated with feelings of disempowerment.

FIGURE 3. 2 DEVELOPMENT OF THE MODEL AFTER THE PHASE 1



3.7 IN SUMMARY

This systematic review indicates that many participants grew up with little or no information concerning their genetic risk. The experience of genetic counselling was either reported as an opportunity for discussing problems or associated with feelings of disempowerment. Emotional outcomes of disclosure did not directly correlate with test results: some mutation carriers were relieved to know their status, however, the

knowledge they may have passed on the mutation to their children was a common concern. Parents appeared to have exerted pressure on their children during the decision making process about testing and risk reduction surgery.

Since one of the central tasks of adolescence is achievement of autonomy and separation from parents, one might suppose that the potential inheritance of a serious condition may hamper this process, therefore prolonging the emotional dependence between children and parents (Erikson, 1968; Collins and Steinberg, 2007). Nevertheless, parent-adolescent relationships may differ according to cultures (Feldman and Rosenthal, 2007). The studies reviewed were performed in countries with similar backgrounds of British cultural influences, therefore information on different socio-cultural contexts are lacking. For instance, in Italian families, women are more likely to have fewer children and give birth while older in comparison to Australian, Canadian and American women (OECD Health Statistics, *2014*), which probably influences family dynamics and parents-children relationships, together with other differences in beliefs and culture. Thus, a study performed in Italy was warranted to examine how young adults or their parents interpret genetic risk information, what their attitudes are, and, eventually, what might best be done to prepare parents to answer their young children's questions and support young adults to make decisions on genetic risk assessment and management.

Please see Appendix 18 for a copy of the published paper of this systematic review (Godino et al., 2016).

CHAPTER FOUR

PHASE 2: QUALITATIVE STUDY

4.1 INTRODUCTION

In this chapter, I will present and discuss the key findings from the qualitative phase of the study. Findings from Phase 1 and Phase 2 will be then compared. The methods used in this phase have been detailed in Chapter Two, Section 2.4.

4.2 AIM AND OBJECTIVES

The aim of this phase was to explore the psychosocial implications of presymptomatic testing for hereditary cancer in young consultands (aged 18-30 years) referred for cancer genetic counselling.

The specific objectives were:

- to explore how young individuals interpret presymptomatic cancer testing
- to explore the basis for the young individuals' decision to undergo testing or not

- to explore the influence that parents have in the choice, with reference to the family dynamics and lifestage theory.

4.3 RESULTS

4.3.1 SAMPLE CHARACTERISTICS

Between November 2014 to December 2015, 89 members of families with a hereditary cancer predisposition but with no personal history of cancer were counselled in the Bologna genetics service. Clients had a mean age of 46±11 (range 8-83) and were counselled and tested for mutations associated with hereditary breast and ovarian cancer (n=62, 69.7%), MEN2 (n=9, 10.1%), hyperparathyroidism-jaw tumour syndrome (n=6, 6.8%), HNPCC (n=4, 4.5%), VHL (n=3, 3.4%), FAP (n=2, 2.2%), PTEN hamartoma syndrome (n=2, 2.2%) and PJS (n=1, 1.1%). Members of this cohort without personal history of cancer aged 18-30 at the time of the first counselling session numbered 17 (18.7%).

Seventeen invitations were sent to potential participants and 14 participants (82.4%) accepted and were interviewed (Figure 3.3). One woman aged 19 years decided to not attend the interviews because she wanted to undergo the genetic test as soon as possible without any other "waste of time". Additionally, a young man aged 27 years declined the counselling session at the clinic because of work problems and subsequently he withdrew from the research. After the first interview, one man aged 28 failed to show up to the appointment for the second research interview and when I re-contacted him, he asked to be withdrawn from the research

because his work did not enable him to take part. In total 42 interviews were conducted with 14 participants. Interviews lasted between 10 and 45 minutes.

Nine hundred and three codes were derived from the interview data and these were organised into 31 categories. A decision was made to stop enrolment after interview number 28. Very few new codes were being generated after the 28th interview and no new categories had emerged since the 37th interview. I felt therefore that saturation had been reached.

4.3.2 DEMOGRAPHIC CHARACTERISTICS OF THE PARTICIPANTS AND THEIR PARENTS

The study sample consisted of 14 young adults: 13 were Italian and one was Polish (living in Italy). The characteristics of the participants are described in Table 4.1. Respondent ages ranged from 18 to 30, with a mean age of 25.26 years. Seven participants were employed full-time, five were students and two were neither employed nor students. One participant had middle school education, six participants had a high school diploma, four had a university degree, and one had a post-graduate degree. Of 12 who were currently in work outside the home, six were paid employees, five were still students and one was a business owner. Twelve participants were single, one was living with a partner and one was married: two participants had children.

The social background of each participant is included in Table 4.2. The characteristics of the participants' parents (based on information provided by the young adults) are presented in Table 4.3.

TABLE 4. 1 DESCRIPTION OF PARTICIPANTS CHARACTERISTICS

ID	GENDER	AGE AT INTERVIEW (YEARS)	COUNTRY OF BIRTH	MOTHER'S LANGUAGE	CONDITION TESTED FOR	AGE AT TEST (YEARS)	RESULT	EDUCATION	DAILY WORK	MARITAL STATUS	CHILDREN	ARE YOU PREGNANT?
Donato	Male	30	Italy	Italian	BRCA2	30	Negative	Post-graduate degree	Business owner	Married	Yes, 1 son	n.a.
Barbara	Female	29	Poland	Polish	BRCA1	29	Positive	High school diploma	Worker	Single	No	No
Morena	Female	25	Italy	Italian	MLH1	25	Negative	University degree	Unemployed	Single	No	No
Mario	Male	26	Italy	Italian	BRCA1	26	Negative	High school diploma	Worker	Single	No	n.a.
Angelica	Female	24	Italy	Italian	BRCA2	24	Negative	University degree	Student	Single	No	No
Paola	Female	25	Italy	Italian	BRCA1	25	Positive	University degree	Employee	Single	No	No
Eleonora	Female	30	Italy	Italian	BRCA2	30	Negative	High school diploma	Unemployed	Single	No	No
Luca	Male	24	Italy	Italian	BRCA1	24	Positive	High school diploma	Worker	Single	No	n.a.
Caterina	Female	29	Italy	Italian	BRCA1	29	Negative	High school diploma	Employee	Living together	Yes, 1 daughter	No
Emma	Female	27	Italy	Italian	BRCA2	27	Negative	University degree	Student	Single	No	No
Patrizia	Female	23	Italy	Italian	BRCA1	23	Negative	University degree	Student	Single	No	No
Dario	Male	20	Italy	Italian	BRCA1	20	Negative	High school diploma	Student	Single	No	n.a.
Matteo	Male	18	Italy	Italian	BRCA1	18	Positive	Middle school qualification	Student	Single	No	n.a.
Saverio	Male	24	Italy	Italian	BRCA2	24	Negative	High school diploma	Employee	Single	No	n.a.

TABLE 4. 2 DESCRIPTION OF THE SOCIAL BACKGROUND OF EACH PARTICIPANT

ID	GENERAL INFORMATION	CARRIER PARENT	COMMUNICATION OF FAMILIAL MUTATION
Donato	His mother was diagnosed with ovarian cancer when he was 26 years old. One maternal aunt had breast cancer some years ago. His mother was the first person in the family to have genetic testing and she discovered her result one year ago. He lives in various countries around the world because of his work. The interviews were very difficult to arrange due to challenges in communication and making time. In fact the last interview was conducted by email.	Mother	Mother
Barbara	Her mother was diagnosed with ovarian cancer when she was 26 years. On her mother's side, her grandmother and one aunt also had breast cancer. Her mother was the first person in the family to have genetic testing and she discovered her result two years ago. Both grandmother and aunt had genetic testing and both have BRCA mutations. She has lived in Italy since she was 20 years old. She gave the impression of being a very strong young woman, however she was accompanied by her mother at both interviews and counselling sessions.	Mother	Mother
Morena	Her mother was diagnosed with colon cancer when she was 8 years old, and with endometrial cancer when she was 19 years old. On her mother's side, her grandfather had colon cancer and his mother was diagnosed with gynaecological cancer. Her mother was the first person in the family to have genetic testing and she discovered her result six years ago. She was accompanied by her mother at counselling sessions. She also texted me to remind me about her interviews.	Mother	Mother
Mario	His mother was diagnosed with breast cancer when he was 13 years old. In the same period, a maternal aunt had breast cancer. Another maternal aunt had breast cancer when he was 20 years old. His maternal grandmother died because of ovarian cancer when he was 22 years old. His grandmother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago.	Mother	Mother

	He lives in a small city in the south of Italy. He texted me to remind me about his interviews, however he was accompanied by his mother and his maternal uncle at the counselling session. His result was collected by his maternal uncle.		
Angelica	Her mother was diagnosed with breast cancer when she was 22 years. Maternal grandmother died because of breast cancer. Her mother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago. She came alone to both interviews and counselling, however she forgot both her first counselling session and our second interview. She only remembered after receiving an appointment reminder.	Mother	Mother
Paola	Two paternal aunts were diagnosed with breast cancer and another paternal aunt had ovarian cancer. grandmother died because of ovarian cancer. Recently her father discovered his genetic status. She came alone to both interviews and counselling sessions.	Father	Father and aunts (father's side)
Eleonora	Her mother died because of breast cancer, as did two maternal aunts. Her grandmother was the first person in the family to have genetic testing and she discovered her genetic status one years ago. She texted me to remind me about her interviews, however she was accompanied by her father both at interviews and at the counselling sessions. Although he was with her during the counselling, she never mentioned this.	Mother (?)	Cousin
Luca	His mother was diagnosed with breast cancer last year. His maternal grandmother was diagnosed with ovarian cancer and breast cancer when he was 20 years. His grandmother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago. He was accompanied by a friend at the counselling sessions.	Mother	Mother
Caterina	Her mother was diagnosed with ovarian cancer when she was 27 years. On her mother's side, two aunts had breast cancer and grandmother had ovarian cancer. One aunt was the first person in the family to have genetic testing and mother discovered her genetic status two years ago. She came alone to both interviews and counselling sessions.	Mother	Mother
Emma	Her mother was diagnosed with breast cancer when she was 25 years. Maternal grandmother died because of breast cancer. Her mother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago. Some months before, her sister (Angelina here) was tested.	Mother	Mother

	She came alone to both interviews and counselling		
Patrizia	Her mother was diagnosed with breast cancer when she was 6 years old and with contralateral breast cancer when she was 20 years old. Her maternal aunt had breast cancer when she was 21. Her mother was the first person in the family who had genetic testing and his mother discovered her genetic status two years ago. She was accompanied by maternal aunt both at interviews and counselling sessions.	Mother	Mother
Dario	His mother was diagnosed with breast cancer when he was 2 years and with contralateral breast cancer when he was 17 years. Both his maternal aunt and grandmother had breast cancer. His mother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago. He was accompanied by his brother both at first interview and first counselling session. He came alone to the post-test counselling and his brother delegated him to collect the brother's genetic test result (in Italy this is not routine, but sometimes happens).	Mother	Mother
Matteo	His mother was diagnosed with breast cancer when he was 17 years. His mother was the first person in the family to have genetic testing and his mother discovered her genetic status one year ago. He was accompanied by a friend both at the interviews and at counselling sessions.	Mother	Mother
Saverio	Two maternal aunts were diagnosed respectively with breast cancer and ovarian cancer. Recently his mother discovered her genetic status. The interviews were very difficult to arrange in terms of communication and time.	Mother	Mother

TABLE 4. 3 DESCRIPTION OF THE CHARACTERISTICS OF THE PARTICIPANTS' PARENTS

ID	FATHER		MOTHER	
	EDUCATION	DAILY WORK	EDUCATION	DAILY WORK
Donato	High school diploma	Retired	University degree	Retired
Barbara	High school diploma	Worker	High school diploma	Worker
Morena	Middle school qualification	Worker	High school diploma	Employee
Mario	Middle school qualification	Employee	Middle school qualification	Homemaker
Angelica	University degree	Employee	Post-graduate degree	Freelance
Paola	Middle school qualification	Business owner	Middle school qualification	Worker
Eleonora	Middle school qualification	Worker	Middle school qualification	Worker
Luca	High school diploma	Freelance	High school diploma	Employee
Caterina	University degree	Employee	High school diploma	Business owner
Emma	University degree	Employee	Post-graduate degree	Freelance
Patrizia	High school diploma	Worker	High school diploma	Worker
Dario	High school diploma	Freelance	High school diploma	Worker
Matteo	Post-graduate degree	Freelance	University degree	Freelance
Saverio	Middle school qualification	Employee	High school diploma	Employee

4.3.3 FINDINGS

Issues emerging from young adults' interviews included family and partner relationships, plans for the future, emotional state and their general approach to life.

Four major themes were identified:

- Knowledge
- Genetic counselling process

- Decision making process
- Dealing with the results.

4.3.3.1 KNOWLEDGE

Young adults' knowledge changed after genetic counselling in terms of awareness of personal cancer risk and options, knowledge about test and obtaining or avoiding knowledge.

Many young adults reported having grown up without awareness or with misinformation about the hereditary cancer running in their family:

> *"It don't feel it's like a coincidence, certainly there is a common thread linking all these events."*
> Paola, age 25 (first interview, test positive BRCA1)

> *"I don't feel I'm competent enough about the issue and this thought is a trouble [...] I think even my mother is confused as well. She has no idea of the meaning, because she isn't a doctor."*
> Caterina, age 29 (first interview, test negative BRCA1)

However, one young woman declared it was her choice to grow up without deep or accurate information:

> "*The information I gathered so far are neither a lot nor accurate. I know (fault gene) considerably increases your chances of developing bowel cancer as well as in other parts of the body. My knowledge stops here [...] in some way I do it intentionally ...*"
> Morena, age 25 (first interview, test negative MLH1)

Following their first counselling appointment, some young adults affirmed that as a result of the counselling session their knowledge had improved:

> *"Although the pathogen gene contaminated the female organs, I thought my mom could have transmitted the pathogen gene to me and it could have contaminated every single organ. At the beginning I was very confused."*
> Mario, age 26 (second interview, test negative BRCA1)

The majority of participants did not talk about the inheritance mode or the predisposition in their family. However, out of three participants who tried to explain their knowledge, two explained it correctly and one incorrectly.

> "*As far as I know, I have a fifty-fifty chance of having it"*
> Morena, age 25 (first interview, test negative MLH1)

> *"It was only an 18% (chance) if I correctly remember"*
> Angelica, age 24 (third interview, test negative BRCA2)

Despite lack of awareness or misinformation, young adults reported that the family history had an important role in terms of awareness and affected their feelings.

> *"Having a family member diagnosed with cancer definitely makes you more aware of cancer"*
> Donato, age 30 (third interview, test negative BRCA2)

> *"I felt very sorry about it. Fortunately, everything was always resolved by chemo. I stayed close to her (mum). What else can you do in cases like that?"*
> Luca, age 24 (first interview, test positive BRCA1)

Another important issue for participants was the need for surveillance. Some had not yet started any additional clinical surveillance that would have been relevant for the familial condition.

> *"I want to prevent [...] I'll do anything to stay healthy [...] I want to live!"*
> Barbara, age 29 (first interview, test positive BRCA1)

Others declared that they had started clinical surveillance.

> *"It's something that we have been doing since the first cancer of my mother".*
> Caterina, age 29 (first interview, test negative BRCA1)

Nevertheless, many participants felt cancer was only a distant possibility because of their young age, in fact they said

> *"Being young, I never worried about having a tumour or not"*
> Morena, age 25 (first interview, test negative MLH1)

However, they expressed fear of the disease:

> *"It is really a taboo, when people talk about this issue everyone turns the other way because there is a lot of fear among young people."*
> Mario, age 26 (first interview, test negative BRCA1)

After genetic counselling, young adults became aware of the options for clinical screening, and the possibility of having more frequent screening without undergoing genetic testing.

> *"They explained to me how the procedure was and which options I could explore: it was not required to proceed to the standard routine of undergoing the exam, wait for the results and then later entering the screening; but you could choose to take up screening in case of having a mother with this genetic mutation and to remain controlled while undergo various visits which maybe were not even necessary because you could have not inherited the mutation."*
> Caterina, age 29 (second interview, test negative BRCA1)

Nevertheless, one young woman thought that cancer could occur even if the mutation was not found and therefore she should have screening because of the family history.

> *"My family history is that, despite the fact of having the syndrome or not."*
> Morena, age 25 (first interview, test negative MLH1)

Knowledge of genetic testing was varied, with some unaware of what testing entailed. For example, Luca (first interview, age 24, test positive BRCA1) said

> "*I have no idea (what genetic testing is). [...] At the end, I think it isn't such a bad thing*."

Before genetic counselling, genetic testing was described as '*just a blood test*' by four of the participants, while three of them thought it was something that showed if a mutation was inherited or not. One said:

> *"You undergo a test to verify if you inherited a genetic predisposition, in particular to breast cancer and ovarian "*
> Angelica, age 24 (second interview, test negative BRCA2)

Waiting for the genetic test result was another point emphasised by young adults. Some reported that was the only thing they wanted to ask the genetic counsellor, in fact a young woman said:

> *"The explanation of Dr. T. was fairly complete. At the end, I had only one question left and it was about the timing. Counselling is unfamiliar to me; therefore I had no idea about the timing in terms of a month or a year. This is the only question I asked. And then I had no doubts (about undergoing the genetic test), but only lack of knowledge"*
> Morena, age 25 (second interview, test negative MLH1)

Also after the genetic counselling, the genetic test was often perceived as 'a *need to wait for result*'.

> *"They told me it takes months: the problem to me is waiting for the result not the result itself. It really takes a lot of time!"*
> Paola, age 25 (second interview, test positive BRCA1)

One young woman, who experienced a pregnancy, had compared '*the need to wait for result*' with her experience of finding out the baby's gender.

> *"They told me it takes about a couple of months to get the result. [...] Once you decide to do something, you immediately want to know the result. It's like being pregnant: you want to know if it's male or female, but you have to wait for it a month, two or three but still you don't know it! I get mad! You do want to know it. Same thing. Since you decide to undergo (the genetic test), you want to know and the sooner you know, the better it is. In fact, when they said, "it will take a little less with you, about two months" I shouted out "two months, you serious?" They explained that normally it takes about five months."*
> Caterina, age 29 (second interview, test negative BRCA1)

Although at first young adults did not really know what genetic testing was, after genetic counselling they declared that they better understood what they were doing or better understood the importance of undergoing genetic testing.

> *"I truly understood (the meaning of all) only after dealing with counselling and questions they asked me."*
> Barbara, age 29 (third interview, test positive BRCA1)

> *"I certainly didn't give all the weight which was given by the doctors [...] in the future it may help you to prevent disease"*
> Paola, age 25 (second interview, test positive BRCA1)

At the same time, young adults regarded the genetic test as a medical test like any other.

> *"An exam like any other. [...] It was an ordinary blood sample."*
> Luca, age 24 (second interview, test positive BRCA1)

Once aware of the family genetic disorder, those who did not understand what it really meant sought information online.

> *"I used Internet to search for information."*
> Barbara, age 29 (first interview, test positive BRCA1)

Others, however, did not want to use the Internet as a source of research:

> *"What is written on the Internet must always be taken with the tongs [...] The Internet problem is always discerning the sources."*
> Morena, age 25 (first interview, test negative MLH1)

Nevertheless, young adults preferred not to speak about this with friends.

> *"Then I sincerely don't want to analyse my private life with my friends too much."*
> Mario, age 26 (first interview, test negative BRCA1)

Almost all young adults were informed of their family genetic status by their mother.

> *"My mum had already told me."*
> Saverio, age 24 (first interview, test negative BRCA1)

When the mother was deceased, the person who had been genetically tested in the family often informed the young adult. For example, Eleonora said:

> *"My cousin informed me of this genetic thing which occurs in our family."*
> Eleonora, age 30 (first interview, test negative BRCA2)

Other young adults experienced different situations: rejecting the information shared by the mother, while others complained they did not receive any information from their parents.

> *"However, my mum had never analysed the subject with me ."*
> Morena, age 25 (first interview, test negative MLH1)

Those who were more conscious of their own risk (or potential risk) arranged the first counselling session to have more information, because they wanted information from a reliable source.

> *"I'd like to be told by a doctor and not by the Internet nor by my mum, who filters a bit, what this syndrome is in detail."*

Morena, age 25 (first interview, test negative MLH1)

4.3.3.2 GENETIC COUNSELLING PROCESS

The experience of the genetic counselling process was explored and young adults explained their motivations to have it, their expectations and experience of it.

Undergoing genetic counselling for some young adults was motivated by curiosity.

> *"Apparently the marker, the antigen, was isolated … whatever it is. I'd like to know a little more about it, for example where it is, etc. Just scientific curiosity."*
> Donato, age 30 (first interview, test negative BRCA2)

While for other it was reported as a source of information.

> *"I wanted to see what this result means for him and how it reflects on me. [...] So far I understand my father would have passed me a 50% chance of getting cancer. So I'd like to better understand this result because I read my father's letter, but I didn't get a word. [...] The only thing that maybe I'd like to understand is if it's really something you could pass on. "*
> Paola, age 25 (first interview, test positive BRCA1)

Still others underwent the genetic counselling to obtain certainty.

> *"I want to be sure if this thing may happen to me"*
> Eleonora, age 30 (first interview, test negative BRCA2)

Others focused their attention on undergoing genetic counselling to understand behaviour and attitudes in terms of prevention of cancer.

> *"I approached the counselling for prevention."*
> Angelica, age 24 (first interview, test negative BRCA2)

The decision to undergo genetic counselling was sometimes not specifically discussed with parents, but the young adult knew that their

relatives had consulted medical professionals and wished to follow a similar pathway.

> *"Basically, this is a course my cousin did and I want to do it as well."*
> Eleonora, age 30 (first interview, test negative BRCA2)

Following the example of relatives was described by young adults, even if they did not approve of the actions of all relatives.

> *"Knowledge helps. [...] my uncle, the brother of my mum, who has two children [...] he said that he will never undergo (the test). I think that information I gather from a doctor is more detailed and precise ... even the position to say 'I don't do it', I think it's a reaction to fear [...]. He said no 'a priori' because he ignores the data as they really are, he says it for selfishness. He has a narrow mind he prefers to say: 'I don't want to know anything' instead of thinking 'I'm aware about it but I don't want to'."*
> Morena, age 25 (second interview, test negative MLH1)

Nevertheless, four participants underwent genetic counselling purely for themselves, for example Mario added that he decided to undergo genetic counselling for his own reasons.

> *"For a more serene future"*
> Mario, age 26 (first interview, test negative BRCA1)

One out of the two young adults with children underwent genetic counselling because of anxiety about her daughter, while some participants without children underwent genetic counselling to understand the risk to their future children.

> *"You know, I have a daughter, so it means more anxiety for her."*
> Caterina, age 29 (first interview, test negative BRCA1)

> *"I am a schematic person. I like to do the right things at the right moment. To be certain of something that might have*

> *makes me more serene, even if tomorrow I could have children."*
> Mario, age 26 (first interview, test negative BRCA1)

The majority of young adults interviewed had no expectations about the counselling, mostly because they did not know what the counselling was.

> *"I didn't consider the problem before. I have no idea about what you have to do. I have a degree in literature and I study music, I have a complete lack of knowledge."*
> Angelica, age 24 (first interview, test negative BRCA2)

Even when young adults were not aware of it, they still expected a blood test, something that genetic counsellors suggested, something they had to do, and that it was something uncomfortable.

> *"Counselling was the prelude of the genetic test [...] I didn't think I could have said 'no' at the end as well as any other person. [...] I thought it was a required step. [...] I expected a psychologist together with Dr. T. I expected to be allowed to do it (genetic testing) by the psychologist. After the genetic counseling, I expect to be told 'okay, in your case you should do it, we do the test' or 'in your case it isn't advisable to take the test, we don't'. I expect them to tell me that I can do it."*
> Morena, age 25 (second interview, test negative MLH1)

Some young adults perceived genetic counselling/genetic testing as a '*need to wait for result*', and they were therefore surprised to have the blood sample taken at the first consultation.

> *"I honestly didn't expect to be tested during the first counselling."*
> Barbara, age 29 (second interview, test positive BRCA1)

For the young adults interviewed, undergoing genetic counselling was reported to have helped them, through the process of discussion with the counsellor. Some positive feelings were expressed about genetic counsellors,

such as the perception of being understood and the feeling that the counsellor was the person who explained the meaning of the testing.

> *"Doctors made me feel comfortable [...] everything was very friendly. They made me feel comfortable. [...] I felt good, I had good impressions [...] I felt welcomed and I think this is the most important thing both for a guy and for any other person [...] Being at one's ease is a great feeling that gives people space to open up and experience problems in a calmer way."*
> Mario, age 26 (second interview, test negative BRCA1)

Many young adults reported that they had not expected to have a choice. They had assumed that, in agreeing to undergo the genetic counselling process, they would have a genetic test and they were surprised after the genetic counselling when they realized that they had a choice.

> *"At the end, they asked me if I wanted to do this thing. I thought counselling ended with the genetic test, instead it wasn't! It was the idea I had for months!"*
> Eleonora, age 30 (second interview, test negative BRCA2)

Young adults who had decided to bring their mothers with them to their genetic counselling appointment spoke of their feelings about this.

> *"I think, one important thing, is the attendance of my mum. I told her to come with me, of course. She was glad to be there and I appreciated it. This was more touching because she is the closest to me, she is the person who has the syndrome and the one I've seen follow the process. I don't know how to explain it, but the fact that she was next to me ... I wasn't worried, I was just excited. That's the matter, the fact that mum was there [...] I felt calm. I mean, I wasn't scared or anything else. I felt good during the counseling because a person who already experienced the course was next to me and this person is my mum. Having her there made me experience the counseling as way more touching."*
> Morena, age 25 (second interview, test negative MLH1)

Although no one rejected the idea of having the blood test during the first counselling session, one young adult felt more aware of the implications of the test when she underwent genetic testing during the second counselling session.

> *"Yes, they proposed to me to take the genetic test right away at the first counselling. I'd have done it right away, however, the doctors advised me to wait and take time to think about it. Actually I was already convinced, but I still read in the previous weeks the things they gave me to be more aware. With hindsight I think the first time I'd have done it unconsciously. I'd have done and that's it. However, today, I'm more conscious about what I'm doing."*
> Paola, age 25 (second interview, test positive BRCA1)

Even if they had already made a clear choice to undergo the genetic test before the consultation, some expressed a desire to have the genetic counsellor give an opinion to guide them. For example, Morena (age 25, second interview, test negative MLH1) said:

> *"At the end of the explanation, they (the genetic clinic staff) asked me if I felt confident to do this thing. [...] I expected an opinion but it didn't come. I was floored by this. By the way I already knew my decision: I still would have liked to do it, whether the psychologist would had said 'yes' or 'not'. [...] yeah, I wanted an opinion."*

4.3.3.3 DECISION MAKING FOR TESTING OR NOT

Although theoretically making an autonomous choice to undergo presymptomatic testing is a fundamental requirement of the process of genetic counselling, some young family members were subject to pressure from their parents to be tested. As a consequence of parental pressure, some young adults underwent genetic testing for the sake of a parent/relative.

> *"Honestly, (I've approached the course) because my mother told me and she did it first... [...] I don't care, I'm doing it as a favour to her."*
> Luca, age 24 (first interview, test positive BRCA1)

However, differences emerged in the extent of parental involvement in the decision making process. In some, the decision to have a presymptomatic test was made autonomously but was congruent with the relatives' point of view.

> *"(Mom) said to us "it would be appropriate that you daughters, of course if you like, do this type of testing [...] When my mother and my aunts underwent the test I was pregnant and I was told by my mother to wait until the end of pregnancy. Currently my daughter is 16 months and now I think it's the right time."*
> Caterina, age 29 (first interview, test negative BRCA1)

> *"I called to have an appointment under pressure from my mother because I didn't want to. [...] As I already told you I'd have done it sooner or later. [...] At the end, I chose to do it, although I would have chosen to wait a bit more."*
> Angelica, age 24 (first interview, test negative BRCA2)

On the other hand, the decision was sometimes at odds with the parent's opinion.

> *"She (mother) has always been very uncertain whether to get me to do the project. She said: 'You have to think more deeply about it, the result doesn't change'."*
> Morena, age 25 (first interview, test negative MLH1)

In one case, a young woman found different influences within the family and wondered whether it was better to listen to her brother, who was a doctor, or her mother.

> *"My brother is a doctor and drives me to undergo the test because he says: 'it is the right thing'. But is it really the right*

> *thing? When I told to my mum what I wanted to do she said: 'Barbara, are you sure? You can't turn back. You will have to deal with the result. Are you sure you don't want to wait a few years?' "*
> Barbara, age 29 (first interview, test positive BRCA1)

However, the family experience in the decision making process was important, especially when one parent had developed cancer.

> *"Because my mom discovered this problem, so I want to know if I have the same problem or not."*
> Saverio, age 24 (first interview, test negative BRCA2)

The participants' decision making process occurred before the first counselling session: no participant reported having genetic counselling to help facilitate their decision about testing. However, it was not clear whose idea it was to undergo genetic testing. Some of them tried to align the decision to have counselling with their perception of the appropriate time to start clinical surveillance. For example, Morena (age 25, first interview, test negative MLH1) affirmed:

> *"At my age is useful do it [...] You should do the test close to the time when you should start the surveillance, shouldn't you?"*

The majority of young adults decided not to share the decision to undergo genetic testing with their friends, although sharing the decision to be tested with friends was reported when their friends had experienced cancer in their own family. For example, Morena (age 25, first interview, test negative MLH1) said

> *"I also spoke with friends. They all think that it is useful to do it for myself. [...] I had extensive discussion even with my friends because some of them have had serious health problems themselves. Talking with them came spontaneously: they began to speak about their health problems, so I told them about my decision on genetic testing. [...] It was not like*

> *talking because I was too anxious and I needed to do that, but because between close friends it's natural to talk to each other about what happens to us. "*

Others decided to share it only with close friends because they felt that other people would not understand the complexities of the situation. As Barbara (age 29, first interview, test positive BRCA1) described:

> *"None of my friends knows (what I'm doing) because I think these are very personal things and, knowing my friends, I'm afraid that some of them might think badly (of me) and then I would feel bad. [...] I prefer not to talk about it, it's my thing and that's that."*

The rationale for taking the test also varied, for example some wanted to avoid unnecessary surveillance, while others took advantage of having a genetic test while it was still available in public institutions without cost.

> *"My mother told me 'I think you should do it now ... They will offer it for free, I inquired how much it costs to do it privately and is an exorbitant amount'. I then told her 'if they offer it for free, let's do it!' It was also for this reason that I chose to do it, I took this advantage."*
> Luca, age 24 (second interview, test positive BRCA1)

Looking back on their experience of having genetic testing, three participants expressed a desire to have had something different from what they had experienced. While Barbara (age 29, third interview, test positive BRCA1) suggested a young adults support group to discuss experiences, share ideas, and provide emotional support, others proposed having more professional psychological support.

> *"Some people that want do this exam, but are a bit ... I mean unprepared for the possibility of having this problem ... maybe in some cases it might be useful to receive psychological help. For instance, a very young woman. So in some specific cases I think it is useful that the doctor proposes this option both before and after the test result."*

Matteo, age 18 (third interview, test positive BRCA1)

4.3.3.4 DEALING WITH THE RESULT

Some participants perceived genetic testing as a source of tension, mostly before they underwent genetic counselling. As Dario (age 20, third interview, test negative BRCA1) described

> *"At the beginning, it is normal to feel a little bit scared or worried because is something unknown ... but when everything is explained one calms down … for example, in my experience it wasn't ... it could be a source of concern just at the beginning …"*

Some young adults expected that the genetic test result would be negative. Others, who believed before testing that they would be mutation-positive, felt relieved when the test had a different outcome. As Barbara (age 29, first interview, test positive BRCA1) described

> *"If I didn't have the gene ... breathe."*

However, the information provided by a genetic test was perceived by young adults as useful in helping to plan their lives, for example Donato said

> *"I know that even if I tested positive, I would have several years left to prepare myself both physically and psychologically or to search for a preventive solution, this reassures me whatever the result will be so if the result is negative, better!"*
> Donato, age 30 (first interview, test negative BRCA2)

Conversely, others did not think that they would change behaviour based upon the possible result.

> *"I think, there is no big difference in my reaction between positive or negative news."*
> Morena, age 25 (second interview, test negative MLH1)

However, when they felt about how they would react, the majority affirmed that they did not know.

> *"How am I supposed to react facing the real result?"*
> Paola, age 25 (second interview, test positive BRCA1)

Once aware of their genetic test result, none of the young adults interviewed reported a catastrophic emotional response to their test result: emotions of relief, happiness and fear were generally reported. Accordingly, participants with negative genetic test results described themselves and their parents as happy to have this knowledge. Regardless of the genetic test result, some young adults felt they had matured as a result of their testing experience.

Moreover, once they had received their genetic test result, they recommended that their relatives, for example sisters or brothers, undergo genetic testing as well. Only one specifically recommended genetic counselling to their relatives.

> *"I will be more directive with my sister. I'll tell her 'I did it and so it's useful that you do it as well, for this and that reasons, so do it. These news may not be pleasant, especially if the result is positive ... But consider your health: you can enter a procedure that allows you to have surveillance.' She definitely needs to be driven ... it's because of her personality... I'll tell her to go. I'll suggest it because of my personal experience."*
> Morena, age 25 (third interview, test negative MLH1)

Although participants with mutation-positive results were more likely to think about their result, no change in behaviour was reported in either mutation-positive or mutation-negative young adults. However, a young woman who was mutation-positive started to pay more attention to her body and possible symptoms.

> *"... Now I pay more attention to my body, I've never been like that before ... but since I've discovered this, even minor pain*

> *during the cycle or pre-cycle ... I've become a little more anxious about this ... but nothing else."*
> Paola, age 25 (third interview, test positive BRCA1)

Young women who were mutation-positive, started their surveillance and one of them described herself as '*a butterfly*' (Barbara, third interview, age 29, test positive BRCA1) after her first screening. She described herself as nervous about her first ultrasound outcome. Fortunately, it was normal and she felt relieved, but she underlined that the relief would last 'until the next follow-up visit'.

4.4 DISCUSSION

In this section, I will discuss the findings in the context of a range of other literature. I will apply existing theories, where applicable, to help interpret the findings, present the theory developed as a result of the findings of Phase 2 and discuss how it emerged inductively from the data.

During the numerous stages of analysis of the data, different relationships between the categories were identified, leading to the development of the theory. With regard to the final phases of analysis, a central category was chosen that linked together the other major categories and logically explained what was evident from the data. I will now provide an overview of these phases to explain how the grounded theory was developed inductively from the data. A number of integrative diagrams were developed during these phases which helped visualise the young adults' experiences and link categories.

During the initial phases of theory development, it was evident from the data that young adults were describing an experience that began when their parents/relatives told them about the family predisposition or they became conscious of the family history. Indeed participants grew up with little or no information about their genetic risk and the communication occurred due to the parents' initiative within one year before testing in 73.3% (n=11) of participants. This is in contrast with findings emerging from the papers reviewed in the systematic review presented in Chapter One where many were informed as young adults several years before testing or clinical actions could be undertaken (Duncan et al., 2007; Patenaude et al., 2013; Hoskins et al., 2014).

This experience was still ongoing at the time of the interview. It became clear during axial coding that, while young adults were describing their experience with psychological and social complexities, there was another process occurring. Young adults were consciously, as well as unconsciously, developing strategies to cope with the experience they were facing. There were a number of key categories considered important to the interpretation of the young adults' experiences. These included 'knowledge', 'genetic counselling process', 'decision making process' and 'dealing with the results'. The decision making process was a central focus of this phase of my research. Yet while 'decision making process' was a central theme, there was, I believed, a more profound young adult experience occurring. In order to achieve a more general explanation, I went back to the data to establish further links among categories, using the paradigm model (Corbin and Strauss, 2014) 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 decision making process and their autonomous choice, and this was a central point. With this in mind I went back to the data to look for clues to help decode what was happening.

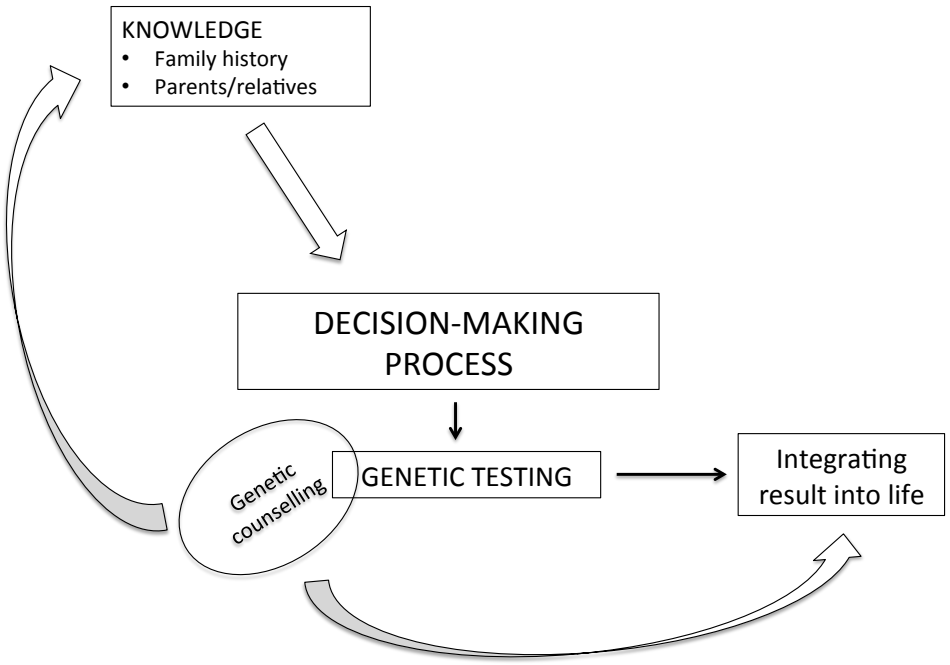
As shown in Figure 4.1, young adults arrived at the decision making process because of their previous knowledge (family history and/or parents/relatives). They had usually been told by one or both parents. Consistent with this finding, a meta-synthesis of the family communication between children and their parents about inherited genetic conditions conducted by Metcalfe et al. (2008) showed that parents were primarily responsible for discussing genetic information with their children. The best source of information was often viewed in the figure of the mother. Although there was a desire by parents to tell their children about their potential genetic risk before others told them (Metcalfe et al., 2008), parents also stressed delaying the disclosure or choosing the right time to talk (Metcalfe et al., 2011).

Once young adults decided to undergo genetic testing they discovered that genetic counselling is an 'anteroom' for the genetic test: a place where information is obtained and the decision making process starts, a place where people are helped by the counsellor to achieve a really informed decision about whether to have testing or not. This is in line with findings by MacLeod et al. (2014), who showed that most participants did not understand that having a presymptomatic test was a choice, but rather something they felt obliged to undergo in order to obtain information about

themselves and to remove uncertainty. Nevertheless, because the decision making process occurred before the first counselling session, none of the young adults interviewed expressed regret regarding the decision to undertake testing, as also reported by Mand et al. (2013), MacLeod et al. (2014) and Gong et al. (2016). However, in a recent paper Mand et al. (2015) analysed the psychosocial context of young people (nine participants of ten were less than 18 years of age) living in families affected by Huntington disease and showed that at the time of interview none had requested a presymptomatic genetic test. This may be due to the difference in condition or because presymptomatic testing for Huntington disease is generally not offered to minors. In my study, the young adults interviewed understood the importance of the genetic counselling both because they increased their knowledge and because they were helped to integrate the result into their lives.

In order to verify the core concept I used the flip-flop technique (Corbin and Strauss, 2014) to obtain a different perspective on the key findings. I asked myself how this experience was different between participants aged less than 24 years and those who were older. I chose this age because it is the median age of my cohort. To help answer this question, I went back to the data to search for an answer. I was particularly interested in re-reading the interviews from young adults who underwent genetic testing before 24 years of age, regarding the decision making process. No differences emerged from the two groups of participants in my study, whereas Hamilton (2012) reported that older young adults were more likely than younger ones to decide autonomously to have genetic testing.

FIGURE 4. 1 THE CENTRAL PHENOMENON



Comments made during the interview with Emma and Morena helped me to analyse the phenomenon further. Emma affirmed that she had thought a lot before making the choice to undergo genetic testing. She then went on to say:

> *"I have postponed this choice so many times."*
> Emma, age 27 (first interview, test negative BRCA2)

This issue was confirmed by Morena who had discussed her sister's decision, in particular how her sister decided to postpone the choice to undergo the genetic test. These and other comments clearly highlighted that the decision making process was something that happened before booking the genetic counselling appointment. Many participants underlined in their interviews they had not undergone genetic counselling to facilitate the decision to be

tested. They arrived for genetic counselling believing that the decision making process was already completed. For example, Angelica said:

> *"They (counsellors) have clearly offered me to think about it (genetic test), but I have already decided to do it."*
> Angelica, age 24 (test negative BRCA2)

Another issue that attracted my attention was the fact that participants tended to talk about themselves in the second person. Self-talk in the form of second person statements is defined by Zell et al. (2012) as 'fragmented self-talk' (Zell et al., 2012, p. 2), who affirmed that fragmented self-talk occurs in situations that require self-control. In particular, it occurs in response to negative events, when people feel autonomous, and when they are currently planning to execute a behaviour. Another potential reason for use of second person self-talk may be in situations in which people feel autonomous and therefore must exercise self-control, as opposed to situations in which their behaviour is externally limited (Ryan and Deci, 2006). When analysing the interviews I found that this splitting of the mind emerged in the presence of negative events (such as when they talked about their family history), autonomous decision making (such as when they highlighted their choice to undergo genetic testing) and action (such as when they affirmed their decision not to tell their friends).

In the final stage of theory development, I decided that I could visualise the genetic counselling and genetic testing as a mirror in which young adults saw themselves (Figure 4.2). The concept of the 'self' has long been argued by religious thinkers, philosophers, and scientist alike (Shaffer, 2005). With reference to the 'mirror' imagery I used in my model, the development of the self was discussed by Cooley (1902) over a century ago,

in the social psychological theory of the '*looking-glass self*' , using an archaic English term for the mirror (Cooley, 1902). Cooley (1902) used an image of the reflection in a mirror of a person looking at him or herself to support understanding the development of the social self. Three principal elements were revealed analysing the looking-glass self process: a) the imagination of our appearance to the other person; b) the imagination of his or her judgement of that appearance, which means imagining what those others must think of them; and c) the self-feeling, meaning that the person experiences an affective reaction to the imagined evaluation of the others (Cooley, 1902). Although the purpose of this social psychological theory is the development of the social self, it might not have applicability to theory developed in this doctoral study because Cooley focused on the development of the social self based on the other's point of view. As I will describe later, in my theory the young adults challenged themselves in front of the mirror, and felt more grown-up. For example a young woman said as a result of the genetic counselling process:

> *"Now I feel like an adult".*
> Barbara, age 29 (third interview, test positive BRCA)

They did not appear to focus their attention on the other person's point of view, but they reflected on themselves. However, it has to be acknowledged that personal self-image and concern about the opinions of others may have changed considerably in the intervening century.

What is thought to be the normal process of genetic counselling was revealed to be peculiar to young adults. The literature states that a client who attends for genetic counselling has personal knowledge that increases during counselling and, after becoming more aware of the limits and

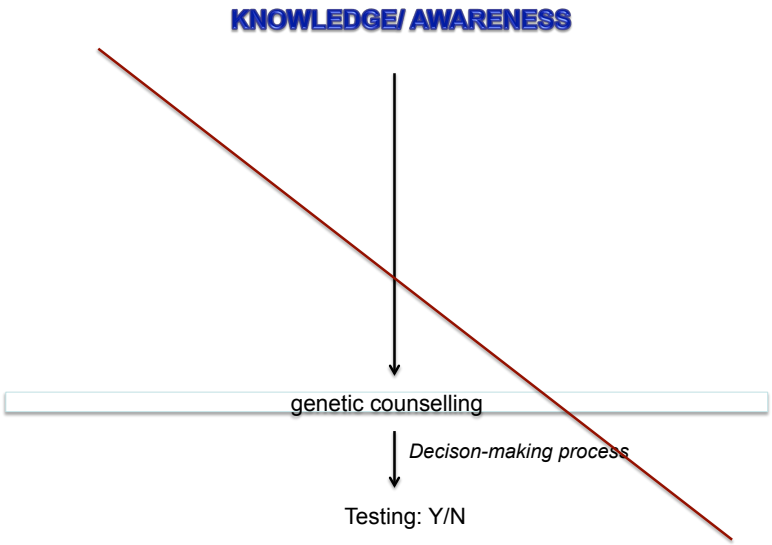
advantages of undergoing genetic testing, decides to be tested or not (Evans, 2006). All of the participants in the current study arrived for genetic counselling with the decision already made. The decision making process began with their background in which both family history and parents/relatives played an important role. Although the decisions were often autonomous and sometimes conflicted with parents' wishes, their backgrounds made young adults draw close to the mirror. Young adults arrived in front of the mirror because of the parents' pressure. The mirror seems to be the role of the genetic counselling. I draw it as a mirror because this determined a bi-directional relationship: young adults challenged themselves in front of the mirror, feeling grown-up in terms of autonomy and integrating the result into their lives. These findings do concur with those of Metcalfe et al. (2011) who suggested that young adults only started to realise and understand the implications of genetic risk only after they underwent genetic testing (Metcalfe et al., 2011). Other authors have also found that young adults felt they were helped to mature and became '*a better person*' (Gong et al., 2016, p. 7) as a result of going through the counselling and testing process. This indicates a need for further guidance on presymptomatic genetic testing in these populations: it is important for health professionals to understand how much the young people involved are really aware of the implications before and after they have been tested (Borry et al., 2009).

