Communication about genetic testing and hereditary cancer management with breast and ovarian cancer patients

Christine June Jacobs

University College London
Department of Clinical, Educational & Health Psychology
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DECLARATION

I, Christine June Jacobs, confirm that the work presented in this thesis is my own. Where information has been derived from other sources, I confirm that this has been indicated in the thesis. My work was funded by a National Institute for Health Research Doctoral Research Fellowship.

This work was carried out at the Health Psychology Research Centre, in the Department of Clinical, Educational, and Health Psychology, University College London, under the supervision of Professor Susan Michie. Professor Stephen Pilling was my second supervisor. Expert supervision was also provided by Professor Jonathan Smith, from Birkbeck University, for Study 5, Chapter 7. Additional support is noted in the acknowledgements.

Edited versions of Chapter 3 (Jacobs et al, 2015) and Chapter 4 (Jacobs et al, 2017) have been published in peer-reviewed professional journals. The results from Study 1, Chapter 3 have also been presented at the UK Society of Behavioural Medicine meeting in December 2012 and the UK/Dutch Clinical Genetics Societies and Cancer Genetics Group meeting in March 2014.

Correspondence concerning this thesis should be addressed to Chris Jacobs,
cj Jacobs@btinternet.com

Signed: Date: 08/01/18

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ABSTRACT

Background: Women with breast or ovarian cancer (patients) are increasingly offered genetic testing shortly after diagnosis to guide management and identify future cancer risks. As a result, new approaches to communication about hereditary cancer are needed to inform decision-making amongst patients and their relatives.

Aims: This thesis aimed to investigate expert opinion and guideline recommendations about the communication needs of patients undergoing genetic testing, the information communicated by genetics health professionals and recalled by patients and their relatives and the experience of patients with newly diagnosed breast cancer who undergo genetic testing.

Methods: A scoping review and five studies were conducted using UK data from patients and health professionals: an observational study, a Delphi survey, a content analysis, a document analysis and mixed methods matrix and a qualitative study.

Results: Accuracy of information recall was low amongst patients and relatives following genetic counselling, especially about hereditary cancer management. Expert health professionals and service users agreed on the key messages required by patients to inform decision-making for themselves and their relatives. However, during genetic counselling, half of the key messages were communicated to patients and fewer key messages were communicated about hereditary cancer management than genetic testing. Recommended information to communicate to patients was identified from international genetics guidelines. Recommendations about hereditary cancer management were infrequently reflected in expert opinion, communicated by genetics health professionals or recalled by patients. Newly diagnosed patients experienced growing uncertainty and concern about inheritance and surgical decision-making, especially when their genetic test result was unexpected.

Conclusions: Patients received insufficient information to understand their future cancer risks and make informed decisions about managing hereditary cancer. Steps are needed to improve information provision, especially when results are unexpected. These findings can help guide refinements to communication as genetic testing is integrated into mainstream oncology.
IMPACT STATEMENT

The impact of this work for policy, practice and research is detailed in Chapter 8 and summarised below.

For policy, the work has the potential to influence genetics and cancer management guidelines by highlighting the information and communication needs of breast and ovarian cancer patients about hereditary cancer and the importance of ensuring that guidelines are relevant and accessible to genetics and oncology health professionals. The findings highlight the importance of educating oncology health professionals about genetics and genetics health professionals about communicating with cancer patients.

The thesis will have implications for change in practice as genetics is integrated into the clinical care pathway in oncology and the 100,000 Genome Project starts to return results to health professionals. The work highlights the importance of communicating key messages and only communicating supplementary messages when they are relevant to the individual or there is the time to do so. The findings demonstrate the need for targeted written information for patients about hereditary cancer management. The thesis suggests that a coordinated multidisciplinary approach to communication about genetic testing is important to ensure that patients who may benefit from enhanced counselling and support are identified prior to testing and those who may benefit from expertise in hereditary cancer management are identified after a pathogenic variant has been found. The work draws attention to the need for genetic counselling to focus on helping cancer patients to understand information in the context of their own risk, prognosis and treatment options, providing psychological support and facilitating dissemination of information within families.

The methods developed for this thesis could be applied to other areas of healthcare research. This work highlights a need for further investigation into the following areas: the communication and support needs of patients with other types of cancer undergoing genetic testing; communication about genetic testing and hereditary cancer management by oncology health professionals; health professionals’ understanding and communication about variants of uncertain significance; the extent to which patients recall information communicated via leaflets; and the development of measures to enhance the use of clinical guidelines to improve interdisciplinary working and provide patients and families with the information they need.
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1.1. INTRODUCTION

Traditionally genetic testing for hereditary cancer has only been available following genetic counselling by specialist genetics health professionals, including medically trained clinical geneticists and non-medical genetic counsellors from a nursing or science background with a Master level degree in genetic counselling. Patients with cancer were generally offered genetic testing after completing definitive cancer treatment, often at the request of relatives without cancer. As a result, genetic counselling research and clinical guidelines have tended to focus on the communication needs of those at risk of hereditary cancer with less attention paid to cancer patients.

The integration of genetic and genomic medicine into mainstream oncology to improve care and save treatment costs is highlighted as a priority in the annual report of the chief medical officer in the United Kingdom (UK) (Davies, 2017). The impact of early identification of BRCA status on cancer treatment has already been demonstrated for breast and ovarian cancer patients (Fong et al., 2009; Ledermann et al., 2014). As a result of these developments, there is an urgent need to increase the availability of rapid and timely genetic testing for patients who are eligible. Analysis of United States of America (USA) national cancer survey data found that only 14.1% of breast and ovarian cancer survivors who were eligible for genetic testing had been tested (Childers, Childers, Maggard-Gibbons, & Macinko, 2017), suggesting a huge number of patients who may yet benefit from testing and who need information about hereditary cancer.

Genetics services do not have the capacity to cope with the high demand for genetic testing of cancer patients (Barlow-Stewart, 2015; Slade, Riddell, Turnbull, Hanson, & Rahman, 2015). As it is no longer feasible for all cancer genetic counselling to be provided by specialist genetics health professionals, oncology health professionals will increasingly be required to communicate with cancer patients about genetic testing and hereditary cancer management.

This thesis investigated existing genetic counselling practice in communication with breast and ovarian cancer patients about BRCA1/BRCA2 genetic testing and the
management of the hereditary cancers associated with pathogenic variants in these
genomes. The purpose of this research was to inform future practice, policy and research
in communication about hereditary cancer with breast and ovarian cancer patients.

1.2. GENETIC COUNSELLING

The role of genetic counselling is to enable discussion of the implications of a family
history of a genetic condition, providing information and support that empowers
individuals to reach the most appropriate decisions for themselves and their families
(Shiloh S., 1996). Genetic counselling is based on the humanist, client-centred
psychotherapeutic approach of Carl Rogers (Veach, Bartels, & LeRoy, 2007). Although
there are psychotherapeutic elements to genetic counselling, for example in the
provision of short-term client-centred counselling, empathic listening and family
dynamics, the profession has tended to steer away from psychotherapy and to adopt
more of a psychosocial approach to counselling (Austin, Semaka, & Hadjipavlou,
2014).

The term ‘genetic counselling’ was first introduced by Sheldon Reed in the 1940s in his
work with parents of children with intellectual disabilities. By the 1950s hereditary
clinics were being established in the USA to counsel patients with a family history of
genetic disease and couples whose children had been born with serious congenital
defects. The concept of non-directiveness was advocated by many geneticists at that
time in acknowledgement of the complexity of the reproductive decision-making and to
avoid the eugenic connotations associated with the Nazi era (Resta, 1997). Non-
directiveness aims to promote the self-determination or autonomy of the individual being
c counselled (Biesecker, 2001) and continues to be a guiding principle in genetic
counselling practice. Several authors however have noted that this principle is not
always adhered to (Michie, Bron, Bobrow, & Marteau, 1997; van Zuuren, 1997) and the
efficacy and appropriateness of non-directiveness for genetic counselling have been
questioned, especially in the context of cancer genetics where evidence-based
targeted treatment, effective risk-reducing options and established surveillance
methods are becoming increasingly available (Koch & Nordahl Svendsen, 2005; Weil,
2003). Partly in response to this debate, the Task Force of the National Society of
Genetic Counsellors developed a new definition of genetic counselling. The definition
does not include the concept of non-directiveness but instead highlights the
educational and facilitative aspects alongside the importance of addressing
psychological issues: ‘Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following: interpretation of family and medical histories to assess the chance of disease occurrence or recurrence; education about inheritance, testing, management, prevention, resources and research; counseling to promote informed choices and adaptation to the risk or condition’ (Resta et al., 2006 p.274).

Communicating information and providing support are at the core of genetic counselling, as reflected by international guidelines and policy documents (Rantanen et al., 2008). There has been much discussion within the profession about the balance of the ‘teaching’ and ‘counselling’ models of genetic counselling. A seminal paper by Seymour Kessler discussed the clear distinction between the goals, philosophies and communication styles associated with each of these approaches (Kessler, 1997). The teaching model is goal-directed, involving the presentation of biomedical information and interpretation of genetic events in an attempt to educate the counsellee. The counselling model is client-centred, with the goals being to empower the individual, foster autonomy and promote competency. Although genetic counselling aims to combine teaching and counselling (Biesecker, 2001) and to some extent achieves this (Ellington et al., 2006), several studies have identified the teaching model as dominant (Meiser, Irle, Lobb, & Barlow-Stewart, 2008; Roter et al, 2006). The heavy clinical caseload and extensive and complex nature of genetic information can result in information-giving becoming the primary focus of genetic counselling consultations at the expense of psychological support (Biesecker B., 2003; J. E. Hartmann, Veach, MacFarlane, & LeRoy, 2015).

Pre- and post-test genetic counselling is usually provided by genetic counsellors and clinical geneticists within the clinical genetics setting. Cancer genetic counselling involves identifying hereditary cancer through family history assessment and genetic testing, explaining inheritance, interpreting genetic test results and facilitating decision-making about risk management options. Genetic counselling also involves facilitating the sharing of genetic risk information within families and providing counselling and support for individuals and families as they come to terms with a diagnosis of hereditary cancer and make decisions about genetic testing, risk management and reproductive options.
1.3. GENES INVOLVED IN HEREDITARY BREAST AND OVARIAN CANCER

Hereditary breast and ovarian cancers are associated with several hereditary cancer syndromes involving high and intermediate penetrance genes. Penetrance is defined as, ‘The production of the expected phenotype by a particular gene or allele; the frequency with which organisms having a particular genotype show the expected phenotype’ (Oxford English Dictionary, 2005). Table 1.1 shows the genes associated with breast and ovarian cancer and the associated cumulative cancer risks.

The most common of the high penetrance breast and ovarian cancer genes are \textit{BRCA1} and \textit{BRCA2}. The \textit{BRCA1} gene is located on chromosome 17q21 and was identified in 1994 (Miki et al.). The \textit{BRCA2} gene is located on chromosome 13q12.3 and was identified in 1995 (Wooster et al.). Both are tumour suppressor genes that maintain genomic stability by facilitating repair of DNA double strand breaks. Pathogenic variants (or mutations) in the \textit{BRCA1}/\textit{BRCA2} genes are autosomal dominantly inherited, so that each child of a carrier has 50\% risk of inheriting the same pathogenic variant. (See section 1.8.5.2 for an explanation of the terminology used throughout this thesis to discuss genetic test results.)

The prevalence of \textit{BRCA1}/\textit{BRCA2} pathogenic variants in the general population is estimated to be 1 in 300 and 1 in 800, respectively (Hampel, Bennett, Buchanan, Pearlman, & Wiesner, 2015). Depending on the population studied, pathogenic variants in \textit{BRCA1} and \textit{BRCA2} are estimated to account for up to 20\% of breast cancer in women (Anglian Breast Cancer Study, 2000; Evans et al., 2011; Wong-Brown et al., 2015) and up to 17\% of high-grade serous ovarian cancers (Alsop et al., 2012; Pal et al., 2005).

Up to 2.5\% of Ashkenazi Jewish individuals are estimated to carry one of three common founder pathogenic variants in the \textit{BRCA1} and \textit{BRCA2} genes that predispose to hereditary breast and ovarian cancer (Hartge, Struwing, Wacholder, Wacholder, Brody & Tucker, 1999; Struwing et al. 1997). Amongst Ashkenazi Jewish women with breast cancer, the prevalence of these founder pathogenic variants is estimated to be up to 10\% (King, Marks, & Mandell, 2003) and in women with ovarian cancer, up to 41\% (Moslehi et al., 2000). Founder pathogenic variants exist in other populations where the prevalence is also increased (Ferla et al., 2007).
Table 1.1. High and intermediate penetrance genes associated with hereditary breast and ovarian cancer showing cancers associated with each gene and the risks for breast (BC) and ovarian cancer (OC) only

<table>
<thead>
<tr>
<th>Types of cancer</th>
<th>Syndrome</th>
<th>Genes</th>
<th>Breast/ovarian cancer risk to age 80 except where shown</th>
</tr>
</thead>
<tbody>
<tr>
<td>Breast cancer</td>
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<tr>
<td>Ovarian cancer</td>
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<td>Prostate cancer</td>
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<tr>
<td>Male breast cancer</td>
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<tr>
<td>Breast cancer</td>
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<tr>
<td>Ovarian cancer</td>
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<tr>
<td>Childhood sarcoma</td>
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<tr>
<td>Adrenocortical carcinoma</td>
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<td>Leukaemia</td>
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<tr>
<td>Brain tumours</td>
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<tr>
<td>Prostate cancer</td>
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<td>Male breast cancer</td>
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<td>Breast cancer</td>
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<td>Childhood cancer</td>
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<tr>
<td>Renal cell cancer</td>
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<tr>
<td>Endometrial cancer</td>
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<tr>
<td>Colorectal cancer</td>
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<tr>
<td>PTEN</td>
<td></td>
<td></td>
<td>BC up to 85% (Tan et al., 2012)</td>
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<tr>
<td>PALB2</td>
<td></td>
<td></td>
<td>BC up to 58% by age 70 (Antoniou et al., 2014; Rahman et al., 2007)</td>
</tr>
<tr>
<td>CDH1</td>
<td></td>
<td></td>
<td>BC 42% (Pharoah, Guilford, Caldas, &amp; International Gastric Cancer Linkage, 2001; van der Post et al., 2015)</td>
</tr>
<tr>
<td>ATM</td>
<td></td>
<td></td>
<td>BC 27% up to age 80 (Renwick et al., 2006; Thompson et al., 2005)</td>
</tr>
<tr>
<td>NF1</td>
<td></td>
<td></td>
<td>BC 26% up to age 50 (Madanikia, Bergner, Ye, &amp; Blakeley, 2012; Sharif et al., 2007)</td>
</tr>
<tr>
<td>STK11</td>
<td></td>
<td></td>
<td>BC 29% (The CHEK Breast Cancer Consortium, 2002)</td>
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<td>BC 29% (The CHEK Breast Cancer Consortium, 2002)</td>
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1.4. GENETIC TESTING FOR HEREDITARY BREAST AND OVARIAN CANCER

Until recently, diagnostic genetic testing of cancer patients involved consecutive testing of single genes using Sanger technology (Sanger, Nicklen, & Coulson, 1977). Many laboratories now use a combination of Sanger sequencing and next-generation sequencing, or massive parallel sequencing, to simultaneously test for panels of multiple genes, thus speeding up the process of genetic testing and increasing the likelihood of finding a pathogenic variant (Crawford et al., 2017). The decision to offer testing of multiple genes or just the BRCA1 and BRCA2 genes depends on the personal and family cancer history. (See section 1.8.5.1 to explain the context of the focus on BRCA1/BRCA2 genetic testing in this thesis.)

Ideally, before genetic testing is offered to a person without cancer (a predictive test), a pathogenic variant first needs to be identified in a family member with the relevant cancer (a diagnostic test). In the UK, direct gene testing may occasionally be offered to high risk individuals without cancer who have at least 10% chance of a pathogenic variant when there is no living relative with cancer available to test (National Institute for Health and Care Excellence, 2013). To inform both the patient and at-risk relatives, it is therefore important to identify those who are eligible for genetic testing.

Although 3 to 4% of cancer patients at high risk of hereditary breast and ovarian cancer have a pathogenic variant in a rare cancer predisposing gene (Desmond et al., 2015; Tung et al., 2016), most hereditary breast and ovarian cancer is associated with pathogenic variants in the BRCA1 and BRCA2 genes. In the UK, diagnostic genetic testing of the BRCA1 and BRCA2 genes is offered to cancer patients when there is a 10% or greater chance of identifying a pathogenic variant (National Institute for Health and Care Excellence, 2013).

There are three possible outcomes of diagnostic testing: a pathogenic variant is detected, a variant of uncertain significance (VUS) is detected or no variant is detected.

When a pathogenic variant is detected, the risk of future cancers can be accurately estimated for the patient and options are available for risk management. Each first-degree relative (child, sibling, parent) of the patient has a 50% risk of having inherited the same pathogenic variant and can be offered predictive testing.

A VUS is a variation in the coding sequence of a gene where the functional effect of the sequence change is unknown. There are classification systems for some genes
and some databases are available to monitor variants that have previously been reported, although the data are sketchy. There is a large variation in the frequency of BRCA1/BRCA2 VUS reports from different laboratories depending on the prevalence of the population tested. The reported rate amongst the African American population is up to 21%, 5-6% in those of European ancestry in USA laboratories and 15% in Europe (Lindor, Goldgar, Tavtigian, Plon, & Couch, 2013; Ready, Gutierrez-Barrera et al., 2011).

Most cancer patients who undergo testing do not have a pathogenic variant or a VUS. This is sometimes called an ‘inconclusive’ or ‘uninformative result’. This result is not a true negative. There remains the possibility that the cancer could be due to another untested or undiscovered cancer predisposing gene or that the cancer is due to a combination of multiple genes of minor effect (polygenic risk). The probability that the cancer is due to a pathogenic variant in the BRCA1/BRCA2 genes is markedly reduced by this result, although the degree of reassurance about hereditary cancer will depend on whether single gene testing or multi-gene panel testing has been undertaken.

In the absence of a pathogenic variant, future cancer risk assessment is based on the age at diagnosis and the likelihood of other cancer-predisposing genes. The decision to offer more extensive treatment to a cancer patient in the absence of a pathogenic variant is generally based on the age at diagnosis and the extent of the family history. Generally, predictive testing and risk-reducing surgery is not offered to relatives of patients with a VUS, although this may occasionally be discussed depending on the interpretation and classification of the variant. For patients in whom a pathogenic variant is detected, options are available to reduce the risk of future cancers and to detect cancer early (see section 1.7).

1.5. PSYCHOLOGICAL ASPECTS OF GENETIC COUNSELLING AND GENETIC TESTING FOR HEREDITARY BREAST AND OVARIAN CANCER

1.5.1. At-risk women

Research into the psychological aspects of genetic counselling and genetic testing for hereditary breast and ovarian cancer has tended to focus on at-risk women. Perception of risk amongst women with a family history of breast cancer is often inaccurate with many over-estimating their risk (Lerman, Kash, & Stefanek, 1994). The world-wide response to the disclosure of a genetic predisposition to breast cancer by Angelina
Jolie in 2013 highlights the concern experienced by women with a breast cancer family history. Ms Jolie’s announcement led to a huge increase in referrals for genetic counselling, mostly from women at low or moderate risk of breast cancer (Evans et al., 2014). For women at risk of hereditary breast and ovarian cancer, genetic counselling and genetic testing has psychological benefits and improves the accuracy of risk perception (Butow, Lobb, Meiser, Barratt, & Tucker, 2003).

1.5.2. Patients tested following cancer treatment

The uptake of genetic testing has consistently been shown to be greater amongst patients with cancer than those at risk (Meiser, 2005). The motivation for genetic testing differs amongst at-risk women who undergo predictive genetic testing and those with cancer who have a diagnostic test. At-risk women are often motivated by concern for their own health and a desire to initiate breast screening (Press, Yasui, Reynolds, Durfy, & Burke, 2001). Prior to the availability of genetic testing to guide cancer treatment, cancer patients were reported to be motivated to undergo testing primarily by a wish to find out the cancer risk for their children but also to understand the cause of their diagnosis (Julian-Reynier et al., 1998). Patients undergoing genetic testing to guide cancer management are primarily motivated by the desire to inform treatment options as well as to protect the family (Gleeson et al., 2013; Meiser et al., 2012; Zilliacus, 2012).

Several studies have reported no increase in distress levels amongst cancer patients following genetic counselling and testing (Lerman et al., 1996; Reichelt, Heimdal, Moller, & Dahl, 2004; Schwartz et al., 2002). Several studies of the psychological impact of genetic testing within one year of breast cancer diagnosis found high levels of breast cancer distress following disclosure of a pathogenic variant result (van Roosmalen et al., 2004; Bonadona et al., 2002). Patients' understanding and interpretation of genetic test results are affected by their personal experiences of cancer rather than the information communicated during genetic counselling (Hallowell, Foster, Eeles, Arderne-Jones, & Watson, 2004; Vos et al., 2011).

Studies have shown that cancer patients need support throughout the process of genetic testing and results disclosure (Farrelly et al., 2013; Augestad, Høberg-Vetti, Bjorvatn, & Sekse, 2017). Some patients experience increased anxiety and depression following genetic counselling, requiring additional support (Nordin et al., 2011). However, although patients with a pathogenic variant experience negative psychological well-being with feelings of distress, anxiety and depression immediately
after disclosure of genetic test result, clinically relevant symptoms do not persist in the intermediate or longer term for most patients (Ringwald et al., 2016).

1.5.3. Patients tested shortly after diagnosis to guide cancer treatment
Prior to the availability of targeted cancer treatment, genetic testing shortly after diagnosis was considered unacceptable and overwhelming by cancer patients and oncology health professionals (Ardern-Jones, Kenen, & Eeles, 2005). Over time, however, with the availability of PARP inhibitors and greater awareness of the risk benefit of contralateral mastectomy at the time of definitive surgery, attitudes have changed. Recent studies have found that testing shortly after breast or ovarian cancer diagnosis is feasible and acceptable to patients (Katz, Kurian, & Morrow, 2015; Sie et al., 2014; Ziliaciuc, 2012) with no increased psychological morbidity (Christie et al., 2012; Wevers et al., 2017; Wevers et al., 2012). However, the number of patients with a pathogenic variant in these studies is small and further evaluation of the short and long term psychological impact of this type of testing is needed before conclusions can be drawn.

1.6. CANCER RISKS ASSOCIATED WITH PATHOGENIC VARIANTS IN THE BRCA1/BRCA2 GENES

1.6.1. Breast cancer
Retrospective studies have estimated the cumulative risk of breast cancer to age 70 years to be between 40% and 80% for BRCA1 carriers and between 27% and 84% for BRCA2 carriers (Antoniou et al., 2003). The results of an international prospective cohort study have recently been reported (Kuchenbaecker et al., 2017). The study included 6036 BRCA1 carriers and 3820 BRCA2 carriers from the United Kingdom, Europe, Scandinavia, the USA, Canada, Australia and New Zealand. Data from 4810 carriers with cancer and 5046 carriers without cancer were included. The peak incidence in breast cancer for BRCA1 carriers was between 41 and 50 years. For BRCA2 carriers, the peak incidence was between 51 and 60 years (Kuchenbaecker et al., 2017). The cumulative risk of breast cancer to age 80 was 72% (95% CI, 65%-79%) for BRCA1 and 69% (95% CI, 61%-77%) for BRCA2 carriers. Pathogenic variants in the BRCA1/BRCA2 genes are not thought to increase the risk of recurrent or metastatic disease (Wesolowski R, 2007).
BRCA1/BRCA2 carriers with breast cancer have up to 68% cumulative risk of developing a further primary breast cancer and the risk is significantly higher in women diagnosed with breast cancer before the age of 40 than in those diagnosed after the age of 50 (Antoniou et al., 2003; Kuchenbaecker et al., 2017; Metcalfe et al., 2011). In the 20 years after a primary breast cancer diagnosis, the cumulative risk of contralateral breast cancer is 40% for BRCA1 carriers and 26% for BRCA2 carriers (Kuchenbaecker et al., 2017).

The estimated cumulative risk of breast cancer up to age 70 years associated with the two Ashkenazi Jewish BRCA1 founder mutations is 65%. The breast cancer risk associated with the BRCA2 founder mutation is 43% (Antoniou et al., 2005).

Breast cancers associated with pathogenic variants in the BRCA1 gene display characteristic pathological features. These tumours are generally higher grade than sporadic tumours and often oestrogen, progesterone and HER-2 negative (triple negative) (Lakhani et al., 2002). Most BRCA1 breast tumours express basal cytokeratins and are classed as being of a basal subtype (Lakhani et al., 2005). Up to 20% of women diagnosed with triple negative breast cancer carry a BRCA1 pathogenic variant, although figures vary between countries and studies (Evans et al., 2011; Wong-Brown et al., 2015). BRCA2-related breast tumours are more heterogeneous, closely resembling sporadic breast cancer (Mavaddat et al., 2012).

1.6.2. Ovarian cancer

The cumulative risk of ovarian and tubal cancer has been estimated at 22% to 65% for BRCA1 carriers and 10% to 35% for BRCA2 carriers (Mavaddat et al., 2013). The peak incidence of ovarian cancer is reported to be between the ages of 61 and 70 for BRCA1 and BRCA2 carriers (Kuchenbaecker et al., 2017). The prospective study found the cumulative risk of ovarian and fallopian tube cancer to age 80 years to be 44% (95% CI, 36%-53%) for BRCA1 and 17% (95% CI, 11%-25%) for BRCA2 carriers (Kuchenbaecker et al., 2017).

The estimated cumulative risk of developing ovarian cancer up to age 70 years for the two BRCA1 Ashkenazi Jewish founder mutations is 14% to 33%. For the BRCA2 founder mutation the estimated cumulative risk is 20% (Antoniou et al., 2005).

In women with a BRCA1/BRCA2 pathogenic variant, ovarian tumours are mostly high-grade serous epithelial cancers (Lakhani et al., 2004; Mavaddat et al., 2012). It is
thought that most ovarian cancers originate at the distal end of the fallopian tube (Dubeau, 2008).

1.6.3. Male breast cancer

Male breast cancer is rare, comprising less than 1% of all male cancers. Due to the small sample sizes in studies, the reported frequencies of \( BRCA2 \)-related male breast cancer vary widely, ranging from 3% to 40% (Abdelwahab Yousef, 2017). A multi-centre US study including 97 male breast cancer patients from 1939 families estimated the cumulative incidence of male breast cancer up to age 70 years for \( BRCA1 \) carriers to be 1.2% (95% confidence interval [CI] = 0.22% to 2.8%) and for \( BRCA2 \) carriers, 6.8% (95% CI = 3.2% to 12%) (Tai, Domchek, Parmigiani, & Chen, 2007). This study also found that the relative risk of male breast cancer was highest in men in their 30s and 40s and that the risk decreased with age. In \( BRCA2 \) carriers the relative risk at age 30 was 22.3 times higher than at age 70. \( BRCA2 \)-related male breast cancers have been found to be more aggressive (of a higher stage and grade) and more likely to be oestrogen and progesterone receptor positive than \( BRCA1 \)-related male breast cancers (Silvestri et al., 2016).

1.6.4. Prostate cancer

Up to age 65 years, the risk of prostate cancer is estimated to be 9.5% in \( BRCA1 \) carriers (Leongamornlert et al., 2012) and 15% in \( BRCA2 \) carriers (Kote-Jarai et al., 2011). A study comparing BRCA carriers and non-carriers with prostate cancer found that node involvement and distant metastases were significantly more common in \( BRCA1/BRCA2 \) carriers. However, treatment programmes for carriers and non-carriers were similar (Castro et al., 2013). Shorter survival amongst \( BRCA2 \) carriers with prostate cancer has also been reported (Edwards et al., 2010).

1.6.5. Other cancers

Several studies have investigated the risk of cancers other than breast, ovarian and prostate cancer in \( BRCA1 \) and \( BRCA2 \) carriers. Tumours of the pancreas and bowel have been associated with \( BRCA1 \) pathogenic variants (Iqbal et al., 2012; Phelan et al., 2013). Uveal melanoma and cancers of the oesophagus, stomach, pancreas, bone, buccal cavity and pharynx, gallbladder and bile duct have been associated with \( BRCA2 \) pathogenic variants (Breast Cancer Linkage Consortium, 1999; Easton, 1997; Moran et al., 2012; van Asperen et al., 2005). In a recent study of 1072 \( BRCA1/BRCA2 \) carriers, no significant increases were identified in cancers other than breast or ovarian cancer for \( BRCA1 \) pathogenic variants (Mersch et al., 2015). However, the occurrence of
pancreatic cancer was 82.5 times higher than expected in male BRCA2 carriers and 14 times higher than expected in female carriers. The authors speculated that this finding could be explained by the high number of smokers in the sample (Mersch et al., 2015). These findings have not been confirmed by large prospective studies.

1.7. MANAGEMENT OF HEREDITARY BREAST AND OVARIAN CANCER FOR CANCER PATIENTS AND THOSE AT RISK

For cancer patients with a BRCA1/BRCA2 pathogenic variant, management options include targeted chemotherapy, enhanced breast surveillance or a double mastectomy at the time of definitive cancer surgery and risk-reducing bilateral salpingo-oophorectomy. Breast screening and risk-reducing surgery is available for unaffected female carriers. Prostate screening for male carriers is under investigation.

1.7.1. Targeted chemotherapy

Identification of a BRCA1/BRCA2 pathogenic variant early in the diagnosis of ovarian cancer can have a major impact on treatment options and outcomes. Patients with BRCA-related ovarian cancer respond better to platinum- and non-platinum-based chemotherapy than those with sporadic epithelial ovarian cancer (Alsop et al., 2012; Bolton et al., 2012). Clinical trials have demonstrated that the use of poly ADP ribose polymerase (PARP) inhibitors in ovarian cancer treatment significantly improves progression-free survival compared to a placebo and that this effect is enhanced in women with a BRCA1/BRCA2 pathogenic variant (Ledermann et al., 2014). PARP inhibitors play an essential role in repairing DNA double strand breaks through the base excision repair pathway. In BRCA tumour cells with homologous recombination deficiency, the inability to repair double strand breaks leads to synthetic lethality (Ashworth, 2008). Treatment with the PARP inhibitor olaparib is now recommended for BRCA carriers with ovarian cancer in many parts of the world, including the UK (Tucker, Charles, Robertson, & Adam 2017).

A proof of concept trial suggested a favourable therapeutic response following treatment with olaparib in 27 patients with advanced breast cancer and a BRCA1/BRCA2 pathogenic variant (Tutt et al., 2010). Several other studies have suggested that PARP inhibitors hold promise for breast cancer treatment (Gelmon et al., 2011; Kaufman et al., 2015) and clinical trials are underway (Livraghi & Garber, 2015).
1.7.2. Breast surveillance

There is evidence that mammography reduces breast cancer-related mortality in women over the age of 50 years (The Independent UK Panel on Breast Cancer Screening, 2012). However, young women have high breast density which limits the sensitivity of mammography as a screening modality (Lehman et al., 2005). The effectiveness of magnetic resonance imaging (MRI) in combination with digital mammography has been demonstrated for young women at high risk of breast cancer in five prospective studies undertaken in Europe and North America (Kuhl et al., 2005; Leach et al., 2005; Lehman et al., 2005; Sardanelli et al., 2011; Warner et al., 2004). A systematic review of the findings across these studies reported that the use of MRI in combination with mammography reached a sensitivity of 83% to 100% (Lord et al., 2007). The increased sensitivity of MRI screening however leads to low specificity and a high false positive rate, resulting in a high recall rate, further imaging and biopsies (Chiarelli et al., 2014). In the UK, BRCA1/BRCA2 carriers with and without cancer are offered annual breast MRI scans from the age of 30 to 49 years and annual digital mammograms from the age of 40 until 69 years (National Institute for Health and Care Excellence, 2013).

1.7.3. Ovarian screening

Whether ovarian cancer surveillance is of benefit to the general population or high-risk women has yet to be determined. Two large studies have been undertaken to assess the mortality benefit of ovarian screening in the general population. The Prostate Lung Colorectal Ovarian (PLCO) Cancer Screening Trial found no difference in cancer deaths amongst the screening group and the control group (Buys et al, 2011). The UK Collaborative Trial of Ovarian Cancer Screening (UKCTOCS) reported high levels of sensitivity and specificity and a relative reduction in mortality of 15% in the trial group who were screened with annual transvaginal ultrasound scan and CA125 assay compared with 11% in the group screened with ultrasound scans alone. Although the mortality reduction was not significant, there appeared to be a late effect on mortality amongst participants 7 to 14 years after screening commenced. The authors concluded that further follow-up to assess the extent of the mortality reduction would be required (Jacobs et al., 2016).

The efficacy of ovarian screening has been studied in high risk women in the UK Familial Ovarian Cancer Screening Study (UKFOCSS). The Phase I study using annual serum cancer antigen 125 (CA-125) and transvaginal ultrasound scans reported that women screened within the year before their diagnosis were more likely
to have lower stage disease, highlighting the need for strict adherence to the screening schedule and a shorter time between screens (Rosenthal et al., 2013). Phase II of the study involved four-monthly CA-125 measurements interpreted using the risk of ovarian cancer algorithm (ROCA), together with transvaginal ultrasound. The study reported that women were significantly less likely to be diagnosed with stage IIIb or IV ovarian cancer during the trial than those diagnosed one year after completion of the trial. The findings suggest that the study triggered earlier and less complex surgery resulting in reduced surgical morbidity and fewer incomplete resections. It remains unknown whether screening improves survival in high risk women (Rosenthal et al., 2017). At present, there is insufficient evidence to offer ovarian screening to high risk women.

1.7.4. Prostate screening

Two large randomised controlled trials of prostate screening have been undertaken in the general population using annual Prostate Specific Antigen (PSA) with at least 10 years of follow up. One study (Schröder et al., 2012) found that although PSA screening significantly reduced the risk of metastatic prostate cancer, some men whose cancer was detected early through screening, still went on to develop metastatic disease. The second study (Andriole et al., 2012) identified no mortality benefit associated with screening. Although several studies have evaluated targeted screening amongst men at high risk, methodological differences between the studies make it difficult to draw conclusions about the efficacy of screening (Bancroft et al., 2014). In the UK, men who are concerned can access PSA testing following discussion with their general practitioner.

The IMPACT study (Identification of Men with a genetic predisposition to ProstAte Cancer: Targeted study screening in BRCA1/BRCA2 carriers and controls) is an international multi-centre study that investigates the role of prostate screening in men at high risk associated with a pathogenic variant. Preliminary results suggest that targeted screening detects clinically significant disease in BRCA2 carriers (Bancroft et al., 2014).

1.7.5. Risk-reducing mastectomy

Breast cancer patients tested shortly after diagnosis may decide to have a double mastectomy with or without immediate reconstruction at the time of definitive cancer surgery. Patients tested after completing cancer treatment may opt for a contralateral mastectomy once a pathogenic variant has been identified. For those at high risk of
metastatic disease or with a previous history of ovarian cancer, surgery may not be suitable (McGee et al., 2017). Bilateral mastectomy reduces the risk of contralateral breast cancer by up to 95% (Metcalfe, 2004; van Sprundel et al., 2005) but does not reduce the risk of metastases from the initial breast cancer (Lostumbo, Carbine Nora, & Wallace, 2010). Retrospective studies have shown a disease free and overall survival benefit to contralateral mastectomy (Herrinton et al., 2005; Kruper, Kauffmann, Smith, & Nelson, 2014), although prospective studies are lacking (Boughey et al., 2016). A Cochrane review found no difference in overall survival or breast cancer specific survival following contralateral mastectomy (Lostumbo et al., 2010). Several studies have found improved overall survival amongst BRCA1/BRCA2 carriers who opted for the surgery compared to those who did not (Heemskerk-Gerritsen, Rookus, et al., 2015; Metcalfe et al., 2014).

In female BRCA1/BRCA2 carriers who have not had breast cancer, there is evidence that bilateral mastectomy reduces the risk of breast cancer by approximately 95% (Hartmann et al., 2001; Meijers-Heijboer et al., 2001; Rebbeck et al., 2004).

1.7.6. Risk-reducing bilateral salpingo-oophorectomy

There is evidence that bilateral salpingo-oophorectomy reduces the risk of ovarian cancer in BRCA1/BRCA2 carriers by approximately 80% (Domchek et al., 2010; Rebbeck, Kauff, & Domchek, 2009). To maximise the risk benefit, it is recommended that BRCA1 carriers have surgery between the ages of 35 and 40 years (Finch et al., 2014). Removing the ovaries before the natural menopause can however have a negative impact on quality of life and health. To overcome this, women who have not had breast cancer are offered hormone replacement therapy (HRT) until the age of 50 years, although this is not an option for cancer patients (National Institute of Health and Care Excellence, 2013).

Risk-reducing bilateral salpingo-oophorectomy has also been shown to reduce all-cause mortality by 77% (Finch et al., 2014; Marchetti et al., 2014). The risk benefit is largely due to the reduction in the incidence of ovarian, tubal and peritoneal cancers but a reduction in breast cancer risk following surgery has also been observed. Several studies have reported a 50% reduction in breast cancer risk when oophorectomy is performed before the age of the natural menopause (Eisen et al., 2005; Rebbeck et al., 1999). Recently however these findings have been questioned, citing biases in the original studies (Heemskerk-Gerritsen, Seynaeve et al., 2015; Kotsopoulos et al., 2017).
1.7.7. Psychological impact of risk-reducing surgery

Most cancer patients who make the decision to have a contralateral mastectomy are satisfied with their choice and do not experience significant adverse psychological effects (Ager, Butow, Jansen, Phillips, & Porter, 2016; Collins, Gee, Clack, & Wyld, 2017). For some patients, there are areas of dissatisfaction, including altered body image, poor cosmesis, the impact of surgery on sexual relationships and complications relating to surgery and reconstruction (Ager et al., 2016; Collins et al., 2017).

The psychological impact of bilateral risk-reducing surgery on women at high risk of breast cancer is well documented with evidence of relief from cancer worry and few regrets (Frost, Schaid, Sellers et al., 2000; Hatcher, Fallowfield, & A'Hern, 2001; Hopwood, Lee, Shenton, Baildam et al., 2000). However, studies consistently report that for many women surgery results in psychological morbidity related to altered body image (Brandberg et al., 2008; Den Heijer et al., 2012; Gopie et al., 2013), reduced sexual pleasure and reduced feelings of attractiveness (Brandberg et al., 2008; Glassey, Ives, Saunders, & Musiello, 2016; Heiniger et al., 2015).

A study of BRCA carriers with and at risk of breast cancer found that levels of general distress and quality of life following risk-reducing salpingo-oophorectomy were similar to controls who did not have surgery and that ovarian cancer specific distress was reduced (Finch et al., 2011). Physical functioning scores were lower for cancer patients who were unable to take HRT than for at-risk women. Young women with breast cancer who had their ovaries removed as part of their cancer treatment are reported to experience significant menopausal symptoms, reduced sexual functioning and more psychological difficulties than those who did not have their ovaries removed (Sayakhot, Vincent, Deeks, & Teede, 2011).
1.8. CURRENT THESIS

1.8.1. Rationale
Increasingly, genetic testing of cancer patients will take place shortly after diagnosis, during chemotherapy treatment, at metastatic relapse or in palliative care. Various methods of delivering pre-test communication about genetic testing in mainstream oncology have been investigated, including face-to-face consultations with genetic counsellors or oncology health professionals, group education and education via written or digital means (George, Kaye, & Banerjee, 2017; Kentwell et al., 2017; Quinn et al. 2016; Sie et al., 2014; Wevers et al., 2017). These studies have mainly focused on the acceptability, feasibility and uptake of testing rather than the process, content or impact of the communication.

The goals of genetic counselling are ‘to promote health enhancing behaviours, to enhance accurate and useful risk perception, to facilitate adaptation to genetic risk and to prevent disease’ (Biesecker, 2001 p.329). These goals remain the same whether the ‘genetic counselling’ or communication takes place within clinical genetics provided by genetics specialists or in oncology provided by oncology health professionals.

Shifting genetic testing into mainstream oncology will inevitably have an impact on patients and families. The urgency of decision-making about testing changes the focus of discussions with patients from the future potential risks for the patient and family to impending cancer treatment. It is imperative that patients are equipped with sufficient information to make decisions about their own treatment and future cancer risks and to alert relatives of the risks to themselves and the choices available to them.

Changes to the delivery of genetic testing will also have a major effect on genetics and oncology health professionals. Although there are many similarities between genetics and non-genetics health professionals in terms of adopting a patient-centred approach, enhancing understanding, facilitating shared decision-making, promoting risk-reducing behaviour and providing emotional support (Smets, van Zwieten, & Michie, 2007), training and priorities amongst these two specialist areas are somewhat different. Genetics health professionals may not be trained in oncology or cancer care or be familiar with the specific concerns or communication needs of cancer patients at vulnerable times in their cancer trajectory. Oncology health professionals are unlikely to have experience at explaining the nuances in the possible outcomes of genetic testing, interpreting and communicating complex or ambiguous genetic test results or considering the cancer risks for the wider family.
The organisation of genetics and oncology clinical services will need to change to accommodate these developments. Decisions will need to be made about the funding of diagnostic testing, the organisation and delivery of patient pathways into specialist genetics and cancer surveillance services and the provision of individual and family support throughout the process of genetic testing, decision-making around hereditary cancer management and living with cancer risk.

