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Supporting Information Sharing in Families at Risk of Bowel Cancer Through a Secure Website

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UNIVERSITY OF
PLYMOUTH

*Supporting information sharing in
families at risk of bowel cancer
through a secure website*

by

SELINA GOODMAN

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

DOCTOR OF PHILOSOPHY

School of Nursing & Midwifery

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Abstract:

Supporting information sharing in families at-risk of bowel cancer through a secure website

Selina Goodman

Background: Bowel cancer is a significant health threat as it is the third most commonly diagnosed cancer worldwide. Screening can detect tumours early, thus allowing treatment to reduce morbidity and mortality. However, many relatives who share a high lifetime risk of bowel cancer remain unaware of their familial diagnosis and so are unable to access genetic testing or screening. Providing family letters and verbal recommendations to patients diagnosed with a vulnerability to bowel cancer has not been sufficient to support effective communication in these families. Little is known about how electronic methods of communication could be used to facilitate communication in families affected by a genetic vulnerability to cancer.

Aim: To investigate whether a secure website could support families with an increased risk of bowel cancer to share information with their relatives.

Methods: A pragmatic mixed methods approach with four phases was used. First, patients at high risk of bowel cancer who had been advised to have regular colonoscopy were invited to participate in an anonymous cross-sectional survey administered online and through NHS clinical services (n=286). Second, more in depth qualitative data were elicited through telephone interviews with a purposive sample of volunteers (n=14). Third, a secure website was designed to help relatives share sensitive documents online, with content informed by the experiences and views of survey and interview participants. Fourth, reactions to the website from 12 volunteers were elicited through three phases of video recorded Think-Aloud interviews, which guided further refinement of the website.

Findings: The survey showed that: 43% of those at risk were first informed of the familial diagnosis by their relatives. The majority of participants needed much more practical information after learning they had an increased risk of cancer. Key issues were: a healthy lifestyle, genetic testing, bowel surveillance and talking to children. These topics were endorsed in the interviews, which also identified four main themes: **impact** of the genetic diagnosis; the need for **practical information**; the importance of **adaptation** to sharing information; and **appropriate communication** in contacts with relatives. Reactions to the website were positive; access to tailored information, plus the opportunity to store and share personal information were all welcomed.

Conclusion: A personalised web-based information resource and document sharing facility like www.familyweb.org/ could improve health communication within families. It requires further research to confirm that it is effective in practice. Such innovations could help improve early detection and treatment though increased awareness of familial cancer risk. Patients need more support from health professionals to adjust and then disseminate information about their diagnosis.

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List of abbreviations

BRCA	Breast cancer gene
DNA	Deoxyribonucleic acid
FSGI	Family system genetic illness
GP	General practitioner
GTM	GoToMeeting
HBOC	hereditary breast and ovarian cancer
HBM	Health Belief Model
HRA	Health Research Authority
IHC	immunohistochemistry
LS	Lynch syndrome
MFDG	Multi-family discussion group
MLH1	MutL homolog 1 gene
MSH2	MutS homolog 2 gene
MSH6	MutS homolog 6 gene
MSI	microsatellite instability
NICE	National Institute of Health and Care Excellence
PHR	Personal Health Record
PMS2	Post meiotic segregation increased 2 gene
NHS	National Health Service
PI	Principle investigator
PIS	Participant information sheet
R&D	Research and Development
RCT	randomized controlled trial
REC	Research ethics committee
REM	Reciprocal Engagement Model
TPB	Theory of Planned Behaviour

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List of presentations at meetings and conferences 2015 to 2018

South West of Britain Clinical Genetics meeting, Exeter	March	2015
Colorectal surgeons breakfast meeting, Plymouth NHS Hospitals Trust	April	2015
Endoscopy departmental meeting, Plymouth NHS Hospitals Trust	February	2016
Peninsula Clinical Genetics Service, RD&E Hospital, Exeter,	March	2016
N.W. Thames Clinical Genetics Service, Northwick Park Hospital, Harrow	March	2016
Lynch Syndrome UK Patient Conference, Bristol	May	2016
Lynch Syndrome information event, Wessex Clinical Genetics Service	May	2016
European Society of Human Genetics conference, Barcelona (Appendix 31)	May	2016
Joint UK/Dutch Clinical Genetics Societies meeting, Cardiff	May	2016
South West of Britain Clinical Genetics meeting, Bristol	June	2016
Polyposis MDT meeting, St Mark's hospital, Harrow	June	2016
Lynch syndrome information event, Northwick Park Hospital, Harrow	June	2016
Mustard Tree Macmillan Centre, Plymouth NHS Hospitals Trust	August	2016
Polyposis information day, St Mark's Hospital, Harrow	November	2016
Bowel Cancer Awareness Month PubhD event, Café Kiss, Plymouth	April	2017
Gynaecological cancers in Lynch syndrome International Consensus Meeting (Invited discussion group member), Manchester	April	2017
European Society of Human Genetics conference, Copenhagen	May	2017
S.W. Thames Regional Genetics service, Guy's Hospital	May	2017
International Society of Nurses in Genetics conference, Dublin (Appendix 32)	August	2017
International Society for Gastrointestinal Hereditary Tumours (InSiGHT) biennial meeting, Florence	July	2017
Joint UK/Dutch Clinical Genetics Societies meeting, Utrecht (Appendix 33)	March	2018
Lynch Syndrome UK Patient Conference, Birmingham	April	2018
Interview for Bowel Cancer West and press release (Appendix 28)	November	2017

Author's declaration

At no time during the registration for the degree of Doctor of Philosophy has the author been registered for any other University award without prior agreement of the Doctoral College Quality Sub-Committee.

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

This study was financed with the aid of a grant from the charity Bowel Cancer West.

A programme of advanced study was undertaken, which included relevant postgraduate courses to gain transferable skills. Seminars and conferences were also regularly attended, at which work was often presented in the form of posters or spoken presentations (listed above).

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Signed:

A handwritten signature in black ink, appearing to read 'S. A. Radman', written in a cursive style.

Date: .16.10.2018

Chapter One

Genetic predisposition to cancer

1.1 Introduction

Cancer is a significant health problem, with an estimated 14.1 million new cases worldwide in 2012 alone (Torre *et al.*, 2016). While cancer can affect anyone, it is now well recognised that a subset of cancers arise due to inherited genetic vulnerability (Vogelstein & Kinzler, 2004). Those individuals who have inherited a susceptibility to cancer may benefit by receiving surveillance or prophylactic surgery to reduce their likelihood of developing a malignancy or to detect the cancer early and thereby lessen morbidity and mortality of, for example, thyroid cancer, breast cancer or colon cancer (Domchek *et al.*, 2010; Guillem *et al.*, 2006; Jarvinen *et al.*, 2000). People with such an increased risk, if not already affected by cancer, would usually learn of their risk through their family (Gilbar *et al.*, 2016; Stol *et al.*, 2010). However, evidence suggests that a substantial proportion of those 'at risk' relatives remain unaware of their potential genetic susceptibility (Hodgson *et al.*, 2014; Sharaf *et al.*, 2013) and are therefore unable to access screening or take steps to reduce their likelihood of cancer. Attention therefore needs to be given to identifying how more people with such a predisposition can be forewarned of their risk and given the opportunity to seek advice and have appropriate cancer surveillance. In this chapter, I will present evidence to demonstrate how significant this problem is, how people at risk are currently identified and why further research is needed in this area. In addition, I will show that there are potential health benefits for individuals who know that they are at increased risk.

In the following chapter I will look in more detail at issues of communication in families affected with a genetic condition, what motivates patients to share information with their relatives and how new technology could help in the process of disclosure. In the subsequent chapter I will describe the systematic review I carried out to find out if there was any evidence around the use of email or websites to share confidential health information securely within families. Then I explain the research methods I used to find out about patients' experiences of sharing information within their families in Chapter Five. I set out the results of each phase of this research (Chapters Six, Seven and Eight) and how it culminated in the creation of a website designed to help families at risk of bowel cancer share information about their familial diagnosis. Finally, the results will be discussed in Chapter Nine and the implications of those results presented in Chapter Ten.

1.2 Biological mechanisms of cancer

Increased understanding of the biological mechanisms that lead to cancer may help reduce the incidence of certain cancers (Vogelstein & Kinzler, 2004). Research has shown that exposure to extrinsic factors, such as teratogens in our environment or lifestyle choices, such as smoking or alcohol consumption, can significantly increase the risk of certain cancers (Torre *et al.*, 2016). The fundamental mechanisms that underpin this process arise from the accumulation of damage to the functional instructions within each cell that are contained in long strands of deoxyribonucleic acid (DNA); where 'somatic' DNA damage in cells builds up during an individual's lifetime (Krogan *et al.*, 2015); the accumulated changes in DNA can eventually result in cells behaving or functioning differently (Vogelstein & Kinzler, 2004). The alteration may not be evident or result in any significant change in health, however in some

situations it does. Most cells within the body have two copies of DNA strand, providing two versions of the biological instructions contained within the DNA (Turnpenny & Ellard, 2016). Studies of the epidemiology of cancers have revealed that certain cancers have huge accumulations of very many alterations or mutations within the different clones of cells that constitute the tumour and are therefore termed 'hypermuted' (Roberts & Gordenin, 2014). What initiates or drives the process of carcinogenesis is not yet fully understood but it is being unravelled through the application of new genomic technologies such as next generation sequencing, which has revealed the highly varied genetic constitution of many tumours (Helleday, Eshtad & Nik-Zainal, 2014).

The hypothesis that, in relation to cancer, cells need to accumulate at least two mutations which result in a fundamental change in the action of certain genes was proposed in a seminal paper by Alfred Knudson in 1971 and since referred to as 'Knudson's Two Hit Hypothesis' (Knudson, 1971). Knudson described this mechanism based on his work on retinoblastoma, a rare tumour of the eye but it has since been shown to apply to many different cancers (Dyer, 2016). What this indicates is that people who are born with, or inherit, a constitutional variant in certain genes can have an increased risk of particular cancers (Turnpenny & Ellard, 2016). This is because all their cells already have a variation in the DNA which makes them less fit to function, in effect the first 'hit' described by Knudson. This then means that the likelihood that the DNA in any one cell will be affected by a second 'hit' or mutation in the course of that person's life is greater than for someone with two functioning copies of the same gene (Dyer, 2016). More recent work has indicated that certain cell types have a propensity to accumulate variations in the DNA due to rates of mutagenesis that are influenced

by intrinsic factors and are usually tissue specific (Wu *et al.*, 2016). Conversely, extrinsic or external factors are thought to make the most major contribution to mutagenesis and cancer development (Wu *et al.*, 2016). Without going into further detail to describe the complex and varied hypotheses of malignancy, the principle remains that a proportion of people who get cancer have an underlying genetic predisposition to certain cancers (Vogelstein & Kinzler, 2004) which greatly increases the risk of them developing cancer.

1.3 Bowel cancer as a focus

Although a large proportion of bowel cancers could be prevented (CRUK, 2017) it remains one of the most common cancers in the United Kingdom (UK) and across other developed countries, with 34,729 new cases in England in 2015 (ONS, 2017) and an estimated 1.4 million new cases of bowel cancer diagnosed worldwide in 2012 (IARC, 2012). Although bowel cancer mortality rates have decreased, there has been an increase in the number of younger people diagnosed with bowel cancer, prompting a campaign by Bowel Cancer UK to improve awareness, particularly amongst GPs, entitled 'Never Too Young' (BowelCancerUK, 2017). The likely cause of an increased incidence of bowel cancer in younger people has been suggested as being connected with increased rates of obesity and alcohol consumption in the young (Siegel, Jemal & Ward, 2009). Clearly more research is needed to investigate the complex interactions between genetic predisposition, environmental factors and epigenetic changes to understand this phenomenon. However, we are already aware that individuals with an inherited vulnerability to bowel cancer tend to be affected by cancer at younger ages than observed in the wider population (Vasen *et al.*, 2008; Vasen *et al.*, 2010; Vasen *et al.*, 2013).

I have worked in clinical genetics services since 2000 and during my work I have repeatedly been told of younger relatives being falsely reassured by their doctors that they were not at an increased risk of bowel cancer, when in fact their family history indicated they were. Although my experience is only anecdotal evidence, this situation was tragically illustrated by the death of Stephen Sutton. He was a young man who died from bowel cancer at the age of 19 in May 2014 but campaigned and raised over £5.5 million for the Teenage Cancer Trust (Harley, 2016). Although Stephen was exceptionally young to develop symptoms at 16, his father had already been diagnosed with a genetic vulnerability to bowel cancer called Lynch syndrome (LS). Consequently the family were well aware of the potentially high risk of cancer in those with the gene variant. Andy Sutton, Stephen's father, reported appealing unsuccessfully to get investigations for his son when Stephen was losing weight and suffering from persistent constipation (Weathers, 2014). However, despite the media interest in Stephen's tragic story, the support group Lynch syndrome UK describe a frustrating lack of awareness amongst clinicians and the general public, as reported by their members via their closed Facebook page. Since then, Bowel Cancer UK, as part of their 'Never Too Young' campaign, have promoted a new risk assessment tool developed for primary care (Stapley *et al.*, 2017) stating that with more than 2,300 people diagnosed under the age of 50 with bowel cancer in the UK in 2014 (CRUK, 2017) these issues need to be urgently addressed.

By contrast, public awareness of the familial nature of breast cancer increased dramatically following the publication of an article by the actress Angelina Jolie in the New York Times in May 2013 (Jolie, 2013), explaining how she had learnt that she had a BRCA1 gene variant giving her a high lifetime risk of breast and ovarian cancer. This

led to significant increases in the number of women seeking genetic testing for BRCA1 or BRCA2 gene variants (Evans *et al.*, 2014). No equivalent high profile announcements have been made about familial bowel cancer yet. However data from Holland (van Lier *et al.*, 2012) now suggests that there are likely to be many more people affected by Lynch syndrome (LS) than previous believed. These authors have reassessed estimates of population incidence based on the results of applying immunohistochemical tests (IHC) of mismatch repair (MMR) deficiency to every case of bowel cancer diagnosed under the age of 70 years. This method of 'reflex' or universal screening of incident cancers was applied prospectively to 1117 consecutive cases of colorectal cancer between 2007 and 2009 from 11 hospitals and has indicated that a high lifetime risk of cancer due to LS is something that is likely to affect many more people than previously believed (van Lier *et al.*, 2012). Current estimates extrapolated from these data now suggest that the prevalence of LS alone is around 1 in 350 (Staffa *et al.*, 2015), making it a much more common condition than previously thought. These data also indicate that probably only 5% of individuals with this condition are currently aware of their risk. Of course, some caution needs to be applied to the evidence from the Netherlands as it may be population specific. To counter that concern, there does not appear to be a high frequency of founder mutations in any of the mismatch repair genes in the Netherlands (van Lier *et al.*, 2012). However the authors do note that they have detected a higher frequency of germline mutations in MSH6 and PMS2 than previously suspected (van Lier *et al.*, 2012). The authors conclude that this result may reflect the fact that variants in MSH6 and PMS2 are of lower penetrance (Baglietto *et al.*, 2010; Caron, 2015; Senter *et al.*, 2008) so giving rise to fewer cancers, than the other mismatch repair genes MLH1 and

MSH2 (Dowty *et al.*, 2013). Therefore families with MSH6 and PMS2 pathogenic variants are less likely to present with a family history of cancer indicative of LS (Møller *et al.*, 2017).

In addition to LS, there are other rarer but well recognised genetic conditions where individuals with high penetrance pathogenic gene variants are at a significantly increased lifetime risk of bowel cancer, such as familial adenomatous polyposis (FAP), MYH associated polyposis (MAP), Peutz Jeghers syndrome (PJS) and juvenile polyposis (JPS) (de la Chapelle, 2004; Gross & Brand, 2015). The proportion of individuals affected by these in relation to all those diagnosed with colorectal cancer is illustrated in Figure 1.1 (taken from Burt 2000 and reproduced online (PDQ® Cancer Genetics Editorial Board, 2018)).

Figure 1.1 image has been removed due to copyright restrictions

Figure 1.1 Proportion of colorectal cancers due to different family risk settings taken from Gastroenterology, Burt (Burt, 2000)

Altogether, evidence of heritability from twin and kindred studies indicates that around a third of all bowel cancers occur because of an inherited vulnerability (Jasperson *et al.*, 2010; Lynch & Shaw, 2013; Papaemmanuil *et al.*, 2008).

Management for these different conditions varies based on their natural history, and it is not appropriate to detail them here, but a principle factor that can lead to some

symptoms of disease being overlooked is the unusually young ages at which pathology can develop in genetic predispositions to cancer (Giardiello *et al.*, 2014). For example, in FAP hundreds of colonic adenomas start to form from the mid-teens onwards. This then necessitates regular bowel screening by colonoscopy from age ten or eleven years and usually leads to colectomy in the early twenties (Vasen *et al.*, 2008) with the average age of diagnosis with cancer (if treatment is not received) of around 39 years (Jasperson *et al.*, 2010). Recently published prospectively collected cumulative cancer incidence data for 1,942 people with Lynch syndrome and proven pathogenic mutations calculated a 15% risk of any cancer by age 40 for women and 14% for men, rising to 75% by age 70 in women and 58% by age 70 for men (Møller *et al.*, 2017). The authors reporting these new data avoided the ascertainment bias that has previously affected estimates of cancer risk, by excluding index cases and starting when patients had their first colonoscopy. However, the data only reflected the experience of those patients receiving regular colonoscopy at those centres participating in their study. Therefore, it is logical to assume that if pre-cancerous polyps are being detected and removed at colonoscopy in their research participants, then the incidence of cancer is likely to be even higher in an unscreened group.

Screening recommendations for high risk groups start at the ages appropriate to that condition, for example, colonoscopy is recommended from 25 years in LS (Cairns *et al.*, 2010). However, such recommendations may be unfamiliar to professionals outside clinical genetic services. Indeed, research into the rising incidence of bowel cancer diagnosed under the age of 50 years have shown that younger people with symptoms are more likely to have to visit their General Practitioner (GP) several times before they are referred for investigation (Stapley *et al.*, 2017). Similar data from an

online survey concur with these findings but it is possible that the survey data could contain bias as the online survey might have attracted more responses from those patients who received delayed care (BowelCancerUK, 2016b). The evidence from the study by Stapley and colleagues (Stapley *et al.*, 2017) is likely to be more robust as their data came from a large case control study comparing the primary care records of 1661 cases of colorectal cancer diagnosed under 50 years with 3979 age matched controls.

In my experience, problems with diagnosis may be partly attributable to the fact that the high risk of cancer and the potentially young age at which individuals can be affected is not widely appreciated outside of the clinical specialties such as gastroenterology, genetics and colorectal surgery. Thus symptoms may not be recognised as significant, which in turn can lead to delays in diagnosis (Barrow *et al.*, 2015). Consequently, charitable groups representing patients are now campaigning for greater awareness, both in the public and amongst health professionals, in the hope that this will improve symptom awareness and reduce times from presentation to treatment (BowelCancerUK, 2016b; Monahan *et al.*, 2017).

1.4 Clinical responsibility to support information sharing

Patients diagnosed with an inherited vulnerability to cancer are encouraged to share information about their diagnosis with their relatives (Adelson *et al.*, 2013; Mendes *et al.*, 2016; Peterson *et al.*, 2003); however, there is evidence that many relatives remain unaware of it (Hodgson *et al.*, 2014; Landsbergen *et al.*, 2005). Clinical guidelines published in the Netherlands in 2013 (Menko *et al.*, 2013) and the UK (Lucassen & Hall, 2012) have both emphasised the responsibility of clinicians to promote and support communication in families with LS, due to the potential benefits to other family

members once a diagnosis had been made. This perspective is not new (Forrest *et al.*, 2007; Godard *et al.*, 2006) but the 2013 Dutch guidelines were preceded in the Netherlands by a qualitative interview-based study of the attitudes of clinical geneticists to this issue. The authors found that all nine of the geneticists interviewed thought it was important to inform family members of their risk but they held with the accepted practice of leaving this process up to the index patient. This was a small sample so may not be generalizable; it is interesting to note that these geneticists were aware that a significant proportion of relatives probably remained ignorant of the familial diagnosis. However, they continued to practice according to their clinical 'mores', citing the relatives right not to know and their uncertainty of their legal position in this respect, plus a lack of resources to support them contacting the wider family (Stol *et al.*, 2010).

The study by Stol *et al.* (2010) provides an insight into the perspectives of a small group of experienced geneticists and may not be representative. However, it could reflect a degree of inertia towards changing clinical practice, despite evidence to suggest that this current approach is not effective and many relatives remain at risk and unaware (Hodgson *et al.*, 2014; Pujol *et al.*, 2013; Sharaf *et al.*, 2013). In fact, the authors argue that there is a moral duty to inform relatives; which they say is shared between the doctor and the patient. They cite Beauchamp and Childress (Beauchamp & Childress, 2001) and claim that the duty to warn described by the interviewed doctors does fulfil ethical criteria described by Beauchamp and Childress of beneficence and justice, describing a moral imperative that: "*1) the person to be helped is at significant risk of harm; (2) assistance is needed to prevent that risk from materialising; (3) there is a high probability that assistance will prevent the harm (4) assistance would not pose*

significant risks, costs or burdens to the person asked to help (5) the benefit for the person to be helped outweighs the costs or burdens to the person asked to help". (Stol et al., 2010)(p.393). Therefore there appears to be an inconsistency in current clinical practice, where clinicians acknowledge that there is a duty to warn relatives, but they do not accept that this responsibility should rest with them, even though they also recognise that the index patient is not always able or willing to discharge this duty.

In France the law has recently changed regarding this issue. The problem of non-disclosure to relatives was considered so significant that there have been changes to the law so that patients are now legally required to inform their relatives of the familial diagnosis (Van Haecke & de Montgolfier, 2015). However this law does allow index patients to delegate their responsibility in this respect to a health professional (Derbez et al., 2017). This change in legislation appears to have already resulted in a shift in practice in France. In an ethnographic survey carried out over eight months in 2014, a sociologist observed altered clinical practice in a cancer genetics department in Paris following the new legislation (Derbez et al., 2017). The authors of that paper saw that clinicians in the department were now introducing the topic of passing on information to relatives at their first appointment, using a different consent form (which acknowledged the responsibility to disclose information) and the discussions were being documented more routinely in patient notes (Derbez et al., 2017).

Clearly patient confidentiality is very important and a fundamental aspect of patient trust and professional responsibility. Previously it has been suggested that a genetic diagnosis could be a 'familial diagnosis' rather than an individual one (Lucassen & Parker, 2010) and this concept should extend to the process of taking consent at the time of any genetic testing. The evidence from France does appear to endorse the

option of including a clause in the patient consent process, which draws attention to the familial nature of a genetic diagnosis (Derbez *et al.*, 2017). This would then help draw attention to the shared responsibility to inform others who might be at risk in the event of a genetic diagnosis being made (Dheensa *et al.*, 2017). Patients' perspectives on this issue have been gauged through a qualitative study (Dheensa, Fenwick & Lucassen, 2016) where patients affected by a genetic condition were interviewed regarding their views about confidentiality, consent and information sharing. Using scenarios about non-disclosure to initiate discussion on these topics, the researcher interviewed 33 patients who had been seen in clinical genetics services within the last two years regarding risk of cardiac conditions or hereditary cancers (Dheensa, Fenwick & Lucassen, 2016). Their analysis revealed two major themes, firstly genetic information is familial and therefore should be disclosed; and secondly that patients prefer to maintain some degree of control over the flow of genetic information. Although their participants broadly supported the stance of a 'relational joint account' model for genetic information (where no one family member should have control over the familial diagnosis) the participants were still mindful of the sensitive nature of some personal information. These findings provide a different perspective to the study by Stol and colleagues (Stol *et al.*, 2010), who elicited the views of clinical geneticists, although Dheensa *et al.* note that the researcher may have subtly influenced the interviews through the selection of questions posed in them (a factor in many qualitative studies). The patients who were interviewed appeared pragmatic and accepting that a familial diagnosis should be available to all family members. The authors acknowledged that each families' circumstances were unique and would influence that actual process of information sharing and that their findings

were based on reactions to hypothetical situations (Dheensa, Fenwick & Lucassen, 2016). This research further strengthens the argument that knowledge of a familial diagnosis should not be constrained by the views of particular individuals and genetics health professionals could be more proactive in the process of disseminating information. Ideally genetics health professionals could provide opportunities for follow-up in order to identify potential barriers to information flow.

One barrier to dissemination of information frequently cited by clinicians is the situation where their patient is reluctant to inform their relatives of their risk. This then presents the clinician with a conflict between their role in facilitating information sharing and the need to avoid breaching confidentiality. Fortunately, the authors of a study of genetic professionals' experience found it was rare that patients refused to inform their relatives (Clarke *et al.*, 2005). When the situation did arise, then some clinicians decided to inform at risk relatives directly in order to avoid harm and alert other family members to the opportunities to have genetic testing or surveillance for early signs of disease. Acting against the wishes of an individual patient to share information with relatives directly is likely to be considered only in exceptional circumstances in the UK but informing relatives directly after a genetic diagnosis is an alternative strategy (Aktan-Collan *et al.*, 2007; Suthers *et al.*, 2006). Direct contact with at risk relatives to inform them of the familial diagnosis is not usual practice in families with an increased risk of cancer. However direct contact by clinicians has been investigated in Finland (Aktan-Collan *et al.*, 2007) and Australia (Suthers *et al.*, 2006) with families at increased risk of cancer and this strategy was found to be acceptable to most families contacted in this manner. However, while cascade testing for pathogenic variants in families at risk of cardiac disease (e.g. hypertrophic

cardiomyopathy, Long QT syndrome or familial hypercholesterolaemia) is sometimes facilitated through direct contact, this is usually with the knowledge of the proband (Newson & Humphries, 2005; Ormondroyd *et al.*, 2014; Sturm, 2016). This is regarded as a legitimate method of contact by specialist clinicians treating these conditions and by those relatives who received notification.

1.5 Lack of awareness amongst relatives under the current system

The correct identification and treatment for people with LS has now been recognised to be an important public health issue (Giardiello *et al.*, 2014), gaining prominence and endorsed in the recent guidelines by the National Institute of Clinical Excellence (NICE) (Gulland, 2017; NICE, 2017). These guidelines draw attention to the increased risk of a range of cancers including: stomach, endometrial, ovarian, small intestine, skin, urinary tract, and brain cancer. The guidance (DG27) also explains that *“expanding testing to all people with colorectal cancer may increase the detection of Lynch syndrome and, because Lynch syndrome is an inherited condition, identify families who could benefit from cascade genetic testing to determine if other family members have Lynch syndrome. This could lead to increased surveillance and consequently improved patient outcomes through earlier diagnosis and treatment, if cancer is present.”* (NICE, 2017)(p.6)

While attention has recently been focussed on LS, it remains equally pressing that people suffering with other inherited cancer syndromes receive appropriate specialist clinical management (de Vos tot Nederveen Cappel *et al.*, 2013; Jong *et al.*, 2006; Vasen *et al.*, 2008).

The health economic argument for pathological testing of incidental bowel cancers described by Snowsill and colleagues (Snowsill *et al.*, 2014) has added weight to the

recommendations published in 2014 by the Royal College of Pathologists (RCPATH) (Loughrey, Quirke & Shepherd, 2015). The Royal College of Pathologists in the UK had updated their dataset for colorectal cancer histopathology reports, advocating universal screening or 'reflex testing' of tumour specimens. The organisation also recommended testing tumour specimens from anyone diagnosed with bowel cancer under the age of 50, regardless of family history, either by microsatellite instability (MSI) or through immunohistochemistry (IHC) tests to distinguish patients who might have Lynch syndrome (Loughrey, Quirke & Shepherd, 2015). Subsequently, NICE published their recommendations in 2017, as referred to earlier (NICE, 2017), indicating that all newly diagnosed colorectal tumours should be tested for MSI or IHC, regardless of the patient's age (Gulland, 2017). It is interesting to note that the health economic calculations were predicated on the potential to diagnose unaffected at risk relatives following the diagnosis of bowel cancer in an index case through cascade testing (Hampel, 2016; Snowsill *et al.*, 2014). Snowsill *et al.* (Snowsill *et al.*, 2015) calculated that the cost of universal tumour screening would still be cost effective even if no unaffected relatives chose to be tested. However, the cost benefit to the NHS was estimated to be substantially improved if healthy relatives were subsequently tested and received screening appropriate to their risk.

Current evidence collected via a systematic review and meta-analysis of uptake of genetic testing has indicated that less than half of at risk first degree relatives were found to access testing (Sharaf *et al.*, 2013). In fact the actual proportion of relatives who choose to have pre-symptomatic testing is likely to be even lower, as the data reviewed and analysed by Sharaf and colleagues (Sharaf *et al.*, 2013) were derived from studies undertaken either at leading cancer centres or disease registries, where

you might expect optimal information and support for newly diagnosed families (Barrow *et al.*, 2013). Whether or not relatives choose to have genetic testing is a matter of knowledge and personal choice and that is not the key issue in this study; what remains a concern is whether relatives are aware of their risk and can exercise that choice.

1.6 Increasing numbers of people diagnosed with a familial risk of bowel cancer

With the changes to colorectal tumour screening reported in Section 1.5, it is anticipated that there will be a substantial increase in the number of families diagnosed with a susceptibility to bowel cancer (Monahan *et al.*, 2017). The drivers for this change come from two areas; firstly, there are new approaches to oncological treatment (Kawakami, Zaanani & Sinicrope, 2015) and secondly, there are changes in guidance (NICE, 2017) regarding disease prevention. Both aspects will potentially result in a rise in the number of patients newly diagnosed with LS, which I will elaborate below.

Treatment for advanced stage colorectal cancer has been dominated for many years by the chemotherapy agent 5-fluorouracil (5FU). However it has now been recognised that this treatment is less effective in tumours that are mismatch repair (MMR) deficient (Ryan *et al.*, 2017a; Sargent *et al.*, 2008) and 5FU may in fact cause harm in these patients as MMR deficient cells are resistant to 5FU (He *et al.*, 2016; Jover *et al.*, 2006). Conversely, MMR deficiency has also been found to be a positive prognostic indicator in studies involving age and stage matched controls (Coppedè *et al.*, 2014; Hewish *et al.*, 2010; Popat, Hubner & Houlston, 2005). In addition, new evidence suggests that MMR deficient colorectal tumours are potentially a good target for treatments that use immunotherapies instead of traditional cytotoxic therapy and

have less morbidity associated with them (Cohen *et al.*, 2017). Therefore, for all these reasons, oncologists are now more likely to be interested in the mismatch repair status of their patient's tumours in order to optimise treatment modalities (Kawakami, Zaanani & Sinicrope, 2015; Ryan *et al.*, 2017a), thus adding weight to the argument for improving methods to diagnose LS in patients with cancer.

Taking a different perspective, authors of the economic evaluation referred to earlier (Snowsill *et al.*, 2014) examined different strategies to diagnose LS in patients with colorectal cancer. They demonstrated both patient and cost benefits when routine molecular testing of tumour samples was performed in any patient diagnosed before 70 years of age (Snowsill *et al.*, 2017). These authors found multiple benefits for this approach including: decreased cost of overall care, increased rates of identification of LS (de la Chapelle, 2004; Lynch & Lynch, 1979), better management for patients known to be at risk of several malignancies, reduced mortality in these patients and their relatives and reduced morbidity for those at lower risk (by releasing their relatives from surveillance colonoscopy) (Snowsill *et al.*, 2017). The economic argument has therefore again endorsed the need to improve existing methods used to identify families at high genetic risk of this cancer, where traditional approaches using family history and clinical presentation have been ineffective (Adelson *et al.*, 2013; Barrow *et al.*, 2015).

The proposal for universal testing of incident colorectal tumours recommended by NICE has been welcomed by geneticists, surgeons, pathologists and physicians caring for this group of patients. However, in a letter recently published in the British Medical Journal, Monahan and colleagues (Monahan *et al.*, 2017) called for a more comprehensive approach to the diagnosis and management of individuals with LS.

They noted that this policy change should result in the prevention of several hundred new cancers each year (Monahan *et al.*, 2017).

However, in order to deliver such a policy there is a pressing need for improved education across clinical specialties and probably a shift in funding. This is because the 2014 Royal College of Pathologists guidelines are not yet being fully implemented, even for tumours in people under 50, according to the results of a freedom of information request carried out by Bowel Cancer UK (BowelCancerUK, 2016a).

Although I might suggest that the slow implementation could be due to lack of knowledge about LS amongst clinicians, it is also conceivable that there is a reticence to test tumours when the patient pathway is not necessarily fully understood and there are cost implications to carrying out these additional tests on tumour specimens. In addition, a plausible perceived barrier to screening incident tumours is the timing and opportunity to consent patients to the testing. Concern about whether patients have been counselled adequately could inhibit pathologists from initiating testing (personal communication with a pathologist). This is because this type of testing of tumour specimens could have implications for family members, so it is important to counsel patients appropriately before doing the immunohistochemistry or alert them to the likelihood of further tests being indicated.

Another limitation to the efficiency of tumour screening as a route to diagnosis is the proportion of people who decide to have diagnostic genetic testing following an abnormal tumour screen. In order to inform this issue, different pathways to universal tumour testing were analysed in the United States of America (USA) (Cragun *et al.*, 2014). This study took place across 15 different institutions with 'patient follow-through' (when patients had germline testing following abnormal tumour screening

results) as one of their primary outcome measures. The researchers used surveys and interviews with genetic counsellors (GCs) to elicit what factors might influence higher levels of patient follow-through (PF) (Cragun *et al.*, 2014). Institutions were categorised into high, medium or low-PF, with frequency of PF as above 40%, between 11% - 40% or below 10% respectively. Qualitative comparative analysis (QCA) was used to identify variables associated with high or low PF. All high PF institutions shared some common conditions for implementation, namely: reflex testing for BRAF and MLH1 promoter methylation happened if tumour tests indicated MMR deficiency, that screen positive patients did not need to be referred to clinical genetics for counselling as genetic counsellors were automatically informed of tumour screening results, or alternatively the GCs helped facilitate clinician referrals, abnormal screening results were always given by a genetic nurse or counsellor and difficulty contacting patients was not given as a barrier to implementation. The adaptations that the high PF institutions made to help overcome barriers to universal testing of tumour specimens were first, the GCs reminding referring clinicians and helping facilitate referral of relevant patients for counselling, and second, the GCs being able to meet patients at follow-up appointments to initiate contact. In summary, the authors of this research report that a key element to increasing PF was a high degree of involvement by GCs in the process of consent and disclosure of results to patients (Cragun *et al.*, 2014). As might be anticipated in the USA, cost or lack of insurance cover was seen as a barrier to testing in some areas, but patient disinterest or failure to appreciate the importance of the test were also reported. Although some of the findings described above (Cragun *et al.*, 2014) may not be applicable to the UK, the importance of having GCs, or other appropriately qualified

clinicians, available to meet and counsel patients following abnormal tumour screening results is a factor that is likely to apply in the UK too. A weakness of this study was that it only sought the opinion of 21 'informants' from 15 institutions and did not necessarily gather a broader view from multiple stakeholders. The barriers were those that were perceived by the GCs and may not have truly reflected all the factors that influenced test uptake by patients or why clinicians from other disciplines found it hard to implement universal tumour testing.

Fortunately, two recent studies by Hunter and colleagues (Hunter *et al.*, 2017; Hunter *et al.*, 2015) in the USA have investigated the perspective of patients with newly diagnosed colorectal cancer about testing tumour samples for signs of LS (Hunter *et al.*, 2017; Hunter *et al.*, 2015). Initially they surveyed the views of 145 patients whose tumours were being tested for signs of LS using microsatellite instability (MSI) testing. Distress was measured using the Impact of Event Scale-Revised (IES-R) and attitudes to different statements were assessed using Likert-type scales. Family history and personal information was captured and both multivariate and multinomial logistic regression calculations were used to analyse predictors of response. Over 90% of respondents indicated that they thought they would cope with the MSI result, they understood the reason for such testing, they wanted information that could benefit their family and they thought the test should be widely available; all of these findings demonstrated the acceptability of the testing. Distress associated with the testing was reported as minimal and was not linked to patient age or stage of cancer (although the numbers in the stage of cancer subgroups were too small to have sufficient statistical power). Overall the study authors concluded that patients recently diagnosed with bowel cancer had a 'positive attitude' towards screening for

LS. However they did recommend that both patients and health care providers would benefit from education to understand more about Lynch syndrome and the fact that a lack of family history of colorectal cancer did not rule out LS (Hunter *et al.*, 2015).

Following on from the study to assess acceptability of tumour testing in patients with newly diagnosed colorectal cancer (Hunter *et al.*, 2015), the same research group looked at the feasibility of screening by gathering data on patient perspectives on both screening and sharing the test results with their relatives (Hunter *et al.*, 2017).

The authors recruited 189 patients recently diagnosed with bowel cancer and administered a survey to assess their attitudes to a range of questions regarding genetic screening. Of the 38 participants who received a high MSI result from the tumour tests, 35 completed a second survey. The results from the first survey broadly endorse the acceptability of tumour screening, with 175 patients (92.6%) who wanted to know if they were at risk of hereditary cancer, learn their genetic risks (85.6%) or understand why they had developed cancer (93%). Patients were also asked to report the likelihood that they would share screening results with their relatives. The majority of patients with at least one close living relative indicated their intention to share test results, with over 95% saying that they would share a result with their sibling or child. Of those who received a high risk result, again the majority felt it was important to share results with their relatives so that relatives could seek to reduce their risk of cancer (78%) and stating that they had a responsibility to inform their relatives of their risk (89%) (Hunter *et al.*, 2017).

Both the studies discussed above (Hunter *et al.*, 2017; Hunter *et al.*, 2015) indicate that screening patients newly diagnosed with colorectal cancer for LS without other selection criteria is acceptable to the majority of patients. They also provide

encouraging evidence that patients are likely to pass on information about test results to their relatives. Clearly intention and action may differ and there may be bias in these findings because participants interested in the issue and more likely to discuss it openly could be more inclined to take part in such a survey. The authors speculate that the provision of educational material at the time of consent could be a key element of uptake. They provided such material to potential research participants but note that authors of another study found that patients did not understand the clinical utility of the screening test (Tomiak *et al.*, 2014). These studies were done in the USA and through a health care provider (Kaiser Permanente Northwest) where the participants were all insured and most were well educated, so while informative they may not be generalizable. In the UK, where care is free at the point of delivery in the NHS (Goddard & Smith, 2001) the financial barriers to implementation are more likely to be institutional rather than personal. Consequently, despite the apparent delay in the implementation of screening for LS in all bowel cancers, it is highly plausible that over time increasing numbers of patients will be diagnosed with an inherited vulnerability to their cancer. Therefore I would advocate again that it is important to inform and support these individuals so they are able to adjust to the diagnosis themselves and disclose relevant information to relatives for whom there are health implications.

1.7 The benefits of knowing about familial risk of bowel cancer

I will now change my focus from those patients already diagnosed with a cancer to their relatives who have not yet had cancer. I would suggest that in the context of an inherited risk of bowel cancer there are clear advantages to knowing about the familial risk because such knowledge can enable relatives to receive regular bowel screening by colonoscopy (Lowery *et al.*, 2011). Alternatively, those relatives who are

tested and found not to have inherited the familial variant can be reassured that their cancer risk is not increased and they do not need to have regular colonoscopy (Cairns *et al.*, 2010; Menko *et al.*, 2013). I will now elaborate further on the potential benefits of such knowledge.

Investigations such as colonoscopy are done on the premise that cancer is likely to be detected early or even prevented through the removal of precancerous polyps or adenomas (de Vos tot Nederveen Cappel *et al.*, 2013; Jarvinen *et al.*, 2009). This form of surveillance has therefore been recommended for individuals at increased risk (Cairns *et al.*, 2010; Vasen *et al.*, 2013) i.e. with a recognised genetic condition in their family or where their family history of cancer has indicated that an increased risk of bowel cancer is likely.

In addition to targeted cancer surveillance, such individuals could take action to influence their personal risk of cancer by following advice regarding aspects of diet and lifestyle that can change the likelihood of developing cancer (van Duijnhoven *et al.*, 2013). This is something that may not be appreciated by people who have a strong family history of cancer and, in my experience, some consider that developing cancer is inevitable. Cancer arises through somatic changes to DNA (Krogan *et al.*, 2015) this means that lifestyle factors (such as smoking, alcohol or high consumption of red or processed meat) that lead to DNA damage are potentially going to result in a higher incidence of cancer (Anand *et al.*, 2008). These theories have been demonstrated through observational longitudinal studies (Song, Garrett & Chan, 2015). Conversely, having a lifestyle that is 'healthy', avoiding or reducing such factors will have particular benefit for people with an inherited predisposition to bowel cancer (van Duijnhoven *et al.*, 2013). In addition, there is now strong evidence that regular use of non-steroidal

anti-inflammatory drugs (NSAIDs) can reduce cancer incidence (Ait Ouakrim *et al.*, 2015). This has been most clearly demonstrated in the case of aspirin, with evidence from a randomised controlled trial indicated there was up to a 50% reduction in bowel cancer incidence in people with LS taking daily aspirin (Burn *et al.*, 2011). It is similarly encouraging that taking regular supplements of calcium and multivitamins have also been shown to significantly reduce the risk of cancer in people with LS (Chau *et al.*, 2016). These findings are exciting and add weight to the perspective that there is a positive benefit from people knowing their risk status. Therefore, there is now an increasing body of evidence that indicates that there are actions individuals can take to reduce their risk of bowel cancer, and these factors apply to people with an inherited vulnerability to this cancer, as well as those in the general population.

1.8 Opportunities to improve health

It would be logical to assume that if a person knew that they could influence their risk of cancer by adopting a healthier diet or lifestyle they would do so, but this is not always the case (Burton *et al.*, 2010; Quillin, 2016; Visser *et al.*, 2017). I would suggest that the process of acceptance and adjustment to living at risk is a complex one (Kenen, Ardern-Jones & Eeles, 2003) but this could be aided by providing counselling and support to those individuals who are experiencing particular difficulties. Burton and colleagues, in their review of evidence relating to health behaviours in patients and families with hereditary bowel cancer (Burton, Hovick & Peterson, 2012), concluded that people who had been affected by cancer demonstrated more healthy behaviours than people who had not had cancer. They also found that being less than 50 years of age, being male and being less well educated were all associated with more 'risk behaviours'. In addition, individuals who perceived themselves to be at high

risk of cancer were likely to anticipate the result of testing and adopt a healthier lifestyle than those who anticipated having a low risk result (Brodersen *et al.*, 2004), so perception of risk does influence behaviour but cannot fully predict it.

How personal risk is understood and acted on is a complex issue. I will touch on this issue to show that it is possible to promote knowledge, but that behaviour that promotes health does not necessarily follow. Using the Health Belief Model (HBM) (Rosenstock, Strecher & Becker, 1988) as a theoretical framework, Visser and colleagues (Visser *et al.*, 2017) conducted a series of focus groups with a total of 29 people with pathogenic variants in LS genes and therefore at increased risk of cancer. They were investigating determinants of, or barriers to, adherence to health and lifestyle recommendations (Visser *et al.*, 2017). Key barriers to adherence to a healthy lifestyle were found to be the desire to continue to 'enjoy life': participants also adopted an attitude where the diagnosis of LS was not allowed to dominate. Those who had had colorectal surgery reported finding it hard to maintain a healthy diet when they craved sugar or salt and had difficulty finding alternatives to processed meat in their diets. Some participants lacked self-efficacy regarding their compliance with health recommendations and several cited a habit of unhealthy eating as being a barrier to adherence. Interestingly, you might assume that the diagnosis of LS could be considered a 'cue to action' as described in the HBM, and for some people it was. However most said that they had not been given any advice about how their diet or lifestyle could alter their risk of cancer when they were given their diagnosis. Despite this lack of guidance, some actively sought out information about how they might improve their lifestyles, particularly after a personal diagnosis of cancer (Visser *et al.*, 2017).

The evidence from the study by Visser et al (Visser *et al.*, 2017) provides insight into some of the competing pressures and factors that can have an impact on people who learn they have a high risk of cancer. Although this group of patients reported not receiving much advice about their diet and lifestyle, this is less surprising when considering that they were diagnosed between 7 and 32 years ago. Severity of threat in terms of potential cancer diagnosis was not felt to be a motivator, which conflicts with the HBM. This may reflect the 'perceived locus of control' (Goldzweig *et al.*, 2016; Rotter, 1966; Visser *et al.*, 2017) over cancer risk which appeared to vary widely between participants; some felt they had no control over their risk of cancer whilst others felt they should do whatever they could to influence their likelihood of getting cancer. A limitation to this study is the older age of the participants (mean age 54 years) and the length of time since their diagnosis. It is possible that a younger group might have expressed different views and received more advice on this issue. However, the findings concur with those reported by Albada and colleagues (Albada *et al.*, 2012) below.

On the issue of whether lifestyle advice was given in genetic counselling I am interested in the evidence from a Dutch study (Albada *et al.*, 2012). This showed that in their cohort of women seeking advice about their risk of breast cancer (between February 2008 and April 2010) lifestyle factors were only discussed in 52 out of 192 (27%) taped genetic counselling sessions. On those occasions when lifestyle factors were raised, the discussion was mostly initiated by the woman seeking advice (56%) rather than the counsellor. How lifestyle factors (such as smoking, diet, alcohol consumption, contraception, exercise and breast feeding) influence breast cancer risk was almost never discussed with women already affected by cancer. Despite this

apparent lack of guidance, after counselling 29% of the affected women attributed lifestyle factors to causing cancer, compared with 19% prior to counselling, which was a significant difference ($p=0.003$). I would infer from this that those women felt the genetic contribution of their risk had been assessed and dismissed as the cause of their cancer, but this is only speculation. The strength of this study is the fact that the researchers analysed consecutive routine genetic counselling sessions regarding familial breast cancer and follow-up appointments. In addition, counselees were asked to complete a questionnaire providing demographic details and giving responses about how they attributed the cause of their cancer, if they had received a cancer diagnosis. Since the genetic counsellors and their clients consented to take part in the research, and were being videotaped, this might have influenced their behaviour in some way but the study design comes as close as possible to capturing data on actual clinical practice. In conclusion, there are other factors that contribute to cancer risk, some with greater weight than genetic vulnerability (Wu *et al.*, 2016). Therefore genetic counselling provides an opportunity to inform and advise patients in ways that can empower them to have greater control over their health.

Given the opportunities for health behaviour change, it is relevant to consider the evidence from the Health Information National Trends (HINTS) survey of 2013. This was a large population based survey in the USA, in which the authors analysed data from 3016 individuals, of which 135 (4%) had had genetic testing for either familial breast and ovarian cancer or LS (Quillin, 2016). These data were analysed to see if there was any difference in reported health behaviours between those who had received a genetic test and those from the general population. Across all the measured factors there was no significant difference between the two groups. In fact,

Quillin draws attention to the fact that of the 135 people with genetic tests, 58% were overweight or obese (with a BMI greater than 25), 24% were current smokers, 18% indicated that they had no regular moderate intensity physical activity and between 18% and 36% consumed less than the recommended daily amount of two and a half cups of fruit or vegetables. However, it was not possible to distinguish from the survey data if participants had received a high risk genetic test result or not and what, if any, genetic counselling they received at the time of their test. The author noted that these were all risk factors for anyone in the population and factors that were within the control of the individual to change. He speculated whether in the USA lifestyle factors are not given the same prominence in genetic counselling, where there is a greater readiness to discuss medical interventions to reduce risk such as screening for cancer through colonoscopy, mammogram or MRI and prophylactic surgery rather than lifestyle factors (Quillin, 2016). He suggested that this reticence amongst genetic counsellors may be influenced by the central tenant of a non-directive approach within counselling, but this assertion may be more valid in the USA and would require research to support it. It may be that discussion around oral contraceptive use is relatively common for women at increased risk of breast cancer, but risk reduction through changing lifestyle has not previously been supported by strong evidence to demonstrate their relevance to people at high risk of bowel cancer. Fortunately that situation is changing with more robust evidence of the benefits of smoking cessation (Pande *et al.*, 2010), healthy weight (Movahedi *et al.*, 2015) and diet being accrued (Song, Garrett & Chan, 2015).

1.9 Conclusions

In this chapter I have focussed on the clinical aspects of familial predispositions to cancer, particularly bowel cancer. I have presented some of the mounting evidence that there are opportunities to reduce the risk of disease for those people living with an increased risk of bowel cancer. There are improvements in diagnosis which are resulting in increasing numbers of families becoming aware of their genetic vulnerability to bowel cancer. However, many individuals still remain unaware of their potential risk of this cancer and what action they could take to reduce their chances of being affected by it. In the following chapter I will consider how different factors can influence communication in families and how this might impact on how relatives share their understanding of a familial diagnosis.

Chapter Two

Communication issues in families affected by a genetic condition

2.1 Introduction

In the first chapter I have explained how there are increasing numbers of people for whom knowledge of their familial diagnosis of a susceptibility to cancer provide an opportunity to access screening or improve their health. In this chapter I will focus on issues beyond the specific relevance of genetic information to families with an increased risk of bowel cancer, as I think many of the same issues are applicable across a wide range of conditions. Therefore I will draw on evidence from a range of different genetic conditions to demonstrate the common factors that influence communication in families. I will present evidence that informs our understanding of the barriers to communication in families and suggest how using new technology may provide an innovative way to improve support to families affected by genetic disease.

2.2 Implications of a familial diagnosis across different conditions

We now recognise that while the familial nature of inherited disease is implicit, it can still create a burden of responsibility for newly diagnosed patients when they recognise that they need to alert their relatives to possible symptoms of disease (de Geus *et al.*, 2014a; Ormondroyd *et al.*, 2014). Alternatively, having a familial diagnosis can provide opportunities for at risk relatives to access screening for the early detection of disease and genetic counselling to understand and empower them, through this knowledge, to make the most appropriate decisions for themselves and their family (Skirton *et al.*, 2013). Therefore, personal understanding of a familial

diagnosis, and the ability to share that knowledge with relatives for whom it also has implications, is an important issue for many people (Gaff *et al.*, 2007).

In order to prepare for a future where genomic information is increasingly incorporated into health care and needs to be understood and assimilated by patients, it is vital to optimise the way in which patients and their families are supported (Mendes *et al.*, 2017). This may involve the wider dissemination of information and knowledge about genetics and heritability from an early age (Harding *et al.*, 2017; Metcalfe *et al.*, 2008; Santerre-Theil *et al.*, 2016). It may also necessitate a greater profile of genetics in the media (Haga *et al.*, 2013). I would also argue that access to genetic information warrants the development of sophisticated web-based information that is available in different formats to suit the needs of readers of all ages and backgrounds.

Individually perceived as rare, in fact genetic diseases together impact the health of many people and are estimated to affect around 24 million people in Europe alone (EURORDIS, 2016). While inherited conditions may occur for the first time in a person, occurring 'de novo' due to a genetic change at conception, they are more commonly passed down through the family (Turnpenny & Ellard, 2016). Where a genetic condition is inherited from a parent, or both parents, there is the potential that it could affect several members of the same family. This means that when one member of the family is diagnosed, then that diagnosis probably has implications for some of their biological relatives as well.

2.3 Personal understanding is necessary before information sharing is possible

It is reasonable that someone diagnosed with a genetic condition is likely to take time to adjust to that diagnosis. They may be aware of the responsibility to inform their

relatives but fearful of the reaction they might receive. Therefore tension may exist between the needs of the individual and the needs of the family (Wiseman, Dancyger & Michie, 2010). Some individuals may lack confidence in what they understand about the condition and this, or actual lack of knowledge, can inhibit information sharing (Bartuma, Nilbert & Carlsson, 2012).

In a longitudinal study, 80 people from 16 families affected by LS were interviewed to learn about their views around the duty to inform relatives and the role of health professionals supporting this communication (Pentz *et al.*, 2005). Although those who agreed to participate may have had a more positive attitude to communication and genetic testing (i.e. individuals who didn't participate may have been more likely to have declined genetic counselling and testing) the majority of participants endorsed the need to inform relatives about their risk. Family members were seen as the most appropriate informants, but the majority of participants also supported information coming from health professionals; who were regarded as being trustworthy and more reliable informants about complex genetic information (Pentz *et al.*, 2005). These findings concur with those by Forrest and colleagues (Forrest *et al.*, 2003) who conducted a qualitative interview based study and found that, particularly in families at risk of hereditary breast and ovarian cancer, patients needed health professionals to 'legitimize' information when they were seeking to inform their relatives about the condition (Forrest *et al.*, 2003). It is therefore important that health professionals are available to families to support their sharing of information about the diagnosis and reduce the chance of incorrect information being circulated, as well as to help those who lack confidence or motivation in the process.

It has also been observed that misconceptions can occur about the way a condition is inherited in the family, leading to errors in who is thought to be at risk. For example, in some families with a dominantly inherited risk of breast cancer with a known BRCA gene pathogenic variant, brothers were not seen as at risk of inheriting the variant (Hallowell *et al.*, 2005). Where uncertainty exists about who needs to know, what they need to know and when it is appropriate to inform them, these factors can all inhibit information sharing about the genetic diagnosis (Wiens *et al.*, 2013).

2.4 The importance of timing

An intervention to facilitate communication is likely to be more successful if it comes at the right time for that individual; typically, information and supporting letters are provided to patients at the time or shortly after they are given their genetic testing result (Barrow *et al.*, 2015). This may not be the best time for patients to consider who they should contact and how they will do so, as they themselves are going to need time to adjust to their diagnosis (Forrest *et al.*, 2008). Interviews conducted with patients and their relatives affected by a range of genetic conditions indicated how important it was for the person with the condition to first seek emotional support for themselves before they could focus on the implications of the diagnosis for others. I think these findings are likely to be generalizable across conditions but the time taken to adapt to the diagnosis will vary between individuals, therefore any supportive intervention would need to be flexible and adjusted to the needs of each patient.

While it is probably inevitable that our understanding of how information is shared in families focusses on the patient or proband, characterising them as the key actor in this process, the culture of their family and the perspectives of other relatives will have a major impact on how such information is disseminated in the family (Ersig,

Hadley & Koehly, 2011; Koehly *et al.*, 2009; Koehly *et al.*, 2003; Peters *et al.*, 2011).

Koehly and colleagues (Koehly *et al.*, 2003) used random graph techniques and social network analysis to explore the relationships within five families with LS. They described how those relationships impacted on the way genetic counselling and testing was discussed within those families. Data were drawn from interviews with 36 people from the five families and described 783 dyadic relationships. They found that, in these families, two relatives were more likely to discuss genetic counselling or testing if they were closely related (first degree relatives or partners); either relative had the familial gene variant; or their relationship was marked by 'positive cohesion', 'leadership' or 'lack of conflict' (Koehly *et al.*, 2003).

The same research group (Koehly *et al.*, 2009) then went further to understand and describe the aspects of family functioning and communication that could impact on disclosure of genetic test results using 'Coloured Eco-Genetic Relationship Map's (CEGRM) to analyse the relationships and personal support networks to 183 women (Koehly *et al.*, 2009). Using this method of analysis they characterised the behaviour of different people within these 124 BRCA gene identified families, describing 'gatherers', 'disseminators' and 'blockers' to the process of sharing health information. Data were collected through semi-structured interviews where participants reflected on the different roles and emotional support provided by different people in their network. The researchers then looked for associations and characteristics associated with these different roles. Frequently 'gatherers' were also 'disseminators', and typically 'disseminators' were female close relatives who had a personal history of cancer and provided emotional or tangible support. In contrast information 'blockers' were more often male first degree relatives, or male partners.

Figure 2.1 1 image removed due to copyright restrictions

Figure 2.1 Example of CEGRM map

Taken from Koehly, L. M., Peters, J. A., Kenen, R., Hoskins, L. M., Ersig, A. L., Kuhn, N. R., Loud, J. T. & Greene, M. H. (2009) '**Characteristics of health information gatherers, disseminators, and blockers within families at risk of hereditary cancer: implications for family health communication interventions**'.

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The CEGRMs provided a way of looking at these family interactions and relationships over time and were used to further investigate the role of blockers in this population of women from families with BRCA gene variants (Peters *et al.*, 2011). A weakness of the 2009 study was the fact that all participants who informed the CEGRM were white women, 91% of whom had experienced higher education. The relationships represented on the CEGRM are described from the perspective of that one informant, so they might not have been truly representative of the situation in the family. Although probably not generalizable across other populations, the roles provided by certain individuals in these families are still the social reality for those women who

reported them. Also, these findings do concur with conclusions drawn from other studies (Patenaude *et al.*, 2006; Young *et al.*, 2017) with respect to the sort of relative who is most engaged with the task of gathering and sharing information about the genetic condition. These individuals are often 'pivotal' in the process of passing on information, they are typically women, and often older women such as mothers or grandmothers (Keenan *et al.*, 2005).

Extrapolating findings from a different type of adult onset genetic disorder, Huntington disease (HD), Forrest Keenan et al (Forrest Keenan *et al.*, 2009) investigated the experiences of young people at risk of HD and how they had learnt that they were at risk. Their research in Scotland involved interviewing 33 children, adolescents or young adults (12 males and 21 females) who ranged in age from 12 to 28 years old, all of whom were at risk of HD. Semi-structured interviews were used to explore the process of disclosure and the results make poignant reading at times. Their analysis showed that there were four types of disclosure, which varied from open discussion (termed 'having always been told') through gradual disclosure, to the diagnosis being kept a secret, and when the diagnosis was new to the family. The findings support the view that the communication norm in the family is likely to be more influential than any information that is provided by an outside agency such as the genetics services (Forrest Keenan *et al.*, 2009). These authors also found that women were often the 'gatekeepers' of the genetic information. However, rules of authority within families influenced disclosure and led in some circumstances to collusion where secrets were kept across generations or people lacked authority to tell others, such as cousins. This emphasises the impact of pre-existing patterns of communication within families for disclosure when new information comes into the

family (Forrest *et al.*, 2008). The evidence from Forrest and colleagues on this topic also demonstrated the temporal nature of disclosure; revealing how patients first seek support for themselves immediately post diagnosis by sharing information with close friends and family and it is only later when they are able to consider the implications for others (Forrest *et al.*, 2008). Their findings further support the importance of providing information in a format that can be revisited or referred to, as it could then be used at whatever time is most appropriate for that person to support their disclosure.

Since 2012 the Memorial Sloane Kettering Cancer Centre Clinical Genetics Service have been providing annual educational workshops to families with LS (Corines *et al.*, 2017). Recently reporting the results of their pre and post workshop participant surveys over that time they indicated how they decided on the range of topics covered at these meetings and the preferences of attendees. While the most frequently requested topics were cancer screening options and chemoprevention, strategies for family communication was a common suggestion as a topic for future lectures. (Corines *et al.*, 2017). In addition to this, they reported that the majority of their survey respondents would like to use online methods of communication but they also wanted more teaching in simple principles of genetics. While clearly beneficial to attendees, with high levels of satisfaction post workshop, it was evident that only a minority of those invited were able to or chose to attend (on average only 24%) with peak attendance at 75 people in the first year. The gender of attendees is not given but the majority the group (57%) had attended a previous workshop, so the responses may not be representative of their LS patient population as a whole and so may not be generalizable. The authors noted that they wished to conduct further research to

quantify the effect of the workshops on family communication. They anticipated that the knowledge gained through the workshops could assist and empower patients. They realised that there is a potential wider benefit to such events, as such workshops could enable family members who might be planning to have genetic testing to be better informed and also help them to understand and support those relatives already living with a diagnosis of LS (Corines *et al.*, 2017).

2.5 Motivations for sharing information with relatives

When a patient tells a health professional that their relative “would not want to know” about the diagnosis, this could be true but it could also stem from that individual feeling protective of themselves and their relatives. Of course it is very difficult to ascertain whether someone would not want knowledge of a disease risk without asking them directly. However a research group at Sheffield University (Heaton & Chico, 2016) attempted to do this amongst students and staff at the University using vignettes to illustrate different hypothetical scenarios. They conducted an online survey of 955 people to investigate attitudes of the public to disclosure of unsolicited information about a genetic condition to those who may be at risk following genetic testing in a relative. A range of 54 vignettes were developed with the help of focus groups and then each participant was presented with four vignettes to consider, these were selected at random. The key issues were: whether an at risk relative would wish to be contacted about their risk, whether the at risk relative’s interests should override patient confidentiality in this circumstance and whether, if they had been tested, they would be willing to give up their confidentiality to inform their relatives. Likert-type scales were used to measure attitudes. Vignettes were designed to represent a range

of disease severity, disease preventability and comparative risk in order to allow comparison about what factors might influence a decision to inform relatives.

What the researchers (Heaton & Chico, 2016) found was that people wanted to be contacted for conditions where there was something that could be done to avoid illness (the majority 91% (CI 88-94%) wanted to know if they were at risk of a fatal disease which was preventable) . Even for less serious diseases the majority indicated that they would like to be informed. However, if a condition was not preventable then 25% to 40% would not want to receive genetic information, however serious the condition. Although the data were derived from a survey of university students and staff, in their analysis the authors reweighted the data demographically to reflect the general population. Using this reweighted data they concluded that the majority of the public would want to know about a condition if some preventative action was possible, overriding the patient's confidentiality if necessary (Heaton & Chico, 2016). Respondents to this survey did not alter their views generally whether they considered the scenario from the perspective of a relative or as a patient. A small proportion would not wish to lose their confidentiality in any circumstance (1% for a fatal and preventable condition). While it would be valuable to have such a survey repeated in another, non-academic environment in case the results were biased by this population, it does provide insight into the factors influencing people's views on disclosure. These survey results do support the view that most people would want to know about their risk, at least if there was some action they could take to ameliorate the risk of illness.

This pragmatic stance to what motivates patients to inform relatives was already evident from the findings of a review by Bleiker et al (Bleiker *et al.*, 2013) which

focussed on the psychosocial issues associated with LS. Those authors synthesised the evidence from studies since 1993, when genetic testing for this condition first became available. They reported that the primary motivations for having genetic testing in people at risk of LS were: '*early detection of cancer*', '*reduction of uncertainty*', '*information on whether (continued) screening is necessary*' and '*knowledge of one's children's risk*' p327 (Bleiker *et al.*, 2013). Against these incentives to be tested, there are other factors, such as concern about impact on health insurance or insurance costs, that are frequently cited by patients as reasons not to have testing, despite changes in law to protect against discrimination of this kind (Wauters & Van Hoyweghen, 2016).

2.6 Information needs of relatives

Undoubtedly, many different variables can influence how and when information is shared within a family. Broad estimates have been made regarding the proportion of relatives informed of a genetic diagnosis and they indicate that from 20% to 40% of relatives remain unaware of a diagnosis (Gaff *et al.*, 2007). In their exploration of the ethical and professional dilemmas around enhancing family communication, Hodgson and Gaff (Hodgson & Gaff, 2013) state that communication within families cannot be forced, but needs to happen at the pace which is appropriate to the proband, or whichever individual has received the new information. If third parties, such as genetic counsellors, try to persuade someone who is reluctant to contact their relatives, they are no longer supporting that person's autonomy.

Returning to the evidence provided by Hodgson and colleagues (Hodgson *et al.*, 2016) from their RCT of an intervention using non-directive follow-up telephone calls, they found that even in families with a diagnosis of genetic disease, only a minority (25.6%

intervention vs 20.9% in control group) communicated with their relatives in such a way as to prompt them to seek genetic advice themselves (Hodgson *et al.*, 2016). Even in the group of patients with the greatest increase in contact (those with treatable disease) only 39% of relatives contacted genetics services within 18 months of the proband being given their diagnosis. It was a key element of this intervention that would infer that either relatives were choosing not to seek genetic advice within this time frame, or the process of adjustment within the family is a slow process for many families. However, it is only possible to speculate as to the motives of someone who does not respond or participate in the research.

One aspect that may influence how families perceive the condition in their family is their comprehension of which relatives are at risk and which are not (Forrest *et al.*, 2003). A family tree, or 'pedigree,' is a visual tool well recognised and utilised by clinicians but also by patients and their families (Bennett, 2012). Skirton (Skirton, 2001) suggested that using the patient's own pedigree would help them comprehend the inherited nature of their condition better than the use of diagrams. Thus, by visually placing the patient within the context of their family, they may understand the implications for themselves but also be better able to identify through the family tree which other relatives are at risk too.

How information is communicated in families also requires further analysis. Metcalfe *et al.* (Metcalfe *et al.*, 2011) explored how parents and children communicate about the genetic condition in their families. In the 33 families studied (affected by a range of genetic conditions) children (from aged 8), their siblings and parents were all interviewed, but separately. This study included families at risk of adult onset disorders (Huntington disease and familial adenomatous polyposis). Amongst their

findings, Metcalfe and colleagues found that young people had difficulty understanding genetic risks and statistics so they suggested that such figures could be represented graphically to facilitate understanding. In addition, their research found that there was a gender difference around which family members shared genetic information, with mothers often playing a key role in disseminating information. The family members involved agreed that children should usually be informed about the diagnosis by their parents. Many of the young people interviewed in this study thought parents should receive help from health professionals to support their disclosure to their children.

In an extension of the study cited above, Metcalfe et al (Metcalfe *et al.*, 2011) have taken their findings and have sought to develop an intervention to try and help families that are experiencing particular difficulties adjusting to their diagnosis and communicating about it. Their collaborative group (The Socio-Psychological Research in Genomics SPRinG Collaboration) (Eisler *et al.*, 2016) carried out a series of focus groups with families affected by a range of genetic conditions and health professionals in clinical genetics. Using the data from those, they then set up and tested a pilot intervention called multi-family discussion groups (MFDGs) where several families came together with a trained facilitator (a genetic counsellor with additional training) to discuss and explore the impact of the condition on themselves and the function of their families (Eisler *et al.*, 2016). Following the pilot MFDGs the participants were very enthusiastic about their experience and what they had gained from it. The facilitators were also excited by the success of the method having witnessed the benefit to families who had participated. The families themselves reported several benefits, these included how helpful it was to explore the common

issues experienced by families with genetic conditions and realise that they were not disease specific. One grandmother who described previously feeling stigmatized said *"I was able to talk about things I have not disclosed to anyone else. It felt a lot had been taken off my shoulders and I can go forward with a much more positive view on life."* p7 (Eisler *et al.*, 2016). I would not wish to detract from the success of the project and the potential benefit that such MFDGs might provide. However, it is perhaps an inevitable limitation of this type of study that the people who came forward to participate, while acknowledging that they had problems, may have been the families already willing to take steps to resolve those problems. The challenge would be to get effective assistance to the families who were really struggling to cope with the issue.

The common difficulty faced by the families involved in the above research (Eisler *et al.*, 2016) was the problem of how to talk about the inherited genetic condition with their children (Metcalf *et al.*, 2008). The MFDG approach appeared particularly apt since it allowed children and young people of similar ages to interact while participating in activities and sharing experiences with each other and also with their parents, other parents and caregivers. This approach has been used in other settings, with chronic diseases affecting families such as diabetes, eating disorders and mental illness and also investigated by a Portuguese group who used this approach to try to help families affected by hereditary colorectal cancer (Mendes *et al.*, 2013). The Portuguese investigators perceived the MFDG approach as working well alongside the delivery of genetic counselling to specific individuals, drawing attention to how the threat posed by a diagnosis of an increased risk of cancer is likely to have repercussions throughout the family; testing resilience, coping mechanisms and the family's ability to adjust to the threat. They set up four semi-structured meetings of

up to two hours held on Saturday mornings for one combined group that was made up of 19 people (ranging in age from 14 to 56) from four families. The success of the programme was then assessed during a focus group of participants one month later. The structure of the meetings had been predetermined with a mixture of education about the medical implications and management of the genetic vulnerabilities (LS and familial adenomatous polyposis) plus support related discussions focussing on a selection of themes such as disclosure, emotional reactions, coping strategies and how to maintain family identity (Mendes *et al.*, 2013).

I would agree with the conclusions of Mendes et al (2013) that since a genetic diagnosis does not only affect the individual in whom it is made, but also has implications for their relatives, there is a need to augment existing genetic counselling provision. Currently this is largely focussed on the individual, but it may be necessary to improve long term post-test follow-up and support adjustment within affected families to what is inevitably a chronic health threat. The results of the post intervention focus groups in Portugal highlighted the benefit felt by participants to the opportunity to share experiences and gain support from other people who were experiencing a similar situation. In fact their participants wanted longer sessions in order to have more time to talk with the other people there. The authors (Mendes *et al.*, 2013) do not comment on the availability of patient led support groups in their area, which might have met this need, but since the MFDG placed emphasis on developing both intra familial and inter familial coping strategies, it could be argued that families could benefit from both types of support and they shouldn't be mutually exclusive. However, the authors did recognise the potential bias within their sample towards people who were positively motivated to take part.

These initiatives then draw into focus how and what guidance should be given to patients and their families to understand and adjust to the familial diagnosis.

Meanwhile Otten (Otten *et al.*, 2014) has described how patients expected a variety of information sources to be available, at a range of complexity that should be tailored to the patient's particular needs. This finding (from their focus groups of patients and genetics health professionals) is now informing the development of a web-based 'recontact-app' to facilitate re-contacting genetics patients in Groningen in the Netherlands (Halbersma-Konings, 2018).

2.7 Barriers to communication within families

As standard clinical practice is to encourage and support patients to inform their family members, sometimes described as a 'family-link' approach (Mesters *et al.*, 2005), it is important to understand the barriers and facilitators to this method.

Research has in the past focussed on the accuracy of the information a proband will take away and potentially pass on to their relatives (Esplen *et al.*, 2014). Several studies have focussed on recall of risk information both for the benefit of the patient's own understanding but also making the point that this is an important indicator of the success of genetic counselling. In addition, a patient's knowledge about their condition has implications for their compliance with medical advice but also their ability to convey appropriate information to their relatives (Wiens *et al.*, 2013).

However, other evidence indicates that the type of information communicated may be quite limited. Mesters (Mesters *et al.*, 2005) found that usually only the key concepts were passed on to relatives, that there was a hereditary "danger" in the family and that there was an opportunity to have a genetic test to determine personal risk (Mesters *et al.*, 2005).

Another complicating factor is the patient's own personal theories of inheritance (PTI) as described by McAllister (McAllister, 2003) These may contradict or deviate from the Mendelian pattern of inheritance described by their health professional (Henderson & Maguire, 2000). If the patient's personal theory of inheritance does differ significantly from accepted Mendelian inheritance then I would argue that it is likely to influence their attempts to forewarn their at risk relatives, as they may perceive some relatives as less vulnerable than they actually are.

However, more pertinent evidence emerged from the Dutch study by Mesters and colleagues (Mesters *et al.*, 2005). Their interview based study with 30 people (8 men, 22 women, average age of 53 years) explored patient perspectives and experiences of sharing information within their family. Participants were selected at random from a registration database of people at risk of hereditary cancers. The authors found that not only was the concept of 'family' really confined to a nuclear family, but if patients had a bad initial experience of attempting to explain the implications of the diagnosis to their relative, this inhibited them in any further attempts to inform other people in the family. Although the findings in this study may have been influenced by recall bias (because the interviews were capturing retrospective experiences) their conclusions are in agreement with my own clinical experience. Previously, patients have described a reluctance to inform relatives where they have anticipated that their message would be rejected (Koehly *et al.*, 2003). Conversely, if patients are endorsed in their efforts and gain sympathy and support from other family members then they are likely to persist in attempting to inform everyone in the family (Gaff *et al.*, 2007; Mesters *et al.*, 2005). These findings have reinforced my conviction that health professionals need to

be alert to those families where difficulties in communication may occur and try to provide support that is tailored to the needs of those patients and their relatives.

2.8 How information sharing can be facilitated

In a study where patients were interviewed about what motivated them to tell their relatives about their diagnosis (Mesters *et al.*, 2005), they described a sense of a moral obligation, with the additional spur of anticipated regret if a family member were to develop a cancer that could have been prevented. If such motives were found to be endorsed by health professionals, this gave patients an additional stimulus to attempt contact and disclosure (Mesters *et al.*, 2005). This then provided evidence that how health professionals encourage or indicate to their patients that they should talk to their relatives about the familial diagnosis is also important, as well as what they say.