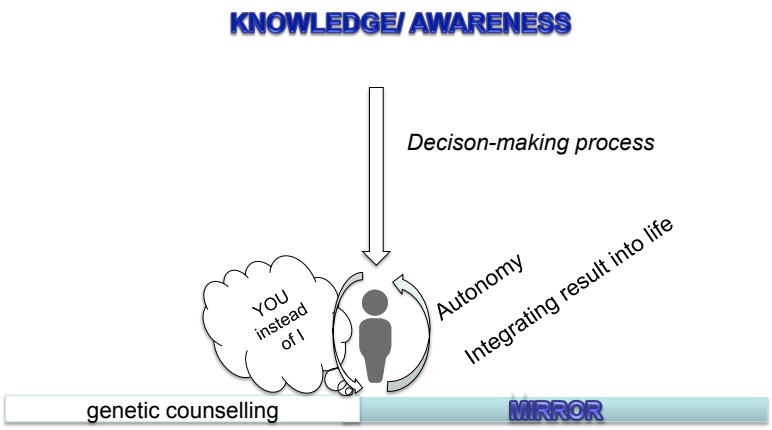
When considering young adults' understanding of the implications of the test before they accept it, it is appropriate to consider what adults in general understand about testing. The UK Huntington's Prediction

Consortium has collected anonymised data on 9407 presymptomatic genetic tests for the UK population at 50% risk of Huntington disease that were performed from 1993 to 2014 (Baig et al., 2016), The authors showed that the median age at testing was 37 years (range 29–47 years). Comparing the first five years of presymptomatic testing (1994–1998) with the last five years (2010–2014) a comparison of the age distributions of participants at 50% risk of Huntington disease between these two periods showed a statistically significant difference: in the earlier years (1994–1998) more older individuals decided to undergo the genetic test compared with the later years (2010-2014) where proportionally more younger individuals were undertaking the genetic test. Although the authors did not focus specifically on the understanding of the implications before testing by adult participants, the most common reasons for presymptomatic genetic testing have been reported by these and other authors. The main reasons reported were to reduce uncertainty (Meissen et al., 1991; Williams et al., 1999; Baig et al., 2016), to need or want to know (Meissen et al., 1991), to plan the future (Meissen et al., 1991; Williams et al., 1999; Baig et al., 2016), to provide information to relatives (Williams et al., 1999; Baig et al., 2016), to inform reproductive decision making (Baig et al., 2016), to clarify risk status of children (Meissen et al., 1991; Williams et al., 1999), as a factor in possible marriage (Meissen et al., 1991), and because of hope for future treatments (Baig et al., 2016). In summary, due to a lack of existing data, it was not possible for me to compare what young adults have understood about the implications of testing with that of older adults. I was therefore not able to conclude if what I have shown about young adults is generalizable for other adults undergoing a presymptomatic test.

Concluding, 18 to 30-year-olds are normally at a stage of life in which they are acquiring knowledge about themselves and the world around them (Arnett and Jensen, 2000; Arnett and Tanner, 2006). As expressed in Chapter One, they may or may not possess the maturity, the foundation of an established career or family trajectory, a realistic set of expectations about what genetic information will allow them to do, or even the health insurance to support risk management decision making (Steinberg and Cauffman, 1996; Richards, 2006). They may or may not fully understand the science behind genetic testing related cancer risk, penetrance, or prevention.

FIGURE 4. 2 FINDING YOURSELF IN FRONT OF THE MIRROR





In my research, at the start of the genetic counselling process participants had not often understood that their choices had serious implications. Instead, as Lindenmeyer et al. underlined (2011), participants did not choose to undergo genetic testing separate from the collective concerns and desires of their families. Parents may exert pressure on young adult children to complete genetic testing (Hoskins et al., 2012; Werner-Lin et al., 2008), however none of the participants reported the same behaviour as the parents in terms of risk management decisions (e.g. surgery rather than screening).

Concerning the impact of test results, overall, our findings do not support any substantial risk of adverse emotional outcome in mutation carriers, which is in agreement with previous findings (Broadstock et al., 2000). By contrast, Gong et al. (2016) showed that the knowledge of being mutation-positive for Huntington disease influenced the young adults' education and career, their relationships, and family planning. This may be partly due to the fact that there is no preventive treatment available at present for that condition, or that it is perceived to have much greater impact

on functioning throughout life.

4.5 COMPARISON OF FINDINGS OF PHASE 1 AND PHASE 2

In closing, based on the systematic review described in Chapter One, I compared the key findings from the systematic review and the qualitative phase. In Table 4.4 I present the main findings from both phases of my research project.

TABLE 4. 4 DIRECT COMPARISON OF THE KEY FINDINGS FROM THE SYSTEMATIC REVIEW AND PHASE 2

SYSTEMATIC REVIEW (PHASE 1)	QUALITATIVE PHASE (PHASE 2)
▪ Many young adults grew up without information or with misinformation concerning their potential genetic risk	▪ Participants grew-up with little or no information about their genetic risk ▪ After genetic counselling young adult's knowledge become more accurate, they became aware of the options for clinical screening, they perceived genetic testing as needed to wait for result and they better understood what they were doing or the importance of undergoing genetic testing
▪ Communication generally occurred due to the parent's initiative several years before testing or clinical actions could be undertaken	▪ Communication occurred due to the parents' initiative within one years before testing in 73.3% of participants
▪ The choice to undergo genetic	▪ Some relatives were reported to

testing constituted a major life event - Parents appeared to have exerted pressure on their young children during the choice to undergo presymptomatic testing	exert pressure on their young family members during the choice to undergo presymptomatic testing - The decision to have presymptomatic test for some of participants was autonomous, but often congruent with relatives' point of view. Sometimes, the decision was at odds with parent's opinion
- The experience of genetic counselling on one hand is reported as an opportunity for discussing problems, on the other hand it has been associated with feelings of disempowerment	- Curiosity, source of information, obtaining certainty, prevention of cancer and parents' influence were main motivations to undergo genetic counselling. - Most participants had no expectations from genetic counselling. - Personal experience of genetic counselling revealed positive feelings about genetic counselling in terms of being understood and clarifying the meaning of the test. They expressed the desire to have an opinion to guide them during the genetic counselling process. - Some participants felt they had matured as a result of their testing experience
- Positive and negative	- Before testing, although

emotional outcomes of personal test result communication are not directly correlated with test result: in both scenarios young adults thought that the best thing was to find out the result anyway, a common concern was related to the knowledge they may have passed on the mutation to their children • In some cases, parents appeared to have exerted pressure on their young children during the choice to pursue risk reduction surgery	participants perceived the information provided by a genetic test as useful in helping to plan their life, they did not think they would change behaviour based upon the possible result. However, when they thought about how they would react, the majority affirmed that they did not know. • None of the participants reported a catastrophic emotional response to their test result: emotions of relief, happiness and fear were generally reported • Neither mutation-positive nor mutation-negative participants reported changes in their behaviour. None considered the choice to pursue risk reduction surgery • Once they became aware about their genetic test result, participants recommended their relatives (e.g. brothers and sisters) undergo genetic testing as well.

As is shown in the table, there are some common points between the two phases, but other aspects were revealed to be different. For example, it was evident from Phase 2 of this research project that the communication between parents and their children occurred within one

year before testing in the majority of cases, while the systematic review (Phase 1) showed that it occurred several years before. Although both phases revealed that young adults felt the pressure during the choice to undergo presymptomatic testing by some relatives, the qualitative phase pointed out other factors in young adults' decision making processes. Very interesting findings emerged regarding their experience of genetic counselling. Young adult participants felt they had matured as a result of their testing experience. Once young adults became aware about their genetic test result, none considered the choice to pursue risk reduction surgery and the parents were not mentioned in this choice, while from Phase 1 it was clear that parents had a role in this decision too. These differences may relate to cultural mores, as none of the papers included in the systematic review reported data from the Italian population.

It is clear from the synthesis of both phases that parents may benefit from greater support from health professionals in helping them to communicate the genetic risk information to their children. Health professionals should be available to help them in choosing appropriate strategies to support their children's understanding. Health professionals can also play an important role regarding helping both parents and their children to manage the emotions evoked by this information at children's different developmental stages. The role of the health professionals in this context was also mentioned by some young adults interviewed (Phase 2), as they expressed the desire to have an opinion to guide them during the genetic counselling process. This need for guidance may conflict with the non-directive philosophical stance of genetic counsellors

(Evans, 2006) and this will be discussed in the final chapter (see Section 6.4).

4.6 IN SUMMARY

In this Chapter I have presented the findings of the Phase 2 of this doctoral study.

At the end of the chapter, I have related the findings from Phase 1 and Phase 2. In the next chapter, I will present the findings from Phase 3 of my research project and discuss how they relate to each other.

CHAPTER FIVE

PHASE 3: QUANTITATIVE STUDY

5.1 INTRODUCTION

In this chapter, I will present and describe the key findings from the quantitative phase. Findings from all three phases will be then compared. The methods used in this phase have been detailed in Chapter Two, Section 2.5.

5.2 AIM AND OBJECTIVES

The aim of this phase was to investigate the psychosocial implications of presymptomatic testing for hereditary cancer in young adults and their parents.

Specific objectives were to investigate:

- how young individuals interpret presymptomatic cancer testing
- the reasons for the young individual's decision to undergo testing
- the experiences of the counselling process of both young adults and parents

- the influence that parents have on the choice to be tested or not
- the influence that parents have on the young adult's decisions after the disclosure of the positive test result
- how the experiences of young adults being tested in Italy and their parents compared with those in other countries.

5.3 RESULTS

5.3.1 SAMPLE CHARACTERISTICS

Of 223 individuals who logged onto the young adult survey site, 199 (89.2%) consented to be involved in the study. Among these, 57 were excluded: 29 (14.6%) did not complete the minimum of 30% of the questions, 25 (12.6%) were index cases (the first person in their family who knew they had the gene mutation), and three (1.5%) were under 18 years of age when they underwent a genetic test. The motivations for excluding individuals logged onto young adult survey have been detailed in Chapter Two, Figure 2.9.

Of 57 individuals who logged onto the parent survey site, 50 (87.7%) consented to be involved in the study. Of these, seven (14.0%) did not complete at least 30% of the questions, two (4.0%) had no children who had undertaken a genetic test, one (2.0%) declared all their children were under 18 years of age, one (2.0%) had no genetic tendency to cancer in either his or his partner's side of the family. The motivations for excluding individuals logged onto parent survey have been detailed in Chapter Two, Figure 2.9.

Overall, the surveys completed by 152 young adults and 42 parents were included in the analysis. The results for the young adult questionnaire will be presented followed by the results for the parent questionnaire.

5.3.2 YOUNG ADULT QUESTIONNAIRE RESULTS

Demographic characteristics of the young adult participants

The demographic information provided by the study participants is shown in Table 5.1: mean age at questionnaire was 29.5 years (18-46); no significant differences were found between those who completed the English questionnaire (PEQ) and those who completed the Italian version (PIQ) with respect to age, education and daily work. Conversely, as shown in Table 5.1, differences between PEQ and PIQ were observed in gender, in paid employment, and marital status. Forty-eight percent of the respondents had children. Among those who had children, 100% (n=23) of the PEQ had both daughters and sons, while three of the PIQ had only daughters, and one had only a son.

All those who answered the questionnaire in Italian were living in Italy. The majority of those who answered the English version lived in either the UK (41.4%) or the US (30.9%), but there were also respondents living in 12 other countries as detailed in the following table.

TABLE 5. 1 SAMPLE CHARACTERISTICS: YOUNG ADULT PARTICIPANTS

	ALL (N=152)	PEQ (N=127)	PIQ (N=25)	*p-value*
Age at questionnaire (years)				
mean±SD	29.5±5.6	29.6±5.9	28.7±3.7	0.463 [a]
Gender				
Male	11 (7.2%)	2 (1.6%)	9 (36.0%)	0.000[b,*]
Female	140 (92.1%)	124 (97.6%)	16 (64.0%)	
I prefer not to say	1 (0.7%)	1 (0.8%)	0	
Country				
Australia	1 (0.7%)	1 (0.8%)	0	-
Austria	1 (0.7%)	1 (0.8%)	0	
Canada	3 (2.0%)	3 (2.4%)	0	
Cyprus	1 (0.7%)	1 (0.8%)	0	
Germany	1 (0.7%)	1 (0.8%)	0	
Ireland	4 (2.6%)	4 (3.1%)	0	
Italy	25 (16.4%)	0	25 (100%)	
New Zealand	1 (0.7%)	1 (0.8%)	0	
Netherlands	1 (0.7%)	1 (0.8%)	0	
Northern Ireland	2 (1.4%)	2 (1.6%)	0	
Poland	1 (0.7%)	1 (0.8%)	0	
South Africa	1 (0.7%)	1 (0.8%)	0	
United Kingdom	63 (41.4%)	63 (49.6%)	0	
United States of America	47 (30.9%)	47 (37.0%)	0	
Education				
Primary school	1 (0.7%)	1 (0.8%)	0	0.191[b]
Secondary school	15 (9.9%)	15 (11.8%)	0	
Post-secondary educ.	49 (32.2%)	38 (29.9%)	11 (44.0%)	
University degree	62 (40.8%)	50 (39.4%)	12 (48.0%)	
Postgraduate degree	25 (16.4%)	23 (18.1%)	2 (8.0%)	

Daily work				
Paid employment	112 (73.7%)	94 (74.0%)	18 (72.0%)	0.176[b]
Voluntary employment	2 (1.3%)	1 (0.8%)	1 (4.0%)	
Student	18 (11.8%)	13 (10.2%)	5 (20.0%)	
Homemaker	15 (9.9%)	15 (11.8%)	0	
Not working not student	5 (3.3%)	4 (3.1%)	1 (4.0%)	
If paid employment (n=112)				
Paid employee	73 (65.2%)	63 (67.0%)	10 (55.6%)	0.013[b]
Manager	11 (9.8%)	10 (10.6%)	1 (5.6%)	
Self-employed	2 (1.8%)	0	2 (11.1%)	
Business owner	4 (3.6%)	4 (4.3%)	0	
Professional	22 (19.6%)	17 (18.1%)	5 (27.8%)	
Marital status				
Single (never married)	48 (31.6&)	33 (26.0%)	15 (60.0%)	0.009[b]
Married	67 (44.1%)	60 (47.2%)	7 (28.0%)	
Divorced	7 (4.6%)	6 (4.7%)	1 (4.0%)	
Living with a partner	30 (19.7%)	28 (22.0%)	2 (8.0%)	
Children				
Yes	73 (48.0%)	69 (54.3%)	4 (16.0%)	0.000[c]
No	79 (52.0%)	58 (45.7%)	21 (84.0%)	

[*] "I prefer not to say" answer was excluded from the analysis
[a] Independent samples T-test

[b] Pearson chi-squared test

[c] Fisher's exact test

Participants underwent predictive genetic testing at a mean age of 24.7±3.7 years (range: 18-30): no significant differences were found between PEQ and PIQ (t(150)=-0.387; p=0.700). One hundred and eleven (73.0%) participants were tested for hereditary breast and ovarian cancer (HBOC), 26 (17.1%) for Lynch syndrome, 14 (9.2%) for familial adenomatous polyposis,

and one (0.7%) for Cowden syndrome. The fraction of PIQ tested for Lynch syndrome (LS) was significantly higher if compared to PEQ (44.0% *versus* 11.8%; χ^2=15.5, df=3, p=0.01). Male participants were tested more frequently for Lynch syndrome than women (8/11, 72.7% *versus* 18/140, 12.9%) while more women underwent hereditary breast and ovarian cancer testing compared to men (107/140, 76.4% *versus* 3/11, 27.3% χ^2=25.792, df=3, p=0.00). A difference in the type of condition tested was also shown across different age strata: the fraction of participants tested at 26 years and older for hereditary breast and ovarian cancer was significantly higher if compared to participants tested at younger age (58/69, 84.1% *versus* 53/83, 63.9%), whereas the fraction of participants tested at 26 years and above for Lynch syndrome was significantly higher if compared to participants tested at younger age (20/83, 24.1% *versus* 6/69, 8.7%, χ^2=1,132, df=3, p=0.17).

The majority of participants (n=142; 93.4%) were mutation-positive, and among those 6.3% (n=9) had been diagnosed with cancer since having their genetic test. The number of female PEQ was significantly higher if compared to men (134/140, 95.7% *versus* 7/11, 63.6%; p=0.003, Fisher's exact test). Among the PEQ the majority were mutation-positive (96.9%) while in the Italian sample there were 19 (76.0%) mutation positive and six (24.0%) mutation negative respondents (p=0.001, Fisher's exact test). No significant differences were found between PIQ and PEQ when the other parameters were considered.

Fifty-two (34.2%) participants reported that their mother was the first person in their family who knew she had the mutation, 11 (7.2%) their father, and three (6.6%) their sister. None reported their brother as the first

person in their family who knew of his genetic status. More distant relatives were mentioned by 77 (50.7%) participants. The number of participants informed by the mother among those tested at young age was significantly higher if compared to participants tested at 26 years and older (35/81, 43.2% *versus* 17/69, 24.6%; χ^2=9.032, df=3, p=0.29).

Table 5.2 shows the demographic characteristics of the parents of young adult participants. The majority of participants had both parents still living. Eight-two participants (53.9%) affirmed the relative who had cancer was the mother, while in 27 (17.8%) cases it was the father. Among parents who were diagnosed with cancer, participants mentioned cancer of the breast (n=24), bowel/colon/rectum (n=8), ovary (n=7), lung (n=4), prostate (n=3), and other organs (bladder, brain, oesophagus, kidney, melanoma, mesothelioma, prostate, throat). The mean age for the first episode of cancer where the respondent's father was affected was 54.8±10.9 years (range: 35-73), and for the second cancer 58.4±6.7 years (range: 52-67). However, the mean age of first diagnosis of cancer in respondents' mothers was lower, at 41.9±8.5 years (range: 22-64) for the first episode of cancer and 47.9±8.6 years (30-63) for the second one. For other episodes of cancer the mean age was 54.1±6.9 (46-68). No significant differences were found between PEQ and PIQ.

TABLE 5. 2 DEMOGRAPHIC CHARACTERISTICS OF THE PARENTS OF YOUNG ADULT PARTICIPANTS

	ALL (N=152)	**PEQ (N=127)**	**PIQ (N=25)**	***p-value***
ABOUT THEIR FATHER				
Is he still living?				
Yes	126 (82.9%)	107 (84.9%)	19 (76.0%)	0.382[c]
No	26 (17.1%)	20 (15.7%)	6 (24.0%)	
Education (n=151)				
No formal education	6 (3.9%)	6 (4.7%)	0	0.000[b]
Primary school	14 (9.2%)	11 (8.7%)	3 (12.5%)	
Secondary school	48 (31.6%)	46 (36.2%)	2 (8.3%)	
Post-secondary educ.	39 (25.7%)	24 (18.9%)	15 (62.5%)	
University degree	32 (21.1%)	29 (22.8%)	3 (12.5%)	
Postgraduate degree	12 (7.9%)	11 (8.7%)	1 (4.2%)	
Daily work				
Paid employment	146(96.1%)	121 (95.3%)	25 (100.0%)	0.541[b]
Voluntary employment	1 (0.7%)	1 (0.8%)	0	
Not working not student	5 (3.3%)	5 (3.9%)	0	
If paid employment (n=146)				
Paid employee	72 (47.4%)	63 (52.1%)	9 (36.0%)	0.033[b]
Manager	18 (11.8%)	18 (14.9%)	0	
Self-employed	22 (14.5%)	14 (11.6%)	8 (32.0%)	
Business owner	11 (7.2%)	9 (7.4%)	2 (8.0%)	
Member of armed forces	6 (3.9%)	4 (3.3%)	2 (8.0%)	
Professional	17 (11.2%)	13 (10.7%)	4 (16.0%)	
ABOUT THEIR MOTHER				
Is she still living?				
Yes	122 (80.3%)	104 (81.9%)	18 (72.0%)	0.276[c]
No	30 (19.7%)	23 (18.1%)	7 (28.0%)	
Education (n=151)				
No formal education	5 (3.3%)	5 (3.9%)	0	0.003[b]
Primary school	8 (5.3%)	7 (5.5%)	1 (4.2%)	

Secondary school	56 (36.8%)	51 (40.2%)	5 (20.8%)	
Post-secondary educ.	34 (22.4%)	21 (16.5%)	13 (54.2%)	
University degree	40 (26.3%)	35 (27.6%)	5 (20.8%)	
Postgraduate degree	8 (5.3%)	8 (6.3%)	0	
Daily work				
Paid employment	104 (68.4%)	88 (69.8%)	16 (64.0%)	0.890[b]
Voluntary employment	1 (0.7%)	1 (0.8%)	0	
Student	1 (0.7%)	1 (0.8%)	0	
Homemaker	27 (17.8%)	21 (16.7%)	6 (24.0%)	
Not working not student	18 (11.8%)	15 (11.9%)	3 (12.0%)	
If paid employment (n=104)				
Paid employee	70 (46.1%)	56 (63.6%)	14 (87.5%)	0.380[b]
Manager	8 (5.3%)	8 (9.1%)	0	
Self-employed	8 (5.3%)	7 (8.0%)	1 (6.2%)	
Business owner	6 (3.9%)	6 (6.8%)	0	
Professional	12 (7.9%)	11 (12.5%)	1 (6.2%)	

[b] Pearson chi-squared test
[c] Fisher's exact test

Finding out about their risk

The first part of the questionnaire explored the young adults' experiences before testing. The first six questions concerned the first time they were told that there might be a greater tendency to develop cancer in their family, compared to other families. Fifty-four (35.5%) were told by their mother, 19 (12.5%) by their father, 16 (10.5%) by both parents together, seven (4.6%) by their sister, 24 (15.8%) by other relatives such as aunts or cousins, 26 (17.1%) by a person outside the family such as a genetic counsellor or a physician. Three participants (2.0%) had suspected it

themselves, and then sought medical advice, because of the family history of cancer and three (2.0%) reported it was already something well known in their family. No significant differences were found between PEQ and PIQ (χ^2=12.523, df=6, p=0.051). In the Table 5.3 I present some quotes by participants who had written more about their experience.

TABLE 5. 3 QUOTES BY PARTICIPANTS WHO HAD WRITTEN MORE ABOUT THEIR EXPERIENCE ON THE QUESTION *"WHO TOLD YOU ABOUT THE POSSIBILITY THAT MEMBERS OF YOUR FAMILY MIGHT BE MORE LIKELY THAN OTHERS TO DEVELOP CANCER?"*

WHO TOLD YOU ABOUT THE POSSIBILITY THAT MEMBERS OF YOUR FAMILY MIGHT BE MORE LIKELY THAN OTHERS TO DEVELOP CANCER?	**QUOTE**
Your mother	*Mum had mentioned it but I didn't act on it until after she had died.* (R. 204)
Both your parents together	*I don't remember who told me - probably my mother and father together.* (R. 60)
Other relatives	*My auntie sent a letter* (R. 63)
A person outside the family	*I was told when I met with my doctor based on family history there is a possibility, that's when I was sent for testing and later found out my paternal aunt had tested positive for BRCA2* (R 133)
I found out another way	*It was just always know in our family* (R.92)

Participants declared they received this information the first time between 5-30 years of age (20.0±5.6); no significant differences were found between PEQ and PIQ (21.1±5.5 *versus* 20.5±6.2; t(145)=0.433, p=0.657). Participants tested for Lynch syndrome were more likely to have been informed at a younger age than those tested for hereditary breast and

ovarian cancer (18.0±5.7 *versus* 22.0±5.3; F=6.134, p=0.001). A significant difference was also observed between who told them about the possibility that members of their family might be more likely than others to develop cancer: participants who were told about the cancer risk in their family by both parents together were younger than those who were told by a person outside their family (15.9±7.2 *versus* 22.9±5.3; F=3.627, p=0.002).

One-hundred and two participants (68.5%) reported they received the information at an unplanned time (75 in a face to face conversation and 27 in a telephone or social media call/message), while 43 (28.9%) received the information in a pre-planned conversation (38 in a face to face meeting and five in a telephone call). Three participants (2.0%) reported they had always known there was a risk in their family and one (0.7%) reported she had asked her father's doctor '*about the possibility of there being a hereditary link on the cancers*' (R 54). Two participants did not remember how they received the information. No significant differences were found between PEQ and PIQ participants (χ^2=3.047, df=4, p=0.550). Table 5.4 in the next page includes quotes by participants on this topic.

TABLE 5. 4 QUOTES BY PARTICIPANTS WHO HAD WRITTEN MORE ABOUT THEIR EXPERIENCE ON THE QUESTION "*HOW DID YOU RECEIVE THE INFORMATION?"*

HOW DID YOU RECEIVE THE INFORMATION?	**QUOTE**
Unplanned face to face conversation	*Informal conversation with my father regarding taking the contraceptive pill* (R 171) *It was just mentioned over dinner one evening* (R 95)
Unplanned telephone call	*Family break up so call was out of the blue and unexpected* (R 222) *I wasn't aware until my father phoned me up to inform me that he was being tested* (R 111)
Pre-planned face to face meeting	*Appointment with consultant at hospital* (R. 102) *Meeting with my mom and genetic counsellor* (R 132)
Pre-planned telephone call	*My maternal aunt has had a large history of both breast and ovarian cancers- she mentioned she would be getting the test before, and when she had the results planned to contact me.* (R 130)
Another way	*My father who I wouldn't have much contact with sent me a text message* (R 56)

In my analysis I investigated whether they became aware that there might be a genetic condition in their family at the same time as becoming aware of their cancer family history. The majority of participants (n=132; 86.8%) were told at that time that the tendency to cancer in their family could be due to a genetic change, while 10 (6.6%) did not remember. No significant differences were found between PEQ and PIQ participants (p=0.361, Fisher's exact test). Some participants who already knew about the genetic predisposition in a family member only seemed to become aware of the meaning of this after receiving their own test result. For example participant 240 wrote "*I was told by my mother about her genetic status [...] I*

became aware that there might be a genetic condition in my family after my positive test result".

Among participants who received the information that a mutation was present in their family at a different time with respect to the awareness of cancer family history, there were participants who mentioned the genetic counselling with or without relatives and others specified the moment when the relatives received their genetic test result. For example participant 206 wrote "*We knew my Dad's history but didn't know it could be genetic until the letter came. We just thought it was possible we may get cancer but didn't know it was in the genes".*

Considering the age at which participants were told of the familial cancer risk, no significant differences were found between PEQ and PIQ participants (χ^2=2.075, df=3, p=0.557). A significant difference was observed in relation to the person who told participants about the possibility that members of their family might be more likely than others to develop cancer: 19 out of the 26 (73.1%) participants who had received the information from a person outside the family underwent genetic testing within one year of obtaining the information (χ^2=19.951, df=9, p=0.018).

The age at which young adults underwent presymptomatic genetic testing was also compared to the age when they were told about the genetic condition in their family. In general, the mean time that elapsed between receiving the information and when they were tested was 3.8±4.8 years (range 0-24 years). In more detail, 72 participants (47.4%) were tested within one year of receiving the information, 29 (19.1%) between two and four years after, 31 (20.4%) between five and 10 years after, and 20 (13.2%)

from 11 years or more later. With the aim of understanding if the attitudes and behaviours to be tested changed in relation to how early participants were informed, I dichotomised the sample comparing those who were told when the test was available for them (18 years and older) and those who were told at an age where the test was not yet available. One-hundred and eleven participants (75.5%) were informed after their 18th birthday, while 36 (24.5%) were informed earlier. Comparing these groups, it was revealed that the mean time that elapsed between receiving the information and when they were tested was 2.0±2.8 years (range 0-11) in the first group and 9.3±5.5 years (range 1-24) in the second one. No significant differences were found between PEQ and PIQ participants (p=1.000, Fisher's exact test) or connected with socio-demographic variables. However, observing the behaviours of participants who were told after 18 years of age, married participants were more likely to wait more time before testing than those who were single (2.8±3.3 *versus* 1.2±1.9; F=2.853, p=0.041). It has been observed that participants tested for hereditary breast and ovarian cancer were more likely to be informed after their 18th birthday than those tested for Lynch syndrome (77.5% *versus* 15.3%; χ^2=7.769, df=3, p=0.051). No significant difference in time elapsed between hearing of the risk and being tested was found based on who was the first person in their family who knew they had the mutation. However, it was revealed that participants who became aware of their potential genetic risk before their 18th birthday were more likely to be tested before 25 years of age than after 26 years of age (69.4% *versus* 30.6%; p=0.053, Fisher's exact test). Instead, there was unequal variance between those who were told after their 18th birthday and the age when they were tested and the independent t-test was not performed.

Observing the behaviours of participants who were told after 18 years of age, participants who were told by other family members were more likely to wait more time before testing than those who were told by a person outside the family (3.1±3.0 *versus* 0.6±0.8 F=4.795, p=0.010), while no such differences were observed comparing participants who were told before their 18th birthday (8.8±1.3 *versus* 9.0±6.2 F=0.012, p=0.988).

For convenience and for fluency in the text I have reported all the KMO measures of sampling adequacy and the Bartlett's test of sphericity results in Table 5.5 for all the items for which factor analysis was performed in below, in order as the items are cited though this result section.

TABLE 5. 5 KMO AND BARTLETT'S TEST PERFORMED ON DATA FROM THE YOUNG ADULTS AND PARENTS QUESTIONNAIRES

YOUNG ADULTS QUESTIONNAIRE

How did you react to the news that there might be a genetic condition in your family?

KMO Measure of Sampling Adequacy.		0.799
	Approx. Chi-Square	248.748
Bartlett's Test of Sphericity	df	21
	Sig.	0.000

How did you feel about the genetic counselling?

KMO Measure of Sampling Adequacy.		0.961
	Approx. Chi-Square	4429.632
Bartlett's Test of Sphericity	df	210
	Sig.	0.000

What were your reasons for wanting to be tested?

KMO Measure of Sampling Adequacy.		0.632
Bartlett's Test of Sphericity	Approx. Chi-Square	752.648
	Df	120
	Sig.	0.000

How did you feel after receiving your genetic test result

KMO Measure of Sampling Adequacy.		0.852
Bartlett's Test of Sphericity	Approx. Chi-Square	1241.939
	Df	136
	Sig.	0.000

How did you feel living with your genetic risk?

KMO Measure of Sampling Adequacy.		0.692
Bartlett's Test of Sphericity	Approx. Chi-Square	929.330
	Df	210
	Sig.	0.000

PARENTS QUESTIONNAIRE

What were your reasons for telling or not telling your children about the family cancer risk?

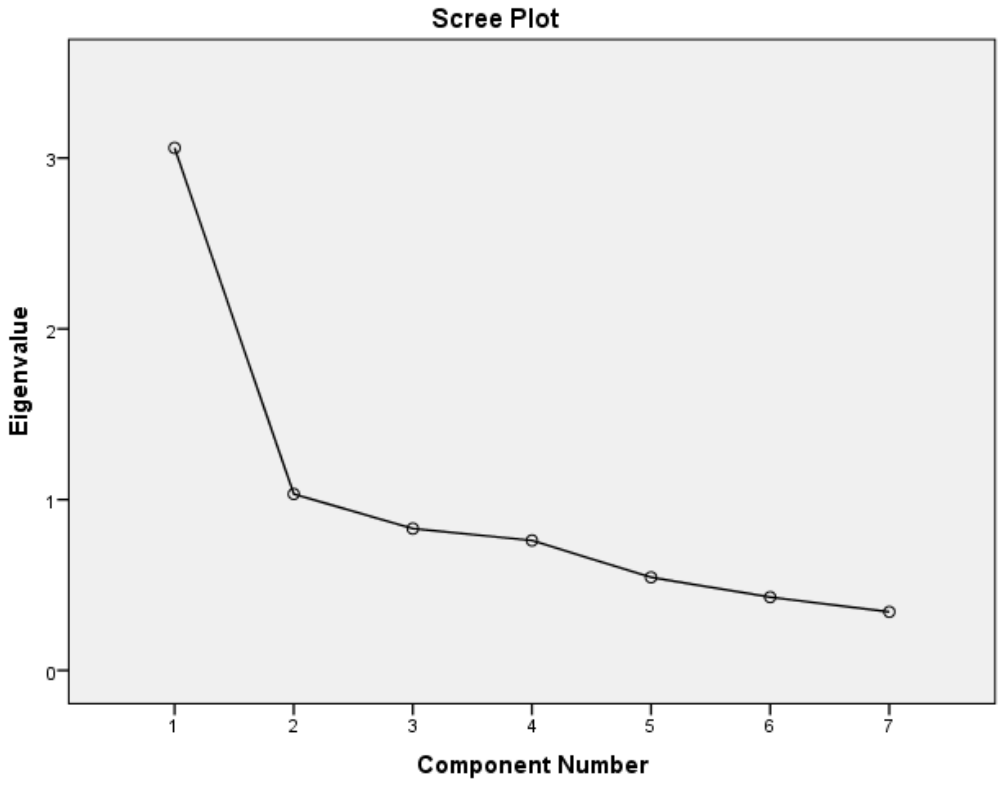
KMO Measure of Sampling Adequacy.		0.782
Bartlett's Test of Sphericity	Approx. Chi-Square	623.426
	df	153
	Sig.	0.000

The following questions concern how young adults reacted after they became aware of the family genetic condition. An exploratory factor analysis

was performed on the question "*How did you react to the news that there might be a genetic condition in your family?*".

The data were then subjected to a principal component analysis using a Varimax rotation. The analysis revealed two factors with Eigenvalues over 1.00. These two factors accounted for 58.1% of the explained variance: the first variable on the two factor rotation accounted for 43.1% of the unique variance and the second variable contributed 15.0% of the variance (Figure 5.1).

FIGURE 5. 1 SCREE PLOT OF COMPONENT AND EIGENVALUE FOR QUESTION "*HOW DID YOU REACT TO THE NEWS THAT THERE MIGHT BE A GENETIC CONDITION IN YOUR FAMILY?*"



Analysis of the structure matrix, which detailed the correlation between the variables and the factor, showed moderate to strong

correlations. Any item with a correlation below 0.40 was discarded. The factor loadings for each item were presented in Table 5.6.

TABLE 5. 6 ROTATED COMPONENT MATRIX QUESTION "*HOW DID YOU REACT TO THE NEWS THAT THERE MIGHT BE A GENETIC CONDITION IN YOUR FAMILY?*"

	COMPONENT	
	1	2
I did not know what it really meant	**0.758**	
I looked for information online	**0.678**	
I was more conscious of my risk	**0.603**	-0.428
I arranged the first counselling session to discuss my risk		0.**838**
I arranged the first counselling session to have a genetic blood test	**0.664**	-0.408
I did not want to know any more about it at the time		**0.575**
I felt it explained things I had been wondering about	**0.729**	

Lastly, I calculated the percentages of the item responses to identify areas with different reaction to the news that there might be a genetic condition in their family (Table 5.7).

TABLE 5. 7 PERCENTAGES OF EACH ITEM DIVIDED BY FACTORS INDIVIDUALIZED QUESTION "*HOW DID YOU REACT TO THE NEWS THAT THERE MIGHT BE A GENETIC CONDITION IN YOUR FAMILY?*"

	STRONGLY OR SOMEWHAT DISAGREE	NEITHER AGREE NOR DISAGREE	STRONGLY OR SOMEWHAT AGREE
Factor 1: Awareness			
I did not know what it really meant	31 (20.7%)	15 (10.0%)	104 (69.3%)
I looked for information online	10 (6.8%)	15 (10.1%)	123 (83.1%)
I was more conscious of my risk	36 (23.8%)	20 (13.2%)	95 (62.9%)
I arranged the first counselling session to have a genetic blood test	34 (54.0%)	24 (16.0%)	45 (30.0%)
I felt it explained things I had been wondering about	43 (28.9%)	46 (30.9%)	60 (40.3%)
Factor 2: Need for information			

I arranged the first counselling session to discuss my risk	81 (22.8%)	13 (8.7%)	102 (68.5%)
I wanted to know some more about it at the time	22 (14.7%)	16 (10.7%)	112 (74.7%)

No significant differences were found between PEQ and PIQ participants in relation to awareness (Factor 1) ($t(143)=0.665$; $p=0.507$), while a significant difference was observed in relation to need for information (Factor 2): PIQ were more likely to seek information/advice once they knew that there might be a genetic condition in their family (3.0±0.8 *versus* 2.7±0.8; $t(148)=-2.008$; $p=0.046$). A significant difference was also observed between participants who received the information at an unplanned or pre-planned time in relation to Factor 2: participants who received the information in a pre-planned conversation/call were more likely to need more information (3.0±0.7 *versus* 2.7±0.8; $t(1)=2.364$; $p=0.042$). No significant differences were found when the other parameters were considered.

In summary, the item "*How did you react to the news that there might be a genetic condition in your family?*" was analysed by means of factor analysis. It revealed two factors: awareness and the need for information.

Experience of genetic counselling

The following results concern participants' experiences of the genetic counselling they received in the genetic clinic. An exploratory factor analysis was performed on the question "*How did you feel about the genetic counselling?*".

Using a Varimax rotation the data were then subjected to a principal component analysis. This produced one factor with an Eigenvalue over 1.00. This one factor accounted for 77.6% of the explained variance (Figure 5.2). The factor loadings for each item were presented in Table 5.8.

FIGURE 5. 2 SCREE PLOT OF COMPONENT AND EIGENVALUE FOR QUESTION "*HOW DID YOU FEEL ABOUT THE GENETIC COUNSELLING?*"

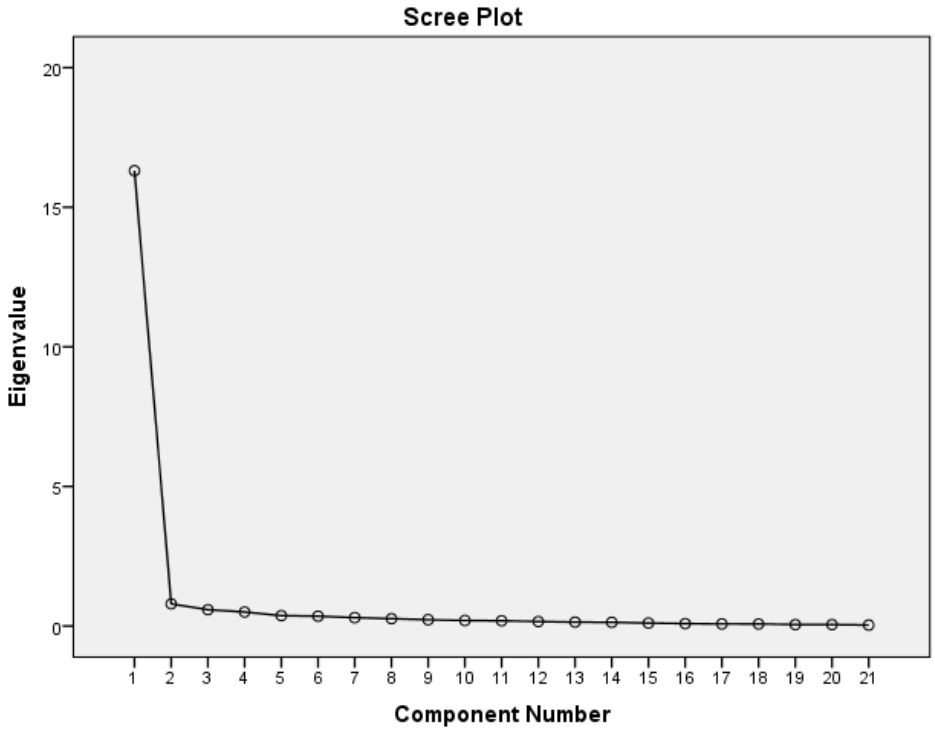


TABLE 5. 8 ROTATED COMPONENT MATRIX QUESTION "*HOW DID YOU FEEL ABOUT THE GENETIC COUNSELLING?*"

	COMPONENT
	1
The doctor or genetic counsellor showed an interest in your personal situation regarding the cancer family history	**0.813**
The doctor or genetic counsellor explained your risk to you clearly	**0.826**
The doctor or genetic counsellor met your expectations of him or her	**0.912**
The doctor or genetic counsellor treated you as an individual	**0.922**
You would be comfortable in calling the doctor or genetic counsellor to ask further questions	**0.881**
The doctor or genetic counsellor listened to what you had to say	**0.906**
The doctor or genetic counsellor was considerate of your emotional state during the meeting	**0.933**
You are satisfied with the way that information was communicated to you	**0.913**
The doctor or genetic counsellor understood what was really concerning you	**0.922**
The doctor or genetic counsellor made you feel you were "in good hands"	**0.922**
The doctor or genetic counsellor made you feel that they knew how to handle situations like your's	**0.913**
The doctor or genetic counsellor gave you enough of their time	**0.892**
The doctor or genetic counsellor was sensitive and tactful during your conversation	**0.936**
The doctor or genetic counsellor seemed to be an expert in the field	**0.854**
The doctor or genetic counsellor helped you deal with any concerns you had	**0.938**
You felt comfortable to talk about yourself during the genetic counselling session	**0.809**
You were satisfied with the length of time you had to wait until your first appointment	**0.603**
You were satisfied with the information your received during the genetic counselling appointment	**0.902**
If a friend needed similar help you would recommend this clinic to him or her	**0.898**
The counselling was given in an appropriate setting	**0.804**
Overall you are satisfied with the genetic counselling service	**0.933**

Lastly, I calculated the percentages of the item responses to identify areas with different experience of the counselling session (Table 5.9).

TABLE 5. 9 PERCENTAGES OF EACH ITEM DIVIDED BY FACTORS INDIVIDUALIZED QUESTION "*HOW DID YOU FEEL ABOUT THE GENETIC COUNSELLING?"*

	STRONGLY OR SOMEWHAT DISAGREE	NEITHER AGREE NOR DISAGREE	STRONGLY OR SOMEWHAT AGREE
Factor 1: Satisfaction with genetic counselling			
The doctor or genetic counsellor showed an interest in your personal situation regarding the cancer family history	14 (9.6%)	10 (6.8%)	123 (83.7%)
The doctor or genetic counsellor explained your risk to you clearly	12 (8.2%)	8 (5.4%)	123 (83.1%)
The doctor or genetic counsellor met your expectations of him or her	19 (13.0%)	14 (9.6%)	113 (77.4%)
The doctor or genetic counsellor treated you as an individual	15 (10.2%)	10 (6.8%)	122 (83.0%)
You would be comfortable in calling the doctor or genetic counsellor to ask further questions	27 (18.4%)	16 (10.9%)	104 (70.7%)
The doctor or genetic counsellor listened to what you had to say	14 (9.7%)	15 (10.3%)	116 (80.0%)
The doctor or genetic counsellor was considerate of your emotional state during the meeting	20 (13.7%)	16 (11.0%)	110 (75.3%)
You are satisfied with the way that information was communicated to you	21 (14.3%)	11 (7.5%)	115 (78.3%)
The doctor or genetic counsellor understood what was really concerning you	21 (14.5%)	14 (24.1%)	110 (75.8%)
The doctor or genetic counsellor made you feel you were "in good hands"	22 (15.1%)	14 (9.6%)	110 (75.3%)
The doctor or genetic counsellor made you feel that they knew how to handle situations like your's	23 (15.6%)	15 (10.2%)	109 (74.1%)
The doctor or genetic counsellor gave you enough of their time	16 (10.9%)	13 (8.8%)	118 (80.3%)
The doctor or genetic counsellor was sensitive and tactful during your conversation	18 (12.2%)	8 (5.4%)	121 (82.3%)
The doctor or genetic counsellor seemed to be an expert in the field	19 (12.9%)	11 (7.5%)	117 (79.6%)
The doctor or genetic counsellor helped you deal with any concerns you had	19 (13.0%)	20 (13.7%)	107 (73.3%)

You felt comfortable to talk about yourself during the genetic counselling session	17 (11.6%)	16 (10.9%)	114 (77.5%)
You were satisfied with the length of time you had to wait until your first appointment	33 (22.6%)	19 (13.0%)	94 (64.4%)
You were satisfied with the information your received during the genetic counselling appointment	22 (15.0%)	11 (7.5%)	114 (77.5%)
If a friend needed similar help you would recommend this clinic to him or her	19 (13.0%)	18 (12.2%)	110 (74.8%)
The counselling was given in an appropriate setting	10 (6.8%)	11 (7.5%)	126 (85.7%)
Overall you are satisfied with the genetic counselling service	18 (12.2%)	14 (9.5%)	115 (78.3%)

Participants answering the English questionnaire were more likely to describe themselves as satisfied with genetic counselling than PIQ, considering the mean of responses (4.2±1.0 *versus* 3.7±1.3; $t(141)=2.121$, $p=0.036$). Participants who were told about the cancer risk in their family by people outside their family were more satisfied with the counselling session than those who were told by siblings (4.5±1.0 *versus* 3.2±1.1; $F=2.734$, $p=0.046$). No significant differences were found when the other parameters were considered.

