UNIVERSITY OF SOUTHAMPTON

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Talking To Relatives About Genetic Testing For BRCA1/2 And Its Risk Implications: An On-going Discussion

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ABSTRACT

FACULTY OF HEALTH SCIENCES

Doctor of Philosophy

TALKING TO RELATIVES ABOUT GENENTIC TESTING FOR BRCA1/2 AND ITS RISK IMPLICATIONS: AN ON-GOING PROCESS

by Kimberley-Clair Chivers Seymour

Background: Access to genetic cancer risk information can be highly dependent on whether familial risks are discussed within the family. Despite its essential role in ensuring family members have access to genetic services, there are a number of gaps in the knowledge available on people's experiences regarding talking to their relatives about genetic testing for *BRCA1/2* and its risk implications. In particular, research to date has focused far more on with whom and why (motivations) family communication regarding genetic testing occurs, rather than when or how it is occurring.

Method: The study is qualitative in nature, employing in-depth interviews and constructing eco-maps as a method of identifying relevant family members and guiding the researcher through the family structure and relationships. These methods were chosen in line with an interpretive description methodology to ensure depth and richness in analysis and reporting of findings.

Results: The Key Findings are as follows:

1. Communication between emotionally close relatives is different to communication with emotionally distant relatives; with emotionally close family and friends it is about sharing and supporting; whereas with emotionally distant family it is about gaining and imparting information.
2. A family's engagement in communication regarding genetic testing is implicitly linked to their experiences of cancer burden, and how openly this is discussed in the family.
3. There is a lack of understanding of risks to men and their offspring based on perceptions of hereditary breast and ovarian cancer being a female disease.
4. Emotionally distant and male relatives are only contacted selectively. Those undergoing genetic testing for *BRCA1/2* are not good at identifying all at-risk family members in order to share the implications of the genetic test with them.
5. As far as the family are concerned, members do not have the right to make an informed decision to decline.
6. Plans for telling people in the future, especially children, is a cause of worry and concern for those undergoing testing and needs further support, especially in the longer term.

Conclusions: Developing interventions to help manage problems associated with family communication regarding genetic testing for cancer risk should be a top research priority, especially as the numbers of people affected by these issues is set to rise as more genes are discovered. The longitudinal view identified gives deep insight into how and when genetic testing for *BRCA1/2* are discussed within these families, allowing future interventions to be targeted where they are most helpful.

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Declaration of Authorship

I, Kimberley-Clair Chivers Seymour, declare that this thesis entitled: *Talking To Relatives About Genetic Testing For BRCA1/2 And Its Risk Implications: An On-going Discussion*, and the work presented in it, is my own and has been generated by me as the result of my own original research.

I confirm that:

1. This work was done wholly or mainly while in candidature for a research degree at this University.

2. Where any part of this thesis has previously been submitted for a degree or any other qualification at this University or any other institution, this has been clearly stated.

3. Where I have consulted the published work of others, this is always clearly attributed.

4. Where I have quoted from the work of others, the source is always given. With the exception of such quotations, this thesis is entirely my own work.

5. I have acknowledged all main sources of help.

6. Where the thesis is based on work done by myself jointly with others, I have made clear exactly what was done by others and what I have contributed myself.

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Signed:

Date:

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Glossary & Abbreviations

Autosomal dominant - a trait that is expressed whenever the gene is present and unrelated to the sex of the individual. The condition will be transmitted to children with a 50 per cent chance of an affected child for each pregnancy.

BRCA1/2 - Normal genes that can carry a mutation that may increase a person's risk of developing breast cancer.

Chromosomes - Long strings of genetic material made up of DNA and accessory proteins. The DNA contains the approximately 30,000 to 100,000 genes that make up the human genome. Human cells contain 23 pairs of chromosomes (46 in total), with mother and father each contributing one chromosome to each pair.

Diagnostic genetic testing (DGT) - Testing individuals to identify defective genes capable of causing heritable conditions after diagnosis of disease.

DNA (deoxyribonucleic acid) - a long linear polymer found in the nucleus of a cell and formed from nucleotides and shaped like a double helix; associated with the transmission of genetic information.

First-degree relative (FDR) - a close blood relative, who includes the individual's parents, full siblings, or children.

Genetic counselling (GC) - The provision of advice to families about the nature and likelihood of inherited disorders and the options available in terms of prevention and management.

Germline - Pertaining to the cells from which gametes are derived. When referring to species, the cells of the germline, unlike somatic cells, bridge the gaps between generations.

HNPCC (Hereditary nonpolyposis colorectal cancer) - An inherited disorder in which affected individuals have a higher-than-normal chance of developing colorectal cancer and certain other types of cancer, often before the age of 50.

Index case - the family member with the earliest documented case of a genetic disease.

Mutation - changes in the DNA sequence of a cell's genome and are caused by radiation, viruses, transposons and mutagenic chemicals, as well as errors that occur during meiosis or DNA replication.

Oophorectomy - the surgical removal of an ovary or ovaries.

Mastectomy - the surgical removal of one or both breasts, partially or completely.

Penetrance - the proportion of individuals carrying a particular variation of a gene (allele or genotype) that also express an associated trait (phenotype).

Predictive genetic testing (PGT)- Presymptomatic genetic testing is used to determine whether persons who have a family history of a disease, but no current symptoms, have the gene alterations associated with the disease.

Second-degree relative (SDR) - a blood relative which includes the individual's grandparents, grandchildren, aunts, uncles, nephews, nieces or half-siblings.

Somatic mutations - Alterations in DNA that occur after conception. Somatic mutations can occur in any of the cells of the body except the germ cells (sperm and egg) and therefore are not passed on to children. These alterations can (but do not always) cause cancer or other diseases.

Tertiary care - specialised consultative care, usually on referral from primary or secondary medical care personnel, by specialists working in a centre that has personnel and facilities for special investigation and treatment.

Third-degree relative (TDF) - a blood relative who includes the individual's first-cousins, great-grandparents or great grandchildren.

Uninformative - A negative test result in an individual where a clearly deleterious mutation has not been found in any family members. The genetic risk status of such an individual must be interpreted in the context of their personal and family history. Also called indeterminate and inconclusive.

Chapter 1 – Introduction

Personal Statement and Thesis Overview

Human genetics, and in particular the genetic basis of disease, has fascinated me since I first learned about Gregor Mendel and his sweet-peas in the first year of secondary school. I went on to read for a BSc in Medical Biochemistry and Human Genetics at the University of Sheffield; and then an MSc in Human Genetics (Biosciences) at the University of Leeds. Throughout my studies it became increasingly apparently to me that my future was not going to be in lab-based genetic research. I was funding my way through University by working as a Health Care Assistant in the NHS, and doing volunteer work with the Samaritans and other charities. It seems much more logical to pursue a career as a health-based researcher utilising my communication and people skills.

My research career started with my appointment as a 'Clinical Researcher' for the Kent and Medway Cancer Network. Two very dynamic consultants in the Colorectal Surgery Department of the East-Kent NHS Hospital Trust had been conducting research into the appropriateness of colonoscopy referrals. I joined their team to conduct a piece of research to assess compliance of the Surveillance Colonoscopy Waiting list across the Cancer Network. We compared referrals against the *Association of Coloproctology of Great Britain and Ireland (ACPGBI)* and the *British Society of Gastroenterology (BSG)* guidelines for colonoscopy follow up and measured the impact of adjusting referrals to be in-line with these guidelines. This study involved a large patient cohort from the Kent and Medway Cancer Network, which included seven hospitals across four NHS Hospital Trusts with an estimated population of 1.8 million. The work was

largely quantitative with a real emphasis on pragmatic research that helped patients by improving services.

In 2006, I joined the University of Southampton, Faculty of Health Sciences' *Cancer, Palliative and End of Life Care (CPELC)* Research Group; funded by a Cancer Research UK studentship. The work of the *CPELC* is focussed on carrying out research to enhance the lives of individuals affected by cancer, other life-limited conditions and those at the end of life. At the time, my experiences were based mainly on lab-based and quantitative research but I was keen to extend my expertise to incorporate knowledge of the qualitative research field.

The overall structure of this thesis takes the form of nine chapters. Chapter One sets the scene and Chapter Two synthesises and critiques the literature available to date, with a view to identifying the gaps within the literate that warrant further study. Chapter Three begins by setting out the research question, aims and objectives, and then goes on to describe the theoretical perspective and methodology. Chapter Four lays out the methods used throughout the study. Chapters Five to Eight present the findings of the research, focusing on the four key stages of talking about genetic testing and its implications with relatives that have been identified during the analysis. Finally, Chapter Nine gives a summary and discussion of the key findings, and looks at the limitations and implications of the work. Appendices and a glossary of key terms and abbreviations can be found towards the end of the thesis.

1.1 Introduction

BRCA1/2 genetic mutations increase the risk of developing cancers, in particular breast and ovarian cancers (Miki *et al.*, 1994;Wooster *et al.*, 1995). These mutations can be carried by men and women without any symptoms. Mutations can be identified through genetic testing before any cancer symptoms appear and thus an individual's risk of developing breast or ovarian cancer can be calculated (McKelvey, Jr. and Evans, 2003). Mutation carriers can then manage this risk by engaging in risk-reducing strategies or regular surveillance (Bennett *et al.*, 1999). However, genetic risk information relating to other members of the family, especially first degree relatives,[1] will be uncovered in this process (Claes *et al.*, 2003). For example, each offspring of a *BRCA1/2* mutation carrier will have a 50% chance of carrying the same mutated gene (Brody and Bowles, 1998).

Individuals tested to see if they carry a *BRCA1/2* mutation in the United Kingdom (UK) are usually encouraged to disclose test results and any potential risk information to other family members so that they, too, can consider genetic testing and risk management strategies (Lucassen, 2007). Those undergoing genetic testing may also share their results with family members for other reasons, for example, to seek emotional support or to help make decisions about risk management (Hughes *et al.*, 2002). It is hoped that educating other family members about their potential genetic risk and providing relevant information about predictive genetic testing will increase the relatives' ability to make informed decisions, thereby leading to early detection and prevention and, hopefully, to fewer breast/ovarian cancer deaths within these families (Sermijn *et al.*, 2004). The limited evidence available suggests that talking to relatives about genetic testing and genetic risks can be difficult for individuals (Green and Thomas, 1997;Julian-Reynier *et al.*, 2000b;Daly *et al.*, 2003;Wagner *et al.*, 2003;Foster *et al.*, 2004b;Van

[1] A close blood relative which includes the individual's parents, full siblings, or children.

Oostrom *et al.*, 2006;D'Agincourt-Canning, 2001). Because of this, there have been many calls for interventions to support family communication regarding genetic testing (refer to section 1.2.8).

This first part of this chapter presents an introduction to genetic testing for hereditary breast and ovarian cancer risk (sections 1.2.1-1.2.3); the importance of family communication regarding genetic testing, including the ethical issues related to non-disclosure (sections 1.2.4-1.2.7); and the calls for further research and development of interventions (section 1.2.8). The second part of the chapter will examine the literature on family communication regarding genetic testing in more detail, with a view to identifying gaps in the literature that might become the target for future interventions to enhance such communication, or where further research is needed. The focus of the literature presented is on *who* those undergoing genetic counselling and genetic testing talk to (section 1.3.1); *why* they talk to family members, and why they do not (section 1.3.2); *when* this communication occurs (section 1.3.3); and *how* the topic is discussed (section 1.3.4).

1.2 Background

1.2.1 Hereditary Breast and Ovarian Cancer

Each individual has 23 pairs of chromosomes, housing almost three billion base pairs of DNA[2] that contain about 30,000-40,000 protein-coding genes (Weidenhammer and Tsongalis, 2005). Every person inherits two copies of each chromosome and, therefore, two copies of each gene –

[2] Deoxyribonucleic acid: the hereditary material in humans and almost all other organisms.

one copy from their mother and one copy from their father (Weidenhammer and Tsongalis, 2005). Genetic alterations within these genes, known as mutations, can cause or predispose an individual to a specific disease and can occur in two ways:

1. They are inherited from a parent - hereditary mutations or germ-line mutations; or
2. They occur in the DNA during an individual's lifetime - somatic mutations.

When a particular type of cancer affects a number of family members across several generations, especially at a younger age than is usual, it is possible that relatives share an inherited alteration in a gene which makes them genetically susceptible to that cancer (Weidenhammer and Tsongalis, 2005). Individuals, who are known to be 'at risk' for certain familial conditions, can now be tested to ascertain whether they have inherited disease-causing mutations (Claus *et al.*, 1996).

Breast cancer has been the most common cancer in the UK since 1997, with over 45,800 new cases diagnosed each year (Cancer Research UK, 2011b). Breast cancer can affect both males and females, although it is rarer in men (Cancer Research UK, 2011b). The familial nature of breast cancer has long been recognised; in fact, one of the earliest recordings of the familial nature of breast cancer can be dated back to literature from Ancient Rome to around 100 AD (Ackerknecht, 1965). Women with a mother, sister or daughter diagnosed with breast cancer have almost double the risk of being diagnosed with breast cancer themselves (Cancer Research UK, 2011b). Approximately one in every ten incidences of breast cancer can be linked to mutations in specific genes, which can be passed down through the family (Claus *et al.*, 1996).

1.2.2 The *BRCA* Genes

In the mid-1990s, two high penetrance[3] breast cancer susceptibility genes called *BRCA1* (Miki *et al.*, 1994) and *BRCA2* (Collins *et al.*, 1995;Wooster *et al.*, 1995) were discovered. Normally, the *BRCA* genes help to prevent cancer by encoding proteins that keep cells from growing abnormally (Ormiston, 1995). However, if the cell growth mechanisms are not regulated, as is the case when a *BRCA1/2* mutation is present, the cell can replicate unrestrictedly leading to tumour development (Bennett *et al.*, 1999). Genetic mutations within these genes are thought to account for approximately 5-10% of breast and ovarian cancers (Claus *et al.*, 1996).

The average lifetime risk for women in the UK general population is a one in nine chance of developing breast cancer (Cancer Research UK, 2011a) and a one in 50 chance of developing ovarian cancer (Cancer Research UK, 2011c). Around 300 cases of male breast cancer are diagnosed each year (Cancer Research UK, 2011b). Women with a mutation in the *BRCA1* gene have between a 45% and 87% risk of developing breast cancer by the age of 70 (Thompson and Easton, 2002), between a 36% and 66% risk of ovarian cancer (Thompson and Easton, 2002), and a 95% risk of developing either in their lifetime (Easton *et al.*, 1994;Ford *et al.*, 1994). Mutations in the *BRCA2* gene are associated with between a 31% and 56% risk of acquiring breast cancer, and a 2% to 19% risk of ovarian cancer (Antoniou *et al.*, 2003).

When compared to the general population risk, the evidence suggests that female carriers of *BRCA1/2* mutations are likely to develop these cancers at a younger age, often before the onset of menopause, and are at a higher risk of bilateral breast cancer (Ford *et al.*, 1998). Furthermore, a study found the risk of breast cancer in male *BRCA2* carriers from multiple case breast and/or

[3] Genetic penetrance describes the frequency, under given environmental conditions, at which a specific phenotype (observable characteristics or traits) is expressed by those individuals with a specific genotype (genetic makeup). Therefore, high penetrance genes are expressed almost irrespectively of environmental factors.

ovarian cancer families to be 80-fold higher than in the general population (Thompson and Easton, 2001).

BRCA1/2 mutations are also thought to be associated with potentially increased risk of other cancers. Thompson & Easton (2002) found that the risk of cancers at alternative sites, including the pancreas, uterine body and cervix, in *BRCA1* carriers was small. However, *BRCA2* mutations have been shown to be associated with 6% of families with moderate and high-risk pancreatic cancer (Couch *et al.*, 2007). In other studies, mutations in *BRCA2* have been found to be associated with increased risk of prostate cancer (Douglas *et al.*, 2007). Specific founder mutations have also been linked to particular ethnic groups, such as Ashkenazi Jews, and to clusters within families in the Netherlands, Iceland, Sweden and Quebec (Vezina *et al.*, 2005;Peelen *et al.*, 1997;Arason *et al.*, 1998;Einbeigi *et al.*, 2001).

Cancer susceptibility due to *BRCA1/2* mutations is inherited in an autosomal dominant pattern. This refers to the fact that only one of a pair of genes needs to be mutated to cause the cancer susceptibility. Since we have two copies of every gene (one inherited from our mother and the other from our father), any individual who has an autosomal dominant condition has a 50% chance of having a child (male or female) that will also carry the genetic mutation (Weidenhammer and Tsongalis, 2005). If a family member tests positive for a *BRCA* mutation, all first degree relatives (FDR) are at a 50% risk of carrying the same mutation, while second degree relatives (SDR) are at a 25% risk (McKelvey, Jr. and Evans, 2003). However, the majority of cancers involve complex, multi-factorial interaction between environmental and genetic components, which is not yet fully understood (Ramos and Olden, 2008). This means that, even when someone carries the mutated gene, there is no guarantee they will actually go on to develop cancer or, if they do, when this is likely to occur (Esplen *et al.*, 2001;Evans *et al.*, 2001).

1.2.3 The Role of Genetic Testing in Determining Cancer Predisposition

Genetic testing is the identification of alterations or mutations in a person's genome associated with an increased risk of disease (McKelvey, Jr. and Evans, 2003). However, it is not suitable in every case: its value is dependent on the nature of the disease being tested, the availability of an effective treatment and/or the cost and efficiency of surveillance and screening measures (Evans *et al.*, 2001). For example, an important predictor for autosomal dominant early-onset familial Alzheimer's disease is mutations in the *PSEN1* and *PSEN2 FAD* genes. However, with the unpredictable psychological consequences, risk of errors in an interpretation of mutation penetrance, as well as there being no effective prevention strategies at this time, testing is deemed by many as unethical and is not recommended (Kowalska, 2004). For conditions such as breast cancer and colorectal cancer, however, where more effective risk management and prevention strategies are available, genetic testing is far more acceptable and more readily available (Esplen *et al.*, 2001;Evans *et al.*, 2001).

BRCA1 is a large gene comprising 5,592 nucleotides that, together with the non-coding regions, spreads over about 100,000 DNA bases (Shattuck-Eidens *et al.*, 1995). To improve the sensitivity of the test, genetic testing usually starts by identifying the family-specific mutation through the testing of a family member who has already been affected with breast or ovarian cancer. This is known as 'mutation search' or 'diagnostic genetic testing' (DGT) (Lerman and Shields, 2004). Then, if a genetic mutation is found, testing can be offered to relatives unaffected by cancer in the form of 'predictive genetic testing' (PGT) as it is now simply a matter of searching for that known mutation, making it possible to accurately distinguish between mutation carriers and non-carriers (Lerman and Shields, 2004). Unlike traditional medical diagnostic testing, which defines something about a patient's current state, this testing can be carried out prior to any

symptomatic disease development to predict the future likelihood of developing a genetic-based disease or disability (Evans *et al.*, 2001).

Those who opt for diagnostic genetic testing either receive a (mutation) positive test result, where they are found to carry a mutated *BRCA* gene, or they receive an uninformative/inconclusive test result, which indicates that a genetic mutation has not been found within the *BRCA* genes but a genetic susceptibility is still suspected (Van Dijk *et al.*, 2006). Those who opt for predictive genetic testing either receive a (mutation) positive test result as proven carriers, or a (mutation) negative test result as a proven non-carrier of that mutation (Lerman and Shields, 2004). Genetic testing for *BRCA1/2* holds considerable potential for reducing morbidity and mortality of breast and ovarian cancers through accurate assessment of an individual's risk, the subsequent targeting of screening to detect cancer early, and preventative strategies, as well as having important implications for blood relatives and future generations (McKelvey, Jr. and Evans, 2003).

Figure 1 outlines the process of genetic counselling and genetic testing for *BRCA1/2* at the Genetic Service where this research was based.

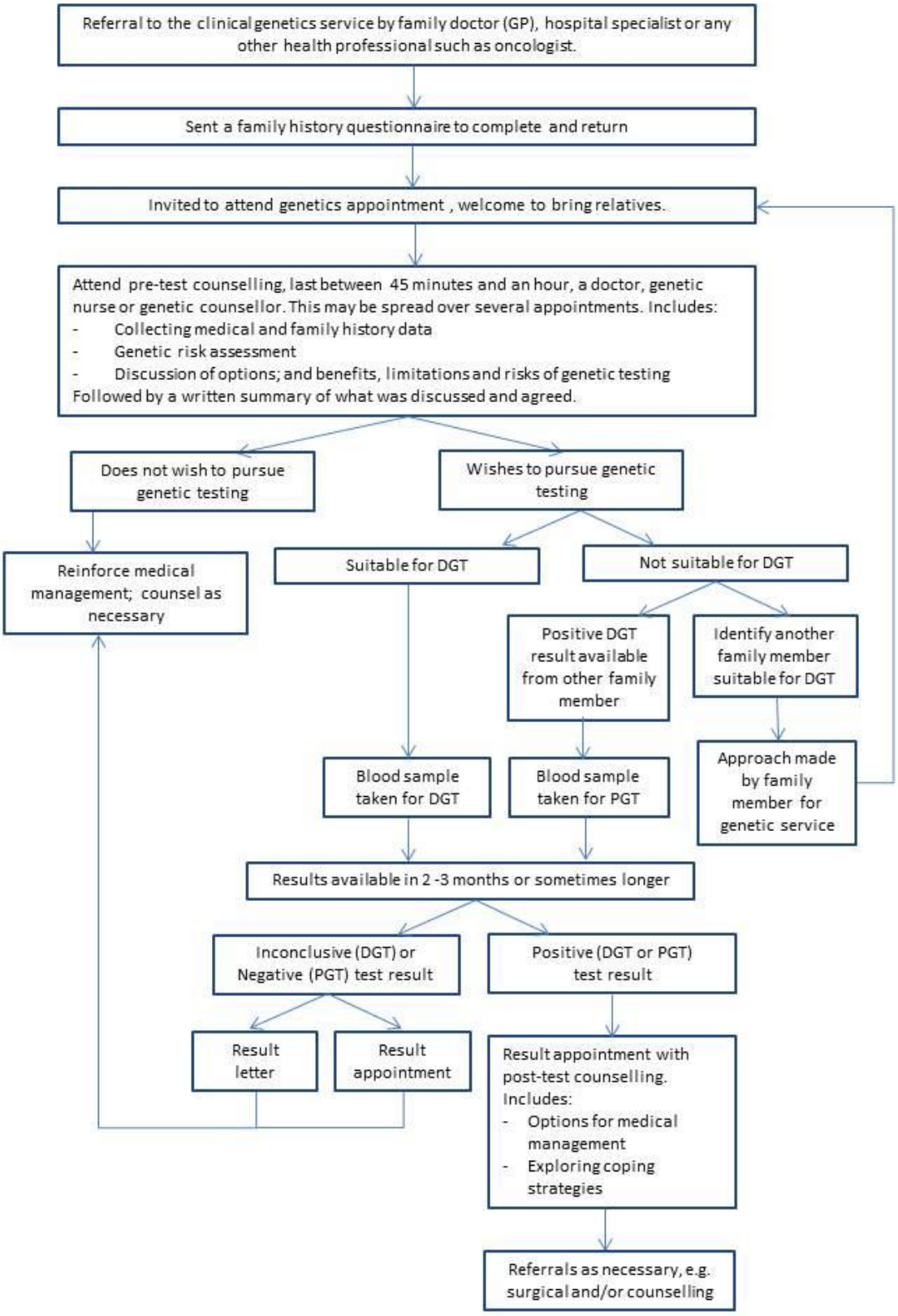

Figure 1: Process of genetic counselling and genetic testing for BRCA1/2

1.2.4 The Importance of Family Communication Regarding Genetic Testing

Family communication regarding genetic testing for cancer risk is important for three reasons:

Firstly, the information about the family history of cancer and other family members provided to the geneticist during the genetic counselling process is vitally important. The geneticist uses this information to assess whether or not there is a risk of a mutation within the family. If the information supplied is inaccurate or has gaps, then the geneticist cannot make an accurate assessment and counsel accordingly. Thus, the information the person undergoing genetic counselling is able to obtain about their family history from other family members is crucial and family communication is of key importance (Green *et al.*, 1997)

Secondly, a genetic test result does not only uncover genetic information about the individual being tested, but also brings to light potential risk for relatives (Claes *et al.*, 2003). Educating other family members about their potential genetic risk and providing relevant information about predictive genetic testing might increase the relatives' ability to make informed decisions, thereby leading to early detection and prevention and, hopefully, to fewer breast/ovarian cancer deaths within these families (Sermijn *et al.*, 2004). Due to patient confidentiality regulations, the usual practice adopted in most clinical settings is to discuss with the individual opting for genetic testing the implications of the test results for other family members; this is so they have the opportunity to contact relatives directly to disseminate the genetic risk information and to encourage at-risk relatives to seek genetic counselling (Finlay *et al.*, 2008;Croyle and Lerman, 1999;Croyle and Lerman, 1999;Patenaude *et al.*, 2006;Wilson *et al.*, 2004).

Finally, there is evidence to suggest that receiving a *BRCA1/2* mutation test result may be emotionally demanding and decisions have to be made regarding risk management; therefore, it is likely some patients will communicate their results to family members in order to gain

emotional support or advice in the next steps such as surveillance and surgical decisions (Hughes *et al.*, 2002;McGivern *et al.*, 2004). A recent study by Van Oostrom (2007)(56) found that communication regarding hereditary cancers and genetic testing within families was vital to the psychological adjustment to cancer susceptibility genetic testing. Those who were reluctant or hesitant to communicate reported significantly more psychological distress up to six months after the disclosure of their genetic test results.

1.2.5 Transmission of Genetic Information

For the most part, the literature suggests that the majority of people will want to discuss their test results within their family. This has been associated with feelings of duty, responsibility and obligation (Foster *et al.*, 2004a) (refer to section 1.3.2. for further discussion). For example, several studies have reported that parents regard the disclosure of genetic information to their children as their personal responsibility rather than the responsibility of health professionals (Claes *et al.*, 2003;Forrest *et al.*, 2003;Hallowell *et al.*, 2005a). Data presented by Green *et al.* (1997) suggests that those undergoing genetic testing recognise a duty upon themselves to inform relatives of genetic risks. Foster *et al.* (2002) also found most of the women in their study that had undergone predictive genetic testing felt they had an obligation to take action to balance their risk of developing cancer, and also had a duty to inform others and encourage others to do the same. According to Foster *et al.* (2002), women were thinking of the test as a 'family affair' rather than as an individual endeavour. In this way, women felt obliged to consider other family members in their decision to have predictive genetic testing and encouraged, or intended to encourage, other relatives to be tested.

On the other hand, disclosure of such risk information to relatives has been described as one of the most complex areas for those undergoing genetic testing for cancer risk, in particular carriers (Clarke *et al.*, 2008). They are faced with the dilemmas of deciding who, what, when, how, and indeed whether, to tell relatives (including children) about their genetic status {Forrest, 2003 12 /id;Green, 1997 457 /id;Hallowell, 2003 10 /id;Skirton, 1998 759 /id;Foster, 2004 427 /id;Hallowell, 2005 1 /id}. The literature suggests individuals can find the process of disclosing test results and potential risk information to their families difficult and that it can cause them considerable anxiety (Foster *et al.*, 2004a;Green and Thomas, 1997;Julian-Reynier *et al.*, 2000b;Daly *et al.*, 2003;Wagner *et al.*, 2003;D'Agincourt-Canning, 2001;Van Oostrom *et al.*, 2006;Forrest *et al.*, 2003). In fact, according to Patenaude *et al.* (2006), given the nature of modern families and the complexity of the information itself, this communication can even be deemed impossible in some circumstances.

1.2.6 Non-disclosure

The literature suggests that relevant genetic information may be withheld from family members, especially more distant relatives, by others in the family (Forrest *et al.*, 2003;Keenan *et al.*, 2005;Wilson *et al.*, 2004). Reasons for not disseminating to certain relatives given in the literature may be described as either *personal* or *disengaged*. For example, personal reasons for not communicating with a particular relative may include feeling concerned that the news of a *BRCA1/2* mutation would alarm or upset others. An example of this would be a woman in one study who reported deliberately lying to her family about her positive mutation status in order to prevent her father from the guilt of having passed the mutation to her (Loud *et al.*, 2006). Such reasons present a dilemma for those undergoing genetic testing between wanting to provide potentially life-saving information to their relatives but without causing any emotional

harm or upset (Hallowell *et al.*, 2003;Hallowell *et al.*, 2005a;Hallowell *et al.*, 2005b;Hughes *et al.*, 2002;Bradbury *et al.*, 2007;Green *et al.*, 1997).

Conversely, disengaged reasons may include: not personally knowing someone, contact difficulties, and not feeling emotionally close to them (McGivern *et al.*, 2004;Claes *et al.*, 2003;Daly *et al.*, 2001;Forrest *et al.*, 2003;Green and Thomas, 1997;Hughes *et al.*, 2002;MacDonald *et al.*, 2007). Factors such as family rifts and tensions, divorce, separation and adoption are likely to exacerbate the reasons for non-disclosure (Forrest *et al.*, 2003;Green *et al.*, 1997). As will be discussed in 1.3.1, there is a substantial amount of evidence that says individuals are significantly more likely to communicate with first-degree relatives and those they feel emotionally close to. However, withholding relevant genetic information to a potentially at-risk relative may result in them being denied the opportunity to seek medical advice when they would have chosen to do so (Keenan *et al.*, 2005).

1.2.7 Ethical Issues for Genetic Health Care Professionals

The fact that the content of your genes may directly affect your mother, father, brother, sister and your children is a relatively new concept in medicine and carries a new set of ethical dilemmas (McKelvey, Jr. and Evans, 2003). As Kenen *et al.* (2004b) state: 'The health care profession's code includes the ethical principles of beneficence - to do good - and nonmaleficence - to do no harm. The professionals in the cancer genetic clinic sometimes have to walk a fine line between these two ethical obligations- to see that relatives of the client receive information about their possibly increased familial risk of developing breast/ovarian cancer without infringing on these same individuals' rights to privacy and their right to 'not know'' (p. 343).

In principle, confidentiality agreements prevent clinicians from disclosing to any relatives health information that becomes apparent in the course of a patient's diagnosis or treatment. However, knowing that access to cancer risk information can be highly dependent on whether *BRCA1/2* test results are discussed within the family (Hughes *et al.*, 2002), does this mean they hold a moral duty to inform such a person of their risk? (Dickens *et al.*, 1996).

Respect for patient confidentiality is an essential feature of good medical practice (Lucassen and Parker, 2004). But for medical genetics, ethical and moral issues arise when defining who the 'patient' is. Practising genetic medicine by definition involves families. Genetic practitioners hold patient confidentiality with the individual they are testing (Dickens *et al.*, 1996;Hallowell *et al.*, 2005a). However, in the process of treating that individual, they are likely to uncover genetic risk information relating to other members of the biological kinship, especially first degree relatives (D'Agincourt-Canning, 2001). This can raise challenges for clinicians in practice. Lucassen *et al.* (2007) argue that patients have a right to privacy, but this is not the same as sole ownership of information.

For many genetic practitioners, difficulties arise when genetic testing of one family member reveals something about the genetic code held by others. If practitioners opt to take an 'individual ownership' stance regarding genetic information, as is traditional in western medicine, risk information may not be passed on to relevant family members, thus potentially causing harm. Alternatively, the 'joint account mode' argues that, since genetic information is shared, confidentiality does not hold in the same way and information should be available to all account holders; in other words, all relevant family members, unless there is a good reason to do otherwise (Lucassen, 2007).

In 1993, UK Nuffield Council on Bioethics(73) recommended that, if genetic counsellors are unable to persuade those undergoing genetic testing to share important information with other family members for whom there may be serious implications, they should be free to override the individuals' desire for confidentiality (Nuffield Council on Bioethics, 1993). Richards and Green (1996) argue that such a principle runs the risk of bringing both genetic counselling and medical confidentiality into disrepute, as well as failing to take family processes into account. It cannot be assumed that family members necessarily want to receive that information (Richards and Green, 1996). The House of Commons Science and Technology Committee's report also rejected the Nuffield Council's conclusions: 'If counselling cannot persuade someone to consent to share information with their relatives the individual's decision to withhold information should be paramount' (paragraph 228) (House of Commons Science and Technology Committee, 1995).

Individuals who are reluctant to pass on information to relatives may be 'persuaded' to do so by professionals who feel that such information should be transmitted by family members (Petersen and Bunton, 2002); however, these actions may be perceived as against the 'non-directive' principles that govern genetic counselling (Norrgard, 2008). Broaching how genetic information will be shared requires sensitivity and relevant communication, both on behalf of the health care professionals and within the family (Lucassen, 2007). However, when families do not agree to, or are unable to, contact others at risk, clinicians may be left knowing there are potential harms to others that might be prevented (Offit *et al.*, 2004;Patenaude *et al.*, 2006;Keeling, 2004;Julian-Reynier *et al.*, 2000a;Leung *et al.*, 2000;Harris *et al.*, 2005).

The issues with confidentiality mean talking within the family about a family history of cancer, associated risks and genetic testing, and it is very important to ensure all family members have access to genetic services (Hughes *et al.*, 2002). It can, however, be potentially difficult for concerned individuals and their relatives. As more genes are discovered, there is likely to be an

increase in the numbers of people affected by these issues and so the development of interventions to help people manage problems associated with the process is an important research priority.

1.2.8 Calls for Research

Understanding how families communicate about hereditary risk information is particularly important when considering the role of providers in enhancing family communication and in planning for genetic services (Patenaude *et al.*, 2006). Wiseman *et al.* (2010) conclude that 'genetic counselling practice could benefit from further understanding of the complex ways in which families communicate about genetic risk information in order to deliver high quality counselling services by enabling clinicians to modify and shape their messages as well as identify and raise communication issues directly with patients' (p. 701).

Early studies on family communication about genetic risk information assumed a sender-receiver model of communication (Wiseman *et al.* 2010). In other words, the individual undergoing genetic testing would simply tell all their family members of the genetic test (Wilson *et al.* 2004). Subsequent research suggests that family communication in this area is highly selective, both in respect of who is told about genetic risk and what they are told (Gaff *et al*. 2007). Much of the research surrounding family communication regarding genetic testing has been exploratory in nature[4] and/or concentrated on communications that occurred shortly after receiving the result (Finlay *et al.*, 2008;Wilson *et al.*, 2004). What is more, the focus is limited to motivations for communication, who was told, and barriers to communication (Wiseman *et al.*, 2010).

[4] Which is not surprising given the nature of the research questions which have largely been about understanding experiences of those undergoing genetic testing. .

There have been many calls for further research on how families communicate and for the development of interventions, and other ways, to support family communication regarding genetic testing (Clarke *et al.*, 2008;Crotser and Dickerson, 2010;Chivers Seymour *et al.*, 2010;Clarke *et al.*, 2005;Douglas *et al.*, 2009;D'Agincourt-Canning, 2001;Mellon *et al.*, 2006;Butow *et al.*, 2003;Claes *et al.*, 2003;Forrest *et al.*, 2010;Patenaude *et al.*, 2006;Forrest *et al.*, 2003;Gaff *et al.*, 2007a;Gaff *et al.*, 2005;Hughes *et al.*, 1999;Hughes *et al.*, 2002;Holt, 2006;Segal *et al.*, 2004;Tercyak *et al.*, 2007;Vos *et al.*, 2011;van Roosmalen *et al.*, 2004;Wagner *et al.*, 2003). For example, Foster *et al.* (2004a), like many others, suggest that women tested for *BRCA1/2* need particular support in how to communicate with relatives about testing and clarification.

According to Campbell *et al.* (2007), an understanding of the context of the research, such as the wider socio-economic background, the health service systems, the characteristics of the population, the prevalence or severity of the condition studied, and how these factors change over time, is crucial to the development and evaluation of any health care intervention. Understanding the context allows researchers to examine how a problem is caused and sustained, whether it is likely to be susceptible to intervention, and how any intervention could work. Therefore, this next section will examine the literature on family communication regarding genetic testing in more detail. The emphasis will be on who those undergoing genetic counselling and genetic testing talk to; why they talk to family members (and why not); when this communication occurs; and what is discussed. The aim is not only to learn about patterns of family communication about genetic test results, but also to determine if there are gaps in the literature where more research is needed, which might become the target for future interventions to enhance family communication regarding genetic testing for cancer risk.

1.3 Family Communication Regarding Genetic Testing Research

1.3.1 Who Do Those Undergoing Genetic Testing For *BRCA1/2* Talk To?

To date, most studies on who those undergoing genetic counselling and genetic testing talk to have focussed on communication with one particular family member, or group of family members. For example, communication with: siblings (Wagner *et al.* 2003), sisters (Hughes *et al.* 2002; Bodd *et al.* 2003); offspring (Tercyak *et al.* 2002; Clarke *et al.* 2008; Tercyak *et al*. 2001; Tercyak *et al.* 2007); young children (Bradbury *et al*. 2007); adult children (Wagner *et al*. 2003); daughters (Bodd *et al.* 2003); those offering support or information (Green *et al.* 1997); or first-degree relatives only (Sermijn *et al*. 2004; Barsevick *et al.* 2008; Patenaude *et al.* 2006; Julian-Reynier *et al*. 2000).

Studies have consistently demonstrated that family communication about genetic counselling and testing is highest with first-degree relatives (FDRs) (Koehly *et al*. 2003; Claes *et al*. 2003; Wagner *et al.* 2003; McGivern *et al.* 2004; Finlay *et al.* 2008; Blandy *et al*. 2003; Patenaude *et al*. 2006). In a retrospective study designed to examine the process of communicating a positive *BRCA1* or *BRCA2* genetic test result to male and female, first, second, and third-degree relatives, McGivern *et al.* (2004) found the proportion of informed parents, siblings, and offspring was nearly twice that of more distant relatives including nieces, nephews, aunts, uncles, grandchildren, and cousins (88% versus 45%; P = 0.02). As well as FDRs, the literature suggest people generally pass on genetic risk information to family members they feel emotionally close to (Chivers Seymour *et al.* 2010; Julian-Reynier *et al.* 2000; Hughes *et al.* 2002; Claes *et al.* 2003; Peterson *et al.* 2003). According to Patenaude *et al.* (2006), demographic, health-, and test-

related factors also predict genetic test result communication to FDRs. For example, gender and/or results status:

Gender

Research has consistently demonstrated that disclosure of *BRCA1/2* genetic test results for cancer risk is highest to female, compared to male, FDRs (Claes *et al.* 2003; Wagner *et al.* 2003; Sermijn *et al.* 2004; Barsevick *et al.* 2008; Julian-Reynier *et al.* 2000). Although the risk of developing breast and ovarian cancer risk is considerably greater for female carriers of *BRCA1/2* mutations, there are a number of cancer risks to male carriers (Douglas *et al.* 2007; Liede *et al.* 2004). There are also risk implications for the female offspring of male carriers due to the autosomal dominant patterns of inheritance (Weidenhammer and Tsongalis 2005). Patenaude *et al.* (2006) did find that brothers were told more often about a sister's *BRCA1/2* test result in families where the mutation was inherited through the paternal side of the family. They propose that this reflects a misperception that *BRCA1/2* hereditary risk differentially affects men in paternal lineage families. However, as the authors note, social reasoning rather than scientific reasoning is not uncommon in the lay understanding of genetics. For example, Richards & Ponder (1996) found that lay people thought that their stronger emotional bonds with their children meant they shared more of their genes with their children than with their siblings. Nevertheless, the gender of relatives is not a complete predictor of family communication, as shown by the fact that 14% of participants' sisters were not told at all (Patenaude *et al.* 2006).

Disclosure of genetic information is often described as a gendered activity (D'Agincourt-Canning, 2001). Women have been described as the 'kin-keepers' of genetic knowledge (Green *et al.*, 1997) and have been shown to play a greater role in communicating about inherited cancers, particularly breast cancer, compared with men (Marteau and Richards, 1996). There is evidence to suggest that women talk about family cancer more than their male relatives, and mothers are

key providers of information, even if their husband is the one at-risk (Green *et al.*, 1997;Koehly *et al.*, 2003;McAllister, 1999).

Results status

The literature indicates that tested individuals typically disclose their results to at least one relative, whether the genetic test results are positive, negative, or inconclusive (McGivern *et al.* 2004; Wagner *et al.* 2003; D'Agincourt-Canning 2001; Patenaude *et al.* 2006). Patenaude *et al.* (2006) found that, for the 273 women who completed a family communication measure four months after receiving their *BRCA1/2* genetic test result, receiving a positive versus true-negative test result was not a significant factor in the telling of FDRs, except in the telling of results to children. However, inconclusive results (variant or negative without known familial mutation) were shared less frequently than conclusive (definitively positive or negative) results, although the difference only reached significance for communication to sisters (Patenaude *et al.* 2006). In a study of 43 pairs of sisters, Hughes *et al.* (2002) also reported that women tested for *BRCA1/2* conveyed positive results to sisters more often than they conveyed inconclusive results. In addition, Claes *et al.* (2003) found distant relatives were more likely to be told about conclusive rather than inconclusive *BRCA1/2* genetic results.

Patenaude *et al.* (2006) theorise that inconclusive results are less likely to be disclosed to relatives, either because the complexity of the message increases as the conclusiveness declines, or because of a perception that telling relatives about an inconclusive result would be of little or no use to relatives. However, a more in-depth study of participants' reasons for not-disclosing would be required to confirm these theories. Koehly *et al.* (2003) suggest that family culture may play a more important role in determining whether or not discussions occur compared with mutation status. In their study using social network analysis of communication about Hereditary Nonpolyposis Colorectal Cancer (*HNPCC*) genetic testing, mutation status was not a significant

predictor of discussions about genetic testing and counselling when kinship (ties) and family functioning relationships (evaluated by the constructs of communication, cohesiveness, affective involvement, leadership, and conflict) were included in the model of analysis.

In summary, the literature suggests that communication is most likely to occur with FDR, those who are emotionally close, and female relatives, but does not explain why this is. Whilst much research on who those undergoing genetic testing for cancer risk communicate with has been published, very few studies have looked at family communication across the whole family kinship. Instead, studies tend to restrict exploration to distinct sub-groups of relatives. Those that have included communication to first, second, and third-degree relatives, such as McGivern *et al.* (2004), have imposed other restrictions, such as only including communication of positive *BRCA1/2* genetic test results.

1.3.2 Why Do Those Undergoing Genetic Testing For *BRCA1/2* Talk To Others?

Several studies have investigated people's motives for undergoing genetic testing for *BRCA1/2* and other late onset cancers, such as *HNPCC*. The literature indicates that these motives are driven by a number of factors, including: a desire to reduce anxiety and uncertainty (Metcalfe *et al.* 2000; Watson *et al.* 2004; Wroe *et al.* 1998); wanting to know if more screening tests are needed (Esplen *et al.* 2001); a desire for increased certainty of own potential risk of developing disease (Foster *et al.* 2002; Metcalfe *et al.* 2000); and understanding risk for children or other family members (Esplen *et al.* 2001; Foster *et al.* 2002; Lerman *et al.* 1995; Lynch *et al.* 1997; Dudok de Wit 1997). It should be noted that most studies examining motivation for genetic testing for cancer risk include small, self-selected and homogenous sample populations,

including predominantly white individuals. Some studies also had several participants from the same family, possibly producing family-specific effects.

That said, pursuing genetic testing with the expressed interest of learning about the potential risk to other family members would also suggest a motive to engage in family communication regarding the genetic testing (Segal *et al.* 2004). In a retrospective study examining the process of communicating a positive *BRCA1/2* genetic test result to male and female first, second, and third-degree relatives, McGivern *et al.* (2004) found the most important reasons for discussing the genetic test results were (1) to inform the relatives of their risk, (2) to suggest that they be tested, and (3) to fulfil a perceived duty to inform.

Much of the literature relates to those undergoing genetic testing for *BRCA1/2* who feel they have some moral duty to inform their family members about their testing and its results (Clarke *et al.* 2008; McGivern *et al.* 2004; D'Agincourt-Canning 2001; Chivers Seymour *et al.* 2010; Gaff *et al.* 2007). For example, Green *et al.* (1997) conducted a study looking at communication issues encountered by women attending a genetic counselling clinic because of a family history of breast and/or ovarian cancer. Their findings suggested that those undergoing genetic testing recognised a duty to inform relatives of their genetic risks and that some were prepared to go to some lengths to meet that responsibility. Ritvo *et al.* (1999) propose that, due to the autosomal dominant inheritance pattern, mothers who carry *BRCA1/2* mutations are particularly sensitive to the vulnerability of their female offspring because of their own experiences in relation to the previous family history of disease, and so are motivated to communicate with them.

Other reasons for communicating with relatives about genetic testing are based on needing information and/or support. Green *et al.* (1997) found that going for genetic counselling may itself be a result of family discussion about the disorder; but, whether or not this was the case,

nearly all those who were undergoing genetic counselling for a family history of breast and/or ovarian cancer had contacted someone in the family for information about the disease. Mothers in particular, if they were still alive, were key figures in supplying family information. This was not only because they often had the requisite information, but also because they were the link with other relatives of the previous generations (Green *et al.* 1997). There is evidence to suggest that open communication regarding hereditary cancer and partner support may be important buffers against hereditary cancer distress (Van Oostrom *et al.* 2007). Previous research has shown that communicating with family members is a strategy used by breast cancer patients to cope with their cancer diagnosis (Hilton 1994). Therefore, needs for social support may motivate family communication on the subject (Hughes *et al.* 2002).

In a recent qualitative systematic review of factors that promote and impede family communication regarding genetic testing for cancer risk (refer to Chapter Two), Chivers Seymour *et al.* (2010) identified that individuals are most likely to engage in family communication if they: undergo genetic testing with the intention of gaining information for other family members as well as for themselves; have a sense of duty to warn others of potential risk; have taken time to process the information before telling others; have close relationships with their relatives; and have been encouraged and supported by his/her genetic practitioner to engage in family communication.

Studies have also been able to give insight into why those undergoing genetic testing may not talk to some, or all, relatives. Green *et al.* (1997) reported that, whilst the majority of their sample, of 46 women attending a cancer genetics clinic, contacted at least one relative regarding counselling, most named a relative with whom they did not feel able to communicate on this subject. In these cases, communication, both obtaining and giving information, was impeded by adoption, divorce and remarriage, family rifts, and large age gaps between siblings (Green *et al.*

1997). Other studies have found that little contact and/or emotionally distant relationships are major barriers to family communication regarding genetic testing for cancer risk (McGivern *et al.* 2004; Chivers Seymour *et al.* 2010; Hughes *et al.* 2002; Finlay *et al.* 2008; Julian-Reynier *et al.* 1996; Forrest *et al.* 2003). There is also balance between the perceived obligations of passing on information with that of not causing alarm which must be overcome (Green *et al.* 1997; Clarke *et al.* 2008; Peterson *et al.* 2003; Gaff *et al.* 2007). According to Chivers Seymour *et al.* (2010), the issue of feeling torn between the responsibilities to inform at-risk relatives but not wanting to cause them any harm or distress poses significant tensions to those undergoing genetic testing for cancer risk. Such tensions may increase the chances of non-disclosure.

1.3.3 When Do Those Undergoing Genetic Testing For *BRCA1/2* Talk To Others?

The literature provides little evidence about when families communicate about genetic testing. Forrest *et al.* (2003) suggest that disclosure of genetic information is best described as a process, thereby implying a collection of actions rather than a single event (Gaff *et al.* 2007). Yet, what literature there is tends to specifically focus on when test results are disclosed rather than communication throughout the whole process of genetic counselling and genetic testing. For example, descriptive and correlation studies indicate that sharing test results occurs most often within a week of receiving the genetic test result with the majority of at-risk adult female family members (Claes *et al.* 2003; Wagner *et al.* 2003; Forrest *et al.* 2003; Hughes *et al.* 2002; McGivern *et al.* 2004; Patenaude *et al.* 2006; Segal *et al.* 2004; Petersen and Bunton 2002; Peterson *et al.* 2003). More importantly, however, few studies have explored the reasons behind why communication may occur at specific time points, or how it may differ throughout the journey of undergoing genetic counselling and genetic testing. Hughes *et al.* (2002) do suggest that *BRCA1/2* test results are likely to be communicated to relatives quickly when support is

needed. In a grounded theory study examining the experiences of *BRCA1/2* carriers in communicating genetic information to their offspring, Clarke *et al.* (2008) found that disclosure happened in several distinct phases. These phases included a pre-disclosure phase, where participants contemplated sharing the news; a disclosure phase, where participants shared the genetic information; and, lastly, the impact of disclosure phase, where participants described their reflections after disclosure occurred (Clarke *et al.* 2008). Hamilton *et al.* (2005) also suggest that disclosures performed later tend to be more carefully planned.

As discussed in 1.3.1, motivations to engage in family communication regarding genetic counselling and testing include the need to gain information and/or emotional support (Hughes *et al.* 2002; Green *et al.* 1997; Van Oostrom *et al.* 2007). The major limitation of looking at discussions with relatives only after the person has actually received their results is that the need for information and/or emotional support is not restricted to post- test result. In fact, because a significant amount of information on the family history of cancer is needed during the early genetic counselling sessions, and decisions then have to be made regarding whether or not to actually have the test, including other relatives in order to gain information, emotional support is most likely to occur early in the process (refer to section 1.2.1). According to Gaff *et al.* (2007), 'There would also be value in considering the family communication processes that lead to an understanding of risk, which may occur over a long period of time rather than in the context of conveying a piece of information' (p. 1009). Further research is needed to reflect this, so interventions to support family communication regarding genetic testing can be appropriately targeted to when they are most useful.

1.3.4 How Do Those Undergoing Genetic Testing For *BRCA1/2* Talk To Others?

Studies have found that uptake of genetic counselling and/or genetic testing by potentially at-risk relatives is consistently low (Blandy *et al.* 2003; Landsbergen *et al.* 2005; Sanz *et al.* 2010; Claes *et al.* 2003; Peterson *et al.* 2003; McGivern *et al.* 2004; Ormond *et al.* 2003; Ayme *et al.* 1979). Of those studies that have determined the rate of uptake of genetic counselling/testing by relatives informed about testing, the percentage of those informed who underwent testing ranged from 13% (Ormond *et al.* 2003) to 57-64% (Peterson *et al.* 2003).

Interestingly, one study in the Netherlands (Landsbergen *et al.* 2005) found uptake levels for predictive *BRCA*-mutation testing of just 36%, by first- and second-degree relatives of 50 female index[5] patients, could not be explained by demographic or counselling characteristics, nor by cancer-related history. Rather, the authors concluded that uptake in BRCA1/2-mutation families is related to emotional and behavioural communication characteristics of index patients (Landsbergen *et al.* 2005). Blandy *et al.* (2003) and Foster *et al.* (2004a) have expressed concerns that, how well the information given by the geneticist is understood and retained by those who are talking to their relatives about genetic testing, may be a barrier to informing relatives. The knowledge about the risk of transmission of *BRCA1/2* mutations by women was found to be positively and significantly associated with the testing decision among first-degree relatives by Blandy *et al.* (2003). These findings reiterate the need to have a full understanding of not only when and with whom information about genetic testing is being shared, but also how it is being done and what is being said.

In 2003, Peterson *et al.* conducted a retrospective, cross-sectional, qualitative study to evaluate how information about the identification of an *HNPCC* gene mutation was disseminated in five

[5] The family member with the earliest documented case of a genetic disease.

families where at least one member had undergone genetic counselling and testing for cancer risk (Peterson *et al.* 2003). This was conducted as part of a multiphase study of psychosocial aspects of genetic counselling and testing for *HNPCC* (Vernon *et al.* 1997). Of the 63 biological relatives invited to take part in the qualitative study, 29 agreed to participate, and ten of the 31 invited spouses agreed to participate (Peterson *et al.* 2003). The primary finding regarding how the families talked about genetic testing was that, provided someone in the family knew how to contact relatives, communication followed the norms used for conveying other non-urgent family news. As was discussed in section 1.3.1, the news was first shared with emotionally close and first-degree relatives, i.e. with their spouses, children, and siblings. None of the families organised a specific effort or event, such as a family letter or a family meeting, to communicate genetic information to other family members. Instead, the information was conveyed on a one-to-one basis through usual contacts with other family members, such as telephone calls and social activities (Peterson *et al.* 2003). It is important to note that, in this study, the probands (the first person in the family to undergo the genetic test), who were instrumental in first bringing news of a mutation to the rest of the family, were cancer survivors themselves. This fact may have mitigated the potentially threatening nature of this news and facilitated more open discussion about it; however, further research is needed to explore this.

Whilst the authors claim that the purpose of the paper is to describe how information about an identification of a mutation was disseminated within the family, the findings actually focus more on when and under what circumstances this information was shared, and how family members reacted to and acted on this information (Peterson *et al.* 2003). As was found in other literature in section 1.3.2, the focus is on dissemination of test results; and, where more general discussions about genetic counselling are included, these focus on how relatives can access testing. No exploration of how the subject is approached and discussed during the earlier stages of the process is included. Exploration about how those undergoing genetic counselling and

genetic testing for *BRCA1/2* communicate with second and third-degree relatives is also important, as there is further evidence to suggest that informing more distant relatives is often carried out in a less systematic and more selective manner (Julian-Reynier *et al.* 2000; Hughes *et al.* 2002; Claes *et al.* 2003).

In a postal questionnaire regarding familial communication of *BRCA1/2* results sent to mutation carriers in Pennsylvania, USA, by Finlay *et al.* (2008), participants (n=115) were asked about: methods of disclosure, including whether they relied on other family members to disseminate risk information; what was included in disclosure discussions; and whether they recommended testing at-risk relatives. The results found 88 (77%) disclosed their positive *BRCA1/2* mutation status to all at-risk family members, while 27 (23%) survey participants disclosed to at least one, but not all, at-risk family members. The most common methods for disclosure of positive test results were discussions in person (87.0%) or over the telephone (76.5%). Writing letters or e-mails describing the results were less frequent methods of disclosure. Interestingly, there were no statistically significant differences between methods used by people who disclosed to all at-risk relatives versus those who disclosed to only some at-risk relatives. In addition, gender did not influence the methods used for disclosure. Topics discussed with family members included: chance of having a mutation (reported as discussed with one or more relatives by 85.2% of participants); cancer risk for people with a mutation (88.7%); screening guidelines for people with a mutation (70.4%); changes in medical care for people with a mutation (69.6%); preventative surgery or medical options to reduce risk (78.3%); and/or information about genetic discrimination (43.5%). A total of 89% of those surveyed either strongly recommended or suggested genetic testing to their relatives during disclosure conversations. Despite these recommendations, uptake of testing in at-risk relatives was only 57% (although uptake data is limited in that they are based on survey participants' knowledge of relatives' testing behaviour, not on actual test results). Due to the research methods chosen, i.e. postal surveys, the work

was unable to discover any factors that significantly contributed to participants' desire to talk about genetic risk with family members, which would be useful information to obtain.

Green *et al.* (1997) also found that information regarding genetic test results for *BRCA1/2* was most likely to occur when family members met face to face, and to be integrated into communication that would be taking place in any case. Concern about the possible alarmist effect of contacting relatives solely for the purpose of telling them of their, or their children's, possible cancer risk, meant letters and phone calls were not considered suitable ways of giving information to relatives who are not in close contact. Instead, people waited until a family gathering occurred for some other reason, or to include the information with a regular communication, like a Christmas card (Green *et al.* 1997). However, this research was conducted well before the widespread introduction of mobile phones and emails, which has no doubt changed the nature of how families communicate (Kennedy *et al.* 2008).

How families talk about cancer and the family history of cancer may also affect how they communicate about genetic counselling and genetic testing for cancer risk. Mellon *et al.* (2006) conducted focus groups with family dyads, consisting of breast or ovarian cancer patients and close female relatives, and found some families talked openly about cancer and risk information, while others did not talk about the cancer at all after the initial diagnosis and treatment was finished (Mellon *et al.* 2006). McAllister (2002) found family communication can have significant effects on engagement with cancer risk; for example, 'When the family does not discuss the family history, family members can forget about it and remain partially engaged' regarding the process of genetic testing (McAllister 2002, p. 497). Further investigation would be warranted to see if there is a link between how families discuss cancer, and their family history of cancer, and how they discuss genetic counselling and genetic testing. For example, it might be hypothesised

that, where families communicate very openly about cancer, they may be more open in talking about genetic testing as well.

1.4 Conclusion

The literature presented in this chapter has demonstrated that, because of the issues with confidentiality, talking about a family history of cancer, associated risks and genetic testing within the family can be very important in ensuring all family members have access to genetic services. It can, however, be potentially difficult for concerned individuals and their relatives. As more genes are discovered, there is likely to be an increase in numbers of people affected by these issues and so the development of interventions to help people manage problems associated with the process should be a top research priority.

Despite a wide range of literature, it is clear that the nature of interactions regarding genetic information remains poorly understood (Gaff *et al.* 2007). Research to date has focused far more on with whom and why (motivations) those undergoing genetic testing talk to their family, rather than when or how it is occurring. What is more, the research tends to focus on communication with specific family members at the point of result disclosure only. During this chapter, the following areas where further research is needed have been identified:

1. Research examining *when* those undergoing genetic testing for *BRCA1/2* talk to their relatives, throughout the whole process of genetic counselling and genetic testing, not just disclosure of test results.
2. Research focusing on *how* those undergoing genetic testing for *BRCA1/2* talk to their relatives about genetic testing for cancer risk, and what is communicated.