There is no consensus about the information that should be communicated to cancer patients about genetic testing or hereditary cancer risk and management or the method of communication. To meet the goals of genetic counselling as the timing and methods of genetic testing shifts, it is important to learn from existing evidence and clinical practice.

1.8.2. Aims and objectives

The overall aim of this thesis was to investigate genetics health professionals’ communication about genetic testing, risk and management of hereditary breast and ovarian cancer with cancer patients. The purpose of the research was to identify areas of good practice and areas for improvement to inform future practice, policy and research in communication about hereditary cancer with cancer patients. The objectives were to identify the information that should be communicated, the way in which information should be communicated, the information genetics health professionals communicate, the information patients and their relatives recall following this communication and the experience and understanding of patients who undergo genetic counselling and testing shortly after breast cancer diagnosis.

1.8.3. Presentation of the research

The studies presented in this thesis build on each other (see Figure 1.1). Although the scoping review is presented in Chapter 2 to provide context and background information for the studies undertaken, this review was completed after the studies in this programme in order to map the breadth of existing research, to ensure the literature is as current as possible and to make recommendations for future research. The studies presented in Chapters 3, 4, 5 and 6 are presented in the order in which they were undertaken. The longitudinal qualitative study presented in Chapter 7 was undertaken over the course of the PhD programme.

Summaries of Chapters 2 to 8 are presented in Section 1.8.4. The context, terminology and support provided for this research are explained in section 1.8.5.
1.8.4. Chapter summaries

1.8.4.1. Chapter 2: Communication about hereditary cancer with breast and ovarian cancer patients - A scoping review

Aims: To identify areas for further research, this scoping review mapped the nature, extent and characteristics of existing research in this field.

Methods: Using a recognised scoping review method, a systematic search of databases was undertaken to identify studies about health professionals’ communication with cancer patients about hereditary cancer.

Results: In total, 29 papers from 25 studies were included in this review. Studies were identified about i) information needs, ii) process and content of genetic counselling, iii) cognitive and psychological impact of communication, iv) risk perception, v) recall, understanding and interpretation of genetic test results, vi) experiences of
communication, vii) communication shortly after cancer diagnosis and viii) alternative methods of communication.

Conclusions: No studies of communication about hereditary cancer by non-genetics health professionals were identified. There are gaps in the knowledge of the process and content of genetic counselling with cancer patients, particularly about the communication of ambiguous results. Few studies have investigated the information and support needs of cancer patients or experiences of those who undergo genetic counselling shortly after diagnosis. Few studies have investigated post-test genetic counselling when it appears that knowledge may be reduced and anxiety increased amongst cancer patients.

1.8.4.2. Chapter 3: Study 1. Tracking the accuracy of information about a BRCA1/BRCA2 pathogenic variant from genetics health professionals to cancer patients and their relatives – A retrospective content analysis

Aims: This observational study aimed to investigate i) the accuracy of information about genetic testing and hereditary cancer management recalled by patients and relatives following genetic counselling and ii) whether accuracy amongst relatives improved when information was provided directly by genetics health professionals.

Methods: Secondary analysis was undertaken of genetic test results consultations with 10 patients with breast or ovarian cancer and semi-structured interviews with the patients and 22 of their at-risk relatives. The information provided in the patients’ consultations was tracked through the families and coded for accuracy. Accuracy was analysed using the Wilcoxon signed-rank test. Sources of the information were identified from the transcripts and tested using a Spearman’s rank order correlation coefficient.

Results: Patients accurately recalled 53% of the information communicated. Relatives made significantly more inaccuracies than patients ($p = .017$). Patients and relatives made significantly more inaccuracies about hereditary cancer management information than genetic testing information ($p = .005$). Relatives in families where information was provided solely by the patient made significantly more inaccuracies than those to whom information was provided by the genetics health professional ($p = .001$) in addition to the patient.

Conclusions: Patients do not always pass on accurate information about a genetic susceptibility to cancer to their relatives, particularly about hereditary cancer management. Accuracy amongst relatives may be improved if targeted information is provided for relatives by genetics health professionals.
1.8.4.3. Chapter 4. Study 2: Consensus amongst health professionals and service users concerning key messages for communicating information about genetic testing and hereditary cancer management to breast/ovarian cancer patients – A Delphi consensus exercise

Aims: This study aimed to i) identify areas of agreement and disagreement between genetics and cancer health professionals and service users about the key messages required by patients with breast/ovarian cancer, ii) investigate the ease/difficulty of reaching agreement and iii) identify areas of agreement about the timing of communicating key messages.

Methods: Participants were 16 health professionals specialising in cancer genetics or oncology and 16 service users with breast and/or ovarian cancer and a BRCA1/BRCA2 pathogenic variant. Up to three rounds of an iterative Delphi survey were circulated to identify which messages were key and the optimal timing of communication.

Results: There was a high level of agreement within and between the groups about 30 key messages and 10 messages considered to be not key. Seven key messages were agreed by ≥ 95% of participants. Both groups agreed that key messages should be communicated before genetic testing and once a pathogenic variant is identified.

Conclusions: Expert health professionals and service users agreed on the key messages about BRCA1/BRCA2 required by patients with breast or ovarian cancer. Both groups agreed that key messages should be communicated before genetic testing and once a pathogenic variant has been identified.

1.8.4.4. Chapter 5: Study 3. Communicating key messages about a pathogenic BRCA1/BRCA2 pathogenic variant to patients with breast and ovarian cancer – A retrospective content analysis

Aims: This study investigated the types of message communicated by genetics health professionals during post-test genetic counselling and whether or not the key messages identified by expert health professionals in Study 2 were communicated.

Methods: The study design was a retrospective content analysis of anonymised transcripts of post-test genetic counselling consultations between 37 women with breast and/or ovarian cancer and 14 genetics health professionals. Transcripts were searched for reference to the key messages. Supplementary messages were also identified. The key and supplementary messages were grouped into messages about genetic testing and those about hereditary cancer management. The types of message communicated were analysed using the Wilcoxon signed-rank test.
Results: The genetics health professionals communicated significantly more key than supplementary messages ($p = .014$). Fifty percent of the key messages agreed by expert health professionals in the field were communicated, of which significantly more key messages were communicated about genetic testing than hereditary cancer management ($p = .004$). The key messages about genetic testing for relatives were frequently communicated. The key messages about the risks and limitations of breast and ovarian cancer screening and risk-reducing surgery were infrequently communicated.

Conclusions: Cancer patients did not receive all the information judged by experts as required to enable decision-making about managing their risk of future cancers. Health professionals should focus on communicating key messages about hereditary breast and ovarian cancer management as well as those about genetic testing. Supplementary messages should be communicated only when they are particularly relevant to the specific patient and there is the time to do so.

1.8.4.5. Chapter 6: Study 4. Investigating the translation of guideline recommendations about hereditary breast and ovarian cancer for women with cancer into expert opinion, health professionals’ communication and patients’ recall: Documentary analysis, content analysis and mixed methods matrix

Aims: This study aimed to investigate the translation of guideline recommendations about hereditary breast and ovarian cancer into expert opinion, genetics health professionals’ communication and cancer patients’ recall.

Methods: Stage I was a documentary analysis of international genetics guidelines and national and European breast and ovarian cancer guidelines. Stage II involved a documentary analysis of clinical protocols and patient leaflets for breast and ovarian cancer patients provided by UK Genetics Centres. Stage III was a content analysis using a mixed methods matrix to identify the extent to which the guideline recommendations were referred to in the guidelines and translated into expert opinion, health professionals’ communication and patients’ recall. To compare the findings across the studies in this programme of research, a comparable ‘matrix score’ was calculated from the range of each dataset. Recommendations were grouped into those scoring ≥ 4 (frequently referred to/translated), 2.1 to 3.9 (intermittently referred to/translated) and ≤ 2 (infrequently referred to/translated).

Results: Stage I: Seventeen guideline recommendations about information to communicate to cancer patients and 15 recommendations about the method of this communication were identified from the guidelines. Stage II: The information
communicated in UK genetics clinical protocols and patient information leaflets was relevant for women with cancer. However, less than half (42.48%) of the guideline recommendations were communicated. Significantly more recommendations about hereditary cancer management were communicated via patient leaflets than via clinical protocols ($p = .016$). Stage III: Fewer than 1.5% of the genetic testing recommendations and 20% of the hereditary cancer management recommendations were referred to in the cancer guidelines. Recommendations about genetic testing and the availability of surveillance and risk-reducing surgery were frequently translated into expert opinion, communicated by health professionals and recalled by patients. Recommendations about the risks for relatives and aspects of hereditary cancer management were infrequently translated into expert opinion, communicated by health professionals’ or recalled by patients.

**Conclusions:** National and international genetics guidelines mainly focus on at-risk women. UK and European breast and ovarian cancer guidelines make little or no reference to genetic testing or hereditary cancer management information. Genetics clinical protocols and patient leaflets for cancer patients communicate less than half of the information recommended by existing guidelines. Cancer patients may not receive or recall all the information they need to make decisions about hereditary cancer management for themselves or to inform their at-risk relatives about their options.

**1.8.4.6. Chapter 7: Study 5. Understanding and experiences over time of patients who undergo genetic counselling and testing for newly diagnosed breast cancer:**

**Interpretative Phenomenological Analysis**

**Aims:** This study aimed to explore understanding and experiences over time of women with newly diagnosed breast cancer who undergo genetic testing for hereditary breast and ovarian cancer.

**Methods:** The sample was 11 women undergoing genetic testing for pathogenic variants in the **BRCA1**/**BRCA2** genes shortly after breast cancer diagnosis. In-depth semi-structured interviews were undertaken at three time points: following genetics referral shortly after breast cancer diagnosis, following genetic counselling and testing and following the genetic test result. Participants were stratified into four groups for analysis according to their expectation of a genetic predisposition to cancer and their genetic test result. Data were analysed using Interpretative Phenomenological Analysis.

**Results:** Amongst those with a strong family history of cancer and a prior expectation of a genetic predisposition, there was a persistent feeling that the cancer was hereditary,
regardless of the genetic test result. Those who did not expect a genetic predisposition were left with uncertainty about a potential hereditary cause for their cancer, even when no abnormality was detected. Participants who received an expected result experienced growing certainty and confidence about surgical decision-making. Those who received an unexpected result experienced growing uncertainty about inheritance and surgical decisions. Feelings of concern and responsibility for the family in relation to the genetics information remained constant regardless of prior expectation, family history or genetic test results. Participants with a pathogenic variant or VUS grew increasingly dissatisfied with interactions with cancer and genetics health professionals.

Conclusions: Incongruence between the expectation of hereditary cancer and the genetic test result negatively affected certainty and confidence about inheritance and surgical decision-making for breast cancer patients tested shortly after diagnosis. The genetics information heightened feelings of responsibility for the family. Those found to have a pathogenic variant or VUS expressed growing dissatisfaction with health professional interactions. Health professionals who counsel breast cancer patients about genetic testing shortly after diagnosis should elicit expectations of inheritance and understanding of the potential impact of genetic testing on the family prior to testing. This may help to identify patients who would benefit from increased support and specialist genetic counselling once the result is known.

1.8.4.7. Chapter 8: General discussion

This chapter summarises and discusses the key findings in the context of the study objectives. The implications of the research findings for practice, policy and future research are considered.

1.8.5. Notes about the research presented in this thesis

1.8.5.1. Context

Over the course of this research, there have been important advances in genetic testing technology and cancer treatment that have changed practice. For example, the availability of olaparib as a clinical treatment for ovarian cancer has heightened the urgency of testing shortly after ovarian cancer diagnosis.

Completion of the Human Genome Project in 2000 (Lander et al.) laid the foundations for genome-wide association studies (GWAS) to identify multiple small genetic effects
that combine to predispose to complex diseases and Single Nucleotide Polymorphisms (SNPs). In the UK, the 100,000 Genome Project was launched in 2012 to translate the benefits of the Human Genome Project to patients in terms of personalised prevention, diagnosis and treatment of disease. These developments led to advances in genetic testing technology, such as the availability of massive parallel sequencing, whole exome sequencing (WES) and whole genome sequencing (WGS). WES and WGS are not yet available in clinical practice in the UK. Massive parallel sequencing has however been introduced in many laboratories over the last few years, enabling many patients to benefit from simultaneous testing of multiple genes rather than consecutive testing of single genes (Jacobs & Pichert, 2016).

Genetic testing using multiple gene panels was not offered when the data in this thesis were collected. The research in this thesis therefore focuses on single gene testing of the \textit{BRCA1}/\textit{BRCA2} genes. In the USA, multiple panel gene testing has become standard practice. However, in other parts of the world testing of the \textit{BRCA1}/\textit{BRCA2} genes is still offered to many patients without testing other intermediate and low penetrance genes. The uncertain clinical utility of intermediate and low penetrance gene variants has led to questions about the appropriateness of testing for genes that are not highly penetrant until more is known about the cancer risks associated with variants (Obeid, Hall, & Daly, 2017).

1.8.5.2. Terminology used to refer to the outcomes of genetic testing

Techniques for sequencing of human DNA have led to identification of a vast number of variants in the DNA sequence. Within the field of cancer genetics, variants are classified according to their likely pathogenicity. Pathogenicity is determined by the level of evidence of the clinical significance of the variant drawn from peer-reviewed literature, large scale cancer variant databases and clinical practice guidelines (Li et al., 2017). To avoid confusion in the reporting of genetic and genomic testing, specific standard terminology is recommended by the American College of Medical Genetics and Genomics (ACMG). The ACMG recommends that the following terms are used to describe gene variants: ‘pathogenic’, ‘likely pathogenic’, uncertain significance, ‘likely benign’ and ‘benign’ (Richards et al., 2015). The ACMG terminology however is not referred to in papers or studies prior to 2015. For clarity, in this thesis the following terminology is used. A pathogenic mutation is referred to as ‘a pathogenic variant’, except in the wording of the key messages in Study 2 where the term ‘faulty gene’ is used in line with evidence of patients’ preference (Wakefield et al., 2007) and in the references when the term ‘mutation’ is used in some studies. When the significance of
a variant is uncertain or unknown the result is termed a variant of uncertain significance or ‘VUS’. When the result shows neither a pathogenic variant nor a VUS, the result is referred to as ‘no variant’ or ‘no variant detected’.

1.8.5.3. Definition of terms used throughout this thesis

- **Genetic testing information** refers to information communicated pre- and/or post genetic testing about the process or implications of the genetic test for the patient and/or the family.
- **Hereditary cancer management information** refers to information about cancer risks, surveillance, risk-reducing surgery or treatment of hereditary cancer.
- **Cancer patients** refers to women with a personal history of breast and/or ovarian cancer.
- **At-risk women** refers to women who are at high risk of inheriting a genetic predisposition to breast or ovarian cancer.
- **Genetics health professionals** includes genetic counsellors who are non-medical and medical clinical geneticists.
- **Oncology health professionals** includes oncologists, breast surgeons, gynaecological oncologists and cancer nurses.

1.8.5.4. Study Interest Group

Chapter 4 (Study 2), Chapter 6 (Study 4) and Chapter 7 (Study 5) refer to ‘The Study Interest Group’. The Study Interest Group was set up to provide clinical and lay advice about aspects of the research to support the research supervision. This advice was mostly provided via email or telephone. The Study Interest Group consisted of Dr Gabriella Pichert (GP) who provided expertise in cancer genetics and oncology, Dr Christine Patch (CP) who provided expertise in genetic counselling, Ms Jackie Harris (JH) who provided expertise in cancer care and W.D. who provided the patient’s perspective (name withheld to protect identity).

Other health professionals and researchers have been involved in other aspects of this body of work. Their contribution is acknowledged in the acknowledgements section of this thesis.
1.8.5.5. The Family Communication Study

Studies 1, 3 and 4 draw on data collected for the ‘Family Communication Study’. The Family Communication Study was funded by the Department of Health to investigate communication within families about genetic testing for hereditary breast and ovarian cancer and Familial Hypercholesterolaemia. The findings have been published (Dancyger, Smith, Jacobs, Wallace, & Michie, 2010; Dancyger et al., 2011; Smith, Dancyger, Wallace, Jacobs, & Michie, 2011).

The Study team for the Family Communication Study consisted of the thesis author (CJ), Professor Susan Michie (SM), Professor of Health Psychology, University College London (supervisor), Dr Caroline Dancyger (CD), Clinical Psychologist, Barts Health NHS Trust (Research Associate) and Professor Jonathan A. Smith (JAS), Professor of Psychology, Birkbeck University of London.

NHS Research Ethics Committee approval (06/MRE02/6) was obtained for the Family Communication Study. Genetics health professionals consented to audio-tape recording of the post-test genetic counselling consultations and analysis of post-consultation clinic letters. Patients consented to audio-taped recording of the consultations and research interviews. Relatives consented to audio-taped research interviews. The participant information sheets and consent forms are shown in Appendix 1.1-1.4.

Eligible participants were patients with breast or ovarian cancer who were informed about a \textit{BRCA1/BRCA2} pathogenic variant following diagnostic testing and their at-risk biological relatives with whom they had shared the result. All participants were over 18 years of age and spoke English.

Data were collected between 2006 and 2008 from patients and health professionals in two Regional Genetics Centres in the UK. Patients receiving \textit{BRCA1/BRCA2} diagnostic genetic test results were seen for pre-test genetic counselling and results were given in person during a subsequent genetic counselling consultation by a genetic counsellor or clinical geneticist. Patients were recruited after blood had been taken for genetic testing but prior to receiving their test result. The patients recruited their at-risk relatives after they had discussed the result with them. These relatives may or may not have undergone predictive testing by the time they were interviewed. Only families where the patient and at least two relatives were interviewed were included.
Semi-structured interviews were carried out with patients and relatives once the result had been communicated. Data were analysed using Interpretative Phenomenological Analysis. The interview schedules are shown in Appendix 2.1.
CHAPTER 2: COMMUNICATION ABOUT HEREDITARY CANCER WITH BREAST AND OVARIAN CANCER PATIENTS - A SCOPING REVIEW

2.1. INTRODUCTION

Increasingly, genetic testing is offered shortly after cancer diagnosis to guide management, leading to a growing need to deliver information about hereditary cancer and to facilitate informed decision-making to greater numbers of patients and within a shorter time frame than has traditionally been the case. Identifying areas of good practice and gaps in existing knowledge is important for developing new approaches to communication with cancer patients. This scoping review was undertaken to identify and map the research into health professionals’ communication with cancer patients about hereditary breast and ovarian cancer.

2.2. BACKGROUND

2.2.1. Genetic counselling about hereditary cancer

Several studies have examined health professionals’ communication about hereditary breast and ovarian cancer with at-risk women (Hallowell, Statham, Murton, Green, & Richards, 1997; Hopwood, Howell, Laloo, & Evans, 2003; Lobb et al., 2002; Pieterse, Ausems, Van Dulmen, Beemer, & Bensing, 2005; Pieterse, Van Dulmen, Ausems, Beemer, & Bensing, 2005; Watson et al., 1998). Studies have found extensive provision of technical information, little engagement in social or emotional issues, a variety of advice and evaluative statements and a focus on communication about biomedical rather than psychological and social issues (Aalfs, Oort, de Haes, Leschot, & Smets, 2006; Ellington et al., 2006; Ellington et al., 2005; Meiser, 2005; Meiser, Irle, Lobb, & Barlow-Stewart, 2008; Michie, Lester, Pinto, & Marteau, 2005; Pieterse et al., 2005; Roter et al., 2006).

Two previous reviews have investigated studies into the process and content of genetic counselling about a variety of genetic conditions (Meiser et al., 2008; Paul, Metcalfe, Stirling, Wilson, & Hodgson, 2015). A critical review documented empirical studies of the content and process of reproductive and hereditary cancer genetic counselling, reviewing studies of audio- or videotaped genetic counselling sessions with patients and those at risk of the condition (Meiser et al., 2008). Six of the eighteen studies included in the review investigated communication with cancer patients and women at risk of hereditary breast and ovarian cancer. The review concluded that genetic
counselling is often provider-driven and that information giving is integral to the genetic counselling, particularly for hereditary cancer. More recently, a systematic review, analysed communication in genetics consultations with patients and those at risk of mixed genetics conditions, including hereditary breast and ovarian cancer (Paul et al., 2015). The largely biomedical and educational content of the genetic counselling was highlighted, together with the lack of focus on psychosocial aspects.

2.2.2. Communication with cancer patients

Studies have consistently shown that cancer patients wish to be fully informed about their condition and treatment (Jenkins, Fallowfield, & Saul, 2001; McPherson, Higginson, & Hearn, 2001) and that satisfaction is increased when information is communicated without the patient needing to ask questions (Ong, Visser, Lammes, & de Haes, 2000; Pieterse, van Dulmen, Beemer, Bensing, & Ausems, 2007; Siminoff, Ravdin, Colabianchi, & Sturm, 2000). UK national guidelines on improving supportive and palliative care for adults with cancer (Nationale Institute for Clinical Excellence, 2004) identified key communication needs for cancer patients to enable understanding, inform decisions, facilitate better health-related quality of life and improve the experience of care. The guidelines identified the need for skilled health professionals with the ability to listen and engage emotionally with patients, promote mutual understanding and facilitate shared decision-making. A close relationship between clear and sympathetic communication and the provision of emotional support was highlighted as important. The recommended communication skills also included assessment of patients’ information needs and information-giving at critical points in the patient pathway, accompanied by opportunities for reflection and questioning and supplemented by written or other materials and telephone communication.

A review of the communication goals and needs of cancer patients concluded that cancer patients have unmet communication needs and that communication outcomes are enhanced when health professionals attend to patients’ emotional needs (Hack, Degner, & Parker, 2005). Several systematic reviews have investigated the communication between oncology health professionals and individuals with cancer. Reviews have identified the need for an individualised approach to patients (Rodin et al., 2009), the influence of clinicians’ personal characteristics on the effectiveness of communication (De Vries et al., 2014; Tay, Hegney, & DNurs, 2010) and good communication skills (Fallowfield & Jenkins, 1999).
2.2.3. Communication with cancer patients about hereditary cancer

A preliminary search was also undertaken for existing systematic and scoping reviews into communication about genetic testing or hereditary cancer management with cancer patients. Other than the reviews discussed above, no previous relevant reviews were identified on the following databases: Joanna Briggs Institute Database of Systematic Reviews and Implementation Reports, Cochrane Database of Systematic Reviews, EPPI and PubMed.

2.3. SCOPING REVIEW METHODOLOGY

Scoping reviews are used to identify the nature, extent and characteristics of existing studies in a particular field, to determine the feasibility of a systematic review, to summarise and disseminate research findings and to identify gaps in the literature (Arksey & O’Malley, 2005). Scoping reviews are particularly useful when no previous systematic reviews have been undertaken or where the diversity of the literature does not lend itself to a systematic review (Peters et al., 2015). Because different study designs can be included in scoping reviews, they do not generally include a quality assessment (Arksey & O’Malley, 2005), although this criterion has been challenged (Brien, Lorenzetti, Lewis, Kennedy, & Ghali, 2010; Daudt, van Mossel, & Scott, 2013). Whereas a systematic review aims to address very specific research questions and to synthesise research findings, a scoping review aims to identify the range and nature of the qualitative and quantitative studies in an area and to develop a visual representation that ‘maps’ or ‘charts’ the data (Peters et al., 2015).

Scoping reviews systematically organise and describe a body of literature, making them useful for disseminating research findings to health professionals, policy makers and other groups. As a result, this method of review is widely used and growing in popularity. There has been criticism of the lack of clarity and consistency in terminology, methodology and reporting within scoping reviews and recommendations to conduct scoping reviews that are methodologically sound (Colquhoun et al., 2014). The method described in this scoping review was therefore informed by the methodology developed by key authors in the field (Arksey & O’Malley, 2005; Levac, Colquhoun, Levac, O’Brien, Straus, Tricco, Perrier, Kastner & Moher, 2010; Peters et al., 2015). The procedure was based on the most recent guidance for conducting scoping reviews (Peters et al., 2015; The Joanna Briggs Institute, 2015).
2.4. METHOD

2.4.1. Objectives

The objective of this scoping review is to summarise and map the range, extent and nature of the published research into communication about hereditary breast and ovarian cancer with cancer patients, providing an overview of the studies in this area. The purpose of the review is to provide background information for this thesis and identify areas for future research.

2.4.2. Review question

In accordance with the methodology (The Joanna Briggs Institute, 2015), the review question was driven by the population, concept and context of the review.

The review question was as follows:
What is known about the communication that takes place about hereditary cancer between genetics/oncology health professionals and patients with breast or ovarian cancer?

2.4.3. Population

2.4.3.1. Inclusion criteria

Types of participants

This review included patients with a personal history of breast or ovarian cancer (cancer patients) who had undergone communication about hereditary breast and ovarian cancer in the genetics or oncology setting. Participants were included if they had experienced this communication after completing cancer treatment or prior to or during cancer treatment.

Health professionals specialising in genetics or oncology were included in the review including genetic counsellors, geneticists, oncologists, surgeons and nurses.

The following topics and settings were not included:

1. Men with breast or prostate cancer with whom hereditary cancer had been discussed,
2. Cancers other than hereditary breast or ovarian cancer,
3. Children below the age of 18,
4. Individuals at risk of hereditary breast and ovarian cancer but without a personal history of the disease,
5. Communication about the risk of hereditary cancer in the primary care setting,
6. Communication about the risk of hereditary cancer in the palliative care setting,
7. Communication within families about hereditary breast and ovarian cancer.

2.4.4. Concept

Types of interventions
Studies were included if they were about the process and/or content of verbal, written
or digital communication, the information needed, knowledge understood and recalled
or the experience of this communication.

The following types of study were not included:
1. Studies about the impact of genetic test results but not the communication,
2. Studies where none of the findings for cancer patients and at-risk women were
   presented separately.

Comparators
Comparators were different participants, contexts, interventions or strategies involving
i) patients with other types of cancer, ii) women at risk of hereditary cancer, iii) health
professionals, iv) health conditions or v) communication methods.

2.4.5. Context

The context of this study was the hospital or community setting, including genetics
services, cancer clinics and oncology departments.

Types of studies
Published quantitative and qualitative studies of all designs were included. Reviews,
editorials, chapters and commentaries were not included.

Types of outcomes
Outcomes expected from the literature about communication with women at risk of
hereditary breast and ovarian cancer were:
- Ratio of health professional-patient talk,
- Accuracy and extent of patients’ knowledge at recall,
- Met and unmet communication needs,
- Patients’ satisfaction,
- Patients’ distress and anxiety,
• Patients’ intention to have a procedure or investigation, such as genetic testing or surgery,
• Patients’ experience and understanding,
• Communication methods.

2.4.6. Search strategy
An initial limited search was undertaken of the MEDLINE and CINAHL databases to identify the extent of the evidence and to ensure key texts were identified. The key words used in the initial search were informed by the review questions: genetic counseling/counselling, genetic testing, breast cancer, ovarian cancer, information, recall, experience, understanding, comprehension. From analysis of the title, abstract and index terms of the identified studies, the search was refined, although the search terms remained broad to capture as many relevant studies as possible. The search strategy was discussed with an information scientist with experience of literature search methodologies in the field of genetics (Cheng Sui). It was agreed that the search terms should be kept as broad as possible to capture the relevant papers. Six key papers were identified to test the search strategy. Once all the test papers were identified the search terms were agreed with a second researcher with experience of conducting scoping reviews and systematic reviews (CP). The time frame for inclusion of studies was from 1994, when the \textit{BRCA1} gene was identified, to October 2017. A second search was undertaken of the databases MEDLINE, CINAHL, PsycINFO and EMBASE for the following search terms adapted for each database: genetic counselling or genetic counseling or genetic testing and ovarian, breast or fallopian tube neoplasms or neoplastic syndromes, hereditary syndromes or hereditary breast and ovarian cancer syndrome or \textit{BRCA1} or \textit{BRCA2}. Forward and backwards citation searches were undertaken on the Web of Knowledge and Scopus for all included papers. Due to time and cost constraints, articles not in English were not translated.

2.4.7. Quality assessment
As the intention of this scoping review was to map the extent, nature and features of existing research (Arksey & O'Malley, 2005), quality assessment of the studies was not undertaken.

2.4.8. Data collection
The qualitative and quantitative data were extracted and documented onto the standardised data extraction tools adapted from the Joanna Briggs Institute (2015). Details of the study method, participants, sample size and interventions and outcomes
significant to the review question were documented onto a flow diagram (Moher, Liberati, Tetzlaff, Altman, & The, 2009).

2.4.9. Data extraction
The data were extracted by one researcher (CJ). A second researcher (CP) independently reviewed 10% of titles, abstracts and articles. An agreement level of 97% was reached. The remaining articles were reviewed by the thesis author and validated by CP.

2.4.10. Data synthesis
The process of synthesising the data drew on the published guidance for conducting a narrative synthesis (Petticrew, 2006; Popay, 2006). The iterative process involved a preliminary synthesis of the study findings, exploration of relationships between the studies and summary of the synthesised findings in narrative form within areas of study.

To identify themes and develop a preliminary synthesis of the findings, the studies were organised according to the main area of study. The important characteristics of each study were transcribed onto a table. Table headings included aims, population and sample, method, intervention, measures, procedure, methodology, results and authors’ conclusions. This process enabled identification of the areas of study.

To identify the relationship between the studies a further table was devised to document the population within each study and the areas addressed by each study. This process enabled identification of studies that addressed more than one area of communication research (Tables 2.2 and 2.3).

Within each study area a narrative description was written up of each of the studies. Finally, relationships between the findings in each area were written up as a narrative synthesis.
2.5. RESULTS

A total of 5458 citations were identified by the search strategy and 30 additional papers were added. Duplicates were removed and 4823 citations were excluded as they were not relevant. A further 30 papers were added following forward and backward searching of the selected articles. In total, 79 full text papers were reviewed of which 50 were excluded as they did not meet the inclusion criteria. A total of 29 papers from 25 studies were included in this scoping review. The PRISMA flow diagram of the number of selected and rejected studies is shown in Figure 2.1 (Moher et al., 2009).

Population

Participants were female cancer patients. In 20 papers from 18 studies, participants were exclusively cancer patients. Nine papers from seven studies included women at risk of breast and ovarian cancer (at-risk women). Eleven papers included patients with breast cancer and those with ovarian cancer, 17 papers included breast cancer patients only and one paper included ovarian cancer patients only. No studies of cancer patients were identified prior to 2000. Since 2008, more studies have focused on cancer patients only than on cancer patients and at-risk women (see Figure 2.2).

Health professionals were mainly genetic counsellors or clinical geneticists. Six papers from three studies included genetics health professionals only (Butow & Lobb, 2004; Lobb et al., 2004; Lobb et al., 2002; Pieterse, Ausems, Spreeuwenberg, & van Dulmen, 2011; Pieterse, van Dulmen, van Dijk, Bensing, & Ausems, 2006). One study included genetics and oncology health professionals (Jacobs, Pichert, Harris, Tucker, & Michie, 2017). One study included satisfaction with information provided by referring physicians at genetics referral but investigated the genetic counselling communication (Vadaparampil et al., 2011).
Figure 2.1. PRISMA Flow diagram of selected and rejected studies
### Figure 2.2. Types of participants in selected studies

<table>
<thead>
<tr>
<th>References in ascending order according to year of publication</th>
<th>Cancer patients &amp; at-risk women</th>
<th>Cancer patients only</th>
<th>Genetics health professionals</th>
<th>Cancer health professionals</th>
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<td>Randall, J., et al. (2001)</td>
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### Areas of research

Areas of research included i) information needs, ii) process and content of genetic counselling, iii) cognitive and psychological impact of communication, iv) risk perception, v) recall, understanding and interpretation of genetic test results, vi) experiences of communication, vii) communication shortly after cancer diagnosis and viii) alternative methods of communication. Figure 2.3 maps the areas of research into communication addressed by each study. Abbreviations in the table refer to information communicated (info), process and content of communication (process), cognitive impact (cog), psychological impact (psych), risk perception (risk), recall and understanding (recall), experience (exp), communication shortly after cancer diagnosis (timing) and alternative methods of communication (method). Papers from the same study are grouped and shown with an asterisked number.
Table 2.3. Areas of research into health professionals’ communication with cancer patients

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Study details

Of the 25 studies, there were two randomised surveys, two retrospective surveys, one Delphi survey and 10 non-randomised longitudinal surveys (including two studies that also included content analysis of transcripts of consultations). Three further studies involved content analysis of transcripts of consultations. Seven studies were qualitative, including five individual semi-structured interview studies and one focus group study.

The studies included in the review were published between 2000 and 2017 and were mainly from the Netherlands and Australia (see Table 2.1).

Details of the included studies are shown in Table 2.2. Some studies that addressed different areas of research are referred to more than once.

Table 2.1. Country and year of publication of the included studies

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<td>Norway</td>
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</table>

2.5.1. Information needs

Five studies addressed the information needs of cancer patients undergoing communication about hereditary cancer. Of these, two studies reported unmet needs following genetic counselling (Lobb et al., 2004; Metcalfe et al., 2000) and three studies investigated the information needs of patients undergoing genetic testing (Gleeson et al., 2013; Jacobs et al., 2017; Meiser et al., 2012).

Following pre-test genetic counselling, patients reported unmet needs for information about their risk of contralateral breast cancer and the cancer risks for their relatives (Lobb et al., 2004). Prior to counselling, 77% of patients had commented that they would like information about their own risk and 98% wanted information about their relatives’ risk. This information was discussed in less than 45% of the consultations (Lobb et al., 2004). Cancer patients also reported more unmet needs than at-risk women about treatment options, including surgery, screening and chemoprevention (Metcalfe et al., 2000).
Thirty key messages were agreed in a Delphi survey by 16 expert genetics and cancer health professionals and 16 service users with cancer and a BRCA1/BRCA2 pathogenic variant. The key messages included information about genetic testing, cancer risks for patients and relatives and the management of hereditary cancer. Participants did not consider the implications of testing on treatment to be a key message. It was agreed that key messages should be communicated prior to genetic testing and repeated if a pathogenic variant was identified (Jacobs et al., 2017) (see Chapter 4).

Qualitative studies with 26 young breast cancer patients diagnosed before age 50 years and 22 ovarian cancer patients identified the information needs of patients tested shortly after diagnosis. Participants preferred brief, personalised, positive and straightforward information without statistics. Most patients considered it important to have information about the purpose of testing, the implications for treatment decisions, the time frame for results and the availability of predictive testing for relatives. For some patients, more detailed information may be required (Gleeson et al., 2013; Meiser et al., 2012).

2.5.2. Process and content
In total, four papers from two studies investigated the process and/or content of genetic counselling about hereditary cancer and reported some findings for cancer patients and at-risk women separately (Butow & Lobb, 2004; Lobb et al., 2004; Lobb et al., 2002; Pieterse et al., 2006). Four papers were from the same study involving analysis of pre-test genetic counselling consultations between 69 cancer patients, 89 at-risk women and seven health professionals (Butow & Lobb, 2004; Lobb et al., 2004; Lobb et al., 2002)

Most of the findings were reported for at-risk women and cancer patients combined. Essential information about hereditary breast cancer was consistently communicated but emotional concerns were not always identified and genetics health professionals infrequently facilitated patients’ involvement in consultations (Butow & Lobb, 2004). Patients and at-risk women were satisfied with the information they received and considered their expectations to be met (Lobb et al., 2004).

Health professionals discussed significantly more aspects of genetic testing ($p < .001$), facilitated active patient involvement ($p < .001$) and used more supportive and counselling behaviours ($p = .02$) with cancer patients than they did with at-risk women.
(Lobb et al., 2002). The percentage of consultations in which specific cancer risks were discussed was similar for cancer patients and at-risk women (Butow & Lobb, 2004). However, the risk of having a pathogenic variant was communicated in just 13.3% of consultations with cancer patients compared to 71.8% of consultations with at-risk women (Lobb et al., 2004).

With cancer patients only, the risk of further cancers in the presence of a pathogenic variant was discussed in 37.3% of consultations. The risk of further cancers in the absence of a pathogenic variant was discussed in 29.3% of consultations respectively (Butow & Lobb, 2004). However, of the 89 breast cancer patients within the sample, 68% did not feel reassured and 57% felt they could have been helped to cope better with their situation (Lobb et al., 2004). Information about their own and their relatives’ risks of cancer was communicated in less than 45% of the consultations with cancer patients. A further analysis of genetic counselling consultations was undertaken between 10 genetics health professionals and 34 patients with breast cancer and 17 at-risk women. This study found that the risk of contralateral or primary breast cancer was communicated to 38% of cancer patients and the risk of ovarian cancer was communicated to 50% of cancer patients (Pieterse et al., 2006).

### 2.5.3. Cognitive and psychological impact of the communication

In total, nine studies addressed the impact of communication. Of these, five studies reported on the cognitive and the psychological impact (Bredart et al., 2016; Christie et al, 2012; Mancini et al., 2006; Quinn et al., 2016; Randall, Butow, Kirk & Tucker, 2001). Two studies reported on the cognitive impact only (Benusiglio et al., 2017; Scherr, Christie, & Vadaparampil, 2016). Two studies reported on the psychological impact only (Pieterse et al., 2011; van Roosmalen et al., 2004).

A significant increase in knowledge was reported following pre-test communication by face-to-face genetic counselling (Bredart et al., 2016; Christie et al, 2012; Randall et al., 2001; Scherr et al., 2016), group education (Benusiglio et al., 2017) and written communication (Mancini, et al., 2006; Quinn et al., 2016). A wide variety of knowledge measures were used, only some of which were validated for patients.

Two studies suggested that the knowledge gained at pre-test counselling may not be retained at post-test genetic counselling (Bredart et al., 2016; Scherr et al., 2016). A significant reduction in hereditary cancer knowledge between pre-and post-test genetic counselling was identified in survey of 243 breast cancer patients (Bredart et al., 2016).
There was a mean of 11 months (SD 3) between consultations. The mean total breast cancer genetics knowledge score dropped from 18.6 (SD 4.2) after pre-test genetic counselling to 17.1 (SD 4.5) after post-test counselling. In patients who over-estimated their risk of a pathogenic variant at pre-test counselling, anxiety was raised following post-test counselling. A further study with 103 breast cancer survivors found that knowledge was significantly reduced in the six months between pre-and post-test genetic counselling (\(p = .003\)) (Scherr et al., 2016).

Pre-test communication did not have a negative impact on cancer related distress, anxiety or depression, intrusive thoughts, decisional conflict, family involvement in decision-making or satisfaction amongst cancer patients (Benusiglio et al., 2017; Christie et al, 2012; Mancini et al., 2006; Quinn et al., 2016; Randall et al., 2001).

Anxiety, depression and cancer related distress was increased and general health decreased amongst cancer patients and at-risk women after disclosure of the genetic test result (Bredart et al., 2016; Pieterse et al., 2011; van Roosmalen et al., 2004). A randomised longitudinal study found that cancer related distress and anxiety was higher amongst patients diagnosed within a year of genetic testing than those tested over a year from diagnosis (\(p = .05\)) (van Roosmalen et al., 2004). In patients who over-estimated their risk of a pathogenic variant at pre-test counselling, anxiety was raised following post-test counselling (Bredart et al., 2016). A longitudinal survey including 44 cancer patients identified that cancer-related distress peaked immediately after post-test counselling (Pieterse et al., 2011).

### 2.5.4. Risk perception

Four papers from three studies addressed the impact of communication on risk perception (Pieterse et al., 2011; Pieterse et al., 2006; Vos, Oosterwijk, et al., 2012; Vos, Stiggelbout, et al., 2011). Two of the papers were from the same longitudinal survey of 248 breast or ovarian cancer patients. Of these, 30 had a pathogenic variant, 16 had a VUS and 202 had no variant (Vos, Siggelbout, et al., 2011; Vos, Oosterwijk, et al., 2012).

Risk perception was affected by health professionals’ communication. Risk was mostly communicated negatively in terms of harm, as a lifetime risk and numerically or qualitatively, (Pieterse et al., 2006). The use of words to convey risk was more successful than other formats. Mirroring risk, for example 80% risk of cancer rather
than 20% chance of not developing cancer, reduced the accuracy of risk perception (Vos, Stiggelbout, et al., 2011).

Risk perception was also affected by patients’ characteristics. Risk perception was more accurate amongst at-risk women than cancer patients. Knowledge was unchanged and anxiety was reduced over time for at-risk women but not for cancer patients (Pieterse et al., 2006). Cancer patients with a pathogenic variant, who considered their cancer to be less severe and who used positive coping styles rather than avoidance strategies had more accurate risk perception than other patients (Vos, Stiggelbout, et al., 2011).

Patients preferred risk to be personalised and presented in general terms, although general lifetime risks were more frequently communicated. Health professionals rarely asked about preferred risk format or existing understanding of risk (Pieterse et al., 2006).

Amongst cancer patients, risk perception was unchanged by the genetic counselling and there was little reduction in anxiety (Pieterse et al., 2011). The only information provided by the genetic counselling that predicted risk perception and risk management intentions concerned the genetic test result, the risk for the patient and the risk for relatives (Vos, Oosterwijk, et al., 2012).