In summary, the item "*How did you feel about the genetic counselling?*" was analysed by means of factor analysis. Satisfaction about genetic counselling was revealed as the only one factor.

Decision making process

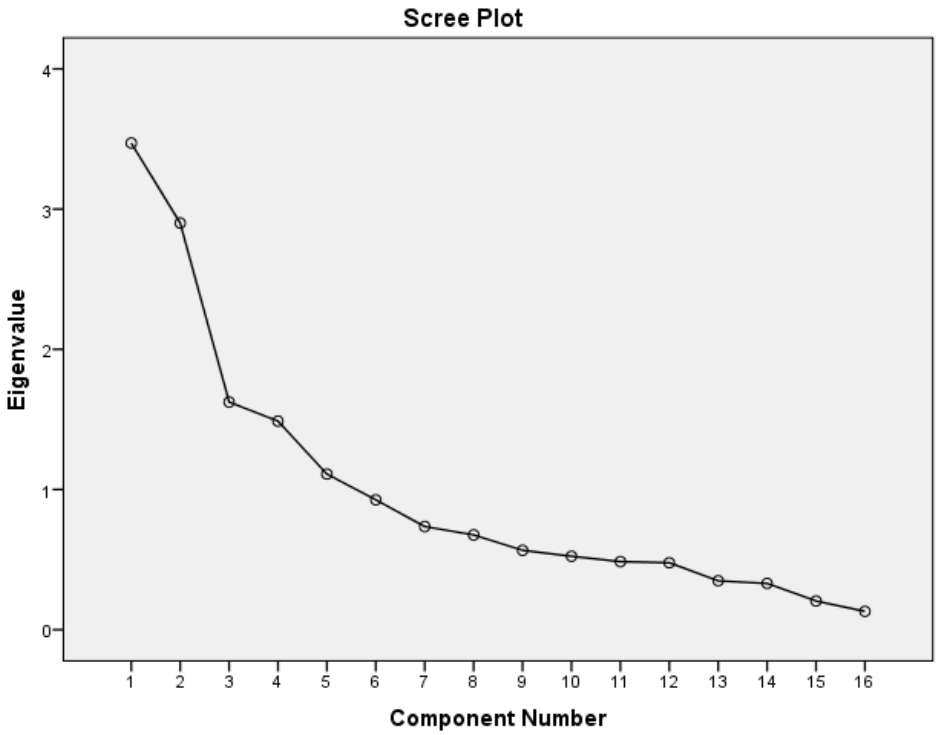
This part of the analysis concerns questions aimed at evaluating young adults' reasons for their testing decisions and their experience of the decision making process. The majority of participants (n=105; 75.5%) responded "myself" when asked about the person who decided that they would be tested, while "both myself and parents" was mentioned by 23 (16.5%), "parents" by four (2.9%), "aunt" by four (2.9%), and genetic counsellor/doctor by three (2.2%). The number of PEQ tested because the decision was made by themselves was significantly higher if compared to PIQ (96/116, 82.8% *versus* 9/23, 39.1%; χ^2=38.715, df=4, p=0.00). Moreover, participants with children were significantly more likely to request testing themselves if compared to participants without children (60/67, 89.6% *versus* 45/72, 62.5%, χ^2=14.663, df=4, p=0.005). Women participants were more likely to decide autonomously than men (101/128, 78.9% *versus* 3/10, 30%; χ^2=14.117, df=4, p=0.007). Participants who decided themselves to be tested were less likely to need more information than those decided by aunts (2.7±0.7 *versus* 4.1±0.9; F=5.173, p=0.001). Consistently, participants who decided themselves were more satisfied with the genetic counselling received than those decided by aunts (4.2±1 *versus* 2.2±1.4; F=3.910, p=0.005). No significant differences were found when the other parameters were considered.

The following questions concern the reasons for wanting to be tested. An exploratory factor analysis was performed on question 44 which was "*What were your reasons for wanting to be tested?*".

Five factors with Eigenvalues over 1.00 resulted from a principal component analysis using a Varimax rotation. These five factors accounted

for 66.2% of the explained variance: the first variable on the five factor rotation accounted for 21.7% of the unique variance, the second variable contributed 18.1% of the variance, the third variable contributed 10.1%, the fourth variable contributed 9.3%, and the fifth variable contributed 7.0%. No other factor contributed greater than 5.6% of the variance (Figure 5.3).

FIGURE 5. 3 SCREE PLOT OF COMPONENT AND EIGENVALUE FOR QUESTION "WHAT WERE YOUR REASONS FOR WANTING TO BE TESTED?"



Analysis of the structure matrix, which detailed the correlation between the variables and the factor, showed moderate to strong correlations. Any item with a correlation below 0.40 was discarded. The factor loadings for each item are presented in Table 5.10.

TABLE 5. 10 ROTATED COMPONENT MATRIX QUESTION "*WHAT WERE YOUR REASONS FOR WANTING TO BE TESTED?*"

	COMPONENT				
	1	2	3	4	5
I wanted to learn about my children's risk or risks to any children I may have				**0.818**	
I wanted to try to help advance research	**0.505**				
I wanted to know if I need to get cancer screening tests more often	**0.620**				
I wanted to be reassured	**0.743**				
I wanted to make a decision about surgery to reduce my risk	**0.762**				
I wanted to make a decision about having (more) children				**0.728**	
My mother strongly encouraged me					**0.640**
My father strongly encouraged me					**0.895**
I had genetic testing because of pressure from my family members			**0.830**		
I had genetic testing because my parent asked me to do it			**0.835**		
I made my own decision	**0.808**				
My decision was influenced by family experience	**0.528**				
My mother warned me about having the test		**0.702**			
My father warned me about having the test		**0.749**			
My mother advised me to wait, but I decided to have it		**0.812**			
My father advised me to wait, but I decided to have it		**0.878**			

Lastly, I calculated the percentages of the item responses to identify areas with different reasons for wanting to be tested (Table 5.11).

TABLE 5. 11 PERCENTAGES OF EACH ITEM DIVIDED BY FACTORS INDIVIDUALIZED QUESTION "*WHAT WERE YOUR REASONS FOR WANTING TO BE TESTED?*"

	STRONGLY OR SOMEWHAT DISAGREE	NEITHER AGREE NOR DISAGREE	STRONGLY OR SOMEWHAT AGREE
Factor 1: Proactivity			
I wanted to try to help advance research	20 (14.5%)	26 (18.8%)	87 (63.0%)
I wanted to know if I need to get cancer screening tests more often	4 (2.9%)	7 (5.1%)	113 (83.1%)
I wanted to be reassured	9 (6.5%)	29 (21.0%)	92 (66.7%)
I wanted to make a decision about surgery to reduce my risk	13 (9.4%)	14 (10.1%)	97 (70.5%)
I made my own decision	5 (3.6%)	8 (5.8%)	95 (68.8%)
My decision was influenced by family experience	14 (10.2%)	21 (15.3%)	88 (67.7%)
Factor 2: Parents' pressure against testing			
My mother warned me about having the test	78 (60.0%)	26 (19.0%)	16 (11.7%)
My father warned me about having the test	88 (64.8%)	25 (18.4%)	8 (5.9%)
My mother advised me to wait, but I decided to have it	95 (68.8%)	16 (11.6%)	9 (6.5%)
My father advised me to wait, but I decided to have it	100 (74.8%)	15 (11.1%)	5 (3.8%)
Factor 3: Parents' decision to be tested			
I had genetic testing because of pressure from my family members	99 (71.8%)	19 (13.8%)	16 (11.6%)
I had genetic testing because my parent asked me to do it	92 (66.7%)	23 (16.7%)	17 (12.3%)
Factor 4: Concern for children			
I wanted to learn about my children's risk or risks to any children I may have	10 (7.3%)	19 (13.9%)	92 (67.1%)
I wanted to make a decision about having (more) children	34 (24.8%)	26 (19.0%)	59 (43.2%)
Factor 5: Parent's pressure for testing			
My mother strongly encouraged me	35 (25.3%)	31 (22.5%)	59 (42.7%)
My father strongly encouraged me	44 (32.3%)	36 (26.4%)	44 (32.3%)

No significant differences were found between PEQ and PIQ participants, considering the mean of responses, in relation to Factor 1 ($t(133)=0.703$; $p=0.484$), Factor 2 ($t(133)=-0.634$; $p=0.527$), Factor 4 ($t(134)=-0.558$; $p=0.578$) and Factor 5 ($t(134)=-1.191$; $p=0.507$). There was unequal variance between PEQ and PIQ in relation to Factor 3 and the independent t-test was not performed.

A significant difference was observed in relation to proactivity (Factor 1) when the timing of the test was examined. Participants who underwent the genetic test within one year of obtaining the information were more likely to be proactive than those who underwent the genetic test between two and four years (3.9 ± 0.9 *versus* 3.2 ± 1.3; $F=2.987$, $p=0.034$). No significant differences were found when the other parameters were considered.

A significant difference was also observed in relation to parental pressure against testing (Factor 2): participants tested between 26-30 years of age were less likely to undergo it because of parental pressure than participants who underwent the genetic test between 18-25 years of age (1.2 ± 0.7 *versus* 1.6 ± 1.1; $t(133)=3.030$, $p=0.003$). Significant differences were also observed in relation to parent's decision (Factor 3) with gender, daily work, age at genetic test, and who decided to be tested. Men were more likely to have undergone genetic testing because of their parent's decision than women (3.4 ± 1.3 *versus* 1.6 ± 1.4; $t(135)=4.640$, $p=0.000$). Also, parents were more likely to influence the decision of those who were tested between 18-25 years of age than those tested at 26-30 years of age (2.0 ± 1.3 *versus* 1.5 ± 1.1; $t(136)=2.008$, $p=0.047$). Consistently, participants who reported they made the decision with their parents were more likely to be influenced by their

parents than those who reported deciding for themselves (2.7±1.4 *versus* 2.7±1.4; F=15.357, p=0.000).

Significant differences were also observed in relation to parents' pressure toward testing (Factor 5) when analysed against having children, condition tested, age at genetic test, and who decided to be tested. Participants without children were more likely to undergo genetic testing because of pressure from parents than participants who had children (3.1±1.4 *versus* 2.5±1.4; t(134)=-2.771, p=0.006). Participants tested for Lynch syndrome were more likely to have a genetic test because of pressure from parents than those tested for hereditary breast and ovarian cancer (3.4±1.4 *versus* 2.6±1.4; F=4.014, p=0.020). Participants tested between 18-25 years of age were more likely to undergo it because of parents' pressure than those who underwent a genetic test between 26-30 years of age (3.0±1.4 *versus* 2.5±1.4; t(134)=2.202, p=0.029).

In summary, the item "*How did you feel after receiving your genetic test result?"* was analysed by means of factor analysis. Five factors were revealed:

- negative feelings
- negative impact on relationships
- uncertainties about the meaning of test result
- worry for relatives
- perceiving the test result as helpful.

Genetic test result

In this section I present the results concerning how young adults felt after receiving their genetic test result. An exploratory factor analysis was performed on the question "*How did you feel after receiving your genetic test result?*".

A principal component analysis (Varimax rotation) indicated there were five factors with Eigenvalues over 1.00. These five factors accounted for 72.2% of the explained variance: the first variable on the five factor rotation accounted for 38.0% of the unique variance, the second variable contributed 13.7% of the variance, the third variable contributed 7.7%, the fourth variable contributed 6.8%, and the fifth variable contributed 6.0%. No other factor contributed greater than 5.6% of the variance (Figure 5.4).

Analysis of the structure matrix, which detailed the correlation between the variables and the factor, showed moderate to strong correlations. Any item with a correlation below 0.40 was discarded. The factor loadings for each item were presented in Table 5.12.

FIGURE 5. 4 SCREE PLOT OF COMPONENT AND EIGENVALUE FOR QUESTION *HOW DID YOU FEEL AFTER RECEIVING YOUR GENETIC TEST RESULT?*

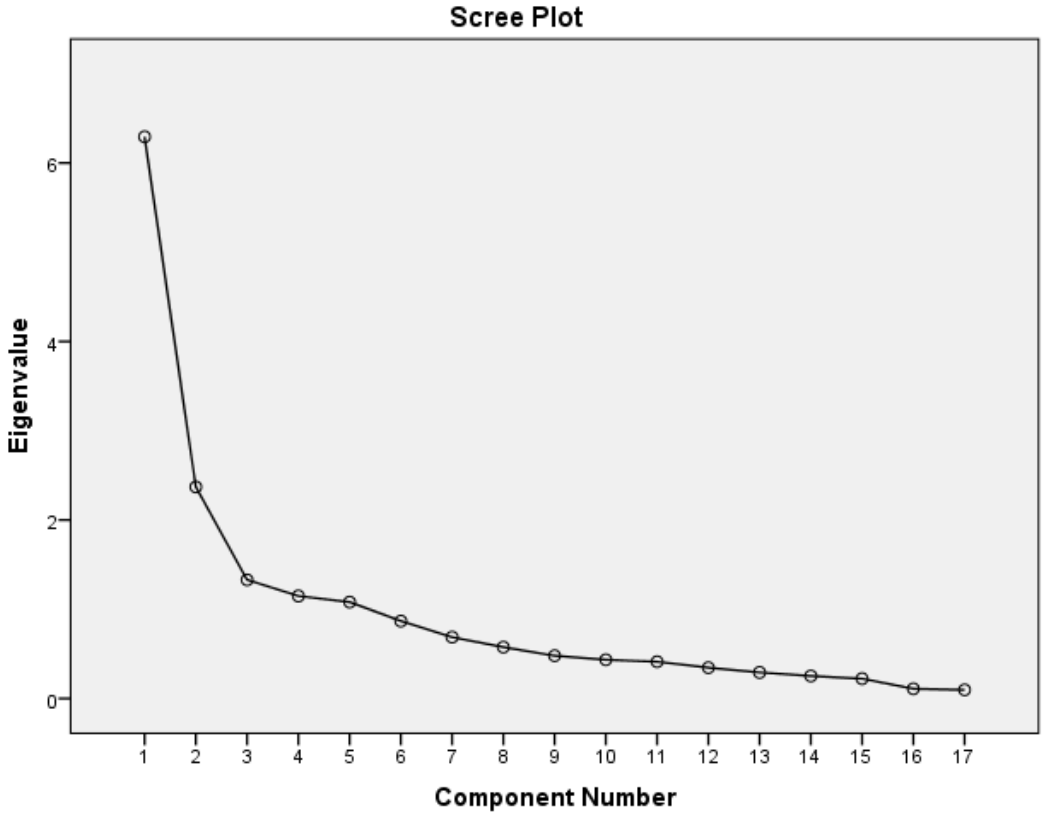


TABLE 5. 12 ROTATED COMPONENT MATRIX QUESTION "*HOW DID YOU FEEL AFTER RECEIVING YOUR GENETIC TEST RESULT?*"

Rotated Component Matrix[a]

	COMPONENT				
	1	2	3	4	5
I felt upset about my test result	**0.907**				
I felt sad about my test result	**0.865**				
I felt anxious or nervous about my test result	**0.810**				
I felt guilty about my test result		**0.514**		0.494	
I felt relieved about my test result	-0.431				**0.602**
I felt a loss of control	**0.575**	0.549			
I had problems enjoying life because of my test result	0.478	**0.640**			
I felt able to plan my future					**0.901**
I was more worried about my risk of getting cancer	**0.753**				

I was uncertain about what my test result meant for my cancer risk		**0.750**	
I was uncertain about what my test result meant for my children or any children I may have		**0.888**	
I was uncertain about what my test result meant for my family's cancer risk		**0.900**	
I was worried other people might discuss this behind my back	**0.795**		
I was worried that other people might think less of me because of my result	**0.763**		
I was worried because of the possibility of passing the mutation to my children or any children I may have			**0.798**
I felt guilty about my family			**0.753**
I felt more distant from family members	**0.637**		

Lastly, I calculated the percentages of the item responses to identify areas with different reasons for wanting to be tested (Table 5.13).

TABLE 5. 13 PERCENTAGES OF EACH ITEM DIVIDED BY FACTORS INDIVIDUALIZED QUESTION "*HOW DID YOU FEEL AFTER RECEIVING YOUR GENETIC TEST RESULT?"*

	STRONGLY OR SOMEWHAT DISAGREE	**STRONGLY OR SOMEWHAT AGREE**
Factor 1: Negative feelings		
I felt upset about my test result	36 (26.7%)	99 (73.3%)
I felt sad about my test result	27 (20.0%)	108 (80.0%)
I felt anxious or nervous about my test result	40 (29.6%)	95 (70.4%)
I was more worried about my risk of getting cancer	28 (20.7%)	107 (79.2%)
I felt a loss of control	70 (51.5)	66 (48.5%)
Factor 2: Negative impact on relationships		
I felt guilty about my test result	95 (69.8%)	41 (30.1%)
I had problems enjoying life because of my test result	84 (62.2%)	51 (37.5%)

I was worried other people might discuss this behind my back	114 (85.1%)	20 (14.9%)
I was worried other people might think less of me because of my result	114 (83.8%)	22 (16.2%)
I felt more distant from family members	115 (84.6)	21 (15.4%)
Factor 3: Uncertainties about the meaning of test result		
I was uncertain about what my test result meant for my cancer risk	112 (82.4%)	24 (17.6%)
I was uncertain about what my test result meant for my children or any children I may have	103 (76.8%)	31 (23.2%)
I was uncertain about what my test result meant for my family's cancer risk	106 (78.5%)	29 (21.5%)
Factor 4: Worry for relatives		
I was worried because of the possibility of passing the mutation to my children or any children I may have	23 (17.0%)	112 (83.0%)
I felt guilty about my family	87 (64.0%)	49 (36.0%)
Factor 5: Perceiving the test as helpful		
I felt relieved about my test result	83 (61.5%)	52 (38.5%)
I felt able to plan my future	42 (31.1%)	93 (68.9%)

Significant differences were observed between PEQ and PIQ, considering the mean of responses, in relation to negative feelings (Factor 1) and worry for relatives (Factor 4) : PEQ were more likely to experience negative feelings than PIQ (2.9±0.9 *versus* 2.4±1.1; t(131)=2.596, p=0.011), and PEQ were more likely to feel worry for relatives than PIQ (2.8±0.9 *versus* 2.2±0.9; t(133)=2.557, p=0.012). No significant differences were found between PEQ and PIQ in relation to Factor 2 (t(131)=1.714; p=0.089), Factor 3 (t(131)=0.688; p=0.493) and Factor 5 (t(132)=-0.049; p=0.961).

A significant difference was observed in relation to negative feelings (Factor 1): participants who received a positive test result, as expected, were more likely to feel negative feelings than those who received a negative test

result (3.0±0.8 *versus* 1.5±0.9; t(131)=4.958, p=0.000). Participants who received the information in an unplanned or pre-planned time: participants who received the information in an unplanned conversation/call were more likely to feel negative feelings (3.0±0.8 *versus* 2.7±0.9; t(125)=2.060; p=0.041). A significant difference was also observed in relation to negative impact on relationships (Factor 2): participants who decided to be tested because of parents' desire were less likely to have negative impact on relationships than those decided by aunts (1.4±0.5 *versus* 2.9±1.4; F=2.492, p=0.046).

Significant differences were also observed in relation to uncertainties about the meaning of test results (Factor 3) with the condition tested and who decided they should be tested. Those tested for Lynch syndrome were more likely to have uncertainties about the meaning of the test result than those tested for hereditary breast and ovarian cancer (2.2±0.9 *versus* 1.6±0.8; F=3.773, p=0.012). Participants who decided to be tested because of parental wishes were less likely to have uncertainties about the meaning of test results than those were influenced by their aunts to be tested (1.3±0.5 *versus* 3.0±1.4; F=3.582, p=0.008).

In relation to worry for relatives (Factor 4) with daily work and their genetic test result, significant differences were also found. Homemaker participants were more likely to be worried for their relatives when compared to those not working and students (3.2±0.9 *versus* 1.8±1.0; F=2.525, p=0.044). Consistently, participants who received a positive test result were more likely to be worry for their relatives than those who received a negative test result (2.7±0.9 *versus* 1.8±0.8; t(133)=3.316,

p=0.001). No significant differences were found when other socio-demographic variables and other parameters were considered.

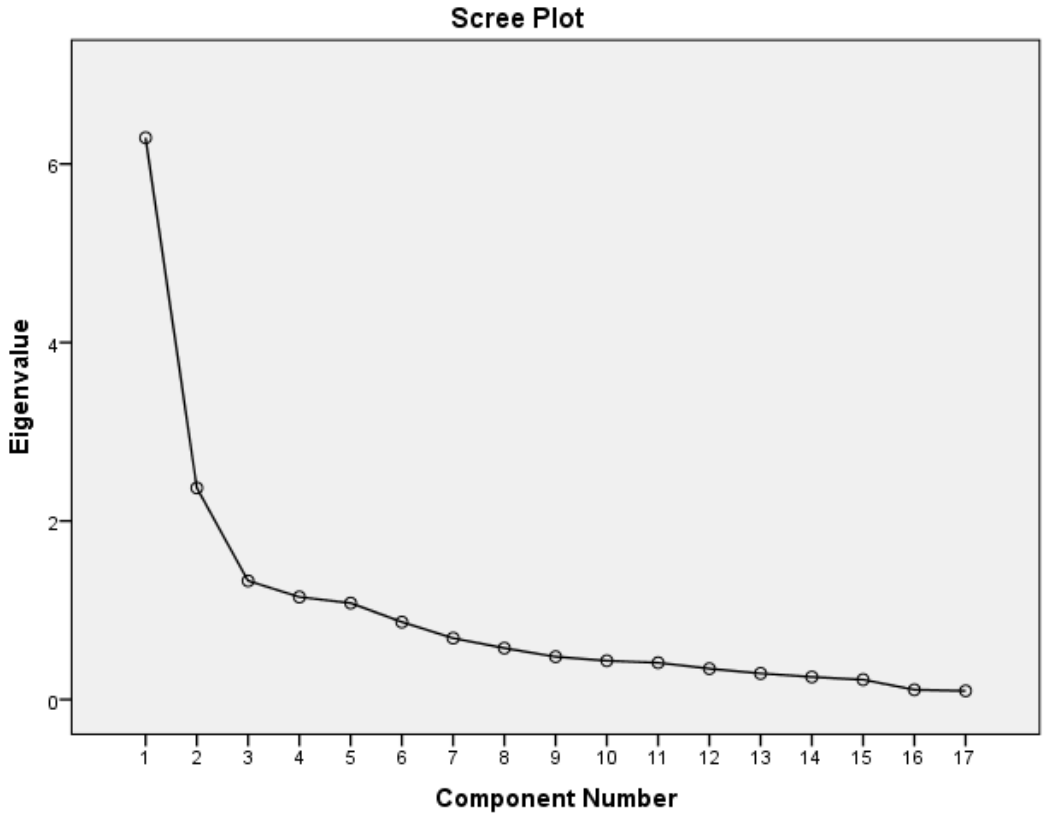
Significant differences were also observed in relation to perceiving the test as helpful (Factor 5), and with the identity of the first person in the family to test positive for the mutation. Those who received a positive test result were more likely to perceive the test as helpful than those who received a negative test result (2.7±0.9 *versus* 1.8±0.8; t(133)=3.316, p=0.001). Participants were more likely to perceive the test as helpful when the first person in their family tested was a first-degree relative, compared to other relatives (2.6±0.8 *versus* 2.3±0.9; t(132)=2.262, p=0.025).

Living with genetic risk

Participants were asked about their experiences of living with genetic risk (Q46) and an exploratory factor analysis was performed on the results.

The data were subjected to a principal component analysis using a Varimax rotation, resulting in seven factors with Eigenvalues over 1.00. These seven factors accounted for 66.7% of the explained variance and one of them contributed greater than 5.6% of the variance. Inspection of the scree plot for this data showed a distinct separation between the fourth and fifth factors, which suggests that four factors should be retained. The scree plot is presented in Figure 5.5.

FIGURE 5. 5 SCREE PLOT OF COMPONENT AND EIGENVALUE FOR QUESTION "*HOW DID YOU FEEL LIVING WITH YOUR GENETIC RISK?*"



In order to further reduce the number of items selected for the question "*How did you feel living with your genetic risk?"*, the data were subjected to another Varimax rotation with the specification that only four factors be extracted. An analysis of the factor rotations showed the four-factor rotation more accurately represented the proposed constructs than the seven factor rotation. The first variable on the four factor rotation accounted for 21.2% of the unique variance, the second variable contributed 13.1% of the variance, the third variable contributed 8.6% of the variance, and the fourth variable contributed 6.8% of the variance. Analysing the structure matrix, which detailed the correlation between the variables and the factor, showed moderate to strong correlations. Any items with a

correlation below 0.30 was discarded. The factor loadings for each item are presented in Table 5.14.

TABLE 5. 14 ROTATED COMPONENT MATRIX QUESTION *HOW DID YOU FEEL LIVING WITH YOUR GENETIC RISK?*

	COMPONENT			
	1	2	3	4
Having time before the regular cancer screening was due to start gave me the opportunity to think about it		**0.592**		
Having time before the cancer screening was due to start gave me the opportunity to think about having surgery to reduce my risk		**0.502**	0.431	
I was having difficulty making decisions about cancer screening or measures to reduce my risk			**0.562**	
I understood my choice for cancer prevention or early detection clearly		**0.494**		
I felt frustrated that there are no ways I can completely prevent cancer		**0.522**		
I thought about having risk -reducing surgery sooner rather than later			**0.485**	
My parents strongly encouraged me to have surgery to reduce my risk of cancer			**0.464**	
Thinking about my test result has affected my work or family life			**0.643**	
I had difficulty talking about my test results with family members			**0.768**	
I felt satisfied with family communication about my genetic test result		**0.588**		
I have wondered about when to share my genetic risk with a new partner	**0.890**			
I have wondered about how early in a relationship to discuss having children	**0.861**			
I have wondered about how early in a relationship to discuss surgery to reduce my risk	**0.867**			
I was worried about the possibility of my children (or any children I may have) getting cancer		**0.708**		
I was feeling guilty about possibly passing on the disease risk to my children or any children I may have		**0.634**		0.469

I decided to limit the number of children I have because I may pass on the mutation		**0.428**	
I regretted my choice to have children			**0.704**
I try to do all I can to stay alive for my children			**0.802**
I have confidence in the cancer screening procedures			**0.431**
I feel anxious waiting for the first or next screening		**0.475**	
I try not to think about the cancer risk because I am too young yet for screening	**0.351**		

Lastly, I calculated the percentages of the item responses to identify areas according to different reasons for wanting to be tested (Table 5.15).

TABLE 5. 15 PERCENTAGES OF EACH ITEM DIVIDED BY FACTORS INDIVIDUALISED QUESTION "*HOW DID YOU FEEL LIVING WITH YOUR GENETIC RISK?*"

	STRONGLY OR SOMEWHAT DISAGREE	STRONGLY OR SOMEWHAT AGREE
Factor 1: Influence on lifestage perception		
I have wondered about when to share my genetic risk with a new partner	46 (35.4%)	42 (32.4%)
I have wondered about how early in a relationship to discuss having children	47 (35.9%)	37 (28.2%)
I have wondered about how early in a relationship to discuss surgery to reduce my risk	46 (35.1%)	42 (32.0%)
I try not to think about the cancer risk because I am too young yet for screening	69 (53.1%)	32 (24.6%)
Factor 2: Impact of test result on own prevention and on relatives		
Having time before the regular cancer screening was due to start gave me the opportunity to think about it	26 (20.1%)	86 (66.7%)
Having time before the cancer screening was due to start gave me the opportunity to think about having surgery to reduce my risk	29 (11.6%)	95 (73.6%)
I understood my choice for cancer prevention or early detection clearly	10 (7.8%)	111 (87.7%)
I felt frustrated that there are no ways I can completely prevent cancer	32 (24.4%)	89 (68.0%)

I felt satisfied with family communication about my genetic test result	24 (18.7%)	96 (74.4%)
I was worried about the possibility of my children (or any children I may have) getting cancer	12 (9.3%)	103 (79.3%)
I was feeling guilty about possibly passing on the disease risk to my children or any children I may have	22 (16.8%)	92 (70.3%)
Factor 3: Anxiety		
I was having difficulty making decisions about cancer screening or measures to reduce my risk	89 (68.4%)	33 (25.4%)
I thought about having risk -reducing surgery sooner rather than later	27 (20.7%)	90 (69.3%)
My parents strongly encouraged me to have surgery to reduce my risk of cancer	78 (59.5%)	30 (23.0%)
Thinking about my test result has affected my work or family life	58 (44.3%)	66 (50.4%)
I had difficulty talking about my test results with family members	95 (73.8%)	30 (23.8%)
I decided to limit the number of children I have because I may pass on the mutation	70 (53.5%)	40 (30.6%)
I feel anxious waiting for the first or next screening	29 (22.4%)	83 (63.8%)
Factor 4: Protection of self and children		
I regretted my choice to have children	73 (56.6%)	12 (9.4%)
I try to do all I can to stay alive for my children	6 (4.6%)	75 (58.2%)
I have confidence in the cancer screening procedures	28 (21.6%)	95 (73.8%)

A significant difference was observed between PEQ and PIQ, considering the mean of responses, in relation to influence of lifestage perception (Factor 1): PIQ were more likely to perceive influence of lifestage than PEQ (2.2±1.1 *versus* 1.4±1.1; t(127)=-2.701, p=0.008); while no significant differences were found between PEQ and PIQ in relation to Factor 2 (t(121)=0.996; p=0.321), and Factor 4 (t(126)=0.235; p=0.815). There was unequal variance between PEQ and PIQ in relation to Factor 3 and the Independent t-test was not performed.

In relation to influence of lifestage perception (Factor 1) with marital status, condition tested, and who decided to be tested, significant differences appeared to be present. Single participants were more likely to perceive influence of lifestage than married participants (2.1±1.1 *versus* 1.4±1.0; F=5.584, p=0.001), while those tested for Lynch syndrome were more likely to perceive influence of lifestage than those tested for FAP (2.1±1.2 *versus* 1.1±1.3; F=3.083, p=0.030). Participants who decided to be tested because of the influence of an aunt were more likely to perceive influence of lifestage than participants who decided after discussion with a genetic counsellor or a physician (3.6±0.7 *versus* 1.3±1.3; F=4.095, p=0.004).

Consistently, a significant difference was observed in relation to impact of test result on own prevention and on relatives (Factor 2): participants who received a positive test result were more likely to have a favourable impact of the test result on their own prevention and on relatives than those who received a negative test result (3.0±0.8 *versus* 1.7±1.2; t(121)=3.343, p=0.001). In relation to the impact of test result on own prevention and on relatives and age at genetic test: participants tested between 26-30 years of age were less likely to have favourable impact of test result on own prevention and on relatives than participants who underwent genetic testing between 18-25 years of age (2.0±0.7 *versus* 2.2±0.8; t(121)=2.127, p=0.035). Significant differences were also observed in relation to anxiety (Factor 3) with genetic test result, and who influenced the testing decision. Consistently, participants who received a positive test result were more likely to feel anxious than those who received a negative test result (2.2±0.7 *versus* 1.2±0.7; t(125)=3.043, p=0.003). Participants whose

parents decided they should be tested were less likely to feel anxious than those influenced by aunts (1.3±0.9 *versus* 3.1±1.2; F=3.030, p=0.020).

A significant difference was also found, as expected, between protection of self and children (Factor 4) when compared to marital status. Married participants were more likely to feel self and children protection than single participants (2.4±0.9 *versus* 1.8±1.0; F=4.000, p=0.009).

In summary, the item "*How did you feel living with your genetic risk?*" was analysed by means of factor analysis. Four factors, were revealed, these were:

- influence on lifestage perception
- impact of test result on own prevention and on relatives
- anxiety
- protection of self and children.

5.3.3 PARENT QUESTIONNAIRE FINDINGS

Demographic characteristics of parent participants

The demographic information of the study participants is shown in Table 5.16: the mean age at time of completing the questionnaire was 52.4 years (39-78); no significant differences were found between English and Italian participants with respect to age, education, daily work and marital status. Twenty-five participants (59.5%) had both daughters and sons, while 12 (28.6%) had only daughters and five (11.9%) had only sons.

TABLE 5. 16 SAMPLE CHARACTHERISTICS: PARENT PARTICIPANTS

	ALL (N=42)	PEQ (N=34)	PIQ (N=8)	*p-value*
Age at questionnaire (years)				
mean±SD	51.9±7.6	51.7±7.3	55.1±3.8	0.211 [a]
Gender				
Male	4 (9.5%)	4 (100.0%)	0	0.572[c]
Female	38 (90.5%)	30 (78.9%)	8 (21.1%)	
Country				
Australia	1 (2.4%)	1 (2.9%)	0	-
Germany	2 (4.8%)	2 (5.9%)	0	
Ireland	3 (7.1%)	3 (8.8%)	0	
Italy	8 (19.0%)	0	8 (100.0%)	
United Kingdom	17 (40.5%)	17 (50.0%)	0	
United States of America	11 (26.2%)	11 (32.4%)	0	
Education				
Secondary school	10 (23.8%)	9 (26.5%)	1 (12.5%)	0.461[b]
Post-secondary educat.	20 (47.6%)	15 (44.1%)	5 (62.5%)	
University degree	7 (16.7%)	5 (14.7%)	2 (25.0%)	
Postgraduate degree	5 (11.9%)	5 (14.7%)	0	
Daily work				
Paid employment	29 (69.0%)	23 (67.6%)	6 (75.0%)	0.392[b]
Homemaker	7 (16.7%)	5 (14.7%)	2 (25.0%)	
Not working not student	6 (14.3%)	6(17.6%)	0	
If paid employment (n=29)				
Paid employee	19 (65.5%)	14 (60.9%)	5 (83.3%)	0.162[b]
Manager	1 (3.4%)	1 (4.3%)	0	

Self-employed	2 (6.9%)	2 (8.7%)	0	
Business owner	1 (3.4%)	0	1 (16.7%)	
Professional	6 (20.7%)	6 (26.1%)	0	
Marital status				
Single (never married)	1 (2.9%)	1 (2.9%)	0	0.378[b]
Married	32 (76.2%)	24 (70.6%)	8 (100.0%)	
Married	8 (19.0%)	8 (23.5%)	0	
Living with a partner	1 (2.4%)	1 (2.3%)	0	

[a] Independent samples T-test
[b] Pearson chi-squared test

[c] Fisher's exact test

The majority of participants (n=25, 59.5%) had been previously diagnosed with cancer and 37 (88.1%) declared that there was a genetic tendency to cancer on their side of the family. Among those, 35 (94.6%) had a presymptomatic cancer genetic test at 47.4±6.2 age of years: 18 (42.9%) after they had cancer and seven (16.7%) before they had cancer. Ten (23.8%) of those who had a presymptomatic test had never had cancer. The majority of participants were tested for hereditary breast and ovarian cancer (n=24, 68.6%), six (17.1%) for Lynch syndrome, four (11.4%) for Cowden syndrome, and one (2.9%) for FAP. All were found to have a mutation. Of the 35 tested, 19 (45.2%) were the first person in their family to have such a test.

Five (11.9%) participants affirmed that the genetic tendency to cancer was on their partner's side of the family.

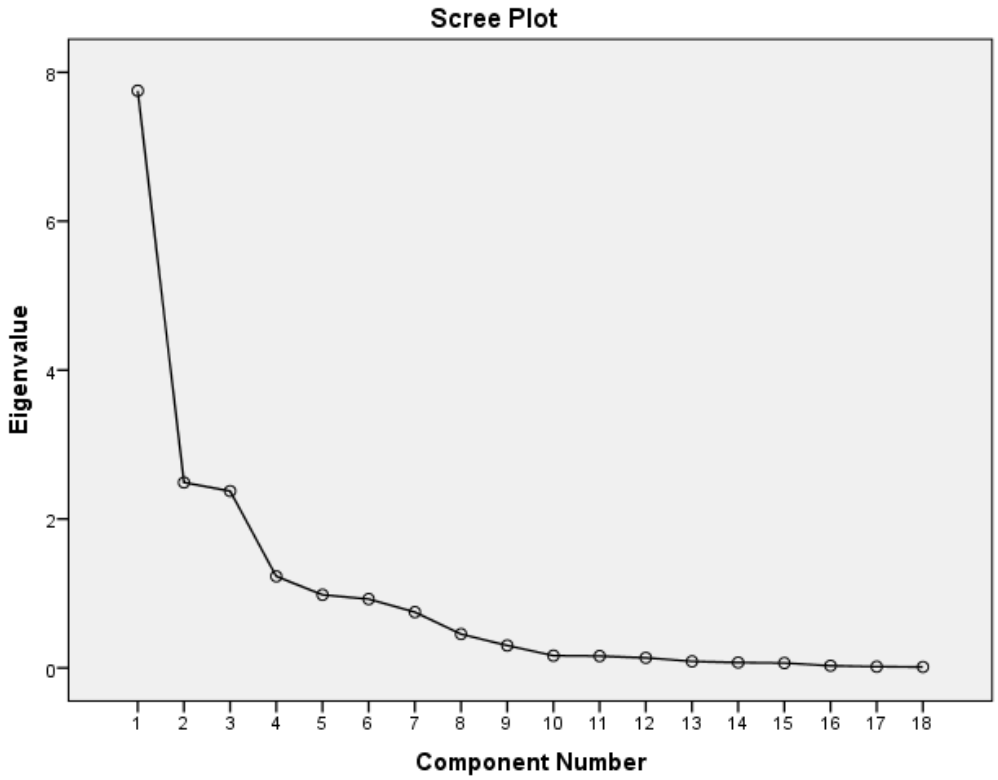
Telling your children

All participants reported that they had told their children about the family risk of cancer, but the age of the children when told ranged from 5-44 years (21.8±6.6). The majority of the parent participants (n=28, 66.7%) decided to disclose the information in a planned conversation with their child(ren), eight (19.0%) told them in a casual way, and six (14.3%) took advantage of a moment when the child raised the issue.

The following results relate to parents' reasons for telling their children about the family cancer risk (Q75). An exploratory factor analysis was performed on question 75 (see Table 5.5 for the KMO and the Bartlett's test).

Four factors with Eigenvalues over 1.00 emerged from a principal component analysis (Varimax rotation). These four factors accounted for 76.9% of the explained variance. Inspection of the scree plot for this data showed a distinct separation between the third and fourth factors, which suggested that three factors should be retained. The scree plot is presented in Figure 5.6. No other factor contributed greater than 5.6% of the variance.

FIGURE 5. 6 SCREE PLOT OF COMPONENT AND EIGENVALUE FOR QUESTION "*WHAT WERE YOUR REASONS FOR TELLING OR NOT TELLING YOUR CHILDREN ABOUT THE FAMILY CANCER RISK?*"



In order to further reduce the number of items selected for question 75, the data were subjected to another Varimax rotation with the specification that only three factors be extracted. An analysis of the factor rotations showed the three-factor rotation more accurately represented the proposed constructs than the four factor rotation. The first variable on the four factor rotation accounted for 43.1% of the unique variance, the second variable contributed 13.8% of the variance, and the third variable contributed 13.2% of the variance. Analysis of the structure matrix, which detailed the correlation between the variables and the factor, showed moderate to strong correlations. Any item with a correlation below 0.40 was discarded. The factor loadings for each item were presented in Table 5.17.

TABLE 5. 17 ROTATED COMPONENT MATRIX QUESTION "*WHAT WERE YOUR REASONS FOR TELLING OR NOT TELLING YOUR CHILDREN ABOUT THE FAMILY CANCER RISK?*"

	COMPONENT		
	1	2	3
I wanted to provide access to information for my children	**0.938**		
I wanted to make my children aware of the risk	**0.938**		
I wanted to share my genetic test results with my children so they could be tested	**0.841**		
I wanted to share my partners genetic test results with my children so they could be tested			**0.896**
I wanted to explain the family history of cancer	**0.933**		
I wanted to share my genetic test results with my children because of my grandchildren or future grandchildren	**0.809**		
I wanted to share my partners genetic test results with my children because of my grandchildren or future grandchildren			**0.882**
I felt it was the appropriate age to tell them	**0.852**		
I didn't intend to tell them but they accidentally found out			**0.618**
I wanted them to be able to have screening	**0.892**		
I thought my children were too young to know	**-0.740**		
I thought it might make my children anxious		**0.881**	
I thought it might increase my children's fear of getting cancer		**0.872**	
I thought it might increase my children's worry about my and my partner's health		**0.884**	
I thought it was unnecessary to make my children aware of the family history			**0.446**
I am still coping with the test results		**0.535**	
I was not ready to share the news	**-0.516**	0.456	0.408
There was no medical reason to tell them	**-0.627**		

Lastly, I calculated the percentages of the item responses to identify areas with different reasons for wanting to be tested (Table 5.18).

TABLE 5. 18 PERCENTAGES OF EACH ITEM DIVIDED BY FACTORS INDIVIDUALISED QUESTION "*WHAT WERE YOUR REASONS FOR TELLING OR NOT TELLING YOUR CHILDREN ABOUT THE FAMILY CANCER RISK?*"

	STRONGLY OR SOMEWHAT DISAGREE	NEITHER AGREE NOR DISAGREE	STRONGLY OR SOMEWHAT AGREE
Factor 1: Making children aware			
I wanted to provide access to information for my children	4 (10.0%)	2 (5.0%)	34 (85.0%)
I wanted to make my children aware of the risk	4 (10.0%)	2 (5.0%)	34 (85.0%)
I wanted to share my genetic test results with my children so they could be tested	4 (10.3%)	1 (2.6%)	36 (90.0%)
I wanted to explain the family history of cancer	4 (10.3%)	3 (7.7%)	32 (80.0%)
I wanted to share my genetic test results with my children because of my grandchildren or future grandchildren	4 (10.0%)	6 (15.0%)	27 (67.5%)
I felt it was the appropriate age to tell them	5 (12.8%)	4 (10.3%)	30 (76.9%)
I wanted them to be able to have screening	4 (10.5%)	1 (2.6%)	32 (88.9%)
I thought my children were too young to know	22 (61.1%)	1 (2.8%)	3 (8.3%)
I was not ready to share the news	26 (70.3%)	2 (5.4%)	4 (10.8%)
There was no medical reason to tell them	28 (77.8%)	1 (2.8%)	1 (2.8%)
Factor 2: Worry about emotional impact on children			
I thought it might make my children anxious	16 (43.2%)	7 (18.9%)	10 (27.8%)
I thought it might increase my children's fear of getting cancer	19 (51.4%)	3 (8.1%)	11 (29.7%)
I thought it might increase my children's worry about my and my partner's health	16 (43.2%)	7 (18.9%)	10 (27.0%)
I am still coping with the test results	21 (56.8%)	3 (8.1%)	8 (21.6%)
Factor 3: Difficulties in communicating own genetic status			
I wanted to share my partner's genetic test results with my children so they could be tested	2 (5.6%)	1 (2.8%)	9 (22.5%)

I wanted to share my partner's genetic test results with my children because of my grandchildren or future grandchildren	2 (5.6%)	1 (2.8%)	9(25.0%)
I didn't intend to tell them but they accidentally found out	19 (52.8%)	1 (2.8%)	1 (2.8%)
I thought it was unnecessary to make my children aware of the family history	27 (73.0%)	1 (2.7%)	4 (10.8%)

No significant differences were found between PEQ and PIQ in relation to Factor 1 ($t(27)=-0.242$; $p=0.811$), Factor 2 ($t(27)=0.237$; $p=0.815$), and Factor 3 ($t(27)=-0.792$, $p=0.435$).