3. Research looking at talking to the whole family, not just with first-degree relatives or specific family members.
4. Research investigating whether there is a link between how openly families talk about cancer in the family; and talking about genetic testing and its risk implications.
5. Research examining whether those undergoing genetic testing have understood the genetic information in order to relay it successfully to other family members.
6. Research looking at whether the use of modern technologies, such as mobile phones and emails, has changed how those undergoing genetic testing for *BRCA1/2* talk to their relatives.
7. Research observing whether those undergoing diagnostic genetic testing, i.e. cancer survivors, communicate differently from those undergoing predictive genetic testing, i.e. who have not had cancer.

Having identified gaps in the literature on how those undergoing genetic testing for *BRCA1/2* talk to their relatives, it is necessary to consider how they may be addressed and the design of the research. These seven areas are more concerned with the process rather than the outcome (Munhall and Boyd, 1993). They are looking to describe, explore, and explain the phenomena being studied (Marshall and Rossman, 1995). This would suggest that a qualitative method of inquiry, that seeks to understand social phenomena within the context of the participants' perspectives and experiences (Merriam, 2002), would be most appropriate for their exploration.[6]

[6] Rather than a quantitative inquiry which, according to Creswell (1994), would be 'based on testing a theory composed of variables, measured with numbers, and analysed with statistical procedures, in order to determine whether the predictive generalizations of the theory hold true' (p. 2).

Qualitative research has the advantage in that it offers rich, subjective accounts of a phenomenon of interest (Munhall and Boyd, 1993). However, designing a study that aims to address all of the seven areas of research identified above risks losing that in-depth exploration. Hence, there was a need to select and refine the research area and research questions.

With this in mind, before going any further, it was necessary to have a complete understanding of what qualitative research had already been conducted concerning family communication regarding genetic testing for *BRCA1/2.* Therefore, in order to inform the selection and development of the research questions underpinning this research, the next activity involved the conduct of a systematic review and meta-synthesis of the qualitative literature. This review addressed the question 'What facilitates or impedes family communication following genetic testing for cancer risk?' and is discussed fully in the next chapter.

Chapter 2 – Systematic Review and Meta-synthesis

What facilitates or impedes family communication following genetic testing for cancer risk?

2.1 Introduction

The literature reviewed in Chapter One demonstrates how access to genetic cancer risk information can be highly dependent on whether familial risks are discussed within the family (Hughes *et al.*, 2002). Despite its essential role in ensuring family members have access to genetic services, there are a number of gaps in the knowledge available on people's experiences talking to their relatives about genetic testing for *BRCA1/2* and its risk implications, as identified in section 1.3.1, which would warrant further investigation.

The objective of this chapter is to systematically review and meta-synthesise the primary qualitative research regarding family communication following genetic testing of cancer risk, in order to:

- Ascertain what qualitative research had already been conducted concerning family communication regarding genetic testing for *BRCA1/2*;
- Identify barriers and/or facilitators that arise when communicating with relatives about undergoing genetic testing for cancer risk;
- Inform the direction and focus of this study and aid refinement of the research question(s).

2.2 Qualitative Systematic Reviews

The decision was taken to pursue a qualitative line of inquiry, because the intention was to further explore the experiences and process of sharing genetic information with relatives and the challenges that can present. Qualitative research, rather than quantitative research, is more suited to this as the emphasis is on meanings, experiences, and views of participants (Pope and Mays, 1995). However, in the past, there have been criticisms regarding incorporating qualitative studies into systematic reviews.

- A systematic review aims to 'apply pre-defined strategies to identify, appraise and synthesise research findings from primary studies to provide empirical answers to targeted clinical questions' (Gysels and Higginson, 2007). They are characterised by specific features, including:
    - an explicit study protocol, addressing a pre-specified, highly focused question(s);
    - explicit methods for searching and identifying studies;
    - an appraisal of the studies' scientific quality; and
    - clear methods, including descriptive summary or meta-analysis, to combine the findings across a range of studies (Egger *et al.*, 1995).

As such, systematic reviews and data synthesis are important tools in developing an evidence base for health care (Jones, 2004).

In 1979, Archie Cochrane published an essay in which he suggested that 'It is surely a great criticism of our profession that we have not organised a critical summary, by specialty or subspecialty, adapted periodically, of all relevant randomised controlled trials' (Cochrane, 1979). Since the 1970s, the gained momentum of the evidence-based policy and practice movement has promoted the use of systematic reviews. However, the 'hierarchy of evidence' approach adopted by the Cochrane Collaboration has historically favoured the randomised controlled trial

(RCT) design, thus excluding consideration of other quantitative and qualitative study designs (Jones, 2004). According to Dixon-Wood and Fitzpatrick (2001), the reluctance to extend the existing remit of systematic reviews to include qualitative research was due to a fear it would signal an unwelcome return to the haphazard and biased era of the traditional review.

One major criticism of the 'hierarchy of evidence' approach is that such systematic reviews are unhelpful and inappropriate for answering complex questions (Dixon-Wood *et al.*, 2006). The limitations of only counting 'evidence' as evidence when it comes from RCT-based systematic reviews, and only when it answers 'what works?' questions, are increasingly recognised (Dixon-Wood and Fitzpatrick, 2001). In response to this, guidelines from the NHS Centre for Reviews and Dissemination (CRD) explicitly recognised the need for systematic review methodology to incorporate more diverse forms of quantitative and qualitative evidence (NHS Centre for Reviews and Dissemination, 2001). Qualitative research has an especially valuable role to play in answering questions that are not easily addressed exclusively by experimental methods (Dixon-Wood *et al.*, 2001).

Sherwood (1999) suggested that combining qualitative studies in a review makes the findings more substantial because they draw on a broader range of participants and descriptions. They have the potential to make an important contribution and influence health care decisions because they explore the beliefs, expectations and understandings of participants (Evans and Pearson, 2001). As a result, a new interpretation the phenomenon under study is produced which contributes to the further understanding of the topic (Evans and Pearson, 2001). However, that is not to say there are not concerns about potential limitations of qualitative systematic reviews, such as:

1. The need for rigour in identifying research. It can be difficult to search for and identify appropriate qualitative studies as there is not an equivalent to the Cochrane controlled

trials register, and because indexes and search filters can be lacking (Dixon-Wood *et al.*, 2001).

2. Appraising quality of qualitative studies. It is difficult to construct a hierarchy of evidences for types of qualitative research studies; and there are no universally agreed criteria for judging a paper's quality, deciding whether or not it should be included and/or how to weight it (Dixon-Wood *et al.*, 2001).
3. Secondary summary and synthesis. This is made difficult by the number of data types and different methods for data collection, widely varying theoretical perspectives and diverse analytic approaches (Dixon-Wood *et al.*, 2001).

However, in the literature there is evidence to support how methods of systematic review and meta-synthesis can be applied to qualitative research and the importance of such work (Dixon-Wood and Fitzpatrick, 2001;Dixon-Wood *et al.*, 2001;Dixon-Wood *et al.*, 2006;Evans and Pearson, 2001;Jones, 2004). To overcome these limitations, researchers have developed and adapted the rationalist systematic review methodologies to allow them to be applied to qualitative research. For example, the analysis and synthesis of qualitative studies is commonly termed meta-synthesis. It refers to the synthesis of findings from different types of qualitative research, allowing critical review and analysis (Sherwood, 1999). Unlike meta-analysis, which is associated with quantitative systematic reviews, the goal is to be interpretive rather than aggregative. It aims to provide increased understanding of a phenomenon rather than providing definitive evidence on the effectiveness of an intervention (Evans and Pearson, 2001). It is this aim that made a systematic review and meta-synthesis of qualitative research appropriate to meet the objectives of this chapter.

2.3 Methods

The methods used for this systematic review and meta-synthesis followed those described by Jones (2004). Relevant studies were appraised for quality and relevance by three independent researchers (the PhD student and two academic supervisors), using a quality assessment tool (Appendix 1) adapted from Hawker *et al*. (2002). These two sources were selected because they provided clear explanations of methods and examples of successful systematic reviews in Cancer, Palliative and End of Life Care research.

2.3.1 Identifying Relevant Studies

Egger and Smith (1998) argue that the prominent feature of rationalist systematic review methodology is an explicit search strategy for identifying relevant evidence. In particular, the account provided of search methods should be easily reproducible. Criteria of eligibility are used during the conduct of systematic reviews to ensure only studies addressing the specific topic of interest are included in the review (Evans and Pearson, 2001). This systematic review identified published primary qualitative research studies where:

- participants or a close family member (spouse, partner, parent or sibling) had undergone genetic testing for cancer risk;
- findings included data relating to aspects which facilitated or impeded family communication following testing; and
- those aspects were either stated by the authors or appeared from the published data to be an important element in the study findings.

Studies using mixed methods were included only if the qualitative findings were reported and discussed separately from the quantitative findings, so the data could be incorporated into the meta-synthesis with the other papers included. Studies from any country were eligible for inclusion, allowing comparisons between countries if appropriate. Studies were excluded if they:

- focused on other forms of genetic testing, such as carrier testing7
- focused on genetic testing for other conditions than adult onset cancer; or
- were not written in English, as resources for translation were not available.

Primary qualitative research studies were taken to be studies which used methodologies associated with qualitative research, such as: phenomenology, ethnography or grounded theory; and those methods more often linked to qualitative research, such as: in-depth interviews, focus groups, observations, and reflective diaries to explore participants' experiences (Jones, 2004).

The dates searched were from the existence of each database until January 2008. The searches have not been repeated to update the content of the systematic review in preparation for thesis submission, as the objective of the review was to inform the future direction of this study during its early stages and to aid the development of the research questions. Any literature published since then is, therefore, not relevant to this chapter; however, such literature was reviewed in preparation for the discussion in Chapter Eight.

2.3.2 Search Strategy

Studies were identified primarily through searches of relevant electronic databases (Table 1). Groups of terms relating to four specific parameters were combined: (1) family (e.g. 'relations',

[7] A test designed to detect carriers of an altered gene for a particular recessive disease, such as sickle cell trait.

'relatives', 'kin', 'children', 'offspring', 'siblings', 'parents'); (2) communication (e.g. 'talking', 'transmission', 'sharing', 'discussing', 'informing'); (3) genetic testing (e.g. 'predictive' or 'diagnostic'); and (4) qualitative (e.g. 'phenomenology', 'ethnography', 'grounded theory', 'interviews', 'observations', 'focus groups', 'field studies', 'case studies'). Some experimentation was required to develop an appropriate search strategy that allowed effective identification of relevant studies (Table 2).

Database	Version	Dates Searched
CINAHL	Ovid/Silver Platter	1982-Dec 2007
Embase	Ovid	1996-Jan 2008
Medline	Ovid ®	1996-Jan 2008
British Nursing Index	Ovid	1994-Jan 2008
PsycINFO	Ovid including PsycARTICLES	2000-Jan 2008

Table 1: Electronic databases searched for systematic review

1	genetic test$.mp.
2	predictive genetic test$.mp.
3	diagnostic genetic test$.mp.
4	1 or 2 or 3
5	communicat$.mp.
6	discuss$.mp.
7	shar$.mp.
8	talk$.mp.
9	inform$.mp.
10	5 or 6 or 7 or 8 or 9
11	4 and 10
12	famil$.mp.
13	kin$.mp.
14	relative$.mp.
15	parent$.mp.
16	sibling$.mp.
17	offspring$.mp.
18	child$.mp.
19	relations.mp.
20	12 or 13 or 14 or 15 or 16 or 17 or 18 or 19
21	11 and 20
22	qualitative.mp.
23	phenomenol$.mp.
24	ethnon$.mp.
25	grounded theory.mp.
26	interview$.tw.
27	observation$.tw.
28	focus group$.tw.
29	field studies.tw.
30	case stud$.tw.
31	22 or 23 or 24 or 25 or 26 or 27 or 28 or 29 or 30
32	31 and 11
33	31 and 21
34	32 or 33

Key to abbreviations, as used in Ovid:

- $, tructionation
- mp= ti, ab, hw, it, sh, tn, ot, dm, mf, nm, tc, id
- hw = word in subject heading
- ti = words in title
- tw = textwords

Table 1: Final OVID search strategy used for systematic review

2.3.3 Sifting and Sorting

Sifting was carried out in three stages: first by title (n=327), then by abstract (n=92), and finally by full text (n=28), excluding studies that did not satisfy the inclusion and exclusion criteria at each stage. Each paper was independently reviewed by the PhD student and at least one supervisor at each stage and data were extracted with the help of a standard pro forma (Appendix 2).

Twenty-eight papers (8.6% of the total number of hits) were read in full; only 10 (30%) of these met the study inclusion criteria (Figure 2). Four further studies were identified by the process of citation pearl-growing (Hartley, 1990), whereby the reference list of all studies which met the inclusion criteria were hand-searched for relevant studies. Therefore, a total of 14 papers are included in the final meta-synthesis (Table 3).

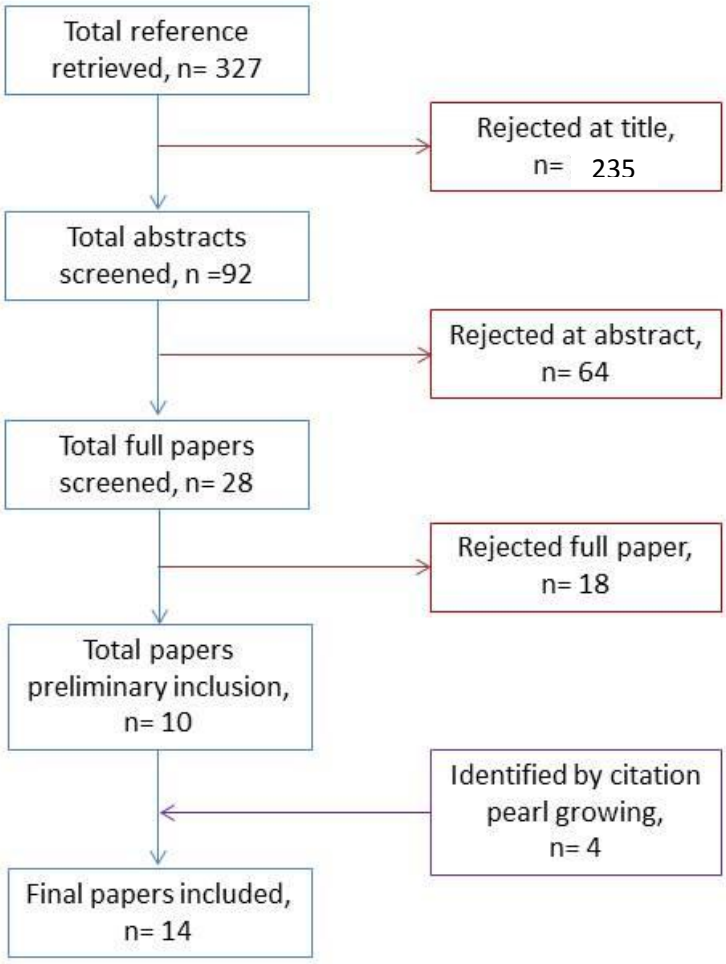

Figure 2: Summary of study selection and exclusion for all electronic literature searches

2.3.4 Assessment of Quality

The quality and relevance of each paper was then assessed using a Quality Assessment Tool (QAT) adapted from Hawker *et al*. (2002). To assess each paper fairly and to ensure consistency, Hawker *et al*. (2002) rate each sub-section as either 'good', 'fair', 'poor', or 'very poor' based on clearly defined criteria. This tool was then further adapted to give a numerical score of 0-3 points to make it easier to display results (Appendix 1). Once again, each was independently assessed by the PhD student and at least one supervisor. The team then met to discuss scores. Generally, there was strong agreement between reviewers, demonstrating that the criteria for each score were well-defined. Any disagreements in scores were resolved by consensus following a group discussion. (The final scores can be found in Table 3.)

There are two points worth noting here. Firstly, one of the supervisors involved, Dr Claire Foster, was an author on several of the papers included in the final review. To ensure fairness, she did not review any of the papers that she had been involved in; in these cases, these reviews were completed by the PhD student and the other supervisor. Secondly, it is important to recognise that the 'transferability and generalisabilty' criterion of the QAT is of limited appropriateness to some methodologies, for example, the first stage of a grounded theory study. However, appraisal of the evidence is an important activity in the systematic review method to reduce the possibility of bias (Dixon-Wood *et al.*, 2001).

Study	Country	Cancer	Data Collection	Total Participants (N=)	Gender		Cancer Status		Participants			Mean Age (y)	Quality assessment tool Rating (n/36)
					Male	Female	DGT / Affected	PGT / Unaffected	Patient	Partner	Offspring		
(Forrest *et al.*, 2003)	UK	HBOC HD	Interviews	28 (56 incl HD)	0	21 +partners	X	✓	✓ (21)	✓ (7)	X	?	**33**
(Hallowell *et al.*, 2005a)	UK	BRCA	Interviews	29	17 +partners	0	X	✓	✓ (17)	✓ (8)	✓ (4)	54	**32**
(Foster *et al.*, 2004a)	UK	HBOC	Interviews	15	0	15	X	✓	✓	X	X	46	**31**
(Gaff *et al.*, 2005)	Australia	HNPCC	Telephone Interviews	12	5	7	✓	X	✓	X	X	41	**24**
(Hallowell *et al.*, 2003)	UK	BRCA	Interviews	30	0	30	✓	X	✓	X	X	53	**24**
(Kenen *et al.*, 2006)	UK	BRCA	Focus Groups	13	0	13	✓	X	✓	X	X	?	**24**
(Hallowell *et al.*, 2002)	UK	BRCA	Interviews	30	0	30	✓	X	✓	X	X	55	**23**
(Peterson *et al.*, 2003)	USA	HNPCC	Interviews	39	15	24	✓ (6)	✓ (?)	✓ (?)	✓ (?)	✓ (?)	53	**22**
(Mesters *et al.*, 2005)	Netherlands	HNPCC	Interviews	30	8	22	✓	✓	✓	X	X	43	**21**
(Carlsson and Nilbert, 2007)	Sweden	HNPCC	Interviews	19	9	10	✓ (8)	✓ (11)	✓	X	X	47	**15**
(Claes *et al.*, 2003)	Belgium	BRCA	Interviews	63	1	62	✓	X	✓	X	X	51	**14**
(D'Agincourt-Canning, 2001)	Canada	BRCA	Interviews	36	5	31	?	?	✓	X	X	53	**14**
(Bonadona *et al.*, 2002)	France	BRCA HNPCC	Interviews	23	6	17	✓	X	✓	X	X		**13**
(Hamilton *et al.*, 2005)	USA, Canada, Denmark	HBOC HD	Interviews (incl email)	17 (29 incl HD)	?	?	X	✓	✓	X	X	44	**13**

HBOC = Hereditary breast and ovarian cancer; HD = Huntington's disease, *BRCA* = High risk breast and ovarian cancer genes; HNPCC = Hereditary non-polyposis colorectal cancer; DGT= Diagnostic genetic testing; PGT = Predictive genetic testing; (?) = Data not provided by author.

Table 2: Summary of descriptive data for studies included in systematic review

The 14 studies included in this review were of variable quality. The graph in Figure 3 shows the number of papers scoring Good (3 points), Fair (2 points), Poor (1 point) and Very Poor (0 points) for each category within the QAT. There is an argument that it is not possible to exclude studies based on methodological flaws as standards of 'good' research change over time, and because a researcher's judgement is biased by her or his own area of research, training and individual preference (Jones, 2004). Therefore, no studies were excluded on the basis of their quality assessment score; however, it should be noted that quality scores ranged from 13 to 33 (n/36).

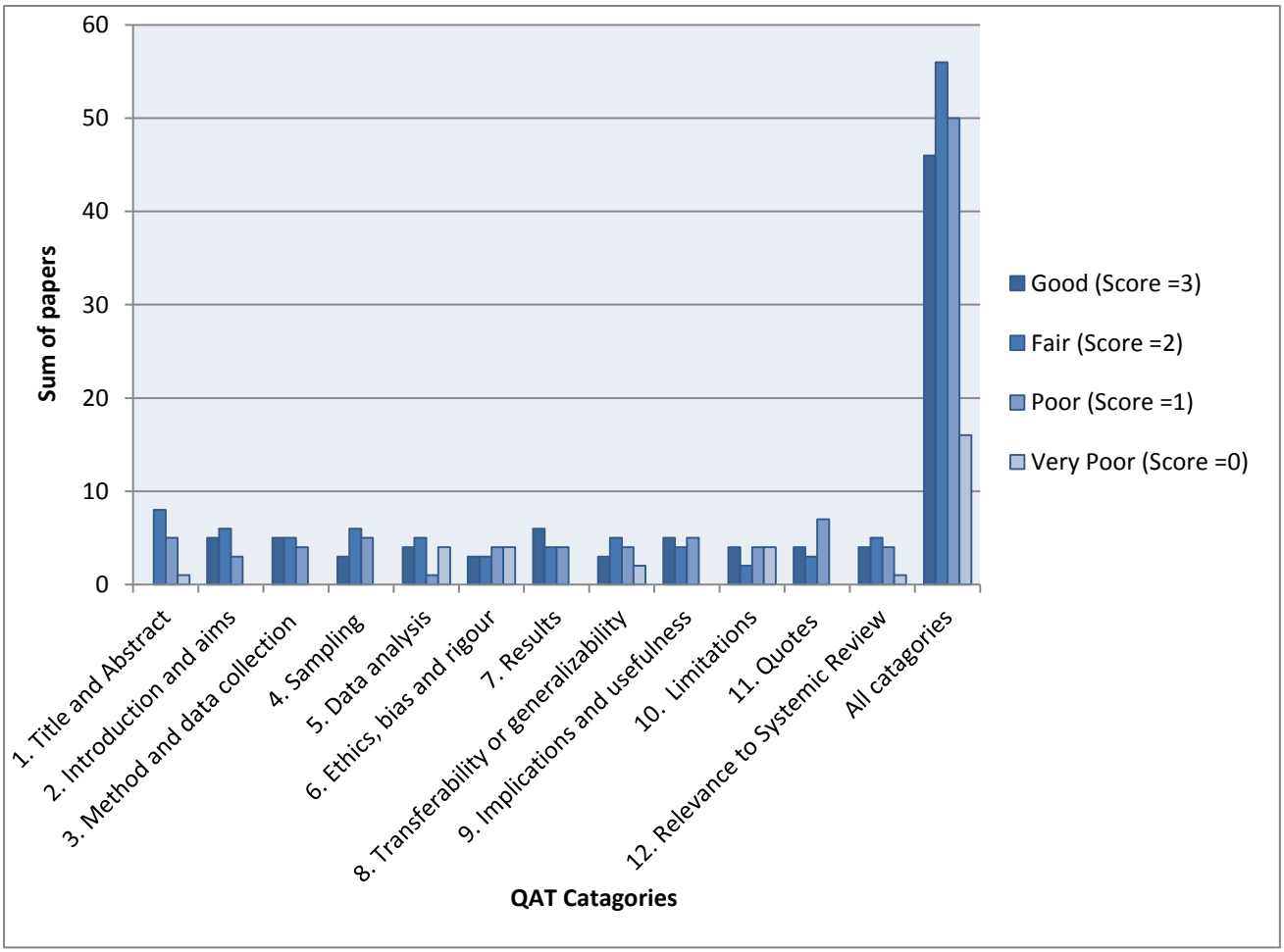

Figure 3: A graph showing the number of papers scoring Good, Fair, Poor and Very Poor for each category of the Quality Assessment Tool

2.3.5 Data Extraction and Analysis

To try to provide the data and the participant's voice in their purest form for meta-synthesis, only the 'results' section of each paper was used. This was felt to be important so as not to strip studies of their original meaning and specific context, as would have been the case if, for example, only participants' quotes had been used. Study findings were meta-synthesised and analysed using Ritchie and Spencer's Framework approach (Ritchie and Spencer, 1994). This method was developed in the 1980s by the Qualitative Research Unit at the National Centre for Qualitative Research (UK) as a method of analysing qualitative research data. It has been successfully used by others for analysis of systematic review data (Jones, 2004;Lloyd Jones, 2005).

This approach was chosen because it provides a clear series of steps, designed to help first-time researchers to manage the large amount and complex nature of qualitative data easily. It is best adapted to research that has specific questions, a limited time frame, a pre-designed sample and a priori issues (Srivastava and Thomson, 2009). This systematic review started with a specific research question designed to inform the study. The time frame was limited in that it needed to be done fairly quickly in order for the study to progress forward. The sample was pre-designed by nature of the research question and objectives; for example, the sample was qualitative research that explored family communication regarding genetic testing. A priori issues had been informed by exploration of the literature when preparing the background to the study (Chapter One).

The key advantages to a Framework approach, making it suitable for this study, are that it is: a) heavily based in, and driven by, the original data collected; b) dynamic: in that it is open to change, addition and amendment throughout the analytical process; c) systematic: allowing

methodological treatment of all similar units of analysis; d) comprehensive: allowing a full, rather than partial or selective, review of the material collected; and e) it allows within-case and between-case analysis (Srivastava and Thomson, 2009). It involves a systematic process of five phases: familiarisation, identifying a thematic framework, indexing, charting and mapping, and interpretation, all of which are described in more depth below. The approach was designed for use in applied qualitative research to inform social policy, allowing for rigorous and transparent data management. It does not purely rely on the emergent properties of a relatively small body of data, but also draws on the aims and objectives of the pre-defined research question and knowledge gained from the existing literature (Ritchie and Spencer, 1994). The stages followed were:

1. Familiarisation

The aim of the familiarisation phase is to gain an overview of the data and to begin listing key ideas and recurrent themes. This was achieved by reading and re-reading each paper's results section several times and noting down the key emerging issues.

2. Identifying a Thematic Framework

The thematic analysis focused on identifiable themes and patterns. Initially, the framework was based on the key issues identified during the familiarisation phase and a priori themes.

3. Indexing

Indexing involved the systematic coding of all the 'results' section of each paper using the thematic framework. Once identified, the key themes and sub-themes were numbered and indexed, thereby creating a thematic framework within which the data were sifted and sorted. During this time, the index was refined and modified. The initial framework was then applied to the results section of each paper and refined to incorporate additional emergent themes.

Indexing references were recorded and stored using the Nvivo8 data management software. The reliability and consistency of the analysis of the data were confirmed by independent assessment of the papers by at least one supervisor.

4. Charting

Following from this, charts (or tables) were created for each theme to show a summary of the data across the included papers. A matrix-based approach was adopted, with the columns of the matrix representing themes and the rows quotes from the paper. The charts display the range of experiences for each main theme across the data as a whole, and in doing so facilitated the task of the final phase of the analysis: mapping and interpretation.

5. Mapping and Interpretation

In this final phase, each theme was explored for patterns and contradictions across the papers' results. A realist approach was used, whereby participants' quotes in the papers were taken to be representative of their true feelings, experiences and beliefs. The analysis then moved beyond description to interpretation, in the context of existing research.

2.4 Results

2.4.1 Methodological Issues and Limitations

As with all systematic reviews, there are limitations based on the scope of the review. First, some relevant papers may have been excluded due to the manner of searching. Second, the quality of any systematic review or meta-synthesis is dependent on the quality of the studies

included (Jones, 2004). There was considerable heterogeneity in the study populations, methods used and style of writing across journals, making it more challenging to assess quality and to pool data for meta-synthesis. However, the impact of this issue was minimised by the standardised quality assessment tool. Fourth, the papers included only relate to hereditary adult onset breast and colorectal cancers, which may present a bias. Four dealt with predictive genetic testing, six with diagnostic genetic testing, three with both, and one did not specify which type. Fifth, the results sections of each paper are subject to the bias of the original authors and will be dependent on their original research questions. For example, the research training and theoretical perspective of each author or research team will have influenced the way in which they carried out their research. Factors such as which journal the paper was published in will have affected the word limit and, therefore, the amount of the collected qualitative data that could be reported on. Finally, while there are 14 articles reviewed, there are only ten different research teams whose views are being presented, as Hallowell and Foster are cited on each other's work, and five of the 14 articles are by one or the other of these authors.

2.4.2 Identified Themes

The data analysis identified six major themes that influence family communication following genetic testing for late-onset hereditary cancer: (1) the informant's feelings about telling relatives about genetic testing; (2) the perceived relevance of the information to other family members and their anticipated reactions; (3) the 'closeness' of relationships within the family; (4) family rules and patterns (e.g. who is best placed to share information with whom); (5) finding the right time and level of disclosure; and (6) the supportive role of health care professionals. The challenges and facilitators are summarised during the discussion as per the research question.

Theme 1: Reactions to Role of 'Informant'

Several studies explored motivations for undergoing genetic testing and reported that participants wanted to gain information for others, for example offspring, siblings and cousins. This seemed especially true for those undergoing diagnostic genetic testing (i.e. the participant had already had cancer):

> Interviewer: 'You said that one of the reasons was to find out information for your daughters, were there any other reasons that you decided to proceed with testing?
>
> Oona: 'Oh no, really just to help my daughters and any further family. I mean to me it's immaterial now I know that I have got breast cancer, or had breast cancer.' (Hallowell *et al*. 2003, p. 75)

Being the recipient of information from the genetics team and then having the responsibility of passing it on to other family members was seen as emotionally demanding. The issue of feeling torn between their responsibilities to inform at-risk relatives…

> Susan: 'I think people have the right to know… if, for example, they hadn't known and then they found it [cancer] later, I think they would be angrier, and say well why didn't you tell me about this? Especially if somebody close to me dies. Well, why didn't I? You know, you basically could have saved a life.' (D'Agincourt-Canning 2001, p. 238)

…but not wanting to cause them any harm or distress poses tensions to the role of the informant:

> Mary: 'Other members of the family I haven't done anything with, because how do you approach them? Because [to] my mother, her sisters, and brothers 'cancer' is a death word… the ones who are further away I haven't approached. One, because I don't see them very often. Two, how do I do it? This is the dilemma. Do you ring people up, write

> to the people, go and see them and say 'look there is this chance'? I think if it was me, I would want to know… because then you can do bits and pieces. But if you are the bearer of this news, I think you are torn. You don't know what to do for the best.' (Hallowell *et al*. 2003, p. 77)

Other feelings included apprehension about the potentially harmful nature of the information; the accurate transmission of technical information; the fact that kin would be faced with difficult risk management decisions; dealing with relatives who did not want to receive such information; feelings of isolation and burden; and being the bearer of 'bad news'. There was evidence that some had not reflected on the implications of passing on information to their relatives until quite late in the process:

> Verity: 'I didn't think backwards I only thought forwards. I only thought about my offspring and their offspring. I really didn't consider my brother or, no I didn't... I didn't give any thought to that really until I spoke to Dr X and they started explaining… and then suddenly I thought 'oh crumbs' you know, that's why I say it's like throwing a stone into a pond and the circles start coming out' (Hallowell *et al*. 2002, box 4).

Not everyone found the process of communicating to relatives troublesome. Some, particularly females, had found it straightforward:

> Female participant: 'It wasn't an issue… This issue has been discussed in many family conversations over a long period of time [20 yrs], as part of my treatment too, it's been a topic of discussion… This has been a long term process for us, it didn't come out of the blue. It's just part and parcel of an ongoing process' (Gaff *et al*. 2005, p. 136).

Theme 2: Perceived Relevance and Anticipated Reactions

Genetic test results were viewed as more relevant to some family members than others. These judgements were based on a number of considerations including gender, age, genetic relationship (for example, cousins, siblings) and anticipated reactions. For example, parents were often considered 'too old' and children considered 'too young' for the burden of information:

> Participant: 'I did not need to notify any one. My children are too young to understand. This question does not apply to me.' (Mesters *et al*. 2005, p. 165)

For *BRCA1/2*, there was also evidence that the information was perceived to be of more relevance to female rather than males relatives:

> Participant: 'I mean obviously if I had daughters I'd have maybe been more anxious about it but… I've had three boys [who don't really need to know].' (Forrest *et al*. 2003, p. 323)
>
> Jane, 55 years: 'I don't even know how it affects them [sons] at all. You know, I don't know whether um, if the gene was there then they may get bowel cancer or whether they could get some other cancer or whether it wouldn't sort of affect them and they might just pass it on. I really don't know. I don't know anything about it at all'(57). (Foster *et al*. 2004, p. 446)

The decision to disclose or not was often based on the anticipated reactions of family members or their perceived receptivity. For example, Hamilton *et al*. (2005) state that participants decided whether to disclose genetic test results by assessing family members' vulnerability and receptivity to the information. Assessments of vulnerability and receptivity were related not to the disease or test outcomes, but to the life situation and personality of the family member.

Parents and children were assessed regarding their vulnerability. One participant talked about her mother:

> 'The previous couple of years had been very difficult (my diagnosis, my grandmother's death, and my father's death all occurred within a 7-month period) and while I don't believe in keeping secrets, I also didn't want to raise my mother's anxiety (and blood pressure) more than necessary.' (Hamilton *et al*. 2005, p. 20)

Theme 3: Closeness of Relationships

Emotional, genetic and demographic distance plays a role in family communication following genetic testing. The desire to help the family become aware of choices that could improve their health commonly motivated people, and the availability of predictive testing was seen as good news. Emotional ties, rather than the genetic relationship, often influenced who was told about predictive testing.

The responsibility for passing on information includes close family members as well as family members who live far away, perhaps in other countries. A particular difficulty can be experienced in conveying information about a hereditary predisposition for cancer to family members with whom one has only sporadic contact.

There was evidence that genetically-close relatives such as children, siblings and parents were more likely to be informed about diagnosis or test results than distant relatives. This was often accompanied by an assumption that someone else would have informed the more distant relatives. Little or superficial contact with distant relatives, and difficulties related to overcoming pre-existing conflicts or rifts in the family, impeded communication:

> Husband: [She] has got two brothers and they all know about it. Well one does, they don't speak to the other one. (Forrest *et al*. 2003, p. 323)

> Participant: My daughter got tested, but she never went back for the test results. This created tension in our family, so we did not take it any further. My brother and his family have no idea. (Mester *et al*. 2005, p. 165)

Theme 4: Family Rules and Patterns

Genetic testing is described as a family affair often influenced by the family's collective experiences of cancer. 'Rules' of family interactions and authority dictate family life and, therefore, communication patterns. There were several examples where the authority to disclose followed 'vertical' patterns through the family (e.g. from parent to child) rather than 'horizontal' (e.g. from cousin to cousin). For example, a grandparent is perceived to have more authority than an aunt or uncle.

> 'You see, when my sister died my brother was completely torn apart… But he just won't allow anything to touch him like that again, and in a way, [it's] his way of dealing with it, he just doesn't go there… So my [deceased] sister's children don't know yet, but my sister's mother-in-law knows, and she has an excellent relationship with them. And when the time is right she will brief them and we'll start moving things forward.' (Forrest *et al*. 2003, p. 321)

Also evident was the gendering of disclosure within families, where women often appeared to take responsibility for initiating contact with a genetic counsellor and for passing on information within the family.

Theme 5: Timings

Genetic testing presented a dilemma about when and how to tell relatives. It was important to find the 'right' time and be able to manage the time and content of disclosure, not only for the benefit of the recipient but also the informant. Death and disease in the family made it difficult to initiate discussions. Some needed time to absorb the information and make decisions, while others found sharing their information helped them do this.

Others wanted to delay talking to their relatives until they had received all the information themselves. For example, some participants would wait until they had received their own test results before telling relatives about their involvement in genetic testing; or, in some cases, waiting until the recipient was in a 'better place' to deal with the information:

> 'I have not told my son. He is 23 - old enough to know-but not ready to hear it yet. He is in a very stressful place trying to finish graduate school. He doesn't need to have further complications to his life right now.' (Hamilton *et al*. 2005, p. 20)

Theme 6: Role of Health Care Professionals

There were mixed views regarding the role of health care professionals in telling relatives about genetic testing. Some felt it was their responsibility rather than that of the health care professional to communicate family risk, as they knew the family better, whereas others wished a professional could speak to their relatives on their behalf:

> Donna: 'I just wish there was somebody—somebody else that could come and talk to them, you know, talk to my family. 'Cause you don't want to keep telling them things that hurts them.' (Kenen *et al*. 2006, p. 156)

However, there was clear evidence that health professionals can aid family communication following genetic testing:

> 'My sister's physician underlined the importance, so we did it together. It felt like doing the right thing.' Thus, although there was an intrinsic motivation to disclose information to one's family, it helped if external cues were present that gave people the feeling that what they were doing was okay. (Mester *et al*. 2005, p. 164)

Several expressed a desire for health professionals' input to legitimise risk information, and a tendency to rely on them for technical information was noted:

> 'I mean I could say to the kids I have these flags and markers and everything which do confuse me. I could be giving them the totally wrong information and leading them up the garden path… I mean you could tell them to a certain degree that you may have this genetic problem, but if you want to know more information, then your best bet is to contact the genetic clinic because they have all the details, they know what they are looking for.' (Forrest *et al*. 2003, p. 320)

2.5 Discussion

This is the first systematic review of qualitative studies exploring communication to potentially at-risk relatives following genetic testing for hereditary adult-onset cancers. The findings indicate six themes, representing both facilitating factors and challenges, that influence family communication: (1) the individual's reactions to the 'role of being the informant'; (2) the perceived relevance of the information for individual family members and anticipated reactions; (3) the closeness of the relationship between the informant and recipients; (4) the family rules

and patterns that influence their interactions; (5) the informant's management of the timing and content of information shared with relatives; and (6) the role played by health care professionals.

In summary, the data included in this systematic review and meat-synthesis would suggest that factors likely to promote family communication regarding genetic testing for cancer risk include: if the motivations for undertaking genetic testing included finding out potential risk information for other family members; regular discussion of the topic as part of the family's collective experiences of cancer; seeing genetic testing as a positive thing and a desire to make other family members aware of choices to improve health; having close relationships; being able to manage the time and content of discussions to allow the informant to feel prepared and in control (interestingly, this also appears in the challenges list); and positive input from health care professionals, in particular their endorsement, legitimising the word of the informant and providing more technical information.

A profile of the contextual factors from this analysis predicts that individuals are most likely to engage in family communication if they:

- undergo genetic testing with the intention of gaining information for other family members as well as for themselves;
- have a sense of duty to warn others of potential risk;
- have taken time to process the information before telling others;
- have close relationships with their relatives; and
- have been encouraged and supported by his/her genetic practitioner to engage in family communication.

Several challenges were also identified that could potentially act as barriers to family communication regarding genetic testing, including: feelings of burden and responsibility; misguided perceptions of who could be affected (for example, perceiving breast cancer as a predominately 'female' disease); not having a close relationship; geographic distance; assumption that others will inform certain family members; death and disease within the family; and a desire that a health care professional would take on the role of informant. These are potential areas where future interventions to aid family communication regarding genetic testing for cancer risk could be targeted.

These findings are consistent with other research not included in this systematic review. As discussed in Chapter One, several studies have demonstrated that there is an increased likelihood that family members discuss genetic counselling and testing with their first-degree relatives and their spouses, whereas discussions are less likely to occur with more distant relatives (Claes *et al.*, 2003;Koehly *et al.*, 2003). Koehly *et al.* (2003) found mutation status was not a significant predictor of whether discussions occurred. This suggests family culture may play a more important role in determining family communication rather than the results of the genetic test itself. The nature of family relationships, in particular, is likely to have an impact on the dissemination of information within the family and the support available to those tested. These may include: the nature of pre-existing relationships; divorce; remarriage; the patterns of interactions; family tensions and rifts (Forrest *et al.*, 2003;Green and Thomas, 1997).

One of the recurrent themes throughout was that of informing children about genetic testing and potential risk. While parents had often made the decision not to inform children at the time because they were deemed 'too young' or perceived unready for the information, several participants reported difficulties in deciding when 'the right time' was. As Forrest *et al.* (2003)

noted, this appears linked to perceptions of when action to prevent cancer was needed, or when the first key life decisions that may be affected by the disease needed to be made.

Whilst the objective of this review was to look at the implications for the research study and its direction, the review findings have implications for clinical practice. While the role of a genetic counsellor is primarily to facilitate informed decision-making about genetic testing, improve adjustment to genetic test results, and aid informed decisions about cancer prevention, surveillance and treatment (Hughes *et al.*, 2002), these findings suggest their role in aiding communication with relatives is vital. There was evidence that the endorsement of the health care professional can be an important stimulus to talking to relatives (Mesters *et al.*, 2005). In these studies, health professionals were also relied upon as a source of technical information or to legitimise the word of the informant. These findings support the use of aids to communication, such as family letters and information sheets, and the development of other interventions to aid family communication.

2.6 Conclusion

This systematic review and meta-synthesis gives insight into the challenges that those engaged in family communication regarding genetic testing for cancer risk face (Chivers Seymour *et al.*, 2010). In particular, it provides information on who is most likely to engage in such activities and why. It supports the fact that research tends to focus on disclosure as a single act at the point of receiving a genetic test result. The findings reinforce the conclusion of Chapter One: that there are still specific gaps in the literature, particularly around how and when families talk about the

topic, that should be addressed before steps can be taken in order to find ways of supporting family communication regarding genetic testing. As Gaff *et al.* (2007b) state:

> '*If genetic services are going to continue to rely on family communication to transmit information about hereditary cancer risk or genetic testing, or conversely if they seek greater participation in this process, it is vital to go beyond identifying factors that influence who in the family will be told and gain an understanding of the process of communication and its consequences*' (p. 1000).

Therefore, the next stage of the research was to conduct a qualitative study of how those who have had a genetic testing for *BRCA1/2* talk to relatives using in-depth interviews with people who had undergone genetic testing. This work goes beyond previous work, which primarily looked at *who* individuals tell and *why*, by focusing on *how* these families discuss genetic testing for cancer risk and *when* this communication occurs. Chapter Three will set out the research question, aims and objectives, and describe the qualitative study in theoretical (Research Methodology) and practical terms (Research Methods).

Chapter 3 – Research Process

3.1 Introduction

Chapter One introduced the topic under investigation and critically examined the literature on family communication regarding genetic testing for *BRCA1/2*. At the end of the chapter, seven areas were identified where further research is needed in order to have a fuller understanding people's experiences of family communication regarding genetic testing for *BRCA1/2* (refer to section 1.3.1). These areas of research relate to gaining further understanding of people's experiences of the phenomenon under study and therefore advocate pursuing a qualitative line of inquiry. In order to inform the selection and development of the research questions, Chapter Two then presented a systematic review and meta-synthesis of the qualitative literature to date, and focused on barriers and facilitators that arise when communicating with relatives about undergoing genetic testing for cancer risk. This review particularly highlights that, although extensive research has been carried out by looking at with whom and why (motivations) those undergoing genetic testing talk to their family, the research to date does not adequately cover how and when families talk about the topic. The 'how' refers to what they talk about; how the topic is introduced and managed; and what methods and patterns of communication are used. The 'when' refers to the time points that these communications occurs and how they differ throughout the whole process of genetic counselling and genetic testing. Therefore, from the seven gaps in the literature identified in section 1.3, it is the 'how' and 'when' that will form the primary focus of this present study.

Building on that work, this chapter begins with the selection and rationale of the research question and aims for this study. The rest of the chapter and the following chapter will then go on to describe the qualitative study in theoretical (Research Methodology) and practical terms (Research Methods).

3.2 Research Question

Research to date has focused far more on with whom and why (motivations) family communication regarding genetic testing occurs rather than when or how it is occurring. What is more, the research tends to focus on communication with specific family members at the point of result disclosure only. Therefore, the work presented in this thesis will address the following research question:

How and when do those undergoing genetic testing for BRCA1/2 talk to their relatives about a family history of cancer, associated risks and genetic testing?

3.3 Research Aims

The aim is to gain insight into participants' experiences of discussing their genetic testing, their test results and potential risk information following genetic testing for *BRCA1/2* with their family (not just with first-degree relatives or specific family members), with particular focus on *how* these families discuss genetic testing for cancer risk and *when*. There will be an

appreciation of family communication as a process rather than as a discrete event. Therefore, the research will examine when, from the perspective of a family member undergoing genetic testing, family communication regarding genetic testing occurs, throughout the whole process of genetic counselling and genetic testing, not just disclosure of test results.

The resultant study, as described in the remaining chapters, was qualitative in nature, employing in-depth interviews as the method for data collection and utilising the technique of constructing eco-maps (Ray and Street, 2005) as a method of identifying relevant family members and guiding the researcher through the family structure and relationships. These methods were chosen in line with the interpretive description methodology to ensure depth and richness in analysis and reporting of findings.

3.4 Research Methodology

According to Joubish *et al.* (2011), the design of any research study begins with the selection of a topic and a paradigm: 'A paradigm is essentially a worldview, a whole framework of beliefs, values and methods within which research takes place. It is this world view within which researcher works' (Joubish *et al.*, 2011) (p. 2083).

The previous chapters have summarised the selection of the topic under study and aligned the research within a qualitative paradigm. All research questions are theoretical in that they advocate positions on what the world is and how it can be known (Silverman, 2000a). Therefore, before engaging with any research, and throughout the process itself, it is important to take the opportunity to consider the foundations of the work. This section, Research Methodology, will

discuss the theoretical assumptions implicit within the research questions and the decision to conduct a qualitative interpretive descriptive study.

When one consults the research literature, much of the terminology in this field is not always used consistently; therefore, it is necessary to outline some of the literature on frameworks for the Process of Research, examining in more detail different epistemological, theoretical and methodological approaches to research. This will lead on to a justification for the study within a qualitative interpretive paradigm and an interpretive description methodology. The methodologies and methods chosen must depend on the nature of the project, the type of information needed, the context of the study and the availability of resources, for example, time, money, and human *(researcher)* input (Silverman, 1997). However, as Denscombe (2003) states: 'The crucial thing for good research is that the choices are reasonable and that they are made explicit as part of any research report' (p. 3).

3.4.1 Qualitative Research Paradigm

In social science, the principal research designs are either qualitative or quantitative in nature, or a combination of the two (mixed methods). Probably the most basic differentiation between a qualitative and quantitative paradigm is the importance of quantifiable data. Generally, quantitative research is associated with numbers, measurements and statistical reasoning. However, the heart of the quantitative-qualitative 'debate' is not methodological but philosophical (Krauss, 2005). Quantitative research expresses the assumptions of a positivist theoretical perspective (refer to section 3.4.4.), which holds that behaviour can be explained through objective facts, and knowledge is discovered and verified through direct observations or measurements of phenomena (Firestone, 1987). Quantitative approaches lend themselves to,

and are more often associated with, larger sample sizes that aim to be representational of the study population and provide research that can be replicated or repeated. As such, it would not be feasible or desirable to obtain vastly in-depth data from such research.

On the other hand, qualitative research is exploratory in nature, offering understanding and answers to the 'how', 'why' and 'what is' questions about human behaviour and the basis that underpin such behaviours (Denzin and Lincoln, 2003). The term 'qualitative research' may encompass a wide range of data, data collection, interpretation and/or analytical approaches and methodologies (Van Maanan, 1979) and it is important for a researcher to select the qualitative research approach that will best answer their own research question (Ploeg, 1999). However, there is fairly wide consensus that it is '*a naturalistic, interpretative approach concerned with understanding the meanings which people attach to phenomena (actions, decisions, beliefs, values etc.) within their social worlds*' (Snape and Spencer, 2003). Qualitative researchers believe that the best way to understand any phenomenon is to view it in its context; therefore, one of the central roles of qualitative research is to provide levels of richness, depth and profundity not necessarily available with other research paradigms, thus making it the most suitable choice for this present study.

According to Mays and Pope (1995), the three main criticisms of qualitative research in the health field are:

1. That qualitative research is merely an assembly of anecdote and personal impressions, strongly subject to researcher bias - in other words, it lacks validity and reliability.

2. That qualitative research lacks reproducibility - the research is so personal to the researcher that there is no guarantee that a different researcher would not come to radically different conclusions.
3. That qualitative research lacks generalisability.

There are those who have argued that terms like 'validity' and 'reliability' come from a positivist quantitative paradigm and should, therefore, be rejected in relation to qualitative inquiry (Altheide and Johnson, 1994;Leninger, 1994;Peck and Secker, 1999;Whittemore *et al.*, 2001). However, others, such as Morse (1999), have cautioned that rejecting these terms would mean rejecting the core scientific concept of rigour, which risks undermining the valued contribution of qualitative research to the advancement of knowledge. Rigour is the means by which researchers demonstrate integrity and competence: 'it is about ethics and politics, regardless of the paradigm' (Tobin and Begley, 2004).

As in quantitative research, the basic strategy to ensure rigour in qualitative research is systematic and self-conscious research design, data collection, interpretation, and communication (Mays and Pope, 1995). So, in order to overcome the criticisms outlined above, the finding of this research must be an authentic and trustworthy reflection of how participants talk to relatives regarding genetic testing for *BRCA1/2*. In line with interpretive description methodology (refer to section 3.5), several strategies were taken to ensure methodological rigour. For example, consideration of Lincoln and Guba's (1985; 1994) framework: credibility, transferability, dependability, confirmability (Lincoln and Guba, 1985) and authenticity (Guba and Lincoln, 1994). The application of this framework will be discussed in detail in section 4.9.

Nordgren *et al.* (2008) argue that generalisability, as we would normally make sense of it in a clinical trial, is impossible in qualitative research, because the phenomena are intimately tied to the times and contexts in which they are found. It may, however, be replaced by concepts of applicability (Guba, 1981), transferability (Lincoln and Guba, 1985) and case-to-case transfer (Firestone, 1993). Generalisability, from this perspective, is analytic rather than empiric (Nordgren *et al.*, 2008) and researchers should supply sufficient detail in the description so the reader can make an informed judgement of whether the original study setting and their own are sufficiently similar in order to 'apply' the findings (Murphy *et al.*, 1998).

For example, it is hoped that the findings of this study will be useful for anyone who has professional encounters with those undergoing genetic testing for *BRCA1/2*. The findings may provide professionals and researchers with a better understanding of how and when these families discuss the family history of cancer, associated risks and genetic testing. The findings may also offer a theoretically informed starting point for clinical questions that merit further investigation (quantitative or qualitative); for example, 'How can we support family communication regarding genetic testing for *BRCA1/2*?' Hence, the findings, which will be derived from the patient's own perspectives, may work as frames of interpretation for those supporting these individuals (Nordgren *et al.*, 2008). The theoretical perspective underpinning this research will be discussed in section 3.4.4.

3.4.2 The Qualitative Research Process

The 'Process of Research' (or 'Research Process') refers to the different approaches to research at different levels. Carter and Little (2007) argue that 'three fundamental facets of research – epistemology, methodology and method – should provide the framework for planning, implementing, and evaluating the quality of qualitative research' (p. 1316).

Epistemology is a branch of philosophy concerned with the nature of knowing. For example, epistemology aims to address questions such as: 'What do we know?', 'How is knowledge acquired?' and 'How do we know what we know?' (Grad, 2009). Methodology provides the justification for the methods by way of analysis of assumptions, principles and procedures in a particular approach to the inquiry (Schwandt, 2001), but are not the methods themselves (Kaplan, 1964). Whereas, methods are the 'research action' (Carter and Little, 2007), or the techniques used to gather evidence (Harding, 1987). As Carter and Little (2007) describe it: 'Methodology justifies method, which produces data and analysis. Knowledge is created from data and analysis. Epistemology modifies methodology and justifies the knowledge produced' (p. 1317). This relationship is depicted in Figure 4[8].

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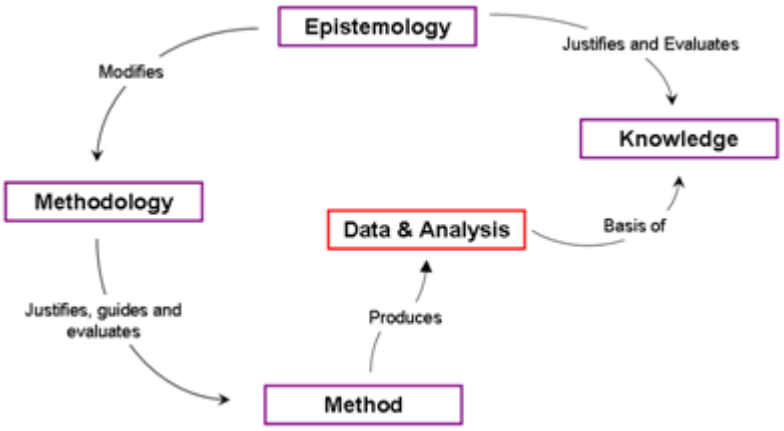

Figure 4: The Simple Relationship between Epistemology, Methodology and Method (Carter and Little 2007: 1317)

Crotty (1998) adds a fourth element for the research process, which is theoretical perspective. This is 'the theoretical stance informing the methodology and thus providing a context for the process and grounding its logic and criteria' (p. 3). As with Little and Carter's framework, Crotty's four elements inform one another.

This section will explore each of these four elements in more detail.

3.4.3 Epistemology: Justifying Knowledge

Epistemology is one of the core branches of philosophy. It concerns itself with the nature, acquirement and limitations of knowledge. For example, epistemology primarily addresses three questions: 'What is knowledge?', 'How is knowledge acquired?' and 'What do people know?'

Oka and Shaw (2000) contend that a lack of appreciation of the philosophical backgrounds of qualitative research often leads to confusion when analysing qualitative data and, therefore, argue it is essential for qualitative researchers to be aware of the influence of philosophy on strategies of research. There are a number of epistemological stances demonstrated in the literature; however, Crotty (1998) identifies the three core ones as:

1. Objectivism: holds that meaning, and therefore meaningful reality, exists as such without the operation of consciousness. For example, an object is that object whether anyone is aware of existence or not. 'Objectively' carries the intrinsic meaning of itself. When we recognise it as a particular object, we are simply discovering a meaning that has been there all along. Objectivism is founded within the principles of positivism.

   Positivism, as a philosophical system, holds that knowledge must be scientific to be valid; therefore, such knowledge can only be derived from the strict scientific methods of the 'natural science'. According to positivism, all research can be conducted objectively and independently of the researcher's values or perspectives. As such, positivism and objectivism are most often associated with quantitative research.

2. Subjectivism: here, meaning is imposed on the object by the subject from somewhere else. In other words, with the exception of existence, the object makes no contribution to the generation of meaning.

3. Constructivism: knowledge and meaningful reality is dependent on human engagement with the world, in and out of which meaning is constructed. Therefore, it is the interaction between object and subject that constructs meaning, and meaning cannot

exist without one or the other. As such, different people may construct meanings of the same phenomenon in different ways and, therefore, there cannot be one 'true' or 'valid' interpretation. An example of constructivism would be Interpretivism or Post-positivism.

As the prefix indicates, post-positivism is a meta-theoretical viewpoint to positivism. It recognises that human beings cannot perfectly understand reality but, with rigorous methods, researchers can reach an understanding of the social world whereby explanations are offered at the level of meanings rather than cause (Snape and Spencer, 2003). The methods of the 'natural science' are deemed inappropriate, as the social world is not governed by law-like regularities, and the researcher and participants are recognised to impact on each other.

Another epistemological debate to be considered is the way in which knowledge is acquired and the relative merits of inductive and deductive methods of scientific enquiry. Snape and Spenser (2003)(142)(17) define the difference as:

'*Induction looks for patterns and associations derived from observations of the world; deduction generates propositions and hypotheses theoretically through a logically derived process*' (p. 14).

Quantitative inquiry uses the latter, with 'emphasis on hypothesis testing, causal explanation, generalization and prediction' (p. 14), whilst inductive approaches to design, fieldwork and analysis are more commonly associated with qualitative inquiry, concentrating on understanding 'rich description and emerging themes and concepts' (p. 14). However, there are no exact rules; indeed, Snape and Spenser (142)(2003) go on to argue that both induction and deduction are involved at different stages of the qualitative research process (p. 24).

The implicit reasoning in this present study, as a piece of qualitative work, is held to be interpretistic and inductive, as an understanding of how people really behave and what people actually mean when they describe their experiences, attitudes, and behaviours is integral to answering the research question (refer to section 3.2). Therefore, this research study was conducted from within an interpretive perspective. Guba and Lincoln (1994), who prefer the label of constructivist, characterises this perspective as commitment to:

- A relativist ontology: where realities exist as situated constructions;
- A subjectivist epistemology: where the findings are recognised to be a product of the interaction between the research subject, be that participants, groups, cultures or situations, and the researcher; and
- A hermeneutic, dialectic methodology: concerned with interpretation and refinement.

3.4.4 Theoretical Perspective: Justifying methodology

A theoretical perspective provides the context for the process and grounds its logic and criteria. Several authors have noted the lack of theoretical models relating to predictive genetic testing and, in particular, high-risk families (Cull *et al.*, 1999;McAllister, 2001;McAllister, 2002;Rees *et al.*, 2001;Reeves *et al.*, 2000).

As discussed in Chapters One and Two, the literature is increasingly recognising that talking to one's family about genetic testing and its risk implications is significantly more than disclosing test results as a discrete activity (Gaff *et al.*, 2007a). Rather, it is a process that may occur throughout the whole journey of genetic counselling and genetic testing. As we move away from the assumed sender-receiver model of communication (Wiseman *et al.* 2010) and towards more

of a 'family affair' (Foster *et al.*, 2002), a family systems perspective is been proposed as a potentially useful framework for understanding family issues in genetic testing (Ross *et al.*, 1990).