Unfortunately there are situations where the support of health professionals or the provision of written information may be of negligible use because significant barriers to communication exist in families (Chivers-Seymour *et al.*, 2009). For example, where relatives have lost touch with each other over time there may be ways of assisting or encouraging family members to overcome such difficulties to make contact again. However, where a family rift has occurred it is unlikely that outside encouragement will be sufficient for relatives to put aside their differences. Some barriers can be explored with patients with the potential to overcome them. This could occur when people feel torn between the competing demands of their responsibility to tell their relatives and their desire to protect them from distress and anxiety (Hallowell *et al.*, 2003). Chivers-Seymour (Chivers Seymour *et al.*, 2010) characterises the feelings felt in response to a need to communicate risk information to relatives as '*reactions to the role of informant*' (p.335), drawing attention to this emotionally demanding task. They

detail feelings of burden and isolation; the anticipated (unpleasant) reactions of relatives; concern about passing on technical information accurately; and worry about being the bearer of bad news (Chivers Seymour *et al.*, 2010). I think, given the anxiety that many patients experience over this task, it remains important that healthcare professionals assist and support them where they can.

A recent randomized controlled trial (RCT) conducted in Australia (Hodgson *et al.*, 2016; Hodgson *et al.*, 2014) was designed to give patients the opportunity to consider and explore the barriers to communication within their families through telephone follow-up with genetic counsellors (Hodgson *et al.*, 2014). Using techniques of non-directive counselling, the counsellors sought to explore with the participants their experience to date of communication in their family about the diagnosis, what their intention was in terms of contacting relatives, and what future plans they had for further communication. It was essential that counsellors avoided trying to persuade or advise participants as this would undermine their autonomy (Gaff & Hodgson, 2014). This complex intervention study was based on the Reciprocal Engagement Model (REM) (Veatch, Bartels & LeRoy, 2007) which postulates that genetic counselling is made up of a dynamic series of interactions between different elements. The interrelationship of three key tenets around a central concept, that the relationship between counsellor and client is integral, and recognises that:

- genetic information (hence education) is vital,
- patients are resilient, need support and their emotions matter,
- outcomes from counselling should enable the patient to manage their condition, adapt to their situation and make decisions appropriate for themselves and their families (Veatch, Bartels & LeRoy, 2007).

Part of the argument put forward by Gaff and Hodgson (Gaff & Hodgson, 2014) is that the patient themselves may have valid reasons for non-disclosure. These could include actively attempting to protect a vulnerable relative from bad news or as a defence mechanism protecting themselves emotionally from the repercussions of the diagnosis. Either way, it may be detrimental for the family for a health professional to try to overrule these reasons by persuasion. However, if the counsellor comes alongside the patient empathically and explores the difficulties or obstacles to disclosure this can promote the self-efficacy of the patient and in the long term is likely to be more effective as a strategy for facilitating information sharing (Gaff & Hodgson, 2014; Hodgson *et al.*, 2014). I think this is a very cogent argument; however, the results of the RCT that this group reported in 2016 were not as significant as anticipated. The intervention for the study (Hodgson *et al.*, 2014) was telephone follow-up by specially trained genetic counsellors at 3, 6 and 12 month intervals post clinic who delivered the intervention in accordance with the REM model. One of their primary outcome measures was the proportion of at risk relatives who contacted the Clinical Genetics Services in Victoria, Australia within 18 months of the participant's recruitment to the trial (Hodgson *et al.*, 2016). Starting with n=95 proband participants, the overall difference between the control group and the intervention group was 20.9% (112/536) of control group at risk relatives compared to 25.6% (142/554) of intervention group relatives contacting genetics. Taking account of the clustering effect within families, the adjusted Odds Ratio (OR), was 1.30; and the 95% Confidence Intervals (CI) were 0.70–2.42, giving a non-significant result of $p=0.40$. However, the group were using contacting genetics as a proxy for communication within the family which makes the assumption that relatives would wish to seek

further professional advice soon after learning about their risk. For this reason the trial would have benefited from a longer follow-up. Added to which, not all at risk relatives might live within the State of Victoria or they might have chosen to seek advice from other sources such as via the Internet or from their general practitioner. Therefore their results could underestimate the actual numbers of relatives contacted and seeking information.

This trial (Hodgson *et al.*, 2016) did find a greater difference between rates of contact in families with conditions which gave a high risk of illness to offspring (such as in X-linked conditions such as Fragile X syndrome and Duchenne muscular dystrophy or chromosomal anomalies such as translocations) with an adjusted OR 24.0, 95% confidence interval 3.4–168.5, $p=0.001$. However, the actual number of contacts for this cluster of families was relatively small, so they were comparing twelve out of nineteen relatives (63.2%) with four out of sixty relatives in the control group (6.7%), which further emphasises the need to replicate or continue this type of trial to give more robust results. Another possible weakness in their trial design was that genetic counsellors did not appear to have provided other tangible supporting information to assist probands when they were communicating with their relatives. Since the purpose of the research was to investigate a method of follow-up and support that could be delivered through existing genetics services in that part of Australia, it is possible that their standard care included the provision of leaflets or other educational resources, but that was not stated. The authors found that the highest level of contact happened following diagnosis of a condition where treatment or active surveillance was available, such as in familial cancer syndromes. For this category of patient around a third of at risk relatives contacted genetics services with only slightly more

from the intervention group compared to the control group: 30.3% (108/356) versus 28.0% (85/304) OR 1.12 (0.50–2.50). This result possibly reflected the greater emphasis that genetics professionals put on informing relatives in these families at the proband's consultation (Hodgson *et al.*, 2016). It may also demonstrate the participant's own motivation to warn their relatives and help give them access to potentially beneficial medical treatments or screening (Heaton & Chico, 2016).

2.9 Some relatives choose not to know

While providing support to those who are attempting to inform their relatives is important, it is also necessary to accept that some relatives may choose to ignore or avoid engaging with genetic services. Cowley, an experienced genetic counsellor who interviewed 15 members (out of 50) from one extended family with LS (Cowley, 2016), found that only people in that family who had received testing would agree to be interviewed. The topic of her research was how people experience genetic testing and whether that influenced family relationships. She found that the interviewees spoke in terms of a genetic test being "common sense" and framed it as a moral imperative. When discussing their untested relatives, they talked about them in negative terms, describing them as "selfish", "silly", or "stupid"; that they were "cowardly" or "fearful of the truth" p.631 (Cowley, 2016); so apparently viewing them as imprudent or morally lacking. Cowley concludes that by promoting genetic testing as a logical way to protect and promote health there is a danger of creating a 'genetic underclass' of those who decide not to participate in genetic investigations. She does also recognize that since she was known to be a genetic counsellor by her study participants this may have influenced the way they presented their opinions. Most critically, the views of the untested relatives were not heard because they did not participate in the research

(Cowley, 2016). While it is important to acknowledge the potential to negatively impact the lives of family members by seeking to disseminate information about the familial diagnosis, there is also a responsibility for health professionals to improve health where possible. This is an important issue where more research is needed to look for practical, pragmatic improvements to the problem of how best to support patients and their families dealing with the implications of a new genetic diagnosis in the family.

2.10 The value of information provided in different formats

Without providing some documentation of the detailed information given at the time of diagnosis, it is reasonable that many probands would find it difficult to recall and pass on information with confidence. The importance of educational materials and how they were used in communicating about a diagnosis of LS was investigated in the USA by Dilzell (Dilzell *et al.*, 2014). The research showed that those relatives who received written information about the condition were significantly more likely to seek further advice. Seventy-four participants (50 of whom were the first person in the family to be tested and 24 who were either their first or second degree relatives) completed online anonymous questionnaires: different questionnaires were used for probands and for relatives. In this group, the probands' reported a high level of disclosure, saying they had informed 88% of first degree relatives and 64% of second degree relatives. The material they used to support this was mainly the letter they received after their genetic counselling (53%) but other material such as letters, personal notes, laboratory details, support group information or online resources were provided to a third of relatives. More of the relatives who were given educational material sought advice from a health professional compared to those that received no

material (74% versus 22%, $p < 0.001$). However this difference could not be corroborated as none of the relatives who did not seek advice or testing were represented amongst the study participants (Dilzell *et al.*, 2014). This demonstrates a weakness in their study in this respect. It is likely to illustrate the possibility that people who took part in the research were the more proactive patients, already receptive to health information and therefore presenting as more engaged with genetics services than might apply to the wider patient population. The study was also retrospective and self-reported so liable to error of recall. However these authors (Dilzell *et al.*, 2014) do detail what types of educational material were shared and what proportion of recipients were thought to have responded to that information, which adds to evidence regarding the utility of providing a post clinic or family letter to relatives. Although this study does provide evidence of an association between receiving educational material and seeking genetic advice, it is not sufficient to determine what benefit relatives got from the material.

2.11 The use of technology and online Personal Health Records (PHRs) to share information

Written information such as letters or leaflets are most commonly provided in clinical practice (Forrest *et al.*, 2010) but making specialist information available online could be considered a logical step, and an economically feasible way to provide information to a greater number of people. It is not unusual for family members to be widely geographically dispersed which, it might be argued, could compromise sharing of information but this could be mitigated against by using online technology.

Although Internet access is not universal and could disadvantage some patients, providing information online could be a practical solution to the need to educate and

inform about genetic disease (Kardashian *et al.*, 2012). Providing information that can be tailored to the patient has been found to be particularly beneficial and more likely to be seen as relevant (Bental, Cawsey & Jones, 1999; Jacobs *et al.*, 2016; Neuhauser & Kreps, 2008). This could be viewed as an appropriate use of technology and one not constrained by the relatively limited availability of genetic counsellors or other trained health care professionals. The application of new technology to the advantage of patients affected with inherited conditions could also be an important and appropriate development in service provision. Alongside such changes, it would be necessary to ensure that those patients without access to the appropriate technology are not disadvantaged (Huxley *et al.*, 2015). Nonetheless, current evidence indicates that the vast majority of the population do regularly use the Internet (ONS, 2014) and this is predictably higher in the young who increasingly use texting, direct messaging, social media and email to communicate (Duggan, 2015). Also, widespread use of mobile technology such as smartphones, tablets and laptops can enable people to access these forms of communication throughout the day and avoid the need to carry printed documents with them (Poushter, 2016). Indeed, some studies have indicated that the opportunity to email general practitioners was particularly valued by some otherwise marginalised groups, such as people who work shifts, those with mental illness, the physically disabled and caregivers (Huxley *et al.*, 2015). While not suggesting that men are a marginalised group, there is evidence that indicates a gender difference in the utilisation of electronic communication with healthcare providers.

In a large European study (Newhouse *et al.*, 2015) seeking to describe the characteristics of people using email to communicate with general practitioners and

other healthcare organizations, their findings indicated that more men than women had used email ((29.11%, 2099/7210 versus 21.42%). They also found that the largest proportion of users was in the 16-24 age group, but the most frequent users were those who reported bad health or who had several health problems (Newhouse *et al.*, 2015). Therefore the opportunity to use email or online communication is likely to particularly appeal to those groups of people who might have more difficulty communicating with their relatives.

An additional point to consider is that of sustainability and ease of data sharing. Reducing the use of paper alongside increased use of information technology (IT) within the NHS has been a goal for over ten years, although with only limited success to date (Cresswell *et al.*, 2011). While government strategy had focussed national implementation of IT improvements across hospital trusts, this has proved unwieldy and ultimately difficult to achieve (Greenhalgh & Keen, 2013). In the interim, different hospitals and primary care trusts will have had to seek their own solutions, leading to a varied provision of electronic records and paperless solutions. Despite these challenges, sustainability is recognised within healthcare as an important aspect of procurement (Grose & Richardson, 2013) so the need to provide information in electronic format instead of paper documents is something that is supported in principle, but not necessarily carried out in practice.

When considering the specific example of patients' opportunity to access their own information and health records, the technology has been developed and provided through a range of platforms or Personal Health Records (PHRs) (Prey *et al.*, 2016). These provide a secure platform for sharing information digitally, which can be viewed

through any device connected to the Internet. This means that information stored in this format could be retrieved much more easily than if it is provided in paper format.

It is some time since evidence emerged from a randomized trial of information provision to patients with cancer indicated that giving patients' access to their own personal information reduced their anxiety and facilitated them showing the information to others (Jones *et al.*, 1999). Unfortunately the initiatives to introduce PHRs in the UK have not all been successful, with both NHS HealthSpace and GoogleHealth withdrawn in 2012 (Davies, 2012). Archer *et al.* (Archer *et al.*, 2011) in his scoping review of what contributes to a successful PHR, described how the functional utility and security of the system is important. However, he also emphasises that the purpose of the PHR and its perceived benefits need to be appreciated by patients and health professionals alike, as their acceptance of the technology is key to its adoption (Archer *et al.*, 2011; Nazi, 2013). Consequently, within an already overstretched NHS where clinical and administrative staff are under pressure to provide high quality clinical care to more people with fewer clinical staff, it is important to recognise that the motivation of health professionals to learn new skills and adopt novel technology may be reduced.

However PHRs such as 'My Health Record' and 'Patients Know Best' (PKB) would provide the opportunity for patients to control and share medical information and evidence of their diagnosis with their relatives, if they wished to do so (Bidmead & Marshall, 2016). This facility of PHRs has not yet been tested, although the use of PHRs and their effect on patient empowerment was investigated at a children's hospital in the UK. The hospital used PKB to encourage participation in health management and share test results with the parents of children with chronic

conditions. (Schneider, Hill & Blandford, 2016). The researchers used a semi-structured qualitative field study approach and they identified four styles of use amongst the families observed and interviewed: 'avoiders', 'co-operators', 'collaborators' and 'controllers'. The authors (Schneider, Hill & Blandford, 2016) used self-determination theory to guide their analysis and they argue that the provision of a PHR does not necessarily empower patients, as predicted, but may be helpful for those with certain coping styles. Although their research looked at PHR use by parents of sick children rather than adult patients seeking information to inform their own care, the issue of coping styles and motivation for accessing health information is something to be considered in the design of any system to promote information sharing (Al-Busaidi, Gray & Fiddian, 2006; Schneider, Hill & Blandford, 2016).

At the moment PHRs are not widely available across the UK, with only a minority of trusts using them (de Lusignan *et al.*, 2014; Mold & de Lusignan, 2015). Despite this, the opportunity to utilise sharing genetic information online through PHRs has not yet been investigated to my knowledge. The creation of comprehensive PHRs would require collaboration with large health institutions so that lies outside the scope of a PhD. However, investigating the acceptability and feasibility of sharing information online using a more modest platform, such as a website, would be possible.

2.12 Websites to support families at increased risk of cancer

Ongoing research at MD Anderson in the USA by Peterson and colleagues has created 'HEALTH4Families' (Peterson, 2017). This website is part of an initiative to develop an eHealth weight management intervention for people at increased risk of cancer (currently families with known BRCA gene variants or Lynch syndrome). The intervention is intended to promote healthy behaviour to help both patients and their

relatives who are overweight (Body Mass Index greater than 25) to reduce their weight and get more exercise using online weight monitoring, an online social network, and coaching by E-mail or via the telephone (Peterson *et al.*, 2017). This programme allows online communication between participants to encourage and provide peer support, so although not directly intended for communication between family members, HEALTH4Families does utilise information technology to optimise health benefit using communication within 'family teams' (Peterson *et al.*, 2014; Peterson *et al.*, 2017).

More specifically targeted at assisting communication, at least one website exists in the USA for the purpose of supporting families at increased risk of cancer (Myers, Conrad & Terdiman, 2014). It was created at the University of California, San Francisco to encourage and support their families with an inherited risk of cancer to share information about the diagnosis using an online platform called 'Kintalk' www.kintalk.org (Myers *et al.*, 2013). The researchers have attempted to measure satisfaction with the website through an online survey with users but so far have not published any results (Myers, personal communication July 2016).

An alternative approach, also in the USA, has been the development of a web-based intervention called the Family Gene Toolkit (Katapodi *et al.*, 2018). This intervention has been designed to provide genetics education about hereditary breast and ovarian cancer (HBOC), coupled with skills building around coping, decision making and family communication. Thus far a prototype of the intervention has been tested with 12 dyads of women (each dyad comprised a woman with a BRCA gene variant and her untested relative). In addition, the authors reported the results of focus groups (n=11) to discuss the two live webinar sessions and follow-up phone call delivered by the

project. Although this research is still ongoing, participants to the pilot study found the intervention acceptable, delivery was feasible and they were 'highly satisfied' with it overall. However, the focus groups valued the content of the modules but thought the live delivery difficult to accommodate in their schedules (Katapodi *et al.*, 2018).

2.13 Conclusions

In this chapter I have considered different aspects of communication in families affected by genetic conditions. The issue of disclosure of a genetic diagnosis with relatives for whom it may have implications is something that could be discussed with patients either during their preparation for genetic testing or in follow-up. Topics that might help determine the patient's perspective around this and any likely barriers to communication include: the family norms of communication around health issues, their emotional and geographical distance to relatives, the appropriate timing of disclosure and what role health professionals can play supporting the process of information sharing (Chivers Seymour *et al.*, 2010). I suggest that in seeking to facilitate family communication health professionals first need to understand the unique circumstances that their patient or client is facing and their perspective about the urgency of the task. With insight about the potential barriers or obstacles that someone is likely to face, and whether their patient feels ready to pass on information to their family members, we are then better placed to help them both practically and psychologically. With new digital technologies and changing norms of communication in families it would be timely to investigate what methods of information provision would be welcomed and feasible. In the next chapter I describe the systematic review that I conducted to find out what evidence had accrued about existing types of

electronic communication being used to disseminate health information within families.

Chapter Three

A systematic review of literature around the use of email or websites to facilitate communication about a health issue

3.1 Introduction

Electronic methods of communication are now ubiquitous in everyday life but utilised less often to communicate between healthcare professionals and their patients (Newhouse *et al.*, 2015). In order to better understand the efficacy and potential pitfalls of using electronic communication to share information about a genetic diagnosis I carried out a systematic review. This was to capture and synthesise research evidence that existed about the issue in order to inform the development of an innovation. In this chapter the rationale, method and results are presented, with a discussion of how the findings informed the subsequent phases of this research.

3.2 Background and rationale

With increasing demands and limited resources in the provision of healthcare, it could be argued that the opportunity to provide information and respond to queries from patients efficiently and sustainably would be desirable. Using electronic methods of communication could enable this increased efficiency as such communication would not be limited to a clinic setting and would not use administrative time or printed-paper. However doctors, nurses and other health professionals fear that responding to emails from patients will adversely impact on their time, and they are also concerned that emails are less secure when sending sensitive, confidential information (Sawmynaden *et al.*, 2012). However, with improving technology providing greater security for the use of email in healthcare (Newhouse *et al.*, 2015)

and alternatives in the form of password protected patient portals and websites (Ammenwerth, Schnell-Inderst & Hoerbst, 2012), such security issues are gradually being addressed.

On the basis that emails can be forwarded and links to websites can be shared, these forms of electronic communication could provide an opportunity for family members to share information that has been provided by their healthcare professional. Given its widespread use, it is credible that some studies have found that patients would like email contact with their doctors (Peleg & Nazarenko, 2012), although in practice the email provided may be used infrequently (Andreassen, 2011). The opportunity to send information by email, or make it available on websites, could be particularly relevant in genetic healthcare where a diagnosis made in one individual very often has relevance to other people in their family (Dheensa, Lucassen & Fenwick, 2017; Wiseman, Dancyger & Michie, 2010).

Although a systematic review and meta-analysis of email for health promotion or disease prevention concluded that the benefits of using email against standard mail were not clearly demonstrated, the reviewers considered the quality of the evidence to be low (Sawmynaden *et al.*, 2012). However, none of the studies included in that review had attempted to measure the impact or ease of transmission of information between relatives that could be facilitated by the use of email or websites.

More positively, the potential benefits to greater utilisation of information and communication technologies (ICT) was investigated within the context of the Cancer Genetic Services in Wales, with over 80% of the 225 patient respondents to their online survey indicating that having an email facility for queries would be "highly

acceptable". In addition, the use of "email and text messaging services" was the most common suggestion in an open-ended item regarding which initiatives should be prioritized by the service (Hilgart, Hayward & Iredale, 2012).

In order to optimise the health benefits to families where there is a shared genetic vulnerability to disease, health professionals (such as clinical geneticists or genetic counsellors) try to provide accurate information and support (Dheensa *et al.*, 2015; Edwards *et al.*, 2008). This is because there are potential significant health benefits to the relatives from doing so, such as symptom awareness, access to targeted screening, treatment and consideration of reproductive options (Menko *et al.*, 2013). Consequently, the issue of how to harness new technology to facilitate communication in families where there is genetic diagnosis or shared genetic vulnerability has implications for many people (Chivers Seymour *et al.*, 2010; Edwards *et al.*, 2008; Gaff *et al.*, 2007).

In practice there are many barriers that can influence and negatively impact on communication between relatives about a health issue. Most of these barriers are unlikely to be influenced by the method of information provision. However, it could be argued that providing information in a digital file (that can be shared electronically) could help facilitate communication where the barrier is one of relatives being emotionally or geographically distant. This would be particularly beneficial if people live in different time zones where the cost or convenience of a phone call could be an issue. Another barrier to communication exists if the patient has difficulties in approaching someone with whom they are in only sporadic or infrequent contact, or if the relationship is dysfunctional in some way, for example following a divorce in the family (Chivers Seymour *et al.*, 2010; Mendes *et al.*, 2017). In both these situations, it

could be postulated that if information was available online or via email that might help facilitate the communication by making the process less intrusive.

Within the context of families with an inherited vulnerability to bowel cancer, it was found that the provision of educational materials led relatives to seek further advice about their own risk of cancer and the opportunity for cancer surveillance (Dilzell *et al.*, 2014). It is understandable that providing timely, appropriate and accurate information about a diagnosis is likely to be important to ensure that correct information is given to relatives. However, balancing the privacy and autonomy of the individual against the need to prevent harm in their relatives remains a dilemma in genetics (Dheensa, Fenwick & Lucassen, 2016; Lucassen & Parker, 2004; Lucassen & Parker, 2010). In the UK, issues of information governance in health are guided by the 2013 Information Governance (or 'Caldicott') Review (Caldicott, 2013). Within that document a new seventh principle is stated: "*that the duty to share information can be as important as the duty to protect patient confidentiality*" (p.119 (Caldicott, 2013)). This draws attention to the need to balance the confidentiality of the individual against their relative's risk of harm when a potentially treatable or preventable condition is known in the family.

I was already aware that Personal Health Record (PHR) systems such as Patients Know Best (PKB) could enable health professionals to provide patient's access to information specific to them and allow it's onward transmission (Prey *et al.*, 2016). In addition, at least one health care provider was using a purpose built website (www.Kintalk.org) to facilitate information sharing, but to date they had only published conference abstracts regarding the purpose and development of the

website without any data about the efficacy of this approach (Myers *et al.*, 2013; Myers, Conrad & Terdiman, 2014).

Without evidence of the efficacy of using email or interactive websites as a means of sharing health related information in families it is unlikely that such methods would be adopted by healthcare professionals. Therefore it was considered appropriate to conduct a systematic review to determine what peer reviewed published evidence existed regarding the impact of using email or interactive websites as alternative ways of providing information to patients and their families, when a diagnosis had been made that was of relevance to more than one individual.

3.3 Aims and objectives of the systematic review

The aim of the systematic review was to investigate what methods of electronic communication were being used by health professionals, in order to support families communicating information about a shared diagnosis or health issue.

The main objectives were to:

- Understand what had already been done in order to identify gaps in knowledge around this topic.
- Gather evidence of the use of electronic communication methods in healthcare in order to inform the development of an innovation.
- Appreciate the potential benefits and barriers to this type of communication.

This led to an overarching question:

'What is the impact of information provided by health professionals either by email, or via interactive websites, on communication within the family about a familial diagnosis or other health care issue?'

3.4 Design

A systematic review is a method of identifying, analysing and synthesising the available evidence on a particular topic. I used the method described by the Centre for Reviews and Dissemination to search, select and analyse the relevant papers (Reviews & Dissemination, 2009). The aim of the search was to identify any peer reviewed empirical published research evidence, which was either qualitative, quantitative or mixed methods. The issues communicated needed to be health related, or a familial diagnosis. The communication was between relatives, so within families, or from a health care professional to their patient and then on to a third party of the patient's choice, but the use of email or a website needed to be assessed within the study.

3.5 Search methods

The first systematic search and review was conducted from January 1990 to January 2015 across ten electronic databases (Cochrane Library, AMED, CINAHL, EMBASE, ERIC, MEDLINE, PsycINFO, SocINDEX, Web of Science (Core Database) & EThOS). These databases had been recommended by the health faculty librarian at the University of Plymouth and although there was overlap between them this ensured a thorough approach. The search was limited to peer reviewed published evidence and not extended to 'grey literature', such as book chapters, reports or commentaries. This was because the grey literature was not considered as academically robust and trustworthy as peer reviewed evidence.

This search was repeated in December 2017 in order to ensure that the review was comprehensive over the period of January 1990 to December 2017. The process of systematic appraisal and rejection is illustrated by a PRISMA diagram (Moher *et al.*, 2009) for the combined search and review (Appendix 1). The syntax of the search terms varied according to the syntax proscribed by the different databases (Figure 3.1 is an example of the syntax used in EBSCO).

The syntax given below is an example of the search terms used in EBSCO for the systematic search of 10 databases.

I combined terms for digital communication such as "Email*, electronic communication, information technology, social media, Facebook, web-based, online mail, web#site, web#based OR interactive web-site", plus "Health, healthcare, health care, diagnos*" with terms for "Family, siblings, children*, relatives, relations, at-risk relatives" or "Famil* disclosure, famil* communication, famil* discussion, famil* discourse, communicat*, inform* relatives, tell* relatives, facilitate communication, shar* information, support tool*, communication* tool, enhance communication, discussion*" The resulting nested groups were combined with AND to link the four concepts of (i) 'family', (ii) 'communication', (iii) by 'email or social media', (iv) within 'healthcare'.

Figure 3.1 Example of syntax used as search terms

Articles were searched by abstract or topic only, in English, and full text articles were obtained for papers that were likely to meet the inclusion criteria, plus manual

ancestral searching was carried out for completeness. All searches were saved and the results imported into an EndNote X8 citation manager (Clarivate Analytics, 2016).

3.6 Inclusion and exclusion criteria

Studies were included if they were:

- focussed on the issue of communication of information regarding a familial diagnosis or shared health issue;
- involved some form of information exchange where information provided by a health professional could be passed on to a third party;
- empirical studies, including observational studies, surveys or trials;
- published in English.

Studies were excluded if they were:

- papers describing communication which was exclusively between healthcare professionals or professionals and hospitals or primary care, and without comment or opportunity to forward that communication to a patient or relative;
- papers that involved relatives or carers in the receipt of information about their unwell relative (the patient) where the information itself had no relevance to the health of that relative or carer;
- papers where the electronic communication under investigation was not using email or access to a website (for example: text messaging, or social media, as

these were unlikely to be used in a clinical context for confidential information);

- papers that provided clinical or professional guidelines on the use of electronic communication within healthcare, or made recommendations for practice;
- conference abstracts, commentaries, literature reviews or systematic reviews which did not contribute any new primary data.

3.7 Search outcome

The process of screening, eligibility and exclusion identified 3587 (2247 articles in the first search, 1340 in the second search) of which 105 were assessed in full text.

Table 3.1 Systematic search outcome by database

Database	Articles retrieved
EMBASE (OVID)	2037
PsycINFO	296
Web of Science	561
AMED	10
CINAHL	112
MEDLINE	300
SocINDEX	20
ERIC	196
Cochrane Library (Trials)	55
EThOS	0
Total	3587

3.8 Results

Only one article was identified that met the criteria:

- '*Communication Among Melanoma Family Members*' (Bowen *et al.*, 2017).

3.9 Quality appraisal

Since there was only one article (Bowen *et al.*, 2017) which met the search criteria it was concomitant that it would be included in the review. Nonetheless the quality of the above article was reviewed and scored independently by two researchers and was scored at 75% on the criteria described by Kmet (Kmet L M, 2004). I had anticipated finding articles with different methodologies, either qualitative, quantitative or mixed methods approaches in the search and Kmet's approach to quality appraisal would have been useful in that situation. This is because it allows the internal validity and comparison of studies with disparate designs (Kmet L M, 2004). However, one criticism of their checklists are that they give equal weight to a variety of factors (such as study design, methodology, sample size, reporting of results) so it would remain important to comment on overall quality.

3.10 Study findings

The initial search in 2015 did not yield any eligible evidence. One article (Crotser & Dickerson, 2010) identified nearly met the eligibility criteria but an email correspondence with the authors revealed that the information shared by family members had not been provided by a health care professional. In their study they investigated the experiences of eight women from families with a diagnosis of hereditary breast and ovarian cancer who had been recruited to the study via a

website (FORCE, 2016) and also learnt more about their risks of cancer via the same website (Crotser & Dickerson, 2010).

The only article that met the search criteria provided evidence from a randomized controlled trial of a web-based intervention to promote communication and support in families affected by malignant melanoma, the 'Suntalk Study' (Bowen *et al.*, 2017; Bowen *et al.*, 2014; Bowen *et al.*, 2012; Harris *et al.*, 2010). The authors of the article '*Communication Among Melanoma Family Members*' (Bowen *et al.*, 2017) presented quantitative data relating to the frequency of communication about family history of cancer and level of agreement about specified topics between different family members. In this study, communication between individuals who had been diagnosed with melanoma ('cases') was compared with either a first degree relative of theirs ('FDR') or another relative who was the parent of a child (aged under 18) described as 'parents'. The family triads (n=313) were either given access to the web-based intervention over the period of a year (intervention group) or given access to the same website at the end of a year (control group). Data were collected via surveys at baseline and at one year follow-up. Frequency of communication was assessed using a Likert scale but simplified to a dichotomized response of either 'frequent' or 'infrequent', thus losing some of the nuanced differences between cases, FDRs and parents. Their data did show an increased frequency of communication at 12 months and a significant difference between the intervention and the control groups. Agreement within pairs (case/FDR and case/parent) regarding a series of statements was measured again at baseline and follow-up with comparison between the level of agreement about statements in the intervention group and control group. A significant alteration in agreement was in response to the statement: "*It is important*

to check their skin for signs of melanoma" which rose from 30% to 78% agreement in the intervention case/FDR pairs ($p=.001$). However, the statement: "*tanning lamps are a good way to get a tan*" also showed a rise in agreement from 24% to 81% ($p=.001$) in the case/parent pairs of the intervention group with no significant difference in the control group. Although the authors interpret their data to support the efficacy of the web-based information and communication aid, the fact that the frequency of communication data is self-reported and only distinguishes between frequent and infrequent weakens this argument. The increase in agreement between family member pairs does appear to support the inference that the health advice and information on the website is being shared between relatives. However, participation in the study could prompt further information seeking behaviour by participants, so might not solely reflect the impact of the website intervention.

3.11 Discussion

This systematic search and review elicited only very limited evidence regarding studies where information about health issues had been provided to patients and their families via electronic means of communication. The data from the Suntalk study did indicate that receiving personalised health information via a secure website, coupled with emailed prompts to view new information on the website, had a significant impact on family members discussing their risk of melanoma. They also found an increase in agreement about statements of belief and what action to take to ameliorate that risk between baseline and follow-up in the intervention group compared to the control group.

No synthesis was possible since only one article was identified that provided empirical evidence of this issue. However, Bowen and colleagues did emphasize the need for further research to more fully understand the processes involved in communicating about a familial diagnosis or shared risk. They drew attention to the importance and potential to alert family members to a health threat that would enable relatives to make appropriate choices for themselves and seek further advice should they wish to do so.

3.12 Limitations

The inclusion criteria were specific to evidence of health professionals' provision of information via methods of electronic communication, where the recipient had the opportunity to share or pass on that information to others. The specificity of the eligibility criteria meant that only one study fulfilled the criteria. The strict inclusion criteria meant that several studies where information was provided to parents or carers about their child's condition (Brown *et al.*, 2016; Hopkins *et al.*, 2016; Osara *et al.*, 2017) were not included, although they did investigate the acceptability of using a secure website to communicate about health issues.

Conducting a systematic review was considered the most rigorous approach and necessary to identify empirical evidence. However, a realist review or scoping review that included 'grey literature' might have found more examples of the use of electronic communication in healthcare.

3.14 Conclusions

With only one relevant article to review, the search clearly indicated the dearth of evidence around this topic. It would therefore have been premature to draw any conclusions regarding the efficacy of providing more extensive and specific health information online and whether this could help those seeking to understand the impact of the diagnosis on themselves and their families. Although it has been recognised for some time that giving cancer patients online access to information that was specific for them reduced anxiety (Jones *et al.*, 1999) it may also assist those patients who wish to accurately inform their relatives. Indeed, evidence suggests that families who talk more openly about health issues appear to cope better with the diagnosis and experience less detrimental effect on family functioning (Metcalf *et al.*, 2008). Nonetheless, I did not wish to make assumptions about what patients might want, or attempt to provoke change without valid empirical evidence. I concluded that it was important to investigate the opinions and preferences of people who were themselves living with a health threat. This was necessary in order to meet their needs when designing an innovation to support communication with their relatives.

Although there are now projects focussed on the development of online tools specifically designed for information sharing (Anderson, 2016; Harris *et al.*, 2010; Myers, Conrad & Terdiman, 2014) or risk assessment (NIH, 2014) there is still very little published data to demonstrate their utility. It has been observed that in countries where government funding and infrastructure has supported national implementation, such as Portugal, Austria and Australia (Prey *et al.*, 2016), PHR systems have been more widely adopted. Therefore, I hoped that my research in this area, combined with that of other researchers, might contribute to our understanding

of how digital technology can help facilitate disclosure in families with genetic conditions and attract support for such systems at national or international level. The next chapter describes my aims and objectives based on a culmination of the evidence described in the preceding three chapters. In Chapter Five I set out the methods I used to achieve those objectives.

Chapter Four

Purpose

4.1 Introduction

The evidence presented in the first two chapters indicated to me that there was more work to be done to help people understand and come to terms with the implications of a genetic diagnosis in the family. It could also be argued that further investigation and innovation is needed to improve services and promote healthy behaviours in individuals living with an increased risk of cancer. Furthermore, the systematic review (Chapter Three) indicated that there was very little evidence relating to the acceptability, functionality and application of using either email or websites to facilitate communication in families with a shared health threat.

Enabling those people living with an increased risk of cancer to better understand their condition and share that knowledge with their relatives was the focus of my research. Looking for original ways to facilitate information sharing in a manner that is appropriate to current and future generations necessitates exploiting new technology. In the previous chapter (Chapter Three) I explained how I reviewed current evidence of health professionals providing health related information in a digital format that was then shared between relatives.

4.2 Aims and objectives

Motivated by the evidence presented in Chapters One and Two, and the limited evidence found in the systematic review, the aim of this research was to investigate whether a secure website could support families with an increased risk of bowel cancer to share information with their relatives.

The research question was therefore:

Can a secure website support families with an increased risk of bowel cancer to share information with their relatives?

In order to achieve this aim my objectives were to:

1. Explore the perspectives of patients, including their experiences of how they received information about the familial diagnosis themselves.
2. Invite patient's suggestions for improvement (if needed) in the way they were told about the familial diagnosis.
3. Investigate patient's preferences for information topics and also how they would like to receive information, including whether these varied by age or gender.
4. Explore patients' views about a secure website that provided a platform for patients to share documents about their diagnosis with their relatives.
5. Create a secure website in accordance with the suggestions of participants.
6. Investigate the acceptability and feasibility of sharing electronic documents regarding a familial diagnosis securely online using the purpose built website.
7. Test the website's function and acceptability with research participants.

4.3 Conclusions

The problems addressed within this research have already been recognised, and the importance of addressing the apparent lack of information dissemination was summarised by Bleiker and her co-authors: "*Without success in these efforts, needless deaths will continue to occur despite our knowledge of the genetic aetiology*" p.331 (Bleiker *et al.*, 2013). I acknowledge that there are likely to be limitations to the utility of providing information to families, whatever the format. However, I considered it both necessary and timely to investigate new ways of supporting families who were grappling with these challenges. In the next chapter I present the methods used to address my aims and objectives.

Chapter Five

Methodology

5.1 Introduction

In the preceding chapters I have provided the background to my research and given the context from which I have approached the issue of communication in families about a genetic susceptibility to bowel cancer. In this chapter I will give a brief background to mixed methods in healthcare research and explain why I thought it was an appropriate method to use in this context. I will present my perspective, how this informed my research questions and what methods I have used to answer those questions.

5.2 Strategy

I have taken a pragmatic approach, both in a practical and a philosophical way (Cherryholmes, 1992) to investigate the factors that influence communication in families about a genetic diagnosis. Realising at the outset that I was examining the issue with the perspective of a health professional, I decided that I needed to learn from people who had greater insight than I did, the patients themselves. In order to help guide any new development I first wanted to know more about the particular difficulties patients had encountered and conversely what approaches had been more successful. This realisation indicated to me that I needed to use an inductive approach. I considered that a qualitative interview based design for at least part of the study would be the most informative way to capture the complex personal perspectives that I wanted to contribute to the development of any proposed innovation (Pope & Mays, 2006).

I wanted to know what research had already been done about how helpful it would be to share digital documents provided by health professionals. It was also necessary to find out more precisely what evidence already existed to inform the method of sharing information electronically I would investigate. Consequently I carried out the systematic search and review of peer-reviewed published literature (Reviews & Dissemination, 2009) already described in detail in Chapter Three. The review was quite specific, looking for evidence about the use of electronic communication (email or websites) to provide information to families regarding a familial diagnosis, where the information had first been provided by a healthcare professional.

In addition to identifying existing evidence regarding the use of digital communication to disseminating health information, I wanted to capture a broad range of different patient views. I also wanted to invite as many patients as possible to comment on the use of information technology (IT) as a potential vector for receiving and sharing health information. Since evidence (Newhouse *et al.*, 2015) has indicated that men are more frequent users of email to communicate with healthcare providers than women, I was interested in whether there were any differences in attitude to this use of IT between the genders. I was also wanted to know if there were variations in perspective across different age groups, since younger people have been recognised as using more IT for communication than older people (Duggan, 2015). I concluded that these questions would be most clearly answered through a quantitative cross-sectional survey (Colton & Covert, 2007). An added advantage to starting my data collection with a survey was that it could be made available online and in a hard copy (Braithwaite *et al.*, 2003). The strategy of making the survey available online was intended to increase awareness about the research (O'Connor *et al.*, 2014). Based on

my clinical experience I also anticipated that I might have difficulties recruiting to my research if I was solely reliant on busy NHS health professionals to introduce the study to patients within their clinical time.

Therefore this combined complementary strategy appeared the most appropriate, combining both post-positivist and constructivist views and using a combination of deductive and inductive approaches (Johnson & Onwuegbuzie, 2004). Combining quantitative with qualitative methods of data collection sat within the increasingly used pragmatic approach (Borglin, 2015; Collins & O’Cathain, 2009; Mayoh, Bond & Todres, 2012) to health care research into complex interventions and was aligned with the Medical Research Council (MRC) Guidance (Craig *et al.*, 2008). The table below (Table 5.1) gives a summary of the pragmatic perspective, illustrating that this approach encompasses different views.

Table 5.1 Overview of the pragmatic world-view (taken from Borglin, p.34 2015)

Pragmatism

<i>Methodology</i>	Mixed- methods design
<i>Ontology</i>	Multiple ways of viewing, hearing and understanding the world.
<i>Epistemology</i>	Knowledge is not neutral as influenced by human interest. Hence knowledge is formed by both objective and subjective values.
<i>Rhetoric</i>	Meta-inference, inference quality and inference transferability.

I subscribe to the argument that mixing approaches to health sciences research can give rise to a more holistic understanding of the phenomena under scrutiny (Ivankova, Creswell & Stick, 2006; Tashakkori & Teddlie, 2010). However I am aware that others would argue that it is not possible to successfully combine approaches that stem from very different research paradigms, since they arise from contrasting sets of assumptions about what reality and knowledge really are (Guba, 1990). Mixed method research runs the risk of being muddled in its approach by seeking to assimilate the opposing ideas of the positivist scientific paradigm of quantitative research and the interpretivist or constructivist paradigms of qualitative research (Mayoh, Bond & Todres, 2012). Clearly there are many different ways methods can be mixed. However it should be possible to remain mindful of the tension that exists between these paradigms in order to apply the alternative approaches as complementary contributions to understanding the research question within a single study (Sale, Lohfeld & Brazil, 2002). Pluye and Hong (Pluye & Hong, 2014) describe the breadth of the discipline of mixed methods research, encompassing as it does many combinations of approach, and they proposed the following definition:

“Mixed methods research is a research approach in which a researcher or team of researchers integrates (a) qualitative and quantitative research questions, (b) qualitative research methods and quantitative research designs, (c) techniques for collecting and analyzing qualitative and quantitative data, and (d) qualitative findings and quantitative results” p. 30 Pluye & Hong 2014.

This definition makes the distinction that the different approaches should be applied to different and appropriate aspects of the research question, forming a kind of ‘jigsaw’ of interconnected parts rather than an undisciplined amalgamation of merged techniques and findings.

5.3 Design

In order to investigate my research questions as stated above in Chapter Four (Section 4.2) I anticipated that the integration of different methods of enquiry would also provide more scope for an iterative process of development of the innovation. This is where the design of the project would allow for feedback loops where there was sufficient flexibility for emerging insights to be incorporated into the investigation. This would also link the process of testing feasibility with development and vice versa as described in the MRC guidance (Craig *et al.*, 2008). The intention was to gather information with both depth and breadth in order to more fully investigate such complex phenomena (Mayoh, Bond & Todres, 2012). This type of convergent mixed method design was utilised by Mayoh *et al.* (Mayoh, Bond & Todres, 2012) when they used a questionnaire to gather broad quantitative data concurrently alongside qualitative interviews conducted with a group of participants drawn from their survey respondents.

Previous studies which have sought to investigate patient experiences and satisfaction with health care have concluded that quantitative survey measures are sometimes insufficiently sensitive to measure small changes or variability in quality of care experienced by patients (Andrew *et al.*, 2011). In the context of this investigation into patient views of how they would like information provided, quantitative data was necessary to distinguish some of the differences in attitudes between different groups. However a cross-sectional survey was unlikely to provide sufficient insight into the direction needed for improvement and therefore integrating qualitative data was necessary to provide that understanding (Andrew *et al.*, 2011)

In conclusion, the design of this research project was a pragmatic mixed methods study (Ivankova, Creswell & Stick, 2006). In order to capture the views and experiences of a range of individuals and achieve triangulation (Creswell & Clark, 2007) I combined both a quantitative approach and a qualitative approach in the different phases of the research. Through triangulation I sought to achieve consistency and convergence of the conclusions reached. I applied these methods in order to attain some complementarity (Borglin, 2015) as the different approaches of the questionnaire and interviews were intended to investigate different aspects of the problem of sharing information in families and thereby lead to deeper interpretations and conclusions (Farquhar, Ewing & Booth, 2011).

5.4 Methods

The first phase was a cross-sectional survey (Colton & Covert, 2007) of individuals who were already aware that they were at increased risk of bowel cancer. The questionnaire was self-completed with a mixture of closed and open questions. I sought to elicit the experiences of participants about how they had learnt of their risk; what information they had received at the time; what further information they had looked for and how they felt this process should be improved. In addition, participants in the survey were asked to reflect on alternative methods of information provision and how they themselves might communicate with their relatives about such issues. The survey consisted of 20 questions, which included six optional open questions with free text boxes which invited participants to comment or suggest improvements to the situation they had experienced. Hence these open questions were intended to capture qualitative data to supplement responses to the multiple choice questions. The survey was launched online via a link on the charity website Lynch syndrome UK

in December 2015. More advertisements were placed online via other organisations in January 2016, which are described in more detail below in section 5.8.3.

The second phase of the Family Web study was gathering qualitative data about the same issues through semi-structured telephone interviews with a purposive sample of survey participants who had volunteered to help with further research. These interviews commenced in April 2016 while the Phase 1 survey was still ongoing. This allowed a convergent approach to the data collection with adaptation of the interview questions to further investigate issues that were emerging from the survey data.

The third phase was development of the website innovation with recently diagnosed patients (diagnosed within two years of receiving their invitation to participate).

Guided by the data gathered through the survey and telephone interviews, website content was written to meet the needs of this patient group. The website was then tested through a series of Think-Aloud interviews with twelve participants, allowing an iterative process of data collection, analysis and adaptation of the website. The people who participated were either patients recruited through the clinical genetics services or respondents to the survey who had volunteered for interview and were diagnosed less than two years before their participation in the survey. The criteria of time since diagnosis was confirmed by me prior to consenting the respondents.

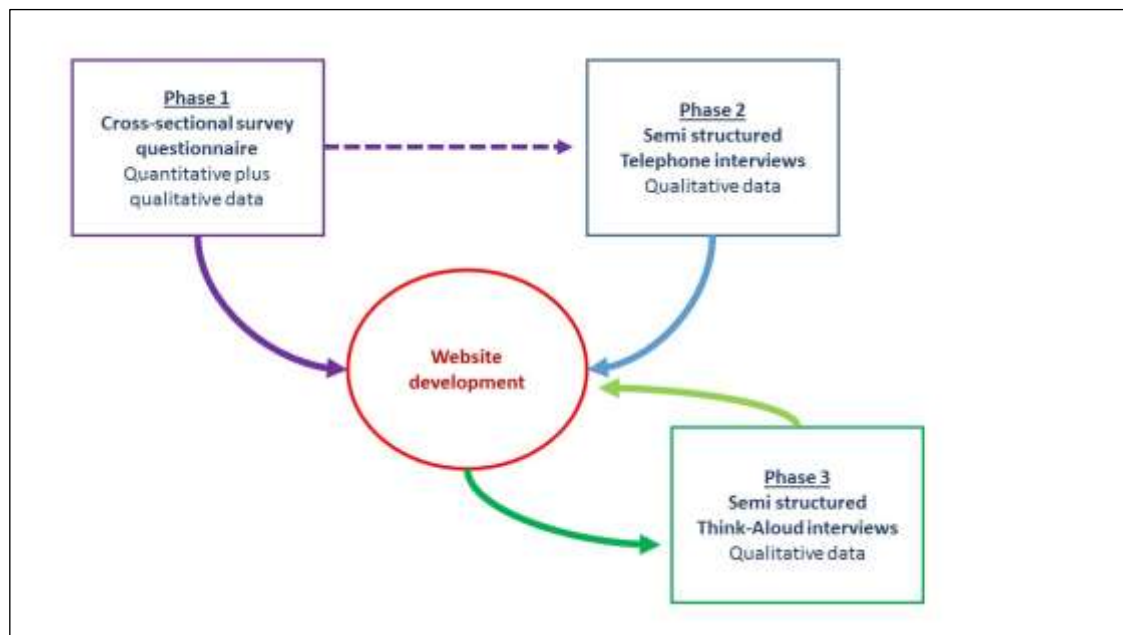


Figure 5.1 Illustration of the interaction between the different phases of the study

See the Family Web Study timeline (Appendix 9) for illustration of the timing of recruitment and data collection across the three phases of the study.

In order to facilitate contact by potential participants a generic email address was set up (familyweb@plymouth.ac.uk) and a mobile phone was purchased. Both the mobile phone number and email address were on the advertisement (Appendices 10 and 13) and Participant Information Sheet (PIS) (Appendix 15) to give a choice of contact method.

5.5 Patient involvement

I wished to involve a group of individuals who were from families with a genetic diagnosis or who had a personal diagnosis of an increased risk of bowel cancer. This was in order to ensure that the study was aligned to the needs of the patient group it was intended to investigate. Therefore an appeal for volunteer patient advisors was displayed at a national meeting of the support group 'Lynch Syndrome UK' in Birmingham in April 2015. This was a printed notice with a sign-up sheet briefly

describing the study and requesting help which was displayed near the coffee area at the conference. Slips with my contact details were also made available and I had been introduced to all participants at the event to encourage those who might want to approach me for more information about the study. Eight people signed the sheet, providing their telephone numbers and email addresses and indicating that they would be interested in being patient advisors. I contacted these people by telephone or email to discuss how best to proceed. Six people agreed to review the draft survey questionnaire, patient information sheets and consent forms, providing advice about the wording of these documents and the questions in the questionnaire. These same six people then completed two questionnaires each of the final version of the questionnaire to enable validation of the online versus the paper copy questionnaire.

In addition to the patient volunteers recruited through Lynch Syndrome UK, the study was registered on the National Institute for Health Research INVOLVE database [<http://www.invo.org.uk/>]. This public access database was seen as a way of encouraging public involvement in the research (Appendix 2). Providing details of the study on this platform (Goodman, 2015) was intended to broaden the opportunity for people who had an interest in this issue to become involved as lay advisors to the project, however no approaches were received via INVOLVE.

5.6 Approvals process

5.6.1 Ethical approval

Application for National Health Service (NHS) ethical approval was submitted on 3rd August 2015, presented to the Health Research Authority (HRA) South West Research Ethics Committee (Appendix 5) on 3rd September 2015 and full HRA ethical approval granted on 8th October 2015 (Appendix 6).

Initially twelve different clinical geneticists had been approached by letter (Appendix 16) or email as potential clinical collaborators, with later face-to-face discussions. Verbal and written agreement was reached with clinicians who were going to be principal investigators (PI) for the study at West Midlands Regional Genetics Service in Birmingham, All Wales Medical Genetics Service in Cardiff, Peninsula Clinical Genetics Service in Exeter, North West Thames Regional Genetics Service and the Guy's Hospital Department of Clinical Genetics in London, plus the Colorectal Surgery and Endoscopy units at Plymouth Hospitals NHS Trust in Plymouth. Plymouth University ethical approval was granted in October 2015 (Appendix 4) on Chairman's action following notification of the HRA approval.

Timeline of ethical & research approvals process in relation to phases of data collection

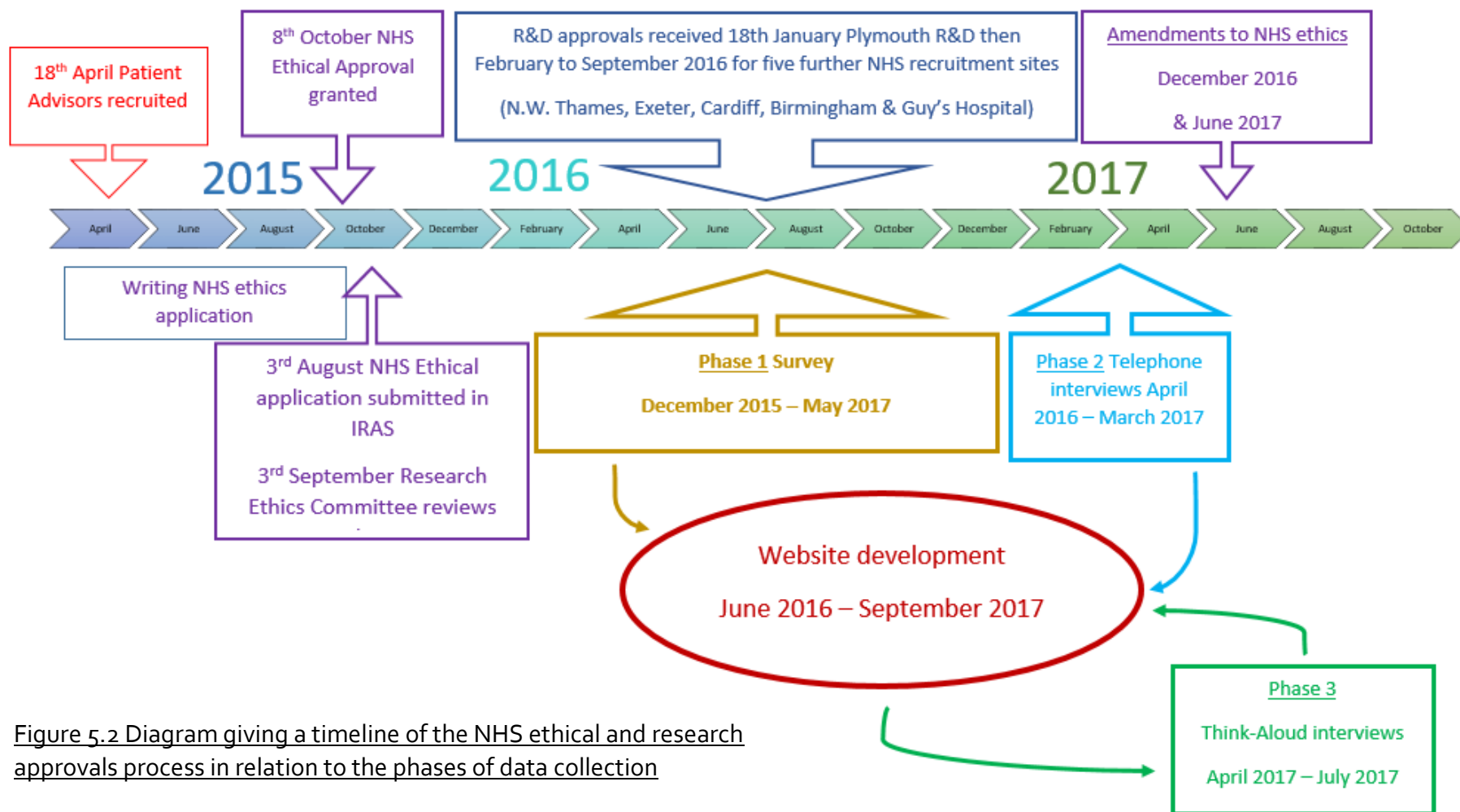


Figure 5.2 Diagram giving a timeline of the NHS ethical and research approvals process in relation to the phases of data collection

5.6.2 Application and amendments

Approval for local site specific research and development (R&D) was obtained on 18th January 2016 at the lead site of Plymouth (Appendix 7). Subsequent applications to the other local sites were made with supporting localised documentation in accordance with HRA guidelines. The site specific forms were submitted via the IRAS online platform. Due to local variations in requirement, approval was obtained across the different recruitment sites between January 2016 and September 2016. An additional application to the Health and Care Research Wales Permissions office was made in order to have authorisation to recruit via the All Wales Clinical Genetics Service (Appendix 8).

A letter of access (LoA) was provided by Plymouth Hospitals NHS Trust in January 2016 to allow me to visit the site and assist clinical staff in their decisions about which patients were eligible and should be approached about the study. This was used by me to assist staff in the Endoscopy unit where eligible patients were not necessarily known to the Peninsula Genetics Service. Subsequently I was granted a Research Passport on 8th February 2016 which allowed me to visit clinical units if necessary to assist local research teams in any of the six recruitment sites.

A substantial amendment to the HRA ethical approval was submitted in November 2016 (as previously arranged with the HRA ethics committee). This amendment (Appendix 22) provided more detailed information regarding the content of the Family Web website and was necessary prior to commencing recruitment to Phase 3. A non-substantial amendment was made in June 2017 (Appendix 23) to extend the period of recruitment to 30th September 2017 as the projected 14 months of

recruitment was due to expire at the end of June 2017 and more volunteers were still needed at that stage.

5.7. Recruitment

5.7.1 Amended recruitment criteria

The amendment to the HRA ethical approval described above also included a change to the protocol (Appendix 3) that allowed broader opportunities for recruitment to Phase 3. This change allowed for the recruitment of people who had already completed the Phase 1 survey, as long as they were diagnosed within the last two years and had indicated that they would be happy to be interviewed.

Recruitment to the study was either online or through NHS clinical centres. More detailed descriptions of each part of the study are given below within the relevant section for each phase.

5.7.2 Participants

The study sought to capture the views, opinions and experiences of family members living with a genetic susceptibility to bowel cancer. Therefore the inclusion criteria were that participants were over the age of 17 years and also one of these below.

- Part of a family which was deemed to have an increased risk of bowel cancer due to their family history, or where a genetic vulnerability to bowel cancer had been found.
- Had themselves been recommended to have bowel screening by colonoscopy on the basis of their family history of cancer, or where they were from a family where regular colonoscopy has been recommended for this reason.

- Or had been diagnosed with cancer, which they had been told was due to an inherited vulnerability to bowel cancer, such as Lynch syndrome, where their close relatives had been recommended to have regular colonoscopy.

Participants needed to have had time to adjust to their diagnosis so they were only eligible if their diagnosis (genetic or cancer) was made three or more months before recruitment. All participants had to be competent in reading and speaking English to take part in the study.

Anyone eligible for this research would have already been given a recommendation to have regular bowel screening on the basis of their increased risk of bowel cancer.

Since the criteria for eligibility was that they should be aware of their risk, the concept should not have been novel or alarming to them. However, prior to potential participation in the study, patients were provided with information sheets explaining whether they were eligible and what the study involved. Every information sheet included my telephone number and email address should participants have any questions. I had expected that potential participants might have had questions about their risk, their eligibility or the study in general. In fact over the course of the study, only six potential participants contacted me by telephone, two to clarify if the survey was still open and four to discuss what would be required in order to take part in the Think-Aloud interviews.

Patients were excluded from taking part in the study if they were receiving active treatment (radiotherapy or chemotherapy) for any cancer or were diagnosed with any cancer within the previous 3 months. However, patients who were considered in

remission or who were taking maintenance medication were considered eligible to take part as long as they were diagnosed at least three months previously. These criteria were to avoid giving greater burden to cancer patients while they were experiencing an acute phase of their illness.

5.8 Phase 1 anonymous cross-sectional survey

5.8.1 Phase 1 Design

In Phase 1, a cross sectional survey design was used (Morris, 2004) administered both online (using SurveyMonkey <https://www.surveymonkey.com/>) and in paper format (Stern, Bilgen & Dillman, 2014). The questionnaire was designed to elicit the views of a broad cohort of individuals. The paper copy version was an eight page A4 booklet and is reproduced in Appendix 18. The overarching objective of the questionnaire was to assess participants' experiences and attitudes to different types of communication both within the family and with health professionals; what information they had received and whether this was all they wanted. I was also interested in participants' views on what could be improved and the topics where they felt they would like more information.

I wanted to look at the influence of gender as my own clinical experience and published evidence both indicated that women are more likely than men to talk about the genetic diagnosis in their family (Bartuma, Nilbert & Carlsson, 2012; Chivers Seymour *et al.*, 2010; Koehly *et al.*, 2009). In addition, men and women have been found to use IT differently to communicate, with men being more task focussed in their communication (Kimbrough *et al.*, 2013). Also men have been found to use email more frequently to contact their GP (Newhouse *et al.*, 2015) therefore I was particularly interested in any gender differences in responses.

Younger people are recognised as being more frequent users of the Internet, email and social media for communication (Newhouse *et al.*, 2015; Poushter, 2016). In order to investigate novel methods of providing information about a genetic diagnosis I had envisaged that younger participants to the survey would indicate a preference for receiving information by email, a website or via social media.

I was also aware that by making the survey available online this might have attracted responses from people who were more confident or comfortable with digital communication (Sax, Gilmartin & Bryant, 2003). The questionnaire was also available in a paper copy format in order to try and elicit the views of all eligible patients.

However, I expected that there might be differences in response to questions about preferences of mode of information provision between those who responded online and those who responded via the paper questionnaire.

Participants were asked if they had been diagnosed with bowel cancer, any other cancer, or no cancer, as I expected that the information needs might be different between these groups. Evidence has shown that people diagnosed with cancer have less anxiety when given information that is personalised or specific to themselves (Jones *et al.*, 1999). Also, relatives who are living with the knowledge of their risk of cancer, but currently well, might have different needs and interests from those people already diagnosed with cancer.

The questionnaire was available online to reach respondents across a wide geographical distribution, to reduce costs and facilitate completion (Fink, 2012). The target for recruitment was 300 participants to the survey phase of the study. I had calculated that if half of the projected sample of 300 were to give clear preferences,

for example for information provided by email, this would give a 95% confidence interval of 44% to 56% for that estimate. This was an acceptable level of precision for the study.

The concurrent triangulation design of this study allowed for data collection from the questionnaire responses to continue while some participants were taking part in the Phase 2 telephone interviews and Phase 3 Think-Aloud interviews.

5.8.2 Phase 1 Participants

Patients were considered eligible if they had already been advised to have regular bowel screening by colonoscopy to prevent or detect cancer in accordance with guidelines (Cairns *et al.*, 2010). The criteria are described above in Section 5.7.2 so it is not necessary to repeat them here in detail, apart from noting that patients were eligible because of their relationship to someone who had been diagnosed with a genetic vulnerability to bowel cancer, whether or not they had chosen to have a molecular genetic test to find out if they had inherited the familial genetic variant. The key aspect of eligibility was whether they or their relative had been advised to have regular bowel screening by colonoscopy. In this phase of the study the experiences of being informed of the diagnosis and sharing information about that in the family was sought from participants. Therefore research teams and clinicians at the recruitment sites were encouraged to approach patients who had been given their diagnosis over two years previously.

5.8.3 Phase 1 Recruitment

Potential participants from across the UK were approached through online advertisements (Appendix 10) and links on the charity websites of:

Lynch Syndrome UK (<https://www.lynch-syndrome-uk.org/research>)
(Appendix 12)

Bowel Cancer West (<http://bowelcancerwest.com/>),

Beating Bowel Cancer via their online community forum
(<http://community.beatingbowelcancer.org/forum/research-and-media-opportunities/>)

FAP Gene Support Group (<http://www.fapgene.com/phdsurvey.html>)

Also cancer charity Macmillan through 'Macmillan Evidence' tweeted a link to the survey (https://twitter.com/mac_evidence?lang=en as @Mac_Evidence)

In accordance with the planned process for recruitment (Appendix 11) A4 printed advertisements (Appendix 13) were put up and information leaflets were distributed in the endoscopy clinic, in colorectal surgical outpatients' clinics and the Macmillan Centre in the local recruitment centre in Plymouth NHS Hospital Trust. In addition, locally headed copies of the PIS (Appendix 15) and invitation letters (Appendix 17) were distributed to eligible patients through the approved recruitment sites at five NHS clinical genetics services in England and Wales. I was recruiting through different regional genetic services, plus online, in order to involve participants from different geographical areas.

Potential participants who were considered eligible by clinical staff after checking against the checklist (Appendix 14) were approached during their clinic visit, posted information about the study with their post clinic letter, or approached by letter via the research administrators at each recruitment site. A reply paid envelope was provided for those participants who used the paper format questionnaire.

The survey questionnaire concluded with an invitation to take part in further research. If participants, having read and understood the PIS and consent form, wished to be interviewed they were asked to contact the research team or provide their contact details on a separate sheet or via a different webpage, thus ensuring participant anonymity was preserved.

5.8.4 Phase 1 Data collection methods

Data were collected through completion of the survey questionnaire, either online or in paper copy. Nominal categorical data were collected using fourteen multiple choice questions. There were also six open questions with free text boxes inviting more detailed responses or elaboration to some answers. In addition, one question had a Likert type scale giving a range of options between “very unhelpful” and “very helpful” to different formats for receiving information (see questionnaire in Appendix 18). The items on the questionnaire were divided into sections. The reliability of the questionnaire was only assessed in terms of the variation in responses by the patient advisors who answered both a paper copy and an online copy of the questionnaire. No attempt was made to assess internal consistency or construct validity of the questions prior to data collection.

5.8.5 Phase 1 Data analysis methods

The data collection strategy allowed for a responsive dynamic and evolving interpretation of the qualitative data in conjunction with the process of gathering more quantitative data. This was a nested analysis (Lieberman, 2005) which utilised the benefits of both methods of data collection simultaneously and allowed for the investigation and interpretation of this complex issue. Qualitative data from free text responses were coded and analysed for recurrent themes (Joffe & Yardley, 2004;

Leech & Onwuegbuzie, 2011) using NVivo qualitative data analysis Software (QSR International Pty Ltd. Version 11, 2015). The quantitative data from the cross sectional survey (Colton & Covert, 2007) was analysed using descriptive statistics in Microsoft Excel (2016 version) and using SPSS software (IBM SPSS statistics version 22).

The majority of the questions in the survey elicited nominal categorical data, with only two questions having ordinal responses and the remainder being free text.

Consequently the statistical tests that were appropriate were bivariate analysis through contingency tables and non-parametric tests. Pearson's Chi Square or Fisher's exact tests were used to give a measure of association between categorical variables. Since Chi Square is calculated based on the sum of the differences between the observed and expected counts in each cell, it is less accurate for small samples where the cell count falls below 5. Therefore when tables contained cells with small values the Fisher's exact test was used to calculate association between variables. These calculations were done using either Excel or SPSS software.

The responses to the Likert type questions were analysed as ordinal data using descriptive statistics to show central tendencies and Chi-squared as a measure of association (Boone & Boone, 2012). In addition, the Likert type responses to question 10 were coded numerically. Then the mean scores for each different part (paper leaflet, follow-up appointment, etc) of the question were calculated. This made it possible to compare the mean scores and then rank preferences for different forms of communication.

5.8.6 Phase 1 Research rigour

Validation was carried out of the online questionnaire versus the paper copy questionnaire with the help of six Patient Advisors, all of whom had a diagnosis of Lynch syndrome. Each patient advisor was sent a paper copy of the questionnaire and the link to the online questionnaire by email. They were equally divided so that half received the online version first and half received the paper copy questionnaire first. Their responses were received and collated to allow comparison in December 2015.

5.8.7 Phase 1 Ethical issues

Phase 1 Freedom from coercion

The survey was designed to avoid being intrusive as it was optional and anonymous. The survey could be completed at a convenient time to participants and they could stop after partial completion of the survey and return to it later. Participants had the option of completing the survey online or via a paper copy (supplied with reply paid envelopes). Survey questionnaires were given out in clinics with a PIS and an invitation letter or alternatively these were sent out by post. These strategies were designed to avoid any coercion to participate and the optional nature of the survey was stressed in supporting information.

Phase 1 Consent

The act of completing the anonymous survey questionnaire and returning it was taken as assent to taking part, therefore explicit consent was not required prior to completion of the survey.

Phase 1 Confidentiality

The confidentiality of participants was maintained throughout the survey. No clinical details were required for participation in the survey and participation was anonymous. Participants who wished to volunteer for further involvement with the research had a separate form or tear off slip in order to maintain the anonymity of their responses and participants could choose to give a pseudonym. Completed questionnaires were kept in a locked filing cabinet in a secure room in the University and online responses were password protected and only accessed by me in order to ensure the confidentiality of participants.

Phase 1 Emotional reactions

It was anticipated that some participants might experience emotional reactions to the survey. These might be feelings of guilt or an increased concern regarding their susceptibility to cancer. The survey questions could have evoked latent cancer fears as participants were reminded of their own increased risk. However, evidence from genetic counselling, and experience from my own clinical practice, suggested that such psychological distress would usually be short lived following genetic counselling or genetic testing (Burton-Chase, Gritz & Peterson, 2013) and to my knowledge there was no evidence to indicate that questionnaires would cause more distress.

Participants might have experienced feelings of guilt in relation to their fears for the health of their children or grandchildren (Chivers Seymour *et al.*, 2010). In addition, these feelings could also have arisen because taking part in the study reminded participants that they had not communicated with all their relatives about the shared risk of cancer. Evidence (Lucassen & Parker, 2010) indicates that people with a genetic vulnerability to cancer do realise that they have a duty to warn their relatives, but they

can experience a conflict between the desire to protect their family from anxiety and distress and the knowledge that their relatives could reduce their risk of cancer through regular screening, taking medication and symptom awareness.

5.9 Phase 2 telephone interviews

5.9.1 Phase 2 Design

This part of the study was conducted using qualitative methods, more specifically a thematic analysis (Boyatzis, 1998; Braun & Clarke, 2006; Vaismoradi *et al.*, 2016) to enhance and provide more in depth information about participants' experiences and their needs in relation to sharing health information in the family.

5.9.2 Phase 2 interview participants

Participants for the telephone interviews were drawn from people who had already responded to the survey, who had indicated that they were willing to take part in further research and who had provided their contact details. The eligibility criteria for this phase was the same as for the survey detailed above in Section 4.8.2 (Phase 1 participants). A purposive sample of respondents to the survey, with maximum variance for age and educational qualification and with equal numbers of men and women (Bryman, 2006) was selected by anonymously sorting the list of interview volunteers using an Excel spreadsheet, sorting by gender, age and qualification.

5.9.3 Phase 2 Recruitment strategy

This phase of the study used a nested sampling design (Onwuegbuzie & Collins, 2007) where participants for the telephone interviews were recruited via an invitation to take part in further research at the end of the survey questionnaire with an optional tick box and tear off slip. These slips were laid out so that they could be separated

from the questionnaire and returned using the Freepost address. This meant that participants in Phase 2 could have known about their diagnosis, or the diagnosis in the family, for some time and were therefore drawn from the same population of patients as for Phase 1.

Participants in Phase 1 who volunteered for Phase 2 were sent an email acknowledgment indicating that their offer to take part in further research had been received. If they had only provided a telephone number then a telephone message was left giving the same message. If they were selected for interview they were approached either by telephone or email (dependent on their preferred method of contact) and given more information about the study and the interview process. If the participant decided that they wished to take part they were asked to give a telephone number and then a mutually convenient time for the interview was agreed.

5.9.4 Phase 2 Data collection methods

Semi structured telephone interviews were used to collect data from a purposive sample of respondents to the survey, with maximum variance for age and educational qualification and with equal numbers of men and women (Bryman, 2006). The digital recordings of the interviews were then transcribed to allow coding and subsequent analysis by content and theme (Joffe & Yardley, 2004). Data were collected from fourteen interviews in order to better understand the difficulties encountered and preferences for information of people of different ages and both sexes. Fourteen interviews (six men and eight women) were sufficient to reach saturation of themes (Ponterotto & Grieger, 2007; Vaismoradi *et al.*, 2016).

5.9.5 Phase 2 Data analysis methods

Phase 2 data analysis was based on a qualitative thematic analysis approach using both deductive and inductive coding (Boyatzis, 1998; Joffe & Yardley, 2004; Strauss & Corbin, 1998). This was done in order to define concepts of interest or concern and develop information most suited to the needs of the potential recipients. Primary analysis was deductive using descriptive coding and content analysis, focussing on how the participant had been informed of their risk, how health issues were communicated in the family and which topics they would like more information about. Secondary analysis was inductive and sought to develop theory regarding what facilitated or impeded communication in the family over health issues.

A coding frame was developed specifically looking at signs of adaptation (Biesecker & Erby, 2008) and was applied to the data then there were a range of signs detectable in what participants said. The coding frame (Chapter Seven, Table 7.3) gives some examples of adaptation. Features that were considered key to demonstrating personal adaptation were included, such as: whether the participant spoke positively about the diagnosis; referring to the benefits of knowing; but also whether they communicated with their relatives or others about it; sought additional information or took action to adjust to the new 'threat'.

5.9.6 Phase 2 Research rigour

Following each interview I wrote reflective notes which captured my thoughts about the experience of the interviews. Although my positioning as a female genetic counsellor (coming with the perspective of a health professional) remained the same,

the way the interaction had gone with the interviewee and any technical problems they had encountered were noted. These notes contributed to the reflexivity of the analysis (Berger, 2015).

5.9.7 Phase 2 Ethical issues

Phase 2 freedom from coercion

In order to avoid coercion, eligible patients were either sent or given information about the study as a PIS. They were provided with consent forms that they could sign at a later date having had the opportunity to consider what participation might involve and discuss this with me. Prior to starting each of the interviews participants were reminded that they are free to be involved and could stop the interview at any time without compromising their care.

Phase 2 consent

At the beginning of each telephone interview, before recording began each participant was asked about the different aspects of the consent form. This was done to ensure that they were completely happy with taking part and aware that the interview was going to be recorded. They were invited to ask any questions they might have about the study and if they were happy to proceed, also being reminded that they could withdraw and stop the interview at any time. Their permission to have the interview recorded was checked.

Phase 2 confidentiality

Maintaining the confidentiality of participants was very important so I was the only person conducting the interviews in Phase 2 and had access to the participants name (or pseudonym) plus their email address or telephone number in order to set up the

interview at a mutually convenient time. The names of participants were changed in the transcripts of the interviews to protect their identity.

Participants who completed the online or paper survey did have the opportunity to contact me directly to express their interest in taking part in an interview before they made a decision about whether to do so. Although some survey respondents were then identified to me by this their responses were not linked to their names so they remained anonymous. When participants indicated that they were willing to be interviewed they provided their preferred method of contact (email or telephone) using the tear off slip at the end of the questionnaire. These personal details could then be posted or sent separately from their survey responses in an additional reply paid envelope. Those participants that said they were happy to be contacted about being interviewed were able to choose to use an alias, pseudonym or username to conceal their identity. Participants' data remained confidential at all times. Consent forms and all data generated by the study were kept in a locked filing cabinet within a secure office.

Phase 2 emotional reactions

I had anticipated that the interview process could provoke unpleasant emotional reactions in some participants. These would be the same emotional reactions described in Section 5.8.7. Therefore I was prepared for them experiencing a heightened anxiety in relation to cancer, and also possible feelings of guilt or remorse if they had been reminded about how the inherited risk of cancer could affect the health of their descendants or other relatives.