A significant difference was found in relation to making children aware (Factor 1): married participants were more likely to make their children aware than divorced participants (3.5±0.5 *versus* 3.2±0.9; $F=4.838$, $p=0.008$) but no differences were found when worry about emotional impact on the child (Factor 2) was compared against most parameters. However, it was observed that participants who underwent presymptomatic genetic testing after having cancer were more likely to worry about the emotional impact on the child than those who underwent it before having cancer (2.3±1.1 *versus* 0.8±0.8; $F=2.944$, $p=0.050$).

In relation to difficulties in communicating own genetic status (Factor 3): participants who mentioned the family cancer risk in a casual way to their children were less likely to have difficulties in communicating genetic status than those who planned a conversation with them (2.3±1.7 *versus* 1.0±1.1; $F=4.164$, $p=0.025$). Consistently, a significant difference was also found between participants with the genetic tendency to cancer in their partner's

side of the family and participants with the genetic tendency in their family: the first group were less likely to have difficulties in communicating genetic status (2.4±1.1 *versus* 0.7±0.9; t(23)=3.952, p=0.001).

In summary, the item "*What were your reasons for telling or not telling your children about the family cancer risk?"* was analysed by means of factor analysis. The three factors revealed were:

- making children aware
- worry about children's emotional impact
- difficulties in communicating own genetic status.

Children's experience of the genetic test

The majority of children of the parent participants were told about the choice to have a genetic test (94.7%). In only one case (2.6%) the participant declared he had not told all his children because one of them had autism.

Parents reported that the request for genetic testing was made by the adult children themselves in 28 cases (73.7%), by the children together with one or both the parents in five cases (13.2%), by the respondent or his partner in four cases (10.5%), and by the doctor in one case (2.6%).

Parents' feelings about genetic testing for their children

In the last part of the questionnaire, parents were asked about their experiences regarding testing for their children. All participants felt their children should be tested. The majority (n=27, 75.0%) felt guilty about the

possibility that the mutation might be inherited by their children: those who did feel guilty were older (mean age 57.3±8.8 *versus* 50.6±5.5; t(34)=2.720; p=0.010) than those who did not. The majority of participants (n=26, 74.3%) also felt they had control over the decision their child made about the test.

5.3.4 SUMMARY OF FACTOR ANALYSIS

In this section, I will present a summary of the factors that emerged from the analysis. Table 5.19 shows factors for each of the questions analysed.

TABLE 5. 19 FACTORS FOR EACH QUESTION ANALYSED

How did you react after you became aware of the family genetic condition?
FACTOR 1: Awareness
FACTOR 2: Need for information
How did you feel about the genetic counselling?
FACTOR 1: Satisfaction about genetic counselling
What were your reasons for wanting to be tested?
FACTOR 1: Proactivity
FACTOR 2: Parents' pressure against testing
FACTOR 3: Parents' decision to be tested
FACTOR 4: Concern for children
FACTOR 5: Parents' pressure for testing
How did you feel after receiving your genetic test result?
FACTOR 1: Negative feelings
FACTOR 2: Negative impact on relationships

FACTOR 3: Uncertainties about the meaning of test result

FACTOR 4: Worry for relatives

FACTOR 5: Perceiving the test result as helpful

How did you feel living with your genetic risk?

FACTOR 1: Influence on lifestage perception

FACTOR 2: Impact of test result on own prevention and on relatives

FACTOR 3: Anxiety

FACTOR 4: Protection of self and children

What were your reasons for telling or not telling your children about the family cancer risk?"

FACTOR 1: Making children aware

FACTOR 2: Worry about children's emotional impact

FACTOR 3: Difficulties in communicating own genetic status

5.4 DISCUSSION

In this section, I will discuss the findings and, where applicable, I will use pre-existing literature to help interpret the findings. Strengths and limitations will be also discussed.

The survey findings suggest that approximately one-third of young adults were told about the potential genetic risk by their mothers, and they received this information the first time when aged between 5-30 years in an unplanned conversation. This is in line with findings by Rew et al. (2009) and Bradbury et al. (2007), who showed that the majority of children of BRCA mutation carriers learnt of their potential genetic risk of cancer many

years before preventive interventions were recommended. Tercyak et al. (2002) evaluated the likelihood and the effect of parent-child factors on communicating about maternal genetic test results for breast and ovarian cancer risk and also found that children at risk of BRCA mutations were told at mean age of 13.5±2.6 (Tercyak et al., 2002). Nevertheless, it seems that understanding the risks for themselves occurs around 12-15 years of age, while the awareness of the reproductive implications occurs many years later (15-17 years of age) (Metcalfe et al., 2011). However, in the research conducted by Metcalfe et al. (2011) it was pointed out that less formal discussion about their potential genetic risk was preferred by children and young people. In more detail, they expressed a preference for the situations in which parents were also doing other activities such as preparing a meal, gardening or riding in the car (Metcalfe et al., 2011).

The communication of genetic information is often a difficult issue. Although parents felt an obligation to pass on their genetic information to their children and other family members as well, they did not wish to cause anxiety or alarm (Green et al., 1997; D'Agincourt-Canning, 2001). It has been reported that geographical distances, family rifts, relational ruptures, adoption, generational gaps or complex family relations are factors that might make it more difficult to transfer information to children or other family members (Borry et al., 2009). Borry et al. (2009) also reported that parents were not able to transmit accurate information to their children regarding their genetic risk. Parents may fail to understand the meaning of their genetic test result and to share appropriate information. However, the data from the Phase 3 study suggest that most parents make the decision to

disclose on their own, and do not frequently involve genetic counsellors. These findings could be the result of the fact that there are no clear guidelines for parents or genetic counsellors as to the best ways for parents to discuss their genetic test results with their children, making it difficult for genetic counsellors to make recommendations to parents. Damage to the child's self-esteem, distortion of the family's perception of the child, loss of future adult autonomy and confidentiality, discrimination against the child in education, employment, or insurance, and adverse effects on the child's capacity to simply be a child are harms regarding parents (Clarke, 1994). It is possible that parents are concerned that some of these harms may also apply through telling their children of their risk. It is also possible that parents have not perceived the existence of support from genetic counsellors, even though Metcalfe et al. (2008) showed that health professionals are increasingly being asked for advice from parents about risk disclosure to their children.

Additionally, these data showed that participants who received the information at a pre-planned time were more likely to need more information. These findings could be the result of the fact that the majority of those who received the information at a pre-planned time were told by a physician or genetic counsellor, it may be that they understood the importance of the information shared and as a result they needed more information.

Young adult participants were satisfied with the genetic counselling they received, a finding consistent with that of MacLeod et al. (2014), who analysed the experiences of young people who had had presymptomatic

testing. However in the systematic review reported in Chapter Three of this doctoral dissertation it came to light that the experience of genetic counselling on the one hand was reported as an opportunity for discussing problems, and on the other hand it was been associated with feelings of disempowerment. Consistent with Gong et al.'s findings (2016) which assessed how testing mutation-positive for Huntington's disease influences young adults' life, genetic counselling on the one hand was appreciated, but on the other hand it was also reported as a hurdle. Moreover, Mand et al. (2013) showed that young adults expressed some negative feelings associated with genetic counsellors, such as the perception of not being understood and the feeling that the counsellor was the person with the power over the testing decision. There was no indication of this in the Phase 3 study results.

Although theoretically an autonomous choice to undergo presymptomatic testing is a fundamental requirement of the process of genetic counselling, my results showed that participants who underwent genetic testing between 18-25 years of age were more likely to do it as a result of their parent's decision. This was corroborated by Hamilton et al. (2009), who found that participants in their late 20's and 30's were more likely than younger ones to decide autonomously to have genetic testing. It has been reported that formal genetic testing should generally wait until the children request it as autonomous adults (Clarke, 1994). This respect for autonomy and confidentiality would involve the postponement of testing until the person is both adult and is able to understand not only the genetic

test result, but also the emotional and social consequences of it (Clarke, 1994).

Almost all of the participants were mutation-positive. This could be the result of the way young adults were recruited. Apart from the recruitment undertaken in the genetic clinic, the web link was posted using group or hashtag on my specific topic. Although I used very general terms (such as breast cancer, BRCA, genetic test), it may be that potential participants who received negative test results were no longer sufficiently interested in the topic to respond, or perceived that the topic was not relevant to them.

Participants who received the information at an unplanned time were more likely to feel negative feelings, as well as participants who received a positive test result. In contrast, in my previous systematic review (Chapter Two) it was observed that positive and negative emotional outcomes were not correlated with test results. In addition, although it is reasonable to hypothesize that undergoing testing at the right time reduces the risk of negative effects of the genetic test result, my results suggested that other parameters must be taken into account as well such as marital status and pressure from parents. While none of the young adults described by Mand et al. (2013) reported a catastrophic emotional response to their test result, conflicting emotions of relief, happiness, guilt, fear and anger have been reported (Macrae et al., 2013).

Overall, it seems clear from my findings that those young adults who were told of the risks by their aunts reacted differently to those in other groups. However, this finding is not generalizable because the sample size

(aunts group) is far too small. Nevertheless, Wisnieski et al. (2015) highlighted that a majority of young adults noted that they had close relationship with aunts, uncle and/or grandparents during their adolescent years. For example, in their study young adults named aunts and grandmothers as family members with whom they were comfortable speaking about romantic relationships. The impact of other family members appears to receive little attention in the literature as underlined by Tingvold et al. (2012). It is important to remember that parents' and other adult family members' discussions with young people have the potential to influence young people's decision and behaviours. In families affected by a genetic condition, the dynamics may be affected, for example, by the early death of one parent. In these cases, other family members may step into a more parental role. Although the group informed of their risk by aunts was small, the findings related to that group could be important and to the best of my knowledge, there is a gap in research regarding the influence of other adult family members on test decisions.

All parent participants reported that they had told their children about their family risk of cancer when their children were aged between 5 years and 44 years. These results concur with reports by young adult participants that they were told between 5 and 30 years of age. As previously discussed, it has been reported that children became aware about their potential genetic risk of cancer many years before preventative interventions were recommended (Rew et al., 2009; Tercyak et al., 2002; Bradbury et al., 2007). Usually, minors are considered unable to fully understand the implications of genetic testing until the development of their ability to conduct abstract

thought at around 11-14 years (Wertz et al., 1994). However, Metcalfe et al. (2008) believed that information about their potential genetic risk is needed by children before a specific life event such as developing their first sexual relationship. Parents were reported to feel a strong sense of responsibility to discuss this information with an open style of communication and it was suggested that this kind of communication empowered the family and increased the family support and care for one another. Instead, where the communication was more closed, children felt upset and frustrated with family secrecy. Therefore open communication of genetic risk information by parents to their children has been suggested to benefit both informed reproductive decision making and family cohesion (Metcalfe et al., 2008).

It has been reported that parents face a difficult job in deciding when, how, and what to tell their children (Metcalfe et al., 2008). The majority of parent participants disclosed the information in a planned conversation, however the majority of young adults reported that discussions were not usually planned. This was consistent with the findings of Metcalfe et al. (2011) who had assessed how genetic risk information was shared between family members and the factors affecting it. In my study, due to anonymity of participants, I was not able to determine if participants (both young adults and parents) belonged to the same families. In addition, it may be that a conversation that was planned by parents may have appeared unplanned to their children.

My findings showed also that participants who mentioned the family cancer risk in a casual way to their children were less likely to have difficulties in communicating genetic status than those who planned a

conversation with them. This may be due to the fact that they are less anxious about disclosure and therefore able to take an opportunity to discuss the familial risk with their child when an opportunity arose. Consistently, a significant difference was also found between participants with the genetic tendency to cancer on their partner's side of the family and participants with the genetic tendency in their family: the first group were less likely to have difficulties in communicating genetic status. Reasons for this might be both the difficulties of the parent dealing with his or her own diagnosis of cancer and guilt in parents for passing the family mutation onto their children, as described by Quinn et al. (2009). Additionally, married participants were more likely to make their children aware of the risk than divorced participants. This is in line with current literature where many studies have shown that a parental divorce has a negative effect on parent-child relations, specifically that divorced parents, especially fathers, have less frequent contact with their children than married parents (Daatland, 2007; Albertini and Garriga, 2011). Data suggest limited involvement of genetic counsellors in parental decisions to disclose results to young family members. The involvement of specialist health professionals could be helpful, as some offspring may not fully understand the information shared and there is also a potential for initial adverse reactions among offspring in response to parent disclosures, as highlighted by Bradbury et al. (2007a), who assessed the parental communication of BRCA results to children under 25 years of age.

Although the majority of the requests for genetic testing were made by young adult offspring, the majority of parent participants felt they had control over the decision their child made about the test and all felt their

children should be tested. This concurs with the findings of the systematic review (Chapter Three), where parents appeared to have exerted pressure on their children during the decision making process about testing. The findings of the review showed that as a consequence of parental pressure, young adults reported feelings of disempowerment and lack of control and declared that they underwent genetic testing because of pressure from family members or "for" a parent. These issues raise the ethical problem of how health professionals can respect young adults' developing autonomy (Duncan et al., 2007; Duncan et al., 2008; Duncan et al., 2010; Hamilton et al., 2009; Werner-Lin et al., 2012). This issue has been addressed by Duncan et al (2007) who explored the experience of presymptomatic genetic testing for Huntington disease from young persons' perspectives and documented the impact that testing has upon various aspects of young peoples' lives. Further work was done by Duncan et al. (2008) to evaluate the potential effects associated with presymptomatic genetic tests in young people, and by Duncan et al. (2010), who assessed some of the key ethical challenges associated with presymptomatic genetic testing for familial adenomatous polyposis in young people. On the other hand, related to health professionals' point of view, Werner-Lin et al. (2015) investigated genetic counsellors' perspectives on counselling clients aged between 18 and 25 years, using an online survey. They found that genetic counsellors reported that they adapted their genetic counselling styles with the age of the consultand, due to experiencing some differences according to whether the consultand was 18-25 years old or older. A primary challenge reported was navigating family dynamics in counselling sessions. However, it has been shown by my findings that young adult participants who were strongly influenced by their parents

to be tested were less likely to feel anxious. This result may confirm that young adults did not completely understand the implications of the genetic test but complied because of parental pressure. However, it could be suggested that genetic counsellors have a responsibility to enable young people to challenge decisions made by their parents that may be inappropriate for them (American Society of Human Genetics Board of Directors and American College of Medical Genetics Board of Directors, 1995). It may be that parents do not always make the best possible decision for their offspring, but usually one that is intended to support them. In the context of presymptomatic genetic testing, where there is uncertainty about the potential harm and/or benefits, Cohen believes that the parent's decision should prevail over their offspring's decision (Cohen, 1998). However, with regard to the principle of decision-making by a surrogate, in this case parents, Buchanan and Brock (1990) provided data on the fact that there may be a failure by parents to make a decision in the best interests of their children.

Acceptance of the parental decision could be a reason why young adults did not completely understand the implications of testing. One reasonable hypothesis would be that those who are very young when informed are not fully able to understand the test and its implications. However this would potentially conflict with the idea that children should be told of their risk. Although the involvement of health professionals in parents' decision to communicate genetic risk to young family members was found to be limited in the systematic review performed (Phase 1), health professionals could have a role in supporting both parents and young adults. However, reluctance by parents to involve health professionals may be

partly due to the parents' wish to undertake this task alone (Gaff et al., 2006; Metcalfe et al., 2008).

The evidence of this study highlights the need for a comprehensive, longitudinal counselling process with appropriate timing and setting, which supports 'parent-to-offspring' risk communication first and young people's decision making about presymptomatic testing and risk management afterwards. This would include emphasising that disclosure of genetic risk is a gradual and dynamic process in the family, and where children are told at an early age, this should be followed with further age-appropriate information.

Strengths and limitations of this phase of the study

The advantages and disadvantages of using an online questionnaire as a data collection method were discussed in Section 4.3.5. Further to this, the strengths and limitations of this phase are discussed in the next Chapter (see Section 6.7 for more details).

The limited number of mutation-negative participants reduced the possibility of observing any significant differences between them and those who were mutation-positive. This could be the result of conducting the recruitment by posting recruitment messages to social network webpages or groups on specific cancer genetic conditions, as I have described in Chapter Two, Section 2.5.4.1. Those who are mutation-negative may not use these sites. Another possible reason that I considered relates to the fact that

individuals are less interested in this topic once they are aware they are mutation-negative regarding the genetic condition in the family.

The limited number of PIQ reduced the possibility of observing any significant differences between groups. This could be the result of difficulties in recruiting mainly via clinics, because, as I have described in Chapter Four, Section 4.3.5, the majority of the Italian population do not use the Internet regularly. When the recruitment route is analysed this is clear: only 39.3% of PIQ had accessed the questionnaire via the Internet, compared to 100% of PEQ. I have wondered about these results, considering that approximately 70% of young adults in Italy are thought to use the Internet regularly, as I have shown in Chapter Two, Section 2.5.4, of this dissertation. One possible reason that I considered relates to the fact that there are many specific Facebook groups in the English speaking world regarding hereditary cancer syndromes, while there are only two Italian ones (one with approximately 20 participants about Lynch syndrome and the other one with approximately 400 participants about BRCA genes). Because the administrator of the biggest group demanded to become one of the authors of the research publications resulting from this doctoral study, I was not able to post the recruitment messages emphasising my research and therefore to recruit potential participants from this group. This could be one of the reasons limiting the number of Italian questionnaire respondents through the Internet. However, I have also found this difference in terms of number of groups and/or pages interested in these themes between the English speaking world and the Italian one on Twitter. Another reason limiting the number of Italian questionnaire respondents could be less interest in the

Italian population regarding sharing information on medical issues via the Internet.

Furthermore, the possibility of generalizing the results of factor analysis could be hampered by the small sample size, particularly for the parent questionnaire. The minimum sample size required is widely discussed in the literature: many papers focused on the importance of a definite minimum sample size. A minimum sample size of 200 was recommended by Guilford (1954), 50 is the number suggested both by Comrey and Lee (1973) and Gorsuch (1974). Other authors focused on the number of cases per variable and recommendations range from 3:1-6:1 (Cattell, 1978) to 20:1 (Hair et al., 1979). Consistently, researchers suggested obtaining the highest number of cases per variable possible in order to minimise the chance of overfitting the data (de Winter et al., 2009). In my young adult questionnaire, where the highest number of items is 21, I recruited 132 cases. This means that I obtained six cases per variable. On the contrary, in my parent questionnaire I had 18 items analysed with factor analysis. This means, considering what Cattell (1978) suggested, I would have needed to recruit 54 cases, while I obtained 42 cases. Although 42 cases are a number correct to perform factor analysis according to Kaiser and Barlett, the ability to generalise the findings of the parent survey could be questioned.

5.5 IN SUMMARY

In this chapter I have presented the results of the Phase 3 study. Finding out about their risk (young adults' and parents' points of view),

decision making process (young adults' and parents' points of view), genetic test result (young adults' point of view) and living with genetic risk (young adults' point of view) were the four themes that emerged.

In the next chapter, I will summarise and discuss the findings from all the three phases of my research project and discuss how they relate to each other. In particular, I will present a model based on the synthesis of findings of all three phases.

CHAPTER SIX

DISCUSSION

6.1 INTRODUCTION

In this final chapter, I will present the specific objectives revisited, and an updating of the literature. An overarching theoretical framework that draws together the findings from all three phases of this doctoral project will be described. I will discuss the relevance of the doctoral study findings within the context of genetic services and professional working in genetics, and provide a reflective assessment of the doctoral study process. Finally, I will make recommendations for practice and future research.

6.2 THE SPECIFIC OBJECTIVES REVISITED

As stated at the beginning of Chapter One, the aims of this doctoral study were to explore the implications of presymptomatic testing for hereditary cancer in consultands aged 18-30 years. To achieve this, the specific objectives were:

- to explore how young adults interpret cancer presymptomatic testing

- to explore the basis for young adults' decisions to undergo testing or not
- to explore the influence that parents have in the choice, with reference to the family dynamics and lifestage theory
- to analyse the psychosocial impact of test disclosure, according to mutation status
- to develop a theoretical model regarding the decision making process in young adults considering pre-symptomatic testing for hereditary cancer
- to inform the process of cancer genetic counselling for young consultands.

Having re-stated the objectives, I will now discuss each one in turn, in relation to the major findings identified during the course of this doctoral study, and the extent to which each objective was achieved.

6.2.1 HOW YOUNG ADULTS INTERPRET CANCER PRESYMPTOMATIC TESTING

In the previous chapters, the implications of presymptomatic testing for hereditary cancer in young adults, and the nature of a genetic testing have been discussed. I will now attempt to look at the evidence from the systematic review of the literature performed (Phase 1) and from empirical data I collected (Phase 2 and Phase 3) to answer this question. From the systematic review of the literature I found that many young adults were described as having grown up without information or with misinformation concerning their potential genetic risk (Phase 1): these findings were confirmed during the qualitative study (Phase 2). Moreover, four of the

participants described genetic testing as '*just a blood test*'. However, after genetic counselling young adults' knowledge became more accurate and they perceived genetic testing as involving a need to wait for a result and they better understood what they were doing and the importance of undergoing genetic testing.

6.2.2 THE BASIS FOR YOUNG ADULTS' DECISIONS TO UNDERGO TESTING OR NOT

Data from all the three phases of this doctoral project enabled me to assess the basis for young adults' decision to be tested or not. Parental pressure was an influence, and this is discussed in the next section. Other influences were found. In the qualitative study (Phase 2) it came to light that a young woman, Barbara, had found different influences within the family and she had wondered whether it was better to listen to her brother, who was a doctor, or her mother. In more detail, during the interview she said:

> *"My brother is a doctor and drives me to undergo the test because he says: 'it is the right thing'. But is it really the right thing? When I told my mum what I wanted to do she said: 'Barbara, are you sure? You can't turn back. You will have to deal with the result. Are you sure you don't want to wait few years?' "*

In this case, the different influences within the family have been characterized both by brother and his profession, which she had decided to emphasize during her interviews. It seems clear from my quantitative results that young adult participants were influenced by their aunts, reacting differently to those than to other relative groups. However these results regarding the doctor brother and the aunts are not generalizable because the sample size is far too small. Further investigation of how the young adults'

close relationship with distant relatives (such as aunts, uncles, grandparents) influence their decision making process could be explored. For example, Wisnieski et al. (2015) pointed out that a majority of young adults had a close relationship with aunts, uncle and/or grandparents during their adolescent years.: young adults named aunts and grandmothers as family members with whom they were comfortable speaking about romantic relationships. However, the impact of distant relatives appears to receive little attention in the literature as also underlined by Tingvold et al. (2012).

Nevertheless, parents' and other adult family members' discussions with young people have the potential to influence young people's decision and behaviours. In families affected by a genetic condition, the dynamics may be affected, for example, by the early death of one parent. In these cases, other family members may step into a more parental role (Tingvold et al., 2012).

6.2.3 THE INFLUENCE THAT PARENTS HAVE IN THE CHOICE, WITH REFERENCE TO THE FAMILY DYNAMICS AND LIFESTAGE THEORY

The debate about the nature of the autonomous decision making process was introduced in Chapter Three. In the context of this doctoral study, I consider it relevant to discuss this issue. As a result of my doctoral study, it was clear that not only the influence of parents must be taken into account, but of other people (such as relatives and persons outside the family) as well. The current generation of young adults have higher levels of student debt and are more likely to experience poverty, unemployment and the 53% of emerging adults aged 18-24 years currently lived with parents

(Pew Research Center, 2012, 2013). Living independently is one of the key developmental tasks of emerging adulthood (Shanahan, 2000). It is reasonable to hypothesise that this style of life has an impact on developmental tasks reducing the autonomy of young adults in their decision making process. Because many young adults are living with their parents, this could slow down the process of achieving autonomy as an adult. I therefore bore this in mind when interpreting the results of my doctoral study, as stated in the previous section.

Revisiting the findings through all the three phases, the influence that parents (or other people) had on testing choices was evident in different scenarios. First of all, it emerged that young adults aged between 18-25 years were more influenced by parents than those aged 26-30 years (Phase 3). Moreover, comparing parent and young adult participants, the quantitative study revealed that parent participants felt they had control over the decision their children made about the genetic test, while the majority of the young adult participants declared that the request for the genetic test was based on their own decision. In addition, in the systematic review, it was evident that in some cases, parents appeared to have exerted pressure on their young children to pursue risk reduction surgery, but this was not indicated by my doctoral study, although I did not focus specifically on this issue.

6.2.4 THE PSYCHOSOCIAL IMPACT OF TEST DISCLOSURE, ACCORDING TO MUTATION STATUS

In order to explore the psychological impact of genetic test disclosure, according to mutation status, it was important to synthesise findings from all three phases. It was clear from the qualitative study (Phase 2) that before testing, although participants perceived the information provided by a genetic test as useful in helping to plan their lives, they did not think they would change behaviour based upon the possible result. However, the majority could not predict how they would react to the result. Nevertheless, neither mutation-positive nor mutation-negative participants reported changes in their behaviour after the result..

Understanding if the emotional outcomes of personal test result communication were correlated with the test result was an important point. None of the young adult participants in the qualitative study (Phase 2) reported a catastrophic emotional response to their test result: emotions of relief, happiness and fear were generally reported. In addition, data from the systematic review of the literature (Phase 1) showed that positive and negative sensation outcomes of young adults' test result communication were not directly correlated with a test result: in both scenarios young adults thought that the most important thing was to have a result.

However, negative feelings, negative impact on relationships, uncertainties about the meaning of the test result, worry for relatives and perceiving the test as helpful were the five factors that emerged concerning the genetic test result from the quantitative study (Phase 3). In more detail, negative feelings were reported both by participants who had a mutation and

those who received the information at an unplanned time, while worry for relatives was reported by participants who had a mutation.

Another important point on the psychosocial impact of genetic test disclosure concerned the relationship between young adults and their relatives in relation to taking measures for cancer prevention, although a favourable impact of the test result on their own prevention and on relatives was more likely in participants tested between 26-30 years of age (Phase 3). It emerged from the qualitative study (Phase 2) that once young adults became aware of their genetic test result, they recommended that their relatives (e.g. brothers and sisters) undergo genetic testing as well. This could be seen to be evidence that they really understood what they had done after their presymptomatic genetic testing experience and realised how to integrate the test result into their everyday life for themselves and their relatives.

6.2.5 THEORETICAL MODEL REGARDING THE DECISION MAKING PROCESS IN YOUNG ADULTS CONSIDERING PRESYMPTOMATIC TESTING FOR HEREDITARY CANCER

By means of a progressive process throughout all three phases, the theoretical model regarding the decision making process in young adults considering presymptomatic testing for hereditary cancer was built. The theoretical model summarised the interlinking relationships of all the phases of this doctoral study. The central aspect of this model was the decision making process. Supporting clients to make appropriate decisions is one of the roles of a genetic counsellor (Evans, 2006; Uhlmann et al., 2009; Harper, 2010) as was discussed in Chapter One. As came to light in my doctoral

research, participants arrived for genetic counselling with the decision making process already made. Because participants arrived for genetic counselling with their own clear decision in their minds, my model has challenged the accepted version of the process. The outcome of this theoretical model then contributes to adjustment theories and subsequently contributes to the area of expertise of health professionals. I will present my full theoretical model in more detail in the Section 6.4 of this Chapter Six.

6.2.6 THE PROCESS OF CANCER GENETIC COUNSELLING FOR YOUNG CONSULTANDS

Analysing the process of cancer genetic counselling for young adults, it is important to keep in mind the process of genetic counselling. While considering that each consultation is unique, it is important to define some common themes in the genetic counselling. The counselling starts with an exploration of the nature of the request and motivation for it, followed by building up a family tree, exploring what is already understood about the disorder before giving any information, and discussing and exploring awareness of the possible consequences of having personal genetic information (Evans, 2006; Uhlmann et al., 2009; Harper, 2010). Based on this, one of the roles of the genetic counsellor is to help the consultand to visualize their possible future reactions according to their mutation status (Evans, 2006; Harper, 2010).

To address the research question, "What is the process of cancer genetic counselling for young adults?" all the three doctoral study phases play an important role. First of all, through the qualitative study it is apparent that although most young adult participants had no expectations of

genetic counselling, they reported their motivations to undergo the genetic counselling as curiosity, to use it as a source of information, to obtain certainty, to prevent cancer and to respond to parental influence (Phase 2).

Personal experience of genetic counselling revealed positive feelings about genetic counselling in terms of being understood and clarifying the meaning of the test (Phase 2): that the experience was satisfactory was further confirmed by young adults who answered the online questionnaire (Phase 3).

However, despite coming with an intention to be tested, some participants became less sure of their decision as a result of genetic counselling and they expressed the desire to have an opinion to guide them during the genetic counselling process. Data from young adults analysed during the systematic review of the literature (Phase 1) showed the experience of genetic counselling as an opportunity for discussing problems, but it has also been associated with feelings of disempowerment. Disempowerment was not reported by participants in Phases 2 and 3.

6.3 AN UPDATE ON THE LITERATURE

In order to understand what has been published after the systematic review that I presented in Chapter Three, I have updated the search, using the same keywords on the eight databases that I previously searched. The search of eight databases produced 26 potential papers published between January 2015 and October 2016. Eleven were duplicates, leaving 15 for

examination. Four of these papers were focused on participants who had been diagnosed with cancer (Mork et al., 2015; Brehar et al., 2016; Porter and Fischer, 2016; Weber et al., 2016), three were not on the topic of my research (Curtin et al., 2015; Woltsche et al., 2015; Kothari et al., 2016), three were not focussed on the testing decision (Mand et al., 2015; Gong et al., 2016; Hamilton et al., 2016), two were based on the parental point of view (Lowe et al., 2015; Hayes et al., 2016), and one on the genetic counsellors' point of view (Werner-Lin et al., 2015). Therefore, following review of the titles and abstracts, two papers were found as potentially relevant: one of those was the systematic review I published based on this doctoral dissertation.

A qualitative study performed by Forrest Keenan et al. (2015) was the only relevant study (apart from my own) I could identify that was published after the previous systematic review (Chapter 3). The aim of this qualitative study was to assess young people's experiences of predictive testing for Huntington disease. Twelve young women aged 17-26 years were recruited and pre- and post-test interviews were conducted with them.

Before commenting on that paper, I will refer back to the findings of the initial review. The issues emerging from the studies included family and partner relationships, plans for the future, emotional state and the general approach to life. Five key themes were identified:

- the period prior to testing
- the experience of genetic counselling
- involvement of parents in decision making
- impact of personal test result communication
- living with genetic risk.

I will now describe the findings reported by Forrest Keenan et al. (2015) based on the structure of the findings of the systematic review.

The period prior to testing

During the period prior to testing the majority of participants reported having grown up with awareness about the genetic disease running in their family (Forrest Keenan et al., 2015). By contrast, the systematic review found out that young adults grow up without awareness and with no information about the genetic disease or the inheritance mode (Duncan et al., 2007; Patenaude et al., 2013; Hoskins et al., 2014). In the paper by Forrest Keenan et al. (2015) it was not possible to ascertain if young adults had knowledge about the inheritance mode and the appropriate age for testing.

Authors did not report in detail how and when the first communication about genetic risk was undertaken. The majority of participants were told by their parents when they were adolescent, while two were told while they were still children (Forrest Keenan et al., 2015). By contrast, findings from the systematic review indicated that none of the young adult participants in the other studies was younger than 12 years of age when informed (Godino et al., 2016).

Once aware of the family genetic disorder, some young adult participants who did not understand what it really meant sought information online according to findings showed by Patenaude et al. (2013), while those who were more conscious of own risk (or potential risk) arranged the first

counselling session to have their blood test according to Hamilton et al. (2009). In one case reported by Forrest Keenan et al. (2015) parents were reported to have encouraged their young adult children to access genetic counselling. Those authors also showed that in another case, a young woman was referred for genetic counselling by a professional because of her anxiety. It is not clear in the study conducted by Forrest Keenan et al. (2015) if at the time when young adult participants were told of their risk, the implications for themselves seemed distant, or whether when they became young adults the fear of developing the adult-onset disease recurring in their family had increased.

When approaching the decision about testing, many young adults understood that having a presymptomatic test was a choice. For example, a young woman decided not to be tested but to postpone testing until later in her life (Forrest Keenan et al., 2015). Overall, this new data adds a new finding to the systematic review (Phase 1) and, more in general, from the findings from qualitative study (Phase 2) where none regretted the choice to undergo presymptomatic genetic testing.

One of the findings from my systematic review indicated that choosing to undergo genetic testing constituted a major life event. This finding was confirmed by Forrest Keenan et al. (2015).

The experience of genetic counselling

The experience of genetic counselling reported by Forrest Keenan et al. (2015) is very similar to that which emerged from the Phase 1 review. For the young adults studied by Forrest Keenan et al. (2015), undergoing

genetic counselling was reported to have helped discussion and acted as a source of information. However, other young adults in that study reported some negative feelings, such as the perception of not being understood and the feeling that the counsellor was the person with the power over the testing decision. Some participants were disappointed to hear that the counsellor believed they were not ready to deal with the psychological consequences of the genetic test and that they needed to take time to reach an autonomous decision. For example, one young woman said '*I just kept going, can you just test me, can you just test me? I don't want to do this. I go through enough bloody counselling*' (Forrest Keenan et al., 2015, p. 4). None focused their attention on the procedure, instead of the meaning of testing, for example with the fear of the needle overshadowing the purpose of the counselling as reported in the findings of the systematic review.

In the earlier review, a consequence of discussion during pre-test counselling was that most young adults had shared their test result with only close friends and family (Godino et al., 2016). However, young adult participants studied by Forrest Keenan et al. (2015) decided not to involve family in the testing process.

Involvement of parents in decision making

In the systematic review it was revealed that some parents were reported to exert pressure on their young adult children's choice whether to be tested or not (Duncan et al., 2007; Duncan et al., 2008; Duncan et al., 2010; Hamilton et al., 2009; Werner-Lin et al., 2012; MacLeod et al., 2014). However, parental involvement in the decision making process was different

based on the specific disease (MacLeod et al., 2014). It was indicated that parents in families with familial cardiomyopathy had a strong influence in favour of testing; in hereditary breast and ovarian cancer the decision was autonomous but usually congruent with the parents' point of view, while in Huntington disease the decision was autonomous and sometimes went against the parents' opinion (MacLeod et al., 2014; Forrest Keenan et al., 2015). Nevertheless, one of the young adult participants thinking about her choice used 'we' instead of 'I', she said *'we've always said we wouldn't get tested'* (Forrest Keenan et al., 2015, p. 5).

In addition, Hoskins et al. (2014), and Forrest Keenan et al. (2015) as well, reported that some young women underwent genetic testing because of a professional's suggestion.

Impact of personal test result communication

Forrest Keenan et al. (2015) did not focus their attention on how young adult participants experienced waiting for the test result. However, none of the participants reported a catastrophic emotional response to their test result, according to previous findings, but the conflicting emotions of relief, happiness, guilt, fear and anger were generally reported in both the earlier review and the paper by Forrest Keenan et al. (2015). In more detail, authors described the emotional impact of both gene-negative and gene-positive test results. From the systematic review it was revealed that positive and negative emotional outcomes were not correlated with test results: in any case interviewees thought that the best thing was to find out the test result with participants describing themselves as happy just to know

their genetic status or as willing to begin enjoying life and to make behavioural changes (Duncan et al., 2007; Mand et al., 2013). Forrest Keenan et al. (2015) pointed out that a positive result created some negative emotions such as shock and disbelief. A young woman said:

> "*Just know I don't know what to do … With the result I feel like I need to something quick, if that makes sense, because I know that I am going to develop it … But just know I don't really have the motivation to do anything … I don't really know how much time I do have.*" (Forrest Keenan et al., 2015, p. 5).

Some young adults whose data were analysed in the systematic review described their shock at finding out that they had not inherited the family mutation and other participants felt able to plan for the future after their negative test result (Godino et al., 2016). Also Forrest Keenan et al. (2015) described similar findings: a negative test result was associated with feeling of relief. Making a '*normal family life*' was a priority of one young woman who was mutation gene-negative (Forrest Keenan et al., 2015, p. 4).

Living with a genetic risk

The systematic review findings suggested that having time before surveillance was scheduled to begin gave young adults the opportunity to think about surveillance protocols or prophylactic surgery, while others expressed frustration at receiving inconsistent, inaccurate, ambiguous or incomplete recommendations by genetic counsellors or doctors (Godino et al., 2016). Forrest Keenan et al. (2015), who focused their study only on Huntington disease, found that although it was difficult to have received a positive test result, a young woman expressed her hope of the possible benefits of clinical trials and started to consider having a family. These

results are in accord with the systematic review findings that showed that none of the participants in the studies included regretted the choice to have children.

As a result of this updated search, it appears that the findings of Forrest Keenan et al. (2015) have added some new information on the issue. However, it is also clear that there is still very little published on the topic of genetic testing for young adults.

6.4 OVERARCHING FINDINGS OF THE DOCTORAL STUDY

I will now discuss the major findings identified during the course of this doctoral study. First of all I will compare the findings between the Phase 1, Phase 2 and Phase 3. I will then discuss the risk-disclosure behaviour of those who surround the young adults and what children have understood of genetic risk. Finally I will explore the genetic counsellor's role in this context and the young adults' decision making process in the presymptomatic genetic testing scenario and other related scenarios.

6.4.1 COMPARISON OF FINDINGS OF PHASE 1, PHASE 2, AND PHASE 3

Setting aside the choice to undergo presymptomatic genetic testing, the transition from childhood to adulthood is an important event in the human life. There are a number of life changing decisions that are made during this period of life including continuing their education, having a career or job, living independently, adopting a particular lifestyle, which may not be the same as their family, taking up new hobbies and entering into intimate relationships (Albritton and Bleyer, 2003; Stern et al., 2010). However,

during the presymptomatic genetic testing experience young adults are still faced with some, or all, of these decisions.

A number of themes were identified during each phase of this doctoral study that fed into the final overarching theory. In this chapter I have included an updated table of major findings (Table 6.1), indicating again the way findings have been enhanced with each successive phase of the research, using different methods. It can be seen that some of the details differ from those in earlier tables of findings; this is due to the exploratory sequential nature of mixed method design. During the systematic review five major themes were identified. These included the period before testing, the experience of genetic counselling, the parental involvement in the decision making process, the impact of test result communication, and living with genetic risk. In the next phase, as a result of analysis of the interviews with young adults, four themes emerged. These included knowledge, genetic counselling process, decision making for testing or not, and dealing with the result. Through the analysis of interviews conducted with young adults, a grounded theory emerged inductively from the data, which I named 'Finding yourself in front of the mirror'. This theory included the process of genetic counselling, decision making and living with genetic risk. All participants arrived for genetic counselling with the test decision already made and saw themselves as in front of the mirror during the genetic counselling. They took distance from themselves and they thought about themselves not in first, but in second person, especially when they spoke about sensitive situations. Furthermore, throughout this process, young adults developed a form of autonomy and realised how to integrate the test result in to their

everyday life. In the third and final phase of this doctoral project, I further corroborated the findings from the first two phases of the doctoral study. These findings supported the experiences reported by young adults during interviews.

TABLE 6. 1 DIRECT COMPARISON OF THE KEY FINDINGS FROM THE SYSTEMATIC REVIEW, THE QUALITATIVE AND THE QUALITATIVE PHASE

SYSTEMATIC REVIEW	QUALITATIVE PHASE	QUANTITATIVE PHASE
▪ Many young adults grew up without information or with misinformation concerning their potential genetic risk	▪ Participants grew-up with little or no information about their genetic risk ▪ After genetic counselling young adults' knowledge become more accurate, they became aware of the options for clinical screening, they perceived genetic testing as needing to wait for a result and they better understood what they were doing or the importance of undergoing genetic testing	▪ The majority of young adult participants (75.5%) were informed after their 18th birthday. The mean time that elapsed between receiving the information and when they were tested was 2.0±2.8 years (range 0-11) in those who became aware at an age when the test was available for them and 9.3±5.5 years (range 1-24) in the others. ▪ It was observed that participants tested for HBOC were more likely to be informed after their 18th birthday than those tested for Lynch syndrome. ▪ Participants who became aware of their potential genetic risk before their 18th

		birthday were more likely to be tested before 25 years of age than after 26 years of age.
▪ Communication of risk generally occurred due to the parent's initiative several years before testing or clinical actions could be undertaken	▪ Communication occurred due to the parents' initiative within one year before testing in 73.3% of participants	▪ Although the majority of young adult participants thought they had been informed in an unplanned conversation/ call, the majority of parent participants reported they had disclosed the information in a planned conversation with their child(ren) ▪ Participants were tested within one year of obtaining risk information in 47.4% cases.
▪ The choice to undergo genetic testing constituted a major life event ▪ Parents appeared to have exerted pressure on their young	▪ Some relatives were reported to exert pressure on their young family members during the process of decision making to undergo presymptomatic testing	▪ Although the majority of the requests for genetic testing were made by young adult offspring, the majority of parent participants felt they had control over the decision their child made about the test and

children during the choice to undergo presymptomatic testing	▪ The decision to have presymptomatic test for some of participants was autonomous, but often congruent with relatives' points of view. Sometimes, the decision was at odds with the parent's opinion	all felt their children should be tested ▪ Parents were more likely to influence the decision of those who were tested between 18-25 years of age than those tested at 26-30 years of age ▪ Participants without children were more likely to undergo genetic testing because of pressure from parents than participants who had children
▪ The experience of genetic counselling on the one hand has been reported as an opportunity for discussing problems, on the other hand it has been associated with feelings of disempowerment	▪ Curiosity, source of information, obtaining certainty, prevention of cancer and parental influence were main motivations to undergo genetic counselling ▪ Most participants had no expectations of genetic counselling	▪ The experience of genetic counselling was reported as satisfactory.

	• Personal experience of genetic counselling revealed positive feelings about genetic counselling in terms of being understood and clarifying the meaning of the test. Participants changed their mind as a result of it and they expressed the desire to have an opinion to guide them during the genetic counselling process • Some participants felt they had matured as a result of their testing experience.	
• Positive and negative emotional outcomes of personal test result communication were not directly correlated with test result: in both scenarios young adults thought that the best thing was	• Before testing, although participants perceived the information provided by a genetic test as useful in helping to plan their lives, they did not think they changed behaviour based upon the possible result. However, the majority	• Negative feelings, negative impact on relationships, uncertainties about the meaning of the test result, worry for relatives and perceiving the test as helpful were the five factors that emerged about

to find out the result anyway. A common concern was related to the knowledge they may have passed on the mutation to their children

- In some cases, parents appeared to have exerted pressure on their young children to pursue risk reduction surgery.

could not have predicted how they would react to the result.

- None of the participants reported a catastrophic emotional response to their test result: emotions of relief, happiness and fear were generally reported
- Neither mutation-positive nor mutation-negative participants reported changes in their behaviour. No one considered the choice to pursue risk reduction surgery
- Once they became aware of their genetic test result, young adults recommended that their relatives (e.g. brothers and sisters) undergo genetic testing as well.

the genetic test result

- Negative feelings were reported both by participants who had a mutation and those who received the information at an unplanned time
- Worry for relatives was reported by participants who had a mutation
- Favourable impact of test result on own prevention and on relatives was more likely in participants tested between 26-30 years of age.

6.4.2 RISK-DISCLOSURE BEHAVIOUR OF THOSE WHO SURROUND THE YOUNG ADULTS

By my findings, it was also revealed that young adults surrounded themselves with other people (such as parents, other family members, a person outside the family) who influenced them with their knowledge and awareness and the decision making process began when they received the information outside the genetic counselling. It is clear from the literature that the main sources of information of potential genetic risk for children are their parents (Metcalfe et al., 2008), more often the mothers (Metcalfe et al., 2008, 2011). Parents in this context could be defined as the main gatekeepers of information for their children. Benkendorf et al. (1997) defined the proband as the gatekeeper of information, but this role may be partially or fully undertaken by parents when the proband is still not ready or able to make a fully autonomous decision. Overall, the gatekeepers of information were motivated to share genetic risk information within the family for multiple reasons. These included their need to increase information from other family members to inform their own risk perception, satisfying their own sense of responsibility to keep family members informed, to promote risk-reducing behaviours between the individuals of the family, and to increase emotional support and guidance for themselves (Daly, 2016). However, it has been shown by some authors that clients do not always pass on this risk-related information to their relatives (European Community Huntington's Disease Collaborative Study Group, 1993; Miesfeldt et al., 2003; Ormond et al., 2003; Wagner Costalas et al., 2003; Clarke et al., 2005). Clarke et al. (2005) assessed the frequency with which genetic counsellors become concerned about the failure of clients to disclose important genetic information to their relatives by means of a prospective

collaborative study. They collected data from 12 Regional Genetic Services in the UK and two in Australia over 12 months. Nearly 40000 genetic counselling sessions were performed annually across all centres. A total of 65 cases of nondisclosure were reported, representing less than 1% of the genetic counselling interactions conducted. A very interesting point in relation to my doctoral study was that those authors showed that in 39 cases of these 65, parents were not passing full information on to their adult offspring. However, this result may not be surprising because, as Metcalfe et al. (2011) reported, parents often find it very difficult and emotionally painful to discuss genetic risk information. In fact, Metcalfe et al. (2011) indicate that parents delay the communication for as long as possible until a particular event occurs. In contrast, young people reported that learning their potential genetic risk as younger children in an informal discussion allowed them to become aware of the risk gradually as they grew up (Metcalfe et al., 2011).