Previous studies have used Family Systems theories in the analysis or explanations of findings relating to genetic testing (Blandy *et al.*, 2003;Foster *et al.*, 2004a;Kenen *et al.*, 2004b;McGivern *et al.*, 2004;Mellon *et al.*, 2006;Peterson *et al.*, 2003;Bakos *et al.*, 2008;Carlsson *et al.*, 2004;Clarke *et al.*, 2008;Wilson *et al.*, 2004;Harris *et al.*, 2010). According to Wiseman *et al.* (2010), the use of Family Systems and other social and political theories has enabled researchers to suggest that individual and wider contexts such as gender, culture and biology affect communication within families (p. 701).

According to Broderick (1993), Murray Bowen's Family Systems Theory is a theory of human behaviour that views the family as an emotional unit and describes the complex interactions within the unit using systems thinking. It assumes that family members are intensely connected and reactive to changes in each other's functioning. As such, the theory suggests that individuals cannot be understood in isolation from one another, but rather as a part of their family (Broderick, 1993;Peterson, 2005). In other words, the Family Systems approach proposes that behaviour may have as much to do with the 'systems', the family, and the patterns that are established within that system, as it does to do with the personality of the individuals within the system. So, behaviours can only be understood by looking at the relationships and interactions among all family members (Broderick, 1993). Critiques of the family systems theory have raised concerns about empirical limitations associated with its generating descriptive rather than explanatory abilities (Gavazzi and Gavazzi, 2011).

Peterson (2005) proposes that a family systems perspective is a most suitable framework for conducting family-based research in hereditary risk and genetic testing because it accounts for the reciprocal nature of family relationships, the broader social context in which families exist, and the multiple dimensions that comprise family functioning. Peterson's version of the family systems model for family-based genetic research uses three domains that, according to Walsh (1998), are characteristic of well-functioning families: organisation and structure of family relationships, family communication, and health-related cognitions and beliefs shared within families. These three domains influence how families respond to external stressors, such as chronic illness; changes in the family unit as whole will in turn affect individuals (Djurdjinovic, 1998;Rolland, 1994). According to Peterson (2005), these domains may help guide the selection and organisation of variables in family-based research and intervention development regarding genetic testing.

For example, Harris *et al.* (2010) used Peterson's version of the family systems model to investigate cancer risk communication within melanoma families by examining the relationship between familial organisation and structures, shared health-related beliefs, and communication in families at increased risk of developing melanoma. Following Peterson's model, they hypothesised that a family's organisational and structural characteristics, especially adaptation, coping and cohesion, plus shared familial beliefs about melanoma, would together predict the frequency and style of family communication about melanoma risk (Harris *et al.*, 2010). According to the authors, all family organisational variables (family coping, adaptation and cohesion) and familial health beliefs were found to be associated with an open style of communication. Those individuals reporting more active styles of coping and higher levels of adaptation and cohesion were more likely to have open communication about melanoma within their family.

Building on this, the theoretical perspective of this doctoral study holds that how and when those undergoing genetic testing for *BRCA1/2* talk to their relatives about genetic testing will be influenced by their family's organisational and structural characteristics, plus shared familial beliefs, as shown in Figure 5.

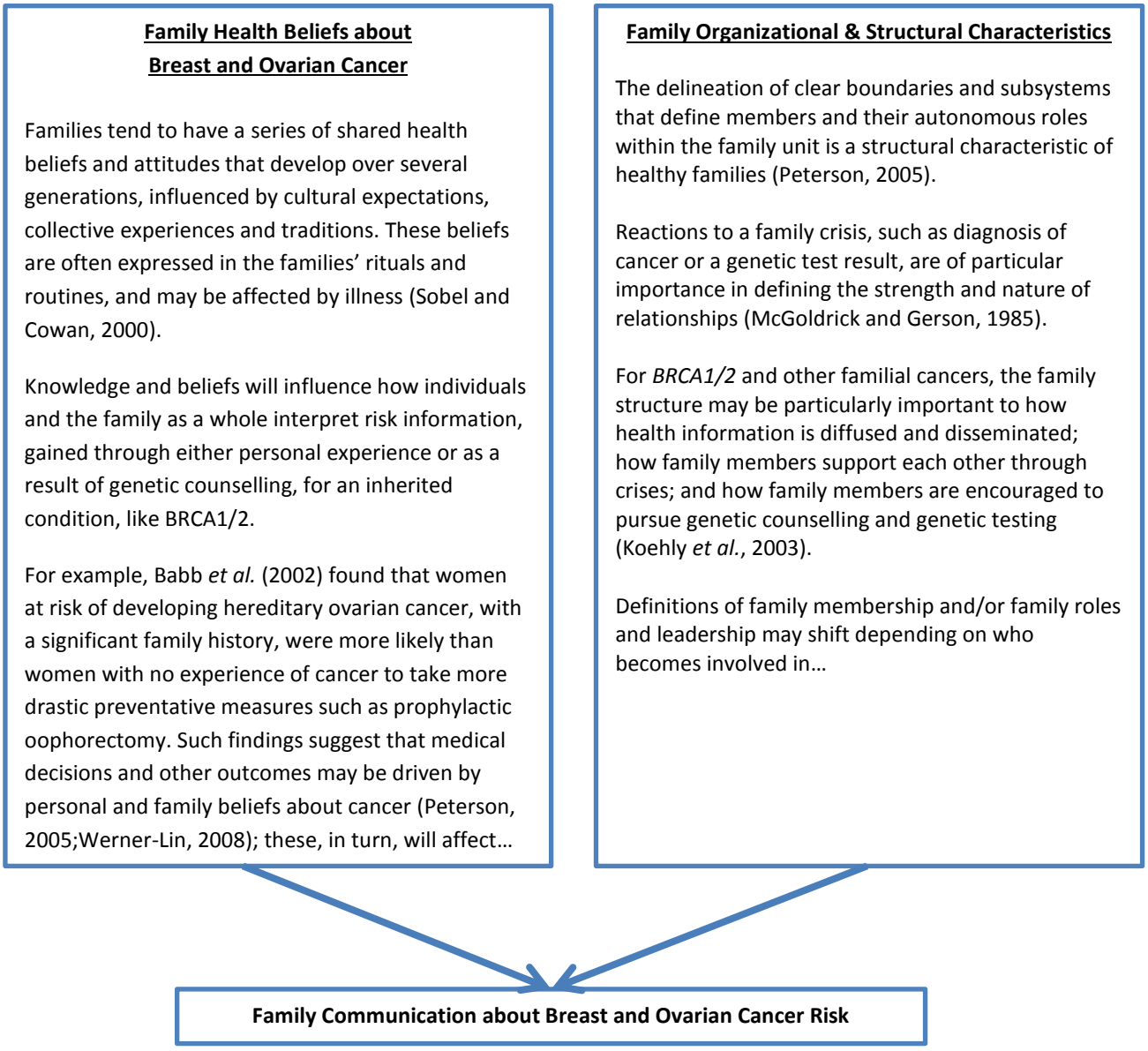

Figure 5: The conceptual model of the relationship between family functioning and family communication; adapted from Harris et al.'s (2010) conceptual model for analysing the relationship between family functioning and family communication about melanoma risk

Given its basis within a Family Systems perspective, it could be presumed that, in order to use such a conceptual model to look at how those undergoing genetic testing for *BRCA1/2* talk to their family, it would be necessary to conduct research with the family as a whole rather than an individual within the family. However, whilst talking about genetic testing and its risk implications may be a 'family affair', as part of the process the individuals undergoing the genetic testing for *BRCA1/2* may need to interact with their families in a new and unfamiliar manner (Peterson, 2005). Moreover, conceptualising families in research poses a number of challenges; for example, the family is a subjective concept that can vary by ethnicity and culture, and its structure can be described in terms of biological as well as social relationships (Peterson, 2005).

Therefore, in order to explore the how and when those undergoing testing talk to their family, it is important to capture the experience of the individual, and their definition of who constitutes their family, within that. Carter and MGoldrick (1989) argue that it is extremely difficult to think of the family as a whole because of the complexities involved; essentially, 'a family is more than just the sum of its parts' (p. 4). The 'Family Life Cycle' theory (Carter and McGoldrick, 1989) offers an understanding of what happens in families in terms of the flow of life over generations, and can be used to examine variables that affect the course of family development including chronic illness. Importantly, at the centre of the 'Family Life Cycle' sits the view that you cannot understand the family without understanding the experience of the individual.

The family life cycle perspective can be used as conceptual framework to study how individuals within a family, and by association the family as a whole, adapt to chronic illness in order to improve care provided by nurses and other health care professionals (Newby, 1996). It provides a conceptual framework from which to view pathology within both the individual and the family. Essentially, the view is that the individual life cycle takes place within the family life cycle, which

is in the primary context of human development. Families are shaped by people who share a history and a future together and, although the family process is by no means linear, it exists in the linear dimension of time. As such, families can be described as 'living systems moving through time'.

The family life cycle is a complex phenomenon, but the theory is based on the emotional and intellectual stages passed through from birth to death as a member of a family. The progress from one life cycle stage to another is referred to as 'transition'. Relationships with parents, siblings and other family members go through transition points as one moves along the life cycle, just as parent-child or spouse relationships do. At each stage, there are challenges to family life that cause the individual family members, as well as the family as a whole, to develop or gain new skills to help them work through and adapt to the changes encountered. These are referred to as 'developmental processes'. The evidence suggests that family stresses, which are likely to occur around life-cycle transition points, frequently create disruptions of the life cycle and produce symptoms and dysfunction. The greater the anxiety generated in the family at any transition point, the more difficult or dysfunctional the transition will be.

According to Dudok de Wit *et al.* (1997), in *BRCA1* families and families with extensive experience of hereditary disease, normative change may be overshadowed by the presence or threat of illness and untimely death. For example, early childhood experiences, or experiences coinciding with the onset of puberty, of a loved one being diagnosed with cancer can have an extensive impact on psychosocial and sexual development throughout adolescence and into adulthood (Werner-Lin, 2008). Typical tasks of early adulthood, such as finding a partner and planning a family, may be contemplated against the backdrop of an expected illness timeline (Werner-Lin, 2007).

Carter and McGoldrick (1989) describe the flow of anxiety as being both 'vertical' and 'horizontal', as shown in Figure 6[9]. The vertical stressors on the system are all the family's attitudes, taboos, expectations, labels and loaded issues which we grow up with, and which are transmitted down the generations of a family; for example, in the case of *BRCA1/2*, how cancer is perceived and dealt with by the family (Werner-Lin, 2008). The horizontal stressors on the system are those that the individual and the family encounter as they move forward through time. These include developmental stressors, for example: coping with the changes and transitions of the family life cycle, and unpredictable events that may disrupt the life cycle process, for example: untimely death, chronic illness, or wars.

According to Carter and McGoldrick (1989), 'the level of anxiety engendered by the stress on the vertical and horizontal axes at the points where they converge is the key determinant of how well the family will manage its transition through life and when the horizontal (developmental) stress intersects with a vertical (transgenerational) stress there is a quantum leap in anxiety in the system' (p. 9). Each family member affects the others and the added element of intergenerational dynamics further complicates the interactions (Ferguson, 1979).

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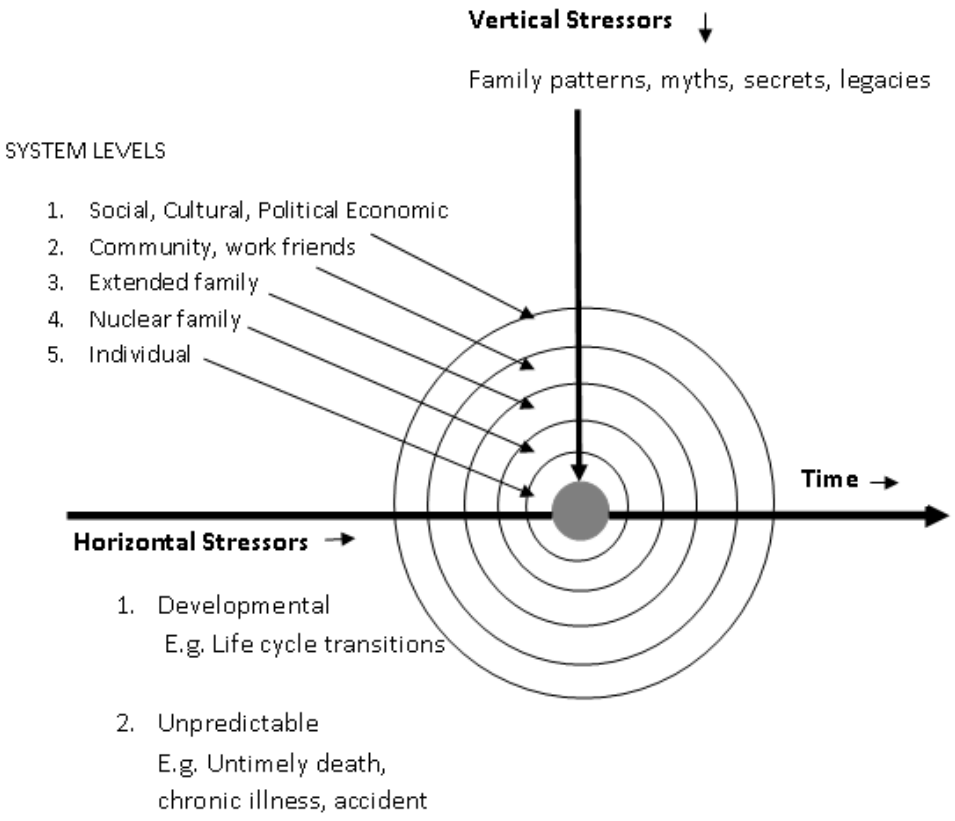

Figure 6: Horizontal and Vertical stressors on the individual and family life cycles. Taken from Carter and McGoldrick (1999, p. 9)

The major strength of the family life cycle perspective is the ability to view the dynamic nature of the family over long periods of time. However, it does make certain assumptions: that it is the stage in family development rather than the age that is important; whilst individual development is important, the development of the family as a group of interacting individuals is more important; transition from one stage to another is going to happen; families and individuals progress through a series of similar developmental stages and face similar transition points and developmental tasks; and, finally, to understand the family we must consider the challenges they face in each stage, how well they resolve them and how well they transition to the next stage. As such, it has been criticised for being unable to account for different family forms, gender, ethnic and cultural differences, and for not being culturally relevant or sensitive to other lifestyle choices.

Although chronic illness has a profound effect upon the individual concerned, an immense responsibility is simultaneously imposed upon the family (Shaw and Halliday, 1992). As such, chronic illness increases family stresses and requires family members to continually adapt. In return, chronic illness creates challenges to health care professionals, who must also adapt to understand and meet the changing needs of both the family as a whole and the individuals involved (Newby, 1996). The family's response to chronic illness will vary according to: the age and the developmental stage of the ill individual; the family's strength and coping mechanisms; and family life cycle stage (Newby, 1996). Families with hereditary breast and ovarian cancer may be confronted with regular cancer diagnosis and untimely deaths from generation to generation (vertical stressors). Because of the autosomal dominant hereditary and penetrance patterns, there is a constant uncertainty about which family individual members cancer may affected, or when or what the outcome will be (horizontal stressor) (Werner-Lin, 2008).

Combrinck-Graham (1985) presented a Developmental Model of Family Systems that emphasised changes in family shape through the individual life cycle. The family system is described as oscillating through time between periods of family closeness (centripetal) and periods of family disengagement (centrifugal). According to Rolland (1987), the concept of centripetal and centrifugal modes is useful in linking the illness life cycle to the individual and family life cycles. In most cases, chronic disease exerts a centripetal pull on the family system. Symptoms, loss of function, the demands of shifting or new illness-related, practical and affective roles, and the fear of loss through death all serve to refocus a family inwardly. If the onset of an illness coincides with a centrifugal period for the family, it can derail the family from its natural momentum. Therefore, the Family Life Cycle perspective brings the individual undergoing genetic testing back to the centre of the study; however, in order to explore their

experiences of talking to their relatives, it is important to recognise, and gather data, on their family context. One way to achieve this is through the introduction of an eco-map or genogram.

Daly *et al.* (1999) constructed genograms for 38 high-risk women to pilot the efficacy of using genograms for assessing family relationships. They compared the degree of family cohesion as depicted by the genogram, with scores obtained on the standardised Social Adjustment Self-Report (SASSR); from this, they reported a positive correlation ($p=0.01$), thus demonstrating the validity of the genogram. That is, 'the higher the percentage of close and very close relationships represented on the genogram, the greater the social adjustment recorded on the SASSR subscales' (Daly *et al.*, 1999). This method allowed them to convert qualitative data into a graphical representation of family dynamics that may be used in comparative and longitudinal research and counselling settings. However, the authors do recognise that there are limitations. Namely, interviewer bias and the subjective nature of the information recorded in the genogram by the counsellor, although some of this may have been limited by standardising both the content and the structure of the questions asked and adhering to defined symbols and nomenclature. There is also the possibility that women involved answered questions about their familial relationships in a socially acceptable way in order to present themselves and their families in a positive light (Daly *et al.*, 1999).

Genogram construction was originally rooted in Murray Bowen's Family Systems Theory (McGoldrick and Gerson, 1985). According to this perspective, behaviour patterns within families are often stable and transmissible over time. Therefore, genogram construction can be used to examine patterns of function and relationships from one generation to the next (Daly *et al.*, 1999). How eco-maps were used in this study will be discussed in section 3.4.4.

To summarise, the resultant study, as presented in this doctoral thesis, was qualitative and grounded in a conceptual model adapted from Peterson's (2005) family systems model, for conducting family-based research in hereditary risk and genetic testing, as well as Carter and McGoldrick's (1989) 'Family Life Cycle', which allows the study of how individuals within a family, and by association the family as a whole, adapt to chronic illness. The technique of constructing eco-maps (Ray and Street 2005) was employed as a method of identifying relevant family members and guiding the researcher through the family structure and relationships.

3.4.5 Methodology: Justifying Method

The goal of qualitative research is the development of concepts that help us to understand social phenomena in natural (rather than experimental) settings, giving due emphasis to the meanings, experiences, and views of all the participants (Pope and Mays, 1995). To achieve this, several theoretical approaches to conducting qualitative research have been developed, for example phenomenology, grounded theory, ethnography and their variations (Avis, 2003). These describe, either explicitly or implicitly, the purpose of the qualitative research, the role of the researcher(s), the stages of research, and the method of data analysis. The aim of this present study is to examine how and when participants talked to their relatives about genetic testing and the risk implications; this could be done via several different methodologies and, thus, methods. However, the choice of methodology will shape the exact nature of the research questions and the study design.

For example, if the research was approached using an ethnographic methodology, then the question would move to studying family communication regarding genetic testing in, or as, a culture, perhaps examining the culture at the genetics service and place of family

communication regarding genetic testing within it. If, on the other hand, a grounded theory methodology was used then the aim would now be to develop a substantive theory. However, with a phenomenological approach, the emphasis would shift to one of seeking to uncover the essence or meaning of the experiences of those people undertaking family communication regarding genetic testing. So, whilst the methodology affects the research questions and the study design, conversely, they themselves will influence the choice of methodology.

Avis (2003) argues that, because positivism encapsulates the epistemological assumption that the foundation of genuine knowledge is from empirical science based in verification, objectivity and reproducibility, qualitative researchers feel obliged to justify their different ethical, ontological and epistemological commitments by adhering to one of three traditional methodological approaches, namely phenomenology, grounded theory or ethnography. According to Thorne *et al.* (1997), in the past, methodological variations from these traditions have not been encouraged, particularly in the field of nursing research, as it was deemed 'sloppy' research (Morse, 1989a). The result is a danger of claiming to reveal lived experience by 'doing phenomenology' but using an 'ethnographic interview' method and then applying 'grounded theory analytical approaches', such as constant comparison (Thorne *et al.*, 1997).

3.5 Interpretive Description as a Methodology

In response to an expressed need for an alternative method for generating grounded knowledge pertaining to clinical nursing context, Thorne *et al.* (1997) developed interpretive description.

Interpretive description, as presented by Thorne *et al.* (1997;2004b), is grounded in an interpretive paradigm. It acknowledges the constructed and contextual nature of much of the health-illness experience, yet also allows for shared realities.[10] As a methodological approach, it is designed to specifically fit the kind of complex experimental questions that nurses and other applied health researchers might be inclined to ask (Thorne *et al.*, 2004b). Key characteristics of naturalistic (qualitative) inquiry, such as those described by Lincoln and Guba (1994), provide the philosophical underpinnings for the research design, including:

- There are multiple constructed realities that can be studied holistically. Thus, reality is complex, contextual, contrasted and ultimately subjective.
- The inquirer and the 'object' of inquiry interact to influence one another; indeed, the knower and the known are inseparable.
- No *a priori* theory could possibly encompass the multiple realities that are likely to be encountered; rather, resulting theory must emerge or be grounded in the data.

The purpose is to capture themes and patterns within subjective perceptions and to generate an interpretive description capable of informing clinical understanding (Thorne *et al.*, 2004b).

An interpretive description methodology has been successfully used in health and social research, including but not exclusively: examining the experiences of women diagnosed with breast or ovarian cancer who received inconclusive *BRCA1/2* genetic test results (Maheu and Thorne, 2008); exploring communications throughout the cancer trajectory from the perspective of patients (Thorne *et al.*, 2010); identifying health care communication issues in multiple sclerosis (Thorne *et al.*, 2004c); mapping patterns of practice of arts therapists working with

[10] An assumption that reality, and hence the phenomenon under study, are constructed both within the individual and through social interaction. Consequently, multiple, dynamic, and potentially contradictory realities are assumed to exist. These realities are best understood through collaboration between researcher and participants in which the social worlds of the participants are brought together through the researcher (Rosenwald, 1988).

older people who have Dementia in the UK (Burns, 2009)[11]; an inquiry into the moral experience of clinicians in humanitarian work (Hunt, 2009a);[12] and exploring Aboriginal women's experiences of cervical cancer screening (Duchcherer, 2010).

Unfortunately, as a relatively new qualitative methodology, there has been little discussion in the literature of the particular merits and limitations of this methodological framework at this time. However, Hunt (2009b) has published an experience of using interpretive description as a methodology that aims to identify and discuss strengths and challenges that can arise in its application. Strengths identified include a coherent logic and structure, an orientation toward the generation of practice-relevant findings, and attention to disciplinary biases and commitments. Challenges include limited examples of its use for situating the methodology because it is new, challenges in employing a lesser-known methodology, and uncertainty regarding the degree of interpretation to seek (Hunt, 2009b).

With regards to this last challenge identified by Hunt, the terms ‘‘interpretive’’ and ‘‘description’’ seem contradictory in qualitative research. Qualitative ‘description’ may be perceived as superficial analysis when compared with the in-depth ‘interpretation’ that qualitative researchers aim for (Pope *et al.*, 2000). Interpretive description should not be mistaken for qualitative description or pattern analysis. The main difference between interpretive description and qualitative description lies in the data analysis, where interpretative description goes beyond a simple description and aims to provide an in-depth conceptual description and understanding of a phenomenon, whereas qualitative description stays closer to the data obtained (Neergaard *et al.*, 2009). The analytic procedures in interpretive description

[11] PhD thesis.
[12] PhD thesis.

capitalise on such processes as synthesising, theorising and recontextualising, rather than simply sorting and coding.

The important thing to note is that an interpretive description does not just provide an in-depth description of the phenomenon under study. As stated by Thorne *et al.* (2004b), interpretive description uses inductive analytic approaches to 'seek understanding of clinical phenomena that illuminate their characteristics, patterns and structure…' (p. 6). In other words, it aims to create a qualitative description that can be characterised as interpretive. There is an interpretive orientation, however, in that it is not intended to yield new theory or high-order abstractions; instead, researchers should pursue the interpretation to the degree that it will yield useful insights to guide clinical practice (Hunt, 2009b).

The general principles of interpretive description as a form of qualitative inquiry, with solid grounding in practice knowledge and nursing science, are embedded within five key components: the analytical framework, the sample selection, the data sources, the data analysis and rigour.

3.5.1 Analytic Frameworks

Unlike traditional phenomenological inquiry, when conducting interpretive description research, 'existing knowledge' should be considered the 'foundational forestructure' to a new inquiry (Schultz and Meleis, 1988), so that findings can be developed on the basis of thoughtful linkage to others' work (Mitchell and Cody, 1993). Interpretive description presumes there will be some theoretical knowledge, clinical pattern observation and scientific basis within which all studies of

human health and illness phenomena are generated. A critical review of current knowledge forms the basis for the preliminary ‘‘analytical framework’’ with which the investigator makes sampling, design and early analytical decisions. An analytical framework of this nature stems from critical analysis of research literature and/or clinical interpretation, and provides a starting point for research design and the inductive reasoning for interpretation of meanings within the data (May, 1989). Because it represents a starting point for the research rather than an organising structure for what is found, it will typically be challenged as the inductive analysis proceeds (Thorne *et al.*, 1997).

For this study, an analytic framework, in the form of a summary of findings and identification of gaps, was developed through a critical analysis of research literature, which is presented in this thesis in the form of background literature (Chapter One) and a systematic review and meta-synthesis (Chapter Two). The searching, reading and critiquing of the literature related to family communication regarding diagnostic and predictive testing for hereditary cancers were the first activities undertaken as part of this research. As discussed at the beginning of this chapter, the findings identified areas where little research evidence was available and subsequently informed the development of the research question, aims and objectives of this study. The knowledge gained during these activities also shaped the interview guide used during the qualitative interviews (Appendix 3) and allows the findings to be discussed in relation to previous research in the field (Chapter Nine).

3.5.2 Sample Selection

Sampling and data collection methods should be derived from specific research questions, informed by the framework of what is already known about the phenomenon from a range of

sources (Thorne *et al.*, 2004b). Samples are purposively, often theoretically, generated, thus reflecting an awareness of expected and emerging variations within the phenomena being studied (Thorne *et al.*, 2004b).

Theoretical sampling is built into the design of interpretive description research, with particular emphasis on obtaining maximal variation on the themes that emerge from the inductive analysis itself (Glaser, 1978;Sandelowski, 1995a), the argument being that usually the positions or experiences that each participant or informant might represent cannot be known until data collection is underway (Thorne *et al.*, 1997). Unfortunately, there are many variations of qualitative sampling described in the literature and much confusion, as the terms 'purposeful' and 'theoretical' are often viewed as synonymous and used interchangeably (Coyne, 1997). Theoretical sampling is the principal strategy for grounded theory methodology, as it calls for building interpretative theories from the emerging data and selecting a new sample to examine and elaborate on this theory (Marshall, 1996).

The decision not to use a theoretical sampling strategy of this nature for this study was taken for two reasons. Firstly, the data protection regulations when conducting research with NHS patients meant that it was not possible for the researcher to access patient information in order to screen for eligibility into the study. For example, if during the ongoing data analysis it had become a requirement to select only those who had a brother, there would have been no way of identifying these patients as the researcher did not have access to patient notes, if indeed that information was even available within the notes. And, whilst it may have been possible for the staff at the genetic service to identify suitable participants, it would have been a time-consuming activity and placed an unnecessary burden on the service. The second reason for not using theoretical sampling was because the nature of the PhD studentship meant there was a time constraint for conducting the interviews. So, a sampling strategy that allowed the identification

of potential participants, recruitment, interviewing and transcribing to happen simultaneously was beneficial.

However, the analytic framework developed as part of the initial literature scoping and critical appraisal activities had already identified several limitations in the research on family communication regarding genetic testing for *BRCA1/2*. Specifically, the research tends to focus on communication at the point of result disclosure by those who had undergone a particular type of testing (for example, predictive genetic testing only), or received a particular genetic test results (for example, including only those who had received a positive *BRCA1/2* test result) with a specific family member (for example, a sister). To really understand how these families discuss genetic testing for cancer risk, and when this communication occurs, it would therefore be necessary to recruit from the following: those who had undergone both diagnostic and predictive genetic testing; both men and women; those with a variety of result statuses; and from a variety of ages. Therefore, a method of purposive sampling was employed to include a sample who could best help understand the studied phenomenon through their personal experiences (Crossley, 2007). The methods used for sampling will be discussed more fully in section 4.2.

3.5.3 Data Sources

In keeping with many qualitative methodologies, interpretive description is based on the belief that people who have lived with certain experiences are the best sources of expert knowledge regarding those experiences (Morse, 1989b). Thorne *et al.* (1997) also suggest that appropriate collateral data sources, such as lay print or other media information, as well as case reports and clinical papers, are often readily available for qualitative researchers interested in expanding the scope of their inquiry and broadening the reach of their theoretical sample, without incurring the excessive monetary and time costs of intensive interviewing and participant observations. Throughout the study, the researcher made regular visits to the Genetics Service where the research was based and collected various patient-information sheets. Popular media items, for example television programmes or newspaper articles on genetic testing for cancer risk, were constantly reviewed. These data sources were not directly incorporated into the study findings; but, instead of regularly reviewing them, for example throughout the data analysis process, the researcher aimed to immerse herself within the world under study and to encourage thinking and interpretation through the eyes of the participants.

For this doctoral study, data were collected in one-time, individual, semi-structured interviews. The in-depth interviews were chosen because they are a relatively flexible data collection method: allowing the researcher to prompt for more information, and the participants to explore their own thoughts, react to questions spontaneously and honestly, and articulate their ideas slowly whilst reflecting on them (Holloway and Wheeler, 2002). Eco-maps (Ray and Street, 2005) were used as a secondary data source in this inquiry. They provided a method of identifying relevant family members and guiding the researcher through the family structure and relationships (refer to section 4.5).

3.5.4 Data Analysis

Interpretive description uses inductive analytic approaches to seek an understanding of clinical phenomena that illuminates the characteristics, patterns, and structure in some theoretically useful manner (Thorne *et al.*, 2004b). The final product from an interpretive description 'is a coherent conceptual description that taps thematic patterns and commonalities believed to characterise the phenomenon that is being studied and also accounts for the inevitable variations within them' (Thorne *et al.*, 2004b). The product should also have application potential, 'in the sense that a clinician would find the sense in them and they would therefore provide a backdrop from assessment, planning and interventional strategies (Thorne *et al.*, 2004b).

Interpretive description should involve a rigorous analytical process that involves carefully navigating within and beyond the analytical framework with which the research started, in order to fully engage the process of inductive reasoning, including testing and challenging preliminary interpretations, and conceptualising an ordered and coherent final product (Thorne *et al.*, 2004b). Many textbooks and papers describe different methods for the analysis of qualitative data often based on different research methodologies; however, there are some common features to analytic practices that may be used across different qualitative research types, a classic set include (Miles and Huberman, 1994):

- Affixing codes to a set of field notes drawn from observations or interviews.
- Noting reflections or other remarks in the margins.
- Sorting and sifting through these materials to identify similar phases, relationships between variables, patterns, themes, distinct differences between subgroups, and common sequences.

- Isolating these patterns and processes, commonalities and differences, and taking them out to the field in the next wave of data collection.
- Gradually elaborating a small set of generalisations that cover the consistencies discerned in the database.
- Confronting those generalisations with a formalised body of knowledge in the form of constructs or theories.

The method used for the data analysis of the in-depth interviews was based primarily on the work of Miles and Huberman (1994). This will be fully outlined during the next chapter (refer to section 4.7).

Interpretive description requires analytical techniques that encourage repeated immersion in the data prior to coding, classifying or creating linkages (Thorne *et al.*, 1997), with an emphasis on synthesising, theorising and recontextualising rather than simply sorting and coding (Morse, 1994). In order to address the dialectic between individual cases and common patterns, the researcher must intimately know the individual cases, and produce explanations of overall findings that can be applied back to the individuals (Thorne *et al.*, 1997). Strategic periods of immersion in the field interspersed with periods of immersion within the data are recommended to encourage refinement of the inquiry, as well as testing of developing conceptualisations and emerging theories (Lofland, 1976;Strauss, 1987).

To allow 'immersion in the field', regular visits were made to the Genetic Service during the time when interviews were being conducted and data analysed. This included observing some genetic clinic consultations (after recruitment had been completed so as not to come into contact with potential participants). This allowed the researcher to keep in touch with the clinical staff and witness first-hand the process of genetic counselling and testing that the participants had been

through in order to put the experiences into context. Searching for and reading of emerging literature also continued during this time.

3.5.5 Rigour and Validity

As with other qualitative methodologies, issues of rigour and credibility are an important consideration when creating an interpretive description (Thorne *et al.*, 2004b). According to Thorne *et al.* (1997), an interpretive description requires the researcher to record their analytical journey, for example keeping a reflective journal and/or field notes. Research reports should provide sufficient information for the reader to be able to follow the analytic reasoning process and to judge the degree to which the analysis is grounded within the data. Steps should also be taken to ensure the theoretical validity of the findings (Brink and Wood, 1989). Typically, repeated interviewing, in which developing conceptualisations can be subjected to challenges or refinements, is built into the design of an interpretive description. Thorne *et al.* (1997) believe that taking the raw data (for example, the interview transcripts) back to participants for a credibility check is generally insufficient for these purposes and may create contradictions (Sandelowski, 1993). Instead, they recommend presenting the initial conceptualisations, representing the entire sample rather than the individual research participant, to individual research participants for their critical consideration. They believe this strategy creates optimal conditions for challenging the emergent theories and refining the theoretical linkages (Thorne *et al.*, 1997).

Time constraints, as well as the lack of ethical approval or participant consent to present the initial conceptualisations to the research participants during the early stages of the study, meant this was not feasible. According to Maheu and Thorne (2008), another way of considering the

validity of findings in an interpretive description study is by reflecting on how they match or enlarge the clinical hunches of expert clinicians familiar with the study phenomenon. Therefore, in order to have confidence in the theoretical validity of the findings, the conceptual framework and research findings were regularly presented to the clinical team at the Genetic Service and at national conferences[13] at various points during the analytical process. Their feedback and discussions helped shape the work and gave insight into how the work related to their and their patients' experiences of family communication regarding genetic testing for *BRCA1/2* in their clinical practice.

3.6 Conclusion

Having established the foundations of the study, decisions have to be made about the research practice; in other words, the methods. As has already been described, the epistemological, theoretical and methodological positions are all interrelated; and all influence, and are influenced by, the choice of the method. There is no one 'correct' method associated with any one type of research; there are, however, options more sympathetic to the type of research question and study aims. For example, interview data would provide a deeper understanding of experiences compared with a questionnaire-based method. The next chapter, Research Methods, will describe in-depth the methods used for data collection and data analysis.

[13] National Cancer Research Institute (NCRI) Conference 2008 & 2009; the British Society of Human Genetics (BSHG) Conference 2008; and the International PsychoOnlogy Society (IPOS) conference 2011.

Chapter 4 - Research Methods

4.1 Introduction

The chapter will systematically describe and justify the methods chosen for the study. The aim of the study is to gain insight into participants' experiences of discussing their participation in genetic testing, their test results and potential risk information following genetic testing for *BRCA1/2* with their family; particular focus will be on *how* these participants discuss genetic testing for cancer risk with relatives and *when* this communication occurs. It was qualitative in nature, employing in-depth interviews as the method for data collection and utilising the technique of constructing eco-maps (Ray and Street, 2005) as a method of identifying relevant family members and guiding the researcher through the family structure and relationships. These methods were chosen in line with the conceptual framework (refer to section 3.4.4) and the interpretive description methodology (refer to section 3.5) to ensure depth and richness in analysis and reporting of findings.

4.2 Population and Sampling

One of the limitations of previous research in the field, as identified in Chapters One and Two, is that studies tend to limit themselves to particular groups of people, for example those who received a positive result from predictive genetic testing. Therefore, the aim was to be as inclusive as possible to gain a wide range of experiences.

The sample was drawn from individuals who had attended a clinical Genetic Service in the South-East of England and undergone diagnostic or predictive genetic testing for *BRCA1/2,* regardless of their result. Evidence in the literature suggests that, on average, it takes eight months for familial communication to occur following predictive genetic testing (Julian-Reynier *et al.*, 2000b). After discussion with the clinical team at the Genetic Service about how many *BRCA1/2* tests they conducted each year and the likely response rate, an upper limit of 18 months was set. It was felt that this timeframe meant the experience of family communication regarding genetic testing for *BRCA1/2* would be recent enough to be recalled accurately. The study, therefore, included those individuals who received their test results eight to 18 months prior to being invited to participate.

Men and women were considered eligible for the study according to pre-set inclusion criteria as follows:

- They had undergone genetic testing (diagnostic or predictive) for *BRCA1/2* mutations;
- With eight to 18 months having passed since receiving their results of the genetic testing;
- They were asymptomatic for breast or ovarian cancer (to ensure their experience is not influenced by any present symptoms);
- They were at least 18 years of age; and
- They were able to understand and speak English (in order to give consent and be able to complete the interview successfully).

No exclusion that might lead to discrimination because of gender, age, or social class was permitted; however, any potential participants who were known by the Genetic Service team to

have a psychiatric illness that was likely to be exacerbated by involvement in the study were excluded.

The study was reviewed and given a favourable opinion by the NHS Trust Research and Development Office (R&D number: CAN0568) and the Isle of Wight, Portsmouth & South East Hampshire Research Ethics Committee (REC reference number: 07/H0501/100). Once all research governance and ethical procedures were completed, potential participants were selected via the clinical genetic laboratory computer database using purposive sampling. This database keeps records of all genetic testing done within the Genetic Service, and it was possible to search for patients according to type and date of testing. This was done by the staff at the Genetic Service, in line with data protection guidelines and stipulations of the Research Ethics Committee. Once the list had been generated, it was presented at the multi-disciplinary team meeting so the practitioners at the Genetic Service could advise as to whether they knew of any reason why the potential participant should not be invited to participate in accordance with the inclusion or exclusion criteria; for example, a recent diagnosis of cancer.

As the aim was not to generalise from the sample to the population, it was not necessary to select representative individuals. Instead, the method of purposive sampling was employed to include a sample who could best help understand the studied phenomenon through their personal experiences (Crossley, 2007). For example, to ensure the experiences of both those who had undergone diagnostic genetic testing and those who had undergone predictive genetic testing were represented, approximately equal numbers of each were invited to participate.

The sampling strategy was to invite potential participants in batches of seven patients, with approximately equal numbers of those who had undergone predictive and diagnostic genetic testing in each batch, until the sample frame of approximately 30 participants was achieved.

Selection started with those closest to eight months having passed since testing. The exception to this was when inviting men. Considerably more women had undergone genetic testing for *BRCA1/2* than men during the timeframe selected. This is not unexpected considering women have a significantly higher chance of developing breast cancer in their lifetime than men (Cancer Research UK, 2011a). Other research also shows that, although variable, rates of predictive testing in men are lower than in women (Bodd *et al.*, 2003;Goelen *et al.*, 1999;Julian-Reynier *et al.*, 2000b). Therefore, in order to include the experiences of men, all potential male participants were invited at once, regardless of who had had their test first, or the type of testing.

Sandelowski (1995b) suggests that determining an adequate sample size in qualitative research is ultimately a balance of judgement and experience in evaluating the quality of the information collected against the uses to which it will be put, the particular research method, the purposeful sampling strategy employed, and the research product intended. Patton (1990) maintains that no guidelines should exist for sample size in qualitative research, while sample sizes differ greatly in qualitative studies. That said, although there are no set rules, research texts often mention some kind of guidelines. For example, Kuzel (1999) suggests that 6-8 participants are needed when the sample consists of a homogeneous group, while 12-20 participants suffice for a heterogeneous sample. Charmaz (2006) suggests that 25 participants are 'adequate for smaller projects' (p. 114). Ritchie *et al.* (2003) suggest qualitative samples often 'lie under 50' (p. 84), whereas Green and Thorogood (2009) suggest little 'new' data 'comes out of transcripts after you have interviewed 20 or so people' (p. 120). In fact, Holloway and Wheeler (2002) report that most often the sample consists of between four and 40 participants; however, certain research projects contain as many as 200.

For the 14 qualitative studies included in the Systematic Review and Meta-Synthesis (Chapter Two), the sample size ranged from 12 to 63 participants, with an average of 30.3 per study.

Therefore, a potential sample size of approximately 30 was determined as appropriate to address the research questions for this study. The aspiration was to have a sufficient number of participants to be able to present and represent a range of experiences, but not to have so many as to lose the required depth to the study. The plan was to allow data collection to reach data saturation (Holloway and Wheeler, 2002).

The interpretive description methodology literature suggest sample size should be evaluated on an on-going basis to identify when sufficient density of the data has been achieved (Hunt, 2009b). 'Sufficient' density of data collection, otherwise known as data saturation (Holloway and Wheeler, 2002), occurs when the researcher judges that no new data is being given by the participant to deepening the understanding of the phenomenon (Carnevale, 2002). Therefore, it may have been necessary to increase the proposed sample size, of approximately 30 participants, if new data were still being introduced at the end of these interviews.

4.3 Recruitment

The process taken is outlined in Figure 7. Potential participants were invited to participate in the study by a letter from the Genetic Service (Appendix 4), outlining the study and enclosing the participant information sheet, which detailed what would be involved in the study (Appendix 5). A telephone number was given to contact the researcher to ask further questions about the research if required. There was an opt-in reply slip to return to the researcher with a stamped-addressed envelope (Appendix 6).

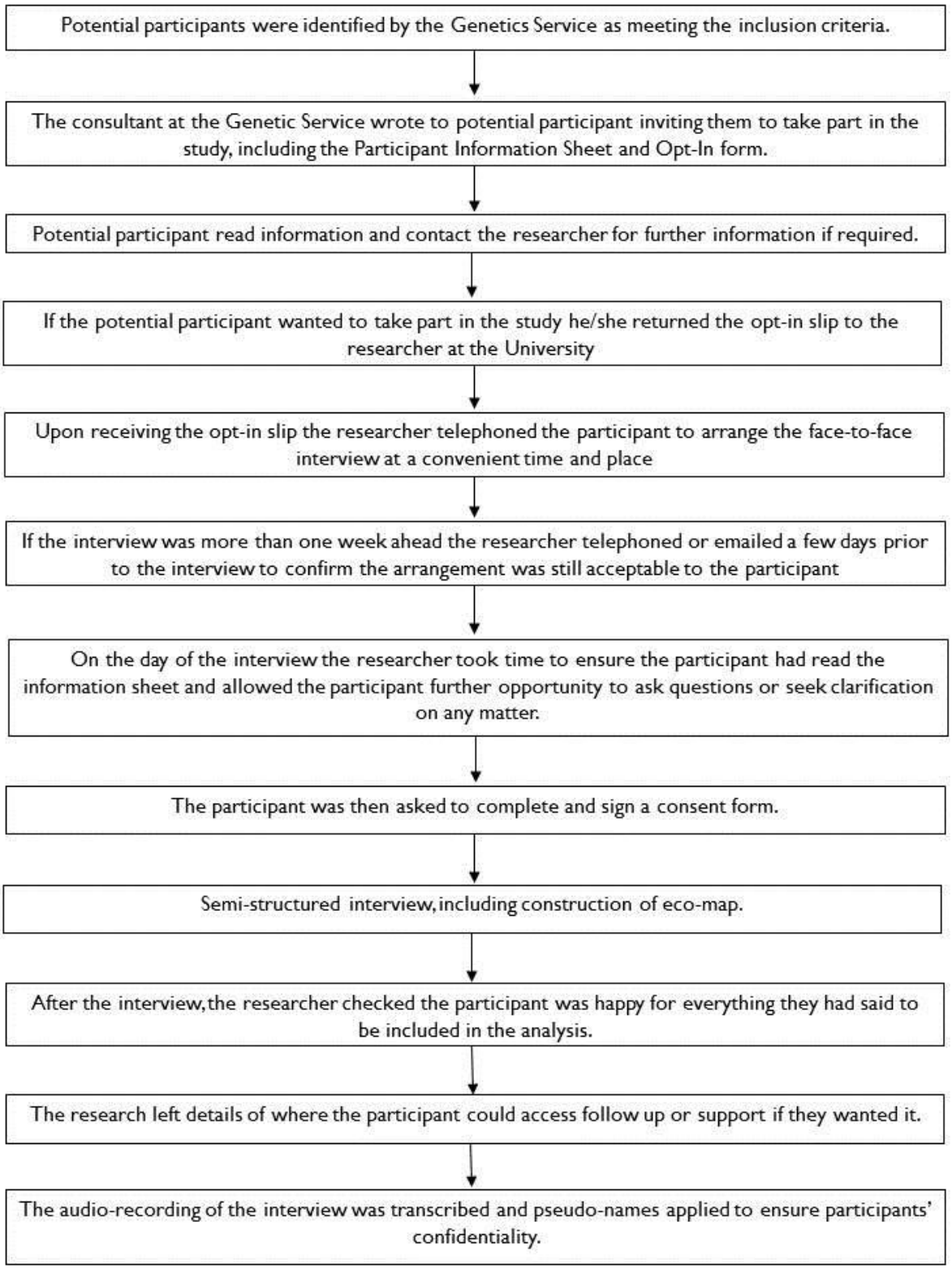

Figure 7: A summary of recruitment and participant interview process

Once the opt-in slip had been received, the researcher rang potential participants to arrange the interview at a time and place convenient to the participant. If a suitable venue, a private and comfortable room with no disruption or interference during the course of the interview, was not available in their homes, they were invited to attend the Faculty of Health Sciences (at either the Highfield or Portsmouth Campus), or another University location, where such rooms were available. Two of the participants chose to be interviewed at the University, both stating that they had busy households at home with teenage children on summer holidays and, therefore, it was unlikely to be a suitable interview environment.

4.4 Consent

On the day of the interview, the researcher ensured the participant had read the information sheet and gave them the opportunity to ask questions or seek clarification on any matter. Participants were advised they may withdraw from the project at any time without having to give a reason. Participants were then asked to complete and sign a consent form which indicated that they were fully informed of the process of participating in the study, provided consent for audio-recording and consented to the material from the interview being used in any subsequent dissemination activities, including publications, while at all times protecting the participant's confidentiality (Appendix 7).

4.5 Data Collection

With participants' consent, the interviews were audio-recorded and lasted between 23 and 106 minutes, with the median length being 54 minutes. Individual interviews were appropriate in order to create a space where participants felt able to talk freely about their views and experiences of the topic under discussion. This was further encouraged by assurances of anonymity and confidentiality for participants. The interviews had a semi-structured nature. This meant that, unlike more structured interview techniques, where the answer is often a choice of predetermined options, there was no forced consistency in people's thinking (Wilkinson *et al.*, 2004). While there was a minimal list of open-ended questions and topics to be addressed, in the form of an interview guide (Appendix 3), there was flexibility for the participant to speak widely on issues and develop ideas (Denscombe, 2003). In line with the interpretive description methodology, the interview guide was developed as part of the analytic framework after an extensive review of the literature and discussion with genetic clinicians. This interview guide acted as a framework only, with the exact order of the questions posed varying according to the flow and direction of the conversation.

Mathieson (1999) describes interviewing as a process of story building; 'a process of two people, the narrator and the listener, with a structure that allows interpretation of meanings' (p. 130). Face-to-face interviews provided in-depth insight into the emotions, experiences and feelings of the participants. Wilkinson, Joffe and Yardley (2004) stress the importance of awareness that the class, race, gender, age and social status of the interviewer will have an impact on the interviewee. While much of this cannot be disguised or avoided, the authors suggest that an open and friendly manner with emphasis on confidentiality and impartiality may overcome this. This was particularly important, as some of the participants may have felt that the issues being discussed were personal and/or sensitive, for example if they had made the decision not to

inform close relatives of their *BRCA1/2* risk; therefore, it was necessary to build up rapport and trust with them through face-to-face contact. The interviewer aimed to be non-judgemental and non-directive at all times to allow open and honest information to be collected.

The interviews were organised into three parts:

4.5.1 Part One of Qualitative Interview

In line with the theoretical basis of the research (refer to section 3.4.4), in order to gain an in-depth understanding of how and when those undergoing genetic testing for *BRCA1/2* discussed their experiences with relatives, it was necessary to gain some understanding of the participant's social context and family relationships. In particular, it was important to examine their family organisational and structural characteristics, as well as health beliefs shared by family members as described in the conceptual model.

Koehly *et al.* (2003) utilised a social network perspective to investigate the relationship between the familial culture and communication about genetic counselling and testing. Family functioning was evaluated by the constructs of communication, cohesiveness, affective involvement, leadership, and conflict. According to Koehly *et al.* (2003), by investigating the interpersonal relationships among a set of individuals, or actors, social network methodology can provide a detailed map of the social environment within which family members interact. This can facilitate an understanding of which aspects of the familial culture influence the discussion and participation in genetic counselling (Koehly *et al.*, 2003). However, social network perspective is a methodology for research rather than a method for collecting data.

A suitable method for data collection may have been to use standardised genogram construction, as demonstrated in previous research concerning genetic testing for cancer risk by

Daly *et al.* (1999). The genogram was first used by Murray Bowen in the late 1970s as part of the Family Systems Theory (McGoldrick and Gerson, 1985) and has been used widely in the practice of the family (Daly *et al.*, 1999). Genograms are designed to provide insight into how the individual functions in the context of the family system and, likewise, how individuals interact as a functional whole (Hartman, 1978). In 1984, McGoldrick and Gerson then standardised genogram construction, thereby providing practitioners with a consistent and reliable structure for recording family dynamics (McGoldrick and Gerson, 1985). Daly *et al.* (1999) found that genograms could be used successfully to identify levels of family cohesion reported by women at high risk of developing familial breast and/or ovarian cancer attending counselling clinics.

However, genograms are widely supported and used in genetic counselling practice (Eunpu, 1997) to record family histories and, as such, are likely to be associated with clinical practice by participants, who will have all been through genetic counselling as part of their genetic testing for *BRCA1/2.* It was important to the researcher to try to separate the research interview from the clinical services offered by the Genetic Service in order to ensure that the research interview was an 'impartial space' where participants could speak freely about their experiences. This was achieved using eco- or communication-mapping:

> 'Eco-maps provide a visual means of facilitating discussions around the structure and strength of networks. Being able to represent the social networks visually through eco-mapping enabled people to identify each member of the network, examine the strength of each relationship and ascertain the sources of nurture and tension over time.' (Ray & Street 2005: 545)(132)(6)

The ecological map, or 'eco-map', was developed and described by Hartman (1978) as a tool to represent social relationships and systems that constitute the interaction between both the

social and physical environments that people live in. The eco-map, designed to be either standalone or be used as a supplement to a genogram, is a tool used to illustrate how the family system is currently connected to outside resources, organisations and agencies. The term 'eco-map' originates from ecology – the study of the connection between a living thing and its environment, and how that connection is maintained and enhanced (Ray and Street, 2005). Traditionally, the three key concepts for the construction of an eco-map are relationships, social networks and support (Ray and Street, 2005); however, this tool was developed further by Martino (2006), who describes the concept of a 'communication eco-map' which aims to merge geno- and eco-maps into one tool based on family communication patterns. The use of genograms and eco-maps in research is well described in the literature.

Peters *et al.* (2006) examined the feasibility and acceptability of the collared eco-genetic relationship map (CEGRM)[14] in a familial cancer genetic research setting. The participant and the researcher constructed the CEGRM together and then the researcher used a semi-structured interview to guide the participant through the process of placing various colour-coded symbols on the pedigree, at the appropriate location. Twenty women (mean age 44 years) from *BRCA1/2* mutations-identified families found including a CEGRM into a research interview to be feasible and comfortable to do, and was efficiently accomplished in usually less than 30 minutes.

Although the focus for this present study is *how* and *when* participants talked to their relatives, visually mapping *who* participants considered to be in their family network allowed: (a) a visual representation on which to base further discussion and exploration of the *how* and *when*; (b) information collected to be placed into a familial context, allowing easier examination of organisational and structural characteristics; (c) insight into how discussions on *BRCA1/2* genetic

[14] A CEGRM is a novel psychosocial assessment tool, which incorporates features of the genetic pedigree, family systems genogram and eco-maps designed to allow the clinically-oriented researcher to visually and conceptually organise information about study participants' social interactions (Peters *et al.*, 2006).

testing differed from normal communication patterns within the family; and (d) deeper understanding of what factors influence the decision to disclose, or not, to certain family members, including health beliefs shared by the family members. It also proved a useful exercise to building up rapport and trust with the participants, as found by Peters *et al.* (2006):

> 'Because the construction is interactive and non-threatening, it was largely engaging and enjoyable for both participant and researcher. This had the positive effect of lowering participants' psychological defensiveness, levelling power differentials inherent in medical settings and increasing empathetic connections.' (Peters *et al.* 2006: 261)

Eco-maps are not dissimilar to geno-maps or genograms used for representing family details in genetic clinics. However, they are not so regimented in their structure and add a dimension of social support systems; they can include any person (not just restricted to blood relatives), friends, family, support groups, communities or institutions (for example, work or church networks) that the participate deems to have a place on the map, depending on what is being explored. In this study, the idea was to build a 'communication map' (Martino, 2006) with the participant identifying those people they consider to be an important part of their life. Whom this constituted was up to the participant.

As Koehly *et al.* (2003) state: 'individuals' perceptions of their family may also include spouses or life partners, adopted children, stepchildren, and, in some cases, very close friends, all of whom may be influential or important in communication and family functioning processes' (p. 305). For the present study, the individual who was being interviewed set the boundaries of who was considered as part of the family, or who was important.

The following section will explain the process of constructing and operating a communication eco-map[15] used within the research interviews. The construction of an eco-map will be used as an example.

Having fully explained the process, the researcher would start by writing the name of the participant in the centre of the map. The participant would then be asked, 'Who do you consider important in your life?' As the participant identified each member, the researcher drew them onto the map – as demonstrated in Figure 8. Geographic distances could be represented by positioning each member near or far away from the centre of the map, while family units that lived together could be enclosed in a circle. The researcher continued to ask follow-up questions such as 'Is there anybody else you might speak to about things going on in your life?[16]' This allowed the researcher to understand who the various family members and friends were, and their relationships, as the participant spoke about his or her experiences of genetic testing and the family communication surrounding it.

[15] Subsequently referred to as an eco-map.
[16] Not specifically about genetic counselling or testing.

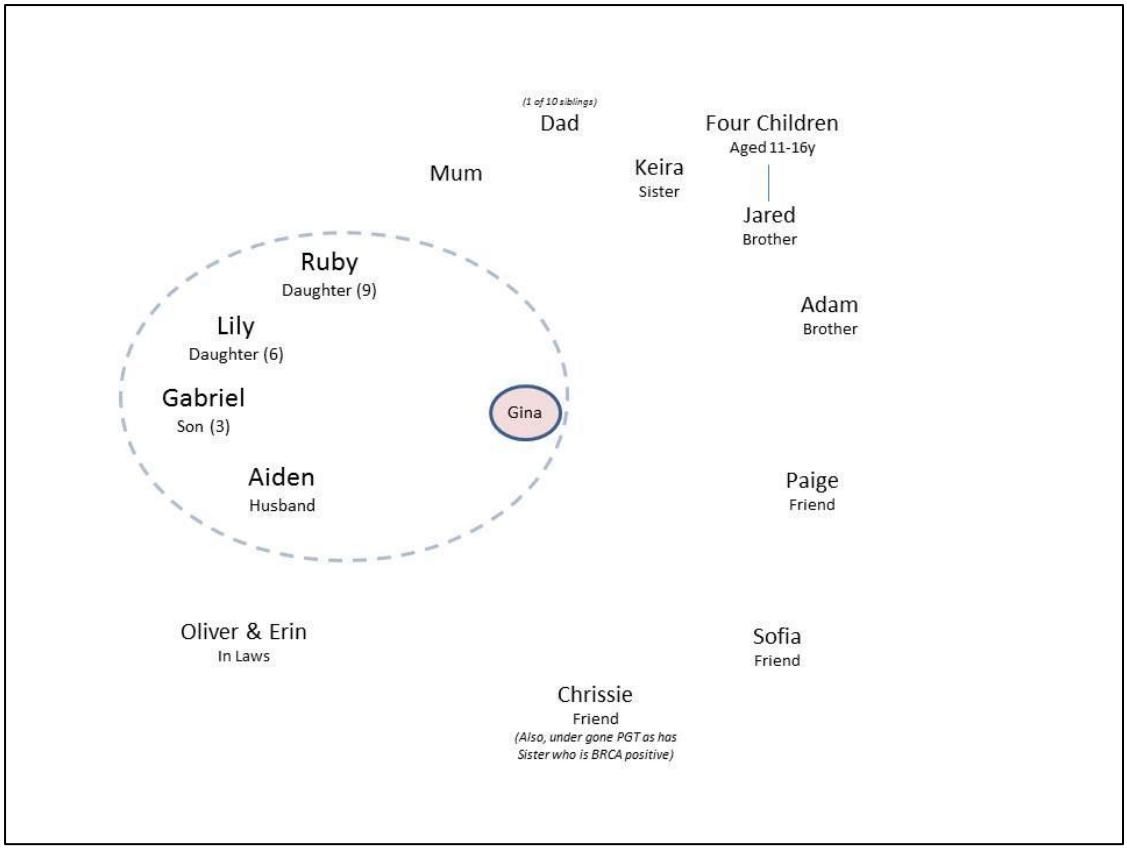

Figure 8: Communication Eco-map one: the participant identifies who is important in their life

In order to get a sense of the organisational and structural characteristics, and to identify the normal patterns of communication between the participant and the people they had identified, the family functioning construct of cohesion, or 'closeness', was measured. Cohesive relationships are supportive relationships that involve those whom the participant feels close to, and are characterised by, behaviours such as support-seeking during a crisis and/or minor everyday upsets, or the sharing of confidences (Koehly *et al.*, 2003). The negative aspect of this construct is defined by a lack of cohesion - those to whom the respondent would not confide in or go to when he or she is upset (Koehly *et al.*, 2003).