Although it would not necessarily be obvious at the time of interview, if any of the participants had become very distressed during their telephone interviews they would

have been asked if they would like the interview to be suspended. This was a planned strategy in order to minimise their distress. As a Registered Genetic Counsellor my clinical experience talking to people about these issues in a sensitive way gave me confidence to respond appropriately if a participant did become upset.

In addition, had any of the participants experienced distress or voiced concern they would have been encouraged to seek the advice of their GP (if they had any physical symptoms that gave them concern). Alternatively, I would have advised them to contact their genetic counsellor or colorectal specialist nurse. If someone had experienced profound and intrusive feelings of guilt, or other negative emotions, they could have sought appropriate referral through their GP for supportive care.

5.10 Website development

5.10.1 Identifying web designers

Six web-development companies (ICO₃, Modern websites, Robert Stillwell, Live IT Solutions, Papertank and Kevin Hamer at University Hospital Southampton) were identified either by personal recommendation or links via NHS websites.

Invitations to quote for the contract to create a website were sent by email to the above companies in October 2015. Attached to the email (Appendix 40) was a PowerPoint file 'Family Web planning' (Appendix 41) which explained the requirements of the planned website and included hyperlinks to other sites which might be relevant such as Kintalk.org and Patients Know Best. From these web development companies Modern Websites <https://www.modernwebsites.co.uk/p-website-design-services> was selected because they provided the most competitive estimate for the work, they also engaged with the concept and the developer had

previous experience working on academic projects. Partial funding for my research, which included funding for the website development, had been secured through a research grant awarded by the charity 'Bowel Cancer West'. The funding provided by the charity limited the cost of website development to £4,500.

5.10.2 Initial development stages

Initially it was necessary to create a domain name that was registered and therefore could not be used by any other organisation or individual. We agreed that this would be www.familyweb.org.uk rather than www.familywebstudy.org although both were available. Registering the domain name incurred a small annual cost and the process was managed by the web developer.

In order to guide the developer regarding the requirements of the website it was first necessary to map out the different ways information could be provided to the patient users and how they might distribute it. This was a visual way of representing the underlying function of the website. Initially we worked from the PowerPoint slides I had provided initially that specified the brief and then I created an image representing the function.

Discussions with the web developer allowed me to emphasise the key issue of data security and confidentiality. Once the quote for work had been accepted and an initial fee paid Modern Websites provided a link to their account at 'Shutterstock' © which is a company providing licenced images. We had email correspondence and some telephone calls to discuss how the website would be structured (examples of an email in Appendix 42). At the recommendation of the webdeveloper we created an initial stage, 'Stage I' as a mock up of the website using dummy data. The Patient Advisors were then given the opportunity to comment on the appearance of the Stage I

website. Subsequently a 'Stage II' live website was set up to reflect the comments of the Patient Advisors. At all stages of the development the content was written by me with the exception of brief functional commands, such as "Click here for more information", which were written by the webdeveloper.

A feature of the website that was necessary to enable it to be viewed on different devices was that it was 'responsive', this meant that the style elements of the website would adapt to a given screen size. For example, the images would be proportionately smaller if viewed on a smartphone, allowing more room on the screen for the written content to be viewed. This was achieved by the developer through an instruction in the software (a script) which checked what type of device the website was being viewed on and changed some of the functionality accordingly.

5.10.3 Testing the prototype website

The developer and I had agreed that to achieve sufficient security it would be necessary to have a free membership system where users created their own login.

This represented the 'front end' and would be preceded by a short explanation about the purpose of the website. In addition to the document sharing function I wanted to provide freely available generic information. The choice of information that appeared on the website in the open access pages was determined by the results of the Phase 1 survey and the Phase 2 interviews. I organised this content using a life course approach entitled 'Your Journey' (see PowerPoint slides (Appendix 43)). I shared and discussed this approach with the developer by telephone and email. The final version of the text on each page of the website was written by me and emailed as Word files to the developer to insert into the various webpages. I also suggested which images should accompany each of the different passages of text.

The priority was to create this front end before going on to Stage II which would be the core functionality of the system with file sharing between health professionals, their patient 'users' and the patients' relatives. We agreed that it was important to keep the amount of personal data required by the system to the absolute minimum. He explained that part of the security would be achieved through the site creating unique passwords to protect digital files in folders. At each point of the process people viewing files would only be able to do so if they had received an encrypted password created by the system which could not be shared with third parties. We acknowledged that once a file had been downloaded from the site by the user (or their relative) then it was up to that person how they protected the confidentiality of their documents.

The purpose of the site was intended to be equivalent to sharing an information leaflet or clinical letter in hard copy but being able to do so in a digital format. The resulting structure of the website was in effect a very secure database with multiple levels of encryption (Appendix 35) which meant the security of documents was well in excess of that prescribed by the NHS (Appendix 34).

The function of the Stage II website was subsequently tested with volunteers of various ages who were happy to test the role of the Patient User with dummy documents. The web developer had already tested the function with his own selection of alternative pseudonyms but he needed to check that he had not missed anything. I created a login for myself as a 'health professional' member and also as a 'patient' user. This enabled me to see the email invitations that the website generated when a health professional was inviting their patient to access the documents they had uploaded. I was also able to see the invitation emails that a patient would generate when sending an encrypted link to their relatives by using my own personal email as if

I were a relative. We amended the wording of the invitation email slightly and some of the volunteers sent me screenshots of what they saw on their computers at each stage to check that it was as planned.

One initial change that we made at this stage was the choice of banner images. The developer had set up the Stage II website with a series of changing images on the banner at the top of the page. I thought this was distracting from the written content so I asked that he make the banner a still image as I thought it would be better at orientating the reader to the different 'pages' of the website. I also chose which images appeared on each page using a selection of images suggested by the developer.

5.10.4 Stage III website testing and iterative development

The 'Family Web' website www.familyweb.org.uk/ (Appendix 26 screenshot of the homepage) was intended to function as an alternative means of sharing health information with at risk relatives. Since the website would enable health professionals to upload documents we made it flexible to allow a range of file formats (e.g. word files, pdfs, jpeg, etc). I intended that this might facilitate sharing health information by patients with their relatives. In order to test the function I created documents in different digital formats (mostly pdfs and word documents) that I could use as dummy documents to upload and share in my tests or to demonstrate the website function with volunteers.

5.10.5 Timescale of development

Although agreement was reached commissioning Modern Websites to build the Family Web website in October 2015, work on it did not start in earnest until June 2016. Over 150 emails were exchanged with the web developer during the course of

creating the website between June 2016 to the end of November 2017 discussing all aspects of the process. My academic supervisors proof read the content and made suggestions for improvements both in the function and content of the website during this period. A more detailed description of the process of website development in response to the Think-Aloud interview data is given in Chapter Eight and a timeline illustrating the iterative process of development is given in Figure 8.7.

5.11 Phase 3 Think-Aloud interviews

5.11.1 Phase 3 Study design

Phase 3 was guided by the results of the survey and interviews in Phases 1 and 2. The Family Web website was developed and tested with eligible participants through an iterative series of twelve Think-Aloud interviews (McDermott *et al.*, 2010). These were semi-structured interviews conducted through an online video conferencing platform called GoToMeeting (LogMeIn, 2017) where the participant navigated through the website and voiced their thoughts about the website during the recorded interview.

5.11.2 Phase 3 Participants

The participants in Phase 3 were patients who had been given a diagnosis of an increased risk of bowel cancer and advised to have regular bowel screening by colonoscopy. The criteria for eligibility was as quoted above (Section 5.7) but in contrast to Phases 1 and 2 the people who were eligible for Phase 3 were more recently diagnosed. I was looking for people who knew about the risk of cancer in their family but I hoped they might still be trying to share information with their relatives. Interviews were conducted at a mutually convenient time but I first clarified if the

participant was recently diagnosed (at the time of invitation to participate they were between three months and two years post their diagnosis).

5.11.3 Phase 3 Recruitment strategy

Eligible patients were approached through their clinical genetics service, either during their clinic appointment (if they were being seen again post-diagnosis) but more often with an invitation letter and PIS (Appendix 24) sent out later. In addition to this some participants were selected from the volunteers who had completed the Phase 1 survey and who had been diagnosed within the last two years. These participants were contacted by telephone or email to ask how long ago they were first diagnosed. If they were recently diagnosed they were sent an invitation letter by email with a PIS and consent form. I contacted anyone who responded positively to explain in more detail what the Think-Aloud interview would entail. Signed consent forms were then returned to the researcher by post or scanned copies sent by email.

5.11.4 Phase 3 Data collection methods

The website was further developed and refined using a series of Think-Aloud interviews (McDermott *et al.*, 2010). In these interviews the participant talked to me via an online link, while the participant explored the website and voiced their thoughts. 'Think-Aloud' interviews were used because they are a method for recording the dynamic interaction of a participant with a computer programme (McDermott *et al.*, 2010; Sadasivam *et al.*, 2011). I used this type of interview in order to find out how participants explored the website while talking about their thoughts and actions (Appendix 25). The activity of the computer and their associated verbalisation was recorded for analysis. The recordings were done remotely following the method of moderated remote usability testing (Barnum, 2010; Wozney *et al.*,

2016) where the participant and interviewer/moderator can be in different locations but the interviewer can talk to the participant, viewing what the participant is seeing on their computer and recording their dialogue. These moderated remote usability interviews were recorded using GoToMeeting online video conferencing software (LogMeIn, 2017) which created a video, linking the participants' comments to what they were viewing in the website at the time. Initially four interviews were conducted with participants who had no prior contact with the website. Subsequent interviews were carried out after the participants had been given time to explore the website in their own time. In total 12 participants were recruited to this phase of the study.

5.11.5 Phase 3 Data analysis methods

The video recordings were played over several times, so that I was able to view them repeatedly while taking notes about the participant's reactions and comments to different aspects of the website. Phrases were transcribed verbatim to provide illustrative quotes. For each participant a matrix was constructed so that their comments were linked to the context of the relevant part of the website. An anonymised copy of one matrix is presented as an illustration in (Appendix 27). At each iterative stage, themes within the interviews were developed and analysed to guide changes to the website.

5.11.6 Phase 3 Research rigour

Participants to this phase of the research were invited to take part through the recruitment sites in clinical genetics services or via the option to volunteer for interview at the end of the Phase 1 survey questionnaire. In order to maximise the variability in perspective of participants a purposive sample of varied age and sex was selected from the interview volunteers. The only other details recorded about the

participants was the time since their diagnosis in order to confirm eligibility to this phase. This design was intended to minimise bias in the selection of participants to this phase of the study.

Since the Think-Aloud interview took place via an online platform (GoToMeeting) this necessitated access to the Internet. The link to GoToMeeting was sent by email so only patients who had access to email were able to participate. These issues were explained to volunteers who had only provided a contact telephone number. However, the fact that the interviews were recorded on video provided the opportunity for independent researchers to check the validity of the findings against the raw data if necessary.

In common with Section 5.9.6, reflective notes were written after each Think-Aloud interview, noting issues that had arisen during the course of the interview and any technical problems that may have influenced the continuity of the interview and recording.

5.11.7 Phase 3 Ethical issues

Phase 3 freedom from coercion

I provided eligible patients with written information and consent forms. The PIS and consent forms were sent by email to potential participants if they had not already been sent these by the NHS recruitment site. The method of recording the Think-Aloud interview was explained to interested patients over the telephone and via email. No pressure was made to try and persuade patients to take part.

Phase 3 consent

Consent forms were distributed with the invitation letter and PIS sent out by recruitment sites. These were either returned in paper copy or scanned and returned by email before the interviews. Any participant who had volunteered for interview following completion of the survey questionnaire was contacted by the researcher. Before starting the Think-Aloud interviews participants were again asked if they were happy to take part and reminded that their participation would be anonymous and would not affect their treatment. Their permission to record the interview was obtained and their right to stop the interview at any time was repeated.

Phase 3 confidentiality

In Phase 3 of the study, participants' details were only passed on by clinical staff with the verbal or written consent of the participant. No details about participants' health or treatment were taken prior to contact. Research or clinical staff provided the researcher with a contact telephone number or email if patients expressed interest in the study in order for the researcher to explain the study in more depth. All information pertaining to participants (name, contact number, email) were kept securely in accordance with data security guidelines from the HRA.

Phase 3 emotional reactions

Just as for the Phase 2 telephone interviews described above (Section 5.9.7) it was anticipated that the content and purpose of the website might potentially distress participants if it was reminding them about their increased risk of bowel cancer. However, the structure of the Think-Aloud interviews was focussed on eliciting participants reactions to the website itself and for them to suggest ways to improve it. This was therefore not considered as sensitive to participants as the more in depth

telephone interviews, but the researcher was still alert to the vulnerability of participants during the Think-Aloud interviews. If participants wanted to explain their circumstances and how they had learnt of their own risk time was given in the interview for this.

5.12 Conclusions

In this chapter I have described the pragmatic mixed methods approach I have taken to the issue of how to investigate communication of a genetic diagnosis within families affected by a susceptibility to bowel cancer. This is a complex issue requiring a range of methods to elucidate it. These data were used to inform the development of an innovation intended to facilitate communication. The novel function of the website was investigated for its feasibility and acceptability with potential users. In the following chapters, I will give the results of the first two phases of the study, describing how those data were used to create the Family Web website.

Chapter Six

Results of the cross-sectional survey in Phase 1

6.1 Introduction

In this chapter I will describe the results of the first phase of data collection, where responses to the survey from across England and Wales provided both breadth and depth of data regarding patients' experiences. These data were analysed using descriptive statistics and calculations of association using Chi square and Fisher's exact tests. Participants did not disclose their identity, so where quotes are given in the text the quotation is followed by a letter and a number (either 'O' for online or 'P' for paper questionnaire, followed by a number) providing a unique identifier.

6.2 Data collection and analysis

The first phase of data collection was a mixed methods survey collecting anonymous data via a questionnaire. The objectives and methods are described in detail in Chapters Four and Five respectively so will not be repeated here. The first part of the questionnaire, questions 1 to 8 (Appendix 18) were intended to elicit information about the participant's experience of what happened to them, how they themselves learnt about the diagnosis of increased risk of bowel cancer in their family. The following questions related to areas of information provision the participant thought should be changed or improved, such as what other information they would like and how they might like to receive that information. The analyses were focussed on aspects that may have influenced participant's responses: gender, age, cancer diagnosis, education, who informed them about their risk and whether a paper or

online version of the questionnaire was used. The data derived from responses to question 10, using the Likert type scale of preferences, were coded numerically and mean scores for different parts of the question were calculated to allow comparison of means and the ranking of preferences.

6.3 Demographic characteristics of the survey participants

6.3.1 Response rate

I recruited to the survey through online links posted on charity websites and also through six NHS clinical services. Recruitment sites did not provide data on the number of eligible patients they had approached due to the different strategies used by clinicians (Chapter Nine, Section 9.10.1) so it was not possible to calculate the response rate for the survey. 281 participants gave their postcode, but only 150 could be linked geographically to one of the recruitment sites (Figure 6.1). The results for Plymouth and Exeter were merged under 'Peninsula' due to shared postcodes.

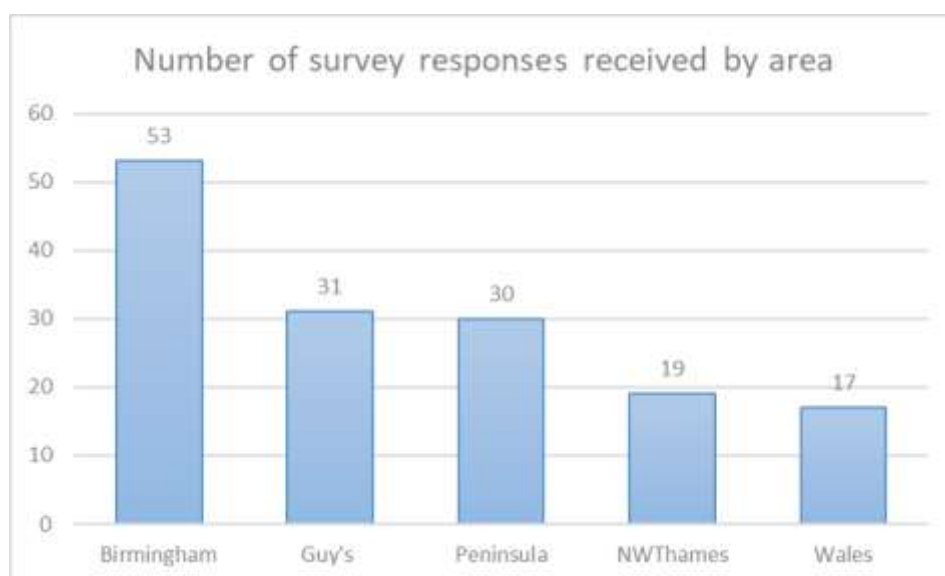


Figure 6.1 Comparison of number of survey responses received by area (n=150)

The demographic characteristics of the 286 participants who took part in the survey are described below, using the variables of gender, age, cancer diagnosis, education and response type.

6.3.2 Gender

The majority of participants were women, 217 (77%) with 66 (23%) male participants; only three participants did not state their gender.

6.3.3 Age of participants

Participants ranged in age from a minimum of 17 -19 years to a maximum of 80 -84 years (two participants did not give their age). Since age was given in age groups, a mean was not calculated, but the mode was 50-54 years of age.

From the age distribution (Figure 6.2) it was calculated that 139/284 (49%) of participants were less than 50 years old and 145 (51%) were 50 years or over. The differences in responses between younger and older participants was of interest, however only 22 (8%) people under the age of 30 participated in the survey, 68 (24%) were under the age of 40 years and in the oldest group, only 14 (5%) were 70 years or over. Therefore, in order to compare younger and older participants' responses, comparison was made between those under 40 years and those 40 years or over (Section 6.7.2).

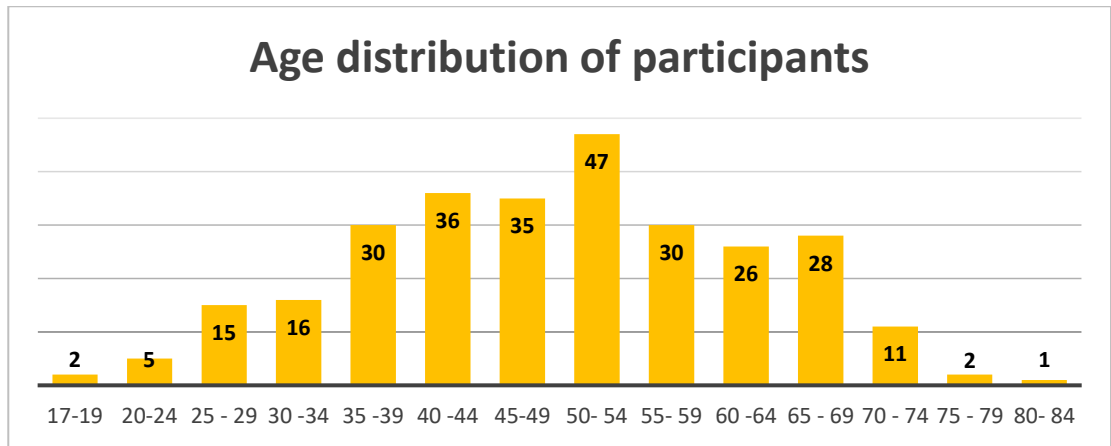


Figure 6.2 Age distribution of participants (n=284)

6.3.4 Cancer diagnosis

Of 284 participants who responded to this question, over half had not been affected by cancer 165/ 284 (58%) but 87 (30%) reported that they had been diagnosed with bowel cancer and 32 (11%) reported being diagnosed with another type of cancer.

6.3.5 Educational qualifications

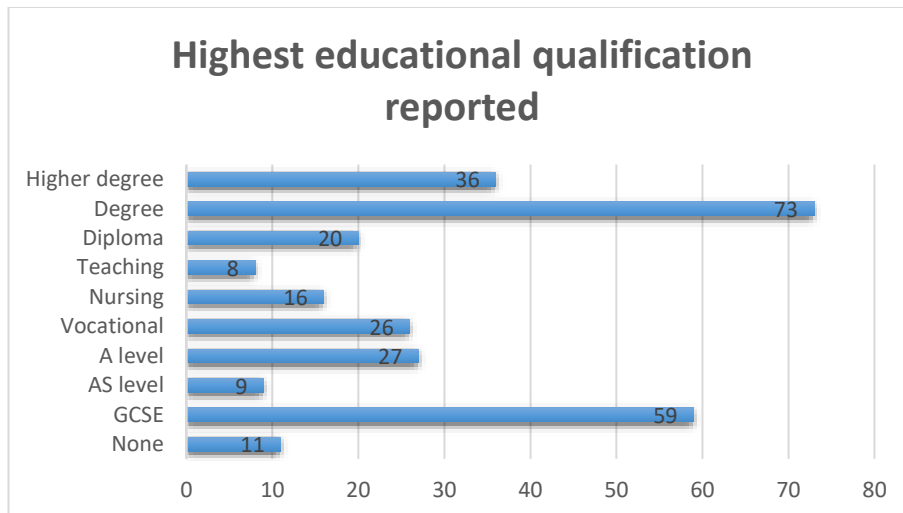


Figure 6.3 Participants' highest reported educational qualifications (n=285)

The participants to the survey were an educationally diverse group (Figure 6.3). The most frequently reported qualification was "first degree", 73/285 (26%) which included graduate membership of a professional institute. The next most frequent category was "GCSE/ O level/ CSE" with 59/285 (21%).

When the data were divided into those participants with or without a degree or higher qualification, 109 (38%) had a first degree or higher degree qualification.

6.3.6 Response type – online or paper questionnaire

Of 286 responses, 183 (64%) were made online via a SurveyMonkey link and 103 (36%) were made via the paper copy questionnaire.

6.4 Participants' experiences

6.4.1 Who first informed participants about their risk of cancer

Firstly, please can you think back to how you first found out about the increased risk of bowel cancer in your family.

1. Who first told you that there was a risk of bowel cancer in your family?

Please tick one:

- Your doctor (General Practitioner "GP")
- Specialist doctor (e.g. surgeon, gastroenterologist, oncologist, etc.)
- Genetics specialist (e.g. medical doctor or genetic counsellor)
- Another healthcare professional
- Your relative, can you tell us who? (e.g. mother, brother, cousin?)
- Other person, please tell us who? (e.g. friend or charity advisor?)
- Can't remember

Figure 6.4 Question 1 from the questionnaire

When asked who had first told the participant about their risk of cancer (Figure 6.4), the largest proportion, 124 (43%) were told by a relative (Table 6.1), most often by their mother. While broadly equal numbers learnt of their risk either through a genetics professional, 79 (28%) or another health professional, 69 (24%) (Table 6.1).

Table 6.1 Who first informed about the risk of cancer (n=286)

Who first informed	Number	%
Doctor (GP)	8	3%
Specialist doctor	56	20%
Genetics specialist	79	28%
Another health professional	5	2%
Relative	124	43%
Other person	12	4%
Can't remember	2	1%
Total	286	100%

6.5 Family awareness of the increased risk

6.5.1 Proportion who were the first to know

Of 286 respondents, 88 (31%) reported they were the first person in their family (to their knowledge) to be told that there was an increased risk of cancer in the family. In clinical genetic terms, this meant that they were the 'index case' and therefore the onus of responsibility to inform other relatives would rest initially with them.

Participant's responses in the free text box (question 13 see Appendix 18) that invited them to provide more detail about any difficulties they had encountered when sharing information had some recurring themes of difficult family dynamics, loss of contact with relatives and family members not wishing to accept the diagnosis, which are summarised in the quote below:

"Lost touch with cousins. Some relatives putting their heads in the sand. Not wanting to have the test." O10

While another participant just said:

"Delivering bad news is difficult" P28

In addition, some participants mention the length of time to get a diagnosis, problems with getting referral to a specialist, needing written information to explain the diagnosis, or relatives being too young to be informed. These themes were ones that recurred, some having already been mentioned in responses to questions 2 and 3.

6.5.2 What proportion of participants' relatives were aware of the risk of cancer

When asked how many of their relatives were aware of the increased risk of cancer in the family, the most common response was "Most" 116/286 (41%).

Comparing the responses from those participants who were the index case in their family to other participants, there were no significant associations in terms of what proportion of relatives were aware of the risk. Of the 88 people who were the 'index case' in their family: 29/88 (33%) said "All" were aware, while 33/88 (38%) said that "Most" were aware (Table 6.2).

Table 6.2 Participants' estimates of the number of their relatives who were aware of the risk

	All	Most	Some	None	TOTAL
Index	29 (33%)	33 (38%)	21 (24%)	3 (3%)	88
Non index	71 (37%)	83 (42%)	36 (18%)	4 (2%)	198
All	100 (35%)	116 (41%)	57 (20%)	7 (2%)	286

However, level of educational attainment was associated with whether participants' relatives were aware of the diagnosis. Comparing participants with a degree or a non-degree level qualification (Table 6.3) participants educated below degree level more frequently believed that their relatives were aware of the diagnosis (39% vs 28%) (Fisher's exact 26.85; $p < .05$).

Table 6.3 Participants' estimates of the number of their relatives who were aware of the risk by participant education level

Participants' estimate of what proportion of the family were aware of the familial risk					Total
Education	All	Most	Some	None	
degree or above	31 (28%)	52 (48%)	22 (20%)	3 (3%)	108
below degree	69 (39%)	64 (36%)	35 (20%)	4 (2%)	172
	100	116	57	7	280

These two groups (education at degree level or above and below degree level) were also compared to see if there were associations between educational level and receiving information about the diagnosis. More participants educated below degree level received no supporting information at the time of their diagnosis (Fisher's exact 10.24; $p < .05$). Conversely, participants educated to degree level or above more often searched for additional information on the Internet (Fisher's exact 11.64; $p < .01$). There were no associations between education level and preference for receiving information either via email, a website or a follow-up appointment.

6.5.3 Genetic testing in the family

Most participants 249 (87%) indicated that genetic testing had been done in their family but nine people did not know if a test was available. I interpreted genetic testing taking place as a proxy indication that a genetic diagnosis had been made in the family. Although potentially inaccurate and dependent on participants' understanding of the outcome of the genetic testing, I considered this the best

indication of a genetic diagnosis in the circumstances and while maintaining participant anonymity. Where a genetic test was available, this would then imply that most respondents should have had access to some specific information about the diagnosis, however I did not know if this was necessarily the case. Therefore questions five and six in the questionnaire were intended to probe the issue of what written information participants had received when they first learnt about the risk in their family (Figure 6.5).

Information you received

5. Please can you tell us what written information you received when you were told about your risk of cancer? Tick all that apply

- None received
- General information about the condition
- Specific information about your family
- A copy of your family tree indicating who had cancer
- A copy of your family tree showing who could have bowel screening
- A 'Dear Relative' or 'To Whom it May Concern' letter to give to your relatives
- Other – please give details

Figure 6.5 Question 5 wording from the questionnaire

The type of information received could then be linked back to the role of the person providing that information, whether they were a health professional or a family member. My expectation was that when people were informed by their relative they might only receive information verbally or have quite general information. However, where families have been provided with a letter specifically designed to be passed to relatives, a 'Dear Relative' or 'To whom it may concern' letter, this was likely to be used. Such letters usually provide practical details about how to access referral to

specialists and screening and give reference numbers that could assist GPs when they are making referrals.

6.5.4 Information received at the time of their diagnosis

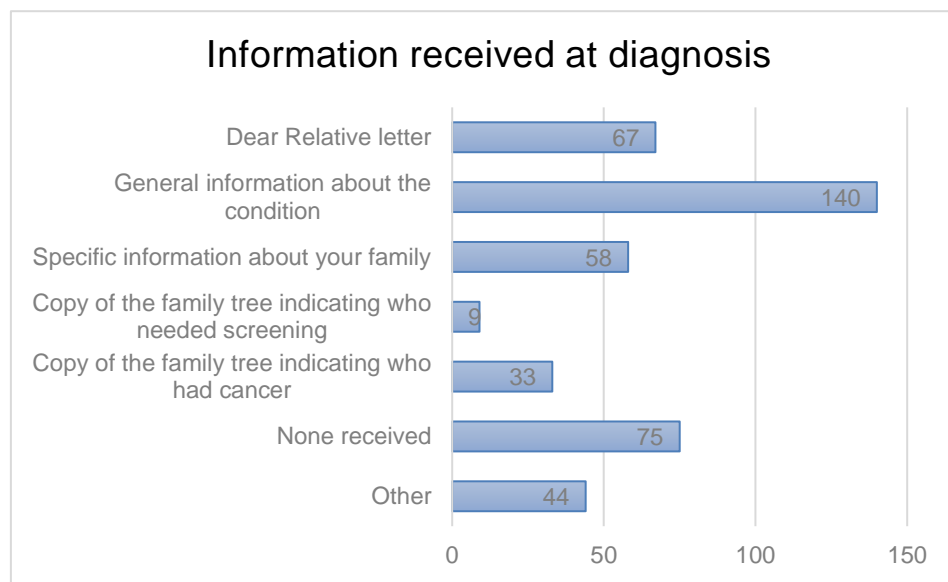


Figure 6.6 Number of participants who had received specified types of information at the time of their diagnosis (n=283)

Those that received information at the time of their diagnosis most often received some general information, 140/283 (49%), or a 'Dear Relative' letter, 67/283 (24%). However, of the 124 participants who first learnt about their risk from their relative, 40/124 (32%) received no written information at all at that time. Types of written information received at the time of diagnosis (Figure 6.6) were all noted, with opportunity for other types to be given in free text.

The types of written information received were analysed against the role of the person who had first informed the participant of their diagnosis. Distinguishing between those participants who had first been informed by a genetics health professional (geneticist or genetic counsellor) or another type of health professional, genetics professionals were more likely to provide general information (65%) and less

likely to give no information at all (8%) than for the other groups of informants (Fisher's exact 23.54; $p < .0001$) (Table 6.4). However, there was weaker association between learning of the diagnosis from genetics professionals and receiving a 'Dear Relative' letter (37%) (Fisher's exact 10.21; $p < .01$).

Table 6.4 Type of information received by person who informed (n=214)

Who informed	What type of information was received (multiple responses allowed per participant)				Total
	Specific	General	Dear Relative	None	
Genetics professional	20 (25%)	51 (65%)	29 (37%)	6 (8%)	79
Another health professional	8 (12%)	36 (52%)	15 (22%)	26 (38%)	69
Relative	26 (21%)	48 (39%)	21 (17%)	40 (32%)	124
Total	54	95	65	72	

No associations were found between being told by another health professional and the type of information received. However if participants' learnt of their diagnosis through their relative they were more likely to receive general information (Pearson's Chi Square = 9.19 with 1 df ; $p < .01$).

6.6 Participants' views of the support they had received

6.6.1 Was the written information received considered sufficient at that time

The question that followed asked participants to reflect on what they had received and whether this was sufficient for them at that time. Being informed by a genetics health professional was found to be associated with receiving sufficient information

(Fisher's exact 18.25; $p < .001$) (Table 6.5). However, being informed by a relative or by another type of health professional were not found to have any significant associations with these variables.

Table 6.5 Whom informed of increased risk of cancer with whether participants thought they had received sufficient information at that time (n=256).

Who informed	Did participants get all the information they wanted?				
	All	Most	Some	None	
Genetics	30 (39%)	21 (27%)	20 (26%)	6 (8%)	77
Another health professional	14 (22%)	9 (14%)	27 (42%)	14 (22%)	64
Relative	19 (17%)	32 (28%)	46 (40%)	18 (16%)	115
Total	63	62	93	38	256

6.6.2 Support received from health professionals

In addition to their information needs, participants were asked to say whether they had received sufficient support at the time of their diagnosis. The responses of those participants who had been informed of the diagnosis by a health professional (general practitioner, specialist doctor, genetic specialist or other healthcare professional) were grouped together. Out of those, the majority 104 /148 (70%) indicated that they had felt well supported at the time they learnt of their increased risk of cancer. However, 24 (16%) said they had not and 17 (11%) indicated that they were "Not sure". Many participants who did not feel well supported at the time they learnt about their risk provided a free text response to the next question, where they were invited to give suggestions about what could have been done better.

Some suggestions on how to improve support to people at the time of their diagnosis were made, but the responses mainly provided insight about what participants felt was bad or lacking in support (Figure 6.7) with some of these issues apparently interacting and contributing to each other.

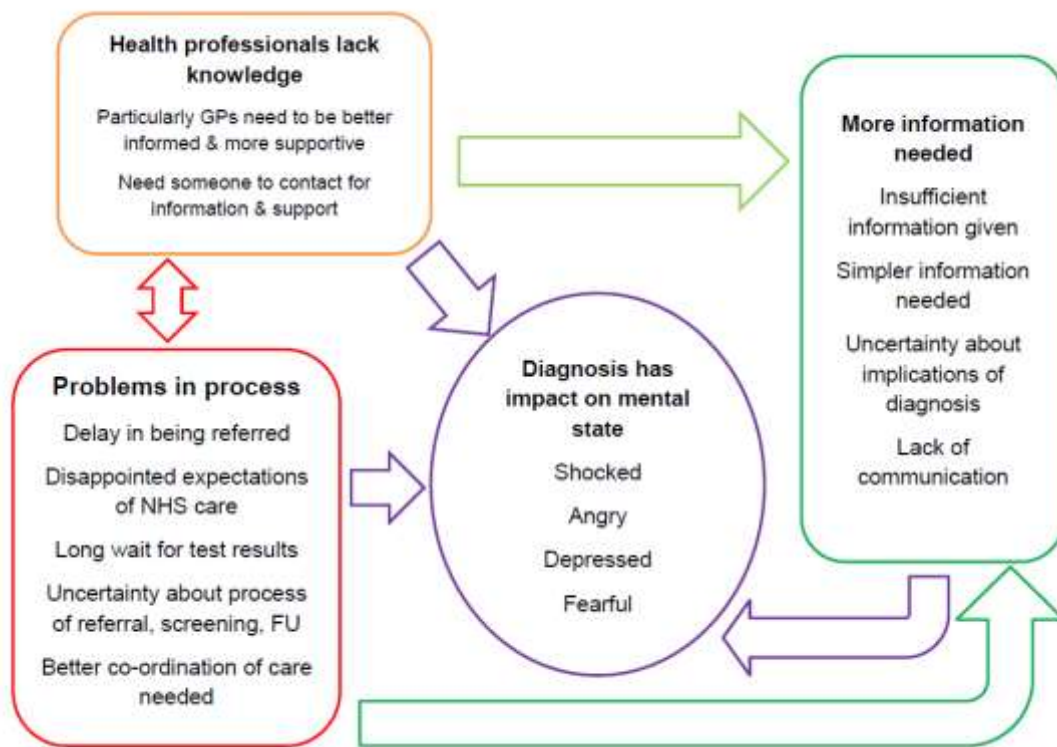


Figure 6.7 Map of themes developed from Phase 1 survey responses (n=286) regarding how support could be improved. *Interrelationships between identified themes of: information needed by patient (green), HPs lack of knowledge (orange), problems in process (red) and the impact on the patient's mental state (purple)*

The data revealed a sense that people's expectations in the care they would receive through the NHS had been disappointed. Many participants reported experiencing delays at each stage of the process; such as when they were being referred to screening or to clinical genetics services; having testing; receiving test results; or receiving screening for cancer. These problems in the process of care appear to have been compounded by a lack of information, leading to uncertainty and confusion

about what they should do themselves to mitigate their risk. The contribution and interaction between the deficiencies in information, lack of support from health professionals, delays in referral and test results were described as impacting on their mental state, as represented in the map of themes above (Figure 6.7).

Some excerpts from the responses are quoted below. In one case the participant described their experience and how they felt isolated after initially experiencing good care:

"I felt well supported during the process of diagnosis which took several months but I was completely unsupported thereafter without any communication at all until I requested it." P49

Another participant stressed the importance of being kept informed:

"It is absolutely essential to explain testing timescales to patients and to keep them fully informed every step of the way. Being given bad news and left to rot is a really bad experience." O72

This participant wanted a better understanding and more realistic expectations:

"A simpler way of explaining it and a realistic time frame to get conclusive results would have benefited me" O163

While this participant also wanted practical guidance:

"It would have been helpful to be given some guidance of the questions/tests I should be now be asking my doctor for." O116

The emotional impact of the genetic diagnosis, or just the awareness that they might have an increased risk of cancer, was described by some as "shocking" or "stressful" and one woman wrote:

"I was confused and mainly angry with my diagnosis (and still am)." P52

Overall, participants perceived that there were many areas where support could be improved.

6.6.3 What other information would have been helpful at the time of diagnosis

Participants were asked to suggest what additional information they thought might have been helpful at the time of their diagnosis. About a quarter, 66/286 (23%) said how they had received all the information they needed at that time but many others described their uncertainty about what to do next and how to manage their increased risk. The quote below describes how that person sought more information to better understand their situation:

"There seemed to be very little information available, and I was only Googling. I found a mix of social media posts and scientific papers. I would have liked something clearly written by health professionals for the lay person." O148

One participant wanted advice about how to approach their relatives, saying:

"Informal advice about how to broach this subject with relatives, some of whom I do not see often and who are not local to my area. The 'To Whom it May concern' letter is rather too formal in my opinion". P49

While another participant noted that they might have made different decisions had they been better informed at the time:

"More information to make better choice instead of being told this is the only way. Knowing what I know now I would have made a different choice". P29

The following comment echoed a recurring problem of insufficient knowledge amongst health professionals about their condition:

"What screening was available and how to get it when even your own doctor hasn't heard of Lynch syndrome". P09

Some of the same themes emerged from a question later in the survey (Figure 6.14) when participants were asked what issues about which they would like more information. These are described in more detail below in Section 6.9.

6.7 Differences and associations between groups

6.7.1 Gender differences in responses

Differences between the male and female participants were significant in how they had responded, online or via the paper questionnaire (Table 6.6) as more women than men had responded online (67% vs 52%) (Fisher's exact 6.58; $p < 0.05$).

Table 6.6 Showing frequency of response type by gender

		Gender		Total
		female	male	
Type	Online	146 (67%)	34 (52%)	183 (64%)
	Paper questionnaire	71 (33%)	32 (49%)	103 (36%)
	Total	217	66	286

Bowel cancer affects both men and women, but in Lynch syndrome women also have a high risk of developing endometrial cancer (Ryan *et al.*, 2017b) so I was expecting that there would be more women in the group defined as "Yes, another cancer" (Table 6.7). This association between gender and cancer diagnosis was significant with many more female respondents having cancer but not bowel cancer (88% vs 9%) (Fisher's exact 14.47; $p < 0.05$).

Table 6.7 Proportions of participants diagnosed with cancer by gender.

Gender	No cancer	Yes, another cancer	Yes, bowel cancer	total
female	130 (79%)	28 (88%)	58 (67%)	216
male	33 (20%)	3 (9%)	29 (33%)	65
Total	163	31	87	281

Another potential difference between male and female participants was regarding the choice of topics where they wanted more information. Since information about a 'healthy lifestyle' was a popular topic across the entire sample of responses, with 140/286 (49%) indicating that they wanted information about this, associations between gender and topic were examined. However no significant association was found between the topic of 'healthy lifestyle' and gender.

Other topics where respondents wanted more information were 'genetic testing' and 'talking to children'. There was no association between gender and 'genetic testing', with 44% of both men and women wanting information on this topic. However 'talking to children' was of greater interest to women than men (39% vs17%)(Fisher's exact 11.84; $p < .005$).

Participants were invited to consider the different ways they could envisage receiving information about the familial diagnosis (Figure 6.8). They were asked to rate different forms of information provision between "very unhelpful" to "very helpful" on a Likert type scale. It was of interest whether there were any gender differences in these responses, particularly if preferences for email or websites were expressed as potential future methods of communication with their geneticist or genetic counsellor.

10. Please indicate how helpful you think this would be for the different ways getting information by making a cross on each of the scales below:

(Likert type scales removed)

a. A **paper leaflet** which has general information about an increased risk of bowel cancer, the implications for relatives and the screening available?

b. A **secure email** which has more specific information about your increased risk, the implications for your relatives and the screening advised?

c. A **password protected website** which has more specific information about your increased risk, the implications for your relatives and the screening advised?

d. A **follow-up appointment** in the hospital clinic where you are given specific information about your increased risk, the implications for your relatives and the screening advised?

e. A **follow-up telephone call** where you are given specific information about your increased risk, the implications for your relatives and the screening advised?

Figure 6.8 Question 10 from the questionnaire

Responses were coded between 1 and 4, with 1 being 'very unhelpful' and 4 being 'very helpful'.

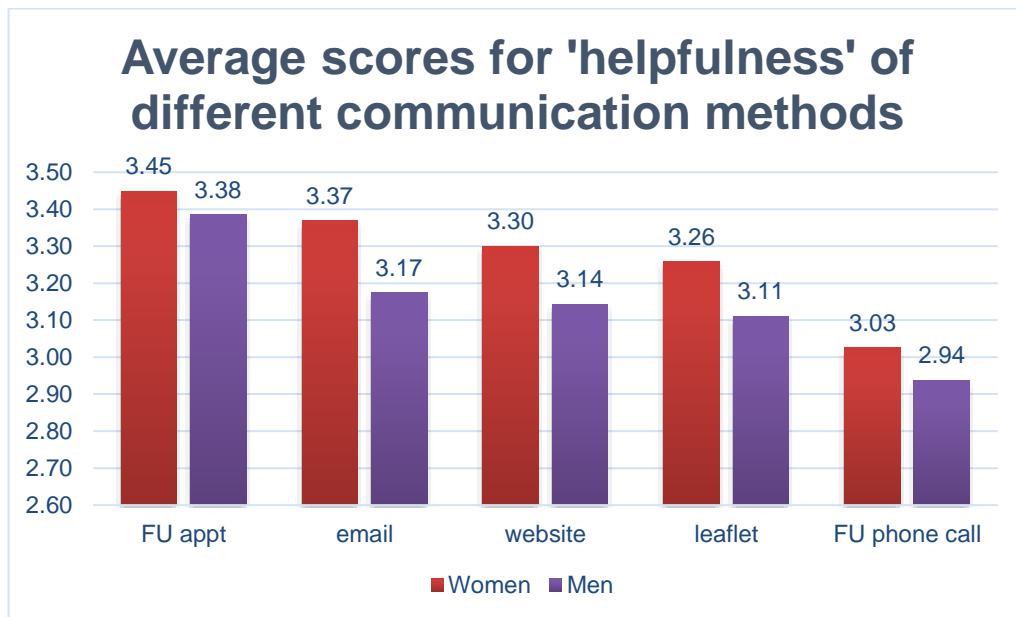


Figure 6.9 Ranking of question 10 responses by gender (n=283)

Figure 6.9 above illustrates how the preferences for different methods of communication from genetics health professionals were ranked similarly by men and

women, with more women indicating that each of the different methods would be helpful than the responses given by male participants.

Although the coding of the responses equated 'helpful' to 3 and 'very helpful' to 4, it is only possible to rank this ordinal data, giving relationship to these different preferences but not to equate the numerical means to actual responses. However, it is possible to infer that the majority of participants of both sexes considered all methods 'helpful' or 'very helpful'. The exception was a follow-up telephone call, which was less often thought to be 'helpful', particularly by male participants.

6.7.2 Age differences in responses

There was a broad range of ages represented among participants to the survey. The modal age group was 50 -54 years (Figure 6.2). When the age distribution was subdivided by response type there was evidence that participants between the ages of 30 and 60 had responded more often online (Figure 6.10) but this difference was not tested statistically.

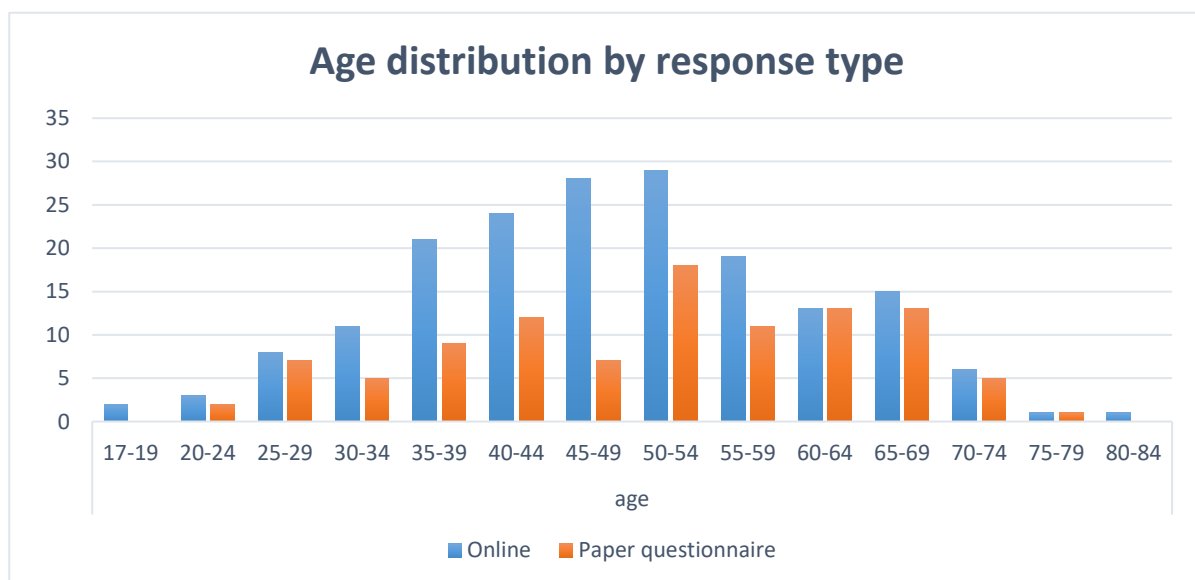


Figure 6.10 Response type by age (n=284)

Participants who were eligible to take part in the survey did not have to have a specific genetic diagnosis to be eligible but the commonly identified conditions which resulted in an increased susceptibility to bowel cancer were familial adenomatous polyposis (FAP) and Lynch syndrome (LS). Since these conditions typically give rise to cancers occurring at young ages (mid 30s and mid 40s respectively) differences in responses were examined between the under 40 year old age group and the 40 years and above group. It was also expected that there would be an association between age and cancer diagnosis amongst respondents.

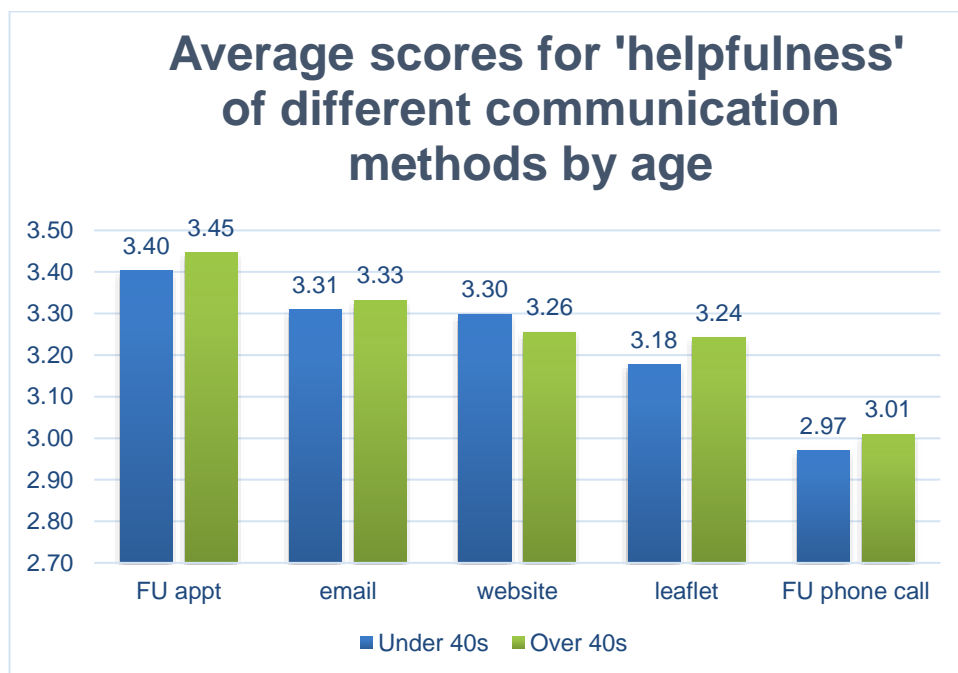


Figure 6.11 Ranking of question 10 responses by age (n=283)

The ranking of preferences did not change between the two age groups (40 years and above and below 40 years) with a follow-up appointment the most popular option for both groups (Figure 6.11).

6.7.3 Differences between groups with and without cancer diagnoses

Table 6.8 Participants' cancer diagnosis by age group (n=284)

		No cancer	Bowel cancer	Another cancer	Total
Age group	40 and over	106	77	31	216
		49%	36%	14%	
	under 40	59	8	1	68
		87%	12%	2%	
		165	85	32	284
	Total	58%	30%	11%	

It was more common for the older participants to have been diagnosed with cancer (50% vs 13%) (Table 6.8) (Pearson's Chi Square 30.75; 4 df; $p < .0005$). This is expected (Torre *et al.*, 2016) but there was also a significant association between having had cancer and being the first person in the family who knew about the risk (the index case) (Chi Square 35.47; 9 df; $p < .0005$). As reported above in Section 6.7.1 (Table 6.7) male gender was also significantly associated with having a cancer diagnosis.

Looking at the data for other associations with cancer diagnosis there was a significant association between wanting to receive information through a follow-up appointment and having had cancer (Pearson's Chi Square 16.69; 6 df; $p < .05$). In contrast, there were no associations found between having cancer and wanting to receive information either by email or through a website. In addition, there was no significant association between wanting information about a healthy lifestyle and having a diagnosis of cancer amongst these participants.

6.7.4 Differences between responses from the online and paper questionnaires

Age distribution by response types (online or paper questionnaire) (Table 6.9)

appeared to show that more of the younger participants had responded online but no significant association was found between responding online and being under 40 years old.

Table 6.9 Frequency of response by age group (n=284)

		type		Total
		Online	Paper	
Age group	over 40	136 (63%)	80 (37%)	216
	under 40	45 (66%)	23 (34%)	68
Total		181	103	284

However, when sources of additional information (Figure 6.12) were considered, participants could tick as many options as they chose to indicate where they had found additional information after learning about their risk of cancer.

<p>7. If you have found additional information about the shared risk of cancer in your family, who provided that information? Tick all that apply</p> <ul style="list-style-type: none"><input type="checkbox"/> Your doctor, surgeon or other health professional<input type="checkbox"/> Other relatives<input type="checkbox"/> Friends<input type="checkbox"/> Support group or charity meeting<input type="checkbox"/> Internet website<input type="checkbox"/> Social media<input type="checkbox"/> Library<input type="checkbox"/> Other source of information – please give details
--

Figure 6.12 Question 7

This allowed comparison between different groups of participants (Table 6.10) and showed that those participants who responded online were much more likely to look for more information on the Internet (Fisher's exact 205.5; $p < .0005$) or via a support group (Fisher's exact 308.6; $p < .0005$).

Table 6.10 Sources of information post diagnosis by response type (n=286)

	support group	Sources of additional information		
		Internet website	other relatives	Total
Online	45 (25%)	102 (56%)	33 (18%)	183
Paper	8 (8%)	33 (32%)	36 (35%)	103
	53	135	69	286

In contrast, those participants who responded via the paper questionnaire were more likely to have found additional information from a relative (35% vs 18%) (Fisher's exact 260.0; $p < .0005$).

6.8 Receiving information about the condition in other formats

Looking at other ways of receiving information, much of the survey was seeking to elicit data regarding participant's preferences for future methods of communication and topics where more information was desired. Following the questions about what information they had received when they learnt about their risk of cancer (questions 5 to 8) participants were asked if they would like to receive information in other ways (Figure 6.13).

9. Would you like to receive information in other ways? Yes / No / Don't know

- If yes, would this be
 - Via a website
 - By Email
 - Social media
 - In a follow-up appointment
 - Other – please state

Figure 6.13 Question 9 from the questionnaire

Table 6.11 Whether information was desirable in other formats

	Receiving information in other ways			
	Yes	No	Don't know	Total
Responses overall	182 (76%)	34 (14%)	24 (10%)	240

The responses to this question (Table 6.11) indicated that the majority of participants would be interested in receiving information in other formats, such as by email or in a follow-up appointment. Since there were more responses to the survey made online, 183/ 286 (64%), different options for receiving information were analysed by response type. Associations were found between responding online and wanting to have more information in a follow-up appointment or via a website. This is described in more detail below in Section 6.10 (Figure 6.18).

6.9 Topics where more information was wanted

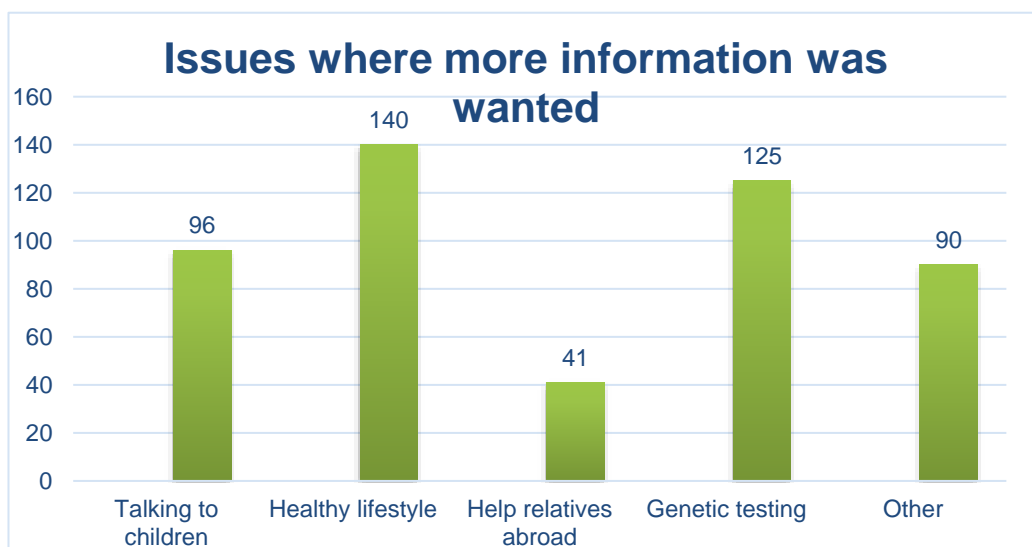


Figure 6.14 Frequency of responses to specific issues that might be of interest to participants (n=286).

Participants were given a list of topics ('talking to children', 'healthy lifestyle', 'helping relatives who live abroad', 'genetic testing' and 'other') where they might have wanted more information and asked to tick all that applied. Most participants 140/286 (49%) indicated that they wanted more advice on how to have a 'healthy lifestyle' (Figure 6.14). In addition to providing answers to the closed question using the predefined topics, 122 participants completed the free text box giving details about other issues that were of concern. The results are presented in a word cloud (Figure 6.15) which illustrates how concerns about cancer screening were frequently cited. Many participants expressed their wish to know how screening was determined, what national guidelines were and whether there was screening for cancers other than bowel cancer. One participant described the need for practical advice on this:

"Regular screening, how often and what it involves and contact names for arranging the screening." Oo8



Figure 6.15 Word cloud illustrating the most frequently cited words in the free text responses describing topics of concern (n=122)

A variety of perspectives were voiced in the free text responses regarding what information would be helpful and indicated many different aspects of how the condition affected people. In addition to questions about screening, a common issue concerned informing other family members about the familial diagnosis, including advice on how to broach what was considered a difficult subject, to not knowing what advice to give about the next steps:

"I have found that relatives want to know about various treatments and always the same question is "What happens next?"" P67

Another person also had a practical request for information to support their conversations with their relatives:

"I would like some written information, maybe a letter or email which I can give to interested relatives. I feel frustrated by the way some of them seem to think that they do not need to listen, as if I am exaggerating the risks, having told them verbally. Maybe a letter would be more official and they would read it, then make an informed choice on proceeding to get tested or not." P75

Many of the participants stated that they had Lynch syndrome and wanted to know more about their risk of cancers other than bowel cancer. Other practical concerns given were: the impact a genetic diagnosis would have on health or life insurance, what symptoms of cancer they needed to be alert to and how they might ameliorate their risk of cancer through changes to their lifestyle, particularly diet. This is illustrated by the quote below from one participant:

"Information about talking to children that will have to be tested by a genetics specialist. What eating habits could help you decrease your chances of your illness turning to cancer e.g. what to eat more of and what to avoid. More in depth exploration about genetics testing." P83

However other issues included starting a family, management of their condition and updated information or research, encapsulated in these three quotes:

"family planning and fertility treatment options" O65

"Who should co-ordinate family screening" P51

"Any breakthrough or what is happening going forward". O12

6.10 Different methods of information provision

To investigate the ways that information could be provided, a Likert type scale was used where several possible options for different methods of receiving information could be classed as "very unhelpful" to "very helpful" (Appendix 18, p.383).

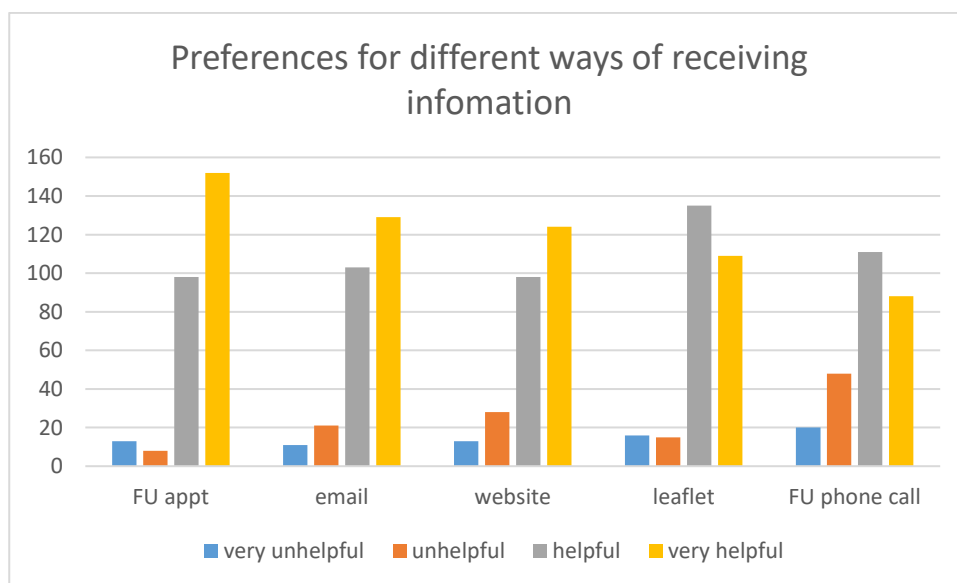


Figure 6.16 Preferences for each method of providing information (n=286)

This question was intended to invite participants to consider how they would prefer to receive "more specific information" about their increased risk. The responses from all participants showed that, for each of the methods given, most participants thought all

the options were either "helpful" or "very helpful" (Figure 6.16). In order to allow comparison of each preference type between different groups of participants, the responses were changed into a numerical value to allow each preference type (paper leaflet, secure email, etc.) to be compared through calculating the mean score for each type. These mean scores were then ranked and the ranking compared between different groups of participants.

Using this method of comparison, it was shown that across all age groups what was considered the most helpful sources of information were:

- A follow-up appointment
- Secure email
- Password protected website.

As stated earlier, there was no change in the order of ranking when the data were analysed by gender (Figure 6.9) or by age group (Figure 6.11).

The option of receiving a 'follow-up phone call' was less popular across several groups; receiving a telephone call to explain specific information about the diagnosis was more often considered "unhelpful" or "very unhelpful" by men, those under 40 years old and participants who had responded online.

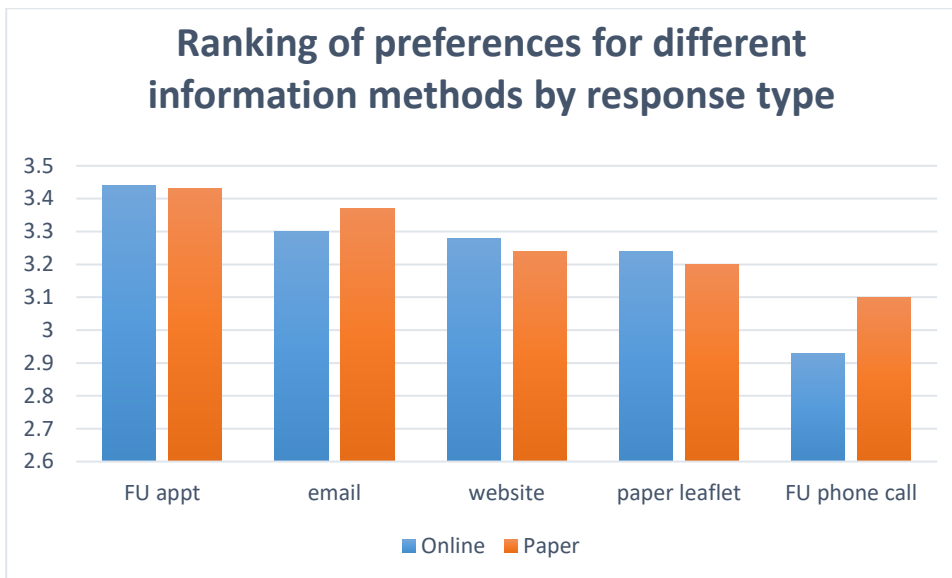


Figure 6.17 Comparison of information preferences according to type of questionnaire response (n= 286, online = 183, paper questionnaire = 103)

It was important to compare responses between the participants who responded online with those who used a paper copy questionnaire, as this could give some indication as to whether the sample was biased in this respect. Using the comparison of mean scores showed how similar the responses of the two groups were to this question (Figure 6.17).

Since the preferences expressed on the Likert type scale by men and women ranked so closely (Figure 6.9), this created a ceiling effect which was not particularly informative. However, the preceding question (Figure 6.13) provided indications for the popularity of different ways of receiving information, such as by email, via a website or through a follow-up appointment.

The proportion of men and women who wanted to receive information in other formats was 44/66 (67%) of men and 135 /217 (62%) of women.

Table 6.12 Preferred methods of information provision by gender (n= 282)

	Preferred method of information provision				
	email	follow-up	website	Social media	Total
Women	93 (43%)	96 (44%)	70 (32%)	31 (14%)	216
Men	29 (44%)	27 (41%)	27 (41%)	12 (18%)	66
All	122 (43%)	123 (44%)	97 (34%)	43 (15%)	282

Although a follow-up appointment was the most frequently cited preference by women (96/216, 44%), and an email was the method most often preferred by men (29/66, 44%) (Table 6.12) there were no significant differences in responses between men and women regarding their preferred methods of information provision.

5.10.2 Preferred methods of receiving health information

When the data from question 9 (Figure 6.13) were analysed for associations according to whether participants answered the survey online or via a paper questionnaire, some differences were observed (Table 6.13).

Table 6.13 Preferred methods of information provision by survey type (n=286)

Survey type	Method of information provision preferred (multiple responses possible)			
	Email	FU appointment	Website	Denominator
Online	79 (43%)	91 (50%)	74 (40%)	n=183
Paper	43 (42%)	34 (33%)	25 (24%)	n=103
Total	122	125	99	

Correlations between response type and preferred method of receiving information were calculated.

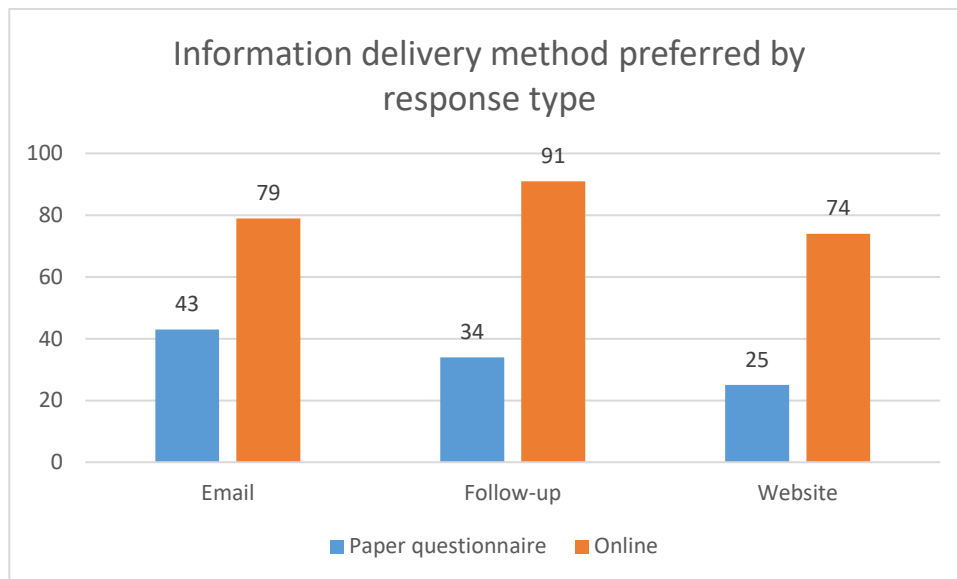


Figure 6.18 Frequency of preferred method of receiving more information by response type, online or via paper questionnaire

It was found that wanting a follow-up appointment was significantly associated with responding online (Fisher's exact 202.2 ; $p < .0005$). A significant but less strong association was found between online responses and wanting to receive information via a website (Chi Square 7.61; 1 df; $p < .01$). No association was found with wanting information provided via email.

6.10.3 Where additional information had been found

In order to further investigate if there were any other associations with response type, Question 7 (Figure 6.12) responses were compared from those people who had responded online to those who had responded on paper.

Searching for information on the Internet and responding online to the survey were correlated (Chi Square 16.43; 1df; $p < .0005$). In addition, when obtaining more

information from a relative was considered, there were significantly more participants who responded via the paper questionnaire who reported receiving more information about their diagnosis from a relative (Chi Square 9.17; 1df; $p < .005$).

6.11 Conclusions

The Phase 1 cross sectional survey was completed by people with a broad range of ages and qualifications. The results of the survey indicated that the majority of participants did feel well supported at the time of their diagnosis or when they first learnt about the risk of bowel cancer in their family. However, there was a desire for more information and support, with certain topics being of particular interest. The free text responses provided a more detailed view of the range of topics where participants wanted specific information, key issues being: screening for cancers and how to reduce their risk of cancer through changes in their lifestyle or diet.

These data therefore gave evidence that this group of patients and their families would also consider receiving or sharing information about their familial susceptibility by email or through a website.

In the next chapter (Chapter Seven) I will set out the results of the second phase of data collection. Qualitative data, gathered through telephone interviews with a purposive sample of volunteers, were interpreted using thematic analysis and the results are described.

Chapter Seven

Results of Phase 2 Qualitative telephone interviews

7.1 Introduction

In this chapter I will present the results of my qualitative analysis of the semi-structured telephone interviews conducted in Phase 2 of the study. These interviews were carried out to inform the development of the proposed website innovation. It was anticipated that the interview data would add to our understanding of participants' experiences of learning about their risk of bowel cancer; how information was shared in the family; what methods of communication they used and what aspects of the condition they would have liked to have more information about. These qualitative data were therefore building on the data collected through the preceding survey in order to enhance and deepen my understanding of what a website might contribute to communication within families. I also wanted to improve the trustworthiness of the findings through triangulation between the survey data and interview data.

The thematic analysis (Boyatzis, 1998; Braun & Clarke, 2006) was carried out using the software package NVivo, version 11 (QSR International, Pty Ltd) which facilitated the identification of multiple codes, amalgamation of codes into themes and enabled data sources to be indexed for retrieval. This process revealed fifteen themes which coalesced into four major themes. Some themes were around participants (the impact of the diagnosis and people's adaptation to it) while others were specific to the

concept of the website. It was key to the development of the website that I captured participants' views about what content they wanted and how they might envisage using the website in order that it aligned with their needs.

All names given are pseudonyms in order to preserve participant anonymity.

7.2 Demographic characteristics of participants

All interviewees had already completed the anonymous survey questionnaire. Many participants had been recruited to take part in the survey via the Lynch syndrome UK support group website, which hosted a link to the online survey with information about the research study (Appendix 15). At the end of the survey there was a request for volunteers and a tick box so that participants could indicate if they were willing to be interviewed. In total 291 people completed the questionnaire, of which 187 (64%) volunteered to be interviewed. Interview volunteers were selected from this group of 187 using purposive sampling to obtain a maximum variance sample (Teddlie & Yu, 2007) based on gender, age and educational attainment.

Demographic characteristics of the interview participants are presented below in tabular form (Table 7.1). The participants were six men and eight women, ranging in age from 20 years to 68 years (this range of ages broadly reflects the characteristics of patients referred to clinical genetics services for assessment of risk of bowel cancer). Educational attainment was varied amongst the participants, although the majority had a degree or higher degree. Twelve out of the fourteen participants had a diagnosis of Lynch syndrome, of which five had been tested and found to have a pathogenic gene variant following their diagnosis with cancer. Six participants had been diagnosed following a pre-symptomatic test (PST) and three had no proven

molecular diagnosis but they had been given a diagnosis of either Lynch syndrome or a high risk of bowel cancer based on their family history of cancer.

Table 7.1 Demographic characteristics of participants in Phase 2 interviews.

Pseudonym	Age	Gender	Cancer	Genetic test	Relatives having PST	Genetic diagnosis
George	68	Male	bowel	Diagnostic	Many relatives	Lynch syndrome
Bill	56	Male	bowel	Not available	NA	Lynch syndrome
Emma	20	Female	none	PST	uncle	Lynch syndrome
Robin	27	Male	none	PST	sibling	Lynch syndrome
Natalie	27	Female	none	PST	parent & siblings	Lynch syndrome
Sandra	55	Female	endometrial	Diagnostic	children	Lynch syndrome
Dave	54	Male	none	PST	Many relatives	Lynch syndrome
Fiona	43	Female	bowel	Diagnostic	sibling	Lynch syndrome
Anne	64	Female	bowel & endometrial	Diagnostic	siblings & children	Lynch syndrome
Bob	36	Male	none	PST	none	Lynch syndrome
Diane	56	Female	endometrial	Diagnostic	sibling & child	Lynch syndrome
John	32	Male	none	PST	parent & siblings	Lynch syndrome
Kay	48	Female	none	Not available	NA	High risk of bowel cancer
Gina	39	Female	none	Not available	NA	High risk of bowel cancer

Key: PST – pre-symptomatic testing; NA- not applicable; child(ren) = adult child(ren) over 18.

7.3 Summary of the four major themes identified

Each of the interviews provided an opportunity for the participants to tell their story. I encouraged them to reflect on what could have been improved in terms of supporting information at the time of their diagnosis. This approach may have encouraged a critical perspective on their experiences. What I found striking was the profound, mostly negative, effect the diagnosis had made to their lives. This led to me identifying the first major theme as the **impact of the diagnosis**. The evidence for this was in the descriptions given by some participants of the shock, a sense of burden and the feelings of isolation they had experienced since their diagnosis. Some spoke of having to become self-reliant, for example: having to keep reminding clinical services about their need for regular surveillance by colonoscopy.

The second major theme was that of **adaptation**; it was evident to me that lack of adaptation to the diagnosis had affected some participants' ability and inclination to share information about the implications of the diagnosis with their relatives.

Conversely, those participants who appeared to have adjusted and adapted to their diagnosis were more accepting and also more engaged with their sense of responsibility to their relatives. The participants wanted more **practical information** to augment their understanding of the familial diagnosis, this was the third major theme. Descriptive coding identified what information they would have liked to receive (and what topics they would want covered on the proposed website). This revealed that advice on how to reduce their risk of cancer through a healthy diet and changes to their lifestyle was most commonly mentioned. Other frequently cited topics were: risks of different cancers and the relevant screening for them, how to

spot the early symptoms of cancer, how to explain the diagnosis to children, where to go for support, and finding advice specific to their particular mismatch repair gene.

The fourth major theme identified was using **appropriate communication** with relatives. Those participants who had shared information about the diagnosis with their family members used ways to contact them that were feasible and that they thought would suit that person. This meant that using a secure website to share information in the family could be utilised as one of several ways of communicating with their relatives.

Table 7.2 Summary of main themes and subthemes identified in Phase 2.