2.5.5. Recall, understanding and interpretation of genetic test results

In total, six studies addressed recall, understanding or interpretation of health professionals’ communication about genetic test results. Of these, two studies included cancer patients and those at risk (Jacobs et al., 2015; van Dijk et al., 2004). The remaining studies were with cancer patients only (Vos, Gomez-Garcia, et al., 2012; Hallowell, 2002; Maheu, & Thorne, 2008; Vos et al., 2008).

Two studies found that cancer patients did not accurately recall information communicated during post-test genetic counselling (Jacobs et al., 2015; Vos, Gomez-Garcia, et al., 2012). In an observational study of recall amongst cancer patients and relatives following disclosure of a BRCA1/BRCA2 pathogenic variant result, the mean percentage of accurate statements recalled by the cancer patients was 53% (Jacobs et al., 2015) (see Chapter 3). A retrospective survey found that although 75% of patients recalled their genetic test result five years after genetic counselling, no more than 30%
of patients were able to accurately recall the associated risks and likelihood of inheritance (Vos, Gomez-Garcia, et al., 2012).

A longitudinal survey of 111 breast cancer patients and 130 at-risk women found no differences in levels of confusion, anxiety, reported understanding of the result, perceived breast cancer risk or psychological distress between cancer patients who received a VUS and those who did not have a variant (van Dijk et al., 2004). A qualitative study of 30 breast and ovarian cancer patients found that those who misinterpreted a VUS result as good news experienced elation or relief, whereas those who correctly understood the result as inconclusive, experienced a range of emotions including disbelief, acceptance, disappointment, anger or frustration (Hallowell et al., 2002). Similar difficulties with interpretation were identified in a qualitative study of 21 breast and ovarian cancer patients who were found not to have a variant. Interpretations of the result varied, with some believing with certainty that they carried a pathogenic variant, some believing that they did not and some expressing uncertainty (Maheu & Thorne, 2008).

In a qualitative study with 24 patients who received a VUS result, 16 recalled the result as not informative and seven recalled the result as a pathogenic variant. Despite factual recall of the result as not informative, 19 participants (79%) interpreted the result to be pathogenic, showing that subjective interpretation of the result was different to factual recall. Amongst the 19 participants who interpreted the result as pathogenic, 10 had undergone risk-reducing surgery in the year following the result (Vos et al., 2008). Receiving a result that showed a pathogenic variant had been detected or that no variant had been detected, directly predicted decisions about surgery or more frequent surveillance. All other decisions that were made as a result of the test were based on interpretation of cancer risk rather the actual communicated risk (Vos, Gomez-Garcia, et al., 2012).

Patients with a pathogenic variant expressed uncertainty about which family members they should communicate genetic risk information to (Hallowell et al., 2002). Low levels of accuracy (30%) were identified amongst the at-risk relatives with whom cancer patients had communicated following disclosure of a pathogenic variant (Jacobs et al., 2015).
2.5.6. Experiences of communication

Five studies investigated experiences of communication about hereditary cancer. Three of these studies (Hallowell et al., 2002; Maheu & Thorne, 2008; Vos et al., 2008) were about recall, understanding or interpretation and the findings are presented in section 2.5.5. Two studies reported the experiences of patients who had received communication about genetic testing shortly after diagnosis (Augestad, Høberg-Vetti, Bjorvatn, & Sekse, 2017; Vadaparampil, Quinn, Brzosowicz, & Miree, 2008).

A qualitative study with nine patients following pre-test genetic counselling prior to (n=6) and after (n=3) definitive surgery, identified lack of understanding about what would be involved and misunderstanding about the inevitability of genetic testing. Patients were unaware even after genetic counselling of the utility of genetic testing and surprised that testing might result in further surgery and heightened emotions (Vadaparampil et al., 2008).

As discussed section 2.5.5., several studies highlighted the confusion and shock experienced by cancer patients upon receiving genetic test results (Hallowell et al., 2002; Maheu & Thorne, 2008; Vos et al., 2008). Patients who underwent genetic testing shortly after diagnosis without pre-test genetic counselling experienced shock, distress and confusion. The emotional turmoil of the cancer diagnosis heightened the difficulty of receiving and comprehending the information communicated in written form only (Augestad et al., 2017) (see section 2.5.8.).

Two studies reported on coping strategies employed by patients receiving genetic test results. Those who were distressed by results that showed no variant had been detected coped by questioning the adequacy of testing, distrusting the result and emphasising the difference between their family history and other higher risk families (Maheu & Thorne, 2008). Those who adopted more positive coping styles had a better understanding of the risks associated with a VUS (Vos et al., 2008).

2.5.7. Communication shortly after cancer diagnosis

In total, 10 studies addressed communication with patients undergoing genetic testing before definitive surgery to guide cancer management. Four studies reported on alternative methods of communication delivered shortly after diagnosis (Augestad et al., 2017; Benusiglio et al., 2017; Quinn et al., 2016; Sie et al., 2014). Three studies investigated the information needs of women undergoing such testing (Gleeson et al., 2013; Meiser et al., 2012; Vadaparampil et al., 2008). Two studies examined
knowledge gained from genetic counselling (Christie, 2012 et al; Scherr et al., 2016) and one study investigated the content of the communication (Vadaparampil et al., 2011).

The quantitative studies found that pre-test communication shortly after diagnosis was acceptable to patients and did not cause distress (Benusiglio et al., 2017; Christie, 2012; Quinn et al., 2016; Scherr et al., 2016; Sie et al., 2014; Vadaparampil et al., 2011).

Three studies indicated that some patients were unprepared for the implications of testing or did not understand the utility of the test (Vadaparampil et al., 2008; Vadaparampil et al., 2011). For some the experience was distressing and overwhelming (see section 2.5.8.) (Augestad et al., 2017).

2.5.8. Alternative methods of communication

Five studies examined alternative methods of communication to face-to-face genetic counselling. Four studies investigated written and/or digital communication with women undergoing genetic testing shortly after breast cancer diagnosis (Augestad et al., 2017; Mancini et al., 2006; Quinn et al., 2016; Sie et al., 2014). One study investigated group education (Benusiglio et al., 2017).

The quantitative studies found that the alternative methods of communication were acceptable and did not cause distress or reduce knowledge (Benusiglio et al., 2017; Quinn et al., 2016; Sie et al., 2014; Vadaparampil et al., 2011). One study reported that groups of up to eight cancer patients received a 20-minute genetics education presentation by a genetic counsellor, followed by 10 minutes for informal questions and answers. The group discussion was followed by a short individual consultation lasting for a mean of 18.4 minutes (range 5-40 minutes). Knowledge and satisfaction were increased (see section 2.5.3.). In addition, genetic counsellor time was saved (Benusiglio et al., 2017).

Two studies involved trials of written information, aimed to supplement (Mancini et al., 2006) or replace genetic counselling (Quinn et al., 2016). Knowledge and satisfaction were increased (see section 2.5.3.)

Two studies (Augestad et al., 2017; Sie et al., 2014) were undertaken as part of the DNA-BONus study investigating genetic testing without face-to-face genetic
counselling (Hoberg-Vetti et al., 2016). A quantitative study compared patients’ satisfaction with genetic testing following written and digital communication with face-to-face genetic counselling. Participants in both groups were satisfied with the amount and quality of the pre-test information they received with 89% stating they would choose the same procedure again and 70% stating that they would recommend the procedure to other patients. No differences were found between the groups for psychological distress, quality of life, or risk perception (Sie et al., 2014).

In a qualitative study involving a small group of DNA-BONus study participants (n=17), four focus groups were undertaken seven to 18 months after completion of genetic testing (Augestad et al., 2017). The study sample included those with and without a pathogenic variant who underwent genetic testing without genetic counselling but with information provided via an information sheet. Participants reported feeling shocked, distressed and overwhelmed by the experience. The emotional turmoil of the cancer diagnosis heightened the difficulty of receiving and comprehending the information. Participants described a feeling of being ‘beside’ themselves. The experience raised issues of altruism and ethical dilemmas and highlighted the need for support and counselling to increase understanding and empower decision-making (Augestad et al., 2017).
Table 2.2. Studies included in the scoping review

<table>
<thead>
<tr>
<th>Author and year</th>
<th>Aims</th>
<th>Sample</th>
<th>Method</th>
<th>Findings relevant to cancer patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>(Augestad, Høberg-Vetti, Bjorvatn, &amp; Sekse, 2017)</td>
<td>To explore experiences of women with newly diagnosed cancer following genetic testing after written information only.</td>
<td>17 breast and ovarian cancer patients</td>
<td>Semi-structured focus group interviews</td>
<td>The experience was shocking, distressing and overwhelming.</td>
</tr>
<tr>
<td>(Benusiglio et al., 2017)</td>
<td>To evaluate group genetic counselling</td>
<td>210 breast and ovarian cancer patients</td>
<td>Longitudinal survey</td>
<td>Knowledge and satisfaction was increased over time.</td>
</tr>
<tr>
<td>(Bredart et al., 2016)</td>
<td>To investigate the impact of genetic knowledge on feelings of personal control.</td>
<td>243 breast cancer patients</td>
<td>Longitudinal survey</td>
<td>Breast cancer knowledge was not retained at post-test genetic counselling.</td>
</tr>
<tr>
<td>(Butow &amp; Lobb, 2004)</td>
<td>To describe the process and content of genetic counselling</td>
<td>158 participants – breast cancer patients (n=69), at-risk women (n=89); genetics health professionals (n=7)</td>
<td>Analysis of audiotaped genetic counselling consultations</td>
<td>Essential information was successfully communicated. Eliciting concerns and facilitating involvement was less successfully communicated. Risk information was infrequently communicated. Knowledge was increased for both groups. Women tested before surgery showed deceased cancer – related stress and intrusive thoughts.</td>
</tr>
<tr>
<td>(Christie et al., 2012)</td>
<td>To investigate changes in cancer-related knowledge and psychological outcomes in women tested before and after definitive surgery.</td>
<td>103 breast cancer patients counselled before (n=16) and after (n=87) definitive breast surgery</td>
<td>Longitudinal survey</td>
<td>Knowledge was increased for both groups. Women tested before surgery showed deceased cancer – related stress and intrusive thoughts.</td>
</tr>
<tr>
<td>(Gleeson et al., 2013)</td>
<td>To identify information and communication preferences about genetic testing shortly after diagnosis for women with ovarian cancer</td>
<td>22 ovarian cancer patients</td>
<td>Semi-structured qualitative interviews</td>
<td>Patients expressed a preference for brief, positive, hope-giving information without statistics early in their diagnosis.</td>
</tr>
<tr>
<td>Study</td>
<td>Objective</td>
<td>Sample</td>
<td>Study Design</td>
<td>Findings</td>
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<tr>
<td>Hallowell et al., 2002</td>
<td>To investigate motivations for testing, information and support needs, and reactions to the test results</td>
<td>30 breast and ovarian cancer patients; carriers (n=10) carriers, no pathogenic variant or VUS result (n=12), awaiting results (n=8)</td>
<td>Semi-structured qualitative interviews</td>
<td>The primary reason for testing was for family members. Waiting for results was not anxiety provoking. Those who misinterpreted a result showing no pathogenic variant or VUS as good news were elated or relieved. Those who correctly interpreted the result as inconclusive felt disbelief, acceptance, disappointment, anger or frustration.</td>
</tr>
<tr>
<td>Jacobs, Dancyger, Smith, &amp; Michie, 2015</td>
<td>To compare the accuracy of information recall amongst patients and relatives following genetic counselling</td>
<td>10 breast and ovarian cancer patients and 22 of their at-risk relatives</td>
<td>Analysis of audiotaped genetic counselling consultations and post consultation interviews</td>
<td>71% of the information communicated during genetic counselling to cancer patients was about hereditary cancer management. Cancer patients accurately recalled 53% of the information.</td>
</tr>
<tr>
<td>Jacobs, Pichert, Harris, Tucker, &amp; Michie, 2017</td>
<td>To identify the key messages about BRCA1/BRCA2 required by women with cancer</td>
<td>16 breast and ovarian cancer patients (service users) and 16 expert genetics and cancer health professionals</td>
<td>Delphi survey</td>
<td>Cancer patients agreed that 35 key messages should be communicated pre-and post-test. Of these, 30 key messages were agreed with health professionals. Disagreements were other cancers associated with BRCA2, diet and lifestyle and risks for non-carriers.</td>
</tr>
<tr>
<td>Lobb et al., 2002</td>
<td>To examine the influence of patients’ individual characteristics on health professionals’ behaviour during genetic counselling</td>
<td>158 participants – breast cancer patients (n=69), at-risk women (n=89); genetics health professionals (n=7)</td>
<td>Analysis of audiotaped genetic counselling consultations</td>
<td>Genetic counsellors discussed more aspects of genetic testing, facilitated patient involvement and used more supportive and counselling behaviours with cancer patients than women at risk.</td>
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<tr>
<td>Reference</td>
<td>Methodology</td>
<td>Participants</td>
<td>Findings</td>
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<tr>
<td>(Lobb et al., 2004)¹</td>
<td>To investigate the effect of communication styles on patient outcomes</td>
<td>As for Lobb et al. (2002)</td>
<td>As for Lobb, et al. (2002) Cancer patients had unmet information needs about risk of contralateral breast cancer and risks for their relatives. Results were shocking and difficult to interpret. Coping strategies included questioning the adequacy of testing, distrusting results and focusing on the similarities and differences with other families.</td>
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<tr>
<td>(Maheu &amp; Thorne, 2008)</td>
<td>To explore the experience of women who receive genetic test result showing no pathogenic variant or VUS has been detected</td>
<td>21 breast and ovarian cancer patients</td>
<td>Semi-structured interviews</td>
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<tr>
<td>(Mancini, Nogues, et al., 2006)</td>
<td>To assess the impact of a standardised information booklet on decision-making</td>
<td>560 breast cancer patients; Trial group (n=297), controls (263)</td>
<td>Quasi-experimental trial, longitudinal survey</td>
<td></td>
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<tr>
<td>(Meiser et al., 2012)</td>
<td>To identify information and communication preferences about genetic testing shortly after diagnosis for breast cancer</td>
<td>26 breast cancer patients diagnosed &lt;50 years</td>
<td>Semi-structured qualitative interviews</td>
<td></td>
</tr>
<tr>
<td>(Metcalfe et al., 2000)</td>
<td>To identify the impact and information needs of women who undergo genetic counselling</td>
<td>79 participants – breast cancer patients (n=46), at-risk women (n=33)</td>
<td>Longitudinal survey</td>
<td></td>
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<tr>
<td>(Pieterse et al., 2006)</td>
<td>To characterise breast cancer risk communication</td>
<td>51 participants - breast cancer patients (n=34), at-risk women (n=17): genetics health professionals (n=10)</td>
<td>Longitudinal survey and video-recording of genetic counselling consultations</td>
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</table>

¹ Lobb, et al. (2002)
<table>
<thead>
<tr>
<th>(Pieterse et al., 2011)</th>
<th>To evaluate outcomes of breast cancer genetic counselling in women with and without breast cancer</th>
<th>77 participants - breast cancer patients (n=44), at-risk women (n=33), genetics health professionals (n=11)</th>
<th>Longitudinal survey and video-recording of genetic counselling consultations</th>
<th>Risk perception improved and anxiety was reduced in at-risk women. Risk perception unchanged and there was less reduction in anxiety for cancer patients.</th>
</tr>
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<tbody>
<tr>
<td>(Quinn et al., 2016)</td>
<td>To evaluate the efficacy of an educational pamphlet in preparing women for decision-making about genetic testing</td>
<td>136 breast cancer patients; Trial group (n=66), controls (n=70)</td>
<td>Randomised controlled non-inferiority trial, longitudinal survey</td>
<td>There were no differences between the groups for variance in knowledge and psychological outcomes.</td>
</tr>
<tr>
<td>(Randall, Butow, Kirk, &amp; Tucker, 2001)</td>
<td>To investigate the psychological impact and knowledge gained from genetic counselling and testing in women with breast cancer</td>
<td>64 breast cancer patients; Genetic counselling group (n=34), controls (n=30)</td>
<td>Analysis of audiotaped genetic counselling consultations</td>
<td>There was no difference in psychological impact or knowledge gain between the groups.</td>
</tr>
<tr>
<td>(Scherr, Christie, &amp; Vadaparampil, 2016)</td>
<td>To explore the impact of genetic counselling on breast cancer survivors' knowledge about hereditary cancer over time</td>
<td>103 breast cancer patients; Counselling before definitive breast surgery (n=16), counselled after definitive breast surgery (n=87)</td>
<td>Longitudinal survey</td>
<td>The knowledge gained following pre-test genetic counselling was not retained at 6 months.</td>
</tr>
<tr>
<td>(Sie et al., 2014)</td>
<td>To compare experiences of patients receiving pre-test information via written/digital formats with usual care</td>
<td>161 breast cancer patients; Trial group (n=95), usual care group (n=66)</td>
<td>Survey</td>
<td>There were no differences in satisfaction, psychological distress, quality of life, breast cancer worry, risk perception for further cancer between groups.</td>
</tr>
<tr>
<td>(Vadaparampil, Quinn, Brzosowicz, &amp; Miree, 2008)</td>
<td>To understand the experiences of breast cancer patients who have genetic counselling and testing prior to or after completing definitive cancer surgery.</td>
<td>9 breast cancer patients; Tested prior to definitive treatment (n=3), tested after definitive treatment (n=6)</td>
<td>Semi-structured interviews</td>
<td>There were no differences in motivation for testing, influence of family on decision-making or expectations of testing between groups. Patients tested before surgery were unprepared for the implications of testing.</td>
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<tr>
<td>Reference</td>
<td>Study Objective</td>
<td>Methods</td>
<td>Results</td>
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<tr>
<td>(Vadaparampil et al., 2011)</td>
<td>To evaluate satisfaction with the timing and strength of recommendations and information received prior to and during genetic counselling among breast cancer patients</td>
<td>Survey</td>
<td>There was high satisfaction about the timing and strength of the recommendation for genetics referral. Patients had low levels of expectations pre-counselling and limited understanding of the process.</td>
<td></td>
</tr>
<tr>
<td>(van Dijk et al., 2004)</td>
<td>To compare breast cancer risk and distress in women who receive different genetic test results</td>
<td>Longitudinal survey</td>
<td>There was no greater confusion or anxiety amongst VUS group than no pathogenic variant or VUS group. No differences were seen between the groups for understanding, perceived risk or distress after result.</td>
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<tr>
<td>(van Roosmalen et al., 2004)</td>
<td>To evaluate the impact of BRCA1/2 testing and disclosure of a positive test result on cancer patients and women at risk</td>
<td>Randomised longitudinal survey</td>
<td>For both groups anxiety, depression and cancer related distress increased and general health decreased over time. Anxiety and cancer related distress was higher amongst cancer patients diagnosed ≤1 year than those tested ≥1 year. Intention to have risk-reducing surgery was higher among patients than at-risk women. Risk perception increased when a pathogenic variant was identified, VUS discussed pre-test, risk communicated in words, cancer perceived to be less severe and positive coping styles used.</td>
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<tr>
<td>(Vos et al., 2008)</td>
<td>To explain why cancer patients inaccurately perceive cancer risks when a VUS is detected</td>
<td>Semi-structured interviews and five-point Likert scales</td>
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<tr>
<td>Study</td>
<td>Objective</td>
<td>Research Design</td>
<td>Methodology</td>
<td>Findings</td>
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<tr>
<td>(Vos et al., 2011)</td>
<td>To investigate accuracy of risk perception following genetic counselling</td>
<td>Longitudinal survey</td>
<td>248 breast/ovarian cancer patients. Pathogenic variant (n=30), VUS (n=16), no pathogenic variant or VUS (n=202)</td>
<td>Medical decisions were influenced by perception of risk. The genetic test result and risk influenced outcomes.</td>
</tr>
<tr>
<td>(Vos, Gomez-Garcia, et al., 2012)</td>
<td>To quantify the effect that perception has in genetic counselling for hereditary breast/ovarian cancer</td>
<td>As for Vos et al. (2011)</td>
<td>As for Vos et al. (2011)</td>
<td>Five years after genetic counselling recall of the result was accurate but perception of the cancer risk and inheritance implications were inaccurate. A pathogenic variant and no pathogenic variant or VUS result were the only factors that predicted decisions about surgery and surveillance.</td>
</tr>
<tr>
<td>(Vos, Oosterwijk, et al., 2012)</td>
<td>To investigate the short-term outcomes of communicating a genetic test result</td>
<td>As for Vos et al. (2011)</td>
<td>As for Vos et al. (2011)</td>
<td>Following genetic counselling risk management decisions were influenced by the perception of risk rather than by the communicated risks. The only counselling information that directly predicted counsellees' perceptions and indirectly predicted outcomes were the DNA test result, the risk for the patient and the risk for her relatives.</td>
</tr>
</tbody>
</table>

1 Papers from the same Australian study
2 Papers from the same Netherlands study
2.6. DISCUSSION

Prior to 2008, most studies of communication about hereditary breast and ovarian cancer focused on at-risk women. This scoping review identified 29 papers from 25 studies in which findings were reported for cancer patients. Patients identified a need for cancer-focused, personalised information. Genetic counselling focused on biomedical information-giving with less attention to psychological aspects. Pre-test genetic counselling increased knowledge without raising anxiety. For some patients however, knowledge was reduced and anxiety increased following genetic test results. Personal interpretation rather than genetic counselling affected risk perception. Inaccurate recall, misunderstanding or incorrect interpretation of ambiguous genetic test results led to inappropriate risk management decisions and hindered family communication. For some patients, the experience of genetic counselling and results disclosure was shocking, confusing and distressing. Pre-test communication shortly after diagnosis was acceptable to most patients. For those who were unprepared or unsupported, the experience was overwhelming. Despite the increasing need to deliver genetic testing in mainstream oncology, all the studies included in this review addressed communication in the clinical genetics setting.

Cancer patients tested before and after definitive surgery expressed the need for information about the risks of further cancers and the implications of genetic testing on treatment and risk management options (Lobb et al., 2004; Metcalfe et al., 2000). Two studies found that patients tested shortly after diagnosis need brief treatment-focused information prior to testing, with family-related information at a later time (Gleeson et al., 2013; Meiser et al., 2012). A further study by the same group confirmed this finding (Zilliacus, 2012). However, neither service users nor expert health professionals considered information about the impact of the genetic test on treatment to be a key message (Jacobs et al., 2017). The need for information about treatment may vary, with some patients wanting more and some wanting less detailed information.

It is less clear why patients tested after completing cancer treatment wanted information about the impact of testing on treatment (Metcalfe et al., 2000). One possible explanation is the high level of information need and desire to be kept informed that many cancer patients express (Jenkins et al., 2001). Information is often regarded as a method of reducing uncertainty and providing a sense of personal control as well as enabling informed decisions about treatment options (Collins et al., 2009; McInnes et al., 2008). However, even amongst cancer patients who consider
themselves to be well informed, there is a desire for more information, indicating that satisfaction with communication does not equate with information needs being met and that for some patients no amount of information is sufficient (Faller et al., 2016).

Genetic counselling of cancer patients focused on biomedical information-giving (Butow & Lobb, 2004; Lobb et al., 2004). Although more attention was paid to psychological support for cancer patients than at-risk women, health professionals infrequently facilitated involvement in the communication and did not always provide the support needed (Lobb et al., 2002). Several previous studies have reported similar findings about genetic counselling with at-risk women (Ellington et al., 2005; Lobb, Butow, Barratt, Meiser, & Tucker, 2005; Meiser et al., 2008; Pieterse, van Dulmen, Ausems, Beemer, & Bensing, 2005; Roter et al., 2006).

Despite the need for cancer-related information (Jacobs et al., 2017; Lobb et al., 2004; Metcalfe et al., 2000), the risk of contralateral breast cancer was infrequently communicated (Lobb et al., 2004; Pieterse et al., 2006). A great deal of information is communicated during genetic counselling (Roter et al., 2006). Concern about overloading patients with information or reluctance to raise issues about future cancer risk may have prevented health professionals from discussing these issues.

Pre-test communication increased knowledge and did not increase anxiety or cancer-related distress (Benusiglio et al., 2017; Christie, 2012; Mancini et al., 2006; Quinn et al., 2016; Randall et al., 2001; Scherr et al., 2016). Similar findings have been reported following pre-test cancer genetic counselling of at-risk individuals (Albada, van Dulmen, Lindhout, Bensing, & Ausems, 2012; Kelly et al., 2004).

For some patients, disclosure of the genetic test result led to a reduction in knowledge compared to pre-test counselling and/or an increase in cancer-related distress (Bredart et al., 2016; Pieterse et al., 2011; Scherr et al., 2016; van Roosmalen et al., 2004). Previous studies have also demonstrated an increase in distress amongst cancer patients following genetic testing, highlighting the impact of the cancer experience on responses to the result disclosure (Hallowell, Foster, Eeles, Ardern-Jones, & Watson, 2004; Meiser, 2005).

Knowledge measures included modified tools that had been validated with at-risk women (Erblich et al., 2005; Lerman et al., 1997; Lerman et al., 1996; Zellerino et al.,
and those developed purposefully for the included studies. The reliability of these measures for cancer patients is not known.

Risk perception following genetic test results was affected by health professionals’ communication style and patients’ characteristics (Pieterse et al., 2006; Vos, Stiggelbout, et al., 2011).

Personal interpretation rather than genetic counselling appeared to influence risk perception amongst cancer patients (Pieterse et al., 2011; Vos, Oosterwijk, et al., 2012). The difficulty of processing uncertain or ambiguous information may lead patients to interpret the result in order to make sense of it (Vos et al., 2011). In contrast, systematic reviews have concluded that genetic counselling improves risk perception in individuals at risk of breast cancer (Butow, Lobb, Meiser, Barratt, & Tucker, 2003; Meiser & Halliday, 2002) and other genetic conditions (Smerecnik, Mesters, Verweij, De Vries, & De Vries, 2009).Unlike testing of cancer patients, predictive genetic testing of at-risk individuals provides a clear result, reducing the need for personal interpretation of risk.

Several studies found that recall, understanding or interpretation of genetic test results was frequently inaccurate, particularly when no pathogenic variant was detected (Hallowell et al., 2002; Maheu & Thorne, 2008; van Dijk et al., 2005; Vos, Gomez-Garcia, et al., 2012; Vos et al., 2008). Even when results were accurately recalled, they were often misunderstood or misinterpreted (Vos, Gomez-Garcia, et al., 2012).

Inaccuracies in understanding of results may lead to inappropriate risk management decisions (Vos, Gomez-Garcia, et al., 2012; Vos et al., 2008). Many breast cancer patients without a pathogenic variant opt for a contralateral mastectomy (Davies et al., 2016). Inappropriate uptake of surgery may be compounded by a lack of understanding of genetic test results amongst breast surgeons (Eccles, Copson, Maishman, Abraham, & Eccles, 2015) and other non-genetics physicians (Richter, Graham, Haroun, Eisen, & Warner, 2012).

Two studies suggested that inaccurate recall or understanding of information about a pathogenic variant may hinder family communication (Hallowell et al., 2002; Jacobs et al., 2015). Several barriers to family communication about genetic information have been identified, including lack of close relationship (Forrest et al., 2003), family culture (Dancyger et al., 2011) and reluctance to distress relatives (Hughes et al., 2002). Lack
of understanding of the risks and benefits of genetic information has been identified as a barrier to family communication in previous studies (Claes et al., 2003; Gallo, 2009).

Qualitative studies found that patients experienced shock and confusion on receiving genetic test results (Hallowell et al., 2002; Maheu & Thorne, 2008; Vos et al., 2008). Some patients tested shortly after diagnosis also experienced feelings of distress (Augestad et al., 2017; Vadaparampil et al., 2008).

For many patients, communication about hereditary cancer shortly after diagnosis was acceptable and did not cause distress (Benusiglio et al., 2017; Christie, 2012; Quinn et al., 2016; Sie et al., 2014; Vadaparampil et al., 2011). Several studies of the acceptability of testing shortly after diagnosis have found that most patients are satisfied with the timing of testing and that testing facilitates patients’ decision-making (Schlich-Bakker et al., 2008; Wevers et al., 2017; Zilliacus, 2012). For patients who are unprepared or unsupported however, genetic testing shortly after diagnosis may be overwhelming and distressing (Augestad et al., 2017; Vadaparampil et al., 2008). Patients and health professionals have expressed concerns about the potential impact of genetic testing close to cancer diagnosis (Ardern-Jones, Kenen, & Eeles, 2005; Lobb, Barlow-Stewart, Suthers, & Hallowell, 2010).

The potential for targeted cancer treatment has led to a sharp rise in the demand for genetic testing amongst cancer patients. Meeting this demand requires the development of new approaches to information delivery. Several studies evaluated alternative methods of information delivery to face-to-face genetic counselling (Augestad et al., 2017; Benusiglio et al., 2017; Mancini et al., 2006; Quinn et al., 2016; Sie et al., 2014). For most patients, these methods were found to be acceptable and one study reported that patients found the alternative form of communication preferable to face-to-face counselling (Sie et al., 2014). Previous studies have also reported no differences in knowledge, distress or satisfaction amongst patients who have received genetics communication within the clinical genetics setting via group education (Calzone et al., 2005; Manchanda et al., 2016), (Doughty Rice, Ruschman, Martin, Manders, & Miller, 2010; Patrick-Miller et al., 2013) or video-conferencing (Bradbury et al., 2016; E. M. Ziliacus et al., 2011). For many cancer patients, pre-test communication without individual face-to-face genetic counselling may be acceptable.

Several trials have investigated the timing, uptake and acceptability of genetic testing in the oncology setting (George et al., 2016; Katz, Kurian, & Morrow, 2015; Kentwell et
al., 2017; Percival et al., 2016; Wevers et al., 2017; Zilliacus, Meiser, Gleeson, Watts, Tucker, Lobb & Mitchell, 2012). These studies have not however investigated the information that is communicated, the method of communication, patients’ and relatives’ recall or understanding following the communication or patients’ experiences of the communication.

2.7. LIMITATIONS

This scoping review maps the current range of evidence specific to health professionals’ communication about hereditary cancer with cancer patients. This review should not be considered a definitive review of the literature in this field as, consistent with the methodology, no quality assessment was made. Due to the breadth of the studies reviewed, some areas of research only include a few studies. It is only possible to draw tentative conclusions about the clinical implications from these findings.

2.8. CLINICAL IMPLICATIONS

The findings of this review suggest that greater attention may be needed to the psychological and supportive aspects of genetic counselling. If pre-test communication is increasingly to be provided via methods other than face-to-face genetic counselling, the post-test genetic counselling appointment may be the only opportunity for cancer patients to interact with a genetics health professional.

Some patients experience misunderstanding, reduced knowledge and raised anxiety at results disclosure. Patients with a pathogenic variant and those with a VUS may need additional help post-test to understand the implications of genetic test results and support with decision-making about cancer management and cancer risk management.

Helping patients and families to adjust to a genetic diagnosis and facilitating dissemination of genetics information within families are likely to become more important for genetics health professionals than delivering biomedical information.
The limited studies of experiences of communication suggest that some patients may need enhanced counselling and support throughout the genetic testing process.

To meet the demand for more genetic testing, new approaches to communication will need to be developed and evaluated. It will be important to learn from current practice in developing new communication methods.

2.9. RESEARCH IMPLICATIONS

This review highlighted gaps in knowledge about the information and support needs of cancer patients and the extent to which these needs are met by health professionals’ communication. Understanding the information that should be provided to cancer patients will be helpful in developing new approaches within the oncology setting.

Although some studies identified in this review did investigate the process and content of genetic counselling with cancer patients and at-risk women, no studies investigated this area exclusively with cancer patients during pre- or post-test genetic counselling. There is a need to understand the information that is communicated and the method of communication about hereditary cancer between cancer patients and oncology health professionals.

This review highlighted the confusion patients experience when they are informed about ambiguous genetic test results, the lack of influence of genetic counselling on patients’ interpretation of VUS results and the potential clinical impact of misinterpretation. As more tests are undertaken, more patients will be found to have a VUS. Research to investigate what is communicated about VUS results and how patients make decisions based on their interpretation of the result will become increasingly important.

Despite the growing number of patients undergoing genetic testing shortly after cancer diagnosis, very few studies identified by this review investigated patients’ experience and understanding of testing at this vulnerable stage in their cancer trajectory. Further research in this area is needed to inform health professionals’ communication.
2.10. CONCLUSIONS

Most studies of communication with cancer patients about hereditary cancer have focused on the cognitive and psychological impact of genetic counselling. Few studies have investigated information needs, process and content, recall and understanding or experiences of communication shortly after diagnosis. Although several studies have investigated alternative methods of communication to face-to-face genetic counselling, no studies of involving communication by non-genetics health professionals were identified. This review found that patients’ need for cancer-focused, personalised information is not always met by genetic counselling. Genetic counselling tends to focus on biomedical information-giving at the expense of psychological support. For most patients, knowledge is increased and anxiety is not raised by pre-test communication. However, some patients experience confusion and distress when results are disclosed. For patients who are unprepared or unsupported, communication about genetic testing shortly after diagnosis may be overwhelming. As genetic testing becomes further integrated into mainstream oncology with less involvement from genetics health professionals, it will be important to find ways to identify patients who need specialist counselling and support throughout the process.
3.1. INTRODUCTION

Once a pathogenic variant has been identified in a patient with breast or ovarian cancer, it is important that accurate information about genetic risk and the risk management options is communicated to at-risk relatives. Disseminating information within the family once a pathogenic variant has been detected falls to the patient. Supporting and facilitating this communication is a key role of the genetics health professional. Yet little is known about the accuracy of the information that is recalled by cancer patients or communicated to relatives by cancer patients following identification of a pathogenic variant.

3.2. BACKGROUND

The responsibility for sharing information about a newly identified pathogenic variant falls to the individual with cancer who receives the initial news (Hayat Roshanai, Lampic, Rosenquist, & Nordin, 2010). Families prefer information to be communicated by the patient rather than by the health professional (Forrest et al., 2003). An international review of guidelines about family communication concluded that families have a moral obligation to pass genetic information on (Forrest, Delatycki, Skene, & Aitken, 2007) and it seems that most families do share this information (Hayat Roshanai et al., 2010).

However, there are several well-documented barriers to family communication, including difficulties with relationships and complexity of information. Several studies have shown that genetic information is more commonly shared with first and second degree relatives, especially sisters (Hughes et al., 2002), or with siblings and children (Claes et al., 2003). Female relatives are more commonly informed about a cancer predisposing BRCA1/BRCa2 mutation among patients and relatives. European Journal of Human Genetics 23(2), 147-151 (Appendix 4.1.)
predisposing BRCA1/BRCA2 pathogenic variant than male relatives (McGivern et al., 2004). Other studies have highlighted the influence of the relationship between the index patient and relatives on family communication, (Dancyger, Smith, Jacobs, Wallace, & Michie, 2010; Forrest et al., 2003; McGivern et al., 2004; Patenaude et al., 2006) and the importance of family culture (Dancyger et al., 2011). Other influences on disclosure of information within families include patients’ perceived risks and benefits of the information (Gallo, 2009), personal beliefs about the causes of genetic illness (McAllister, 2003; Michie, Smith, Senior, & Marteau, 2003) and reluctance to upset relatives (Hughes et al., 2002). The information that relatives receive and understand about the result is likely to influence their risk perception and decision-making about screening and genetic testing (Forrest et al., 2003; Peterson et al., 2003). The rate of uptake of genetic counselling by relatives of individuals who carry a pathogenic variant is reported to be between 20% and 40% (Barsevick et al., 2008). One study found that in families with low uptake, index patients expressed a retrospective need for greater support with family communication (Landsbergen, Verhaak, Kraaimaat, & Hoogerbrugge, 2005), highlighting the important facilitative role of genetics health professionals.

Genetic information is complex and difficult to understand (Cypowyj et al., 2009), yet giving information is an integral component of genetic counselling to enable informed decision-making (Resta et al., 2006; Shiloh, Avdor, & Goodman, 1990). Awareness and knowledge about cancer genetics amongst the general population has increased since the 1990s (Bluman et al., 1999; Mogilner, 1998), particularly about hereditary breast and ovarian cancer. However, a survey of US citizens found that only 40.2% of adults had heard of genetic testing for cancer risk and only 50% of those at the highest risk had heard of genetic testing. Awareness was lowest amongst males and minority ethnic groups (Baer, Brawarsky, Murray, & Haas, 2010). The same study also found that primary care physicians often did not discuss hereditary cancer risk and genetic testing with their at-risk patients, highlighting the importance of accurate information being passed on within the family.

Although several studies have examined accuracy of recall amongst patients following genetic counselling for conditions such as Down Syndrome (Abramovsky, Godmilow, Hirschhorn, & Smith, 1980; Swerts, 1987), there have been few studies of accuracy amongst cancer patients following genetic counselling or amongst their relatives following family communication. In a Belgian study of 107 first-degree relatives of 14 patients with a BRCA1/BRCA2 pathogenic variant, participants were asked about their
knowledge of the variant, cancer risks, risk-reducing options and genetic testing (Sermijn et al., 2004). Less than 47% of participants were aware of the existence of hereditary breast and ovarian cancer, cancer risks, the options for reducing cancer risks, the possibility of predictive testing. Less than 14% of participants were aware of autosomal dominant inheritance and less than 4% of women under age 50 and men of any age were aware of the possibility of prenatal diagnosis. Awareness of hereditary breast and ovarian cancer in the family was also low. The study found that women, younger participants and those with a close relationship to the patient were better informed. The researchers found no statistical difference between the awareness of the original 14 patients and the family members regarding knowledge of inheritance and attitudes to genetic testing and counselling. The authors concluded that the transfer of information from patients to their relatives was highly defective and recommended that at-risk relatives of carriers of a pathogenic variant should be systematically informed of the genetic test result by letter.

A more recent study (Vos, Jansen, et al., 2011) examined how BRCA1/BRCA2 genetic test results, were communicated and perceived within families. The researchers hypothesised that as the information passed through the family it would ‘fade out’. The study investigated how information changed and whether the transfer of information from patient to relatives was influenced by socio-demographic factors, relationship and cancer history. Twenty-five patients who had received a BRCA1/BRCA2 genetic test result between 1998 and 2008 and 70 of their first and second degree at-risk relatives agreed to participate. Participating relatives were sent a questionnaire asking about recollections and interpretations of cancer risks and the likelihood of inheritance. The information recalled by the patients reflected the information provided by the genetic counsellor, but there were few similarities between relatives’ perception of the information they received and the information actually communicated by the genetic counsellor to the patient. The authors concluded that the information was re-interpreted at each stage of the information transfer.

Despite the fact that a key role of genetic counselling is to encourage and facilitate family communication (Chivers Seymour, Addington-Hall, Lucassen, & Foster, 2010; Hayat Roshanai et al., 2010), an international review found that none of the genetics guidelines about family communication detailed how this information should be communicated or what information should be communicated (Forrest et al., 2007). A survey of genetics health professionals worldwide found that, although 90% of participants stated that they always identify at-risk relatives and encourage family
communication, 41% never write a letter specifically for at-risk relatives. The main reason given was lack of time and administrative support (Forrest, Delatycki, Curnow, Skene, & Aitken, 2010).

3.3. AIMS AND RESEARCH QUESTIONS

This observational study aimed to track the information communicated in genetics consultations with patients through to the families to address the following research questions.

Following post-test genetic counselling where a cancer predisposing BRCA1/BRCA2 pathogenic variant was discussed,

1. How accurate is the information reported by patients and their at-risk relatives?
2. Is there a difference in the accuracy of information about genetic testing and hereditary cancer management reported by patients and their relatives?
3. Is there an association between the accuracy of the information reported by relatives and the source from which they received the information (i.e. information received solely from the patient or directly from genetics health professional as well as from the patient)?

3.4. METHODS

This study was part of the Family Communication Study (see section 1.8.5.5).