Once members were identified, a semi-structured interview technique was used to guide the participant through the process of recoding cohesiveness, as demonstrated in Figure 9.

Participants were asked to score how close they felt they were to each person on the map and how open they felt their communication was with them. This was depicted using a multiple-line technique (Ray and Street, 2005;McGoldrick and Gerson, 1985), where three lines meant really close with open and honest communication and no lines meant did not really communicate. A straight line represented positive relationships, while dashed lines represented tenuous, stressful or conflict-laden relationships (Martino, 2006;McGoldrick and Gerson, 1985). At this stage, the emphasis was on whom they talked to as part of their normal life, rather than about their genetic testing. The exercise concluded by asking the participant if there was anything else he or she wanted to add. The constructed communication map provided an effective visual summary of complex qualitative data.

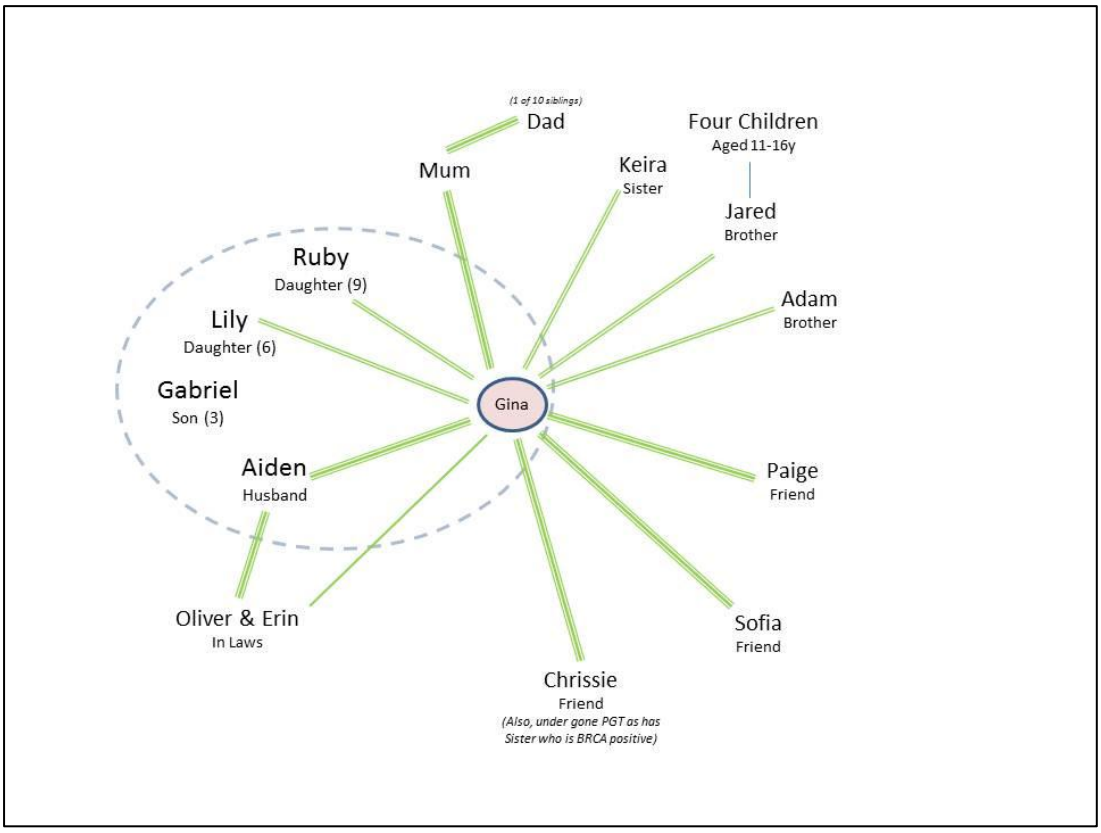

Figure 9: Communication Eco-map two: cohesion and openness of communication

4.5.2 Part Two of Qualitative Interview

Once the map was completed, the researcher moved to the main (second) section of the interview. Participants were asked, '*Could you tell me about your genetic testing and how that came about? I am particularly interested in how and when you spoke to others about it*.' The participants were invited to begin where they liked, with the promise of no interruptions from the researcher. In some cases, people were willing to talk for some time, while others requested or required prompts and guidance from the researcher. This initial question deliberately focused on their genetic testing so as to recognise 'family communication' as one part of the larger experiences of undergoing genetic counselling and testing, and not just about sharing test results.

As the participant told their story, the researcher mapped any important structural and organisational characteristics, as well as the communication patterns about the genetic testing, on to the communication map. For example, in Figure 10, the orange arrows signify that Gina had shared their result information with that person; the arrowhead indicates the direction of information flow, and the numbers the order. The blue arrows are communications not initialised by the participant themselves; for example, Gina's mother introduced *BRCA1*/*2* and genetic testing to Gina and her siblings. The absence of an arrow would indicate no such communication has taken place; for example, Gina did not talk to her children.

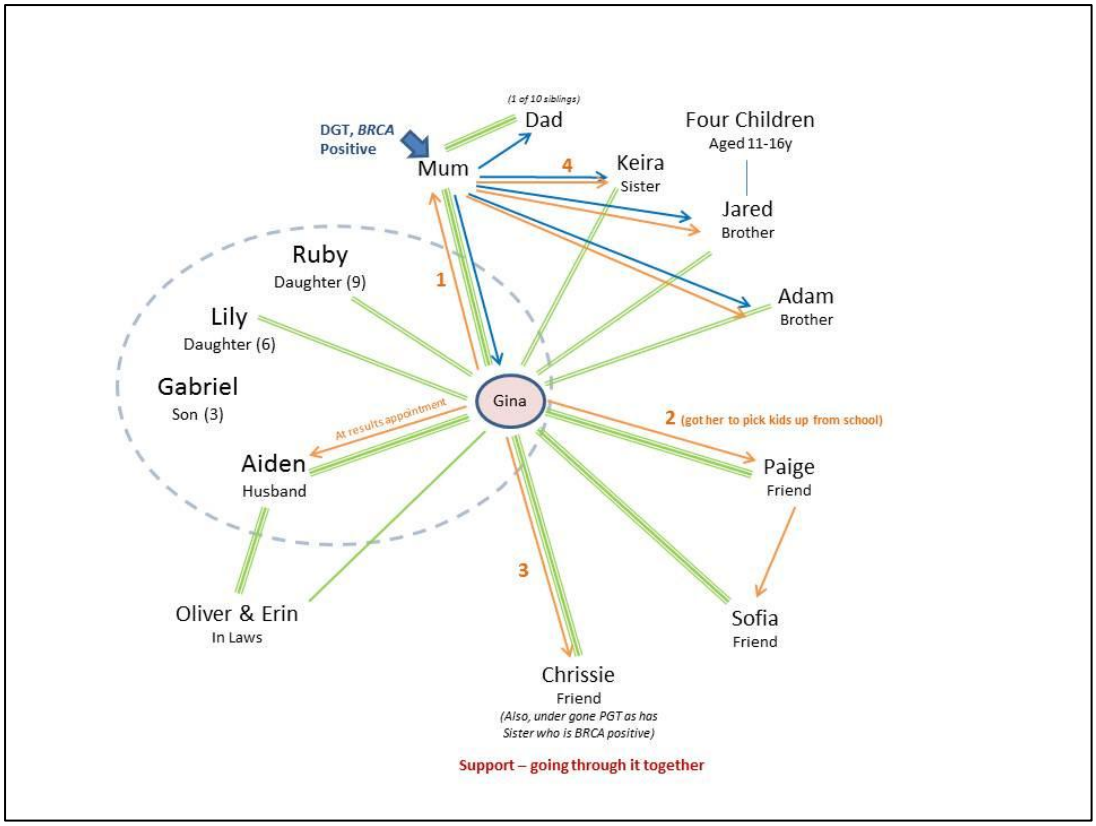

Figure 10: Communication Eco-map three: adding communication patterns about the genetic testing and structural/organisational characteristics

However, in practice, during the first two interviews it became clear that inserting the arrows through the interview was unnecessary and often distracting. It shifted the focus away from allowing the participant to describe their experiences in depth to an almost quantifiable approach of whom on the map told whom and when, with no information about how or why it came about. Therefore, drawing the arrows was abandoned for the remainder of the interviews.

4.5.3 Part Three of Qualitative Interview

The third part of the interview began when the participant had finished telling their story. This involved the researcher seeking more detail, clarifying points and asking for examples about

particular issues that they may already have talked about, such as how they talked to certain family members and any difficulties or challenges they encountered. The researcher also explored family functioning, based on the theoretical perspective of the research, by probing health beliefs, coping strategies, family communication patterns, and affective climate.

Immediately after the interview, the researcher made detailed field notes to capture the richness of data, which the transcript may not have conveyed; for example, tone, pace, non-verbal communication, and subsequent meaning, such as humour, emotion, intensity (Carey and Smith, 1994). The researcher also made notes reflecting on the interview itself - how the researcher felt, what the interview was like, etc. These field and reflective notes were later attached to the interview transcript and reviewed alongside it during the data analysis. Supervision sessions were organised regularly throughout the data collection stages, so that the researcher had the opportunity to further debrief and reflect on the process.

Following the interviews, the audio recordings were transcribed and pseudonyms allocated to protect participant and family members' identity.

4.6 Avoidance of Harm or Distress

There was no anticipated risk of significant harm to participants taking part in this research, although it was recognised by the researcher that some of the participants may have felt that the topics covered were sensitive and/or personal. For example, some may have felt vulnerable exposing their decisions about whether or not to disclose test results and how they went about this, details of their family relationships and/or personal attitudes. During the consent process

and throughout the interview itself, participants were encouraged to only talk about things they were comfortable sharing and assurances were given about the confidentiality of participant's contributions. It was also possible that a participant may have accidently disclosed something they had not considered or planned to disclose before the interview. Therefore, the researcher made a point of checking they were comfortable with everything they had discussed at the end of the interview and took a verbal consent (audio-recorded) from the participant that they were still happy for the things they had said to be included. The researcher also left a thank you letter with the participant with suggested sources of follow-up or support,[17] should the participant require it, as well as contact details for the researcher in case there was anything they wished to discuss at a later date.

During some of the interviews, participants became upset and/or started to cry whilst sharing their experiences. On these occasions, the researcher tried to be empathic and kind. Offers were made to stop the interview, either for a brief time or in its entirety, but all participants wished to continue.

4.7 Data Analysis

Within an interpretive description methodology, the focus for data analysis should be on situating the findings within a framework of the existing body of knowledge and ensuring explanatory factors arising from the analysis are also located within that larger perspective. This can be achieved by strategies such as constant comparative analysis (Glaser, 1965) and iterative

[17] The Genetic Service, Samaritans and Macmillan Cancer Support.

analysis (Thorne *et al.*, 2004b). The following section will describe in depth how the qualitative interview data were analysed and the theories behind them. The process used was based primarily on the work of Miles and Huberman (1994).

Miles and Huberman (1994) define analysis as consisting of three concurrent flows of activity: data reduction, data display, and conclusion drawing and verification.

1. Data Reduction

This is the process of selecting, focusing, simplifying, abstracting and transforming the data that appears in written-up field notes and/or transcriptions. Miles and Huberman reason that data reduction is not something separate from analysis, but rather is part of the analysis. The decisions the researcher makes at this stage (for example, which segments of data to code and extract, which patterns best summarise a number of segments, which evolving stories to tell) are all analytical choices. Therefore, they describe data reduction as a form of analysis that sharpens, sorts, focuses, discards and organises data in such a way that ‘‘final’’ conclusions can be drawn and verified.

2. Data Display

Miles and Huberman argue that, when researchers use only the extended text of written-up field notes or interview transcripts for data analysis, it can be easy to jump to hasty, partial, unfounded conclusions because the data is dispersed, sequential, poorly structured and often extremely bulky. ‘Humans are not very powerful as processors of large amounts of information; our cognitive tendency is to reduce complex information into selective and simplified gestalts or easily understandable configurations’ (Miles and Huberman, 1994). Therefore, it is essential to display the organised information into an immediately accessible, compact form so that the analyst can see what is happening. Typically, qualitative data are displayed in two ways:

networks, with a series of ‘nodes’ with links between them, or ‘matrices’, with defined rows and columns. The advantage of the matrix approach is that it lends itself to within-case and between-case (for example, between those undergoing diagnostic versus predictive genetic resting; or receiving a negative versus positive result analysis (Ritchie and Spencer, 1994). Again, like data reduction, this enterprise is not to be considered separate from the analysis: deciding how the data are best displayed is an analytic activity.

3. Conclusion drawing and verification

It is during this final analytical activity that the researcher begins to decide what things mean. This is done by first noting regularities, patterns (differences/similarities), explanations, possible configurations, causal flows, and propositions; and then testing the emerging meanings for their plausibility, sturdiness and validity. Essentially, the researcher is trying to look at what is going on and how things are proceeding, but also why things occur as they do so they can understand and explain the phenomenon under study. Therefore, there is an analytical progression from describing to explaining the data. ‘Naturally there is no clear or clean boundary between describing and explaining; the researcher typically moves through a series of analysis episodes that condense more and more data into more and more coherent understanding of what, how and why’ (Miles and Huberman, 1994).

4.7.1 Building a Conceptual Framework

In order to reduce and display the data, it was necessary to first build a conceptual framework.

Familiarisation by immersion in the data

This activity began with listening to the recordings of the interviews and the repeated reading and re-reading of the data transcripts, simultaneously creating a list of key ideas, emerging issues, key words and phrases, as well as noting similarities and differences between and within participants' accounts. This is comparable with the method of constant comparison described by Strauss and Corbin (1990) and recommended for interpretive description by Thorne *et al.* (2004b).

The constructed eco-maps were used as a secondary data source in this inquiry. It was important to keep returning to these throughout the process of data analysis. It allowed the analysis to be mindful of the whole of each participant's story and its grounding within their family context, and not lose the coherence of each narrative during the process of comparative analysis. It also helped make visible the way their family make-up, including friends and support networks, might shape their experience of the phenomenon. The objective was to remain attentive to individual cases, while seeking to identify inductively what was common among the experiences of the participants.

Identifying a conceptual framework

This list was then printed out and cut up into individual strips of paper, a strip for each item. These were manually grouped together into separate categories, which allowed reflection as to what were the major components of the participants' experiences and the relationships between them. This activity was informed by the analytical framework (Thorne *et al.*, 1997;Thorne *et al.*, 2004b). However, it was important to remember that, if the original analytical structure was permitted to overwhelm the data collection and analysis processes, the research product would become nothing more than a ''topical survey'' (Thorne *et al.*, 2004b). Thus, this initial analytic stage had to recognise the nature and shape of the preliminary,

theoretical scaffolding that had been used to construct the study, and gradually move away from it as an alternative conceptual emphasis and intrigue arose (Thorne *et al.*, 2004a).

During this activity, it became clear that the chronological flow of events was an important factor in the family communication regarding genetic testing. At each stage of the genetic counselling and testing process, including before and after, there were explicit patterns of communication. A conceptual framework of the data set was thus developed based on four distinct stages of the participants' experiences, both in describing the process of undergoing genetic testing and the communication within the family (Figure 11). The four stages depicted a longitudinal view of family communication regarding genetic testing, which was particularly helpful for understanding the flow, location and connection of events surrounding family communication regarding genetic testing.

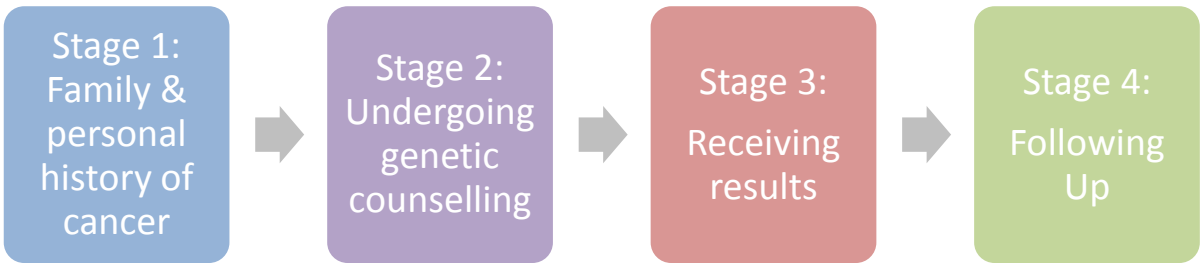

Figure 11: Diagrammatic representation of the conceptual framework developed during the initial stages of the data analysis process. The framework was further developed and refined as the data analysis continued

Developing a coding frame

Once identified, the four stages and the key components or themes within each were numbered and indexed, thereby creating a coding frame of the conceptual framework (Figure 12). This allowed data to be sifted and sorted.

Stage 1	**Family and personal history of cancer**	***Code***
Key components / themes	➢ General family awareness	*1.1*
	➢ Motivations to pursue genetic testing	*1.2*
	➢ Experience of cancer	*1.3*

Stage 2	**Undergoing genetic counselling**	***Code***
Key components / themes	➢ Gathering details of family history	*2.1*
	➢ Accessing DGT through PGT	*2.2*
	➢ Making decisions about GT (getting support & advice)	*2.3*
	➢ Materials to support communication	*2.4*
	➢ Learning about genetic risk	*2.5*

Stage 3	**Receiving results**	***Code***
Key components / themes	➢ Telling those who are waiting to know	*3.1*
	➢ Keeping those in the know in the loop	*3.2*
	➢ Personal reaction to results and telling people	*3.3*

Stage 4	**Following Up**	***Code***
Key components / themes	➢ Distant relatives	*4.1*
	➢ Those who did not want to know	*4.2*
	➢ Children	*4.3*
	➢ Talking to male relatives	*4.4*

Figure 12: The final coding frame. Each key component or theme of the conceptual framework (central column) was assigned a unique code (far right column) ready for data coding

4.7.2 Data Reduction and Display

In order to effectively reduce and display the data, a matrix was created based on the conceptual framework to display the data across all participants, similar to those described by Miles and Huberman (1994) and Ritchie and Spencer (1994).

Coding the data

Coding consists of researchers effectively conducting a detailed, taxonomic process of sorting and tagging data (Green *et al.*, 2007). Basically, each transcript was re-read and each sentence, paragraph or section was assigned a code from the coding frame described above. During this time, the index was constantly refined and modified to incorporate additional emergent themes until every part of each transcript had been assigned a code from the conceptual framework.

Charting

Each matrix displayed one stage, with its columns representing the key-components or themes present in that stage. Each participant was assigned a row within each matrix. The coded sections were charted into the matrices, first as verbatim text from the original transcripts and then each cell was summarised (Figure 13). Whilst time-consuming, this two-step process of charting allowed this analytical activity to remain grounded in the participants' experiences by using their own words. An extended example of the data matrix for four participants can be found in Appendix 8:

Stage One: Family and personal history of cancer			
Participant	**1.1** General Family awareness	**1.2** Motivations to pursue genetic testing	**1.3** Experience of cancer
128	- FH *"a bit grim"* - Thinks mother lived a high-risk life style - So has adjusted own life style to lower risk, e.g. veg, exercise, low stress, not smoking - Always lived convinced she will get br ca one day	- Mother died so young, felt at even greater risk - Small family with very little info on FH so wanted more info	- *"all had breast ca on maternal side"* - Mum died br ca 42y - Felt mum's was very out of blue (never really understood => ↑fear) but later discovered secret letters to grandmother saying had found cysts 10y previously - Couldn't deal w mothers death, could stay in hospital & watch her die (NB Young) - Grandmother had br ca in early 60ys
129	- very aware of - Partaking in regular mammograms - Being a nurse constantly reading up on stuff - Always kept track of family tree and updated it w new ca - Own ca made it more real and felt important to pursue GT - Very close family who openly discuss such things	- Logical isn't it - For interests sake and for research (would be good to get whole family tested for complete picture) - Good to have evidence in place so people don't think you are a time waster - Considering preventative double mastectomy so GT provided more evidence	- Had br ca 2002 - Mum br ca - Uncle died of ca - g/mother died ca - always had regular mammograms
130 (Sister of 127)	- When dad was diagnosed w ca he was given a choice to pursue GT - Sister did a lot of research, weighed up pros and cons so he could make an informed decisions (GP) - Whereas being a lay person she just said she thought her should do it so he did.	Felt it was important to know	Dad br ca

Figure 13: A segment of one of the final matrix displays used to sort and order the data as part of the data analysis process. The columns each represent a key-component or theme and each row a participant

4.7.3 Conclusion Drawing and Verification

The organisation of the data into matrices facilitated in-case and between-case examination, allowing relationships between key components and explanations for patterns within the data to be explored as well as outlining potential patterns, connections and relationships within the data. In line with the interpretive descriptive methodology, the aim was to take the analysis beyond qualitative description to an interpretive piece of work capable of informing clinical

practices. This was executed by detailed within-case analysis, comparisons between cases, repeated interrogation of the data and by generating hypotheses and testing them within the data, such as: 'For one participant, discussion about there being something in the family causing these cancers occurred more often with siblings than parents. Is this true of all cases?'

The emphasis at this stage was focussing on moving the analysis beyond just description to interpretation. To achieve this, the aims were to:

1. Explain specific phenomena within the data. This was done by asking questions such as:
    - What underpins attitudes and health beliefs about cancer, familial risk and genetic testing?
    - What are the implications for a particular behavior, for example, not including young children in discussions?
    - What contributes to different outcomes or impacts? For example, are there certain groups of relatives consistently not being told about the genetic testing and, if so, why?

2. Explain associations between two or more key components. This was done by asking questions such as:
    - Are there linkages in cases, such as two attitudes; an attitude and behaviour; an event and the factor underpinning it? For example, is there a link between feeling it is important to talk openly and honestly with family members about cancer in the family, and feeling it is important to make potentially at-risk relatives aware of a genetic test result?
    - Are there linkages between cases? For example, do two participants, or two groups, show some of the same characteristics and, if so, why? For example, are there

similarities between how those receiving a negative test result share results with their first-degree relatives?

4.8 Data Management

All details of participants, including copies of reply slips and the consent forms, were kept in locked filing cabinets in a secure office space at the Faculty of Health Sciences (previously the School of Nursing and Midwifery). With the permission of the participants, all interviews were audio-recorded with a digital recorder. Written field notes and memos were also kept to record non-verbal communication and reflections on the interviews. Interview recordings were transcribed into text for data analysis. All recordings and transcriptions were kept on a password protected computer, which was backed-up daily. Written field notes, memos and printed transcriptions were kept in a locked filing cabinet separate from any identifying data. All primary data (audio-recordings, written field notes, memos and transcriptions) will be kept for 10 years from the end of the study in accordance with University policy.

4.9 Qualitative Rigour

The importance of rigour in qualitative research was discussed in section 3.3.1. The strategies taken to ensure methodological rigour in the study will be discussed here, as it is seen through 'key qualitative research concepts' identified by Lincoln and Guba (1985; 1994), namely credibility, transferability, dependability, confirmability and authenticity, which have been

fundamental to the development of standards used to evaluate the quality of qualitative inquiry (Morse *et al.*, 2002). These criteria are presented to be parallel with positivist criteria of rigour.

Credibility (comparable with internal validity) relates to how well the researcher's explanation fits the views provided by the study participants (Tobin and Begley, 2004;Schwandt, 2001). According to Lincoln (1995), credibility can be demonstrated through member checks, peer debriefing, prolonged engagement, persistent observation, and comprehensive audit trails. However, the appropriateness of procedures, such as member or dependability checks, have been challenged as being philosophically contradictory to the idea of multiple realities (Gallagher, 1995;Silverman, 2000b).

As was discussed in section 3.5, one of the recommended means of considering the validity of findings in an interpretive description study is to compare them with the clinical hunches of expert clinicians familiar with the study phenomenon (Maheu and Thorne, 2008). Therefore, the conceptual framework and research findings for this study were presented to the clinical team at the Genetic Service and at national conferences at various points during the analytical process. This gave the researcher confidence in the credibility of the findings.

Transferability (comparable with external validity) refers to the generalisability of inquiry (Tobin and Begley, 2004). It is important to recognise that 'external validity' is substantially different in qualitative inquiry than quantitative inquiry, as the naturalistic paradigm holds that there is no single correct or 'true' interpretation. As discussed in 3.4.1, to ensure transferability, the objective will be to supply a description of the research process in sufficient detail so the reader

can make an informed judgement of whether the original study setting and their own are sufficiently similar to ‘‘apply’’ the findings (Murphy *et al.*, 1998).

Dependability (comparable with reliability) describes the researcher's responsibility to substantiate that every part of the research is transparent and methodical (Tobin and Begley, 2004), as well as logical, traceable and clearly documented (Schwandt, 2001). Dependability was demonstrated through a rigorous audit trail, including a series of research note books, supervision records and comprehensive descriptions of all methods and procedures, where others can examine the researcher's documentation of data, methods, decisions and end product.

Confirmability (comparable with objectivity or neutrality) is concerned with establishing that data and interpretations of the findings produced are not exaggerated or fabricated by the researcher, but are clearly derived from the data (Tobin and Begley, 2004). Auditing, as described for dependability, can be, and was also, used to authenticate confirmability (Lincoln, 1995).

Authenticity is regarded as a feature unique to naturalistic inquiry (Schwandt, 2001). In 1994, Guba and Lincoln reworked their framework to incorporate a fifth criterion of authenticity, which relates to fairness (presenting all value differences, views, and conflicts); knowledge sharing (ontological and educative authenticity); and social action (catalytic and tactical authenticity). During the analysis and write-up, every effort was made to represent the views and experiences of each participant. Direct participant quotes from interview transcripts are

used to demonstrate points and to ensure findings are grounded within what the participants actually said.

4.10 Conclusion

The last two chapters have introduced the research question and then described the study in theoretical (Research Methodology) and practical terms (Research Methods). The demographics of the research participants and the findings are presented in the next chapter.

Chapter 5 – Characteristics of Participants; Analysis of Eco-maps; and Stage One Findings: Cancer in the Family

5.1 Introduction

The next four chapters present the results of data analysis. This was a qualitative study using an interpretive description methodology as described in Chapter Three. This inductive approach aims to capture themes and patterns within subjective perceptions in order to generate an interpretive description capable of informing clinical understanding (Thorne *et al.*, 2004b). The objectives are to gain an insight into the participants' experiences of discussing their participation in genetic testing, their test results, and potential risk information regarding genetic testing for *BRCA1/2* with their family (not just with first-degree relatives or specific family members), with particular focus on *how* these families discuss genetic testing for cancer risk and *when*. There will be an appreciation of family communication as a process rather than as a discrete event. Therefore, the research will examine when family communication regarding genetic testing occurs, throughout the whole process of genetic counselling and genetic testing, not just disclosure of test results.

A predominant feature of the data set that emerged during analysis was that participants' experiences could be sub-divided into four distinct stages, to both describe the process of undergoing genetic testing and communication within the family. The presentation of the findings in these chapters will, therefore, follow these stages: Stage One: Cancer in the Family (Chapter Five); Stage Two: Undergoing Genetic Counselling (Chapter Six); Stage Three: Receiving

the Test Result (Chapter Seven); and Stage Four: Following up Longer Term (Chapter Eight). The four stages depict a longitudinal view of family communication regarding genetic testing. The aim of presenting the findings in this way is to reflect the flow of events and experiences of participants as they occurred. This will facilitate exploration of how and when these participants talked about their family history of cancer, their genetic testing and its implications to others around them.

5.2 Characteristics of Participants

Figure 14 outlines the number of potential participants at each stage of the sampling and recruitment process. One hundred and seventy-one people were identified through the Genetic Service as having had a genetic test for *BRCA1/2* within the designated time frame.[18] Fourteen were excluded, as they did not meet the inclusion criteria (refer to section 4.2): four had passed away, two lived abroad, and eight were identified by the Genetic Counsellors as having recently received a cancer diagnosis or having some other condition that made them potentially too vulnerable, for example, suffering from Alzheimer's. From the remaining 158, invitations to participate were sent out in batches of seven, following the sampling strategy described in 4.2, until the required sample size of approximately 30 participants was reached.

Seventy-seven patients from the Genetic Service who met the inclusion criteria were sent letters inviting them to join the study. Twenty-nine individuals completed interviews, giving a response rate of 37.7% (29/77). Unfortunately, the audio recorder failed in one instance, so only the

[18]The study included individuals who had received their genetic test results eight to 18 months prior to being invited to participate (refer to section 4.2).

researcher's hand-written notes are available from this interview. This participant's experiences were compared to the others during the data analysis, but no direct quotes are used as they are not available.

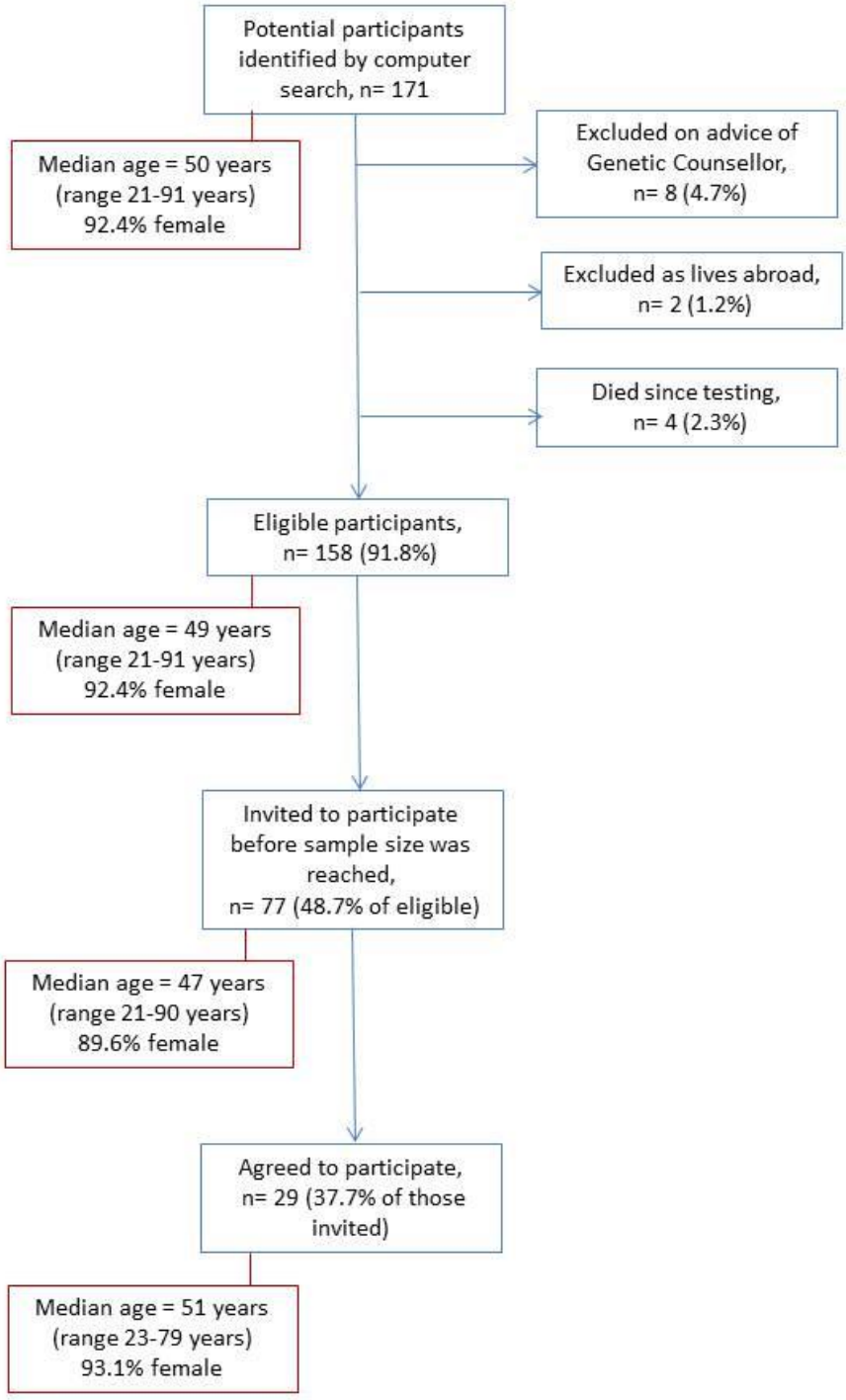

Figure 14: Summary sampling and recruitment numbers

Figure 15 provides a summary of participants' characteristics and Table 4 provides a summary of participants' demographics. Of the 29 final participants, 27 were female and two male. The age range was 23-79 years with a mean age of 51.2 years. Thirteen had undergone diagnostic genetic testing: of which one had received a *mutation-positive* test result and 12 had received an inconclusive result (meaning a negative result in the absence of a known *BRCA* mutation, but a yet unknown genetic mutation is possible). Sixteen participants had undergone predictive genetic testing: of which six were found to be proven carriers *(mutation-positive)* and ten to be proven non-carriers *(mutation-negative)*.

Twenty-six out of the 29 had children. Between them, they had 18 adult daughters (over the age of 18 years old) whose mean age was 28 years (range 18-55 years old). They had 14 daughters under the age of 18 years, whose mean age was 12.6 years old (range 6-17 years old). There were 17 adult sons, whose mean age was 31.5 years (range 18-58 years old) and ten sons under the age of 18 years whose mean age was 9.7 years (range 2-15 years old).

Twenty-four of the participants were married (including one civil partnership), two were divorced and two were single. There were three sets of relations: two sets of sisters (Maya and Nicole; Gillian and Kerry), and one aunt and niece (Kim and Faye).

The average time from receiving their test result to being invited to participate was 402.1 days (13.4 months). Non-responders were of a similar age (median and range) and the ratio of females to males compared to responders (see Figure 14). Unfortunately, no further information is available on non-respondents due to data protection legislation.

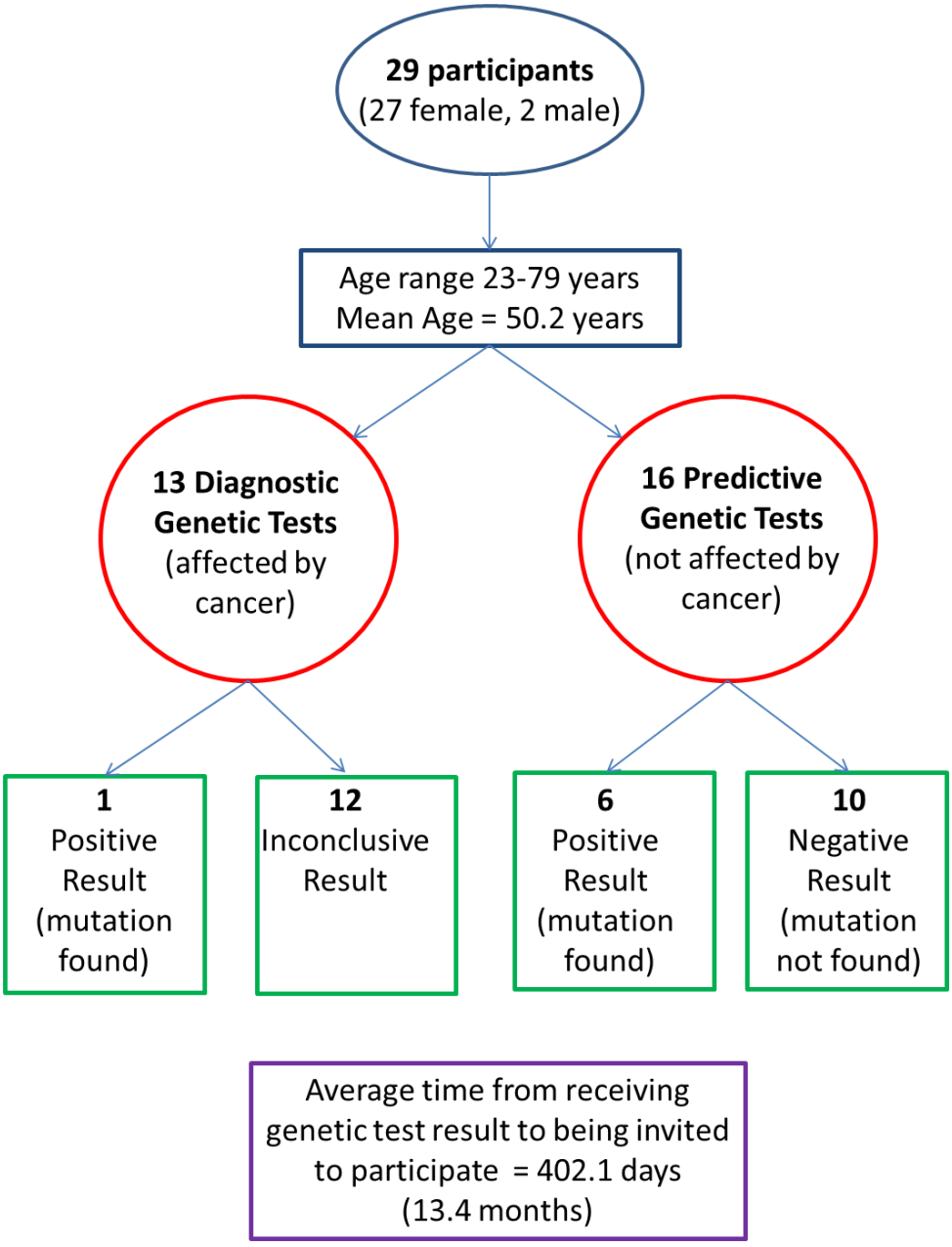

Figure 15: Summary of participants' characteristics

Gender	
Male	2
Female	27
Age Group	
20-29 years	2
30-39 years	2
40-49years	11
50-59 years	8
60-69 years	4
70-79years	2
Mean (years)	*51*
Range (years)	*23-79*
Relationship Status	
Married	23
Civil Partnership	1
Divorced	3
Single	2
Time between test and being invited to participate	
<10 months	6
10-12 months	6
13-15 months	11
>17 months	6
Average	*13*
Occupation Group	
Management	4
Education Professional	3
Sales Assistant	4
Support Worker (Health, Social or Education)	3
Health or Social Care Professional	3
Office/Administrative Support	5
Media and Communications	1
Retired	4
Other	2

Table 4: Summary of participants' socio-demographics

5.3 Analysis of Eco-maps

In line with the theoretical basis of the research (refer to section 3.4.4), it was necessary to gain some understanding of the participant's social context and family relationships. This included their usual communication patterns and how close and open they perceived those relationships to be (cohesion); family organisational and structural characteristics; and any health beliefs shared by family members. Therefore, eco-maps were included in the research interview to identify people that the participant communicated with during their day-to-day life, to assess how close their relationship was, and how open they perceived the communication to be. This allowed the researcher to understand who the various family members and friends were, and their relationships, as the participant spoke about his or her experiences of genetic testing and the family communication surrounding it.

After discussion with the PhD supervisors and member of the Faculty of Health Science's Ethics Committee, the decision was taken not to include copies of the eco-maps in this thesis. This is done to ensure confidentially for the research participants. Even though all the eco-maps have been anonymised and pseudonyms have been given to each person identified, the amount of information present on the family make up, for example number of children and their ages, number of siblings, etc., does significantly increase the chance of someone working out who the eco-map belongs to. This is coupled with the fact that participants were asked to score how open they perceived their communication with each person to be, which is potentially sensitive information that participants would probably prefer not to be shared with family members. Instead, where necessary, example eco-maps have been created based on a simulated participant.

Eco-maps were completed with 26 out of the 29 participants during the research interview. In the three cases where this activity was not done, this was because the participant started talking about their experiences without the interviewer having the opportunity to introduce the eco-map. The interviewer made the decision not to stop the participant because she felt it may make the participants feel uncomfortable, or as if they had done something wrong. This may have altered the atmosphere of trust and openness the interviewer was trying to create (refer to section 3.4.5). The researcher felt confident that the same issues, as laid out in the interview guide (refer to appendix three), had been covered as in the other interviews.

These three interviews were fairly well spread out throughout the data collection period, as seen by their research numbers, which were issued consecutively – participant 104, participant 109 and participant 128. This would suggest that this deviation from the data collection protocol was not due to the interviewer having a lack of confidence in the interview guide (as might have been suggested if all three had been early interviews). Alternatively, it may be due to a heightened confidence that made the interviewer feel she did not need the security of introducing the eco-map as a way to get the interview started (as may have been suggested if all three had been later interviews).

5.3.1 Who Do Participants Communicate With In Their Day-to-Day Lives?

When asked 'Who are the important people in your life?' and 'Is there anybody else you might speak to about things going on in your life?', the 26 participants identified 468 contacts[19] (average 18 per participant, range 6-44). Table 5 presents an overview of the composition of

[19] Individuals or groups; when a participant mentioned a group of people as one, for example, a bible group, these were counted as one contact.

each study participant's personal network as they identified them during the construction of the eco-map. As summarised in Figure16, 138 (30%) of these were first-degree relatives,[20] 105 (22%) second-degree,[21] 46 (10%) third-degree relatives,[22] 75 non-blood relatives (16%) and 104 (22%) friends/colleague. This comprised an average of 5.3, 4.0, 1.8, 2.9 and 4.0 per participant respectively.

As previously described in section 3.4.5, the emphasis at this stage of the research interview was on identifying who participants talked to as part of their normal life rather than about their genetic testing. To facilitate this, the questions posed by the interviewer were purposely phrased as 'Who do you consider important in your life?' and 'Is there anybody else you might speak to about things going on in your life?' However, as was revealed during the second part of the interview, many participants automatically included individuals in the construction of the eco-map who they were not necessarily emotionally close to, but who had played a role in their genetic testing. For example, this was particularly true of first cousins (a third-degree relative). This will be further explored in section 6.2.

Despite the careful wording of the questions, the participants' choice of who was included in their eco-maps was likely to have been influenced by the way the interviewer introduced the research topic to the participants during the recruitment and consent processes. The

[20] A first-degree relative is defined as a close blood relative, which includes the individual's parents, full siblings, or children.

[21] A second-degree relative is defined as a blood relative, which includes the individual's grandparents, grandchildren, aunts, uncles, nephews, nieces or half-siblings.

[22] A third-degree relative is defined as a blood relative, which includes the individual's first-cousins, great-grandparents or great grandchildren.

participants knew the researcher was there to interview them about their genetic testing and, more specifically, how they talked to their relatives about it and so were presumably motivated to cover the topic from the outset.

It is also possible that, because the topic under discussion related to family genetics, some participants were influenced by their experiences of having attended multiple genetic counselling appointments, where it is common to give a full family history and create a genogram. For example, Elizabeth (participant 103) and Annie (participant 105) both described their communication patterns in terms of their family tree and, perhaps due to her background in human genetics, the interviewer automatically sketched it out in the layout of a genogram rather than a true eco-map. These were also two of the first interviews to be completed when, perhaps, the interviewer was not quite so confident in the data collection protocol and was more easily led by the participant.

Participant	First degree relatives	Second degree relatives	Third degree relatives	Non blood relatives (including partners & in-laws)	Friends & work colleagues	Total number of individuals identified by participant
Molly	5	5	2	1	4	17
Eloise	5	4	4	2	2	17
Elizabeth	9	10	17	7	1	44
Arthur	4	5	3	2	2	16
Faye	6	2	0	4	1	13
Annabelle	4	5	0	1	3	13
Brenda	4	1	2	3	3	13
Laura	5	2	0	1	2	10
Martine	6	4	0	4	4	18
Joanna	3	2	0	1	0	6
Julia	5	13	0	3	6	27
Zena	6	7	1	1	9	24
Karen	3	0	1	1	3	8
Gina	8	4	0	3	3	18
Sara	5	2	1	0	3	11
Rachel	5	0	0	1	10	16
Katherine	5	4	0	5	2	16
Viv	5	3	3	13	8	32
Robert	1	2	0	5	3	11
Christina	0	1	7	0	3	11
Carolyn	5	5	3	1	4	18
Maya	8	6	0	3	1	18
Gillian	9	9	1	2	8	29
Tina	8	4	1	7	11	31
Nicole	7	5	0	3	4	19
Kerry	7	0	0	1	4	12
TOTAL	**138**	**105**	**46**	**75**	**104**	**468**
Average	**5.3**	**4.0**	**1.8**	**2.9**	**4.0**	**18.0**

Table 5: Summary of family and social networks as reported by participants, showing the number of individuals identified broken down by relationship

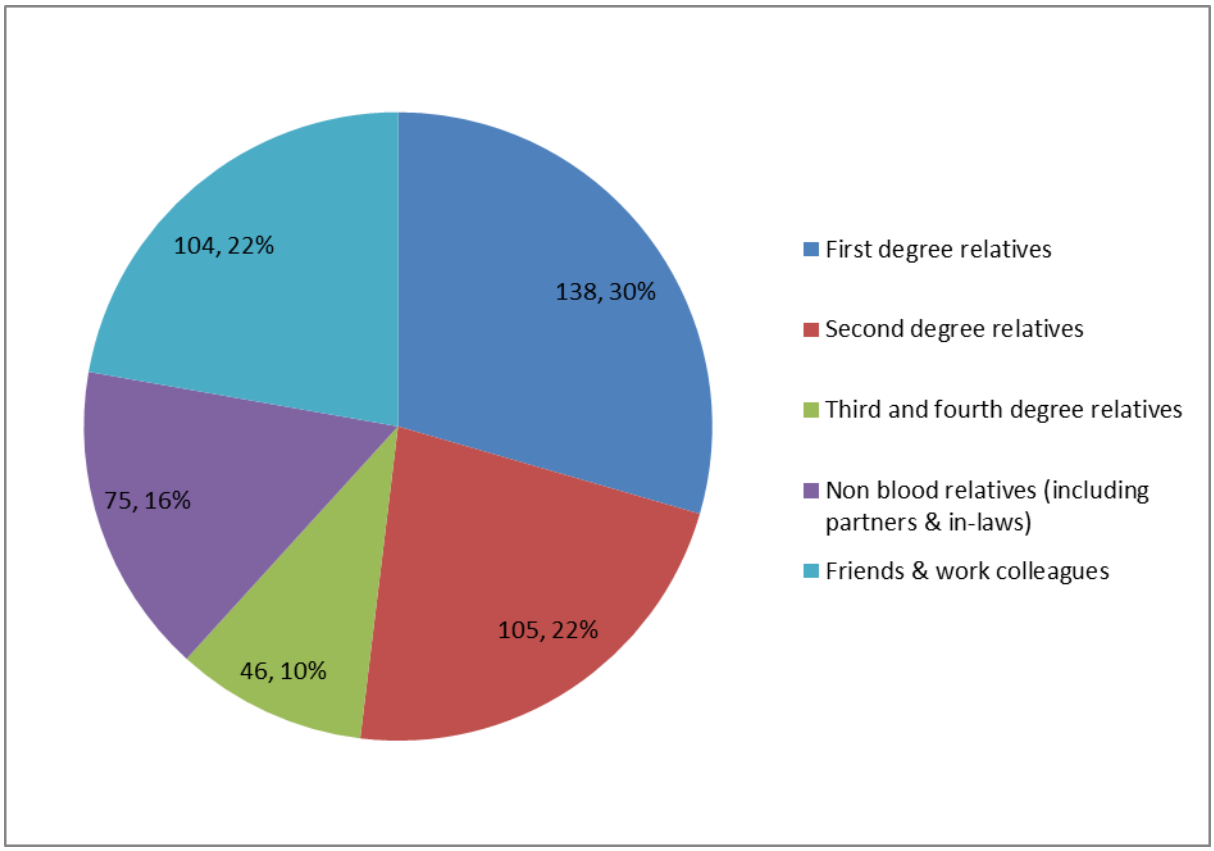

Figure 16: A breakdown, by relationship, of the 468 contacts participants identified as important people in their lives in their eco-maps

5.3.2 How Open Is That Communication?

Having identified those people participants felt were important in their day-to-day lives, they were then asked to score how close they felt to each person on the map and how open they felt their communication was with them (as a measure of cohesion). This was depicted using a multiple line technique (Ray and Street, 2005), where three lines meant really close with open and honest communication, and no lines meant did not really communicate. A dashed line represented a tenuous, stressful or conflict-laden relationship (Martino, 2006). Table 6 shows

the breakdown of how the cohesion with family and friends was scored by participants. The breakdown of scores for an individual participant's eco-maps can be found in Appendix 9

	Total[β] (average per participant[¥])	Scores of 'closeness' and 'openness' of communication[μ].					
		3	2	1	0	Disrupted[π]	Not alive
All participants							
First degree relatives	**138 (5.3)**	46 (1.8)	42 (1.6)	23 (0.9)	3 (0.1)	11 (0.4)	11 (0.4)
Second degree relatives	**105 (4.0)**	0 (0.0)	4 (0.2)	9 (0.3)	84 (3.2)	3 (0.1)	5 (0.2)
Third degree relatives	**46 (1.8)**	3 (0.1)	9 (0.3)	4 (0.2)	29 (1.1)	0 (0.0)	1 (0.0)
Non blood relatives (including partners & in-laws)	**75 (2.9)**	23 (0.9)	16 (0.6)	14 (0.5)	22 (0.8)	0 (0.0)	0 (0.0)
Friends & work colleagues	**104 (4.0)**	33 (1.3)	41 (1.6)	28 (1.1)	2 (0.1)	0 (0.0)	0 (0.0)
Total	**468 (18.0)**	**105 (4.0)**	**112 (4.3)**	**78 (3.0)**	**140 (5.4)**	**14 (0.5)**	**17 (0.7)**

[β] Total number of individuals or contacts included on eco-maps by all participants.
[¥] Total number reported divided by 26 participants who completed an eco-map as part of research interview.
[μ] A score of three (lines on the eco-map) meaning really close with open and honest communication and zero meaning do not really communicate.
[π] Tenuous, stressful or conflict laden relationship

Table 6: Summary of family and social networks with scoring of closeness of relationship and openness of communication as a measure of cohesion, reported by participants

Figure 17 shows the breakdown of scores for all persons on the maps. One hundred and five (22%) were scored with three lines; 112 (24%) with two lines; 78 (17%) with one line; 140 (30%) no line; 14 (3%) with a dashed line; and 17 (4%) of those identified were dead and were not scored.

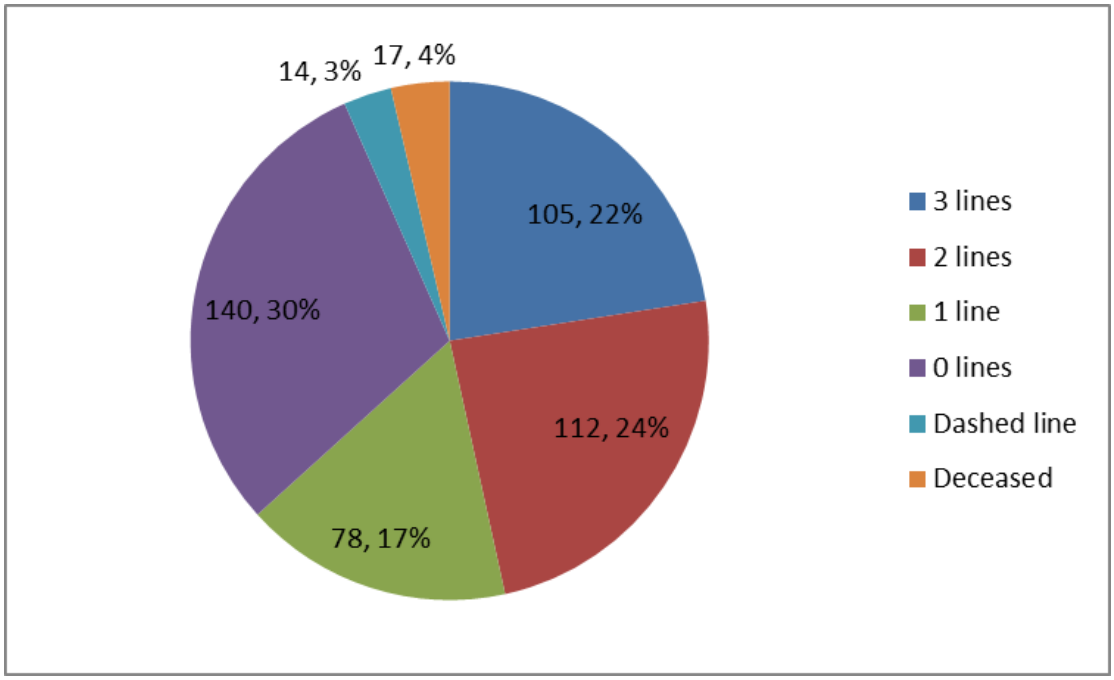

Figure 17: A breakdown of scores for how open participants felt their communication is using a multiple line technique

As before, because the questions to identify these contacts were phrased as 'Who do you consider important in your life?' and 'Is there anybody else you might speak to about things going on in your life?', 30% being scored as little or no communication (no line) seems surprising. However, when the scores are broken down according to degrees of relationship, as in Figure 18, the majority of these are second-degree relatives. These are largely accounted for by the

presence of either nieces or nephews, who were mentioned in association with their parents, rather than as a contact in their own right, and were therefore not really part of the participant's ''normal'' communication pattern. Likewise, grandchildren were sometimes mentioned as being important, but were given a score of zero because they were too young for the communication to be considered open or not.

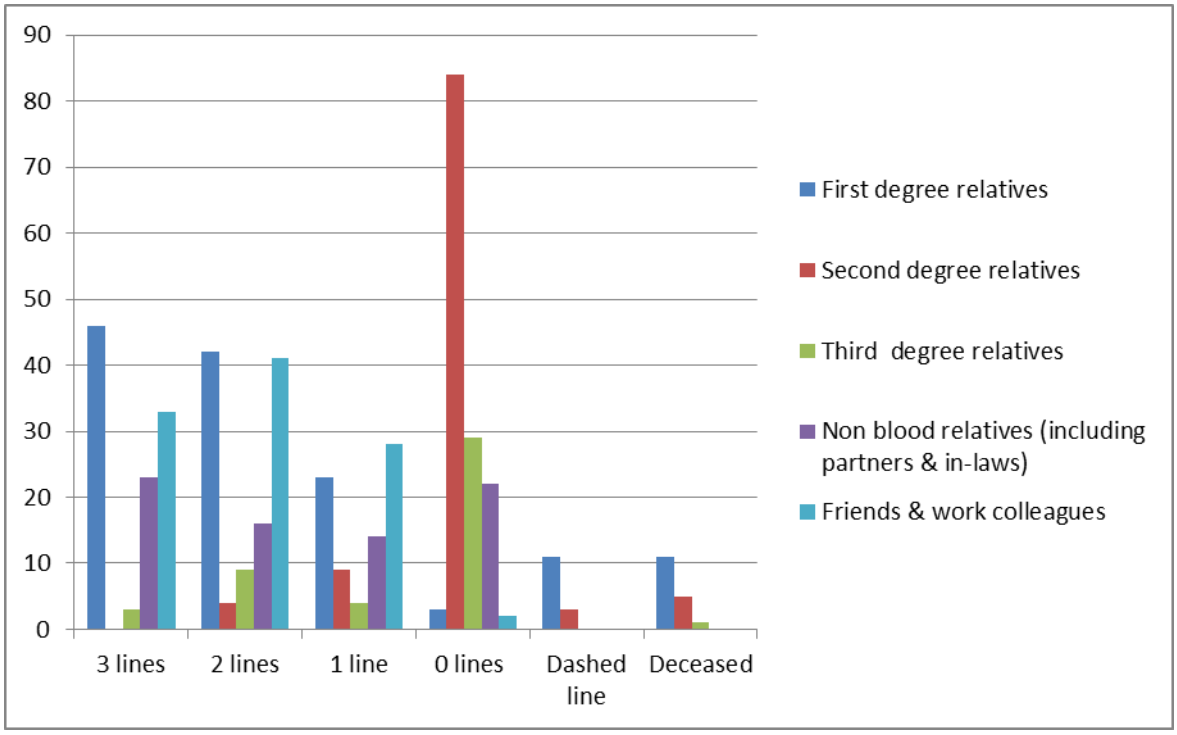

Figure 18: Scores for 'openness of communication' broken down by degrees of relationship

5.4 Stage One: Cancer in the Family

The first stage of family communication begins well before genetic testing and is based on people's experiences of cancer (Figure 19). Discussion in the family about the family history of cancer was prompted by personal experiences, for example, death and disease within the family, or undergoing regular screening. For participants, cancer was an ongoing theme throughout their lives, even if it is not a constant focus, and so was discussed regularly. This continued discussion about cancer and the family history is the first step to open discussion about genetic testing for *BRCA1/2.* It tended to include emotionally close relatives, such as first-degree relatives, partners and close friends.