Theme	Subthemes	Examples
<i>Impact of the diagnosis</i>		
	Emotional burden	Role of being informant; fears for self and others
	Initial understanding	Shock or some expectation
	Feelings of isolation	Self-reliance, fight for treatment, ignorance of GPs
	Practical implications of diagnosis	Colonoscopy screening, responsibility to family, aware of symptoms of cancer.
<i>Adaptation</i>		
	Acceptance, coping & self esteem	Adjusting to things unable to change, positive perspective, forward looking.
	Social integration	Closer to loved ones, joined support group
	Regaining control	Taking action, role of family co-ordinator, information seeking, sharing information
	Family adjustment	Family talk about diagnosis, relatives access screening
	Lack of adaptation	Denial, avoid talking about diagnosis
<i>Practical information</i>		
	Information to support understanding	Written in plain language, education for GPs and other health professionals
	Information that gave hope	Less clinical and more positive
	Website content requirements	Healthy lifestyle advice , gene specific risks, talking to children
<i>Appropriate communication</i>		
	Using existing methods of communication	Different methods for different relatives; Facebook, email, telephone.
	Positive attitudes to website idea	Could access information at anytime; store screening reminders; ideal for younger relatives.
	Limitations of information via a website	Not appropriate for all relatives; concern re security

7.4 Impact of the diagnosis

7.4.1 Initial understanding

Most of the participants either learnt about their risk of cancer through their parents, often their mother, or through their own diagnosis of cancer. Several had experienced multiple relatives being affected with bowel cancer or other cancers. This was the case with Sandra, who was not surprised by her diagnosis:

"my maternal grandmother had cancer of the womb and then she had bowel cancer twice. My mum had cancer of the womb. (Sandra, S6 line 11).

This indicated to Sandra that she was likely to develop cancer too:

So I always expected..it" (Sandra, S6 line 12).

The exception to this was Robin, who in his early 20s realised that his father's diagnosis with bowel cancer at 40 years was unusual. He sought the advice of his GP, which eventually resulted in his father being tested and found to have a pathogenic gene variant, giving him a diagnosis of Lynch syndrome. Robin appeared to have had a key role in his family regarding the diagnosis. Nonetheless, he did not know whether his aunt had talked to his cousin about the diagnosis:

"her daughter is obviously aware of that operation and whether she's tested I am not sure, and she has kids and I am not sure if they are [aware] either" (Robin, S4 line 158).

Quite unusually Fiona (S8) learnt about the condition in the family through a letter from her genetics service, which she found profoundly shocking:

"I knew nothing about that at the time. And so it was, you know, it was a real shock" (Fiona, S8 line 62)

However, all other participants were told either in person or over the telephone by their relative or a health professional.

Thinking beyond the mechanism of how people found out, the way that participants described their experience of learning about their familial risk of cancer indicated the profound effect it had on them.

7.4.2 The profound effect of the diagnosis

None of the participants reported regretting having a pre-symptomatic test or being given a diagnosis. However, the data revealed what a major impact the diagnosis had made on the participants' lives. The need to adjust and come to terms with their increased risk of cancer appeared to have had a profound effect on them. Some participants emphasised the role chance played in their diagnosis, and this appeared to reinforce their sense of uncertainty. Some reported that their cancer was only detected by chance or was entirely unexpected. For others, the participant or their family had realised that there was some genetic predisposition in their family, but this had not been recognised before by health professionals.

Some of the participants found the diagnosis frightening or shocking initially. This is most evident for Fiona who did not have contact with her late mother's family and learnt of the familial diagnosis by letter from her genetics service:

"a complete bolt out of the blue really" (Fiona, S8 line 59).

Several participants felt it was better to know of the risk and benefit from regular screening. However in almost all, the diagnosis led to concern and the burden of responsibility of having to inform their relatives. In addition, there was anxiety that their attempts to inform other people in the family might be met with indifference or

negative reactions. Anne explained how difficult it was when her son was tested following her own diagnosis:

"it was a really horrible thing to go through to see the devastation with my son, when he realised he had it. It was very traumatic" (Anne, S9 line 285)

Some participants alluded to the emotional burden they felt by being constantly aware of the condition. Others described the trauma they experienced through witnessing the effects of cancer on people they cared for.

7.4.3 Feelings of isolation

Feelings of isolation appeared common and were expressed by the majority of participants; Fiona said:

"I feel that nobody is looking after me" (Fiona, S8 line 531).

While Anne reflected that after her genetic counsellor retired she felt unsupported:

"I felt like a little bit abandoned" (Anne, S9 line 199)

These feelings were compounded by her GP retiring:

"he was absolutely amazing but he's retired now and I don't feel that there is anybody that's left" (Anne, S9 line 209).

Responses from Bill and Fiona reflected their pain and sense of powerlessness; Bill described how he felt his cancer could be worse if it recurred:

"I have got these thoughts, you know, of the ..you know, the death sentence and that type of thing and the thoughts of if it does come back, you know, it's going to be a lot more difficult to manage, the consequences are going to be worse and that type of stuff" (Bill, S2 line 321).

Another common issue reported by participants was having to be self-reliant, particularly needing to be the expert patient because the health professionals may

know less than the patient. Diane, Robin, Dave and Fiona all described how they have to “push and push” to get the appropriate treatment or screening. Dave reported:

“it seems to be all the time I am battling to (get) my colonoscopy, colonoscopies done yearly rather than three yearly.” (Dave, S7 line 127).

Similarly, Diane believed that her cancer would not have been found but for her persistence:

“I had to push and push and push each stage of the way” (Diane, S11 line 73)

and

“I would possibly be dead by now or had a really serious, you know, end stage cancer before they got to me. I don’t think I would have been taken seriously. ...I have had to really, really push...particularly about my bowel screening.” (Diane, S11 line 158).

Several participants spoke of their frustration and dismay that very few health professionals they met had heard of their condition; some suggested that they felt it was important that GPs, in particular, received education about LS. The difficulties of having to be self-reliant and become an expert patient appeared to add to the burdens experienced by participants.

7.4.4 The burden of the diagnosis

Some participants voiced a sense of burden associated with the diagnosis. There was an emotional burden sometimes described in terms of a fear for themselves:

“..sometimes it does feel as though there is a bloody sword hanging over your head” (Sandra, S6 line 175)

The emotional burden was sometimes expressed as feelings of guilt, and the trauma of watching loved ones suffer:

"it is the watching people with it, or er, the predation of people um ..through cancer, which is probably the most painful thing" (Bob, S10 line 152).

Bob also described how he felt he had become a burden to others when he tried to talk about his condition; this may have contributed to his feelings of isolation:

"I get the sense, that you know, I'm becoming a burden, I am becoming boring. I know I shouldn't be, but it is just something that gets glazed over really and I don't actually know why. Because it is quite a big deal, I would have thought" (Bob S10 line 183)

Other aspects of the diagnosis that created a sense of burden were their efforts to inform relatives, their anticipation of negative reactions in relatives and how participants expressed feeling a responsibility to others. These feelings were most frequently voiced in relation to children, so they appeared fearful about their own health and the health of their family members. In addition, there were also the practical burdens of taking time off work to have screening or prophylactic surgery:

"It was a complete waste of time because they didn't actually pass any of the information on. Yes it was a complete waste of time and effort. And then because when I have to go for these appointments I have to take time off work" (Kay S12 line 214)

'Access to information' was also a recurring minor theme where lack of access to information was a factor that inhibited communication, like the condition not being openly discussed in the family, having insufficient knowledge to be confident explaining the diagnosis to others, or having problems being referred to genetics because of inadequate information to support that with the GP. However, some positive experiences were described by participants such as: access to genetics follow-up or information found through a support group, also greater awareness of research and strategies to reduce risk of cancer.

7.5 Theme 2 - Adaptation to the diagnosis

7.5.1 Adaptation was associated with sharing information

Moving beyond the manifest to the latent meaning of the interviews, secondary analysis of the data indicated that coming to terms with, and adjusting to, the diagnosis was likely to be necessary before patients could fully engage with sharing information about the diagnosis with their relatives. The coding frame (Table 7.3) provides some examples of adaptation taken from the data from seven of the fourteen interviews. The signs that I took to indicate adaptation also appeared to be associated with how engaged participants were communicating with their families about their test result or diagnosis.

7.5.2 Lack of adaptation

Amongst the participants in this study, two people stand out as lacking peace of mind and did not appear well adapted to their diagnosis, these are Bill (S2) and Bob (S10). Bill had spoken to his sons about the diagnosis of Lynch syndrome but could hardly contemplate attempting to speak to one brother and had no intention of trying to contact his other brother. He returned repeatedly to his complaint that his family received very little psychological support 20 years earlier when he was diagnosed with cancer. He also admitted to feeling troubled by his more recent diagnosis of LS:

"I guess I come up with trying to make sense of nonsense, you know, and just try to make what is going on a little bit more manageable" (Bill S2, line 282).

Bill was also fearful for his own future:

"on me bad days, it is like um.. a death sentence" (Bill S2, line 247).

Bob presented a different perspective but with only tentative signs of adaptation. He sounded isolated and was the only person in his family to be tested; he was still struggling to come to terms with his positive pre-symptomatic test eight years earlier:

"I have not got any family. I mean, you know, I am not going to lie to you, since I was diagnosed it has totally affected my interaction with other people, forming relationships" (Bob, S10 line 114).

While showing some indications that adjusting to their situation was still difficult, Bill's and Bob's involvement in this study could be interpreted as demonstrating a degree of adaptation.

7.5.3 Signs of psychological adaptation

In contrast to Bill and Bob, all the other participants appeared to demonstrate more signs of psychological adaptation to their diagnosis and some were actively engaged in trying to inform and support their relatives about aspects of the familial diagnosis. One of the commoner signs of adaptation noted in this group was actively seeking information about the condition. For example, John and Gina both described looking on 'Google' for more information, while Fiona, Emma, Robin and Natalie all sought information and support through the Lynch syndrome UK Facebook group and website.

Another aspect that comes across in some of the data is the positive view of their circumstances that some participants appeared to have. A few of the participants described how their familial gene variant was in some way advantageous compared to other mismatch repair gene variants. They regarded their particular gene variant as being, as Dave says: "probably one of the better ones to have" (Dave S7 line 186). Dave (gene variant in MLH1) shared this perspective with Robin (gene variant in

PMS2) and Sandra (gene variant in MSH6) and this optimistic view was interpreted by me as evidence of coping efficacy.

7.5.4 Which relatives are informed

The closeness of family members seemed to have a bearing on how likely they were to be informed or share information, although this may not hold true for some families with dysfunctional relationships. For most participants the people they talked about as 'family', and for whom they felt a responsibility, were their children or siblings.

The younger participants who didn't have children, such as Emma, Natalie and John, were all recipients of information provided by their parents and appeared not to have felt a responsibility to share any information with relatives. In contrast, it was Robin, who at age 27 years and married, did encourage his brother to seek genetic advice and have testing, despite his brother's apparent indifference:

"he's had the test. Er he was very.. not reluctant, but not bothered is probably too strong a word, but it wasn't something that he was particularly interested in. But he has had the test and luckily he hasn't got it.." (Robin, S4 line 123).

Initially Robin had recognised the pattern of cancers in the family as unusual.

Subsequently he appeared to adopt the role of a family co-ordinator regarding the diagnosis, following the instigation of genetic investigations in his father.

Other participants with adult children (George, Gina, Kay, Anne, Bill, Diane, Dave, Anne and Sandra) focussed on the importance of protecting their children from cancer and supporting their children through genetic testing. This is illustrated by Sandra who had children ranging in age between 36 and 22 years:

"that's where I think ...it's alright supporting them, 'cause I...I.. I...for me, I knew about me, that wasn't my issue, the issue now is that I have got four children" (Sandra, S6 line 64).

7.5.5 The role of family co-ordinator – taking action

Another aspect of adaptation was the type of actions participants described following their diagnoses. In response to questions about how they had shared information about the diagnosis, most indicated that they had talked to their close family but few mentioned relatives in the wider family. One clear exception was George, who had taken it upon himself to be a family co-ordinator and had created a spreadsheet where he logged what testing or screening each relative had received:

"I do check up to make sure that the colonoscopy teams in their local hospital are responding and that there's no delays." (George, S1 line 680).

Diane's behaviour also illustrates that she took this role with regard to sending information to her relatives, asking other family members to send on information to those for whom she lacked contact details:

"..getting people to share stuff and sending information and you know, if I haven't got an up to date address its getting someone else and they have to send it off" (Diane, S11 line 414).

Diane also went with her sister and son to their genetic counselling appointments, which was a supportive strategy also used by Sandra.

Emma mentioned how her uncle looked into the family history first following her mother's diagnosis and Fiona described how she was asked by the genetics service to inform her brother and to make sure he had screening. Natalie had a different perspective, mentioning several times the importance of her partner having access to

information about her diagnosis. She wanted him to have information from which he could draw his own conclusions and not be influenced by her interpretation:

"I felt it was very important for my partner to know about Lynch syndrome, because I didn't know um about Lynch syndrome because I didn't um, I was not sure if I was explaining it well enough to him , so I felt that reading the letters would be a good way for him to um, to have a better insight" (Natalie, S5 line 54) .

7.5.6 Family adjustment interacts with personal adaptation

Assessment of family adjustment was through indicators such as how openly the condition was discussed in the family and whether relatives were reported as having testing or screening. This revealed that some families did not appear to have adjusted to the diagnosis, even when the diagnosis had been known for several years. Most notably, Bill did not appear to have adapted to his genetic diagnosis and there was little evidence of adjustment in his family, with his sons both struggling with the diagnosis and Bill estranged from his brothers.

In contrast, Emma, Natalie and Dave all had supportive families with open communication around the diagnosis and they themselves appeared to have adapted to the diagnosis. Passages of each of their dialogue could be interpreted as showing evidence of social integration, coping, acceptance and improved self-esteem.

Table 7.3 Evidence of participants' adaptation coding

Adaptation domain*	Indicated by these aspects	Examples derived from the study participants
Coping efficacy, Self-esteem or Positive self-concept	Acceptance of diagnosis, positive perspective, or appears forward looking	"Technically your kind of.. safer having it, and getting the regular screening and the regular tests, than... not knowing and not getting that help, then if they can catch it early then that's amazing.." line 278 S3 Emma
Social integration	Describes relationships positively	"..a lot of my concerns I have brought up with my mum prior to going anyway, because we have a.. a very good relationship" line 50, S5 Natalie
	Joined support group	" the genetic side of it and emotionally I go on the Lynch syndrome UK Facebook" line 52 S4 Robin
	Aware of support available from others	I still call mum like er..for three times a week, because I don't know, because I think we are as a family quite used to it now I'm in which it needs talk about it that much says there is anything like worrying me then I would just call her and tell her. Line 162 S3 Emma
	Informing relatives about diagnosis	"I said "Take my letter with you because it will explain it easier than you trying to explain it", so I had to give him my personal clinic letter to take to his GP and his GP, did refer him" line 291 S8 Fiona
	Raising awareness of the condition	"If I am telling someone, I do tell a lot of people about it, because I would rather them know about it, so if they say if someone in my family had it young, and my brother had it young, then it raises awareness" line 414 S4 Robin
Regaining control (actions or planned action)	Information seeking	"I did a kind of Googling it in the early days I think when my mum had first given me the information I sort of started to dig around to find what is it what does it mean" line 204 S14 John "I have got ..got a profound interest in genetics of all sorts of things now. So I am forever reading up the latest things they have found out" line 75 S7 Dave
	Participating in research	"I get a phone call from my counsellor every six months but that's because I'm on the CaPP3 trial" line 183 S3 Emma
	Taking action to adjust to new threat	"I have done my own research and I have spoken to the Lynch experts at the conference I have just been to are very much advocating HRT and it is potentially, it has a protective role particularly in Lynch" line 415 S8 Fiona
Family adjustment	Relatives accessing screening or testing	"Yes, my brother's had testing and he didn't have the gene, um.. my uncle had testing and he didn't have the gene so that wouldn't have passed on to my cousins" line 203 S5 Natalie
	Family talk about the condition or diagnosis	"As soon as my mum found out, everyone, everyone in the family, both sides of the Atlantic were informed straight away by my mum" line 356 S7 Dave
*Adaptation domains taken from Biesecker et al 2013 "Development and validation of the Psychological Adaptation Scale (PAS): use in six studies of adaptation to a health condition or risk"		

7.6 Theme 3 – Practical information

7.6.1 Practical information to support their understanding of the diagnosis

When participants reflected on what supporting information they had received at the time of their diagnosis most reported receiving some written information. However, this was often limited to a clinical letter from the genetics service that was not necessarily regarded as appropriate material to share with other family members. Some spoke of receiving standard leaflets but almost all described how they had looked for information themselves, mostly via the Internet:

"I have looked at a lot of different websites particularly once my sister was diagnosed" (Kay, S12 line 260).

John described how he was given plenty of information after his pre-symptomatic test result but he found the information factual but too clinical; he wanted more practical information guiding him about the repercussions of the diagnosis:

"..too much about the medical aspects I guess rather than what you really want to know is what is the real likelihood that you have this gene, what are the things that you need to do in terms of you know protecting yourself, really what is the impact on your life, do you need to worry about it, is it something you can manage with lifestyle changes" (John, S14 line 214).

Robin commented how he would have found it helpful to have been given simpler written information. This would have enabled him talk about LS and its implications with his cousins, and people outside the family, in order to raise awareness of the condition. Robin also described how it would be a practical benefit to him to be given a record of his colonoscopies to help him keep track of when the next one was due.

Following her initial contact with clinical services, Gina described how she would have liked some additional supporting information:

“Some information sheets, as soon as the doctor said we need this doing it would have been nice to read something about it and why, because all he kept saying was” you are really high risk” so it would have been nice to read something then when we first found out” (Gina, S13 line 200).

Although different participants at times described which particular issues were of concern to them, it became apparent that the issues they wanted more information about changed as their understanding of the diagnosis deepened. They had questions for themselves and they also wanted information resources to support their conversations with their relatives.

7.6.2 Website content requirements

In the interviews participants were encouraged to suggest issues where they felt more information would be helpful. The coding then identified numerous different topics and the majority of participants did describe areas where they thought more information would be beneficial. The desired topics identified were:

- risk reduction through diet and lifestyle
- gene specific information
- cancer risks and relevant screening
- advice on talking to children
- symptoms of cancer
- inheritance and starting a family
- research
- where to go for support
- insurance implications.

These topics were mentioned numerous times across the interviews, which gave an indication of the broad spectrum of issues where the participants felt they needed more information. The most frequently cited topic of interest was risk reduction

through diet and lifestyle, which was specified by many of the participants. Gene specific information, cancer risks, screening and starting a family were all cited by several different people. Robin listed several which encapsulated the key issues:

"talking to other people about Lynch syndrome um where to go for support, um like you said, healthy eating, keeping up to date with your scans, symptoms of bowel cancer" (Robin S4, line 194),

Whereas Diane focused on information about the specific gene variant found in her family:

"some information that is for your specific gene marker would be quite helpful I think, because we all approach this quite differently" (Diane S11, line 462).

George was interested in:

"encouraging people to adopt a better lifestyle" (George, S1 line 425).

More information about a healthy lifestyle was the most frequently cited topic of interest, being mentioned by the majority (Anne, Bob, Fiona, George, John, Kay, Natalie and Robin). Advice that might help them in a practical way:

"That might potentially ward off or slow down the effects" (Robin, S4 line 201).

In more general terms, most participants described how the format of supporting information could be improved. John, Robin, Natalie, Sandra and Fiona all spoke about how information provided should be simpler or tailored to the age of the recipient. While Diane, George, Emma, Dave and Anne all expressed the view that it would be helpful to have information that was *"more positive"* or more sensitive to people's feelings. Emma described this aspect, saying:

"a little bit more.. sort of sensitive and encouraging about it, rather than just listing symptoms" (Emma, S3 line 114).

Information that was less clinical and focussed more on the benefits of knowing about the condition in the family, was considered desirable.

What was apparent across the interviews was that participants wanted information that was practical for their needs. They needed to apply the information they received to taking action in their lives. The issue of being able to make some difference to their risk of cancer came up many times, as did the importance of being able to access bowel surveillance at the recommended intervals.

7.7 Theme 4 - Appropriate communication

7.7.1 Existing methods of communication

When asked how they usually communicated with their family members it was apparent that participants used a range of different methods of communication with their relatives. They appeared to choose a way of contacting their relative that was appropriate or familiar for the family member concerned. Thus, Facebook messenger was used communicating with a niece (George) but a printed letter was better for an elderly parent (Fiona). Accessing information via websites was a common experience, so storing and sharing information via a secure website was envisaged as useful, if it was appropriate for the recipient.

For most participants communication about the familial diagnosis was initially either in person or over the telephone and this was seen as the ideal. For example, Dave's mother telephoned multiple family members:

"As soon as my mum found out, everyone, everyone in the family, both sides of the Atlantic were informed straight away by my mum" (Dave, S7 line 357).

Many participants reported using texting or social media to keep in touch with their family (Gina, John, George, Emma, Natalie, Sandra & Anne) and texting was described by Anne as less upsetting than communicating verbally. Therefore it was apparent that consideration was taken by participants to use a method of communication that was appropriate for the relative and the content of the message being conveyed.

7.7.2 Attitudes to the proposed website

Although most participants did not consider it appropriate to use email to make an initial disclosure about the familial diagnosis they did regard a secure website as a useful tool in disseminating information. It was clear that the majority did welcome the idea and they could perceive using such a facility to share information with their relatives. Many were enthusiastic, describing which different topics they would like to see covered on the website. As noted earlier, several already reported having searched the Internet for reliable information relating to their diagnosis.

Participants were almost unanimous in engaging with the idea of the website, including the function and the topics they would like covered in the resources. Only Bill appeared disinterested, although he did think information about diet and lifestyle would be helpful. In contrast, several people engaged thoroughly with the idea, commenting on how equality of access and improved public awareness could be a significant benefit to them and other people with the condition. Kay imagined how her brother might use the website as he was likely to take time to face the risk to himself, she said how providing information online might be really helpful to him:

"once he gets to that point where he feels "I can face it now" he can go and have a look and he can find the information but he needs to do it in his own time and in his own way, and I think having that ability would be the key thing to it. But it has not got to be immediate but if it is there and you have got access to it you can do it in

your time and you can do it as privately as you want to, to deal with it, and then discuss things once you have sort of taken that on board” (Kay, S12 line 379).

The novel function of the website, as a way of facilitating sharing information about the diagnosis, was possibly less tangible when raised as a theoretical option but still some of the participants considered how this might work for them. Natalie imagined the website:

“I don't suppose that I would mind having information personally on the website if it was password protected” (Natalie S5 line 355).

However, she insisted that she should remain in control of who could see what information due to the confidential nature of some documents, saying:

“I do feel that if we didn't have that control over what we could put, what could be seen on the website, I do feel I would be reluctant to want to take part” (Natalie S5 line 416).

John was more positive, he thought about the potential efficiency of using it:

“I think the more you can share information together the better certainly I was doing the same thing with my family it would be a lot easier if you can go on line to access and upload so that aspect sounds really good. Yes I think it sounds very sensible” (John S14, line 365).

Diane considered the justice of making it easier to access relevant information, not only for herself but for others:

“I think it's just everybody having equal access to the information or having ease of access where to go for it” (Diane S11, line 592).

An alternative perspective was expressed by Robin; he imagined how the website might help him manage his condition, saying:

“(if) you had a little portal yourself, and you had an area where new guidelines, new screenings, when your screening might be coming up, I think it would be helpful” (Robin S4, line 384).

Overall, the concept of a website which allowed confidential information to be shared securely was viewed positively, but as a way of reinforcing a disclosure that had already been made verbally.

7.7.3 Perceived limitations of providing information via a website

Some participants recognised the limitations of providing and sharing information via a website. Fiona described how electronic documents would not suit her father:

"I would use whatever was appropriate for the person I was talking to. So, you know, if I was trying to explain to my father then I would use a patient information leaflet. If I was trying to explain to my nieces, then I would say "Look at this website"" (Fiona S8 line 597)

Almost all participants explained how they would prefer to first inform their family members face to face but would telephone relatives who lived further away. Providing written information was described as helpful but needed to be sufficiently simple to be interpreted. The clinic summary letter was considered too detailed, personal and clinical to be ideal for sharing, as John said it was:

"probably too technical, too much about the medical aspects I guess rather than what you really want to know is what is the real likelihood that you have this gene, what are the things that you need to do in terms of you know protecting yourself, really what is the impact on your life, do you need to worry about it"(John, S14 line 212)

However, for several people the clinic letter was the only written information they appeared to have received. This meant that 'written information' was coded both within the barriers to sharing information theme and as a facilitating factor in accessing information.

Eight of the participants reported using the Internet to search for more information about their diagnosis, although one person (Emma, S3) was specifically warned by her

genetic counsellor not to search the Internet. She reported that her genetic counsellor had told her that the information she might find could be “scary”, “exaggerated” and might make her “feel even worse” (Emma, S3 line 100).

Therefore, using a secure website to share information was perceived as an acceptable approach but was regarded as not without its limitations.

7.8 Conclusions

The participants interviewed in this part of the research study were generally engaged with the need to improve information and support to themselves and their families.

They were able to imagine the potential uses of a website designed to provide information. Most of them were also able to envisage a function that would also enable them to share documents securely with their relatives. They had numerous suggestions regarding how information should be presented and what content it could contain, often qualifying these needs with how they wished to help themselves. However, some of the participants appeared to be struggling with their own psychological adaptation to their diagnosis and could only conceive of barriers to communicating with their relatives at this time.

Therefore the data from the interviews was very informative regarding the function and content of the proposed website. In addition, it provided insights into why some patients might find a website of this kind of limited use. I interpreted that some might look at the open access information on the site. However, they might not use the document sharing function if they themselves were not at an appropriate stage of psychological adaptation to be ready to share information with their relatives.

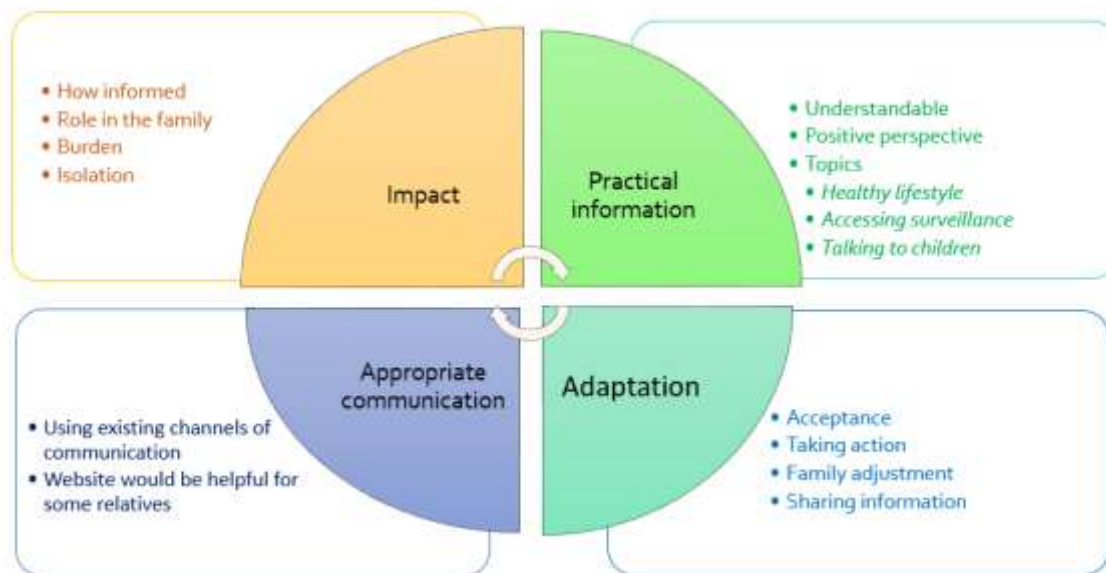


Figure 7.1 Phase 2 - major themes and their suggested interactions

The four major themes identified of **impact**, **adaptation**, **practical information** and **appropriate communication** could be regarded as interacting with each other as illustrated above (Figure 7.1). The postulated interrelationships between these themes and how they are relevant to familial communication are discussed in more detail in Chapter Nine.

In the next chapter (Chapter Eight) I describe the results of 12 Think-Aloud interviews, which were conducted with volunteer users from across England and Wales. These interviews enabled the website content and function to be viewed and tested by people who had been diagnosed with a high risk of bowel cancer within the last two years. The three cycles of interviews contributed to the development of the website and tested its usability.

Chapter Eight

Phase 3 Think-Aloud interviews and website development

8.1 Introduction

As described earlier, patients' views were investigated regarding which supplementary methods of providing information were most likely to be welcomed through a cross-sectional survey (Phase 1, Chapter 6) and telephone interviews (Phase 2, Chapter 7). The concept that providing information in alternative formats could support patients when they were sharing information in their families had culminated in the creation of a website. Whether the website approach and what it provided were both acceptable and feasible were then explored through a series of Think-Aloud interviews. These interviews involved volunteer users visiting the website while being interviewed via an online video conferencing platform called GoToMeeting (LogMeIn, 2017). This meant that volunteers could be in a location of their choice and using a computer, laptop or tablet that they were familiar with during the interview.

The process of website development was an iterative one with three different stages. In the first stage volunteer users were given the link to the website at the start of the interview so that their immediate reactions to the site could be recorded. In the next stage, the group of four different users were each provided a link to the website via their email two days prior to the interview and encouraged to view the website and sign up as a user to it. This allowed for more reflective reactions to the site to be

recorded. The final group were given a link to the website several days before the interview and encouraged to try the functions of signing up and sharing documents.

Recording the Think-Aloud interviews using GoToMeeting allowed the interviews to be transcribed into a matrix (Appendix 27) where the participant's remarks were linked to the specific area of the website which they were viewing at the time. As in the previous chapter, all names of participants have been changed to preserve their anonymity and only pseudonyms have been used in this text.

8.2 How the survey and interview results informed website content

The survey conducted as Phase 1 of this study provided evidence that the participants wanted to receive additional supporting information about the familial diagnosis in a range of formats (Chapter Six). The participants were all either living with a high risk of bowel cancer, or from a family where a genetic susceptibility to bowel cancer had been found. They had indicated their need for information on a range of topics relevant to their condition. The cross-sectional survey captured data regarding which issues were of particular interest. This then enabled information on these topics to be included on the website.

The 14 telephone interviews that followed the survey also contributed data regarding what issues interested participants (Chapter Seven). These were: their cancer risks, symptoms and relevant screening; how changes in diet or lifestyle could influence cancer risk; information about specific genes; how to talk to children about the diagnosis; starting a family and inheritance; research opportunities; insurance implications, and where to go for support. In addition to the choice of topics covered in the open access resources, the interview participants had been asked to envisage

whether they would use the proposed website and how they would use it. All but one of the interviewees responded very positively and provided examples of how they might use it. The main considerations that they described were the ability to control who saw what personal information and how to educate their GP or specialists about their condition. However, several participants thought it was important to have information that described their condition more favourably, stating the benefits to knowing the diagnosis. This positive perspective they described as generally lacking in what was currently available to them. Participants wanted to be able to present information about the familial diagnosis in such a way as to explain the potential benefits of knowing about it; this was perceived as an important incentive in informing their relatives.

Questions around how they found additional information following their diagnosis indicated that searching the Internet, talking to their relatives and interacting with other people with the same condition via an online support group, were the most significant sources of information for survey respondents. Whilst most interview participants described how they thought it was important to communicate initially in person or over the telephone with their relatives about the diagnosis, they also described how they used email or social media to communicate with their relatives at other times. Therefore using a secure website to share personal information relating to the familial diagnosis was endorsed as an acceptable approach.

8.3 Initial website structure and design

The purpose of the website had been described to the web developer including the importance of creating a website with a high degree of security in order to maintain the confidentiality of documents. Modern Websites (www.modernwebsites.co.uk)

was chosen following a process of informal tender, competing against four other web development companies. This professional web developer was chosen because he engaged with the concept of the website, he was experienced creating commercial websites, he had undertaken other academic projects before and his quote was within the allocated budget.



Figure 8.1 Screenshot of Homepage banner

The choice of images, as well as the functional aspects of the website, were all discussed as part of the development of the initial website (Figure 8.1). Since some of the funding for the website development was provided by the charity Bowel Cancer West, their logo was placed on the title banner alongside Plymouth University who were the academic sponsor. The 'Family Web' logo was designed by the webdeveloper, again following discussion with the researcher.



Figure 8.2 Screenshot of the original 'About Family Web' page

It was considered important to explain to users why the website had been created (Figure 8.2) and that the study had received NHS Health Research Authority approval. A small photograph of the researcher was put at the bottom of the 'About the Family Web Study' page in order to highlight that this study was being conducted as part of a PhD but the researcher had clinical experience in the NHS (as a Registered Genetic Counsellor) that had informed her approach.

In the preliminary stages, and before the website was live and available to users, the structure and images were assembled in a draft form to help the researcher envisage the website and identify where content was needed for the different pages of the site (Figure 8.3). Therefore the construction of the website was through a close collaboration between the researcher and the webdeveloper.



Figure 8.3 Screenshot of a basic web page prior to addition of content

It was decided that people accessing the site could be at different stages of their understanding about the familial diagnosis. Some might be aware of their family history of bowel cancer but without having received specialist advice, while others might have known of the diagnosis for some time and be concerned about the current management of their condition. Although the focus of the website related to the

importance of making relatives aware of the diagnosis and the potential health implications, it was acknowledged that users might have a range of different information needs. Therefore the organisation of the open access resources was considered important and if possible needed to be logical and intuitive. The life cycle approach that was used to organise the information available on the website into 'Your Journey' (Figure 8.4). This resulted in a series of pages on the website which included information to suit different perspectives depending on whether someone was a concerned relative or someone living with a diagnosis. These were entitled:

- **Before Diagnosis**
 - Being seen by the Genetics Team
 - What is Bowel Cancer
- **Sharing the news**
 - Why telling your family is important
 - What do your family need to know
 - Talking to children (Figure 8.5)
- **Living your life** (Figure 8.11)
 - Healthy Lifestyle: Diet
 - Healthy Lifestyle: Alcohol
 - How to get the right screening for you?
 - How could a genetic test affect your insurance?

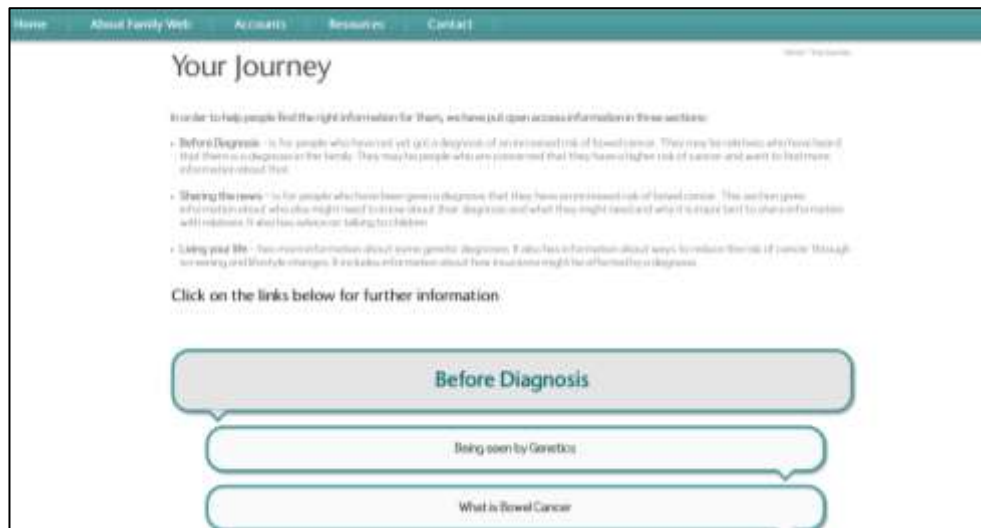


Figure 8.4 Screenshot of 'Your Journey' introduction and first topics

The different topics covered in the open access resources were guided by the participant responses to both the cross-sectional survey and the telephone interviews (Section 8.2).

In addition, a 'Useful websites' page (Figure 8.22) had a brief description and hyperlinks to a variety of websites that provided more detailed information about some topics.



Figure 8.5 Screenshot of the topic 'Talking to Children'

The views of participants captured in both the survey and interviews had indicated that giving a positive or optimistic perspective would be welcome, therefore the images and content were written to reflect this.

8.4 Think-Aloud Interview volunteers

Participants in the Phase 1 survey who had indicated that they would consider being interviewed were contacted by email or telephone. In addition, eligible patients were approached by letter (Appendix 17) or in a clinical setting by their health professionals at the six NHS recruitment sites and given an information sheet (Appendix 20). The process of the Think-Aloud interview was explained in more detail and the fact that they would need to have access to a computer, laptop or tablet plus a telephone for the duration of the interview in order to have their reactions to the website recorded.

Volunteers who wished to be interviewed were asked to sign a consent form (Appendix 19) and return it. Before recording each interview their decision to take part was confirmed verbally.

Table 8.1 Characteristics of interview volunteers and duration of interview

Number	Alias	Gender	Age	Interview date	Duration
First Phase		No prior experience – first impressions			
#1	Oliver	Male	34	13.04.2017	56 minutes
#2	Luke	Male	39	26.04.2017	31 minutes
#3	Jane	Female	42	27.04.2017	1 hour 6 minutes
#4	Freya	Female	25	27.04.2017	1 hour 7 minutes
Second Phase		2 days prior access to website before interview			
#5	Theo	Male	63	04.05.2017	1 hour 17 minutes
#6	Mike	Male	65	07.06.2017	1 hour 30 minutes
#7	Annie	Female	48	13.06.2017	55 minutes
#8	Harry	Male	69	14.06.2017	13 minutes recorded
Third Phase		~ 1 week prior access to website before interview			
#9	Mark	Male	48	17.07.2017	1 hour 34 minutes
#10	Stella	Female	60	19.07.2017	1 hour 16 minutes
#11	Jenny	Female	48	31.07.2017	1 hour 19 minutes
#12	Keith	Male	47	31.07.2017	53 minutes

The average age of the men taking part in interviews was 52 years and for the women it was 45 years. The participants in the interviews were selected as a purposive sample with a range of ages. The intention was to have equal numbers of men and women taking part in each of the three iterative phases of development.

8.5 Generalised responses to the Think-Aloud interviews

8.5.1 First Iterative phase responses

The first iterative phase of testing the website with four volunteer users (Table 8.1) sought to capture immediate impressions of the site. The users all expressed interest and commented on how much they liked the look of the website:

"This is exactly the.. type of resource that I would have liked to have access to when I first got the diagnosis to be honest and there wasn't anything like this made available to me." Oliver (#1)

"I think the content looks really good and the way it is laid out is really good, it is really clear." Luke (#2)

"It is nice and clear, it looks like a good page, I would look at this. I can see what it is all about". Jane (#3)

"Looks nice, looks good.. will it get an NHS badge?" Freya (#4)

However, they did not immediately grasp what the function of the website was intended to be, Luke (#2) said:

"It's not instantly clear what it is regarding.. .. is this just about people with bowel cancer awareness?" Luke

Whilst Freya (#4) did not know how to categorise herself, whether as a 'patient', 'family member' or 'professional' since she could identify with all three terms. She explained that because she had not been diagnosed with cancer herself, she was not sure she felt like a 'patient':

..from this bit I can tell it is for a number of different people, I presume I click on the patient's thing? Although I also guess I am a family member so I am not 100% sure where I would, where I would go upon first getting here. Because I wouldn't, I think I wouldn't think I was a patient for someone who is at risk as opposed to someone having cancer is.. I don't know if I like that, I am not sure.. Freya (#4)

All the users in this phase liked the resources, two users drew attention to the way the material was arranged in sections in 'Your Journey' (Figure 8.4) saying:

"I like the fact that it is simple, I want to read it quickly. If I don't want to hear about it just yet I can go back to it". Jane (#3)

"I think it has a got a lot more information than anything I have seen before, even just looking quickly, just what you need to know, this bit here why telling your family is important, talking to children".. Luke (#2)

They all particularly liked the information about diet in the 'Living your Life' section:

"This is my favourite page of it all because these are the questions that I have had to go away and find answers to myself" Oliver (#1)

and

"I think the content looks really good and the way it is laid out is really good, it is really clear... I like the way you have listed about getting the right screening and the insurance". Luke (#2)

These and other comments by the users confirmed that the appearance of the website was acceptable and the way the resources were presented made sense to them.

Freya (#4) made a very pertinent suggestion that in her view more prominence needed to be given to the potential beneficial effects of following dietary advice.

When looking at the Living your Life page she said :

"I think I would put that right at the top." Freya (#4)

and then repeated:

"I think I would put that right at the top. Something like.. .healthy diet, exercise, everything, that is good for everyone but you are at a higher risk and it makes even more of a difference for you. Some people might think, I am at a high risk of getting it due to this problem with my mismatch repair, I am going to get it anyway, it doesn't matter whether I am healthy or not so why should I bother changing my life? But if your behaviour can have even more kind of.. if your behaviour can impact and have even more so then it is probably good to hear."
Freya (#4)

8.5.2 Second Iterative phase responses

The next group of volunteers (Table 8.1) were provided with the link to the website two days before their interview and encouraged to sign up to the website so they could comment on that process within the interview. One chose to have their interview at the University (Theo, #5) but technical difficulties were encountered with the recordings for two of the interviews (#5 and #8) which then limited the data captured during this phase of development.

The two other users (Mike, #6 and Annie, #7) had problems signing up to the website and did not understand that they needed to activate the account before using the document storage and file sharing facilities. Although Mike acknowledged that he had not read the instructions:

"I had not read instructions about how to activate the account". Mike (#6)

Another user (Harry, #8) found the process "easy" and did not have problems but then expressed his concern that because changing a password was easy to do he thought this might make the website less secure.

Again, in interviews #5, #7 and #8 the users commented about how much they liked the look of the website. One person explained how they understood the purpose:

"Quite straightforward. Initially I would take it as a sharing site where I can add to it and other people can add. The initial feel I get is that it is like a one stop shop" Theo (#5)

While others liked the information resources:

"I think you have pitched it pretty much spot on, it's clear it is uncluttered, you are not constantly having to open documents." Harry (#8)

"Particularly liked resources. Sat and watched videos. Really good, I did like the resources.. I think this is good, very useful information, very clear" Annie (#7)

However, 'Mike' (#6) was critical of many aspects of the website saying that in general he found the text "too wordy" and questioning who it was intended for:

"What audience are you looking at? Some of the things are way beyond layman level? People are not going to plough through lots of stuff that is going to frighten them." Mike (#6)

He didn't like the text being in dark grey as he felt it was harder to read. He was particularly critical of the diagram of biological mechanisms (Figure 8.13) and thought people would not get to the most vital information:

"probably not hitting the crux of the thing about passing on knowledge of risk of cancer." Mike (#6)

When the document sharing function was demonstrated during the interview Mike was more positive, as he liked that aspect but he did say that it was still too complicated. The others were more optimistic about using the site, thinking how they might share documents with their relatives:

"This seemed fine, that seemed straightforward... I know my brother would be very keen I wanted to send him a link" Annie (#7)

One aspect that seemed to be confusing was how to share files that were uploaded and it became apparent that users were not interpreting the blue folder icon (Figure 8.24).

8.5.3 Third Iterative phase responses

In the last phase of development the volunteers were sent the website link in advance of their interview (Table 8.1). This ranged between 4 days (#9) and 13 days (#12) in advance and they were encouraged to sign up and try out the site. In fact none of the users had attempted to share documents with their relatives by the time they were

interviewed. Two of the users (#10 and #11) expressed their enthusiasm for the concept and endorsed the fact that their relatives were likely to benefit from it and probably would engage with it. One said:

"I liked the fact that you can add only people from your family that you want to add. So they can get to your information and if they don't want to sort of or they feel uncomfortable talking to me, they can access the information without having to approach me so that they have got their own privacy with it. I think that is good for the grandchildren more than anything." Stella (#10)

and another said:

"I would send a link to the website to friends and family because it is very informative... My son would much prefer to receive an email with a link on it than a letter in the post." Jenny (#11)

Although a different user was concerned that the approach would not suit all relatives and some might still prefer a printed document:

"It is the modern way isn't it. It will probably help people like me and in my age group. Whether it helps people of an older age group would be different. They are more inclined to want a paper version aren't they?" Keith (#12)

The document sharing function was demonstrated during the interviews by the researcher, through pretending to be their health professional and connecting with each user to share a variety of documents. Two of the three for whom this worked successfully were apparently delighted, saying:

"Oh, my documents!" Stella (#10)

"I love the icons, this is really good!" Mark (#9)

Although the final volunteer Keith was less enthusiastic, he did comment:

"I thought it was all pretty straight forward really. There wasn't anything that I thought "Oh that shouldn't be there."" Keith (#12)

and when discussing the website overall he remarked:

"It is a good database isn't it?" Keith (#12)

Since the volunteers had not attempted to connect with their relatives before the interviews, it was necessary to explain the function and what their relatives would encounter, such as the email invitation (Figure 8.6).

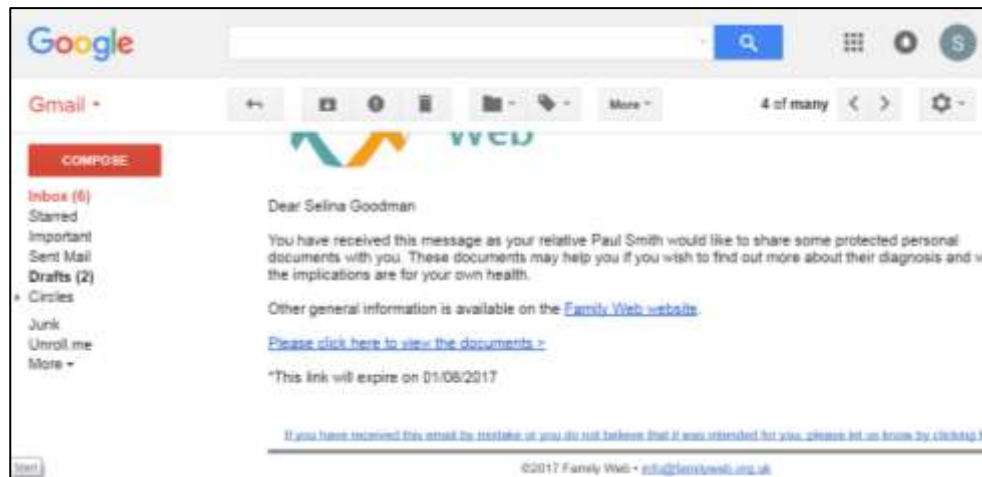


Figure 8.6 Screenshot of the email message received inviting a relative to view documents

One volunteer user (#10) was particularly excited by the website and how it was specifically designed to meet her needs:

"The actual look of the site is brilliant. I think it's modern, I mean the colours are great. I know that sounds silly but it's all visual isn't it, things like that. I think the links are good, and you tend to read things and then all of a sudden you think oh.. you go into it and think oh... I don't always do that with a lot of websites if I am looking for stuff... so it did make me want to explore a little bit more" Stella (#10)

Qualifying that she found it particularly helpful to be able to read information at her own pace, adding about the open access resources:

"I think it is much easier to cope with than when you are actually talking to somebody. I tend to get much more emotional with people. And I prefer to have something that I can read and digest and think about, and it doesn't get so muddy, and it's sort of then you can start making informed choices for yourself and rationalising whether it is important for you and whether there are things you can improve for yourself.." Stella (#10)

While another, Mark (#9) liked the resources section and endorsed the importance of informing his relatives:

"It is very factual it is very good. The pitch is right..."

"One of the biggest things for me is when do I tell my family, what do I tell them"
Mark. (#9)

8.6 How the interview results translated into changes to the website

For each volunteer user a matrix was constructed linking their comments to specific areas of the website as they interacted with it during the interview. These data were amalgamated and summarised (Table 8.2) to illustrate how suggestions for improvement led to changes in the website.

8.6.1 Matrix of the changes made to the website

Table 8.2 Changes made to the website linked to suggestions given in the Think-Aloud interviews

Interview	Area of site	Suggestions for improvement	Changed
#4, #6, #9, #10	Home page	Need to say what people can do, what they can find on the site, at a glance list showing relevance to users.	Bullet points added with short cuts to provide quick access to different parts of the site. <i>(Figure 8.8 to Figure 8.9)</i>
#4, #7 #2, #4	About Family Web	Banner picture too big, you have to scroll down to see text. More explanation needed about what website function is.	Some pictures removed or reduced in size <i>(Figure 8.11)</i> . Graphic created to show function of the website. <i>(Figure 8.10)</i>
#4	Account information	Banner picture unnecessary, just obscures information	Banner picture removed <i>(Figure 8.17)</i>
#7, #12 #1, #6	Patient sign up	Problems with creating username Need more directions for sign-up	Added statement that username must be in lower case letters <i>(Figure 8.17)</i> Text added to explain account activation needed before use. <i>(Figure 8.16 and Figure 8.17)</i> More detail added to the instructions. <i>(Figure 8.23)</i>
#9 #4, #5	Document sharing	No partner as option in drop down list of relatives Blue folder icon meaningless	'Partner' added as an option to drop-down list <i>(Figure 8.20)</i> Change blue folder icon to 'Share Files' <i>(Figure 8.24 to Figure 8.25)</i>
#4, #6, #6, #10, #11 #2 #7	Living your life	Disliked biological mechanisms graphic. Screening very important so wanted more information about it. Information about aspirin & CAPP3 Some pictures distract from the text.	Biological mechanisms graphic moved to another page. <i>(Figure 8.13 to Figure 8.14)</i> Added more information about colonoscopy <i>(Figure 8.12)</i> Information about aspirin added <i>(Figure 8.15)</i> Picture reduced in size <i>(Figure 8.21)</i>
#6	Useful websites	More links to different sites wanted	More websites added, including link to CAPP3 study and NICE guidelines re tumour testing. <i>(Figure 8.15)</i>

Think-Aloud Interviews

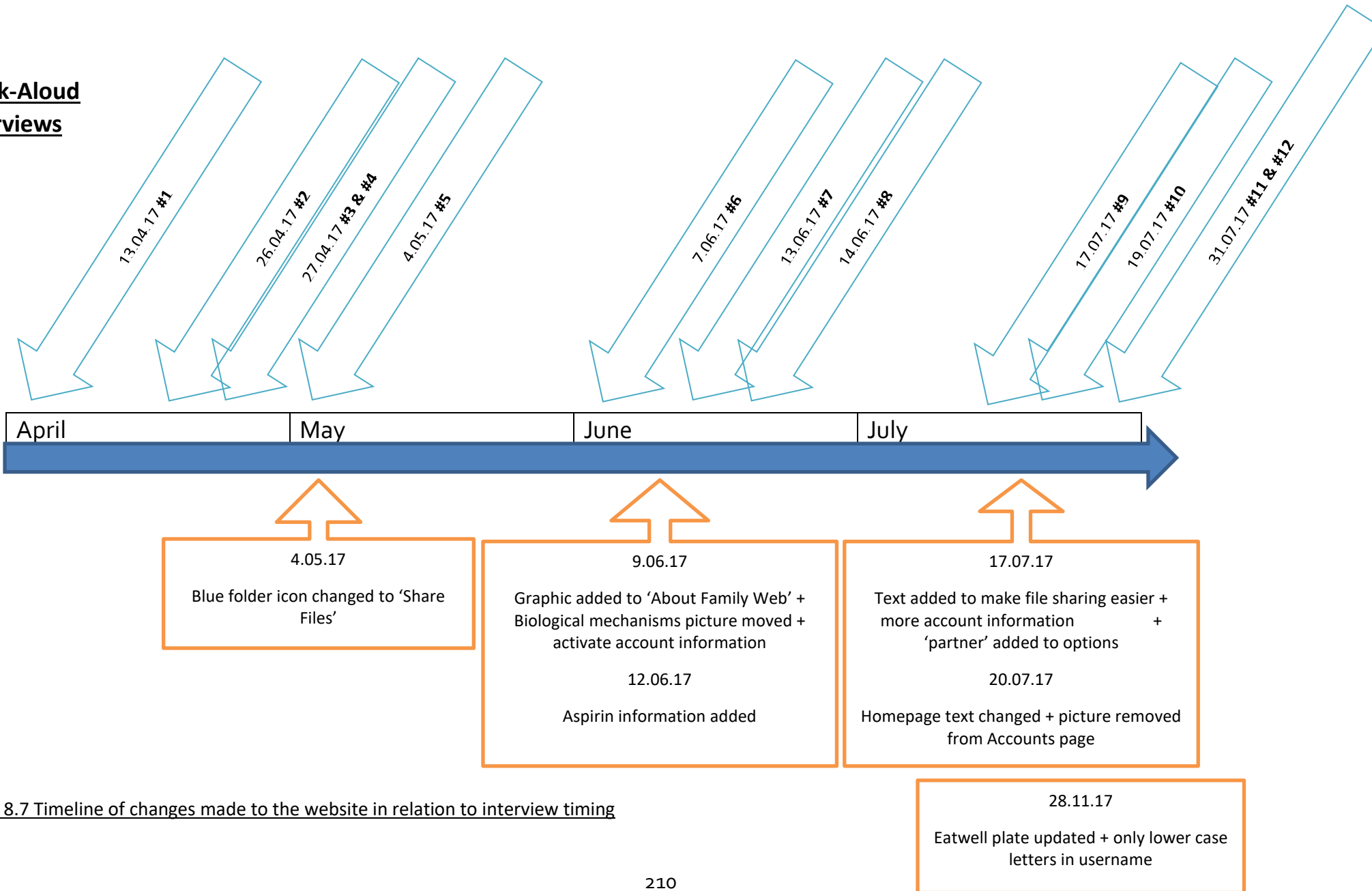


Figure 8.7 Timeline of changes made to the website in relation to interview timing

8.6.2 Homepage

The picture of a cheerful family used in the banner to this page (Figure 8.1) was broadly welcomed by the volunteer users. One commented:

"I liked the feel of it. It is really good, it is quite a positive feeling" Mark (#9).

While another said she liked the picture and the test was sufficient:

"Nice picture, not too much information at one go. Little bits and not too much. Very happy, very straightforward" Annie (#7).



Figure 8.8 Screenshot of the original Homepage layout

However, both Mike (#6) and Mark (#9) said that they thought the information (Figure 8.8) needed to be simplified, so bullet points were added to help people orientate towards the areas of the site that would be of most interest to them (Figure 8.9).



Figure 8.9 Screenshot of the updated Homepage with bullet points and links

8.6.3 About Family Web

This page on the website explained why the website had been created and provided a brief description of who the researcher was and the NHS ethical approval. When volunteer users were asked to read and comment on this page many still asked questions about the purpose of the site. This led to changes being made and a diagram representing the file sharing function of the site was inserted to make the function clearer to users (Figure 8.10).



Figure 8.10 Screenshot of the updated 'About Family Web' page

This graphic was well received by the later phase users and did appear to contribute to understanding. One user commented that they didn't like the pastel colours but no other negative comments were recorded.

8.6.4 Living your Life resources

Lifestyle resources welcomed

The Healthy lifestyle sections (Figure 8.11) were almost unanimously praised by the volunteer users in their interviews, one spoke of the reassurance he felt reading the information:

"The living your life bit, which I have just read, that is just brilliant!"... "I don't know, instantly, I just felt a sense of confidence and reassurance about my own situation by looking at this website." Oliver (#1)



Figure 8.11 Screenshot of the original 'Living your Life' page

Although one volunteer was less enthusiastic, saying that much of the advice was 'common sense':

"A lot of it to me is common sense. Life is like a pair of scales, what you eat and drink can balance your outcomes in life. Aspirin is an interesting one. I was asked to go on to aspirin once as a study but I wasn't keen. It is everything in moderation really" Keith (#12).

While another volunteer felt there should be even greater emphasis on the ways that people could reduce their risk of cancer through changes to their lifestyle:

"This is what I was particularly interested in, the prevention angle". Freya (#4).

Information about screening

There was some criticism of this section of the website that there could have been more detailed information about screening and colonoscopy. One user said what an important aspect of her condition this was:

"How to get the right screening is quite important. Obviously diet, lifestyle, alcohol but the screening is what is most important. People want to know what they have to do." Jenny (#11)

She also suggested adding practical advice about how best to manage the preparation taken prior to the colonoscopy:

"I learnt that the prep needs to be ice cold, drink it through a straw, drink extra fluid like ginger ale, some people eat jelly, clear broth. You can still drink black coffee and it is important to keep hydrated" Jenny (#11)

While another volunteer user made the suggestion that it would be helpful to explain what a colonoscopy involves, with diagrams, in order to show people that it is:

"nothing to be afraid of" Mike (#6)

These comments led to the inclusion of a short animated video (Figure 8.12) of what a colonoscopy involved.



Figure 8.12 Screenshot of the updated screening information page

Dietary advice pages

Several users commented on the diagram representing the biological mechanisms linking food consumed to bowel cancer risk (Figure 8.13). One person particularly liked it but qualified that by saying it was probably because he was a scientist. Most disliked it and thought it would be confusing or daunting to most people:

"I think for a patient website I think this graphic is a bit much. I have a degree in biology and I think this is a bit much. I don't know what glucosamine is.. I think the summary of the paper is nice and the link to the paper but the use of acronyms and words that patients won't understand is a bit overwhelming."
Freya (#4)

"Diagrams and explanation about how food is metabolised that right over my head." Mike (#6)

Therefore this diagram was moved to a separate page and a link to that page was put below the advice about eating red and processed meat (Figure 8.14).

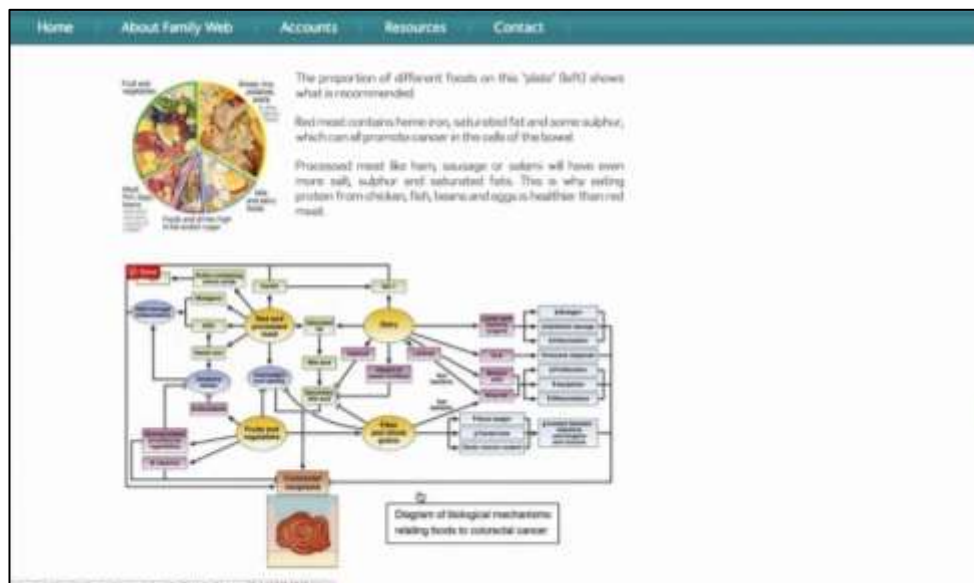


Figure 8.13 Screenshot of the diagram of biological mechanisms and 'Eatwell plate'

Another image that was changed was the 'Eatwell plate' image, which illustrated the best proportions of foods that should be consumed. Freya (#4) was aware that this advice had recently been updated, so the plate of food image was changed to the

most recent 'Eatwell Guide' (Figure 8.14) published by Public Health England in February 2017.



Figure 8.14 Screenshot of the updated dietary advice and 'Eatwell plate'

Information about aspirin

Initially the website did not include information about the risk reducing properties of taking regular aspirin. However, several of the people who took part in the Think-
Aloud interviews were surprised by this as they were already taking part in a dose optimisation trial of aspirin, the CaPP3 trial.

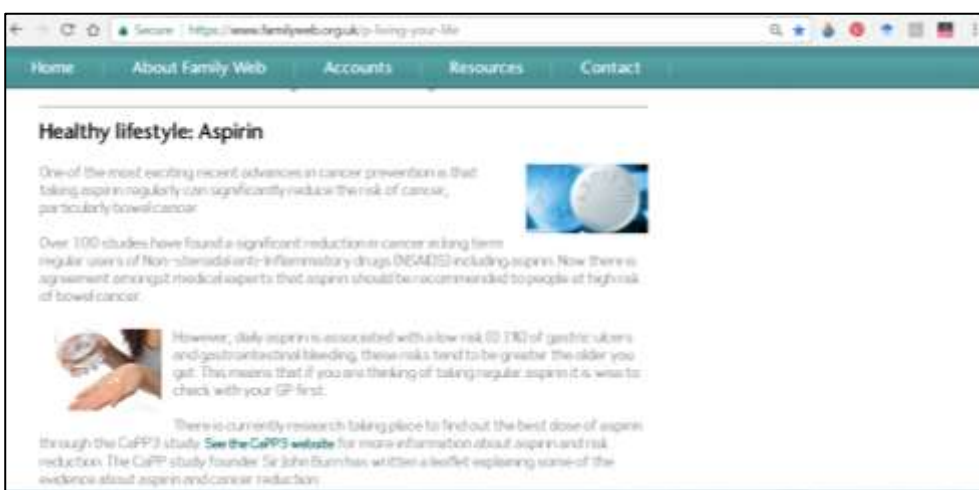


Figure 8.15 Screenshot of information about aspirin added to the website

This led to the inclusion of an additional section with a hyperlink to the trial website (Figure 8.15). This page was positively received in the later Think-Aloud interviews, particularly by Mark (#9) and Stella (#10).

One person emphasised how important it was for patents and their relatives to be aware of medical advances and what research they could contribute to:

"We are part of the CAPP3 study, so these studies are going on. It would be nice to know what else is going on behind the scenes" Luke (#2).

8.7 How the website function was improved

Problems with the function of the website that were observed were difficulties in the process of signing up to become a 'member' (Figure 8.16) and therefore have access to the secure document storage and file sharing facility. The difficulties were in two main aspects, the choice of username and understanding that the account had been activated. The webdeveloper had specified that usernames could only include lower case letters but this wasn't immediately clear so this was then stated below the username entry box (Figure 8.17).



The screenshot shows a web browser window with the URL <https://www.familyweb.org.uk/patient/sign-up>. The page has a teal header with navigation links: Home, About Family Web, Accounts, Resources, and Contact. The main content area is titled "Patient Sign Up" and "Sign up to Family Web as a Patient". Below the title, there is a paragraph of instructions: "If you would like to register to Family Web as a patient, please fill in all the fields below and click 'Sign Up Now'. After that, you will be sent a password and a link by email. You must click on the message 'Click here to activate account' before any other action is possible." The form consists of four input fields: "Name", "Surname", "Email", and "Repeat Email". The "Repeat Email" field has a red "X" icon next to it, indicating an error. The page also includes a small link "Sign Up Accounts Patient Sign Up" in the top right corner.

Figure 8.16 Screenshot of the 'Patient Sign Up' page

The other recurrent issue around the sign-up process was that it was not immediately clear to the volunteer users that they needed to activate their account by clicking on a link in their email. Therefore this aspect was explained in the instructions for sign-up (Figure 8.23) and a sentence was added at the bottom of the sign-up pages for both patients and health professionals (Figure 8.17).

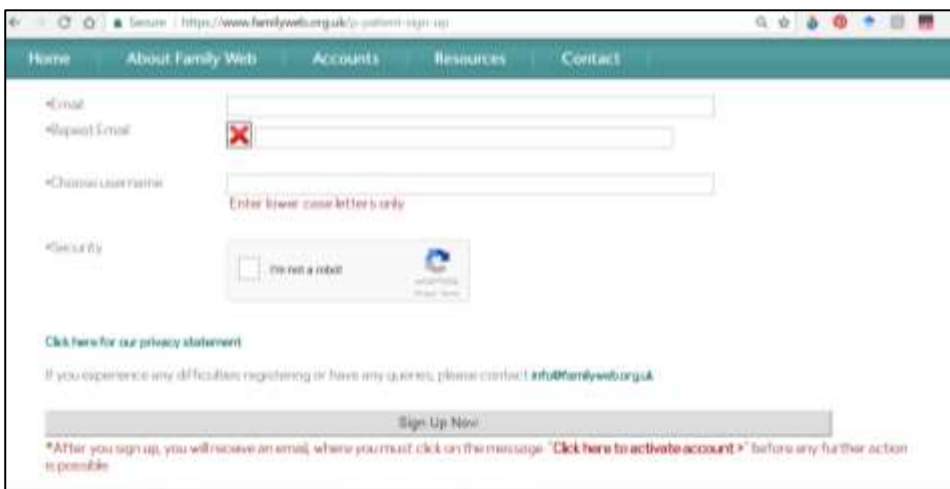


Figure 8.17 Screenshot showing additional information on the Patient Sign Up page

Two users (#4 and #7) drew attention to the size of the banner pictures on each page of the website. They complained that due to the size of the picture this obscured some of the text and made it necessary to scroll down the page to understand what the page was about.

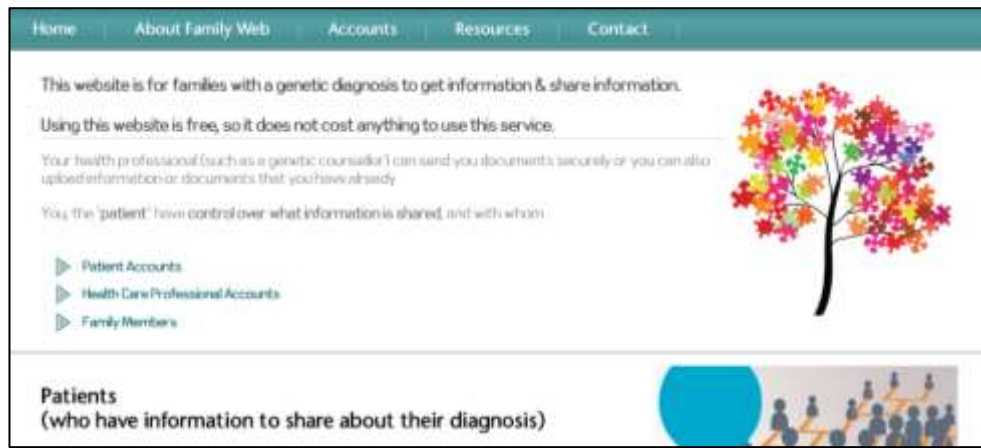


Figure 8.18 Screenshot of the account information page

This aspect was modified on one page with the banner picture removed (Figure 8.18) but was retained on other pages as the website was designed using 'responsive' technology where the image displayed adapts appropriately to the physical size of screen.



Figure 8.19 Screenshot of the Contact page banner

The researcher also wished to maintain a variety of images within the website to keep a colourful appealing appearance (Figure 8.19).

It was only when volunteers were engaging with the site and utilising the file sharing function that one (#9) pointed out that there was no option of 'partner' on the drop down list of family members. The function had been envisaged as a way to help families share information about the familial diagnosis with those relatives potentially at risk of the condition. However, passing on files to a partner could also be useful, so this option was added (Figure 8.20).



Figure 8.20 Screenshot of the Sharing Documents Member page

In response to the interest that nearly all the volunteer users showed in the 'Living your life' section, more hyperlinks were added to the content (Figure 8.21).



Figure 8.21 Screenshot of part of the updated 'Living your Life' page

To accommodate the increase in text, the size of some images were reduced (Figure 8.21 and Figure 8.11).

Another aspect of the website which was welcomed by the users were the useful website links. These were increased in number during the course of development in response to some of the questions posed by users (Figure 8.22) but not due to any specific requests.

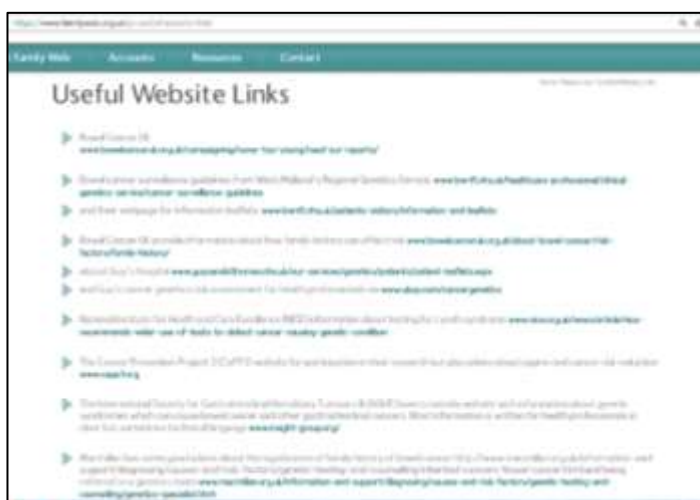


Figure 8.22 Screenshot of the updated Useful Website Links page

Although the webdeveloper had designed the sign up process to as intuitive as possible, several of the volunteer users (#1, #2, #7, #12) had problems getting signed up as a member to the site. In order to maintain the confidentiality of files being shared via the website it was necessary to create password protected



Figure 8.23 Screenshot showing additional instructions to assist sign up process

identities for users of the file sharing facility. Therefore more comprehensive step by step guides were written for both the patient users and health professionals (Figure 8.23).

Two of the volunteers (#4 and #5) questioned what the blue folder icon (Figure 8.24) meant when they had received files during the demonstration of file sharing with the researcher. However, it was apparent from the behaviour of the earlier users that they also found this icon uninformative and this icon was changed to 'Share Files' (Figure 8.25) which was a functional link and a necessary step when sending links to relatives to view specific files.

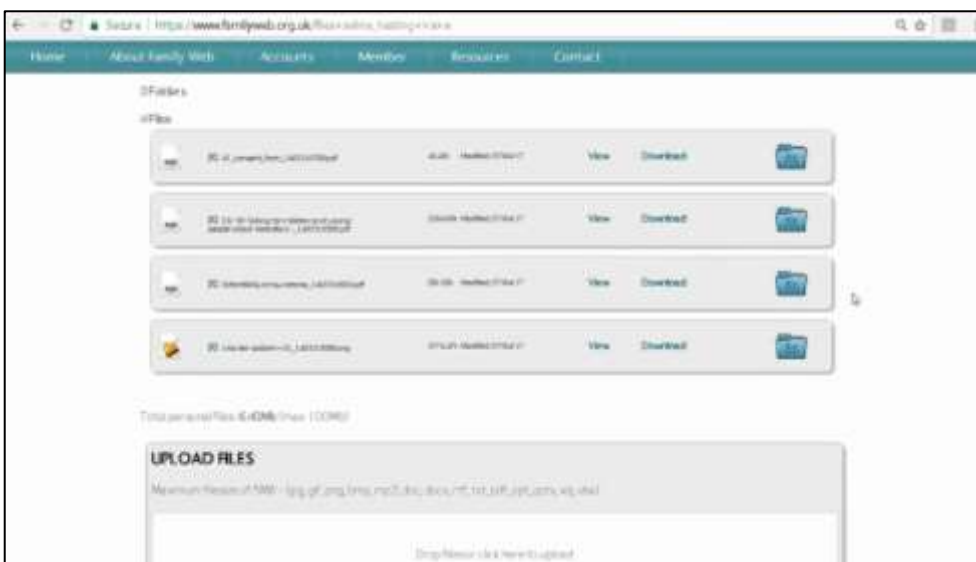


Figure 8.24 Screenshot of the original Member documents page showing the blue folder icon



Figure 8.25 Screenshot showing change from the blue folder icon to 'Share Files'

8.8 Suggestions for changes that were not implemented immediately

All the interview volunteers made comments and had creative ideas about how they envisaged using the website and what changes they felt would be helpful. Two of the volunteers (#4 and #6) made particularly numerous suggestions and followed up their interviews with emails to the researcher regarding their ideas. However, it was not possible to act on all the suggestions made due to limited resources and time constraints.

The website had been designed to provide a very high level of data security, exceeding the criteria specified by NHS Information Governance (Appendix 34) and following recommendations from the National Data Guardian and the Care Quality Commission (Caldicott, 2013; National Data Guardian, 2016). Nonetheless, Harry (#8) worried about the security of the site and wanted some indication that the documents were encrypted. However, the web developer had advised against putting overt descriptions about the data security on the website. This was because such statements were more likely to provoke interest from hackers who target websites that they perceive to be challenging. Instead he provided a detailed description the security mechanisms that could be shared with interested users or clinicians if necessary (Appendix 35).

Some of the ideas expressed would be developments that would align with the objectives of the website and therefore justify future consideration, such as adding a calendar option for patients to store a record of their colonoscopy or other screening dates (#7) and adding stories or video testimonies from users (#6). Additional content that was suggested included advice about how to break the news of the diagnosis to

elderly parents or adult children (#9 and #11) and more advice on where to access support, such as links to counselling services or what to do if users were feeling very distressed (#4).

A couple of the volunteer users spoke about aspects of the site which were particularly important to them and where they thought that the website should put more emphasis. Luke (#2) regarded the main benefit of the website to be in enabling him to connect online with his GP and specialists. He wanted to share reports with them, such as his most recent colonoscopy report where he had concerns about the size and number of the polyps found. Luke also really wanted information about current research:

"Just really.. what is happening from the medical side... What stories you read in the papers what is true and what is not... Just 'cos it gives you some kind of.. hope, more hope in the future about what is possible" Luke (#2).