6.4.3 CHILDREN'S UNDERSTANDING OF GENETIC RISK

There is a lack of literature on children's understanding of genetic risk based on their parents' descriptions of the information. Young people were reported to have understood the risks for themselves around 12-15 years of age, while they realised the reproductive implications around 15-17 years of age (Metcalfe et al., 2011). On the other hand, parents were reported by Metcalfe et al. (2008) to experience anxiety, worry, concern and lack of support or advice from health professionals regarding their experience of discussing genetic conditions with their children. In fact, in a recent paper,

Hodgson et al. (2016) analysed the improved family communication based on a specifically designed telephone genetic counselling intervention in 95 probands/parents. This was a randomised controlled trial study conducted in six public hospitals, where participants were randomised into the control (current practice) or the intervention group (dedicated telephone follow-up and telephone contact at three, six and 12 months). All participants were examined 18 months after recruitment with the aim of enhancing their ability to identify and overcome the barriers regarding the communication of genetic information within the family. Although no major differences were identified, they concluded that health professionals may want to consider additional ways of supporting families in communicating genetic information. Similar conclusions were drawn by Metcalfe et al. (2011) and by Paneque et al. (2015): family members' coping and adjustment to genetic risk information could be helped by more family-centred care from health professionals. This is considered as a part of providing a high standard of quality in the presymptomatic genetic counselling session (Paneque et al., 2015).

6.4.4 THE GENETIC COUNSELLOR'S ROLE

In a recent systematic review of literature, Skirton et al. (2015) assessed the role of genetic counsellors and showed that genetic counsellors undertake a significant amount of work associated with patient care. Although the genetic counsellor's role in supporting families in communicating genetic information was not mentioned by the papers they analysed (Skirton et al., 2015), the importance of the health professional

being appropriately trained and possessing the relevant competences to provide presymptomatic genetic testing was underlined by Skirton et al. (2010). Health professionals must be able both to recognise their own limitations and to make referrals to other health professionals if more support is required (Skirton et al., 2013). A lack of counselling skills is considered a barrier to effective communication (Faulkner, 1997): this is particularly important in the context of presymptomatic genetic testing where making the decision and coping with the results may have an impact on the entire life of the consultand.

Although it has been shown that clients benefit from clinical genetic services in terms of discussion of risk, feeling of control, and helping management of the emotional effects (McAllister, Davies, et al., 2007; McAllister, Payne, et al., 2007; McAllister, Payne, Macleod, et al., 2008; McAllister, Payne, MacLeod, et al., 2008; McAllister et al., 2011), some young adults in families with a hereditary cancer predisposition decided not to be tested without undergoing the genetic counselling, although I have no data about this population. Empowerment is the term used by McAllister, Payne, Macleod, et al. (2008) for describing the feeling of control over and hope for the future in a person from a family with a genetic condition. In that study the authors found that clients perceived feelings of control when they underwent genetic testing. Empowerment is also a useful client-reported outcome both to assess interventions in clinical genetics and to generate data for the quality improvement in clinical practice (McAllister et al., 2011). A reliable and valid measure of genetic counselling outcome is provided by the Perceived Personal Control tool developed by Berkenstadt et al. (1999).

They showed that the main counselling factor that influenced the perceived personal control of the client is the simple fact of being at risk and not the level of risk. It is clear that the perceived personal control of clients depends on the nature of the specific genetic condition and it must be remembered that Berkenstadt's study was undertaken in a specific cultural environment, in Israel, and its findings may reflect the particular social norms of that country. However, improving the clients' sense of control by emphasizing specific issues by means of the concept of perceived personal control is one of the skills that genetic counsellors may use. Even if the presence of the genetic condition in the family and the anxiety of being at risk cannot change with the genetic counselling, the genetic counsellor's sensitivity to issues of the perceived personal control can be important and may be helpful in many scenarios.

After completing the three phases of my study, I started to think deeply about the role of the genetic counsellor in respect to clients who belong to this specific age group. Generally, it is important that the genetic counsellor understand the complex emotions that a client can experience during the process of a genetic counselling session (Evans, 2006; Uhlmann et al., 2009; Harper, 2010). The counsellor has to be sensitive to each of these emotional states and understand the effect on the client. They can help to prepare the client for the possible psychosocial consequences of genetic testing (Evans, 2006). During the period in which the client is considering whether or not to undertake genetic testing, the genetic counsellor can explore whether the client has discussed the option of genetic testing with other members of her family who might be affected by the test results, and whether she or he has considered if and how the results will be shared

among relatives (Sadler et al., 2004). To arrange individualized care for clients who are having the test, or who have already undergone the test, the genetic counsellor should pay attention to the client's interests and ask questions that probe their emotions and expectations (Hutson, 2003). A systematic review of available trials suggests that genetic breast cancer risk assessment helps to reduce distress, improve the accuracy of the perceived risk of breast cancer and increase the client's knowledge of it (Hilgart et al., 2012). The role of the health professionals in this context was mentioned by some young adults interviewed (Phase 2), as they expressed the desire to have an opinion to guide them during the genetic counselling process. Although this need for guidance may conflict with the non-directive philosophical stance of genetic counsellors (Evans, 2006; Uhlmann et al., 2009; Harper, 2010), health professionals may not have clarity regarding if, how or when to provide more directive genetic counselling (Werner-Lin et al., 2015). One third of genetic counsellors reported using '*somewhat directive techniques*' (Werner-Lin et al., 2015b, p. 83) during their genetic counselling sessions with 18-25 years old clients. However, the authors of that study showed that one genetic counsellor affirmed that her counselling was more directive when clients were older adults:

> *"[…] I am more directive with a cancer-free woman who is age 45-50 who has a BRCA mutation in her family to get tested because the test result would directly impact a decision for surgical risk reduction (ovaries) in the very near future. But for a 20-25 years old, I am much less directive because those decisions are not necessary for her immediate care*" (Werner-Lin et al., 2015b, p. 83).

Other genetic counsellors in the study reported that non-directive counselling is no longer valid, especially in the cancer genetic context.

Although they underlined that the client point of view was to be respected, their recommendations were provided in a directive way (Werner-Lin et al., 2015). Nevertheless, genetic counsellor participants emphasised the need to conduct appropriate assessment of young adult clients, tailoring their counselling to the needs of the younger clients. They also reported that they referred young adult clients from families affected by hereditary breast and ovarian cancer to advocacy and support organisations for psychosocial information and support. It was apparent that genetic counsellor participants in Werner-Lin et al.'s study (2015b) were focussing less on counselling and more on genetic information in their sessions. This means that they used less time to explore psychosocial issues with clients and their families. Therefore, in the context of different approaches to genetic counselling, it is not surprising that I found in the systematic review that young adults reported the experience of genetic counselling either as an opportunity for discussing problems or associated with feelings of disempowerment (Phase 1).

6.4.5 THE DECISION MAKING PROCESS

A recent systematic review on genetic testing decisions conducted by Sweeny et al. (2014) showed that the relationship between age and the decision making process regarding genetic testing is complex. Authors showed that, for example, younger men are less interested in testing for prostate cancer susceptibility than older men, while they revealed that younger people were more interested in a hypothetical genetic test for obesity. However, the interest in the genetic testing for Alzheimer's disease,

psychiatric conditions, lung cancer risk, general genetic testing, and general cancer risk did not seem correlate with age.

Although the systematic review (Phase 1) findings showed that the choice to undergo genetic testing constituted a major life event, parents appeared to have exerted pressure on their young children regarding the choice to undergo presymptomatic testing. Similar results came to light from the qualitative phase. The decision making process and the parental influence are not widely discussed in the literature, even in other health care contexts. However, to demonstrate the relevance of parents' influence on adolescent behaviour a Social Interaction model of parenting was developed by Dishion and McMahon (1998). As shown in Figure 6.1, the parent-child relationship is at the centre of the model, while the parent's motivations are at the apex.

FIGURE 6. 1 SOCIAL INTERACTION MODEL OF PARENTING DEVELOPED BY DISHION AND MCMAHON (1998)

The purpose of this model is to explain the connections between the quality of the parent-adolescent relationship, parental monitoring, parental management of adolescent behaviour, and parents' norms, goals and values. For example, a review by Hayes et al. (2004) showed that parents' behaviours have a direct influence on adolescent alcohol use. Parental monitoring is showed as the most significant parenting behaviour regarding both adolescent alcohol use (Hayes et al., 2004) and sexual behaviours (Buhi and Goodson, 2007). There is some evidence that shows that when the parent-adolescent relationship is of poorer quality, the influence of peers starts to have an important role on the decision making process of adolescents (Armsden and Greenberg, 1987). Generally, parents were involved in decisions concerning vocation or money, and peers in decisions about clothes, social activities, and entertainment (Hayes et al., 2004). In addition, the effects of peer presence on both risk taking and risky decision making was more clearly seen during middle and late adolescence than during adulthood (Gardner and Steinberg, 2005). In Western society, many adolescents are confronted with the choice of deciding whether to use or not to use available addictive substances, such as cigarettes, volatile substances which can be inhaled, marijuana, some chemical substances (amphetamines, ecstasy, ice), psychotropic substances (magic mushrooms, datura, LSD), and hard drugs (cocaine and heroin) (Geldard and Geldard, 2009). Some of these available addictive substances are legally available for use by adults and others are not, while other substances may not be legally used by young people below certain ages in most countries. Adolescents face dilemmas related to their behaviour

due to their natural tendency to explore and experiment (Geldard and Geldard, 2009), but several researchers have investigated the effects of family and peer influence on young people in this context. Bahr et al. (2005) assessed the effect of peer and family characteristics on the risk of adolescent drug use. They found that the family characteristics have a direct influence on adolescent drug use, distinct from any peer influences. The choice of peers was found to be correlated with family characteristics: adolescents were more likely to have friends who use drugs when parents were tolerant of drug use. A negative association was also observed between parental monitoring and having friends who use drugs, while adolescents who were attached to their parents were less likely to have friends who used drugs. Frauenglass et al. (1997) provided data on the fact that young people were influenced by peers using tobacco, alcohol, and marijuana, but they reduced the use of tobacco and marijuana when they were supported by the family. Parental attachment is therefore an important feature when considering how young adults make decisions that affect their health. In the genetic testing context, Metcalfe et al. (2011) found that parents encouraged and supported their young children to make their own decisions. Although this attitude appeared to support young people to discuss the information received and their thoughts with their parents, the authors showed that young people did not discuss either the genetic information or their concern with parents. In this scenario, parents started to make decisions and to take a more authoritative role (Metcalfe et al., 2011).

Other contexts will now be considered to support further understanding of young adults' decision making processes. These findings will be compared with the presymptomatic genetic testing scenario that I have analysed in this doctoral study. First of all I considered the diagnosis of cancer in young people. The experience of cancer in teenagers and young adults occurs at a significant stage of human life, a stage where young people make the transition from children to adults. While cancer treatment takes place, many activities normally experienced by young adults are suspended: for example, the process of making independent decisions may be halted completely (Davies et al., 2015). To be more specific, it has been shown that young adults interrupted planning for their future adult life because health care issues had taken priority (Stern et al., 2010): they also curtailed everyday activities because of the effect of treatment, although they wanted to have a social life that was similar to their peers (Davies et al., 2015). In terms of decision making regarding the suspension of social interaction, this was often made in agreement with parents. The experience of cancer in this age group has the potential to have an effect on the decision making process (Wicks and Mitchell, 2010). Young adults also go through a process of obtaining control over their health care and support needs, due to becoming socially independent (Kirk, 2008). Kirk (2008) interviewed 28 young people with the aim of analysing the experience and perspectives of the disabled child population, particularly in relation to transition into adulthood. The author concluded that young people were not well prepared by the children's services for becoming more frequently involved in the decision making process regarding their health

care choices. On the other hand, Vandemheen et al. (2010) identified the decisional needs of adult cystic fibrosis patients considering referral for lung transplantation. Their decision making process was characterised by some decisional needs such as wanting more information, and how to cope with anxiety and depression when making the decision (Vandemheen et al., 2010). Another different point of view is shown by Henderson et al. (2006) who analysed how individuals (aged between 18 to 34 years) make decisions about presymptomatic genetic testing in a simulated decision task. Authors recruited people without family history of a known genetic condition for which presymptomatic genetic testing was available and none of their participants had considered the issue of presymptomatic testing before taking part in their study. It was pointed out that few participants in that study (Henderson et al., 2006) considered both options, either to proceed with presymptomatic genetic testing or not to proceed, in their hypothetical decision scenario. Until they found an acceptable solution, they decided to consider both decision alternatives because they continued to search for information. Instead, three quarters of their participants used the evaluation of the consequences of the various decision options as a strategy in their decision making process. These findings were in accord with what was found in seminal work by Lippman-Hand and Fraser (1979a, 1979b, 1979c), who explored the decision making process in prenatal genetic counselling. Authors showed that clients constructed scenarios about what could happen: in this way they were able to isolate the consequences that they considered important to them and to think how they felt about each option. The decision making process guided by

mental concepts (such as the construction of scenarios) was not reported by any of the participants whose data were analysed in Phases 2 and 3 of my doctoral study, nor did it emerge from the systematic review conducted as Phase 1.

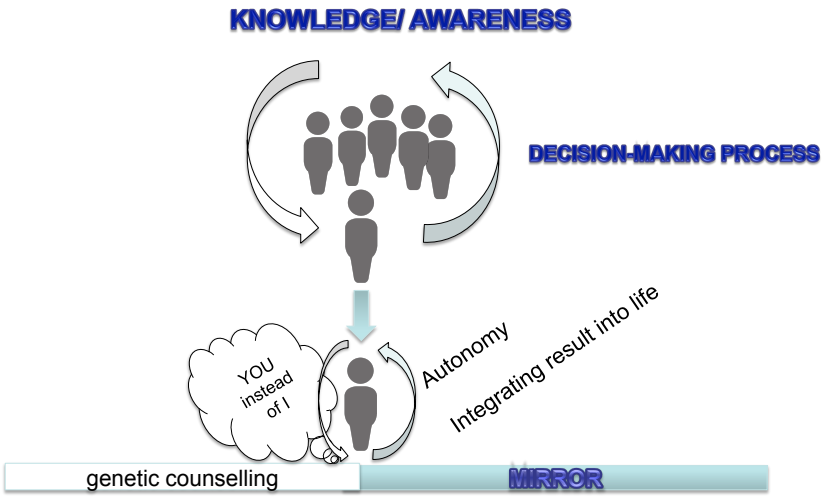
Regarding presymptomatic genetic testing, Richards (2008) pinpointed that there is no specific age when a person is able to give autonomous consent, but it is important to consider psychological maturity that is cumulative with age, life experience and cognitive development (Steinberg and Cauffman, 1996). In Italy and in the UK, parents do not have legal responsibility over their offspring once they achieve the age of 18 years old. Therefore, they have no legal control over the decisions made by their offspring (Davies et al., 2015). Although young adults may still need support at this stage of their lives because they could be vulnerable and inexperienced, parents may also feel the need to take some control over their young adult offspring who are considered legally adults (Grinyer, 2004). The role of parents (and other people) in the young adult's journey has been reported in this doctoral study. As previously indicated, young adults currently are more likely to be living at home with their parents, due to the economic situation (Pew Research Center, 2012, 2013). It is a reasonable hypothesis that living with parents for an extended period has an impact on developmental tasks and reduces the autonomy of young adults in their decision making processes: not only in their daily lives, but also when medical decisions need to be made.

6.5 THE DEVELOPMENT OF THE MODEL

I will now present an updated figure of the conceptual model that emerged after the qualitative doctoral study phase (Figure 4.2). Figure 6.1 shows that some of details differ from those in the earlier figure, due to the exploratory sequential nature of the mixed method design that I used.

Constant re-reading of the data with each successive set of findings has clarified some of the findings and led me to re-organise others, whilst new findings have been identified and included. What I first considered should be the ideal path for the genetic counselling process was a client who approached genetic counselling having basic knowledge of his situation and increased his knowledge after the counselling, thereafter making a decision to be tested or not.

FIGURE 6. 2 FINAL MODEL OF YOUNG ADULT GENETIC COUNSELLING PROCESS



However, it appears that based on qualitative interviews performed in the previous phase, all participants arrived for genetic counselling with the decision already made. They saw themselves as in front of the mirror during the genetic counselling. They distanced themselves from the person they see reflected and talked about themselves not in first but in the second person, especially when they spoke about sensitive situations. At the end, they obtain a kind of autonomy and realise how to integrate the test result into their everyday life (see Figure 4.2 and Section 4.5 in Chapter Four for more details). However, in the last phase of my research project it was revealed that the young adults surround themselves with other people (such as parents, other family members, persons outside the family) who influence them with their knowledge and awareness. Young adults' decision making process started with these people and only those who decided to be tested arrived for genetic counselling. Those who decided not to be tested declined genetic counselling as well, and I have no data about this group of young adults.

My model has challenged the accepted wisdom that genetic counselling is about the decision making process. As illustrated by the model that emerged during my doctoral research, participants arrived at genetic counselling with the decision already made. Because of this, the role of genetic counselling in young adults could shift so that the main aim was to facilitate understanding. To elaborate, the genetic counsellor needs to understand the knowledge and awareness levels of the person in front of them and only after eliciting the client's beliefs can the genetic counsellor

help them to reflect upon and potentially amend their decision regarding testing.

Decisions in genetic counselling lead to significant actions such as testing for hereditary cancer predisposition. Theoretical models of health behaviour underline factors that are relevant for understanding health-related actions, and explain why some people take protective actions and others do not. For example, Becker et al. (1977) described the health belief model. The principal predictors of the likelihood of engaging in a specific health behaviour associated with taking the action are the perceived susceptibility to a health threat, its perceived severity, and the perceived benefits and barriers. These four major constructs of perception are modified by other variables, such as culture, education level, past experiences, skills, and motivation. These are individual characteristics that influence personal perceptions. One of the individual characteristics to take into account is autonomy. It is clear from the literature that there is not a specific age when a person is able to give autonomous consent (Richards, 2008). Genetic counsellors may consider the psychological maturity of the client in front of them (Steinberg and Cauffman, 1996). In the light of these considerations, I could conclude that the final theoretical model could be applied to other genetic counselling clients, for example it could be suitable within a context of prenatal genetic counselling, where the decision making process is a major component. Moreover, because there is not a specific age when a person has defined autonomy, it is possible that this model could be applied to all clients who are involved in a decision making process in which other people could be influencing their choice. Due to the familial nature of

many genetic conditions, decisions by one person are likely to have an impact on others and it is perhaps not surprising that family members would seek to influence their relatives. However, further research would be needed to test whether the model does apply to a wider cohort of clients accessing genetic counselling.

6.6 A REFLECTIVE ASSESSMENT AND IDENTIFICATION OF STRENGTHS AND LIMITATIONS OF THE STUDY PROCESS

In this section I will report a general reflexive account followed by a reflective assessment and identification of strengths and limitations of the study process in detail for each phase of this doctoral study. The recruitment process will also be addressed. This section will be followed by the identification of strengths and limitation of the mixed methods sequential exploratory design.

6.6.1 A GENERAL REFLEXIVE ACCOUNT

Reflexivity is defined by Bryman (2016) as the awareness by researchers of the implications and power relations of their methods, values and decisions during the research process. In other words, the researcher is someone who both extracts knowledge and is viewed as implicated in the interpretation of knowledge (Bryman, 2016).

First of all, it is salient to briefly summarise some of the key things which have shaped me as a researcher during this three year journey. I decided to undertake this doctoral research project four years ago because

after the European Meeting on Psychosocial Aspects of Genetics I started to think about the psychosocial implications of presymptomatic testing for hereditary cancer in young adults. Much of the published literature aims to analyse adults' or children's experience. I wondered what happened with those in the population who are neither adults nor children. To be more specific, how young adults interpreted cancer presymptomatic testing, what the basis was for their decision to undergo testing or not, parental influence, and how young adults react once aware of their genetic status.

6.6.2 A REFLECTIVE ASSESSMENT AND IDENTIFICATION OF STRENGTHS AND LIMITATIONS OF THE QUALITATIVE INTERVIEWS CONDUCTED (PHASE 2)

During this research project, I experienced conducting face to face qualitative interviews for the first time. First of all, it is important to remember that the research interview is a social occasion and this will have had an influence on what people said or felt they could say to me, the researcher. The social nature of this interaction will have both released and constrained some aspects of the conversation, so even though there may have been no intention to conceal or deceive, participants will have only disclosed certain information in this specific social context. For example my background, age, and gender may have influenced the relationship between me and the participants. On one hand, they may have felt a form of friendship bond (due to my age and friendly manner), that enabled them to disclose some types of information but restrained them from talking about other topics. During interviews, I felt that it was important to reiterate that I was a researcher doing a PhD at Plymouth University, but that I also worked

as a genetic nurse at the Oncology Genetic Clinic at the Genetic Units of Bologna University Hospital Authority St. Orsola-Malpighi Polyclinic, and that I am a young adult as well. Talking about my own background and interest in the subject and being a young adult 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 was positive in that it made participants feel comfortable talking to me, and made me feel more confident talking to them. I have gained personally by embracing the idea of a participatory approach where young adults are viewed as people with complex individual lives rather than merely subjects of research. By meeting participants personally at the recruitment and interview stage and working hard to develop rapport, across six months, this was invaluable to me later when absorbed in analysis, as a reminder of why I had originally started the journey. In this context, a relationship of trust as a researcher was established with each participant.

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 the first interviews, in fact, young adults spent considerable amount of time during the interview wondering what the presymptomatic genetic test meant and the impact of it on themselves. Although this was not strictly in line with the guide, I felt that because the subject was clearly an emotional one, it would be inappropriate to interrupt.

I also felt that they had perhaps mistaken me for a professional at that moment. After this, I made sure at the beginning of interviews that it was clear I was a researcher for them and not a genetic counsellor in that moment, and I was unable to offer any medical advice or information regarding what they would like to know. It is possible that, participants felt more comfortable talking to me than they may have been with an older person. For this reason, it is possible that participants revealed to me something that would not have come to light if the interviewer were an older adult.

6.6.3 A REFLECTIVE ASSESSMENT AND IDENTIFICATION OF STRENGTHS AND LIMITATIONS OF THE ANALYSIS OF QUALITATIVE INTERVIEWS (PHASE 2)

In reflecting on the method chosen to analyse interview data, I believed that the grounded theory approach worked well for the qualitative phase of this doctoral project as it provided a good framework for key themes to surface from the data. In addition, the findings and theory derived from this approach have contributed to the literature. Although it is important to declare that my own values, interest and experiences (both personal and working) will have shaped my interpretation of the findings, I hope that by using the validation techniques described in grounded theory that my personal 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. However, because a systematic review on presymptomatic genetic testing in young adults had already been conducted, a number of key themes had been

identified before qualitative research began. On one hand, this information gave me a good understanding of the key issues, on the other hand it was important to ensure that knowledge did not exert too much influence over my coding. For this reason, I tried to remain as open as possible to new categories and themes, identifying them inductively from the data in line with the grounded theory approach. Creating hundreds of codes (free nodes) before attempting to group them together into high-level categories was the method I used to achieve this. To further ensure the emerging theory was grounded in the data, I labelled codes and categories using words or phrases directly taken from participants in the doctoral study.

6.6.4 A REFLECTIVE ASSESSMENT AND IDENTIFICATION OF STRENGTHS AND LIMITATIONS OF THE QUANTITATIVE STUDY (PHASE 3)

Regarding the quantitative phase of this doctoral project, exploring young adults' and parents' experiences, I have developed organizational skills and learned to apply statistics in order to understand patterns in the data. I have also improved my skills in statistical inference, sampling methods and data analysis. By the end of this journey, I was able to derive an estimator and its properties, to use estimating methods, to define and verify parametric and non-parametric statistical hypotheses in simple context, and to build confidence intervals. I was also able to conduct stratified and probability sampling, to derive the estimators and associated standard errors of population in the different sampling strategies, to correct estimation by the ratio principle, and to understand the difference between observational and experimental studies. I was also able to apply and interpret methods of

dimension reduction, methods for cluster analysis and discrimination, and interpret the output of R procedures for multivariate statistics. I have also continued to develop my academic writing and presentation of my work to a variety of audiences as described in the Author's declaration of this dissertation.

6.6.5 A REFLECTIVE ASSESSMENT AND IDENTIFICATION OF STRENGTHS AND LIMITATIONS OF THE RECRUITMENT PROCESS

One of the biggest challenges I faced was recruitment of PIQ which differed from the approaches I had to use with PEQ. First of all, there are many specific Facebook groups in the English speaking world regarding hereditary cancer syndromes, while there are only two Italian ones. I was not able to use one of them during the recruitment process because the administrator of the group demanded to become one of the authors of the research publications resulting from this doctoral study. I was very embarrassed about this request. I discussed it with my Director of Studies and other supervisors and decided it was not ethical, and the request was refused. This could be one of the reasons limiting the number of Italian questionnaire respondents. In fact, only the 39% of PIQ were recruited from the Internet, compared with the 100% of PEQ. On the other hand, one of the strengths of this study phase was that I have administered the surveys and the interviews in Italian, which enabled me to recruit all those in Italy who spoke Italian, not just Italians who were English speakers. More generally, instead, I noted that the recruitment by Google+ failed. It is possible that because it is a more recent social network, fewer people use it regularly. This

result is consistent with the data in the literature. In fact, it seems that 2.2 billion are the active profiles, but only 6% of those have any post activity in 2015 (Barrie, 2015) (see Section 2.5.4.1 for more details). This could be another general reason that limited the number of PIQ and PEQ.

6.6.6 A REFLECTIVE ASSESSMENT AND IDENTIFICATION OF STRENGTHS AND LIMITATIONS OF THE MIXED METHODS DESIGN

I have used a mixed methods sequential exploratory design by incorporating the systematic review, the qualitative study and the quantitative study. In this way, I was able to achieve the aim of this programme of doctoral study, which was to explore the implications of presymptomatic testing for hereditary cancer in consultands aged 18-30 years. However, this was not entirely satisfactory because, although similar findings were identified using the different methods, the quantitative study phase involved very few questionnaires that were completed by parents of young adults who underwent presymptomatic genetic testing. Because of this, the possibility of generalizing the results, specifically the results of the factor analysis, could be hampered by the small sample size. In the literature the minimum sample size required is largely discussed. The importance of a definite minimum sample size was focused upon by many authors (Guilford, 1954; Comrey and Lee, 1973; Gorsuch, 1974), while other authors focused on the number of cases per variable (Cattell, 1978; Hair et al., 1979). Consistently, in order to minimise the chance of overfitting the data, researchers suggested obtaining the highest cases per variable possible as I have already described in Section 5.4 in Chapter Five. While I achieved

sufficient data from young adults, the argument that there were adequate numbers of parents participants is less convincing. The involvement of parents in the presymptomatic genetic testing experience of their young adults offspring therefore needs further exploration. Nevertheless, my results of Phase 3 of this doctoral study showed some interesting points that I have already discussed in Chapter Five.

Additionally, a grounded theory for assessing the implications of presymptomatic genetic testing for cancer in young adult clients was proposed. This qualitative phase was able to provide more detail about the lived experience of young adults who decided to undergo genetic testing. In particular the importance of pre-existing awareness/ knowledge, the parental pressure on the decision making process, the experience of genetic counselling and dealing with the result were explored.

In conclusion, by combining qualitative and quantitative methods in this doctoral project, I was able to cross-check the findings using the sequential exploratory mixed method design. Overall, 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 the following phase. By using a mixed method design, I was also able to develop a richer and more complex model of what was occurring.

6.7 CONTRIBUTIONS OF THE STUDY TO THE BODY OF SCIENTIFIC KNOWLEDGE

The results of the research presented within this dissertation provide several theoretical and practical contributions, which are described in the subsections below.

6.7.1 THEORETICAL CONTRIBUTION

Whilst genetic testing of minors has been largely analysed by other researchers, the testing of young adults has not been well-addressed by researchers. What is novel in this doctoral project is that I have identified a multi-faceted and dynamic process of young adult decision making, regarding whether to be tested or not. Moreover, I have developed a new model to describe the psychosocial implications of presymptomatic testing for hereditary cancer in consultands aged 18-30 years.

6.7.2 METHODS

Whilst the methods employed in this doctoral project have been used widely in health research, the mixed methods design formed a robust methodology to develop client information. By combining qualitative and quantitative methods I was able to verify the findings. To the extent of my knowledge, this is the first time that a mixed method design has been used to investigate experiences in a sample of young adults and their parents regarding genetic testing. The approach used in this doctoral study can be adopted by others as a guide to acquire evidence in other areas of genetic

health care and other situations where health-related decision making is required.

6.7.3 PRACTICE

There are a number of recommendations that can be made to inform service providers in light of the findings from this doctoral study.

The evidence of this doctoral project indicates that many young adults grew up without information or with misinformation concerning their potential genetic risk, but their knowledge became more accurate after genetic counselling. Parents seem to be the major source of information. In addition, there is little information regarding the role of genetic counselling in supporting young adults and providing information, whether or not they wish to be tested.

It is therefore important to publicise the supportive and educational role of genetic services. One way to achieve this could be by forming partnerships with patient support groups so that they can help to convey this message. Another strategy would be to include information about this aspect of the service in clinic literature.

The Internet is generally well-used by young adults (Roger, 2002) and is an important source of information for them (Vance et al., 2009). In response to the findings of this doctoral study, the website of the Genetic Units of Bologna University Hospital Authority St. Orsola-Malpighi Polyclinic was searched and assessed. It was noted that much of the information on the clinic website focussed on which tests were performed, with no information

about the role of the counselling service. It would therefore be useful to add some information about genetic counselling and that testing is a choice. Young adults need to be aware that they can undergo genetic counselling regardless of their decision about testing or if they have not yet decided. In the light of my findings, it has been revealed that young adults are influenced by others and only those who wish to be tested utilise genetic counselling. Counsellors therefore need to be aware of these issues when testing young adults. Appropriate communication of genetic risk information by parents to their children is highly desirable, since it has been shown to have long-term consequences (Metcalfe et al., 2008). To achieve this, health professionals could have a role in both supporting parents and young adults, but their involvement in the parents' decision to communicate genetic risk to young family members was found to be limited in the systematic review performed (Phase 1) and previous reports (Rew et al., 2009; Bradbury et al., 2007; Werner-Lin et al., 2015). Although this may be partly due to the parents' wish to undertake this task alone, it is reported that some parents desired health professionals to be available in a supporting role, but found that this support was limited (Gaff et al., 2006; Metcalfe et al., 2008).

According to the General Medical Council's guideline (General Medical Council, 2012), it is the clinician's duty to assess about the adolescent's level of understanding and maturity and to act accordingly. However, it may be unrealistic to expect genetic counsellors to be able to assess the adolescent's level of understanding and maturity. This could be addressed by involving appropriate trained professionals who have specific skills, such as child/adolescent psychologists in the genetic counselling process (Binedell,

1998). This evidence highlights the need for a comprehensive, longitudinal counselling process with appropriate timing and setting, which supports 'parent-to-offspring' risk communication first and young people's decision making about presymptomatic testing and risk management afterwards. Concluding, it is clear that counselling approaches to this population may require modification both for young adults and their parents. From both the qualitative and quantitative phase it has been shown that on one hand, young adults may benefit from a multi-step approach for undergoing genetic testing. On the other hand, parents need to be more informed that genetic counselling is a place where information is obtained and young adults can talk about the decision, regardless of whether they want to be tested or not.

Some young adults felt they could benefit from an appropriate support group for them to contact because it could provide general information and support to young adults who are living with or experienced the presymptomatic genetic testing choice. Nevertheless, data shows that a parent support group could be helpful as well. More specifically, parents could benefit from the information provided by organisations for particular hereditary cancer predisposition.

In summary, my recommendations are:

1. To publicise the supportive and educational role of genetic services forming partnerships with patient support groups so that they can help to convey this message or include information about this aspect of the service in clinic literature
2. To add some information about genetic counselling and that testing is a choice on the relevant webpages on the hospital website

3. To modify the counselling approach to this population. This evidence highlights the need for a comprehensive, longitudinal counselling process with appropriate timing and setting, which supports 'parent-to-offspring' risk communication first and young people's decision making about presymptomatic testing and risk management afterwards. For these reasons, young adults may benefit from a multi-step approach for undergoing genetic testing, and parents need to be more informed that genetic counselling is a place where information is obtained and young adults can talk about the decision, regardless of whether they want to be tested or not.
4. To have an appropriate support group for young adults and parents.

6.8 RECOMMENDATIONS FOR FUTURE RESEARCH

This section highlights several areas of research that could be investigated further as a result of this research, but which were not addressed during the PhD due to the timescale of the study. As discussed in Section 5.4 (Chapter Five), to determine whether the findings related to the influence of parents are more generalizable, further investigation with a larger sample is required, both of PIQ and parents of young adults. Furthermore, as a result of conducting this doctoral study I would recommend the following future studies:

1. A study designed to obtain more data from parents of a child who had a genetic test for cancer when they were aged between 18-30 years to enable a more detailed exploration of the complicated inter-relationship identified between them and their children.

2. To add to the current findings, a mixed methods study exploring the decision making process from the professionals' perspective would be helpful. In-depth interviews with genetic service professionals could be conducted. Areas for exploration include: professionals' views regarding young adults and their parents, what their experience is of the decision making process of this populations, how they facilitate communication between the parent and their young adult children, and professionals' experience of counselling both parents and young adults. A quantitative study would be performed to systematically assess the most relevant findings emerging from the qualitative study in a wider population. To achieve this aim, based on the results of the previous qualitative study, some specific variables would be identified, and the most appropriate tools would be chosen to measure those variables. Specific questionnaires would be designed.
3. To determine how the role of the genetic counselling service is presented to clients, written information materials provided by genetic counsellors could also be analysed to see if and how they promote the genetic counselling as a place where the information is obtained and the decision making process starts.
4. A qualitative study looking at the experience of young adults who decided not to be tested. Particular attention would be paid to where and how they became aware of their potential genetic risk and why they declined the opportunity for genetic counselling. Grounded theory might be a suitable method to analyse the data in this exploratory type study.

6.9 CONCLUSION

In this doctoral study I have used a range of methods to explore the psychosocial implications of presymptomatic testing for hereditary cancer in consultands aged 18-30 years. As a result of the findings, I have been able to develop a new theoretical model of decision making and impact on young adults who underwent presymptomatic genetic testing, showing it as a dynamic process. Counselling approaches to this population may require modification both for young adults and their parents. From the qualitative Phase I learnt that young adults may benefit from a multi-step approach for undergoing genetic testing (for instance through a multi-step approach as for Huntington disease). From both qualitative and quantitative phases it came to light that parents need to be more informed that genetic counselling is a place where information is obtained and young adults can talk about the testing decision, regardless of whether they want to be tested or not. My data suggest that the traditional 'wait until they come to us' approach adopted by many genetic services may be failing to meet the educational and emotional needs of this population. This is particularly true for the professional community dealing with young adults at high risk, rooted in the ethics of respect for parents' privacy. 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 health care regarding young adults.

My research journey has not only produced information about the experience of presymptomatic genetic testing in young adults, it has also impacted upon me as a researcher and a genetic nurse. In this chapter I

sought to justify the importance of a reflexive underpinning in the research process and this has helped me reflect on how the process has changed me. I feel I have been very aware that the process has shaped me as a genetic nurse because by being open to findings, which were unpredictable at the start, my preconceptions have been challenged and I have sought to embrace the challenges. In conclusion, by being reflexive at all stages of the research process and attempting to learn from the challenges I have developed as a researcher and also a genetic nurse.

LIST OF APPENDICES

APPENDIX 1 APPROVAL BY FACULTY RESEARCH ETHICS COMMITTEE BY PLYMOUTH UNIVERSITY (12TH NOVEMBER 2014)

RESEARCH WITH PLYMOUTH UNIVERSITY

12th November 2014

CONFIDENTIAL

Lea Godino
UO Genetica Medica, Pad.11, 2°piano
Policlinico Sant'Orsola-Malpighi
via Massarenti, 9
40138 Bologna, Italy

Dear Lea

Application for Approval by Faculty Research Ethics Committee

Reference Number: 14/15-324
Application Title: **THE IMPACT OF PRESYMPTOMATIC GENETIC TESTING FOR CANCER ON YOUNG ADULTS**

I am pleased to inform you that the Committee has granted approval to you to conduct this research.

You may wish to consider whether information about possible sources of psychological support can be given to participants prior to the commencement of the interview.

Please note that this approval is for three years, after which you will be required to seek extension of existing approval.

Please note that should any MAJOR changes to your research design occur which effect the ethics of procedures involved you must inform the Committee. Please contact Sarah Jones (email sarah.c.jones@plymouth.ac.uk).

Yours sincerely

Professor Michael Sheppard, PhD, FAcSS
Chair, Research Ethics Committee -
Faculty of Health & Human Sciences and
Peninsula Schools of Medicine & Dentistry

Faculty of Health & Human Sciences
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Professor Michael Sheppard
CQSW BSc MA PhD AcSS
Chair, Faculty Research Ethics

APPENDIX 2 APPROVAL BY ST. ORSOLA-MALPIGHI ETHICAL COMMITTEE (29TH OCTOBER 2014)

SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA
Azienda Ospedaliero - Universitaria di Bologna

Policlinico S. Orsola-Malpighi

Comitato Etico Indipendente dell'Azienda Ospedaliero-Universitaria di Bologna, Policlinico S. Orsola-Malpighi

Bologna, 29/10/2014
Prot. n. 3413/2014

Ns. riferimento da citare sempre nella corrispondenza:
n° 132/2014/O/Oss approvato il 07/10/2014

Chiar.mo Prof. Marco Seri
Direttore
U.O. Genetica Medica
Pad. 11 – S.Orsola
Policlinico S.Orsola-Malpighi

E p.c.

Gent.mo Dott. Primoz Juric
Investigational Drug Service
c/o U.O. Farmacia – Puggioli
Pad. 19 – S.Orsola
Policlinico S.Orsola-Malpighi

Gent.ma Dott.ssa Cristina Puggioli
Direttore
U.O. Farmacia – Pad. 19
Policlinico S.Orsola-Malpighi

Oggetto: 132/2014/O/Oss – Studio osservazionale "Studio qualitativo sull'impatto dei test genetici di predisposizione al cancro nei 'giovani adulti'" – Prot. GIGA - Studio promosso dall'Unità Operativa di Genetica Medica del Policlinico S.Orsola-Malpighi di Bologna
Sperimentatore Responsabile: Dr.ssa D. Turchetti
Direttore: Prof. M. Seri

- Modulo di "Richiesta di parere al Comitato Etico per Studio osservazionale di coorte prospettico" sottoscritto dallo Sperimentatore Responsabile e dal Direttore dell'Unità Operativa il 04/07/2014
- Richiesta di autorizzazione sottoscritta dal Promotore e Responsabile dello studio il 04/07/2014
- Lettera di accompagnamento della documentazione da parte dello Sperimentatore datata 04/07/2014
- Protocollo di studio versione finale del 04/07/2014
- Sinossi del protocollo versione del 04/07/2014
- Interviste semi-strutturate (Allegato 1) versione del 04/07/2014
- Dichiarazione studio no profit datata 04/07/2014
- Dichiarazione no compenso datata 04/07/2014
- Foglio informativo per l'utente- Partecipazione allo studio versione del 04/07/2014
- Foglio informativo per l'utente- Trattamento dati sensibili versione del 04/07/2014
- Consenso informato- Partecipazione allo studio versione del 04/07/2014

COMITATO ETICO INDIPENDENTE DELL'AZIENDA OSPEDALIERO-UNIVERSITARIA DI BOLOGNA
POLICLINICO S. ORSOLA – MALPIGHI
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Sito WEB: http://www.aosp.bo.it/content/comitato-etico

SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA
Azienda Ospedaliero - Universitaria di Bologna

Policlinico S. Orsola-Malpighi

Comitato Etico Indipendente dell'Azienda Ospedaliero-Universitaria di Bologna, Policlinico S. Orsola-Malpighi

- Consenso informato- Trattamento dati sensibili versione del 04/07/2014
- Scheda Raccolta Dati versione del 04/07/2014
- Dichiarazione del proponente sulla natura osservazionale dello studio datata 04/07/2014
- Curriculum dello sperimentatore

Documentazione integrativa:

- Lettera di trasmissione della documentazione integrativa, inviata dallo Sperimentatore il 02/10/2014
- Protocollo di studio, versione emendata del 02/10/2014
- Documentazione relativa all'approvazione del progetto triennale di PhD dello Sperimentatore
- Modulo di consenso informato – Trattamento Dati Sensibili per l'utente, versione del 04/07/2014
- Modulo di consenso informato – Partecipazione allo Studio per l'utente, versione del 04/07/2014

Il Prof. Marco Seri esce temporaneamente dalla seduta.

Acquisita la documentazione ed i chiarimenti richiesti, il Comitato Etico, nella seduta del giorno 07/10/2014, ritiene che il protocollo di studio ed i suoi obiettivi soddisfacciano i criteri etici e scientifici che ne giustificano l'esecuzione. Le modalità del consenso informato sono corrette ed adeguate allo scopo. Il Comitato, pertanto, all'unanimità esprime **parere favorevole** alla conduzione dello studio.

Si segnala, tuttavia, di valutare la possibilità, contestualmente alla presentazione di un eventuale futuro emendamento, di inserire nell'obiettivo del protocollo un riferimento alla valutazione della variabile psico-sociale in linea con quanto dichiarato negli "*Aims and Objectives*" del progetto triennale di Dottorato di Ricerca.

Si ricorda che lo studio potrà essere avviato solo dopo aver ricevuto l'autorizzazione all'attivazione da parte della Direzione Generale dell'Azienda.

Si precisa che, come previsto dalla normativa vigente in materia, lo **Sperimentatore Responsabile** dovrà comunicare al Comitato Etico le seguenti informazioni relative all'andamento dello studio: data di inizio arruolamento, data di fine arruolamento e data di conclusione dello studio. Inoltre, a partire dalla data di approvazione, dovrà essere fornito un **rapporto annuale** sullo stato di avanzamento dello studio. **Per le suddette comunicazioni è possibile fare riferimento al modello disponibile sul sito web del Comitato Etico**.

Cordiali saluti.

IL PRESIDENTE
(Prof. Nicola Montanaro)

NOTA: per qualsiasi comunicazione relativa all'oggetto (compresi eventuali successivi emendamenti ed eventi avversi), è indispensabile, sia da parte dello sperimentatore che dello sponsor, fare sempre riferimento alla data della presente approvazione, nonché al numero indicato a margine dell'Oggetto.

Allegato: elenco Componenti Comitato Etico Indipendente del Policlinico S.Orsola-Malpighi

COMITATO ETICO INDIPENDENTE DELL'AZIENDA OSPEDALIERO-UNIVERSITARIA DI BOLOGNA
POLICLINICO S. ORSOLA – MALPIGHI
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APPENDIX 3 INFORMATION SHEET OF QUALITATIVE STUDY

Research Study Information Sheet

GENETIC TESTING IN YOUNG ADULTS

What is the study about?

Young adults with a family history of cancer may be offered a genetic test. Some people wish to be tested, others decide against it. This study is being done to explore genetic testing for hereditary cancer in young adults (aged 18-30 years) undergoing cancer genetic counselling. The results of the study will be used to help us understand how we can support young adults who are making decision about genetic testing.

Does this study concern me?

If you are aged between 18-30 years and have a family history of cancer, we would value your help with this study. It doesn't matter if you will undergo a genetic test or not, or if you have not decided. We are interested in the views of all young adults who are thinking about a genetic test..

What am I being asked to do?