3.4.1. Sample

The Study 1 sample consisted of ten patients with breast and/or ovarian cancer and 22 of their relatives including at least two at-risk first, second or third-degree relatives of each patient. The mean age of the index patients was 55.5 years (range 34 to 71). The mean age of the relatives was 37.1 years (range 20 to 65). BRCA status, type of cancer and age at diagnosis for patients and relatives, and the relatives’ relationship to the patient is shown in Table 3.1.
Table 3.1. Demographic data: showing patients' cancer type, age at diagnosis, gene affected and age at genetic testing; relatives' relationship to patient, personal history of cancer, age and genetic status

<table>
<thead>
<tr>
<th>Family</th>
<th>Patient</th>
<th>Relative 1</th>
<th>Relative 2</th>
<th>Relative 3</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Fallopian tube cancer (ovarian) diagnosed 53y</td>
<td>Sister age 50y Breast cancer age 45y</td>
<td>Daughter age 28y No personal history of cancer</td>
<td>n/a</td>
</tr>
<tr>
<td></td>
<td>BRCA1 Age 55y</td>
<td>BRCA1 untested</td>
<td>BRCA1 untested</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>Breast cancer diagnosed age 28y</td>
<td>Niece age 38y No personal history of cancer</td>
<td>Sister age 50y No personal history of cancer</td>
<td>n/a</td>
</tr>
<tr>
<td></td>
<td>BRCA2 Age 38y</td>
<td>BRCA2 –ve</td>
<td>BRCA2 +ve</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>Breast cancer diagnosed age 37y &amp; 59y</td>
<td>Daughter age 41y No personal history of cancer</td>
<td>Paternal cousin (female) age 64y Ovarian cancer diagnosed 55y</td>
<td>n/a</td>
</tr>
<tr>
<td></td>
<td>BRCA2 Age 60y</td>
<td>BRCA2 +ve</td>
<td>BRCA2 –ve</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>Breast &amp; ovarian cancer diagnosed age 70y</td>
<td>Niece age 34y No personal history of cancer</td>
<td>Daughter age 33y No personal history of cancer</td>
<td>n/a</td>
</tr>
<tr>
<td></td>
<td>BRCA1 Age 71y</td>
<td>BRCA1 untested</td>
<td>BRCA1 awaiting result</td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>Breast cancer diagnosed 59y</td>
<td>Son age 39y No personal history of cancer</td>
<td>Son age 35y No personal history of cancer</td>
<td>n/a</td>
</tr>
<tr>
<td></td>
<td>BRCA2 Age 64y</td>
<td>BRCA2 untested</td>
<td>BRCA2 –ve</td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>Breast cancer diagnosed 34y &amp; ovarian diagnosed 63y</td>
<td>Sister age 65y Oral cancer diagnosed 63y</td>
<td>Brother age 60y No personal history of cancer</td>
<td>Brother age 49y No personal history of cancer</td>
</tr>
<tr>
<td></td>
<td>BRCA1 Age 64y</td>
<td>BRCA1 +ve</td>
<td>BRCA1 awaiting result</td>
<td>BRCA1 awaiting result</td>
</tr>
<tr>
<td>7</td>
<td>Ovarian cancer diagnosed 60y</td>
<td>Son age 37y No personal history of cancer</td>
<td>Son age 35y No personal history of cancer</td>
<td>n/a</td>
</tr>
<tr>
<td></td>
<td>BRCA2 Age 63y</td>
<td>BRCA2 untested</td>
<td>BRCA2 untested</td>
<td></td>
</tr>
<tr>
<td>8</td>
<td>Breast cancer diagnosed 40y</td>
<td>Daughter age 20y No personal history of cancer</td>
<td>First cousin age 37y No personal history of cancer</td>
<td>Breast cancer diagnosed 51y</td>
</tr>
<tr>
<td></td>
<td>BRCA1 Age 43y</td>
<td>BRCA1 untested</td>
<td>BRCA1 untested</td>
<td>BRCA1 –ve</td>
</tr>
<tr>
<td>9</td>
<td>Breast cancer diagnosed 33y</td>
<td>Sister age 21y No personal history of cancer</td>
<td>Sister age 22y No personal history of cancer</td>
<td>n/a</td>
</tr>
<tr>
<td></td>
<td>BRCA1 Age 34y</td>
<td>BRCA1 untested</td>
<td>BRCA1 untested</td>
<td></td>
</tr>
<tr>
<td>10</td>
<td>Breast cancer diagnosed 49y &amp; ovarian diagnosed 60y</td>
<td>Daughter age 26y No personal history of cancer</td>
<td>Daughter age 29y No personal history of cancer</td>
<td>n/a</td>
</tr>
<tr>
<td></td>
<td>BRCA1</td>
<td>BRCA1 untested</td>
<td>BRCA1 untested</td>
<td></td>
</tr>
</tbody>
</table>
3.4.2. Procedure

3.4.2.1. Data collection
For the patients, demographic information was documented about the pathogenic variant, the type of cancer, age at diagnosis and the age at interview. For the relatives, information was documented about the age at interview, gender, relationship to the patient, cancer history, experience of genetic testing and the sources of their information.

Semi-structured interviews were carried out with the patients approximately four weeks after receiving the genetic test result. Patients were asked about their understanding of their genetic risk and implications for themselves and family, whether they had informed relatives of the result, how and what information that had given to relatives and how this was received. Semi-structured interviews were subsequently carried out with the relatives. Relatives were asked what they were told about the genetic test result by the patient, how the information had been communicated to them, how they reacted to the information, their perceptions of their own risk, their intended actions based on the information and the sources of their information. The interview schedules are shown in Appendix 2.1.

3.4.2.2. Identifying information provided by genetics health professionals to patients
The transcripts of the clinic consultations with the patients and the letters summarising the main points of the consultation sent by genetics health professionals to the patients were systematically searched for ‘chunks’ of information communicated. These ‘chunks’ of information were documented verbatim on a matrix for each family. Once all the information communicated by the health professionals had been identified, similar chunks of information were grouped into categories and each category was summarised as a statement. All information communicated by health professionals was categorised in this way, even if it had been communicated to just one patient. Figure 3.1 shows an example of the method of developing a category statement drawing on the information communicated in clinical consultations and summary letters. This process was used to develop all category statements.
Figure 3.1. Development of the category statement ‘Breast cancer risk is increased by the pathogenic variant’

<table>
<thead>
<tr>
<th>Patient 1</th>
<th>Patient 2</th>
<th>Patient 3</th>
<th>Patient 4</th>
<th>Patient 5</th>
</tr>
</thead>
<tbody>
<tr>
<td>There is for female family members a very substantial increased risk for breast cancer. It’s anywhere between 50% and 80% (consultation)</td>
<td>For women who carry these types of mistakes in one of these two genes, the risk of getting breast cancer is high (consultation)</td>
<td>The lifetime chance of someone developing breast cancer who has a BRCA2 alteration is up to about 80% (consultation)</td>
<td>Girls would be at increased risk of developing breast and ovarian cancer (letter)</td>
<td>Not mentioned</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Patient 6</th>
<th>Patient 7</th>
<th>Patient 8</th>
<th>Patient 9</th>
<th>Patient 10</th>
</tr>
</thead>
<tbody>
<tr>
<td>Not mentioned</td>
<td>The risk to a woman who had never had breast cancer who carries this gene fault is up to 80% lifetime risk (letter)</td>
<td>It means that you’ve got a higher risk of developing breast cancer… and ovarian cancer as well (consultation)</td>
<td>If she has inherited that fault she would have an increased breast cancer risk kind of starting from the age of 30 (consultation)</td>
<td>(The) chances … can be up to about 80% for breast cancer. (consultation)</td>
</tr>
</tbody>
</table>

The category statements were grouped into similar domains of information. The domains of information were grouped into two main domains. Within the main domain of ‘Genetic testing information’ were: ‘inheritance’, ‘the gene’ and ‘genetic counselling and testing for relatives’ (see Figure 3.2.). Within the main domain of ‘Hereditary cancer management information’ were: ‘cancer risk for women who have had BRCA1/BRCA2 related cancer’, ‘cancer risk for individuals who have not had BRCA1/BRCA2 related cancer’ and ‘cancer risk management options’ (see Figure 3.3.).
Figure 3.2. Category statements and domains within ‘Genetic testing information’

<table>
<thead>
<tr>
<th>Category statements</th>
<th>Domain</th>
</tr>
</thead>
<tbody>
<tr>
<td>Each child and sibling of a person with the pathogenic variant has 50% risk of inheriting the pathogenic variant</td>
<td>Inheritance</td>
</tr>
<tr>
<td>Identifying the side of the family that is at risk</td>
<td></td>
</tr>
<tr>
<td>The pathogenic variant doesn’t skip a generation</td>
<td></td>
</tr>
<tr>
<td>The pathogenic variant explains the family history</td>
<td></td>
</tr>
<tr>
<td>The pathogenic variant is in BRCA1/BRCA2</td>
<td></td>
</tr>
<tr>
<td>Not everyone with the pathogenic variant will develop cancer</td>
<td></td>
</tr>
<tr>
<td>The role of the gene</td>
<td></td>
</tr>
<tr>
<td>Predictive testing is available</td>
<td></td>
</tr>
<tr>
<td>Individuals who have a negative predictive test are not at increased risk of cancer</td>
<td></td>
</tr>
<tr>
<td>There is no obligation for family to have a predictive test</td>
<td></td>
</tr>
<tr>
<td>It is important to inform relatives about the pathogenic variant</td>
<td></td>
</tr>
<tr>
<td>Genetic testing is not recommended at a young age</td>
<td></td>
</tr>
<tr>
<td>Genetic counselling is required before genetic testing</td>
<td>Genetic counselling/testing for relatives</td>
</tr>
<tr>
<td>Genetic testing is a personal choice</td>
<td></td>
</tr>
<tr>
<td>Genetic test results will not affect insurance</td>
<td></td>
</tr>
</tbody>
</table>
**Figure 3.3. Category statements and domains within ‘Hereditary cancer management information’**

<table>
<thead>
<tr>
<th>Category statements</th>
<th>Domain</th>
</tr>
</thead>
<tbody>
<tr>
<td>Women who have had breast cancer are at increased risk of a second breast cancer</td>
<td>Cancer risk for women who have had breast or ovarian cancer</td>
</tr>
<tr>
<td>The patient can outlive her risk</td>
<td></td>
</tr>
<tr>
<td>The patient's risk is still rising</td>
<td></td>
</tr>
<tr>
<td>It is important to wait a while after cancer treatment before undergoing risk-reducing surgery and important to balance the risks of new cancer and recurrence</td>
<td></td>
</tr>
<tr>
<td>Most BRCA related Breast cancer is ER-ve</td>
<td></td>
</tr>
<tr>
<td>The patient is at increased risk of ovarian and primary peritoneal cancer</td>
<td></td>
</tr>
<tr>
<td>Much still unknown re risks</td>
<td></td>
</tr>
<tr>
<td>Treatment trials available</td>
<td></td>
</tr>
<tr>
<td>Chemo will be required for further cancer and further cancer will be no more difficult to treat</td>
<td></td>
</tr>
<tr>
<td>The mutation does not cause recurrence of cancer</td>
<td></td>
</tr>
<tr>
<td>The mutation doesn't change things for the patient</td>
<td></td>
</tr>
<tr>
<td>Breast cancer risk is increased</td>
<td>Cancer risk for women who have NOT had breast or ovarian cancer</td>
</tr>
<tr>
<td>Breast cancer risk starts to rise at 30/occurs at young age</td>
<td></td>
</tr>
<tr>
<td>Older relatives have lived through some of their risk</td>
<td></td>
</tr>
<tr>
<td>Ovarian cancer risk is increased</td>
<td></td>
</tr>
<tr>
<td>Ovarian cancer risk starts to rise at 40</td>
<td></td>
</tr>
<tr>
<td>Prostate cancer risk is not increased/is increased</td>
<td></td>
</tr>
<tr>
<td>Male breast cancer risk is not increased/is increased</td>
<td></td>
</tr>
<tr>
<td>Pancreatic cancer risk increased</td>
<td></td>
</tr>
<tr>
<td>Diet and lifestyle can help to protect from cancer</td>
<td></td>
</tr>
<tr>
<td>Cancer risk starts to rise in late 20s/early 30s</td>
<td></td>
</tr>
<tr>
<td>Risk-reducing mastectomy is an option</td>
<td>Cancer risk management options</td>
</tr>
<tr>
<td>Reconstruction is an option</td>
<td></td>
</tr>
<tr>
<td>Reconstruction is cosmetically better nowadays</td>
<td></td>
</tr>
<tr>
<td>Risk-reducing mastectomy reduces breast cancer risk</td>
<td></td>
</tr>
<tr>
<td>Risk-reducing mastectomy can be done at a young age</td>
<td></td>
</tr>
<tr>
<td>Breast screening is available</td>
<td></td>
</tr>
<tr>
<td>There are limitations to breast screening</td>
<td></td>
</tr>
<tr>
<td>Breast awareness is important</td>
<td></td>
</tr>
<tr>
<td>No effective ovarian screening</td>
<td></td>
</tr>
<tr>
<td>Bilateral salpingo-oophorectomy is an option</td>
<td></td>
</tr>
<tr>
<td>Bilateral salpingo-oophorectomy reduces ovarian cancer risk (but primary peritoneal cancer risk remains)</td>
<td></td>
</tr>
<tr>
<td>Bilateral salpingo-oophorectomy before menopause reduces breast cancer risk</td>
<td></td>
</tr>
<tr>
<td>Bilateral salpingo-oophorectomy causes surgical menopause</td>
<td></td>
</tr>
<tr>
<td>Hormone Replacement Therapy is recommended after surgical menopause</td>
<td></td>
</tr>
<tr>
<td>Prostate screening is available for men</td>
<td></td>
</tr>
<tr>
<td>Chemoprevention is being investigated</td>
<td></td>
</tr>
<tr>
<td>Reproductive options are available</td>
<td></td>
</tr>
<tr>
<td>Taking the Oral Contraceptive Pill and having children protects from ovarian cancer</td>
<td></td>
</tr>
<tr>
<td>Multidisciplinary discussion is available</td>
<td></td>
</tr>
</tbody>
</table>
3.4.2.3. Identifying information recalled by participants

Participants’ interview transcripts were systematically searched for reference to the category statement in families where the information had been communicated to the patient. Each statement made by the participant referring to the category statement was documented verbatim onto a matrix for each family. Where no reference was made to information communicated by the health professional, this was documented as ‘not mentioned’. Figure 3.4. shows an example of the matrix from Family 3 with verbatim references from the clinical consultation, the health professional’s letter to the patient following the consultation and references made by the patient and two of her relatives to the category statement ‘Each child and sibling of a person with the pathogenic variant has 50% risk of inheriting the pathogenic variant’. The category statement is grouped under ‘genetic testing information’ in the domain of ‘inheritance’.

Figure 3.4. Example from Family 3 showing all participants’ references to the category statement ‘Each child and sibling of a person with the pathogenic variant has 50% risk of inheriting the pathogenic variant’

<table>
<thead>
<tr>
<th>Consultation</th>
<th>Letter</th>
<th>Patient</th>
<th>Relative 1</th>
<th>Relative 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Every single one of your egg cells will have one copy of <em>BRCA2</em> and it could be 50% of them have the alteration, 50% of them of one that’s not altered</td>
<td>Your daughter has 50/50 chance of inheriting the <em>BRCA2</em> gene alteration from you…. Each of your brothers and sisters has a 50/50 chance of having inherited the gene change</td>
<td>I could have had the gene, the mutated gene passed down to me if I had it, um, by either my father or my mother, um, and I would have a 50%, if I had it, I had a 50% chance of passing it on to my children</td>
<td><em>(Patient)</em> told me that my chances of inheriting this genetic fault was 50/50</td>
<td></td>
</tr>
<tr>
<td>So it’s a 50/50 chance for you <em>(daughter)</em> whether you’ve inherited it or not</td>
<td>Because although there’s a 50% chance of her having it, there’s also a 50% chance that she doesn’t</td>
<td></td>
<td>What I would like to find out of course is whether or not it can be, I suppose it can be passed onto boys who would then pass it onto daughters if they had it so I suppose it’s important that <em>(Relative 1)</em> has… her boys are checked.</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td><em>(Patient)</em></td>
<td></td>
<td>I think the probability is quite high</td>
</tr>
</tbody>
</table>
3.4.2.4. Coding the accuracy of participants' statements compared to the information provided in the consultation and/or summary letter

The Family Communication Study research team (see 1.8.5.5) agreed on the coding framework and definitions of accuracy. The accuracy coding framework is shown in Figure 3.5. Using this coding framework, statements made by the patients and relatives were coded for accuracy compared to the information communicated in the consultation and/or summary letter. Where a participant made more than one reference to a category statement, references were grouped together and coded once. For example, if the participant had made two references about a category statement, one accurate and one inaccurate, the code for that category statement was ‘inaccurate’ (see Figure 3.6.).

Figure 3.5. Accuracy coding framework

<table>
<thead>
<tr>
<th>Accuracy</th>
<th>Definition of accuracy</th>
</tr>
</thead>
<tbody>
<tr>
<td>Accurate</td>
<td>Correct: The information stated by the participant is correct compared to the information provided in the consultation/summary letter</td>
</tr>
<tr>
<td>Inaccurate</td>
<td>Incorrect: The information stated by the participant is incorrect compared to the information provided in the consultation/summary letter</td>
</tr>
<tr>
<td>Unknown</td>
<td>The information stated by the participant is unknown/not mentioned/incomplete compared to the information provided in the consultation/summary letter</td>
</tr>
</tbody>
</table>
**Figure 3.6. Example from Family 3: Patients and relatives’ references to the category statement, ‘Each child and sibling of a person with the pathogenic variant has 50% risk of inheriting the pathogenic variant’ showing coding for accuracy and inaccuracy**

<table>
<thead>
<tr>
<th>Patient</th>
<th>code</th>
<th>Relative 1</th>
<th>code</th>
<th>Relative 2</th>
<th>code</th>
</tr>
</thead>
<tbody>
<tr>
<td>I could have had the gene, the mutated gene passed down to me if I had it, um, by either my father or my mother, um, and I would have a 50%, if I had it, I had a 50% chance of passing it on to my children. Because although there’s a 50% chance of her having it, there’s also a 50% chance that she doesn’t</td>
<td>Accurate</td>
<td>(Patient) told me that my chances of inheriting this genetic fault was 50/50</td>
<td>Accurate</td>
<td></td>
<td>Inaccurate</td>
</tr>
</tbody>
</table>

### 3.4.2.5. Sources of information provided to relatives

A coding framework for identifying the relatives’ sources of information was agreed by The Family Communication Study research team (see 1.8.5.5). The sources of information coding framework are shown in Figure 3.7. Using this coding framework, the sources of information referred to by the relatives were identified from the relatives’ transcripts.

**Figure 3.7. Sources of information coding framework**

<table>
<thead>
<tr>
<th>Code</th>
<th>Sources of the information provided to relative</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Patient only</td>
</tr>
<tr>
<td>2</td>
<td>Patient AND genetics consultation OR letter from genetics</td>
</tr>
</tbody>
</table>
3.4.5. Analysis

3.4.5.1. Accurate and inaccurate recall of information

Accurate and inaccurate statements were counted using Content Analysis. This method involves systematically establishing a set of precisely defined categories and then counting the number of instances that fall into each group (Silverman, 2006).

Because there were different numbers of relatives in each family (either two or three) the mean number of inaccuracies for the relatives in each family were calculated. Accuracy of recall of information for patients was operationalised as the number of accurate statements made during the interview divided by the total number of accurate and inaccurate statements so that if there were 5 accurate statements and 5 inaccurate statements, the accuracy score was 0.5 (5/10). Accuracy of recall of information for relatives involved calculating the accuracy score for each relative interviewed, and then calculating the mean score for the relatives as a whole. Thus, if there were two relatives in the family and one had an accuracy score of 0.5 and the other had a score of 0.3, the score for the relatives would be 0.4. Accuracy of recall scores were calculated separately for genetic testing information and hereditary cancer management information and for the two combined.

A priori hypotheses concerning differences in accuracy between patients and relatives and between genetic testing information and hereditary cancer management information were tested using the Wilcoxon signed-rank test. This evaluated differences between matched pairs of numbers with no assumption about the underlying distribution of those numbers. The alpha was set to 0.05, 2-tailed. Although the hypotheses were directional, it is rare to see use of 1-tailed tests in this area so convention was followed.

3.4.5.2. Sources of information:

The a priori hypothesis concerning whether accuracy of recall of information by relatives was greater with a greater number of sources of information was tested using a Spearman’s rank order correlation coefficient. The alpha was set to 0.05, 2-tailed.
3.5. RESULTS

3.5.1. Information provided by health professionals
The mean number of ‘chunks’ of information provided by health professionals was 21 (range 16 to 26). Six domains of information were identified. Each of these domains was discussed during the results consultations.

3.5.2. Accuracy
Independent coding of accuracy by two members of the research team (CJ and CD) was 94% (627/667) prior to discussion. All disagreements were readily resolved (see Table 3.2).

Table 3.2. Mean number of accurate (Acc) and inaccurate (Inacc) statements for each family according to type of family member and category of information. The relatives’ score shown is the mean score for the relatives in each family.

| Family no. | Patient  |  |  | Relatives  |  |  |
|------------|----------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|
|  | (One patient per family)  | (Mean for relatives per family)  |  |  |  |  |  |
|  | Genetic testing  | Hereditary cancer management  | Genetic testing  | Hereditary cancer management  |  |  |  |  |
|  | Acc  | Inacc  | Acc  | Inacc  | Acc  | Inacc  | Acc  | Inacc  |
| 1 | 4.00  | 3.00  | 9.00  | 9.00  | 2.00  | 5.00  | 2.00  | 16.00 |
| 2 | 2.00  | 2.00  | 5.00  | 8.00  | 2.00  | 2.00  | 4.00  | 9.00 |
| 3 | 7.00  | 1.00  | 10.00 | 8.00  | 4.50  | 3.50  | 6.50  | 11.50 |
| 4 | 4.00  | 3.00  | 6.00  | 7.00  | 7.00  | 0.00  | 6.00  | 7.00 |
| 5 | 3.00  | 3.00  | 5.00  | 8.00  | 4.00  | 2.00  | 1.50  | 11.50 |
| 6 | 2.00  | 2.00  | 4.00  | 12.00 | 2.67  | 1.33  | 2.67  | 13.33 |
| 7 | 5.00  | 1.00  | 10.00 | 7.00  | 3.00  | 3.00  | 6.50  | 10.50 |
| 8 | 4.00  | 1.00  | 11.00 | 4.00  | 3.00  | 2.00  | 3.67  | 11.33 |
| 9 | 2.00  | 3.00  | 4.00  | 7.00  | 2.00  | 3.00  | 0.00  | 11.00 |
| 10 | 6.00 | 3.00  | 7.00  | 7.00  | 4.00  | 5.00  | 3.00  | 11.00 |
| Mean  | 3.90  | 2.20  | 7.10  | 7.70  | 3.42  | 2.68  | 3.58  | 11.22 |
| Median  | 4.00  | 2.50  | 6.50  | 7.50  | 3.00  | 2.50  | 3.33  | 11.17 |
| SD  | 1.73  | 0.92  | 2.69  | 2.00  | 1.55  | 1.56  | 2.21  | 2.38 |
The Wilcoxon signed-rank test showed that the accuracy of recall of information overall (in relation to genetic testing and hereditary cancer management combined) was significantly lower in the relatives (30%) than in the patients themselves (53%), \((z = 2.40, p = 0.017, 2\text{-tailed})\). Overall accuracy of patients and relatives is shown in Table 3.3.

*Table 3.3 Mean number (and percentage) of accurate and inaccurate statements recalled about all information by patients and relatives*

<table>
<thead>
<tr>
<th></th>
<th>Patients n (%)</th>
<th>Relatives n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Accurate</td>
<td>Inaccurate</td>
</tr>
<tr>
<td>110.00 (53%)</td>
<td>99.00 (47%)</td>
<td>70.00 (30%)</td>
</tr>
</tbody>
</table>

The accuracy for patients and relatives combined was greater for genetic testing information (60%) than for hereditary cancer management information (36%) \((z = 2.80, p = 0.005)\). The difference in accuracy between patients and relatives appeared to be greater for hereditary cancer information than for genetic testing information. However, this just missed statistical significance by the Wilcoxon signed-rank test \((z = 1.89, p = 0.056)\) (Table 3.4).

*Table 3.4. Mean number (and percentage) of accurate and inaccurate statements recalled about genetic testing and hereditary cancer management information by patients and relatives*

<table>
<thead>
<tr>
<th>Domains</th>
<th>Patients n (%)</th>
<th>Relatives n (%)</th>
<th>Patients and Relatives combined n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic testing</td>
<td>39.00 (64%)</td>
<td>22.00 (36%)</td>
<td>34.00 (56%)</td>
</tr>
<tr>
<td>Hereditary cancer</td>
<td>71.00 (48%)</td>
<td>77.00 (52%)</td>
<td>36.00 (24%)</td>
</tr>
<tr>
<td>management</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><em>Mean</em></td>
<td>55.00</td>
<td>49.50</td>
<td>35.00</td>
</tr>
<tr>
<td><em>SD</em></td>
<td>22.63</td>
<td>38.39</td>
<td>1.41</td>
</tr>
</tbody>
</table>
3.5.4. Sources of information

The Spearman’s rank order correlation coefficient showed a strong positive correlation between the accuracy of recall by relatives with increasing number of sources of information ($R = 0.88$, $p = 0.001$) (see Table 3.5). This was true both for hereditary cancer management ($R = 0.83$, $p = 0.003$) and genetic testing information ($R = 0.72$, $p = 0.02$).

Table 3.5. Mean accuracy scores for relatives receiving information from different sources

<table>
<thead>
<tr>
<th>Source of information</th>
<th>Information level</th>
<th>Family no.</th>
<th>Mean scores for relatives in all families</th>
<th>Mean</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>Accurate n (%)</td>
<td>Inaccurate n (%)</td>
<td></td>
</tr>
<tr>
<td>Patient + letter + genetics appointment</td>
<td>3</td>
<td>3, 4</td>
<td>24.00 (52%)</td>
<td>22.00 (48%)</td>
<td>23.00</td>
</tr>
<tr>
<td>Patient + letter/genetics appointment</td>
<td>2</td>
<td>2,5,6,7,8,10</td>
<td>40.00 (33%)</td>
<td>82.00 (67%)</td>
<td>61.00</td>
</tr>
<tr>
<td>Patient only</td>
<td>1</td>
<td>1, 9</td>
<td>6.00 (15%)</td>
<td>35.00 (85%)</td>
<td>20.5</td>
</tr>
</tbody>
</table>

3.6. DISCUSSION

Accuracy of recall of information about genetic testing and hereditary cancer management following post-test genetic counselling about a pathogenic variant was significantly lower for relatives than for patients. Accuracy of recall amongst patients and relatives was significantly lower for hereditary cancer management information than for genetic testing information. Accuracy of recall was greater amongst relatives who received information from the genetics health professional in addition to the patient.

The accuracy of recall of information amongst relatives was significantly lower than the accuracy amongst patients, although it should be noted that accuracy amongst the patients was only 53%. The reduction in accuracy of recall as information is
communicated through families supports the findings of previous studies (Sermijn et al., 2004; Vos, Menko, et al., 2011).

It is possible that the important information may have been ‘lost’ in the amount and complexity of the information communicated at each stage in the information process. There is evidence that the amount of information communicated has a negative effect on recall (Cowan, 2005; Howard, 1983), although a systematic review of interventions to improve recall of medical advice in healthcare, found no trials exploring the effect of restricting the amount of information given at any one time (Watson & McKinstry, 2009). Information about genetics and hereditary cancer can be particularly complex. A study of 150 audiotaped simulated genetic counselling sessions found that the use of technical jargon and complex terminology was even more pronounced than during other medical discussions with an average of 11 genetics-related technical words, such as syndrome, mutation and susceptibility, being used an average of 10 times per consultation. In the same study, the videotaped simulated consultations were shown to individuals with a similar family history and recall of information was shown to decrease as the amount of complex terms increased (Roter et al, 2006).

Learning that a pathogenic variant has been identified is likely to create anxiety for all the family. High and low states of anxiety have been shown to negatively impact on recall ability (Kessels, 2003; Ley, 1979), although a study of recall of information in genetic counselling found no association between knowledge and anxiety (Michie, McDonald, & Marteau, 1997).

In this study, further inaccuracies appear to have occurred when the information was passed onto the relatives. The information may not have been fully understood by the patients or relatives when it was communicated or individual interpretations may have led to inaccuracies as suggested by a previous study (Vos, Menko, et al., 2011). The index patients’ perceived lack of relevance of the information, for example for male, young or unwell relatives, may have led to information being edited or withheld in the relatives’ perceived best interests (Dancyger et al., 2011). Even when the information is considered relevant and cancer patients feel adequately informed following genetic counselling, the emotional legacy of cancer may influence the dissemination of information within the family (Landsbergen et al., 2005) and negatively affect accuracy amongst patients and relatives.
This study’s finding that there is a lower level of accuracy amongst patients and relatives about hereditary cancer information than about genetics information is contrary to previously reported findings from studies into knowledge and accuracy following genetic counselling. A previous study found high levels of accuracy about various aspects of hereditary cancer but lower levels of knowledge about inheritance amongst first-degree relatives of patients with cancer or a known BRCA1/BRCA2 pathogenic variant (Sermijn et al., 2004). In a randomised intervention study of patients with cancer or with a family history of cancer, a high level of knowledge was reported about both genetics and hereditary cancer, although cancer risk was least accurately recalled (Roshanai, Rosenquist, Lampic, & Nordin, 2009).

Visual representations have been shown to improve recall of health information (Flocke & Stange, 2004; Houts, Doak, Doak, & Loscalzo, 2006). Visual representations are often used in genetic counselling to establish the family history and family relationships. This may have helped with recall of genetic information, for example in understanding how the pathogenic variant might have been inherited within the family, the pathogenic variant and who the information is important for. Hereditary cancer information is less easily presented in a visual format.

More information was communicated about hereditary cancer management than genetic testing during the consultations. The patients would have been more familiar with the genetic testing information from the pre-test genetic counselling. Detailed information about hereditary cancer management, such as the availability of surveillance or risk-reducing surgery, may not have been discussed at pre-test genetic counselling. Relatives may also have been more familiar with the genetic testing information as the letters given to the patients for their relatives mainly focused on the gene involved, who might be at risk and what they could do about it.

The National Institute for Clinical Excellence (2004) published guidelines highlighting the importance of effective communication with cancer patients and making recommendations for communication skills training for all health professionals involved in the care of cancer patients. Whilst many genetics health professionals are trained in communication skills and have expert skills in communicating about genetics issues, there may be gaps in the skills and knowledge of genetics health professionals about the communication needs of cancer patients, which may have contributed to this finding.
Accuracy of recall amongst relatives improved when information was received from a greater number of sources. Whether this was because information was provided in several different ways, or because information was provided directly by the genetics health professional, is not known. Studies of family communication in genetics have consistently recommended that health professionals provide patients with letters about genetic test results to pass onto their relatives (Hallowell, 1998; Sermijn et al., 2004). Yet the findings from this study suggest that the provision of several sources of information may help to improve accuracy. This is consistent with recommendations in health education that written information is better remembered than verbal information and that presenting information through multiple channels and in different formats improves recall (Kessels, 2003). A study of the communication preferences of patients and relatives about a pathogenic variant identified a need for educational and supportive materials and a preference for brief written information or web-based resources (Ratnayake et al., 2011), suggesting that more sources of information would be helpful to family communication.

Direct contact with relatives by genetics health professionals has been shown to significantly increase the uptake of genetic tests (Suthers, Armstrong, McCormack & Trott, 2006), although whether this improved accuracy of recall amongst relatives was not studied. The ethics and feasibility of genetics health professionals routinely communicating directly with relatives with whom they have not had any previous contact has been the subject of much debate (Parker & Lucassen 2003; Lucassen, 2007; Clarke, 2007; Suthers et al., 2006). Patients and relatives prefer information about a pathogenic variant to be communicated by the patient with support from genetics (Forrest et al., 2003). However, it can be emotionally difficult for cancer patients to communicate this information to relatives (Hallowell et al., 2002, Hallowell et al., 2004). The emotional barriers preventing family communication have been suggested as an explanation for the low uptake of genetic testing (Landsbergen et al., 2005). The information provided directly to relatives by genetics health professionals is likely to involve less interpretation and emotion than that provided by patients (Vos, Menko, et al., 2011), possibly improving accuracy of recall. The provision of targeted information for relatives by health professionals may improve the accuracy of information.

Early studies of communication assumed a simple model, whereby information was transferred from the sender to the receiver (Wilson et al., 2004). It was assumed that information was passively received, processed and reproduced in the same form and
quality as it was communicated (Kessler, 1989). A critical review of communication in healthcare challenged the assumption that changing knowledge, attitudes and beliefs will translate into changes in health behavior (Gravois Lee & Garvin, 2003) and argued for a move in health communication from information transfer, based on a one-sided relationship between communicator and receiver privileging the expert over the lay perspective, to information exchange, based on a two-way dialogue between the health professional and patient. Since then much work has been done to improve communication with patients, acknowledging the central role of the patient as the expert in his or her own health.

Northouse and Northouse’s model of health communication (1998) emphasises the impact of factors such as relationships, transactions and contexts on interactions in a healthcare setting and stresses the ongoing transactional and interactive nature of health communication with the different participants influencing each other as the interaction progresses. The Family Systems Theory (White, 2002) provides a framework for better understanding of the complexity of the interactions between family members. Within this theory, the family is regarded as a complex and interactive social system influenced by family structure, change and development. The illness influences and is influenced by the individuals within the family who interpret the illness and manage interactions about the illness, or in this case the genetic test result. This framework may help to explain the inaccuracies of information seen in both the patients and the relatives in this study and may also help to explain why relatives’ accuracy was improved when information was received directly from the genetics health professional in addition to the patient.

3.7. CLINICAL IMPLICATIONS

Whilst responsibility for informing family members about genetic test results remains with cancer patients, facilitating family communication will continue to be a key role of genetics health professionals. The low levels of accuracy amongst relatives in this study highlights the need for genetics health professionals to be more proactive in helping patients to identify which relatives to inform, what information to pass on and how to approach this. The use of genograms to identify social exchanges between patients and at-risk relatives has been found to be helpful for exploring and overcoming barriers to communication about hereditary breast and ovarian cancer within families (Peters, et al, 2011).
The low level of accuracy about hereditary cancer amongst patients and relatives suggest that genetics health professionals may benefit from a better understanding of the issues that are important to cancer patients and their families and that patients and families may benefit from direct contact with health professionals specialising in both genetics and cancer. Closer working between genetics and cancer health professionals, for example in multidisciplinary clinics (Bancroft et al., 2010; Pichert et al., 2010), may result in greater shared knowledge amongst health professionals and improved understanding amongst patients and families.

If barriers such as lack of administrative support prevent health professionals from providing information for relatives (Forrest et al., 2007), there may be other ways to communicate this information to relevant family members, such as access to a website or provision of an audio-tape or DVD of the consultation. The findings from this study suggest that multiple sources of information and direct contact with a genetics health professional may improve the accuracy of information recalled by relatives. The current funding and organisation of genetics services in the UK makes sending a personalised appointment to all relatives impossible without a GP referral. However, alternative approaches, such as a letter from the genetics health professional targeted at specific relatives, inviting relatives to an information session or providing access to individualised online or telephone information may be helpful. It is also important for genetics health professionals to identify families or relatives where there may be barriers to family communication and to work with patients to ensure that information is communicated accurately and directly to those relatives.

3.8. LIMITATIONS

This was a small self-selected sample and the design of the study means that participants were not assessed on their recall of specific information. Quantifying qualitative data raises its own challenges in terms of simplifying the data and reducing it to organised categories. With qualitative data this reduction can result in the loss of richness and meaning (Bowling, 2009; Silverman, 2006). The data analysed in this study, however, involved direct comparison between the information recalled by the participants and the information provided by the health professionals, enabling the data to be quantified without losing the meaning. Approaching this question using a different methodology, such as a survey asking specific questions about information recall, may
have resulted in more accurate recall. However, it was not possible to identify all the sources of information that patients in this study had access to.

This study did not consider the clinical significance or accuracy of the information that was communicated by health professionals. The study also did not identify whether information that might impact on the patient or at-risk relative were recalled regardless of the inaccuracy. In addition, the study did not evaluate other factors that could influence recall, such as educational level, meaning, context, experience, emotion or world view of the participants and the impact those factors can have on understanding, interpretation and communication.

3.9. CONCLUSIONS
In conclusion, the findings suggest that patients with a pathogenic variant in the BRCA1/BRCA2 genes do not always pass on accurate information to their at-risk relatives and that accuracy of recall amongst relatives may be improved if they have more direct contact with genetics health professionals. These findings highlight the importance of accurate communication of information about genetic testing and hereditary cancer management by health professionals to patients and relatives and suggest a need for further research to understand the key information required by patients and the extent to which key information is communicated by health professionals and recalled by patients.
CHAPTER 4. STUDY 2: CONSENSUS AMONGST HEALTH PROFESSIONALS AND SERVICE USERS CONCERNING KEY MESSAGES FOR COMMUNICATING INFORMATION ABOUT GENETIC TESTING AND HEREDITARY CANCER MANAGEMENT TO BREAST/OVARIAN CANCER PATIENTS - A DELPHI CONSENSUS EXERCISE


4.1. INTRODUCTION

Specialist genetics services do not have the capacity to meet the demand for genetic counselling and testing of cancer patients (Slade, Riddell, Turnbull, Hanson, & Rahman, 2015). Genetics health professionals are increasingly required to communicate with patients at vulnerable times in their cancer trajectory such as shortly after diagnosis, at metastatic relapse or during palliative care. Oncology health professionals will increasingly be required to counsel cancer patients about genetic testing and the management of hereditary cancer. To inform current and future practice it is important to identify the information needs of breast and ovarian cancer patients, the views of health professionals specialising in genetics and cancer about the information to communicate and the optimal timing of communicating key information.

4.2. BACKGROUND

4.2.1. Information required by breast and ovarian cancer patients

An international survey of policy documents and guidelines found that during genetic counselling, information is communicated about many topics including the condition and the patient’s risk of having the condition, available treatment options, the purpose, nature and content of the genetic test being considered, risks and limitations of testing, alternatives options available to the patient, the potential harms of genetic testing, risks to relatives and information about available support groups (Rantanen et al., 2008).

The provision of medical information has a positive impact on patient satisfaction (Roter, 2003). Several studies have investigated the information communicated during genetic counselling about BRCA1/BRCA2 (Butow & Lobb, 2004; Ellington et al., 2005;
Roter et al., 2006). The scoping review presented in Chapter 2 found that few studies have investigated the information needs of cancer patients about hereditary cancer. The studies that have been undertaken identified a need for information about risk-reducing surgery, chemoprevention, screening, (Metcalfe et al., 2000) and cancer risk for the patient and family (Butow & Lobb, 2004). Among patients tested shortly after diagnosis, two qualitative studies identified a need for brief information about increased risk of cancer without statistics and hope-giving information about the options available to address the risks (Gleeson et al., 2013; Meiser et al., 2012).

4.2.2. Communication of information about genetic testing and hereditary cancer management by genetics and cancer health professionals

Whereas traditionally genetic counselling has been delivered by genetics health professionals, the shift in genetic testing will mean that cancer health professionals will need to be more involved in communication about aspects of hereditary cancer with cancer patients. It is not clear who will be best placed to deliver such information in the future. In a study considering the hypothetical possibility of genetic testing shortly after breast cancer diagnosis, there was general agreement amongst health professionals and patients that the information should be discussed by surgeons and oncologists (Ardern-Jones, Kenen, & Eeles, 2005). More recently however, women with newly diagnosed ovarian cancer have expressed a preference for the information to be communicated by their medical oncologist, citing relevance of the information for treatment planning, established trusting relationship and convenience as reasons for the preference (Gleeson et al., 2013). In a recent survey of the views of breast oncology health professionals there was a preference for the information to be communicated by the breast surgeon (Burcher et al., 2013).

There are differences in the approach, focus and training of cancer and genetics health professionals which may impact on the information communicated. Genetic counselling involves a non-directive and person-centred approach (Hough, 2002) with the goal of helping people to ‘understand and adapt to the medical, psychological and familial implications of genetic contributions to disease’ (Resta et al., 2006 p.274). Genetic counselling includes helping the patient to think about the implications for the family and rehearsing how they wish to communicate this information to relatives. The ‘work’ of genetic counselling involves information exchange and the provision of support (Smets, van Zwieten, & Michie, 2007). For cancer health professionals however, the main ‘work’ involves initiation of treatment. A further distinction between the two specialties is that the focus of clinical genetics is on the family, whereas oncology and cancer care focuses on the individual (Middleton, Hall, & Patch, 2015). Although
training in genetics has been recommended for some time for oncologists (Robson et al., 2015), cancer health professionals are not always confident to make a risk assessment (Metcalfe, Pumphrey, & Clifford, 2010), may be concerned about causing distress to families by referral (Van Riel, Warlam-Rodenhuis, Verhoef, Rutgers, & Ausems, 2010), are not always clear whose responsibility it is to make the referral (Lanceley et al., 2012) and do not consistently refer patients even if they have been identified (Daniels, Urbauer, Stanley, Johnson, & Lu, 2009; Grover, Stoffel, Bussone, Tschoegl, & Syngal, 2004; Meyer et al., 2010).

4.2.3. Lay and professional understanding of disease
Differences in lay and professional understanding of disease have long been recognised (Popay & Williams, 1996; Popay, Williams, Thomas, & Gatrell, 1998). Lay people construct their understanding to make sense of the experience and orientate their behaviour towards it accordingly (Popay et al., 1998). Lay understanding of inheritance is demonstrated in cancer genetics by a study examining the way in which patients make sense of a risk estimate and integrate the information into their worldview. The authors found that the patients focused more on the benefits associated with being at risk, such as seeking ways of organising health care resources (surveillance and risk management options) around their needs, than the potential negative outcomes of the genetic risk or the genetic disease itself (Scott, Prior, Wood, & Gray, 2005).

4.2.4. Timing of communicating hereditary cancer information to patients
Clinical guidelines recommend pre-test genetic counselling so that patients understand the implications of testing for themselves and their families and are able to give informed consent (National Institute for Health and Care Excellence, 2013; Robson et al., 2015). As genetic testing becomes more closely integrated into mainstream services, there is likely to be increasing discussion about whether full disclosure of the implications of a pathogenic variant should take place prior to genetic testing or whether information should be withheld until after the result is known to prevent unnecessary anxiety. Non-genetics health professionals and women with cancer may have different views about whether pre-test counselling is necessary. In a survey of 81 non-genetics health professionals ordering genetic tests in the USA, approximately half reported scheduling a pre-test counselling session (Vadaparampil, Scherr, Cragun, Malo, & Pal, 2015). A further US study found that just 36.8% (1334) of 3628 women with and without cancer whose physicians ordered commercially available genetic
testing reported receiving genetic counselling from a genetics health professional prior to testing (Armstrong et al., 2015).