Another aspect that needed a greater priority was awareness around the actions that people could take for themselves that would reduce their risk of cancer. Freya (#4) eloquently explained how this information could be transformative for some. She told me that she had not realised that healthy eating advice would influence her risk since she had inherited a susceptibility to cancer.

8.9 Conclusions

The website was an acceptable way of receiving, storing and sharing health information to this group of people. Several people recognised that using a secure website to share information was an approach that would not suit everyone; either because individuals were avoiding communication about the condition; or they were

someone who would find using the technology challenging. However, it was also acknowledged that providing information in a digital file would enable more people, particularly younger relatives, to access the information.

Almost all the volunteers were engaged with the idea and they enthused about different aspects of the site, particularly the information resources, but also how it enabled increased scope for sharing information. Two users (#1, #4) immediately recognised the opportunity the website provided to store and retrieve their own health information online when they were away from home, whether that was on holiday abroad or when they were seeing their GP.

The process of testing and refining the website as a tool for sharing information was possible with volunteers from across England and Wales through the use of an online platform for remote usability testing. Although this presented its own technical challenges at times, it meant that user's reactions to specific aspects of the website could be captured. This process identified problems that would have been a barrier to its use and allowed for the refinement of the website to more closely meet the needs of potential users.

Chapter Nine

Discussion

9.1 Introduction

My intention throughout this research was to investigate through a pragmatic approach an additional method of providing information and support to patients following a diagnosis of an increased vulnerability to bowel cancer in their family. In this chapter I will draw together and discuss the findings of the different phases of my research. I have organised this chapter into sections reflecting the four major themes identified in Phase 2 of **impact, practical information, appropriate communication** and **adaptation** (Figure 7.1 p.194) and I discuss how my findings complement existing theories.

I will consider in particular detail how the impact of learning about the familial diagnosis appeared to influence participants' ability to share information with their relatives. I review how the narratives of some participants could be interpreted as an indication of their adaptation to the diagnosis and show that this would be congruent with the Theory of psychological adaptive modes (Lehti, 2016). I will discuss how the Family Web website could be used to the benefit of patients. I will relate some of its functions to constructs within the Theory of Planned Behaviour (Ajzen, 2002) where the provision of information that encourages a healthy lifestyle could give patients greater 'perceived behavioural control' over their risk of cancer.

9.2 Impact of the genetic diagnosis

9.2.1 Impact on the family

I would like to elaborate on how profoundly the diagnosis affected some of the participants. This was not only something that affected the individual but an understanding that reverberated around the family and caused pain and anxiety to their relatives too. One participant described the effect it had on her and her son as “*devastating*” when they realised he had inherited the genetic vulnerability and “*very traumatic*” (Anne, S9 line 285). This illustrates the potential for patients to suffer compounded or disenfranchised grief (Doka, 1989).

The long-term consequences of genetic testing to influence and change family relationships were explored by Cowley (Cowley, 2016). She found through interviews with 15 members of one LS family that those relatives who had declined a genetic test were characterised negatively, being described as ‘selfish’, ‘ostriches’ or ‘frightened’ of the testing. Since none of the family members who declined testing agreed to be interviewed, their perspectives were not captured. Cowley points out that if the view point of those promoting genetic testing is accepted as the moral imperative then the right not to know is undermined. This in turn is likely to contribute to the long-term impact on family dynamics if not all family members share the same perspective. One of the interview participants (Bob) describes his feelings of isolation because he alone was the one person in his family to have a pre-symptomatic test. It was evident from his narrative that the genetic diagnosis has had a profound, and largely negative, impact on his family. Although the family dynamics were different to that described

by Cowley, as the tested individual was in the minority, my findings appeared to show evidence of long lasting disruption to relationships in this family.

9.2.2 Impact on the individual

In Phase 1, survey participants were asked how they thought support could be improved (Appendix 18, question 3). My analysis of their free text responses is illustrated earlier (Chapter Six, Figure 6.7). That analysis showed that several different issues may have contributed to a negative impact on some participants' mental state. Again, the impact of the diagnosis appeared central to the problems encountered and what aspects of care participants felt could be improved.

I would suggest that improved access to relevant information via a secure website could potentially reduce that impact. This is because part of the impact of an illness, as experienced by an individual, could relate to their uncertainty around how the condition will affect them in the future. In line with this view, Skirton and Bylund (Skirton & Bylund, 2010) argue that uncertainty management theory can be applied in genetic conditions. Using the model described by Lau and Hartman (Lau & Hartman, 1983) they suggest that 'controllability' is one of the five key domains that help people conceptualise the impact of the illness. The other domains are 'disease identity', duration, consequences and causes. Taken together, the illness representation that an individual constructs then enables that person to develop strategies to combat the illness. If this is the case, then it is quite logical that focussing on the controllable or malleable aspect of the risk of cancer is likely to provide hope and a sense of empowerment to patients.

A website provides the opportunity to provide more information to patients that is specific to them or generic about their condition. The flexibility of content that can be updated and expanded also gives the chance to provide more simple information, written to suit different reading ages or address specific questions. Where negative emotional reactions could be attributed to feelings of uncertainty, grief, or loss, but I think those feelings could be reduced or ameliorated by improved follow-up post diagnosis. I propose that a website like Family Web could be a useful tool in such follow-up.

9.2.3 Factors influencing communication in families

Other factors can also impact on whether communication is likely to occur and the clarity and accuracy of the communication (Keenan *et al.*, 2005). The map of factors (Chapter Six, Figure 6.7) was constructed as part of the thematic analysis of the Phase 2 interview data and illustrates the complexity of the influences on communication. Important factors previously identified include: the social norms of communication (Forrest *et al.*, 2003), understanding and knowledge (McAllister, 2003), anticipated reactions (Lafreniere *et al.*, 2013) and feelings of responsibility towards relatives (Dheensa, Fenwick & Lucassen, 2016).

It is recognized that within a given family social norms will vary, but typically 'vertical' communication is more likely, that is between parent and child, or vice versa (Forrest *et al.*, 2003). The identity of the family authority figure is also important as their views are likely to be particularly influential and the usual channels of communication in a family will influence communication about a genetic condition (Dilzell *et al.*, 2014; Koehly *et al.*, 2003). Families with less open communication around health are less likely to inform their relatives about the familial diagnosis or their potential risk of

illness (Bartuma, Nilbert & Carlsson, 2012; Petersen *et al.*, 2014; Rowland & Metcalfe, 2013). Often there is an assumption by family members that someone else will have informed their more distant relatives (Gaff *et al.*, 2007). For whatever reason, when communication about the diagnosis does not occur, this means that some relatives can be denied the opportunity to obtain information and take measures to reduce their risk of cancer. In order to address this I included in the Family Web website open access information in the 'Before Diagnosis' section that explains about bowel cancer, symptoms of bowel cancer and how to access clinical genetics services. I also wrote some basic guidance about communication in the 'Sharing the News' section.

9.3 The need for practical information

9.3.1 How patients are supported when sharing information about their familial diagnosis

Through the survey and telephone interviews I gathered data about what information participants had received initially and whether they had wanted more information.

These data revealed that participants had a range of experiences, with the majority reporting initial satisfaction with how they had learnt of their risk. Many also described how they needed more information about the condition and further ongoing support.

The survey data reflected how difficult genetic information was to assimilate initially. Most participants indicated that a follow-up appointment would be 'helpful' and three out of four participants would have wanted more information at the time of their diagnosis. The survey data were consistent with the results of an earlier survey (Lapointe *et al.*, 2013) with 246 men and women who had received genetic test results relating to BRCA gene variants. Lapointe found that over half (52%) were "highly

interested” in having access to an educational website to help support them communicate information about the genetic diagnosis to other family members. Their participants were also interested in receiving a personalised family letter (38%) or an educational booklet (50%) but only 35% of their participants were interested in a ‘family information session’.

In my study, the responses to both the survey and interviews confirmed my understanding about the current provision of information following a genetic diagnosis in the UK. This is that information is largely provided either through ‘family letters’ (Dheensa, Lucassen & Fenwick, 2017; Stol *et al.*, 2010) or with general information about the condition. Evidence from the study by Dheensa, Lucassen and Fenwick suggested that providing family letters was not necessarily effective. They used focus groups to elicit the views of 80 health care professionals and interviews to gather the opinions of 35 patients. They found that providing letters specifically written for distribution to at risk relatives was not ideal, the letters were difficult for clinicians to word appropriately and patients experienced problems distributing them. These authors concluded that providing *letters* was not sufficient to support sharing information in many families. They (Dheensa, Lucassen & Fenwick, 2017) advocated that HPs should discuss with patients what additional support they might need and consider contacting relatives directly if patients wanted this. Aligning with my own perspective, these authors state their intention to develop an “online resource to facilitate communication” which they regard as justified and appropriate in the context of the digitization of healthcare services (Dheensa, Lucassen & Fenwick, 2017).

Several Phase 1 survey participants commented on the letters they received; they found them too technical, medical and detailed. They also described how the information they received lacked guidance on what the impact would be for them, how they could protect themselves, and what they needed to be concerned about. The free text responses in the survey revealed tension between the needs of the individual and their sense of responsibility to their relatives. This tension has previously been described (Chivers Seymour *et al.*, 2010) and in addition, several authors (Mendes *et al.*, 2017; Stol *et al.*, 2010; Wiseman, Dancyger & Michie, 2010) have drawn attention to the potential disruption to family relationships when patients try to encourage their relatives to have a genetic test. Consequently content was included in the website that was intended to meet these needs. Different 'layers' of information were provided and guidance was given in the 'Sharing the News' section about how information might be received by relatives.

9.3.2 Methods of communication

Nearly half of survey participants reported searching for more information on the Internet and most of the participants in the survey and telephone interviews had reported using email, text messaging or social media to communicate with family members. Providing information in an electronic format could potentially help at least those individuals who already used information technology to communicate with their relatives. One interview participant, George, was frank that he used whichever method of contact that was preferred by the family member. He explained how he used Facebook to contact some younger relatives, although he admitted that he thought it was not an ideal way to convey complex information.

When investigating a method of digital communication that might be appropriate for development within the NHS I had decided that neither SMS texting nor social media (such as Facebook) were likely to meet the stringent data security requirements of the NHS (Caldicott, 2013; National Data Guardian, 2016; NHS Digital, 2017). Since that time the social media platform WhatsApp has introduced end-to-end encryption to ensure users privacy but this was not available at the inception of this project. In my experience, communication by email between health professionals and patients is controlled and often restricted or encrypted in many NHS trusts. Therefore, the preferences expressed by participants for digital technology endorsed the creation of a secure website as currently the best viable IT option available for information sharing.

9.3.3 The importance of receiving accurate and trustworthy information

A quarter of survey participants reported receiving no supporting information at the time they were informed of their risk. This could have presented these individuals with problems, both in accessing appropriate screening or referral for more detailed advice. The fact that these people were not given written information meant that they were more reliant on what they had been told verbally. Potentially these people could have had an inaccurate understanding of what the familial diagnosis meant for them (Sustersic *et al.*, 2017) which in turn might have inhibited sharing information or affected what they told others.

The perceived reliability and trustworthiness of information provided through the NHS in the UK (Williams, Nicholas & Huntington, 2003) was specifically mentioned by two participants who wanted to see the website “endorsed by the NHS” (Oliver #1) or

“NHS badged” (Freya #4). While not in the UK context, an interview based study of CF carriers in Australia (Gorrie *et al.*, 2017) noted that 18/21 of their participants were in favour of having an online source of information. Their participants acknowledged that most people do look for information on the Internet now but they can find it difficult to distinguish which sources of online information to trust (Gorrie *et al.*, 2017). The Family Web website has an explanation of its purpose (which refers to the NHS Research Ethics approval and recruitment through six NHS Trusts) and this may have been interpreted as endorsement for the content by interview volunteers. Without specifically probing this issue it is difficult to know how far the NHS ethical approval might have influenced their responses.

9. 4 Appropriate communication with relatives

9.4.1 Positive framing of information

Several authors have documented the wish to protect relatives (Chivers Seymour *et al.*, 2010; Mendes *et al.*, 2013; Metcalfe *et al.*, 2011). Similarly, my data showed that participants felt motivated to help family members to access appropriate medical services. Volunteer users liked the positive stance of the information provided. I would speculate that following their own diagnosis and before they could attempt to inform their relatives, patients needed to understand the benefits of knowing about the diagnosis. This was so they could share that hope rather than be ‘harbingers of doom’. I would argue that in order to convey a message of hope patients need to have the necessary information to give. The open access resources pages were written with this in mind. For example, the webpages under ‘Living your life’ describe the benefits of having a healthy diet and the opportunity to take aspirin to reduce cancer risk (Section 8.6.4, p.211).

One unexpected finding from the qualitative analysis illustrated how participants were taking a positive perspective, where some viewed their gene variant as being somehow better than the other gene variants. This finding, as far as I know, has not been reported before and it is interesting to speculate whether this could have been part of their coping mechanism? It appeared that participants compared the effects of different gene variants and distinguished an advantage in the one that affected their families.

9.4.2 Open communication about health issues

While most research focusses on the individual, some studies have examined how cancer survivors and their family members communicated about their genetic risk. Breast cancer patients and their unaffected relatives were interviewed in focus groups by Mellon and colleagues (Mellon *et al.*, 2006), who found that communication styles varied both within and between families. They found that those families with more open communication styles about cancer, and particularly where relatives had concordant styles of open communication, reported greater satisfaction with their interactions. Another of their findings was that email was used as a common method of communication for some families where they shared information they had gleaned from the Internet (Mellon *et al.*, 2006). This evidence endorsed the way that Family Web was set up to send invitations to relatives via email. In addition, some content on the 'Sharing the News' webpages encouraged readers to be open with their relatives and not to delay talking about the familial diagnosis.

Providing supporting information for patients for their own understanding, but also to help them explain the health implications to their relatives, has been found to be an important component of adequate support (Barsevick *et al.*, 2008; Daly *et al.*, 2016).

PHRs could provide the same type of information sharing function as the Family Web website. The advantage of using an existing PHR system is that it would have technical support associated with it and already be in use within a health care setting. Unfortunately not all health care providers have adopted this technology and evidence suggests that professionals need to view such systems positively if they are to utilise them (Nazi, 2013). Family Web could address this gap, although it can be used by families without the involvement of a health professional, it is potentially a more powerful tool when HPs upload documents that are specific to their patients.

9.4.3 Information that gave hope or empowerment

Almost half of survey respondents wanted more information about how to have a 'healthy lifestyle'. Several of the interview participants and Think-Aloud volunteers also spoke about their interest in adopting a healthier lifestyle and particularly eating food that might reduce their risk of developing bowel cancer. Considered from the perspective of the Health Belief Model (Rosenstock, Strecher & Becker, 1988) cancer risk reduction was a strong 'perceived benefit' in response to the 'perceived threat' to health posed by their inherited vulnerability. Dependant on the individual's self-efficacy in relation to dietary change (Visser *et al.*, 2017) improved understanding might have provided a 'cue to action'. Certainly risk reduction through changes to diet or lifestyle was one of the topics frequently mentioned across the telephone interviews (Section 7.6.2).

Information on the website regarding risk reduction through a healthy lifestyle was a particular focus in two Think-Aloud interviews (Chapter Eight, Section 8.5.1). One volunteer spoke of his "sense of confidence and reassurance" (Oliver, #1) when reading this content on the website. He described how he had now made changes to

his diet but at the time of his pre-symptomatic test he had struggled with the knowledge of his elevated risk. Another (Freya, #4) was particularly eloquent, explaining how she thought it was really essential to have information about the *benefits* of knowing genetic status. She told me that being given the opportunity to alter her risks of cancer through her own actions was a very important; it gave her hope and a greater sense of control. Both of these volunteers talked about how they had not understood whether general guidance on a healthy diet would apply to them. Therefore, the opportunity to educate or improve knowledge for those at risk was seen as vital. I was told that lifestyle advice needed more emphasis on the website. A reason given was that it would give those who might otherwise have a fatalist attitude about their risk of cancer greater confidence in their ability to alter their risk.

The feelings of hope elicited by such knowledge could be attributed to a greater 'perceived behavioural control' as defined by Ajzen (Ajzen, 2002; Ajzen, 2011) in the Theory of Planned Behaviour (TPB). This theory predicts that the controllability of certain behaviours are individual as the perception of the difficulty of performing the behaviour will be dependent on both internal factors (e.g. knowledge of what foods to avoid) and external factors (e.g. opportunity to exercise) which are specific to an individual and their circumstances.

The TPB construct of perceived behavioural control could also be applied to other issues. My findings suggested that where people's understanding of the potential benefits of being aware of what advice applied to them was a factor influencing their intentions. This could relate to how they themselves might access specialist services and cancer surveillance. Such understanding might also motivate patients when considering when, or if, they chose to discuss the diagnosis with their relatives.

Another construct within TPB theory is 'subjective norms' (Ajzen, 2002) where an individual's behaviour is influenced by their perception of the approval the behaviour would provoke with significant others. This is illustrated by one Phase 2 interview participant (Fiona, S8) who described how she needed to know what practical advice to give her brother. She didn't feel it was sufficient to just tell him about the diagnosis of LS because he wanted to know what he needed to do next.

It was evident when considering all data sources in this study that many of the participants were looking for some type of practical guidance and ways to deal with the uncertainty that was inherent in their genetic diagnosis. Consequently, 'practical information' was identified as one of the four major themes that emerged from the qualitative analysis. What participants apparently desired was information that could be applied in their lives; practical in format (simply written, accessible) and practical in content (covering issues of relevance and concern). Again, the way that information might be utilised could be seen as another example of providing improved 'perceived behavioural control' within TPB theory.

9.4.4 Using a website to facilitate information sharing in families

I recognised that viewing or using a website would be restricted to those patients who had access to the Internet. However, access to the Internet is now widespread as the majority of adults (88% of adults in the UK) are using the Internet at least weekly (ONS, 2018a). Plus 98% of young adults (aged 16 to 24 years) were using a portable device to access the Internet in 2017 (ONS, 2018b). It was estimated that over 60 million people in the UK used the Internet in 2016 (92.6% of the population)(Internet Live Stats, 2016). Evidence which is more specific to family communication showed that in the United States (US) 86% (n=1322) of parents reported using email to

communicate with non-resident family about parenting issues (Rudi *et al.*, 2015).

Therefore I judged that a website could be useful to most people. However, having the capacity to print and make a hard copy of information accessed via a website would still be important to ensure that the opportunities to share web-based information was maximised.

Another unanticipated finding was that volunteers envisaged using the website as a secure online repository for their own health information. This was not a primary function that I had considered when designing the website but it seemed logical when it was suggested. Two users (Freya, #4 and Mark, #9) explained to me how this could potentially help them to access their own records when away from home or if they had forgotten to bring printed copies to their appointments. They acknowledged that it would then be their choice if they shared any of their records with their health professionals outside genetics, family members, friends or employers. This function is one that most personal health record (PHR) systems would provide since password protected access enables patients to effectively take ownership of their health records when registered with a PHR (Chapter Two, section 2.11).

9.4.5 Difficulties or barriers to communication in families

Communicating about a health issue can be a difficult task within families where it is not the norm to discuss health or illness (Keenan *et al.*, 2005; Metcalfe *et al.*, 2011).

Talking about the risk of cancer can provoke painful memories in some when they have lost multiple family members to cancer. Therefore, providing a means of communication which allows relatives to view that information at a time of their choosing may be an advantage. One Think-Aloud interview participant noted how

giving relatives' access to their personal information online might particularly suit her adult grandchildren. She thought they might also appreciate the privacy this afforded and avoided the potential embarrassment of discussing issues related to bowel cancer with their grandmother (Chapter Eight, p.12 Stella, #10). Studies have shown that patients generally acknowledge their responsibility to pass on information about their diagnoses (Dheensa, Fenwick & Lucassen, 2016; Mendes *et al.*, 2013) but some information is considered more private or sensitive.

9.5 Adaptation to the genetic diagnosis

9.5.1 Theories of psychological adaptation

I analysed the Phase 2 interview data for signs of 'adaptation' and these were coded separately, in order to examine which individuals appeared to have adapted to their diagnosis and in what way. The issue of how well adjusted or adapted participants were to their diagnosis appeared to be fundamental to the process of sharing information. If adaptation is considered as a personal and dynamic process in response to change (Biesecker & Erby, 2008; Lehti, 2016), it is likely to be uniquely expressed by different participants. Rolland and Williams (Rolland & Williams, 2005) postulated that a long-term adaptation phase was part of the non-symptomatic stage in genomic disorders in their Family Systems Genetic Illness (FSGI) model. They described adaptation as following the test and post-testing phases in "at risk" individuals (Figure 9.1) and following an initial adjustment period.

Figure 9.1 image has been removed due to copyright restrictions

Figure 9.1 Non-symptomatic time phases of genetic disorders from Family Systems Genetic Illness model (FSGI) Rolland & Williams 2005.

Yellow circle highlights phases of adjustment and adaptation to the genetic diagnosis occurring over a time period.

Lehti (Lehti, 2016) describes a theory of psychological adaptive modes which postulates that an adaptive struggle in response to a stressor is necessary before the individual can achieve a new level of equilibrium. The adaptive struggle interferes with normal functioning. However, through the utilisation of coping skills such as seeking information or asking for help a successful adaptation can occur which results in a raised sense of well-being. This theory appeared to be consistent with some of my findings and could explain why adaptation might be a precursor to disseminating information. Such a theory could also explain the findings by Forrest (Forrest *et al.*, 2008) that seeking social support through confiding in close relatives comes before sharing information with the wider family, as described below (Section 9.5.4).

Although Figure 9.1 shows adaptation in relation to someone who is not symptomatic, I would suggest that adaptation is likely to be as important to those individuals who only receive their diagnosis of a genetic condition after their diagnosis of cancer. For symptomatic patients there may be the additional burden of coping with their cancer, both physically and psychologically, and then learning that their illness has health implications for their family members. One survey participant described how shocked she felt having seen a genetic counsellor in order to “do the right thing” for her family but not anticipating that this would be more than a logical conclusion to her cancer

experience. This illustrates how patients may experience a form of compounded grief or threat when they are given a genetic diagnosis on top of a life threatening diagnosis like cancer.

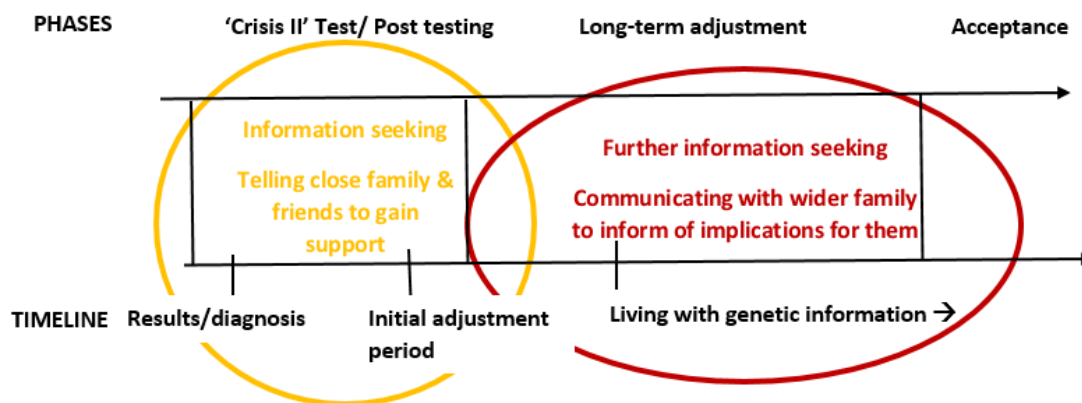


Figure 9.2 How adjustment or adaptation might influence information seeking and sharing

[Phases of adjustment modified from Rolland & Williams (2005) FSGI model Figure 9.1] Yellow circle highlights the initial adjustment period when close family are informed in order to seek support. Red circle highlights longer term adaptation where communication happens with the wider family and information is shared to explain implications of the diagnosis for others.

I postulate that the function of the Family Web website could assist patients in their longer term adjustment to their genetic diagnosis (Figure 9.2) as well as enabling them to share information with greater ease. I envisage that when a health professional provides information about the diagnosis to their patient via the website it will give that patient several choices. They can access the information at any time they have internet access, they can print the documents if they wish and they can share them with any number of relatives or health professionals at times of their choosing.

Initially sharing information may be associated with informing family members and seeking support for themselves. As they adapt and adjust to their diagnosis they can use the website to share information about the condition with more distant relatives. Since the website also allows text documents to be shared alongside other documents, it gives another level of non-urgent contact that could augment other methods of communication.

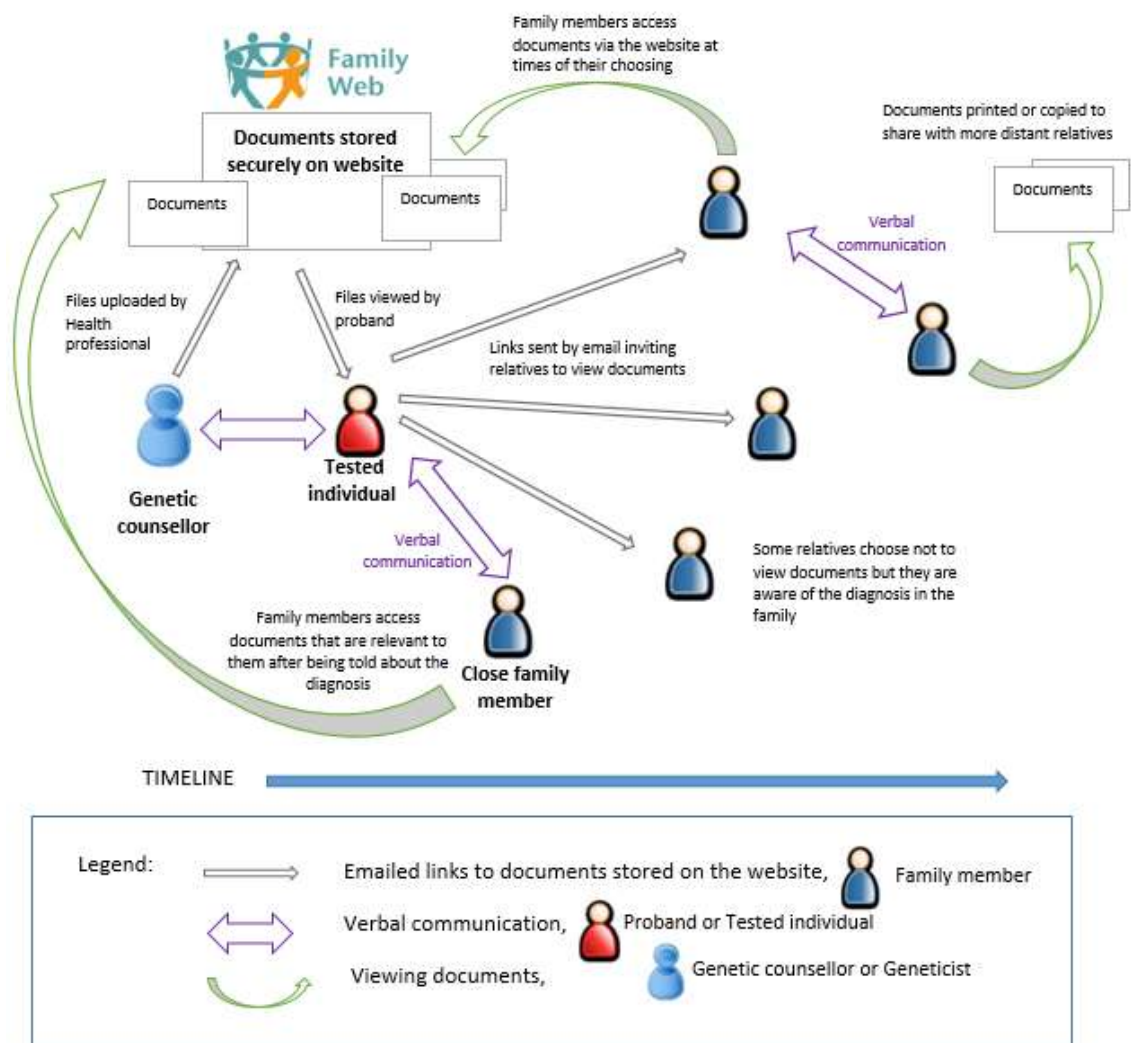


Figure 9.3 Diagram representing the document sharing function of the website

The function of the website as a tool to facilitate communication in families does require further investigation. The only available evidence regarding a similar initiative,

'The Suntalk study' (Bowen *et al.*, 2017) did demonstrate a significant increase in family members discussing their risk of melanoma in the group that received personalised health information via the secure website. This supports my hypothesis that giving patients access to personalised health information via a secure website is likely to facilitate patients sharing that information with their relatives and may also promote adaptation.

9.5.2 Long term adaptation

The process of long term adaptation in women with Hereditary Breast and Ovarian Cancer (HBOC) (Hamilton *et al.*, 2009) was studied in the USA. Their study showed that over a period of four years, the women interviewed (n=7) reported a transition from dealing with the immediate impact of their diagnosis to an adjustment to the consequences of knowing about their increased risk. The women appeared to learn to live with their choices. Initially they were unsure of the efficacy of their lifestyle choices on their risk. However, four years later they were making lifestyle changes to their diet and physical activity with greater confidence and determination. Another process the researchers observed was how the diagnosis had influenced family relationships. At the first interviews some women were still informing relatives about their potential risk. Four years later they had perceived how the diagnosis could negatively impact relationships in the family and were working to protect their relationships with those relatives (Hamilton *et al.*, 2009).

This research, I think, illustrates the necessity of allowing time for patients to process and adjust to their new knowledge. It also shows how the long term outcomes of

testing are not always clear at the outset. Sometimes the adjustment may be quite intangible but nevertheless is an important part of coming to terms with a new familial diagnosis. Skirton (Skirton, 2001) found in her longitudinal study of families who received genetic counselling for Huntington disease that commonly client's spoke of gaining "peace of mind" p.324 (Skirton, 2001) although they had not reported any specific changes in their plans or relationships following genetic counselling.

9.5.3 Engagement with the diagnosis

Behaviours described by participants during the Phase 2 interviews were interpreted as demonstrating how engaged participants were with communicating with their family about their diagnosis. These associations indicated to me that participants needed to understand the diagnosis and its implications for themselves before they could extend that to explaining the implications to their family members. A theory of engagement in relation to a genetic risk in LS has been described by McAllister (McAllister, 2002; McAllister, 2003; McAllister *et al.*, 2007). She defined it as "the degree of cognitive and emotional involvement with one's increased risk of developing cancer as a result of one's family history of cancer" (McAllister, 2003, p.180) and therefore related it to the individual only.

The qualitative interview data that McAllister used to develop the theory of engagement was longitudinal with participants interviewed before, and six months after, their pre-symptomatic genetic tests. McAllister described how those patients who were 'intensely engaged' (cognitively and emotionally) prior to their test

appeared to cope better with learning they had inherited the pathogenic gene variant than those patients who were only 'partially engaged' (cognitively) prior to testing. She postulated that the intensely engaged patients had 'rehearsed' their mutation status and worked through some of the implications before their test. Using this theory of engagement I would interpret that at least one participant (Oliver, #1) had cognitive (partial) engagement prior to his test but this meant that he experienced more anxiety about his risk of cancer post test. Therefore I would agree, based on my own experience and interview data, that the concept of engagement is relevant to how patients adjust to their test result.

What then is the consequence of engagement on the process of sharing information with relatives? Within her theory, McAllister puts forward the idea that the benefits of screening for cancer ameliorate people's reactions to their predictive test result, at least in LS where screening and treatment is available (McAllister, 2002). This would tie in with my observation that participants wanted to focus on and communicate the potential benefits of knowing genetic status.

The action of attempting to inform relatives can be met with denial, disagreement or anger (Chivers Seymour *et al.*, 2010; Koehly *et al.*, 2009) and this can challenge someone's engagement and test their coping strategies. So how individuals act on their knowledge of their genetic risk will be influenced by the reactions they have from their family members; their actions are not happening in a vacuum. There is evidence that when a patient first attempts to inform their relative about the familial diagnosis, the response they get from that family member may influence subsequent attempts to tell other relatives. If they are received positively and listened to this will encourage and reinforce the action but if they are rebuffed, or their communication is

met with a negative response, this can inhibit further attempts to share information (Lafreniere *et al.*, 2013). The relevance of this to the provision of information via a secure online portal is that the provider is distanced both in time and place from the recipient. This might provide some protection from negative reactions for the informant and also give the recipient the opportunity to view and return to the information at a later date. Providing information via a website could help individuals in both their 'decision making' and the 'disclosure' phases (Figure 9.4).

Figure 9.4 image has been removed due to copyright restrictions

Figure 9.4 Framework for understanding and guiding the process of communicating genetic test results to family members from Lafreniere et al 2013

My findings also showed that the initial disclosure to a relative about the familial diagnosis tended to occur in a step-wise fashion. The majority of the interview participants, when asked, did qualify that the initial 'disclosure' they made to their

relatives were either face to face or over the telephone. Subsequent communication might have utilised email, letters or social media in order to share more in depth information or provide updates regarding screening or diagnoses.

9.5.4 Importance of timing when attempting to share information with relatives

As described earlier, when an individual is first diagnosed with a genetic condition they are likely to experience a variety of emotional sequelae. Some of which may relate to a sense of loss and be characterised by evidence of mourning (McAllister *et al.*, 2007; Sobel & Cowan, 2003). How patients experience their adjustment to their diagnosis will probably influence the way they attempt to pass on information about it. This was reflected in the data from the surveys and interviews where some participants described feelings of shock and disbelief when they first learnt of their diagnosis. The impact of the diagnosis is discussed in more detail above (Section 9.2) but what also emerged from the qualitative data in Phases 1 and 2 were participants efforts to find out more information about the health implications of the diagnosis for them personally. It is only after these first reactions had subsided that participants appeared to attempt to communicate with relatives outside their immediate family about what the diagnosis might mean for them. These findings have led me to conclude that providing follow-up, repeated contact, or better ways that patients can communicate with health professionals, are all likely to be as important as support for sharing information. Put another way, it is necessary for health professionals to be considerate to patients regarding the *timing* of their encouragement to inform the wider family about the diagnosis which will depend very much on the individual.

This approach is also supported by the findings of earlier research; “Health first, genetics second” by Forrest and colleagues (Forrest *et al.*, 2008) and would be consistent with Lehti’s theory of psychological adaptive modes (Lehti, 2016). Forrest’s research investigated families’ experiences around communicating genetic information. The themes that emerged from their qualitative analysis of 12 interviews described a process of emotions, information seeking and communication. Interviewees talked about their initial shock immediately following the diagnosis which was followed soon after by contacting close family to seek emotional support. The interviewees’ main concern then centred on the health implications of the diagnosis and the needs of their child (or themselves). Subsequently couples looked for more information to help them understand the diagnosis but communication with family members continued, reaching the wider family sometime later, to inform them about the inheritance and what that implied. [This process is represented by a diagram (Appendix 29)] Their findings (across several different genetic conditions) together with my own, indicate that it would be valuable to have a resource which could be accessed whenever that person felt they wished to view it. Such a facility would also provide flexibility to suit different individuals who were adapting to their diagnoses at different rates.

9.6 Prior research into interventions to facilitate family communication

Interventions to improve family communication and disclosure about a genetic diagnosis have been set up and tested in Australia, the Netherlands, Portugal, the UK, and the USA (Bodurtha *et al.*, 2014; de Geus *et al.*, 2014b; de Geus *et al.*, 2016; Eisler *et al.*, 2016; Hodgson *et al.*, 2016; Hodgson *et al.*, 2014; Kardashian *et al.*, 2012; Katapodi *et al.*, 2018; Mendes *et al.*, 2010; Mendes *et al.*, 2013; Montgomery *et al.*, 2013; Smith,

2007). What is common to these interventions is the perspective that patients may benefit from guidance in how to approach and explain to their relatives about the diagnosis (Appendix 37). Patients may also benefit from more tailored information which was specifically written for them (Kardashian *et al.*, 2012)

The effectiveness of such interventions can be difficult to quantify. Outcomes have included patients reporting which relatives they had informed (Montgomery *et al.*, 2013) and assessing what proportion of relatives have been referred to the relevant genetics services (Hodgson *et al.*, 2016). What constitutes an effective intervention would depend on the objectives of the research. No significant differences were found in rates of communication with relatives about HBOC (the stated objective) between cases and controls but sharing test results were associated with higher perceived control (in conveying information) and subjective norm variables across all subjects in one study (Montgomery *et al.*, 2013). These authors interpreted their findings through the lens of the Theory of Planned Behaviour (Ajzen, 2002) which can be applied to the action of sharing health information.

It is also important to understand why some relatives are receptive to this information while others are not. The constructs of the Health Belief Model (Rosenstock, Strecher & Becker, 1988) could be applied to predict the behaviour of individuals in relation to their own health by assessing the perceived threat, benefits, barriers and their self-efficacy and whether these are in conflict. I would argue that the key issue is whether relatives are *aware* of the condition and its implications for their own health. It is then their choice whether they seek more advice through a specialist, such as provided by a clinical genetics service, or access genetic testing. As stated earlier, people's perception of the level of threat may be dependent on their understanding of it and

their self-efficacy. It has been difficult to obtain data about people who may be aware of their risk if they themselves do not wish to engage with health services (Cowley, 2016) and this remains a challenging area in which to establish empirical evidence. However, capturing data on relatives who have not sought the advice of health services would be an area where a website like Family Web could provide a useful tool to research. This is because the website encourages patients to share documents with their relatives. When this has happened it would give an opportunity to create an anonymous log of activity, such as document views or downloads by relatives. This would then indicate not only how many relatives viewed documents per index case but it would also show which documents were most popular or most frequently downloaded. Overall views and basic website activity within the period of this study are presented in Appendix 38.

It should be noted that the type of genetic condition does appear to have a bearing on the frequency of communication within families. Kardashian (Kardashian *et al.*, 2012) observed in their small pilot study of providing tailored information and educational material to women with BRCA pathogenic variants (n=19) that very little difference was seen between the intervention group and the control group regarding the number of relatives seeking genetic testing. Although the sample size was small, they interpreted this as indicating how motivated these women were already to inform their relatives, prior to receiving the intervention. Kardashian noted that the participants who scored higher on a knowledge of family history test also reported a greater frequency of sharing information about the BRCA gene variant with their relatives. Given the small sample size, tests of statistical significance were not informative. However, Kardashian's findings could be an indication that better

knowledge of family history might translate into greater confidence and motivation in sharing information. Motivational and inhibiting factors were conceptualised in a mind map (Appendix 36) during the process of analysis of the Phase 2 interviews. Many different factors were identified that appeared to interrelate and could potentially influence someone's ability to share information with their relatives.

Building on the evidence accrued through these intervention studies which were intended to promote family communication (Appendix 37) it would be logical to design future research that investigated strategies to facilitate communication which could be applied across different genetic conditions. The Family Web website could be developed to research information sharing in many diverse conditions by extending the content to serve different patient groups.

9.7 The influence of gender

The issue of gender may be relevant. Typically women are reported to be better communicators with their relatives and it has been shown that women are most often the ones to share health information about a diagnosis in the family (Bartuma, Nilbert & Carlsson, 2012; Keenan *et al.*, 2005). Conversely, men might be less likely to communicate with their relatives about a health issue. This was inferred from a study which demonstrated that the children of mutation positive women were three times more likely to seek testing than the children of mutation positive fathers (Aktan-Collan *et al.*, 2011). The most effective communication in families appears to happen when there is a co-ordinator, usually a mother, sister, or female spouse who also provides emotional support within the family (Koehly *et al.*, 2003).

In the survey data few significant gender differences were found. One significant difference was that female respondents were more often interested in information about talking to children. Another gender difference was regarding the type of response, with a greater proportion of women responding online to the survey than men. However, the order of ranked preferences to different methods of receiving information (follow-up appointment, email, website, leaflet, phone call) were the same for both sexes.

I had anticipated that providing information in digital format might be regarded as of particular benefit to younger people and to men who had to initiate communication. This was because other studies have found that men were more likely to use email to correspond with their doctor than women (Newhouse et al., 2015) and digital communication is often preferred by younger people (Duggan, 2015). Therefore a purposive sampling technique with maximum variance was used to ensure that equal numbers of men and women (across several age groups) were interviewed during the course of the study in order to gather data on a broad range of experiences and attitudes.

The Think-Aloud interviews were focused on the acceptability and feasibility of the website but within them most volunteers took the opportunity to talk about their experiences relating to the diagnosis. I had anticipated that men might be more willing or interested in the website as a method for sharing information. What I heard during the interviews was that, irrespective of gender, the participants were enthusiastic about a new and additional way of receiving and sharing information. They nearly all appeared very keen to access more information about their condition. Based on my results I do not have evidence to support gender as a major contributing

factor in why information about familial bowel cancer is not always shared and it was beyond the scope of this study to specifically address this in more detail.

9.8 Prior knowledge of the genetic condition

Differences in participant knowledge of disease at outset was considered to be a factor that explained the disparity in response to the intervention trialled in Australia (Hodgson *et al.*, 2016). The intervention involved providing non-directive telephone follow-up calls to 95 newly diagnosed patients. The proportion of relatives contacting the genetics service in the state of Victoria, Australia was the measured outcome in this randomized controlled trial. Amongst the different case vs control groups, the largest difference in response was in the rarer conditions where 39% cases versus 10% controls at risk relatives sought advice. The investigators (Hodgson *et al.*, 2016) speculated that if patients were diagnosed with a rarer condition, providing a supportive telephone call could be particularly helpful to patients and give them greater confidence to share information about their diagnosis. This implies that previous knowledge of a genetic condition does influence patients' capacity to discuss it with their family members. This concurs with my findings where an important factor reported by several interview participants (George, Robin, Sandra, Dave and Fiona) was the lack of knowledge or health literacy relating to familial bowel cancer amongst members of the public. This made some of them less confident in explaining the implications of the diagnosis to their relatives. They also described how unsupported they felt, particularly by their GPs, who they perceived as knowing very little about their condition.

A recent UK collaboration (Eisler *et al.*, 2016) investigating ways of providing support in multi-family discussion groups (MFDGs) were clear that there was an advantage to

bringing together families affected by different genetic conditions. They found that this removed the focus from the condition itself and allowed participants to consider instead the impact of the diagnosis on family functioning and how to cope better with the diagnosis. What all the families had in common was the challenges of dealing with the immediate risks of the condition in affected individuals, plus what the implications of the diagnosis were for the whole family. A very common concern amongst parents was the decision of when and how to tell their children about the diagnosis. This concurred with my study findings as talking to children was also a frequently cited topic of interest. This resulted in a section on the website about 'Talking to children' with tips and links to other websites, including a recommendation to a leaflet that explained how keeping secrets within the family in order to protect children from fear and anxiety has been shown to be disruptive to family life (Metcalf *et al.*, 2008).

9.9 Ethical considerations

Without making attempts to contact patients it is difficult to know what their experiences are in the months and years following their diagnosis. The conclusions I have drawn were that a subset of my study participants felt unsupported and anxious about how their ongoing care was being managed. One theme that emerged was participants having to be self-reliant; to take responsibility for their own care and needing to "push" for screening. This appeared to be the consequence of feeling unsupported and isolated following their diagnosis.

Previous research has sought to identify risk factors for which patients are most likely to suffer serious psychological sequelae following a genetic test (Aktan-Collan *et al.*, 2013; Burton-Chase, Gritz & Peterson, 2013). Within current clinical practice there

appears to be insufficient emphasis given to post-test follow-up or long term support and consequently patients are being offered testing but they are not necessarily receiving any further contact with their genetic counsellor or geneticist after receiving their test result. The high proportion of families referred to family therapy that have a genetic diagnosis (Alison Metcalfe personal communication August 2017) is indicative of the profound and sometimes destabilising effect that a genetic diagnosis can have.

Earlier work by Skirton and colleagues (Skirton *et al.*, 2013) following a consensus meeting of health professionals from across Europe, developed guidelines for pre-symptomatic genetic testing. In these guidelines it was proposed that HPs counselling patients prior to pre-symptomatic testing need to explore the relevance of the result for other relatives. They should assist their patients to make plans regarding disclosure to other family members; family communication being a core component of a patient's personal management decisions relating to such testing. Subsequently, a systematic review of empirical research concluded that while patients have a responsibility to inform their relatives about the implications of a genetic diagnosis in the family, it is also the health professional's duty to assist their patients in the process of sharing information (Dheensa *et al.*, 2015). I would argue that concomitant in that duty to assist is the ethical principle of non-maleficence (Beauchamp & Childress, 2001).

9.10 Strengths and limitations of this research

In order to properly consider the impact of the findings from this study it is first necessary look at what might have either strengthened or limited the research, in terms of both intrinsic and extrinsic aspects.

9.10.1 Recruitment through Clinical Services

In undertaking a proof of principle study for an innovation suitable for use in the NHS I thought it was important to recruit through NHS clinical services in order to demonstrate that the innovation was suitable in that context. Typically patient satisfaction surveys show high levels of satisfaction with clinical genetics services (Nordin *et al.*, 2002). This may be related to a ceiling effect in such surveys where nuanced responses are lost in the overall positive feedback (Andrew *et al.*, 2011). Participants in this study said that they would have liked more support and specifically more information. Thus providing some guidance about how services could be improved in the future.

At my lead recruitment site in Plymouth I was supported by the surgical (colorectal) and endoscopy services. At all other sites, recruitment was through the clinical genetics services (Peninsula Service in Exeter, All Wales Service in Cardiff, West Midlands Service in Birmingham, North West Thames Service and South East Thames Service in London). In order to make minimal demands on health professionals' clinical time the process of identifying, approaching and recruiting eligible patients was designed to be as simple as possible. I had discussed the process with colleagues in these services at preliminary meetings in Plymouth (Appendix 30) and Exeter in order to design a study that was viable in a busy clinical environment. I had envisaged that health professionals (whether genetic counsellors, clinical geneticists, endoscopists or surgeons) would approach their patients in clinic to give them information leaflets about the study.

What happened at each phase of the study was that designated research staff identified eligible patients, checked with the clinical staff and then sent letters with

information sheets to those patients. This did protect clinicians from any burden of this work but meant that health professionals did not engage with the website in the way that was important to test the function with volunteer users. Consequently, there was greater consistency during the website development phase as all the Think-Aloud interviewees had the website function demonstrated in the same way and received the same generic documents during the GoToMeeting video call. Nevertheless, health professionals' participation as providers of documents for upload was not demonstrated so that is something I would wish to address in future research.

In the final phase of the study I had originally only intended to recruit through the five sites with clinical genetics services because I wanted clinicians to upload appropriate documents for any participating patients. Unfortunately the rate at which eligible patients were being recruited and taking part in the survey through these clinical sites was slower than I had anticipated. Consequently, I applied for and was granted a substantial amendment to my NHS ethical approval which enabled me to extend recruitment beyond the original end date of June 2017 to 30th September 2017. In an earlier non-substantial amendment I had already modified my recruitment strategy to allow recruitment through online advertising. This enabled me to recruit survey respondents who had volunteered to be interviewed and to include patients who had been informed of their diagnosis within the last two years (rather than one year).

9.10.2 Recruitment across England and Wales

The survey was designed to capture data on the issue of family communication in a broad group of patients who had all been recommended to have regular colonoscopy on the grounds of family history or genetic vulnerability. Having eligibility criteria that encompassed a large number of patients was intended to make recruitment easier but

it was outside the scope of this doctoral study to sample a statistically representative number of patients from this large population of patients.

Postcodes were collected in the survey so that information on the number of patients who participated could be fed back to each of the recruitment sites. Once the recruitment sites were active it was difficult to distinguish if participants submitting online responses to the survey had initially been informed about the study through their genetics service or via an online link. Consequently, only online responses received before the NHS recruitment sites were active could be logged as non-NHS responses. In addition, I was only aware of the number of paper questionnaires that I had sent to each recruitment site and not the actual number of patients who had been approached by NHS staff at the recruitment sites, this meant data on response rates were not calculated.

9.10.3 Investigation through families at risk of bowel cancer

In order to create and test the website as a tool for sharing documents I decided to focus on families at increased risk of bowel cancer. I selected an increased risk of bowel cancer as it was a condition where there were potential benefits to relatives who learnt of their risk, since they could then access bowel surveillance and make changes in their lifestyle to reduce their risk. I perceived there would be more motivation for patients to inform their relatives than in some incurable conditions. I acknowledge that by restricting the eligible patients to those with a vulnerability to bowel cancer the findings from the study may be limited. This may lessen the impact of the research even though the function of the website could be applied to any condition where the diagnosis in one individual has health implications for their relatives.

For this reason I intend to look at alternative approaches in my future research. I want to investigate the efficacy of the website as a way of informing relatives about the genetic condition in their family. I could extend the research by seeking to trial the document sharing function with families with a range of different conditions, similar to the approach by Hodgson and colleagues in their family communication intervention (Hodgson *et al.*, 2016).

Another approach would be to incorporate the website function in applied research investigating the pathway for consenting and support to newly diagnosed patients with bowel cancer who are offered tumour analysis to screen for LS. This applied research would be focussing on a more defined population of interest, restricting the research to LS families, but using the website in the broader context of informing individuals who are undergoing genetic testing or seeking more information about the cancer diagnoses in their family. Hampel (Hampel, 2016) described the potential utility of using such a tool in her discussion of methods of cascade testing in LS, referring specifically to the California based website: Kintalk.org. Given the differences in health care provision between the USA and the UK I think it would still be necessary to investigate an approach developed specifically for use within the NHS.

9.10.4 Gender bias

Another reason for focussing on a vulnerability to bowel cancer was that this cancer affects both men and women. Previous research in family communication regarding genetic diagnoses has shown that men encounter more difficulties communicating about such health issues than women (Forrest Keenan *et al.*, 2009; Metcalfe *et al.*, 2008). In the survey more responses were received from women than men (77% vs

23%). This was something I had anticipated as gender bias in response rates is a common issue in survey participation (Sax *et al.*, 2008). In order to mitigate this I used a purposive sampling technique with maximum variance, interviewing an equal number of men and women across the two later phases of the study.

It was interesting to me that four of the eligible female volunteers decided not to take part in a Think-Aloud interview once I had discussed with them what the interview would involve. Only one man declined to be interviewed after a similar conversation. With such a small number of participants it is impossible to draw any conclusions from this and I recognise that the volunteers were a self-selecting sample.

9.10.5 A 'Hawthorne Effect'

In common with other research I acknowledge that the way questions were framed (Appendix 21 interview guide) could have precipitated a more positive response regarding the concept of a secure website and therefore my data may not be a true reflection of participants' views. Whether this potential distortion of response should be described as a 'Hawthorne effect' is debateable, as the term is usually applied in quantitative research (McCarney *et al.*, 2007). The Hawthorne Effect has been used to describe the modification in behaviour by research participants in response to their knowledge and the psychological stimulus of being involved in a research study. This term was originally applied to increased worker productivity under trial conditions in 1928 (Adair, 1984; McCarney *et al.*, 2007).

A strength of qualitative research is that it investigates the personal and subjective views of participants. I acknowledge that their responses were likely to be influenced by our interaction at that time and that influence would have been difficult to

eliminate. As part of that interaction I do recognise that participants may have been more likely to endorse my idea about using a secure website to share information (and less likely to challenge me) because I had suggested it

9.10.6 Strengths and limitations of the survey design

How individuals defined their family was not explored in the questionnaire but evidence suggests that commonly patients consider 'family' to be their nuclear family of first degree relatives (Chivers-Seymour *et al.*, 2009) but can extend to include friends (Koehly *et al.*, 2003). Although 'family' was a key concept in this research, it was potentially ambiguous, as the term 'family' was not defined but left to each individual's subjective interpretation. This meant there would be variation in what participants considered to be their family which could introduce inconsistency into the data. However, this issue would mainly be confined to a few questions, such as in questions 4, 12 and 15. For example, in question 12 participants were asked if "all" or "most" of their relatives had been informed. This was intentionally vague as I was interested in whether participants considered that the process of informing relatives had been completed or not. Since the implications of a genetic vulnerability can extend into the wider family the potential reach of information about a genetic diagnosis would be a valuable area for future research.

The survey was initially tested with volunteer patient advisers to check the wording of questions and validate the consistency between the online and paper copy survey.

The order of question choices was fixed within the survey and this could have influenced the responses for some questions since there is evidence that participants more frequently tick the first available option where multiple options are given (Stern, Dillman & Smyth, 2007).

As none of the survey questions were obligatory some questions were left blank by participants and some indicated that they had received more than one type of information at the time of their diagnosis. These factors may be confounding the calculations, but it is still evident that the majority of people, who received their diagnosis and information from a genetics professional, did at least receive general information about the condition and only a minority received no information at all.

9.11 Summary

In summary, in this chapter I have looked again at what was revealed regarding participants experiences of receiving and sharing information about the inherited vulnerability to cancer in their family. I have discussed how a website could facilitate communication in families and whether this is likely to be helpful given some of the barriers to communication that exist. I have suggested that information should not only be available in a digital format but it should be clear, accurate and trustworthy. Information that conveys a positive benefit to knowing genetic status is likely to help patients when they attempt to share information with their relatives. This is particularly the case if the content can provide some hope for the future. In addition, the timing of support may be important since patients need to adjust to their diagnosis before feeling able to share information about the implications of it with their relatives.

I would consider that I have met my aims and objectives in this study. I did establish that a secure website, such as Family Web could '*support families with an increased risk of bowel cancer to share information with their relatives*' as stated in my aim. Without

demonstrating that the website was directly instrumental in promoting communication in families, the study did establish the proof of principle that such information exchange was possible using this type of tool. It would be appropriate to conduct further research to determine if this would facilitate family communication for families with an increased risk of bowel cancer.

I considered it fundamental to the application of the website for future users that I first needed to gather data on patient experiences. So, in order to meet my objectives to investigate: 1). *The perspectives of patients, their experiences of how they received information about the familial diagnosis themselves.* 2). *Invite patient's suggestions for improvement in the way they were told about the familial diagnosis,* and 3). *Investigate patient's preferences for information topics and also how they would like to receive information, including whether these varied by age or gender,* I conducted a survey and telephone interviews.

In addition, I explored patients' views on the idea of a secure website through the Phase 2 interviews and also in the Think-Aloud interviews in Phase 3, thereby meeting my fifth objective. Finally, I tested '*website function and acceptability*' and '*investigated the feasibility of sharing documents securely*' via the website in accordance with my sixth and seventh objectives. In my last chapter I will draw conclusions from what I have learnt through this research. I will consider the implications of this research for clinical practice, for future research and for policy makers.

Chapter Ten

Implications of this research and conclusion

10.1 Introduction

In this final chapter I will consider the impact of this research for clinicians, researchers and policy makers and on my ongoing collaboration with other researchers to investigate how to facilitate communication in families affected by genetic disease. I will describe the opportunities for future research particularly focussing on the integration of the secure website into further studies.

10.2 Reflection and assessment of the strengths and limitations of my approach

10.2.1 Positioning as an NHS health professional and genetic counsellor

I was aware that as a researcher I could not disassociate myself from my identity as a genetic counsellor, a woman and someone who usually seeks information in new situations. My personality and experiences have inevitably influenced my views and have acted as a filter and lens through which I have interpreted the data. The themes that I identified in the interviews and survey were what I considered important to the issue of communicating genetic diagnoses in families from a pragmatic stance and from my perspective as a health professional with experience working in this field.

Throughout the study I have been open about my background as a genetic counsellor. I realise that by positioning myself in this way it was likely to have influenced participants' responses (Mays & Pope, 1995; Morrow, 2005). Participants might have regarded me as representing the NHS genetics service and this may have influenced

what they expressed about their experience of genetics services. However, I think that by positioning myself in this way I was being genuine and congruent, describing my motivation to conduct the research as derived from the insights I had gained through my clinical practice. I also recognise that by identifying myself as an NHS health professional this may have conferred a degree of trust towards me by participants. Conversely, the opposite could be argued that those patients who had negative experiences of their encounters with health professionals could be deterred from taking part in the study or some might take part in order to voice their frustrations.

10.2.2 Qualitative interviews

In this research I was conducting telephone interviews and Think Aloud interviews for the first time. In the PIS and on the questionnaire I had identified myself as a PhD student and Registered Genetic Counsellor as I wanted to indicate to participants that I had professional credibility to research their views. With my clinical experience I was confident that I could develop a rapport with the participants and draw them out to talk about their subjective experiences and views. I considered it important to establish such a rapport in order that participants would feel comfortable discussing issues that might have been distressing to them.

As a novice researcher I think my interviewing style developed over the course of the interviews and I became more confident keeping the participant on the topic. My confidence was helped by checking my interview topic guides and lists of planned questions (Appendices 21 and 25) but mainly it developed with experience and reflection. In retrospect I think that my style of questioning may have been too informal in some interviews and this is likely to have further impacted on whether

participants felt free to express themselves candidly. I also suspect that participants might not have wished to offend me during the interviews and therefore they may have expressed more enthusiasm for the idea of the website than would otherwise have been the case.

Talking to participants during interview gave me pleasure as I enjoyed the time of engaging with them and learning about them and their families. It also felt to me that the interviews gave an opportunity for most participants to tell their story and maybe get some therapeutic benefit from the process of explaining what they experienced to an interested third party. I realised that participants were self-selecting; I perceived that some had a message they wished to convey, for example 'Fiona' who had learnt about the diagnosis by letter initially and found this a profoundly shocking experience.

I had not anticipated my own feelings of sadness when I said "goodbye" at the end of each interview. It felt awkward to me saying goodbye as I had no reason to talk with them again, unlike in my role as a genetic counsellor when I had an ongoing responsibility to my patients. The contrast between my role as a genetic counsellor and my role as a researcher was evident to me during a few of the interviews. This was most obvious when participants made comments, which showed an inaccurate understanding of something about their condition, and I had to stop myself from correcting them.

10.2.3 Recruitment process and collaboration

I found that when talking to people prior to consenting them for the Think Aloud interviews that some of the volunteers did not meet the eligibility criteria. This meant that it was disappointing for them, and for me, having to explain that they could not

take part. It reminded me that I was very dependent on the research staff and clinicians interpreting my criteria correctly and I would need to make such criteria very clear in the future. On other occasions I have realised during an interview that because the participant had known about the diagnosis in their family for so long they were unable to remember much about what information they had first received. This initially has made me feel frustrated that my eligibility criteria were not more specific and limited to people who had only recently learnt about the condition in their family. I then realised that their inability to remember what information they had been given further demonstrated the need for more enduring sources of information than printed leaflets.

Although I already considered the research to be worthwhile, it was very affirming when participants endorsed what I was doing. They told me how important they regarded the issue of improving support to families affected by an increased risk of bowel cancer. What was harder to hear were the criticisms of how some genetic services had appeared to abandon or restrict support to some people. I realised that these comments had been provoked by me asking what participants thought could be improved. Nonetheless some accounts that I heard were very sobering; what I heard made me feel angry and upset that patients were suffering. I also felt naive that I had not been more perceptive in my clinical role and contacted my patients more proactively as follow-up to their appointments. Some survey participants did praise the service they had received (in their free text responses) but the majority provided comments about what could be improved and were quite critical.

At times, reading the comments and reflecting on the interview transcripts, I have felt inadequate to the pain being expressed. In addition, I have felt powerless in my

attempts to improve clinical services supporting patients in this position. However I have acknowledged that by conducting this research I am contributing to a body of knowledge that will over time potentially lead to changes.

I have been encouraged by meeting other researchers and clinicians who have similar interests in the subject of family communication or the needs of individuals living with a genetic vulnerability to bowel cancer. I realise that my perspective has matured over the course of this research; having commenced in the somewhat isolated role of a PhD student, the process of conducting a multi-site study has reinforced to me how vital it is to develop good working relationships with collaborators. Looking forward, I remain hopeful that this PhD study will feed into larger studies and together provide evidence to inform how clinical services can be changed for the better.

10.3 Implications for clinical care

10.3.1 Web-based information

The potential health benefits concomitant with improved provision of information and support using these methods should be of interest to health service managers and policy makers as well as clinicians. As stated before, if patients feel able to pass on information about their diagnosis to their relatives this has the potential to save lives. There are many reasons why information may not be shared within a family in a timely manner. In my view it is fundamental to this process that patients can access clear, simple, accurate information about their diagnosis, so that they themselves can be confident that they understand their own diagnosis. This then gives them the tools to pass on reliable information to their family.

Providing open access information via a website can augment traditional methods of providing information verbally and in paper format. The advantage of web-based information is that it can be easily augmented and updated by health professionals. Having documents available via a secure website doesn't restrict them to being viewed online as such documents can be printed out to share with family members who may not use this technology. Most significantly, providing web-based health information means that it is more readily available to young adults who might not otherwise retain paper copies of leaflets or letters. I remain concerned that younger relatives at risk of cancer may not have access to evidence of the diagnosis in their family. This then may result in greater difficulty for them being referred for the recommended surveillance. There is also the risk that they might not receive adequate support from their primary care team if their degree of risk is not appreciated by their GP (Weathers, 2014).

10.3.2 Topics where patients wanted more information

I was aware from my own clinical practice and from recent evidence (Dheensa, Lucassen & Fenwick, 2017) that the information provided to patients at diagnosis was often limited to generalised information about the condition and a letter to pass on to their relatives informing them of the new diagnosis in the family. Therefore, I included questions in the survey questionnaire, and within the telephone interviews, to enquire about what information participants received at the time of their diagnosis and what further information they thought would have been helpful.

The results from the survey are detailed in Chapter Six, section 6.9. Different topics which related to the practical implications of a diagnosis of a high lifetime risk of bowel cancer were most frequently cited. Of particular note was the issue of a 'healthy

lifestyle' which was mentioned by many participants in each phase of the study. These data therefore have major implications for clinical practice; health professionals may not realise that their patients are unclear about whether they can reduce their risk through lifestyle changes. Recommendations to maintain a healthy weight, limit alcohol intake, eat plenty of fruit and vegetables but eat less processed meat and stop smoking, are all messages that are widely available to the public. The relevance of these messages to families with a genetic diagnosis needs to be emphasised in clinical encounters in my view.

10.3.3 Testimonies or stories to encourage other patients

Videos are used in many fields to engage users and promote understanding. The support group Lynch Syndrome UK use a series of videos to help explain the diagnosis to younger family members . These were incorporated into the Family Web website in response to the survey data that showed there was substantial interest in the issue of talking to children. One suggestion that was made in the telephone interviews (but not yet acted on) was that the website could incorporate video testimonies from patients. This was perceived as both reassuring and informative for newly diagnosed patients (Kirk *et al.*, 2013). Another advantage to providing open access video resources via a website is that a visual explanation with British Sign language signing could make it more accessible to hearing impaired patients (Middleton *et al.*, 2010) and the audible facility could help visually impaired patients.

10.3.4 Changing the balance of clinical time

Drawing on my findings with regard to the importance of adaptation to a diagnosis, I suggest that it is likely to benefit patients in the future if health professionals could give individuals more time and support to help them adjust to their diagnosis.

Critically, this support should be provided before encouraging patients to discuss the health implications of the diagnosis with their relatives.

Currently clinical encounters within clinical genetics are generally organised and funded in relation to date of referral. This means that the systems in place to book clinic appointments are focussed on the needs of the new patient. The flexibility to arrange follow-up contact appears to have been eroded due to increasing pressure to see more new patients. Pre-symptomatic testing protocols were developed in relation to the needs of individuals at risk of Huntington disease (Harper, Lim & Craufurd, 2000) but have been extended to a range of conditions (Skirton *et al.*, 2013). These put emphasis on how patients prepare for having a pre-symptomatic test and whether it is the appropriate time for them to have such a test. While these considerations are still important to individuals at risk of cancer in my experience many patients come for genetic counselling already clear that they want to be tested. However, they may not be able to perceive what the full impact of that testing will be until they have had their result. Consequently I would advocate a shift in how health professionals' time is allocated in order to make provision for follow-up contact with patients and provide better support to families in the medium to long term.

10.3.5 Optimal timing of encouragement to disclose

If adaptation to their diagnosis is likely to influence how and when people share information with their relatives then health professionals need to be mindful of this. I suggest that this has implications for the health professionals and at what point they suggest to their patients that the diagnosis should be shared with family members. It may remain the most practical solution that information about the condition and letters for relatives are provided post-test result. Where possible I would recommend

that later contact by telephone or letter would be a more appropriate time to discuss how their patients plan to pass on information to their relatives. Each individual will have unique circumstances and support needs to be relevant to them if health services wish to optimise the benefit and minimise the harm of a genetic diagnosis.

10.4 Opportunities for future research

10.4.1 Planned future projects

Providing adequate, appropriately worded and specific information to patients following their diagnosis with a life threatening condition is a reasonable intention at any time. Given the growing and widespread use of digital technology, the Family Web website has demonstrated that this is a feasible way of providing such information in digital format for flexible access at any time. The Think Aloud interviews tested the website's acceptability with volunteer patients but how much this innovation could impact on information sharing in families has yet to be investigated. Therefore I am developing grant proposals intending to extend this research to look at the efficacy of this approach as a way of disseminating information about a diagnosis in families. This is being developed in collaboration with academics in the UK and in Europe as part of randomized controlled trials.

10.4.2 Utilising opportunities provided by Personal Health Record systems

The opportunity to provide accurate and trustworthy information in a digital format to patients would be supported by a secure website like Family Web but this could also be done using pre-existing Personal Health Record (PHR) systems such as 'My Health Record' (Dheensa, Lucassen & Fenwick, 2017) or 'Patients Know Best'. I understand that research is ongoing at a university hospital in the UK to investigate the utility of

providing information to patients enrolled in a gene-sequencing project via their existing PHR system. Within the South West I am only aware of one NHS trust which currently uses a PHR system but previous discussions with clinicians and managers at that trust were very positive regarding the use of the system for genetics patients. I perceive that there would be further opportunities to explore whether the findings from this study could apply to a wider group of patients.

It would be feasible as part of service development to create an extension to routine clinical care within existing clinical genetics services to upload clinical letters, test results and pedigrees for patients on to a PHR system. This could be followed with patient satisfaction surveys or more in depth investigations through interviews with patients to determine how beneficial patients found it to use the system and whether health professionals experienced any problems using it. Evidence regarding PHR use has shown that such systems are particularly helpful to patients with complex or chronic care needs, such as patients with cystic fibrosis, but they require the engagement and support of health professionals (Archer *et al.*, 2011; Nazi, 2013).

10.4.3 Proposed project to use the website in support of bowel cancer patients

As a further application of the website, I have been developing ideas and considering another grant proposal with other researchers in the UK to set up a study to improve support, information and informed consent for patients with colorectal cancer whose tumours are being tested for microsatellite instability (MSI). This is following the NICE guidance (NICE, 2017) which recommended that all bowel tumours are tested for microsatellite instability (MSI). As a member of this project group I want to investigate novel ways of supporting patients newly diagnosed with Lynch syndrome which would be an improvement on current clinical care. A recent Freedom of Information request

by Bowel Cancer UK in 2018 revealed that only a minority of bowel cancer patients are having their tumours tested for MSI in the UK (Bowel Cancer UK, 2018). One aspect of this problem appears to be a shortage of appropriately trained staff who are knowledgeable about Lynch syndrome and can therefore provide information, take informed consent prior to tumour testing, counsel and consent prior to germline testing and give results to these patients.

In this proposed research project we intend to integrate the provision of information to patients in digital formats via the Family Web website. The website could be incorporated into a care pathway and provide the function of helping patients understand their diagnosis and disseminate information about the diagnosis within their family. In the longer term such a resource could serve health professions outside genetics by improving awareness and could provide opportunities for education within surgical and oncology services in accordance with Department of Health goals to 'mainstream' genetics (Bennett *et al.*, 2010; Davies, 2017; Kirk, Tonkin & Burke, 2008).

10.4.4 Capturing anonymous data on information views

The function of the Family Web website could be used as a tool in future research. This is because the website will automatically collect anonymised data on usage via web analytics, logging which parts of the website are visited and how frequently. It is challenging to gather data regarding receipt of information when family communication is being investigated. I could envisage a project where there is an option to access information via the website and therefore how many times certain information resources were accessed, downloaded or shared could be captured by such a site.

10.4.5 Opportunity to interview research participants using a video conferencing platform

One of the novel methods used in this research was the use of the video conferencing platform GoToMeeting (GTM) in the remote online usability testing. Previous published evidence of online remote usability testing has been limited to audio recorded moderated sessions (Wozney *et al.*, 2016). The use of GTM software meant that participants could be interviewed while interacting with the Family Web website wherever they were in the country. GTM is a relatively simple system to use and does not require every person who 'joins' the meeting to be registered with their system. Using GTM the interviewer (or moderator) was able to transfer the role of 'presenter' to the volunteer user. This meant that the user shared their computer screen with the interviewer while they explored the website and commented on what they saw.

The Think-Aloud (Nielsen, 2012) interaction was recorded as video which in turn enabled the recording to be played back and analysed later, linking the users comments directly to their inter-actions with the website. Being able to see the interviewer may also have encouraged the users, although the prospect of using unfamiliar systems, even in the familiar situation of their own home, may have inhibited other potential volunteers from taking part.

This method of collecting data using a video conferencing platform could be applied in many other research settings and would be particularly valuable when eligible participants are dispersed over a wide geographical area. This method could be used in other genetic research where there are only small numbers of eligible patients or any research setting where participants are widely geographically distributed. I can therefore envisage GoToMeeting or a similar platform being used in online remote

usability testing of eHealth interventions. It could also be used to record the interactions of remote online focus groups or to assess student's use of online learning tools.

10.5 Implications for policy makers

I would acknowledge that further research needs to be carried out using this website (or others like it) to refine it, expand its application and test to what degree this method of information provision assists families. If there is evidence available then I believe policy makers should be recommending such innovations. However, without technical support and availability to training, plus a clear incentive to clinical staff to incorporate this type of innovation into their practice, it is unlikely to be utilised fully. I acknowledge that the inertia inherent in any health care system is likely to mitigate against change even when patients and their relatives want information to be provided to them in digital format.

10.6 Conclusions

This research study has made a unique contribution to knowledge on several counts.

- New **theories** have been developed that contribute to our understanding of what factors influence dissemination of information in families. A map of four major themes (impact, adaptation, practical information and appropriate communication) showed their suggested interactions. In addition, a model was postulated indicating the importance of a patient's psychological adaptation to their genetic diagnosis and how this influenced the timing of disclosure to other relatives.
- This research has made a contribution to clinical **practice**; indicating that people living with a high risk of bowel cancer do want more practical

information and support that is tailored to their needs; with emphasis on what they can do themselves to mitigate their increased risk of cancer. It has also shown that the timing of when clinical staff talk to their patients about sharing information is important and needs to come at a time when patients have adjusted to their own diagnosis.

- This study has also contributed to research **methods** development in healthcare through the novel use of online video conferencing to interview and record usability testing with participants across a wide geographical area.

Finally, I believe that many patients would use the type of web-based file sharing facility that I developed if it was available to them. In order to circulate information to the wider family patients need to be given time and support to adapt to their own diagnosis first. These original findings will be disseminated through peer-reviewed publications, scientific meetings and to a wider audience through the media, and I hope will be the catalyst for new research within this important field.