If you agree to help us with this study, we will interview you in person on three occasions. Each interview will last approximately 15-20 minutes. You will be asked to give your opinion on genetic testing for hereditary cancer.

The interview will be recorded so we make sure your views are recorded accurately but we will change all names or other details so you cannot be identified in the final report. Your personal details will remain completely confidential.

Your health care will not be in any way affected if you decide not to be involved in the study.

I would be very happy to answer any further questions you might have about the study before you decide about being involved. If you want to ask a question, please contact me:

Telephone: +393935234941(daytime)
OR Email: lea,godino@students.plymouth.co.uk

Can I change my mind about being involved?

Even if you agree to help, and start the study, you can change your mind at any time without giving a reason. You can ask us to remove your interview data from the study at any time up to four weeks after the interview.

How do get involved?

Please contact the researcher (Lea Godino) and she will arrange a time that is convenient for you for the interview.

Telephone: +393935234941(daytime) OR Email: leagodino@gmail.com

Where do I go if I have a complaint?

If you have a complaint about the conduct of this research, this should be directed to Daniela Turchetti, MD, Assistant Professor of Medical Genetics, Department of Medical and Surgical Sciences (DIMEC), University of Bologna.

daniela.turchetti@unibo.it

Finally ....thank you for reading this information sheet.

Lea Godino

APPENDIX 4 ENGLISH VERSION OF INTERVIEWS (PHASE 2)

Interview 1

About you

Gender
- □ Female
- □ Male

Age: _____

What is your highest educational qualification?
- □ No formal education
- □ Elementary school qualification
- □ Middle school qualification
- □ High school diploma
- □ University degree
- □ Post-graduate degree

What is your daily work?
- □ Unemployed
- □ Student
- □ Homemaker
- □ Worker
- □ Employee
- □ Manager
- □ Freelance
- □ Artisan
- □ Business owner
- □ Service person
- □ Other _____

Married status:
- □ Unmarried
- □ Married
- □ Divorced
- □ Widowed
- □ Living together
- □ Other _____

Do you have any children?
- □ Yes
- □ NO

If *Yes*, how many children do you have? ____
how many daughters do you have? __
how many sons do you have? _____

Are you pregnant?
- □ Yes
- □ NO

Country of birth:
_____

Mother's language
_____

About your parents

Father
What is his highest qualification?
- □ No formal education
- □ Elementary school qualification
- □ Middle school qualification
- □ High school diploma
- □ University degree
- □ Post-graduate degree

What is his daily work?
- □ Unemployed
- □ Student
- □ Homemaker
- □ Worker
- □ Employee
- □ Manager
- □ Freelance
- □ Artisan
- □ Business owner
- □ Service person
- □ Other _____

Mother
What is her highest qualification?
- □ No formal education
- □ Elementary school qualification
- □ Middle school qualification
- □ High school diploma
- □ University degree
- □ Post-graduate degree

What is her daily work?
- □ Unemployed
- □ Student
- □ Homemaker
- □ Worker
- □ Employee
- □ Manager
- □ Freelance
- □ Artisan
- □ Business owner
- □ Service person
- □ Other _____

About the genetic test *(indicative questions)*

How did you reach the decision to have an appointment for genetic counselling?

Can you tell me more about your motivation to have an appointment?

Do you know what happens during genetic counselling?

What do you expect to obtain from genetic counselling?

How do you feel about discussing your family history?

Have you ever thought about undertaking genetic testing?

Can you tell me what you know about genetic testing?

What would you like to understand better?

What are your feelings about genetic testing for yourself?

Have you spoken with, or are you going to speak with anyone else (such as family or friends) about genetic testing? If so, what do they think?

Thank you very much for helping us with this study, we appreciate you time and effort. If you agree, I would like to talk to you again after your genetic counselling session.

……

Interview 2 (after 2 weeks from counselling session) (indicative questions)

Can you tell me what happened during your genetic counselling session?

How did you feel during the consultation? Did you have all your questions answered to your satisfaction?

Did they offer you a genetic test? How did you feel at that moment?

Did anything unexpected happen in the counselling session?

What have you been thinking about since the consultation? How do you feel now?

Have you discussed your decision about having about a genetic test with anyone else?

Thank you very much for helping us with this study, we appreciate you time and effort. If you agree, we will recontact you again in six months.

……

Interview 3 (after 6 months from counselling session) (indicative questions)

Can you tell me what has happened during this period?

Did you decide to have genetic testing?

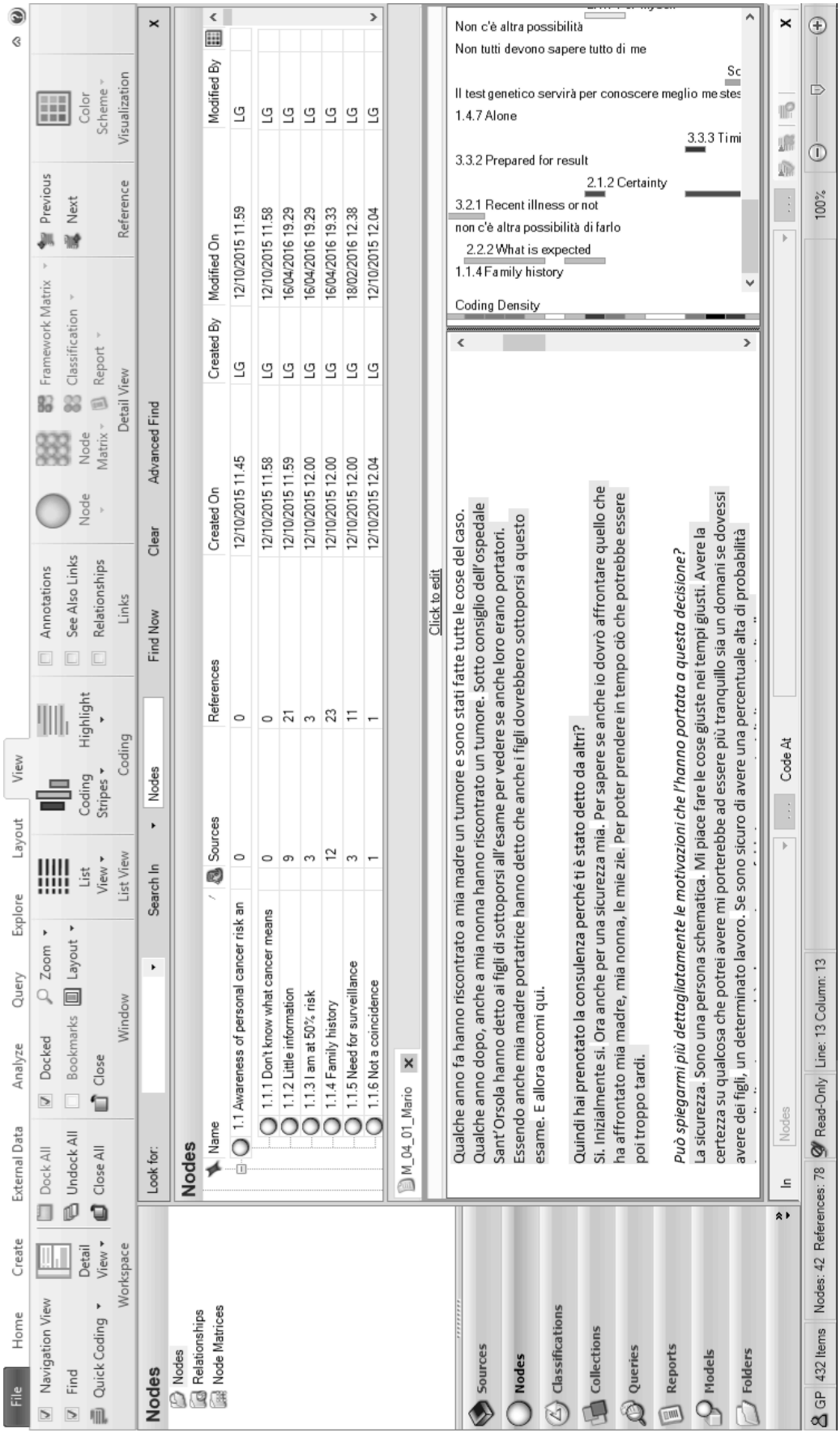
Can you tell me if anyone has influenced your decisions/ thoughts?

Can you tell me more about your experience? What do you think about emotions of young people like you may have regarding genetic testing?

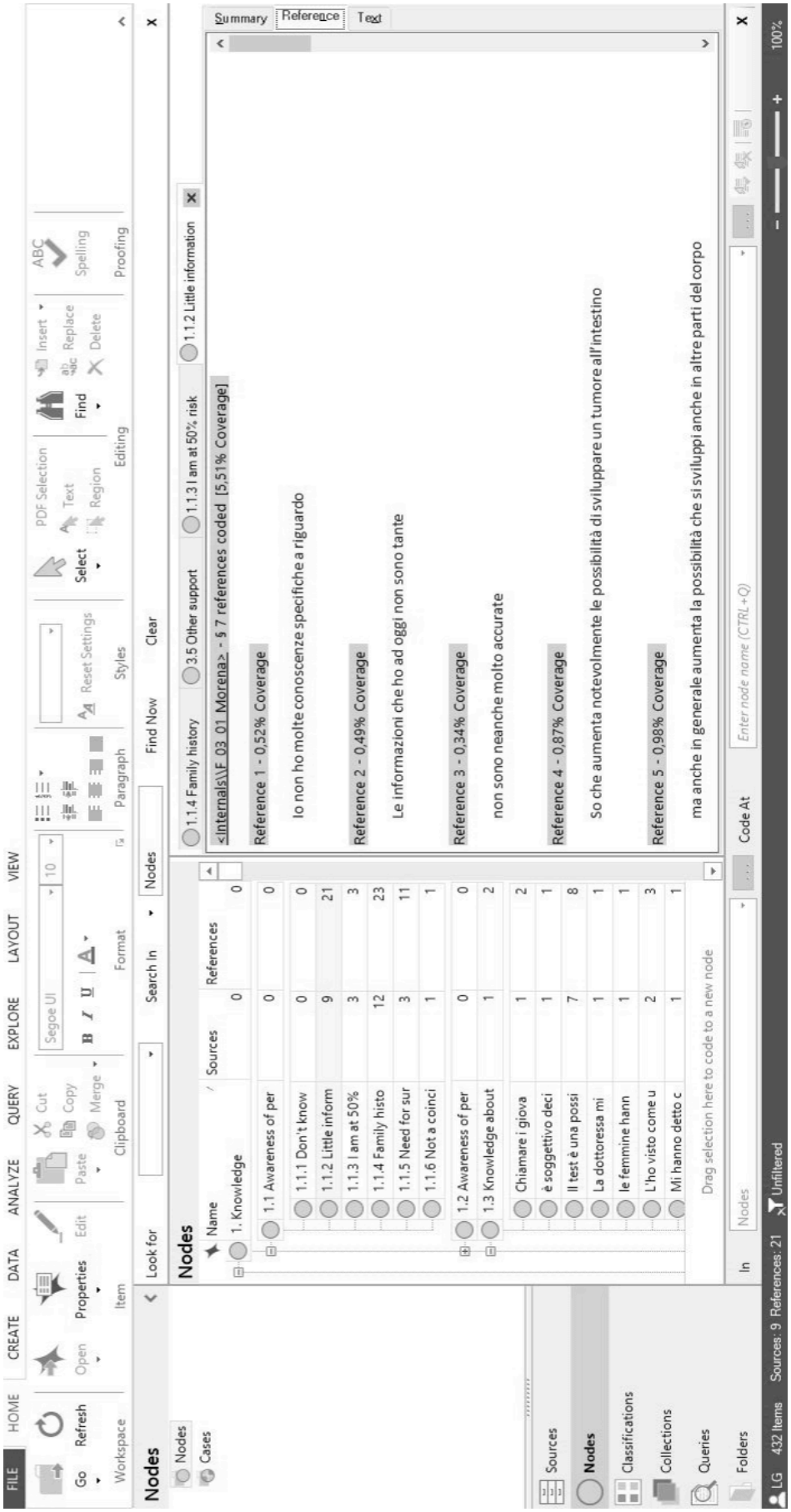
Is there any advice you can give the genetic service to help them improve their service for young people?

Thank you very much for helping us with this study, we appreciate your time and effort.

APPENDIX 5 EXAMPLE OF CODING UNDERTAKEN USING NVIVO



APPENDIX 6 EXAMPLE OF AXIAL CODING UNDERTAKEN USING NVIVO



APPENDIX 7 ENGLISH VERSION OF ALL THREE INTERVIEWS CONDUCTED WITH A YOUNG WOMAN (BARBARA)

Interview 1

ABOUT YOU

AGE: 29 YEARS OLD

GENDER:

□ MALE

þ FEMALE

WHAT IS YOUR HIGHEST EDUCATIONAL QUALIFICATION?

□ NO FORMAL EDUCATION

□ ELEMENTARY SCHOOL QUALIFICATION

□ MIDDLE SCHOOL QUALIFICATION

þ HIGH SCHOOL DIPLOMA

□ UNIVERSITY DEGREE

□ POST-GRADUATE DEGREE

WHAT IS YOUR DAILY WORK?

□ UNEMPLOYED

□ STUDENT

□ HOMEMAKER

þ WORKER

□ EMPLOYEE

□ MANAGER

□ FREELANCE

□ ARTISAN

□ BUSINESS OWNER

□ SERVICE PERSON

□ OTHER _____

MARRIED STATUS

þ UNMARRIED

□ MARRIED

□ DIVORCED

□ WIDOWED

□ LIVING TOGETHER

□ OTHER _____

DO YOU HAVE ANY CHILDREN?

□ YES

þ NO

~~**IF YES, HOW MANY CHILDREN DO YOU HAVE?**~~

~~**HOW MANY DAUGHTERS DO YOU HAVE?**~~

~~**HOW MANY SONS DO YOU HAVE?**~~

ARE YOU PREGNANT?

□ YES

þ NO

COUNTRY OF BIRTH:

KOZIENICE, POLAND

SHE HAS LIVED IN ITALY SINCE 2003.

LINGUA MADRE : POLISH

ABOUT YOUR PARENTS:

FATHER

WHAT IS YOUR HIGHEST EDUCATIONAL QUALIFICATION?

□ NO FORMAL EDUCATION

□ ELEMENTARY SCHOOL QUALIFICATION

□ MIDDLE SCHOOL QUALIFICATION

þ HIGH SCHOOL DIPLOMA

□ UNIVERSITY DEGREE

□ POST-GRADUATE DEGREE

WHAT IS YOUR DAILY WORK?

□ UNEMPLOYED

□ STUDENT

□ HOMEMAKER

þ WORKER

□ EMPLOYEE

□ MANAGER

□ FREELANCE

□ ARTISAN

□ BUSINESS OWNER

□ SERVICE PERSON

MOTHER

WHAT IS YOUR HIGHEST EDUCATIONAL QUALIFICATION?

□ NO FORMAL EDUCATION

□ ELEMENTARY SCHOOL QUALIFICATION

□ MIDDLE SCHOOL QUALIFICATION

þ HIGH SCHOOL DIPLOMA

□ UNIVERSITY DEGREE

□ POST-GRADUATE DEGREE

WHAT IS YOUR DAILY WORK?

□ UNEMPLOYED

□ STUDENT

□ HOMEMAKER

þ WORKER

□ EMPLOYEE

□ MANAGER

□ FREELANCE

□ ARTISAN

□ BUSINESS OWNER

□ SERVICE PERSON

L: Okay, now we have finished collecting information about you and your family. Let's go on with the interview.
B: I'm ready!
L: Fist of all, how did you reach the decision to make an appointment for genetic counselling? Can you tell me more about your motivation to make an appointment?
B: I decided to book a counselling session because my mother underwent a serious operation for ovarian cancer two months ago. I faced the same experience with my grandmother ten years ago and before that my aunt had the same disease. I wish to prevent this: I'm aware that I can have the gene, too, but I feel strong.
L: Do you know what happens during genetic counselling?
B: I'll undergo a blood sample and I'll wait for the results.
L: What do you expect to obtain from genetic counselling?
B: On one hand, if I had the mutation like both my mother and my aunt, I would anyway feel strong. On the other hand, I also know that if I didn't have the mutation, I could develop cancer for other reasons like pollution, diet and stress.
L: How do you feel about discussing your family history?
B: I feel prepared and calm. I've got used to this issue because I have already faced the diagnosis of cancer in my family. I'm quite prepared also for the blood sample. I can say that I'm serene.
L: Have you already thinking about genetic testing?
B: Yes, I have already thought about the genetic test. I think it will be offered to me during the counselling because of my family history. I feel prepared. I think that it's the right moment to know, I feel strong and in a good health. I'm not afraid. I think it's better to know this kind of things sooner than later, for instance when I've already got cancer … without being prepared … something occurs inside me and then … better to prevent, better to know. We are all different: someone prefer deny the disease ... I want to live! I want to fight.
L: Can you tell me what you know about genetic testing?
B: The genetic test will tell me whether I have the gene, like my mother, or not.
L: What would you like to understand better?
B: I'm searching on the web to see if something has been discovered to prevent. I'm keen on this issue. I try to understand why tumours grow ... and then I talk a lot with friends. I would also like to know what exactly will be done with my blood in the laboratory.
L: What are your feelings about genetic testing?
B: I'm very curious. Right now I think positive. Everything looks simple. When the result will be available I will probably react or feel in a different way but at the moment I don't know anything. For sure it's not an easy thing. I try to be strong, but honestly I don't know how I'll react. I don't want to think too much about the test. I know that it consists of a blood sample and a counselling session with a doctor. The only thing that matters now is the result, It allows me to prevent. Even If I won't receive a positive answer for me, I have to accept and go on. I must to do everything to stay healthy. I try to be more careful. If I wouldn't have the gene … breathe.
L: Have you spoken with, or are you going to speak with anyone else (such as family or friends) about genetic testing? If so, what do they think?
B: I haven't talked with friends about the genetic test. It's a thing of mine and my family. There's no need that everybody knows everything about me. I talked only with my relatives. For example I've been talking with my sisters and brother because they are having the test in Poland too. My brother is a doctor and drives me

to undergo the test because he says: 'it is the right thing'. But is it really the right thing? When I told my mum what I wanted to do she said: "Barbara, are you sure? You can't turn back. You will have to deal with the result. Are you sure you don't want to wait few years?' I'm curious and I want to know. If I know, I could do something to prevent.
L: Thank you for answering to my questions and thank you for your time. We will meet again after the counselling session for the second interview. Do you agree?
B: Oh yes. See you soon.

Interview 2

L: Can you tell me what happened during your genetic counselling session?
B: I don't remember much of what actually happened. I remember they handed me three sheets to read by myself and then finally they decided to take a blood sample. We have not talked so much indeed.
L: How did you feel during the consultation?
B: During the counselling I wasn't scared at all. I was definitely willing to have the test. I honestly didn't expect to be tested during the first counselling. I thought Dr D. would decide whether to test me or not in the next session while it has been decided at that moment: I was very happy when I was told "now we are preparing for the blood sampling". And then when I went out, I felt good and I felt strong.
L: Did you have all your questions answered to your satisfaction?
B: I had no questions actually because I was quiet. Dr D. actually didn't ask me if I had any other questions. It was all very fast.
L: How did you feel when they offered you the genetic test?
B: I was happy that they offered the test to me.
L: Did anything unexpected happen in the counselling session?
B: No. Dr D. is a very good person in every sense. All has made like I wanted.
L: After the consultation have you discussed your decision of having genetic test with anyone else?
B: After the consultation I went out with my mother, who was present during the counselling. When I talked with my aunt and with my sister, I felt very happy and calm. I was told "you did very well". I have to thank you, at the beginning I just wanted to give the blood sample and go home, but thanks to our chat and the counselling now I feel more prepared and I feel better. This pathway has helped me much. Thank you, again.
L: Thank you too. See you in six months for the last interview.

Interview 3

L: Can you tell me what has happened in this period?
B: After two months I received the result. Sincerely when they called me to tell me that the result was ready, I wasn't so quiet, but I tried to think positive, as usual. I went to the hospital with my mom. On the way, she told me " Barbara be quiet, I'm sure you don't have it". I thought so, too. Anyway, whatever the result, never give up. Then, the result moment came. I looked the doctor in the eyes and I understood that I have the mutation. I was quiet and positive, I thought " I'm not alone". I have a very good mother who does not leave me alone.
L: How did you feel when you were given the result?
B: At that moment I was not quiet, a bit worried. But I told myself that I had to move on. Since I have got the result, I'm constantly thinking about the implications, but always in a positive way. I also started to have surveillance and everything went well.

L: Can you tell me more about your experience?
B: When the doctor told me that I have the BRCA gene with mutation, it was really ... there are no words to explain what I felt. She was direct and friendly, I still have in my eyes that expression. I'm glad we have faced this situation with people like that. Moreover, she explained me that I should not worry because there are women who get to 70 and older without having developed any cancer. I hope it's the same for me. Anyway, I have examinations every six months. Some fear exists, especially when I'm approaching examinations. Recently I had my first gynaecological exam and it went very good, the doctor told me that everything was fine. I felt like a butterfly.
L: And what about the test?
B: As for the test I'm very glad that I made this choice. We are young and we are not very careful with the lifestyle: with the food, smoking, drinking. Now I know so many things, I'm reading and I can tell. Now I feel an adult and I feel I have made the right choice.
L: Can you tell me if anyone has influenced your decisions/ thoughts?
B: My mother pushed me a little bit to do so. She said, "Barbara you must be strong and determined, if you do not feel ready, we can also wait a few years until you feel prepared. Nobody forces you to do the test right now". She had told me that when you have this kind of information you have to live with this. So you need to be psychologically prepared for this. Nobody of my friends knows (what I'm doing) because I think these are very personal things and, knowing my friends, I'm afraid that some of them might think badly and then I would feel bad. I prefer keeping the thing for my close relatives and myself. When I got the result, I spoke with two or three people. But I feel strong and so far all is gone well. But I prefer not to talk about it, it's my thing and that's that.
L: How did the friends you have told react?
B: Amazed. Very sorry. How many words...
L: Do you think that there is a specific age to get tested? It is hard to say. Perhaps 18 years is too young to deal with the result. It would be too heavy. I think that it depends also on what kind of cancer. Some tumours are diagnosed even when you're very young. For example, if I look at my family, cancer mainly occurred at about 50 years. I've done it at 29 years, so I have time to do the surveillance. If my family were sick before, I would have definitely taken the test before. We have to look at the age of onset of cancer in the family to understand the better age to take the test.
L: What do you think about feelings that young people like you may have regarding genetic testing?
B: I think that young people see it just like a blood test. I first saw it like a blood test, but I truly understood (the meaning of all) only after dealing with counselling and questions they asked me. At first I was not very happy, when I lived the clinic but I definitely wanted to do it.
L: Is there any advice you would give to help the genetic service to improve the service for young people?
B: To me it would be very helpful if there was a group within you can talk to get information. I used the Internet to search for information. Sharing information with someone else who is facing or has already faced the same experience could be useful especially for young people, because a young person is curious. Such a group could also help people who have difficulties to speak about the diagnosis, in order to let them come out and feel better. But also for me: I have friends, but I'm not at ease talking with them about my emotions and my fears, while it would be easier to talk with people experiencing your same emotions. Maybe they understand better.
L: Good idea! Thank you for answering at all three interviews.

APPENDIX 8 ENGLISH VERSION OF ALL THREE INTERVIEWS CONDUCTED WITH A YOUNG MAN (MARIO)

Interview 1

ABOUT YOU

AGE: 26 YEARS OLD

GENDER:

þ MALE
□ FEMALE

WHAT IS YOUR HIGHEST EDUCATIONAL QUALIFICATION?

□ NO FORMAL EDUCATION
□ ELEMENTARY SCHOOL QUALIFICATION
□ MIDDLE SCHOOL QUALIFICATION
þ HIGH SCHOOL DIPLOMA
□ UNIVERSITY DEGREE
□ POST-GRADUATE DEGREE

WHAT IS YOUR DAILY WORK?

□ UNEMPLOYED
□ STUDENT
□ HOMEMAKER
þ WORKER
□ EMPLOYEE
□ MANAGER
□ FREELANCE
□ ARTISAN
□ BUSINESS OWNER
□ SERVICE PERSON
□ OTHER _____

MARRIED STATUS

þ UNMARRIED
□ MARRIED
□ DIVORCED
□ WIDOWED
□ LIVING TOGETHER
□ OTHER _____

DO YOU HAVE ANY CHILDREN?

□ YES
þ NO

~~**IF YES, HOW MANY CHILDREN DO YOU HAVE?**~~

~~**HOW MANY DAUGHTERS DO YOU HAVE?**~~

~~**HOW MANY SONS DO YOU HAVE?**~~

ARE YOU PREGNANT?

□ YES
þ NO

COUNTRY OF BIRTH:

CROTONE, ITALY

LINGUA MADRE : ITALIAN

ABOUT YOUR PARENTS:

FATHER

WHAT IS YOUR HIGHEST EDUCATIONAL QUALIFICATION?

□ NO FORMAL EDUCATION
□ ELEMENTARY SCHOOL QUALIFICATION
þ MIDDLE SCHOOL QUALIFICATION
□ HIGH SCHOOL DIPLOMA
□ UNIVERSITY DEGREE
□ POST-GRADUATE DEGREE

WHAT IS YOUR DAILY WORK?

□ UNEMPLOYED
□ STUDENT
□ HOMEMAKER
□ WORKER
þ EMPLOYEE
□ MANAGER
□ FREELANCE
□ ARTISAN
□ BUSINESS OWNER
□ SERVICE PERSON

MOTHER

WHAT IS YOUR HIGHEST EDUCATIONAL QUALIFICATION?

□ NO FORMAL EDUCATION
□ ELEMENTARY SCHOOL QUALIFICATION
þ MIDDLE SCHOOL QUALIFICATION
□ HIGH SCHOOL DIPLOMA
□ UNIVERSITY DEGREE
□ POST-GRADUATE DEGREE

WHAT IS YOUR DAILY WORK?

□ UNEMPLOYED
□ STUDENT
þ HOMEMAKER
□ WORKER
□ EMPLOYEE
□ MANAGER
□ FREELANCE
□ ARTISAN
□ BUSINESS OWNER
□ SERVICE PERSON

L: Okay, now we have finished collecting information about you and your family. Let's go on with the interview.
B: Ready!
L: How did you reach the decision to make an appointment for genetic counselling?
M: Both my mother and my grandmother have been diagnosed with cancer and have undergone specific analyses. The genetic service has suggested that also the children should undergo the same analyses to see if they were carrier. So, here I am.
L: So you've booked the counselling session because you've been told by others, haven't you?
M: Yes, I have, at least at the beginning. But now, I also think of my health. To know if I'll have to deal with what my mother, my aunts and grandmother have faced. To be able to take in time what lately might be life-threatening.
L: Can you tell me more about your motivation to make an appointment?
M: My health. I describe myself like an pragmatic person, so I like to do the right thing at the right time. Being certain of something I may develop, makes me more comfortable if I would have children or a specific job. If I were sure to have an increased risk of develop cancer, I will avoid working in a farm where one is exposed to dangerous material.
L: Do you know what happens during genetic counselling?
M: I think it consists of a blood sample and a short interview. I think that a doctor will explain something to me and at the same time I will undergo the test.
L: What do you expect to obtain from genetic counselling?
M: Actually, I have no expectations. I'm just waiting for the results. I'll face whatever will be. I have decided not to have expectations because I don't want to imagine something different from the reality.
L: How do you feel talking about your family history of cancer?
M: I feel sad. It's not easy deal with this family story. You don't know if you can understand what your relative with cancer feels like. You don't have certainties about what is going on, you live expecting that something happen. So you wait and you hope everything is fine. I feel different emotions at the same time: sadness, anger and resignation. I'm sad because I have to see the people next to me, with whom I grew up and shared many beautiful moments, who are suffering and you can do nothing. You feel angry as you cannot actually do anything except being with them when they do examinations. You also feel disheartened because the problem already exists and you have just to face it.
L: Have you already been thinking about genetic testing?
M: Not much. I try not to think about the test or other related things. I just have to have the test, so let's do it.
L: Would you like to tell me more about who told you that "you have to" do this thing?
M: My father, my mother and my aunts. They said, "Your mother has this medical problem and, in theory, you should be tested, too. Clearly, the decision is yours". I said " if I must have the test, I'll have it. There is no any other possibility."
L: There may be the possibility not to have the test ...
M: No, there isn't. If it gives me a certainty, I will absolutely do it. I don't want to see myself in the situation of ignoring something, living with the anxiety, fearing what might be. If you are aware of the risk, you know that the problem is quite real and so you can start having specific examination. Thanks to this information, you could develop healthy behaviours in order to prevent cancer.

L: Can you tell me what you know about genetic testing?
M: A blood sample. The laboratory counts the percentage of the mutant cells in the blood.
L:What would you like to understand better?
M: I've always thought: I go there and I'll listen to what they have to tell me. I haven't thought about more details I could need.
L: What are your feelings about genetic testing?
M: Resignation: I have to do it for myself and for a more calm future. I don't have any specific expectations.
L: Have you spoken with, or are you going to speak with anyone else (such as family or friends) about genetic testing? If so, what do they think?
M: I became aware of the diagnosis in my family in a casual way, just bringing some documents to the doctor on behalf of my mother. Then I spoke with my closest friends, but this issue is quite a taboo in the small town where I live. The 80% of the population in my area is at risk of developing cancer, but none wants to talk about that. It's really a taboo, when people talk about this issue everyone turns away the other way because there is a lot of fear among young people. And also I'm a reserved person, I don't like to share it with my friends.

Interview 2

L: Can you tell me what happened during your genetic counselling session?
M: I entered the room and there was a female doctor with two co-workers. The doctor started explaining to me what this genetic is, and how it could affect me, then explained the privacy policy, and that further research could be done on this issue. Doctor made me feel comfortable. Thanks to her I've understood the issue better, initially I thought it was more like a genetic problem that could affect only me, while my children couldn't inherit it. Although the pathogen gene contaminated the female organs, I thought my mom could have transmitted to me the pathogen gene and it could have contaminated every single organ. At the beginning I was very confused. Now I know that the test will be useful in the future for my potential daughters. Regarding me, they said that I have to do the standard surveillance but starting it at age 40 rather than 50.
L: How did you feel during the consultation?
M At first, there was a big question mark on my head about what they would do and would say. Then those questions dissolved as soon as I started talking to the doctor because she put me immediately at ease and comfortable.
L: Have you had all your questions answered to your satisfaction?
M: Yes absolutely. Everything was explained to me softly, with a particular attention to the emotional aspects.
L: What do you mean by "softly"?
M: As I've already said last time, people working in an hospital like this, are different from those working in my area: here psychological aspects are something important, all the team try to make the patient feel comfortable immediately. So I can say that I was very satisfied. Where I live, when someone has to give you a news, good or bad one, it's not common that he sees you as a person, the psychological and emotional aspect are not really considered. Here, it seems that there is more attention for the psychological than the physical side. I don't mean they do not care

about the body, but the psychological side is treated more. This fact was the most surprising to me.
L: What you mean? Could you explain me more?
M: I was expecting something more direct, rude. I was expecting fewer smiles, less kindness. It is totally different instead: it was all explained in detail, I could understand all the aspects and there is always someone who smiles at you. There was also a moment for jokes. This is the great thing, the difference between a small town like mine and big centres.
L: Do you think this attitude is more related to the hospital or to the particular subject?
M: I think it is related more to the hospital. The same consultation in my area would have been performed differently. Probably, where I live the consultation might not have been made, as over there we have a different attitude toward these issues. In my area the doctors usually say " thing are like this" and that's all. There is not a study behind, why this has happened and what could be done.
L: So, during the consultation did they offer you a genetic test?
M: Yes, obviously.
L: How did you feel at that moment?
M: I was satisfied and very happy.
L: Did anything unexpected happen in the counselling session?
M: Two things surprised me. First, as I said before, the moral aspect that I didn't expect to be so accentuated. Secondly a specific question that they have asked: "if some blood remained, can we use it for further research?". I was so happy and surprised that I told a joke saying " If you want you can take more blood. Here is my other arm!". This thing stunned me. Usually you do a test for yourself or for someone that is quite close to you and that's all. This case was different and it was a good thing because it helps the research and the will to understand what is not yet known. I'm happy to be part of the research, I'm grateful to help someone even if it turns out that there is a disease in me. Maybe I'm narcissistic but that's what I thought.
L: After the consultation, have you discussed about your decision of having a genetic test with anyone else?
M: Now, this kind of topics are quite normal in my family because all my maternal aunts are at risk: they did the surgery and they also did the blood sample. I sincerely don't want to share too much of my private life with my friends. None asks me more. But honestly it's better, I'm a reserved person.

Interview 3

L: Can you tell me what happened during this period?
M: After we spoke, I received a letter and I opened it. Apart from the first impression of the letter there was nothing (no emotions).
L: You had decided to undergo the test. What was the result?
M: Negative. It went quiete well, I'm not a carrier of the mutation.
L: Did you expect this?
M: No, I didn't.
L: How did you feel?
M: I tried to disguise the tension. It wasn't easy knowing that all my family has this genetic defect.
L: Tension related to what specifically?

M: The tension was linked to the result, the idea of opening that envelope and finding out the result made me very anxious. I felt the anxiety in all of my body. I think that it is quite normal to be anxious when you read something about your future. But let's call it tension rather than anxiety
L: And once you have discovered the result?
M: All the tension suddenly disappeared and I started laughing. I was so happy. My mother and my father too. It wasn't a nice thing to know to have this "baggage", we can call it this way.
L: Has someone influenced your choice or your thinking?
M: No. As soon as I knew there was this possibility I immediately said let's do it. I haven't been influenced or other.
L: Can you tell me your experience about genetic counselling and genetic testing?
M: I was expecting something more complicated. It consisted in a talk with a doctor and a blood test, nothing more. I was expecting a more complicated test. All this experience was quite easy thanks to the ability of the doctor to make me feel comfortable, to laugh and joke with me. I didn't expect something so calm. As I said last time, the place where I live is different, it's not like a big hospital. Everything you do there, makes you anxious. Here all the people are able to put you at ease.
L: What do you think other young people like you can feel about a genetic test?
M: This is a very interesting question. Talking with other people who have a family history like mine (parents or relatives with cancer) I had some difficulties, I found talking about genetic heritage extremely complicated, it's a misunderstood topic. I understood that is a simple thing, other people felt afraid when they listened about it. Especially young people. They were afraid that their lives could change radically. They did not even want to hear.
L: At the end of our interviews, have you any advice for us to improve our cancer genetic counselling service, particularly for young people?
M: No, because there is no need to give tips to improve the service. The service is already excellent. I felt good, I had a good impressions and the doctors gave me just the right directives. I felt welcome and I think this is the most important thing both for a young man and for any other person. Really, I don't have any advice because it's a very good service. Being at one's ease is a great feeling that gives people space to open up and experience problems in a calmer way.
L: Thank you Mario for answering to my questions and thank you for your time.

APPENDIX 9 ITALIAN VERSION OF ALL THREE INTERVIEWS CONDUCTED WITH A YOUNG WOMAN (BARBARA)

Intervista 1

CARATTERISTICHE SOCIO-DEMOGRAFICHE DELL'INTERVISTATO

ETÀ: 29 ANNI

SESSO:

□ MASCHILE
þ FEMMINILE

TITOLO DI STUDIO:

□ NESSUNA EDUCAZIONE FORMALE
□ LICENZA ELEMENTARE
□ LICENZA MEDIA
þ DIPLOMA DI SCUOLA MEDIA SUPERIORE
□ LAUREA
□ SPECIALIZZAZIONE POST LAUREA

LAVORO:

□ DISOCCUPATO
□ STUDENTE
□ CASALINGA
þ OPERAIO/A (PARRUCCHIERA)
□ IMPIEGATO/A
□ DIRIGENTE
□ LIBERO PROFESSIONISTA
□ ARTIGIANO/A
□ COMMERCIANTE
□ MILITARE
□ ALTRO(SPECIFICARE):

STATO CIVILE:

þ CELIBE/NUBILE
□ SPOSATO/A
□ SEPARATO/A O DIVORZIATO/A
□ VEDOVA/O
□ CONVIVENTE
□ ALTRO(SPECIFICARE): _____

HA FIGLI?

□ SI
þ NO

~~**SE SI, QUANTI FIGLI HA?** _____~~
~~**QUANTE FIGLIE FEMMINE?** _____~~
~~**QUANTE FIGLI MASCHI?** _____~~

- **É ATTUALMENTE IN GRAVIDANZA?**

□ SI
þ NO

CITTÀ/NAZIONE DI NASCITA:

KOZIENICE, POLONIA

VIVE IN ITALIA DA 10 ANNI.

LINGUA MADRE : POLACCO

CARATTERISTICHE SOCIO-DEMOGRAFICHE DEI GENITORI:

IL PADRE

TITOLO DI STUDIO:

□ NESSUNA EDUCAZIONE FORMALE
□ LICENZA ELEMENTARE
□ LICENZA MEDIA
þ DIPLOMA DI SCUOLA MEDIA SUPERIORE
□ LAUREA
□ SPECIALIZZAZIONE POST LAUREA

LAVORO:

□ DISOCCUPATO
□ STUDENTE
□ CASALINGA
þ OPERAIO/A (ELETTRICISTA)
□ IMPIEGATO/A
□ DIRIGENTE
□ LIBERO PROFESSIONISTA
□ ARTIGIANO/A
□ COMMERCIANTE
□ MILITARE
□ ALTRO(SPECIFICARE):

LA MADRE

TITOLO DI STUDIO:

□ NESSUNA EDUCAZIONE FORMALE
□ LICENZA ELEMENTARE
□ LICENZA MEDIA
þ DIPLOMA DI SCUOLA MEDIA SUPERIORE
□ LAUREA
□ SPECIALIZZAZIONE POST LAUREA

LAVORO:

□ DISOCCUPATO
□ STUDENTE
□ CASALINGA
þ OPERAIO/A (CUOCA)
□ IMPIEGATO/A
□ DIRIGENTE
□ LIBERO PROFESSIONISTA
□ ARTIGIANO/A
□ COMMERCIANTE
□ MILITARE
□ ALTRO(SPECIFICARE):

L: Okay, Finita le domande socio demografiche ora iniziamo con le domande centrali dell'intervista.
B: Sono pronta!
L: Prima cosa, perché hai deciso di prenotare una consulenza genetica? Puoi spiegarmi più dettagliatamente le motivazioni che ti hanno portata a questa decisione?
B: Ho deciso di prenotare la consulenza perché mia madre due mesi fa ha subito un intervento molto pesante per il tumore ovarico, ho passato la stessa esperienza con mia nonna dieci anni fa, anche mia zia. Io vorrei prevenire. In aria gira questa cosa. Anche potrei avere il gene. Io mi sento forte.
L: Sei a conoscenza di come si svolge una consulenza genetica?
B: Mi verrà fatto un prelievo di sangue e aspettare il risultato.
L: Quali sono le tue aspettative circa la consulenza genetica?
B: Se ho il gene come la mamma e la zia sono tranquilla, mi sento forte. So anche che se non ho questo gene comunque il tumore può venire lo stesso per altri motivi: inquinamento, alimentazione, stress.
L: Quali sono le emozioni/sensazioni che provi parlando della tua storia familiare di malattia?
B: Mi sento più preparata e mi sento tranquilla a parlarne perché l'ho vissuto. Mi sento più preparata anche per il prelievo del sangue. Mi sento più tranquilla.
L: Hai già avuto modo di pensare al test genetico?
B: Si ci ho già pensato al test genetico. Penso che mi verrà proposto. Mi sento preparata. Mi sento forte e bene. Io lo posso fare subito. Non mi spaventa. Anzi! Meglio saperlo prima piuttosto che fra qualche anno che poi mi viene e non sono preparata. Poi mi si forma dentro qualcosa e poi dopo … meglio prevenire, meglio sapere. È il mio carattere. Non siamo tutti uguali, qualcuno durante la malattia non vanno avanti… Io invece voglio vivere! Vorrei combattere.
L: Puoi spiegarmi quelle che sono le tue conoscenze sul test genetico?
B: Il test genetico mi dirà se ho il gene come mia madre oppure no.
L: Che cosa ti piacerebbe approfondire?
B: In questo momento mi sto informando su Internet per capire se è stato scoperto qualcosa per prevenire. Mi butto su questo tema. Cerco di capire per quale motivo si formano i tumori … e poi parlo tanto con gli amici. Mi piacerebbe anche sapere cosa effettivamente verrà fatto al mio sangue in laboratorio.
L: Quali sono le emozioni/sensazioni che provi pensando al test genetico per te stessa?
B: Io sono molto curiosa. Io ora guardo tutto positivo. Per me è tutto più semplice adesso. Quando arriverà questa risposta saprò solo allora come realmente mi sentirò o reagirò. Per il momento non so nulla. Dipende. cerco di essere forte. Per il momento mi sento forte, poi non so come reagirò. Sinceramente non ci voglio nemmeno pensare ora al test genetico. Non è facile. Ora so che lo voglio fare il prelievo di sangue e sapere. Sapere mi permetterà di prevenire. Anche se riceverò una risposta non positiva per me devo accettare e andare avanti. Devo fare tutto per stare bene e cerco di stare più attenta. Se invece non avrò il gene … respiro.
L: Hai già avuto modo, o lo farai in futuro, di parlare con qualcuno (per esempio familiari, amici etc) circa il test genetico? Se SI, che cosa ne pensano? Cosa ti spinge verso questa scelta?
B: Non ho parlato del test genetico con amici. È una cosa mia. È una cosa familiare. Non tutti devono sapere tutto di me. Ne ho parlato solo con i parenti. Per esempio ne

sto parlando con le mie sorelle e fratello che anche loro in Polonia lo stanno facendo. Mio fratello è medico e mi spinge a fare il test perché dice che è la cosa giusta. Ma sarà veramente la cosa giusta? La mia mamma invece quando le ho detto che volevo il fare il test mi ha detto "Barbara sei sicura? Non si torna più indietro poi. Poi ci dovrai convivere. Sei sicura che non vuoi aspettare ancora qualche anno?". Io sono curiosa e voglio sapere. Se so posso poi fare qualcosa per prevenire.
L:Grazie per aver risposto alle mie domande e del tempo dedicatomi. Se sei d'accordo, proseguiremo la nostra intervista dopo la consulenza genetica.
B: Okay! Alla prossima

Intervista 2

L: Mi puoi raccontare cosa è successo durante la consulenza genetica?
B: Non mi ricordo tanto in realtà di quello che è successo. Mi ricordo che mi hanno consegnato tre fogli da leggere per me stessa e poi finalmente hanno deciso di prelevarmi il sangue. Non abbiamo parlato tanto in verità.
L: Quali sono le emozioni/sensazioni che hai provato durante il consulto?
B: Durante la consulenza non ero spaventata per niente. Ero decisa al 100% che volevo fare il test. Sinceramente non mi aspettavo che il test me lo facessero durante il primo colloquio. Pensavo che la dott.ssa D. avrebbe deciso prossimamente se farmi fare o no il test. Invece è stato deciso subito: ero molto contenta quando mi è stato detto ad un certo punto "ora ci prepariamo per il prelievo di sangue". E poi quando sono uscita mi sentivo bene, mi sentivo forte.
L: Tutte le tue domande hanno trovato risposta? Ne sei rimasto soddisfatto?
B: Non ho fatto nessuna domanda in realtà perché ero tranquilla. La dott.ssa poi in realtà non mi ha chiesto se avevo domande, è stato tutto molto veloce.
L: Durante la consulenza genetica ti hanno offerto il test genetico? Come ti sei sentita in quel momento?
B: Durante la consulenza genetica mi hanno offerto il test genetico e volevo che andasse tutto così.
L: È successo qualcosa di inaspettato durante la consulenza?
B: No. Non è successo nulla. La dott.ssa D. è una persona molto brava in tutti i sensi. Volevo veramente che andasse tutto così.
L: Dopo la consulenza, hai deciso di condividere con qualcuno la scelta di sottoporti o no al test genetico?
B: Dopo la consulenza sono uscita con mia madre che era presente durante la consulenza. Ho parlato poi con mia zia e mia sorella. Mi hanno detto "hai fatto benissimo!". Loro sono anche molto felici che io stia facendo un percorso di preparazione al test e non subito e solo il prelievo di sangue. All'inizio in realtà io volevo solo fare il prelievo e di sangue e basta. Ora sono molto felice invece che il percorso preveda degli spazi di chiacchere perché aiuta veramente tanto a livello psicologico. Anche quando sono da sola, ora ho gli strumenti per pensare. Grazie, veramente grazie.
L: Grazie per aver risposto alle domande. Se è d'accordo, concluderemo la nostra intervista fra sei mesi.