Patients who are found to have a pathogenic variant in a cancer predisposing gene, will usually be offered post-test genetic counselling. This counselling provides a second opportunity to discuss the issues raised by the genetic test result and moves the discussion from the hypothetical to the actual implications, such as risk management options, access to psychological support and family communication. The proportion of at-risk relatives who make contact with a genetics service has been shown to significantly increase when post-test genetic counselling support is provided to the index patient (Forrest, Burke, Bacic, & Amor, 2008).

There have been few studies investigating the views of patients about the optimal timing of information about hereditary cancer. A qualitative study of the views of women with newly diagnosed ovarian cancer about genetic testing shortly after diagnosis reported a preference for a ‘one-step-at-a-time’ approach whereby initial information about genetic testing in the context of the cancer is presented prior to testing with the specifics of other non-treatment related information given once a pathogenic variant has been detected (Gleeson et al., 2013). In-depth interviews with 26 women with newly diagnosed breast cancer aged 50 and younger found that 16 women preferred a brief discussion of all clinically relevant information and seven women preferred detailed information about surgical options prior to testing but three women preferred that details about surgical options only if a pathogenic variant was identified (Meiser et al., 2012). One study comparing the timing of a decision aid (video and brochure) presented to women with and without cancer following genetic testing but before and after learning about a BRCA pathogenic variant, found no statistical differences in well-being, decision and information related outcomes or treatment choices (van Roosmalen et al., 2004).
4.3. AIMS AND RESEARCH QUESTIONS

This study aimed to:
1. Identify areas of agreement and disagreement amongst and between health professionals and service users about messages that are key and not key;
2. Investigate how easy or difficult it is to reach agreement amongst and between health professionals and service users about messages that are key and not key;
3. Identify areas of agreement amongst and between health professionals and service users about the timing of communication of key messages.

4.3.1. Key messages
I. According to health professionals, which messages are a) agreed as key, b) not agreed as key or not key or c) agreed as not key?
II. According to service users, which messages are a) agreed as key, b) not agreed as key or not key or c) agreed as not key?
III. Which messages do health professionals and service users a) agree are key, b) agree are not key?

4.3.2. Reaching agreement
I. How many rounds of survey were required for health professionals to reach agreement about a) key messages and b) messages that were not key?
II. How many rounds of survey were required for service users to reach agreement about a) key messages and b) messages that were not key?
III. In which round of the survey did health professionals and service users agree on a) key messages and b) messages that were not key?

4.3.3. Timing of communication
I. According to health professionals when should key messages be communicated a) before testing and once a pathogenic variant has been identified or b) once a pathogenic variant has been identified?
II. According to service users when should key messages be communicated a) before testing and once a pathogenic variant has been identified or b) once a pathogenic variant has been identified?
III. About which key messages do health professionals and service users agree on the timing of communication?
4.4. METHODS

4.4.1. Design
The study design was a Delphi consensus exercise (Dalkey & Helmer, 1963; Pill, 1971) with health professionals and service users who had expert professional or personal experience in this field.

4.4.2. Delphi Consensus method
The Delphi consensus method involves several rounds of a survey with a group of experts who anonymously respond and then receive feedback on the group response before being sent a subsequent survey to complete. The goal is to reduce the range of responses with a view to achieving consensus. The method is particularly useful for obtaining the knowledge and opinions of a diverse group of experts in areas that are not supported by a strong evidence-base (Jones & Hunter, 1995; Pill, 1971) and has been used extensively in health and social science research, in particular for developing consensus guidelines (Berk, Jorm, Kelly, Dodd, & Berk, 2011; Jefferson et al., 2016; Paneque, Sequeiros, & Skirton, 2015; Skirton, H., Barnoy, S., et al. 2013).

The main premise of the method is that the opinion of the group has greater validity than the opinion of individuals (Snape et al., 2014). The systematic and structured process of anonymous survey and controlled feedback enables inclusion of a wide range of participants, allowing divergence from publicly expressed views and limiting the dominating influences of individuals or groups, as can occur in face-to-face consensus exercises. There are no universally agreed requirements for using the Delphi method (Hasson, Keeney, & McKenna, 2000) but one of two approaches are generally employed: the basic Delphi procedure (involving multistage iteration and controlled feedback via anonymous surveys) and the modified Delphi procedure (whereby the survey method is combined with a physical meeting of experts). Four key concepts and assumptions have been described that distinguish the basic Delphi procedure from other research methods: anonymity, the use of experts, multi-stage iteration and controlled feedback and exploration of consensus using a statistical group response (Snape et al., 2014).
4.4.3. Phase 1 pilot study: developing the questionnaire

4.4.3.1. Developing the online questionnaire
An online questionnaire was developed for feasibility and comprehensiveness. The Phase 1 pilot study aimed to develop the questionnaire. The thesis author and seven experienced UK genetics health professionals (four cancer geneticists and three cancer genetic counsellors) were involved. The health professionals were approached through personal contact and asked to help with developing the questionnaire. This phase involved developing a definition for what constitutes a key message, refining the criteria for a key message and deciding on the information messages to be used in the survey.

4.4.3.2. Defining a key message for breast and ovarian cancer patients about hereditary cancer and refining the criteria for a key message
The study definition of a key message was derived from the cognitive and behavioural aspects of the widely accepted definitions and published goals of genetic counselling (Resta et al., 2006) (see section 1.2).

To achieve clarity and consistency in the selection of key messages in the study, protocol specific key message criteria were set. Following feedback in the Phase 1 pilot study, the definition of a key message was simplified into one sentence to assist with reviewing the messages and the criteria were bulleted and examples provided. The final agreed definition and key message criteria for this study was as follows:

A key message is ‘information required by the individual with cancer in order to understand the risks, implications and options for themselves and their relatives and to decide on a course of action that is appropriate for them’ (Jacobs et al, 2017, p2). If an individual did not accurately receive this information they would be less likely to: Inform at-risk relatives; seek/have genetic counselling/genetic testing; take measures to detect cancer early, such as breast awareness or surveillance; take measures (appropriate to the prognosis) to reduce the risk of cancer, such as surgery; or seek other medical advice or information related to the risks, such as contraception, reproductive options or Hormone Replacement Therapy.

4.4.3.3. Refining the information messages
The Phase 1 pilot study involved review of the 58 information messages identified in Study 1. The process of identifying the information messages in Study 1 is described in Chapter 3 (Jacobs, Dancyger, Smith, & Michie, 2015). The purpose of the Phase 1
pilot study was to eliminate any inaccurate or nonsense information and refine the wording of the messages. Five information messages were removed at this stage.

4.4.4. Phase 2 pilot study: testing the questionnaire
The Phase 2 pilot study tested the online questionnaire for comprehension, readability and usability. This was undertaken with members of the Study Interest Group (see section 1.8.5.4.). Minor changes were made to the online questionnaire as a result of the Phase 2 pilot study. The preliminary questionnaires to assess suitability to participate in the study are shown in Appendix 2.2-2.3. The online questionnaire sent to all participants once accepted as eligible is shown in Appendix 2.4.

4.4.5. Recruitment, sample and participation
Purposive sampling methods were used to identify 16 health professionals with expert knowledge and 16 service users with personal experience of genetic testing for hereditary breast and ovarian cancer within the National Health Service (NHS). All participants were required to have an interest in the topic, capacity, willingness and time to contribute to the study, be accessible by email and able to communicate in English.

Complete anonymity of health professional participants from the researcher was not possible as selecting for appropriate expertise required a direct approach from the researcher. The service users were not known to the researcher. Participants were anonymous to each other.

4.4.5.1. Health professionals
4.4.5.1.1. Recruitment
Potential health professional participants included cancer geneticists, cancer genetic counsellors, clinical oncologists, breast surgeons, gynaecological oncologists and breast care nurses. Potential participants were identified by personal contact, recommendation of senior colleagues or participation at a senior level in professional organisations, such as the British Society for Genetic Medicine (BSGM) or the British Association of Surgical Oncologists (BASO). Approach was made via personal email, in which potential participants were informed about the study and invited to take part.

Health professionals who agreed to take part were sent the preliminary questionnaire to confirm eligibility and to gather demographic data.
4.4.5.1.2. Sample

Four clinical geneticists, four genetic counsellors, two cancer clinical nurse specialists, two gynaecological oncologists, two breast surgeons and one clinical oncologist agreed to participate at the first request.

Six clinical oncologists were approached before a further clinical oncologist was recruited: one declined due to a busy workload, two agreed but did not complete the online questionnaire and two did not respond to the request.

All eight genetics health professionals reported counselling or managing cancer about a *BRCA1/BRCA2* pathogenic variant at least once a week, with six participants reporting this activity two to three times a week.

Seven of the cancer health professionals reported managing, caring for or counselling women with cancer about *BRCA1/BRCA2* at least once a month, with three participants reporting this activity at least once a week. One cancer health professional reported discussing genetics referral and *BRCA1/BRCA2* with patients two to three times a week. The health professionals’ experience and expertise is shown in Table 4.1.

4.4.5.1.3. Participation

All 16-health professionals who proceeded with the study completed the round 1 questionnaire. One geneticist and one clinical oncologist did not continue beyond round 1. The other 14 participants completed round 2 and round 3.
Table 4.1 Health professionals' experience of communicating about BRCA1/BRCA2 with breast/ovarian cancer patients

<table>
<thead>
<tr>
<th>Participant no.</th>
<th>Area of expertise</th>
<th>Type of experience</th>
<th>Frequency of experience</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Senior cancer genetic counsellor</td>
<td>Counsel patients</td>
<td>2-3 times/week</td>
</tr>
<tr>
<td>2</td>
<td>Cancer genetic counsellor</td>
<td>Counsel patients</td>
<td>2-3 times/week</td>
</tr>
<tr>
<td>3</td>
<td>Cancer genetic counsellor</td>
<td>Counsel patients</td>
<td>2-3 times/week</td>
</tr>
<tr>
<td>4</td>
<td>Senior cancer genetic counsellor</td>
<td>Counsel patients</td>
<td>2-3 times/week</td>
</tr>
<tr>
<td>5</td>
<td>Consultant cancer genetics</td>
<td>Counsel patients</td>
<td>2-3 times/week</td>
</tr>
<tr>
<td>6</td>
<td>Consultant cancer genetics</td>
<td>Counsel patients</td>
<td>2-3 times/week</td>
</tr>
<tr>
<td>7</td>
<td>Consultant cancer genetics</td>
<td>Manage patients</td>
<td>At least once/week</td>
</tr>
<tr>
<td>8</td>
<td>Consultant cancer genetics</td>
<td>Counsel patients</td>
<td>Once/week</td>
</tr>
<tr>
<td>9</td>
<td>Consultant breast surgery</td>
<td>Manage/care for/ counsel patients</td>
<td>2-3 times/week</td>
</tr>
<tr>
<td>10</td>
<td>Consultant breast surgery</td>
<td>Manage/care for/ counsel patients</td>
<td>2-3 times/month</td>
</tr>
<tr>
<td>11</td>
<td>Senior nurse breast care</td>
<td>Manage/care for/ counsel patients</td>
<td>Once/week</td>
</tr>
<tr>
<td>12</td>
<td>Senior nurse breast care</td>
<td>Refer patients</td>
<td>2-3 times/week</td>
</tr>
<tr>
<td>13</td>
<td>Sub-specialty doctor oncology</td>
<td>Manage/care for/ counsel patients</td>
<td>Once/month</td>
</tr>
<tr>
<td>14</td>
<td>Consultant oncology</td>
<td>Manage/care for/ counsel patients</td>
<td>2-3 times/month</td>
</tr>
<tr>
<td>15</td>
<td>Consultant gynae-oncology</td>
<td>Manage/care for/ counsel patients</td>
<td>2-3 times/month</td>
</tr>
<tr>
<td>16</td>
<td>Sub-specialty doctor gynae-oncology</td>
<td>Manage/care for/ counsel patients</td>
<td>2-3 times/week</td>
</tr>
</tbody>
</table>
4.4.5.2. Service users

4.4.5.2.1. Recruitment

Service users included women with a BRCA1/BRCA2 pathogenic variant who were tested in the UK following genetic counselling, who had been the first in their family to be diagnosed with a pathogenic variant and who received their result after publication of the NICE guidelines for familial breast cancer (McIntosh A, 2004; updated 2006). All participants were aged 18 or over and English speaking. Potential participants were identified via voluntary organisations that provide peer support and information for people with a BRCA1/BRCA2-related cancer: Breast Cancer Care, Cancer Help UK, Macmillan Cancer Support, Ovacome, Ovarian Cancer Action and FORCE UK. An information sheet was provided for the voluntary organisations to distribute to their membership. The Breast Cancer Care Helpline details were provided and additional support was available via the recruiting voluntary organisations. Potential participants were advised to contact their General Practitioner and/or local Regional Genetics Centre should personal questions arise from participation in the study.

Potential participants who expressed an interest in the study were telephoned to explain the study, determine eligibility and answer any questions. Eligible participants were sent a patient information sheet, consent form (see Appendix 1.5-1.6) and preliminary questionnaire to confirm eligibility and gather demographic data.

4.4.5.2.2. Sample

Twenty-seven potential service user participants responded to the recruitment call, of which 25 agreed to a telephone discussion. Following the phone call one service user declined to continue with the study, six were not eligible and two did not complete the round 1 online questionnaire.

Of the 16 service users who proceeded with the study, five were BRCA1 carriers and 11 were BRCA2 carriers. The mean age of the service users was 53 (range 43 to 69 years). Nine service users had breast cancer, three had ovarian cancer and four had breast and ovarian cancer. Two participants underwent genetic testing before definitive cancer surgery and the remaining 14 underwent testing after surgery. Ten participants were educated to degree level or above, three completed education at age 18 and three completed their education at age 16. Service users’ experience and educational level are shown in Table 4.2.
4.4.5.2.3. Participation

All 16 service users who proceeded with the study completed the round 1 questionnaire. Fourteen participants completed the round 2 questionnaires. Twelve participants completed the round 3 questionnaires.

Table 4.2. Service users’ BRCA1/BRCA2 experience and educational level

<table>
<thead>
<tr>
<th>Participant no.</th>
<th>Type of cancer</th>
<th>Gene involved</th>
<th>Highest educational level</th>
</tr>
</thead>
<tbody>
<tr>
<td>17</td>
<td>Breast cancer</td>
<td>BRCA1</td>
<td>High school</td>
</tr>
<tr>
<td>18</td>
<td>Ovarian cancer</td>
<td>BRCA1</td>
<td>Graduate</td>
</tr>
<tr>
<td>19</td>
<td>Breast and ovarian cancer</td>
<td>BRCA2</td>
<td>Post-graduate</td>
</tr>
<tr>
<td>20</td>
<td>Breast and ovarian cancer</td>
<td>BRCA2</td>
<td>High school</td>
</tr>
<tr>
<td>21</td>
<td>Breast cancer</td>
<td>BRCA2</td>
<td>Graduate</td>
</tr>
<tr>
<td>22</td>
<td>Breast and ovarian cancer</td>
<td>BRCA2</td>
<td>Post-graduate</td>
</tr>
<tr>
<td>23</td>
<td>Breast and ovarian cancer</td>
<td>BRCA1</td>
<td>High school</td>
</tr>
<tr>
<td>24</td>
<td>Ovarian cancer</td>
<td>BRCA2</td>
<td>High school</td>
</tr>
<tr>
<td>25</td>
<td>Breast cancer</td>
<td>BRCA2</td>
<td>Graduate</td>
</tr>
<tr>
<td>26</td>
<td>Breast cancer</td>
<td>BRCA2</td>
<td>Graduate</td>
</tr>
<tr>
<td>27</td>
<td>Ovarian cancer</td>
<td>BRCA1</td>
<td>Post-graduate</td>
</tr>
<tr>
<td>28</td>
<td>Breast cancer</td>
<td>BRCA2</td>
<td>Post-graduate</td>
</tr>
<tr>
<td>29</td>
<td>Breast cancer</td>
<td>BRCA2</td>
<td>Graduate</td>
</tr>
<tr>
<td>30</td>
<td>Breast cancer</td>
<td>BRCA2</td>
<td>High school</td>
</tr>
<tr>
<td>31</td>
<td>Breast cancer</td>
<td>BRCA2</td>
<td>Post-graduate</td>
</tr>
<tr>
<td>32</td>
<td>Breast cancer</td>
<td>BRCA1</td>
<td>High school</td>
</tr>
</tbody>
</table>

4.4.6 Procedure

UCL ethics approval was obtained for this study (4513/001). Consent was obtained from service users. To save time for the health professionals, return of the preliminary questionnaire was accepted as consent to take part in the study.

On receipt of the preliminary questionnaire, eligible participants were sent the online questionnaire using the Qualtrix software (UCL license). Hard copies of the questionnaire were available. Non-responders were contacted on up to three occasions. The initial online questionnaire included information about the purpose of the study, instructions on completion of the questionnaire, the study definition of a key message and the information messages. The questionnaire was circulated to three groups: service users, genetics health professionals and cancer health professionals.
In round 1, the same questionnaire was circulated to all participants. In rounds 2 and 3 the questionnaire was amended for each group according to feedback from the previous rounds. Messages where no agreement was reached were recirculated, together with the median score, the range of responses and summarised anonymised comments from the group. Participants’ responses from earlier rounds were not sent back to them in subsequent rounds as the aim of the study was to gather views in the light of comments from the whole group and not to measure consistency of responses.

4.4.6.1. Key messages
Participants were asked to decide if each message was key or not key using a 5-point Likert scale. Messages with a definite response were scored higher than those with a less definite response to capture the extent of certainty about the message. Options were as follows (score shown in brackets):
- A key message (2)
- Probably a key message (1)
- Not sure or don’t have a view (0)
- Probably not a key message (-1)
- Not a key message (-2)

4.4.6.2. Reaching agreement
At each round, messages agreed as key or not key with ≥75% agreement from each group were removed from circulation. Remaining messages were re-circulated up to three times or until agreement was reached. An agreement level of ≥75% was selected in line with findings from a systematic review of Delphi surveys (Boulkedid, Abdoul, Loustau, Sibony, & Alberti, 2011).

The first time a message was circulated, comments were invited on the wording of the message, the reasons for the selection and potential additional key messages. For feasibility, changes to wording were only accepted when suggested by two or more participants or for consistency with other messages. The same method was employed for suggested additional messages. The Study Interest Group reviewed the anonymised participants’ comments, the rationale for inclusion and the re-worded and suggested additional messages. Minor changes to the message wording were made following this process. The final agreed messages and message numbers are shown in full in Table 4.3. Subsequent tables show abbreviated messages and message numbers.
<table>
<thead>
<tr>
<th>No.</th>
<th>Message</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>The children of a person with a <em>BRCA1</em> or <em>BRCA2</em> gene fault each have a 50% (1 in 2) risk of inheriting the gene fault.</td>
</tr>
<tr>
<td>2</td>
<td>Identifying which side of the family is at risk of hereditary cancer is important.</td>
</tr>
<tr>
<td>3</td>
<td>A <em>BRCA1</em> or <em>BRCA2</em> gene fault does not ‘skip’ a generation.</td>
</tr>
<tr>
<td>4</td>
<td>It is possible that breast/ovarian/prostate cancers in the family can be explained by a <em>BRCA1</em> or <em>BRCA2</em> gene fault.</td>
</tr>
<tr>
<td>5</td>
<td>The fault is in the (<em>BRCA1</em> or <em>BRCA2</em>) gene (i.e. specifying the name of the gene involved).</td>
</tr>
<tr>
<td>6</td>
<td>If a person inherits a <em>BRCA1</em> or <em>BRCA2</em> gene fault their risks of breast/ovarian/prostate cancer will be significantly increased but it is not inevitable that they will develop cancer.</td>
</tr>
<tr>
<td>7</td>
<td><em>BRCA1</em> and <em>BRCA2</em> genes are DNA damage repair genes. A fault in one of these genes results in a loss of their function and increases the risk of breast, ovarian and prostate cancer.</td>
</tr>
<tr>
<td>8</td>
<td>Predictive (targeted) genetic testing is available for relatives once a <em>BRCA1</em> or <em>BRCA2</em> gene fault has been identified. This will show whether or not the person has inherited the known faulty gene, and so predicts whether they might be at risk (this is called a predictive test).</td>
</tr>
<tr>
<td>9</td>
<td>If a person does not inherit a known <em>BRCA1</em> or <em>BRCA2</em> gene fault, their risks of breast/ovarian/prostate cancer will be similar to other people in the general population.</td>
</tr>
<tr>
<td>10</td>
<td>In England and Wales, female relatives at 50% (1 in 2) risk of inheriting a gene fault who do not wish to have a genetic test can have the same breast screening as women who are known to have inherited the gene fault. This may be different in other parts of the UK.</td>
</tr>
<tr>
<td>11</td>
<td>It is important to try and inform all your relatives who are at risk that genetic testing is available.</td>
</tr>
<tr>
<td>12</td>
<td>Predictive (targeted) genetic testing for a <em>BRCA1</em> or <em>BRCA2</em> gene fault is not generally offered before the age of 18.</td>
</tr>
<tr>
<td>13</td>
<td>Before having a genetic test, it is important to discuss the implications and possible outcomes</td>
</tr>
<tr>
<td>14</td>
<td>Once a gene fault has been found in a family it is up to each individual to decide if they want a genetic test or not. Some relatives may decide not to be tested.</td>
</tr>
<tr>
<td>15</td>
<td>A personal diagnosis or family history of cancer may affect insurance. There is however currently an agreement between the British Government and the Association of British Insurers which means that people who have had a predictive (targeted) genetic test are not required to disclose the results in order to obtain insurance. There are financial limits to these policies. The agreement is in place until 2017 and it is likely to be extended but this cannot be guaranteed.</td>
</tr>
<tr>
<td>16</td>
<td>Women with breast cancer who have a <em>BRCA1</em> or <em>BRCA2</em> gene fault are at increased risk of developing further primary breast cancers</td>
</tr>
<tr>
<td>17</td>
<td>Women who have/have had ovarian/tubal cancer and have a <em>BRCA1</em> or <em>BRCA2</em> gene fault are at increased risk of developing breast cancer.</td>
</tr>
<tr>
<td>18</td>
<td>Women who have had cancer treatment are advised to discuss the timing of risk-reducing surgery with their oncologist and surgeon before the surgery.</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>---</td>
<td>---</td>
</tr>
<tr>
<td>19</td>
<td>Most BRCA1 related breast cancer is ER-ve and so does not respond to tamoxifen treatment.</td>
</tr>
<tr>
<td>20</td>
<td>Women with breast cancer who have a BRCA1 or BRCA2 gene fault are at increased risk of ovarian/tubal cancer.</td>
</tr>
<tr>
<td>21</td>
<td>In the future, it is likely that research will lead to greater understanding about the role of BRCA1/BRCA2 genes, their interaction with other genes and the role of lifestyle factors in the development of cancer.</td>
</tr>
<tr>
<td>22</td>
<td>Treatment trials may be available for women with cancer who have a BRCA1 or BRCA2 gene fault.</td>
</tr>
<tr>
<td>23</td>
<td>If a woman with cancer and a BRCA1 or BRCA2 gene fault was to develop further breast or ovarian cancer, the treatment may still involve chemotherapy.</td>
</tr>
<tr>
<td>24</td>
<td>Further cancers may be treated differently if a woman has a BRCA1 or BRCA2 gene fault.</td>
</tr>
<tr>
<td>25</td>
<td>A BRCA1 or BRCA2 gene fault does not increase the risk of cancer recurrence or metastases (secondaries).</td>
</tr>
<tr>
<td>26</td>
<td>Breast cancer risk is increased for women without cancer who have a BRCA1 or BRCA2 gene fault.</td>
</tr>
<tr>
<td>27</td>
<td>For women with a fault in BRCA1 or BRCA2, the risk of developing breast cancer between age 25 and 30 may be higher than for women in the general population, but most of the risk occurs after the age of 30.</td>
</tr>
<tr>
<td>28</td>
<td>For women who have a BRCA1 or BRCA2 gene fault, ovarian cancer (including fallopian tube and primary peritoneal cancer) risk is increased.</td>
</tr>
<tr>
<td>29</td>
<td>For women with a fault in BRCA1, the risk of developing ovarian/tubal cancer before age 40 may be increased but most of the risk occurs after the age of 40. For women with a fault in BRCA2, most of the risk occurs after the age of 45.</td>
</tr>
<tr>
<td>30</td>
<td>Prostate cancer risk is increased for men who have a BRCA1 or BRCA2 gene fault. The risk for men with BRCA2 fault is higher than for men with a BRCA1 fault.</td>
</tr>
<tr>
<td>31</td>
<td>Male breast cancer risk is not increased by a BRCA1 gene fault.</td>
</tr>
<tr>
<td>32</td>
<td>The breast cancer risk for men with a BRCA2 fault is 5-10% throughout their lifetime. Men are advised to check their chest and underarms for changes such as lumps, nipple discharge or skin changes and report any changes promptly to their GP but no regular breast screening is recommended.</td>
</tr>
<tr>
<td>33</td>
<td>Pancreatic cancer risk is not increased by a BRCA1 gene fault.</td>
</tr>
<tr>
<td>34</td>
<td>A BRCA2 gene fault may slightly increase the risk of other cancers, such as pancreatic, gall bladder and bile duct cancer. However, the risks are small and there is no screening available.</td>
</tr>
<tr>
<td>35</td>
<td>Diet and lifestyle can make a difference to the risk of cancer generally but the impact is likely to be small compared with the risk associated with the BRCA1/BRCA2 gene fault.</td>
</tr>
<tr>
<td>36</td>
<td>Risk-Reducing Mastectomy (surgery to remove the breasts in order to reduce the risk of cancer) is an option for women who have a BRCA1 or BRCA2 gene fault.</td>
</tr>
<tr>
<td>37</td>
<td>Breast reconstruction is an option after Risk-Reducing Mastectomy or surgery for breast cancer.</td>
</tr>
<tr>
<td>38</td>
<td>Techniques for breast reconstruction have improved in recent years. Women who wish to consider breast reconstruction are advised to see an oncoplastic breast surgeon (i.e. a surgeon specialising in breast cancer and plastic surgery).</td>
</tr>
<tr>
<td>39</td>
<td>Risk-Reducing Mastectomy reduces the risk of breast cancer (but a small risk of breast cancer remains).</td>
</tr>
</tbody>
</table>
40 Risk-Reducing Mastectomy can be offered to women without cancer at a young age (usually from the mid-twenties when the risk of breast cancer starts to increase).

41 Annual breast screening is available from the age of 30 for women who have a *BRCA1* or *BRCA2* gene fault and women at 50:50 (1 in 2) risk of having a gene fault but who have not had a genetic test.

42 There are limitations to breast screening, for example sometimes screening finds breast cancer that would never have caused a woman harm and rarely cancer may be missed by screening.

43 Breast awareness is important.

44 Ovarian screening has not yet been shown to be effective. Therefore, no NHS ovarian screening programme is available. However, women who have symptoms such as fatigue, bloating, loss of appetite or unexplained weight loss are advised to see their GP.

45 Once the risk of ovarian cancer starts to rise, Risk-Reducing Bilateral Salpingo-Oophorectomy (surgery to remove the ovaries and fallopian tubes in order to reduce the risk of cancer) is an option for women who have a *BRCA1* or *BRCA2* gene fault.

46 Bilateral Salpingo-Oophorectomy reduces the ovarian cancer risk (but a small risk of primary peritoneal cancer remains).

47 Bilateral Salpingo-Oophorectomy before the natural menopause may reduce the risk of breast cancer by up to 50% in women with a *BRCA2* gene fault who have not previously had breast cancer.

48 Having a bilateral-salpingo-oophorectomy will result in an early menopause, if a woman has not already gone through this.

49 Following Bilateral Salpingo-Oophorectomy, women without breast cancer are usually offered Hormone Replacement Therapy (HRT) until age 50/51 (ie the average age of the natural menopause). Women who have had breast cancer are advised to discuss the options available for managing menopausal symptoms with their oncologist.

50 There is no UK prostate cancer screening programme. However, a PSA (prostate specific antigen) test can be performed on request following discussion with a General Practitioner about the risks and benefits of testing. Men who have symptoms, such as increased need to pass urine especially at night, reduced urine flow or blood in the urine, are advised to see their General Practitioner.

51 There are tests that can be done before or during pregnancy to reduce the risk of a child being born with the gene fault. These tests are not available or suitable for everyone and the risks, benefits and surrounding issues would need to be discussed with the genetics team before making a decision to proceed.

52 Taking the Oral Contraceptive Pill and having children help to protect a woman from ovarian cancer. However, the Pill increases the risk of breast cancer. It is not suitable for women who have already had breast cancer. Women at high risk are advised to consider alternative methods of contraception.

53 Once a *BRCA1* or *BRCA2* gene fault has been identified, it may be helpful to have a discussion with other specialists in the multidisciplinary team (e.g. oncoplastic breast surgeon, gynaecologist and/or clinical psychologist) in order to understand all the options available.

54 Both men and women can inherit a *BRCA1* or *BRCA2* gene fault. Therefore, if either parent carries a gene fault, each child will have a 50% (1 in 2) chance of inheriting it from them.
| 55 | There are three possible results of a BRCA1 or BRCA2 genetic test for a person with cancer: a cancer-causing gene fault is found which means that a predictive (targeted) genetic test is available for ‘blood relatives’; no cancer-causing gene fault is found, meaning that it is very unlikely that the cancer is due to a BRCA1 or BRCA2 gene fault and a predictive genetic test is not available for relatives; a gene change is found that may or may not cause cancer. This is called a variant of unknown significance (VUS) or an unclassified variant (UV). This result means that a predictive (targeted) genetic test is not available for relatives. |
| 56 | In the general population, approximately 1 woman in 8 develops breast cancer in her lifetime and 1 woman in 50 develops ovarian cancer. Most breast and ovarian cancer occurs after the age of 50 and is not due to a faulty BRCA1 or BRCA2 gene. |
| 57 | Genetic testing may provide helpful risk and management information for women with cancer, as well as for relatives who have not had cancer. |
| 58 | Cancer risks may vary for each individual with a gene fault depending on genetic, environmental and lifestyle factors as well as personal and family history of cancer. |
| 59 | Genetic testing can lead to complex emotions which may be unexpected, like shock, fear, sadness and upset, especially close to the result. The health care team can provide information about the support that is available to help with this. |
| 60 | Some people feel guilty about the possibility that their children and grandchildren may inherit the faulty gene from them. |
| 61 | Identifying a faulty gene can help people to be more aware of the symptoms of cancer. |
| 62 | For a woman who has had breast or ovarian cancer, a Risk-Reducing or Contralateral Mastectomy will reduce the risk of a future new primary breast cancer but will not reduce the risk of metastases from the initial cancer. |
| 63 | Women who have not had cancer may wish to consider medication to reduce the risk of breast cancer (chemoprevention). The benefits, side effects and limitations of this will be discussed with the health professional undertaking the testing if appropriate. |
4.4.6.3. Timing of communication

For messages judged as key or probably key, participants were asked to decide the optimal timing of communication. The score for each time point were equally scored as one point (1). The timing options were:

- The information should be communicated before genetic testing
- The information should be communicated once a pathogenic variant has been detected
- Not sure or don’t have a view. (The neutral option was removed after the first circulation of each message to increase positive or negative responses.)
- The information should be communicated before genetic testing and once a pathogenic variant has been detected
- The information should be communicated at another time altogether.

4.5. ANALYSIS

4.5.1. Key messages

The mean score and its standard deviation (SD) for each message at each round and for each group was calculated using these functions in SPSS. The final mean and SD for each message was calculated once agreement was reached or, for messages where no agreement was reached, at the end of Round 3. The genetics health professionals and cancer health professionals were surveyed separately in each round. Once each message had been agreed as key or not key by each group of health professionals, the responses were combined to calculate the final score for each message. Where participants did not continue with the study, their last recorded score was counted as their final score.

Mean scores for each message judged by health professionals and service users were organised in descending order, enabling identification of messages with ≥ 75% agreement as key messages. These messages were grouped into those with ≥ 95%, 85-94% and 75-84% agreement as a key message, messages where no agreement was reached and messages with ≥ 75% agreement as not key.

4.5.2. Reaching agreement

The number of messages circulated and agreed as key or not key at each round was documented for health professionals and service users. For health professionals, the final assessment of each message was calculated once agreement was reached by the genetics and the cancer health professionals. For example, a key message agreed
in round 1 by the cancer health professionals but in round 3 by the genetics health professionals was recorded as having reached agreement in round 3.

4.5.3. Timing of communication
Analysis of the optimal timing of communication involved identifying the number and percentage of responses in each time point for each key message. Agreement was reached when $\geq 75\%$ of participants agreed on the timing of communication.

4.6. RESULTS

4.6.1. Key messages
4.6.1.1. Health professionals: key/not key messages
The health professionals reached agreement about 52 messages in total (82.5%): 34 messages (54%) were agreed as key. Of these, eight key messages (24%) reached $\geq 95\%$ agreement. Eighteen messages (29%) were agreed as not key. There was no agreement about 11 messages (17%) (see Table 4.3).

4.6.1.2. Service users: key/not key messages
The service users reached agreement about 46 messages (73%): 35 messages (56%) were agreed as key. Of these, 20 key messages (57%) reached $\geq 95\%$ agreement) and 11 key messages (31%) reached 75-84% agreement. 11 messages (17%) were not key. There was no agreement about 17 messages (27%) (see Table 4.4).

4.6.1.3. Health professionals and service users: agreement about key/not key messages
Health professionals and service users agreed about 40 messages (63.5%): 30 messages (48%) were key and 10 messages (16%) were not key (see Table 4.5). Seven key messages reached $\geq 95\%$ agreement within both groups:

- Message 1. The children of a person with a $BRCA1$ or $BRCA2$ gene fault each have a 50% (1 in 2) risk of inheriting the gene fault.
- Message 8. Predictive (targeted) genetic testing is available for relatives once a $BRCA1$ or $BRCA2$ gene fault has been identified. This will show whether or not the person has inherited the known faulty gene, and so predicts whether they might be at risk (this is called a predictive test).
• Message 13. Before having a genetic test, it is important to discuss the implications and possible outcomes

• Message 26. Breast cancer risk is increased for women without cancer who have a BRCA1 or BRCA2 gene fault.

• Message 28. For women who have a BRCA1 or BRCA2 gene fault, ovarian cancer (including fallopian tube and primary peritoneal cancer) risk is increased.

• Message 36. Risk-Reducing Mastectomy (surgery to remove the breasts in order to reduce the risk of cancer) is an option for women who have a BRCA1 or BRCA2 gene fault.

• Message 45. Once the risk of ovarian cancer starts to rise, Risk-Reducing Bilateral Salpingo-Oophorectomy (surgery to remove the ovaries and fallopian tubes in order to reduce the risk of cancer) is an option for women who have a BRCA1 or BRCA2 gene fault.

4.6.1.4. **Health professionals and service users: disagreement about key/not key messages**

There was disagreement between health professionals and service users about the following three messages (see Table 4.6):

• Message 9. ‘If a person does not inherit a known BRCA1 or BRCA2 gene fault, their risks of breast/ovarian/prostate cancer will be similar to other people in the general population.’ Health professionals viewed message 9 as key (mean 1.5, SD 1.03) whilst service users viewed this as not a key message (mean 0.5, SD 1.46).

• Message 34. ‘A BRCA2 gene fault may slightly increase the risk of other cancers, such as pancreatic, gall bladder and bile duct cancer. However, the risks are small and there is no screening available.’ Service users viewed message 34 as key (mean 1.63, SD 0.62) but health professionals viewed this as not a key message (mean 0.38, SD 1.15).