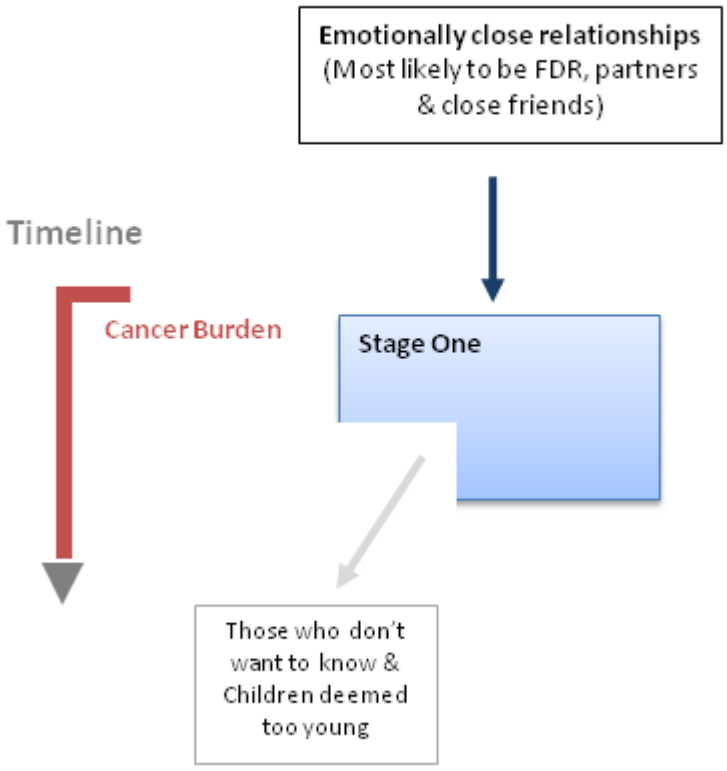

Figure 19: Stage one: Family and personal history and experiences of cancer

5.4.1 Family and Personal History and Experiences of Cancer

Experiences of cancer and, in particular, their family history of cancer, shaped how the participants and their families talked about the disease and genetic testing.

A striking feature of the data was just how prominent a role cancer plays in many of the participants' lives. At a basic level, there was a sense that ''everyone'' in the family, especially the females, had had cancer…

> *'Erm…well first off…erm, it was my grandma, my aunt, my mum and…my aunt would be like her sister. Erm, they all died of cancer.'*
>
> (Molly, 49y, PGT, Negative)

Sara, the youngest participant, spoke about how she remembers cancer always being part of her family life as a child. Her Nan (maternal side) had died from ovarian cancer when she was 10 years old and she was aware that several of her aunts (her mother's sisters) had died of breast and ovarian cancer by that time:

> *We are all just like – we remember Nan being ill and she came to live with us as soon as she found out she was ill. So she lived for about two years while she was dying, so we were all like really aware of it and I think we are quite an open family anyway, so we all discussed things'.*
>
> (Sara, 23y, PGT, Negative)

She remembers various phone calls her mother made to the Genetic Service about the genetic testing, but admitted *'I never really understood it when I was younger*'. Sara felt her family were very open when it came to discussing cancer, particularly between herself, her two sisters and her mum, all of whom she gave three lines to on her eco-map. From her perspective, there was no evidence of her being excluded from these conversations because she was a child: *''…me and my Mum and my sisters all just spoke about it all the time''.* It must be recognised, however, that, if asked, her mother may say these conversations were censored in some way that Sara was not aware of; however, there is no way to verify this within this research method. On the other hand, Sara was experiencing first-hand the death of a close relative due to cancer because her Nan came to live with them when she got sick, so some conversation on the topic was to be expected.

Other participants also spoke about being aware of the family history from a young age and that they would potentially get cancer in the future:

> *'It was something that I'd kind of, from a teenager, always been told there's a chance that you might get breast cancer later on as well. So it was something that I'd kind of grown up with.'*
>
> (Annabelle, 51y, PGT, Negative)

For many participants, there had always been an assumption that they themselves would get cancer one day. For some, this assumption was linked to genetics, often those with some kind of

medical background such as being (or had been) a doctor (n=1), nurse (n=3), midwife (n=1) and physiotherapist (n=1). But, for others, it was expressed more as ‘‘always knowing’’ there was ‘‘something’’ in the family. This family trait was most often discussed horizontally across the family; in other words, between siblings, rather than vertically.[23]

So often in the interviews, this perceived knowledge that they were destined to get cancer was stated in a ‘matter of fact’ manner, as if it was just part of being a member of that family. And yet, when questioned further, the participants revealed just what a huge emotional burden this could be. For example, Zena’s mother had died of breast cancer when she was 16 years old (and her grandmother when her mother was 9 years old) and she was left with a very visual image of the risk from the way her father talked about it:

> *‘So we knew already (my sister and I) that there was a very strong genetic link of some sort within our family. And, whilst my Dad didn’t talk a lot about it, the odd times that he did say anything, he just used the symbol of the Sword of Damocles hanging over you, basically, and that’s that vision I’ve always had growing up really and moving away.’*
>
> (Zena, 49y, DGT, Inconclusive)

Like Sara before, many had experiences of caring for relatives with cancer and witnessing relatives dying from the disease. Many of the participants had experienced their mothers being diagnosed with cancer and, in many cases, die from it. For about half of these, maternal death

[23] Vertical communication: up or down, the family as laid out in a genogram, for example down to offspring or up to parents.

had occurred when the participants were very young. These experiences could have an important psycho-social impact on the individual and the family as a whole and, as such, shaped the way participants' felt about the disease and how they talked about it. For example, Eloise felt she had never really understood why her mother had died and so quickly when, in her young eyes, so little treatment was given:

> *'I suppose mum was the, the first, that I was really, that really affected me, coz she died, very soon after her diagnosis. Which I never really understood because, she was diagnosed, she had a lumpectomy, radiotherapy and tamoxifen. And I couldn't understand why just over a year later she'd gone. And she was full of cancer… I was eleven when she died… So, so when mum got it, you know, there was nothing to suggest anything sinister, but I could never understand why, if it was such a trivial matter, that she only ever needed to have the lump removed, why she died'.*
>
> (Eloise, 42y, PGT, Positive)

During the interview, Eloise came across as a very warm and bubbly person who spoke very articulately about her family history and her genetic testing for *BCRA1/2.* She described having very close relationships with very good communication with those she identified on her eco-map (all being scored as either three or two-line, with the exception of one male cousin). At times, she obviously found the topics under discussion, such as her mother's death, upsetting, but these experiences seemed to have given her a drive to do everything she can to protect herself and her family from this disease, including talking very openly about her experiences to her friends and family: *'In fact, I would stand on the roof tops',* she said.

For some, the emotional burden of the family history can make talking about it really challenging. This was highlighted by the fact that, during the interviews, several of the women became tearful when talking about relatives who had been diagnosed with cancer or had died, and the emotional burden of a family history of cancer was evident. It does not seem unreasonable to assume that this may be even harder when talking to close relatives, where both parties are emotionally involved, compared to talking to an independent researcher. However, all the participants had spoken to some, if not the majority, of the people identified on their eco-map about their family history of cancer and genetic testing. For many, tensions arose between wanting to care for and protect loved ones whilst, at the same time, having to discuss a potentially threatening and emotionally laden topic.

One participant, Elizabeth, had been aware that her family history was worth investigating since her breast cancer diagnosis in 1997. For her, pursuing genetic testing was about calming her teenage daughter Grace's anxieties and fears. However, with this came the responsibility of making sure she timed it right so as not to cause her daughter more stress:

> *'It's very much been in the back of my mind for quite a while that it may be something we needed to look at. I tended to sit on it when – there have been times when Grace is saying "I must know, I must know" and then "Mum, "cause I need to do something" you know. She's quite a reactive sort of person, and so as she was going through the sort of teenage phases I was just trying to keep the lid on it really and say "well let's just – we'll leave it a bit and we'll talk about it when you are a bit clearer". Because one day she would be "Yes, let's go for it" and the next day would be "No I'll bury my head*

in the sand and I don't want anything". So I think when she was coming up last year to – you know, she was almost through University and she felt – what she was saying felt a lot more stable and she said "Yes I think I would like to know". So we felt that if I approached the GP who had talked with her as well as with me over the years (the same GP for both of us) perhaps we would see if I could be tested.'

(Elizabeth, 54y, DGT, Inconclusive)

Elizabeth felt she often had to mediate when and how the topic was discussed. To avoid unnecessary upset and to keep the matter from blowing out of proportion, she took her cues on when to discuss it from Grace. However, she was very aware that, during her teenage years, Grace regularly changed her mind about whether or not she wanted to know if they carried the gene; therefore, Elizabeth tended to placate her fears rather than pursue it until she felt Grace was emotionally ready. That is not to say she avoided talking about the topic as she felt it was important to keep it out in the open, but it gave her a way of dealing with the tension she felt between wanting to be open about it and also wanting to protect her child from harm.

5.4.2 Reactions to Family History

Generally, the significant family history of cancers was dealt with in three ways within the family: normalising it; increased engagement with it; and/or disengagement. These could be from individuals or by the family unit as a whole, and were based on experiences of cancer in the

family and their shared health beliefs. Some communicated differently with different relatives and/or at different time points.

1. Normalising it

Despite it being an emotional topic close to their hearts, with cancer playing such a prominent role in their lives it was a regular topic of conversation within these families. Really, for these participants and their families, this is an on-going experience, the effects of which they live with throughout their lives. This resulted in them talking openly about it, as just a normal part of being a member of that family:

> *'So – but me and my Mum and my sisters all just spoke about it [family history of cancer[24]] all the time; we were just all quite savvy to cancer I think, like in our family because everybody's got it. It's like a family heirloom!'*
>
> (Sara, 23y, PGT, Negative)

Everything from a new diagnosis to engaging in regular screening could provide the stimulus for family communication about the topic, as described by Tina:

> *'I've always kept track of the family tree, so you know, I make a note every time someone has had it done, you know, and we talk about it. So it's just – we just talk about it whenever it comes up really.'*

[24] Square brackets denote additional information inserted by researcher for clarification.

(Tina, 49y, PGT, Positive)

These were the kinds of discussions that occurred when the family sat around the table for dinner rather than as planned meetings or activities. In some cases, younger family members were excluded - or, more accurately, not engaged with - so as to protect them from something they would not understand or need to worry about. Or, if there were certain family members who did not want to discuss it (see disengagement below), then these discussions simply happened when that family member was not around.

2. Increased engagement

One way of combating this risk of cancer and the associated feelings was to engage in regular surveillance, such as yearly mammograms. This was habitually coupled with a hyper-awareness of potential symptoms, which was also impressed on other family members, particularly offspring. This appeared to be a subconscious coping mechanism to deal with their fear of developing cancer. This hyper-awareness and attention to self-examination opened the dialogue about the potential risks with their children from a young age and increased their awareness of the family history.

> *'It's like my son will say something "I feel I've got a pain here, I've got a lump there". I always say to him "You do check yourself don't you?" and he'll say "Yes, yes, Mum I do!" That's the sort of thing, and I say to Sarah [daughter] "Make sure you check yourself" and she'll say "Yes, Mum I do!" So we do talk about it periodically; we do think when it comes up or she might say "Oh I've*

got a funny feeling" I say "Well, make sure if you do find anything just go to the doctors and get it done straight away.'

(Julia, 58y, DGT, Inconclusive)

'I have always told them [her daughters], even when they first started getting their breasts and that, that you've got to check yourself. I mean, they've always done it and I've always done it. I've always made it a thing and I've always said to them, you know if you feel anything different or you think "Oh it's not right", I said, you let me know – coz when they were at home, obviously younger, I'd say "You tell me and we'll be straight up the doctors". I'm probably a bit paranoid about it. So, no, they've always checked themselves from a young age and they still do now.'

(Molly, 49y, PGT, Negative)

3. Disengagement

Disengagement tended to happen only with or by certain relatives. There were several accounts of relatives or certain groups of family members who did not like to discuss such things, as demonstrated by Elizabeth, who knew one side of her family really did not like to talk about medically related things, especially cancer:

'But from a communication point of view, that [side of the] family [points to eco-map] are really funny about medical stuff, you know. You don't use the 'C' words still… Well yeah, they have a very different communication style within that family. You know, the parents and the three children and their

families, they just don't talk health at all. They don't talk health in a way that gives you anything you can sort of get your fingers into, you know. It's all very vague, and I mean I didn't know for years that my uncle had prostate cancer; I mean, I knew he took tablets and he was often off having little operations and things. But it wasn't discussed, and he told me about his treatment after I'd had mine, which was fascinating.'

(Elizabeth, 54y, DGT, Inconclusive)

This was often associated with older generations, especially when it came to something as modern as genetics. Brenda's experience represents that of many participants who had older relatives, who felt uncomfortable talking openly about the potential family risk and genetics:

'I've got an aunt (my mother's sister) we tend not to talk about this because of the generation really; she's gone into her 80s. I sort of keep her very loosely up to date if there are any developments, but tend not to talk about the genetics thing because I found, you know, they don't really – they are not so open to that way of thinking about breast cancer!'

(Brenda, 58y, DGT, Inconclusive)

To overcome this disengagement, participants simply did not include these family members and, in some cases, friends, in those discussions, but sought other people to discuss the family history of cancer and its implications with. At this stage in the process, before genetic testing, there was no need to 'rock the boat' by forcing others to talk about something they found distressing. This changed when they needed specific information on the family history for their genetic

counselling appointments (refer to section 6.2.2 in the next chapter), or once they had their test results and felt it was important to pass on risk information (refer to Chapter Eight).

Other challenges came, not from the topic of discussion, but from individuals' personalities and communication style, which made such discussions difficult. Certain family members were often accused of being hard to communicate with, such as Zena's father below, because they had a reserved, shy or private personality:

> *ZENA: [Dad] is a terrible communicator, absolutely awful; always has been, all my life. Better if I ring him up and talk to him, he's better than face to face.*
>
> *INT: Why do you say he's a bad communicator?*
>
> *ZENA: He's a very, very private person. He's not very good at showing his emotions, never has been. Never ever has been, and he'd be the first to admit that you know. You know, I was talking to my brother a while back about that (a few years back about it, now) and it's funny, he's got a way of hurting us all. He's got a way of hurting all four children by showing very little interest in us, as it seemed when we were growing up. But I think that was his coping mechanism; he couldn't show how he felt or anything and I just think he shut himself off quite a bit and he's not able to communicate freely with us that well, except on neutral things really, yeah. And still isn't, but we're older now and accept that sort of thing. It's not that he's not interested, he doesn't want to pry, you know, I think, he's just such a private person.'*

(Zena, 49y, DGT, Inconclusive)

For Brenda, although she was interested in the implications of the family history of cancer and potentially genetic testing, it was not until her mother passed away that she felt she was able to pursue it, as she did not approve. Her mother's different view meant Brenda avoided discussing it with her.

> *'My mother was never keen on that at all; she said breast cancer is something that you tend to get when you're older; many old people develop it, just 'cause they get older. There's nothing wrong with our family, nothing wrong with our genes! And she really didn't support us (or me) having any genetic testing.*
>
> *And it wasn't until once she'd died that I felt I was able to – I was more free, in a way, to go ahead with genetic testing.'*
>
> (Brenda, 58y, DGT, Inconclusive

5.4.3 Stimulus to Pursue Genetic testing

Typically, there was ''one more'' diagnosis in the family that seemed to confirm their feelings about the significance of the family history of cancer, and which triggered either one member or several family members to take the matter further. This was certainly the case for Eloise, whose cousin's diagnosis, especially at a relatively young age, really prompted her to consider seeking advice about her family history.

'Erm, so we'd had Nan, and my aunt, and then Mum with it. They had all died from it and then Lyn [cousin] was diagnosed… erm, in 2004. And she was 41. I started to think about it then. I thought 'Well hold on, Mum was early 50s'. I mean, well, she was... 50… she would have been 50… she was six weeks away from her 53rd birthday. So, which I thought was young.'

(Eloise, 42y, PGT, Positive)

Alternatively, rather than a new diagnosis specifically, it could be that further information about the family history was discovered that prompted further exploration.

'Dad started doing the history of the family tree thing and they managed to track down her Mum's death certificate and it said on there that she'd had ovarian cancer. I was already been screened for breast cancer, you know because of the family thing. So I was having mammograms every year and all of that, so they were watching me with that anyway, but as soon as the ovarian cancer came up we were far more interested then about taking it further.'

(Kerry, 45y, PGT, Positive)

Deciding to investigate genetic testing followed similar communication patterns to those described above. They engaged with those friends and family whom they thought would be interested, and invited any relevant family members to join them, while disengaging with those they knew or thought would not be interested or would disapprove.

Other participants were introduced to the genetic service as a result of another family member receiving a positive test result for *BRCA1/2.* Learning that a family member had been tested and received a positive result often changed the way the matter was discussed. Rather than talking about cancer and the family history, the focus shifted to genetics and specific risk. Family members tended to fall into three groups: those who automatically decided they wanted to be tested and openly talked about it; those who were undecided about undergoing genetic testing, but were willing to undergo genetic counselling for more information and to discuss it further; and those who were not interested and did not want to discuss the matter at all.

> *'My sisters rang me up. They just said that Rebecca [sister] had been found to carry this gene, but the gene increased the likelihood of breast cancer by 80% or prostate cancer by 10% in a man and that, if it went on through, if we were carriers, it could go on to our children and our children's children. And I mean there was never a question to me. I immediately wanted to be tested to find out whether or not. I knew at that stage, and I knew I would have to have the test, so I contacted the people immediately because my son was getting married in late September/early October and I really wanted to be able to tell him whether he had to be aware of this risk in his life.'*
>
> (Arthur, 61y, PGT, Negative)

> *'And Frank [her cousin] phoned me up [after his daughter had been found to be a carrier] and said "Look Annie…" (and he felt really bad about it). He obviously felt really guilty, saying to me "Look, you'll have to go– it would be*

a good idea if you went to be tested" and I said "Well, I have no problem with that at all" and I didn't. When we saw each other and shortly after that we talked a lot about it.'

(Annie, 65y, PGT, Negative)

5.5 How those undergoing genetic testing for *BRCA1/2* talk to their friends and family during Stage One.

Communication during this first stage, Cancer in the Family, concerns how participants and their family talk about cancer before genetic counselling and/or genetic testing even became an issue to them. These were unobtrusive conversations, which followed normal communication patterns within the family, and were prompted by diagnosis, disease and death within the family. For example, they talked about how common cancer was in the family, what that might mean for family members and where it might come from. Communications were often coupled with a hyperawareness of risk and cautioning relatives, especially offspring, to the importance of self-awareness, such as, regularly checking for lumps in the breast.

5.6 Conclusion

This, the first of four results chapters, has reported the participant characteristics and the main features of the eco-maps. In order to answer the research question on when those undergoing genetic testing talk to their relatives, the primary findings of the qualitative interview have been

split into four stages, spread over four chapters. The first investigates how participants talked about their experiences of cancer and its implications, before their genetic testing.

The key findings in this chapter show that, before genetic testing, participants talked about their family history of cancer and what it might mean for the family in a variety of ways: from discussions about being a normal part of life in a family where cancer is so prominent, to disengaging from those who did not want to talk about it or disapproved.

Experiences of cancer, in particular their family history of cancer, and how the family viewed the cancer as a whole (shared health beliefs), shaped how the participants and their families talked about the disease and genetic testing. For some family members, it encouraged open communication to increase awareness of the dangers, particularly in the form of hyper-awareness of symptoms, as a strategy to hopefully catch any disease early. For other family members, in particular older relatives, it made them nervous and reluctant to talk about such distressing things. There was evidence of participants mediating how and when the topic was discussed in order to protect family members from distress. Discussion was often prompted by diagnosis and treatment, but there was a need to negotiate personalities and communication styles.

The next chapter will look at how participants talked about their experiences and genetic testing during their genetic counselling.

Chapter 6 – Stage Two: Undergoing Genetic Counselling

6.1 Introduction

This chapter investigates how those undergoing genetic testing for *BRCA1/2* talked to their relatives when the participants or their relatives went to the Genetic Service for their genetic counselling (Figure 20). During this stage, communication opens up to include more distant relatives due to the need for information or assistance.

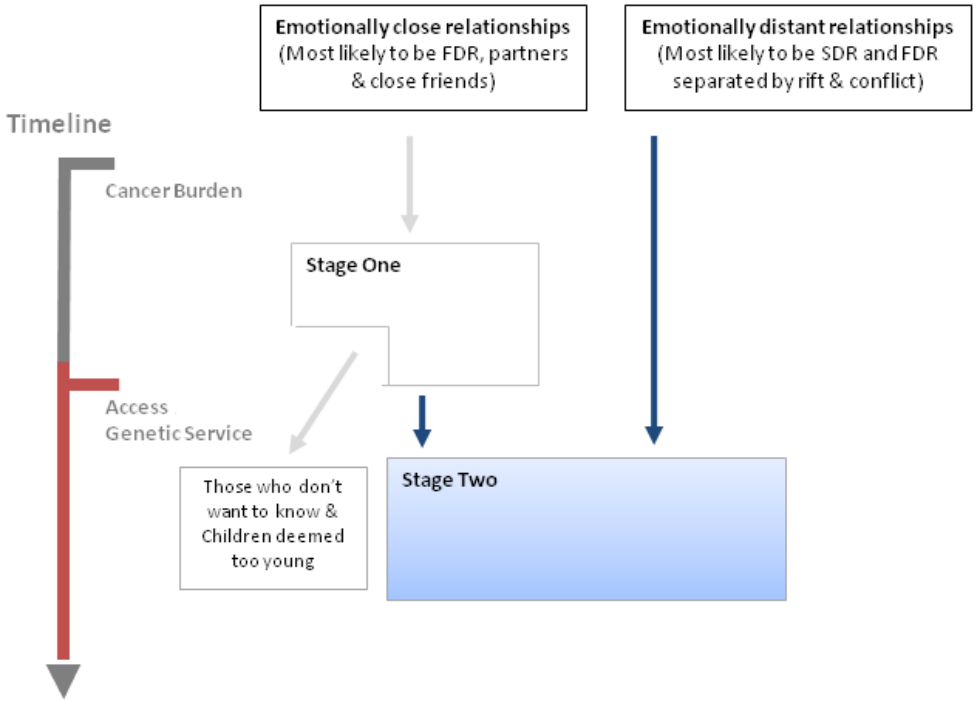

Figure 20: Stage two: Undergoing genetic counselling

6.2 Stage Two: Undergoing Genetic Counselling

6.2.1 Initial Experiences of Communication About Undergoing Genetic Counselling

In order to have genetic testing, you have to partake in genetic counselling. This process potentially opened up communication on the subject, especially with those who were already part of the stage one communications about cancer in the family.

> *'I mean everybody in the family read the booklet and what they sent, all that stuff, and the literature I had. And, you know, went "Ooooh oooh oooh" but you know "don't let's worry about this until I've had the blood test and then we can see what the options are"'.*
>
> (Annie, 65y, PGT, Negative)

Annie, like many, involved her family in her genetic testing by sharing the news and the literature she received. But she also played a role in managing the family's reactions to it by encouraging them to wait for the results, as shown in the quote above.

The data illustrated that the people identified as close and important (two or three lines on the eco-map, mainly first-degree adult relatives, partners and close friends) by the participants knew about the genetic counselling, and the opportunities for genetic testing, at this stage because it was perceived to be 'something going on in their lives', so it was discussed with others. For many, it was not so worrying, as they were not expressly seeking emotional support by

discussing their genetic counselling with others; rather, they found it as something different and interesting to talk about:

> *'And also you just generally talk – I would talk to people because people would say "How's your Dad?" and I would say "Fine, but he's going for genetic testing". "Oh why?" der, der, der, der. And "Oh you might – Oooh and then what?" and it sows a seed, doesn't it? (INT: What kind of people?) Friends and family mainly.*
>
> (Nicole, 39y, PGT, Negative)

For people like Eloise, who had had cancer,[25] going for genetic counselling felt like, and was perceived by others as, an extension of her treatment and, therefore, was discussed in much the same way.

> *INT: You mentioned that your family knew at the various stages?*
>
> *ELOISE: Yeah*
>
> *INT: Did you talk to them at each stage?*
>
> *ELOISE: Yeah they all…I mean, with things like going for an MRI scan, going for a mammogram. They'd all know when I was going and they'd ring up to say, you know, 'What's happening next?'. And I'd say 'Well next I've got to go for counselling'. 'Well when's that?' And I'd tell them. And then it might go quiet, especially like from Dad. He might go quiet. But then on the day, he would ring up. 'How did you get on?' Or if he didn't, then I would ring*

[25] Although Eloise had had cancer, she was not the first person in her family to have a *BRCA1/2* test, hence she had a predictive genetic test rather than a diagnostic genetic test.

him. It was very open. Right from the start. Even when it was just like, 'Oh, my doctor referred me' initially to the Breast Team at [city]. They all knew.'

(Eloise, 42y, PGT, Positive)

There did not seem to be a sense of secrecy around partaking in genetic counselling or genetic testing, unless a conscious decision was taken to not tell a particular individual because they were deemed too young or too vulnerable. One exception to this was Maya, who had concerns about telling her workplace in light of contract renewals, and so managed the time of disclosure until after her new contract had been approved:

'The only thing that slightly frightened me was work. I was just going on to a new contract and I needed my new contract to start in the January and I was having the test in December; I wanted to get my contract sorted because I thought I'd never been sick, but if I have to go for a mastectomy I need to make sure my contract… So that was the only thing that I held back slightly, on telling them all the details till I knew my contract was all right. But otherwise, no there was no problem; there wasn't anyone that we hid it from.'

(Maya, 47y, PGT, Negative)

The genetic counselling process provided new information that the family were not aware of regarding cancer and what the risks might be to family members. For example, for Molly and her family, the knowledge that they could also be at increased risk for other cancers, in particular ovarian cancer, came as quite a shock to them. They were very anxious about their considerable

family history and experiences of breast cancer as it was and this new information was further cause for concern.

> *'[The Genetic Counsellor] said to my sister, because she had the gene, that she was at more risk of getting, erm, cer…cer… 'cervaian' cancer and did she know about that. And we thought, we didn't, because we'd always assumed it was just breast cancer. Never worried about anything else, it was always, sort of, breast cancer. So they said that her options would be to have the other breast removed and have a full hysterectomy. Which was a bit of a kick in the stomach, because none of us thought about that, you know.'*
>
> (Molly, 49y, PGT, Negative)

The link with ovarian cancer was also a surprise to Elizabeth. As mentioned previously in Chapter Five, she was undergoing genetic testing because of the concerns of her teenage daughter. As far as she was concerned, she'd already had breast cancer so there were no major consequences for her.

> *INT: Did that surprise you?*
> *ELIZABETH: It did, yeah, because then she explained why there was an implication for me obviously and that I should – if it came back, as that then I had to think about my ovaries, which were no longer required! I suppose I was thinking about it in terms of Grace [her daughter] and I wasn't – I thought, you know, the deed was done as far as I was concerned and it was – I thought I was in a watch, wait and see and I didn't appreciate that there*

might be something that I could actively do to prevent further events… I didn't think that it was going to affect me at all, you know, in terms of my future health, and it was only that comment… 'Your ovaries may –'. But it made me realise that perhaps I'd been a bit narrow on this, but I just didn't have that knowledge.

(Elizabeth, 54y, DGT, Inconclusive)

This 'new' information had several implications for communication. Firstly, new information was perceived as interesting and, therefore, when they were discussing how their genetic counselling appointment had gone with close family and friends, it was recalled and discussed. Secondly, it sometimes adjusted the participants' perception of who in the family the information was relevant to; for example, relatives who had previously had breast cancer now needed the information about ovarian cancer risk as well.

6.2.2 The Need To Gain a Complete and Accurate Family History Opened New Lines of Communication.

As part of the initial stages of any genetic counselling, patients are asked to complete a family history form, which acts as a prompt for discussion. In order to do this, they often sought the advice of other family members who had more knowledge of the exact details of the family history.

'Rosa [cousin] seemed to sort of know more about the aunts than I did, because obviously she's 10 years older and she sort of just remembered things. And she has – when my Mum died, she took all the paperwork and all sorts of things connected to my Mum because she was the eldest so she takes charge! She took charge, so she sort of knew a lot more than what we ever did.'

(Julia, 58y, DGT, Inconclusive)

'[My niece] rang me one day and explained that, because it was in the family for so many generations, they wanted to know if someone could give the details you see. So of course I wracked my brain and wrote it all down as far as I could and sent it off to her. And she took it to them.'

(Shirley, 78y, DGT, Inconclusive)

Sometimes this information could easy gained by asking emotionally close relatives who were already involved in stage one discussions. However, especially in cases where matriarchal family members such as mothers had died, that was not always possible. This often opened communication with family members who the participant did not feel that, under normal circumstances, they were as close to, or had no regular contact with; but rather, they were included because they had knowledge of the information required. These included contacting either:

- Otherwise emotionally close relatives who did not like to talk about the family history of cancer (refer to 'Disengagement' in section 5.4.2) and so were not normally involved in such discussions; or
- Emotionally distant relatives who were not part of participants' normal communication patterns.

Getting emotionally close relatives who were not usually involved in family discussion about cancer to engage.

For some participants, gathering details of the family history was a hard subject to discuss with certain relatives, especially parents, as they did not want to cause them distress. As the quote below demonstrates, Zena found it hard to talk to her father; tensions arose between needing to know the information and not wanting to upset him.

> *'And I tell you what is hard, is finding out the family history! That's quite tough if you are going too far back, you know, or sideways as well. That's quite hard, especially with a dad that doesn't tell you very much, that was very hard. And that's where my sister did well. She actually got all the forms from her genetic team and actually took them all round to my dad the same day and said "Right give me all the family history".*
> *She's a sort of two feet in sort of person, you know, so we do laugh. Yeah, she'll do that and she won't worry that she's ruffled feathers or anything you know whatever, but I don't think he had much choice there. So that was quite good 'cause she got all the information I didn't know.*
>
> (Zena, 49y, DGT, Inconclusive)

Clearly, as well as finding it hard to talk to her father because he 'wasn't the sort of person' she was comfortable asking, Zena's own reported shyness played a significant part in making this a challenging situation for her. Luckily, her sister's more outgoing personality allowed them to overcome this. This is a good example of negotiating with another to do the 'hard' talking, a strategy adopted by many of the participants.

Conflict and rift within the family and family secrets made gathering details of the family history and generally discussing genetic testing harder. Discord in the family habitually meant genetically close relatives, i.e. first and second-degree relatives, were not in contact and often not willing to be in contact. Jan found that her parents' divorce and subsequent estrangement from her father made gathering details of the family history even more challenging:

> *'I've found it very difficult to get the history, because my parents were divorced and my father's in America. We had no contact with him after they divorced. So getting that information was difficult; I only actually started gleaning information once relatives started to die, which I suppose is probably how it happens. I don't know. But three years ago my grandfather (my maternal grandfather) died. He was living in America so I went to deal with his estate and it was at that point that I found out information that I had a Jewish heritage… I had Hungarian Jewish maternal lineage, which I remember being brought up in the interview with the genetics lady. She asked did I have a Jewish background, and I came back to her and said 'Actually I did! I do have a Jewish background' and she explained to me that*

they believe that part of the breast cancer gene comes from that background'.

(Jan, 44y, DGT, Inconclusive)

It was only after the death of her grandfather that Jan discovered he had changed his surname in order to disguise his Jewish heritage, which was something that, as far as she was aware, had never been discussed within the family.

Asking emotionally distant relatives for family history information.

These contacts were more formal in nature than the ad hoc discussions referred to in stage one (Chapter Five) and tended to require a specific plan about who to contact and the best way to approach them.

'I wrote. Because my Dad hadn't kept in touch with Mum's side, you know, I wasn't too sure whether they were still living at the address I had, so I wrote and got a reply back more or less straight away with lots of information in it [from maternal cousin, Nancy]. And then I think I must have rung Nancy just to say I'd got it, and ask her what she thought of the genetic testing (because obviously with her being in lineage she was quite interested in the family tree side of things, as well). So she said "Right, I'm going to go to Records Office and see what I can find basically" and that's when she found my Mum's grandmother having breast cancer, so she sent me over a

photocopy death certificate with it on there, so it was written on there. So she and I communicated quite a lot by letter and email and phone.'

(Tina, 49y PGT, Positive)

In this way, the family history form acted as a facilitator of family communication. It was common to write to more distant relatives. In many cases, this was because the participant, or another family member, only had an address for the relative they needed to contact; but also a letter was less intrusive than a telephone call. Writing a letter gave them time to plan how they would introduce the topic and allowed them to structure their message. Once the channels of communication were open, then other, faster, forms of communication, such as telephone calls and emails, were used.

Once contacted, that relative informed others, so information about the genetic testing naturally cascaded through the family.

'I rang him up, my uncle. Because he had information about my mum's side of the family. Because you have to go back don't you, like Grandma. So he helped me with all that, obviously because he knew more details. You sort of get, you've got to get all the dates and all that correct. So really in the process of gathering information people got invited.'

(Molly, 49y, PGT, Negative)

For others, needing specific information gave them a 'way in' to discussions regarding the significance of the family history and potential risk information. For Elizabeth, being able to give her cousin a 'task' greatly relieved the pressure of having to open up the communication on the subject:

> *'Like talking to Olivia [cousin] and getting her to get the information on my grandmother, we were able to discuss it sensibly because there was actually something that she could do (she could contribute). She could scuttle off down to the cemetery and you know find dates because nobody has got death certificates. So that was involving her.'*
>
> (Elizabeth, 54y, DGT, Inconclusive)

6.2.3 Experiences of Approaching a Relation about Diagnostic Genetic Testing

When conducting genetic testing for *BRCA1/2,* it is usual to begin the testing in a family member who has already been affected by cancer (diagnostic genetic testing). In some cases, the person who initiated contact with the genetics team was not eligible for predictive genetic testing, as diagnostic testing had not yet been done. So, they needed someone else in the family who had already had cancer to agree to be tested first.

MAYA: I phoned my GP and we filled in some forms; and we were all called [to the Genetic Centre] at the same time. So we all went to the appointment together: my Mother and Father, Leon [husband] and I, and Nicole [sister].

INT: And how was that?

MAYA: It was really helpful actually. Very helpful. We all reacted very differently to it… Quite early on it was obvious that Dad needed to have the test first because he was the one [who had had the cancer].

(Maya, 47y, PGT, Negative)

Maya's father was directly involved in the initial consultation at the Genetic Service but, in many cases, the person eligible for diagnostic genetic testing had to be contacted following the genetic counselling appointment and their consent sought. For some, this was their mother or another first-degree relative who had been involved in the stage one discussions; for others, however, it was necessary to contact more distant relatives that they were not normally in close contact with. As a result, when collecting details of the family history, making contact with such a relative opened up the communication to the wider family not involved in stage one and caused a subsequent cascade through another branch of the family.

'The Hospital in [City] said to my niece that it would be useful if I, because I was the last living person [who had had cancer], could give the blood to be tested to see if I was carrying the line you see, the gene. So she rang me up, we aren't normally in that close contact – just the odd Christmas card. I was very, very willing to do that, particularly as she was terribly anxious, so I

said yes I was willing to do it. Now, a lady like yourself, a very nice person, came along and we had a lovely morning, and then I gave the blood and it was all very easy... I told Daphne [daughter] what I was doing… and because of the conversation that sometimes sons can be affected, I rang my son up'.

(Shirley, 78y, DGT, Inconclusive)

Within this study population, approaching the family member about potentially undergoing diagnostic genetic testing was done by the person that had sought the genetic counselling, rather than by a health care professional in all but one case. Many, including Molly below, felt it was their responsibility to talk to that person directly because they knew them, so it was more appropriate:

'At the time they [the Genetic Service] said, yes there is a test that they could do but they needed a person that was alive that had the cancer. 'Course then there wasn't - my mother had already died. But in the meantime, Tonya [sister] got cancer and had her breast removed and blah, blah, blah. So I contacted them again, coz they said any changes to your family let us know. So obviously I did. So I went back with my husband. And they said they could do the test now if Tonya was in agreement. So I said, ok then, I'll speak to her - because they couldn't contact her just out of the blue. I said I'd talk to her. So I went over and spoke to my sister'.

(Molly, 49y, PGT, Negative)

While contacting more distant relatives for family history, information tended to be initiated by letter, while contacting someone to ask them to undergo diagnostic genetic testing was done over the phone (as in Shirley's case) or in person (as in Molly's case). The closer the relationship, the more personal the contact tended to be. For example, Molly approached her sister who, whilst she reported not being that close to (one line on the eco-map), she does have contact with; therefore, she made the request face to face. Whereas, it was Shirley's niece who made the request of her, whom she was not normally in contact with – *'just the odd Christmas card'* – and this was done over the telephone.

Numerous tactics were used to encourage relatives to undergo diagnostic genetic testing. For example, when Molly went to ask her sister Tonya, who had had cancer, if she would be tested first, she used emotive arguments by drawing on the potential risk to her sister's offspring. For example:

> *'I said: would she have this blood test done? I said: if not for yourself, because she's got three boys and a girl, then for your daughter.'*
>
> (Molly, 49y, PGT, Negative)

In another case, Kerry felt that her sister purposely 'played down' the significance of what a positive test result would mean when approaching their mother, who was dying from ovarian cancer at the time and was highly confused:

> *'There was no point talking to Mum about figures (percentages or anything like that) because (a) she wasn't very well and (b) she didn't really understand I think mentally.... [My sister] Gillian said to her: "It's only a*

blood test Mum that's all it is", that sort of thing and: "they'll only go and find out whether – it's just a blood test and it will show whether the breast cancer is linked to the ovarian cancer".'

(Kerry, 45y, PGT, Positive)

Following this, Kerry and her sister had made a pact not to tell their mother they had received their results as they felt she was too vulnerable and likely to be upset by them. Despite this, when their mother asked, Kerry disclosed both her and her sister's results to her. At this stage, her mother was in the last stages of her life after a difficult battle with ovarian cancer and Kerry reported she was highly confused and so did not really react to the news. It could be hypothesised that the guilt Kerry felt about encouraging her to undergo diagnostic genetic testing for them, without really understanding what she was doing, made her want to be as open as possible with her at the later stages.

Many expressed concerns about how best to approach the matter of diagnostic genetic testing with relatives. Often, these concerns were linked to their assumptions about the relative's emotional and or physical state and how they would react. Some relatives were not even considered, as the participant felt there was no way they would agree and, therefore, not worth the tension that would potentially occur if they were to react badly and refuse to be involved. Eloise had concerns about approaching her cousin, as she perceived her to be too emotionally frail and without support in the form of a partner. Once again, a conflict arose between needing the information and not wanting to cause harm or distress:

'They did explain that, if it was to go any further, Lyn [her cousin who had had cancer] would have to be the one to be tested and whether she would agree to that. I did find it quite difficult approaching Lyn because… she, erm… she was still sort of coming to terms with her own illness and getting over it and she doesn't have a partner. So she is alone with it and, you know, she lived alone. I sort of felt like she'd been through enough. She doesn't like hospitals, and she is not comfortable in hospitals … And so I thought "Oh God… I want to know, but how do I ask her?" And in the end I just thought you have just got to ask her, explain. So I did. And she was absolutely fine. She said "of course".'

(Eloise, 42y, PGT, Positive)

From the perspective of Eloise, it was challenging to raise the subject with Lyn because of the difficult personal circumstances, but it went better than she had anticipated because she was able to explain fully the reasons for her request.

6.2.4 Experiences of Being Approached about Diagnostic Genetic Testing

As discussed above, those participants that needed to ask another to undergo testing first preferred to do it themselves rather than have the geneticist do it. However, Julia was on the receiving end of such a request; rather than being contacted in person, she received a letter 'out of the blue' from the genetic service asking her if she would come to discuss diagnostic genetic testing so more distant relatives could access predictive genetic testing. To this date, she does

not know who started the process, or which relative that information was for. She reported that she had been happy with the way she was approached and willing to go along and be tested. However, when asked if, in the case of her immediate family, she felt it was better for information to come from a health professional or from a family member, she said she felt that ideally a family member should make the initial contact, followed by a professional, so that the person had the option to say no.

INT: Is there a particular reason you think it's important that you do it initially?

JULIA: Yes I think it is because it's your family, so then they've got a choice to say to you 'I don't want to know anything about it' or 'yes' they do want to go. But then, if they still got seen by a professional, then they would explain it more. I don't think people can explain it better than a professional; they know what they are talking about – you get the gist of it, but really I can't explain to my children what it's all about. All I can say is that it's a gene in that blood that will tell you if you've got cancer or you haven't got cancer, that's the basic I know, where they can probably explain a lot more and they probably understand more than I did.

I think initially you talk to them, but I think that the doctors or professionals explain more. I think so, well personally anyway, I would like that.

(Julia, 58y, DGT, Inconclusive)

In fact, Julia had limited understanding of the genetic testing and, in the discussion, she admitted she was not one hundred per cent sure of the details:

> *'I went there and she talked to me about the testing. I did not really quite understood it; I understood you take the blood and they can find out if it's in the children or not (that's how I sort of understood it).'*
>
> (Julia, 58y, DGT, Inconclusive)

Although she was able to convey the essential information to her son and daughter, Julia was not confident in her ability to relay it accurately. She relied heavily on giving them the information booklets and them going to see the genetic counsellors themselves to ensure they received accurate information. This may explain why the aforementioned follow-up by a professional was so important to her. In fact, even those that reported they were confident they had understood the genetic information still used information leaflets and letters summarising the discussion with professionals to back up what they were saying to relatives (refer to section 6.2.6). This approach increased their confidence in their abilities to discuss genetic testing for *BRCA1/2* with relatives.

Being asked by a relative to have the test done could be emotionally demanding. For Katherine, being asked by her daughter triggered feelings of guilt about potentially being a carrier and passing the mutation on to her children:

> *'It was our second daughter, Marian. She is a nurse and she approached me about it. I don't know where she heard about it, but then she went to [her local hospital] and saw someone there and then she got in touch with me.*

She asked if I minded if I [had the genetic test], because she said with [the risk of] ovarian cancer she hasn't got children and if it proved that I had the gene for it, she wouldn't mind having her ovaries removed because it didn't matter to her. I felt a bit upset about it at first, because I hadn't thought that I might have passed the gene on to my children, and it suddenly hit me I could be a carrier and my other daughter died from breast cancer, so it could be me that had passed the gene on to her. Or, I could have passed it on to my other children. Even my son, you know, he could have been – or grandchildren.'

(Katherine, 79y, DGT, Inconclusive)

Two separate participants spoke of the immense pressure they felt put under by close family members to have a diagnostic genetic test almost as soon as their cancer was diagnosed. Brenda felt her younger sister and female cousin were putting a lot of pressure on her to go and have the genetic test done as soon as possible so they would know if they were eligible for testing. When they were all together at a wedding, her cousin used the opportunity to have a face-to-face discussion, but that just added to the pressure Brenda felt under.

INT: So how did you feel about that?

BRENDA: Uuum – slightly not annoyed –I didn't want that sort of pressure, 'cause I knew I was going to have it done; I wanted to have it done when it suited me, not when it suited others. And it may have only been a matter of a few months or whatever. You know, they're the sort of people who down tools and do something now, straight away when it's got to be done and I'm

not. I would sort of fit it in a bit more carefully with other things that are going on in my life, which may make them think that perhaps I'm delaying. But that's not what it was – if any of that makes sense!

INT: Yes, it does. Don't worry. So was it a regular topic of conversation?

BRENDA: Yes, especially with the cousin... At Kai's wedding she collared me and I really couldn't get away very easily and she had a really in-depth discussion about the breast cancer in the family. And I know why she chose that time because we don't see each other a great deal, but it was the opportunity there that, you know – son's wedding!

(Brenda, 58y, DGT, Inconclusive)

Zena had a similar experience with her sister, who wanted to start genetic testing as soon as she had received her cancer diagnosis. However, she was not ready and needed to deal with her cancer treatment first.

'It was my sister, really. She was not causing problems, I wouldn't say it was problems as such, but it was causing a bit of hassle because when I told my sister [about her cancer diagnosis] she then went to her GP and started flapping about all the genetic tests and everything ('cause obviously she's concerned for herself and her daughter and her granddaughter)... So Faye wanted some sort of genetic testing going on and she felt now that somebody living was diagnosed that they could get the ball rolling... So she's trying to get things sorted from that point of view, and I'm trying to deal with chemotherapy and she's on the phone, and it wasn't a good time

because I couldn't cope with that at the time, I can only do one thing at a time. I wanted to get through the initial diagnosis, the initial treatment, and once you've gone through that, if everything was going ok, then it was time to sit back and look at the genetics. And I felt that that was the right time for me rather than right at the beginning; it was too much to cope with, too much at once.'

(Zena, 49y, DGT, Inconclusive)

Both Brenda and Zena needed time to come to terms with their cancer diagnoses and deal with their treatments before pursuing genetic testing. However, they found it very hard to express these feelings to family members who, they felt, were putting them under pressure. This lack of understanding actually acted as a barrier to family communication regarding genetic testing for *BRCA1/2,* as both Brenda and Zena started to experience feelings of resentment and annoyance towards their family members.

6.2.5 Communications about the Decision to Proceed With Testing

During and following genetic testing, there was also a decision-making process: they had to decide if they actually wanted the test. The process of genetic counselling is designed to make patients think: to consider all the options and likely outcomes so that they can make a well-informed choice. As a result, it was often discussed with the family as a source of support and guidance, for example, asking others for their opinions. When deciding whether or not to actually have the genetic test, participants drew on a range of experiences and family

discussions on the topic. Talking through the option of genetic testing and weighing up the pros and cons was common; this was especially with partners, who did not tend to express strong opinions of whether or not the participant should do the test, but acted more in a supportive role, and siblings and/or parents, who were also considering testing or had already been tested, and who did have strong opinions and tended to use the conversations to influence the decision.

For example, Gina discussed having the genetic test with her husband, Aiden. He was happy to let her make the decision; however, she still found having the conversations with him helpful and supportive:

> *INT: You mentioned that, when you were making the decision about whether to have the testing or not, you spoke to Aidan [husband] about that. Could you tell me some more about that?*
>
> *GINA: Oh yeah, yeah, absolutely because you know it has an impact on all our family life, but Aidan was very much – 'well, if you think that's the right thing to do, then that's the right thing to do'. And I went away and I did my research and I got booklets and pamphlets and everything else. I used to say to him 'we need to sit down so you know what you're letting yourself in for'… It was very helpful because, you know, we are a close couple and we go through everything together – there's no way, I would never have entertained doing this without him being fully in agreement.*
>
> (Gina, 41y, PGT, Positive)

On the other hand, when she spoke to her mother and sister, they were far more opinionated and directive about telling her what she should do. They presented, what they perceived to be, the positive side of the testing. For example, that knowledge is power and that they were being given an opportunity not available to everyone. These persuasive arguments influenced Gina's decision to be tested:

INT: Was there anybody else you talked to about the option of having the test with?

GINA: My Mum, I talked to my Mum (I talked to my Mum and Dad) and to her there was kind of like no debate, 'You should go and have it, because information, knowledge is power isn't it? You can then make decisions based on the knowledge that you have.'

I also talked to my sister about it, and she was very much 'Oh yeah, you know, I don't see why you wouldn't.'

I think everyone was very positive [about having the test done]– and again it kind of brought home to me how many people out there [are] walking around who don't know that they've got inherited genetic conditions or BRCA1 or BRCA2; and, you know, you have to come at it from that perspective I think. You have to think 'Yes I'm very lucky that actually I know this and I'm being given the chance to do something about it and to potentially prolong my life, you know, because otherwise the outcome might be very different.'

(Gina, 41y, PGT, Positive)

Molly found it particularly helpful to talk to friends outside of her family, as they were removed from the situation and did not know any of the people involved. This meant they could give her impartial advice as well as a level of confidentiality that what she said would not get back to the family:

> *'I think, because they were outside and they only know me. They haven't met any of my family. They only know about me, same about you at work if you talk about your family, that's the only part of the family that they know. So I think because they didn't know any of them, it did help me. I think because they let me talk and blubber, they might have only just said, 'Oh yeah yeah, you'll be alright, you'll be alright'. But I think it was just being able to talk to them without them knowing anybody else in the family that I knew, it didn't matter what I said to them, no one else would find out about it.'*
>
> (Molly, 49y, PGT, Negative)

6.2.6 Materials To Support Communication Regarding Genetic Counselling

Throughout the genetic counselling process, the participants increased their knowledge base about genetics; they were being given information from the genetics team including leaflets, booklets and letters documenting their discussions. This was repeatedly described as useful and

was often used as a tool for sharing the process with family members or friends. For example, letters were often photocopied and passed on. This ensured that people were receiving the correct information, rather than the informant relying on memory-recall of what can be quite complex genetic information for a lay person.

> *ZENA: So the fact that the letters were all written out as well, I was able to give a copy to my sister, because I felt that helped her along as well and she could take that back to her GP... the good thing was that I was able to have a copy of the letter which I was able to pass on to my sister, which makes it a lot easier if you've got it in writing, and then she can take that information and deal with it as she wants to deal with it.*
>
> *INT: Were they specific letters for her or literally copies of your letters?*
>
> *ZENA: No, I copied it for her. At least I felt I was able to give her all my information and not be secretive. I don't want to be secretive about it all.*
>
> (Zena, 49y, DGT, Inconclusive)

It was common for participants and family members to seek other sources of information, for example, searching on the internet. Several participants talked of the importance of only seeking information from 'trusted' sources, such as well-known cancer charities like Cancer Research UK, Macmillan Cancer Support and Breast Cancer Campaign.

6.3 How those undergoing genetic testing for *BRCA1/2* talk to their friends and family during Stage Two.

For discussions during Stage Two, participants described how they considered the personal attributes and characteristics of family members so they could adapt how they approached the subject as required. For example, Christina spoke about how she played a "*bit of a game*" to draw out of people their own perceptions of genetics and genetic testing were before she moved on to sharing with them, so she had some idea of how they were likely to feel about it. She reported saying things like "*well did you see such and such a thing on Panorama the other night when they were talking about ……*" in order to find out what their perceptions of the issues were.

It was important for participants to make discussion about genetic counselling "*a positive thing, don't make it into a doom and gloom thing!*"(Eloise). This was a serious issue for participants; however, they acknowledged it was important to talk to family members about their experience of visiting the genetic service in ways that were going to make them listen. For example, Gillian spoke about "*selling it*". She used examples of selling the benefits of genetic testing to her family on the basis that medicine was always moving on and how, in her opinion, the chances of them having to be bothered about this gene in ten or 15 years was minimal, but the information was available now and it could be so useful to the family. For her, it was about giving people the positive before the negative: "*sell the top end first, talk about medicine first and that they are doing some fantastic things*" (Gillian). In fact, many participants emphasised how medicine was constantly moving on and they often expressed to family members a confidence that this would

not be an issue in the future. These kind of persuasive arguments were frequently used in discussions with relatives.

Generally, participants felt that it was better if people were forewarned so no information-, like a positive result- came as complete shock ,therefore they saw a great advantage of including as many relatives, with whom they were close, to during this stage. In addition to which, they found it easier to talk to those family members who were aware of the genetic testing from the beginning, rather than those they contacted afterwards.

Often, despite their initial concerns and preparations, participants were surprised at how well their requests for information or involvement were received by family members during this stage. For example, Eloise was worried about asking her cousin Lynn if she would undergo diagnostic genetic testing because she felt Lynn "*was still sort of coming to terms with her own illness and getting over it and she doesn't have a partner….. And I felt like, she doesn't like hospital, and she is not comfortable in hospitals*". Eloise said she did not know how to ask Lynn, but in the end, "*I just thought you have just got to ask her, explain. So I did, I explained calmly why it was important and how it would help Barry [brother] and I, and our children. And she was absolutely fine. She said 'of course' "*.

6.4 Conclusion

This second findings chapter has reported how participants talked to others about their genetic testing during the genetic counselling process. This stage of family communication regarding

genetic testing takes place in the context of participants understanding and relaying the complexity of genetic information. Discussions also started with more distant relatives, specifically for information or help.

Barriers to family communication during this stage included:

- Participants, or their relatives, making a conscious decision not to discuss genetic counselling with certain family members;
- Not wanting to cause distress;
- Conflict and rift within the family;
- Participants, or their relatives, making judgements on whether individual relatives would want to know and/or anticipating their reactions as to how they would handle the information;
- A lack of understanding and/or support from relatives (sometimes leading to feelings of resentment, which acted as a further barrier to communication).

However, factors such as perceiving the genetic counselling as new and/or interesting information that was worth sharing; receiving information booklets and summary letters from the genetic counselling appointments; and needing further information from specific relatives, such as details of the family history or asking a relative to undergo diagnostic genetic testing, acted as facilitators. Those undergoing genetic counselling were also more likely to talk about their situation if they wanted support and/or guidance to help them make decisions about whether or not to pursue genetic testing.

The findings presented specifically look at how participants communicated. For example, with close friends and family the conversations were an extension of those happening in Stage One. Genetic counselling and the information learnt through the process were something interesting going on in their lives to be talked about. For those who had had cancer, it was often perceived as an extension of their treatment and was discussed in the same manner as that had been. Often, the participant took on the task of managing people's reactions, for example offering reassurances and encouraging them to wait and see what the genetic test results would be before acting.

When information was needed, for example in order to complete the family history form, from more emotionally distant relatives or from relatives who found it hard to talk about these things, a more strategic and proactive approach was adopted. This may include writing a letter or negotiating with other family members to broach the subject.

Conversely, a more personal approach was adopted when asking relatives to undergo diagnostic genetic testing so other family members could access predictive genetic testing. This was relative to how close they were to that person to start with. For example, with emotionally close relatives, this approach would be done face-to-face rather than over the telephone; whereas, with emotionally distant relatives, this approach would be done over the telephone rather than by letter. If possible, family events, such as weddings, were used as an opportunity to have face-to-face discussions with those they were not normally in contact with.

Participants felt the best approach was to be open and honest when discussing genetic counselling and to explain everything as fully as possible to relatives. However, there was

evidence of participants ‘playing up’, for example, by using emotive language and giving examples or risks to offspring, or ‘playing down’ the significance in order to get family members to agree to be involved. Clinical information leaflets and summary letters of genetic consultations, as well as online sources of information, were regularly used to support explanations and to provide family members with further information. Using these resources significantly increased participants’ confidence in their ability to relay the information accurately and in the best possible way.

The next chapter will look at how participants talked about genetic testing when they, and others, received their results.

Chapter 7 – Stage Three: Receiving the Test Result

7.1 Introduction

Receiving the test result prompted family communication regarding the genetic testing as participants shared their results. This occurred in two distinct waves (Figure 21). The first wave was motivated by a sense of urgency and involved telling those who were 'waiting to hear'. To be included in the group of those relatives told, it was not enough just to know about the testing. These people had to have some emotional investment in the process; specifically, those first-degree relatives and close friends involved in Stage One and Two communications were told. The second wave, involved telling those who were 'in the know' and needed to be 'kept in the loop' but with whom the participant was not particularly close. For example, those relatives contacted for information or to access predictive genetic testing during Stage Two.

Some participants had to wait a long time for their results. The testing system has changed in the last few years (due to the patent expiring) and through-put time has decreased dramatically. These participants had all received their genetic test results 8-18 months prior to the interview; however, some of them had had their blood taken for the test up to two years previously.

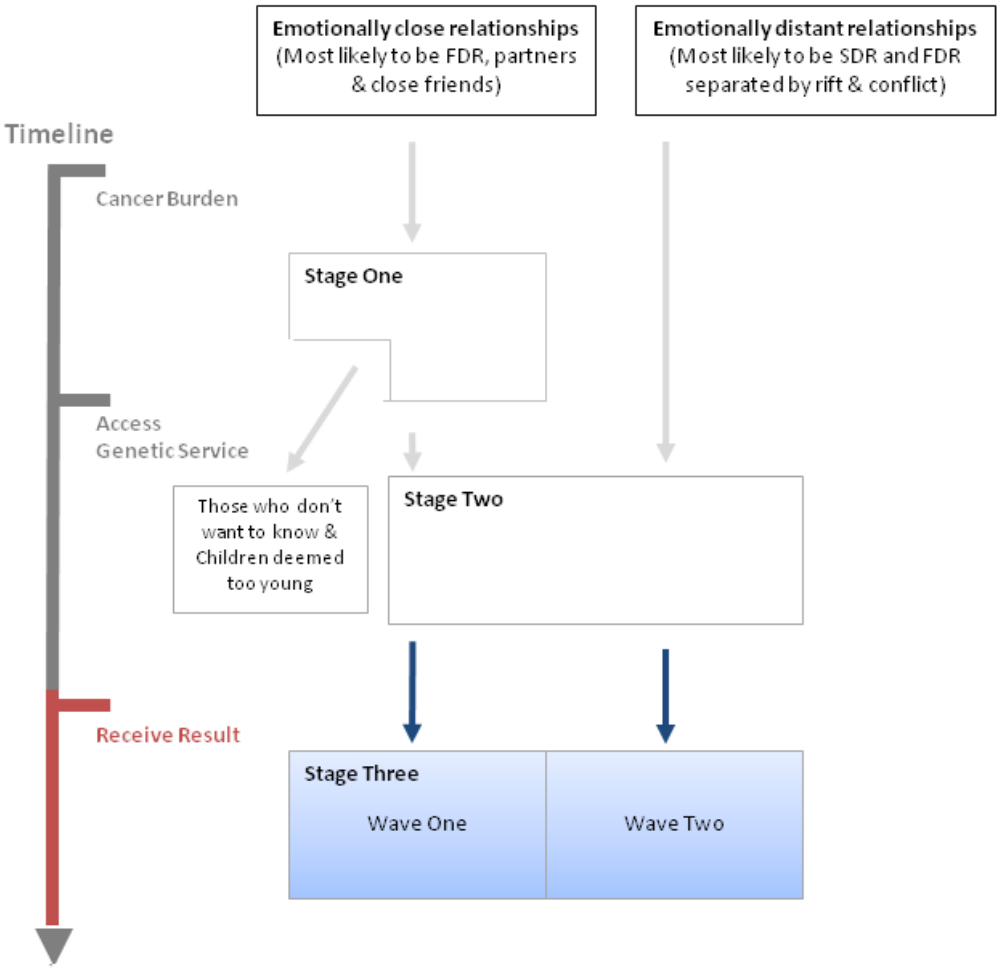

Figure 21: Stage three: Receiving the test result

7.2 Stage Three: Receiving The Test Result

7.2.1 Wave One: Telling Those Who Are Waiting

All participants who tested positive (a mutation was found in their *BRCA1/2* gene) were informed of their result during a specific 'results' appointment at the Genetic Service. Whereas, those receiving an inconclusive result (in the case of diagnostic genetic testing, no known BRCA

mutation was found) or negative result (in the case of predictive genetic, the specific mutation known to exist in the family was not found) were informed either during a 'results' appointment or by letter.