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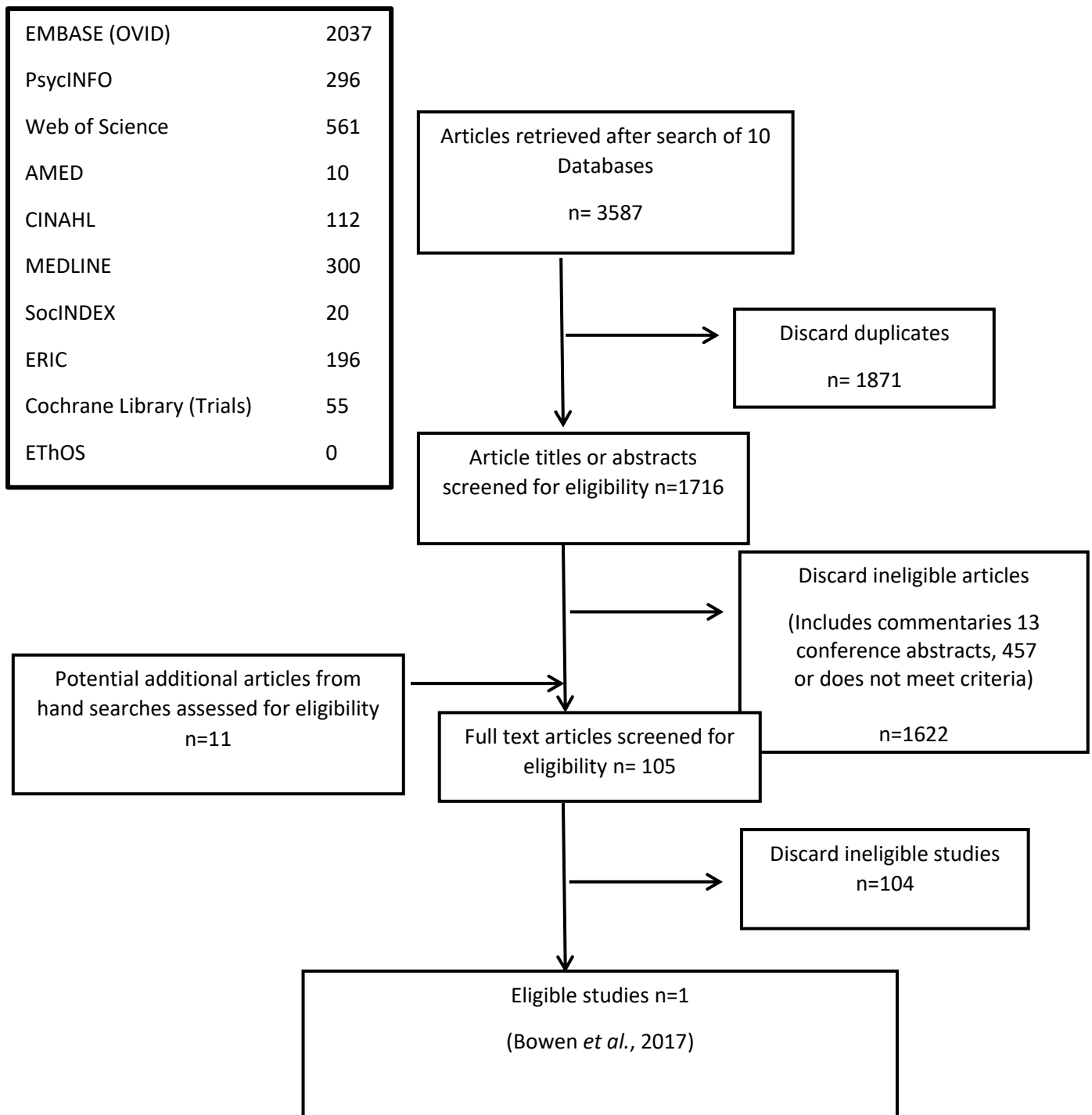
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Appendix 1 Systematic Review Combined Search PRISMA Diagram



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INVOLVE

Project Preview

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Title: Investigation into the impact of email or interactive websites for sharing information in families with an increased risk of bowel cancer

Project timescale: From 01 October, 2015 to 01 October, 2017
(**Added to website on:** 06 July, 2015 - **Date last updated:** 30 July, 2015)

Source of funding:
self-funded

Aims:

The aim of the project is to improve information provision and support to families who have a genetic vulnerability to bowel cancer so that they are better able to share information about the diagnosis and its implications within the family. In order to achieve this, we will be seeking the views and opinions of people who have been advised to have regular bowel screening by colonoscopy on the basis of their family history of cancer, or their known genetic risk of cancer. If our survey respondents and interviewees think it is appropriate, we intend to develop a web-based method of providing family-specific information in a secure but easily accessible way.

Research designs used:
Study of views/experiences
Other:

Methods used to collect data:
Interviews
Questionnaire survey
Other (please specify):

Research project description:

Email is now a common form of communication although it is still used less frequently in healthcare than in other arenas. However improving technology has enabled greater data security for the use of email in healthcare. Now alternatives to email such as password protected patient portals and websites could provide a way for family members to share information that has been provided by their doctor in a quick and secure manner. The issue of how information is shared in families who have a vulnerability to disease is a focus of attention and debate. Many different factors can make communication difficult but in families with an increased risk of cancer, there are significant potential benefits to relatives if that information can help them access appropriate screening or be alert to early symptoms of disease. This study will focus on families with an increased risk of bowel cancer. Those people who have been advised to have regular bowel screening by colonoscopy, will also have been told to warn their relatives of their risk. However, evidence indicates that only a minority of relatives access screening or genetic testing (Sharaf et al 2013, Snowsill et al 2014). We will try to develop improved methods of information provision through this

research. Phase 1: A cross-sectional survey will be given to people at increased risk of bowel cancer via websites, following genetics advice and at screening clinics. Phase 2: Semi-structured interviews will be conducted by telephone with a sample of respondents to the survey. Phase 3: A pilot website will be developed by an iterative process of Think-Aloud interviews and tested by users. Results and conclusions from the study will be made available via participating hospitals and charity websites. We hope that this research will establish new, secure and validated sources of information which can be shared electronically within families and therefore help relatives inform and support each other.

Stages at which the public were involved:

Prioritising topic areas

Planning the research

Managing the research

Designing the research instruments

(eg questionnaires, patient information sheets)

Other:

Description of public involvement in research stages:

This study is still in its design phase and applications for NHS research ethics are currently in process. Members of the public have acted as advisers on the project by providing their perspective and contributing ideas about issues to be addressed within the study. Since the study is seeking patient views and opinions one major contribution by our advisers has been reading and commenting on the draft survey. We also anticipate seeking their advice regarding patient information sheets and interview format.

Training and support provided for either members of the public or researchers involved in the project:**Examples of ways the public have made a difference to the research project:**

So far, members of the public have guided the research design and provided the perspective of potential users to a proposed website. Their input has made us aware of the issues most important to these families.

Evaluating the impact of public involvement in the research:**Details of publications or reports resulting from the research:****Links to Reports:**

Was/is your project user controlled: No

For further information on the project, please contact:

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Family Web Study



Protocol

Investigation into the use of emails and interactive websites for the provision of information by health professionals to families at increased risk of colorectal cancer to facilitate sharing information by relatives

Introduction and summary

Email is now a common form of communication; although it is still used less frequently in healthcare than in other arenas. However improving technology has enabled greater data security for the use of email in healthcare ¹. In addition, alternatives to email such as password protected patient portals and websites ² could now provide an opportunity for family members to share information that has been provided by their healthcare professional in a quick and secure manner.

The issue of how information is communicated in families who have a shared vulnerability to disease is a focus of attention and debate ^{3,4}. Many different factors can impede communication ⁵ but in families with an increased risk of cancer, there are significant potential benefits to relatives if that information can help them access appropriate screening or be alert to early symptoms of disease ^{6,7}.

This study will focus on families at an increased risk of bowel cancer. Those individuals who have been advised to have regular bowel screening by colonoscopy will have been told to warn their relatives of their risk. However, evidence indicates that only a minority of relatives access screening or genetic testing ^{8,9}. We will seek to develop improved methods of information provision through this research.

Phase 1 A cross-sectional survey will be administered to around 300 people at increased risk of bowel cancer via websites, following genetics advice and at screening clinics.

Phase 2 Semi-structured interviews will be conducted by telephone with a purposive sample of approximately 20 respondents to the survey.

Phase 3 A pilot website will be developed by an iterative process of Think-Aloud interviews with about 30 participants and tested by users.

Background

In the United Kingdom (UK), colorectal cancer is now the third most common cancer for both men and women, with over 34,000 new cases diagnosed in 2012 [Office of National Statistics] and 1.4 million new cases diagnosed worldwide in 2012 ¹⁰. Of these cases of cancer, 3.3% diagnosed under the age of 70 years can be attributed to an autosomal

dominantly inherited vulnerability to cancer called Lynch syndrome (LS, previously hereditary non-polyposis colorectal cancer or HNPCC)¹¹, 1% will be due to another autosomal dominant condition, familial adenomatous polyposis (FAP)¹² and a further 20% - 25% will have arisen because of an inherited vulnerability of usually unknown cause¹³. In addition, recent data from Europe indicate that Lynch syndrome is much more common than previously believed, probably affecting 1 in 300 people¹¹ so the implications of such genetic diagnoses will affect many people.

In Lynch syndrome the lifetime risk of colorectal cancer is up to 80% and there are significantly increased risks of multiple malignancies including endometrium, ovary, gastric and other cancers¹⁴, while in FAP the risk of colorectal cancer approaches 100% by age 60¹². However, in individuals known to have inherited an increased vulnerability to colorectal cancer, targeted bowel screening by colonoscopy can significantly reduce the morbidity and mortality of the disease^{6,15}.

Unfortunately, current methods used to diagnose the families with these conditions are only identifying a fraction of the families affected^{9,16}. Recent research in the UK and Australia has looked into why individuals with colorectal cancer, or their relatives with a significant family history of cancer, are not being identified. The evidence suggests that primary care physicians, as well as medical and surgical specialists are reluctant to refer to genetics services for a variety of reasons, with one main barrier being a lack of knowledge about Lynch syndrome^{17,18}.

Once a diagnosis has been made in an individual it is important that they are supported by healthcare professionals in understanding the implications of that disease for themselves and their families¹⁹. However, even in families with Lynch syndrome where clear recommendations for management exist, evidence suggests that information is only shared with less than half of the relatives for whom it would be relevant⁸. This may be because those with the information are not always confident or feel able to disseminate what they know to their relatives who are also at risk²⁰ despite the encouragement of their health care professional²¹. Such inadequate or delayed communication can result in relatives developing cancers that could have been prevented²².

Therefore it is likely that many relatives remain uninformed and unable to access the appropriate advice and risk reducing surveillance available to them. We postulate that the current methods of patient support and information provision (with paper based leaflets) may be inadequate to meet the needs of families¹⁷. We therefore hope to develop new ways to help families who are confronted with this burden of risk and the need to share health advice. In doing so it would seem logical to use methods of communication that reflect the changes that have occurred over the last decade in the ways family members communicate with each other²³. Although a website based in the United States exists (www.kintalk.org) to promote education and communication in families at risk of cancer²⁴, there is as yet no published data on the acceptability and efficacy of such electronic communication, either by email or through interactive websites, in healthcare. A systematic review carried out in January 2015, to explore what evidence there was around

the impact of electronic communication by healthcare professionals on information sharing in families, found only one paper ²⁵ out of 1720 that provided evidence on this issue.

Objectives

The aim of this study is to investigate how health professionals could support and facilitate information sharing in families at an increased risk of bowel cancer, in seeking to optimise the health benefit in those families. More specifically we aim to investigate what the impact would be of providing information about a genetic diagnosis, and the health implications of that diagnosis, in an electronic format such as email or via an interactive website.

More specifically the objectives are to:

1. Explore the perspectives of patients and their relatives about the acceptability and desirability of providing health information about a familial diagnosis by email or through an interactive website.
2. Set up and test a password protected interactive website to facilitate information sharing in families with an increased risk of bowel cancer, to determine if this is feasible.
3. Ascertain the impact of providing information electronically on information sharing with relatives.
4. Make recommendations regarding methods of information sharing, timing and the type of information most useful to families with an increased risk of bowel cancer, based on the results of the study.

Eligibility for participation

Patients who are:

Over 17, living in England or Wales, competent in reading and speaking English and:

- Come from a family with a known genetic condition giving an increased risk of bowel cancer.

OR

- Have been advised to have regular bowel screening because of an increased risk of bowel cancer due to their family history of cancer.

OR

- Have had a cancer and they were told that it was due to a genetic vulnerability which included an increased risk of bowel cancer.

Therefore, men and women will be eligible who are from a family with a known genetic vulnerability to bowel cancer such as Lynch syndrome or familial adenomatous polyposis (FAP), but also those with other inherited vulnerabilities to bowel cancer **where regular bowel surveillance is indicated**.

Participants need to have had time to adjust to their diagnosis so they will be eligible if their diagnosis was made 3 or more months before. All participants would need to be competent in reading and speaking English to take part in the study.

Potential participants would be **excluded** from taking part in the study:

- if they are unable to give informed consent,
- do not comply with the criteria above or
- if they are receiving active treatment (radiotherapy or chemotherapy) for cancer or were diagnosed with cancer within the last 3 months.

However patients considered in remission who are taking maintenance medication would be eligible to take part as long as they were diagnosed at least 3 months before. This is to avoid giving greater burden to cancer patients while they are in the acute phase of their illness.

Recruitment

Recruitment to the anonymous online survey will be via a SurveyMonkey link to UK residents who fulfil the eligibility criteria. Participants will be invited to take part in the survey through promotional material on charity websites such as Macmillan, Lynch Syndrome UK and Bowel Cancer West. The same survey questionnaire will be available in paper copy format to participants who request a copy via the study email or leave a phone message.

Recruitment to Phases One and Two of the study will also be to eligible patients attending colorectal surgical outpatient clinics and Endoscopy clinics in Plymouth and Truro. These patient will be given participant information sheets (PIS) and invited to contact the study for more information and a consent form if they are interested in taking part.

Eligible patients who are known to Clinical Genetics Departments in any one of ten NHS Trusts in England and Wales (Exeter, Southampton, Cardiff, Bristol, Birmingham, Oxford, London, Harrow, Manchester and Newcastle) will be provided with the appropriate PIS for recruitment to each of the three Phases of the study.

Through this strategy we hope to recruit participants from across each of our eligible patient groups.

Experimental design and methods

Research design: this will be a mixed-methods study²⁶. In order to capture the views and experiences of a range of individuals and achieve triangulation²⁷ we are combining both a quantitative approach and a qualitative approach in the different phases of the study. The purpose of this method is to attempt to gather a broad collection of people's opinions and suggestions for improvement. Through triangulation we will attempt to achieve consistency and convergence of the conclusions reached. We hope that by applying these methods it may also be possible to attain some complementarity²⁸ as the different approaches of the questionnaire and interviews are intended to investigate different aspects of the problem of sharing information in families and thereby lead to deeper interpretations and conclusions²⁹.

In Phase 1, we will use a cross-sectional survey³⁰ administered either online (using SurveyMonkey) or in paper format³¹, to elicit the views of a broad cohort of individuals. These participants will have already been advised to have regular bowel screening to

prevent or detect cancer in accordance with guidelines³². The questionnaire will be available online to reach respondents across a wide geographical distribution, to reduce costs and facilitate completion³³.

The concurrent triangulation design of this study allows for data collection from the questionnaire responses to continue while some participants may already be proceeding to Phase 2 interviews. This allows for a responsive dynamic and evolving interpretation of the qualitative data in conjunction with the process of gathering more quantitative data. This nested analysis³⁴ is intended to utilise the benefits of both methods simultaneously and we hope will allow for the investigation and interpretation of this complex issue.

For those participants who use the paper format questionnaire a reply paid envelope will be provided. The quantitative data from this cross-sectional survey³⁵ will be analysed using descriptive statistics and SPSS software. Qualitative data from free text responses will be coded and analysed for recurrent themes using NVivo software. The survey concludes with an invitation to take part in further research. If participants, having read and understood the consent form, wish to be interviewed they are asked to contact the research team or provide their contact details.

Phase 2 will be based on a qualitative grounded theory approach³⁶ in order to define concepts of interest or concern and develop information most suited to the needs of the potential recipients. This is designed to enhance and provide more in depth information about their experiences and their needs in relation to sharing health information in the family. We will use semi-structured telephone interviews to collect data from a purposive sample of respondents from the survey³⁷. In this way we hope to understand the difficulties encountered and preferences for information provision of both men and women, and people of different ages and experiences.

Phase 3 involves the creation and testing of an interactive password protected website with potential users who will be recruited through clinical genetics services. The proposed website will be developed in an iterative manner using a series of Think-Aloud interviews³⁸ where the participant and the researcher sit together while the participant explores the website and voices their thoughts. These interviews will be recorded and then transcribed to allow coding and subsequent analysis by content and theme³⁹.

The subjects of this phase will be people who have been given a diagnosis of an increased risk of bowel cancer and advised to have regular bowel screening by colonoscopy. The interviews should be conducted between three months and one year after they were given their diagnosis in the genetics clinic. The efficacy of the website will be tested by logging the number of occasions that relatives access the website. This will be done anonymously but people who access the website can also in turn complete an anonymous survey to provide feedback regarding the website.

It is our intention to explore this method of information provision via the website and password protected portal. We propose further validation and work to test its efficacy as a tool for information sharing between relatives as part of post-doctoral research.

Confidentiality and Data Protection

The researcher conducting the interviews in Phase 2 (Selina Goodman) will have access to the participants name, their email address or telephone number in order to set up the interview at a convenient time for the participant. Participants who complete the online or paper survey will have the opportunity to contact the researcher direct to express their interest in taking part in an interview before they make a decision of whether to do so. Therefore the survey will remain **anonymous**.

If participants do tick that they are willing to be interviewed and provide their preferred method of contact (email or telephone) via the tear off slip, these personal details will be posted or sent separately from their survey responses in an additional reply paid envelope.

Those participants that indicate they are happy to be contacted about being interviewed can choose to use an alias, pseudonym or username to conceal their identity. All participants data will remain confidential at all times.

In each phase of the study where participants are introduced to the study by NHS clinical staff, the participants details will only be given to the researcher by clinical staff with the verbal consent of the participant. **No information about the participants health or treatment is required by the study.**

However, in order to contact potential participants and explain the study in more depth, providing them with written information and consent forms, it will be necessary for clinical staff to pass on the telephone or email address of their eligible patients who are interested in taking part.

The details of patients who decide not to take part will be removed and deleted as soon as they have expressed that decision. If patients receive information about the study but do not get in touch with the research team within a month, they will be sent a reminder about the study but not contacted again after that.

The study will remain compliant throughout with the Data Protection Act 1998 and information governance standards as set out in the Caldicott Review of 2013.

Analysis

The three phases of this study are intended in an explanatory, sequential design using both quantitative and qualitative methods to investigate a complex problem²⁹. In order to understand the issues deeply we are hoping to recruit a mixed sample of participants across a range of ages and with different experiences. Although much of the data generated by the questionnaire will be numeric, due to the limitations of funding in this study we are only attempting to recruit a relatively small sample of 300 participants to the Phase 1 survey, approximately 20 in the Phase 2 interviews and 50 people to the Phase 3 interviews.

Therefore our analysis will be confined to descriptive statistics, with bivariate analysis to examine covariance or measures of dependence between different variables and comparison of means but assisted by the application of SPSS software. The responses to the Likert type questions will be analysed as ordinal data using descriptive statistics to show

central tendencies and Chi-square as a measure of association ⁴⁰. In this descriptive survey we are seeking a sample with maximum variation of age, geography and educational qualification. However if half of the projected sample of 300 were to give clear preferences, say for information provided by email, this would give a 95% confidence interval of 44% to 56% for that estimate. This is acceptable precision for this study.

The free text parts of the questionnaire and the subsequent interviews will be capturing qualitative data, but we intend to attempt some data reduction and data correlation between the qualitative and quantitative data as part of this process of mixed-method analysis ⁴¹.

The audio-taped recordings of the interviews will be transcribed and read several times prior to any coding. The analysis of the qualitative data both from the free text boxes in the questionnaire and from the interviews will follow a grounded theory approach ³⁶. In order to develop recurrent themes, all statements will be coded and then the codes will be arranged into categories and themes. To make sure that there is no bias in coding, transcripts will be coded independently by the researcher and her supervisor and then they will meet to discuss their findings. Consensus will be achieved between the researchers following discussion about any discrepancies. Eventually the resultant categories and codes will be compared across the participants to arrive at recurrent themes that reflect their experience of and opinions about the topic.

The Think-Aloud interview transcripts will be analysed both by content and by thematic analysis ³⁹ to achieve more comprehensive interpretation of the interaction of the participants with the website.

Ethical considerations

Anyone eligible for this research will have already been given a recommendation to have regular bowel screening on the basis of their increased risk of bowel cancer. Since this is the criteria for eligibility they should be aware of this risk and the concept should not be novel or alarming to them. However prior to potential participation in the study, patients will be provided with information sheets explaining whether they may be eligible and what the study involves. Every information sheet includes a telephone number and email address should they have questions of any sort about the study. We anticipate that potential participants may have questions about their eligibility or the study in general.

Participants will be directed towards appropriate support services should they experience intrusive or burdensome thoughts at any time during the study. They will have read and signed a consent form before taking part in the study. They will retain a copy of the consent form and the original will be kept by the researchers in the university in a locked filing cabinet. An additional copy of the consent will be held in the site file if the participant is recruited through one of the NHS study sites. Participants will be reassured that their involvement and all their responses will remain confidential at all times. Participants will be provided with a letter to give to their GPs should they wish to make them aware of their involvement with the study. At each stage of the study, participants will be offered a list of helpful contacts should they wish to access support in addition to that provided by their GP.

There are a number of possible issues that may arise for participants through their involvement in any phase of this study. Participants are likely to have heightened anxiety in relation to cancer and we are aware that participation in either the survey or interviews may trigger some emotional reactions in relation to the questions asked.

Phase 1

The most likely reactions we anticipate would be feelings of guilt and an increased concern regarding their risk of cancer. When participants answer the questionnaire or when they are interviewed, the questions may evoke latent cancer fears in participants as they will be reminded of their own increased. However evidence from genetic counselling suggests that such psychological distress is usually short lived following genetic counselling or genetic testing ⁴²⁻⁴⁶.

Guilt could be in relation to their fears for the health of their children or grandchildren, who may have also inherited the increased risk of cancer from their parent. Feelings of guilt may also arise because participation in the study could remind them that they have not communicated with all their relatives about the shared risk of cancer ⁵. Evidence indicates that people with a genetic vulnerability to cancer do realise that they have a duty to warn their relatives, but they experience a conflict between the desire to protect their family from anxiety and distress and the knowledge that their relatives could reduce their risk of cancer through regular screening, taking medication and symptom awareness ⁴⁷.

If participants experience concern or distress we would encourage them to seek the advice of their GP (if they have any physical symptoms that give them concern), or alternatively, they should contact their genetic counsellor, or colorectal specialist nurse. If someone is experiencing profound and intrusive feelings of guilt, or other negative emotions, they could seek appropriate referral by their GP for supportive care. However, if the participant would prefer, the researcher can refer the participant to an appropriate health care professional.

We considered the convenience of completing the survey online, and in someone's chosen time and place, to be preferable to completing a paper copy. However paper copy surveys with reply paid envelopes will be offered to participants. We anticipate the paper survey will be the most likely method of completing the survey at the time of a colonoscopy, as there is unlikely to be easy access to the Internet in the Endoscopy unit.

Phase 2

The proposed questions to be used in the semi-structured interviews have been ethically approved (NHS REC ref: 15/SW/0250) but could be amended based on the survey responses. However, there will potentially be overlap between Phase 1 and 2. This is because Phase 2 interviews will be conducted in a purposive sample with maximum variation with people who have already completed the survey. We will seek to avoid a sense of coercion, reminding respondents that they are free to be involved, or not, and can withdraw from the study at any time without compromising their care. Informed consent will be sought at each phase of the study. We are aware that participants may be sensitive in relation to the topic and its association with cancer. We plan to interview people by

telephone for their convenience and the fact that this will enable the researcher to interview participants from across England and Wales. The researcher is an experienced genetic counsellor and in that role she has undertaken many telephone calls regarding sensitive subjects.

If a participant becomes upset or distressed during an interview the interview will be suspended. The interviewer will then give the participant the choice to decide whether to continue or to stop the interview then, or potentially withdraw from the study altogether. A similar approach, responsive to the vulnerabilities and sensitivities of the participants will be used for the Think-Aloud interviews in Phase 3 of the project. In each case, when an interview has been suspended for the above reason, the researcher will offer to refer the participant to an appropriate health care professional for support.

We hope that participants may experience a sense of satisfaction being involved in a study which is seeking to elicit their views and take them into account in developing a better service and better support for families like their own. In a broader sense, health professionals have a duty of care to assist families in sharing information²¹ but data suggests that it is only a minority of relatives in high risk families who are currently receiving bowel screening⁹ so it could be argued that there is currently an urgent need to improve the support for families in this situation.

All data (both collected electronically and that in paper format) will be kept securely in Plymouth University in a passcode protected secure office and locked filing cabinet. The only individuals with access to this data will be the Chief Investigator and her academic supervisors.

Benefits of the study

This original research is therefore being done to try and improve the way in which healthcare professionals inform, support and facilitate the dissemination of relevant information within families at increased risk of bowel cancer, in seeking to optimise the health benefits to those families. If setting up an interactive website is acceptable and feasible, then we would seek to test whether the use of the website does result in an increased uptake of bowel screening or genetic testing in families with an increased risk of bowel cancer. We hope that the proposed website could be set up, with proven efficacy and so established in a way that could be maintained by the NHS through Health Education England or another agency for the long term and future benefit of such families. The proposed website would not be intended for profitable purposes. In addition, it is possible that the principles of providing health information in an electronic format can be applied to help families with other genetic conditions.

Chief Investigator

Selina Goodman BA (Hons), MSc, Registered Genetic Counsellor (GCRB 192)

This research is being undertaken as part of a full time PhD programme. There are no conflicts of interest to declare.

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Appendix 4 Plymouth University letter of ethical approval

**RESEARCH
WITH
PLYMOUTH
UNIVERSITY**

12th October 2015

CONFIDENTIAL

Ms Selina M A Goodman
Plymouth University
4 Portland Villas
Plymouth University, Drakes Circus
Plymouth
PL4 8AA

Dear Selina

Application for Approval by Faculty Research Ethics Committee

Reference Number: 15/16-477

Application Title: Investigation into the use of emails and interactive websites for the provision of information by health professionals to families at increased risk of colorectal cancer to facilitate sharing information by relatives.

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

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Professor Michael Sheppard
CQSW BSc MA PhD FAcSS
Chair, Faculty Research Ethics
Committee

Appendix 5 NHS ethical application form submitted via IRAS

NHS REC Form

Reference:
15/SW/0250

IRAS Version 4.0.0

Welcome to the Integrated Research Application System

IRAS Project Filter

The integrated dataset required for your project will be created from the answers you give to the following questions. The system will generate only those questions and sections which (a) apply to your study type and (b) are required by the bodies reviewing your study. Please ensure you answer all the questions before proceeding with your applications.

Please complete the questions in order. If you change the response to a question, please select 'Save' and review all the questions as your change may have affected subsequent questions.

Please enter a short title for this project (maximum 70 characters)
Impact of email/websites for sharing information in families - V1

1. Is your project research?

Yes No

2. Select one category from the list below:

- Clinical trial of an investigational medicinal product
- Clinical investigation or other study of a medical device
- Combined trial of an investigational medicinal product and an investigational medical device
- Other clinical trial to study a novel intervention or randomised clinical trial to compare interventions in clinical practice
- Basic science study involving procedures with human participants
- Study administering questionnaires/interviews for quantitative analysis, or using mixed quantitative/qualitative methodology
- Study involving qualitative methods only
- Study limited to working with human tissue samples (or other human biological samples) and data (specific project only)
- Study limited to working with data (specific project only)
- Research tissue bank
- Research database

If your work does not fit any of these categories, select the option below:

Other study

2a. Please answer the following question(s):

- a) Does the study involve the use of any ionising radiation? Yes No
- b) Will you be taking new human tissue samples (or other human biological samples)? Yes No
- c) Will you be using existing human tissue samples (or other human biological samples)? Yes No

3. In which countries of the UK will the research sites be located?(Tick all that apply)

- England
- Scotland
- Wales
- Northern Ireland

Date: 03/09/2015

1

181861/825894/1/372

3a. In which country of the UK will the lead NHS R&D office be located:

- England
 Scotland
 Wales
 Northern Ireland
 This study does not involve the NHS

4. Which review bodies are you applying to?

- NHS/HSC Research and Development offices
 Social Care Research Ethics Committee
 Research Ethics Committee
 Confidentiality Advisory Group (CAG)
 National Offender Management Service (NOMS) (Prisons & Probation)

For NHS/HSC R&D offices, the CI must create Site-Specific Information Forms for each site, in addition to the study-wide forms, and transfer them to the PIs or local collaborators.

5. Will any research sites in this study be NHS organisations?

- Yes No

5a. Are all the research costs and infrastructure costs for this study provided by an NIHR Biomedical Research Centre, NIHR Biomedical Research Unit, NIHR Collaboration for Leadership in Health Research and Care (CLAHRC) or NIHR Research Centre for Patient Safety & Service Quality in all study sites?

- Yes No

If yes, NHS permission for your study will be processed through the NIHR Coordinated System for gaining NHS Permission (NIHR CSP).

5b. Do you wish to make an application for the study to be considered for NIHR Clinical Research Network (CRN) support and inclusion in the NIHR Clinical Research Network (CRN) Portfolio? Please see information button for further details.

- Yes No

If yes, NHS permission for your study will be processed through the NIHR Coordinated System for gaining NHS Permission (NIHR CSP) and you must complete a NIHR Clinical Research Network (CRN) Portfolio Application Form immediately after completing this project filter and before completing and submitting other applications.

6. Do you plan to include any participants who are children?

- Yes No

7. Do you plan at any stage of the project to undertake intrusive research involving adults lacking capacity to consent for themselves?

- Yes No

Answer Yes if you plan to recruit living participants aged 16 or over who lack capacity, or to retain them in the study following loss of capacity. Intrusive research means any research with the living requiring consent in law. This includes use of identifiable tissue samples or personal information, except where application is being made to the Confidentiality Advisory Group to set aside the common law duty of confidentiality in England and Wales. Please consult the guidance notes for further information on the legal frameworks for research involving adults lacking capacity in the UK.

8. Do you plan to include any participants who are prisoners or young offenders in the custody of HM Prison Service or who are offenders supervised by the probation service in England or Wales?

Yes No

9. Is the study or any part of it being undertaken as an educational project?

Yes No

Please describe briefly the involvement of the student(s):

Sellina Goodman is the principle investigator and will carry out any interviews and administer the survey. She is a full time doctoral student and Registered Genetic Counsellor (GCRB 192) and she is supervised by Professor Heather Skirton, a professor of Applied Health Genetics and also a Registered Genetic Counsellor.

9a. Is the project being undertaken in part fulfilment of a PhD or other doctorate?

Yes No

10. Will this research be financially supported by the United States Department of Health and Human Services or any of its divisions, agencies or programs?

Yes No

11. Will identifiable patient data be accessed outside the care team without prior consent at any stage of the project (including identification of potential participants)?

Yes No

Integrated Research Application System**Application Form for Research administering questionnaires/interviews for quantitative analysis or mixed methodology study****Application to NHS/HSC Research Ethics Committee**

The Chief Investigator should complete this form. Guidance on the questions is available wherever you see this symbol displayed. We recommend reading the guidance first. The complete guidance and a glossary are available by selecting [Help](#).

Please define any terms or acronyms that might not be familiar to lay reviewers of the application.

Short title and version number: (maximum 70 characters - this will be inserted as header on all forms)
Impact of email/websites for sharing information in families - V1

Please complete these details after you have booked the REC application for review.

REC Name:
South West, Exeter

REC Reference Number:
15/SW/0250

Submission date:
03/09/2015

PART A: Core study information**1. ADMINISTRATIVE DETAILS****A1. Full title of the research:**

Investigation into the use of emails and interactive websites for the provision of information by health professionals to families at increased risk of colorectal cancer to facilitate sharing information by relatives.

A2-1. Educational projects

Name and contact details of student(s):

Student 1

	Title	Forename/Initials	Surname
	Ms	Selina M A	Goodman
Address	4 Portland Villas Plymouth University, Drake Circus Plymouth		
Post Code	PL4 8AA		
E-mail	selina.goodman@plymouth.ac.uk		
Telephone	01752586584		
Fax			

Give details of the educational course or degree for which this research is being undertaken:

Name and level of course/ degree:
Doctor of Philosophy (full time)

Name of educational establishment:
Plymouth University, England

Name and contact details of academic supervisor(s):

Academic supervisor 1

	Title	Forename/Initials	Surname
	Professor	Heather	Skirton
Address	Faculty of Health and Human Sciences 3 Portland Villas Plymouth University Drake Circus Plymouth		
Post Code	PL4 8AA		
E-mail	heather.skirton@plymouth.ac.uk		
Telephone	01752586569		
Fax			

Academic supervisor 2

	Title	Forename/Initials	Surname
	Professor	Ray	Jones
Address	Faculty of Health and Human Sciences 3 Portland Villas Plymouth University, Drake Circus Plymouth		
Post Code	PL4 8AA		
E-mail	ray.jones@plymouth.ac.uk		
Telephone	01752586532		
Fax			

Please state which academic supervisor(s) has responsibility for which student(s):

Please click "Save now" before completing this table. This will ensure that all of the student and academic supervisor details are shown correctly.

Student(s)	Academic supervisor(s)
Student 1 Ms Selina M A Goodman	<input checked="" type="checkbox"/> Professor Heather Skirton <input checked="" type="checkbox"/> Professor Ray Jones

A copy of a current CV for the student and the academic supervisor (maximum 2 pages of A4) must be submitted with the application.

A2-2. Who will act as Chief Investigator for this study?

- Student
 Academic supervisor
 Other

A3-1. Chief Investigator:

	Title Forename/Initials Surname
	Ms Selina M A Goodman
Post	PhD Student
Qualifications	Human Sciences BA Hons Human Molecular Genetics with Counselling MSc Registered Genetic Counsellor
Employer	Plymouth University
Work Address	4 Portland Villas Plymouth University, Drakes Circus Plymouth
Post Code	PL4 8AA
Work E-mail	selina.goodman@plymouth.ac.uk
* Personal E-mail	selina.goodman@plymouth.ac.uk
Work Telephone	01752586584
* Personal Telephone/Mobile	07740927816
Fax	

** This information is optional. It will not be placed in the public domain or disclosed to any other third party without prior consent.
A copy of a current CV (maximum 2 pages of A4) for the Chief Investigator must be submitted with the application.*

A4. Who is the contact on behalf of the sponsor for all correspondence relating to applications for this project?
This contact will receive copies of all correspondence from REC and R&D reviewers that is sent to the CI.

	Title Forename/Initials Surname
	Ms Selina M A Goodman
Address	4 Portland Villas Plymouth University, Drakes Circus Plymouth
Post Code	PL4 8AA
E-mail	selina.goodman@plymouth.ac.uk
Telephone	01752586584
Fax	

A5-1. Research reference numbers. *Please give any relevant references for your study:*

Applicant's/organisation's own reference number, e.g. R & D (if available):

Sponsor's/protocol number:

Protocol Version:

Protocol Date:

Funder's reference number:

Project website:

Additional reference number(s):

Ref.Number	Description	Reference Number

Registration of research studies is encouraged wherever possible. You may be able to register your study through

your NHS organisation or a register run by a medical research charity, or publish your protocol through an open access publisher. If you have registered your study please give details in the "Additional reference number(s)" section.

A5-2. Is this application linked to a previous study or another current application?

Yes No

Please give brief details and reference numbers.

2. OVERVIEW OF THE RESEARCH

To provide all the information required by review bodies and research information systems, we ask a number of specific questions. This section invites you to give an overview using language comprehensible to lay reviewers and members of the public. Please read the guidance notes for advice on this section.

A6-1. Summary of the study. *Please provide a brief summary of the research (maximum 300 words) using language easily understood by lay reviewers and members of the public. Where the research is reviewed by a REC within the UK Health Departments Research Ethics Service, this summary will be published on the website of the National Research Ethics Service following the ethical review.*

A proportion of those affected by bowel cancer have inherited a genetic predisposition to the condition, and members of their families will have an increased risk of bowel cancer. Individuals who have been advised to have regular bowel screening by colonoscopy will have been told to warn their relatives of their risk. However, evidence indicates that only a minority of relatives access screening or genetic testing (Sharaf et al 2013, Snowsill et al 2014).

Many different factors can impede communication (Seymour et al 2010) but in families with an increased risk of cancer, there are significant potential benefits to relatives if that information can help them access appropriate screening or be alert to early symptoms of disease (Menko et al 2013).

Email is commonly used to communicate; although it is still used less frequently in healthcare than in other arenas. Improving technology has enabled greater data security for the use of email in healthcare (Newhouse et al 2015). Also, password protected patient portals and websites (Ammenwerth et al 2012) could provide new opportunities for family members to share information that has been provided by their healthcare professional in a quick and secure manner.

We will seek to develop improved methods of information provision through this research, focussing on providing information electronically to facilitate sharing.

Phase 1 A cross-sectional survey will be administered via websites, following genetics advice and at screening clinics to find out the experiences and opinions of people at increased risk of bowel cancer about methods of sharing information.

Phase 2 Semi-structured telephone interviews of a purposive sample of respondents to the survey will seek to capture more detailed opinions about how to improve supportive information.

Phase 3 A website will be developed and tested by users through Think-Aloud interviews to check acceptability and uptake.

A6-2. Summary of main issues. *Please summarise the main ethical, legal, or management issues arising from your study and say how you have addressed them.*

Not all studies raise significant issues. Some studies may have straightforward ethical or other issues that can be identified and managed routinely. Others may present significant issues requiring further consideration by a REC, R&D office or other review body (as appropriate to the issue). Studies that present a minimal risk to participants may raise complex organisational or legal issues. You should try to consider all the types of issues that the different reviewers may need to consider.

For the purpose of this application, all study participants, whether they are patients who are recruited through an NHS clinic or those people recruited to the study through charity websites, will be referred to as a 'participants'.

Anyone eligible for this research will have already been given a recommendation to have regular bowel screening on the basis of their increased risk of bowel cancer. Since this is the criteria for eligibility they should be aware of this risk

and the concept should not be novel or alarming to them. Prior to potential participation in the study they will be provided with information sheets explaining whether they may be eligible and what the study involves. Every information sheet includes a telephone number and email address should they have questions of any sort about the study. We anticipate that potential participants may have questions about their eligibility or the study in general

UK residents will be eligible for the Phase 1 survey. Recruitment will be via advertisements on charity websites such as Bowel Cancer West, Macmillan and Lynch Syndrome UK. Other advertisements will be put up and distributed in NHS clinics in surgery, endoscopy or clinical genetics in England or Wales. The results of the study will be disseminated via the websites which have hosted links to the study and also to the participants via their local NHS Trust if they have been recruited through the Endoscopy service, Colorectal outpatients clinic or Clinical Genetics clinic.

Participants will be directed towards appropriate support services should they experience intrusive or burdensome thoughts at any time during the study. If they are taking part in the survey alone, they will have read and assented to taking part in the study if they proceed with the survey. Since participants can choose to remain anonymous contributors to the survey no written or explicit consent will be taken.

For those participants who offer to take part in the telephone interviews, they will have been posted or emailed an information leaflet and consent form. Participants can choose to conceal their identity by using a pseudonym or username if they wish to when providing their preferred contact details.

They will have the opportunity to read the information leaflet in their own time. They will be asked if they have any questions relating to the study before any discussion of consent. Then their consent will be agreed verbally with the researcher prior to commencement of the telephone interview. Participants will be offered a copy of the consent form signed by the researcher for their own records.

If participants wish to provide their General Practitioner (GP) details so that their GP can be informed of their involvement in the study we will write to their GP after completion of the interview. However it is not obligatory that we inform their GP if participants do not wish us to. The telephone conversation will be recorded, including the consent process to allow for internal audit. These recordings will be destroyed once the interviews have been transcribed for analysis. Participants will be reassured that their involvement and all their responses will remain confidential at all times.

Participants who are recruited via NHS clinics will be provided with an information leaflet and consent form. If they choose to take part in the study these participants will sign a consent form, retain a copy of that form for their own records, a copy will then go in their hospital notes and the original will be kept by the researchers in the university in a locked filing cabinet. An additional copy of the written consent will be held in the local site file of that NHS study site.

At each stage of the study, participants will be offered a list of helpful contacts should they wish to access support, in addition to that provided through their GP.

We anticipate that there are a number of possible issues that may arise for participants through their involvement in any phase of this study. Participants are likely to have heightened anxiety in relation to cancer and we are aware that participation in either the survey or interviews may trigger some emotional reactions in relation to the questions asked.

Phase 1

The most likely reactions we anticipate would be feelings of guilt and an increased concern regarding their risk of cancer. When participants answer the questionnaire or when they are interviewed, the questions may evoke latent cancer fears as they will be reminded of their own increased risk. However evidence from genetic counselling, and experience from my own clinical practice, suggests that such psychological distress is usually short lived following genetic counselling or genetic testing [1-5].

Guilt could be in relation to their fears for the health of their children or grandchildren, who may have also inherited the increased risk of cancer from their parent. Feelings of guilt may also arise because participation in the study could remind them that they have not communicated with all their relatives about the shared risk of cancer [6]. Evidence indicates that people with a genetic vulnerability to cancer do realise that they have a duty to warn their relatives, but they experience a conflict between the desire to protect their family from anxiety and distress and the knowledge that their relatives could reduce their risk of cancer through regular screening, taking medication and symptom awareness [7].

If participants experience concern or distress we would encourage them to seek the advice of their GP (if they have any physical symptoms that give them concern), or alternatively, they should contact their genetic counsellor, or colorectal specialist nurse. If someone is experiencing profound and intrusive feelings of guilt, or other negative emotions, they could seek appropriate referral by their GP for supportive care. However, if the participant would prefer, the researcher can refer the participant to an appropriate health care professional.

We considered the convenience of completing the survey online, and in someone's chosen time and place, to be preferable to completing a paper copy. However paper copy surveys with reply paid envelopes will be offered to participants.

Phase 2

The questions used in the semi-structured interviews with participants will be developed following the survey results. Phase 2 interviews will be conducted in a maximum variance purposive sample of people who have already completed the survey. However we will avoid a sense of coercion, reminding respondents that they are free to be involved, or not, and can withdraw from the study at any time without compromising their care. Informed consent will be sought at each phase of the study. We are aware that participants may be sensitive in relation to the topic and its association with cancer.

We plan to interview people by telephone for their convenience and the fact that this will enable the researcher to interview participants from across England and Wales. The researcher will seek further training in telephone interview techniques prior to conducting interviews but, she is an experienced genetic counsellor and in that role she has undertaken many telephone calls regarding sensitive subjects.

If a participant becomes upset or really distressed during an interview, they will be asked if they would like the interview to be suspended. The interviewer will then give the participant the choice to decide whether to continue, to stop the interview then, or potentially withdraw from the study altogether. A similar approach, responsive to the vulnerabilities and sensitivities of the participants will be used for the 'Think-Aloud' interviews in Phase 3 of the project. In each case, when an interview has been suspended for the above reason, the researcher will offer to refer the participant to an appropriate health care professional for support.

['Think-Aloud' interviews are a method for recording the dynamic interaction of a participant with a computer programme. In our study this type of interview will be used to find out how participants explore the proposed website at will while talking about their thoughts and actions. The activity of the computer and their associated verbalisation will be recorded for analysis. This recording can be done remotely following the method of moderated remote usability testing where the participant and interviewer/moderator are in different locations but the interviewer is able to talk to the participant, view what the participant is seeing on their computer and record their dialogue.]

We hope that participants may experience a sense of satisfaction being involved in a study which is seeking to elicit their views and take them into account in developing a better service and better support for families like their own. In a broader sense, health professionals have a duty of care to assist families in sharing information [8] but data suggest that it is only a minority of relatives in high risk families who are currently receiving bowel screening [9] so it could be argued that there is currently an urgent need to improve the support for families in this situation.

All data (both collected electronically and that in paper format) will be kept securely in Plymouth University in a passcode protected secure office and locked filing cabinet. The only individuals with access to this data will be the Chief Investigator and her academic supervisors.

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2. Burton-Chase, A.M., E.R. Gritz, and S.K. Peterson, Genetic counseling and testing for hereditary cancers: Psychosocial considerations, in *Psycho-oncology*. 2013, American Psychiatric Publishing, Inc., Arlington, VA. p. 145-169.
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7. Lucassen, A. and M. Parker, Confidentiality and sharing genetic information with relatives. *Lancet*, 2010. 375(9725): p. 1507-9.
8. Dheensa, S., et al., Health-care professionals' responsibility to patients' relatives in genetic medicine: a systematic review and synthesis of empirical research. *Genetics in Medicine*, 2015.
9. Snowsill, T., et al., A systematic review and economic evaluation of diagnostic strategies for Lynch syndrome. *Health Technol Assess*, 2014. 18(58).

A6-3. Proportionate review of REC application *The initial project filter has identified that your study may be suitable for*

proportionate review by a REC sub-committee. Please consult the current guidance notes from NRES and indicate whether you wish to apply through the proportionate review service or, taking into account your answer to A6-2, you consider there are ethical issues that require consideration at a full REC meeting.

Yes - proportionate review No - review by full REC meeting

Further comments (optional):

Note: This question only applies to the REC application.

3. PURPOSE AND DESIGN OF THE RESEARCH

A7. Select the appropriate methodology description for this research. Please tick all that apply.

- Case series/ case note review
- Case control
- Cohort observation
- Controlled trial without randomisation
- Cross-sectional study
- Database analysis
- Epidemiology
- Feasibility/ pilot study
- Laboratory study
- Metanalysis
- Qualitative research
- Questionnaire, interview or observation study
- Randomised controlled trial
- Other (please specify)

A10. What is the principal research question/objective? Please put this in language comprehensible to a lay person.

The aim of this study is to investigate how health professionals could support and facilitate information sharing in families at an increased risk of bowel cancer, in seeking to optimise the health benefit in those families. More specifically we aim to investigate what the impact would be of providing information about a genetic diagnosis, and the health implications of that diagnosis, in an electronic format such as email or via an interactive website.

The objectives are to:

1. Explore the perspectives of patients and their relatives about the acceptability and desirability of providing health information about a familial diagnosis by email or through an interactive website.
2. Set up and test an electronic method of information provision (either by secure email or password protected interactive website) to facilitate information sharing in families with an increased risk of bowel cancer, to determine if this is feasible.
3. Ascertain the impact of providing information electronically on information sharing with relatives.
4. Make recommendations regarding methods of information sharing, timing and the type of information most useful to families with an increased risk of bowel cancer, based on the results of the study.

A11. What are the secondary research questions/objectives if applicable? Please put this in language comprehensible to a lay person.

If providing information electronically (either via an interactive website or by email) is acceptable and feasible, then we would seek to test whether this does result in an increased uptake of bowel screening or genetic testing in families with an increased risk of bowel cancer. We hope that whatever method is set up, such as the proposed website, could be with proven efficacy and so established in a way that could be maintained by the NHS through Health Education

England or another agency for the long term and future benefit of families. Any website created through this study would not be intended for profitable purposes.

In addition, it is possible that the principles of providing health information in an electronic format can be applied to help families with other genetic conditions.

A12. What is the scientific justification for the research? *Please put this in language comprehensible to a lay person.*

In the United Kingdom (UK), colorectal cancer is now the third most common cancer for both men and women, with over 34,000 new cases diagnosed in 2012 [Office of National Statistics www.ons.gov.uk] and 1.4 million new cases diagnosed worldwide in 2012 [1]. Of these cases of cancer, 3.3% diagnosed under the age of 70 years can be attributed to an autosomal dominantly inherited vulnerability to cancer called Lynch syndrome (LS, previously hereditary non-polyposis colorectal cancer or HNPCC) [2], 1% will be due to another autosomal dominant condition, familial adenomatous polyposis (FAP) [3] and a further 20% - 25% will have arisen because of an inherited vulnerability of usually unknown cause [4]. In addition, recent data from Europe indicate that Lynch syndrome is much more common than previously believed, probably affecting 1 in 300 people [2] so the implications of such genetic diagnoses will affect many people.

In Lynch syndrome the lifetime risk of colorectal cancer is up to 80% and there are significantly increased risks of multiple malignancies including endometrium, ovary, gastric and other cancers [5], while in FAP the risk of colorectal cancer approaches 100% by age 60 [3]. However, in individuals known to have inherited an increased vulnerability to colorectal cancer, targeted bowel screening by colonoscopy can significantly reduce the morbidity and mortality of the disease [6, 7].

Unfortunately, current methods used to diagnose the families with these conditions are only identifying a fraction of the families affected [8, 9]. Recent research in the UK and Australia has looked into why individuals with colorectal cancer, or their relatives with a significant family history of cancer, are not being identified. The evidence suggests that primary care physicians, as well as medical and surgical specialists are reluctant to refer to genetics services for a variety of reasons, with one main barrier being a lack of knowledge about Lynch syndrome [10, 11].

Once a diagnosis has been made in an individual it is important that they are supported by healthcare professionals in understanding the implications of that disease for themselves and their families [12]. However, even in families with Lynch syndrome where clear recommendations for management exist, evidence suggests that information is only shared with less than half of the relatives for whom it would be relevant [13]. This may be because those with the information are not always confident or feel able to disseminate what they know to their relatives who are also at risk [14] despite the encouragement of their health care professional [15]. Such inadequate or delayed communication can result in relatives developing cancers that could have been prevented [16].

Therefore it is likely that many relatives remain uninformed and unable to access the appropriate advice and risk reducing surveillance available to them. We postulate that the current methods of patient support and information provision (with paper based leaflets) may be inadequate to meet the needs of families [10]. We therefore hope to develop new ways to help families who are confronted with this burden of risk and the need to share health advice. In doing so it would seem logical to use methods of communication that reflect the changes that have occurred over the last decade in the ways family members communicate with each other [17]. Although a website based in the United States exists (www.kintalk.org) to promote education and communication in families at risk of cancer [18], there is as yet no published data on the acceptability and efficacy of such electronic communication, either by email or through interactive websites, in healthcare. A systematic review that we carried out in January 2015 to explore what evidence there was around the impact of electronic communication by healthcare professionals on information sharing in families found only one paper [19] out of 1720 that provided evidence on this issue.

This original research is therefore being done to try and improve the way in which healthcare professionals inform, support and facilitate the dissemination of relevant information within families at increased risk of bowel cancer.

1. Ferlay, J., et al., GLOBOCAN 2012 v1. 0, Cancer Incidence and Mortality Worldwide: IARC CancerBase No. 11. Lyon, France: International Agency for Research on Cancer; 2013. Visit: <http://globocan.iarc.fr>, 2014.
2. van Lier, M.G., et al., Yield of routine molecular analyses in colorectal cancer patients \leq 70 years to detect underlying Lynch syndrome. *The Journal of pathology*, 2012. 226(5): p. 764-774.
3. Vasen, H., et al., Guidelines for the clinical management of familial adenomatous polyposis (FAP). *Gut*, 2008. 57(5): p. 704-713.
4. de la Chapelle, A., Genetic predisposition to colorectal cancer. *Nature reviews. Cancer*, 2004. 4(10): p. 769.
5. Lynch, H. and A. de la Chapelle, Genetic susceptibility to non-polyposis colorectal cancer. *Journal of Medical Genetics*, 1999. 36(11): p. 801-818.
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7. Newton, K., et al., Colonoscopy screening compliance and outcomes in patients with Lynch Syndrome. *Colorectal*

- Dis, 2014.
8. Adelson, M., et al., UK colorectal cancer patients are inadequately assessed for Lynch syndrome. *Frontline Gastroenterology*, 2013.
 9. Snowsill, T., et al., A systematic review and economic evaluation of diagnostic strategies for Lynch syndrome. *Health Technol Assess*, 2014. 18(58).
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 11. Tan, Y.Y. and L.J. Fitzgerald, Barriers and Motivators for Referral of Patients with Suspected Lynch Syndrome to Cancer Genetic Services: A Qualitative Study. *Journal of Personalized Medicine*, 2014. 4(1): p. 20-34.
 12. Lucassen, A. and A. Hall, Consent and confidentiality in clinical genetic practice: guidance on genetic testing and sharing genetic information. *Clin Med*, 2012. 12(1): p. 5-6.
 13. Sharaf, R.N., et al., Uptake of Genetic Testing by Relatives of Lynch Syndrome Proband: A Systematic Review. *Clinical Gastroenterology and Hepatology*, 2013. 11(9): p. 1093-1100.
 14. Forrest, K., et al., To tell or not to tell: barriers and facilitators in family communication about genetic risk. *Clinical Genetics*, 2003. 64(4): p. 317-326.
 15. Dheensa, S., et al., Health-care professionals' responsibility to patients' relatives in genetic medicine: a systematic review and synthesis of empirical research. *Genetics in Medicine*, 2015.
 16. Mesters, I., et al., Informing one's family about genetic testing for hereditary non-polyposis colorectal cancer (HNPCC): a retrospective exploratory study. *Familial Cancer*, 2005. 4(2): p. 163-167.
 17. Rudi, J., et al., Parents' use of information and communications technologies for family communication: differences by age of children. *Information, Communication & Society*, 2015. 18(1): p. 78-93.
 18. Myers, M., P. Conrad, and J. Terdiman, Kintalk.org: Helping Families Communicate Their Genetic Information. *Current Oncology*, 2014. 21(2): p. e363.
 19. Crotser, C.B. and S.S. Dickerson, Learning About a Twist in the Road: Perspectives of At-Risk Relatives Learning of Potential for Cancer. *Oncology Nursing Forum*, 2010. 37(6): p. 723-733.

A13. Please summarise your design and methodology. *It should be clear exactly what will happen to the research participant, how many times and in what order. Please complete this section in language comprehensible to the lay person. Do not simply reproduce or refer to the protocol. Further guidance is available in the guidance notes.*

Design and methodology

Research design: this will be a mixed-methods study [1]. In order to capture the views and experiences of a range of individuals and achieve triangulation [2] we are combining both a quantitative approach and a qualitative approach in the different phases of the study. The purpose of this method is to attempt to gather a broad collection of people's opinions and suggestions for improvement. Through triangulation we will attempt to achieve consistency and convergence of the conclusions reached. We hope that by applying these methods it may also be possible to attain some complementarity [3] as the different approaches of the questionnaire and interviews are intended to investigate different aspects of the problem of sharing information in families and thereby lead to deeper interpretations and conclusions [4].

In Phase 1, we will use a cross-sectional survey [5] administered either online (using SurveyMonkey) or in paper format [6], to elicit the views of a broad cohort of individuals. Potential participants from across the UK will be approached through advertisements on charity websites. Other advertisements will be put up or distributed in NHS clinics in endoscopy, colorectal surgical outpatients and clinical genetics in England and Wales. In additional potential participants thought to be eligible by clinical staff could be approached during their clinic visit or posted information about the study with their post-clinic letter.

Most participants will have already been advised to have regular bowel screening to prevent or detect cancer in accordance with guidelines [7]. The questionnaire will be available online to reach respondents across a wide geographical distribution, to reduce costs and facilitate completion [8]. We hope to recruit at least 350 participants in total to the study with approximately 300 respondents to the survey.

The concurrent triangulation design of this study allows for data collection from the questionnaire responses to continue while some participants may already be proceeding to Phase 2 interviews. This allows for a responsive dynamic and evolving interpretation of the qualitative data in conjunction with the process of gathering more quantitative data. This nested analysis [9] is intended to utilise the benefits of both methods simultaneously and we hope will allow for the investigation and interpretation of this complex issue.

For those participants who use the paper format questionnaire a reply paid envelope will be provided. The quantitative data from this cross-sectional survey [10] will be analysed using descriptive statistics and SPSS software. Qualitative data from free text responses will be coded and analysed for recurrent themes using NVivo software. The survey concludes with an invitation to take part in further research. If participants, having read and understood the consent form, wish to be interviewed they are asked to contact the research team or provide their contact details on a separate sheet or via a different web-page, thus ensuring participant anonymity is preserved.

Phase 2 will be based on a qualitative grounded theory approach [11] in order to define concepts of interest or concern and develop information most suited to the needs of the potential recipients. This is designed to enhance and provide more in depth information about their experiences and their needs in relation to sharing health information in the family. We will use semi-structured telephone interviews to collect data from a purposive sample of maximum variance respondents from the survey [12]. We intend to recruit approximately 20 people to these interviews but this number may vary and depends on how many interviews are needed to achieve saturation using a grounded theory approach. In this way we hope to understand the difficulties encountered and preferences for information provision of both men and women, and people of different ages and experiences.

Phase 3 will be guided by the results of the survey and interviews in Phases 1 and 2. Currently we anticipate the creation and testing of an interactive website with potential users who will be recruited through clinical genetics services. The proposed website will be developed in an iterative manner using a series of Think-Aloud interviews [13] where the participant and the researcher talk to each other via an online link, while the participant explores the website and voices their thoughts. These moderated remote usability interviews will be recorded and then transcribed to allow coding and subsequent analysis by content and theme [14]. We are seeking to recruit about 30 people to these interviews, which will be conducted with 5 participants per round, with 5-6 rounds of development.

The subjects of this phase will be people who have been given a diagnosis of an increased risk of bowel cancer and advised to have regular bowel screening by colonoscopy. The interviews should be conducted between three months and one year after they were given their diagnosis in the genetics clinic. The efficacy of the website will be tested by logging the number of occasions that relatives access the website. This will be done anonymously but people who access the website can also in turn complete an anonymous survey to provide feedback regarding the website. It is our intention to investigate this method of information provision via the website and password protected portal. We propose further validation and work to test its efficacy as a tool for information sharing between relatives as part of post-doctoral research.

1. Ivankova, N.V., J.W. Creswell, and S.L. Stick, Using mixed-methods sequential explanatory design: From theory to practice. *Field Methods*, 2006. 18(1): p. 3-20.
2. Creswell, J.W. and V.L.P. Clark, *Designing and conducting mixed methods research*. 2007.
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4. Farquhar, M.C., G. Ewing, and S. Booth, Using mixed methods to develop and evaluate complex interventions in palliative care research. *Palliative medicine*, 2011. 25(8): p. 748-757.
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10. Colton, D. and R.W. Covert, *Designing and constructing instruments for social research and evaluation*. 2007: John Wiley & Sons.
11. Strauss, A. and J. Corbin, *Basics of qualitative research: Procedures and techniques for developing grounded theory*. ed: Thousand Oaks, CA: Sage, 1998.
12. Bryman, A., Integrating quantitative and qualitative research: how is it done? *Qualitative research*, 2006. 6(1): p. 97-113.
13. McDermott, L., et al., Developing a computer delivered, theory based intervention for guideline implementation in general practice. *BMC family practice*, 2010. 11(1): p. 90.
14. Joffe, H. and L. Yardley, 4 Content And Thematic Analysis. *Research methods for clinical and health psychology*, 2004. 56.

A14-1. In which aspects of the research process have you actively involved, or will you involve, patients, service users, and/or their carers, or members of the public?

- Design of the research
- Management of the research
- Undertaking the research
- Analysis of results
- Dissemination of findings
- None of the above

Give details of involvement, or if none please justify the absence of involvement.

The researcher has worked alongside a group of Patient Advisors who were recruited from across the UK via the Lynch Syndrome UK conference via a voluntary sign up sheet. Their views and opinions have been sought via email and telephone discussions. These Patient Advisors views have helped formulate the questions being asked on the survey and they have read and commented on the survey, patient information and consent forms. In addition, the researcher has discussed the aims, objectives and design of the study in depth with genetic counsellors, surgeons and consultant geneticists who would be involved in the recruitment of participants.

The survey in Phase 1 of the study will be advertised via charity website and patient support groups such as Bowel Cancer West, Macmillan and Lynch Syndrome UK. The results of the study will be disseminated via the websites which have hosted links to the study and also to the participants via their local NHS Trust if their have been recruited through the Endoscopy service, Colorectal outpatients clinic or Clinical Genetics clinic.

4. RISKS AND ETHICAL ISSUES

RESEARCH PARTICIPANTS

A17-1. Please list the principal inclusion criteria (list the most important, max 5000 characters).

Inclusion criteria:

Anyone over the age of 17

- who is part of a family which is deemed to have an increased risk of bowel cancer due to their family history, or where a genetic vulnerability to bowel cancer has been found.
 - Or who have themselves been recommended to have bowel screening by colonoscopy on the basis of their family history of cancer, or where they are from a family where regular colonoscopy has been recommended for this reason.
 - Or who has been diagnosed with cancer, which they have been told was due to an inherited vulnerability to bowel cancer, such as Lynch syndrome, where their close relatives will have been recommended to regular colonoscopy.
- Participants need to have had time to adjust to their diagnosis so they will be eligible if their diagnosis (genetic or cancer) was made 3 or more months before.

All participants would need to be competent in reading and speaking English to take part in the study.

A17-2. Please list the principal exclusion criteria (list the most important, max 5000 characters).

Exclusion criteria

Men and women will be excluded if they:

They are unable to read and speak English.

They are not a UK resident for Phase 1.

They are not resident in England or Wales for Phases 2 & 3.

They are unable to give informed consent.

They do not have either

-a diagnosis of an inherited vulnerability to bowel cancer,

-or they have been advised by a specialist to have regular bowel screening by colonoscopy on the basis of their family history,

-or they are from a family where there is an inherited vulnerability to bowel cancer.

They would not be eligible if they had received a diagnosis of an inherited vulnerability to bowel cancer within the last 3 months.

They are receiving active treatment (radiotherapy or chemotherapy) for cancer or were diagnosed with cancer within the last 3 months. However patients considered in remission who are taking maintenance medication would be eligible to take part as long as they were diagnosed at least 3 months before. This is to avoid giving greater burden to cancer patients while they are in the acute phase of their illness.

RESEARCH PROCEDURES, RISKS AND BENEFITS

A18. Give details of all non-clinical intervention(s) or procedure(s) that will be received by participants as part of the research protocol. These include seeking consent, interviews, non-clinical observations and use of questionnaires.

Please complete the columns for each intervention/procedure as follows:

1. Total number of interventions/procedures to be received by each participant as part of the research protocol.

2. If this intervention/procedure would be routinely given to participants as part of their care outside the research, how many of the total would be routine?
3. Average time taken per intervention/procedure (minutes, hours or days)
4. Details of who will conduct the intervention/procedure, and where it will take place.

Intervention or procedure	1	2	3	4
Completion of survey questionnaire	1	0	20-30 minutes	Questionnaire to be completed by the participant in their own time and where convenient to them.
Telephone Interview	1	0	30-60 minutes	Researcher will conduct the interview by telephone at an agreed time which is convenient to the participant.
Think-Along interview	1		30-60 minutes	Interview will take place in the participants home via a live moderated remote usability online interview or if the participant prefers at a Plymouth University office with computer access.

A21. How long do you expect each participant to be in the study in total?

The length of time a participant will be in the study will vary depending on whether they take part in the survey, the survey and telephone interview, or the Think-Along interview. Participants taking part in the survey alone would be in the study for the time it took them to complete the survey, so approximately 30 minutes.

Participants who were interested in taking part in the telephone interviews, having completed the survey, could be in the study for between a few weeks to a maximum of 3 months. The participants indication to be available for interview would be acknowledged. Then if they were selected for interview they would be approached either by telephone or email (dependent on their preferred method of contact) and given more information about the interview process and the study. If they chose to take part and provided a telephone number then a time for the telephone interview would be agreed with them ideally within two weeks of their decision to take part. Participants will therefore be made aware that their involvement in the study has ended if they have not been chosen for interview.

Participants taking part in the Think-Along interviews will be a new group of participants recruited via genetics clinics or charity websites. These participants would be in the study for the duration of that interview, around 60 minutes. Once they have contacted the researcher by email or telephone message and consented to taking part in the Think-Along website development interview we would hope to arrange that interview within 2-3 weeks of receiving their written consent.

A22. What are the potential risks and burdens for research participants and how will you minimise them?

For all studies, describe any potential adverse effects, pain, discomfort, distress, intrusion, inconvenience or changes to lifestyle. Only describe risks or burdens that could occur as a result of participation in the research. Say what steps would be taken to minimise risks and burdens as far as possible.

The most likely adverse effects that we think participants might experience would be emotional, such as a sense of heightened anxiety in relation to cancer, which could occur at any stage of the study. Participation in the study may also provoke feelings of guilt because the survey questions or interviews could remind them how the inherited risk of cancer could affect the health of their children or grandchildren.

A different sense of guilt could arise because participants are reminded that they may not have communicated with all their relatives about the shared risk of cancer in the family. Some individuals may experience a conflict between the desire to protect their family from anxiety and the knowledge that their relatives could take action to lower their risk of cancer. All these emotional sequelae would be anticipated by the researcher as she has many years experience working with patients who have an increased risk of cancer. The burden of these emotional reactions would be minimised through careful and sensitive listening by the researcher during interviews but participants would be directed to other sources of information and support, or alternatively encouraged to seek the support and advice of their GP as the researcher could not act as their counsellor.

We hope that participants in the survey would not find this too intrusive as they can choose to complete the survey at a location of their choice, at a convenient time to them and they can stop after partial completion of the survey and return to it later if they wish to. Similarly those taking part in the interviews will have agreed an appropriate and convenient time with the researcher to conduct the interview. However those participants taking part in the Think-Along interviews in their own home may find it mildly intrusive, but all will have had time to consider this prior to consenting to take part. We hope that by providing the option to be interviewed via an online link this will minimise any intrusion

and also enable participants from across England or Wales to take part.

If someone was experiencing profound and intrusive feelings of guilt, or other negative emotions, they could seek appropriate referral by their GP for supportive care. Referral to their local genetics health service might also be appropriate if they had not received genetic counselling before and they wanted further advice about the risk to themselves or their relatives after taking part in the study.

In order to minimise the burden of taking part in the study we decided that the convenience of completing the survey online, and in someone's chosen time and place, would be generally preferable to completing a paper copy. However, in order to be as inclusive as possible, paper copy surveys with reply paid envelopes will be offered. Similarly, taking part in a telephone interview we hope will be minimally intrusive to participants. Participants in the Think-Aloud interviews will have the choice to take part at home (online) or in Plymouth University.

If a participant becomes very distressed during any of the interviews (either telephone or Think-Aloud) they will be asked if they would like the interview to be suspended, in this way seeking to minimise the distress. If an interview has been suspended because of the participant's distress, the researcher will offer to refer the participant to an appropriate health care professional for support.

A23. Will interviews/ questionnaires or group discussions include topics that might be sensitive, embarrassing or upsetting, or is it possible that criminal or other disclosures requiring action could occur during the study?

Yes No

If Yes, please give details of procedures in place to deal with these issues:

The focus of the interviews and questionnaires is about the methods of information exchange in families and as such should not be particularly upsetting. However, since the underlying reason for information exchange is the shared diagnosis of an increased risk of cancer, being reminded of this may be a sensitive subject for participants which they might find upsetting. However the interviewer is trained and experienced working with this group of patients, having worked as a registered genetic counsellor and her primary supervisor is also a registered genetic counsellor and experienced researcher working with patients in health care research. Therefore the research team has a wide experience of discussing sensitive topics and participants will be told at interview that they do not have to disclose information if they do not wish to.

Participants in the survey will be provided with a contact number and email address at which they can leave messages if the survey questions arouse concerns about their own health or the health of their relatives. The research team will read and respond to those messages appropriately in accordance with current clinical practice. Therefore if a respondent has already been seen by their local genetics service they will be encouraged to re-contact that service, but if they have not, the participant will be advised to go to their GP and seek their advice and referral to genetics if it is appropriate.

A24. What is the potential for benefit to research participants?

Taking part in the research may not directly benefit participants however participants may feel empowered by helping develop new resources that could help many other people in a similar situation. They may feel that their opinions are being heard and this will lead to improvements in service provision in the future. For those that take part in the interviews, they might find it therapeutic to talk over their experiences regarding how the diagnosis was shared in their family.

In addition, research participants may benefit by having the opportunity to think about how they and their family are able to reduce their risk of cancer through regular bowel screening. If they participate in the interviews or website development they may learn new information about how to reduce their risk of cancer (for example by taking regular doses of aspirin) or what sort of information to provide to relatives.

A26. What are the potential risks for the researchers themselves? (if any)

The researchers themselves may find it distressing to hear some of the accounts of their experiences given by participants at interview. However the researcher conducting the interviews is an experienced genetic counsellor and able to listen sensitively to people while maintaining an appropriate professional boundaries. The researcher is also supervised and supported by another very experienced genetic counsellor and researcher.

The website development 'Think-Aloud' interviews will be conducted face to face with the researcher. If those interviews are done in the participants own home this will involve the lone worker safe practice guidelines for clinical visits in the community with a third party in the Plymouth University aware of the time and location of the interview. The researcher will phone an agreed number before commencing the interview and at the end of the interview to verify that the interview has taken place safely. The researcher will keep a mobile phone on and available during the interview should any threatening behaviour take place.

In addition, the researcher will make it clear for both the telephone interviews and the Think-Aloud interviews that her role is as an independent researcher. Therefore any requests for clinical advice or guidance from participants (mindful of her previous work as a genetic counsellor) would be declined by the researcher and participants directed to their own local clinical genetics team. In this way the researcher would maintain appropriate boundaries within the research.

RECRUITMENT AND INFORMED CONSENT

In this section we ask you to describe the recruitment procedures for the study. Please give separate details for different study groups where appropriate.

A27-1. How will potential participants, records or samples be identified? Who will carry this out and what resources will be used? *For example, identification may involve a disease register, computerised search of GP records, or review of medical records. Indicate whether this will be done by the direct healthcare team or by researchers acting under arrangements with the responsible care organisation(s).*

Potential participants will be identified through a variety of sources.

Firstly some participants will identify themselves as being eligible through a series of self assessment questions on the websites that will be hosting links to the online version of the survey questionnaire. This self assessment will question whether they have been assessed by a health professional (such as GP, surgeon, or genetic counsellor) that they are at an increased risk of bowel cancer due to their family history of this cancer or other cancers and received advice about their risk. Inclusion criteria would also be met if that individual had only received advice from their relative about the familial risk and accessed regular bowel screening as a result either privately or through an NHS provider.

It is very important that patients who have not been assessed and received the appropriate clinical advice do not take part. We anticipate interest in the survey by people who consider themselves to have a significant family history of cancer but have not been referred to a specialist or received any clinical advice about the implications of their family history for their health. These individuals will be directed to consult their GP for advice and referral as appropriate. We do not wish to surprise anyone with the idea that their individual risk of cancer may also apply to their relatives due to the genetic or familial nature of the condition.

Participants identified through their clinical team will be identified as eligible either because:

- 1) They are part of a family where a known genetic vulnerability to bowel cancer has been found. They themselves do not have to be receiving regular bowel screening by colonoscopy if they are not the appropriate age or have been tested and found not to have the familial mutation. They would be eligible because they will have received advice about the condition and have had an opportunity to pass on that advice to other family members.
- 2) They have been advised to have regular bowel screening by colonoscopy on the basis of their family history and they have been told or counselled about the familial nature of the risk and its relevance to other people in their family.

A27-2. Will the identification of potential participants involve reviewing or screening the identifiable personal information of patients, service users or any other person?

Yes No

Please give details below:

People will be identified as eligible for the study by their clinical team in genetics, endoscopy, or surgery, that is where ever the recommendation has been made for regular bowel screening by colonoscopy in that individual. This means that the identification will not be done by the researcher. Participants to the online survey may be self identified as eligible following responses to initial 'gate-keeper' questions that will find out if someone has received a clinical recommendation to have regular bowel screening on the basis of their family history of cancer. It is not necessary for the purpose of this study to have any detailed clinical information about participants as we are seeking their opinions and ideas regarding how services can be improved. We would like to stratify responses to the survey based on whether someone has been diagnosed with cancer or not and also whether they know that there is a confirmed genetic diagnosis in the family, but their responses should remain anonymous and these details will be used in the analysis to look for associations and not to identify individuals.

A28. Will any participants be recruited by publicity through posters, leaflets, adverts or websites?

Yes No

If Yes, please give details of how and where publicity will be conducted, and enclose copy of all advertising material (with version numbers and dates).

Recruitment to the (Phase 1) survey will be via advertisements and links on charity websites related to cancer, bowel cancer or patient support such as Macmillan and Lynch Syndrome UK. Additional recruitment to both phases of the study will be via paper copy advertisements given out by research nurses, colorectal nurse specialists, surgeons, geneticists and genetic counsellors or A4 posters on display within waiting areas for appropriate clinics.

Phase 2 participants will have already completed the survey, so the last page of that survey invites them to consider taking part in further research by being interviewed on the telephone. This means that participants will be recruited to both Phase 1 and Phase 2 through the same publicity.

Recruitment to Phase 3 of the study will be following genetics assessment within Clinical Genetics clinics. Information leaflets about the study will be given in clinic or sent out with post clinic summary letters inviting interested potential participants to contact the research team for further information and consent forms. Since the Think-Along interviews can be conducted remotely and moderated by the interviewer, participants can be recruited from genetics clinics across England and Wales.

A29. How and by whom will potential participants first be approached?**Phase 1**

Initially the clinical staff member (nurse, genetic counsellor, surgeon, geneticist) will approach potential participants with cancer at no less than 3 months post cancer diagnosis. Participants who are unaffected by cancer and eligible because they have a family history or genetic vulnerability to bowel cancer and have been recommended to have regular screening colonoscopy could be approached at any stage after that recommendation has been made. In all cases the potential participants will be given written information about the study and asked to take their time and consider taking part in the research if they wish to do so. If they do, they will access the survey online, or return a reply slip to the researcher requesting a paper copy survey. Many participants will find out about the survey through advertisements posted on charity websites so will not be approached by a third person.