• Message 35. ‘Diet and lifestyle can make a difference to the risk of cancer generally but the impact is likely to be small compared with the risk associated with the BRCA1 or BRCA2 gene fault.’ Service users viewed message 35 as key (mean 1.56, SD 1.03) whilst health professionals viewed this as not a key message (mean 0.06 SD 1.34).
Table 4.4. Number and abbreviated messages a) agreed as key, b) where no agreement was reached, c) agreed as not key by health professionals

<table>
<thead>
<tr>
<th>No.</th>
<th>Messages agreed as KEY (≥ 95% agreement)</th>
<th>Mean</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>AD inheritance</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>26</td>
<td>Increased risk BC (at-risk women)</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>36</td>
<td>RRM available</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>45</td>
<td>BSO available</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>8</td>
<td>Predictive test available</td>
<td>1.94</td>
<td>0.25</td>
</tr>
<tr>
<td>28</td>
<td>Increased risk OC (at-risk women)</td>
<td>1.94</td>
<td>0.25</td>
</tr>
<tr>
<td>13</td>
<td>Importance of discussion</td>
<td>1.94</td>
<td>0.25</td>
</tr>
<tr>
<td>44</td>
<td>No OC screening</td>
<td>1.94</td>
<td>0.25</td>
</tr>
</tbody>
</table>

**Messages agreed as KEY (85% to 94% agreement)**

<table>
<thead>
<tr>
<th>No.</th>
<th>Messages agreed as KEY (85% to 94% agreement)</th>
<th>Mean</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td>37</td>
<td>Breast reconstruction option</td>
<td>1.88</td>
<td>0.34</td>
</tr>
<tr>
<td>39</td>
<td>RRM reduces the risk of BC</td>
<td>1.88</td>
<td>0.34</td>
</tr>
<tr>
<td>54</td>
<td>Male inheritance</td>
<td>1.86</td>
<td>0.36</td>
</tr>
<tr>
<td>41</td>
<td>Breast screening available</td>
<td>1.81</td>
<td>0.54</td>
</tr>
<tr>
<td>16</td>
<td>Risk 2nd primary BC</td>
<td>1.81</td>
<td>0.75</td>
</tr>
<tr>
<td>46</td>
<td>BSO reduces OC/FTC risk</td>
<td>1.81</td>
<td>0.75</td>
</tr>
<tr>
<td>55</td>
<td>Diagnostic testing outcomes</td>
<td>1.79</td>
<td>0.43</td>
</tr>
<tr>
<td>30</td>
<td>Increased risk of PC</td>
<td>1.75</td>
<td>0.45</td>
</tr>
<tr>
<td>47</td>
<td>BSO may reduce BC risk</td>
<td>1.75</td>
<td>0.58</td>
</tr>
<tr>
<td>5</td>
<td>The gene involved</td>
<td>1.75</td>
<td>0.78</td>
</tr>
<tr>
<td>62</td>
<td>Risk from initial cancer CM</td>
<td>1.71</td>
<td>0.47</td>
</tr>
</tbody>
</table>

**Messages agreed as KEY (75% to 84% agreement)**

<table>
<thead>
<tr>
<th>No.</th>
<th>Messages agreed as KEY (75% to 84% agreement)</th>
<th>Mean</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td>20</td>
<td>Risk OC for BC pts.</td>
<td>1.69</td>
<td>0.79</td>
</tr>
<tr>
<td>48</td>
<td>BSO will result in menopause</td>
<td>1.69</td>
<td>0.79</td>
</tr>
<tr>
<td>57</td>
<td>Information for patient</td>
<td>1.64</td>
<td>0.84</td>
</tr>
<tr>
<td>10</td>
<td>Breast screen at 50% risk</td>
<td>1.63</td>
<td>0.81</td>
</tr>
<tr>
<td>3</td>
<td>Gene fault does not 'skip' a generation</td>
<td>1.56</td>
<td>0.51</td>
</tr>
<tr>
<td>27</td>
<td>Age of BC risk</td>
<td>1.56</td>
<td>0.51</td>
</tr>
<tr>
<td>29</td>
<td>Age of OC risk</td>
<td>1.56</td>
<td>0.51</td>
</tr>
<tr>
<td>4</td>
<td>Gene fault may explain family history</td>
<td>1.56</td>
<td>0.81</td>
</tr>
<tr>
<td>6</td>
<td>Cancer not inevitable for carriers</td>
<td>1.56</td>
<td>0.89</td>
</tr>
<tr>
<td>2</td>
<td>Identifying the side of the family at risk</td>
<td>1.56</td>
<td>1.03</td>
</tr>
<tr>
<td>17</td>
<td>Risk BC for OC patients</td>
<td>1.56</td>
<td>1.03</td>
</tr>
<tr>
<td>14</td>
<td>Testing is an individual decision</td>
<td>1.50</td>
<td>0.52</td>
</tr>
<tr>
<td>42</td>
<td>Breast screening limitations</td>
<td>1.50</td>
<td>0.82</td>
</tr>
<tr>
<td>9</td>
<td>Population risk if gene fault not inherited</td>
<td>1.50</td>
<td>1.03</td>
</tr>
<tr>
<td>11</td>
<td>Inform at-risk relatives</td>
<td>1.50</td>
<td>1.03</td>
</tr>
<tr>
<td></td>
<td>Messages where no agreement was reached</td>
<td></td>
<td></td>
</tr>
<tr>
<td>---</td>
<td>----------------------------------------</td>
<td>---</td>
<td>---</td>
</tr>
<tr>
<td>12</td>
<td>Predictive is not offered &lt;18</td>
<td>1.44</td>
<td>1.03</td>
</tr>
<tr>
<td>38</td>
<td>Discuss breast reconstruction with specialist</td>
<td>1.40</td>
<td>0.63</td>
</tr>
<tr>
<td>32</td>
<td>Male breast cancer BRCA2</td>
<td>1.31</td>
<td>0.87</td>
</tr>
<tr>
<td>56</td>
<td>Population risk BC and OC</td>
<td>1.29</td>
<td>1.27</td>
</tr>
<tr>
<td>15</td>
<td>Insurance</td>
<td>1.25</td>
<td>0.86</td>
</tr>
<tr>
<td>49</td>
<td>HRT after BSO</td>
<td>1.25</td>
<td>1.00</td>
</tr>
<tr>
<td>53</td>
<td>MDT discussion</td>
<td>1.13</td>
<td>0.96</td>
</tr>
<tr>
<td>59</td>
<td>Psychological impact</td>
<td>1.07</td>
<td>0.99</td>
</tr>
<tr>
<td>51</td>
<td>Reproductive options</td>
<td>1.00</td>
<td>0.89</td>
</tr>
<tr>
<td>63</td>
<td>Chemoprevention</td>
<td>1.00</td>
<td>1.18</td>
</tr>
<tr>
<td>43</td>
<td>Breast awareness</td>
<td>0.75</td>
<td>1.48</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th></th>
<th>Messages agreed as NOT key (≥ 75% agreement)</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>58</td>
<td>Personal cancer risks</td>
<td>0.50</td>
<td>1.09</td>
</tr>
<tr>
<td>60</td>
<td>Guilt</td>
<td>0.50</td>
<td>1.56</td>
</tr>
<tr>
<td>31</td>
<td>Male breast cancer risk BRCA1</td>
<td>0.44</td>
<td>1.31</td>
</tr>
<tr>
<td>34</td>
<td>Other cancer risks BRCA2</td>
<td>0.38</td>
<td>1.15</td>
</tr>
<tr>
<td>40</td>
<td>RRM offered at a young age</td>
<td>0.31</td>
<td>1.30</td>
</tr>
<tr>
<td>22</td>
<td>Treatment trials</td>
<td>0.25</td>
<td>1.57</td>
</tr>
<tr>
<td>35</td>
<td>Diet and lifestyle</td>
<td>0.06</td>
<td>1.34</td>
</tr>
<tr>
<td>52</td>
<td>Oral Contraceptive Pill</td>
<td>0.00</td>
<td>1.37</td>
</tr>
<tr>
<td>50</td>
<td>No PC screening programme</td>
<td>-0.13</td>
<td>1.26</td>
</tr>
<tr>
<td>25</td>
<td>No increased risk of metastases</td>
<td>-0.19</td>
<td>1.38</td>
</tr>
<tr>
<td>21</td>
<td>Future research and understanding</td>
<td>-0.31</td>
<td>1.35</td>
</tr>
<tr>
<td>18</td>
<td>Timing of risk-reducing surgery</td>
<td>-0.50</td>
<td>1.32</td>
</tr>
<tr>
<td>61</td>
<td>Symptom awareness</td>
<td>-0.64</td>
<td>0.93</td>
</tr>
<tr>
<td>7</td>
<td>Role of BRCA1 and BRCA2</td>
<td>-0.69</td>
<td>1.35</td>
</tr>
<tr>
<td>33</td>
<td>Other cancer risks BRCA1</td>
<td>-0.69</td>
<td>1.14</td>
</tr>
<tr>
<td>23</td>
<td>Future chemotherapy</td>
<td>-0.88</td>
<td>1.09</td>
</tr>
<tr>
<td>24</td>
<td>Impact on treatment</td>
<td>-0.94</td>
<td>1.29</td>
</tr>
<tr>
<td>19</td>
<td>Tamoxifen</td>
<td>-1.19</td>
<td>1.05</td>
</tr>
</tbody>
</table>

Abbreviations: AD (autosomal dominant), BC (breast cancer), OC (ovarian cancer), FTC (fallopian tube cancer), PC (prostate cancer), RRM (Risk-Reducing Mastectomy), BSO (Bilateral salpingo-oophorectomy), CM (contralateral mastectomy), HRT (hormone replacement therapy), MDT (multidisciplinary team), FH (family history)
Table 4.5. Number and abbreviated messages a) agreed as key, b) where no agreement was reached, c) agreed as not key by service users

<table>
<thead>
<tr>
<th>No.</th>
<th>Abbreviated message</th>
<th>Mean</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td><strong>Messages agreed as KEY (≥ 95% agreement)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1</td>
<td>AD inheritance</td>
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<td>0.00</td>
</tr>
<tr>
<td>2</td>
<td>Identifying the side of the family at risk</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>6</td>
<td>Cancer not inevitable for carriers</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>8</td>
<td>Predictive test available</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>13</td>
<td>Importance of discussion</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>16</td>
<td>Risk 2nd primary BC</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>17</td>
<td>Risk BC for OC patients</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>20</td>
<td>Risk OC for BC patients</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>26</td>
<td>Increased risk BC (at-risk women)</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>28</td>
<td>Increased risk OC (at-risk women)</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>32</td>
<td>Male breast cancer BRCA2</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>36</td>
<td>RRM available</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>39</td>
<td>RRM reduces the risk of BC</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>41</td>
<td>Breast screening available</td>
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<td>0.00</td>
</tr>
<tr>
<td>43</td>
<td>Breast awareness</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>45</td>
<td>BSO available</td>
<td>2.00</td>
<td>0.00</td>
</tr>
<tr>
<td>27</td>
<td>Age of BC risk</td>
<td>1.94</td>
<td>0.25</td>
</tr>
<tr>
<td>37</td>
<td>Breast reconstruction option</td>
<td>1.94</td>
<td>0.25</td>
</tr>
<tr>
<td>48</td>
<td>BSO will result in menopause</td>
<td>1.94</td>
<td>0.25</td>
</tr>
<tr>
<td></td>
<td><strong>Messages agreed as KEY (85% to 94% agreement)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>11</td>
<td>Inform at-risk relatives</td>
<td>1.88</td>
<td>0.34</td>
</tr>
<tr>
<td>4</td>
<td>Gene fault may explain family history</td>
<td>1.81</td>
<td>0.40</td>
</tr>
<tr>
<td>44</td>
<td>No OC screening</td>
<td>1.81</td>
<td>0.40</td>
</tr>
<tr>
<td>62</td>
<td>Risk from the initial cancer CM</td>
<td>1.77</td>
<td>0.44</td>
</tr>
<tr>
<td></td>
<td><strong>Messages agreed as KEY (75% to 84% agreement)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>15</td>
<td>Insurance</td>
<td>1.69</td>
<td>0.79</td>
</tr>
<tr>
<td>46</td>
<td>BSO reduces OC/FTC risk</td>
<td>1.69</td>
<td>0.79</td>
</tr>
<tr>
<td>34</td>
<td>Other cancer risks BRCA2</td>
<td>1.63</td>
<td>0.62</td>
</tr>
<tr>
<td>14</td>
<td>Testing is an individual decision</td>
<td>1.63</td>
<td>0.81</td>
</tr>
<tr>
<td>10</td>
<td>Breast screen at 50% risk</td>
<td>1.63</td>
<td>0.89</td>
</tr>
<tr>
<td>57</td>
<td>Information for patient</td>
<td>1.62</td>
<td>0.51</td>
</tr>
<tr>
<td>47</td>
<td>BSO may reduce BC risk</td>
<td>1.56</td>
<td>0.51</td>
</tr>
<tr>
<td>5</td>
<td>The gene involved</td>
<td>1.56</td>
<td>0.81</td>
</tr>
<tr>
<td>29</td>
<td>Age of OC risk</td>
<td>1.56</td>
<td>0.81</td>
</tr>
<tr>
<td>35</td>
<td>Diet and lifestyle</td>
<td>1.56</td>
<td>1.03</td>
</tr>
<tr>
<td></td>
<td>Message</td>
<td>Agreement</td>
<td>Disagreement</td>
</tr>
<tr>
<td>---</td>
<td>-------------------------------------------------------------------------</td>
<td>-----------</td>
<td>--------------</td>
</tr>
<tr>
<td>42</td>
<td>Breast screening limitations</td>
<td>1.50</td>
<td>0.82</td>
</tr>
<tr>
<td></td>
<td><strong>Messages where no agreement was reached</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>54</td>
<td>Male inheritance</td>
<td>1.46</td>
<td>0.97</td>
</tr>
<tr>
<td>7</td>
<td>Role of BRCA1 and BRCA2</td>
<td>1.44</td>
<td>0.73</td>
</tr>
<tr>
<td>50</td>
<td>No PC screening programme</td>
<td>1.44</td>
<td>0.51</td>
</tr>
<tr>
<td>3</td>
<td>Gene fault does not 'skip' a generation</td>
<td>1.38</td>
<td>0.81</td>
</tr>
<tr>
<td>22</td>
<td>Treatment trials</td>
<td>1.31</td>
<td>1.01</td>
</tr>
<tr>
<td>59</td>
<td>Psychological impact</td>
<td>1.31</td>
<td>0.86</td>
</tr>
<tr>
<td>25</td>
<td>No increased risk of metastases</td>
<td>1.25</td>
<td>1.18</td>
</tr>
<tr>
<td>55</td>
<td>Diagnostic testing outcomes</td>
<td>1.15</td>
<td>1.28</td>
</tr>
<tr>
<td>31</td>
<td>Male breast cancer risk BRCA1</td>
<td>1.13</td>
<td>0.96</td>
</tr>
<tr>
<td>40</td>
<td>RRM possible at a young age</td>
<td>1.13</td>
<td>0.96</td>
</tr>
<tr>
<td>63</td>
<td>Chemoprevention</td>
<td>1.08</td>
<td>1.04</td>
</tr>
<tr>
<td>38</td>
<td>Discuss breast reconstruction with specialist</td>
<td>1.06</td>
<td>0.93</td>
</tr>
<tr>
<td>12</td>
<td>Predictive is not offered &lt;18</td>
<td>0.94</td>
<td>1.12</td>
</tr>
<tr>
<td>49</td>
<td>HRT after BSO</td>
<td>0.81</td>
<td>1.17</td>
</tr>
<tr>
<td>53</td>
<td>MDT discussion</td>
<td>0.81</td>
<td>0.91</td>
</tr>
<tr>
<td>51</td>
<td>Reproductive options</td>
<td>0.69</td>
<td>1.54</td>
</tr>
<tr>
<td>56</td>
<td>Population risk BC and OC</td>
<td>0.69</td>
<td>1.44</td>
</tr>
<tr>
<td></td>
<td><strong>Messages agreed as NOT key (≥ 75% agreement)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>9</td>
<td>Population risk if gene fault not inherited</td>
<td>0.50</td>
<td>1.46</td>
</tr>
<tr>
<td>18</td>
<td>Timing of risk-reducing surgery</td>
<td>0.44</td>
<td>1.37</td>
</tr>
<tr>
<td>24</td>
<td>Impact on treatment</td>
<td>0.44</td>
<td>1.55</td>
</tr>
<tr>
<td>58</td>
<td>Personal cancer risks</td>
<td>0.38</td>
<td>1.45</td>
</tr>
<tr>
<td>61</td>
<td>Symptom awareness</td>
<td>0.38</td>
<td>1.71</td>
</tr>
<tr>
<td>19</td>
<td>Tamoxifen</td>
<td>0.25</td>
<td>1.29</td>
</tr>
<tr>
<td>21</td>
<td>Future research/understanding</td>
<td>0.19</td>
<td>1.38</td>
</tr>
<tr>
<td>33</td>
<td>Other cancer risks BRCA1</td>
<td>0.13</td>
<td>1.50</td>
</tr>
<tr>
<td>52</td>
<td>Oral Contraceptive Pill</td>
<td>0.06</td>
<td>1.48</td>
</tr>
<tr>
<td>23</td>
<td>Future chemotherapy</td>
<td>-0.06</td>
<td>1.61</td>
</tr>
<tr>
<td>60</td>
<td>Guilt</td>
<td>-0.23</td>
<td>1.24</td>
</tr>
</tbody>
</table>

Abbreviations: AD (autosomal dominant), BC (breast cancer), OC (ovarian cancer), FTC (fallopian tube cancer), PC (prostate cancer), RRM (Risk-Reducing Mastectomy), BSO (Bilateral salpingo-oophorectomy), CM (contralateral mastectomy), HRT (hormone replacement therapy), MDT (multidisciplinary team), FH (family history)
Table 4.6. Agreement and disagreement between health professionals and service users about key/not key messages showing message numbers

<table>
<thead>
<tr>
<th>Key messages</th>
<th>Not key messages</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Health professionals</strong></td>
<td><strong>Service users</strong></td>
</tr>
<tr>
<td>1. AD inheritance</td>
<td>9. Population risk if gene fault not inherited</td>
</tr>
<tr>
<td>2. Identifying the side of family at risk</td>
<td>18. Timing of risk-reducing surgery</td>
</tr>
<tr>
<td>4. Gene fault may explain FH</td>
<td>19. Tamoxifen</td>
</tr>
<tr>
<td>5. The gene involved</td>
<td>21. Future research/understanding</td>
</tr>
<tr>
<td>6. Cancer not inevitable for carriers</td>
<td>23. Future chemotherapy</td>
</tr>
<tr>
<td>10. Breast screen at 50% risk</td>
<td>33. Other cancer risks BRCA1</td>
</tr>
<tr>
<td>11. Inform at risk relatives</td>
<td>52. Oral Contraceptive Pill</td>
</tr>
<tr>
<td>13. Importance of discussion</td>
<td>58. Personal cancer risks</td>
</tr>
<tr>
<td>14. Testing is an individual decision</td>
<td>60. Guilt</td>
</tr>
<tr>
<td>16. Risk 2nd primary BC</td>
<td>61. Symptom aware</td>
</tr>
<tr>
<td>17. Risk BC for OC patients</td>
<td></td>
</tr>
<tr>
<td>20. Risk OC for BC patients</td>
<td></td>
</tr>
<tr>
<td>26. Increased risk BC (at-risk)</td>
<td></td>
</tr>
<tr>
<td>27. Age of BC risk</td>
<td></td>
</tr>
<tr>
<td>28. Increased risk OC (at-risk)</td>
<td></td>
</tr>
<tr>
<td>29. Age of OC risk</td>
<td></td>
</tr>
<tr>
<td>30. Increased risk PC</td>
<td></td>
</tr>
<tr>
<td>36. RRM available</td>
<td></td>
</tr>
<tr>
<td>37. Breast reconstruction option</td>
<td></td>
</tr>
<tr>
<td>39. RRM reduces BC risk</td>
<td></td>
</tr>
<tr>
<td>41. Breast screening available</td>
<td></td>
</tr>
<tr>
<td>47. Breast screening limitations</td>
<td></td>
</tr>
<tr>
<td>44. No OC screening</td>
<td></td>
</tr>
<tr>
<td>45. BSO available</td>
<td></td>
</tr>
<tr>
<td>46. BSO reduces OC/FTC risk</td>
<td></td>
</tr>
<tr>
<td>47. BSO may reduce BC risk</td>
<td></td>
</tr>
<tr>
<td>48. BSO will result in menopause</td>
<td></td>
</tr>
<tr>
<td>57. Information for patient</td>
<td></td>
</tr>
<tr>
<td>62. Risk from initial cancer CM</td>
<td></td>
</tr>
<tr>
<td>34. Other cancer risks BRCA2</td>
<td></td>
</tr>
<tr>
<td>35. Diet and lifestyle</td>
<td></td>
</tr>
</tbody>
</table>

Abbreviations: AD (autosomal dominant), BC (breast cancer), OC (ovarian cancer), FTC (fallopian tube cancer), PC (prostate cancer), RRM (Risk-Reducing Mastectomy), BSO (Bilateral salpingo-oophorectomy), CM (contralateral mastectomy), HRT (hormone replacement therapy), MDT (multidisciplinary team), FH (family history)
4.6.1.5. Messages agreed as key by health professionals/no agreement amongst service users
Key messages agreed by health professionals but not service users were about the possible outcomes, limitations and informativeness of testing, male inheritance and that a pathogenic variant cannot ‘skip a generation’.

4.6.1.6. Messages agreed as key by service users/no agreement amongst health professionals
Key messages agreed by service users but not health professionals were about genetic testing and the impact on insurance, the breast cancer risk for men with a BRCA2 pathogenic variant and the importance of breast awareness.

4.6.2. Reaching agreement
4.6.2.1. Health professionals: reaching agreement
18 key messages (53%) were identified in round 1; eight key messages (23.5%) were identified in round 2 and eight key messages were identified (23.5%) in round 3. Three of the key messages (54, 55 and 62) agreed in round 2 had been circulated for the first time in round 2 following suggestions from participants.

Fourteen messages agreed as not key (78%) were identified in round 1; two of these messages (11%) were identified in round 2 and two (11%) were identified in round 3. The number of survey rounds completed by health professionals in order to reach agreement about key messages and messages that are not key are shown in Figure 4.5.

4.6.2.2. Service users: reaching agreement
Amongst service users, 22 key messages (63%) were identified in round 1; five key messages (14%) were identified in round 2 and eight key messages (23%) were identified in round 3. One key message (57) agreed in round 2 had been circulated for the first time in round 2 following suggestions from participants.

Seven messages agreed as not key (64%) were identified in round 1, three not key messages (27%) were identified in round 2 and 1 (9%) not key message was identified in round 3. The number of rounds completed by service users in order to reach agreement about key messages and messages that are not key are shown in Figure 4.1.
# 4.6.2.3. Health professionals and service users: reaching agreement

Fourteen of the key messages (47%) agreed by both health professionals and service users were agreed following round 1, a further six key messages (20%) were agreed following round 2 and the remaining 10 key messages (33%) were agreed following round 3.

Message 10 was agreed as key in round 2 by both groups. Messages 14 and 42 were agreed as key in round 3 by both groups.

Eight messages (12, 38, 49, 51, 53, 56, 59 and 63) failed to reach agreement by either group. The number of rounds in which health professionals and service users reached agreement about key messages is shown in Figure 4.1. Figure 1 shows a flow diagram of the Delphi exercise showing messages agreed and re-circulated at each round by health professionals and service users and messages where there was no final agreement.

# 4.6.3. Timing of communication

## 4.6.3.1. Health professionals: timing of communication

For 18 (53%) of the 34 key messages agreed by the health professionals, there was ≥75% agreement about the timing of communication. For 17 (94%) of these messages it was agreed that the optimal timing of communication is before genetic testing and once a pathogenic variant has been identified. Health professionals agreed that message 5 ‘The fault is in the [BRCA1 or BRCA2] gene [i.e. specifying the name of the gene involved]’ should be communicated once a pathogenic variant has been identified. For the remaining 16 key messages (47%) there was no agreement about the timing of communication amongst health professionals.

## 4.6.3.2. Service users: timing of communication

For 25 (71%) of the key messages agreed by the service users, there was ≥75% agreement about the timing of communication. For all these key messages, it was agreed that the optimal timing of communication is before genetic testing and once a pathogenic variant has been identified. For the remaining 10 key messages (29%) there was no agreement about the timing of communication amongst service users.
Figure 4.1: Flow diagram of the Delphi exercise showing the messages agreed as key messages (KM) and not key messages (NKM) and timing of communication in each round of the survey; the number of circulated messages and the message numbers agreed in each round.
4.6.3.3. Health professionals and service users: timing of communication

Of the 30 key messages agreed by health professionals and service users, both groups agreed that 13 key messages should be communicated before genetic testing and once a pathogenic variant has been identified. Table 4.5 shows agreement and lack of agreement between health professionals and service users about the timing of communication of key messages.

Table 4.7. Agreement and lack of agreement between health professionals and service users about the timing of communication of key messages, showing message numbers

<table>
<thead>
<tr>
<th>Health professionals</th>
<th>Service users</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Timing agreed</strong></td>
<td><strong>Timing not agreed</strong></td>
</tr>
<tr>
<td>6. Cancer not inevitable for carriers</td>
<td>2. Identifying the side of the family at risk</td>
</tr>
<tr>
<td>8. Predictive test available</td>
<td>4. Gene fault may explain FH</td>
</tr>
<tr>
<td>16. Risk 2nd primary BC</td>
<td>13. Importance of discussion</td>
</tr>
<tr>
<td>17. Risk BC for OC patients</td>
<td>27. Age BC risk</td>
</tr>
<tr>
<td>20. Risk OC for BC patients</td>
<td>29. Age of OC risk</td>
</tr>
<tr>
<td>26. Increased risk BC (at-risk)</td>
<td>30. Increased risk PC</td>
</tr>
<tr>
<td>28. Increased risk OC (at-risk)</td>
<td>39. RRM reduces the risk of BC</td>
</tr>
<tr>
<td>36. RRM available</td>
<td>48. BSO will result in menopause</td>
</tr>
<tr>
<td>37. Breast reconstruction option</td>
<td>57. Information for patient</td>
</tr>
<tr>
<td>41. Breast screening available</td>
<td>1. AD inheritance</td>
</tr>
<tr>
<td>44. No OC screening</td>
<td>5. The gene involved</td>
</tr>
<tr>
<td>45. BSO available</td>
<td>41. Testing is an individual decision</td>
</tr>
<tr>
<td>46. BSO reduces OC/FTC risk</td>
<td>10. Breast screen at 50% risk</td>
</tr>
<tr>
<td>1. AD inheritance</td>
<td>11. Inform at-risk relatives</td>
</tr>
<tr>
<td>5. The gene involved</td>
<td>42. Breast screening limitations</td>
</tr>
<tr>
<td>41. Testing is an individual decision</td>
<td>47. BSO may reduce BC risk</td>
</tr>
</tbody>
</table>
| 62. Risk from initial cancer CM | **Abbreviations:** AD (autosomal dominant), BC (breast cancer), OC (ovarian cancer), FTC (fallopian tube cancer), PC (prostate cancer), RRM (Risk-Reducing Mastectomy), BSO (Bilateral salpingo-oophorectomy), CM (contralateral mastectomy), HRT (hormone replacement therapy), MDT (multidisciplinary team), FH (family history)
4.7. DISCUSSION

4.7.1. Key messages

It has been suggested that there are significant differences between genetics and non-genetics professionals that may impact on the way in which genetics information is communicated to patients (Middleton et al., 2015). This study found a high level of agreement about messages that are key and not key within and between the groups. This finding indicates that, despite differences in professional background, training, approach and focus, there is a shared professional knowledge amongst these two types of health professionals.

This study supports the findings of a review of genetic and non-genetics interactions (Smets et al., 2007) which showed that both types of professional interaction deal with the challenge of attuning the patient’s agenda with enhancing understanding, providing emotional support and discussing risk-reducing behaviour. The finding suggests that expert health professionals specialising in cancer genetics and cancer are closely aligned in their knowledge about \textit{BRCA1}/\textit{BRCA2} and share similar views about the information required by women with breast or ovarian cancer.

The high level of agreement amongst the service users suggests a level of consistency in the information provided during genetic counselling in the UK. Lay understandings are inclined to incorporate medical concepts and ideas, even if the form of the knowledge changes slightly (Shaw, 2002). This, together with raised general awareness about hereditary breast and ovarian cancer amongst health professionals and the general public over recent years (Evans et al., 2014), may have contributed to the high level of agreement between the health professional and service user groups.

The messages agreed as key by the health professionals but not the service users refer to the potential implications of testing and concepts of inheritance which may not have been familiar or obviously relevant to service users. Health professionals would have been aware that the meaning of a negative predictive test result (message 9) and the outcomes of diagnostic testing (message 55) are both included within the NICE familial breast cancer guidelines (National Institute for Health and Care Excellence, 2013) within the recommendation: ‘Eligible people and their affected relatives should be informed about the likely informativeness of the test (the meaning of a positive and a negative test).’ (p.85). Service users are unlikely to have been aware of the consequences of not knowing this information. Information that inheritance does not
‘skip a generation’ (message 3) and male inheritance (message 54) would be considered important for informed decision-making by health professionals, although arguably provided the information about dominant inheritance (message 1) has been understood, these two messages are not required. It is documented that concepts of inheritance are constructed from family experience and relationships (Hopwood, 2000; Richards, 1996) and so this information may not have had any meaning for at least some of the service users.

The messages agreed as key by the service users and not the health professionals refer to information that empowers women to protect themselves and their families. These messages provide information that may be more relevant to the family than to the patient. Information about insurance (message 15) and male breast cancer (message 32) for example may be helpful information for patients to communicate to relatives. Information about the risk of other cancers (message 34) provides information about cancer awareness, even when no surveillance or risk-reducing measure are available. For the patients, information about diet and lifestyle (message 35) and breast awareness (message 43) provide information about potential ways to reduce risk or detect cancer early, even with limited evidence. This finding is consistent with research that shows that the vast majority of cancer patients prefer to be kept well informed about their illness (Jenkins, Fallowfield, & Saul, 2001). Having information needs met is empowering for cancer patients (Bakker, Fitch, Gray, Reed, & Bennett, 2001).

Previous studies have demonstrated that protecting the family is a powerful motivator for genetic testing amongst women with breast cancer (Julian-Reynier et al., 1998). The health professionals may have been mindful that the information communicated by these messages is of greater relevance to unaffected relatives than to the patients and may also have wanted to manage the amount and complexity of information communicated. Studies have shown that the vast majority of medical information communicated is not recalled and that the more information given, the lower the proportion recalled (Kessels, 2003). Low recall of information amongst women with cancer and relatives following genetic counselling about a \textit{BRCA1/BRCA2} pathogenic variant has been demonstrated in several studies (Sermijn et al., 2004, Jacobs et al., 2015; Vos, Menko et al., 2011).

Eliciting the patient’s communication needs and tailoring information accordingly is fundamental to effective clinician-patient communication (Hack, Degner, & Parker, 2005). Some of the key messages identified in this study will not be relevant to all women and some women will want more or less information. It may help health
professionals to be aware that some information considered key by health professionals may not be considered key by cancer patients and vice versa. There was at least 95% agreement amongst both groups that patients making decisions about breast and ovarian cancer genetic testing and hereditary cancer management require information about dominant inheritance, the availability of predictive testing for relatives, the importance of discussing the implications and possible outcomes of testing prior to having a test, increased risk of breast and ovarian, tubal and primary peritoneal cancer and the option of risk-reducing mastectomy and risk-reducing bilateral salpingo-oophorectomy. One message about the effectiveness and availability of ovarian screening and symptom awareness (message 44) was agreed by at least 95% of health professionals but did not reach this level of agreement by service users.

Where time or the ability to assimilate information is limited, for example for patients undergoing genetic testing shortly after diagnosis or at other vulnerable times, these key messages provide health professionals with a minimum set of information to communicate. Given the large number of key messages required, there is little justification for communicating the 10 messages agreed as not key by both groups, unless the information is specifically requested by the patient.

4.7.2. Reaching agreement

Amongst health professionals and service users, most of the key and not key messages were agreed the first time they were circulated. All the messages that reached the highest level of agreement amongst service users were agreed in round 1. One message (44) that reached the highest level of agreement as a key message was not agreed by the health professionals until round 2. The most likely reason for this was that the wording of the message (about the effectiveness and availability of ovarian screening) was changed between rounds 1 and 2 following comments from participants. Messages that failed to reach agreement by either group may not have been relevant to all women or may have been more general in nature. The ease of reaching agreement within and between the groups and the high response and retention rate help validate the findings of this study.

4.7.3. Timing of communication

For key messages where timing of communication was agreed, the preferred option was before genetic testing and once a pathogenic variant has been detected, underlining the importance of repeating key information. The only message where a different timing of communication was agreed by either group was message 5 (the
name of the gene in which a pathogenic variant has been detected). Health professionals agreed that this message should be communicated once a pathogenic variant is identified. The wording of this message may have caused confusion amongst service users who did not reach agreement on the timing of communication.

There is limited research investigating the optimal timing of communication of specific information. This is an area that will become increasingly important as genetic testing becomes more integrated into mainstream medicine. As the survey of the practices of non-genetics healthcare providers in the USA demonstrates, pre-test counselling may not always be considered important outside of clinical genetics (Vadaparampil et al., 2015).

4.8. LIMITATIONS

The information messages circulated in this study were developed inductively from transcripts of post-test genetic counselling consultations with women with breast or ovarian cancer about a newly diagnosed BRCA1/BRCA2 pathogenic variant. The messages were refined through two pilot studies and the Delphi process. To capture information that would only be communicated during pre-test genetic counselling, participants were asked to suggest additional messages. Additional messages were added to the questionnaire when suggested by two or more participants. As a result of this requirement, one message that is not evidence-based was added (message 61) and some information that is recommended by clinical guidelines (National Institute for Health and Care Excellence, 2013) was not added; such as information about the meaning of a diagnostic test result that showed that no variant was detected, the timescale of being given the results and confidentiality of genetic testing.

It was not the intention of this study to develop the wording of messages to be communicated but rather to identify the gist of the messages that were key or not key. However, agreeing the wording of the messages was important to the decision-making of the participants. Actual risk figures were avoided for most of the messages to keep them clear, short and simple. As a result, the messages are generic, not personalised and use non-quantifiable words. However, they do provide a consensus-based wording which may be useful for clinical practice.
The key messages identified in this study are not intended as a didactic list but rather as a guide for health professionals who communicate with women who have breast or ovarian cancer about the information required and when it is needed. This study has not addressed the specific information needs of women undergoing genetic testing who do not have cancer, women specifically undergoing treatment focused genetic testing, women having multi-gene panel testing or the post-test information needs of women who do not have a pathogenic variant in BRCA1/BRCA2. The views of women about how, what and who communicates about treatment focused genetic testing have been investigated in other studies (Gleeson et al., 2013; Meiser et al., 2012).

The information requirements for women with breast or ovarian cancer will inevitably change over time. For example, in the England and Wales the decision to approve the drug Olaparib for women with BRCA1/BRCA2-related ovarian cancer as a third-line treatment in January 2016 (http://www.nice.org.uk/guidance/ta381 accessed 16.02.16) means that women with ovarian cancer will also require this information. The increasing availability of multi-gene panel testing means that some women with breast or ovarian cancer will require additional information about other genes being tested. Further research will be necessary to investigate these information needs once such testing is widely available.

4.9. CONCLUSIONS

This study draws on current practice and expert opinion to provide evidence of the information required by breast or ovarian cancer patients about genetic testing and hereditary cancer management and the optimal timing of communication. Expert health professionals and service users agreed on the key messages required, with seven of the key messages reaching ≥ 95% agreement by both groups. Both groups agreed that key messages should be communicated before genetic testing and once a pathogenic variant has been identified. These findings are helpful for guiding health professionals’ communication with patients prior to genetic testing of the BRCA1/BRCA2 genes and following identification of a pathogenic variant.
CHAPTER 5. STUDY 3: COMMUNICATING KEY MESSAGES ABOUT A PATHOGENIC VARIANT TO BREAST/OVARIAN CANCER PATIENTS - A RETROSPECTIVE CONTENT ANALYSIS

5.1 INTRODUCTION

Breast and ovarian cancer patients who are found to have a pathogenic variant face challenging and complex decisions about cancer treatment, managing the risk of future cancer and informing at-risk relatives. The face-to-face post-test genetic counselling appointment provides an important opportunity for patients to learn about the implications of the result once it is a reality. Helping patients to negotiate their way through the mass of available information and ensuring they are equipped with the messages they need to enable informed decision-making is an important aspect of the post-test genetic counselling consultation.

As genetic testing becomes integrated into mainstream cancer services and new approaches to delivering pre-test information are explored (Benusiglio et al., 2017; Hoberg-Vetti et al., 2016; Quinn et al., 2016; Sie et al., 2016), the post-test genetic counselling consultation is likely to become an increasingly important opportunity for cancer patients to interact with a genetics health professional. To plan the future content of post-test genetic counselling for patients with breast or ovarian cancer and a pathogenic variant, it is helpful to understand what information is communicated and to investigate whether the information communicated is the information patients need.

5.2. BACKGROUND

5.2.1. Communicating medical information

Most patients with cancer express a preference to be kept well informed (Jenkins, Fallowfield, & Saul, 2001) and many express a desire for more information than they receive (McPherson, Higginson, & Hearn, 2001). Satisfaction with communication amongst cancer patients in general (Ong, Visser, Lammes, & de Haes, 2000; Siminoff, Ravdin, Colabianchi, & Sturm, 2000) and about genetic testing in particular (Pieterse, van Dulmen, Beemer, Bensing, & Ausems, 2007) is higher when information is delivered without the patient needing to ask questions.
Given the wealth of information available about genetic testing and hereditary cancer management, it can be challenging for health professionals to decide what to communicate and what to leave out. There may be an assumption that it is better to communicate more rather than less information and that the patient can just ignore what is not useful (Peters, Klein, Kaufman, Meilleur, & Dixon, 2013). However, too much information can overwhelm cognitive abilities resulting in inferior and less well understood choices and low levels of recall (Ley, 1988; McGuire, 1996). Information recall may be compounded by uncertain or emotionally challenging information (Bradshaw, Ley, & Kincey, 1975), ageing (Jansen et al., 2008) or low levels of education (Kessels, 2003). Prioritising the order of presentation of key information (Kessels, 2003; Peters, Dieckmann, Dixon, Hibbard, & Mertz, 2007) and limiting information (Selic, Svab, Repolusk, & Gucek, 2011; Zikmund-Fisher, Angott, & Ubel, 2011) can have a positive impact on recall. By limiting information, highlighting relevant information and avoiding communicating distracting additional information, cognitive workload is reduced, increasing the likelihood of focusing on relevant information (Peters et al., 2013).

For cancer patients, there are particular issues that can impact on recall. For a sub-set of patients with cancer, there is evidence of short, medium and long-term cognitive impairment due to adjuvant chemotherapy (Ahles, Root, & Ryan, 2012; Asher & Myers, 2015) which may impact on information processing. Those with a poor prognosis may recall less than those with a better prognosis (Jansen et al., 2008). Study 1 (Chapter 3) found that accuracy of recall of information amongst at-risk women with a newly identified BRCA1 or BRCA2 pathogenic variant and their relatives was low (Jacobs, Dancyger, Smith, & Michie, 2015). Recall does not necessarily equate with understanding (Braithwaite, Emery, Walter, Prevost, & Sutton, 2006). However, to begin to facilitate understanding and recall, it is essential that the information communicated is the information that patients need.

5.2.2. Information needs of breast/ovarian cancer patients at post-test genetic counselling once a pathogenic variant has been identified

There is limited published evidence about the information needed by women with breast or ovarian cancer at post-test genetic counselling, although general guidelines do exist. A one-day workshop involving 12 health professionals specialising in aspects of genetics from across Europe produced guidelines for genetic counselling practice across a range of conditions. The guidelines mainly focus on pre-test genetic counselling. However, it is recommended that post-test counselling should include...
‘appropriate disclosure of results, as previously agreed with patient, discussion of plan for future contact, re-iteration of information on support groups, surveillance, treatment, referrals to other practitioners or services and, where appropriate, participation in research projects, the availability of follow-up and support and ... some of the topics covered in the pre-test session... for example, the implications for the patient and relatives and discussion of the psychological impact of changed genetic status.’ (Skirton, Goldsmith, Jackson, & Tibben, 2013, p.259).

The National Society for Genetic Counsellors in the USA have published consensus guidelines about what should be addressed during genetic counselling about all types of hereditary cancer (Riley et al., 2012; Trepanier et al., 2004). According to the guidelines, the post-test consultation should include disclosure of the genetic test result without interpretation, explanation of the specificity, sensitivity and limitations of the specific genetic test performed, the provision of cancer risk re-assessment and medical management guidelines and recommendations including surveillance and risk-reducing options for the patient and family, referral to appropriate health care providers, identification of at-risk family members and the provision of tools to inform and educate family members (Riley et al., 2012).

UK clinical practice guidelines address the management of women with breast cancer who are found to have a BRCA1/BRCA2 pathogenic variant (National Institute for Health and Care Excellence, 2013). The guidelines focus on recommendations for management and do not specify the information to be communicated at pre- or post-test genetic counselling.

5.2.3. Extent to which the information needs of breast/ovarian cancer patients with a pathogenic variant are met during post-test genetic counselling

There are a small number of studies about the extent to which the information needs of women with cancer are met by genetic counselling. These studies are discussed in Chapter 2. A further study investigated the extent to which support needs of 124 women with early onset breast or ovarian cancer and a BRCA1/BRCA2 pathogenic variant were met. The participants were surveyed prior to and after pre- and post-test genetic counselling using a measure created for women at high risk of breast cancer. Only 9% of participants reported that their needs were met. The most frequently reported unmet needs were dealing with uncertainty about the future, fears about developing cancer and dealing with the impact of a pathogenic variant on the family (Farrelly et al., 2013).
No previous studies, to the author’s knowledge, have investigated the content of the post-test genetic counselling consultation with cancer patients at which a pathogenic variant is first disclosed or discussed or the extent to which the information judged to be required is communicated by health professionals. To enable patients to make the best possible use of post-test genetic counselling in the future, it is important to learn from current practice.

5.3. AIMS AND RESEARCH QUESTIONS

This study aimed to evaluate the extent to which the key messages identified by experts in the field in Study 3 (Chapter 4) were communicated to breast/ovarian cancer patients during post-test genetic counselling about a newly identified BRCA1/BRCA2 pathogenic variant.

The study addressed the following research questions:

1) What proportion of the distinct messages communicated by genetics health professionals were (i) key and supplementary; and (ii) about genetic testing and hereditary cancer management?

2) Is there a difference in the proportion of messages judged as key by expert health professionals that were communicated about (a) genetic testing and (b) hereditary cancer management?

3) Which key messages about (a) genetic testing and (b) hereditary cancer management were frequently and infrequently communicated?

5.4. METHOD

5.4.1. Design

The study design was a retrospective content analysis of anonymised transcripts of post-test genetic counselling consultations.
5.4.2. Sample

The study sample consisted of transcripts of genetic counselling consultations between 37 breast and/or ovarian cancer patients and 14 genetics health professionals, including clinical geneticists and genetic counsellors.

Data were collected between 2006 and 2008 in one of two Regional Genetics Centres in the UK as part of a The Family Communication Study outlined in section 1.8.5.5. (Dancyger, Smith, Jacobs, Wallace, & Michie, 2010; Dancyger et al., 2011; Jacobs et al., 2015).

Thirty of the patients (81%) had breast cancer only, three (8%) had ovarian or fallopian tube cancer only and four (11%) had breast and ovarian cancer. Nineteen (51%) patients had a BRCA1 pathogenic variant and 18 (49%) had a BRCA2 pathogenic variant. At the time of the consultation, 33 patients (89%) had remaining breast tissue and 27 (73%) had ovaries and fallopian tubes in situ (see Table 5.1). Each patient had undergone pre-test genetic counselling before the blood sample was taken. However, as these data are anonymised, the time between pre-test genetic counselling and the genetic test result is not known.

The health professionals were working in one of two Regional Genetics Centres in the UK. As these data are anonymised, the type of genetics health professional providing the genetic counselling is not known.

5.4.3. Ethics

NHS Research Ethics Committee approval was obtained for the original study for which these data were collected (see section 1.8.5.5.). Further analysis of the pre-existing, completely anonymous, non-sensitive data for this study did not require ethics approval as indicated by the UCL Research Ethics Office document.
Table 5.1. Study participants

<table>
<thead>
<tr>
<th>Health professional no.</th>
<th>Patient /Transcript no.</th>
<th>Type of cancer</th>
<th>Gene involved</th>
<th>Breast tissue remaining</th>
<th>Ovarian/tubal tissue remaining</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>01 FTC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td></td>
<td>05 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>06 BC &amp; OC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td></td>
<td>09 BC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>28 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>03 BC</td>
<td>BRCA2</td>
<td>No</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>10 BC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>18 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>19 BC &amp; OC</td>
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<td>No</td>
<td></td>
</tr>
<tr>
<td></td>
<td>20 OC</td>
<td>BRCA1</td>
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<td>No</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>21 BC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>34 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>07 OC</td>
<td>BRCA2</td>
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<td></td>
</tr>
<tr>
<td></td>
<td>11 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>17 BC &amp; OC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td></td>
<td>22 BC</td>
<td>BRCA2</td>
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<td>No</td>
<td></td>
</tr>
<tr>
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<td>26 BC</td>
<td>BRCA1</td>
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<td>Yes</td>
<td></td>
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<tr>
<td></td>
<td>31 BC</td>
<td>BRCA1</td>
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<td>Yes</td>
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<tr>
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<td>BRCA2</td>
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<td></td>
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<tr>
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<td>BRCA1</td>
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<td>Yes</td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>04 BC &amp; OC</td>
<td>BRCA1</td>
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<td>No</td>
<td></td>
</tr>
<tr>
<td></td>
<td>13 BC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
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<tr>
<td></td>
<td>33 BC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td>7</td>
<td>02 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>14 BC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>23 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td></td>
<td>27 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>30 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>36 BC</td>
<td>BRCA1</td>
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<td>Yes</td>
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<tr>
<td>8</td>
<td>16 BC</td>
<td>BRCA1</td>
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<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>24 BC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td>9</td>
<td>25 BC</td>
<td>BRCA2</td>
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<tr>
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<td>29 BC</td>
<td>BRCA1</td>
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<td></td>
</tr>
<tr>
<td>11</td>
<td>12 BC</td>
<td>BRCA2</td>
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<td>Yes</td>
<td></td>
</tr>
<tr>
<td>12</td>
<td>15 BC</td>
<td>BRCA2</td>
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<td>Yes</td>
<td></td>
</tr>
<tr>
<td>13</td>
<td>08 BC</td>
<td>BRCA1</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td>14</td>
<td>37 BC</td>
<td>BRCA2</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
</tbody>
</table>

FTC fallopian tube cancer, BC breast cancer, OC ovarian cancer
5.4.4. Procedure

5.4.4.1. Developing the coding scheme

A coding scheme was developed from the list of key messages identified in Study 2. Codes were also included to identify supplementary messages. The term ‘supplementary messages’ describes the messages that were not judged to be key by the health professionals in Study 2. The term supplementary message was further defined during the process of this study as information volunteered by the health professional that is not judged to be a key message or communicated in response to a patient’s question or specific individual circumstances.