Having a pre-booked 'results' appointment meant that close family and friends, specifically those involved in the stage one discussions, were likely to know of the event in advance. Participants spoke of how these people were 'waiting' to hear from them, often reporting them as waiting in a state of anxiety or worry.

> *'And I know [Mum] was just waiting for the result. In fact, I didn't tell her the time of the appointment, I just told her it was on that day 'cause I didn't want her sitting there waiting for the phone to ring. You know, if I told her it was say 10.30, you know 11 o'clock, she would have been sitting there waiting by the phone. And I didn't want her to do that, I wanted her to just sort of carry on as normal, so I said "I'll tell you what day it is but I'm not telling you the time".'*
>
> (Annabelle, 51y, PGT, Negative)

The anxiety being shared by others meant the participants felt a huge sense of urgency to contact these people as soon as possible and let them know the outcome of the appointment. This was usually done over the phone at the first possible opportunity.

'Well, Aidan [husband] was with me when I found out. The next person I spoke to in the car [from her mobile] in the hospital car park was my mum (because I knew she was waiting by the phone) and I had to tell her.'

(Gina, 41y, PGT, Positive)

Because those participants receiving an inconclusive or negative result had the added motivation of being able to alleviate their relatives' anxiety or worry, one might expect them to communicate their results faster than those receiving a positive result. In reality, result status had little or no effect on speed or style of disclosure. Rather, the fact that close friends and family were waiting in a state of anxiety meant there was no less sense of urgency from those receiving a positive result.

'I let my parents know straight away; a phone call straight away. Actually, it was in the lift outside the [Genetic Service] office, yeah. They were delighted because they were obviously on tenterhooks. Because I think it must be a horrible thing as a parent, a horrible thing to feel that you've given something horrible to your children.'

(Maya, 47y, PGT, Negative)

'And, you know, the day that I went to get the results, of course, they were just sort of sitting by the phone waiting for me to ring them. In fact, I went to [Hospital] to get the results and went straight to… My daughter has a house in [Town], so I went straight round there and we had a glass of wine together to celebrate!'

(Arthur, 61y, PGT, Negative)

These people were contacted in a variety of ways, often as soon as the appointment had finished; for example, ringing them from the car-park or hospital coffee shop; texting them from the elevator at the hospital; and popping round to see them on the way home. Not a huge amount of thought was given to how the news was delivered, or when the news was delivered, but rather the importance was that it was shared as soon as possible.

Despite getting different results, both Faye and Annabelle reported feeling afterwards that perhaps they should have told their mothers their results face-to-face, but the sense of urgency took over:

ANNABELLE: We were going to go and see my Mum after the appointment but Tim [Husband] said to me 'Phone her up and tell her'. So I'm sitting in the car and he's driving along and I phoned her up... So yeah, I told her by phone, unfortunately; I should have told her face-to-face. But we did go and see her straight afterwards and make sure she was all right.

INT: You say you suppose you should have done it face-to-face?

ANNABELLE: Yeah, I should have really told her face-to-face. I mean, she was thrilled to bits; it didn't matter because I then got there quite soon afterwards and we talked about it, anyway. But the initial sort of 'guess what, Mum, I haven't got this gene' it was probably the wrong way to do it. But we were just so pleased that we wanted to tell her. And I know she was just waiting for the result.

(Annabelle, 51y, PGT, Negative)

'I'd just come back from the hospital and rang Mum at work. She said: "Oooh are you all right?" and I said: "Yeah, yeah, well I've got it". And she just burst into tears and I was just like "Oh, don't be upset, I'm fine, I'm fine, honestly you know..." And then I just said to Nicki [Sister-in-Law who drove her to appointment], "Oh, I'll have to walk round and see her 'cause she's upset now" and I thought "Well, I should have maybe waited until later" but you know, I knew she would have been conscious of the time when she hadn't heard anything.'

(Faye, 29y, PGT, Positive)

In both of these cases, the urgent news of the result was followed up by face-to-face discussions to 'make sure she was alright'. There was much evidence of participants working to manage the responses of family members; for example, offering reassurance and comfort if someone got upset. This was particularly true when people experienced feelings of guilt (see section 7.2.4).

Receiving the genetic test result by letter did not seem to lessen the sense of urgency to share the results immediately. Those participants that received their results by letter also contacted close friends and family that had been involved in the Stage One communications in order to share their results immediately.

> *'It was a weekday and I think the first person I told – I think I emailed my cousin, because obviously, I mean, there's like five kids on the other side so she was worried. I think I emailed her and then we talked about it when everybody else [husband and children] got in that night.'*
>
> (Karen,52y, DGT, Inconclusive)

> *ROBERT: The letter arrived and told me I was not carrying the gene anyway. So immediately I told my daughter. She was in the process of arranging to be tested... Having got the result I also let Nick [Son] know because he was waiting, I let me daughter know, I let Deborah [former sister in law] know, I let my niece know as they were waiting too.*
>
> *INT: Ok, and how did you do that?*
>
> *ROBERT: Well, the phone first, and I sent a copy of the result actually.*
>
> *INT: And was that the next time you happened to speak to them, or was it a specific phone call to give them the results?*

ROBERT: No it was specific. It was specific, yes, and almost straight away.

(Robert, 68y, PGT, Negative)

Even though there was no specific date by which the results would arrive, Robert still reported his children as 'waiting'. Once again, like those receiving their results in an appointment at the genetic service, the first urgent contact to share the result was followed up by the participant; in Karen's case, by a phone call and, in Robert's case, by sending a copy of the results.

There was no notable difference between how those who had undergone diagnostic shared their results compared to those who had undergone predictive genetic testing, nor between the status of those results. Consequently, it can be theorised that neither the type of testing (diagnostic or predictive), result status (positive, negative or inconclusive), nor how the result is delivered[26] impacts this wave of communication.[27] Rather, at this stage at least, family communication is motivated by a sense of urgency to share the results with people who know about the testing and are emotionally invested in the outcome and are subsequently 'waiting' to hear about the results, almost as much as the participant themselves. This emotional investment was predetermined by the closeness of their relationship with the participant and principally included those who had been involved in the earlier stage one discussion, explicitly friends and family with whom the participant had a close personal relationship and open communication (those scoring two or threee lines on the eco-map).

[26] Although only negative and inconclusive results were received by post.
[27] However, these factors may affect how things are communicated in stage four, and this will be discussed later (see Chapter Eight).

'I told most people over the phone. My brother because he knew I was going for the test so they were itching for me to get the results. So I told my brother over the phone. Told my dad over the phone. Wendy [her best friend] over the phone. Wendy [her neighbour], I told face to face coz she's always over. My step-dad I called straight away. Lyn [close cousin, who had had DGT for her] I did over the phone. I would say all on the same day. It would have been the same day. Coz they were the ones that knew I was going for the test and they'd all been with me, all before I'd gone for the test. So they were all like waiting for me to come home with the result.'

(Eloise, 42y, PGT, Positive)

7.2.2 Wave Two: Keeping Those Who Know In The Loop

The next group to be told were those who were 'in the know' about the participants' involvement with genetic testing and who needed to be 'kept in the loop'. Unlike the first wave, this communication tended to take place the next time there was normal communication pattern contact, rather than as specific contact in order to share the results. There was not the same sense of urgency as with the first group, but it was still seen as important to share the results with those who knew about the test; these were likely to be the distant relatives who had been contacted for information, or to access diagnostic genetic testing in stage two, as in the case of Elizabeth and her cousin:

'Olivia, my cousin who helped get the family info, and I speak every few weeks, so I just told her when I was on the phone to her. It wasn't a special call to tell her, nothing dramatic like that!'

(Elizabeth, 54y, DGT, Inconclusive)

First-degree relatives were not automatically told in the first wave. Therefore, it was emotional rather than genetic distance/closeness that determined whether relatives were told in this wave or previous wave. For example, despite being first-degree relatives, Molly and her sister are not very close, so she was told about Molly's result in the second wave:

'I didn't really speak to Tonya [sister] until later. She's... well, she's difficult... I mean, this sort of circle [points to other sisters and daughter on eco-maps] is very close, anyway. I mean, we are close to Tonya but she's not, sort of, in our club, so I didn't phone her up straight away. In fact, I can't remember if I told Tonya or if my dad told her, 'cause he sees her once a week so he might have told her. She definitely knows, though.'

(Molly, 49y, PGT, Negative)

This quote illustrates another point, in that the motivation for this wave of communication was to ensure that the participant's test result was shared with others who previously knew they had opted for genetic testing. This did not mean that the participant themselves necessarily engaged in the direct communication with those family members. There were several examples where

the responsibility to keep those relatives involved was assigned to someone else, usually either the person perceived to be the matriarch of the family and/or someone who had had cancer and was pivotal to the whole genetic testing process…

> *GINA: I said to my Mum, 'Tell Keira [Sister]. Tell the family'. So, gradually, after that it kind of just filtered down and it wasn't necessarily me telling people.*
>
> *INT: So your Mum played quite a central role in it, then?*
>
> *GINA: Oh yeah, yeah, very much so. Mum's been very central because I think obviously because she's been through it [cancer] so she probably above anybody else can totally understand... She's great, my Mum. She's a Mum and, like she always says to me, 'it doesn't matter how old you all are, you are still my children, you always will be'. She always wants what's best, so she is happy to do things like that for us.*
>
> (Gina, 41y, PGT, Positive)

…or to a relative thought to be in a better position, usually because they were in more regular contact:

> *'I told Pat [Cousin she was in the most contact with] and I said "Oh make sure that Joanne and Claire [other Cousins] know about this".*
>
> *Yes, so I told Pat and it was left to Pat to tell the other cousins and her daughters. I am not in constant communication with my cousins, so it made more sense for her to do it.'*

(Viv, 58y, PGT, Negative)

7.2.3 Perceptions of a 'Female Disease'

Generally, those family members who participants reported as being emotionally close to, with open communication (two or three lines on the communication map), were involved in stage one conversations about cancer in the family (see Chapter Five) and were told about test results in wave one. The exception to this was male relatives, in particular sons. Although those participants with sons reported being emotionally close to them and having open communication, they were less likely to be told in the first wave than daughters or other female relatives. For example, Annabelle told her sons, both of whom she scored with three lines on her eco-map, as part of her wave two communications:

> *'When I got the results I just said to my boys, "You'll be pleased to know that there's no chance that you'll have it 'cause I haven't..." I didn't go out of my way to tell them on the phone. I waited until the next time I saw them, so it was sometime within the next week. They weren't sort of waiting for the results themselves. I mean, their lives are so full they probably had forgotten anyway! So it was "Oh by the way I've got my test results" and they said "Oh yeah? How did you get on?" and that was it. "Yeah, ok, fine".*
>
> (Annabelle, 51, PGT, Negative)

Annabelle, like many other participants who had sons, attributes only telling her sons as part of this second wave to their apparent disinterest. In fact, the data suggests that both this disinterest, and the disparity between sharing with female and male relatives, may be due to the fact that male relatives are less likely to be involved in stage one and/or two communications as a result of perceptions on both sides of this being a 'female disease'.

Participants were often motivated to undergo genetic testing by a wish to provide risk information for relatives, but in particular to provide information to female relatives because they saw it as directly affecting them. This meant they were more likely to talk to the female family members than the male relatives about their genetic testing for *BRCA1/2*.

> *'I mean, I told my son what was happening and he's very supportive you know, but obviously I'm more concerned for my daughter, and she's more concerned, you know. It is something that's more likely to affect her than it would him.'*
>
> (Martine, 65y, DGT, Inconclusive)

When they went for their genetic counselling, many participants reported being surprised to learn that the family history of cancer in men was relevant; and that, if found, the presence of a mutation in the family could also mean an increased risk of cancer to males.

'I was quite surprised when – almost, I wasn't prepared for it when [Genetic Counsellor] said that Jack's [cousin] prostate cancer made her concerned that there might be a BCRA2; because, prior to me revealing that (which I'd almost forgotten to do), she was not going to give a testing. She said she didn't think there was enough, but she changed her view during our discussion... I would never have had that knowledge. And I wonder in the general population whether that knowledge is common. It's very much perceived as a female disease, isn't it?'

(Elizabeth, 54y, DGT, Inconclusive)

'I filled in the thing all about our medical history throughout the family as much as I could remember. And, they came back and said: 'yes, we think it would be worth discussing'. Erm, so I went and had an appointment with [the Genetic Service] and that's when I found out that the boys could be at risk, coz I didn't know at that point that they could also be at risk.'

(Eloise, 42y, PGT, Positive)

Even having learnt that men could also carry a mutated BRCA1/2 gene, which would put them at increased risk of cancer, the participants were more likely to engage in communications about their genetic testing with female rather than male relatives.

'I've got one other cousin, Neil, who is the son of my aunt. He's aware of it but he's not been involved in the discussions. I know you can get male breast cancer, but the view of the others was that he's not really – you

know, for us as girls they all felt it was very personal to us rather than to him. I mean, he discussed it with his mother, you know, and that's as far as it's gone.'

(Christina, 49y, PGT, Positive)

When male relatives were included in discussions, participants often justified this by stating that the male relatives had daughters who may be affected. During the course of the research interview, Gina became aware of the fact that she had not discussed the topic with her brother;[28] however, her attention was automatically drawn to the fact that there might be implications for his daughter, her niece, rather than him:

'I haven't really spoken to my brother about it. I actually don't even know if Lara [niece] is aware; I don't even know if they've had that conversation with her, so I probably would need to talk to my brother about that. It's important that she knows.'

(Gina, 41y, PGT. Positive)

When male family members were included, several participants reported either a lack of interest from them or a belief that the information was not really relevant to them. Where female participants had decided that the information was relevant to male and female family members, it was often necessary to be quite proactive to engage them. For example, Annabelle made copies of her mother's letter and directed her brother to take them to his GP:

[28] Because his name appeared on her eco-map.

'Like with my brother, you know, it was "wow, it doesn't really affect me" and we both (myself and my Mum) said to him "well, it does because you could have passed it on to your children. And men do get breast cancer but you've got to think of your daughter as much as anything". I made photocopies of my mum's letter and sent one up to him in the post and said "take that to your doctors" and he said "Oh, I don't need to do that, I'll keep it". I said "No, take it to your doctors and show them".'

(Annabelle, 51y, PGT, Negative)

7.2.4 Feelings of Guilt

One of the challenges faced by several participants when sharing their test results was dealing with feelings of guilt, which could impact on how they discussed their testing with other family members. For some, this centred on having received a negative test result when other close family members had received a positive test result.

In Annabelle's case, the guilt she felt at having received a negative test result when her cousins received positives ones led to her making contact with them, even though they were outside of her normal communication patterns. This contact was facilitated though a letter, a method regularly used by participants to contact someone they did not already have a relationship with, which was passed on by her Aunt and Uncle.

'And after [my cousins] had had their tests and they were positive and I had my negative result, I wanted to contact them. I could have phoned them, but because I hadn't spoken to them since they were little, there was no actual contact there between me and them… So I sent them a letter saying how brave I thought they were because they decided they were going to have the double mastectomies and everything. [My Aunt and Uncle] passed the letter on to one of my cousins and she phoned me up and we had… It was really quite nice 'cause we'd never spoken to each other since she was like, that high, and she was really lovely. Absolutely lovely about it, and I think there is still a certain amount of guilt there with me.'

(Annabelle, 51y, PGT, Negative)

Molly had been the one in her family who had initiated contact with the Genetics Centre because of her fears following her mother's early death from breast cancer. Having spoken to the genetic counsellor, she approached her sister, Tonya, who had had breast cancer, to ask her if she would be willing to undergo diagnostic genetic testing first. Both her sisters received a positive *BRCA* result and she herself a negative one. During her interview, Molly became quite upset several times when talking about her feelings of guilt associated with this:

MOLLY: I still feel guilty because my other sisters have got it and I made them do it. And they said to me, yeah but you shouldn't feel like that. It's not your fault if you haven't got it. Which I know makes sense, but at

the time you don't think about sense, do you, or anything like that. And even when… I don't think I felt guilty at the time, in that five minutes or whatever it was when I was in the room and they told me. I don't think I felt guilty. But now I do and when I went up to see Tonya [her sister, BRCA positive]… sorry [BEGINS TO CRY AGAIN]

INT: It's okay, don't worry. Are you okay talking about it?

MOLLY: Yeah.

INT: Okay. Just take your time. Don't worry.

MOLLY: Yeah, so I know I did [feel guilty] when I spoke to my sister and my niece because she'd been told she'd got it. And I was all happy and she came out crying.

INT: Did it ever make you think you would have preferred not to have told them?

MOLLY: No, I don't think so. Because I think… well, they said they were pleased for me, but I will never know if that's true or not, obviously. But I would, I would always have told them.

(Molly, 49y, PGT, Negative)

The different results within Molly's family had an impact on future communications. Molly reported how she tries to steer family conversations away from her negative results.

For others receiving a positive result, guilt was associated with knowing they could have passed the *BRCA1/2* mutation on to future generations; or, that their positive result would reaffirm the guilt felt by their parent at having passed the gene onto them.

FAYE: I think I mostly feel bad for telling – like my Mum. I mean, my Mum just burst into tears. I said 'Oooh, don't be upset, there's nothing anyone can do about it' and I remember talking to Mum's younger sister. I don't talk to her much really, but she 'Oh well, your Mum probably feels guilty because, you know, she's passed it to you'. And I just thought 'What a stupid thing to say' and I said 'Well, at the end of the day what's the point of being guilty?' Because then that would have to go right back to Nan, because she's passed it on to Mum, so there's just no point. And, you know, it's not going to change it. That was decided the moment I was conceived, so you know, what is the point of trying to pass guilt? To me, I don't feel guilty, I don't think. My Mum shouldn't feel guilty, she hasn't done it purposely.

(Faye, 29y, PGT, Positive)

INT: Were you worried about telling anyone?

GINA: My Mum. Really worried about my Mum, because I knew that she would be devastated and, you know, just knowing what she's like, I knew the guilt that she was feeling. I so wanted to phone her up and say 'Mum, I haven't got it' but, you know, it would have been fantastic! But yeah, I think my Mum was the one I was really worried about, just because I knew that she would react very badly, which she did. But that's because of her own – she feels that she was the one that passed it on. It doesn't matter that it's come, you know, from generations back or

where it started, but… I got in the car and my Mum was kind of (my Mum being the devout catholic she is) lit every candle that she had in the house; and I remember phoning her and my Mum never swears, but she did swear that day when I told her.

(Gina, 41y, PGT, Positive)

Both Faye and Gina reported 'feeling bad' and being 'worried' respectively about sharing their positive results with their mothers, who also carried the gene. Yet, they both followed the communication patterns discussed in section 7.2.1; rather than considering the best optimum way by which to share the news, they both made contact immediately as part of their wave one communications because they knew their mothers were waiting anxiously for news. In both cases, this was followed up by an immediate face-to-face visit to calm and reassure their upset mothers.

As well as feelings of guilt, participants often reported that they found the task burdensome or challenging, largely because they felt they were passing on bad news.

'It's quite a burden because you are passing on bad news, aren't you, you know, to a certain extent… You are passing the responsibility on to them, and it's not yours anymore as long as you are the one passing it on. It can be quite a burden at times.'

(Tina, 49y, PGT, Positive)

Eloise, like many participants, felt better sharing her results if she knew that the person she was telling had a partner or other family members who could support them:

> *ELOISE: 'I mean, they've all got somebody. Barry [her brother] with his Jan. You know, that little family, they've got each other they could chat to. Erm, Lyn [her cousin] had me, coz we're like sisters. My step-father had remarried so he was with somebody. Dad is with a partner. Wendy's married. So I felt, you know, if they'd had a problem with it they didn't have to talk to me about it.*
>
> *INT: So you felt everyone was supported?*
>
> *ELOISE: Yeah. And if they had any sort of wobbles going on in the background they didn't show it to me.*
>
> (Eloise, 42y, PGT, Positive)

Eloise also felt she made a point of always looking on the positive side of the experience and making sure she sold it to others in that light, which helped her share her results with her family and friends:

> *'I didn't ring them up in floods of tears. I rang up and said "I'm going to get a new pair of knockers [LAUGHS] and then my periods will stop. Can*

someone still tell me where the downside is?" And I'm still trying to find where the downside is. It's just fantastic.'

(Eloise, 42y, PGT, Positive)

7.3 How those undergoing genetic testing for *BRCA1/2* talk to their friends and family during Stage Three.

As has been discussed, during this stage participants felt a sense of urgency to share their genetic test result with emotionally close relatives who were waiting in a state of anxiety. As a result, not a huge amount of thought was given to how the news was delivered, or when the news was delivered, but rather the importance was that it was shared as soon as possible. This was then followed up with more in-depth discussions about what the results meant and what should happen next.

According to Molly, it was important to share genetic test results "*in order*". She, and many other participants, felt there was a natural pecking order in the family and it was important to make sure they informed people accordingly. For example, it was important to tell those you were close to, people outside the family and siblings should be told in order of their age.

Several participants expressed that they had had concerns about transmitting complex genetic information accurately to their relatives, as it was "*very, very easy to misunderstand*" (Sara).

There was evidence that some participants had taken the time to think about which aspects of their genetics appointments they thought were important and how much of the information they needed to give people so they could understand everything. Annabelle reported trying not to give relatives too much detail: "*I would have just virtually told them 'Look there's 50% chance that you could have this gene as well and I think it would be sensible of you to go and have the genetic testing, but I'm not going to make you'. I would have given them as much information as they wanted: like the implications if they had been tested positive, like they are at a higher risk of getting breast cancer or they could pass the genetic fault on to their children*".

Those asked, found it was important to have lots of information at their disposal when talking about their visit(s) to the genetic service and their genetic testing. A common strategy used was to photocopy the information from the genetic centre, such as letters from the appointments, and give that to family members to read. There was also an emphasis on advising family members to contact the genetic service and talk to a professional as to the best way to proceed.

When asked for tips for talking to families about genetic testing for *BRCA1/2* common words used by study participants included "Honesty", "Openness" and "Respect". Whilst, it may seem obvious, much emphasis was put on the way to communicate with family members by participants. They felt their family histories of cancer, and more significantly seeing loved ones suffer with these diseases, made the whole topic very emotive and, as such, often needed to be approached gently. Many people said that the most important thing was to be as open and as honest as possible. As Brenda described it: ""*I would want to know that the person telling me wouldn't hide anything from me and they would be fairly straightforward in what they said. You know, they would give me facts and information but in a very sympathetic and really kind way*."

7.4 Conclusion

This third findings chapter has reported how participants talked about their genetic testing when they received their *BRCA1/2* test results. Sharing one's test results was driven by a sense of urgency (from close relationships and emotional investment); wanting to give female relatives potential risk information; and male relatives having female offspring who may be at risk.

Participants used strategies such as assigning the task of talking to certain relatives to another family member, and having a positive frame of mind and using positive language to make these communications easier. First wave disclosures were done quickly, usually over the telephone, in order to relieve feelings of anxiety with little thought being given to when or how would be best. This was then followed up by a more personal contact to discuss the implications and offer reassurance and support.

Many of the challenges to family communication during this stage were associated with negative feelings. For example, feeling the burden of having to share 'bad news' or feelings of guilt for having received a negative result when other relatives have received a positive test result; passed (or potentially) passing the mutation on to children; and/or to confirm to a parent that they had passed on the mutated gene to their child. These feelings could impact future communications with family members, censoring conversations so as not to upset other people.

Male relatives were often left out of discussions; if they were included, this was usually as part of wave two communications. There was evidence of a lack of appreciation of the potential risks

to male relatives and their offspring. This was often accompanied by assumptions that male relatives would not be interested information about genetic testing for breast and ovarian cancer, which also hindered family communication on the topic.

The next chapter will look at how participants talked about genetic testing when following up longer term.

Chapter 8 – Stage Four: Following up Longer Term

8.1 Introduction

Receiving the genetic results inspired participants to immediately contact those who were emotionally invested, and who were waiting to hear about the results, and then to keep others, who were aware that the testing was taking place, informed regardless of whether the result was positive, negative or inconclusive. The final stage involved communicating with those who required a specific plan of action on how best to approach the topic (Figure 22). For example:

- Distant relatives with whom contact was infrequent and had not needed to contact until now.
- Those that did not want to know or from whom a negative response had been received.
- Telling children, who were deemed too young to be told at the time of testing.

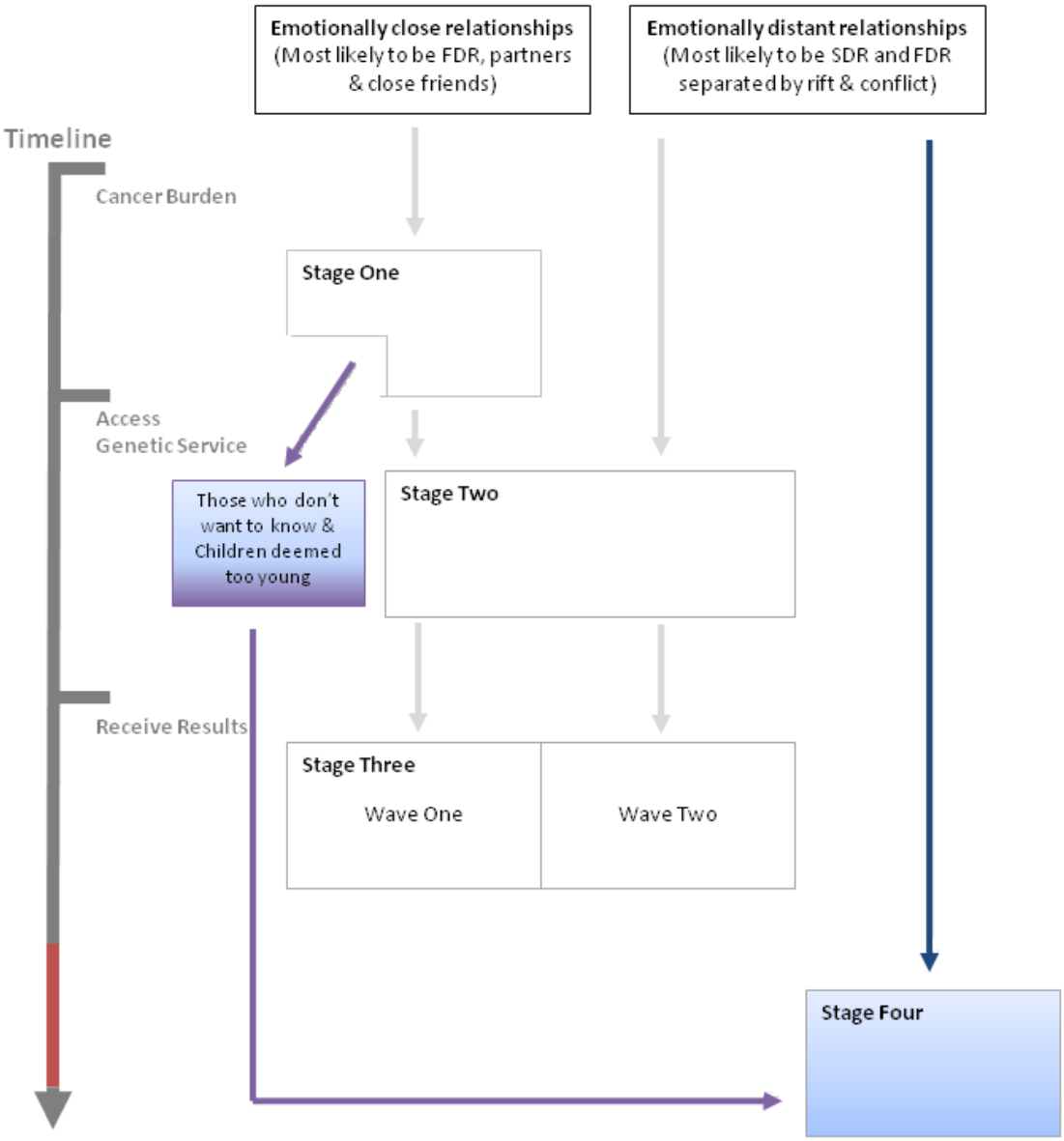

Figure 22: Stage Four: Following up

8.2 Stage Four: Following Up Longer Term

This stage was more commonly associated with receiving a positive result. For those undergoing diagnostic genetic testing, the confirmation that a *BRCA* mutation was present in the family

meant that others may carry it also and participants felt it was important to warn them. A positive result from the predictive genetic testing of further relatives tended to reinforce this, probably because the more relatives that received a positive result somehow made the threat seem more real. However, receiving an inconclusive or negative result did not mean individuals were no longer involved in the family communication. At this stage, the process tended to become a 'family affair', with each member playing their role in ensuring the job got done, regardless of their own risk status. What is more, a negative result from predictive genetic testing often had a similar effect in encouraging the family to tell other relatives about the mutation and/or encouraging them to undergo testing. This may be because participants often held a [misguided] belief that, for example, an individual receiving a negative result would automatically mean their sibling would have an increased risk of receiving a positive result; therefore, it became even more important to the family that that person was tested, especially if they had children who could also be at risk.

> '*Erm, I wasn't that bothered about telling Lianne [her younger sister] because at the time she hadn't had the test done. So I thought, if I haven't got it… so she's got two sisters and one's got the gene and one hasn't got the gene, her chances are still quite good of not having it. That, that… it made sense to me at the time. I think I actually said to her, well you know you're still half and half coz you've got one sister that's got the gene and one sister that's hasn't, so it might have missed you as well. So I thought, well there's a little bit of hope somewhere, you know, in that little part. But she had the test done and she has got the gene.*'
>
> (Molly, 49y, PGT, Negative)

The time span for this stage could be over many weeks, months or potentially years in the future. Generally, the participants felt these were the hardest groups to communicate with as, rather than naturally evolving over time, the information needed to be shared at a discrete event. More thought went into how these people should be contacted: When was the right time? What they should be told? Who should the information come from?

8.2.1 Dealing With More Distant Relatives

In this study, there were three measures of distance:

1. Geographic distance (for example, how far away a relative lived).
2. Genetic distance (for example, differences between first or second-degree relatives).
3. Emotional distance (for example, how close the relationship is between two family members).

No one form necessarily excluded another; for example, modern technology, such as email and telephone, could mean a participant could be emotionally closer to a sister that lived in Australia than she was to a sister who lived five miles down the road. The data suggest that it is, in fact, the emotional closeness of being first-degree relatives, for example living or growing up in the same house, rather than the genetic closeness itself that predicts communication about genetic testing. For instance, being a first-degree relative did not automatically mean genetic information was discussed, as it all depended on the pre-existing relationships. By the same token, more genetically distant relatives were included just as much as first-degree relatives if

there was an emotional closeness at the start of the process. A good example would be where one participant was much closer to her cousins than she was to her brother.

Emotional closeness meant people were included in earlier stage discussions; whereas emotional distance (in other words, the *absence* of emotional closeness or a pre-existing relationship at the start of the process) meant relatives were unlikely to be contacted before stage four, if at all. The exception to this rule would be when one contacts a more distant (emotionally, geographically or genetically) relative for something specific, such as for family history information, or to access predictive genetic testing through diagnostic genetic testing, as seen in stage two. Here, no pre-existing relationship or emotional closeness at the start of the process was necessary. Instead, stage two contact actually created a transitional relationship, meaning that, despite this, test results and risk information was likely to be shared with these family members during stage three rather than stage four.

Therefore, during stage four, communications regarding genetic testing were instigated with relatives who fell into two groups:

1. Genetically close *but* emotionally distant.
2. Genetically *and* emotionally distant.

Many felt it was harder to contact relatives they were not normally in contact with compared to those they had a closer relationship to; as a result, this was frequently left until after they had received their results (stage four). Habitually, they had developed strategies to make this easier,

such as delegating the job to the family member who had the most contact with those relatives, or by photo-copying the letters they had received from the Genetic Service and posting them to them.

> *BRENDA: I always write to Julia [Cousin] at Christmas and she to me, you know, just general things about what people in the family are doing. And that would be an opportunity to let her know (if I felt I should), but Bonnie [Other Cousin] is very good at keeping in touch with her so I leave it to Bonnie.*
>
> *INT: Why is that?*
>
> *BRENDA: Because Bonnie knows Julia best because they're sisters, and she is in close contact, even though she's half way round the world! And I don't myself have instant phone access to Julia. I've got her phone number, but I wouldn't pick up the phone and start talking breast cancer with her! Or genetics, or anything.*
>
> (Brenda, 58y, DGT, Inconclusive)

> '*I was put in charge of telling my Uncle. We've never been that close to him. But I do see him more often than any of the others do. So I just sort of gave him the letter. I mean, all the information was there, all the phone numbers was there. And I did say to him, they are very good. They're very good. If you just want to go and talk to them for a chat, they don't mind. And, erm, so I said, you know if you tell Pat and Jenny, the cousins'.*

(Molly, 49, PGT, Negative)

Having passed the information on to one distant relative, they often relied on the news being filtered down through that branch of the family. There was also likely to be little or no follow-up to see if the information had been passed on or acted upon.

> *'I think it was 12 in my mother's family. She was the eldest, but all the Aunts are dead now (bless them), but there are cousins knocking around up in Northern Ireland. I told my cousin Frank. Now Frank's dealing with that because he's still in full contact with the cousins in Northern Ireland. You know, he goes over there every now and then. It's up to him now, I just need to worry about my brood'.*

(Annie, 65y, PGT, Negative)

The data suggests that contact with more distant relatives was more likely to occur if prompted by a health care professional, such as the genetic counsellor.

> '*I only spoke to my Uncle [to tell him that her mum and sisters had been found to carry a BRCA mutation] really, erm, because when we went for the last meeting with the genetics people they said [that he may be at risk also]... So they said, well yeah you can sort of let him know. 'Course after you've been to see them, they write all the notes down and they write to you to confirm what they told you at the meeting and*

whatever. So I showed that to my uncle, because he was mentioned in it. So he went to his doctors.'

(Molly, 49, PGT, Negative)

In fact, for several participants, even just discussing the topic of more distant relatives during the research interview lead to them identifying family members who may be at risk and who had not been informed. In each case, they reported an intention to follow them up after the interview. This suggests that some kind of follow-up by a health care professional may be advantageous to ensuring more distant relatives who may also be at risk are told.

ZENA: I don't talk to my two brothers very much. They're not so – they just don't figure highly in my day to day life, really; I think that's just distance... Do you know, I don't know whether I actually told them about the genetic test. They all know I've had another mastectomy that I elected to have. Percy [Brother] hasn't had a copy of the letter. I know he hasn't had that. Maybe he should 'cause he has two girls, but then he's not that close to his two girls now. It's difficult isn't it? They all split up and everything. I think you've shown me that there's a gap there that I haven't followed up, to be honest yeah! Yeah!

INT: How do you feel about that?

ZENA: I would hope I would have. I do speak to his ex-wife (well, I send her Christmas cards and what have you). It might be an idea to drop her a line, just to give her a copy of [the letter], so she has the info for the

girls. That's why it's useful to have the letter 'cause it's all set out and it's all clear, it's all in the proper language and everything. And then the ball is taken out of your court and it's given to them; what they do with it is up to them, you know. And I don't think I have passed it on to them. I think there is a bit of a gap. Thank you very much! That needs to be done, actually, yeah.

(Zena, 49y, DGT, Inconclusive)

In some cases, the health care professionals played a more direct role by contacting the relatives on the participant's, or their family's, behalf. For example, an ongoing rift in Gina's family meant that neither she nor her mother felt comfortable contacting one branch of the family; however, they felt there was a moral obligation that they should be informed. So, instead, they gave consent to the Genetic Service to do this.

'*I think my Mum had my Aunt's address and somebody from the genetic department wrote a letter. I think it might even have been a doctor. You know, written a letter just saying that you need to be aware of this; there is this risk that has been identified and you and members of your family should seek having the test... So yeah, I think Mum and I felt a huge responsibility, even though we're not speaking to them and I don't really know where they all are or what they're doing. I just couldn't sleep at night knowing that I'm in possession of this information and they are not. Well, I think most decent human beings would, you know.*'

(Gina, 41y, PGT Positive)

There were several examples of estranged relationships in the dataset. Two participants in particular spoke quite candidly about why the hurt and/or pain caused by the breakdown in the relationship with a first-degree relative meant they were not willing to contact a particular relative.

> *'There's no way I would contact Jonathan [Estranged Brother]. Um – which is a long story… I don't care whether it affects him.'*
>
> (Annie, 65y, PGT, Negative)

Jan, on the other hand, felt that even though her brother and she were estranged, the information was too important not to be shared, even if this meant the health care professionals made contact with him without her consent.

> *'Well, certainly if I had come out positive, and the consultant had said it's important that our family know that, I would just do it. No matter that we are estranged... It's possibly life-sustaining information and I think, as far as the consultant is concerned, that they should have a right to tell the family without my consent because it's like possibly (this sounds dramatic, but it's the only expression I can think of) a "loaded gun". If we have that gene that's our loaded gun, and if I say "No, I'm estranged, I don't talk to anybody, I don't want to tell anybody" I think that's holding important medical information and it should be shared.*

So, I personally think that even without my consent the medical side should be able to go ahead and tell my family. I think that's important.'

(Jan, 44y, DGT, Inconclusive)

Unfortunately, few of the other participants were aware that the Genetic Service team could write to family members who the person being tested was not comfortable contacting for any reason.

8.2.2 Dealing With Relatives Who Did Not Want To Know

Interestingly, when talking about more distant relatives, many participants expressed the sentiment that, once the information about genetic risk in the family had been passed on, it was up to the recipient to act on, that it was an individual choice and that their opinions regarding genetic testing should be respected. However, when discussing members of their immediate family who had opted not to follow up with the genetic counselling, the consensus was that they were 'wrong'. This was especially true if they had children (in particular, daughters) who could potentially be at risk, and so discussions would continue until they could be persuaded. In other words, participants held a theoretical principle of what was ethical and right, but when it came to their own family that rule fell apart.

'*This comes down to the freedom of the individual; I think everyone has a right to decide what's best for them. But I still do feel very strongly*

that he [Brother] should be tested and I am going to go on keeping the pressure on.'

(Arthur, 61y, PGT, Negative)

SARA: Oh, like Sue was positive as well and her daughter doesn't want to know. She doesn't want to find out 'cause she's already got children (which I think is a stupid idea).

INT: What makes you say that?

SARA: Because she's got a child, so she should be tested so that she knows what she's got to do, and if she has got it she can take the precautions to prolong her life. But I think her Mum and my Mum are encouraging her to get tested.

(Sara, 23y, PGT, Negative)

Many participants found it hard to understand why relatives would not want to know.

'*When I told my daughter, Daphne, what I was doing and she was horrified (which is absolutely amazing). I tell you, it doesn't matter how close you are to people, you never know how they are going to react to things. She even phoned my niece and was angry with her for even approaching me about the genetic testing! But anyway, I explained how I felt. I explained that I was really quite pleased to do it for them, my*

own family, and pleased to think that I could maybe help in research generally. So Daphne said "Well, Mum, I don't want to know anything about it whatsoever". So that was that. I was worried about Daphne, but I thought "Well, I'll deal with it again when the opportunity arises. I'll approach it gently when things have simmered down and see what it's all about". But she was adamant the whole way through. Now, of course, immediately a psychiatrist would say it was fear, and I would think it's a deep-down fear really. Because she's a most practical, outgoing, busy kind of person. She's not a fearful sort of person. But I think with cancer and remembering her grandmother and everybody dying – and, of course, in those days the illness was horrific –it is really hard for her. But there's help now.'

(Shirley, 78y, DGT, Inconclusive)

'*My older brother just went into, and still remains in, complete denial. He doesn't acknowledge it at all. And we've had big discussions about it – arguments even. He's older than me so doesn't think it affects him. But you know, his daughter and his son (his son has two children already) are desperate for him to have the test... Ella, his daughter, talks to me about the fact that her father won't have the test, and she's the one that's probably pushing me all the time saying "Please try and get my Dad to do it, will you?" But I got the feeling that she [the Genetic Counsellor] said it was quite common for people to do what Percy is doing. I was surprised. I was really surprised and I still am, because I can't see the reason for it,*

especially as a fella (a man), you know. I think you haven't got a great deal to lose, but you've got a lot to gain for your children.'

(Arthur, 61y, PGT, Negative)

In fact, Arthur expressed concern that perhaps the negative reaction from his brother was as a direct result of how they communicated the issue with him initially.

ARTHUR: I'm not sure – I mean, maybe we mishandled it with him, I don't know the answer to that.

INT: What makes you say that?

ARTHUR: Well, I don't know. I don't know how to tell someone. I've never been trained! I mean, I have not got a clue what to do or how we should have told him.

(Arthur, 61y, PGT, Negative)

Whereas, Annie wondered if her willingness to be tested meant she loved her family more than her siblings, who refused to be tested.

'*But as I say, none of them [her siblings] have done anything about it except myself. But maybe as I said because I worry about my grandsons and my son more than Catherine does about hers or something. But I can't understand why Catherine didn't go and do it, because she could*

have taken the pressure off the whole family - she's got 14 grandchildren, grown up now, you know... I want to say to Catherine 'Well, I really feel that you ought to have done this, you know it's your responsibility as the matriarch of your family', but that would go down like a lead balloon, I can tell you! '

(Annie, 65y, PGT, Negative)

Many participants were particularly concerned about siblings (brothers in the majority of the cases) who refused to engage in communications about genetic testing, thus blocking the information from reaching their children.

'*My brother's reaction, not wanting to know, surprises me because he's got three daughters and I think it's important for him to know whether he could have passed it on to them.*'

(Nicole, 39y, PGT, Negative)

Yet, when these participants were asked if they thought there would ever be a situation where they would consider telling their nieces or nephews themselves, the answer was always no. The argument being that it would be inappropriate to side-step the parents, as this would be likely to cause upset in the family. These were clear examples of family 'rules' about who can tell what to whom, as seen in the systematic review (refer to Chapter Two).

'*Uuuum. I doubt it because I think it's probably – I feel that it's a conversation they [her parents] should have with her [niece]. I would be very open to discuss it with her, but I think probably she should hear it from her parents. That's my own personal view. But I would happily discuss it with her, after she'd been told, but I don't think I would be in a position to tell her... But I don't know, oooh, I don't know. If he came to me and said "Look, could you sit down and tell her?" Then yeah I would. But I wouldn't go over his head, you know, I'd always go through him first.*'

(Gina, 41y, PGT, Positive)

'*No. No. Not in a million years... If Isaac [brother] doesn't listen, I might talk to their step mother, Lucy, because she would listen... You run the risk of upsetting the family and blowing it apart, you can't do that. I wouldn't want someone to go and tell Anne or Claire [her own daughters] something. You have to work on that principle*.'

(Gillian, 49y, PGT, Negative)

However, although Gillian felt it was inappropriate for her to tell her niece about the potential risk in the family, she had a back-up plan involving her daughters if she had not been told by a certain age:

'*But I think by the time she [niece] is 25 [years old], if they've [her parents] not told her, I would get one of her cousins to tell her! But I won't do it. I will get one of the girls to tell her by accident, but not at 19 [years old]. She doesn't need it right now.*'

(Gillian, 49y, PGT, Negative)

Communicating with relatives who did not want to know was acknowledged to be particularly difficult. Various tactics were engaged, such as repeated attempts to engage with them and/or passing on written information, which they could process in their own time. It often became a family affair with many relatives, siblings and parents, in particular, targeting that individual.

'*I think probably try harder to talk to them if they don't want to know. But you have to. I think perhaps a letter is the answer to explain a little bit, to make them a little bit more aware and just sort of talking to them more. But if somebody's absolutely adamant that they're not going to do it, I don't think there's any way you can coax them into it. You just have to hope that the more you explain, and the more you talk to that person, that eventually they will perhaps go and have the test. Just keeping on being a pain! I don't think it's an overnight job, I think it's just picking your times and just talking nicely.*'

(Julia, 58y, DGT, Inconclusive)

8.2.3 Telling Younger Children

One further group that would fit into these stage four communications would be those sharing positive results with children who were deemed too young at the time of testing to be told, but then who needed to be told at a later date (for example, when they are at the age of making life-decisions).

Almost all adult offspring (those aged over 18 years old) were informed about their parent's – check this I am not sure parents' genetic test results and were included in discussions about cancer in the family to some degree, although it is important to note that, when told, sons and daughters did show the same level of interest in the information. Table 7 gives a breakdown of those participants with children under the age of 18 years old, including the children's ages, and whether or not they were informed about their parent's genetic test result. Six of the 24 children, aged 14-17, were told about their mother's genetic testing and were included in communications about cancer in the family, although one of these, Kerry's daughter Katie, was not told intentionally (refer to section 8.2.3). Eighteen children, aged 2-15, were not included in such conversations and were not informed about their mother's genetic testing. There is no difference in the disclosure of test results by gender as, in all cases, except Kerry's, the information was either given to all siblings under the age of 18 years or none.

Participant	Age (years) of sons 17 years old and under	Age (years) of daughters 17 years old and under	Told about parent's genetic test for BRCA1/2
Eloise	15	10	No
Faye	2	10	No
Laura		17	Yes
Karen	14	16	Yes
Gina	3	6, 9	No
Rachel	8		No
Maya	10	12, 13, 16	No
Gillian	10, 11		No
Tina		15, 17	Yes
Nicole	11, 13	8	No
Kerry		12, 15	No, Yes
Total =	**n = 10** **Average age = 9.7 y**	**n =14** **Average age = 12.6 y**	**6 told** **18 not told**

Table 7: A breakdown of those participants with children under the age of 18 years old

For many mothers, telling younger was one of the biggest challenges they faced and one that started early in the process:

> *MAYA: The thing that I found the hardest about all of this was the fact that I might have to then have that conversation with the children, and I think that was always the thing that got everybody. I think that is the one that gets everyone, isn't it? And you can, from a distance (that's probably not the right word) but you can say 'Ok, well they are in their teens now, and what's happened in the last 10 years and what is going to be happening in the next 10 years, and how well breast cancer is doing and what else are they going to find meanwhile'. You can do all that, but when it actually comes to the emotion of actually having to tell*

your children, then you've got an 80% chance of this or they need to have – you know, where do you go with it? And it's just as well we didn't have to, really. But it was quite a… We hadn't thought out how we'd do it.

(Maya, 47y, PGT, Negative)

'*And I remember, when I went to the initial appointment, when we were doing the family tree, and I said then "Oh, I am so concerned about my children" and the nurse was lovely and she did totally put it in perspective and she said "Look, they're children, they're not going to get breast cancer. Medicine moves on, genetics moves on so quickly that by the time they're adults it will be a whole different ball game". And she said "they will be adults and they will have to make their own decisions". Ok, fair enough!*'

(Gina, 41y, DGT, Positive)

For Maya (quote above), and many others, receipt of a negative test result meant they no longer had to have these difficult conversations with their children, which was a great relief to them. However, all the participants with young children (pre-teen,) as well as many with teenage children, who had received a positive result had taken the decision not to tell their children at this time, the reason being that they were too young to emotionally cope with or understand the information. There was an emphasis on childhood being important and that it should be protected from this news. Many also found comfort in the fact that science is constantly changing and, therefore, what they told their children now may not be relevant by the time they

reached adulthood. Several participants reported difficulties in deciding when 'the right time' was. Eloise's comments echo those of many:

> *'Erm, for my kids I think, well, I don't want to have this around them until they're of an age when they can cope with it. And I want them to have their childhood and I want them to have their fun years, and then I don't know when the time will be right. Depends what they want to do with their lives, I mean if they are going to go on into university. Shani doesn't need to worry about it before she is 30, anyway. I would like to tell her… in an ideal situation, if she gets in, I'd love her to go off to university, get herself sorted out with what she wants to do with her life, and then maybe at that point tell her before she meets somebody, so then she is equipped to tell her partner. I don't want her to meet somebody and then to give it to her, coz it's like, oh my god, you're just trying to break-, you know, I don't know what that would do to their relationship. I just think if she knows from the start then it's up to her what she does with it. But I don't want it hanging over her when she should be having fun and enjoying herself, there is plenty of time. And, besides which, when she needs to know, who knows what her options are going to be? And that's why I don't want to, the same with Ben, you know. I'd like to get him to go through, do what he wants to do. They're both bright kids. I hope they go on, but whatever they want to do I don't want their young lives ruined with something that they're not really going to understand anyway, yet. And, as I say, it just changes all the*

time. So what I sit down and tell them today might be completely irrelevant even in five years' time.'

(Eloise, 42y, PGT, Negative)

When asked, many reported they had thought about what they would say when the time comes, albeit in general terms:

INT: Have you had any thoughts about how you will tell Shani and Ben when the time comes?
ELOISE: Erm… I have. I've thought about it a lot. And I just think that there's nothing yet I can do other than be matter of fact. Say 'Now look, do you remember when I had this surgery? Can you understand why?' And then just say 'Well, you both are at a 50% risk of inheriting the same gene'. But then I would like to back it up with some more information… I'd have to sit and say to Shani, 'Look honey, you know, you may want to consider this in the future, what I've had done'. It's not a big deal… I mean… I didn't have any pain. I didn't have anything. I mean, it's uncomfortable. But it wasn't horrendous; I have sunburn and hangovers worse than that. I really have [LAUGHS].

(Eloise, 42y, PGT, Negative)

However, having made that decision not to tell their children at this time, it did not mean it was necessarily an easy secret to keep. Eloise's brother had received a negative result and her sister-in-law had shared the information about the family genetic testing with her own children. For

Eloise, this resulted in new fears that perhaps the cousins would tell her children before she was ready:

> *'She did tell hers why I was having the op. And so I was a bit cross about that. But… I said I don't care, they are fine to know, but if they let on to my children then I will be cross because I want to tell them when I think they're ready to handle it. And I don't want their childhood ruined. Because they won't understand it in an adult way. They are not adults yet. It's bad enough when you are an adult to sort of get your head round things… I just wanted to slap her, frankly. And I am not a violent person, I just wanted to say 'oh for heaven's sake, nobody's been given a death sentence here'. I just pray her two don't tell them!'*
>
> (Eloise, 42y, PGT, Negative)

For Kerry, it was her risk-reducing surgery and her daughter overhearing adult conversations that brought the subject out in the open. She had been discussing her upcoming surgery on the phone with her sister, not realising she was being overheard by her youngest daughter.

> *'I went back up and I looked at her and she just looked at me and I said 'I've got to talk to you, haven't I?' and she just burst out crying.*
>
> *And she thought I had breast cancer; she thought that's why I was going to the hospital all the time, she thought that's why I was having all the mammograms and suddenly I was faced with that 'what do I tell her? What do I say?' and nowhere along the line has anybody talked to me,*

told me how, when, what. All I was told before we had the testing was that having daughters (and a son, obviously, because he can have it as well) they won't even consider screening them or doing anything until they are in their 30s, early 30s and whatever.

But how do you explain it? I don't know. I just – it doesn't – it didn't help me at all, and it is a dilemma that I had to make at that time, and she thought I had breast cancer and I looked at her and I said "Oh Katie no, I don't, I don't" and in the end I told her and her first question to me was "Well, will I get it?" So, you know, how can you tell a 16 year old girl who's just starting in her own life… but I am so worried, so concerned that she's holding all of that and I don't know what to say to her. I don't know who to talk to or just to leave it now and just hope that it will sit in the back of her mind and it won't affect her.'

(Kerry, 45y, PGT, Positive)

In fact, Kerry was very surprised by the lack of discussion and support available to her on the matter. In light of her experience with her daughter, it is understandable why she thought this was an area that would warrant further support:

KERRY: And I don't think [Genetics Counsellor], I don't think anybody helped me at all…You need to – definitely something – some advice on what to say to the children. I know each person is different, but if – I don't know. I don't know. That's my biggest fear all the way along. Not about me dying, or me getting anything, it's about what am I going to tell the girls? I don't want them to go through their teens and their early life; they don't deserve that, they need to have a normal life, like I

did. ...kept asking about the kids. I kept asking 'Well, what do we do? How do we tell the children? What do we say? What sort of effect will it have on their life? Will it be on my records? Will it be' – You know, all those sort of questions that just kept coming up.

No. I naively assumed there would be like (not four of us), but I assumed there'd be like groups where you'd go and talk to people who'd been through it before and they'd be able to tell you, you know, 'This is how I did it'.

INT: Do you think that's something that would be useful?

KERRY: On reflection, yeah. Probably, not straight afterwards, but definitely now, I think as you go further along the chain, definitely.'

(Kerry, 45y, PGT, Positive)

8.2.4 Identifying Who Should Be Told

The findings show that those undergoing genetic testing for *BRCA1/2* are not good at identifying who they should include in discussions in order to pass on relevant information to at risk relatives. Instead, communication is based around normal patterns of communication with emotionally close relatives and a need for information from more distant relatives. As a result, male relatives and more distant relatives are often excluded.

For example, Figure 23 shows a family tree for Jane, as it would be drawn in a genetic counselling appointment. It maps out all the family members who may be affected by Jane's

positive *BRCA1* result. This is the tool a genetic counsellor would use to identify who, in Jane's family, is at risk and should ideally be informed about genetic counselling and testing.

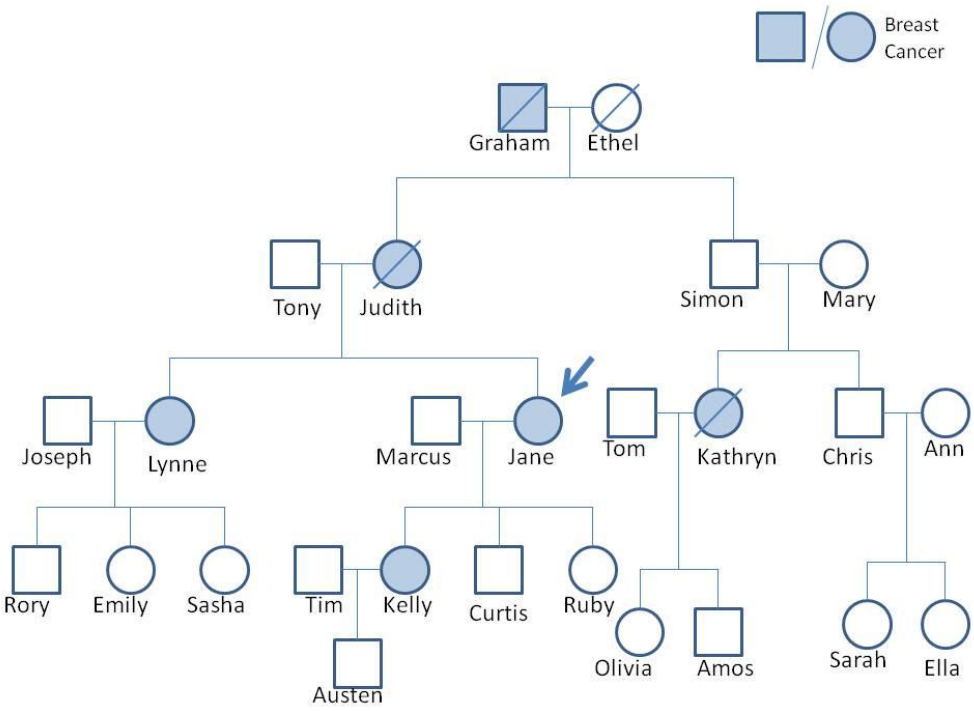

Figure 23: Jane's family tree for a BRCA1 family as drawn in a medical genetics clinic

However, Figure 24 shows Jane's communication eco-map in which Jane reports having close emotional relationships and good communication (three green lines) with many family members. The orange arrows show the flow of communication about Jane's *BRCA* test result. This shows the reality of who is actually included in discussions.