Intervista 3

L: Mi puoi raccontare cosa è successo durante questo periodo?

B: Dopo due mesi ho ricevuto il risultato e sinceramente quando mi hanno chiamata per dirmi che era pronto il risultato non ero tanto tranquilla, però ho sempre pensato positivo. Sono andata con mamma al mio fianco e andando verso la clinica mia madre mi diceva "Barbara tranquilla, non ce l'hai". Anche io credevo così. Poi qualunque fosse stato il risultato non bisogna mollare mai. Ma poi è arrivato il risultato. Ho guardato la dottoressa negli occhi e ho capito che c'era la mutazione. Ero tranquilla e positiva però ho pensato che non ero da sola. Ho una bravissima mamma che non mi lascia da sola.
L: Come ti sei sentita quando ti hanno dato l'esito?
B: In quel momento non ero tranquilla, un po' preoccupata. Però mi sono detta che dovevo andare avanti. Da quando mi hanno dato il risultato, ci sto pensando, ma sempre in positivo. Ho già iniziato anche a fare i controlli ed è andato tutto bene.
L: Mi puoi raccontare la tua esperienza circa la consulenza genetica ed il test genetico?
B: Quando la dottoressa mi ha spiegato che ho il gene BRCA, è stata veramente … non ci sono patole. È stata veramente bravissima. Anche per dirmi il risultato non ha aspettato, è stata diretta e accogliente. Ce l'ho ancora davanti i miei occhi questa espressione. Sono contenta di aver affrontato questo periodo con delle persone così. Poi mi ha anche spiegato che non devo preoccuparmi perché ci sono donne che arrivano a 70 anni e oltre ma che non si sono mai ammalate, però ogni 6 mesi sono sotto controllo. Mi ha detto che "devo stare tranquilla". Io ho la speranza che anche per me sia così. La paura c'è. Sono andata a fare il primo controllo ginecologico e appena è finito e la dottoressa mi ha detto che andava tutto bene, sono uscita come una farfalla. Non posso dire che non ci sia paura quando si affronta il controllo. Ma è una paura che si limita al controllo.
L: Per ciò che riguarda il test?
B: Per ciò che riguarda il test sono molto contenta che ho fatto questa scelta. Siamo giovani e non siamo molto attenti con gli ingredienti, con il cibo, fumare, bere. Adesso so tante cose, sto leggendo e mi informo. Ora mi sento adulta e sento di aver fatto la scelta giusta.
L: Qualcuno ha in qualche modo influenzato la tua scelta/ il tuo pensiero?
B: Mia madre un pochino mi ha spinto a farlo. Mi ha detto "Barbara devi essere forte e decisa, se non ti senti meglio che non lo fai. Possiamo aspettare anche fra qualche anno quando ti senti. Non è detto che si debba fare in questo momento". Lei mi aveva spiegato che poi ci dovevo convivere con la risposta e devi essere psicologicamente pronta per avere questo risultato. Però dei miei amici non lo sapeva nessuno perché secondo me queste cose sono molto personali e conoscendo le amiche ed i miei amici, qualcuno potrebbe pensare proprio male e poi dopo anche io ci sto male. Ho preferito tenere per me la cosa. Poi quando ho ricevuto il risultato a due tre persone l'ho detto. Io sono forte e per ora va tutto bene. Però preferisco non parlarne, è una mia cosa e basta.
L:Questi due tre amici ai quali hai raccontato la cosa come l'hanno presa?
B: Stupiti. Molto dispiaciuti. Quante parole…
L: Pensi che esista un'età specifica per fare il test? È difficile dirlo. Forse a 18 anni si è troppo giovani per affrontare il risultato. Sarebbe troppo pesante. Poi bisogna vedere di che tumore parliamo. Alcuni tumori vengono anche quando si è molto giovani. Io per esempio se guardo la mia famiglia, l'età più a rischio è sui 50 anni. L'ho fatto a 29 anni e ho tempo per fare i controlli. Se nella mia famiglia si fossero

ammalate prima, avrei fatto il test sicuramente prima. Bisogna guardare l'età di insorgenza dei tumori nella famiglia per capire l'età in cui fare il test.
L: Che cosa ne pensi delle sensazioni che persone giovani con te possono percepire circa un test genetico?
B: Penso che il giovane lo veda come un prelievo di sangue. Io per prima lo vedevo come prelievo di sangue, ma solo dopo aver affrontato la consulenza e le domande che mi hanno fatto, ho capito veramente. All'inizio non ero molto contenta dopo essere uscita dalla porta, però avevo voglia di farlo.
L: A conclusione della nostra intervista, hai qualche consiglio da darci per migliorare il nostro servizio di consulenza genetica oncologica, in particolare per i giovani?
B: Per me sarebbe molto utile che ci fosse un gruppo nel quale parlare per informarsi. Io ho guardato su Internet per cercare informazioni. Quindi confrontarsi con qualcuno che sta vivendo le stesse cose o le ha già vissute potrebbe essere utile per un giovane perché un giovane vuole sempre conoscere. Poi c'è chi non si apre e questo gruppo potrebbe essere utile per quelle persone. Ma anche per me, per esempio. Io con le mie amiche non posso raccontare le mie emozioni e le mie paure, invece confrontarsi con persone che vivono le tue stesse emozioni è più facile. Forse ti capiscono meglio.
L: Bella idea! Grazie per aver risposto alle domande e del tempo che mi hai dedicato in questo periodo!

APPENDIX 10 ITALIAN VERSION OF ALL THREE INTERVIEWS CONDUCTED WITH A YOUNG MAN (MARIO)

Intervista 1

CARATTERISTICHE SOCIO-DEMOGRAFICHE DELL'INTERVISTATO

ETÀ: 26 ANNI

SESSO:

þ MASCHILE
□ FEMMINILE

TITOLO DI STUDIO:

□ NESSUNA EDUCAZIONE FORMALE
□ LICENZA ELEMENTARE
□ LICENZA MEDIA
þ DIPLOMA DI SCUOLA MEDIA SUPERIORE
□ LAUREA
□ SPECIALIZZAZIONE POST LAUREA

LAVORO:

□ DISOCCUPATO
□ STUDENTE
□ CASALINGA
þ OPERAIO
□ IMPIEGATO/A
□ DIRIGENTE
□ LIBERO PROFESSIONISTA
□ ARTIGIANO
□ COMMERCIANTE
□ MILITARE
□ ALTRO(SPECIFICARE): _____

STATO CIVILE:

þ CELIBE/NUBILE
□ SPOSATO/A
□ SEPARATO/A O DIVORZIATO/A
□ VEDOVA/O
□ CONVIVENTE
□ ALTRO(SPECIFICARE): _____

HA FIGLI?

□ SI
þ NO

~~**SE SI, QUANTI FIGLI HA?** _____~~
~~**QUANTE FIGLIE FEMMINE?** ____~~
~~**QUANTE FIGLI MASCHI?** _____~~

~~**É ATTUALMENTE IN GRAVIDANZA?**~~
~~□ SI~~
~~þ NO~~

CITTÀ/NAZIONE DI NASCITA:

CROTONE, ITALIA

LINGUA MADRE : ITALIANO

CARATTERISTICHE SOCIO-DEMOGRAFICHE DEI GENITORI:

IL PADRE

TITOLO DI STUDIO:

□ NESSUNA EDUCAZIONE FORMALE
□ LICENZA ELEMENTARE
þ LICENZA MEDIA
□ DIPLOMA DI SCUOLA MEDIA SUPERIORE
□ LAUREA
□ SPECIALIZZAZIONE POST LAUREA

LAVORO:

□ DISOCCUPATO
□ STUDENTE
□ CASALINGA
□ OPERAIO/A (ELETTRICISTA)
þ IMPIEGATO/A
□ DIRIGENTE
□ LIBERO PROFESSIONISTA
□ ARTIGIANO/A
□ COMMERCIANTE
□ MILITARE
□ ALTRO(SPECIFICARE):

LA MADRE

TITOLO DI STUDIO:

□ NESSUNA EDUCAZIONE FORMALE
□ LICENZA ELEMENTARE
þ LICENZA MEDIA
□ DIPLOMA DI SCUOLA MEDIA SUPERIORE
□ LAUREA
□ SPECIALIZZAZIONE POST LAUREA

LAVORO:

□ DISOCCUPATO
□ STUDENTE
þ CASALINGA
□ OPERAIO/A (CUOCA)
□ IMPIEGATO/A
□ DIRIGENTE
□ LIBERO PROFESSIONISTA
□ ARTIGIANO/A
□ COMMERCIANTE
□ MILITARE
□ ALTRO(SPECIFICARE):

L: Okay, Finita le domande socio demografiche ora iniziamo con le domande centrali dell'intervista.
M: Pronto!
L: Perché hai deciso di prenotare una consulenza genetica?
M: Qualche anno fa hanno riscontrato a mia madre un tumore e sono stati fatte tutte le cose del caso. Qualche anno dopo, anche a mia nonna hanno riscontrato un tumore. Sotto consiglio dell'ospedale Sant'Orsola hanno detto ai figli di sottoporsi all'esame per vedere se anche loro erano portatori. Essendo anche mia madre portatrice hanno detto che anche i figli dovrebbero sottoporsi a questo esame. E allora eccomi qui.
L: Quindi hai prenotato la consulenza perché ti è stato detto da altri?
M: Si. Inizialmente si. Ora anche per una sicurezza mia. Per sapere se anche io dovrò affrontare quello che ha affrontato mia madre, mia nonna, le mie zie. Per poter prendere in tempo ciò che potrebbe essere poi troppo tardi.
L: Puoi spiegarmi più dettagliatamente le motivazioni che ti hanno portato a questa decisione?
M: La sicurezza. Sono una persona schematica. Mi piace fare le cose giuste nei tempi giusti. Avere la certezza su qualcosa che potrei avere mi porterebbe ad essere più tranquillo sia un domani se dovessi avere dei figli, un determinato lavoro. Se sono sicuro di avere una percentuale alta di probabilità tumorali, di certo no andrò a lavorare in una fabbrica con materiali di un certo livello.
L: Sei a conoscenza di come si svolge una consulenza genetica?
M: Penso che si tratti di un prelievo ed un piccolo colloquio. Penso che un medico mi spiegherà qualcosa e contemporaneamente si farà l'analisi.
L: Quali sono le tue aspettative circa la consulenza genetica?
M: Non ho aspettative in verità. Aspetto solo che mi diano l'esito. Quello che viene, affronterò. Ho comunque deciso di non farmi aspettative perché non voglio illudermi che una cosa sia come ho pensato io.
L: Quali sono le emozioni/sensazioni che provi parlando della tua storia familiare di malattia?
M: Tristezza. Tutte queste cose in una sola famiglia non è facile. Si può avere un piccolo conforto fra le persone che hanno questa patologia. La restante parte della famiglia però non può capire come si sente il paziente. Quindi tu stai li ad aspettare un qualcosa che sai che oggi c'è e che un domani potrebbe essere ancora peggio. Quindi aspetti con ansia per vedere come si svolgerà la cosa, sperando sempre nel meglio. E poi dipende sempre dagli esiti. Se dovessi elencare le mie emozioni sono tristezza, rabbia e rassegnazione. Tristezza perché vedi persone accanto a te, con cui sto crescendo e ho passato tutti i momenti belli della vita, che stanno soffrendo e tu non puoi fare nulla, rabbia perché non puoi materialmente fare niente se non accompagnare il familiare ai vari controlli, rassegnazione perché oramai si sa che c'è e bisogna affrontarlo.
L: Ha già avuto modo di pensare al test genetico?
M: Non molto. Non mi sto facendo domande. Come viene viene. La cosa è che si deve fare questa cosa, quindi facciamola!
L: Ti andrebbe di spiegarmi meglio a chi ti riferisci quando mi dici che ti hanno detto che "devi" fare questa cosa?
M: Un po' mio padre, mia madre e le mie zie. Hanno detto "visto che tua madre è così, in teoria dovresti fare questo. È chiaro che però la decisione è tua". Io ho risposto "se devo farlo, lo faccio. Non c'è altra possibilità".

L: Potrebbe esserci la possibilità di non farlo …
M: Non esiste questa possibilità perché io sono schematico. Se mi da una certezza in più io lo faccio. Non voglio trovarmi più nella situazione di non sapere se c'è qualcosa e poi sentirsi male ed andare in ospedale e sentirsi dire cosa c'è. E quindi stare con l'ansia e la paura di chissà che cosa è. Se invece lo sai, sai che al 90% è quello il problema ed allora inizi a fare i controlli specifici prima. E poi magari cerchi di avere meno contatti con cose che ti possono portare a sviluppare la malattia come per esempio il lavoro, come dicevo prima.
L: Puoi spiegarmi quelle che sono le tue conoscenze sul test genetico?
M: Un prelievo di sangue ed il risultato che viene fatto attraverso l'analisi delle cellule malate, o meglio di cellule mutanti. La percentuale che potrebbe esserci di queste cellule nel sangue.
L: Che cosa ti piacerebbe approfondire?
M: Ho sempre pensato: vado li e vedo cosa mi devono dire. Non ho pensato nello specifico a cose che vorrei approfondire.
L: Quali sono le emozioni/sensazioni che provi pensando al test genetico per se stesso/a?
M: Rassegnazione: lo devo fare per me stesso e per un futuro più tranquillo. Non spero ne nel bene ne nel male.
L: Hai già avuto modo, o lo farai in futuro, di parlare con qualcuno (per esempio familiari, amici etc) del il test genetico?
M: Sono entrato a conoscenza di questa cosa nella famiglia perché sono stato io a portare ai medici la liberatoria. Proprio così sono entrato a conoscenza della sua malattia. Io però ne ho parlato anche con i miei amici più intimi. L'argomento è tabù per una realtà dove vivo io. L'80% della popolazione del mio paese è a rischio di sviluppare tumori. È veramente un argomento tabù, quando si parla di questa cosa tutti girano lo sguardo e si girano da un'altra parte perché c'è molta paura tra i giovani. I familiari invece sono ben consapevoli. Poi sinceramente sono anche io che non voglio approfondire più di tanto con i miei amici sulla mia vita privata, sono un tipo riservato.
L:Grazie per aver risposto alle mie domande e del tempo che mi hai dedicato. Se sei d'accordo, proseguiremo la nostra intervista dopo la consulenza genetica.
B: Okay! Alla prossima

Intervista 2

L: Mi puoi raccontare cosa è successo durante la consulenza genetica?
M: Sono entrato nella saletta con la dottoressa e due assistenti. La dottoressa ha iniziato a spiegarmi che cosa fosse questa genetica, che cosa comportava, l'informativa sulla privacy, che c'era la possibilità di studiare altre cose. Mi ha messo molto a mio agio. Io pensavo che fosse più una cosa genetica che potevo avere solo io e non che potessi trasmettere ai miei figli. Cioè io pensavo che mia madre mi trasmetteva il gene patogeno e questo gene patogeno poteva collegarsi ad ogni mio organo e non fosse riferito a quell'organo femminile. Questa cosa non mi era ben chiara all'inizio. Mi è stato invece chiarita questa cosa: più che per me è per, in un futuro prossimo, dovessi avere delle figlie femmine. Per me invece mi hanno solo detto che devo fare i controlli standard, ma iniziarli a 40 anni piuttosto che 50.
L: Quali sono le emozioni/sensazioni che hai provato durante il consulto?

M: All'inizio avevo un grande punto interrogativo sulla testa: che cosa faranno che cosa mi diranno. Poi questo punto interrogativo si è dissolto non appena ho iniziato a parlare con la dottoressa perché mi ha messo subito a mio agio, mi ha messo comodo.
L: Credi che tutte le tue domande hanno trovato risposta? Ne sei rimasto soddisfatto?
M: Assolutamente si. Mi è stato messo tutto davanti in modo "soft". Tutte le cose mi sono state esposte con cura però curando il lato emotivo dell'analisi.
L: Cosa intendi per "modo soft"?
M: Guardando il lato psicologico della cosa, cioè come dicevo già l'altra volta, questo denota la differenza abissale fra la realtà della mia zona e un ospedale di quelle dimensioni: non è la stessa. Mentre li ti mettono subito a tuo agio, qui da noi purtroppo non è così. Sono quindi rimasto molto soddisfatto, se così si può dire. Qui nella mia realtà quando ti devono dare delle notizie, belle o brutte, non viene considerato il lato psicologico ed emotivo della persona. Mentre li viene curato più il lato morale che quello fisico. Non che non venga curata la persona, intendiamoci, ma viene curato di più il lato psicologico. Questa è poi la cosa che mi ha lasciato più di stucco.
L: Cioè? Me ne puoi parlare?
M: Mi aspettavo una cosa più cruda. Abituato a delle realtà che sono totalmente diverse, mi aspettavo qualcosa di più crudo senza sorrisi, senza spiegazioni o con spiegazioni sommarie. Invece è totalmente diverso: mi è stata spiegata la cosa nel dettaglio, nel dettaglio che potevo capire e sempre con il sorriso. C'era il momento della battuta della serietà. Questa è la cosa bella, la differenza sostanziale di un piccolo centro e di un grande centro.
L: Questo atteggiamento lo riconduci più all'ospedale come centro o all'argomento trattato?
M: All'ospedale come centro. La stessa consulenza nel mio paese, sarebbe stata fatta con un atteggiamento diverso. Anzi, forse non sarebbe proprio stata fatta perché c'è una cultura diversa su questi temi. Qui non c'è la mentalità di studiare una cosa: qui ti dicono: "è così" punto e basta. Non c'è uno studio dietro sul perché è successo questo e cosa si potrebbe fare.
L: Durante la consulenza genetica ti hanno offerto il test genetico?
M: Si, ovviamente si.
L: Come ti sei sentito in quel momento?
M: Io ero soddisfatto e molto contento.
L: È successo qualcosa di inaspettato durante la consulenza? Se si, me ne pui parlare?
M: Le cose che mi hanno stupito sono due. La prima, come dicevo prima il lato morale della cosa che non mi aspettavo così accentuato. In secondo luogo una domanda specifica che mi hanno fatto, cioè "nel caso in cui dovesse rimanere un po' del sangue prelevato, se era possibile studiarlo per altro patologie di nuova scoperta". Li per lì sono rimasto contento, perché ero contento, e ho fatto anche la battuta "se volete qui c'è l'altro braccio, prendete pure dell'altro sangue!". Mi ha lasciato molto di stucco questa cosa, perché di solito uno fa un prelievo, studiamo questa cosa e poi basta, quello che resta lo buttiamo. Invece in questo caso no, è stata una bella cosa perché stimola la ricerca ed il voler sapere quello che ancora non si conosce. Sono felice di poter far parte della ricerca, per aiutare qualcuno

magari se si scoprisse una malattia in me ed una cura. Magari sono megalomane, però questo è quello a cui ho pensato.
L: Dopo la consulenza, hai deciso di condividere con qualcuno la scelta di sottoporti o no al test genetico?
M: Ormai nella mia famiglia, questo è un argomento all'ordine del giorno. Perché tutte le sorelle di mia madre sono a rischio, hanno fatto l'intervento ed il prelievo. Con amici non ne ho parlato, ho solo detto che avrei fatto questa visita. Non approfondito perché nessuno ha chiesto, ed è meglio così. Sono una persona riservata.
L: Grazie per aver risposto alle domande. Se sei d'accordo concluderemo la nostra intervista fra sei mesi.

Intervista 3

L: Mi puoi raccontare cosa è successo durante questo periodo?
M: Da quando abbiamo parlato l'ultima volta mi è arrivata la lettera e ho aperto la busta. A parte la prima impressione della lettera non c'è stato nulla (nessuna emozione).
L: Tu avevi deciso di sottoporti al test. Qual è stato l'esito del test?
M: Negativo. È andato bene, non risultato portatore della mutazione.
L: Te lo aspettavi?
M: No, non me lo aspettavo.
L: Come ti sei sentito?
M: Sicuramente cercavo di smorzare la tensione perché non era una cosa facile sapendo che tutta la mia famiglia ha questo difetto genetico. Smorzavo la tensione per come mi era possibile.
L: Tensione legata a cosa nello specifico?
M: La tensione era legata al risultato: aprire la busta e scoprire il risultato. L'ansia che mi aspettavo è arrivata in quel momento. Anche perché è qualcosa legato al tuo futuro, l'ansia c'è sempre. Ma chiamiamola tensione più che ansia, era tensione.
L: Una volta scoperto il risultato?
M: Tutta la tensione che avevo è scesa di botto ed è scattata la risata nevrotica, la felicità di mia madre, di mio padre. Perché chiaramente non è una bella cosa sapere di avere questo bagaglio, chiamiamolo così.
L: Qualcuno ha in qualche modo influenzato la tua scelta/ il tuo pensiero?
M: No, questo no. Come ho saputo che c'era questa possibilità ho detto subito si facciamolo. Non sono stato influenzato o altro.
L: Mi puoi raccontare la tua esperienza circa la consulenza genetica ed il test genetico?
M: Mi aspettavo qualcosa di più particolare. Tra virgolette è stata una passeggiata perché è stato tutto guidato. Mi aspettavo un test più particolare, non una semplice analisi del sangue. Poi vedendo che si trattava solo di un prelievo di sangue e li le dottoresse mi hanno messo a mio agio, si scherzava e si rideva. Non pensavo fosse una cosa così tranquilla. Come ti ho detto l'altra volta, la realtà in cui vivo non è la stessa cosa, non è come un grande ospedale. Qui si fanno le cose che ti mettono loro l'ansia. Li invece no, è stato tutto molto cordiale. Mi hanno messo a mio agio.
L: Più in generale, che cosa ne pensi delle sensazioni che persone giovani come te possono percepire circa un test genetico?

M: Questa è una domanda molto interessante. Parlando anche con altre persone che hanno avuto in famiglia persone in cui è stato riscontrato un tumore, parlare di patrimonio genetico è stato tra virgolette frainteso. Mentre io quando ho visto che è una cosa semplice, le altre persone appena hanno sentito questo hanno avuto paura, almeno i ragazzi. Hanno avuto paura che la loro vita potesse cambiare radicalmente. Erano molti lascivi al discorso, facevano "orecchie da mercante" come si suol dire. Non volevano nemmeno sentire.
L: A conclusione della nostra intervista, hai qualche consiglio da darci per migliorare il nostro servizio di consulenza genetica oncologica, in particolare per i giovani?
M: No, perché non c'è bisogno di dare consigli per migliorare il servizio. Il servizio è già ottimo. Mi sono trovato bene, ho avuto delle buone impressioni e delle buone direttive da parte dei dottori. Mi sono sentito accolto e questa credo che sia la cosa più importante sia per un ragazzo sia per qualsiasi altra persona. Non c'è bisogno di dare consigli perché è già ottimo come servizio. L'essere messi a proprio agio è una sensazione bellissima che da spazio alle persone di aprirsi e vivere determinati problemi in modo più tranquillo.
L: Grazie per aver risposto alle domande e del tempo che mi hai dedicato.

APPENDIX 11 APPROVAL BY ST. ORSOLA-MALPIGHI ETHICAL COMMITTEE (13TH NOVEMBER 2015)

SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA
Azienda Ospedaliero - Universitaria di Bologna

Policlinico S. Orsola-Malpighi

Comitato Etico Indipendente dell'Azienda Ospedaliero-Universitaria di Bologna, Policlinico S. Orsola-Malpighi

Bologna, 13/11/2015
Prot. n. 3449/2015

Ns. riferimento da citare sempre nella corrispondenza:
n° 198/2015/O/Oss approvato il 13/10/2015

Chiar.mo Prof. Marco Seri
Direttore
U.O. Genetica Medica
Pad. 11 – S. Orsola
Policlinico S. Orsola-Malpighi

E p.c.

Gent.mo Dott. Primoz Juric
Investigational Drug Service
c/o U.O. Farmacia – Puggioli
Pad. 19 – S.Orsola
Policlinico S.Orsola-Malpighi

Gent.ma Dott.ssa Cristina Puggioli
Direttore
U.O. Farmacia – Pad. 19
Policlinico S.Orsola-Malpighi

Oggetto: 198/2015/O/Oss – Studio osservazionale "Studio quantitativo sull'impatto dei test genetici di predisposizione al cancro nei 'giovani adulti' e nei loro genitori" – Prot. PreGAGe – A.O.U. Policlinico S. Orsola-Malpighi di Bologna/U.O. Genetica Medica
Sperimentatore Responsabile: Dr.ssa D. Turchetti
Direttore: Prof. M. Seri

- Lettera di trasmissione inviata dallo Sperimentatore il 04/09/2015
- Richiesta Autorizzazione alla Direzione Generale dell'Azienda per la conduzione dello Studio osservazionale trasversale da parte del Promotore e Responsabile dello studio datata 04/09/2015
- Richiesta Parere per Studio osservazionale trasversale sottoscritta dallo Sperimentatore Responsabile e dal Direttore dell'Unità Operativa, datata 04/09/2015
- Protocollo di studio versione emendata del 04/09/2015
- Sinossi del Protocollo, versione del 04/09/2015
- Progetto Phd Dott.ssa Godino (Allegato 1)
- Questionario A (Allegato 2)
- Questionario B (Allegato 3)
- Foglio informativo per l'utente- Partecipazione allo studio, versione del 04/09/2015
- Foglio informativo per l'utente- Trattamento dati sensibili, versione del 04/09/2015
- Consenso informato- Partecipazione allo studio, versione del 04/09/2015
- Consenso informato- Trattamento dati sensibili, versione del 04/09/2015

COMITATO ETICO INDIPENDENTE DELL'AZIENDA OSPEDALIERO-UNIVERSITARIA DI BOLOGNA
POLICLINICO S. ORSOLA – MALPIGHI
Sede: Padiglione 3 – Via Albertoni, 15 – 40138 BOLOGNA
Tel.: 051/6361346 – 051/6361384 – Fax: 051/6361249
Indirizzo posta elettronica: cometico@aosp.bo.it
Sito WEB: http://www.aosp.bo.it/content/comitato-etico

SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA
Azienda Ospedaliero - Universitaria di Bologna

Policlinico S. Orsola-Malpighi

Comitato Etico Indipendente dell'Azienda Ospedaliero-Universitaria di Bologna, Policlinico S. Orsola-Malpighi

- Dichiarazione del proponente sulla natura osservazionale dello studio, sottoscritta dal Coordinatore il 04/09/2015
- Curriculum dello sperimentatore
- Scheda di raccolta dati – Giovane adulto, versione del 04/09/2015
- Scheda di raccolta dati – Genitore, versione del 04/09/2015
- Dichiarazione sulla natura no-profit dello Studio sottoscritta dallo Sperimentatore il 04/09/2015
- Dichiarazione di non ricevere alcun compenso per lo Studio, sottoscritta dallo Sperimentatore il 04/09/2015

Il Prof. Marco Seri esce temporaneamente dalla seduta.

Il Comitato Etico, nella seduta del giorno 13/10/2015, ritiene che il protocollo di studio ed i suoi obiettivi soddisfacciano i criteri etici e scientifici che ne giustificano l'esecuzione. Le modalità del consenso informato sono corrette ed adeguate allo scopo. Il Comitato, pertanto, esprime all'unanimità **parere favorevole** alla conduzione dello studio.

Si ricorda che lo studio potrà essere avviato solo dopo aver ricevuto l'autorizzazione all'attivazione da parte della Direzione Generale dell'Azienda.

Resta inteso che, ai fini del monitoraggio dell'andamento dello studio in oggetto, lo **Sperimentatore Responsabile** dovrà comunicare al Comitato Etico le seguenti informazioni **relativamente a questo singolo centro sperimentale**: data di inizio arruolamento, data di fine arruolamento e data di conclusione dello studio. In ogni caso, **a partire dall'anno di approvazione dello studio** e fino alla sua conclusione, **almeno una volta all'anno e comunque entro e non oltre il 31 dicembre**, dovrà essere fornito un rapporto annuale sullo stato di avanzamento dello studio. Per le suddette comunicazioni è possibile utilizzare il modulo disponibile sul sito web del Comitato Etico all'indirizzo http://www.aosp.bo.it/content/comunicazioni-dandamento-studio. Il modulo compilato, firmato e datato, andrà inviato **esclusivamente in formato elettronico all'indirizzo e-mail dedicato** monitoraggiostudi@aosp.bo.it indicando nell'oggetto il nome dello Sperimentatore Responsabile locale ed il codice assegnato dal Comitato allo studio.

Cordiali saluti.

IL PRESIDENTE
(Prof. Nicola Montanaro)

NOTA: per qualsiasi comunicazione relativa all'oggetto (compresi eventuali successivi emendamenti ed eventi avversi), è indispensabile, sia da parte dello sperimentatore che dello sponsor, fare sempre riferimento alla data della presente approvazione, nonché al numero indicato a margine dell'Oggetto.
Allegato: elenco Componenti Comitato Etico Indipendente del Policlinico S.Orsola-Malpighi

COMITATO ETICO INDIPENDENTE DELL'AZIENDA OSPEDALIERO-UNIVERSITARIA DI BOLOGNA
POLICLINICO S. ORSOLA – MALPIGHI
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APPENDIX 12 APPROVAL BY FACULTY RESEARCH ETHICS COMMITTEE BY PLYMOUTH UNIVERSITY (22ND DECEMBER 2015)

RESEARCH WITH PLYMOUTH UNIVERSITY

22nd December 2015

CONFIDENTIAL

Lea Godino
UO Genetica Medica, Pad.11, 2°piano
Policlinico Sant'Orsola-Malpighi
via Massarenti, 9
40138 Bologna, Italy

Dear Lea

Application for Approval by Faculty Research Ethics Committee

Reference Number: 15/16-519
Application Title: **THE IMPACT OF PRESYMPTOMATIC GENETIC TESTING FOR CANCER ON YOUNG ADULTS**

I am pleased to inform you that the Committee has granted approval to you to conduct this research.

Please note that this approval is for three years, after which you will be required to seek extension of existing approval.

Please note that should any MAJOR changes to your research design occur which effect the ethics of procedures involved you must inform the Committee. Please contact Sarah Jones (email sarah.c.jones@plymouth.ac.uk).

Yours sincerely

Professor Michael Sheppard, PhD, FAcSS
Chair, Research Ethics Committee -
Faculty of Health & Human Sciences and
Peninsula Schools of Medicine & Dentistry

Faculty of Health & Human Sciences
Plymouth University
Drake Circus

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E sarah.c.jones@plymouth.ac.uk

Professor Michael Sheppard
CQSW BSc MA PhD FAcSS
Chair, Faculty Research Ethics

APPENDIX 13 ENGLISH VERSION OF THE FLYER ADVERTISING STUDY

RESEARCH WITH PLYMOUTH UNIVERSITY

SURVEY

CANCER GENETIC TESTING IN YOUNG ADULTS: TWO POINTS OF VIEW

Does this study concern you?

- ✔ We would value your help with this study If you are a member of family where there is a genetic predisposition to cancer and
  - ✔ you had a genetic test for cancer when you were aged between 18-30 years

    OR
  - ✔ you are the parent of a child who had a genetic test for cancer when they were aged between 18-30 years.

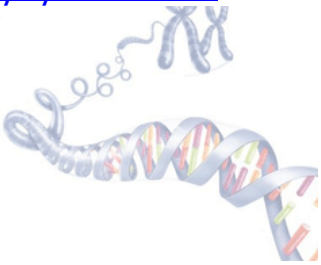
If you are in either group,
your help with this study will be valuable to us!

I'm a PhD student of Plymouth University and my project is aimed to explore motivations and the impact of genetic testing for hereditary cancer in young adults.

PLEASE HELP ME and complete the survey, it will take you only FEW MINUTES
(link below)

https://www.surveymonkey.com/r/PhDENG

THANK YOU



Info:
Lea Godino, PhD student, Plymouth University
lea.godino@students.plymouth.ac.uk

APPENDIX 14 ENGLISH VERSION OF QUESTIONNAIRES TO YOUNG ADULTS (PHASE 3)

ABOUT YOU

4. Are you male or female?

- ☐ Male
- ☐ Female
- ☐ I prefer not to say

5. In which year were you born?

6. The country where you were born?

7. The country where you are now living?

8. Your first language?

9. What is your highest educational qualification?

- ☐ No formal education
- ☐ Completed primary school
- ☐ Completed secondary school (GCSE level)
- ☐ Completed post secondary education (e.g. A level or HND or BTEC or apprenticeship)
- ☐ University degree
- ☐ Postgraduate degree

10.Please tell us about your current work:

- ☐ I have paid employment
- ☐ I have voluntary employment
- ☐ I am a student
- ☐ I am a homemaker
- ☐ I am not working and am not a student

11. If you are in paid employment, are you:

- ☐ Paid employee
- ☐ Manager
- ☐ Self-employed
- ☐ Business owner
- ☐ Member of armed force
- ☐ Professional
- ☐ Other (please specify)

12. Your marital status:

- ☐ Single (never married)
- ☐ Married
- ☐ Divorced
- ☐ Widowed
- ☐ Living with a partner
- ☐ Other (please specify)

13. Do you have any children?

14. How many daughters do you have?

15 How many sons do you have?

YOUR 'GENETIC' HISTORY

16. Have you ever had predictive cancer genetic testing? This is a test to find a known gene fault that increase the change of cancer in a healthy person. Other people in your family may have been tested before you to find the exact gene fault in your family.

- ☐ Yes
- ☐ No

17. How old were you when you were tested?

18. Which condition were you tested for?

- ☐ Hereditary breast and ovarian cancer
- ☐ Lynch syndrome (hereditary bowel cancer)
- ☐ I don't know
- ☐ Other (please specify)

19. What was your test result?

- ☐ A gene fault (mutation) was found
- ☐ A gene fault (mutation) was not found
- ☐ I am not sure what the result was

20. *Have you had cancer since having your genetic test*?

- ☐ Yes
- ☐ No

21. *Who was the first person in your family who knew they had the faulty gene?*

- ☐ Your mother
- ☐ Your father
- ☐ Your sister
- ☐ Your brother
- ☐ Other relatives (please say which relatives)

ABOUT YOUR PARENTS

If you know the following information about your parents, it will help us with the study. If you do not know the answers, you can leave the question blank. We are interested in the information, even if your parent has passed away.

22. Is your father still living?

- ☐ Yes
- ☐ No

23. What is/was your father's highest educational qualification?

- ☐ No formal education
- ☐ Completed primary school
- ☐ Completed secondary school (GCSE level)
- ☐ Completed post secondary education (e.g. A level or HND or BTEC or apprenticeship)
- ☐ University degree
- ☐ Postgraduate degree

24.Please tell us about your father's work:

- ☐ He has/had paid employment
- ☐ He is/was in voluntary employment
- ☐ He is/was a student
- ☐ He is/was a homemaker
- ☐ He is/was not working

25. If your father is/was in paid employment, is/was he:

- ☐ Paid employee
- ☐ Manager
- ☐ Self-employed

☐ Business owner
☐ Member of armed force
☐ Professional
☐ Other (please specify)

26. Has your father ever had cancer?

☐ Yes
☐ No
☐ Don't know

27. If your father has ever had cancer, what type of cancer?

- First episode of cancer:
- Second episode of cancer:
- Other:

28. If your father has ever had cancer, what age was he when he had the cancer? (Give all ages if more than one episode of cancer)

- First episode of cancer:
- Second episode of cancer:
- Other:

29. Is your mother still living?

☐ Yes
☐ No

30. What is/was your mother's highest educational qualification?

☐ No formal education
☐ Completed primary school
☐ Completed secondary school (GCSE level)
☐ Completed post secondary education (e.g. A level or HND or BTEC or apprenticeship)
☐ University degree
☐ Postgraduate degree

31.Please tell us about your father's work:

☐ She has/had paid employment
☐ She is/was in voluntary employment
☐ She is/was a student
☐ She is/was a homemaker
☐ She is/was not working

25. If your mother is/was in paid employment, is/was she:

☐ Paid employee
☐ Manager
☐ Self-employed
☐ Business owner
☐ Member of armed force
☐ Professional
☐ Other (please specify)

26. Has your mother ever had cancer?

☐ Yes
☐ No
☐ Don't know

27. If your mother has ever had cancer, what type of cancer?

- First episode of cancer:
- Second episode of cancer:
- Other:

27. If your mother has ever had cancer, what age was he when he had the cancer? (Give all ages if more than one episode of cancer)

- First episode of cancer:
- Second episode of cancer:
- Other:

FINDING OUT ABOUT YOUR RISK

Thank you for answering the questions about yourself and your parents.

Now we would like to ask you some questions about your experience before testing.

These first questions concern the first time you were told that there might be a greater tendency to develop cancer in your family, compared to other families.

36. Who told you about the possibility that members of your family might be more likely than other to develop cancer?

- ☐ Your mother
- ☐ Your father
- ☐ Both parents together
- ☐ Your sister
- ☐ Your brother
- ☐ Other relative (please say what relation they are to you)
- ☐ A person outside the family (please say who this person was, e.g. friend, doctor)
- ☐ I found out another way (please give information about this)

37. How old were you when you first received this information?

38. How did you receive the information?

- ☐ In an unplanned face to face conversation (please tell us more)
- ☐ In an unplanned telephone call (please tell us more)
- ☐ In a pre-planned face to face meeting (please tell us more)
- ☐ In a pre-planned call (please tell us more)
- ☐ Other (please tell us more)

39. Were you told at that time that the tendency to cancer in your family could be due to a genetic change?

- ☐ Yes
- ☐ No
- ☐ I can't remember

40. If you were told then that cancer could be due to a genetic change, when did you become aware that there might be a genetic condition in your family?

These questions concern how you reacted after you became aware of the family genetic condition. We are interested in your true reactions, there are no 'right' or 'wrong' answers to these questions, so please answer honestly.

Please read each of the following sentences carefully and tick the column that describes your reaction best.

41. How did you react the news that there might be a genetic condition in your family?

	Strongly disagree	Disagree somewhat	Neither sagree nor disagree	Agree somewhat	Strongly agree
	1	2	3	4	5

I did not know what it really meant
I looked for information online
I was more conscious of my risk
I arranged the first counselling session to discuss my risk
I arranged the first counselling session to have a genetic blood
I wanted to know some more about it at the time
I felt it explained things I had been wondering about

YOUR EXPERIENCE OF GENETIC COUNSELLING

Thank you for telling us about your experience before testing.

In the following questions we would like to ask you about your experience of the genetic counselling you received in the genetic clinic. We are interested in your true feelings, there are no 'right' or 'wrong' answers to these questions, so please answer honestly.

Please read each of the following sentences carefully and tick the column that describes your reaction best.

42. How did you feel about the genetic counselling?

	Strongly disagree 1	Disagree somewhat 2	Neither sagree nor disagree 3	Agree somewhat 4	Strongly agree 5
The doctor or genetic counsellor showed an interest in your personal situation regarding the cancer family history					
The doctor or genetic counsellor explained your risk to you clearly					
The doctor or genetic counsellor met your expectations of him or her					
The doctor or genetic counsellor treated you as an individual					
You would be comfortable in calling the doctor or genetic counsellor to ask further questions					
The doctor or genetic counsellor listened to what you had to say					
The doctor or genetic counsellor was considerate of your emotional state during the meeting					
You are satisfied with the way that information was communicated to you					
The doctor or genetic counsellor understood what was really concerning you					
The doctor or genetic counsellor made you feel you were "in good hands"					
The doctor or genetic counsellor made you feel that they knew how to handle situations like your's					

The doctor or genetic counsellor gave you enough of their time
The doctor or genetic counsellor was sensitive and tactful during your conversation
The doctor or genetic counsellor seemed to be an expert in the field
The doctor or genetic counsellor helped you deal with any concerns you had
You felt comfortable to talk about yourself during the genetic counselling session
You were satisfied with the length of time you had to wait until your first appointment
You were satisfied with the information your received during the genetic counselling appointment
If a friend needed similar help you would recommend this clinic to him or her
The counselling was given in an appropriate setting
Overall you are satisfied with the genetic counselling service

YOUR DECISION-MAKING PROCESS

Thank you for telling us about your experience of the genetic counselling.

In the following questions we would like to ask you about your reasons and feelings on your experience on the decision-making process.

43. Who decided that you would be tested?

- ☐ Yes
- ☐ No
- ☐ I can't remember

44. What were your reasons for wanting to be tested?

These questions concern what were you reason for wanting to be tested. We are interested in your true feelings, there are no 'right' or 'wrong' answers to these questions, so please answer honestly.

Please read each of the following sentences carefully and tick the column that describes your reaction best.

	Not applicable to me	Strongly disagree	Disagree somewhat	Neither sagree nor disagree	Agree somewhat	Strongly agree
	0	1	2	3	4	5
I wanted to learn about my children's risk or risks to any children I may have						
I wanted to try to help advance research						
I wanted to know if I need to get cancer screening tests more often						

I wanted to be reassured
I wanted to make a decision about surgery to reduce my risk
I wanted to make a decision about having (more) children
My mother strongly encouraged me
My father strongly encouraged me
I had genetic testing because of pressure from my family members
I had genetic testing because my parent asked me to do it
I made my own decision
My decision was influenced by family experience
My mother warned me about having the test
My father warned me about having the test
My mother advised me to wait, but I decided to have it
My father advised me to wait, but I decided to have it

YOUR GENETIC TEST RESULT

Thank you for telling us about your experience of the decision-making process.

In the following questions we would like to ask you about your experience after receiving your genetic test result.

We are interested in your true feelings, there are no 'right' or 'wrong' answers to these questions, so please answer honestly.

Please read each of the following sentences carefully and tick the relevant column.

45. How did you feel after receiving your genetic test result?

	Strongly disagree	Disagree somewhat	Agree somewhat	Strongly agree
	1	2	4	5
I felt upset about my test result				
I felt sad about my test result				
I felt anxious or nervous about my test result				
I felt guilty about my test result				
I felt relieved about my test result				
I felt a loss of control				
I had problems enjoying life because of my test result				
I felt able to plan my future				
I was more worried about my risk of getting cancer				

I was uncertain about what my test result meant for my cancer risk
I was uncertain about what my test result meant for my children or any children I may have
I was uncertain about what my test result meant for my family's cancer risk
I was worried other people might discuss this behind my back
I was worried that other people might think less of me because of my result
I was worried because of the possibility of passing the mutation to my children or any children I may have
I felt guilty about my family
I felt more distant from family members

YOUR GENETIC TEST RESULT

Thank you for telling us about your experience of the decision-making process.

In the following questions we would like to ask you about your experience after receiving your genetic test result.

We are interested in your true feelings, there are no 'right' or 'wrong' answers to these questions, so please answer honestly.

Please read each of the following sentences carefully and tick the relevant column.

45. How did you feel after receiving your genetic test result?

	Not applicable to me	Strongly disagree	Disagree somewhat	Neither sagree nor disagree	Agree somewhat	Strongly agree
	0	1	2	3	4	5
I felt upset about my test result						
I felt sad about my test result						
I felt anxious or nervous about my test result						
I felt guilty about my test result						
I felt relieved about my test result						
I felt a loss of control						
I had problems enjoying life because of my test result						
I felt able to plan my future						
I was more worried about my risk of getting cancer						
I was uncertain about what my test result meant for my cancer risk						
I was uncertain about what my test result meant for my children or any children I may have						
I was uncertain about what my test result meant for my family's cancer risk						

I was worried other people might discuss this behind my back	
I was worried that other people might think less of me because of my result	
I was worried because of the possibility of passing the mutation to my children or any children I may have	
I felt guilty about my family	
I felt more distant from family members	

APPENDIX 15 ENGLISH VERSION OF QUESTIONNAIRES TO PARENT OF YOUNG ADULTS (PHASE 3)

ABOUT YOU

49. Are you male or female?

- ☐ Male
- ☐ Female
- ☐ I prefer not to say

50. In which year were you born?

51. The country where you were born?

52. The country where you are now living?

53. Your first language?

54. What is your highest educational qualification?

- ☐ No formal education
- ☐ Completed primary school
- ☐ Completed secondary school (GCSE level)
- ☐ Completed post secondary education (e.g. A level or HND or BTEC or apprenticeship)
- ☐ University degree
- ☐ Postgraduate degree

55.Please tell us about your current work:

- ☐ I have paid employment
- ☐ I have a voluntary employment
- ☐ I am a student
- ☐ I am a homemaker
- ☐ I am not working and am not a student

56. If you are in paid employment, are you:

- ☐ Paid employee
- ☐ Manager
- ☐ Self-employed
- ☐ Business owner
- ☐ Member of armed force
- ☐ Professional
- ☐ Other (please specify)

57. Your marital status:

- ☐ Single (never married)
- ☐ Married
- ☐ Divorced
- ☐ Widowed
- ☐ Living with a partner
- ☐ Other (please specify)

58. How many daughters do you have?

59 How many sons do you have?

60? How old are your children?