The coding scheme is shown in Appendix 3.1. The key messages were organised into messages about genetic testing and messages about hereditary cancer management.

Using the coding scheme, the key and supplementary messages communicated in each transcript were identified. A message was coded as key if it was judged as key by the expert health professionals in Study 2, with two exceptions: (I) one message was not key for the sample as it was only relevant prior to genetic testing; (II) three messages depended on the type of cancer the patient was diagnosed with and whether or not they had remaining breast or ovarian tissue and so were coded as key in some transcripts and supplementary in other transcripts.

Transcripts were systematically searched for reference to the key messages and any supplementary messages communicated and coded against the coding scheme. These messages were grouped into key and supplementary messages. The key and supplementary messages were grouped into messages about (a) genetic testing and (b) hereditary cancer management (see Table 5.2.).
<table>
<thead>
<tr>
<th>Type of message</th>
<th>Issues discussed</th>
<th>Example quotes of supplementary messages</th>
</tr>
</thead>
<tbody>
<tr>
<td>DNA, genes, chromosomes, inheritance and the genetic test result</td>
<td>• Inheritance</td>
<td>• ‘That’s what’s known as a frame shift, you know, shifts the way the body reads the code.’ (HP7)</td>
</tr>
<tr>
<td></td>
<td>• DNA, genes and chromosomes</td>
<td>• ‘Your genetic code is, is in every cell of the body, so in every cell that makes the gene product from <em>BRCA2</em>, this error will be [present]. (In) a sample of your blood … we were looking at cells that were representative of your whole body.’ (HP10)</td>
</tr>
<tr>
<td></td>
<td>• How rare it is to have a mutation</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Presence of a normal allele</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Role &amp; function of the <em>BRCA1</em>/<em>BRCA2</em> gene and differences</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• What research has shown and what is still to be learnt</td>
<td></td>
</tr>
<tr>
<td>Process and implications of genetic testing for the family</td>
<td>• Timing of predictive testing</td>
<td>• ‘Testing people in their early 20’s, at the moment, may not be that great an idea.’ (HP9)</td>
</tr>
<tr>
<td></td>
<td>• Reproductive choices</td>
<td>• First of all you need to harvest the eggs, then you need to choose the embryos, then you need to implant the embryo and hope … that the embryo will stay in the womb and grow.’ (HP1)</td>
</tr>
<tr>
<td></td>
<td>• Benefits and difficulties of predictive testing</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• What information the test does and does not provide</td>
<td></td>
</tr>
<tr>
<td>Follow up and support for woman with cancer</td>
<td>• Practical issues around the genetic test result</td>
<td>• ‘Sometimes people find it difficult … if we find that they carry a gene, and then their children are tested, and they carry it as well…But … you have no control over which egg’s fertilised.’ (HP5)</td>
</tr>
<tr>
<td></td>
<td>• Emotional issues around the genetic test result</td>
<td>• ‘It’s not your fault…we all carry genetic faults, ….it doesn’t matter if we pass on to our children, because some don’t express unless the other copy is also faulty.’ (HP8)</td>
</tr>
<tr>
<td></td>
<td>• Genetics follow up and next steps</td>
<td></td>
</tr>
<tr>
<td>Cancer development, diagnosis, treatment and outcomes</td>
<td>Biology of BRCA-related tumours</td>
<td>‘Sometimes a part of that tumour can break off and travel in your blood stream or your lymphatic system and go somewhere else... to the liver or ... to the spine.’ (HP13)</td>
</tr>
<tr>
<td>---</td>
<td>---</td>
<td>---</td>
</tr>
<tr>
<td>Cancer risks for women and men</td>
<td>Population risks of breast/ovarian cancer</td>
<td>‘Ovarian cancer doesn’t occur in the 20s, or if it does, it’s a different type of ovarian cancer, not one that’s associated with this gene mistake.’ (HP4)</td>
</tr>
<tr>
<td>Cancer awareness,</td>
<td>Lifetime and age-related risk for women</td>
<td>‘Your genetic risk ...becomes higher as you go into your 40s and then that plateaus ...but your population risk of getting breast cancer...increases as you get older.’ (HP2)</td>
</tr>
<tr>
<td>Risks/limitations of breast surveillance</td>
<td>Breast cancer risks for men</td>
<td>‘Prostate cancer is highly unusual before the age of 60.’ (HP 12)</td>
</tr>
<tr>
<td>Effectiveness of breast surveillance</td>
<td>Prostate cancer risks/age of onset</td>
<td>---</td>
</tr>
<tr>
<td>Availability/process of breast surveillance for women</td>
<td>Health risks that do not affect men</td>
<td>---</td>
</tr>
<tr>
<td>Availability/lack of availability of surveillance for men</td>
<td>Factors that affect risk</td>
<td>---</td>
</tr>
<tr>
<td>Non-surgical risk-reducing measures</td>
<td>Risk management options</td>
<td>‘They can see through the dense tissue with that technique (MRI), a lot better than they can with the x-rays that are used in mammography.’ (HP3)</td>
</tr>
<tr>
<td>Timing of risk-reducing surgery</td>
<td>Cancer awareness,</td>
<td>‘But if you've never had anything removed down there, and you're not overweight, ... then it's a straightforward operation, and it can be done with keyhole surgery.’ (HP14)</td>
</tr>
<tr>
<td>Benefits/difficulties of surgery</td>
<td>Risk management options</td>
<td>---</td>
</tr>
<tr>
<td>Decision-making</td>
<td>Surgical procedures</td>
<td>---</td>
</tr>
<tr>
<td>Psychological impact of surgery</td>
<td>---</td>
<td>---</td>
</tr>
</tbody>
</table>
5.4.4.2. Inter-rater reliability

Two researchers (CJ and JH) undertook independent coding of the transcripts, revising the coding method until the coding method was able to produce results with good inter-rater reliability. The inter-rater reliability of the extent to which the independent coders identified the same messages was assessed using percentage agreement and Cohen’s kappa co-efficient (Cohen, 1968).

Percentage agreement alone is usually not an acceptable measure of inter-rater reliability as agreement can be over-estimated in certain circumstances (Hallgren, 2012). Cohen’s kappa aims to overcome biases by adjusting for the possibility of agreement occurring by chance but has been shown to under-estimate the level of agreement (Byrt, Bishop, & Carlin; Hoehler, 2000). The prevalence and bias adjusted kappa (PABAK) statistic adjusts for coding bias and takes into account a high prevalence of negative agreement. Several authors advocate this over Cohen’s kappa (Abraham et al., 2015; Wood et al., 2015), although others have suggested that PABAK may result in misleading conclusions (Chen, Faris, Hemmelgarn, Walker, & Quan, 2009; Hoehler, 2000). It has been suggested that for healthcare studies the Cohen’s kappa and percentage agreement should be reported (McHugh, 2012). This approach was therefore used for this study.

Independent coding of the first two transcripts (T01 and T02) showed 80.1% agreement about whether the messages were communicated (kappa 0.615, SE 0.064, p <.000). This process highlighted differences between coders. This was resolved by agreeing that it was the main gist of the message rather than the actual wording of the message on the coding scheme that was relevant for coding. After the first round of coding, the definition of a supplementary message was amended to exclude information communicated in response to individual questions or particular circumstances. The coding guidelines were changed to reflect this. As the inter-rater reliability for round one was below 0.7, which is the conventionally regarded acceptable level of agreement (Nunnally, 1978), four further transcripts were independently coded. An agreement level of 88.5% (kappa 0.757, SE 0.42 p <0.0) was reached and deemed an acceptable level of agreement for this study. The remainder of the data set were coded by CJ only. Table 5.3 shows the order of coding the transcripts and by whom.
Table 5.3. Order of coding patients’ transcripts and initials of researcher undertaking the coding

<table>
<thead>
<tr>
<th>Patient/Transcript no.</th>
<th>Order of coding</th>
<th>Coded by</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Pilot coding</td>
<td>CJ and JH</td>
</tr>
<tr>
<td>2</td>
<td></td>
<td>CJ and JH</td>
</tr>
<tr>
<td>4</td>
<td>Independently coded following discussion of pilot coding</td>
<td>CJ and JH</td>
</tr>
<tr>
<td>5</td>
<td></td>
<td>CJ and JH</td>
</tr>
<tr>
<td>9</td>
<td></td>
<td>CJ and JH</td>
</tr>
<tr>
<td>11</td>
<td></td>
<td>CJ and JH</td>
</tr>
<tr>
<td>3, 6, 7, 8, 10 and 12-33</td>
<td>Coded once reliable coding method agreed</td>
<td>CJ only</td>
</tr>
</tbody>
</table>

5.4.4.3. Analysis

The frequency of messages communicated according to category were analysed using Content Analysis (Silverman, 2006).

All statistical analyses were conducted in SPSS version 23. First, the proportion (n) of messages which were key and supplementary, and classed as communicating genetic testing and hereditary cancer management, were calculated for each transcript. These were then used to determine the mean (SD) proportion for each health professional.

The assumption of normality was violated and so a priori hypotheses concerning differences in the percentage of supplementary messages communicated about genetic testing and hereditary cancer, and the communication of key messages about genetic testing and hereditary cancer were tested using the Wilcoxon signed-rank test. Alpha was set to 0.05, 2-tailed.
5.5. RESULTS

5.5.1. Proportion of the distinct messages communicated by genetics health professionals that were (i) key and supplementary and (ii) about genetic testing and hereditary cancer management

(i) On average, 57.02% \((SD \ 8.28)\) of the distinct messages communicated by each health professional were key messages and 42.98% \((SD \ 8.28)\) were supplementary messages (see Table 5.4). A Wilcoxon signed-rank test showed that genetics health professionals communicated significantly more key messages than supplementary messages \((Z = 13.50, \ p = 0.014)\).

(ii) On average, 47.86% \((SD \ 9.80)\) of the distinct messages communicated by each health professional were about genetic testing and 52.14% \((SD \ 9.80)\) were about hereditary cancer management (see Table 5.4). A Wilcoxon signed-rank test showed no significant difference between the mean percentage of distinct messages communicated about genetic testing and hereditary cancer \((Z = 60.00, \ p = 0.311)\).

Table 5.4. Communicated distinct messages per health professional: mean percentage \((n)\)

<table>
<thead>
<tr>
<th>Type of information</th>
<th>Key messages</th>
<th>Supplementary messages</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic testing</td>
<td>51.58 (305)</td>
<td>41.00 (226)</td>
<td>47.86 (531)</td>
</tr>
<tr>
<td>Hereditary cancer management</td>
<td>48.69 (307)</td>
<td>59.00 (264)</td>
<td>52.14 (571)</td>
</tr>
<tr>
<td>Total</td>
<td>57.02 (612)</td>
<td>42.98 (490)</td>
<td></td>
</tr>
</tbody>
</table>

5.5.2. Proportion of messages judged as key by expert health professionals that were communicated about (a) genetic testing and (b) hereditary cancer management

On average, health professionals communicated 49.93% \((SD \ 10.06)\) of the messages judged as key by experts in the field. Of these key messages a mean of 61.93% \((SD \ 17.79)\) were about genetic testing and 41.73% \((SD \ 11.10)\) were about hereditary cancer management (see Table 5.5). A Wilcoxon signed-rank test showed that genetics health professionals communicated significantly more key messages about genetic testing than about hereditary cancer management \((Z = 60.50, \ p = 0.004)\).
Table 5.5. Key messages communicated/not communicated: percentage (n)

<table>
<thead>
<tr>
<th>Type of information</th>
<th>Communicated</th>
<th>Not communicated</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic testing</td>
<td>61.93 (305)</td>
<td>38.07 (176)</td>
</tr>
<tr>
<td>Hereditary cancer</td>
<td>41.73 (307)</td>
<td>58.27 (395)</td>
</tr>
<tr>
<td>management</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>49.93 (612)</td>
<td>50.08 (571)</td>
</tr>
</tbody>
</table>

5.5.3. Key messages about (a) genetic testing and (b) hereditary cancer management that were frequently and infrequently communicated

Five of the key messages about genetic testing were communicated by over 79% of the health professionals in this study and were the most frequently communicated of all the key messages. Among these five messages were the three key messages about genetic testing that reached ≥95% agreement amongst the health professionals in Study 2: ‘the decision to have a genetic test is up to each individual’, ‘the availability of predictive testing and what it will show’ and ‘autosomal dominant inheritance’.

Three of the key messages about genetic testing: ‘risk of cancer for non-carriers’, ‘cancer is not inevitable for carriers’ and ‘a pathogenic variant does not skip a generation’ were communicated in just over a third of the consultations. The least frequently communicated key message about genetic testing was about the benefit of the test to women with cancer. Table 5.6 shows the mean percentage of times each key message about genetic testing was communicated across all health professionals (full messages are shown in Table 4.3, Chapter 4).
Three of the key messages about hereditary cancer management that reached ≥ 95% agreement amongst the health professionals in Study 2 were communicated by over 55% of the health professionals in this study: ‘the risk of breast cancer’, ‘the risk of ovarian cancer’ and ‘the option of risk-reducing mastectomy’. However, two of the key messages that reached ≥95% agreement in Study 2 were communicated in less than half of the consultations in this study: ‘the option of risk-reducing salpingo-oophorectomy’ and ‘the limitations of ovarian screening’.

Key messages about the benefits and limitations of breast screening and surgery were communicated in less than half of the consultations. The key message about the risk of metastases associated with the initial cancer after risk-reducing mastectomy was the least frequently communicated key message. Table 5.7 shows the mean percentage of times each key message about hereditary cancer was communicated across all health professionals.
Table 5.7. Key messages about hereditary cancer management

<table>
<thead>
<tr>
<th>Message no.</th>
<th>Key message * Messages agreed as key by &gt;95% expert health professionals in Study 2</th>
<th>Mean %</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td>16</td>
<td>Risk 2\textsuperscript{nd} primary BC</td>
<td>75.17</td>
<td>38.88</td>
</tr>
<tr>
<td>41</td>
<td>Breast screening available</td>
<td>68.57</td>
<td>37.55</td>
</tr>
<tr>
<td>26</td>
<td>Increased risk BC (at-risk women) *</td>
<td>66.73</td>
<td>35.27</td>
</tr>
<tr>
<td>20</td>
<td>Risk OC for BC patients</td>
<td>62.24</td>
<td>44.60</td>
</tr>
<tr>
<td>30</td>
<td>Increased risk of PC</td>
<td>61.67</td>
<td>48.12</td>
</tr>
<tr>
<td>28</td>
<td>Increased risk OC (at-risk women)*</td>
<td>57.50</td>
<td>42.78</td>
</tr>
<tr>
<td>36</td>
<td>RRM available *</td>
<td>55.48</td>
<td>44.52</td>
</tr>
<tr>
<td>17</td>
<td>Risk BC for OC patients</td>
<td>50.00</td>
<td>50.00</td>
</tr>
<tr>
<td>44</td>
<td>No OC screening *</td>
<td>48.63</td>
<td>41.17</td>
</tr>
<tr>
<td>45</td>
<td>BSO available *</td>
<td>48.04</td>
<td>40.23</td>
</tr>
<tr>
<td>39</td>
<td>RRM reduces the risk of BC</td>
<td>40.30</td>
<td>39.81</td>
</tr>
<tr>
<td>46</td>
<td>BSO reduces OC/FTC risk</td>
<td>38.87</td>
<td>38.31</td>
</tr>
<tr>
<td>47</td>
<td>\textit{BSO may reduce BC risk}</td>
<td>38.87</td>
<td>38.31</td>
</tr>
<tr>
<td>27</td>
<td>Age of BC risk</td>
<td>31.37</td>
<td>36.38</td>
</tr>
<tr>
<td>48</td>
<td>BSO will result in menopause.</td>
<td>27.08</td>
<td>43.26</td>
</tr>
<tr>
<td>37</td>
<td>Breast reconstruction option</td>
<td>26.43</td>
<td>41.15</td>
</tr>
<tr>
<td>42</td>
<td>Breast screening limitations</td>
<td>24.94</td>
<td>35.70</td>
</tr>
<tr>
<td>29</td>
<td>Age of OC risk</td>
<td>16.49</td>
<td>30.77</td>
</tr>
<tr>
<td>10</td>
<td>Breast screen at 50% risk</td>
<td>13.04</td>
<td>30.29</td>
</tr>
<tr>
<td>62</td>
<td>Risk from initial cancer CM</td>
<td>0.89</td>
<td>3.34</td>
</tr>
</tbody>
</table>

Abbreviations: BC (breast cancer), OC (ovarian cancer), FTC (fallopian tube cancer), PC (prostate cancer), RRM (Risk-Reducing Mastectomy), BSO (Bilateral salpingo-oophorectomy), CM (contralateral mastectomy)

5.6. DISCUSSION

The genetics health professionals communicated significantly more key messages than supplementary messages. However, a third of the messages communicated were supplementary and therefore not required for decision-making according to the expert health professionals in Study 2. There was no difference between the number of distinct key and supplementary messages communicated about genetic testing and hereditary cancer. Overall, only half of the key messages judged by expert health professionals as required by breast/ovarian cancer patients were communicated. Of the key messages that were communicated, significantly more were about genetic testing than hereditary cancer management. The key messages about genetic testing for relatives were frequently communicated. The key messages about the risks and limitations of breast and ovarian cancer screening and risk-reducing surgery were infrequently communicated.
Most of the messages communicated were key, suggesting that much of the information communicated was required by the patients for decision-making about their own management and to inform at-risk relatives. This finding is reassuring as communicating information to facilitate informed decision-making is one of the fundamental goals of genetic counselling (Meiser, Irle, Lobb, & Barlow-Stewart, 2008). This concurs with the findings of studies that overall genetic health professionals are good at communicating information about BRCA1 and BRCA2 (Butow & Lobb, 2004; Butow, Lobb, Meiser, Barratt, & Tucker, 2003).

A third of the messages communicated were supplementary. Some of the supplementary messages may have been important for individual patients. The decision about what to communicate is affected by the characteristics of the patient (Ellington et al., 2005; Lobb et al., 2002) and the counselling skills and communication style of the health professional (Braithwaite et al., 2006; Ellington et al., 2005; Lobb, Butow, Barratt, Meiser, & Tucker, 2005; Michie, Bron, Bobrow, & Marteau, 1997). As all the women in the study had already undergone pre-test genetic counselling several months or years before the post-test genetic counselling consultation, the decision to communicate some of the supplementary messages may have been based on prior knowledge of individual’s circumstances or the perceived need for more detailed information.

Health professionals may have communicated the supplementary messages because they felt the need to offer reassurance and hope or that they should communicate as much information as possible. However, two seminal studies of clinician-patient communication found that too much information can result in inferior, less well-understood choices and low levels of recall (Ley, 1988; McGuire, 1996), suggesting that communication of supplementary messages may not only reduce the time for communicating key messages but may also be detrimental to decision-making.

There was no difference between the proportion of distinct key and supplementary messages communicated about genetic testing and hereditary cancer. According to the findings of Study 2, a greater proportion of key messages are required about hereditary cancer management than genetic testing. Other studies have also found that the most frequently unmet need amongst women with breast cancer following genetic counselling is for information about screening and risk-reducing surgery (Culver et al., 2011; Metcalfe et al., 2000).
Only half of the key messages required by cancer patients in this study were communicated. This finding is in line with studies that have shown that health professionals do not always communicate information that cancer patients need or desire at pre- and post-test genetic counselling (Farrelly et al., 2013; Lobb et al., 2004; Metcalfe et al., 2000). Similarly, studies of communication between oncologists and cancer patients about phase 1 clinical trials have found that health professionals do not always communicate the information that patients require (Jenkins et al., 2010; Tomamichel et al., 1995). Possible explanations for the non-communication of information by health professionals are lack of awareness of the information needed, lack of time in the consultation or the well-intentioned concern not to cause distress (Gaston & Mitchell, 2005).

The key messages about hereditary cancer management were less frequently communicated than those about genetic testing. Guidelines about the management of hereditary risk for patients with breast or ovarian cancer have only recently become available in the UK (National Institute for Health and Care Excellence, 2013) and in Australia (https://guidelines.canceraustralia.gov.au/guidelines/guideline_17.pdf). However, although there were no such guidelines at the time of data collection, the evidence underpinning the key messages was already available in the literature (Antoniou et al., 2003; Breast Cancer Linkage Consortium, 1999; Ford et al., 1998; Hartmann, 1999; Hartmann et al., 2001; Kauff et al., 2002; Kirchhoff et al., 2004; Lostumbo, Carbine, Wallace, & Ezzo, 2004; Meijers-Heijboer et al., 2001; Olivier et al., 2004; Rebbeck et al., 2004; Rebbeck et al., 1999; Rebbeck et al., 2002). This finding may therefore be due, at least in part, to the difficulty for health professionals of accessing, interpreting and filtering research findings about hereditary breast and ovarian cancer in the absence of clinical guidelines.

The lower proportion of key messages communicated about hereditary cancer management than genetic testing suggests that genetics health professionals may have been more familiar with genetic testing information and/or less aware of, or less comfortable with, the information about hereditary cancer management. An explanation of Mendelian inheritance is often central to any genetic counselling consultation (Gale, Pasalodos-Sanchez, Kerzin-Storrar, Hall, & MacLeod, 2010) and discussing the implications of a pathogenic variant for the family is an important aspect of the post-test consultation (Jacobs, Haque & Scott, 2014; Skirton et al., 2013).
The key messages about genetic testing for relatives were communicated by over 79% of the health professionals. The frequency of communication of the messages about inheritance, predictive testing and discussion before testing is consistent with the high level of agreement reached about these messages in Study 2. Several studies have found that women with cancer are mainly motivated to undergo genetic testing for the benefit of the family (Brandt, Hartmann, Ali, Tucci, & Gilman, 2002; Julian-Reynier et al., 1998; Van Asperen et al., 2002), suggesting that women desire this information. However, although the guidelines that refer to post-test genetic counselling refer to the implications for relatives (Skirton et al., 2013), guidelines do not specify what information should be communicated about genetic testing for relatives.

The least frequently communicated key messages about genetic testing: ‘the pathogenic variant does not skip a generation’, ‘the risk of cancer for non-carriers’ and ‘the benefit of testing for affected women’ may be more relevant to communicate to patients with cancer at pre-test genetic counselling.

The key messages about cancer risk, breast screening and risk-reducing mastectomy were communicated by 55% to 75% of the health professionals in this study. Although these messages were the most frequently communicated of the key messages about hereditary cancer management, they were not communicated as frequently as the key messages about genetic testing for relatives. This suggests that genetics health professionals in this study were more focused on the genetic testing aspects of the communication than the hereditary cancer management aspects.

The messages about breast and ovarian cancer risk and risk-reducing mastectomy were among the messages that reached ≥ 95% agreement amongst the health professionals in Study 2. These messages were communicated by most of the health professionals in this study. Two key messages about ovarian cancer risk management also reached ≥ 95% agreement amongst the health professionals in Study 2 but were infrequently communicated in this study. The health professionals in this study may not have considered the messages to be key or they may have felt uncomfortable about raising the issue of further possible cancer risk with women who have already had breast cancer. Feelings of worry about causing distress to patients or lack of skills and knowledge amongst health professionals have been identified as barriers to clinician-patient communication (Baile, Lenzi, Parker, Buckman, & Cohen, 2002; Quinn et al., 2009).
Key messages about the risks and limitations of screening and surgery were communicated by less than half of the health professionals in this study. The message about the continued risk of metastases from the initial cancer following risk-reducing mastectomy was the least frequently communicated of all the key messages. It is possible that the health professionals considered the messages to be unnecessary to communicate at the same time as disclosing the result for fear of overwhelming the patients with information not needed until a later stage (Culver et al., 2011). Alternatively, health professionals may have lacked the knowledge or expertise to discuss these issues or assumed that health professionals specialising in oncology or surgery would communicate the information at another time.

5.7. CLINICAL IMPLICATIONS

As genetic testing becomes further integrated into mainstream cancer services, it is likely that the post-test genetic counselling consultation will be the only opportunity for women with cancer to interact with a genetics health professional. Fewer individual appointments and shorter appointment times are likely to make it increasingly difficult for genetics health professionals to provide the information, support and counselling needed by cancer patients once a BRCA1/BRCA2 variant is detected. It is therefore imperative that the information communicated during post-test genetic counselling is focused on the information needed, freeing up more time for the counselling and supportive aspects of the consultation.

The findings of this study suggest that post-test genetic counselling does not always provide cancer patients with all the information they need, particularly about the risks and limitations of screening and risk-reducing surgery. Health professionals providing post-test genetic counselling for cancer patients need to be aware of the key messages required and may benefit from training in this area.

Guidelines about what information to communicate and when, would help to inform clinical practice in genetics, oncology and cancer surgery, as well as patient support and information services. Written and online patient information that reiterate the key messages may be helpful in consolidating the messages communicated.

Health professionals that provide post-test genetic counselling to breast and ovarian cancer patients should ensure that key messages about hereditary cancer
management as well as genetic testing are communicated, and that supplementary messages are only communicated when they are particularly relevant to the individual and there is the time to do so.

5.8. LIMITATIONS

Understanding about the genetics and management of hereditary cancer has grown enormously between the time of data collection for this study and identification of the key messages in Study 2. However, the evidence-base for the key messages was already available in the literature at the time of data collection. For one message about breast cancer risk reduction associated with pre-menopausal risk-reducing bilateral salpingo-oophorectomy, new evidence has emerged since Study 2. There is currently debate in the literature about whether early studies that estimated a 50% reduction in breast cancer risk among women who had a risk-reducing salpingo-oophorectomy may have been subject to bias (Chai, Domchek, Kauff, Rebbeck, & Chen, 2015; Heemskerk-Gerritsen, Seynaeve et al., 2015). Emerging evidence suggests that premenopausal bilateral salpingo-oophorectomy reduces the risk of primary breast cancer in women with a BRCA2 pathogenic variant who have not previously been affected with breast cancer risk (Kotsopoulos et al., 2017). However, recent data suggest that there is no reduction of breast cancer risk as a result of surgery for at-risk women with a BRCA1 variant or cancer patients with a BRCA1 or BRCA2 pathogenic variant who develop contralateral breast cancer (Basu et al., 2015). This message was considered to be true (Kauff et al., 2002) for patients and at-risk relatives with a BRCA1 and BRCA2 pathogenic variant at the time of data collection. Therefore, to avoid biasing the study, no adjustment was made for the recently published evidence.

The lack of available clinical practice guidelines for breast and ovarian cancer patients at the time of data collection meant that awareness of the evidence was dependent on the time, ability and willingness of the individual health professional to be up to date and their understanding and interpretation of the evidence.

The sample size was small and that there was variation in the number of patients seen by each health professional which was accounted for in the analysis. However, the data were collected across two different regional genetics services and were firmly grounded in clinical practice with the key messages that formed the coding scheme having been developed from analysis of transcripts with the patient group and identified
by experts in the field. Other studies which have evaluated the extent to which information needs were met (Farrelly et al., 2013; Lobb et al., 2003) or knowledge increased (Scherr, Christie, & Vadaparampil, 2016) have either adapted tools developed for a different purpose or applied tools which have been developed to analyse different aspects of communication. To the author’s knowledge this is the first study to examine post-test genetic counselling with cancer patients in detail.

This study was conducted in the UK with cancer patients undergoing genetic testing after completing cancer treatment. The findings may not be therefore be generalisable to other countries or other patient groups.

5.9. FUTURE RESEARCH

Further research is required to investigate the recommendations made by existing clinical guidelines about the information that should be communicated to patients with breast or ovarian cancer at pre- and post-test genetic counselling and the extent to which those recommendations are translated into clinical practice.

Genetic testing is increasingly required immediately after diagnosis to guide treatment. Research will be needed to identify the key messages required by women with breast or ovarian cancer who undergo genetic testing immediately after cancer diagnosis and those being tested with multiple gene panels. In addition, the information communicated to women with cancer by health professionals specialising in oncology and cancer surgery will require investigation.

As non-genetics health professionals become increasingly involved in communicating with patients undergoing genetic testing, further research will be necessary to evaluate the effectiveness of this communication. The methods applied in this programme of research may be helpful in investigating the key messages required and communicated as genetic testing becomes increasingly integrated into mainstream medicine.
5.10. CONCLUSIONS

This study provides a baseline for understanding the information communicated to patients with breast or ovarian cancer at post-test genetic counselling following identification of a BRCA1/BRCA2 pathogenic variant. Although the genetics health professionals communicated significantly more key than supplementary messages, only half of the messages judged as key by expert health professionals were communicated. Of these, significantly more of the key messages were about genetic testing than hereditary cancer management. The most frequently communicated key messages were about genetic testing for relatives and the least frequently communicated key messages were about the risks and limitations of screening and risk-reducing surgery. As a result, patients did not receive all the information they required in order to make decisions about managing their risk of future cancers.
6.1. INTRODUCTION

Traditionally, genetics health professionals have provided genetic counselling to individuals with cancer once treatment is complete. For women with a personal history of breast or ovarian cancer, genetic testing following cancer treatment is often motivated by the wish to help family members (Hallowell et al., 2002; Julian-Reynier et al., 1998). In the last five years, there has been a growing increase in the number of women referred for genetic testing with newly diagnosed breast or ovarian cancer or metastatic relapse. For these women, the motivation for testing is often to make decisions about whether to have a unilateral or bilateral mastectomy or to access chemotherapy (Meiser, Gleeson, Watts, et al., 2012). Unlike other tests involved in cancer diagnosis, genetic tests have the potential to have an impact on the wider family as well as on the individual with cancer. Those who undergo genetic testing therefore need sufficient information to provide informed consent and facilitate decision-making, raising challenges for genetics and oncology health professionals about what should be communicated to women with cancer (patients) about genetic testing and hereditary cancer management and how this communication should take place.

6.2. BACKGROUND

Counselling patients about genetics shortly after diagnosis is challenging for both genetics and oncology health professionals. The rapidly evolving field of genetics and genomics will increasingly require health professionals to rely on clinical guidelines to synthesise research findings and guide practice (Schully et al., 2015).

Existing genetics guidelines that provide recommendations about hereditary breast and/or ovarian cancer mainly focus on the management of those at risk. Existing breast and ovarian cancer guidelines address issues around diagnosis, treatment or follow up, with some providing guidance about cancer communication. Identifying recommendations from existing guidelines about the information that should be communicated and the
method of communication will help inform health professionals’ interactions with patients and future guideline development and as genetic testing becomes increasingly integrated into mainstream cancer services.

Within genetics clinical practice, as in other areas of healthcare, clinical protocols and patient information leaflets help with local dissemination of guideline recommendations. The availability of these documents to guide practice does not guarantee that guideline recommendations are communicated. Clinical documents can vary widely in content, quality and style, as highlighted by a recent systematic review of reviews of patient information (Sustersic, Gauchet, Foote, & Bosson, 2017). Little is known about the extent to which clinical protocols and patient leaflets used in the management of patients with hereditary breast or ovarian cancer reflect clinical practice guidelines.

Guidelines have the potential to improve both the delivery and outcomes of healthcare, although guideline recommendations are frequently not translated into clinical practice (Grimshaw et al., 2001; Grol & Grimshaw, 2003). Barriers to implementation can occur at many levels, from the individual health professional to the organisation (Grol & Grimshaw, 2003). A systematic review of 76 studies identified knowledge, attitudes and behaviour to be barriers to doctors’ adherence to guidelines (Cabana et al., 1999). The extent to which guidelines for hereditary breast and ovarian cancer are implemented in practice is largely unknown. However, the recommendation to prescribe preventative therapy to women at risk of hereditary breast cancer (National Institute for Health and Care Excellence, 2013) has been investigated in two studies. In a UK study, multiple barriers to implementation were identified amongst primary care and genetics health professionals concerning lack of knowledge of the evidence base, lack of time for discussion and lack of clarity in the guidelines (Smith et al., 2016). In an Australian study, barriers included lack of knowledge amongst health professionals, lack of confidence in the evidence, the legality of ‘off-label’ prescribing and the costs of the drug (Keogh, Hopper, Rosenthal, & Phillips, 2009). Understanding the extent to which recommendations are implemented in current genetic counselling practice will help to highlight areas for further research as new approaches to genetic counselling for cancer patients are developed.
6.2. AIMS AND RESEARCH QUESTIONS

This study aimed to investigate the translation of guideline recommendations about genetic testing and hereditary cancer management information for cancer patients into expert opinion, genetics health professionals' communication and patients' recall drawing on the findings of Studies 1 to 5 in this programme of research. The following research questions were addressed:

Stage I
A. What are the recommendations of clinical guidelines concerning:
   a) The information that should be communicated to patients about i) genetic testing and ii) hereditary cancer management;
   b) The method of this communication?

Stage II
A. In clinical protocols and patient leaflets about hereditary breast and ovarian cancer provided for patients, what proportion of the recommendations communicated were:
   a) Judged to be relevant and not relevant for patients;
   b) About genetic testing and hereditary cancer management;
   c) Referred to in clinical protocols and patient leaflets?
B. Is there a difference in the proportion of guideline recommendations judged to be relevant for patients that were:
   a) Communicated about genetic testing and hereditary cancer management;
   b) Referred to by the clinical protocols and patient leaflets?
C. Which recommendations about a) genetic testing and b) hereditary cancer management were frequently and infrequently communicated in i) clinical protocols and ii) patient leaflets?

Stage III
A. To what extent is the recommended information for patients referred to in genetics and cancer guidelines?
B. Which guideline recommendations were frequently and infrequently:
   a) Referred to by genetics and cancer guidelines and
   b) Translated into expert opinion, communicated by health professionals and recalled by patients?
6.3. METHODS OVERVIEW

Study 5 was conducted in three stages. As each stage builds on data gathered in the previous stage, the procedure and results will be presented for each stage, followed by discussion of the overall findings:

Stage I involved a systematic search and documentary analysis of national and international clinical guidelines about genetic testing and UK guidelines about breast and ovarian cancer management. Documentary analysis involves identifying, selecting, appraising and synthesising data from documents and organising these data using content analysis (Bowen, 2009).

Stage II was a documentary analysis (Bowen, 2009) of clinical protocols and patient leaflets about hereditary breast and ovarian cancer provided by UK Genetics Centres for women with cancer.

Stage III was a content analysis (Silverman, 2006) of data from the published and unpublished studies conducted within this programme of research. The findings of the secondary content analysis from each study were documented onto a mixed methods matrix (O'Cathain, Murphy, & Nicholl, 2010). A comparable score was calculated across all datasets (Cresswell, 2011) to enable analysis of the extent of the translation of each recommendation.

Ethics approval was not required as the study involved anonymised secondary analysis of data gathered throughout this programme of research. The ethics approval for each of these studies is explained in the relevant chapter of this thesis.

6.4. METHODS AND RESULTS

6.4.1. Stage I: Systematic search and documentary analysis of clinical guidelines

6.4.1.1. Stage I: Procedure

6.4.1.1.1. Systematic search to identify guidelines

English language clinical guidelines about genetic testing and hereditary cancer management were identified from consultation with three expert members of the Study Interest Group (see section 1.8.5.4.). In addition, senior genetics health professionals
from all UK Genetics Centres were asked to provide information about the guidelines used within their practice.

A systematic search was conducted of the websites of bodies that publish guidelines in English speaking countries identified through the National and International Cancer Organisations Cancer Index.

A systematic search was undertaken of the Medline and CINAHL databases, using the following search terms: ‘guidelines’ or ‘recommendations’ and ‘breast cancer’ or ‘ovarian cancer’ and ‘hereditary’ or ‘familial’ or ‘genetic’ and ‘cancer communication’ or ‘clinician-patient communication’ or ‘genetic counselling’. The relevant MeSH (medical subject headings) and free text were searched for each concept and filters and Boolean logic terms applied.

Clinical guidelines were included if they were published in hard copy or on the Worldwide Web, written in the English language, were reported as based on evidence and applied a described method. The most recent version of the guideline available was selected. Rescinded or published guidelines identified in the database search but not present on the websites of the bodies that published them, and therefore not current, were not included.

As there are few clinical guidelines about hereditary breast and ovarian cancer for patients with cancer, the search for the information that should be communicated included those for patients and for women at risk of cancer (genetics guidelines). To identify the method of communication for patients, the search also included guidelines about breast or ovarian cancer management (cancer guidelines).

Genetics guidelines were eligible for inclusion if they were published between 2013 and 2017. This time frame was selected to ensure that the guidelines were as up to date as possible, given the rapid developments in the field of cancer genetics. Due to the limited number of guidelines about hereditary breast and ovarian cancer, this search included guidelines that met the inclusion criteria from any English-speaking country.

Cancer guidelines were eligible for inclusion if they were published between 2001 and 2017. This time frame was selected following consultation with the expert members of the Study Interest Group to ensure all guidelines in current use were identified. Due to the large number of guidelines available about breast and ovarian cancer management, the
search was limited to those developed within the UK or Europe, or including experts from the UK or Europe.

In total, 688 potentially relevant sets of guidelines were identified by the searches. After removal of duplicates and screening by the thesis author (CJ), 76 sets of guidelines met the inclusion criteria and were subjected to full review. The selected guidelines were checked by a second researcher (CP) for validation. Twenty-eight sets of guidelines were included in the analysis (see Table 6.1). Rejected guidelines are shown in Appendix 3.5. The flow diagram (Moher, Liberati, Tetzlaff, Altman, & The, 2009) for the selection of guidelines is shown in Figure 6.1.
Figure 6.1. Guidelines review flow diagram
6.4.1.1.2. Sample

Nine genetics guidelines were included in the review. Of these, one was from the UK, three were from Europe, four were from the USA and one was from Australia. Four guidelines focused on genetic testing and hereditary cancer management, four focused on genetic testing only and one focused on hereditary cancer management only (see Table 6.1).