Phase 2 participants will have already completed the survey, so in effect the last page of that survey makes the approach about the telephone interviews and no additional approach will be made by clinical staff.

In Phase 3, potential Think-Along interview candidates will need to read and consider the patient information leaflet and then complete and return the consent form before any arrangements can be made for the Think-Along interview.

A30-1. Will you obtain informed consent from or on behalf of research participants?

Yes No

If you will be obtaining consent from adult participants, please give details of who will take consent and how it will be done, with details of any steps to provide information (a written information sheet, videos, or interactive material). Arrangements for adults unable to consent for themselves should be described separately in Part B Section 6, and for children in Part B Section 7.

If you plan to seek informed consent from vulnerable groups, say how you will ensure that consent is voluntary and fully informed.

Phase 1

Patients who are identified as eligible by clinical staff will be given a written participant information sheet (PIS) as an invitation to take part in the study. Potential participants will have the opportunity to ask questions about the research. At the beginning of the questionnaire participants will be asked to tick a box indicating that they have understood the information about the study and are consenting to taking part. Completion and posting of the paper copy questionnaire will constitute another indication of consent.

Where potential participants are responding to the online advertisement of the study, and following the links from charity websites, they will be presented with an online version of the PIS. If they are interested in the study having read the PIS then such potential participants will be asked to complete a short series of screening questions to determine if they are eligible. These questions rely on the honesty of the participant as there will be no way of corroborating their answers. If they are eligible potential participants will be presented with an online consent form to clarify that they have read and understood the purpose of the study and their involvement with it. If they choose they will be able to save and print the consent form for their own records but this will not be a requirement to take part. Since the decision to complete the questionnaire is voluntary and they can choose to stop or abandon the questionnaire at any time, completion and submission of the questionnaire will be an additional indication of consent.

Phase 2

Participants to the survey who have indicated that they wish to consider taking part in telephone interviews will be contacted by their preferred method (telephone or email) to give them information about what the interviews would involve. By email they will be sent a PIS and over the telephone the researcher will read out the PIS and invite questions. The participant will then be asked to consider if they wish to take part and invited to contact the research team by telephone or email to indicate their decision.

All those participants who choose to be interviewed will have the study explained to them again at the start of the interview and their verbal consent will be recorded. They will also be asked if they wish to be sent a letter which they can give to their GP explaining their involvement with the study. This means of informing the GP avoids the participant having to disclose their identity to the researcher and limits the amount of personal details given to the study.

Phase 3

Potential participants for the Think-Aloud interviews will be given a participant information sheet. They will have the opportunity to ask questions about the study and what it might involve for them. If they are interested in being interviewed they will be sent a consent form and asked to sign and return it. Before commencement of the interview the interviewer will go through the consent form again and invite any other questions. A copy of their signed consent form will be provided to the participant for their own records. The interviewer will also ask these participants if they wish to be given a letter to forward to their GP so that their GP is informed of their involvement with the study. If they do want this, the letter will be sent along with the copy of their consent form.

If you are not obtaining consent, please explain why not.

Please enclose a copy of the information sheet(s) and consent form(s).

A30-2. Will you record informed consent (or advice from consultees) in writing?

Yes No

A31. How long will you allow potential participants to decide whether or not to take part?

Participants will have an indefinite time to decide whether to take part in the study but that would be limited by the time period that the survey is 'live' and available online. In practical terms, the survey is likely to remain live for 18 months from the date when all ethical approvals have been granted. Participants will assent to completing the survey once they have read and completed the self-assessment screening questions to check their eligibility.

In Genetics clinics, Surgical outpatient's clinics and in Endoscopy clinics eligible patients will be given an information leaflet at their clinic visit or afterwards enclosed with their clinic summary letter. It would then be up to them to decide to access the survey online or respond with the reply paid slip to request a paper copy survey.

In Phase 3, patients will be identified by clinical staff and given information about the study in their appointment or afterwards with their clinic summary letter. Those patients interested in being involved in the Think-Aloud interviews will respond to the study with a reply slip. They will then be sent further information and a consent form to complete in their own time. They will be encouraged to email or ring the researcher if they have any specific questions relating to the research. There is no specific time-scale between being given information about the study and deciding to take part. However we hope to complete data collection by the end of June 2017 so again an upper limit of 18 months would be practical for the researcher.

A33-1. What arrangements have been made for persons who might not adequately understand verbal explanations or written information given in English, or who have special communication needs?(e.g. translation, use of interpreters)

The survey questions and interviews all be conducted in English and unfortunately it is not within the scope of this PhD research project to employ interpreters or have information sheets translated into many languages. However, a generic email address will be available for questions and enquiries at the commencement of the survey questionnaire, so that if a participant has problems completing the survey then they can email any queries to that address or telephone and leave a message which the researcher will respond to as soon as possible. People who are visually impaired or have literacy issues could use text to speech (TTS) software to provide an audio version of the information and survey questions to help comprehension. In Phases 2 and 3, prior starting any interviews, the researcher will read out the information sheet and go through the consent form with participants and answer any questions they may have, at that point the researcher can check whether the participant has any special communication needs. If they do, every appropriate effort will be made to enable the participant to take part, but in effect the study will be limited to people sufficiently literate in English to take part due to the financial constraints and limited scope of the study.

A35. What steps would you take if a participant, who has given informed consent, loses capacity to consent during the study? Tick one option only.

- The participant and all identifiable data or tissue collected would be withdrawn from the study. Data or tissue which is not identifiable to the research team may be retained.
- The participant would be withdrawn from the study. Identifiable data or tissue already collected with consent would be retained and used in the study. No further data or tissue would be collected or any other research procedures carried out on or in relation to the participant.
- The participant would continue to be included in the study.
- Not applicable – informed consent will not be sought from any participants in this research.
- Not applicable – it is not practicable for the research team to monitor capacity and continued capacity will be assumed.

Further details:

Since the study involves participants for discreet events such as the survey or the interviews it is unlikely that any participant would lose capacity during their involvement. However, for a participant who was able to give informed consent to be interviewed but then prior to the interview lost their capacity, then the interview would be cancelled and would not proceed. The researcher will go over the information and consent questions prior to each interview and within that conversation they would anticipate that they would become aware of a loss of capacity. Contemporaneous notes will be taken and kept by the researcher noting any issues of this kind.

If you plan to retain and make further use of identifiable data/tissue following loss of capacity, you should inform participants about this when seeking their consent initially.

CONFIDENTIALITY

In this section, personal data means any data relating to a participant who could potentially be identified. It includes pseudonymised data capable of being linked to a participant through a unique code number.

Storage and use of personal data during the study

A36. Will you be undertaking any of the following activities at any stage (including in the identification of potential participants)? (Tick as appropriate)

- Access to medical records by those outside the direct healthcare team
- Electronic transfer by magnetic or optical media, email or computer networks
- Sharing of personal data with other organisations
- Export of personal data outside the EEA
- Use of personal addresses, postcodes, faxes, emails or telephone numbers
- Publication of direct quotations from respondents
- Publication of data that might allow identification of individuals
- Use of audio/visual recording devices
- Storage of personal data on any of the following:
- Manual files including X-rays
 - NHS computers
 - Home or other personal computers
 - University computers
 - Private company computers
 - Laptop computers

Further details:

It will be necessary to store personal contact details for the duration of the study in order to arrange interviews but this

data will be destroyed after the project is completed. Any personal details will not be linked to the interview data so it will not be possible to identify which individual made specific comments. However direct quotes may be published to provide validity to the data but only once such data has been anonymised. All anonymised transcripts from the interviews will be kept for 10 years but the recordings of the interviews will be destroyed after the completion of the project as these may include identifying details of participants.

A38. How will you ensure the confidentiality of personal data? Please provide a general statement of the policy and procedures for ensuring confidentiality, e.g. anonymisation or pseudonymisation of data.

The researcher conducting the interviews in Phase 2 (Selina Goodman) will have access to the participants name, their email address or telephone number in order to set up the interview at a convenient time for the participant. Participants who complete the online or paper survey will have the opportunity to contact the researcher direct to express their interest in taking part in an interview before they make a decision of whether to do so. Therefore the survey will remain anonymous. If they do tick that they are willing to be interviewed and provide their preferred method of contact (email or telephone) via the tear off slip these personal details will be posted or sent separately from their survey responses in an additional reply paid envelope. Those participants that indicate they are happy to be contacted about being interviewed can choose to use an alias, pseudonym or username to conceal their identity. All participants data will remain confidential at all times.

In Phase 3 of the study, participants details will only be given to the researcher by clinical staff with the verbal consent of the participant. We are not seeking any details about their health or treatment but in order to contact potential participants and explain the study in more depth, providing them with written information and consent forms, it will be necessary for clinical staff to pass on the telephone or email address of their eligible patients who are interested in taking part. The details of patients who decide not to take part will be removed and deleted as soon as they have expressed that decision. If patients receive information about the study but do not get in touch with the research team within a month, they will be sent a reminder about the study but not contacted again after that.

A40. Who will have access to participants' personal data during the study? Where access is by individuals outside the direct care team, please justify and say whether consent will be sought.

The researcher conducting the interviews in Phases 2 and 3 (Selina Goodman) will have access to the participant's name, telephone number or email address for the purpose of arranging the interviews. These details will only be passed on to the researcher by clinical staff with the consent of the participant.

As part of the consent process participants will be asked whether they agree to their interview being recorded. If they do, the recordings of their interviews will be transcribed and anonymised. Once transcribed the audio recordings will be destroyed. Their words may be used to illustrate the findings of the research but those excerpts will have any identifying details removed to make them anonymous to any reader. If participants decline to have their interview recorded the interviewer will take notes of their comments but explain that this falls below the desired standard for thematic analysis and it is possible that some of their comments will be missed.

Storage and use of data after the end of the study

A43. How long will personal data be stored or accessed after the study has ended?

- Less than 3 months
- 3 – 6 months
- 6 – 12 months
- 12 months – 3 years
- Over 3 years

INCENTIVES AND PAYMENTS

A46. Will research participants receive any payments, reimbursement of expenses or any other benefits or incentives for taking part in this research?

Yes No

A47. Will individual researchers receive any personal payment over and above normal salary, or any other benefits or incentives, for taking part in this research?

Yes No

A48. Does the Chief Investigator or any other investigator/collaborator have any direct personal involvement (e.g. financial, share holding, personal relationship etc.) in the organisations sponsoring or funding the research that may give rise to a possible conflict of interest?

Yes No

NOTIFICATION OF OTHER PROFESSIONALS

A49-1. Will you inform the participants' General Practitioners (and/or any other health or care professional responsible for their care) that they are taking part in the study?

Yes No

If Yes, please enclose a copy of the information sheet/letter for the GP/health professional with a version number and date.

A49-2. Will you seek permission from the research participants to inform their GP or other health/ care professional?

Yes No

It should be made clear in the participant's information sheet if the GP/health professional will be informed.

PUBLICATION AND DISSEMINATION

A50. Will the research be registered on a public database?

Yes No

Please give details, or justify if not registering the research.

The Study is registered on the National Institute for Health Research INVOLVE database. This is a public access database which encourages public involvement in health research. We have registered the study on this database because we would welcome contact from members of the public who have an interest in this issue. We hope that by providing information regarding the aims and a summary of the project on this platform then those people who have an interest and feel that they would like to contribute towards improving the provision of information to families at risk of bowel cancer will contact the Principle Investigator and become involved as lay advisers.

Registration of research studies is encouraged wherever possible.

You may be able to register your study through your NHS organisation or a register run by a medical research charity, or publish your protocol through an open access publisher. If you are aware of a suitable register or other method of publication, please give details. If not, you may indicate that no suitable register exists. Please ensure that you have entered registry reference number(s) in question A5-1.

A51. How do you intend to report and disseminate the results of the study? Tick as appropriate:

- Peer reviewed scientific journals
- Internal report
- Conference presentation
- Publication on website

- Other publication
- Submission to regulatory authorities
- Access to raw data and right to publish freely by all investigators in study or by Independent Steering Committee on behalf of all investigators
- No plans to report or disseminate the results
- Other (please specify)

A PhD thesis will be written based on the results of this research. In addition a plain English summary will be written of the results and main findings to be disseminated to the research participants if they have asked to be sent the results.

A53. Will you inform participants of the results?

- Yes No

Please give details of how you will inform participants or justify if not doing so.

A summary of the results will be published via the charity websites that hosted links to the survey questionnaire and all collaborators within clinical departments will be informed and provided with copies of any published articles and the results generated by the study. This intention will be explained in the information about the study at the outset. Participants will be encouraged to access the results of the study via the charity websites but they would still be able to contact the researchers via the generic email and request a copy of the results should they choose to do so at the completion of the study. Participants will be informed about the likely duration of the study so that they will only anticipate results seeing results around that time.

5. Scientific and Statistical Review

A54. How has the scientific quality of the research been assessed? Tick as appropriate:

- Independent external review
- Review within a company
- Review within a multi-centre research group
- Review within the Chief Investigator's institution or host organisation
- Review within the research team
- Review by educational supervisor
- Other

Justify and describe the review process and outcome. If the review has been undertaken but not seen by the researcher, give details of the body which has undertaken the review:

Prior to commencement of the PhD, the topic of this research was appraised and considered appropriate for study as a PhD within the University.

Since then, the PhD supervisors Professor Heather Skirton and Professor Ray Jones have reviewed and validated the systematic literature review that was undertaken by the researcher in January 2015. In that literature review, ten electronic databases were searched and a total of 1720 peer reviewed articles were found (excluding duplicates) in response to the enquiry 'What is the impact of information provided by email, or via interactive websites, by health professionals on communication within the family about a familial diagnosis or health care issue?' From those articles, only one was found to meet the criteria. We therefore concluded that with evidence from only one study, although interesting, it indicated that there was a lack of published evidence on this issue.

In accordance with University policy, the research proposal has also been reviewed by an educator outside of the supervisory team and considered valid.

For all studies except non-doctoral student research, please enclose a copy of any available scientific critique reports, together with any related correspondence.

For non-doctoral student research, please enclose a copy of the assessment from your educational supervisor/ institution.

A56. How have the statistical aspects of the research been reviewed? Tick as appropriate:

- Review by independent statistician commissioned by funder or sponsor
- Other review by independent statistician
- Review by company statistician
- Review by a statistician within the Chief Investigator's institution
- Review by a statistician within the research team or multi-centre group
- Review by educational supervisor
- Other review by individual with relevant statistical expertise
- No review necessary as only frequencies and associations will be assessed – details of statistical input not required

In all cases please give details below of the individual responsible for reviewing the statistical aspects. If advice has been provided in confidence, give details of the department and institution concerned.

	Title	Forename/Initials	Surname
	Professor	Ray	Jones
Department	Faculty of Health and Human Sciences		
Institution	Plymouth University		
Work Address	Faculty of Health and Human Sciences 3 Portland Villas Plymouth University, Drake Circus Plymouth		
Post Code	PL4 8AA		
Telephone	01752586532		
Fax			
Mobile			
E-mail	ray.jones@plymouth.ac.uk		

Please enclose a copy of any available comments or reports from a statistician.

A57. What is the primary outcome measure for the study?

The primary outcome will be an understanding of the impact of providing health information about a familial diagnosis by email or through an interactive website, in the context of families at an increased risk of colorectal cancer. Thus the intention is to set up a website to facilitate information sharing and help families but only if that is feasible and indicated as desirable to this group.

A58. What are the secondary outcome measures? (if any)

Identification of the preferences to methods of information provision and support by healthcare professionals to this patient group.

A59. What is the sample size for the research? How many participants/samples/data records do you plan to study in total? If there is more than one group, please give further details below.

Total UK sample size: 350
 Total international sample size (including UK): 350
 Total in European Economic Area:

Further details:

For Phases 1 and 2 of the study, participants will be recruited from three different sources representing different groups of people who might access and benefit from improved information sources. These three groups are: 1) patients and their families with a known genetic vulnerability to colon cancer (such as families with Lynch syndrome and familial adenomatous polyposis) recruited through genetics clinics, 2) patients receiving regular bowel surveillance by colonoscopy on the basis of their family history recruited through Colorectal surgery and Endoscopy, 3) people who are aware of their increased risk of bowel cancer recruited through charity websites. Therefore for the

purpose of the Phase 1 survey we aim to recruit 300 people of mixed ages and genders to the study. For Phase 2 of the study we aim to recruit 20 people to the semi-structured telephone interviews. For Phase 3 of the study we aim to recruit 30 people to undertake Think-Aloud interviews while exploring the pilot interactive website. It will be particularly informative if interviewees seek the participation and consent of an eligible relative to test out the information sharing function of the website but each participant will be interviewed singly to gain maximum individual responses. Therefore, in total, we anticipate recruiting 350 people to the study.

A60. How was the sample size decided upon? *If a formal sample size calculation was used, indicate how this was done, giving sufficient information to justify and reproduce the calculation.*

We decided upon the size of the sample as an estimate of the numbers which would be sufficient to give meaningful qualitative data. We hope to recruit participants of both sexes and across the age ranges. However, families diagnosed with a known genetic vulnerability are relatively rare so we intend to commence recruitment initially through local NHS Trusts covered by the Peninsula Clinical Genetics Service (i.e. Plymouth, Truro, Exeter and Torbay) and then extend the study to include other NHS Trusts with clinical genetics services should that be necessary to recruit sufficient participants. These genetic services are aware of the proposed study as presentations have been made at regional meetings where the researcher sought the advice and support of her peers and clinical colleagues regarding the study. The participants recruited through surgical clinics and endoscopy clinics are likely to be recruited through clinics in Derriford Hospital in Plymouth due to the financial constraints of this project as a self-funded PhD project, although that recruitment may extend to Truro and Exeter if necessary.

In Phase 2, approximately 20 semi-structured telephone interviews will be conducted. This should be a sufficient number to achieve saturation and reveal recurrent themes using a Grounded Theory approach.

Similarly, in Phase 3, an iterative process of website development through Think-Aloud interviews will be conducted with each stage of the website explored and tested individually by 5 different interviewees. This process will be repeated over 5- 6 rounds of development until no further enhancement of the website is achieved. Therefore we anticipate needing to recruit about 30 participants to this phase of the study. This participants will be recruited through clinical genetics services across England and Wales.

A61. Will participants be allocated to groups at random?

Yes No

A62. Please describe the methods of analysis (statistical or other appropriate methods, e.g. for qualitative research) by which the data will be evaluated to meet the study objectives.

The three phases of this study are intended in an explanatory, sequential design using both quantitative and qualitative methods to investigate a complex problem [1]. In order to understand the issues deeply we are hoping to recruit a mixed sample of participants across a range of ages and with different experiences. Although much of the data generated by the questionnaire will be numeric, due to the limitations of funding in this study we are only attempting to recruit a relatively small sample of 300 participants to the Phase 1 survey, approximately 20 in the Phase 2 interviews and 50 people to the Phase 3 interviews.

Therefore our analysis will be confined to descriptive statistics, with bivariate analysis to examine covariance or measures of dependence between different variables and comparison of means but assisted by the application of SPSS software. The responses to the Likert type questions will be analysed as ordinal data using descriptive statistics to show central tendencies and Chi-square as a measure of association [2]. In this descriptive survey we are seeking a sample with maximum variance of age, geography and educational qualification. However if half of the projected sample of 300 were to give clear preferences, say for information provided by email, this would give a 95% confidence interval of 44% to 56% for that estimate. This is acceptable precision for this study.

The free text parts of the questionnaire and the subsequent interviews will be capturing qualitative data, but we intend to attempt some data reduction and data correlation between the qualitative and quantitative data as part of this process of mixed-method analysis [3].

The audio-taped recordings of the interviews will be transcribed and read several times prior to any coding. The analysis of the qualitative data both from the free text boxes in the questionnaire and from the interviews will follow a grounded theory approach [4]. In order to develop recurrent themes, all statements will be coded and then the codes will be arranged into categories and themes. To make sure that there is no bias in coding, transcripts will be coded independently by the researcher and her supervisor and then they will meet to discuss their findings. Consensus will

be achieved between the researchers following discussion about any discrepancies. Eventually the resultant categories and codes will be compared across the participants to arrive at recurrent themes that reflect their experience of and opinions about the topic.
The Think-Aloud interview transcripts will be analysed both by content and by thematic analysis [5] to achieve more comprehensive interpretation of the interaction of the participants with the website.

1. Farquhar, M.C., G. Ewing, and S. Booth, Using mixed methods to develop and evaluate complex interventions in palliative care research. *Palliative medicine*, 2011. 25(8): p. 748-757.
2. Boone, H.N. and D.A. Boone, Analyzing likert data. *Journal of Extension*, 2012. 50(2): p. 1-5.
3. Onwuegbuzie, A.J. and C. Teddlie, A framework for analyzing data in mixed methods research. *Handbook of mixed methods in social and behavioral research*, 2003: p. 351-383.
4. Strauss, A. and J. Corbin, *Basics of qualitative research: Procedures and techniques for developing grounded theory*. ed: Thousand Oaks, CA: Sage, 1998.
5. Joffe, H. and L. Yardley, *4 Content And Thematic Analysis. Research methods for clinical and health psychology*, 2004. 56.

6. MANAGEMENT OF THE RESEARCH

A63. Other key investigators/collaborators. *Please include all grant co-applicants, protocol co-authors and other key members of the Chief Investigator's team, including non-doctoral student researchers.*

A64. Details of research sponsor(s)

A64-1. Sponsor

Lead Sponsor

Status: NHS or HSC care organisation Commercial status: Non-Commercial
 Academic
 Pharmaceutical industry
 Medical device industry
 Local Authority
 Other social care provider (including voluntary sector or private organisation)
 Other

If Other, please specify:

Contact person

Name of organisation Plymouth University
 Given name Bridie
 Family name Kent
 Address Level 4, Rolle Building
 Town/city Drake Circus
 Post code PL4 8AA
 Country UNITED KINGDOM
 Telephone 01752 586566
 Fax
 E-mail bridie.kent@plymouth.ac.uk

Is the sponsor based outside the UK?

Yes No

Under the Research Governance Framework for Health and Social Care, a sponsor outside the UK must appoint a legal representative established in the UK. Please consult the guidance notes.

A65. Has external funding for the research been secured?

- Funding secured from one or more funders
 External funding application to one or more funders in progress
 No application for external funding will be made

What type of research project is this?

- Standalone project
 Project that is part of a programme grant
 Project that is part of a Centre grant
 Project that is part of a fellowship/ personal award/ research training award
 Other

Other – please state:

PhD that is not dependent on external funding to proceed, although applications for charitable funding currently in process.

A67. Has this or a similar application been previously rejected by a Research Ethics Committee in the UK or another country?

Yes No

Please provide a copy of the unfavourable opinion letter(s). You should explain in your answer to question A6-2 how the reasons for the unfavourable opinion have been addressed in this application.

A68-1. Give details of the lead NHS R&D contact for this research:

	Title Forename/Initials Surname
	Dr Lisa Vickers
Organisation	Plymouth Hospitals NHS Trust R&D Office
Address	Level 2, MSCP, Bircham Park Offices, Morlaix Drive Plymouth
Post Code	PL6 8BQ
Work Email	lisa.vickers@nhs.net
Telephone	01752 431776
Fax	
Mobile	

Details can be obtained from the NHS R&D Forum website: <http://www.rdforum.nhs.uk>

A69-1. How long do you expect the study to last in the UK?

Planned start date: 01/10/2015

Planned end date: 01/05/2018
 Total duration:
 Years: 2 Months: 6 Days: 1

A71-2. Where will the research take place? (Tick as appropriate)

England
 Scotland
 Wales
 Northern Ireland
 Other countries in European Economic Area

Total UK sites in study 12

Does this trial involve countries outside the EU?
 Yes No

A72. What host organisations (NHS or other) in the UK will be responsible for the research sites? Please indicate the type of organisation by ticking the box and give approximate numbers of planned research sites:

NHS organisations in England 11
 NHS organisations in Wales 1
 NHS organisations in Scotland
 HSC organisations in Northern Ireland
 GP practices in England
 GP practices in Wales
 GP practices in Scotland
 GP practices in Northern Ireland
 Social care organisations
 Phase 1 trial units
 Prison establishments
 Probation areas
 Independent hospitals
 Educational establishments
 Independent research units
 Other (give details)

Total UK sites in study: 12

A76. Insurance/ indemnity to meet potential legal liabilities

Note: in this question to NHS indemnity schemes include equivalent schemes provided by Health and Social Care (HSC) in Northern Ireland

A76-1. What arrangements will be made for insurance and/or indemnity to meet the potential legal liability of the sponsor(s) for harm to participants arising from the management of the research? Please tick box(es) as applicable.

Note: Where a NHS organisation has agreed to act as sponsor or co-sponsor, indemnity is provided through NHS schemes. Indicate if this applies (there is no need to provide documentary evidence). For all other sponsors, please describe the arrangements and provide evidence.

- NHS indemnity scheme will apply (NHS sponsors only)
 Other insurance or indemnity arrangements will apply (give details below)

University of Plymouth insurance and indemnity arrangements apply.
 Zurich Municipal Insurance Policy NHE- 05CA02-0013

Please enclose a copy of relevant documents.

A76-2. What arrangements will be made for insurance and/ or indemnity to meet the potential legal liability of the sponsor(s) or employer(s) for harm to participants arising from the design of the research? Please tick box(es) as applicable.

Note: Where researchers with substantive NHS employment contracts have designed the research, indemnity is provided through NHS schemes. Indicate if this applies (there is no need to provide documentary evidence). For other protocol authors (e.g. company employees, university members), please describe the arrangements and provide evidence.

- NHS indemnity scheme will apply (protocol authors with NHS contracts only)
 Other insurance or indemnity arrangements will apply (give details below)

University of Plymouth insurance and indemnity arrangements apply
 Zurich Municipal Insurance Policy NHE- 05CA02-0013

Please enclose a copy of relevant documents.

A76-3. What arrangements will be made for insurance and/ or indemnity to meet the potential legal liability of investigators/collaborators arising from harm to participants in the conduct of the research?

Note: Where the participants are NHS patients, indemnity is provided through the NHS schemes or through professional indemnity. Indicate if this applies to the whole study (there is no need to provide documentary evidence). Where non-NHS sites are to be included in the research, including private practices, please describe the arrangements which will be made at these sites and provide evidence.

- NHS indemnity scheme or professional indemnity will apply (participants recruited at NHS sites only)
 Research includes non-NHS sites (give details of insurance/ indemnity arrangements for these sites below)

University of Plymouth Insurance and indemnity arrangements apply
 Zurich Municipal Insurance Policy NHE- 05CA02-0013

Please enclose a copy of relevant documents.

PART C: Overview of research sites

Please enter details of the host organisations (Local Authority, NHS or other) in the UK that will be responsible for the research sites. For NHS sites, the host organisation is the Trust or Health Board. Where the research site is a primary care site, e.g. GP practice, please insert the host organisation (PCT or Health Board) in the Institution row and insert the research site (e.g. GP practice) in the Department row.

Research site		Investigator/ Collaborator/ Contact	
Institution name	Plymouth Hospitals NHS Trust	Title	Mr
Department name	Colorectal Surgery	First name/ Initials	Mark
Street address	Derriford Road	Surname	Coleman
Town/city	Plymouth		
Post Code	PL6 8DH		
Institution name	Royal Devon & Exeter Hospital (Heavitree)	Title	Dr
Department name	Peninsula Clinical Genetics Service	First name/ Initials	Charles
Street address	Gladstone Road	Surname	Shaw-Smith
Town/city	Exeter		
Post Code	EX1 2ED		
Institution name	University Hospital Southampton NHS Trust	Title	Dr
Department name	Wessex Clinical Genetics Service	First name/ Initials	Anneke
Street address	Princess Anne Hospital,	Surname	Lucassen
Town/city	Southampton		
Post Code	SO16 5YA		
Institution name	University Hospitals Bristol NHS Foundation Trust	Title	Dr
Department name	Bristol Clinical Genetics Service	First name/ Initials	Alan
Street address	St Michaels Hospital, Southwell Street	Surname	Donaldson
Town/city	Bristol		
Post Code	BS2 8EG		
Institution name	University Hospital of Wales	Title	Dr
Department name	All Wales Medical Genetics Service	First name/ Initials	Ian
Street address	Heath Park	Surname	Frayling
Town/city	Cardiff		
Post Code	CF14 4XW		
Institution name	Oxford University Hospitals NHS Trust	Title	Mr
Department name	Oxford Clinical Genetics	First name/ Initials	Peter
Street address	The Churchill, Old Road, Headington	Surname	Risby
Town/city	Oxford		
Post Code	OX3 7LJ		

Institution name	Birmingham Women's NHS Foundation Trust	Title	Ms
Department name	West Midlands Regional Genetics Service	First name/ Initials	Anna
Street address	Edgbaston	Surname	Considine
Town/city	Birmingham		
Post Code	B15 2TG		
Institution name	Guy's & St Thomas' NHS Foundation Trust	Title	Dr
Department name	Guy's Hospital Department of Clinical Genetics	First name/ Initials	Vishakha
Street address	7th Floor, Borough Wing, Great Maze Pond	Surname	Tripathi
Town/city	London		
Post Code	SE1 9RT		
Institution name	Royal Cornwall Hospitals NHS Trust	Title	Ms
Department name	Colorectal Surgery	First name/ Initials	Claire
Street address	Treliske	Surname	Ferris
Town/city	Truro		
Post Code	TR1 3LJ		
Institution name	St Marks and Northwick Park Hospital	Title	Ms
Department name	North West Thames Regional Genetics Service	First name/ Initials	Demetra
Street address	Watford Road	Surname	Georgiou
Town/city	Harrow		
Post Code	HA1 3 UJ		
Institution name	Newcastle upon Tyne Hospitals NHS Foundation Trust	Title	Professor Sir
Department name	Northern Genetics Service	First name/ Initials	John
Street address	International Centre for Life, Central Parkway	Surname	Burn
Town/city	Newcastle upon Tyne		
Post Code	NE1 3BZ		
Institution name	Central Manchester University Hospitals	Title	Dr
Department name	Manchester Centre for Genomic Medicine	First name/ Initials	Fiona
Street address	St Mary's Hospital, Oxford Road	Surname	Lalloo
Town/city	Manchester		
Post Code	M13 9WL		

PART D: Declarations**D1. Declaration by Chief Investigator**

1. The information in this form is accurate to the best of my knowledge and belief and I take full responsibility for it.
2. I undertake to abide by the ethical principles underlying the Declaration of Helsinki and good practice guidelines on the proper conduct of research.
3. If the research is approved I undertake to adhere to the study protocol, the terms of the full application as approved and any conditions set out by review bodies in giving approval.
4. I undertake to notify review bodies of substantial amendments to the protocol or the terms of the approved application, and to seek a favourable opinion from the main REC before implementing the amendment.
5. I undertake to submit annual progress reports setting out the progress of the research, as required by review bodies.
6. I am aware of my responsibility to be up to date and comply with the requirements of the law and relevant guidelines relating to security and confidentiality of patient or other personal data, including the need to register when necessary with the appropriate Data Protection Officer. I understand that I am not permitted to disclose identifiable data to third parties unless the disclosure has the consent of the data subject or, in the case of patient data in England and Wales, the disclosure is covered by the terms of an approval under Section 251 of the NHS Act 2006.
7. I understand that research records/data may be subject to inspection by review bodies for audit purposes if required.
8. I understand that any personal data in this application will be held by review bodies and their operational managers and that this will be managed according to the principles established in the Data Protection Act 1998.
9. I understand that the information contained in this application, any supporting documentation and all correspondence with review bodies or their operational managers relating to the application:
 - Will be held by the REC (where applicable) until at least 3 years after the end of the study; and by NHS R&D offices (where the research requires NHS management permission) in accordance with the NHS Code of Practice on Records Management.
 - May be disclosed to the operational managers of review bodies, or the appointing authority for the REC (where applicable), in order to check that the application has been processed correctly or to investigate any complaint.
 - May be seen by auditors appointed to undertake accreditation of RECs (where applicable).
 - Will be subject to the provisions of the Freedom of Information Acts and may be disclosed in response to requests made under the Acts except where statutory exemptions apply.
 - May be sent by email to REC members.
10. I understand that information relating to this research, including the contact details on this application, may be held on national research information systems, and that this will be managed according to the principles established in the Data Protection Act 1998.
11. Where the research is reviewed by a REC within the UK Health Departments Research Ethics Service, I understand that the summary of this study will be published on the website of the National Research Ethics Service (NRES), together with the contact point for enquiries named below. Publication will take place no earlier than 3 months after issue of the ethics committee's final opinion or the withdrawal of the application.

Contact point for publication*(Not applicable for R&D Forms)*

NRES would like to include a contact point with the published summary of the study for those wishing to seek further information. We would be grateful if you would indicate one of the contact points below.

- Chief Investigator
 Sponsor

- Study co-ordinator
- Student
- Other – please give details
- None

Access to application for training purposes *(Not applicable for R&D Forms)*

Optional – please tick as appropriate:

I would be content for members of other RECs to have access to the information in the application in confidence for training purposes. All personal identifiers and references to sponsors, funders and research units would be removed.

This section was signed electronically by Ms Selina Goodman on 03/08/2015 11:53.

Job Title/Post: PhD Student
Organisation: Plymouth University
Email: selina.goodman@plymouth.ac.uk

D2. Declaration by the sponsor's representative

If there is more than one sponsor, this declaration should be signed on behalf of the co-sponsors by a representative of the lead sponsor named at A64-1.

I confirm that:

1. This research proposal has been discussed with the Chief Investigator and agreement in principle to sponsor the research is in place.
2. An appropriate process of scientific critique has demonstrated that this research proposal is worthwhile and of high scientific quality.
3. Any necessary indemnity or insurance arrangements, as described in question A76, will be in place before this research starts. Insurance or indemnity policies will be renewed for the duration of the study where necessary.
4. Arrangements will be in place before the study starts for the research team to access resources and support to deliver the research as proposed.
5. Arrangements to allocate responsibilities for the management, monitoring and reporting of the research will be in place before the research starts.
6. The duties of sponsors set out in the Research Governance Framework for Health and Social Care will be undertaken in relation to this research.

Please note: The declarations below do not form part of the application for approval above. They will not be considered by the Research Ethics Committee.

7. Where the research is reviewed by a REC within the UK Health Departments Research Ethics Service, I understand that the summary of this study will be published on the website of the National Research Ethics Service (NRES), together with the contact point for enquiries named in this application. Publication will take place no earlier than 3 months after issue of the ethics committee's final opinion or the withdrawal of the application.
8. Specifically, for submissions to the Research Ethics Committees (RECs) I declare that any and all clinical trials approved by the HRA since 30th September 2013 (as defined on IRAS categories as clinical trials of medicines, devices, combination of medicines and devices or other clinical trials) have been registered on a publically accessible register in compliance with the HRA registration requirements for the UK, or that any deferral granted by the HRA still applies.

This section was signed electronically by Professor Brigid Kent on 03/08/2015 12:40.

Job Title/Post: Associate Dean/Head of School
 Organisation: University of Plymouth
 Email: bridie.kent@plymouth.ac.uk

D3. Declaration for student projects by academic supervisor(s)

1. I have read and approved both the research proposal and this application. I am satisfied that the scientific content of the research is satisfactory for an educational qualification at this level.
2. I undertake to fulfil the responsibilities of the supervisor for this study as set out in the Research Governance Framework for Health and Social Care.
3. I take responsibility for ensuring that this study is conducted in accordance with the ethical principles underlying the Declaration of Helsinki and good practice guidelines on the proper conduct of research, in conjunction with clinical supervisors as appropriate.
4. I take responsibility for ensuring that the applicant is up to date and complies with the requirements of the law and relevant guidelines relating to security and confidentiality of patient and other personal data, in conjunction with clinical supervisors as appropriate.

Academic supervisor 1

This section was signed electronically by Professor ray jones on 03/08/2015 14:43.

Job Title/Post: Professor Health Informatics
Organisation: Plymouth University
Email: ray.jones@plymouth.ac.uk

Academic supervisor 2

This section was signed electronically by Professor Heather Skirton on 03/08/2015 12:13.

Job Title/Post: Professor Of Applied Health Genetics
Organisation: Plymouth University
Email: heather.skirton@plymouth.ac.uk

Appendix 6 NHS Research Ethics approval letter October 2015



Health Research Authority South West - Exeter Research Ethics Committee

Whitefriars
Level 3
Block B
Lewins Mead
Bristol
BS1 2NT

Telephone: 0117 342 1335
Fax: 0117 342 0445

08 October 2015

Ms Selina M A Goodman
PhD Student
Plymouth University
4 Portland Villas
Plymouth University, Drakes Circus
Plymouth
PL4 8AA

Dear Ms Goodman

Study title: Investigation into the use of emails and interactive websites for the provision of information by health professionals to families at increased risk of colorectal cancer to facilitate sharing information by relatives.

REC reference: 15/SW/0250
IRAS project ID: 181861

Thank you for your email of 8 October 2015. I can confirm the REC has received the documents listed below and that these comply with the approval conditions detailed in our letter dated 16 September 2015

Documents received

The documents received were as follows:

<i>Document</i>	<i>Version</i>	<i>Date</i>
Participant consent form [consent form]	Version 2	07 October 2015
Participant information sheet (PIS) [PIS Phase 1]	Version 2	23 September 2015

Approved documents

The final list of approved documentation for the study is therefore as follows:

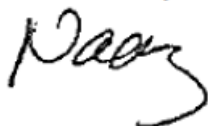
<i>Document</i>	<i>Version</i>	<i>Date</i>
Copies of advertisement materials for research participants [Clinic advert]	version 1	08 August 2015
Copies of advertisement materials for research participants [online advert]	version 1	08 August 2015
Covering letter on headed paper [covering letter]	version 1	08 August 2015
Evidence of Sponsor insurance or indemnity (non NHS Sponsors only)	Version 1	06 July 2015
GP/consultant information sheets or letters [GP letter]	version 1	08 August 2015
GP/consultant information sheets or letters [Clinical collaborator letter]	version 1	08 August 2015
Interview schedules or topic guides for participants [Interview schedule]	version 1	08 August 2015
IRAS Checklist XML [Checklist_03082015]		03 August 2015
IRAS Checklist XML [Checklist_11082015]		11 August 2015
Letter from sponsor [Sponsor letter]	version 1	05 August 2015
Letters of invitation to participant [Phase 3 invitation letter]	version 1	08 August 2015
Letters of invitation to participant [Phase 1&2 invitation letter]	version 1	08 August 2015
Non-validated questionnaire [Survey questionnaire]	Version 1	03 August 2015
Non-validated questionnaire [Survey questionnaire]	version 1	08 August 2015
Other [Online screening questions Phase 1]	Version 1	08 August 2015
Other [Eligibility criteria for clinical staff Phases 1 & 2]	Version 1	08 August 2015
Participant consent form [consent form]	Version 1	03 August 2015
Participant consent form [consent form]	version 1	08 August 2015
Participant consent form [consent form]	Version 2	07 October 2015
Participant information sheet (PIS) [PIS Phase 2]	version 1	08 August 2015
Participant information sheet (PIS) [PIS Phase 3]	version 1	08 August 2015
Participant information sheet (PIS) [PIS Phase 1]	Version 2	23 September 2015
REC Application Form [REC_Form_03082015]		03 August 2015
Referee's report or other scientific critique report [Referee's report]	Version 1	03 August 2015
Research protocol or project proposal [Protocol]	Version 1	08 August 2015
Summary CV for Chief Investigator (CI) [CV Selina Goodman]	version 1	10 July 2015
Summary CV for student [CV Selina Goodman]	version 1	10 July 2015
Summary, synopsis or diagram (flowchart) of protocol in non technical language [Flowchart phases 1 & 2]	Version 1	03 August 2015
Summary, synopsis or diagram (flowchart) of protocol in non technical language [Phase 3 flowchart]	Version 1	03 August 2015
Summary, synopsis or diagram (flowchart) of protocol in non technical language [Flowchart phases 1 & 2]	version 1	08 August 2015
Summary, synopsis or diagram (flowchart) of protocol in non technical language [Phase 3 flowchart]	version 1	08 August 2015

You should ensure that the sponsor has a copy of the final documentation for the study. It is the sponsor's responsibility to ensure that the documentation is made available to R&D offices at all participating sites.

15/SW/0250

Please quote this number on all correspondence

Yours sincerely



Naazneen Nathoo
REC Manager

E-mail: nrescommittee.southwest-exeter@nhs.net

Copy to: Dr Lisa Vickers, Plymouth Hospitals NHS Trust R&D Office

Appendix 7 NHS R&D approval letter from lead site at Plymouth



Plymouth Hospitals **NHS**
NHS Trust

Mr M Coleman
General Surgery & Colorectal Consultant
Derriford Hospital
Plymouth
PL6 8DH

Research Office
Level 2, MSCP
Bircham Park Offices
Moriaix Drive
Derriford, PL6 8BQ

18/01/2016

Tel: (01752) 432196/432197
Fax: (01752) 430919

Dear Mr Coleman

Re: NHS R&D Permission for research project

Eudract: N/A
MREC: 15/SW/0250
UKCRN: N/A
R&D ref: 15/P/157

Study title: Investigation into the use of e-mails and interactive websites for the provision of information by health professionals to families at increased risk of colorectal cancer to facilitate sharing information by relatives.

This letter confirms that the study named above has Plymouth Hospitals NHS Trust R&D permission to proceed. The governance review carried out included the following documents:

Document	Version	Document Date
Protocol	2	08 October 2015
Participant Information Sheet (PIS phase 2 & 3)	1	08 August 2015
Participant Information Sheet (PIS phase 1)	2	23 September 2015
Consent Form	2	07 October 2015
GP Letter	2	01 December 2015

Note: R&D approval extends to all documents that have received a favourable ethical opinion from the relevant Research Ethics Committee, whether or not they have been referenced in this letter.

Please note that the Trust's funding is contingent upon research studies recruiting their first patient within 30 calendar days of R&D permission. We therefore encourage you to be in a position to recruit as soon as possible.

Yours sincerely

Dr Lisa Vickers
R&D Manager



Working in partnership with the Peninsula Medical School

Chairman: Richard Crompton Chief Executive: Ann James



Appendix 8 Email correspondence of application to Health and Care Research Wales

Selina Goodman

From: Health and Care Research Wales - Research Permissions <Research-permissions@wales.nhs.uk>
Sent: 11 February 2016 11:16
To: Selina Goodman
Cc: Pam Baxter; Ray Jones; Heather Skirton
Subject: Wales - Ref IRAS 181861 - Complete R&D Application
Attachments: Clinical Research Portfolio - Information for researchers 23112015.pdf; IRAS 181861 Documents Received 11022016.docx

Dear Ms Goodman,

Re: Impact of email/websites for sharing information in families - V1 (IRAS 181861) – Complete R&D Application

Thank you for your application for NHS research permission in Wales. I am pleased to confirm that your R&D application is complete. Please find attached a list of the study-wide documents you have submitted.

What Happens Next?

- Your application will be reviewed as a multi-NHS organisation study.
- The Permissions Service will undertake the study-wide governance checks for the study and will let you know the outcome. Permissions Service staff may need to contact you if any queries arise during the review.
- NHS organisation R&D offices will undertake the local governance checks on receipt of an SSI application.
- When local governance checks are complete for a particular research site, and the study-wide governance checks are complete, the NHS organisation R&D office will issue a letter to confirm NHS research permission for that site.

Please note that you cannot commence the study at a particular site until you have received written confirmation from the NHS R&D office of NHS Research Permission for that site.

What you need to do now

Please ensure the SSI forms for sites in Wales are submitted to research-permissions@wales.nhs.uk. The following documentation is required:

- SSI Form PDF version
- SSI Form XML file
- SSI checklist (and all documents listed on the checklist)

Your research study will be assessed for eligibility for adoption onto the Clinical Research Portfolio (CRP). Please see the attached leaflet for further information.

Please contact research-permission@wales.nhs.uk should you require any further information or decide not to proceed with this study.

Kind regards,

Graham

Graham Mallaghan

Permissions Service Coordinator / Cydgysylltydd y Gwasanaeth Caniatadau

Health and Care Research Wales Support Centre / Canolfan Gymorth Ymchwil Iechyd a Gofal Cymru

Selina Goodman

From: Health and Care Research Wales – Research Permissions <Research-permissions@wales.nhs.uk>
Sent: 24 February 2016 17:11
To: Selina Goodman
Cc: ABB_R&D; Cwm Taf R&D; HDd Research-Development (Hywel Dda UHB – R&D Hywel Dda Health Board); Pam Baxter; Heather Skirton; Ray Jones
Subject: Wales – Ref IRAS 181861 – Request for further information

Dear Ms Goodman,

Re: Impact of email/websites for sharing information in families - V1 (IRAS 181861) – Study-wide Governance Review: Request for further information.

A review of your study has now been undertaken and in order to complete the study-wide governance checks, the following information is required:

- **2.1 Patient Information and Consent:**
 - o Please send us a copy of Participant Information Sheet questionnaire Phase 1 – Clean version of v2, dated 23 Sep 2015
 - o We'd be grateful for clarification over whether or not the Gillick Principle will be applied to 17 year old participants, or whether there are specific consent forms for their parents – the consent form supplied does not mention children, and there is not a specific one for young people.
- **4.3 Funding:** please send us a copy of your award letter from Bowel Cancer West.
- **5.3 Compliance with the Welsh Language Act:**
 - o In order to meet the requirements of the Welsh Language Act, we require confirmation that Participant Information Sheets and Consent Forms will be translated/ provided bilingually should this be requested by a research participant.

PCU runs a translation service to provide Welsh translations when requested by a participant. This service is available for studies sponsored by a non-Welsh organisation and is free of charge for studies adopted onto the Clinical Research Portfolio (confirmation of this would be required). Further information on the Clinical Research Portfolio is available from their e-mail: portfolio@wales.nhs.uk.

Alternatively, non-portfolio studies may use the PCU translation service, for which a charge will be made, or the Sponsor may make their own arrangements. Please confirm your agreement to use the PCU translation service and to a charge being made if translation is required, or confirm that translations will be provided by the sponsor if requested.

- o The implication of the above legal requirement for your study is that you will be required to provide copies of all printed PIS and ICF documents in the Welsh language upon request, and will need to put at statement at the top of the online ICF and PIS documents stating that a Welsh language translation will be provided upon request and that they need not immediately complete the English version online if they prefer to use a Welsh language version. Contact details should be provided at this point for requesting a translation from the research team. Please send us a copy of the text for PIS and ICF documents that will be used online – with the above statement included – as we will need to review this in order to satisfy the Welsh Language check. Please note that under the terms of the Welsh Language Act, all persons living in Wales have a legal right to request all official documents in Welsh, regardless of their abilities with the English language.

- **6.1 REC correspondence:** Please send us a copy of the Response to REC from Researcher dated 08 Oct 2015 (the email in which you respond to the REC Favourable Opinion with extra conditions dated 16 Sep 2015, that is mentioned in the REC Acknowledgement of receipt of documents as evidence of compliance dated 08 Oct 2015).

Please send your response to research-permissions@wales.nhs.uk. On receipt of your response, the study-wide governance review will be continued.

Please contact us should you require any further information or assistance.

Kind regards,

Graham

Graham Mallaghan

Permissions Service Coordinator / Cydgysylltydd y Gwasanaeth Caniatadau

Health and Care Research Wales Support Centre / Canolfan Gymorth Ymchwil Iechyd a Gofal Cymru

Tel / Ffôn: 029 2023 0457

Email / Epost: Graham.Mallaghan@wales.nhs.uk

Website / Gwefan: gov.wales/healthandcareresearch / lyw.cymru/ymchwiliachydagofal

Twitter: [@ResearchWales](https://twitter.com/ResearchWales) / [@YmchwilCymru](https://twitter.com/YmchwilCymru)

For NHS research permission applications, amendments and related correspondence, email us here: Research-permissions@wales.nhs.uk.

Please note our email address has changed, could you please update your contact lists.

Selina Goodman

From: Health and Care Research Wales - Research Permissions <Research-permissions@wales.nhs.uk>
Sent: 08 March 2016 12:08
To: Selina Goodman
Cc: Research Development (Cardiff and Vale) UHB - R & D; Heather Skirton; Ray Jones; Pam Baxter
Subject: Wales – Ref IRAS 181861 – Study-wide Governance Checks Complete
Attachments: UK Amendments leaflet – v1_1.pdf; IRAS 181861 Documents Reviewed 08032016.docx

Dear Ms Goodman,

Re: Impact of email/websites for sharing information in families - V1 (IRAS 181861) – Study-wide Governance Checks Complete for Wales

I am pleased to confirm that all the study-wide (global) governance checks for Wales have been completed for your study, however this does not constitute permission to proceed at research sites. When local governance checks are satisfied for a participating research site, the NHS organisation R&D office will issue a letter to confirm NHS research permission for that site.

The study-wide governance review was satisfied using the Research Protocol [v2, 08 Oct 2015] that received REC favourable opinion on 08 Oct 2015. Please find attached a list of study-wide documents that have been approved.

The study is currently under review for NHS permission at Cardiff and Vale University Health Board.

Please note that you cannot commence the study at a particular site until you have received written confirmation of NHS Research Permission for that site.

All amendments made during your study, after NHS research permission has been gained, should be notified to research-permissions@wales.nhs.uk. Please see the attached leaflet "UK process for handling UK study amendments" for further information.

Please contact research-permissions@wales.nhs.uk should you require any further information or assistance.

Kind regards,

Graham

Graham Mallaghan

Permissions Service Coordinator / Cydgysilydd y Gwasanaeth Caniatadau

Health and Care Research Wales Support Centre / Canolfan Gymorth Ymchwil Iechyd a Gofal Cymru

Tel / Ffôn: 029 2023 0467

Email / E-bost: Graham.Mallaghan@wales.nhs.uk

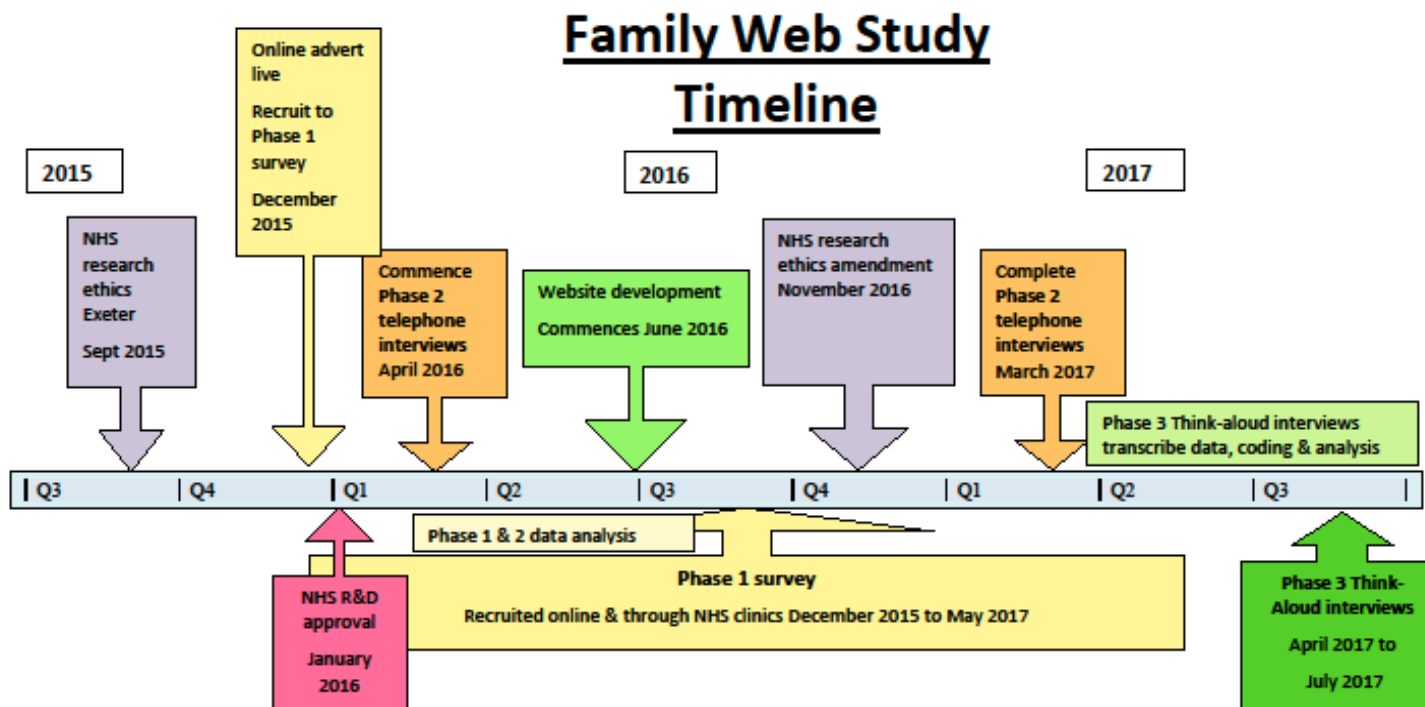
Website / Gwefan: gov.wales/healthandcarenhsresearch / hw.cymru/ymchwiliachydagofal

Twitter: [@ResearchWales](https://twitter.com/ResearchWales) / [@YmchwilCymru](https://twitter.com/YmchwilCymru)

For NHS research permission applications, amendments and related correspondence, email us here: Research-permissions@wales.nhs.uk.

Please note our email address has changed, could you please update your contact lists.

Appendix 9 Timeline of research phases submitted with application for NHS ethical approval



Appendix 10 Advertisement about the Family Web Study shown online via charity websites

Online advertisement

Family Web Study

RESEARCH
WITH
PLYMOUTH
UNIVERSITY

Are you interested in helping in our research?

Some families have an inherited vulnerability to bowel cancer which runs in the family. When someone is found to have an increased risk of cancer like this, their doctor may suggest that they tell their relatives. This is because the health advice given to one person may apply to other people in the family.

If you have an increased risk of bowel cancer in your family we are very interested in what you tell us. Your views will guide us to provide better health care to families like your own. You may have experience of sharing information in the family, or you may not; we are interested to learn from everyone's experiences.

We are conducting a survey (taking 20 -30 minutes) to gather the views of as many people as possible who have an increased risk of bowel cancer in their family.

If you are interested, please click on the link below to read an information leaflet and answer a few questions to check that you are eligible to take part. Survey questionnaires can be completed online via

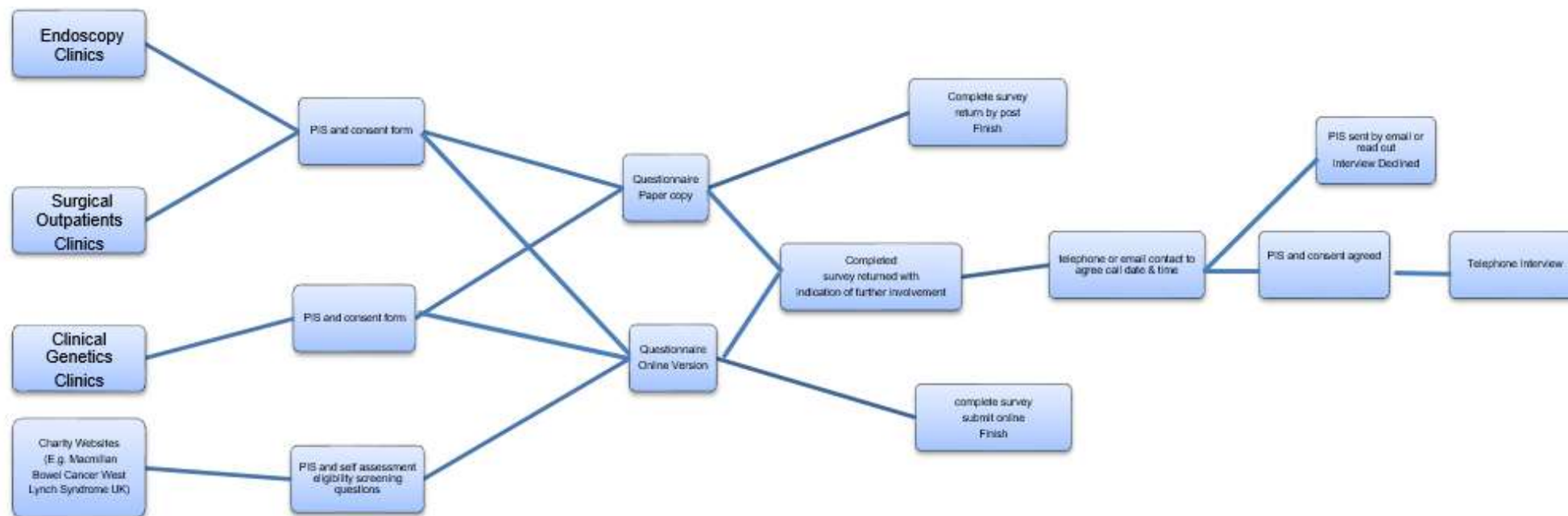
<https://www.surveymonkey.co.uk/r/familywebstudy> at any time, or if you prefer, they are available in a paper copy.



If you have questions about this survey please email: familyweb@plymouth.ac.uk text or leave a message on 07784785368.

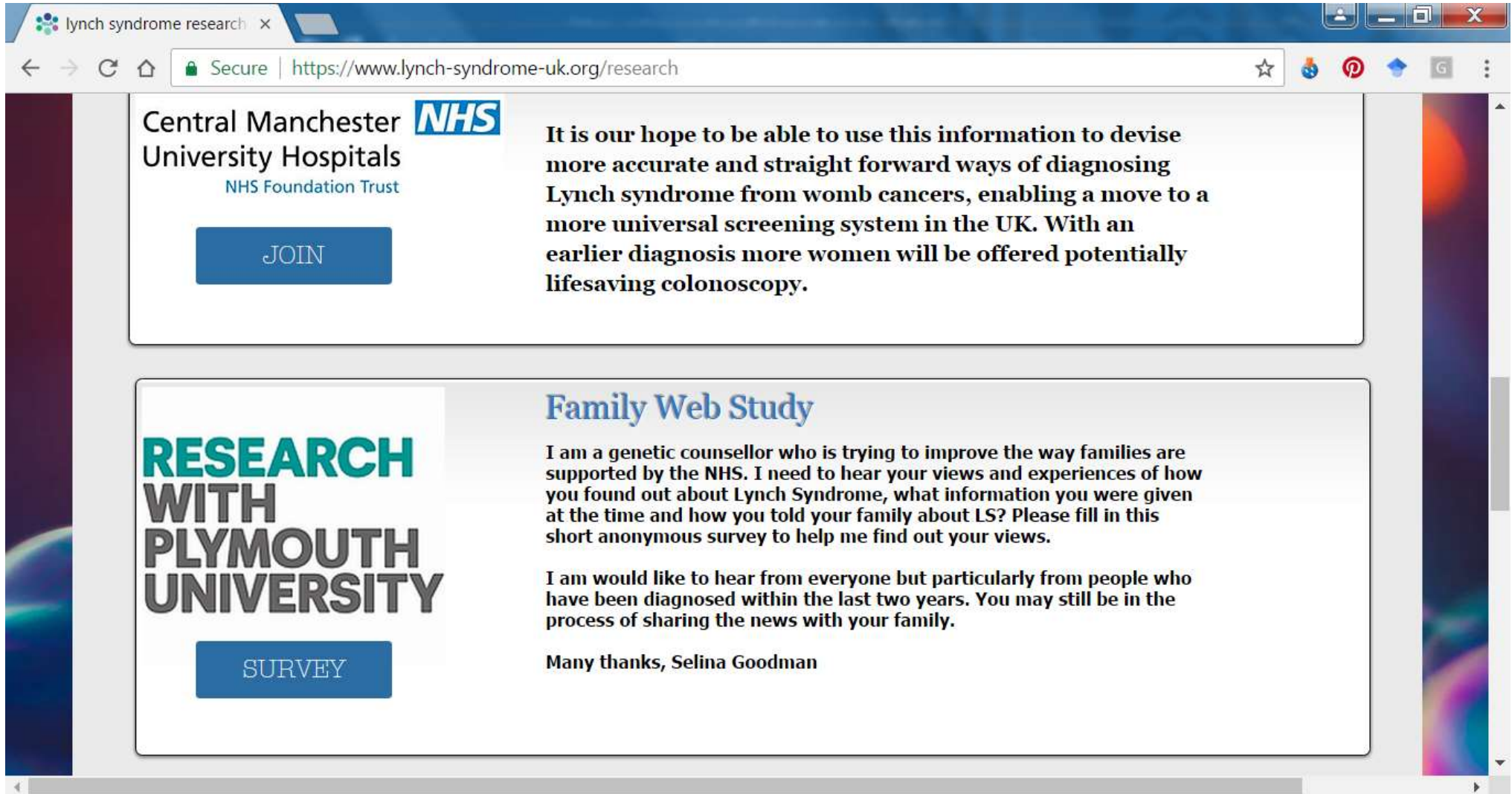
Selina Goodman, PhD student & Genetic Counsellor

Appendix 11 Flowchart of Phases 1 and 2 of the Family Web Study



Phases 1 & 2 Flowchart Family Web Study

Appendix 12 Screenshot showing the link to the online survey on the Lynch syndrome UK website



Family Web Study

**RESEARCH
WITH
PLYMOUTH
UNIVERSITY**

Are you interested in helping in our research?

Some families have an inherited vulnerability to bowel cancer which runs in the family. When someone is found to have an increased risk of cancer like this, their doctor may suggest that they tell their relatives. This is because the health advice given to one person may apply to other people in the family.

If you have an increased risk of bowel cancer in your family we are very interested in what you tell us. Your views will guide us to provide better health care to families like your own. You may have experience of sharing information in the family, or you may not; we are interested to learn from everyone's experiences.

We are conducting a survey (taking 20 -30 minutes) to gather the views of as many people as possible who have an increased risk of bowel cancer in their family.

If you are interested, please ask a member of staff for an information leaflet. Survey questionnaires are available on paper or can be completed online via <https://www.surveymonkey.co.uk/r/familywebstudy>

If you have questions about this survey please email: familyweb@plymouth.ac.uk text or leave a message on 07784785368 .



Selina Goodman

Selina Goodman, PhD student & Genetic Counsellor

Family Web Study

We are conducting a survey to find out the views of people with an increased risk of bowel cancer in their family. We want to learn from people's experiences so that we can improve support to families.

To find out if your patient would be eligible to take part in this research, please answer the questions below? Firstly, they need to be living in the UK and aged 17 or over. Patients would not be eligible if they have been diagnosed with cancer within the last 3 months or they are receiving radiotherapy or chemotherapy for cancer. This is to avoid giving any greater burden to these patients.

Then if you answer "Yes" to any of the following questions, your patient would be eligible:

- **Have they been advised to have regular bowel screening because of an increased risk of bowel cancer due to their family history?**

Yes	<input type="checkbox"/>
No	<input type="checkbox"/>

OR

- **If they have had a cancer, were they told that it was due to a genetic vulnerability?**

Yes	<input type="checkbox"/>
No	<input type="checkbox"/>

OR

- **Do they come from a family with a known genetic condition giving a risk of bowel cancer?**

Yes	<input type="checkbox"/>
No	<input type="checkbox"/>

If you have answered **Yes** to at least one of the questions above, please give your patient a participant information sheet (PIS). They would be eligible to take part in the survey and telephone interviews if they wish to. **We do not ask for any personal details or clinical information about your patient.**

If after reading the PIS your patient wishes to take part in the study they can access the survey online or they may ask you for a paper copy of the survey questionnaire.

If you have any questions, please contact us on **07784785368** or email familyweb@plymouth.ac.uk.

Thank you for your help.



Family Web Study

Study of information given to families with an increased risk of bowel cancer and their information preferences

We would like you to take part in this study

- Please read the following information carefully. Take as much time as you need to think about it. Ask us for more information if you would like it or if anything is not clear.
- Before you decide whether you wish to take part, it is important that you know why the research is being done and what it will involve.
- If you choose not to take part this will not affect your healthcare in any way. You are free to decide what to do.

We are conducting a survey to find out the views of people with an increased risk of bowel cancer in their family. We are aware that it is sometimes difficult to pass on information to relatives about the shared risk of cancer in the family. We think that information could be given in different ways but we would like people's views on this. We also want to learn from people's experiences so that we can improve support to families like yours.

Why am I being asked to take part?

You will be invited to take part in this survey if:

- You have been advised to have regular bowel screening because of an increased risk.

OR

- You have had a cancer and you were told that it was due to a genetic vulnerability.

OR

- You come from a family with a known genetic condition giving a risk of bowel cancer.

What is involved?

- You will be asked to fill in a questionnaire either online or on paper.
- This will take about 20-30 minutes but you can stop at any time and go back to it later.
- There is space in the survey if you want to give extra information.
- This is an anonymous and confidential survey.

What are the risks or benefits of taking part?

- You will be helping us to find out how to help families at increased risk of bowel cancer

- You may find it helpful to think about the situation in your own family
- You may feel anxious or upset when you think about the cancer in your family.
- You may worry about your family and what they understand about their risk.
- You may have new questions for your health professional about your risk.

What happens to my answers?

- The information you provide in the survey will be kept securely on a password protected Plymouth University computer and only seen by the researcher and her supervisors.

We hope that by providing your views you will be helping us work towards a better service for you, your family and other families like yours.

This survey has been reviewed and approved by the NHS research ethics committee and Plymouth University. There will be no financial gain to anyone involved in this research. This research is being undertaken as part of a full time PhD programme. There are no conflicts of interest to declare.

This survey is the first part in the Family Web Study. It will be followed by telephone interviews (which will be audio recorded) where we are hoping to gather more detailed information about this issue. We hope to recruit a total of 350 people to this study. The Family Web Study will continue until Spring 2018 to provide enough time to gather people's views, analyse and report the results.

After that time, all participants will be able to see a summary of the results as they will be available online through website links at Bowel Cancer West, Macmillan, Lynch Syndrome UK or Plymouth University. We can also send the results out by post or email on request.

If you would be happy to complete our survey at a time that suits you, please ask a member of staff for a survey form and reply paid envelope or go to the survey online at

<https://www.surveymonkey.co.uk/r/familywebstudy>.

For any more information about the survey or the Family Web Study please contact us.

How to contact us?

Please email: familyweb@plymouth.ac.uk, text or leave a message on 07784785368 if you have any questions.

**Thank you for taking the time
to read this**



Selina Goodman

PhD student & Registered Genetic Counsellor

*School of Nursing & Midwifery
Faculty of Health & Human
Sciences
Plymouth University, Plymouth,
PL4 8AA*

Family Web Study

<Name and address>



<date to be inserted>

Dear <name to be inserted>

Family Web Study - A Study of information given to families with an increased risk of bowel cancer and their information preferences

I am writing to you regarding the research that I am conducting. I am a full time independent student researcher working towards my PhD at Plymouth University. I am motivated to conduct this research based on my experiences working as a Registered Genetic Counsellor. I have worked for many years in the field of cancer genetics with a particular interest in bowel cancer. During that time I have come to the conclusion that better methods of information provision need to be developed, to assist and support families at risk of this disease.

That conviction is shared by many of my clinical colleagues and we believe that this research is both timely and necessary. You may have patients who would be eligible and interested in being involved, therefore I enclose a copy of the study protocol for your information. Please let me know if you have any questions about the study and whether you would consider supporting this research?

I look forward to hearing from you.

Yours faithfully

A handwritten signature in black ink, which appears to read 'Selina Goodman', is positioned below the 'Yours faithfully' text.

Selina Goodman

PhD Student & Registered Genetic Counsellor

8 Kirkby Place
School of Nursing & Midwifery
Faculty of Health & Human Sciences
Plymouth University
Plymouth
PL4 8AA

Telephone: 01752 586584

email: selina.goodman@plymouth.ac.uk

Family Web Study



Date as postmark

Dear

Re: Study of information given to families with an increased risk of bowel cancer and their information preferences

We are asking people if they would like to take part in our research study. You have been sent or given this letter because you may be eligible to take part. We would like to give you some information about what might be involved. Please read this Information Sheet which is given to all participants to read before deciding whether or not they would like to take part in this study.

This letter has been sent by your own health professional. The research team do not have your contact details or any information about you. We therefore need you to contact us if you would like to help with the study.

If you would like to be involved, and help us in our research to help families, please follow the instructions at the end of the leaflet.

Yours faithfully

A handwritten signature in black ink that reads 'S Goodman'.

Selina Goodman

PhD Student & Registered Genetic Counsellor

8 Kirkby Place
School of Nursing & Midwifery
Faculty of Health & Human Sciences
Plymouth University
Plymouth
PL4 8AA

Telephone: 07784785368

Study email: familyweb@plymouth.ac.uk



Family web study

Thank you for helping with this survey for people with an increased risk of bowel cancer in their family. Your views will guide us to provide better health care to families like your own.

We would like you to answer every question, but if you cannot answer a question, please pass over to the next one.

Firstly, please can you think back to how you first found out about the increased risk of bowel cancer in your family.

1. Who first told you that there was a risk of bowel cancer in your family?

Please tick one:

- Your doctor (General Practitioner “GP”)
- Specialist doctor (e.g. surgeon, gastroenterologist, oncologist, etc.)
- Genetics specialist (e.g. medical doctor or genetic counsellor)
- Another healthcare professional
- Your relative, can you tell us who? (e.g. mother, brother, cousin?)

- Other person, please tell us who? (e.g. friend or charity advisor?)

- Can't remember

Please answer the next questions (2, 3 & 4) only if you were told about the increased risk by a healthcare professional

2. Did you feel well supported at that time? Yes / No / Not sure

3. Please give suggestions below if you think that this could have been done

4. Are you the **first person** in your family to be told that there is an increased risk of cancer in the family?

Yes / No / Don't know

Information you received

5. Please can you tell us what written information you received when you were told about your risk of cancer? Tick all that apply

- None received
- General information about the condition
- Specific information about your family
- A copy of your family tree indicating who had cancer
- A copy of your family tree showing who could have bowel screening
- A 'Dear Relative' or 'To Whom it May Concern' letter to give to your relatives
- Other – please give details

6. Did you get the information you wanted?

- None** of what I wanted.
- Some** of what I wanted.
- Most** of what I wanted.
- All** of what I wanted.

6a. If you didn't get all the information you wanted at that time, what other information might have been helpful? Please tell us...

7. If you have found additional information about the shared risk of cancer in your family, who provided that information? Tick all that apply

- Your doctor, surgeon or other health professional
- Other relatives
- Friends
- Support group or charity meeting
- Internet website
- Social media
- Library
- Other source of information – please give details

- Did not find out more information.

8. If you found out more information via the Internet, what websites or social media were particularly helpful?

Please give details below:

Information you might like

9. Would you like to receive information in other ways? Yes / No / Don't know

- If yes, would this be
 - Via a website
 - By Email
 - Social media
 - In a follow-up appointment

We would like to know if other forms of information for patients could make it easier to share information in the family. Below, we ask you to think about other ways that your doctor or genetic counsellor could give you information. Then please can you rate how helpful these might be to you and your relatives?

10. Please indicate how helpful you think this would be for the different ways getting information by making a cross on each of the scales below:

a. A **paper leaflet** which has general information about an increased risk of bowel cancer, the implications for relatives and the screening available?

————— ————— —————

very unhelpful unhelpful helpful very helpful

b. A **secure email** which has more specific information about your increased risk, the implications for your relatives and the screening advised?

————— ————— —————

very unhelpful unhelpful helpful very helpful

c. A **password protected website** which has more specific information about your increased risk, the implications for your relatives and the screening advised ?

————— ————— —————

very unhelpful unhelpful helpful very helpful

d. A **follow-up appointment** in the hospital clinic where you are given specific information about your increased risk, the implications for your relatives and the screening advised?

————— ————— —————

very unhelpful unhelpful helpful very helpful

e. A **follow-up telephone call** where you are given specific information about your increased risk, the implications for your relatives and the screening advised?

————— ————— —————

very unhelpful unhelpful helpful very helpful

11. What issues would you like more information about? Please tick all that apply

- Talking to children
- Healthy lifestyle
- How can I help my relatives who live abroad
- How to find out more about genetic testing
- Other issues

Please give us more details about any of the issues that concern you in the box below:

Please continue....

Now it would be helpful to us to know a bit about what difficulties there may be in your family about sharing information about the increased risk of cancer. This is so we can better understand how to help and support families like yours in the future.

12. How many of **your relatives** are aware of the increased risk of cancer in the family?

So, as far as you know

- All
- Most (please tick the one which applies)
- Some
- None
- I don't know

13. If you have experienced any difficulties sharing information with your relatives about the increased risk can you tell us what those difficulties were?