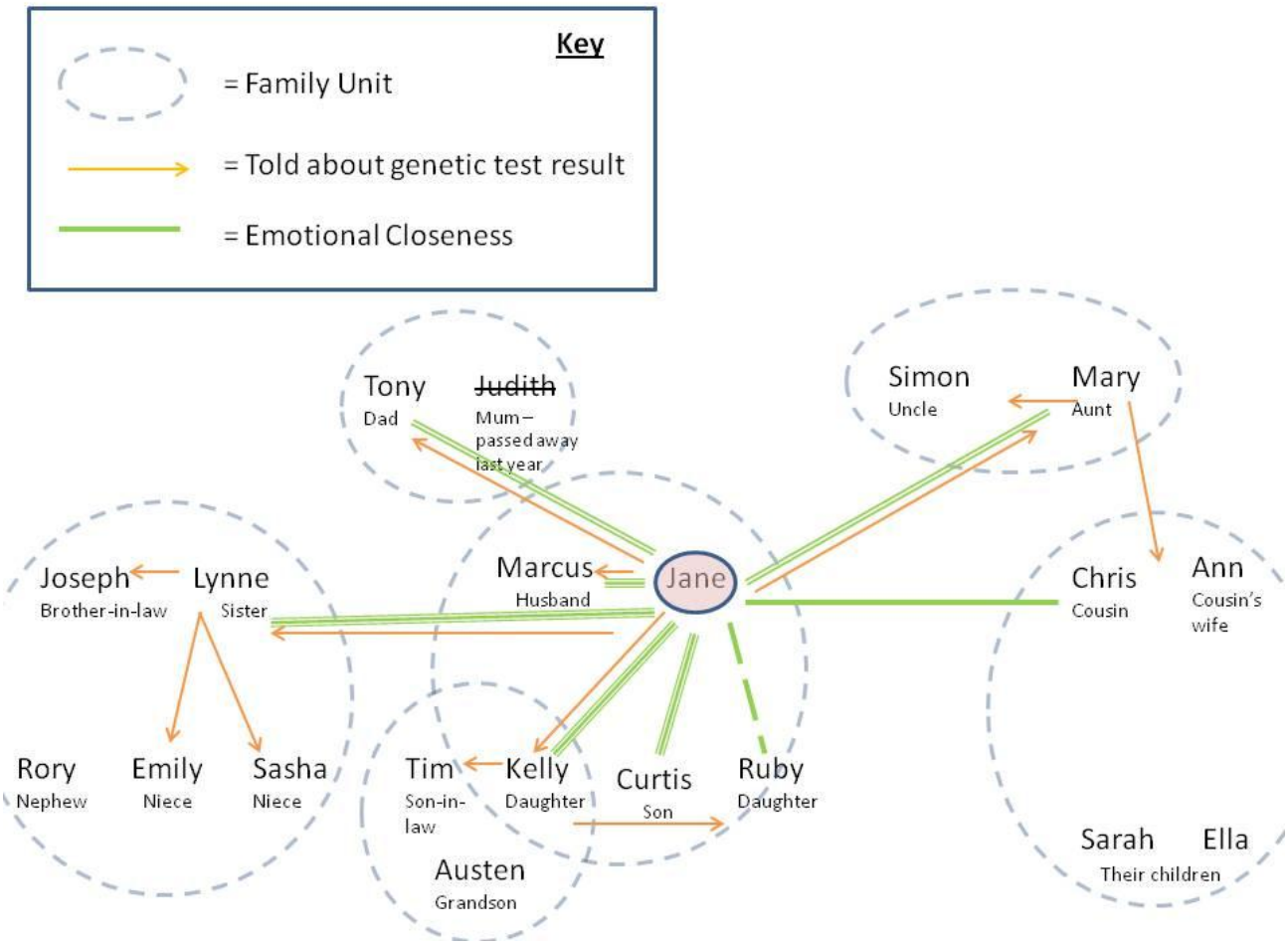

Figure 24: Jane's communication eco-map

Much is revealed when the communication eco-map is laid over the family tree, as seen in Figure 25. All those highlighted in red are relatives that the information about the genetic testing was not shared with. The important thing that this study shows us is that the relatives being missed out are not random, but are clear patterns. For example, Jane communicates readily with those she is emotionally close to; those that are missed are men and relatives who she is not emotionally close to, namely her cousins.

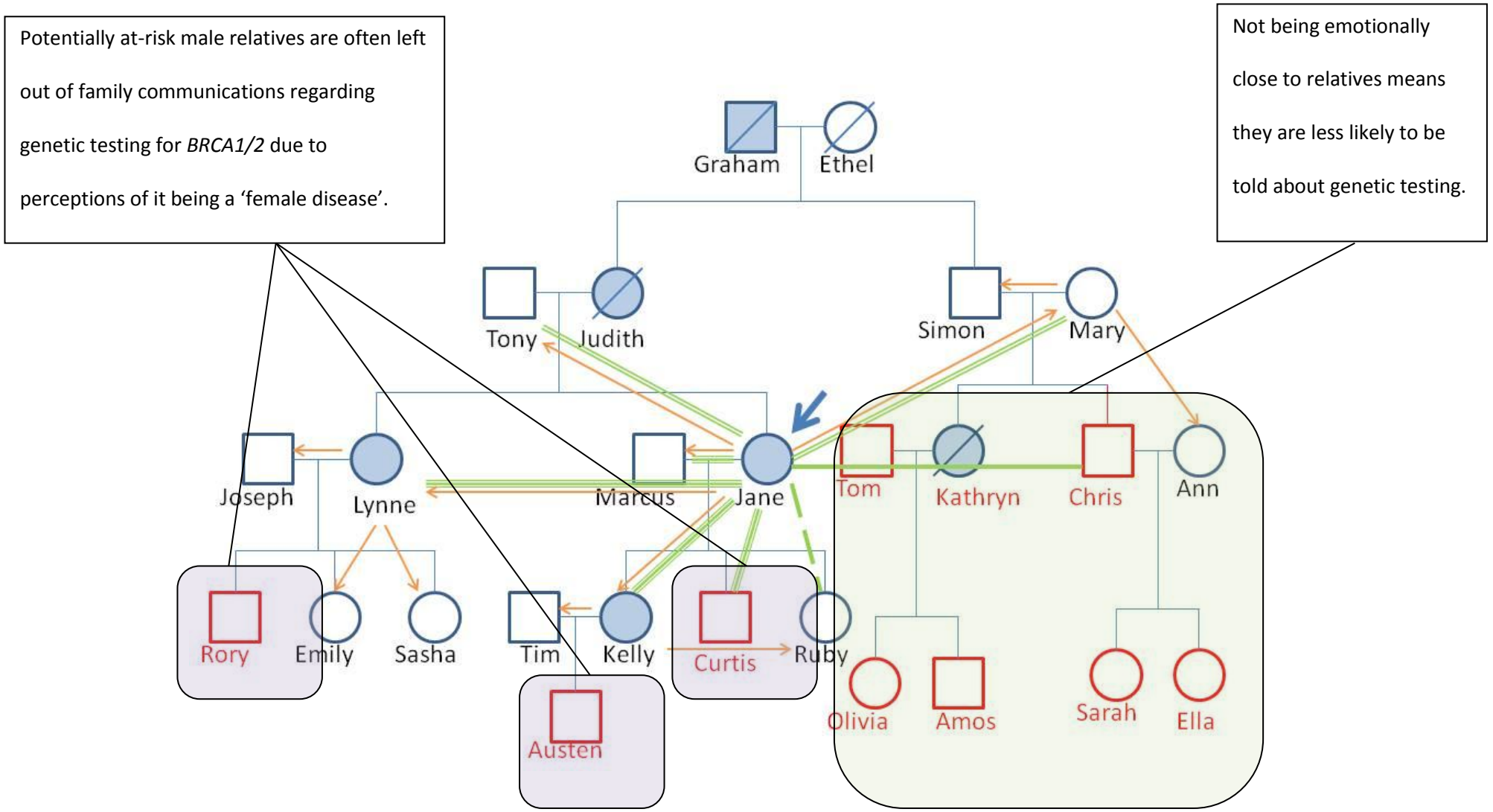
Potentially at-risk male relatives are often left out of family communications regarding genetic testing for *BRCA1/2* due to perceptions of it being a 'female disease'.
Not being emotionally close to relatives means they are less likely to be told about genetic testing.
Graham
Ethel
Tony
Judith
Simon
Mary
Joseph
Lynne
Marcus
Jane
Tom
Kathryn
Chris
Ann
Rory
Emily
Sasha
Tim
Kelly
Curtis
Ruby
Olivia
Amos
Sarah
Ella
Austen

Figure 25: Jane's communication eco-map laid over her family tree identifies those at-risk relatives who were not engaged in family communication regarding genetic testing.

It is clear that one of the ways to improve family communication regarding genetic testing for *BRCA1/2* is to help those undergoing genetic testing to use their family tree rather than their normal communication patterns (shown on the eco-map) when identifying who, in the family, the information is relevant to.

8.3 How those undergoing genetic testing for *BRCA1/2* talk to their friends and family during Stage Four.

It is harder to give specific examples of how those undergoing genetic testing for *BRCA1/2* talk to their friends and family during this final stage because for many these were conversations planned for the future. For example, how they would share their results with their children when they were older.

However, this stage also involved trying to pass information on to relatives that are more distant and with those whom had not wanted to be involved at the earlier stages. One common technique used to engage such relatives was for participants to use their own stories, in other words using their own personal experiences. They felt their lived examples with actual people providing expert testimony and made the information more accessible for relatives. For example, Eloise made an effort to speak very positively but honestly and openly about her

genetic testing with her relatives. She told them how, from the age of 11, her life had been affected by cancer; how losing her mum at such a young age had a deep impact on her and then to have her sister diagnosed with the same disease at a young age was very distressing. She told them participating in genetic counselling and having a genetic test for *BRCA1/2* gave her some understanding about her family history and closure about her mum's death; and allowed her to take matters into her own hands and manage her increased risk of cancer. Eloise reported how by telling her story in this way she put her decision to undergo genetic testing in perspective others could understand and relate to. In her own words, "*it's a real human experience rather than some abstract science!*". As a result of communicating in this way, she found relatives were very receptive to her discussions.

8.4 Conclusion

At this stage, communication became more of a family affair, with each family member playing their role to ensure at-risk relatives were aware of the situation; this made the process simpler for some participants. Techniques such as delegating the task of contacting more distant relatives to one family member and photocopying letters from the Genetic Service to send also made family communication easier. Other facilitators included: being prompted to engage with family communication regarding genetic testing by a health care professional, or the Genetic Service making direct contact with those relatives that the individuals or the family did not feel they could contact (however, not being aware of this option was also a potential barrier). For some, feeling that they had a moral obligation to inform potentially at-risk relatives fostered family communication, despite a family rift.

Barriers to family communication seen at this stage included: not feeling they could make contact with certain relatives, either due to emotional distance or family rift and estrangement; reliance of information filtering down through more distant branches of the family, with little or no follow-up; family 'rules' about who was allowed tell what to whom; and children being too young at the time of testing, but then having to make decisions about when and how to share information with them later. Participants were particularly concerned by how to approach or engage those family members who had previously said they were not interested, especially if participants were worried that that person's offspring may also be at risk and access to risk information for them was being blocked. In some cases, there was a perceived lack of discussion and support from the Genetic Service; and many were not made aware of what support and guidance the Genetic Service could have given (for example, directly contacting certain relatives on their behalf).

Future interventions could be developed to help families identify emotionally, geographically and genetically distant relatives who may potentially be at risk and encourage them to consider how they may be informed about that risk. There may also be merit in the Genetic Service offering a later 'follow up' appointment with those receiving a positive test result (and their family, if acceptable) to review who has been told and offer support and guidance with the task (and/or offering more direct involvement). There should also be regular long-term contact with mutation carriers with children.

Chapters Five to Eight have presented the findings of an interpretive descriptive, qualitative study that explored individuals' experiences of talking to relatives regarding genetic testing for *BRCA1/2*. The findings have been reported as a longitudinal view, providing an explanation of

the process of family communication with a focus on when, how and why such communications occur. The key barriers and facilitators to family communication regarding genetic testing for *BRCA1/2* have been highlighted for each stage and potential areas of support identified. The final chapter will provide a discussion and conclusions to this research.

Chapter 9 – Discussion & Conclusion

9.1 Introduction

This chapter will present a discussion around the six Key Findings and examine the contribution this study makes to the existing literature. The chapter will also provide a critical review of the strengths and weaknesses of the work. It concludes by looking at the implications of the work and suggestions for future research that is needed.

9.2 Summary of Study

Research to date has focused far more on with whom and why (motivations) family communication regarding genetic testing occurs, rather than when or how it is occurring (refer to Chapters One and Two). What is more, the research tends to focus on communication with specific family members at the point of result disclosure only. Therefore, the work presented in this thesis set out to address the following research question:

How and when do those undergoing genetic testing for BRCA1/2 talk to their relatives about a family history of cancer, associated risks and genetic testing?

The aim of the work was to gain insight into participants' experiences of discussing their participation in genetic testing, their test results and potential risk information following genetic testing for *BRCA1/2* with their family (not just with first-degree relatives or specific family members), with particular focus on how these families discuss genetic testing for cancer risk and when.

This work is a top research priority, because, how those undergoing genetic testing for *BRCA1/2* talk to their relatives about their family history of cancer, associated risks and genetic testing can be critical in ensuring all family members have access to genetic services. The existing literature shows that talking about such things can be potentially difficult for concerned individuals and their relatives and, despite a wide range of literature, it is clear that the nature of interactions regarding genetic information remains poorly understood (Gaff *et al.* 2007).

The resultant study, as described in Chapters Three and Four, was qualitative in nature, employing in-depth interviews as the method for data collection and utilising the technique of constructing eco-maps (Ray and Street 2005) as a method of identifying relevant family members and guiding the researcher through the family structure and relationships. The work was grounded in a conceptual model adapted from Peterson's (2005) family systems model, for conducting family-based research in hereditary risk and genetic testing; and Carter and McGoldrick's (1989) 'Family Life Cycle', which allows the study of how individuals within a family and, by association, the family as a whole, adapt to chronic illness. These methods were chosen in line with the interpretive description methodology (Thorne *et al.*, 1997;Thorne *et al.*, 2004b) to ensure depth and richness in analysis and reporting of findings.

There has been a call for research that acknowledges family communication regarding genetic testing as a process rather than a discrete event (Forrest *et al.*, 2003;Hallowell *et al.*, 2005a;Gaff

et al., 2007a). This has been achieved in this study by analysing the longitudinal view of family communication; examining when family communication regarding genetic testing occurs, throughout the whole process of genetic counselling and genetic testing and not just disclosure of test results. The longitudinal view presented in Chapters Five to Eight gives a deep insight into how and when certain family members were included in family communications regarding genetic testing for *BRCA1/2*.

Whilst family communication regarding genetic testing can be intellectually and emotionally challenging (Sermijn *et al.*, 2004), for the participants in this study it was seen as a positive thing; spreading the word meant more awareness by family and friends, leading to increased detection and, thereby, less disease. However, there were a series of issues that arose for these participants, which will be discussed within this chapter.

9.3 Discussion

In order to discuss the findings of the study, it is important to explore them in relation to both the existing literature, as examined in Chapters One and Two, and the broader literature which exists beyond genetic testing for *BRCA1/2*. Therefore, this discussion section will identify the six Key Findings of the study and position them within the current evidence. Whilst some of the findings from the data reflect the existing literature, there are a number of new findings, which both challenge and add to the existing body of knowledge.

The Key Findings are as follows:

1. Communication regarding genetic testing from *BRCA1*/2 between emotionally close relatives is different to communication with emotionally distant relatives; whilst family communication regarding genetic testing for *BRCA1/2* with emotionally close family and friends is about sharing and supporting; communication with emotionally distant family is about gaining and imparting information
2. A family's engagement in communication regarding genetic testing is implicitly linked to their experiences of cancer burden, and how openly this topic is discussed in the family.
3. There is a lack of understanding of risks to men and their offspring based on perceptions of hereditary breast and ovarian cancer being a female disease.
4. Emotionally distant and male relatives are only contacted selectively. Those undergoing genetic testing for *BRCA1/2* are not good at identifying all at-risk family members in order to share the implications of the genetic test with them.
5. As far as the family are concerned, members do not have the right to make an informed decision to decline.
6. Plans for telling people in the future, especially children, is a cause of worry and concern for those undergoing testing and needs further support, especially in the longer term.

Key Finding One: Communication regarding genetic testing from *BRCA1*/2 between emotionally close relatives is different to communication with emotionally distant relatives.

The literature reviewed in Chapters One and Two has already demonstrated that people are more likely to share their *BRCA1/2* genetic test results with first-degree relatives (FDRs) (Koehly *et al.* 2003; Claes *et al.* 2003; Wagner *et al.* 2003; McGivern *et al.* 2004; Finlay *et al.* 2008; Blandy *et al.* 2003; Patenaude *et al.* 2006) and those they have close emotional relationships with (Chivers Seymour *et al.* 2010; Julian-Reynier *et al.* 2000; Hughes *et al.* 2002; Claes *et al.* 2003;

Peterson *et al.* 2003). However, the findings of this doctoral study go beyond telling us that participants are simply more likely to communicate with these groups by showing *how* that communication differs.

In order to get a sense of the organisational and structural characteristics, and to identify the normal patterns of communication between the participant and family members, the family functioning construct of cohesion, or 'closeness', was measured. According to Koehly *et al.* (2003), cohesive relationships are supportive relationships that involve those whom the participant feels close to, and are characterised by behaviours such as support-seeking during a crisis and/or minor everyday upset, or the sharing of confidences. Therefore, as part of their eco-map construction, participants were asked to score how close they perceived their relationship to be and how open they felt the communication was with each person they had identified. The findings reveal that participants were more likely to involve those they scored three or two (cohesive relationships) in discussion about their genetic testing and cancer risk. This supports previous research; however, further analysis of qualitative data revealed much more.

Emotionally close friends and family

In this sample, family communication regarding genetic testing for *BRCA1/2* occurs with emotionally close friends and family through normal communication patterns and evolves over time. Koehley *et al.* (2003) suggest that family functioning may play a more important role than mutation status or type of testing in determining when these discussions occur. For example, patterns of communication associated with discussions about genetic counselling and testing may be characterised by support-seeking and advice-seeking relationships (Koehly *et al.*, 2003) .

This is supported in the findings of this doctoral study. For these participants, patterns of communications started, long before genetic testing, with conversations about cancer and what

it means to, and for, family members and was stimulated by their shared experiences of cancer within the family. These were the kind of conversations that happened around the dinner table and during evenings together, and were prompted by diagnosis, treatment and death in the family. Cancer played a huge role in the lives of the participants, both because of its personal impact and its impact on family relationships and functioning. Cancer has already been shown to shape families' norms and expectations about the family life cycle. For example, Peterson *et al. (2005)* found that, as an individual approached the age when cancer was diagnosed in previous generations, their own sense of vulnerability may increase which, in turn, may often lead to them seeking support and advice from others.

As one, or more, family member(s) entered into genetic counselling, these conversations continued, but the focus shifted to sharing the experience of, and knowledge gained, during genetic appointments. At this stage, the person being tested did not seek out family members the results may be relevant to and inform them of potential risk information, but rather shared their experience with those who offered advice and/or support as part of their pre-existing relationship. This further supports the thinking that family functioning dictates who is included; for example, Kenen *et al.* (2004a) found that women may be less likely to talk to their brothers or spouses about hereditary breast and ovarian cancer risk, possibly because women expect limited support from these male relatives in terms of coping with cancer risk information.

Participants in the present study communicated the genetic information though sharing leaflets and consultation summary letters. Whilst they may have been disappointed or surprised that someone did not want to know or disagreed with the activity, they did not force their involvement but simply found others to talk to about it.

Inclusion during these stages meant these family members knew about the genetic testing and became emotionally invested. According to the study participants, with that investment came a level of anxiety regarding the outcome of the genetic test. Knowing that others were waiting to hear in a state of anxiety prompted rapid disclosure of results, regardless of what that result may be, with little thought as to how, when or where it was shared. For example, this happened from the hospital car-park or on the way home from the genetic service, usually by telephone. There was no discussion or explanation about what the result meant, what had happened before and continues after; communication at that moment was about a dichotomised 'I carry the gene' or 'I don't'. Other studies, such as Gaff *et al.* (2005), have described how participants reported sharing their actual result with close relatives as being straightforward, because it was just part of an on-going process that was regularly discussed. However, it is important to note that many participants in the present study later expressed regret at sharing their results quickly with close family members in this manner because they were worried they had caused distress and/or were not able to offer the right amount of support or information. This may be an area where genetic counsellors, if they are not already, should build time into their consultations to encourage patients to consider how they may share their results and how they will feel about it afterwards.

Generally, having an answer to a genetic test did not end the conversations for the study participants regardless of their result. A positive result gave some insight into the family history, but brought the risk to others into sharper focus. Support and advice-seeking discussions became about risk-reducing strategies and informing others about the mutation within the family. Those who had not wanted to be involved before now became the focus and conflicts may arise as other family members feel not being tested is no longer an option. As Peterson (2005) also found, families may influence testing decisions through support or coercion, and may

also affect decisions indirectly by influencing attitudes related to testing outcomes (refer to Key Finding five).

An inconclusive diagnostic genetic testing (DGT) result left a level of uncertainty. Dorval *et al. (2005)* found no evidence that women receiving an inconclusive *BRCA1/2* genetic test result felt falsely reassured, compared to those receiving a true negative result from predictive genetic testing. The findings from this present study suggest that this may be because the family still share their significant experiences of cancer within the family, but were now without the means to explain it; therefore, conversations may return back to a pre-genetic testing status of wondering where the cancer in the family comes from and how it will impact future generations. This may be a cause for concern, as little is known about the psychosocial consequences of receiving an inconclusive *BRCA1/2* genetic test result (Dorval *et al.*, 2005).

A negative predictive genetic testing (PGT) result may have brought relief to the individual receiving it and mean their offspring are not at risk, but that good news sits within a family where others are receiving positive results and where cancer still dominated the conversations. Other researchers have reported non-carriers describing themselves as feeling guilty, often termed 'survivor's guilt', that they had been found not to carry a *BRCA1/2* mutation when their relatives had been identified as carriers (Dorval *et al.*, 2000;Ardern-Jones *et al.*, 2010;Hallowell *et al.*, 2006).The present study suggests this can create tension and feelings of guilt, which may impact future communications . In the most extreme case, this may mean that those receiving a negative result, whilst others in the family receive positive results, may feel they no longer 'belong' and begin to distance themselves; meaning they are no longer giving or being given emotional support.

The findings of this study demonstrate that family communication regarding genetic testing for *BRCA1/2* with emotionally close friends and family is an on-going process, starting with, returning to and always situated within the shared experience of cancer within the family. This is depicted in Figure 25.

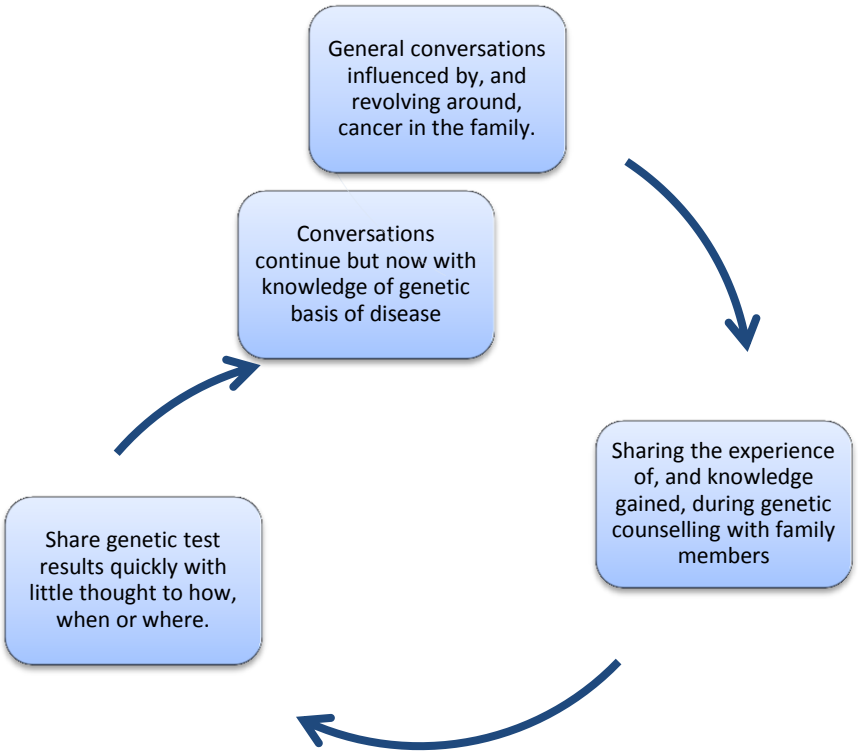

Figure 26: Family communication regarding genetic testing for BRCA1/2 with emotionally close friends and family is an on-going process; starting with, returning to and always situated within the shared experience of cancer within the family.

Of course, emotional closeness does not automatically mean completely open communication with all family members. For some, emotional closeness could create as much of a barrier as emotional distance. For example, some participants found it hard to talk to particular relatives,

despite emotional closeness, in case they upset them or caused them distress meaning it was hard to discuss their genetic testing for *BRCA1/2*.

As one, or more, family members(s) go through genetic counselling and genetic testing, they share their experiences, and the family's knowledge and understanding increases and shifts the conversations. However, having the results of the genetic test may change who is affected: those receiving a positive result may feel the burden falls more on them, while those receiving a negative or inconclusive result may experience feelings of guilt, or that they are no longer entitled to have a say. Actually, the familial nature of the conversations continues because it is rooted within health-related cognitions and beliefs shared within a family that are shaped by the collective experiences and the traditions of its members (Peterson, 2005).

The exceptions to this pattern of communication are male relatives, especially sons, and young children who, despite participants reporting being emotionally close to, were often not included in discussion (see Key Findings Three and Six).

Emotionally distant family

Many studies have reported that communication about genetic testing and inherited cancer risk is less likely to occur in relationships that were both relationally and emotionally more distant (Claes *et al.*, 2003;McGivern *et al.*, 2004;Peterson *et al.*, 2003). Whilst the findings of the present study support that, for example, participants were less likely to involve those they consider themselves less emotionally close to (scoring one or zero on their eco-maps), the findings go beyond that, by demonstrating how and why communication with more distant relatives differ. In summary, whilst family communication regarding genetic testing for *BRCA1/2* with

emotionally close family and friends is about sharing and supporting, communication with emotionally distant family is about gaining and imparting information.

As family members became involved with genetic counselling, they needed to gain information, for example about family history, in some cases asking more distant relatives who had already had cancer if they would be willing to have a diagnostic genetic test. Such requests required them to seek outside of their normal communication patterns, or to have conversations with those who may have preferred not to discuss such a topic with. Many felt it was harder to contact relatives they were not normally in contact with, compared to those they had a closer relationship to. This required a plan, not only for how the contact would be made, and by whom, but also what would be said. Letters were commonly used, as they were reported as being less intrusive and gave the opportunity to plan and structure the message. The family member with the most contact, even if it was just a yearly Christmas card, or the most outgoing personality, was often nominated to make the approach. Participants reported the most success when they were honest and open about their needs.

Sobel and Cowan (2000) describe how genetic testing for Huntington's disease can change notions of family membership. Participants in the present study reported similar experiences. Providing information, or agreeing to have a diagnostic genetic test, brought family members into the loop and created a transitional relationship whereby they, too, were invested in the genetic counselling/testing process. During this time, participants adjusted, albeit temporarily, who they considered to be family, so definitions of family membership and/or family roles and leadership shifted depending on who became involved.

When the genetic test results came in, it was important to share them with these people, but not with the same sense of urgency as close friends and family. It was done at the next point of

contact or by sending copies of letters from the genetic service. Once the test result had been shared, the transitional relationship normally ended and contact returned to its previous status before genetic counselling. Periodically, more substantial relationships and friendships would arise, but this was not common.

Those who had received a positive *BRCA1/2* result may then inform other more distant relatives, such as cousins or aunts, especially if prompted to do so by a health care professional such as the genetic counsellor. Other studies have shown that the endorsement of the health care professional can be an important stimulus to talking to relatives (Mesters *et al.*, 2005;Chivers Seymour *et al.*, 2010), and health professionals may be relied upon as a source of technical information or to legitimise the word of the informant (Mesters *et al.*, 2005;Forrest *et al.*, 2003). Participants in the present study often felt obliged to make contact, usually with the closest family member, impart the information, and then rely on them to pass the information on to *their* side of the family; what they chose to do with the information was then their business. This task could be delegated to another family member, either the matriarch or the person with the closest relationship; strategies such as sending photocopies of letters from the genetic service were used to ensure accurate transmission of information. However, unlike nuclear families (McGivern *et al.*, 2004;Wilson *et al.*, 2004), this is not done in a systematic way, which often means some potentially at-risk distant relatives are just never contacted. If potentially at-risk distant relatives are not told they may not have access to genetics services and subsequently the ability to make informed decisions about their own health (see Key Finding Four).

Key Finding Two: A family's engagement in communication regarding genetic testing is implicitly linked to their experiences of cancer burden, and how openly cancer is discussed in the family.

Previous studies have examined how beliefs about a family's risk of an inherited condition, gained either through personal experience or as a result of genetic testing and often shared across families, may influence health-related decisions and other outcomes (Peterson, 2005). One of the key findings of this present study was how a family's engagement in communication regarding genetic testing was implicitly linked to their experiences of cancer burden and how openly this is discussed in the family. Foster *et al.* (2002) describe 'Cancer burden' as the experiences of having cancer in the family. It relates to the nature of the family history and the emotional burden of witnessing relatives with cancer undergoing treatment or dying. Foster *et al.* (2002) suggest that experiences of cancer in the family play an important role in formulating beliefs about one's own risk and motivation for predictive genetic testing. This study takes that concept further and proposes that these experiences and, importantly, how openly they are discussed, also play an important role in how family members talk about genetic testing and cancer risk information.

To be eligible for diagnostic genetic testing for *BRCA1/2*, a person must have been diagnosed with an associated cancer, for example breast, ovarian or prostate cancer. There must also be a relevant family history that alerts the genetic practitioner to the fact that there might be a mutation within one of the known BRCA genes. As *BRCA1/2* mutations are inherited in an auto-somal dominant pattern, one would expect to see multiple cancers across several generations in the family history. Once a *BRCA1/2* mutation is identified in this family member, then the testing can be extended to other family members who could potentially be at risk in the form of predictive genetic testing (Lerman and Shields, 2004). Therefore, by definition, the participants

in this study have had some experience of cancer, either because they themselves have had cancer or because relatives have had cancer.

For so many of the study participants, cancer had a huge emotional and psychosocial impact. It was an omnipresent feature of their lives and so was part of their normal conversation. The high emotional burden of being a member of a family where hereditary breast and ovarian cancer is present, especially for female, has been discussed in the literature. Foster *et al. (2002)* attribute this to their familial experiences of cancer, high bereavement rates and their own fears of developing the disease.

Crotser and Dickerson (2010) found that white, young and middle-aged women, from New York, who had received news of a family *BRCA1/2* mutation from a biological relative, felt family risk communication began early in their life, before the technology of genetic testing was even available. They lived with the expectation that they would share the fate of their mothers and grandmothers before them, and recalled their mothers ingraining a sense of risk and need for vigilance from an early age so cancer was caught early (Crotser and Dickerson, 2010). These findings are also similar to those of Hamilton and Bowers (2007), whose theory of genetic vulnerability places 'experiencing the family disease' as a key concept in the experience of adult testing.

For the participants of this present study, their experiences of and attitudes towards cancer and how comfortable they felt discussing the family history with certain family members mirrored their attitudes towards family communication regarding genetic testing. For relatives with whom participants felt cancer in the family was a regular, open topic of conversation, talking about genetic testing and its implications was much easier, mainly because it was about sharing their experience of genetic counselling and testing (see Key Finding one). However, for relatives with

whom it was difficult to discuss or acknowledge the family history of cancer, it was equally hard to engage them in discussions about genetic counselling and testing. It is important to acknowledge that experiencing difficulties discussing these topics was not restricted to emotionally-distant relatives or those participants were not in regular contact with. In some cases, the close nature of the relationship itself created barriers, for example not wanting to cause relatives harm or distress, meant cancer in the family was not an open topic of conversation.

Kenen *et al.* (2004b) note that families often follow family scripts, which guide their interactions and communication about a range of topics, and a family history of cancer may be such a topic. Other studies have found that risk communication was more straightforward when the family history of cancer had been a regular topic of family communication (Gaff *et al.*, 2005;Crotser and Dickerson, 2010). Another way to express this would be to say that the degree of engagement in open dialogue about cancer in the family, and what it might mean, influences the approaches and reactions to family communication regarding genetic testing for *BRCA1/2*.

This relates to McAllister's (2002) Theory of Engagement (TE), which suggests that a process of engaging with cancer risk occurs in *HNPCC* family members over time as they interpret their family history and thus become cognitively and emotionally involved with their risk of cancer. Also, the degree of engagement that has occurred at the time of genetic testing influences the approaches and reactions to test results.

The theory of engagement (TE) is constructed around the core category of 'engagement' and the associated 'engaging' with cancer risk. McAllister (2002) defines engagement as a constructed concept reflecting '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' (p. 496). Figure 27 depicts

engagement and action in relation to *HNPCC* risk[29]**.** Engagement is a 'dynamic process that occurs initially at the cognitive level (partial engagement). With the passage of time and events, engagement may progress at the affective level leading to intense engagement (and sometimes disengagement)' (McAllister, 2002). As a theoretical construct, 'engagement' may have explanatory power with regards to variations in attitude towards one's risk status, and the degree of engagement may vary over time and in relation to cancer-related events in family life (McAllister 2002). It may involve dealing with painful memories and so may be avoided or take place slowly over one's lifetime. The study identifies a series of psychological factors that influence the process of engagement with *HNPCC* risk, including:

1. Causal conditions: such as personal experience of the family history of cancer and family communication on the topic.
2. Intervening conditions that block the process: such as ignoring the family history, other life stress and experiences of sporadic cancer.
3. Individual psychological factors: theories of how cancers are inherited through the family and individual coping strategies.

One limitation of McAllister's work is that TE emerged from a grounded theory study with a sample size of only 12 participants and, therefore, it is difficult to generalise to other populations. However, the phenomena identified as potentially influencing attitudes towards predictive genetic testing for *HNPCC* are well supported in the current literature; for example, family experiences of the disease, higher perceived risk, and lay beliefs of how the disease is inherited (Richards, 1996;Dudok de Wit, 1997;Geller *et al.*, 1999;Rolland, 1994). More research is needed to allow further development and confirmation of its value.

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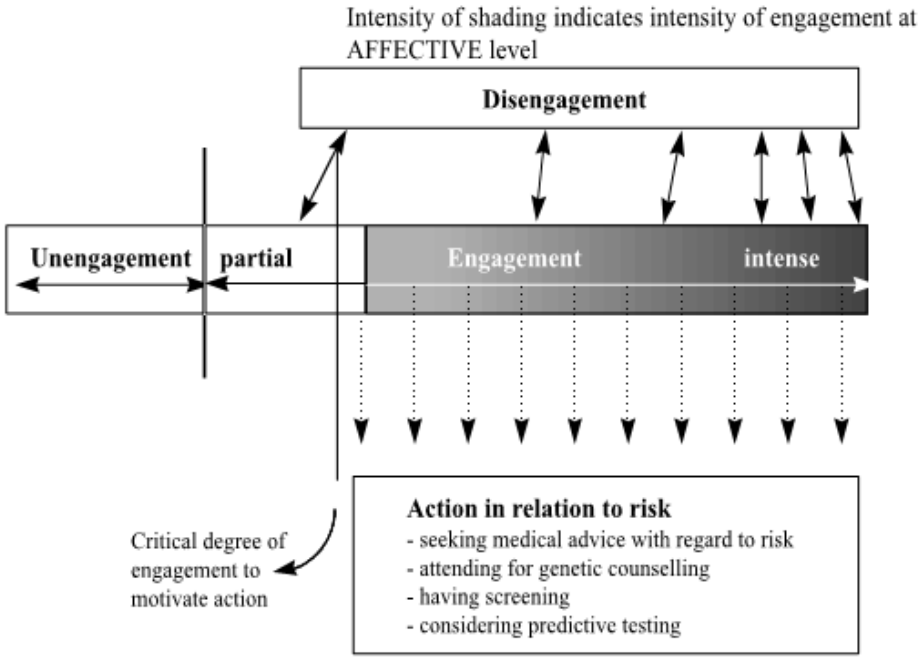

Figure 27: Engagement and action in relation to cancer risk (McAllister 1999; McAllister 2002).

McAllister proposes that the degree of engagement with cancer risk at the start of genetic testing is consistently associated with approaches and responses to predictive genetic testing for *HNPCC*. For example, those who receive a mutation positive test result, and who have been only partially engaged with the cancer risk, will have poor adjustment one to two weeks post-test; whereas, those who receive a positive result, and who have been intensely engaged, will have good adjustment.

Similarly, this present study suggests the degree of engagement at the time of genetic testing may influence family communication on the topic; and that the degree of engagement may be measured by how openly cancer is discussed within the family. All the participants except one in this study reported positive experiences of genetic testing (perhaps this is not surprising, given

that they each self-selected themselves to be involved in a study exploring their experiences). From this, although no specific data were collected, it does not seem unreasonable to assume that this group of individuals were generally intensely engaged with the process of genetic testing. They also reported their overall experiences of talking to their families and friends about their genetic testing and risk information as being positive. There were individual examples of challenges, such as certain relatives not wanting to be involved or worries related to telling their younger children in the future, and wider issues of lack of understanding about potential risk for male relatives and who in the wider family was a risk. Essentially, however, the research participants in this group cannot be described as having struggled with family communication regarding genetic testing.

Therefore, these data may provide preliminary support that the two are linked. In other words, that someone who is intensely engaged with the cancer risk, through open discussion, is more likely to become actively involved with sharing risk information – and, essentially, promoting genetic counselling and testing - with other family members, whereas someone who is only partially engaged may be less involved with these activities. The hypothesis may be further supported by the example of one participant, Kerry, who reported only getting involved with genetic testing because her sister 'rail-roaded' her into it and she 'just went along with it' (in other words, Kerry could be described as only partially engaged with the process). At the time she was interviewed, Kerry was feeling totally overwhelmed by having to talk to her daughter about her forthcoming risk-reducing surgery and felt that no one from the genetic service had offered her any support or prepared her for her positive result. Obviously, further research would be needed for further evidence-based research and development to confirm whether this has the potential to provide a psychosocial model for explaining variations in behaviour around family communication for cancer risk. However, TE does provide a useful framework for this Key Finding.

If further research is able to support this key finding, then there would be implications for offering different types of support to those undergoing genetic testing for *BRCA1/2*, depending on the level of engagement. This subsequently raises the issue of how genetic professionals, such as genetic counsellors, measure clients' 'engagement'. It may be necessary to look at developing a measurement tool or questionnaire that could be introduced into clinical practice to stratify those undergoing genetic testing to their required level of support.

Key Finding Three: There is lack of understanding of risk to men and their offspring based on perceptions of female disease

Cancer risk for male carriers

In reality, perceptions of female disease is a major theme affecting family communication regarding genetic testing for *BRCA1/2*. Perhaps it is not surprising that participants perceived hereditary breast and ovarian cancer as being a woman's issue, given that, in 2006, there were 45,822 new cases of breast cancer diagnosed in the UK, of which over 99% (*n*= 45,508) were women and less than 1% (*n*= 314) were men (Cancer Research UK, 2011b). In this study, participants reported 72 cases of cancer in their families. Of these, only nine occurred in men (of which only three were male incidences of breast cancer).

While men and women have an equal chance of inheriting a mutation in one of their *BRCA* genes, the risks of developing cancer are much greater in women (Hallowell *et al.*, 2005b). Table 8 summarises the estimated cancer risk to male *BRCA1/2* mutation carriers available in the

literature.[30] Prostate cancer risk is the most consistent finding for male carriers of *BRCA1/2* mutations in families with cancer (Liede *et al.*, 2000).

	BCRA 1	BRCA 2
Breast Cancers	*BRCA1* mutation-positive men have an additional 1.2% risk of developing breast cancer (Brose *et al.*, 2002;Evans *et al.*, 2010;Ottini *et al.*, 2003;Tai *et al.*, 2007;Thompson and Easton, 2002)	*BRCA2* mutation-positive men have an additional 6-10% risk of developing breast cancer until age 70 (Lux *et al.*, 2006;Easton *et al.*, 1997;Levy-Lahad and Friedman, 2007;Evans *et al.*, 2010;Ottini *et al.*, 2003;Tai *et al.*, 2007;Thompson and Easton, 2001)
Prostate Cancers	Male *BRCA1* gene carriers have an estimated relative risk [RR] of developing prostate cancer of 3.33% 91.78-6.20). (Ford *et al.*, 1994) Although accepted for non-Jewish *BRCA1* and *BRCA2* carriers, several studies of male Ashkenazi Jewish *BRCA1/2* carriers have not shown an increased risk for prostate cancer (Nastiuk *et al.*, 1999;Vazina *et al.*, 2000)	*BRCA2* mutation-positive men have an additional lifetime risk of prostate cancer of between 6 and 35% in the age group of 65-70 years (Lux *et al.*, 2006;Easton *et al.*, 1997;The Breast Cancer Linkage Consortium, 1999;Levy-Lahad and Friedman, 2007) A more rapid progression of prostate cancer has also been reported in men with *BRCA2* mutations (Mitra *et al.*, 2011;Narod *et al.*, 2008;Mitra *et al.*, 2008)
Other Cancers	*BRCA1* mutations in men have been associated with elevated risks of colorectal, pancreatic, and male breast cancers (Ford *et al.*, 1994;Borg *et al.*, 2000;Moslehi *et al.*, 2000)	Male *BRCA2* gene carriers have been shown to have an increased risk of developing pancreatic, stomach, bile-duct and gall-bladder cancers, and of cutaneous malignant melanoma (The Breast Cancer Linkage Consortium, 1999)

Table 8: The estimated cancer risk to male BRCA1/2 mutation carriers

[30] Section 1.2.2 in Chapter One described the estimated cancer risk to female *BRCA1/2* mutation carriers.

Men and *BRCA1/2* Genetic Testing

Although variable, rates of predictive testing in men are lower than in women (Bodd *et al.*, 2003;Goelen *et al.*, 1999;Julian-Reynier *et al.*, 2000b). For example, male participants accounted for only 24% of participants in a UK nationwide study of predictive *BRCA1/2* testing (Foster *et al.*, 2002). However, it is not just the testing itself that men are less engaged in; evidence suggests male members of breast/ovarian cancer families are less likely to participate at every level of the genetic counselling, testing, and communication process (Daly, 2009;Finlay *et al.*, 2008). In fact, Evans *et al.* (2002) estimate that 50% of eligible women take a *BRCA1/2* genetic test when available compared to only 11% of eligible men.

Hallowell *et al.* (2005b) propose this gender difference may reflect not only the fact that the risks of developing breast cancer are much lower in men than women, but also the limited preventative measures available to men. Media portrayal of female breast cancer and 'breast and ovarian cancer genes' also often reinforces the common misconception that hereditary breast and ovarian cancer is primarily a 'gendered' disease (Claes *et al.*, 2003).

Alternatively, significant differences in general health practices reported between men and women may explain the differences in attitudes towards genetic testing for *BRCA1/2* between males and females. Women have traditionally assumed the role of health maintenance within the family, including genetics (D'Agincourt-Canning, 2001;Hallowell *et al.*, 2005a;Hallowell *et al.*, 2005b). They are more likely to actively seek health-promoting behaviours, whereas men are more likely to avoid them (Courtenay, 2000;Marteau *et al.*, 1997;Schofield *et al.*, 2000). Such avoidance in men has been associated with social perceptions of masculinity, and male illness has been linked to a sign of personal weakness (Beare and Priddy, 1999;Connell and Messerschmidt, 2005;Courtenay, 2000;Hyde, 2005).

In 1996, Dudok deWit *et al.* reported on the psychological impact of undergoing predictive testing on four men from families with breast cancer. All four participants reported difficulties with the genetic-counselling process, exhibiting avoidance behaviours and a tendency to either miss appointments or withdraw from testing. McAllister *et al.* (1998) and Dudok deWit *et al.* (1996) suggest such avoidance is linked to a fear of developing cancer and is a coping strategy for men in hereditary breast and ovarian cancer families. However, evidence concerning psychological implications in men is limited by the few, small-scale, mostly qualitative studies (Shiloh *et al.*, 2011).

Research does suggest that female relatives often initiate the male counselling and genetic testing process (Liede *et al.*, 2000). It has also been suggested that explicit pressure exerted by family members may mean, in some cases, men undergo genetic testing against their will (Hallowell *et al.*, 2005b). Many studies have reported men describing their reason for having predictive testing as 'family recommendation' (Hallowell *et al.*, 2005b;Liede *et al.*, 2000) and that women have a strong influence upon male decision-making regarding genetic testing (Strømsvik *et al.*, 2011). For example, Hallowell *et al.* (2005b) reported that all female partners of men who were offered *BRCA1/2* predictive genetic testing indicated that they felt they had a right to help make the decision because it was their children (or their partner) who were directly implicated by the test outcome.

The literature available suggests that men primarily undergo genetic testing out of an obligation to their children, in particular daughters (Liede *et al.*, 2000;Daly *et al.*, 2003;Goelen *et al.*, 1999;Lodder *et al.*, 2001). A qualitative study of 22 men from 16 high-risk families in Ireland revealed that men who have a family history of hereditary breast and ovarian cancer were particularly worried about their daughters' risk status, and that more men in the study with daughters were tested than men without daughters (McAllister *et al.*, 1998). This makes sense in

that, as seen in Table 8, males with a *BRCA1/2* mutation are not at greatly increased risk for cancer compared to female relatives who carry the mutation (Lodder *et al.*, 2001). However, as carriers they have a 50% chance of passing that significant increased risk on to their daughters (and sons), who in turn if gene carriers have a 50% chance of passing on the gene mutation to their offspring.

Male involvement in family communication about cancer risk and genetic testing

Like the female participants of this current study, the published literature suggests that cancer amongst their family members may influence how male relatives feel about cancer risk (McAllister *et al.*, 1998). McAllister *et al.* (1998) concluded that men from families with hereditary breast cancer are affected emotionally by their female relatives' diagnoses. However, these men reported little communication with relatives about the illness, with some men feeling excluded from discussion about cancer among female family members (McAllister *et al.*, 1998). McAllister (1999) found that family cancers, including colorectal cancer, were discussed more by women than men in families with *HNPCC*, suggesting that their exclusion is not because of the female nature of breast cancer. Given that Key Finding two suggests that there is a relationship between level of engagement with cancer risk and how openly it is discussed, it may be that not including male relatives in early discussion about cancer in the family is why they are not so likely to undergo genetic counselling and/or testing compared to women.

In contrast, in a study of 59 men testing positive for a *BRCA1*/*BRCA2* mutation, Liede *et al.* (2000) found that the majority (52/59) of men had participated in past family discussions of risk of development of breast and/or ovarian cancer. Hallowell *et al.* (2005a) also found there was little evidence that men had been excluded from discussions about cancer in their family in their study exploring the influences on male patients' genetic test decisions, with 29 carrier and non-carrier men and immediate family members (17 male patients, 8 female partners, and 4 adult

children). In all cases, the men reported that the cancer in the family was common knowledge (Hallowell *et al.*, 2005b).It is worth noting, however, that the studies reporting male involvement in family discussion about cancer risk before genetic testing is self-reported by men, who subsequently went on to be tested. There is no evidence available from those who do not go on to have testing, either because they decline or are simply not informed, as to whether they feel they are included in such discussions. Significantly more women take up genetic testing then men and so it may be those men involved in family discussion are over-represented within these samples.

It has been proposed that women are the 'housekeepers' of genetic knowledge (Richards, 1996). Early studies looking at family communication about inherited cancers describe women as being the 'kin-keepers' (Richards and Green, 1996;Green *et al.*, 1997) and that the key providers of information are mothers (Green *et al.*, 1997). In 2009, Daly (2009) conducted a literature review on the experiences of males in families with positive *BRCA1/2* mutations. While acknowledging that the data are limited, Daly concluded that men are considerably less likely to participate in communication regarding genetics at every level, including the counselling and testing process, compared to female relatives (Daly, 2009). Studies involving patients undergoing genetic testing and/or counselling for hereditary breast and ovarian cancer have consistently highlighted that responsibility for communicating information within families is more likely to be taken by women rather than men, even if they are not at risk themselves (Claes *et al.*, 2003;Foster *et al.*, 2004a;Wagner *et al.*, 2003;Forrest *et al.*, 2003;D'Agincourt-Canning, 2001). Women are more likely to communicate genetic test results (d'Agincourt-Canning 2001; Forrest *et al.* 2003; Hughes *et al.* 1999; Lerman *et al.* 1998) and female relatives are more likely to be informed about hereditary breast and ovarian cancer (Hughes *et al.* 1999; Julian-Reynier *et al.* 2000; Lerman *et al.* 1998). Similar findings have also been seen with communication about carrier-testing in Haemophilia A families, a chromosome X-linked disorder carried by females (Sorenson

et al., 2003;Varekamp *et al.*, 1992). So, it may be that female family members are not necessarily well-informed of the risks to males, meaning they are less likely to include them in discussions (Green *et al.*, 1997).

In a qualitative interview study exploring how information about *BRCA1/2* genetic testing is disseminated within the families of at-risk men who undergo genetic testing, Hallowell *et al.* (2005a) found that, although both parents reported sharing the responsibility for initially telling their children about their father's intention to undergo testing and/or disclosing the genetic test results, on-going discussions about the health implications for offspring tended to take place between children (particularly daughters) and their mothers. This held true regardless as to whether the father was found to be a mutation carrier or not (Hallowell *et al.*, 2005a).

Authors have suggested that the reason women play such a dominent role in family communication is because these are 'female' diseases. However, studies of family communication regarding genetic diseases that are carried by and affect both sexes offer no conclusive evidence to support this. For example, some studies looking at family communication and genetic testing for cystic fibrosis (Ormond *et al.*, 2003) and hereditary non-polyposis colon cancer testing (Peterson *et al.*, 2003), have suggested dissemination of genetic information within the family is undertaken by both men and women relatively equally. Whereas, other HNPCC communication studies have reported women as being the key providers of information, even if their husband is the one at-risk (Koehly *et al.*, 2003;McAllister, 1999).

It is clear that something needs to be done to increase the awareness of risks to male relatives and their offspring so they are included in family communications regarding genetic testing (see Key Finding four). Further research is also needed into the best way to present the information to male relatives so they can make informed decisions about genetic risk. Gaff *et al.* (2005)

looked into how the genetic counselling process and communication aids could be utilised to help those undergoing genetic testing for cancer risk in order to inform relatives that predictive genetic testing is available. They found there were clear gender differences. For example, women reported that it was normal for family members to communicate about these issues, whereas no men reported that it was normal to communicate about these issues. Most of the male participants expressed a need for guidance or professional support in communicating to relatives, and they found advice about which family members should be informed helpful (Gaff *et al.*, 2005).

Key Finding Four: Emotionally distant and male relatives are only contacted selectively. Those undergoing genetic testing are not good at identifying all at-risk family members in order to share the implications of the genetic test with them.

The conceptual model for the theoretical basis of this work, introduced in section 3.4.4, held that family functioning, namely family health beliefs about hereditary breast and ovarian cancer and family organisational and structural characteristics, would affect family communication regarding *BRCA1/2* genetic testing. This has consistently been supported in the findings of the study.

This work also shows that, because of the shared health beliefs and structural characteristics, those undergoing genetic testing for *BRCA1/2* are not good at identifying who they should include in discussions in order to pass on relevant information to at-risk relatives. Instead, communication is based on existing norms and patterns within a family that establish how its members generally interact with each other.

However, as Peterson (2003) points out, from a clinical point of view the function of communication may be focussed on accurately disseminating health risk and disease information among potentially at-risk family members; from the perspective of the individual undergoing genetic testing for BRCA1/2, it may also serve as a social support function to build interpersonal ties and to facilitate coping. The findings of this present study show that, for participants, it was their emotionally close, female relatives who offered such social support and regular dialogue. As a result, male relatives and more distant relatives are often excluded.

In section 8.2.4, laying the communication eco-map (which showed who was actually told) over the family tree (which showed who should ideally be told) revealed potentially at-risk relatives who were not given information about genetic testing. The important thing this doctoral study demonstrates is that the relatives being missed out are not random, but that there are clear patterns. For example, those undergoing genetic testing for *BRCA1/2* readily talk to relatives they are emotionally close to; those that are missed are men (refer to Key Finding three) and more emotionally distant relatives (refer to Key Finding one).

It is clear that one of the ways to improve family communication regarding genetic testing for *BRCA1/2* is to help those undergoing genetic testing use their family tree rather than their existing norms and communication patterns within the family when identifying who, in the family, the information is relevant to in terms of genetic risk. That is not to say they will not continue to talk to particular relatives in order to seek support throughout the process, but rather, in addition to that, they are aware of who in the family may also be at risk.

The findings in Chapters Five to Eight, and the Systematic Review presented in Chapter Two, would suggest that endorsement from the clinical staff at the genetic services leads to increased family communication. Those participants who remembered discussions about communicating

with relatives during the genetic counselling process described it as an important, necessary task. However, others reported not remembering talking to the genetic practitioners about communicating their results to family members; in these cases, there was less evidence of discussions with people outside of their normal communication patterns. This would suggest that the encouragement of the genetic practitioner was an important factor in influencing whether relatives were informed. Therefore, one recommendation for the practical implementation of this work would be to encourage genetic practitioners to actively engage patients in more discussions about the consequences of the genetic test results for other family members, and how this may be communicated with them. That is not to say that genetic practitioners should directly tell clients they must inform all at-risk relatives, which would be contradictory to the non-directive principles of genetic counselling. But rather, the evidence suggests that, by introducing the topic and offering support to those undergoing genetic counselling to identify who, in the family, the information may be relevant to, it would be hugely beneficial in promoting family communication regarding genetic testing for *BRCA1/2.* According to Peterson (2005), practitioners can play an important role in helping encourage prospective consideration of barriers and difficulties in sharing genetic information, as well as to support identification of strategies for addressing potential problems.

The success of using the eco-maps in this study would also endorse using an eco-map or genogram in a therapeutic way in clinical practice to point out support networks or possible difficulties within the family. Daly (1999) found that the genogram can serve as a tool to members of a multidisciplinary clinical genetics team to provide a recorded memoir of a family's past and present attitudes and beliefs about genetic risk, as well as a record of the quality of relationships and dynamics within the family. Eunpu (1997) also supports the use of the genogram to incorporate the exploration of family relationships within the genetic counselling

setting. It may be that other, external tools,[31] such as leaflet or an online resource, could be developed to support those undergoing genetic testing to identify potentially at-risk relatives.

Key Finding Five: As far as family are concerned, members do not have the right to make an informed decision to decline.

The literature consistently reports one of the primary motivations for undergoing genetic testing is to learn more about their health risks for other family members (Hallowell *et al.*, 2003). During their family communication regarding genetic testing, almost all participants came across at least one family member who did not want to know about the testing, the results, or who was not prepared to act on the news. This was often a cause for concern. In these instances, the decision was made, sometimes in consultation with the clinician, to wait until the results came back before pursuing it further. Participants in this study were interviewed eight to 18 months after receiving their *BRCA1/2* test result and, for many of them, the family were still struggling with immediate family members, mostly sons and brothers, who did not want to go for genetic counselling or genetic testing, but who they believe should. As mentioned in Chapter Eight, this often became a family affair, with several generations of family members getting involved with talking that person round.

Paradoxically, when talking about more distant relatives, many participants expressed the sentiment that, once the information about genetic risk in the family had been passed on, it was up to the recipient to act on, that it was an individual choice and that their opinions regarding genetic testing should be respected. However, when discussing members of their immediate

[31] Accessible outside genetic counselling appointments.

family who had opted not to follow up with the genetic counselling, the consensus was that they were 'wrong'. This was especially true if they had children (in particular, daughters) who could potentially be at risk, and so discussions would continue until they could be persuaded. As far as family are concerned, members do not have the right to make an informed decision to decline.

Limited data are available about at-risk individuals who decline genetic testing. Foster *et al.* (2004b) conducted a cohort study of 315 adults eligible for predictive genetic testing for *BRCA1/2* from nine UK centres. The aim of the cohort study was to investigate the psychosocial impact of predictive genetic testing for *BRCA1/2*. However, 34 (11%) of the 315 cohort declined the offer of predictive genetic testing, allowing the authors to conduct some research around this group. Seventy-nine per cent of the test decliners were women; 76% were married or living with a partner; 53% had a college or university education; and 76% had children. There was no difference in sex distribution, marital status or employment status among test decliners and test acceptors. However, the decliner group were significantly younger than the test acceptors ($p=0.03$; MW), and had fewer ($p=0.001$; MW) and younger ($p=0.006$; MW) children. However, as the authors note, due to the small numbers, this should be interpreted with caution (Foster *et al.*, 2004b).

While 78% of test decliners felt that their health was at risk, they reported not wanting a genetic testing because, if they were found to carry a *BRCA1/2* mutation, they would worry: about their children's health (76%), their life insurance (60%), and their own health (56%). When asked about whether or not they would think about having a *BRCA1/2* test in the future, 77% said that they might want the test in the future, 18% were undecided, and only one (5%) reported that she definitely never wanted the *BRCA1/2* test (Foster *et al.*, 2004b).