- First child:
- Second child:
- Third child:

- Fourth child:
- Other children:

61. Have you ever had cancer yourself?

☐ Yes
☐ No

62. Is there a genetic tendency to cancer in your side of the family (from your own birth family)?

☐ Yes
☐ No
☐ I don't know

63. Have you ever had predictive cancer genetic testing? This is a test to find a known gene fault that increases the chance of cancer in a healthy person. Other people in your family may have been tested before you to find the exact gene fault in your family.

☐ Yes, before I had cancer
☐ Yes, after I had cancer
☐ Yes, but I have never had a genetic test
☐ No, I have never had a genetic test
☐ I'm not sure if I have ever had a genetic test

YOUR 'GENETIC' HISTORY

64. How old were you when you were tested?

65. Which condition were you tested for?

☐ Hereditary breast and ovarian cancer
☐ Lynch syndrome (hereditary bowel cancer)
☐ I don't know
☐ Other (please specify)

66. What was your test result?

☐ A gene fault (mutation) was found
☐ A gene fault (mutation) was not found
☐ I am not sure what the result was

67. Are you the first person in your family who was tested to find the faulty gene?

☐ Yes
☐ No
☐ Not sure

67s. Please give us more information if you wish

68. Is there a genetic tendency to cancer in your partner's side of the family?

☐ Yes
☐ No
☐ I don't know

69. Have your partner ever had cancer genetic testing? This is a test to find a known gene fault that increases the chance of cancer in a person.

☐ Yes, before he/she had cancer
☐ Yes, after he/she had cancer
☐ Yes, but he/she has never had a genetic test
☐ No, he/she has never had a genetic test
☐ I'm not sure if he/she has ever had a genetic test

70. Which condition was your partner tested for?

- ☐ Hereditary breast and ovarian cancer
- ☐ Lynch syndrome (hereditary bowel cancer)
- ☐ I don't know
- ☐ Other (please specify)

71. What was your partner's test result?

- ☐ A gene fault (mutation) was found
- ☐ A gene fault (mutation) was not found
- ☐ I am not sure what the result was

TELLING YOUR CHILDREN

Thank you for answering questions about yourself and your family.

Now we would like to ask you some questions about your decision to tell your children about the cancer family history.

72. Have you told your children about the family risk of cancer?

- ☐ Yes, I have told all my children
- ☐ Yes, I have told some of my children
- ☐ No, they do not know
- ☐ I can't remember
- ☐ Another person told them (please told us who told them)

73. If you have told your children about the family cancer, how old were they when you told them?

- First child:
- Second child:
- Third child:
- Fourth child:
- Other children:

74. If you have told your children about the family cancer risk, how did you tell them?

- ☐ I planned a conversation with them
- ☐ I took advantage of a moment when they raised the topic
- ☐ I mentioned it in a casual way
- ☐ Other (please specify)

TELLING YOUR CHILDREN: REASONS

Can you please tell us about your reasons for telling or not telling your children about the family cancer risk. We are interested in your true reasons, there are no 'right' or 'wrong' answers to these questions, so please answer honestly.

Please read each of the following sentences carefully and tick the column that is most relevant.

75. What were your reasons for telling or not telling your children about the family cancer risk?

	Not applicable to me 0	Strongly disagree 1	Disagree somewhat 2	Neither sagree nor disagree 3	Agree somewhat 4	Strongly agree 5

I wanted to provide access to information for my children

I wanted to make my children aware of the risk

I wanted to share my genetic test results with my children so they could be tested

I wanted to share my partners genetic test results with my children so they could be tested

I wanted to explain the family history of cancer

I wanted to share my genetic test results with my children because of my grandchildren or future grandchildren

I wanted to share my partners genetic test results with my children because of my grandchildren or future grandchildren

I felt it was the appropriate age to tell them

I didn't intend to tell them but they accidentally found out

I wanted them to be able to have screening

I thought my children were too young to know

I thought it might make my children anxious

I thought it might increase my children's fear of getting cancer

I thought it might increase my children's worry about my and my partner's health

I thought it was unnecessary to make my children aware of the family history

I am still coping with the test results

I was not ready to share the news

There was no medical reason to tell them

YOUR CHILDREN'S EXPERIENCE OF THE GENETIC TEST

Thank you for answering questions about telling your children.

Now we would like to ask you some questions about your children's experience of the genetic test.

76. Have you told your children it is possible to have a genetic test?

☐ Yes, I have told all my children

- ☐ Yes, I have told some of my children
- ☐ No, they do not know
- ☐ I can't remember
- ☐ Another person told them (please told us who told them)

77. If you have told your children about the genetic test, how old were they when you told them?

- First child:
- Second child:
- Third child:
- Fourth child:
- Other children:

78. Have any of your children had a genetic test?

- ☐ Yes
- ☐ No
- ☐ I don't know

79. If your child decided to have a genetic test, who requested it? (if more than one child has been tested, answer in relation to the first child who was tested)

- ☐ My child requested the test
- ☐ Myself or my partner requested the test for my child
- ☐ My child requested it with either me or my partner
- ☐ Other (please tell us more)

YOUR FEELINGS ABOUT GENETIC TESTING FOR YOUR CHILDREN

Thank you for telling us about your experience about testing for your children.

In the following questions we would like to ask you about your feelings about genetic testing for your children.

The are no 'right' or 'wrong' answers to these questions, so please answer honestly.

Please read each of the following sentences carefully and tick the column that describes your reaction best.

80. How did you feel when your child decided to have a genetic test?

	Strongly disagree 1	Disagree somewhat 2	Agree somewhat 4	Strongly agree 5
I felt my child should be tested				
I felt guilty when I thought the gene fault might be inherited by my child				
I felt I did not have control over the decision my child made about the test				

APPENDIX 16 ENGLISH VERSION OF THE FIRST WEBPAGE ON SURVEY MONKEY®

Cancer genetic testing in y ×
SurveyMonkey Inc. [US] https://www.surveymonkey.com/r/PhDENG
Apps
Save to Mendeley
RESEARCH WITH PLYMOUTH UNIVERSITY
Cancer genetic testing in young adults: two points of view
Information about this research study
What is the study about?
Young adults with a family history of cancer may be offered a genetic test. Some people wish to be tested, others decide against it. This study is being done to explore genetic testing for hereditary cancer in young adults (aged 18-30 years) undergoing cancer genetic counselling and how their parents played a part in this scenario. The results of the study will be used to help us understand how we can support young adults, who are making decision about genetic testing, and their parents.
Does this study concern me?
We would value your help with this study If you are a member of family where there is a genetic predisposition to cancer and
- you had a genetic test for cancer when you were aged between 18-30 years OR
- you are the parent of a child who had a genetic test for cancer when they were aged between 18-30 years.
What am I being asked to do?
If you agree to take part in the study, we will ask you to fill out a questionnaire about your experiences. This will take between 15 and 20 minutes. All your answers will be anonymous and we will not be able to identify you in any way.
I would be very happy to answer any further questions you might have about the study before you decide about being involved.
If you want to ask a question, please contact me:
Lea Godino, PhD student, Plymouth University
Email: lea.godino@students.plymouth.ac.uk
1. How do I complete the survey if I am interested? Please click one of the choice below. This will take you to a webpage with a questionnaire for you to complete.
I underwent cancer genetic testing when I was aged between 18-30
I am the parent of a child who had a genetic test for cancer when they were aged between 18-30 years

APPENDIX 17 CHARACTERISTICS OF FULL-TEXT EXCLUDED PAPERS

AUTHORS AND TITLE	AIMS	METHOD	SAMPLE		REASON FOR EXCLUSION
			AGE	FROM	
Aktan-Collan *et al*. (2011) "Sharing genetic risk with next generation: mutation-positive parents' communication with their offspring in Lynch Syndrome."	To assess how parents share knowledge on genetic risk with children in Lynch Syndrome families	Quantitative research	Parents	Finland	On communication to children of parents results, not on testing
Holt (2006) "What Do We Tell the Children? Contrasting the Disclosure Choices of Two HD Families Regarding Risk Status and Predictive Genetic Testing."	To assess parents and children views on HD, including presymptomatic testing	Qualitative research	6 children 10-21 Plus parents	Minneapolis, United States of America	On communication to children of parents results, not on testing
Bradbury *et al*. (2012) "When parents disclose BRCA1/2 test results: Their communication and perceptions of offspring response."	To assess prevalence and predictor of communication of BRCA testing results to the offspring	Quantitative research	253 parents (28-66yo) and 505 offspring (3-35)	Philadelphia, United States of America	On communication to children of parents results, not on testing

Bradbury *et al.* (2007) How often do BRCA mutation carriers tell their young children of the family's risk for cancer? A study of parental disclosure of BRCA mutations to minors and young adults		Qualitative research	Parents with at least one child younger than 25	Chicago	On communication to children of parents results, not on testing
Bradbury *et al.* (2009) Learning of your parent's BRCA mutation during adolescent or early adulthood: a study of offspring experiences	To understand the content and method of disclosure of learning of a parent's BRCA mutation, their understanding and perceptions of hereditary risk and the psychosocial and health related impact of this communication	Qualitative research	18-33 (26.0)	United States of America	On communication to children of parents results, not on testing

Fisher *et al.* (2014) Talking about familial breast cancer risk: topics and strategies to enhance mother–daughter interactions	To capture what (cancer-related) topics elevated-risk mothers reported discussing with their daughters. To explore challenges mothers perceived to further complicate such discussions. To assess strategies mothers perceived can enhance mother–daughter communication about these topics.	Qualitative research	Mothers	New York	Views of mothers of young people, not the young people themselves
Hallowell *et al.* (2005) Communication about genetic testing in families of male BRCA1/2 carriers and non-carriers: patterns, priorities and problems	To assess the experiences of cancer and genetic testing, decision making about testing and the communication of test results and genetic information within the family	Qualitative research	Adult (39-75) and children (19-37)	United Kingdom	On communication to children of parents results, not on testing

Hamann *et al.* (2000) Attitudes toward the genetic testing of children among adults in a Utah-based kindred tested for a BRCA1 mutation	To asses attitudes toward BRCA1 testing for children under among parents who had received BRCA1 tests result	Cross-sectional analysis	Parents (18-82; 46.9) and children (≤18)	Northern European	About testing children
Heimler and Zanko (1995) "Huntington disease: a case study describing the complexities and nuances of predictive testing of monozygotic twins."	To evaluate the co-twin's diagnosis and autonomy of participation at genetic counselling, when a candidate for presymptomatic testing for the Huntington disease gene is a monozygotic twin.	Case study	Young man	United States of America	Focus is on twins
Hoskins and Werner-Lin (2013) A multi-case report of the pathways to and through genetic testing and cancer risk management for BRCA mutation-positive women aged 18-25	To assess a rich description of the experiences of women undergoing BRCA1/2 mutation testing and initiating risk management	Case report	18-25y	New York	Good paper without ethical approval, consent, etc.

Hoskins *et al*. (2012) "Toward a new understanding of risk perception among young female BRCA1/2 "previvors".	To assess how young women's life course decision and relationship are shaped by their knowledge of themselves as BRCA mutation-positive	Qualitative research	29.6 (21-36) at interview and 24.5 (18-35) at genetic testing	United States of America	Age up 35 year
Ormondroyd *et al.* (2014) Pre-symptomatic genetic testing for inherited cardiac conditions: a qualitative exploration of psychosocial and ethical implications	To explore perceptions of the cascade process, impact of pre-symptomatic genetic testing and attitudes towards direct contact as an alternative to family-mediated dissemination for inherited cardiac conditions	Qualitative research	<30->60	Oxford, United Kingdom	Not only on young people
Peshkin *et al.* (2009) Brief assessment of parents? Attitudes toward testing minor children for hereditary breast/ovarian cancer genes: Development and validation of the Pediatric BRCA1/2 Testing Attitudes Scale (P-TAS)	To improve and evaluate a new measure for use in genetic research and consultation	Cross-sectional analysis	Mothers (30-59y) of children (8-21y)	Washington, New York, Boston	About testing children

Rew *et al.*(2010) Cool, but is credible? Adolescents' and parents' approaches to genetic testing	To assess the level of general knowledge and the methods of decision making about genetic testing of adolescent and their parents	Qualitative research	Adolescent (14-21 y) and their parents (31-61 y)	Texas,United States of America	Not at genetic risk
Sparbel *et al.* (2008) Experiences of teens living in the shadow of Huntington disease	To explore the experiences of teens living in families with Huntington disease	Qualitative research	Adolescent (14-18 y)	United States of America, Canada	Not on presymptomatic testing; only teens.
Tercyak *et al.* (2001) "Psychological issues among children of hereditary breast cancer gene (BRCA1/2) testing participants."	To assess experience distressing thought patterns over positive test results	Cross-sectional	Adolescent (11-17)	Washington, United States of America	Not on presymptomatic testing

Tercyak *et al*. (2013) "Decisional outcomes of maternal disclosure of BRCA1/2 genetic test results to children	To assess the prevalence of patient disclosure of BRCA genetic test results to children; to evaluate demographic, clinical, decision making, and psychological predictors of this outcome; to identify patients' satisfaction with their disclosure choice.	Prospective observational study	Mothers of children aged 8-21 y	United States of America	On communication to children of parents results, not on testing
Werner-Lin (2007) Danger zones: risk perceptions of young women from families with hereditary breast and ovarian cancer	To assess beliefs about risk and susceptibility to HBOC of young women with elevated genetic risk	Qualitative research	22-36	Eastern and Western European	Age up to 35 years and did not focus on testing decision

APPENDIX 18 IMPACT OF PRESYMPTOMATIC GENETIC TESTING ON YOUNG ADULTS: A SYSTEMATIC REVIEW

European Journal of Human Genetics (2015), 1–8

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REVIEW

Impact of presymptomatic genetic testing on young adults: a systematic review

Lea Godino*[,1,2], Daniela Turchetti[1], Leigh Jackson[2], Catherine Hennessy[2] and Heather Skirton[2]

Presymptomatic and predictive genetic testing should involve a considered choice, which is particularly true when testing is undertaken in early adulthood. Young adults are at a key life stage as they may be developing a career, forming partnerships and potentially becoming parents: presymptomatic testing may affect many facets of their future lives. The aim of this integrative systematic review was to assess factors that influence young adults' or adolescents' choices to have a presymptomatic genetic test and the emotional impact of those choices. Peer-reviewed papers published between January 1993 and December 2014 were searched using eight databases. Of 3373 studies identified, 29 were reviewed in full text: 11 met the inclusion criteria. Thematic analysis was used to identify five major themes: period befeore testing, experience of genetic counselling, parental involvement in decision-making, impact of test result communication, and living with genetic risk. Many participants grew up with little or no information concerning their genetic risk. The experience of genetic counselling was either reported as an opportunity for discussing problems or associated with feelings of disempowerment. Emotional outcomes of disclosure did not directly correlate with test results: some mutation carriers were relieved to know their status, however, the knowledge they may have passed on the mutation to their children was a common concern. Parents appeared to have exerted pressure on their children during the decision-making process about testing and risk reduction surgery. Health professionals should take into account all these issues to effectively assist young adults in making decisions about presymptomatic genetic testing.
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INTRODUCTION

Presymptomatic and predictive genetic testing are available for a number of heritable genetic disorders including hereditary cancer syndromes, inherited cardiac conditions and neurodegenerative genetic disorders.[1] The terms 'presymptomatic' and 'predictive' genetic testing refer to the possibility of detecting a genetic mutation that causes a particular condition before the presentation of symptoms. The first term generally refers to those diseases in which a positive test result will inevitably lead to the development of the disease later in life (eg, Huntington disease (HD)); the second term refers to a broader range of diseases in which the risk for a disorder is increased but without necessarily implying any degree of certainty (eg, hereditary breast and ovarian cancer (HBOC)). However, these terms are often used in a broadly interchangeable manner. A substantial difference is that cancer disorders can be monitored through a surveillance protocol or prevented by surgical intervention, while no prevention is currently available for diseases such as HD or cerebellar ataxia. Therefore, the choice to undergo a presymptomatic test for disorders with incomplete penetrance and where there are preventive measures could have a highly different psychological and social impact when compared with testing for disorders with complete penetrance and no preventive options, particularly in young adults. In this review, the term 'presymptomatic' will be used to indicate both predictive and presymptomatic tests, but the different impact will be considered whenever appropriate.

A presymptomatic diagnosis of a serious genetic illness can have a profound impact on the person and family and should be managed using an individualized counselling process.[1] Presymptomatic genetic testing of minors (under the age of 18 years) is not usually recommended unless effective clinical actions are available.[2–4] Generally, there are three key arguments against presymptomatic genetic testing in adolescents or young people: that it (1) fails to respect the future autonomy of the young person; (2) breaches confidentiality; and (3) may cause psychosocial harms.[5]

The age at which young people should undergo presymptomatic genetic testing for adult-onset disorder is a matter of debate.[2–5] Key challenges have to be faced during the transition from adolescence to adulthood, such as marriage, finishing education, beginning full-time employment and becoming a parent, and the impact of testing may affect, and be affected by each of these events. In the light of the above-mentioned issues, it would be appropriate to ask what health information and counselling young adults need to make prudent decisions about genetic testing. The purpose of this systematic review was therefore to systematically identify and analyse factors influencing young adults' or adolescents' choices to have a presymptomatic test and the emotional impact of those choices.

MATERIALS AND METHODS

Design

A systematic review is a method of amassing, assessing and synthesizing a body of evidence on a particular topic.[6] This systematic review was conducted in accordance with the Centre for Reviews and Dissemination methods for undertaking reviews in health care[6] with the aim of assessing which factors influence young adults' or adolescents' choices to have a presymptomatic test for a genetic condition and the emotional impact of those choices.

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Search strategy

Eight databases used for indexing medical and psychosocial research (Embase, Medline, PsychoInfo, Pubmed, SocIndex, The Cochrane Library, Cumulative Index of Nursing Allied Health Literature (CINAHL) and Web of Science) were searched for papers published between January 1993 and December 2014. We chose to start at 1993 because presymptomatic testing based on mutation analysis (ie, not based on linkage) became available for HD that year (Harper *et al*,[7]) and this was a landmark in presymptomatic testing for adult onset conditions. As the first authors are bilingual, papers published in either English or Italian were eligible (there were no papers identified that were written in Italian). The literature search employed variations and Boolean connectors of the key terms. An exploratory search with the terms 'genet*' or 'predict* test*' or 'presymptom* test*' or 'young*' or 'adult*' or 'adolescent*' or 'decision*' or 'choic*' or 'communicat*' or 'psycho*' resulted in 976 studies, which failed to include some papers already known on this theme, therefore a further search was conducted by using the key terms: 'adolescent*' or 'young*' or 'BRCA' or 'APC' or 'Lynch' or 'Huntington' or 'genetic* test*'. Targeted internet searching using Google Scholar and reference lists of relevant papers were also examined for any additional studies of interest.

Inclusion and exclusion criteria

The criteria for inclusion in this systematic review were papers: (a) published in English or Italian; (b) published in peer-reviewed journals between 1993 and 2014 and reporting original research (using any methods); (c) where the study sample explicitly included young adults or adolescents (14–30 years); (d) focused on presymptomatic or predictive testing in young people; (e) focused on the factors influencing young adults' or adolescents' choices to have a presymptomatic or predictive test and the emotional impact of those choices. Papers were excluded from the review if they were (a) guidelines for testing; (b) educational or opinion papers; (c) focused on perceptions and attitudes of college students/young adults who were not at known risk of a specific adult-onset genetic condition.

Selection of studies

Three review authors (LG, LJ and DT) independently screened the titles and abstracts of articles identified in the first search against the inclusion criteria and decided which papers should be retrieved. Articles were rejected at this stage if the title or abstract did not focus on our topic, was not in English or Italian, or was not original research. We reviewed selection decisions and resolved disagreements by consultation with a third review author (HS).

Search outcome

The Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA)[8] flowchart showing the consecutive methodological steps of this systematic review is displayed in Figure 1. The search of eight databases initially produced 976 potential papers. With the second search, 2397 papers were found. On the total of 3373, 755 were duplicates, leaving 2618 for examination. Following review of the title and abstract, 29 papers were assessed as potentially relevant. These papers were read in detail by the authors. Eleven papers were included in the review. The main characteristics of the included studies are described in Table 1.

Quality appraisal

All papers considered for inclusion in the review were subjected to independent analysis by two authors (LG and HS) using standard quality assessment criteria for evaluating original research papers from a variety of fields.[9] This evaluation method allows the systematic evaluation of both quantitative and qualitative original research and across a broad range of study designs. Specific aspects of the paper relating to methodology and reporting of results are assessed and assigned 0 points (not addressed), 1 point (partially addressed) or 2 points (satisfactorily addressed). A summary percentage score was calculated by dividing total score summed across all applicable items by the highest possible score total after excluding non-applicable items.[9] Any disagreements about scoring of papers were discussed. Kmet *et al*[9] do not enforce a minimum score for inclusion in a review, although they suggest 60% as a reasonable cutoff point. In the present review two papers scored 50%, but it was decided to include them, since they contributed relevant information that was not included in the other papers (see Table 1). Thus, all papers contributed to the synthesis and development of themes.

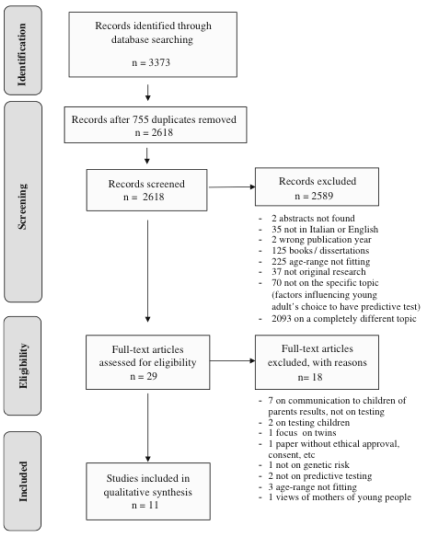

Figure 1 PRISMA flowchart of study selection. This PRISMA flowchart demonstrates the process of identifying and screening relevant studies. The screening process identified 11 studies from an initial pool of 3373 as being relevant to the current review.

Data abstraction and analysis

It was not possible to undertake a meta-analysis of the data due to the heterogeneity of the methods and samples. We therefore prepared a narrative description of the findings[6] using thematic analysis based on the methods described by Braun and Clarke,[10] in order to employ a clear, replicable and transparent methodology. The thematic analysis was confirmed by three authors (LG, DT and HS).

RESULTS

Characteristics of included studies

In total, the systematic review included 11 qualitative studies. Methods adopted by the authors were interpretative phenomenological analysis,[11] thematic analysis,[12–16] a combination of interpretative content analysis and thematic analysis,[17] or grounded theory.[17–21] Patenaude *et al*[16] also included a quantitative analysis of their data.

All the included studies were published between 2007 and 2014 and were focused on few specific heritable disorders, namely autosomal dominant cerebellar ataxia,[15] familial adenomatous polyposis (FAP),[13,17] familial cardiomyopathy,[11] HBOC,[11,14–16,18–21] hereditary diffuse gastric cancer,[15] HD[2,12,15,17] and Lynch syndrome.[15] Samples included participants within an age range of 12–39 years, thus including, but not limited to, the age range of 14–30 years identified by the authors as the focus of the search. Eligible studies were conducted in Australia,[12,13,15,17] Canada,[14,18,19] United Kingdom,[11] and the United States.[16,18–20]

Table 1 Main characteristics of included studies

Study	*Country*	*Number of participants*	*Age*	*Condition*	*Aim*	*Analysis*	*Main findings*	*Kmet et al*[10] *score and quality issues*
Duncan *et al*[12]	Australia	8	17–25 years	Huntington disease	To explore the experience of predictive genetic testing for HD from young persons' perspectives and document the impact that testing has upon various aspects of young peoples' lives	Thematic analysis	For some of the young people interviewed, uncertainty about their genetic status constituted a barrier in their lives and prevented them from moving forward. Testing in similar circumstances may therefore allow other young people to move forward with their lives	Score = 80%
Duncan *et al*[17]	Australia	18	14–26 years	Huntington disease and familial adenomatous polyposis	To evaluate the potential effects associated with predictive genetic tests in young people	Grounded theory	The results were analysed in two categories: harms and benefits. These categories have been separated into three sub-categories: (a) experiences relating to a gene-positive test result; (b) experiences relating to a gene-negative test result; (c) experiences relating to the testing process in general	Score = 70%
Duncan *et al*[13]	Australia	10	12–25 years	Familial adenomatous polyposis	To evaluate some of the key ethical challenges associated with predictive genetic testing for FAP in young people	Combination of interpretative content analysis and thematic analysis	Five themes emerged: (1) the significance of the test; (2) young people's lack of involvement in the decision to be tested; (3) young people's limited understanding; (4) provision of the blood test at the first visit; (5) group testing of family members. These themes highlighted key ethical challenges. From these themes, authors draw eight recommendations for future practice to provide developmentally appropriate care to young adults undergoing predictive genetic testing	Score = 50% Sampling strategy verification procedures to establish credibility of the study were not well described
Hamilton *et al*[18]	United States of America and Canada	44	18–39 years	Hereditary breast and ovarian cancer	To describe the decisional process of young women with increased risk for HBOC	Grounded theory	Four life trajectories that influenced the decision in young women to have genetic testing and subsequent risk reduction decisions after receiving a positive mutation result: (1) long-standing awareness of breast cancer in the family; (2) loss of one's mother to breast cancer at a young age; (3) expression of concern by a health-care provider; (4) personal diagnosis of breast cancer. Understanding possible influences behind decision-making for genetic testing and risk reduction in young women may assist health-care providers in offering age appropriate guidance and support	Score = 90%
Hamilton[19]	United States of America and Canada	44	18–39 years	Hereditary breast and ovarian cancer	To explore how young women live with a BRCA mutation	Grounded theory	Among 13 unmarried women, issues of when to disclose information about their genetic risk with their partners were	Score = 65% The sampling strategy, data

Table 1 (Continued)

Study	*Country*	*Number of participants*	*Age*	*Condition*	*Aim*	*Analysis*	*Main findings*	*Kmet et al*[10] *score and quality issues*
							discussed. 24 women who had children reported 'staying alive' for their children as a primary goal; on the other hand women without children reported an urgency to have children. Several of the 21 who had a breast cancer diagnosis said knowledge of their genetic risk influenced their decision to undergo prophylactic mastectomy.	collection methods and analytic methods are not well described.
Hoskins *et al*[21]	United States of America	32	21–25 years	Hereditary breast and ovarian cancer	To explore patient-centred perspective on the dilemma faced by 18–24 years old as they considered BRCA1/2 genetic testing and risk management		Young adults expressed needs for greater clarity in recommendations for screening and prevention before age 25, and ongoing contact with providers to discuss risk management protocols	Score=90%
Macrae *et al*[14]	Canada	8	22–37 years	Hereditary breast and ovarian cancer	To assess the experiences of those mutation-negative young women	Thematic analysis	To investigate the experience of BRCA mutation-negative young women, eight themes were analysed: (1) timing; (2) disclosure; (3) risk perceptions; (4) cancer worry; (5) cancer burden; (6) hope; (7) plans for the future; (8) explanatory models for mutation status. These young women were likely still affected by the degree of cancer history in their family, even with their understanding of the genetic contribution to disease	Score=90%
MacLeod *et al*[11]	United Kingdom	36	15–31 years	Huntington disease, hereditary breast and ovarian cancer, hypertrophic cardiomyopathy or dilated cardiomyopathy	To evaluate the motivation of young adults to be tested when young, their experiences of the counselling process and the advice they would offer to health professionals and other young adults considering testing	Interpretative phenomenological analysis	Young adults saw the value of pretest counselling not in facilitating a decision, but rather as a source of information and support. Differences emerged between the disease groups in terms of parental attitudes to testing. Parents in families with familial cardiomyopathy were a strong influence in favour of testing, in HBOC the decision was autonomous but congruent with the parents' view, and in HD the decision was autonomous and sometimes went against the opinions of relatives	Score=90%
Mand *et al*[15]	Australia	9	17–21 years	Huntington disease, autosomal dominant cerebellar ataxia, Lynch syndrome, hereditary breast and ovarian cancer, hereditary diffuse gastric cancer	To assess the experiences of young people who request a predictive genetic testing	Thematic analysis	Three themes emerged: (1) life before the test; (2) the battle to be tested; (3) living with the knowledge. The results convey young adults, from families affected by genetic conditions, might possess task-specific competence relating to decision-making about predictive testing	Score=50%. The design and connection to a theoretical framework is not completely described, and the verification procedures to establish credibility of the study were not described

Table 1 (Continued)

Study	Country	Number of participants	Age	Condition	Aim	Analysis	Main findings	Kmet et al[10] score and quality issues
Patenaude *et al*[16]	United States of America	40	18–24 years	Hereditary breast and ovarian cancer	To evaluate what daughters understand about their 50% chance of carrying BRCA mutation and about risk reduction or management options for mutation carriers. To assess the extent and nature of daughters' cancer-related distress and the effects of knowing mother's mutation status on daughters' future plans	Thematic analysis	Daughters of mothers who tested positive for a mutation in BRCA genes showed scarce genetic knowledge. Also, the genetic information was raised by young women regarding their future plans, such as childbearing	Score=70%
Werner-Lin *et al*[60]	United States of America	32	18–24 years	Hereditary breast and ovarian cancer	To build on and increase findings of experiences of BRCA 1/2 mutation carriers in their reproductive years by focusing on those challenges specific to 18–24 year olds	Grounded theory	Feeling vulnerable to a cancer diagnosis were described by participant. Also, they described a quandary regarding their care, a wide range of genetic and health literacy. Several young women contemplated risk-reducing mastectomy before age 25. Parents were a primary source of emotional and financial support for young adults	Score=80%

Findings of the analysis

Issues emerging from young people interviews included family and partner relationships, plans for the future, emotional state and the general approach to life.

Five major themes were identified: the period before testing, the experience of genetic counselling, involvement of parents in decision-making, impact of personal test result communication and living with genetic risk.

The period before testing. Many participants reported having grown up without awareness or with misinformation about the genetic disease running in their family or its inheritance mode:[12,16,21] they also lacked information about the appropriate age for testing.[12,16]

Two sets of authors reported that the first communication about the genetic risk was made by parents.[14,16] None of the participants was younger than 12 years of age when informed, about half experienced disclosure before they were 18 years old and half between 18 and 21 years old. Many participants stated that the disclosure was made during an occasional encounter and in a casual moment (ie, while driving) or by telephone.[16] Almost all daughters were informed of their mother's test result in a private conversation with their mother, and it was rare for both parents to participate in the disclosure.[14,16] In some studies, participants expressed a preference toward being informed by both parents, although they knew that the information given by parents was limited and sought genetic counselling almost immediately after disclosure.[14] In other cases, once aware of the family genetic disorder, those who did not understand what it really meant sought information online or in professional journals,[16] while those who were more conscious of own risk (or potential risk) arranged the first counselling session to have their blood test.[18]

However, interviewees described the disclosure of a positive parental test result as the most important information of their lives,[15] reporting concerns about their mother's health[15,16] and, only secondarily, their own.[15] In the quantitative sub-study by Patenaude *et al,*[16] one-third of the daughters of BRCA1/2 mutation carriers reported normal levels of general distress but high cancer-related distress, which was not significantly different from distress levels of women with known BRCA1/2 mutations.

Some participants reported that at the time they were told of their risk, the implications for themselves seemed distant, but now, as young adults, the fear of developing the adult-onset disease recurring in their family had increased.[16] Conversely, others felt that early disclosure of the family disease gave them the time to digest the information.[14] However, the knowledge of being at risk of a disorder such as HD for some participants involved engagement in risk behaviours such as drugs use, trouble with the police or difficulty at school.[12]

When approaching the decision about testing, in the study by MacLeod *et al*[11] most of the participants did not understand that having a presymptomatic test was a choice, but rather something they felt obliged to undergo in order to obtain information about themselves and to remove uncertainty. For example, a young woman said '*I knew, I had to*' (p 397).[11] By contrast, those who perceived there was a choice prepared themselves for the result; some prepared themselves for the worst possible outcome because then they would not be surprised by bad news,[11,12,15] while other participants were scared that receiving the test result would be devastating.[16] Some study participants expected to test positive because of identification with a gene-positive family member.[11,14,15]

Choosing to undergo genetic testing constituted a major life event,[13] so important that participants reported it had a significant impact on their outlook and sense of self.[15,17] For example, a young woman said

'that was the day the clock stopped; that was the day the uncertainty began' (p 644).[15] Nevertheless, while Mand *et al*[15] and MacLeod *et al*[11] report that no interviewees expressed regret regarding the decision to undertake testing, the timing for testing emerged as important because of potential interference with schooling: one young woman tested during her final year, said that looking back, she wished she had been tested at a different time of her life.[17] Another factor relevant to the timing of testing was childbearing planning,[11,14] with participants split on the issue of undergoing presymptomatic testing before *versus* after having children.[14]

The experience of genetic counselling. Undergoing genetic counselling for the young adults studied by Duncan *et al*[17] was reported to have helped discussion of problems, for example a young woman who was mutation positive for HD said that the counsellor helped her with every type of problem in her life. Even when the genetic counselling was a source of information and support, it did not appear to facilitate the decision to be tested.[11] However, Duncan *et al*[13] showed that when counselled and tested at the same time as their siblings, participants felt this had limited the individual attention and support during the counselling process.

Some negative feelings were reported about counsellors,[15] such as the perception of not being understood and the feeling that the counsellor was the person with the power over the testing decision. Some participants were disappointed to hear that the counsellor believed they were not ready to deal with the psychological consequences of the genetic test and that they needed to take time to reach an autonomous decision. The need to wait for genetic testing increased the feeling of disempowerment raised by uncertainty: a girl said *'I wanted the maybe to become yes or no; I was over maybe'* (p 645).[15] Others focused their attention on the procedure, instead of the meaning of testing, with the fear of the needle overshadowing the purpose of the counselling.[13,17]

A consequence of discussion during pre-test counselling was that most young adults had shared their test result with only close friends and family, because they felt that other people would not understand the complexities of the process from decision-making to the result.[11]

Involvement of parents in decision-making. Although theoretically an autonomous choice to undergo presymptomatic testing is a fundamental requirement of the process of genetic counselling, some parents were reported to exert pressure on their young adult children.[11–13,17,18,20] In addition, Hoskins *et al*[21] reported that a young woman underwent genetic testing because of her gynaecologist's suggestion. As a consequence of parental pressure, interviewees conveyed feelings of disempowerment and lack of control and declared that they underwent genetic testing because of pressure from family members or 'for' a parent, which also raised the ethical problem of respecting young adults' developing autonomy.[12,13,17,18,20] A young boy said *'I was 12 when I was told that I had to have the test, I didn't want to have it, but then I sort of had to'* (p 30);[13] a girl said that her parents did not ask her if she wanted to do, they just said *'you know, you have to go get a blood test'* (p 30).[13]

Older participants were more likely than younger ones to decide autonomously to have genetic testing, so much so that some described it as *'a way to take control and not to be like their mothers'* (p 152).[19]

Even when the decision-making was autonomous and pragmatic, still the family experience was important, especially when parents had developed cancer.[18] Some of these young adults had lost a parent in their adolescence or earlier, so they grew up without a parent and with the knowledge that their parent's death was due to 'the gene' and that they may carry the same risk. In this way, the study conducted by Hamilton *et al*[18] showed that participants both desired and feared genetic testing. Another key motivation was the perception that they were doing something to alter the course of a disease that had led to the death of affected relatives.[11] A young woman said *'I just thought that you know if she (mum) would have had the opportunity to have the test, then things could have been a lot different'* (p397).[11]

However, differences emerged in parental involvement in the decision-making process, based on the specific disease.[11] Parents in families with familial cardiomyopathy had a strong influence in favour of testing; in HBOC the decision was autonomous but usually congruent with the parents' point of view, while in HD the decision was autonomous and sometimes went against the parents' opinion.[11]

Impact of personal test result communication. Once the test was undertaken, waiting for test result was associated with anxiety.[17] At the time of the communication of the genetic test result, participants generally had to face the idea of disease.[14] Usually at their age young adults do not think about disease; their attention is focused on plans for their future such as university and/or job plans.[14] Nevertheless, none of the participants of the study by Mand *et al*[15] reported a catastrophic emotional response to their test result, but conflicting emotions of relief, happiness, guilt, fear and anger were generally reported.[14] In more detail, authors described the emotional impact of both gene-negative and gene-positive test results. Surprisingly, positive and negative emotional outcomes were not correlated with test results: in any case interviewees thought that the best thing was to find out the result.[17] Accordingly, participants described themselves as happy just to know their genetic status or as willing to begin enjoying life and to make behavioural changes.[12,15] Specifically, a positive result led participants to feel able to move forward and to understand what was important (or not) in their lives.[17] Although some participants stated their positive result induced a change of lifestyle, others showed no reaction to testing positive; this lack of reaction sometimes created uneasiness because the counsellors failed to understand the underlying feelings, which are well explained by a girl: *'I kept on the same direction I was already going'* (p 646).[15] In others, a gene-positive result created some negative emotions such as depression and anxiety, either in general or related to potential gossip by other people,[15,17] connected with employment,[17] related to the possibility of passing on the mutation to their future children[15] or because of a different test result in other family members.[11] A young boy said *'when I first found out I didn't want be too happy around them because it's still not the best of situations because my mum's still poorly with it[…], I still upset about my mum'* (p 399);[11] a girl said that she had been only thinking of herself during the decision-making process, but now, receiving a negative test result, she wondered *'what does she (sister) feel about me now because I haven't got it and she has'* (p 399).[11] In addition, some interviewees described their shock at finding out that they had not inherited the family mutation:[11,15] they had prepared themselves for something and then it just did not happen.[11,12] This was particularly true for young adults receiving an HD result.[11] One young man, in the study by Macrae *et al*,[14] received a negative test result but clearly expressed the desire to have been mutation positive. Also, negative test results generated unexpected negative emotions in some participants, such as guilt and feeling distanced from family members. Moreover, some interviewees expressed the desire to receive additional screening regardless of their results, because of their familial cancer experience and residual cancer worry.[14] Nevertheless, in other participants the negative test result was associated with feeling able to plan for the future.[17]

Living with a genetic risk

Although the authors of seven papers analysed risk management in terms of behaviour and attitudes, six of those papers were focused on BRCA1/2 carriers or daughters of BRCA1/2 carriers[11,14,16,18–21] and only one on familial cardiomyopathy and HD.[11]

Even though interviewees stated that having time before the beginning of surveillance gave them the opportunity to think about surveillance protocols or prophylactic surgery,[14] younger participants were more likely to feel out of place in the health-care system and frustrated at their inability to access screening, so much so that some described themselves as '*paralyzed*': one young woman underwent bilateral mastectomy at age 22, believing it was the only way to manage her cancer risk.[21] Others expressed frustration at receiving inconsistent, inaccurate, ambiguous or incomplete recommendations by genetic counsellors or doctors, during the initial phase of their mutation-positive experience.[21] They complained that each doctor explained only their own discipline-specific perspective and knowledge base.

Others wondered about when to share with a new partner their genetic risk or how early in a relationship to discuss having children[11,16,19–21] or plans for prophylactic surgery.[16,19,20]

Some participants with children described the impact of knowing they may have passed on the mutation to their children in terms of feeling guilty, worried and, in some cases, leading to a decision to limit the number of children. However, none regretted the choice to have children;[19] they declared that they first considered the possibility of avoiding having children, but then realized that there were many options and over time there will hopefully be more.[19] However, participants with children thought also about '*staying alive*' related to their children (p 28)[19] or of being not the next in the family.[18] As a consequence of the wish to stay alive, young women were making the choice to have prophylactic surgery sooner rather than later.[18,19] On the other hand, those opting for surveillance did not feel confident in surveillance protocols and reported of being anxious waiting for the next screening.[18] A 35-year-old woman said '*I admire women who can live with surveillance, but that was not for me*' and she felt herself as being a '*ticking time bomb*' (p 153).[18]

Werner-Lin *et al*,[20] also, showed that some parents exerted pressure on their children to pursue risk reduction surgery, while other young adults erected a barrier, because of their young age, to address aspects of cancer risk, for example in terms of being too young for surveillance.

DISCUSSION

Although this systematic review focused on presymptomatic testing, one major issue emerging from the papers reviewed is when and how at-risk individuals are informed of their genetic risk. Although many participants grew up with no or scarce information concerning their potential genetic risk, communication generally occurred due to the parents' initiative and in a casual manner, several years before testing or clinical actions could be undertaken.[12,16,21] This is in line with findings by Rew *et al*,[22] which showed that the majority of children of BRCA mutation carriers learnt of their potential genetic risk of cancer many years before preventive interventions were recommended. Indeed, intra-familial communication is a highly complex process, especially when an inherited genetic condition is involved, thus it is understandable that parents face the dilemma of when, how and what to tell their children about it.[23,24] On the other hand, appropriate communication of genetic risk information by parents to their children is highly desirable, since it has been shown to have long-term consequence in terms of informed reproductive decision-making and better family cohesion.[25] To achieve this, health professionals may have a role in both supporting parents and young people, but their involvement in parents' decisions to communicate genetic risk to young family members was found to be limited in both our search and previous reports.[22,26,27] Although this may be partly due to the parents' wish to undertake this task alone, it is reported that some parents desired health professionals to be available in a supporting role, but found that this support was limited.[25,28] This evidence highlights the need for a comprehensive, longitudinal counselling process with appropriate timing and setting, which supports 'parent-to-offspring' risk communication first and young people's decision-making about presymptomatic testing and risk management afterwards. Accordingly, participants perceived that their lack of emotional experience at the time of testing had made it difficult for them to envisage the possible psychological impact of a test result.[11] Furthermore, establishing a deeper and long-standing relationship with the counsellor may reduce the feelings of disempowerment reported by some study participants about the experience of genetic counselling. Such an approach would also help limit parents' pressure toward testing or risk-reducing surgery, which was a relevant issue in the studies reviewed.[11–13,17,18,20]

Concerning the impact of test results, overall, our findings do not support any substantial risk of adverse emotional outcome in mutation carriers, which is in agreement with previous findings.[29] However, possible reactions to being tested *per se* should be explored before undertaking testing, instead of focusing only on the potential effects of specific test results.

Nevertheless, there is general concern that undergoing presymptomatic testing too early in life may increase the risk of unfavourable impact, and, therefore, the right age to undergo presymptomatic testing is still a matter of debate.[30–32] In most of the papers analysed, the age at which participants had undergone genetic testing was not specified,[13,14,16,18–20] whereas Duncan *et al*,[17] who included in their analysis 10 individuals who were aged 10–17 years at the time of their genetic test for FAP, concluded that harms observed in younger persons were no different in nature from those described in adults. According to UK guidelines, people aged 16 or 17 are presumed to be capable of consenting to their own medical treatment, and, in specific cases, children under 16 years who have sufficient understanding and intelligence to enable them to fully understand what is involved in a proposed intervention will also have the capacity to consent to that intervention.[33] Conversely, according to international guidelines,[4] presymptomatic testing for adult-onset disorders is recommended to be made available to those aged 18 years and older, unless there it is in a child's best interest either in terms of immediate relevance for their health or of psychological or social benefits. Nevertheless, Richards[31] argued that young persons who are considered as adults on the age-based criterion of 18 years are not all necessarily truly autonomous. She pinpointed that the most important aspect of the decision-making process is the recognition that the knowledge obtained from the test result is irreversible. There is not a specific age when a person is able to give autonomous consent, but it is important to consider psychological maturity[31] that is cumulative with age, life experience and cognitive development.[34] Therefore, future studies should aim at defining the optimal moment when to undergo presymptomatic genetic tests, not only on the basis of the age, but also considering psychosocial maturity.[35] In any case, genetic health-care professionals, in the context of presymptomatic counselling, should support young adults to become aware of their own individual needs and capacities and of the fact that, sometimes, waiting to be tested may be helpful to better understand potential harms and benefits of testing.

In addition, although it is reasonable to hypothesize that undergoing testing at the right time reduces the risk of negative effects, it is

important to consider the influence of the specific disease considered: the perception and experience of harms and benefits from the test result for a potentially treatable condition (such as BRCA and FAP, etc) may not be the same as for conditions for which there are no preventive treatment or cure (such as HD).

A potential limitation of this systematic review is that all the papers analysed are based on studies conducted in only four countries with similar British historical and cultural legacies, thus the findings may not generalize to other countries with different sociocultural backgrounds, supporting the need for further studies in other contexts. On the other hand, the papers analysed spanned across several diseases, while considering similar age ranges, thus providing a comprehensive overview of how young adults deal with genetic testing overall and according to the specific disease.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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