Please give details below, or pass on to the next question if there were no difficulties

14. Can you suggest ways in which your health professional could help you or your family more? Please give suggestions below, or pass on to the next question if none:

(For example: These may be ideas of ways to help you overcome difficulties with communication, they may be ideas about getting screening or advice, or they may be things that were done well that could benefit other families.)

And finally, it would be helpful to know a little about you and your circumstances so that we know that different people have given us their views

15. Have you, or to your knowledge has anyone else in the family, had a genetic test for bowel cancer genes?

Yes / No / Don't know

16. Have you ever been diagnosed with cancer yourself?

Yes, bowel cancer / Yes, another type of cancer / No cancer

17. Are you: Male / Female / prefer not to say

18. Can you tell me the highest educational or school qualification you have obtained?

Please tick one only

- GCSE/O Level/ CSE / Standard/Ordinary (O) Grade / Lower (Scotland)
- AS Level / Higher Grade/Advanced Higher (Scotland)/ Certificate of sixth year studies
- A Level / Welsh Baccalaureate / International Baccalaureate
- Nursing or other allied health professional qualification (not degree level)
- Teaching qualification (excluding PGCE)
- Diploma in higher education
- First degree level qualification including foundation degrees, graduate membership of a professional Institute, PGCE
 - University Higher Degree (e.g. MSc, PhD)
 - Other vocational qualification not yet mentioned
 - None of the above

19. What age are you? Please tick

17-19		30-34		45-49		60-64		75-79	
20-24		35-39		50-54		65-69		80-84	
25-29		40-44		55-59		70-74		85+	

Or if you prefer not to say, please tick here

20. Where do you live? Please give the first part of your postcode (e.g. PL)

Please continue....

We want to improve the service and support that is offered to families in your situation, so would like to hear all your views on this issue. If you have anything further to add, we would be grateful if you could tell us in the blank box below. For example, you may like to give us more information about the things that you found helpful, or the things that you think could have been done better.

A large, empty rectangular box with a thin black border, intended for the respondent to provide their views and feedback on the service and support.

Please continue....

Thank you very much for taking part in this survey!

As the next part of this research, we would like to interview a number of people to find out more about their experiences of sharing information in the family about the risk of cancer. We are trying to develop a better service and provide support that more closely meets the needs of families. For this we need the help of people who have experience of this situation. We would like to improve things guided by families and their ideas. We think that more information could be given by email or from special websites but we would like people's views on this.

We are aiming to interview men and women (living in England or Wales) with different experiences about this type of communication. If you are interested, we will send you more information and a consent form. Then if you are happy to take part, we can arrange a time for a telephone interview lasting around 30 -40 minutes.

It may be that not everyone is needed for interviews, but we will contact you within two months to explain whether or not you would be offered an interview. Of course anyone taking part is free to withdraw at any time if they change their mind.

Please tick here if you are interested in further supporting this research

We can send you more information and consent forms, if you would consider being interviewed in this way. Please tear off the slip below and return it to us with your preferred contact details so we can get in touch with you and give you more information.

If you have any questions about this survey, or being interviewed by telephone, please contact Selina Goodman by email at: familyweb@plymouth.ac.uk, text or leave a message on **07784785368**.

All the information we receive will remain strictly confidential.



Family web study

I would consider being interviewed by telephone, please contact me with more information about what that might involve

Name/ alias	
Email or	
Telephone	

Appendix 19 Consent form for Phase 2 and Phase 3

Family Web Study



Participant Identification Number:..... CONSENT FORM:

Study of information given to families with an increased risk of bowel cancer and their information preferences

Name of Researcher: **Selina Goodman**

Please initial each box

1. I confirm that I have read the information sheet coloured:..... (date:..... version.....) for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily.
2. I agree that if I am interviewed as part of the study, that interview can be audio recorded. I understand that this is for the purpose of analysis and any recording will be destroyed once what I say has been put into writing for analysis.
3. I understand that my participation is voluntary and that I am free to withdraw at any time without giving any reason, without my medical care or legal rights being affected.
4. (If appropriate) I understand that data collected during the study, may be looked at by individuals from regulatory authorities or from the NHS Trust, where it is relevant to my taking part in this research. I give permission for these individuals to have access to my records.
5. (If appropriate) I understand that the information collected about me will be used to support other research in the future, and may be shared anonymously with other researchers.
6. (If appropriate) I agree to my General Practitioner being informed of my participation in the study.
7. I agree to take part in the above study.

Please initial in each box

_____	_____	_____
Name of Participant	Date	Signature
_____	_____	_____
Name of Person taking consent	Date	Signature

When completed: 1 for participant; 1 for researcher site file; 1 (if appropriate) to be kept in medical notes, 1 (original) to study Cf file.

Impact of email/websites for sharing information in families (Ethics ref: 15/SW/0250)

07/10/2015 Version 2

Family Web Study

Study of information given to families with an increased risk of bowel cancer and their information preferences

We would like you to take part in this study

- Please read the following information carefully. Take as much time as you need to think about it. Ask us for more information if you would like it or if anything is not clear.
- Before you decide whether you wish to take part, it is important that you know why the research is being done and what it will involve.
- If you choose not to take part this will not affect your healthcare in any way. You are free to decide what to do.

We are interviewing people with an increased risk of bowel cancer in their family to find out their views. If you have already taken part in our survey we would like to interview you on the telephone to hear more about your experiences.

We want to interview people with a range of experiences so we are aiming to interview a mixed group of men and women with different experiences about this type of communication. This is because we want to learn as much as we can so that we can improve support to families like yours.

We are aware that it is sometimes difficult to pass on information to relatives about the shared risk of cancer in the family. We think that information could be given in different ways but we would like people's views on this. We are interested in people's opinions about what could be improved or what has been done well.

Why am I being asked to take part?

You will be eligible to take part if are an adult (17+) you live in England or Wales and:

- You have been advised to have regular bowel screening because of an increased risk.
- OR
- You have had a cancer and you were told that it was due to a genetic vulnerability.
- OR
- You come from a family with a known genetic condition giving a risk of bowel cancer.

What is involved?

- You will be asked to provide a telephone number that you are happy to be contacted on.
- You will be interviewed on the telephone for about 30-60 minutes, but the interview can be stopped at any time if you do not wish to continue.
- The interview will be recorded so that what you say can be studied and analysed later.

- These interviews are completely confidential and they will be made anonymous once they have been transcribed (recording put into writing).

What are the risks or benefits of taking part?

- You will be helping us to find out how to help families at increased risk of bowel cancer
- You may find it helpful to think about the situation in your own family
- You may feel anxious or upset when you think about the cancer in your family.
- You may worry about your family and what they understand about their risk.
- You may have new questions for your health professional about your risk.

What happens to my answers?

- The information you provide in the telephone interview will be kept securely on a password protected Plymouth University computer and only seen by the researcher and her supervisors.

We hope that by providing your views you will be helping us work towards a better service for you, your family and other families like yours.

This study has been reviewed and approved by the NHS research ethics committee and Plymouth University. There will be no financial gain to anyone involved in this research. This research is being undertaken as part of a full time PhD programme. There are no conflicts of interest to declare.

The interviews are the second part of the Family Web Study. If the results of the survey and interviews indicate that it would be helpful, we plan to create a special website for patients to provide and share information with their relatives. We hope to recruit a total of 350 people to this study. The Family Web Study will continue until Spring 2018 to provide enough time to gather people’s views, analyse and report the results.

After that time, all participants will be able to see a summary of the results as they will be available online through website links at Bowel Cancer West, Macmillan, Lynch Syndrome UK or Plymouth University. We can also send the results out by post or email on request.

If you would be happy to be interviewed on the telephone at a time that suits you, please ring or email us.

How to contact us? Please email: familyweb@plymouth.ac.uk, text or leave a message on **07784785368** if you have any questions or wish to be interviewed. We will then contact you to ask for your consent and, if you are still happy to, we will arrange a time for the interview.

Thank you for taking the time to read this



Selina Goodman

PhD student & Registered Genetic Counsellor

*School of Nursing & Midwifery
Faculty of Health & Human Sciences
Plymouth University
Plymouth
PL4 8AA*

Family Web Study

Study of information given to families with an increased risk of bowel cancer and their information preferences

Telephone interview to commence with introduction by researcher.

- Check identity (or pseudonym).
- Check PIS read by participant
- Ask if any questions relating to PIS.
- If participant wishes to proceed, read through consent form.
- Read each item and request response. Tick each item, signed & dated.
- Double check that participant is happy for interview to be recorded.

Likely interview questions re semi-structured interview:

- Experience of sharing information
 - Can you describe how you first learnt about your increased risk of bowel cancer/ risk of cancer in the family?
 - How long ago was that?
 - What information about this were you given then?
 - How were you supported at the time?
 - Can you tell me more about your experience?
- Information sources, type and method
 - What information have you felt able to share with your/other relatives?
 - Can you tell me more about that?
 - What other sources of information have you found, if any?

- How useful were they?
- What would you like changed or improved in the way information was provided to you about the diagnosis/ increased risk?
- Changes to how healthcare professionals provide information
 - We had questions on the survey about other ways of providing information, if you imagine that it was possible to give you information about the diagnosis in the family in other ways (e.g follow-up phone call, email, etc) how much difference do you think that would make?
 - Why is that?
 - What do you think you would use if it was available?
 - Leaflet, email, website, follow-up appointment, etc?
- What information is needed and topics
 - What sort of information do you think would be helpful?
 - e.g. healthy lifestyle,
 - symptoms to be alert to,
 - talking to children, etc
- Genetic testing (if there is a genetic test in the family)
 - What is the situation regarding genetic testing?
 - Have you or your family had any problems being seen in genetics? If there were problems, what improvements would you like to see?
 - Do you think that your GP / surgeon/ gastroenterologist has had all the support or information they needed?
 - (If “no”) Can you tell me more about that?

- Communication within the family
 - How do you usually contact your family (e.g. by phone, talk, email, Skype)? Can you tell me a little more about how you and your family communicate normally?
 - What if any difficulties have you had sharing information with your relatives about the diagnosis?

- Other suggestions
 - Can you suggest anything else that you think might help other families in the same situation in the future?
 - Is there anything else you would like to add?

Thanks and close. Offer to send letter which they can send to their GP. Provide contact number and email address again for future contact if needed.

Appendix 22 Research Ethics Committee favourable opinion letter to substantial amendment.



South West - Exeter Research Ethics Committee

Whitefriars
Level 3
Block B
Lewins Mead
Bristol
BS1 2NT

Please note: This is the favourable opinion of the REC only and does not allow the amendment to be implemented at NHS sites in England until the outcome of the HRA assessment has been confirmed.

09 December 2016

Ms Selina M A Goodman
PhD Student
Plymouth University
8 Kirkby Place
School of Nursing & Midwifery
Faculty of Health & Human Sciences
University of Plymouth
Plymouth
PL4 8AA

Dear Ms Goodman

Study title: Investigation into the use of emails and interactive websites for the provision of information by health professionals to families at increased risk of colorectal cancer to facilitate sharing information by relatives.

REC reference: 15/SW/0250

Amendment number: Amendment 1, 10th November 2016

Amendment date: 10 November 2016

IRAS project ID: 181861

The above amendment was reviewed by the Sub-Committee in correspondence.

Ethical opinion

The members of the Committee taking part in the review gave a favourable ethical opinion of the amendment on the basis described in the notice of amendment form and supporting documentation.

This substantial amendment was to request ethical approval for the following:

- 1.To contact those patients who volunteered to be interviewed for Phase 2 but were not, and ask if they would like to be interviewed in Phase 3.

A Research Ethics Committee established by the Health Research Authority

- 2.To extend eligibility of time since diagnosis from 1 to 2 years.
- 3.To proceed with website development with content guided by participant's responses.
- 4.To invite feedback from health professionals who engage with the website.

Approved documents

The documents reviewed and approved at the meeting were:

<i>Document</i>	<i>Version</i>	<i>Date</i>
GP/consultant information sheets or letters [Phase 3 clinical collaborator letter]	2	08 November 2016
Notice of Substantial Amendment (non-CTIMP) [AmendmentForm_ReadyForSubmission181861]	Amendment 1, 10th November 2016	10 November 2016
Other [PIS Think-Aloud interview Phase 3]	2	24 October 2016
Participant information sheet (PIS) [ScreenshotPreviewWebsite181861]	1	10 November 2016
Research protocol or project proposal [Family Web Study Protocol]	3	08 November 2016

Membership of the Committee

The members of the Committee who took part in the review are listed on the attached sheet.

Working with NHS Care Organisations

Sponsors should ensure that they notify the R&D office for the relevant NHS care organisation of this amendment in line with the terms detailed in the categorisation email issued by the lead nation for the study.

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

We are pleased to welcome researchers and R & D staff at our Research Ethics Committee members' training days – see details at <http://www.hra.nhs.uk/hra-training/>

15/SW/0250:	Please quote this number on all correspondence
--------------------	---

Yours sincerely



PP
Dr Denise Sheehan
Chair

E-mail: nrescommittee.southwest-exeter@nhs.net

Enclosures: *List of names and professions of members who took part in the review*

Copy to: *Dr Lisa Vickers, Plymouth Hospitals NHS Trust*
Ms Selina M A Goodman

A Research Ethics Committee established by the Health Research Authority

South West - Exeter Research Ethics Committee

Attendance at Sub-Committee of the REC meeting by correspondence

Committee Members:

<i>Name</i>	<i>Profession</i>	<i>Present</i>	<i>Notes</i>
Mrs Joan Ramsay	Retired Associate Director of Nursing (Women and Children)	Yes	
Dr Denise Sheehan	Consultant Oncologist	Yes	

Also in attendance:

<i>Name</i>	<i>Position (or reason for attending)</i>
Miss Frances Race	REC Assistant

Notification of Non-Substantial/Minor Amendments(s) for NHS Studies

This template

33**must only** be used to notify NHS/HSC R&D office(s) of amendments, which are **NOT** categorised as Substantial Amendments.

If you need to notify a Substantial Amendment to your study then you MUST use the appropriate Substantial Amendment form in IRAS.

1. Study Information

Full title of study:	Investigation in the use of emails and interactive websites for the provision of information by health professionals to families at increased risk of colorectal cancer to facilitate sharing information by relatives – Family Web Study.
IRAS Project ID:	181861
Sponsor Amendment Notification number:	N-SA-FHHS-181861-SG-02
Sponsor Amendment Notification date:	15 th June 2017
Details of Chief Investigator:	
Name [first name and surname]	Selina Goodman
Address:	8 Kirkby Place, Plymouth University, Drakes Circus, Plymouth
Postcode:	PL4 8AA
Contact telephone number:	01752 586584
Email address:	selina.goodman@plymouth.ac.uk
Details of Lead Sponsor:	
Name:	University of Plymouth, Drake Circus, Plymouth, Devon PL4 8AA
Sponsor Representative Contact email address:	pam.baxter@plymouth.ac.uk Tel: 01752 437326 / 07484 869104
Details of Lead Nation:	
Name of lead nation <i>delete as appropriate</i>	England
If England led is the study going through CSP? <i>delete as appropriate</i>	N/A
Name of lead R&D office:	Plymouth Hospitals NHS Trust

2. Summary of amendment(s)

This template **must only** be used to notify NHS/HSC R&D office(s) of amendments, which are **NOT** categorised as Substantial Amendments.
If you need to notify a Substantial Amendment to your study then you MUST use the appropriate Substantial Amendment form in IRAS.

No.	Brief description of amendment <i>(please enter each separate amendment in a new row)</i>	Amendment applies to <i>(delete/ list as appropriate)</i>		List relevant supporting document(s), including version numbers <i>(please ensure all referenced supporting documents are submitted with this form)</i>		R&D category of amendment <i>(category A, B, C)</i> <i>For office use only</i>
		Nation	Sites	Document	Version	
1	Amendment required to extend study end date: To continue recruitment to Phase 3 Think-Aloud interviews to complete iterative process of website development for www.familyweb.org.uk. Recruitment to this study to continue until 30 th September 2017.	England Wales	All sites All sites	All documentation remains unchanged to those already agreed by REC approval, ref: 15/SW/0250		
2						
3						
4						
5						

[Add further rows as required]

3. Declaration(s)

Declaration by Chief Investigator

- I confirm that the information in this form is accurate to the best of my knowledge and I take full responsibility for it.
- I consider that it would be reasonable for the proposed amendment(s) to be implemented.



Signature of Chief Investigator:

Print name: SELINA GOODMAN

Date: 15th June 2017

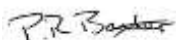
Optional Declaration by the Sponsor's Representative (as per Sponsor Guidelines)

The sponsor of an approved study is responsible for all amendments made during its conduct.

The person authorising the declaration should be authorised to do so. There is no requirement for a particular level of seniority; the sponsor's rules on delegated authority should be adhered to.

- I confirm the sponsor's support for the amendment(s) in this notification.

Signature of sponsor's representative:



Print name: Ms Pam Baxter

Post: Research Governance Specialist

Organisation: University of Plymouth

Date: 15th June 2017

Family Web Study

Study of information given to families with an increased risk of bowel cancer and their information preferences

We would like you to take part in this study

- Please read the following information carefully. Take as much time as you need to think about it. Ask us for more information if you would like it or if anything is not clear.
- Before you decide whether you wish to take part, it is important that you know why the research is being done and what it will involve.
- If you choose not to take part this will not affect your healthcare in any way. You are free to decide what to do.

We are asking people with an increased risk of bowel cancer in their family to help us test and adapt a new website that is aimed to help them. We have already carried out a survey of other people in this situation across the UK. We have also interviewed some of those people to give us more detailed information about their experiences and their information preferences.

We are aware that it is sometimes difficult to pass on information to relatives about the shared risk of cancer in the family. We think that information could be given in different ways but we would like people's views on this. We are interested in people's opinions about what could be improved or what has been done well. If you have been advised that you have an increased risk of bowel cancer due to an inherited vulnerability within the last year then we would like your help.

Why am I being asked to take part?

You will be eligible to take part if are over 17, you live in England or Wales and:

- You come from a family with a known genetic condition giving a risk of bowel cancer.
- OR
- Or you have been advised to have regular bowel screening because of an increased risk.
- OR
- Or you have had a cancer and you were told that it was due to a genetic vulnerability.

What is involved?

- You will be interviewed through an online link via your computer, laptop or tablet. This will take about 30-60 minutes, but the interview can be stopped at any time if you do not wish to continue. In the interview you will be asked to explore and comment on a new website.

- This type of interview is called a 'Think-Aloud' interview. It will be recorded so that what you say about the website can be studied and analysed later.
- You will be asked to provide a telephone number and email address that you can be contacted on. These interviews are completely confidential and they will be made anonymous once they have been transcribed (recording put into writing).

If you wish to take part in the Think-Aloud interview from a location of your choice, you will need to have access to a computer, laptop or tablet and be able to go online. Alternatively, we can arrange an interview to take place in Plymouth University if you don't mind travelling there.

What are the risks or benefits of taking part?

- You will be helping us to find out how to help families at increased risk of bowel cancer
- You may find it helpful to think about the situation in your own family
- You may feel anxious or upset when you think about the cancer in your family.
- You may worry about your family and what they understand about their risk.
- You may have new questions for your health professional about your risk.

What happens to my answers?

- The information you provide in the telephone interview will be kept securely on a password protected Plymouth University computer and only seen by the researcher and her supervisors.

We hope to recruit a total of 350 people to this study. The Family Web Study will continue until Spring 2018 to provide enough time to gather people's views, analyse and report the results. After that time, all participants will be able to see a summary of the results as they will be available online through website links at Bowel Cancer West, Macmillan, Lynch Syndrome UK or Plymouth University. We can also send the results out by post or email on request.

This study has been reviewed and approved by the NHS research ethics committee and Plymouth University. There will be no financial gain to anyone involved in this research. This research is being undertaken as part of a full time PhD programme. There are no conflicts of interest to declare.

If you would be happy to take part in a Think-Aloud interview at a time that suits you, please ring or email us.

How to contact us? Please email: familyweb@plymouth.ac.uk, text or leave a message on **07784785368** if you have any questions or wish to be interviewed. We will then contact you to ask for your consent and, if you are still happy to, we will arrange a time for the interview.

Thank you for taking the time to read this

Selina Goodman
PhD student & Registered Genetic Counsellor



*School of Nursing & Midwifery
Faculty of Health & Human Sciences
Plymouth University
Plymouth PL4 8AA*

Appendix 25 Think-Aloud Interview plan – Phase 3 Family Web Study

Send by email link to website www.familyweb.org/home prior to their meeting time. 10 minutes prior in first round of interviews.

Once GoToMeeting is launched, introduce yourself and ask if they have read the information sheet, consented and are happy to proceed?

Starting with the home page

- What are your first impressions?
- What do you like about it? (images, text, how to locate or navigate onwards)
- What don't you like?
- Why do you think that is?
- What would you change?
- Scrolling down?

Moving on to About Family Web

- What are your first impressions?
- What do you like about it? (images, text, how to locate or navigate onwards)
- What don't you like?
- Why do you think that is?
- What would you change?
- Scrolling down? out of Family Web? Or another part of the site, if so why?

Resources

Accounts

Member

See how you get on with sharing a document with a health professional

At sign up, stop recording.

After sign up Press RECORD again to record rest of interview.

What type of computer are they on? Are they on Mac or PC? What browser? What is their screen?

Appendix 26 Screenshot of Family Web homepage

Cookies | FAQ | Terms | Login

Family Web

BCW
BOWEL CANCER WEST
AWARENESS-RESEARCH-TRAINING

RESEARCH WITH PLYMOUTH UNIVERSITY

Providing Education and Tools to Share Genetic Information with Family Members

Home | About Family Web | Accounts | Resources | Contact

Share genetic information with your family members?

Existing Members Log in Here:

Username/ Email:

Password:

Remember my details [?]

[Patient Sign Up](#)

[Professional Sign Up](#)

[Forgotten Login Details](#)

Welcome to Family Web Study

For many genetic conditions, a family can have several people at risk of the same disease. Passing on that news is important but can sometimes feel difficult if you don't see your relatives very often, or you don't know how they are going to react to the news. We wanted to help make sharing information about the diagnosis easier by providing some advice and a way to share information electronically.

This website is aimed at helping people to understand about the diagnosis in their family. With clear and correct information we think relatives can make the choices that are right for them. These might be about what other information they need, if they want to go for screening or if they are interested in genetic testing.

[Click here for more information about The Family Web Study >](#)

How to Use Family Web:

Health Care Professionals

As a health professional, if you would like to set up or engage with an account for a patient, [click here to sign up now >](#)

Patients

As someone with a diagnosis, you can set up an account and share documents with your relatives. You will also be able to send a message to your Health Professional for them to access and engage with your documentation. [Click here to sign up now >](#)

Family Members

You will be contacted directly by your relative. You should receive an email with a link to log in. If it does not arrive, please check your spam filter. You may also like to read some of the information resources on this website.

Resources

We have compiled a range of freely available documents and resources which we hope will be of use to you. [Click here to view all resources >](#)

©2016 Family Web | info@familyweb.org.uk | Website design by Modern Websites

The above screenshot was provided as a preview of the 'home' page of the website to inform the Research Ethics committee regarding the application for a substantial amendment prior to conducting the Think-Aloud interviews in Phase 3.




Appendix 27 Matrix of Think-Aloud interview with 'Freya' #4 showing analysis by website area

Area of site	Positive	Negative	My reaction	Change
Overview		Had to zoom out as felt right "in there" so picture appeared too large.	Probably how the website is viewed alters this aspect. Adaptive viewing needs to be flexible for use on small screens like smartphones as well as larger monitors. Discuss banner with web developer	
Home page	<i>Yes, it looks nice, it looks good.</i> <i>Will it get an NHS badge?</i>	Not 100% sure where to go as not sure whether to click on patient or family member. Doesn't like the term 'patient' for people at risk. <i>And Bowel Cancer West, is that? What organisation is this? I just don't know...</i>	Need more explanation about what website is about.	Create graphic to illustrate function of the website.
About Family Web	OK	<i>The other thing I wondered was about the login, what is.. what happens when someone logs in? Why do people need a login?</i>		
Account information	<i>But it all looks pretty necessary, it is not as if there are big junks of text, it is just that I have become accustomed to having about three lines to read.. yes, I think it is all pretty clear</i>	<i>Quite a lot of text. Used to other websites with big words and not many of them.</i>	Typo "online online" needs to be corrected	Correct typo
Patient sign up	Apparently no problem			
Document sharing	This would be really useful to keep documents that are relevant to me and access them where ever I am. Successfully tested function of sharing documents and creating text file. <i>That's really cool!</i>	Didn't understand use of blue folder icon	Another user who doesn't understand the relevance of the blue folder icon. It's an important feature so needs to be modified.	Change blue folder icon to some words to indicate sharing files.
Member				
Resources		<i>"I think the pictures are pretty cheesy"</i>		

		<i>"Big and cheesy pictures"</i>		
Your journey				
Before diagnosis		<i>Once I am a patient I already know about this stuff</i>	Add headings that direct family members to the before diagnosis stuff	
Sharing the news		<i>I think there may be too much text, but some people like knowing what there is. Who can leave comments here?</i>	Could add instructions near comments boxes	
Living your life	<i>This is what I was particularly interested in, the prevention angle.</i>	Diagram about effects of food too complicated Put more emphasis on how healthy lifestyle can have a really important effect on you if you have an inherited vulnerability. <i>This is really interesting</i>	Add new text to top of lifestyle page to emphasise the relevance of these factors to people with genetic diagnosis.	Move or change diagram
Useful websites	<i>Oh good, you have some links</i>	Are there any counselling links? Useful to have somewhere if you are feeling distressed talk to your genetic counsellor		
Contact		Doesn't like image. The banner pictures are too big	Could add content to invite feedback to the contacts page.	
Comments		Uncertain what sort of comments are expected. Worried about people leaving inappropriate comments.	Check method of moderator to check comments.	
Not area specific	Very thoughtful throughout. Works for CRUK so very familiar with website design. Wonder if a feedback section would be useful?		Useful concept that website could act as safe storage of medical documents for access abroad or away from home.	

Appendix 28 Plymouth University press release to promote the Family Web website 30th November 2017

The screenshot shows a web browser window with the URL <https://www.plymouth.ac.uk>. The page header features the University of Plymouth logo on the left, a search bar in the center, and a 'Share' button on the right. Below the header is a 'Daily showcase' section with three featured articles:

- Article 1:** 
Your applicant journey - apply now for 2018 entry
[#undergraduate](#) [#courses](#) [#study](#)
- Article 2:** 
New website to help families spot bowel cancer risks
[#research](#) [#news](#) [#students](#)
- Article 3:** 
Will machines soon be thinking for themselves?
[#research](#) [#technology](#) [#robotics](#)


A green question mark icon is located in the bottom right corner of the showcase area.

New website to help fam x

Secure | <https://www.plymouth.ac.uk/news/new-website-to-help-families-spot-bowel-cancer-risks>


PhD student Selina Goodman explains why the website is so important:

“ In its inherited form, bowel cancer can affect people at a younger age, so this website could help to identify links much more quickly. In some families, the risk of developing the disease can be as high as 80 per cent. ”

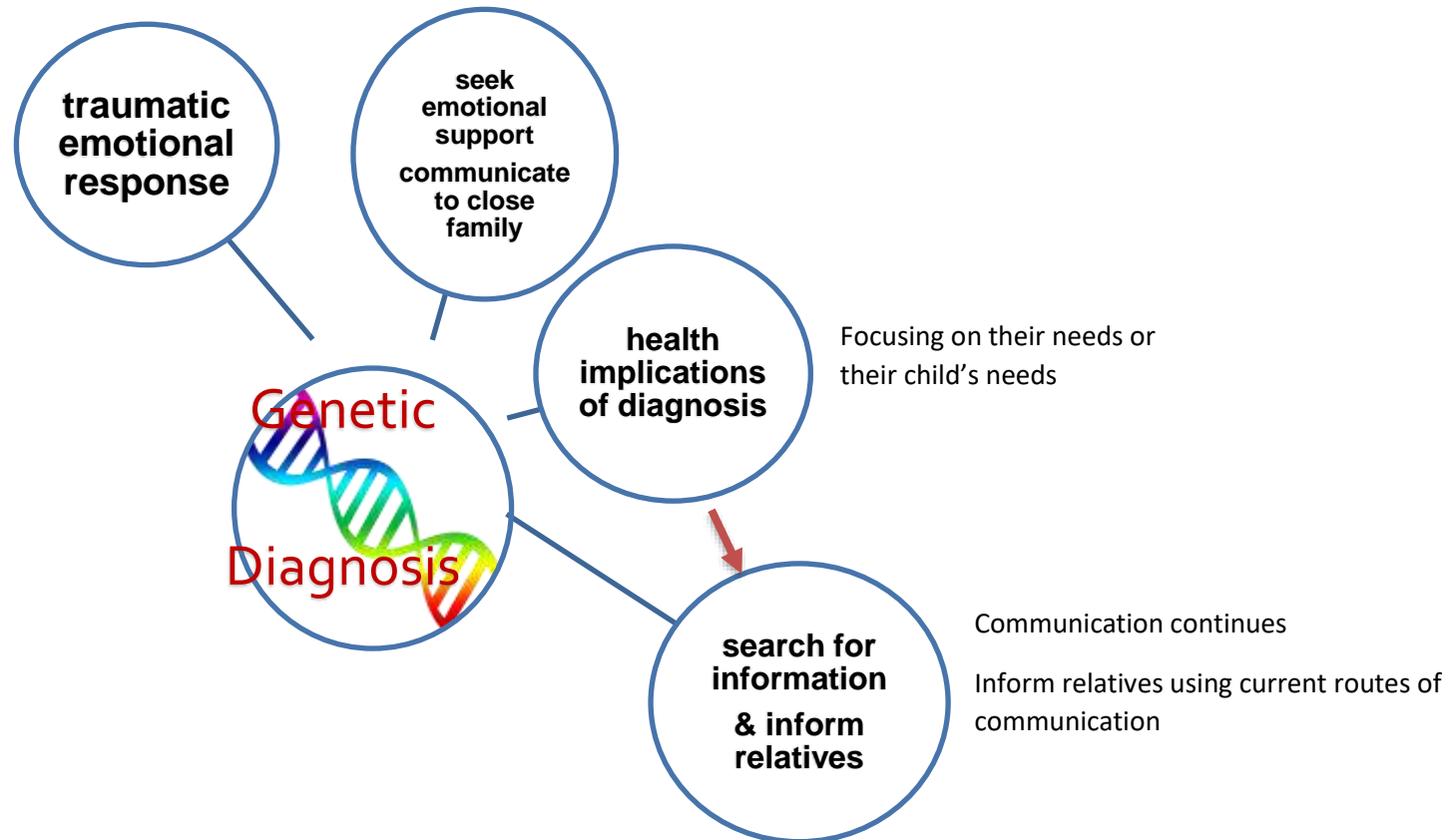


[Visit Family Web to see how families can share genetic information](#)

New research at the University of Plymouth has led to the creation a new website to help families at a high risk of bowel cancer.



Appendix 29 Process of communication developed from Forrest et al 2008 used in presentation to clinical colleagues



Representation of the process of communication with families developed from Forrest et al 2008

Forrest, L. E., et al. (2008). "Health first, genetics second: exploring families' experiences of communicating genetic information."

Communicating Genetic Diagnoses within Colorectal Cancer Families

Why is better communication with relatives needed?

- Less than half Lynch Syndrome first degree relatives sought genetic testing. (Sharaf, Myer et al. 2013)
- 20 -40% of relatives may be unaware of diagnosis (Hodgson, et al. 2014; Landsbergen, Verhaak et al. 2005).
- Relatives could benefit from CRC surveillance (Jarvinen, et al. 2009)
- CRC units are not following guidelines and so are failing to diagnose people with Lynch Syndrome (Adelson, Pannick et al. 2013).

Can communication be facilitated by email or a web-based patient portal?

First phase:

Online Survey + telephone interviews

Second Phase:

Prospective case vs control study of Intervention vs delayed intervention

Primary Endpoint:

Relative acknowledges receipt of:
Standard information (control s)
New style information (cases)

83% of UK adults use email & 86% of parents in the USA used email at least weekly to communicate with non-resident family (Rudi et al. 2015)

Patients would like email contact with their doctors (Peleg and Nazarenko 2012)

Concerns re email or websites:

- Clinicians fear email will increase their workload.
- Concern around privacy and security of content of emails in healthcare. (Sawmynaden et al. 2012)
- Data protection does not preclude sharing information with relatives
"There is a new Caldicott principle, that the duty to share personal confidential data can be as important as the duty to respect service user confidentiality"
(Caldicott Review 2013)



RESEARCH
WITH
PLYMOUTH
UNIVERSITY

Selina Goodman selina.goodman@plymouth.ac.uk

Results from a survey of UK patients at risk of bowel cancer, their experiences and information preferences.



Selina Goodman, Heather Skirton, Ray Jones
Plymouth University, Plymouth PL4 8AA UK



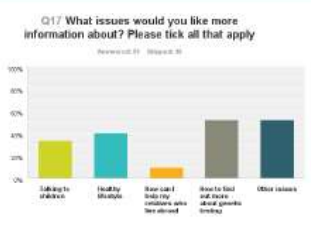
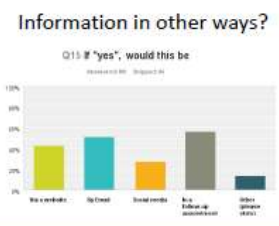
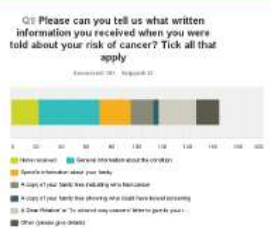
Introduction
Individuals at increased risk of familial bowel cancer are advised to have regular colonoscopy and to discuss the implications of their diagnosis with relatives. However, only a minority of relatives access screening or genetic testing (Sharaf et al 2013). This can be due to lack of information provided about the risks (Chivers Seymour et al 2010) and/or failure of effective communication within families. We are investigating the information preferences of people at risk of bowel cancer and asking for their experiences of the information provided to them at their diagnosis. The aim of this study was to find out if information was provided in an electronic format, with more tailored content, could improve sharing of information (and subsequent uptake of bowel surveillance) amongst at risk relatives. This study is in process and so we present the preliminary results of the survey below.

Methods
Using a cross-sectional survey design, a questionnaire is currently being administered in a paper copy format or online via the link: <https://www.surveymonkey.co.uk/r/familywebstudy>. Participants are eligible if they: have been advised to have regular colonoscopy on the basis of family history or are from a family with an increased risk of bowel cancer. Some may have had bowel cancer. We are recruiting patients via charity websites, clinical genetics, endoscopy clinics and colorectal clinics. Full NHS ethical approval has been obtained.

Results
To date the majority (n=84, 80%) of the 105 responses have been from women. Almost half of respondents (n= 51, 49%) have had a cancer diagnosis, mainly affecting the bowel (n= 45, 43%). The vast majority (n=98, 93%) reported that the familial mutation was known and therefore a genetic test was available in the family. Respondents were drawn from many parts of the UK and ranged in age from 20 to 74 years old, with a diverse spread of education and qualifications.

- Most people were informed of the familial risk by a health professional (n= 72, 69%) commonly by a genetic specialists(n=49, 65%) and the majority felt well supported at that time (n=52, 76%). Those informed of the familial risk by a relative were most often told by their mother (n=19, 35%).
- However 80% of the respondents indicated that they would like to receive more information through other sources (n=78, 80%).
- When participants searched for further information themselves, internet websites (n=57, 56%) were the most popular sources of information.

When asked about difficulties in sharing information about genetic risk within the family, some relatives were reported as being unwilling to talk about the issue, while others had lost contact or were not on good terms. 66 (63%) respondents had suggestions for how their health professional could help them more, including ensuring all health professionals had a better knowledge of the condition and treated them as individuals. Further information required from health professionals included risks of cancer, options for screening and preventive measures. Some reported having to fight for screening.



Discussion
While providing additional information may not directly assist all situations in which sharing of information amongst relatives is restricted, it is clear that participants feel there is a need to improve the knowledge of health professionals outside genetics and to provide a more comprehensive range of information for families at risk of familial bowel cancer.

Our results indicate that a pragmatic approach may be needed to help relatives share information. Using current technology, there is the opportunity to provide information in a format that requires little personal interaction, and could be accessed by relatives in their own time and at their own pace, which may be particularly helpful to those who are finding it difficult to deal with the information about their familial diagnosis.

Building on these data, telephone interviews of a purposive sample of respondents will guide development of a website. We propose to use and evaluate digital technology to enhance support to patients and facilitate information sharing within families. These results may be applicable to genetic conditions beyond cancer.

References:
Sharaf, R.N., et al., Uptake of Genetic Testing by Relatives of Lynch Syndrome Probands: A Systematic Review. *Clinical Gastroenterology and Hepatology*, 2013. 11(9): p. 1093-1100.
Chivers Seymour, K., et al., What facilitates or impedes family communication following genetic testing for cancer risk? A systematic review and meta-synthesis of primary qualitative research. *Journal of genetic counselling*, 2010. 19(4): p. 330-42.



Project partly funded by a grant awarded by Bowel Cancer West. No conflict of interests to declare.

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Are online personal health records useful for patients with genetic conditions to share information with family members?



Selina Goodman, Heather Skirton & Ray Jones
Plymouth University, Plymouth PL4 8AA UK

Introduction

For many genetic conditions, a family can have many people at risk of the same disease. Therefore, for some patients it may be important to share information about the diagnosis with relatives to enable testing, better understanding and improved self-management (Lucassen & Parker, 2010). This could be supported through information provided electronically to patients, but this is still not commonly done, despite widespread use of electronic information by families to share information in other situations.

Patient organised personal health records (PHRs) can provide this facility, allowing individuals to decide what they share and with whom. They provide a secure mechanism for sharing information digitally which won't be lost or destroyed, unlike paper leaflets or letters. Prior to a planned intervention study to support patients at risk of familial colorectal cancer, we looked at current usage of PHRs.

Three examples of currently available PHR platforms which could assist patients with genetic conditions:

Microsoft Health Vault



South London and Maudsley NHS Foundation Trust (SLaM) *myhealthlocker* & University Hospital Southampton NHS Foundation Trust (UHS) *My Health Record*

...which suggests patients register to:

- allowing them to co-manage their healthcare online.
- facilitate online consultations.
- Allows them and their clinicians to share information online & with relatives.

Patients Know Best (PKB) – “friendly and similar to Facebook”



Dr Mohammad Al-Ubaydli, a UK doctor who is the founder of Patients Know Best, explained: “Something like cystic fibrosis involves complex care with multiple specialists and this system will integrate that care by allowing all those involved from GPs to community nurses, local hospitals and specialist centres to access the same information.” (Prasad 2011)

This provides a facility for patients to share data with family members.

Kintalk.org – website to facilitate information sharing (University of California)



Kintalk mission statement specifies that “In an effort to reduce the burden of cancers in families with a hereditary cancer syndrome, our mission is to increase family communication and awareness of hereditary cancer syndromes.”

Users register and create an account to upload electronic files on to their password protected web space and invite relatives to access this information (Myers et al 2013).

Key attributes of successful PHRs for information exchange (Archer et al 2011): Security – maintain confidentiality and trust. Content – both clear generic information and patient specific. Functionality – ease of use in collecting data, sharing data and self-management of data. Purpose – clear, necessary, and perceived benefits. Acceptance – used by clinicians and patients, plus sustainable technology.

It is necessary for the healthcare professionals engaging with the system to reinforce the benefits for successful adoption (Nazi, 2013).

- NHS HealthSpace closed in 2012. Set up with the intention of providing a secure, free online platform within the NHS for people to store and manage their health information, its uptake had been “insufficient to justify its continued maintenance”.
- GoogleHealth withdrawn in 2012 because of “low levels of adoption” but data had to be entered manually.
- Less utilisation of PHRs in the USA with only about 7% of American adults using them in 2010, but 91 commercial PHR platforms in the USA (Jones et al 2010).
- However there has been greater adoption where there is national implementation due to government funding and infrastructure e.g. Australia, Portugal and Austria (Prey et al 2016).

Conclusion

We believe that these technologies have the potential to empower patients and their relatives if care is given to their security, content, functionality, purpose and acceptability for both patients and clinical staff. Genetic services therefore can benefit from the integration of this technology into patient care, as its use has important implications for current and future clinical practice.

References:

- Archer et al (2011) 'Personal health records: a scoping review'. *Journal of the American Medical Informatics Association*, 18 (4), pp 515-522.
- Lucassen, A. & Parker, M. (2010) 'Confidentiality and sharing genetic information with relatives'. *Lancet*, 375 (9725), pp 1507-1509.
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- Nazi (2013) 'The Personal Health Record Paradox: Health Care Professionals' Perspectives and the Information Ecology of PHR Systems in Organizational and Clinical Settings'. *Journal of Medical Internet Research*, 13 (4) pp70.
- Prasad 2011 How 'Patients Know Best' is putting your health back into your hands. (cited 2015 21.09.2015); Available from: <http://www.wired.co.uk/magazine/archive/2011/03/start/power-to-the-patient>
- Prey et al (2016) 'International perspectives on sharing clinical data with patients'. *International Journal of Medical Informatics*, 86 pp 135-141.

Family web study



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Investigation of an innovative method for facilitating communication between patients and their at risk relatives in families at an increased risk of bowel cancer: the Family Web Study



Selina Goodman¹, Ray Jones¹, Leigh Jackson² & Heather Skirton¹

¹Plymouth University, Plymouth PL4 8AA UK

²University of Exeter Medical School, Exeter EX4 4SB UK

Introduction

Relatives of people diagnosed with a genetic susceptibility to bowel cancer may also have a high lifetime risk of this cancer. Therefore, for these families it is important that information is shared about the diagnosis with relatives to enable testing, better understanding and self-management [1]. However, evidence indicates that less than half of at risk relatives access genetic testing or screening colonoscopy[2]. Information provided electronically to patients by health professionals could help communication in families, but this is still not commonly done.

Aim

To investigate whether a secure website helps families with an increased risk of bowel cancer share information.

Methods

Patients at risk of colorectal cancer were recruited online or via genetics clinics at six NHS hospitals in England and Wales. Data from a cross-sectional survey (n=286) and 14 semi-structured telephone interviews were analysed and used to guide the structure and content of the Family Web website.

Website acceptability and feasibility was tested by 12 volunteer users using Think-Aloud interviews conducted through the GoToMeeting platform. This recorded user comments alongside their interaction with the website and allowed for an iterative process of analysis and further website development.



Topics where more information was wanted; Word cloud of most frequently cited words by survey participants

Results

Most survey participants (76%) would like information to be in other formats in addition to the letter they received. A follow-up appointment was rated most desirable but communication by email or via a website were also wanted.

Issues of particular interest were:

1. Healthy lifestyle.
2. Genetic testing.
3. Talking to children.

Barriers to communication included family dynamics and a lack of adaptation to the diagnosis.

Participants welcomed the opportunity to store and share personal information on the website but desired more support from health professionals, reporting the profound effect of the diagnosis on them and their family relationships. They wanted more information on a variety of topics to support themselves and inform their relatives.



Conclusion

The website was demonstrated to be both feasible and acceptable to patients to help them share information about their diagnosis with their relatives. Health professionals contribute documents via secure links but patients decide what they share and with whom. This innovation has the potential to save lives through improving awareness of risk and access to appropriate surveillance by utilising current technology. Storing and sharing information via this website could benefit many families as the function is not specific to a condition. Further research is planned to incorporate the website into patient care as part of a trial of specialist support.

References:

1. Lucassen, A. and M. Parker, Confidentiality and sharing genetic information with relatives. *Lancet*, 2010. 375(9725): p. 1507-9.
2. Sharaf, R.N., et al., Uptake of Genetic Testing by Relatives of Lynch Syndrome Probands: A Systematic Review. *Clinical Gastroenterology and Hepatology*, 2013. 11(9): p. 1093-1100.

www.familyweb.org.uk

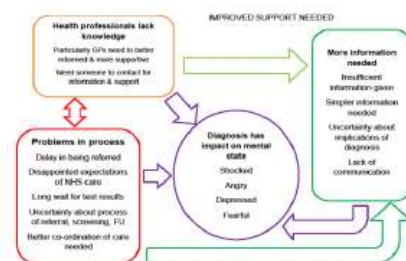


Figure representing the key themes from the survey free text responses inviting suggestions about what support could have been better



Any correspondence please contact Selina Goodman, PhD student & Registered Genetic Counsellor selina.goodman@plymouth.ac.uk

Guidelines on use of encryption to protect person identifiable and sensitive information

1. Introduction

David Nicholson, NHS Chief Executive, has directed that there should be no transfers of unencrypted person identifiable data held in electronic format across the NHS. This is the default position to ensure that patient and staff personal data are protected. Any data stored on a PC or other removable device in a non-secure area or on a portable device such as a laptop, PDA or mobile phone should also be encrypted. This is also now a requirement across all public sector organisations set by the Cabinet Secretary.

It is recognised however that this may take some time to achieve in the NHS where patient care is our highest priority. NHS bodies will need to make a local judgement on the balance of risk to patient care against risk to personal data security in determining whether use of unencrypted devices should continue as an interim measure. Where it is felt that continued reliance upon unencrypted data is necessary for the benefit of patients, the outcome of the risk assessment must be reported to the organisation's Board, so that the Board is appropriately accountable for the decision to accept data vulnerability or to curtail working practices in the interests of data security.

2. Data encryption applications

NHS Connecting for Health is already implementing a robust NHS information governance architecture that contains strong in-built encryption functionality for those core services it provides. Security services implemented within this architecture protect the flows of patient information between component parts of connected national and local applications, and automatically encrypt transmission of emailed information communicated through the NHSmail service between NHSmail endpoints. Tools are also provided within applications provided by NHS CFH for encrypting removable media as explained at Annex A.

For those other systems under local NHS organisation control, there is a requirement that the owners of those systems should consider, select and where relevant implement similar security protections that comply with expected NHS Information Governance policy, standards and legal requirements¹. Guidance on potential encryption tools is provided at Annex B.

¹ The NHS Code of Practice on Information Security can be found at http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_074142

NHS Information Governance

NHS organisations should adopt a structured approach to the identification, implementation and management of their local data encryption needs. This will normally comprise five stages:

- Perform risk assessment and identify outline data encryption needs;
- Develop a local data encryption policy;
- Establish local roles and responsibilities;
- Define how data encryption will operate within the local infrastructure and with business partners including business impact analysis;
- Implement and monitor deployed solution effectiveness.

An encryption requirements control form is provided at Annex C to supplement this guidance and will be helpful in locally developing these stages.

3. NHS Information Governance data encryption standards

For those systems under local NHS control, the Electronic Government Interface Framework (E-gif) Technical Standards Catalogue version 6.2 identifies current technical security standards, including those for data encryption that should be applied. This catalogue is available to download at http://www.govtalk.gov.uk/schemasstandards/egif_document.asp?docnum=957

In brief summary, the NHS IG data encryption algorithms currently applicable are:

- **3DES (168bit)**
- **AES 256**
- **Blowfish**

These algorithms should be used with a recommended minimum key length of 256 bits where available. This is the standard we are moving towards and whilst tactical deployments of less robust encryption are acceptable for now this should be kept under review and stronger encryption introduced when practicable.

Where data is to be transferred across the internet or by removable media it is recommended that AES256 encryption is employed. This standard is available when using applications such as PGP or WINZIP version 9. With these products the data can be put into a Self Decrypting Archive (SDA) as the software that created the archive does not need to be installed on the recipients' computer. The pass phrase for the archive must be of an appropriate length and complexity. To ensure the safety of data in transit the pass phrase should be communicated to the recipient separately from the encrypted data so that the intended recipient is the only one able to decrypt the data.

NHS Information Governance

A comprehensive technical good practice guideline overview of Approved Cryptographic Algorithms, including Secure Sockets Layer (SSL) and Transport Layer Security (TLS) has been produced by NHS Connecting for Health and is available for download at

<http://www.connectingforhealth.nhs.uk/infrasec/gpg/acs.pdf>

NHS Connecting for Health has completed the national procurement of an encryption solution for removable media and full disk encryption on behalf of the NHS. For all the latest information relating to the NHS encryption tool initiative please see the encryption tool website, at:

<http://www.connectingforhealth.nhs.uk/systemsandservices/infogov/security/encryptiontool>

Any further queries can be directed to cfh.encryptedtool@nhs.net


Digital Information Policy
Department of Health
31 January 2008

Appendix 35 Specification of Family Web website security

Family Web Study Website Security

(described by Damien, web developer from Modern Websites)

Site Security

All connections to the website are secure and use https  protocol.

That is, all communication from browser to website and back is encrypted.

The site is verified as secure by cPanel Inc with SSL (Secure Sockets Layer) certificates.

Email security

Emails from the website are sent using SSL from the website.

There is no guarantee that recipients have SSL connection set up their end. Some users may have badly set up accounts. These options are available whenever you set up the connection details for a new email account.

It is likely that all NHS health professionals have SSL security set up on their email accounts.

Website passwords are encrypted using a 512 bit hash and cannot be viewed by anybody (even me).

eg the password 'Hell0ee' is stored as

```
5eb93e3a849b3c41ee0f96430296e44e5becb6f6314b63f59e96e945d755a1b2abbf4dfda721edf93503e0c16e2f2f410b7161376f35fc44b422c0f8b8671794
```

We do not store any personal information about any user (HP or Patient) other than name, email address and the encrypted password, although their uploaded files and any connection to health professionals and family members are potentially sensitive. Patients will only enter name, relationship type and email address for their family members.

File Security

The files themselves are protected from external access by obscured location and a 10 digit alphanumeric code, stored in the database, which is never exposed on the site.

Even when files are viewed, the real location is still disguised; the contents are recreated as required on a single 'viewer' page and only if the current user has appropriate privileges. If this link is shared, it will be useless to any external person.

Once the files are saved to disk by a family member or other user, they can then be shared beyond the website, this is therefore outside the control of the system.

There are multiple tiers of access requirement within the code for each document and account.

Login

The login script uses 'pre-prepared sql statements' to avoid sql injection. (SQL means Structured Query Language).

Internal password for critical functions

Most of the critical functions within the membership system have an extra layer of security and can only be performed from the appropriate, specific page on the website.

Therefore it is impossible to perform anything like upload files, add family members etc from anywhere other than the appropriate web page on the site and when logged in as an appropriate member. Each function has it's own hidden password that is passed from page to page. This is for added protection against direct access by robot scripts, even if they did manage to gain access to the membership system itself.

Appendix 36 Factors influencing communication in families



Map of factors influencing communication and their relationships to other factors taken from qualitative analysis of Phase 2 interviews (n=14)(Chapter Seven)

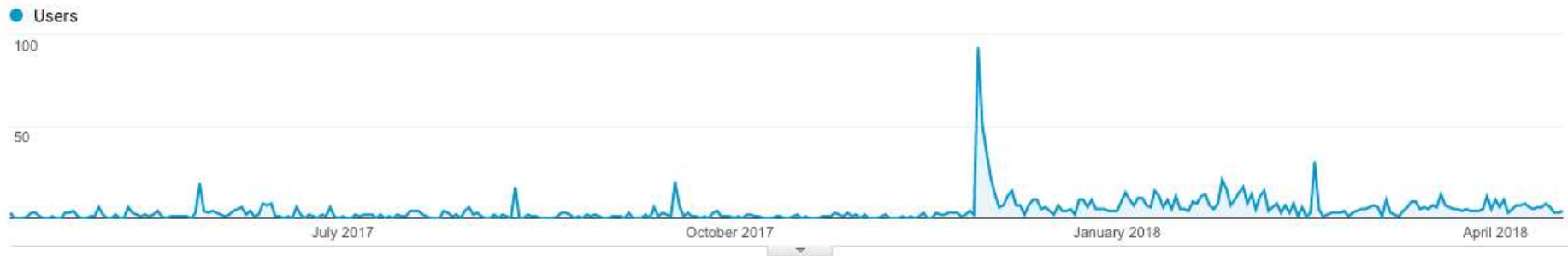
Lower cluster represents motivational factors (green), inhibiting factors (red), influences that can be either motivational or inhibiting depending on circumstance (blue) and how the method of information provision (top) feeds into these factors (orange and purple).

Appendix 37 Table of familial communication interventions

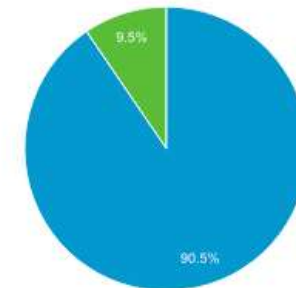
Reference by First Author; Year; origin	Study Hypothesis	Population type & size	Design & outcomes	Findings relevant to this review	Comments on findings
(Kardashian <i>et al.</i> , 2012) USA	Sharing Risk Information tool (ShaRIT) provided to BRCA carriers to support and enhance discussion of their result and its implications with relatives.	Nineteen female BRCA carriers, 10 controls, vs. 9 receiving ShaRIT tool	Case v control study. ShaRIT folder of educational material & personal info (in CD format) provided at clinic appointment when receiving test result. Control group receive standard care. Outcome: Proportion of relatives contacted and tested for BRCA. Semi structured phone interview survey at 2 months post clinic.	Outcomes primarily focussed on feasibility and acceptability of ShaRIT to patients and staff. Number of relatives told of BRCA test and seeking testing both reported by patient. Both groups shared info with first degree relatives (88% control vs 90% cases) but ShaRIT group shared info with more of their second degree relatives (38% controls vs 75% cases) but control group reported 67% of second degree relatives being tested for BRCA. None of differences reached significance.	Small pilot study with intervention now incorporated into standard care. Used both paper and electronic formats for educational tool with personalised risk information and pedigree. Control group requested more info and suggested info in an electronic format to facilitate sharing by email. Study developed a pedigree knowledge scoring system and showed association of increased knowledge with increased sharing about BRCA.
(de Geus <i>et al.</i> , 2014) (de Geus <i>et al.</i> , 2016) Netherlands	Additional telephone counselling by psychosocial workers enhances disclosure of genetic cancer info with relatives.	Two hundred and sixty four patients with relatives at risk of familial cancer (n=132 control vs n=132 intervention) randomly assigned consecutive patients.	Multi-centre RCT comparing effects of additional telephone counselling to enhance disclosure to at-risk relatives. Web based questionnaires at T1 after clinic summary letter, T2 immediately after intervention and T3 at 4 months post T1. Intervention is in directive client centred counselling style (Motivational Interviewing) with prescribed steps. All will be recorded + proportion analysed.	Design and feasibility reported but RCT ongoing – results not yet reported. n=144 received intervention to test acceptability & feasibility. Telephone counselling acceptable mode of delivery. However, psychosocial workers delivering it only asked about client understanding in 11%. In 27%, they assumed counselees had not informed their relatives. 54% counselees reported intervention helped understand which relatives needed to inform.	Intervention based on motivational interviewing so it is directive but client centred and aims to elicit behaviour change. Targets ambivalence towards informing relatives. Delivered by social work trained specialists. Further training needed to deliver intervention as more client centred.
(Hodgson <i>et al.</i> , 2016; Hodgson <i>et al.</i> , 2014) Australia	Non-directive genetic counselling telephone follow-up to support patients in disclosure to relatives.	Ninety-five patients + 10 counsellors trained in GIF intervention. 1090 possible contactable at risk relatives (intervention + control group.)	RCT comparing genetic counselling follow-up intervention with standard care to enhance family communication about genetic diagnosis. Primary outcome: proportion of relatives making contact with genetics service following intervention in each arm of trial.	Intervention group showed small increase in number of relatives seeking genetics advice 142/554 (25.6% intervention vs 20.9% in control), overall. Rarer conditions showed significant difference 39% vs 10% relatives seeking advice, so intervention more effective in this group.	Endpoint may not have captured all relatives as only logged if within Victoria, Australia and sought genetic advice within 18/12 months. Also, the standard care included a follow-up phone call. Intervention of 3 calls over 1 year may not have changed outcomes so much compared to control group as they also received one call.
(Metcalfe <i>et al.</i> , 2011) & (Plumridge <i>et al.</i> , 2012) (Eisler <i>et al.</i> , 2016) UK	Multi focus discussion groups (MFDG) with parents and children with genetic diagnosis.	12 hours over 2 days pilot MFDG co-facilitated by 2 specially trained genetic counsellors working with 6 families.	Observational study Intervention to support family communication and how to cope better with diagnosis. Developing family story/narratives about genetic condition enables parents to develop method and opportunity for communication.	Prototype MFDG tested feasibility prior to RCT. Thematic analysis of interview data found MFDG feasible and acceptable. Families involved were enthusiastic. Exercises helped facilitate communication within the close family. Advice was gained from other families on how to manage challenges. Some reported a therapeutic benefit for their own coping.	No data yet on how intervention influenced communication at home. Fears that illness in other participants would scare children were not evident in prototype MFDG. Activities enabled participants to share personal thoughts. Participants found MFDG beneficial.

(Montgomery <i>et al.</i> , 2013) USA Communication skills building session or wellness control session	Communication skills building intervention to prepare BRCA probands to explain their results with first-degree relatives. Intervention group will experience less distress sharing results than controls.	Four hundred & twenty two women (n=219 intervention vs n=203 control) but data only analysed from 137 intervention vs 112 control	RCT of communication skills intervention based on 'breaking bad news' training for clinicians. Control arm received wellness education intervention before and after test results. Both groups received standard care of 3 counselling sessions. Probands completed surveys at three points, before education session, before test result and 3 months after disclosure, plus questionnaires re anxiety and depression.	Overall, 249 probands shared test results with 838 first-degree relatives (80% of possible). No significant differences between intervention and control groups. Probands more likely to tell female than male relatives.	Communication skills intervention did not impact on sharing results with relatives. Perceived control and subjective norm variables associated with sharing test results across all subjects. High baseline intention to share genetic test results across all subjects may indicate highly motivated group.
(Bodurtha <i>et al.</i> , 2014) USA	20-minute KinFact intervention of interactive presentation reviewing risk to individual and communication tools.	Four hundred & ninety women recruited through primary care clinics (n=224 control group vs n=211 intervention completed surveys)	RCT of intervention to promote gathering & sharing family history information in the family. Intervention: 20-minute session giving tailored risk information, review of cancer risk, communication skills coaching and plan for family communication. Controls had booklet on reducing cancer risk.	Outcome measures of family history gathering & frequency of communication collected via survey at 4 time points (baseline, 1, 6 & 14 months). Intervention did promote family communication about cancer risk. Significant differences found between intervention & control groups in increasing genetic literacy, improved reported gathering family history information and sharing cancer risk information (42% intervention vs 25% controls at 14 months).	Participants were young (average age 33.4yrs) and majority black (59%). Increased gathering, sharing and frequency of communication about cancer risk with family members over time in intervention group vs little change in control group. Participants were not blinded & survey completion could have acted as a prompt or reminder of ongoing task. Authors consider adapting intervention to delivery online or link to electronic patient portals.
(Katapodi <i>et al.</i> , 2018)	Web-based intervention 'Family Gene Toolkit' designed to enhance coping, decision making & family communication	Focus groups (n=11) and 12 dyads of BRCA carriers + relative were recruited to the pilot study. 12 case dyads vs 4 control dyads.	RCT of Family Gene Toolkit educational & skills building intervention of 4 modules delivered by webinar: HBOC genetics, testing, coping with cancer, family communication over 4 weeks + FU phone call. Outcomes measure by baseline survey & FU survey in week 5.	Pilot study so no data relating to family communication. Low recruitment rate attributed to time since test result meant most relatives already tested. Timing of the intervention impacted on its usefulness. Optimal timing shortly after positive test result. Live webinar was acceptable but often difficult to fit into people's lives. Suggested tailored information delivered via an interactive website.	Statistical evaluation not given due to small sample size. Only 1 dyad in control group returned FU surveys. Participants all white, well educated & affluent. Web based patient decision aid was rated as acceptable.

Appendix 38 Google analytics data on the views made to www.familyweb.org.uk from April 2017 to April 2018



■ New Visitor ■ Returning Visitor



Appendix 39 Email sent to web developers inviting quotation for creating website.

Selina Goodman

From: Selina Goodman
Sent: 21 October 2015 10:49
To: -----
Subject: website development
Attachments: Family web planning.pptx
Dear '-----'

You were recommended to me by [REDACTED] at Southampton University. I am currently looking for a webdeveloper to build a website that will function as part of my PhD investigating the use of electronic communication methods for sharing information in families where there is an increased risk of bowel cancer. One of the main reasons this is not occurring already is due to concerns regarding data security and breach of confidentiality.

Therefore I have made some investigations into existing platforms that provide facilities for sharing data securely re health. I have spoken to people at both Southampton and Torbay NHS trusts regarding their use of the PHR platforms HealthVault and Patients Know Best (PKB) respectively, as both these allow sharing with third parties and an ability to choose what level of access different individuals have, plus they are already in use in the NHS.

Currently different NHS trusts have different rules about email and some don't allow any email traffic between health care professionals and patients. I would like to design something that is easy to use but secure enough to meet the NHS criteria. On that basis it may be that a patient needs to register with a site first and then 'invite' their health care professional to upload files to their area within the website, which the patient can then choose to share with trusted others, ie relatives. However what I would prefer is where a health professional could pre-prime a site with patient specific information and then invite the patient to access it. The advantage of the patient taking ownership of the information and who can access it, is that then the responsibility for that data confidentiality passes to the patient.

My research question is:

"Can the use of emails or interactive websites for the provision of diagnostic information by health professionals to families at increased risk of bowel cancer facilitate information sharing by relatives?"

This project aims to improve the outcomes of individuals with an increased risk of bowel cancer by seeking to increase bowel cancer awareness, screening uptake and early cancer detection through facilitating information sharing in these families, via secure electronic communication methods.

Summary: A significant proportion of people with bowel cancer will have inherited a genetic predisposition to their cancer. Other members of their families will also have an increased risk of bowel cancer if they have inherited the vulnerability too. Those individuals who have been recommended to have regular bowel screening by colonoscopy will have been advised to warn their relatives of their risk. However, evidence indicates that only a minority of relatives access screening or genetic testing.

Many different factors can impede communication, but in families with an increased risk of cancer, there are significant potential benefits to relatives if that information can help them access appropriate screening or be alert to early symptoms of disease.

Email is commonly used to communicate; although it is still used less frequently in healthcare than in other arenas. Improving technology has enabled greater data security for the use of email in healthcare. Therefore, password protected patient portals and websites could provide new opportunities for family members to share information that has been provided by their healthcare professional in a quick and secure manner.

Through this research we are seeking to develop improved methods of information provision, focussing on making information available electronically in order to facilitate sharing.

1

Phase 1 : A cross-sectional survey will be administered via websites, following genetics advice and at screening clinics to around 300 adults to find out the experiences and opinions of people at increased risk of bowel cancer about methods of sharing information.

Phase 2 : Semi-structured telephone interviews of a purposive sample of 20 respondents to the survey will seek to capture more detailed opinions about how to improve supportive information

Phase 3 : A website will be developed and tested by 30 users through 'Think-Aloud' interviews to check acceptability and uptake. More comprehensive assessment of the impact of the website is anticipated as part of post-doctoral work, but hit rates and conversion ratios will be reported within the scope of this project.

We intend to disseminate a summary of the results online through Macmillan, Lynch Syndrome UK, Bowel Cancer West, Plymouth University and participating NHS Trusts in October 2017.

Therefore in anticipation of the website development I have been investigating possible PHR platforms which are currently in use within the NHS. The specific requirements we need is that:

- Clinicians (in genetics, surgery or gastroenterology) could upload information about the diagnosis in the individual.
- Patients with a diagnosis can choose to share that information with specific relatives electronically.
- Relatives could download, print or share (on a device such as a smart phone or tablet) the information to show their GP.
- This is secure, maintaining the confidentiality of the patient.

I now have NHS research ethical approval and I am about to commence the online survey phase of my study. In terms of budget, I have recently been awarded a small research grant, which allows £4.5K for website development. I attach a short PowerPoint which gives some back additional information regarding my ideas for the website. In conclusion, I am now looking for tenders from potential collaborators/ web developers with whom I can work to set up an appropriate website, would you be interested?

Best wishes

Selina

Appendix 40 Example of an email sent to the web developer.

Selina Goodman

From: Selina Goodman
Sent: 04 November 2015 14:48
To: [REDACTED]
Cc: Ray Jones; Heather Skirton
Subject: RE: Website
Attachments: SGoodman _Family Web timeline.docx
Dear [REDACTED]

Thank you very much for your email; you just beat me to it, I was planning to email you today and I am sorry if I had gone quiet on you.

Ray and I had a long discussion last week and we agreed what we need to do in the process of developing the website (I had been jumping a few steps). Since we are trying to develop something very much in response to user (families at increased risk of bowel cancer) needs and preferences we will have to wait for the results of the online survey and initial interviews before setting up too much function in the website. However we will need to mock up (stage1) a website and present that to my current group of patient advisors, with a view to showing the resultant (stage2) mock up to the interviewees to gauge their opinion of it. I will have to do the mock up but I would welcome some input from you regarding attractive design. I will need to ask my patient advisors to look at the first mock up during December/January with a view to having something available for interviewees to comment on by February.

Then following the results of the interviews we could set up the first interactive/functioning website (stage3) for our first phase of user testing. Ray has advised me that in these early stages we do not need to have the function of the clinician uploading or populating the website with documents but we need to set up the front end and establish how many and what functions are key to the users.

We have planned six phases of user testing, with the feedback via 'think-aloud' interviews carried out remotely so that we can recruit participants from across England and Wales. All the participants in the user testing of the website will have been recruited from clinical genetics services so they will be people who are genuinely experiencing the issue of how to inform their relatives about the diagnosis in their family. We may also wish to involve some of the clinicians who would be contributing documents to the website, although that would require a change in my protocol and an application to the ethics committee. We would also like to create a short feedback survey from the website which could capture the views of users who could be either patients or their relatives.

I am currently gathering responses from my patient advisors in the process of validating the survey questionnaire prior to it going live online. I attach a copy of my timeline for the study for your information. In a nutshell, there is not much to do just yet although I would welcome a chance to talk to you, even by telephone.

Would you like to plan a time for me to call you?

Best wishes

Selina

1

From: [REDACTED]
Sent: 04 November 2015 13:16
To: Selina Goodman
Subject: Website

Hi Selina

I thought I would just jot you a message to say that I am here if you need any further information or would like to discuss your website further.

Please let me know.

I am confident that I can make you a great website that meets your requirements.

If you would like to move forward with the site, it would probably be beneficial to arrange a SKYPE chat to discuss the development in further detail.

Kind regards

[REDACTED]

--



www.modernwebsites.co.uk info@modernwebsites.co.uk

Skype: [damiensoskin](#)

[01273 509762](#)

[07752 222144](#)

Twitter: [modernwebsites](#)

Facebook: [webdesignbrighton](#)

Appendix 41 PowerPoint slides that were part of website design brief

Family web

Website design & planning ideas from Selina Goodman

WITH PLYMOUTH UNIVERSITY

Kintalk.org

- Website started in USA
- Provides information and advice
- Opportunity to share information securely

'Kintalk.org: Helping Families communicate their genetic information' (Myers, Conrad et al. 2014)

NHS Trusts who share data:

Southampton via My health record on the Microsoft 'HealthVault' platform

...and South Devon at Torbay via 'Patients Know Best' (PKB)

Patients Know Best (PKB) is a collaboration with Torbay Health Trust and Devon Partnership NHS Foundation Trust. It is a secure online platform for patients to access their health records and share information with their healthcare providers. The platform is designed to be user-friendly and accessible to all patients, including those with limited digital skills. It allows patients to view their medical history, test results, and prescriptions, and to share this information with their GP and other healthcare professionals. The platform also provides a secure way for patients to communicate with their healthcare providers, and to request appointments and referrals. Patients Know Best is a key part of the NHS Digital strategy to improve patient care and reduce costs. It is a secure and reliable platform that is used by millions of patients across the UK. Patients Know Best is a secure and reliable platform that is used by millions of patients across the UK. It is a key part of the NHS Digital strategy to improve patient care and reduce costs. It is a secure and reliable platform that is used by millions of patients across the UK.

Existing websites for online health information sharing

- <http://www.liverpoolcommunityhealth.nhs.uk/health-services/children-and-families/eredbook.htm> Liverpool
- <http://www.uhs.nhs.uk/AboutTheTrust/Myhealthrecord/Myhealthrecord.aspx> Southampton
- <https://www.myhealthlockerlondon.nhs.uk/> London
- <http://web.patientsknowbest.com/south-devon-healthcare-nhs-foundation-trust.html> Torbay
- <https://patient.emisaccess.co.uk/Account/Login?ReturnUrl=%2f> Primary Care
- <https://www.patientslikeme.com/> Patient data sharing

Family web - website for families

- Online space for sharing information
 - With relatives
 - With health professionals
 - Advice about what information may be needed
 - How to approach relatives
- Information resource on:
 - Bowel cancer
 - Surveillance recommendations
 - colonoscopy
 - Inherited cancer syndromes
 - Lynch syndrome
 - FAP
 - MutYH (MAP)
 - POLE & POLD1
 - Peutz Jeghers syndrome

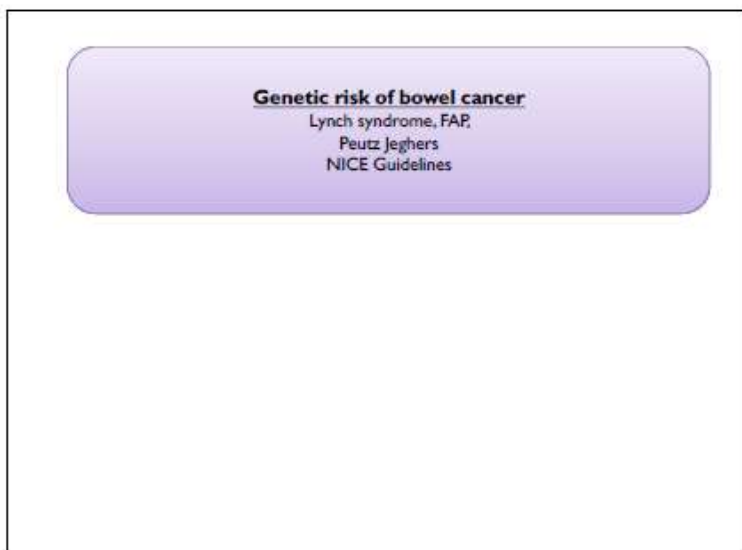
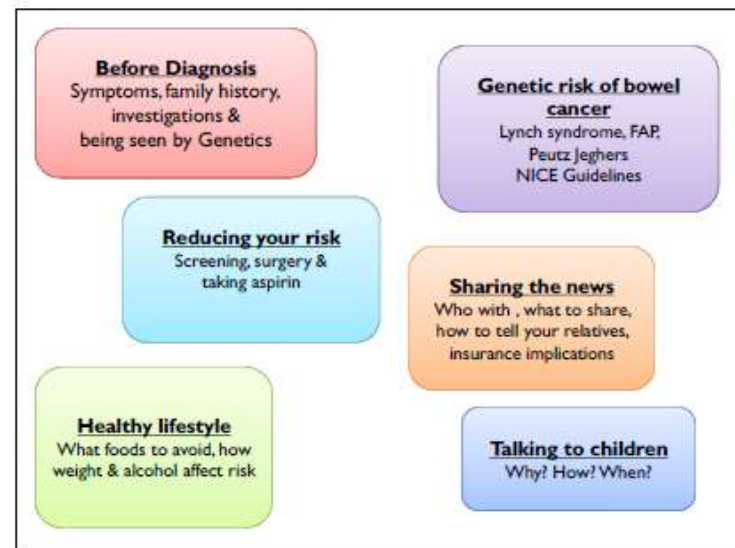
Family web - website for families

The specific requirements we need is that:

- Clinicians (in genetics, surgery or gastroenterology) could upload information about the diagnosis in the individual.
- Patients with a diagnosis can choose to share that information with specific relatives electronically.
- Relatives could download, print or share (on a device such as a smart phone or tablet) the information to show their GP.
- This is secure, maintaining the confidentiality of the patient.

Design brief

- Key intervention design objectives
 - Inform and guide users
 - Provide encouragement / motivation for sharing information
 - Practical steps to achieve information sharing
- Key distinctive features
 - Secure place to share information with trusted third parties.
 - Source of validated information
 - Links to other sources of validated information and guidance
- Video information
 - Patient stories
- Visually appealing



Living with an increased risk

Lifestyle

How to reduce risk
through what you eat
& drink

Surveillance through screening

Colonoscopy

Talking to children

Why? How? When?

Surgery

Hysterectomy or
colectomy – are they
needed

About Family Web