Few studies have prospectively compared cancer-related distress in decliners and testers and the research to date is limited by small decliner groups and/or uncontrolled analyses. Lodder *et al.* (2003) reported no differences in cancer-related distress between decliners and testers using nine of the 15 items of the impact of event scale (IES)[32], where all participants had received counselling or education before deciding to decline testing. The sample, however, only included 13 decliners (Lerman *et al.*, 1998). Foster *et al.* (2004) found test decliners had lower levels of cancer worry, using the 6-item Cancer Worry Scale-Revised,[33] compared to those women who had accepted the test. Reichelt *et al.* (1999) found unaffected women (n = 301) who declined *BRCA1/2* testing had significantly lower levels of HADS[34]-defined depressive symptoms, compared to testers, although other potential confounders were uncontrolled. In contrast, two studies (Thompson *et al.*, 2002;Lerman *et al.*, 1998) found lower distress in decliners; however, in these cases, the decliners either did not receive counselling/education, or measures were completed before counselling sessions.

For example, Lerman *et al.* (1998) reported higher rate of cancer worry (Intrusion Subscale of the Revised Impact of Event Scale) and depression (the Centre for Epidemiologic Studies Depression (CES-D) Scale[35]) amongst men and women participating in a research study who did not wish to learn their *BRCA1/2* predictive test result, compared to those that requested testing. Likewise, Thompson *et al.* (2002) found African-American women who declined both counselling and testing for *BRCA* reported the lower levels of cancer-specific distress (Intrusion Subscale of the Revised Impact of Event Scale) compared to those who underwent counselling. However, the participants who declined counselling did have: significantly less knowledge of breast cancer

[32] Impact of event scale (IES) [Horowitz *et al.* 1979].
[33] The 6-item Cancer Worry Scale-Revised (Foster *et al.* 2002b; Lerman & Schwartz 1993).
[34] Anxiety and Depression Scale (HADS).
[35] The Centre for Epidemiologic Studies Depression Scale (CES-D; (Radloff 1977)).

genetics; significantly higher perceived barrier scores; greater anticipation of negative emotional responses to testing; and more concern about stigmatisation.

Questions remain as to why individuals decline genetic testing for *BRCA1/2* and what the consequences of this action are. Barriers for cancer genetic counselling and reasons to decline genetic testing reported in the literature include: anxiety; anticipation of negative emotional reactions to the test result; travelling to the genetics clinic; taking time away from work/family; concerns for health insurance; no perceived benefit; and time commitment (Codori *et al.*, 1999;Decruyenaere *et al.*, 1997;Foster *et al.*, 2004b;Geer *et al.*, 2001;van der Steenstraten *et al.*, 1994). Of course, such barriers can be discussed in genetic counselling. However, it is important to appreciate that not all eligible individuals reach genetic services in order to discuss them. This was certainly the case for the participants' relatives, who decided not to pursue genetic testing themselves in this present study; in most cases, their decision was based on the second-hand information provided to them by other relatives who had attended genetic counselling sessions.

The genetic counselling process is designed to allow those individuals eligible for predictive genetic testing to make informed risk management decisions and minimise psychological distress experienced, whether or not they proceed with predictive genetic testing (Foster *et al.*, 2004b). However, if these relatives are not even reaching genetic counselling, can they be said to making an 'informed' decision? The 'declining' relatives, with whom the participants in this study, and their family, are battling with because they do not agree with their decision not to pursue genetic testing, are largely those who have not had direct contact with the genetic service. Instead, their information and knowledge has been communicated to them though the family members that have attended and opted for genetic testing.

It may be that they feel they have had enough accurate information from their family to make an 'informed' decision and, therefore, from a professional opinion that is their choice and should be respected. However, if, as these findings suggest, the information is being communicated haphazardly and some individuals are declining testing for less well-informed reasons, then further research is needed on how to support this group and to improve family communication regarding genetic testing. Also, as Foster *et al.* (2004b) point out, 'whilst genetic counselling aims to be non-directive, relatives may be far from impartial' (p. 25).

Similar to the findings of this present study, Foster *et al.* (2002) reported that relatives with children not interested in *BRCA1/2* testing were described as selfish by individuals tested, and women have described encouraging relatives to have *BRCA1/2* testing. Tension may arise within these families and continue long-term, potentially resulting in some individuals eventually feeling coerced into testing against their will. This is an area where further research is needed; the problem being that those who do not reach genetic services are a difficult, if not impossible, group to assess (Foster *et al.*, 2004b), thus making future research challenging.

Key Finding Six: Plans for telling people in the future, especially children, is a cause of worry and concern and needs further support, especially long term.

Participants in this study had 35 adult children (over the age of 18 years old). Almost all of them were informed about their parent's genetic test result and were included in discussions about cancer in the family to some degree (refer to Chapter Five). Twelve of the 29 participants had offspring under the age of 18 years, totalling 24 children (refer to section 5.2). The mean age was 12.6 years for girls and 9.7 years for boys. Six of the 24 children aged 14 to 17 were told

about their mother's genetic testing (giving a disclosure rate of 25%), compared to 18 children, aged 2-15, who were not informed.

From the findings presented in Chapter Five, it could be hypothesised that older children (those over 15 years old) are more likely to be told than younger children (those under 14 years old). Also, the children's gender has little impact on disclosure of test results as, in all cases except one, the information was either given to all siblings under the age of 18 years or none. However, as there were only a few children reported in this study, it is difficult to draw any definite conclusions. There is evidence in the current literature to support this thinking.

A number of studies have reported on whether parents communicated *BRCA1/2* genetic test results to at-risk offspring (Wagner *et al.*, 2003;Patenaude *et al.*, 2006;Claes *et al.*, 2003;Bradbury *et al.*, 2007;Hughes *et al.*, 1999;Tercyak *et al.*, 2001;Tercyak *et al.*, 2002;McGivern *et al.*, 2004;Hallowell *et al.*, 2005a). These studies give an average disclosure rate of approximately 50% for offspring aged between 4 and 25 years old. However, few studies break down disclosure rates by age, e.g. adult versus child offspring, which may explain the higher rate than found in this present study, which only includes offspring aged 17 years old and younger. Only a handful of studies specifically discuss disclosure to young children (Tercyak *et al.*, 2002;Bradbury *et al.*, 2007;Bradbury *et al.*, 2012;Tercyak *et al.*, 2001).

Several studies have shown that the age of offspring is an important factor in parental decisions as to whether or not to disclose test results. Segal *et al.* (2004) found that, out of 31 mothers disclosing their BRCA test results to offspring, 50% of offspring aged 20 to 29 years were informed of the results, whereas approximately 25% of those aged 19 years or younger were told (similar rates found in this present study). However, unlike this study, sons and daughters were not notified equally (Segal *et al.*, 2004). Likewise, Bradbury *et al.* (2007) reported 83% of

offspring over 18 years old were told of their mother's positive BRCA mutation results, compared to only 21% of those aged 13 years or younger.

Interestingly, Bradbury *et al.* (2007) reported that almost half the parents reported that their child did not appear to understand the significance of a positive *BRCA1/2* test result, with older children seeming to have a better understanding. Level of understanding was measured by parents' qualitative perception rather than quantitatively, using a standard tool.

In 2012, Bradbury *et al.* conducted semi-structured interviews with 253 parents (61% response rate), who had *BRCA1/2* testing and at least one child under the age of 25 years. Of the 505 offspring, 334 (66%) were told about their parent's test result. Children were more likely to be told if they were older ($P \leq .01$), were female ($P = .05$), or if the parent's test result was negative ($P = .03$). Parents most frequently reported their offspring's initial response was neutral (41%) or relief (28%). However, 13% of offspring were reported to experience concern or/and 11% distress; this was associated with parents receiving a *BRCA1/2* positive or inconclusive result (Bradbury *et al.*, 2012).

Hallowell *et al.* (2005a) observed that parents adopted one of three strategies when communicating information about a father's genetic risk for *BRCA1/2* to their children. These were complete openness, limited disclosure, or total secrecy, in which the timing and content of disclosure varied. Parents justified the adoption of a particular strategy by reference to children's rights to information versus parental duty to avoid causing children. The authors found parents were more hesitant about discussing genetic testing and its implications with sons and younger children (those less than 18 years old) prior to receiving the result. However, unlike the findings of this study, which found that whilst the majority of adult children were informed, only 25% of child offspring were, by the time Hallowell *et al.*'s research interviews occurred

(median time since receipt of results = 26 months; range 8-74 months) nearly all of the offspring, including child offspring, had been informed about their father's test result. It is worth noting that only eight out of 16 fathers included in the study had children under the age of 18 at the time (Hallowell *et al.*, 2005a).

Tercyak *et al.* (2002) found that participants who told their children younger than 13 years about their genetic test result said their children had increased distress, compared to participants that did not tell their young children, who experienced a slight decrease in distress.

The findings presented in Chapter Eight show that those participants with young children, and who had received a positive *BRCA1/2* test result, had given some consideration as to how and when their children should be told in the future; and this matter caused them anxiety to a greater or lesser degree, depending on the personality and how much support they perceived themselves to have.

Several studies have shown that parents regard the disclosure of genetic information to their children as their personal responsibility rather than the responsibility of health professionals (Claes *et al.*, 2003;Forrest *et al.*, 2003;Hallowell *et al.*, 2005a). This was true of these participants; however, several suggested that they would seek advice from a genetic counsellor when they decided the time was right to share the information.

Previous studies have looked at the kind of support people undergoing genetic testing would like when talking to their children about the genetic test and its implications. For example, Tercyak *et al.* (2007) found mothers undergoing *BRCA1/2* testing, who had children aged 8-12 years, most-to-least frequently cited information resource needs regarding communication to be: literature (93.4%), family counselling (85.8%), prior participants (79.0%), support groups (53.9%)

and other (28.9%). Similarly, Segal *et al.* (2004) found that those who had disclosed their *BRCA1/2* results and those who had not, respectively, indicated that they would have appreciated further follow-up meetings with a genetic counsellor (50.0% vs. 46.2%), family counselling (38.9% vs. 38.5%), peer support groups of carriers and their children (33.3% vs. 38.8%), professional-led support group (22.2% vs. 38.5%), educational forum for families (46.2% vs. 38.8%), or information pamphlets on 'how to disclose and cope' with genetic test results.

Many of the participants in this situation felt that the 'right' time to share their genetic test results and its implications was when their children were 'grown up' and emotionally equipped to deal with the information. Predictive genetic testing for adult-onset diseases, such as breast and ovarian cancer, is generally discouraged until the age at which interventions, such as risk-reduction measures and screening, are believed to be helpful, which is usually not before 25 years old (Bradbury *et al.*, 2007;Bradbury *et al.*, 2012).

This Key Finding would support a call for further research looking at the experiences of parents telling their young children about their genetic testing, both at the time and, for those who choose to, at a later date; and looking at how they could best be supported by genetic professionals.

9.4 Limitations of Work

Naturally, there are some limitations to this study. The work presented in this thesis represents not only the development of knowledge on the topic of how those undergoing genetic testing for *BRCA1/2* talk to their family, but also the development of the author's competence and

capabilities as a researcher. The work was undertaken by an apprentice researcher and, as such, the learning curve associated with it was huge. The strengths and weaknesses of the work are inextricably linked to these two parallel processes and the journey of learning that took place over the years of its development.

The sample was purposively sampled and, therefore, does not represent the entire population for whom genetic testing for *BRCA1/2* would be appropriate. However, as this was a piece of exploratory qualitative research, this was never the intention of this work. Individuals who were considered by the geneticists as too vulnerable to participate were excluded (refer to 4.3), and affected families who did not come into contact with clinical genetic services were not represented. The response rate was 37.7% of those eligible patients invited to participate (see section 5.2). Many of the participants reported to the interviewer that their relatives had also received an invitation to participate and intended to do so. However, only three sets of relations were included in the final interviews (two sets of sisters, and one aunt and niece). This suggests that, despite potentially showing interest to their family, many potential participants did not return their forms to the researcher. As a term of the ethical approval, no reminder letters were sent to individuals who did not send back a reply slip, which may have limited the response rate.

The participants were predominately Caucasian women with at least one child. Most participants had an inconclusive or negative mutation status. Although not evident in the findings, it is possible that those receiving a positive result found talking to their relatives more challenging, which would be under-represented in this population. Only two men were included in this sample (although this is proportional to the number of men who undergo genetic testing for *BRCA1/2* at the Genetics Centre compared to women). Even so, Gaff *et al.* (2005) found that men had a greater need for professional support than women when communicating genetic

testing results for Hereditary Non-Polyposis Colorectal Cancers (*HNPCC*), suggesting that this population would warrant further research.

From the outset, potential participants were aware that the study aimed to examine family communication following genetic testing, and so it may have appealed more to those who felt they had had positive experiences of this, were more confident, and/or willing to talk. Also, there were complicated cases of family communication at the time this study was being conducted as reported by the staff at the Genetics Service. It may be that some or all participants wanted to present themselves in certain ways, and therefore under-reported complicated or difficult situations. As such, those who found the experience of communicating with relatives more challenging may be under-represented, thereby introducing a sample bias.

As with all studies that employ a retrospective design, it is possible that the participants' accounts may have been tainted by hindsight or the need to present themselves as responsible parents.

The work could be criticised for the lack of triangulation given that only one data source was used. However, the focus of the study was to concentrate on the experiences of the individual's themselves within their own life world. The inclusion of multiple data sources would have introduced alternative perceptions, which would have been contradictory to this focus. Had multiple sources been used, then the depth of the data that was attained could not have been achieved due to the breadth which would have been required. Also, the utility of other sources of data, with the exception perhaps of other family members, would not have contributed to the aims of the study.

This work is arguably weakened by not adhering solely to a predefined and validated method of data analysis. The difference between reading and understanding how to conduct qualitative data analysis, and actually putting that into practice without losing either the depth or breadth of the data, was one of the hardest challenges to overcome as a novice researcher, as describing how to conduct qualitative data analysis is so often over simplified in the reporting in the literature. That said, although the data analysis was influenced by many sources, the final method used was logical and transparently reported, meaning that it could be repeated by another researcher.

If the study had been conducted with family, rather than individual, interviews it might have yielded different results and recruited a different sample. To meet the family together would allow the researcher to observe the interactions between members, including their roles, agreements, disagreements, efforts to expose or protect each other (Sobel and Cowan, 2000). This would have been in keeping with a systems framework, which recognises that the whole family is more than the sum of its parts and would have meant the family as a unit would be the focus of the analysis. However, this approach was not chosen because, as discussed in section 3.4.4, whilst talking about genetic testing and its risk implications may be viewed as a 'family affair', the individuals undergoing the genetic testing for BRCA1/2 may need to interact with their families in a new and unfamiliar manner (Peterson, 2005). In order to explore how and when those undergoing testing talk to their family, it was important to capture the experience of the individual, and their definition of who constitutes their family, within that. Therefore, a conceptual model, adapted from Peterson's (2005) family systems model , and Carter and McGoldrick's (1989) 'Family Life Cycle', which allows the study of how individuals within a family, and by association the family as a whole, adapt to chronic illness, was used as theoretical basis of this work.

Finally, the extent to which this study has succeeded in meeting its objectives depends not only on the sample of participants, but also the data collection and analysis strategy. This study was conducted from within an interpretive paradigm utilising an interpretive descriptive methodology. However, it is important to acknowledge that a different theoretical perspective, such as a phenomenological or ethnographical perspective, might have resulted in different findings.

9.5 Contributions to Knowledge, Implications and Future Research

The longitudinal view of family communication regarding genetic testing for *BRCA1/2* presented in this thesis is a new way of examining how those undergoing the genetic testing talk to their family as an on-going process. Gaining an understanding of the process of how and when, those undergoing genetic testing for *BRCA1/*2, talk to their relatives means the development of future interventions to support such family communication can be specifically targeted, not only to the most appropriate time point(s), but also to be in accordance with how these families are already communicating. Presenting the longitudinal view in this way, it endorses the need for prospective, longitudinal research looking at the experiences of individuals and families at each stage of the process.

Section 9.3 has identified some specific areas, relating to the six Key Findings of this work, where further research is needed. For example:

1. Further evidence-based research and theoretical development would be required to support whether the Theory of Engagement (McAllister 2002) has the potential to

provide a psychosocial model for explaining variations in behaviour around family communication for cancer risk (see Key Finding two).

2. Work is needed to increase the awareness of risks to male relatives and their offspring, and accuracy of transmission, so they are included in family communications regarding genetic testing (see Key Finding three).
3. The findings support that how those undergoing genetic testing for *BRCA1/*2 talk to their relatives is influenced by preexisting structures and belief systems within the family. Future research is necessary to examine the most effective way to appropriately utilise these in order to improve family communication on the topic (Harris *et al.*, 2010).
4. It is also necessary to identify the most suitable way(s) for those undergoing genetic testing for *BRCA1/2* to present the information about genetic testing and its implications to male relatives, so they can make informed decisions about genetic risk (see Key Finding three).
5. It is clear that one of the ways to improve family communication regarding genetic testing for *BRCA1/2* is to help those undergoing genetic testing use their family tree rather than their normal communication patterns (as identified on their eco-maps) when identifying who in the family the information is relevant to (see Key Finding four).
6. Little is known about those family members who decline a genetic test and whether that decision is a fully informed one; also, whether some decliners eventually feel coerced into testing against their will by other family members (see Key Finding five).
7. Plans for telling people in the future once regular contact with the genetics service has stopped, especially children, is a cause of worry and concern and needs further support, especially long term (see Key Finding six).
8. The findings would support a call for further research looking at the experiences of parents telling their young children about their genetic testing, both at the time of

testing and, for those who choose to, at a later date; and looking at how they could best be supported by genetic professionals (see Key Finding six).

Very few of the research participants were aware of the support mechanisms available to them from the Genetics Service; for example, contacting relatives directly on their behalf or producing customised letters to send to family members. Subsequently, this should be highlighted as an area where extra support and future research should be targeted. It is important that genetic practitioners take time to identify if this is a service that could be useful to the individual and offer it accordingly so a family member does not slip through the net. These findings are supported by Barsevick *et al.* (2008), who emphasise the need for genetic counsellors to devote more time and attention to help prepare individuals to communicate genetic test results to those relatives from whom they are distanced or estranged. The findings of Wiseman *et al.* (2010) also suggest that genetic practitioners would benefit from obtaining a clear understanding of the 'personal beliefs of those undergoing genetic testing about sharing genetic risk information and the relationships that that person has with each relative in order to identify areas of difficulty and support accurate communication' (p. 701).

Overall, the findings suggest that the developing interventions to help manage problems associated with family communication regarding genetic testing for cancer risk should be a top research priority, especially as the numbers of people affected by these issues is set to rise as more genes are discovered. The longitudinal view identified gives deep insight into how and when genetic testing for *BRCA1/2* and its implications are discussed within these families. This understanding will allow future interventions to be targeted where they are most helpful.

9.6 Conclusion

This research presented in this PhD thesis is particularly important because, as has been demonstrated, family communication regarding genetic testing for *BRCA1/2* can play a major role in ensuring relatives get access to genetic services and risk information. Nevertheless, the evidence suggests it can cause considerable distress and poses many challenges for those undergoing genetic testing. As a result, there have been numerous calls for interventions and support mechanisms from the clinical and academic communities. However, the critical review of the existing literature at the beginning of this PhD journey suggested this is not yet achievable because the nature of interactions regarding genetic information in families remains poorly understood. Therefore, this work strives to fill some of the gaps identified in the literature, which would also allow the improvement and development of future genetic services.

The work presented provides three key contributions that develop existing knowledge further. Firstly, this is the first piece of qualitative research that looks at how those undergoing genetic testing for *BRCA1/2* talk to their friends and family throughout the whole process before, during and beyond genetic counselling and genetic testing. This longitudinal view of family communication regarding genetic testing will allow future interventions to be specifically targeted where they will be most useful and to give the necessary level of support at the right time during what is now recognised as an on-going process.

Secondly, the work goes beyond the current literature by demonstrating how communication between emotionally close- and emotionally distant relatives differs, and why. In summary, with the first group, communication is focused around sharing and supporting; whereas with the latter it is about gaining and imparting information. As a result, the findings also demonstrate

the differences in expectations and follow up. Communication with emotionally close relatives is an on-going process that may have long last effects on the family; for example, relatives may not have the right to decline genetic testing in the eyes of other family members. On the other hand, communication with emotionally distant relatives is more haphazard and largely relies on information being cascaded down through the family.

Finally, despite participants being engaged and open to informing potentially at-risk relatives, there are clear pattern of whom those undergoing genetic testing for *BRCA1/2* are not communicating with. In particular male and more distant relatives with whom they are not normally in regular contact with. It would appear that the current model of genetic counselling offered to these participants is not enough to overcome participants' reliance on their normal communication patterns and pre-existing health beliefs and attitudes often shared by the family when discussing genetic testing and genetic risk to at-risk relatives. From this, the findings can be used to make specific recommendations on how to improve support, for example the development of interventions that encourage those undergoing genetic counselling/testing for *BRCA1/2* to use their family tree rather than their normal pattern of communication, as shown on their eco-maps, to identify who the information is relevant to.

The original research question for this PhD thesis, as described in Chapter Three, was focussed on how and when those undergoing genetic testing for *BRCA1/2* talk to the relatives about family history of cancer, associated risk and genetic testing. The extent to which the thesis addresses the research question should be considered. The longitudinal view presented in the findings does examine family communication at four difference stages throughout an on-going process and so does address the 'when' element of the research question directly.

The findings also give insight in to how communication differs at each stage. For example, stage one communications are based on normal communication patterns with emotionally close relatives and are influenced by cancer burden, in particular by death, diagnosis and life stage. During stage two communications with those same relatives continues in the form of sharing experiences and now knowledge from genetic counselling because it is viewed as new and interesting. This is also the stage where family communication with emotionally distant relatives may begin in the form of needing family history information and/or accessing predictive genetic testing through the diagnostic genetic testing of a relative who has already had cancer. Stage three communications occurred in two distinct waves: firstly rapid disclosure of test results to emotionally close relatives to alleviate anxiety with little thought or preparation; followed by keeping those in the known in the loop. Stage four communication could be an issue years in to the future and revolved around sharing a positive result, however was not limited to the person receiving that result, but rather became a family affair. Participants relied on sharing leaflets and clinic summary letters to ensure accurate transmission of genetic information; and often-delegated tasks to certain family members to ensure the job got done.

The discussion provided in the final chapter of this PhD thesis focusses on six key findings that are over-arching themes across how those undergoing genetic testing for *BRCA1/2* talk to their relatives rather than 'how' the information was transmitted, in terms of who said what and how it was received. This slight shift in focus away from the original research question strengthens the work in two strategic ways:

1. It provides evidence that data analysis went beyond just describing the events that occurred as reported by participants to providing an interpretive understanding that exposes the characteristics, patterns and structure in some clinically and theoretically useful way.

2. It allows the researcher to make significant and meaningful recommendations for future research and implications for clinical practice and intervention development that will help families talk about these issues.

The work was always intended to have a pragmatic emphasis. The very nature of “here is a problem that real people using the genetic services in the National Health Services are facing and something could be done to improve that situation” was the thing that appealed most as an area of study. In fact, the initial PhD proposal was for a two-phase study: a short exploratory study looking at the how those undergoing predictive genetic testing shared their test results with their at-risk relatives, followed by the development and piloting of an intervention to support these activities. Unluckily for the plans for a short PhD, but fortunately for the author’s continued development and training in qualitative methods, during the literature and proposal development it became clear that the first phase, if it was to be done rigorously with the required depth, would be ample to produce "an original and substantial contribution to knowledge”.

Although the majority of this PhD thesis represents a piece of qualitative research, there is evidence of a researcher whose roots lie in quantitative and lab-based research. For example, the eco-maps have been analysed quantitatively resulting in a measure of cohesion with the family. The methods chosen, such as: - Jones’ methods of conducting systematic reviews and meta-synthesis, Miles and Huberman’s sourcebook of data analysis and Ritchie and Spencer’s Framework Approach, all provide a clear and logical, practically step-by-step instructions, for a novice qualitative researcher to work through in what otherwise could have been a unruly depth of data. As well as giving structure and order to the proceedings, these methods have the major benefit of providing a very clear and coherent audit trail for the researcher, and others. That is

not to say that purist qualitative researchers do not work logically or are unable to provide an audit trail to their work, but rather to acknowledge how the personal qualities of this, now most definitely a 'mixed methods' researcher, bring strength to the work.

Appendices

Appendix 1: Quality Assessment Tool for Qualitative Papers.

1. Abstract and title: Did they provide a clear description of the study?

Rating	Criteria	Score
Good	Structured abstract with full information and clear title.	**3 points**
Fair	Fair abstract with most of the information.	**2 points**
Poor	Inadequate abstract.	**1 point**
Very poor	No abstract.	**0 points**

2. Introduction and aims: Was there a good background and clear statement of the aims of the research?

Good	Good full but concise background to discussion/study containing up-to date literature review and highlighting gaps in knowledge. Clear statement of aim AND objectives including research questions.	**3 points**
Fair	Some background and literature review. Research questions outlined.	**2 points**
Poor	Some background but no aim/objectives/questions, OR Aims/objectives but inadequate background.	**1 point**
Very Poor	No mention of aims/objectives. No background or literature review.	**0 points**

3. Method and data collection: Is the method appropriate and clearly explained?

Good	Method is appropriate and described clearly (e.g. interview guide included). Clear details of the data collection and recording.	**3 points**
Fair	Method appropriate, description could be better. Data collection described.	**2 points**
Poor	Questionable whether method is appropriate. Method described inadequately. Little description of data collection.	**1 point**
Very Poor	No mention of method, AND/OR method inappropriate, AND/OR no details of data collection.	**0 points**

4. Sampling: Was the sampling strategy appropriate to address the aims?

Good	Details (age/gender/race/context) of who was studied and how they were recruited. Why this group was targeted. The sample size was justified for the study. Response rates shown and explained.	**3 points**
Fair	Sample size justified. Most information given, but some missing.	**2 points**
Poor	Sampling mentioned but few descriptive details.	**1 point**
Very Poor	No details of sample.	**0 points**

5. Data analysis: Was the description of the data analysis sufficiently rigorous?

Good	Clear description of how analysis was done. Description of how themes derived/ respondent validation or triangulation.	**3 points**
Fair	Descriptive discussion of analysis.	**2 points**
Poor	Minimal details about analysis.	**1 point**
Very Poor	No discussion of analysis.	**0 points**

6. Ethics, bias and rigour: Have ethical issues been addressed, and what has necessary ethical approval gained? Has the relationship between researchers and participants been adequately considered?

Good	Ethics: Where necessary issues of confidentiality, sensitivity, and consent were addressed. Bias: Researcher was reflexive and/or aware of own bias. Rigour: Attempts made to ensure the rigour of the research	**3 points**
Fair	Lip service was paid to above (i.e. these issues were acknowledged).	**2 points**
Poor	Brief mention of issues. At least, evidence that ethical approval has been sought.	**1 point**
Very Poor	No mention of issues.	**0 points**

7. Results: Is there a clear statement of the findings?

Good	Findings explicit, easy to understand, and in logical progression. Tables, if present, are explained in text. Discussion of results relate directly to aims. Sufficient data are presented to support findings.	**3 points**
Fair	Findings mentioned but more explanation could be given. Data presented in discussion relate directly to results.	**2 points**
Poor	Findings presented haphazardly, not explained, and do not progress logically from results. Qualitative data presented with stats or percentages with only limited suggestion that results were used within a qualitative paradigm.	**1 point**
Very Poor	Findings not mentioned or do not relate to aims. Qualitative data presented as stats or percentages only (e.g. 4/8, 50% participants said…).	**0 points**

8. Transferability or generalisability: Are the findings of this study transferable (generalisability) to a wider population?

Good	Context and setting of the study is described sufficiently to allow comparison with other contexts and settings, plus high score in Question 4 (sampling).	**3 points**
Fair	Some context and setting described, but more needed to replicate or compare the study with others, PLUS fair score or higher in Question 4.	**2 points**
Poor	Minimal description of context/setting.	**1 point**
Very Poor	No description of context/setting.	**0 points**

9. Implications and usefulness: How important are these findings to policy and practice?

Good	Contributes something new and/or different in terms of understanding/insight or perspective. Suggests ideas for further research. Suggests implications for policy and/or practice.	**3 points**
Fair	Two of the above.	**2 points**
Poor	Only one of the above.	**1 point**
Very Poor	None of the above.	**0 points**

10. Limitations: Are the limitations of the study discussed?

Good	Clear description of limitations with critical analysis of impact.	**3 points**
Fair	Descriptive list of study limitations.	**2 points**
Poor	Minimal details of study limitations.	**1 point**
Very Poor	No mention of study limitations.	**0 points**

11. Quotes: Are direct quotes of participants used to illustrate qualitative findings?

Good	Directs quotes used with full explanation of context and meaning and who they were made by (e.g. *male, carrier*). Quotes linked back to results to clearly illustrate points.	***3 points***
Fair	Direct quotes used with some explanation of meaning.	***2 points***
Poor	Minimal quotes used with little or no explanation	***1 point***
Very Poor	No quotes used.	***0 points***

12. Relevance to Systematic Review research question.

Good	Study explicitly based on family communication following GT with at least one aim to investigate factors that facilitate or impede family communication following GT.	***3 points***
Fair	Study based on experiences family communication following GT.	***2 points***
Poor	Study based on experiences of genetic testing generally, which included issues/experiences of with family communication following GT.	***1 point***
Very Poor	Study based on experiences of genetic testing where issues of family communication arise but were not explicitly asked about by the researcher.	***0 points***

Appendix 2: Standard Pro form used to review full papers for inclusion or exclusion in Systematic Review

Systematic Review – Filter for Full Papers					
Paper Reference		Primary Author		Year of Publication	

Inclusion Criteria	**Details**	**Meets Criteria**
Published primary qualitative research studies. Mixed methods included only if the qualitative findings are reported and discussed separately from the non-qualitative findings.		
The participants or a close family member (partner, parent or sibling) had undergone genetic testing for cancer risk.		
The outcome measures included data relating to factors which facilitated or impeded family communication following the testing.		
These factors are either stated by the authors or appeared from the published data to be an important element in the study findings.		
Exclusion Criteria	**Details**	**Meets Criteria**
Include other forms of genetic testing, such as carrier testing, or genetic testing for other conditions than late onset cancer.		

Paper Included		**Paper NOT Included**	

Appendix 3: Interview Guide for Researcher

1. Welcome and thank them for participating. Introduce researcher and study (including assurances of confidentiality).

2. Ask if the participant has any questions.

3. Ask participant to sign consent form (three copies: for hospital notes, for researcher, for themselves).

   [TURN AUDIO-RECORDER ON]

4. Demographic Data
   'I would like to start with a bit of background, so can you tell me a bit about yourself?'
   - Age
   - Occupation
   - Marital status
   - Children
   - General health status

5. Construct Communication Eco-Map:
   - Explain what map is and its purpose (stress this is about everyday life, not necessary genetic testing).
   - *'Who would you say are the most important people in your life?'*

6. Experiences of genetic testing and family communication
   - '*Could you tell me about your genetic testing and how that came about? I am particularly interested in who you spoke to about it, when and why*.'

7. Clarification and follow up
   May include details on:
   - Family communication patterns
   - Type of testing/Mutation status
   - Motives/reasons for GT
   - Coping strategies
   - General process of GT
   - Introduction by genetic service
   - General emotions towards telling family
   - Thought/preparation beforehand
   - People's reactions
   - Health beliefs
   - Anyone who didn't want to know
   - Help from Genetic Service
   - Thoughts now (regrets?)
   - Support would have liked
   - Top tips to others in situation

8. Round up interview, anything they would like to add?

9. Thank participant for time and effort. Reassurances of confidentiality. Remind them that support available from genetic service if they wish.

Appendix 4: Letter of Invitation from the Genetics service

[To be printed on headed note paper]

Dear [name of participant]

Re: Invitation to participate in a research study

I am writing to you on behalf of the School of Nursing and Midwifery at the University of Southampton to invite you to take part in a research study looking at whether and how people who have had genetic testing for hereditary breast or ovarian cancer talk to their relatives about their results and its implications, and how the health service could support them in doing this.

A researcher from the University, Kim Chivers, would like to talk to you about your experience of talking to your relatives about your genetic test results. This interview will last about one hour, depending on how much you have to say, and with your permission it will be audio-recorded. It will take place at a time and place suitable for you, probably in your own home. Anything you say will be treated with complete confidence. Please read the enclosed information sheet, which provides further detail about the study.

Once you have read the information sheet, if you choose to take part in this study please complete the form included and return it to the researcher, Kim Chivers, in the stamped address envelope provided. Once Kim has received your form, she will ring you to ask if you have any further questions about the research and to arrange a good time for the interview.

In the meantime, please do not hesitate to contact Kim if you have any further questions about the research. Her telephone number can be found at the end of the Information Sheet.

Yours sincerely

Professor Anneke Lucassen
Consultant & Professor in Clinical Genetics

Appendix 5: Participant Information Sheet

[To be printed on headed note paper]

Title of Project: Family Communication Following Genetic Testing

Why am I receiving this information sheet?
We would like to invite you to take part in a research study. Before you decide, you need to understand why the research is being done and what it would involve for you. Please take time to read the following information carefully. Talk to others about the study if you wish. Part I tells you the purpose of this study and what will happen if you take part. Part II gives you more detailed information about the conduct of the study. Please ask if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part.

Part I

What is the purpose of the study?
Some people have said that it can be difficult to talk about genetic testing with their family. We would like to understand how people who have had genetic testing for hereditary breast and/or ovarian cancer talk to their family about their results so that people can be helped to do this in the future.

Why have I been invited?
You have been invited to take part in this study as you have been identified by the Genetic Centre as having had a genetic test for hereditary breast and/or ovarian cancer within the last 6-18 months. We hope to talk to up to 30 men and women.

Do I have to take part?
You do not have to take part; it is up to you to decide if you want to. This information sheet describes the study so you can make your decision. If you do decide to take part, you can withdraw at any time, without giving a reason. This would not affect the standard of care you receive.

What will I have to do?
Taking part in this study will involve one face-to-face interview with our researcher, Kim Chivers, at a place of your choosing. This interview should last about one hour and will explore whether you talked to your family about your genetic testing results; your experiences of doing so; whether you would have liked more support with this and, if so, what kind of support would have been helpful. During the interview, you and the researcher will create a 'communication map' – this is similar to a family tree, but may include friends and family members not related by blood. This will help us see who, if anyone, you have shared your genetic information with.

What should I expect if I decide to take part?
If you choose to take part in this study we will telephone you to arrange the interview at a time and place that is good for you. If the interview is more than one week ahead, we will telephone you a few days before the interview to check the arrangement is still suitable.

On the day of the interview we will check you have read the information sheet and give you a chance to ask any more questions you may have. We will then ask you to sign a consent form which says you are happy to take part, that we can audio-record the

interview and can use the information you give, which will be made totally anonymous so you cannot be recognised, in our write-up of the study.

Expenses and payments
If you have to travel to the interview we will reimburse all your travelling expenses.

What are the possible disadvantages and risks of taking part?
While we do not foresee any harm or disadvantages to you by taking part in this study, we do understand that you may feel the things we discuss are sensitive and/or personal. So we aim to be supportive and open with you and you will be encouraged to only discuss the things which you are happy to talk about. We will also make sure that, should you need it, support is available to you afterwards.

What are the possible benefits of taking part?
This study may not benefit you directly, but the information you give may help improve the service provided for people undergoing genetic testing for hereditary breast and/or ovarian cancer in the future.

What happens when the research study stops?
We hope to be able to use the information provided by you and the other participants to develop a way of helping patients talk to their families about genetic risk if they want it.

What if there is a problem?
Any complaint about the way you have been dealt with during the study or any possible harm you might suffer will be addressed. The detailed information on this is given in Part II.

Will my taking part in the study be kept confidential?
Yes. We will follow ethical and legal practice and all information about you will be handled in confidence. The details are included in Part II.

This completes Part I. If the information in Part I has interested you and you are considering participation, please read the additional information in Part II before you make any decisions.

Part II

What will happen if I don't want to carry on with this study?
It is totally within your right to withdraw from the study at any time without having to give any reason. If you withdraw from the study, we will destroy any contact details.

What if there is a problem or I have a complaint?
If you have a concern about any aspect of this study, you should speak to the researcher, Kim Chivers, who will do her best to answer your questions. If you remain unhappy and wish to complain formally, you can do this through the NHS or University Complaints Procedure. Details can be obtained from the hospital.

In the event that something does go wrong and you are harmed during the research and this is due to someone's negligence, then you may have grounds for a legal action for compensation against the University of Southampton, but you may have to pay your legal costs. The normal NHS complaints mechanisms will still be available to you.

Will my taking part in the study be kept confidential?
Yes. We will follow ethical and legal practice and all information about you will be handled in confidence. All procedures for handling, processing, storage and destruction of data will match the Caldicott principles and the Data Protection Act 1998. All information which is collected about you during the course of the research will be kept strictly confidential, and any information about you which leaves the hospital will have your name and address removed and pseudonyms or codes will be used so that you cannot be recognised.

All details, including copies of your consent forms, will be kept in locked filing cabinets in secure office space within the School of Nursing and Midwifery. All recordings and transcriptions will be kept on a password protected computer which is backed up daily. Written field notes, memos and printed transcriptions will be kept in a locked filing cabinet separate from any identifying data. All primary data (audio-recordings, communication maps, written field notes, memos and transcriptions) will be kept for 15 years in accordance with University policy.

What will happen to the information I give?
With your permission, all interviews will be audio-recorded with a digital recorder. Then the interview recordings will be transcribed into text for data analysis.

What will happen to the results of the research study?
We have several plans for dissemination activities for this research, including:

- A PhD thesis
- Presentations (or similar event) within departmental research group at the University of Southampton
- Presentations at local, national and international conferences
- Papers for publication in service user, professional and peer-reviewed academic journals.

Who is organising and funding the research?
This work is funded by a studentship grant from Cancer Research UK and will contribute towards the researcher's PhD.

Who has reviewed the study?
All research in the NHS is looked at by an independent group of people called a Research Ethics Committee to protect your safety, rights, wellbeing and dignity. This study has been reviewed and given favourable opinion by the Research and Development Office and the Isle of Wight, Portsmouth & South East Hampshire Research Ethics Committee.

Can I get independent advice about taking part in research?
Yes, if you have any concerns or need independent advice, you can contact the Patient Advice and Liaison Service (PALS) by calling 023 8079 8498 or by calling into the Information Point just inside the main entrance of the General Hospital. Alternatively, you can email them at PALS@suht.swest.nhs.uk

Further information and contact details of the researcher

Kim Chivers
Doctoral Research Student
Nightingale Building (67)
School of Nursing and Midwifery
University of Southampton
Highfield Campus
Southampton
S017 1BJ

Tel: 023 8059 8203
Email: kc02@soton.ac.uk

This information sheet is for you to keep. If you decide to participate you will be asked to sign a consent form and given a copy of this to keep for your records

Thank you for taking the time to read this information sheet

Appendix 6: Opt-In Form

[To be printed on headed note paper]

Title of Project: Family Communication Following Genetic Testing

Name of Researcher: Kim Chivers

I am returning this form to indicate that I am willing to consider taking part in the above study.

I understand the researcher will contact me to ask if I have any further questions about the research and to arrange a suitable time for the interview.

Name:	
Address:	
Telephone Number:	
Most convenient time to be contacted:	

Name of Participant *(Block capitals)* | Date | Signature

When completed please return in the self-addressed envelope provided or to Kim Chivers, Nightingale Building (67), School of Nursing and Midwifery, University of Southampton, Highfield Campus, Southampton, S017 1BJ.

Appendix 7: Consent Form

[To be printed on headed note paper]

Title of Project: Family Communication Following Genetic Testing

Name of Researcher:		Study identification code:	

Please complete this form by placing your initials in the box next to each question. It is a statement that you fully understand the study and are willing to take part.

1. I confirm that I have read and understood the information sheet dated Sept 2007 (Version 5 – 1st Sep 2007) for the above study. I have had the opportunity to consider the information and ask questions, to which I have received satisfactory answers. ☐
2. I understand that my participation is voluntary and that I am free to withdraw at any time without giving any reason, without my medical care and legal rights being affected. ☐
3. I understand that data collected during the study may be looked at by individuals from the University of Southampton, from regulatory authorities or from the NHS Trust. I give permission for these individuals to have access. ☐
4. I give permission for anonymous quotes of things I say in this interview to be used in dissemination activities related to this study. ☐
5. I agree for the interview to be audio-recorded. ☐
6. I agree to take part in the above study. ☐

Name of Participant	Date	Signature (before interview)
		Signature (after interview)
Name of Person taking consent	Date	Signature

When completed: 1 for patient; 1 for researcher site file; 1 (original) to be kept in medical notes

Appendix 8: An extended example of the data matrix for four participants

Code	*Molly*	*Eloise*	Gillan Sister of 131	*Jan*
1.1: General family awareness	- Good to have other people's opinions when deciding whether or not to be tested. - some people said she shouldn't do it for her girls but for herself - not an easy decision - Evidence of hyper br awareness due to FH, passed on to daughters	- happened to mention concerns re FH to GP during routine smear - regular MRI/mammograms due to FH => family awareness & discussions	- Talked about FH w sister & "*penny dropped"*	- FH "*a bit grim"* - Thinks mother lived a high-risk life style - So has adjusted own life style to lower risk, e.g. veg, exercise, low stress, not smoking - Always lived convinced she will get br ca one day
1.2: Motivations to pursue	- final decision came when daughters said they would have test anyway - decided to be "brave"	-	-	- Mother died so young, felt at even greater risk - Small family with very little info on FH so wanted more info
1.3: Experience of cancer	- grandma, aunt, mum, sister	- mum died when 13y => huge ps impact - nan, aunt, mum, sister	- Mum br ca (52y) and then ov ca 10y later - "*tended to tolerate mummy & forget she'd been through br ca"* - She would mention check up & everyone would brush away as clear for 10 years - Never really understood she was living w fear of it coming back - Died from the ov ca, terrible quality of life w chemo & pain - Went into help w caring as needed to help & have chance to be w her	- "*all had breast ca on maternal side"* - Mum died br ca 42y - Felt mum's was very out of blue (never really understood => ↑fear) but later discovered secret letters to grandmother saying had found cysts 10y previously - Couldn't deal w mothers death, could stay in hospital & watch her die (NB Young) - Grandmother had br ca in early 60ys

2.1: Gathering details of family history	- rang Uncle for info on FH - during info gathering => people got invited - genetic team suggested "family" meeting	-	- Sister and her got referred to WGC	- Difficult to gather FH - little contact with dad after parents divorce (living in US) - "*only actually started gleaming info once relatives started to die"* - Death of maternal grandfather revealed Hungarian-jewish maternal linage which everyone had denied
2.2: Accessing DGT through PGT	- needed someone for DGT - sister got ca, she would speak to her so gen team didn't contact out of blue - "if not for yourself, for 3 sons and daughter EMOTIVE ARGUMENT - Found gene => PGT for others	- Needed sister for DGT - Found it difficult to approach as felt she was still dealing with ca diagnosis and had no partner importance of support	- Know needed mum to consent to DGT but not sure what she would say - When GC came out to explain testing, mum was so focused on her own diagnosis/treatment felt she didn't really get it was about daughters risk - When blood was taken was confused about what it was for - Just happy it wasn't about her being worse - ?Is this really informed consent	- Read that needed blood from relative w ca - Mum had died and was only child - G/mother elderly & never very well - Think she may have been DGT despite not having had ca, is this an option when testing privately??
2.3: Making decisions about GT (getting support &	- support from dad,hasband, daughters, family friend re decision - FF about so included NOT SECRETIVE - friends useful as outside family and only knew her Not so emotionally invested	- Husband often came to appointments, especially initial info and results (not test)	- Mums result came back positive so sister and her decided to be tested	- Paid to have test done privately - Discussed w wife & step father -
2.4: Learning	- explanations in "plain English" - Letters not as clear - Had not heard of BRCA before counselling	- Found out men could be at risk too - Written info helpful for processing at home - Had plan for positive results as"	All done with a lot of humour (as is normal in family) – not sure GC	- Not eligible for GT on NHS - But personally felt there wasn't enough evidence to rule out mutation in family (very few

about genetic risk	- still not clear what gene is - "cervarian" ca risk came as shock, "kick in the stomach" - All 99% sure of being carrier - Neg result hadn't entered head - Firm plans for mastectomy/hysterectomy ?mental prep for bad news, arming oneself - Awareness of risk of prostate ca (but not br ca) by hcp so specifically followed up uncle (4.2) ?because HCP didn't discuss br ca or not seen as important as prostate ca	had time to prepare"	approved - Convinced statistically one sister had to be positive - Wanted it to be her as "*can cope with it better"* than sister - Worked out in mind how to reduce risks by RRS - Worked out in mind "*worst case scenario"* - Sister said she wanted it to be both of them or neither ("*She hadn't really looked at the percentages")* Own	-
2.5: Materials to support	- Daughter printed off extra info from websites but couldn't understand	- Trusted websites only	-	-
3.1: Telling those who are waiting to know	- 1. Husband present, 2. Rang daughters outside hospital ("phones ready"), 3. Went to ward to tell sister (DGT), 4. dad came round when got home as waiting	- Brother knew going so waiting to hear - All over phone Normal CP - Reassured knowing everyone had "someone" – importance of support	- Told daughters on the phone asap "*stop worrying, I'm not carrying the gene"*	- All waiting for results - Told step-parents straight away - Such a relief - NB very small family so hardly anyone to tell
3.2: Keeping those in the know in the	- "difficult sister" and cousin not "in our club" so no rush to tell NORMAL PATTERNS OF COMMUNICATION	-	- Telephoned brothers and GC sent a letter with details	
3.3: Personal reaction to results and telling people	- Guilt re: other positive family vs. relief daughters were fine - "still got to go through it" with sisters - Knew she would feel bad about being neg before got results	- Brief "crumble" at result - Though positive result answered so many questions (re mum etc) - Would have been worse to have been told got ca - ↑ contact w brother during process but now back to normal - SinL very neg, had to deal with that on top of all else	- Sister wanted a joint appointment - She didn't, saw her role as support, so secretly rang GC for her results the week before - Neg result => "*shit, that means sister has it"* - Thinks it would have been better if GC had given result to each on paper rather than verbally one after the other - **Sister said she saw her relief she was neg, but that's not true as she already knew & was just sad her sis was pos**	- Received neg result by letter - Very surprised but please. - Not really thought about it since - **Has changed feelings about ca, now thinking she may not get it as opposed to all consuming fear she would**

4.1 Distant relatives	- Rang uncle re: results as had more contact with him that sisters - Prompted to do so by HCP and reinforced by mention in letter !!! - Told him to tell cousins - No follow up though ?not emotionally invested	- Sister spoke to cousin as although not close has slightly more contact = nominated person - Cousin doesn't want test, causes worry as has two daughters but not following up ?not enough emotionally invested		Knows of some paternal cousins but never been in contact Wrote to her father so he could pass on details -
4.2: Those who did not want to know	-	-	- All agreed they wouldn't tell mum so she wouldn't worry **(See notes about her confusion in 2.2)** - But Sister actually did. No huge reaction but thinks she doesn't really understand the implications especially for brothers or grandkids - Would never talk to brothers kids without his consent - "*too young and not my place"* - "*you run the risk of upsetting the family and blowing it apart"* - *"I wouldn't want someone to tell my girls and I work on that principle"* - However if by the time she's 25 they've not told her, "*I would get one of the cousins to tell her! I wont do it but I will get one of my girls to tell her by accident!"*	- Estranged brother - Step father acts as a go between and may have mentioned it Not made any effort to tell him about testing as "*<u>suppose I didn't think of him as a likely candidate fro br ca because I think of it more of an female thing</u>"*
4.3: **Children** **Talking to male relatives**		- Children too young, (10,15) wait until age when they can cope w info - There's plenty of time - Don't want to ruin childhood by something they wont understand yet anyway - SinL told her children (n&n) vague concern a cousin will tell her children - Will revisit gen team for more info when time comes	- Its not really about the grandchildren at the moment (they've got 10y before they need to worry). - At the moment its about sister (pos), she is "*carrying all this"*	-

Appendix 9: Number of individuals and frequency of scores of cohesion given by each participant during construction of Eco-maps.

	Total	Cohesion Score					Deceased
		3	2	1	0	Disrupted	
114							
First degree relatives	3	0	0	2	0	1	0
Second degree relatives	0	0	0	0	0	0	0
Third and fourth degree relatives	1	1	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	1	0	0	0	1	0	0
Friends & work colleagues	3	1	2	0	0	0	0
Total	8	2	2	2	1	1	0

125							
First degree relatives	5	3	1	1	0	0	0
Second degree relatives	5	0	0	0	5	0	0
Third and fourth degree relatives	3	0	3	0	0	0	0
Non-blood relatives (including partners & in-laws)	1	1	0	0	0	0	0
Friends & work colleagues	4	2	1	1	0	0	0
Total	18	6	5	2	5	0	0

127							
First degree relatives	9	1	6	2	0	0	0
Second degree relatives	9	0	0	0	9	0	0
Third and fourth degree relatives	1	0	0	1	0	0	0
Non-blood relatives (including partners & in-laws)	2	1	0	0	1	0	0
Friends & work colleagues	8	2	2	3	1	0	0
Total	29	4	8	6	11	0	0

131							
First degree relatives	7	5	1	1	0	0	-1
Second degree relatives	0	0	0	0	0	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	1	1	0	0	0	0	0
Friends & work colleagues	4	1	1	2	0	0	0
Total	12	7	2	3	0	0	-1

110							
First degree relatives	6	2	0	1	0	0	3
Second degree relatives	4	0	0	0	4	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	4	0	1	0	3	0	0
Friends & work colleagues	4	1	3	0	0	0	0
Total	18	3	4	1	7	0	3

118							
First degree relatives	5	3	1	1	0	0	0
Second degree relatives	0	0	0	0	0	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	1	1	0	0	0	0	0
Friends & work colleagues	10	1	4	5	0	0	0
Total	16	5	5	6	0	0	0

111							
First degree relatives	3	1	1	0	0	1	0
Second degree relatives	2	0	0	0	2	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	1	1	0	0	0	0	0
Friends & work colleagues	0	0	0	0	0	0	0
Total	6	2	1	0	2	1	0

123							
First degree relatives	1	0	0	0	0	1	0
Second degree relatives	2	0	1	1	0	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	5	1	1	2	1	0	0
Friends & work colleagues	3	1	2	0	0	0	0
Total	11	2	4	3	1	1	0

119							
First degree relatives	5	2	1	0	0	1	1
Second degree relatives	4	0	0	2	2	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	5	1	2	2	0	0	0
Friends & work colleagues	2	0	0	2	0	0	0
Total	16	3	3	6	2	1	1

129							
First degree relatives	8	5	0	1	0	1	1
Second degree relatives	4	0	0	2	2	0	0
Third and fourth degree relatives	1	0	0	0	1	0	0
Non-blood relatives (including partners & in-laws)	7	3	0	2	2	0	0
Friends & work colleagues	11	5	6	0	0	0	0
Total	31	13	6	5	5	1	1

117							
First degree relatives	5	3	0	0	0	2	0
Second degree relatives	2	0	0	0	1	1	0
Third and fourth degree relatives	1	0	0	0	1	0	0
Non-blood relatives (including partners & in-laws)	0	0	0	0	0	0	0
Friends & work colleagues	3	1	1	1	0	0	0
Total	11	4	1	1	2	3	0

130							
First degree relatives	7	1	4	1	0	1	0
Second degree relatives	5	0	1	0	4	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	3	1	1	0	1	0	0
Friends & work colleagues	4	0	2	2	0	0	0
Total	19	2	8	3	5	1	0

126							
First degree relatives	8	1	6	0	0	1	0
Second degree relatives	6	0	1	2	3	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	3	1	0	1	1	0	0
Friends & work colleagues	1	0	1	0	0	0	0
Total	18	2	8	3	4	1	0

115							
First degree relatives	8	1	5	0	2	0	0
Second degree relatives	4	0	0	0	4	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	3	1	0	2	0	0	0
Friends & work colleagues	3	3	0	0	0	0	0
Total	18	5	5	2	6	0	0

112							
First degree relatives	5	3	2	0	0	0	-1
Second degree relatives	13	0	0	0	13	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	3	0	1	1	1	0	0
Friends & work colleagues	6	0	4	1	1	0	0
Total	27	3	7	2	15	0	-1

113							
First degree relatives	6	3	0	1	1	1	0
Second degree relatives	7	0	0	0	7	0	0
Third and fourth degree relatives	1	0	0	0	1	0	0
Non-blood relatives (including partners & in-laws)	1	1	0	0	0	0	0
Friends & work colleagues	9	7	2	0	0	0	0
Total	24	11	2	1	9	1	0

122							
First degree relatives	5	2	2	0	0	0	1
Second degree relatives	3	0	0	0	3	0	0
Third and fourth degree relatives	3	0	0	0	3	0	0
Non-blood relatives (including partners & in-laws)	13	2	2	3	6	0	0
Friends & work colleagues	8	2	3	3	0	0	0
Total	32	6	7	6	12	0	1

101							
First degree relatives	5	2	2	1	0	0	0
Second degree relatives	5	0	1	0	4	0	0
Third and fourth degree relatives	2	0	0	0	2	0	0
Non-blood relatives (including partners & in-laws)	1	1	0	0	0	0	0
Friends & work colleagues	4	1	0	3	0	0	0
Total	17	4	3	4	6	0	0

106							
First degree relatives	6	0	0	6	0	0	0
Second degree relatives	2	0	0	0	0	2	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	4	0	3	1	0	0	0
Friends & work colleagues	1	1	0	0	0	0	0
Total	13	1	3	7	0	2	0

102							
First degree relatives	5	2	1	1	0	0	1
Second degree relatives	4	0	0	0	3	0	1
Third and fourth degree relatives	4	1	0	1	2	0	0
Non-blood relatives (including partners & in-laws)	2	1	1	0	0	0	0
Friends & work colleagues	2	1	1	0	0	0	0
Total	17	5	3	2	5	0	2

105							
First degree relatives	4	1	1	1	0	0	1
Second degree relatives	5	0	0	0	1	0	4
Third and fourth degree relatives	3	0	1	0	1	0	1
Non-blood relatives (including partners & in-laws)	2	1	1	0	0	0	0
Friends & work colleagues	2	1	1	0	0	0	0
Total	16	3	4	1	2	0	6

103							
First degree relatives	9	2	1	0	0	1	5
Second degree relatives	10	0	0	1	9	0	0
Third and fourth degree relatives	17	0	0	1	16	0	0
Non-blood relatives (including partners & in-laws)	7	1	1	0	5	0	0
Friends & work colleagues	1	0	1	0	0	0	0
Total	44	3	3	2	30	1	5

107							
First degree relatives	4	1	2	1	0	0	0
Second degree relatives	5	0	0	0	5	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	1	1	0	0	0	0	0
Friends & work colleagues	3	0	1	2	0	0	0
Total	13	2	3	3	5	0	0

108							
First degree relatives	4	1	3	0	0	0	0
Second degree relatives	1	0	0	1	0	0	0
Third and fourth degree relatives	2	0	1	0	1	0	0
Non-blood relatives (including partners & in-laws)	3	1	2	0	0	0	0
Friends & work colleagues	3	1	2	0	0	0	0
Total	13	3	8	1	1	0	0

109							
First degree relatives	5	1	2	2	0	0	0
Second degree relatives	2	0	0	0	2	0	0
Third and fourth degree relatives	0	0	0	0	0	0	0
Non-blood relatives (including partners & in-laws)	1	1	0	0	0	0	0
Friends & work colleagues	2	0	0	2	0	0	0
Total	10	2	2	4	2	0	0

124							
First degree relatives	0	0	0	0	0	0	0
Second degree relatives	1	0	0	0	1	0	0
Third and fourth degree relatives	7	1	4	1	1	0	0
Non-blood relatives (including partners & in-laws)	0	0	0	0	0	0	0
Friends & work colleagues	3	1	1	1	0	0	0
Total	11	2	5	2	2	0	0

	Total	Cohesion score					Deceased
		3	2	1	0	Disrupted	
All participants							
First degree relatives	**138**	46	42	23	3	11	11
Second degree relatives	**105**	0	4	9	84	3	5
Third and fourth degree relatives	**46**	3	9	4	29	0	1
Non-blood relatives (including partners & in-laws)	**75**	23	16	14	22	0	0
Friends & work colleagues	**104**	33	41	28	2	0	0
Total	**468**	**105**	**112**	**78**	**140**	**14**	**17**

18

All participants – average							
First degree relatives	**5.3**	**1.8**	**1.6**	**0.9**	**0.1**	**0.4**	**0.4**
Second degree relatives	**4.0**	**0.0**	**0.2**	**0.3**	**3.2**	**0.1**	**0.2**
Third and fourth degree relatives	**1.8**	**0.1**	**0.3**	**0.2**	**1.1**	**0.0**	**0.0**
Non-blood relatives (including partners & in-laws)	**2.9**	**0.9**	**0.6**	**0.5**	**0.8**	**0.0**	**0.0**
Friends & work colleagues	**4.0**	**1.3**	**1.6**	**1.1**	**0.1**	**0.0**	**0.0**
Total	**18.0**	**4.0**	**4.3**	**3.0**	**5.4**	**0.5**	**0.7**

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