Nineteen cancer guidelines were included in the review. Of these, 11 were from countries within the UK, seven were European and one was international with input from experts from the UK or Europe. Fifteen guidelines focused on breast cancer, three focused on ovarian cancer and one focused on communication about cancer (see Table 6.1).
<table>
<thead>
<tr>
<th>No.</th>
<th>Guidelines</th>
<th>Publishing body where relevant</th>
<th>Publication (updated to be updated)</th>
<th>Focus of guideline</th>
<th>Country of origin</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>Prevention and screening in BRCA mutation carriers and other breast/ovarian hereditary cancer syndromes: ESMO Clinical Practice Guidelines for cancer prevention and screening (Paluch-Shimon et al., 2016)</td>
<td>European Society for Medical Oncology (ESMO)</td>
<td>Aug 2018</td>
<td>Genetics and hereditary cancer management</td>
<td>Europe</td>
</tr>
<tr>
<td>4</td>
<td>SEOM Clinical guidelines in Hereditary Breast and Ovarian Cancer, 2015 (Lort et al., 2015)</td>
<td>Sociedad Española de Oncologia Médica (SEOM)</td>
<td>Dec 2015</td>
<td>Genetics and hereditary cancer management</td>
<td>Spain</td>
</tr>
<tr>
<td>5</td>
<td>Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer in Women- U.S. Preventive Services Task Force Recommendation Statement (Moyer, 2014)</td>
<td>US Preventative services task force</td>
<td>Dec 2013</td>
<td>Genetics</td>
<td>USA</td>
</tr>
<tr>
<td>8</td>
<td>NSGC Practice Guideline- Risk Assessment and Genetic Counseling for Hereditary Breast and Ovarian Cancer (Berliner et al., 2013)</td>
<td>National Society of Genetic Counsellors (NSGC)</td>
<td>2013</td>
<td>Genetics</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Recommendation/Resource</td>
<td>Organization/Source</td>
<td>Date</td>
<td>Type of Management</td>
<td>Location</td>
</tr>
<tr>
<td>---</td>
<td>----------------------------------------------------------------------------------------</td>
<td>-------------------------------------------------</td>
<td>------------</td>
<td>--------------------------</td>
<td>--------------</td>
</tr>
<tr>
<td>11</td>
<td>NICE CSG4 Improving Supportive and Palliative Care for Adults with Cancer [<a href="https://www.nice.org.uk/guidance/csg4">https://www.nice.org.uk/guidance/csg4</a>](accessed 23.09.17)</td>
<td></td>
<td>Mar 2004</td>
<td>Cancer communication</td>
<td></td>
</tr>
<tr>
<td>12</td>
<td>NICE CG80 - Early and locally advanced breast cancer: diagnosis and treatment [<a href="https://www.nice.org.uk/guidance/cg80">https://www.nice.org.uk/guidance/cg80</a>](accessed 23.09.17)</td>
<td></td>
<td>Feb 2009</td>
<td>Breast cancer management</td>
<td></td>
</tr>
<tr>
<td>14</td>
<td>NICE CG122 Ovarian cancer and Initial Management [<a href="https://www.nice.org.uk/guidance/cg122">https://www.nice.org.uk/guidance/cg122</a>](accessed 23.09.17)</td>
<td></td>
<td>Apr 2011</td>
<td>Ovarian cancer management</td>
<td></td>
</tr>
<tr>
<td>No.</td>
<td>Title</td>
<td>Organization/Access</td>
<td>Date</td>
<td>Country</td>
<td></td>
</tr>
<tr>
<td>-----</td>
<td>----------------------------------------------------------------------</td>
<td>-------------------------------------------------------------------------------------</td>
<td>--------</td>
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<td></td>
</tr>
<tr>
<td>19</td>
<td>BGCS Epithelial Ovarian / Fallopian Tube/ Primary Peritoneal Cancer Guidelines: Recommendations for Practice</td>
<td>British Gynaecological Cancer Society (BGCS)</td>
<td>Mar 2017</td>
<td>Ovarian cancer management</td>
<td></td>
</tr>
<tr>
<td>21</td>
<td>Primary Breast Cancer: ESMO Clinical Practice Guidelines for diagnosis, treatment and follow-up (Senkus et al., 2015)</td>
<td>European Society for Medical Oncology (ESMO)/ European School of Oncology (ESO)</td>
<td>Sep 2015</td>
<td>Breast cancer management</td>
<td></td>
</tr>
<tr>
<td>22</td>
<td>3rd ESO-ESMO International Consensus Guidelines for Advanced Breast Cancer (ABC3) (Cardoso et al., 2017)</td>
<td>European Society for Medical Oncology (ESMO)/ European School of Oncology (ESO)</td>
<td>Dec 2016</td>
<td>Breast cancer management</td>
<td></td>
</tr>
<tr>
<td>23</td>
<td>Locally recurrent or metastatic breast cancer: ESMO Clinical Practice Guidelines for diagnosis, treatment and follow-up (Cardoso et al., 2012a)</td>
<td>European Society for Medical Oncology (ESMO)</td>
<td>Jun 2012</td>
<td>Breast cancer management</td>
<td></td>
</tr>
<tr>
<td>24</td>
<td>The EUSOMA recommendations for the management of young women with breast cancer (Cardoso et al., 2012b)</td>
<td>European Society of Breast Cancer Specialists (EUSOMA)</td>
<td>Oct 2012</td>
<td>Breast cancer management</td>
<td></td>
</tr>
<tr>
<td>26</td>
<td>SEOM clinical guidelines in early-stage breast cancer, 2015 (Garcia-Saenz et al., 2015)</td>
<td>Sociedad Española de Oncología Médica (SEOM)</td>
<td>Oct 2015</td>
<td>Breast cancer management</td>
<td></td>
</tr>
<tr>
<td>27</td>
<td>2008 update of the guideline: early detection of breast cancer in Germany (Albert et al., 2009)</td>
<td>Sociedad Española de Oncología Médica (SEOM)</td>
<td>Jul 2008</td>
<td>Breast cancer management</td>
<td></td>
</tr>
<tr>
<td>28</td>
<td>Second international consensus guidelines for breast cancer in young women (BCY2) (Paluch-Shimon et al., 2016)</td>
<td>Endorsed by ESO and EUSOMA</td>
<td>Feb 2016</td>
<td>Breast cancer management</td>
<td></td>
</tr>
</tbody>
</table>
6.4.1.1.3. Development of coding guidelines for identifying the recommended information to be communicated and method of communication for cancer patients and those at-risk

The coding guidelines and method to extract, document and organise the guideline recommendations were developed iteratively by two researchers (CJ and CP). Table 6.2 shows the order in which the guidelines were coded, who they were coded by and the number of rounds of coding required to develop a reliable coding method.

Table 6.2. Order of coding clinical guidelines and initials of researcher undertaking the coding (clinical guideline numbers are shown in Table 6.1)

<table>
<thead>
<tr>
<th>Clinical guideline no.</th>
<th>Type of clinical practice guideline</th>
<th>Order of coding and number of rounds of coding</th>
<th>Coded by</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Genetics</td>
<td>Pilot round of coding</td>
<td>CJ and CP</td>
</tr>
<tr>
<td>28</td>
<td>Cancer</td>
<td>Second round of coding</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>Genetics</td>
<td>Coded once reliable coding method agreed</td>
<td>CJ</td>
</tr>
<tr>
<td>3, 4, 5, 6, 7, 8, 9,</td>
<td>Genetics</td>
<td></td>
<td></td>
</tr>
<tr>
<td>10, 11, 13, 14, 15,</td>
<td>Cancer</td>
<td></td>
<td></td>
</tr>
<tr>
<td>16, 17, 18, 19, 20,</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>21, 22, 23, 24, 25,</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>26</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>27</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

To develop the coding guidelines and method, one set of genetics guidelines (guideline 1) and one set of cancer guidelines (guideline 28) were initially coded by the thesis author (CJ). These two sets of guidelines were purposively selected to represent the variety of types of guidelines included in the study. The NICE guidelines for Familial Breast Cancer (guideline 1) is published on the Web in the standard NICE format. This is the most familiar of the genetics guidelines for UK health professionals, making it a useful starting point for developing the coding method. Guideline 28 was a published in a journal as an article, as were several of the other cancer guidelines, making it a typical example of a cancer guideline.

Each recommendation was coded according to whether it referred to information to be communicated or the method of communication.
To identify information to be communicated, two types of recommendation were highlighted: those that directly referred to information to be communicated and those that referred to actions to be taken by the health professional which would require information to be communicated.

To identify recommendations about the method of communicating information, references to the communication style or approach to be taken by the health professional were highlighted, for example advice to use appropriate language when communicating with the patient or advice to refer a patient for a specialist opinion.

To refine the coding guidelines, a second researcher (CP) independently coded the same two sets of guidelines (1 and 28) using the coding guidelines. This was done using sections which were compared against the author’s codes. If good agreement was not achieved, the discrepancies were discussed, the coding guidelines were amended and a further section coded. This was repeated twice, at which point good agreement was reached. Good agreement was taken to be a Cohen’s kappa co-efficient of 0.60 to 0.74. Subsequent to achieving this reliability level, the coding was undertaken by one researcher (CJ). The coding guidelines are shown in Appendix 3.2.

*Inter-rater coding reliability* was measured using percentage and Cohen’s Kappa co-efficient. Agreement following the pilot round of coding for guidelines 1 and 28 was 79.4% (Kappa 0.608 SE 0.051, p < 0.001). Discussion of the disagreements revealed confusion between coders in identifying recommendations about how communication should take place, as this type of recommendation was made by both the genetics and cancer guidelines. To resolve the confusion, both coders agreed that recommendations from the genetics guidelines about how communication should take place with women with or at risk of cancer about hereditary breast and ovarian cancer would be coded separately from recommendations made by the cancer guidelines about how communication should take place about cancer in general. To test the revised coding guidelines, clinical practice guidelines 2 and 12 were selected by CJ for independent coding. These two sets of clinical practice guidelines addressed genetics and cancer recommendations as well as those published on websites and in the literature. Agreement between the independent coders for these two sets of guidelines was 90% (Kappa 0.869, SE 0.55 p < 0.001). All disagreements were resolved.
6.4.1.1.4. Documentary analysis of clinical practice guidelines

Identifying the information to be communicated to cancer patients and the method of communication involved first identifying the recommendations for patients and for those at risk.

The recommendations from the genetics and cancer guidelines were documented onto separate tables. The titles of the clinical guidelines were documented onto the rows of the tables. The columns were populated with the recommendations made by the guidelines. Each new recommendation identified was documented onto a new column. Similar recommendations were documented onto existing columns.

6.4.1.1.5. Synthesis of recommendations

Once all the clinical guidelines had been analysed and documented, similar recommendations identified in the genetics and cancer guidelines were grouped together and re-labelled using wording close to that used within the guidelines. Expert health professionals within the Study Interest Group (GP, CP and JH) agreed the synthesis and labelling of the recommendations.

6.4.1.1.6. Development of coding guidelines to identify the relevant recommendations for cancer patients

Coding guidelines were developed to identify which of the recommendations were judged to be relevant a) for at-risk women but not for patients, b) patients but not for at-risk women and c) patients and at-risk women.

Recommendations about information be communicated to patients were included if they: i) were made by both the genetics and cancer guidelines or by the cancer guidelines only; ii) were agreed as key messages by expert health professionals and/or service users in Study 2 (Chapter 3) (Jacobs, Pichert, Harris, Tucker, & Michie, 2017) unless judged to be not key by the other group or iii) did not meet criteria i. or ii. (Coding guidelines are shown in Appendix 3.3)

Recommendations about the method of communicating the information to women with cancer were included if they were i) made by the cancer guidelines; ii) concerned the way in which genetic testing should be offered to women with breast or ovarian cancer and those at risk or iii) did not meet criteria i or ii. (See Appendix 3.3)
Inter-rater coding reliability: The recommendations were independently coded against the coding scheme by two researchers (CJ and CP) until a good level of agreement was reached. Agreement about the selection of recommendations for patients was 96% (Kappa 0.947, SE 0.037 p < 0.001). All disagreements were resolved through discussion.

6.4.1.2. Stage I: Results

6.4.1.2.1. Information to be communicated

In total, 45 recommendations concerning information to be communicated about hereditary breast and ovarian cancer were identified: 37 from the genetics guidelines and eight from the cancer guidelines. Synthesis of these 45 recommendations resulted in 31 recommendations of information to be communicated (see Table 6.3). Of these, 14 recommendations were judged to be relevant for women at risk but not for patients, 17 were relevant for patients, of which eight were about genetic testing and nine were about hereditary cancer management. Of the 17 recommendations relevant for patients, two were relevant for patients but not for women at risk and 15 were relevant for patients and for women at risk.

The recommendations judged as relevant for patients but not for women at risk were:
- Possibility of hereditary breast and ovarian cancer and potential risk to relatives.
- Clear quantification of the risk of contralateral breast cancer and the timing, risks and benefits of contralateral mastectomy.

6.4.1.2.2. Method of communication

In total, 38 recommendations were identified about the method of communicating information about hereditary breast and ovarian cancer: 17 from the genetics guidelines and 21 from the cancer guidelines. Synthesis of these 38 recommendations resulted in 17 recommendations about the method of communication (see Table 6.3). Of these, two recommendations were judged to be relevant for women at risk but not for patients and 15 were relevant for patients. Of the 15 recommendations for patients, four were for patients but not women at risk and 11 were for patients and for women at risk.

The recommendations judged as relevant for patients but not for women at risk were:
- Ensure continuity, avoid unnecessary repeated assessments from different health professionals aiming to elicit similar information and provide contact details for a named point of contact in genetics and oncology.
• Involve relevant members of the multidisciplinary team, provide timely referral for expert discussion about surgical options, and offer a follow up plan and second opinion where required.
• Where possible offer genetic testing to individuals with breast or ovarian cancer and at least a 10% probability of a pathogenic variant before testing unaffected relatives.
• Offer genetic testing during initial cancer management to inform decision-making and treatment. If women are not ready to consider testing at diagnosis, offer again at follow up.
Table 6.3 Information to be communicated and the method of communication for patients and at-risk women, showing recommendations referred to in the genetic testing guidelines 1, the cancer guidelines 2 and the genetics and cancer guidelines 3

<table>
<thead>
<tr>
<th>Type of recommendation</th>
<th>Information for at-risk women but not patients</th>
<th>Information for patients but not at-risk women</th>
<th>Information for patients and at-risk women</th>
</tr>
</thead>
</table>
| Information to be communicated about genetic testing | What to expect from risk assessment and genetic counselling. 1  
Personal genetic risk assessment the uncertainties of risk estimation & advice to seek re-assessment if the family history changes or symptoms develop. 1  
Cancer in the general population (including breast & ovarian cancer and age as a risk factor). 1  
Potential reproductive choices for carriers 1  
Privacy, confidentiality & possible consequences related to disclosure of the result. 1  
Psychosocial consequences of genetic testing for the individual and family, including information about support groups and voluntary organisations. 1  
Modifiable hormonal & reproductive risk | Possibility of hereditary breast & ovarian cancer & potential risk to relatives 3 | Potential risks & benefits of genetic testing specific to the type of test being offered, including the implications for the individual and the family and the purpose of testing 1  
Potential cancer risks, including penetrance & variable expression 1  
Potential options for prevention, early diagnosis & surveillance 1  
Autosomal dominant inheritance pattern 1  
Reliability, limitations & informativeness of the genetic test, the probability of finding a mutation, the possibility of a VUS & the likely timescale of results (For panel testing - the potential for incidental & secondary germline information specific to the test being offered, the relevance & potential benefits & the limitations for the individual and family including the possibility & implications of incidental findings) 1  
Legal ethical & social implications of genetic testing including the right to decide not to know. 1  
Informing at-risk relatives about the availability of genetic testing and/or surveillance 1 |
| Information be communicated about hereditary cancer management | Breast awareness & clinical breast examination. \(^1\)  
Limitations & availability of surveillance for other BRCA1/BRCAl2-related cancers. \(^1\)  
Symptom awareness & screening for male carriers. \(^1\)  
Psychosocial & sexual consequences of risk-reducing bilateral salpingo-oophorectomy. \(^1\)  
Options, risks & benefits of PGD & prenatal testing options for BRCA1/2 carriers. \(^1\)  
Risks & benefits of OCP and HRT use for women at high risk of hereditary breast and ovarian cancer. \(^1\) | Clear quantification of the risk of contralateral breast cancer & the timing, risks & benefits of contralateral mastectomy, including relief of anxiety about developing breast cancer, the likely prognosis of breast cancer & the risk of distal recurrence of their previous breast cancer. (For women who have breast conserving treatment - adjuvant endocrine therapy should be used when appropriate based on hormone receptor status to reduce the risk of ipsilateral & contralateral events.) \(^3\) | Type & frequency of breast surveillance available according to age and risk. \(^3\)  
Risks, benefits and limitations of breast surveillance (For TP53 carriers with breast cancer - the risks of malignancy associated with radiation). \(^1\)  
Limitations & availability of ovarian surveillance. \(^3\)  
Ovarian symptom awareness. \(^3\)  
Risks & benefits of risk-reducing mastectomy, including possible outcomes, risk reduction, the possibility of cancer being diagnosed histologically, possibility of immediate or delayed reconstruction & differences in the look and feel of the reconstructed breast. \(^3\)  
Options, risks and benefits of immediate and delayed reconstruction should be discussed with a member of a surgical team with specialist skills in oncoplastic surgery or breast reconstruction. \(^1\)  
Psychosocial & sexual consequences of surgery, including adjustment to the reconstructed breast & changed body image, loss of sensation, emotional well-being & quality of life & details of local & national support services. \(^3\)  
Risks, benefits & timing of bilateral salpingo-oophorectomy including early menopause, ovarian & breast cancer risk reduction & fertility issues. \(^3\) |
| --- | --- | --- | --- |
| Method of communication about genetic testing and hereditary cancer | Offer genetic testing in specialist genetic clinics to a person with no personal history of breast or ovarian cancer | Ensure continuity, avoid unnecessary repeated assessments from different | Ensure information is of evidence-based clear & consistent and does not contradict messages from other service providers. \(^3\)  
Ensure information in a suitable format that is accessible, |
if their combined \textit{BRCA1} and \textit{BRCA2} mutation carrier probability is 10\% or more & an affected relative is unavailable for testing. 

Offer predictive genetic testing to adults after pre-test genetic counselling & with informed consent. 

Offer genetic testing during initial cancer management to inform decision-making and treatment. If women are not ready to consider testing at diagnosis, offer again at follow up. 

understandable, culturally appropriate & tailored to the individual. 

Provide written information to support discussions. 

Provide help with navigating & understanding and information & disseminating genetic information to relatives. 

Facilitate shared decision-making & informed consent. 

Provide adequate time for decision-making & respect the decisions made. 

Offer timely psychological & social support & further counselling to patients concerned about genetic risk or risk-reducing surgery. Provide the opportunity to talk to women who have undergone the procedure. Where appropriate offer support to carers & families. 

Ensure health professionals are appropriately trained in genetic risk assessment, genetic counselling, genetic testing, psychosocial needs assessment. 

Establish an environment conducive to good communication, including honesty and transparency, confidentiality & privacy & respect for the views of the individual. 

Assess risk using a validated model & present personal risk assessment in several ways to facilitate understanding. 

Provide interpretation of genetic test results by a trained health professional with experience &expertise in cancer genetics.
6.4.2. Stage II: Documentary analysis of clinical protocols and patient leaflets from UK Genetics Centres

6.4.2.1. Stage II: Procedure

6.4.2.1.1. Data collection and sample

Senior geneticists and genetic counsellors from each UK Genetics Centre were contacted on up to three occasions and asked to provide written sources of information concerning hereditary cancer for breast/ovarian cancer patients (see Table 6.4).

Table 6.4. Participating Genetics Centres

<table>
<thead>
<tr>
<th>England</th>
<th>Scotland</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bristol</td>
<td>Nottingham</td>
</tr>
<tr>
<td>East Anglia</td>
<td>Peninsula</td>
</tr>
<tr>
<td>Leeds</td>
<td>West London</td>
</tr>
<tr>
<td>Leicester</td>
<td>Sheffield</td>
</tr>
<tr>
<td>Manchester</td>
<td>South East Thames</td>
</tr>
<tr>
<td>North East Thames</td>
<td>South West Thames</td>
</tr>
<tr>
<td>North West Thames</td>
<td>Wessex</td>
</tr>
<tr>
<td>Northern</td>
<td>West Midlands</td>
</tr>
</tbody>
</table>

Clinical protocols (including standard operating procedures, flow charts, pathways, checklists and clinical guidelines) and patient leaflets (including standard letters and paragraphs) were accepted. Documents concerning predictive testing, indirect diagnostic testing of individuals without cancer, men with breast or prostate cancer or individuals with other types of hereditary cancer were excluded from the study.

Documents were received from 20 of the 23 centres contacted. Of the 150 documents received, 88 met the inclusion criteria. Of these, 19 were rejected by the thesis author (CJ) because they were duplicates, concerned genetics referral only, concerned family history only or were not relevant for women with breast or ovarian cancer considering genetic testing or hereditary cancer risk management. Sixty-nine documents were included in the review (see Figure 6.2). The flow diagram (Moher, Liberati, Tetzlaff, Altman, & The, 2009) for the selection of clinical protocols and leaflets is shown in Figure 6.2
Figure 6.2 Document review flow diagram

PRISMA 2009 Flow Diagram

Identification:

150 documents received

Screening:

146 documents screened after duplicates removed

58 documents excluded

Eligibility:

19 documents excluded

Reasons for exclusion:

Concerning genetics

Concerning family history

Not relevant for women with breast or ovarian cancer

Included:

88 documents accepted

42 documents about genetic testing included in review

18 clinical protocols

24 patient leaflets

27 documents about hereditary cancer management included in review

8 clinical protocols

19 patient leaflets

175
The documents were organised into clinical protocols and patient leaflets about genetic testing before the test was undertaken or those about hereditary cancer management for women with a BRCA1/BRCA2 mutation or an equivalent level of risk. The latter were usually relevant following genetic testing. Clinical protocols and leaflets that addressed genetic testing and hereditary cancer management were grouped under ‘genetic testing’ on the assumption that the information was intended to be communicated prior to the test being undertaken. An exception was made for two clinical protocols that clearly addressed pre-test genetic counselling and post-test management of patients and/or at-risk women with a BRCA1/BRCA2 pathogenic variant (see Table 6.5).

Of the 69 documents analysed, 42 were about genetic testing and 27 were about hereditary cancer management; 26 were clinical protocols and 43 were patient leaflets. Forty-eight of the documents were for patients and those at risk. Twenty-one documents were for patients but not those at risk; of these, 19 were about genetic testing and two were about hereditary cancer management.

All the documents were produced by the Genetics Centres for their own use apart from two patient leaflets and three clinical protocols that had been published on the Web by the centres that produced them. No documents were used by more than one centre. Fifty-two documents were dated; the median date was July 2015 (range July 2011 to April 2016). The study sample is shown in Table 6.5.
Table 6.5 Clinical protocols and patient leaflets, showing Centre number and summarised document titles

<table>
<thead>
<tr>
<th>Centre</th>
<th>Genetic testing</th>
<th>Hereditary cancer management</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>• Information for women with an increased lifetime risk of breast and ovarian cancer (patient leaflet)</td>
<td></td>
</tr>
</tbody>
</table>
| 2      | • BRCA genes and inherited breast and ovarian cancer (patient leaflet)  
|        | • Cancer protocol for health professionals (clinical protocol)  
|        | • Breast/Ovarian Cancer Post-Consultation check list (clinical protocol) | • Management of cancer risks (patient leaflet) |
| 3      | • Diagnostic testing for BRCA1/BRCA2 genes (patient leaflet)  
|        | • Protocol for diagnostic testing (clinical protocol) |                              |
| 4      | • Genetic Testing for breast and ovarian cancer (patient leaflet)  
|        | • The Inherited Cancer Exome (patient leaflet)  
|        | • Management of breast and ovarian cancer referrals (clinical protocol) | • Risk-reducing surgery: Bilateral Salpingo Oophorectomy (patient leaflet) |
| 5      | • BRCA diagnostic test - standard paragraphs (patient leaflet) |                              |
| 6      | • Update letter to BRCA carriers (patient leaflet) |                              |
| 7      | • Family history of breast or ovarian cancer (patient leaflet)  
|        | • Breast cancer and genetics: Jewish population (patient leaflet)  
|        | • Breast cancer and genetics: Polish population (patient leaflet) | • Carrier of a BRCA1/BRCA2 pathogenic variant (patient leaflet) |
| 8      | • Genetic testing for hereditary breast and ovarian cancer (patient leaflet)  
|        | • BRCA1/BRCA2 gene testing for ovarian cancer protocol (clinical protocol)  
|        | • Information sheet for patients with cancer (BRCA gene testing) (patient leaflet)  
|        | • Receiving a normal BRCA1/BRCA2 test result (patient leaflet)  
|        | • Receiving a BRCA1 and BRCA2 test result that identifies a variant of unknown significance (patient leaflet)  
<p>|        | • Receiving a BRCA1/BRCA2 test result that identifies a |                              |</p>
<table>
<thead>
<tr>
<th>Page</th>
<th>Pathogenic variant (patient leaflet)</th>
<th>Management of familial breast cancer and ovarian cancer * (clinical protocol)</th>
</tr>
</thead>
<tbody>
<tr>
<td>9</td>
<td>Gene panel testing for inherited cancer susceptibility (patient leaflet)</td>
<td>Management of familial breast cancer and ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td></td>
<td>Management of Familial breast and ovarian cancer * (clinical protocol)</td>
<td>Management of Familial breast and ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td></td>
<td>Breast cancer genetic testing NICE guidelines (clinical protocol)</td>
<td>Management of Familial breast and ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td></td>
<td>Familial breast cancer (clinical protocol)</td>
<td>Management of Familial breast and ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td>10</td>
<td>Ovarian cancer BRCA testing (clinical protocol)</td>
<td>Guidelines for the genetic management of a family history of breast and/or ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td></td>
<td>Breast cancer BRCA testing (clinical protocol)</td>
<td>Guidelines for the genetic management of a family history of breast and/or ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td></td>
<td>BRCA genes and inherited breast and ovarian cancer (patient leaflet)</td>
<td>Guidelines for the genetic management of a family history of breast and/or ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td></td>
<td>Guidelines for the genetic management of a family history of breast and/or ovarian cancer * (clinical protocol)</td>
<td>Guidelines for the genetic management of a family history of breast and/or ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td>11</td>
<td>Pathway for BRCA diagnostic testing (clinical protocol)</td>
<td>Guidelines for the genetic management of a family history of breast and/or ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td></td>
<td>Breast cancer genes: looking for a pathogenic variant (patient leaflet)</td>
<td>Guidelines for the genetic management of a family history of breast and/or ovarian cancer * (clinical protocol)</td>
</tr>
<tr>
<td>12</td>
<td>Protocol 2 BRCA testing guidelines and FAQ (clinical protocol)</td>
<td>Protocol 3 BRCA carrier guidelines and FAQ (clinical protocol)</td>
</tr>
<tr>
<td></td>
<td>Protocol 4 TP53 testing and management guidelines and FAQ (clinical protocol)</td>
<td>A beginner’s guide to BRCA1/BRCA2 (patient leaflet)</td>
</tr>
<tr>
<td>13</td>
<td>Summary of clinical consultation - new referral breast/ovarian cancer (clinical protocol)</td>
<td>BRCA1 diagnostic positive result letter (patient leaflet)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>BRCA2 diagnostic positive result letter (patient leaflet)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Breast/ovarian guidelines (clinical protocol)</td>
</tr>
<tr>
<td>14</td>
<td>Genetic testing BRCA1/BRCA2 (clinical protocol)</td>
<td>Risk-reducing mastectomy pathway (patient leaflet)</td>
</tr>
<tr>
<td></td>
<td>Genetic testing for women affected by ovarian cancer (patient leaflet)</td>
<td>Surgery to reduce the risk of ovarian cancer (patient leaflet)</td>
</tr>
<tr>
<td>15</td>
<td>Diagnostic BRCA genetic testing (patient leaflet)</td>
<td>BRCA1/BRCA2 pathogenic variant carriers’ pathway (clinical protocol)</td>
</tr>
<tr>
<td></td>
<td>Diagnostic genetic testing - Ashkenazi Jewish BRCA alterations (patient leaflet)</td>
<td>Invitation to attend the hereditary cancer carrier clinic (patient leaflet)</td>
</tr>
<tr>
<td></td>
<td>Diagnostic genetic testing - Polish BRCA alterations (patient leaflet)</td>
<td>Invitation to attend the hereditary cancer carrier clinic (patient leaflet)</td>
</tr>
<tr>
<td></td>
<td>Information leaflet for women with ovarian cancer (patient leaflet)</td>
<td>Invitation to attend the hereditary cancer carrier clinic (patient leaflet)</td>
</tr>
<tr>
<td>16</td>
<td>Breast and ovarian cancer checklist (clinical protocol)</td>
<td>Psychology services for patients considering Risk-Reducing Mastectomy (patient leaflet)</td>
</tr>
<tr>
<td>Page</td>
<td>Document Details</td>
<td></td>
</tr>
<tr>
<td>------</td>
<td>------------------</td>
<td></td>
</tr>
<tr>
<td>17</td>
<td>Cancer genetics guidelines (clinical protocol) • BRCA Family service information sheet (patient leaflet) • BRCA risk management options and family matters (patient leaflet) • Protocol for risk-reducing bilateral salpingo-oophorectomy for women with a BRCA1/BRCA2 pathogenic variant (clinical protocol)</td>
<td></td>
</tr>
<tr>
<td>18</td>
<td>BRCA genes and inherited breast and ovarian cancer (patient leaflet) • Management guideline for families with potential BRCA1/2 pathogenic variants (clinical protocol) • Care pathway for women considering risk-reducing mastectomy (clinical protocol) • Risk-reducing mastectomy (patient leaflet) • Risk-reducing oophorectomy (patient leaflet)</td>
<td></td>
</tr>
<tr>
<td>19</td>
<td>Hereditary breast and ovarian cancer (patient leaflet) • Management guidelines for BRCA1 carriers (patient leaflet) • Management guidelines for BRCA2 carriers (patient leaflet)</td>
<td></td>
</tr>
<tr>
<td>20</td>
<td>• BRCA1 and BRCA2 Screening guidelines and management options (patient leaflet)</td>
<td></td>
</tr>
</tbody>
</table>

* Document includes genetic testing and hereditary cancer management information and therefore included in analysis of both groups

6.4.2.1.2. Development of guidelines for coding the clinical protocols and patient leaflets against the clinical guideline recommendations

The variety of clinical protocols and patient leaflets was such that not all recommendations were relevant to code in all documents. For example, it would not be relevant to code a recommendation about breast surveillance as 'not communicated' in a leaflet about bilateral salpingo-oophorectomy. To identify the exclusion criteria for coding each document, coding guidelines were iteratively developed through the coding process. The coding guidelines are shown in Appendix 3.4. To validate the coding guidelines and method, 10% of the documents were randomly selected using SPSS. These seven documents were independently coded against the coding guidelines by a second researcher (CP). The documents were then re-coded by CJ.

Using the coding method developed for the content analysis of transcripts of clinical consultations in Studies 1 and 3, reference to a guideline recommendation judged to be relevant for patients with cancer was accepted as communicated if the main subject of the recommendation was referred to, even when all aspects were not mentioned.
Inter-rater coding reliability was assessed using percentage and Cohen’s Kappa coefficient. Agreement following independent coding was 94% (Kappa 0.861, SE 0.65, \( p < 0.001 \)). Disagreements were discussed and resolved. No changes were made to the coding guidelines. The remaining coding was undertaken by CJ and checked for validation by a second researcher (CP). There were no further disagreements.

### 6.4.2.2. Stage II: Analysis

The frequency of recommendations according to category communicated in clinical protocols and patient leaflets provided by UK genetics centre was analysed using Content Analysis (Silverman, 2006).

Sixteen of the guideline recommendations about the information that should be communicated to patients with cancer identified in Stage I were included in the content analysis. One recommendation, ‘ovarian cancer symptom awareness’ was excluded because the median date of the clinical protocols and patient leaflets was prior to the recommendation being made by the guidelines.

Communicated guideline recommendations were scored as one point. Recommendations not communicated were scored as zero. Where a genetics centre did not provide a document about genetic testing or hereditary cancer management, those recommendations were scored as not communicated. Where a document was provided but the recommendation was not applicable to the document, no score was given.

To address the questions, ‘What proportion of the recommendations communicated were a) relevant and not relevant for patients with cancer; b) about genetic testing and hereditary cancer management; c) referred to in clinical protocols and patient leaflets?’ and ‘Is there a difference in the proportion of guideline recommendations judged to be relevant for women with cancer that were a) referred to in the clinical protocols and patient leaflets; b) about genetic testing and hereditary cancer management?’, the proportion of recommendations (\( n \)) classed as communicating about genetic testing and hereditary cancer management was calculated for each document. These were then used to determine the mean (SD) proportion for each genetics centre. All statistical analyses were conducted in SPSS version 24.

To address the question, ‘Which recommendations were frequently and infrequently communicated to patients with cancer in clinical protocols and patient leaflets?’, the
6.4.2.3. Stage II: Results

6.4.2.3.1. Proportion of guideline recommendations about genetic testing and hereditary cancer management judged to be relevant and not relevant for patients with cancer

A total of 437 guideline recommendations were communicated in 69 clinical documents from 20 genetics centres. Of these, 66.8% (n = 292) were about genetic testing and 33.2% (n = 145) were about hereditary cancer management. In Table 6.6, the mean number of recommendations judged to be relevant and not relevant for patients with cancer are tabulated as a function of message type and document type.

Table 6.6. Proportion (n) of clinical protocol and patient leaflet recommendations communicated per Centre judged to be relevant and not relevant for patients with cancer according to message type and document type

<table>
<thead>
<tr>
<th>Written material from clinics</th>
<th>Clinical protocol and patient leaflet recommendations relevant for patients with cancer % (n)</th>
<th>Clinical protocol and patient leaflet recommendations not relevant for patients with cancer % (n)</th>
<th>Total % (n)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic testing (n=42)</td>
<td>75.32 (220)</td>
<td>24.68 (72)</td>
<td>63.92 (292)</td>
</tr>
<tr>
<td>Clinical protocols (n=17)</td>
<td>75.99 (67)</td>
<td>24.01 (26)</td>
<td>42.59 (93)</td>
</tr>
<tr>
<td>Patient leaflets (n=25)</td>
<td>77.85 (153)</td>
<td>22.15 (46)</td>
<td>57.42 (199)</td>
</tr>
<tr>
<td>Hereditary cancer management (n=27)</td>
<td>83.63 (113)</td>
<td>16.37 (32)</td>
<td>36.08 (145)</td>
</tr>
<tr>
<td>Clinical protocols (n=8)</td>
<td>83.01 (29)</td>
<td>16.99 (6)</td>
<td>27.22 (35)</td>
</tr>
<tr>
<td>Patient leaflets (n=19)</td>
<td>82.1 (84)</td>
<td>17.90 (26)</td>
<td>72.78 (110)</td>
</tr>
<tr>
<td>Total</td>
<td>77.53 (333)</td>
<td>22.47 (104)</td>
<td>437</td>
</tr>
</tbody>
</table>
6.4.2.3.1.1. Proportion of clinical protocol and patient leaflet recommendations judged to be relevant and not relevant for patients

On average, 77.53% (SD 7.92) of clinical protocol and patient leaflet recommendations were judged to be relevant for patients and 22.47% (SD 7.92) were not relevant (see Table 6.6). A Wilcoxon signed-rank test showed that significantly more of the recommendations were relevant than not relevant for patients (Z = -3.92, p < 0.001; r = 0.88).

6.4.2.3.1.2. Proportion of clinical protocol and patient leaflet recommendations about genetic testing and hereditary cancer management

On average, 63.92% (SD 38.11) of clinical protocol and patient leaflet recommendations were about genetic testing and 36.08% (SD 38.11) were about hereditary cancer management (see Table 6.6). A Wilcoxon signed-rank test showed no significant difference between the proportion of guideline recommendations about genetic testing and hereditary cancer management (Z = -1.913, p = 0.056; r = 0.43).

6.4.2.3.2. Proportion of guideline recommendations for patients that were and were not communicated in the clinical protocols and patient leaflets

A total of 333 (42.48%) guideline recommendations judged to be relevant for patients were communicated and 425 (57.52%) were not communicated. In Table 6.7, the mean number of guideline recommendations communicated and not communicated are tabulated as a function of message type and document type.
Table 6.7. Proportion (n) of guideline recommendations for patients communicated and not communicated according to message type and document type

<table>
<thead>
<tr>
<th>Written material from clinics</th>
<th>Guideline recommendations communicated % (n)</th>
<th>Guideline recommendations not communicated % (n)</th>
<th>Total % (n)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic testing (n=42)</td>
<td>47.46 (220)</td>
<td>52.54 (210)</td>
<td>56.7 (430)</td>
</tr>
<tr>
<td><strong>Clinical protocols (n=17)</strong></td>
<td>33.85 (67)</td>
<td>66.15 (130)</td>
<td>47.05 (197)</td>
</tr>
<tr>
<td><strong>Patient leaflets (n=25)</strong></td>
<td>55.81 (153)</td>
<td>44.19 (80)</td>
<td>52.95 (233)</td>
</tr>
<tr>
<td>Hereditary cancer management (n=27)</td>
<td>32.79 (113)</td>
<td>67.21 (215)</td>
<td>43.27 (328)</td>
</tr>
<tr>
<td><strong>Clinical protocols (n=8)</strong></td>
<td>20.08 (29)</td>
<td>79.92 (127)</td>
<td>49.96 (156)</td>
</tr>
<tr>
<td><strong>Patient leaflets (n=19)</strong></td>
<td>48.86 (84)</td>
<td>51.14 (88)</td>
<td>50.04 (172)</td>
</tr>
<tr>
<td>Total</td>
<td>42.48 (333)</td>
<td>57.52 (425)</td>
<td>758</td>
</tr>
</tbody>
</table>

6.4.2.3.2.1. Proportion of guideline recommendations for patients communicated via clinical protocols and in patient leaflets

On average, 27.52% (SD 28.11) of the guideline recommendations were communicated via clinical protocols and 53.10% (SD 28.11) via patient leaflets. A Wilcoxon signed-rank test showed that significantly more recommendations were communicated via leaflets than via protocols (Z = -2.408, p = 0.016, r = 0.54).

6.4.2.3.2.2. Proportion of guideline recommendations for patients communicated about genetic testing and hereditary cancer management

On average, 47.46% (SD 24.45) of the guideline recommendations about genetic testing and 32.79% (SD 28.21) of those about hereditary cancer management were communicated (Table 6.7). A Wilcoxon signed-rank test showed no significant difference between the mean number of recommendations communicated about genetic testing and hereditary cancer management (Z = -1.389, p = .165, r = 0.31).
6.4.2.3.3. Guideline recommendations for patients frequently and infrequently communicated in clinical protocols and patient leaflets

The mean percentage (SD) of guideline recommendations communicated via clinical protocols, patient leaflets and overall are tabulated in Table 6.8 in descending order of overall mean.

The genetic testing recommendation about the possibility of hereditary breast and ovarian cancer was frequently communicated. The recommendation about the importance of informing relatives about genetic risk and surveillance was infrequently communicated.

The hereditary cancer management recommendation about the risks, benefits and timing of bilateral salpingo-oophorectomy was frequently communicated. The recommendation about the risk of contralateral breast cancer and risks and benefits of contralateral mastectomy was infrequently communicated.
Table 6.8 Mean percentage (SD) of guideline recommendations communicated via clinical protocols and patient leaflets

<table>
<thead>
<tr>
<th>Guideline recommendations</th>
<th>Clinical protocols</th>
<th>Patient leaflets</th>
</tr>
</thead>
<tbody>
<tr>
<td>Possibility of hereditary breast and ovarian cancer and potential risk to relatives</td>
<td>Mean %</td>
<td>Mean %</td>
</tr>
<tr>
<td>Options for prevention, early diagnosis and surveillance of the genetic test</td>
<td>50</td>
<td>50</td>
</tr>
<tr>
<td>Reliability, limitations and informativeness of the genetic test</td>
<td>49</td>
<td>49</td>
</tr>
<tr>
<td>Potential risks, benefits of genetic testing</td>
<td>36.67</td>
<td>46</td>
</tr>
<tr>
<td>Cancer risks, including penetrance and variable expression</td>
<td>33.33</td>
<td>46</td>
</tr>
<tr>
<td>Autosomal dominant inheritance pattern</td>
<td>0.49</td>
<td>0.46</td>
</tr>
<tr>
<td>Risks and benefits of bilateral salpingo-oophorectomy</td>
<td>44.43</td>
<td>48</td>
</tr>
<tr>
<td>Sonic and benefits of risk reducing mastectomy</td>
<td>4.1</td>
<td>4.1</td>
</tr>
<tr>
<td>Limitations and availability of ovarian surveillance</td>
<td>36.63</td>
<td>36.63</td>
</tr>
<tr>
<td></td>
<td>Mean %</td>
<td>Mean %</td>
</tr>
<tr>
<td>Possibility of hereditary breast and ovarian cancer and potential risk to relatives</td>
<td>13.56</td>
<td>13.56</td>
</tr>
<tr>
<td>Options for prevention, early diagnosis and surveillance of the genetic test</td>
<td>4.42</td>
<td>4.42</td>
</tr>
<tr>
<td>Reliability, limitations and informativeness of the genetic test</td>
<td>17.38</td>
<td>17.38</td>
</tr>
<tr>
<td>Potential risks, benefits of genetic testing</td>
<td>21.51</td>
<td>21.51</td>
</tr>
<tr>
<td>Cancer risks, including penetrance and variable expression</td>
<td>20.63</td>
<td>20.63</td>
</tr>
<tr>
<td>Autosomal dominant inheritance pattern</td>
<td>21.21</td>
<td>21.21</td>
</tr>
<tr>
<td>Risks and benefits of bilateral salpingo-oophorectomy</td>
<td>23.86</td>
<td>23.86</td>
</tr>
<tr>
<td>Sonic and benefits of risk reducing mastectomy</td>
<td>17.69</td>
<td>17.69</td>
</tr>
<tr>
<td>Limitations and availability of ovarian surveillance</td>
<td>22.88</td>
<td>22.88</td>
</tr>
<tr>
<td>Type and frequency and of breast surveillance $^2$</td>
<td>17.5</td>
<td>37.26</td>
</tr>
<tr>
<td>Reconstruction to be discussed with specialist surgeon $^2$</td>
<td>17.5</td>
<td>37.26</td>
</tr>
<tr>
<td>Psychosocial and sexual consequences of surgery $^2$</td>
<td>15</td>
<td>36.63</td>
</tr>
<tr>
<td>Legal ethical and social implications of genetic testing $^1$</td>
<td>5</td>
<td>0.22</td>
</tr>
<tr>
<td>Informing at risk relatives about genetic testing/surveillance $^1$</td>
<td>14.17</td>
<td>0.32</td>
</tr>
<tr>
<td>Contralateral breast cancer and contralateral mastectomy $^2$</td>
<td>10</td>
<td>30.78</td>
</tr>
</tbody>
</table>

$^1$ Genetic testing recommendations  
$^2$ Hereditary cancer management recommendations
6.4.3. Stage III: Translation of guideline recommendations for patients into clinical guidelines, expert opinion, health professionals’ communication and patients’ recall

6.4.3.1. Stage III. Procedure

The sample for Stage III included the datasets gathered for Studies 1 to 5. Where required, secondary content analysis of each dataset was undertaken. The findings from each analysis were translated onto the columns of a matrix. The guideline recommendations judged as relevant for patients with cancer were translated into the rows of the matrix. Where further content analysis was required, the method referred to in Stage II (see 6.4.2.1.2.) was applied.

To address the question, ‘To what extent is the recommended information for patients with cancer referred to by the genetics and cancer guidelines?’ all recommendations were included for all guidelines with the following exceptions. One genetics guideline focusing on hereditary cancer management was excluded from analysis of genetic testing recommendations; four genetics guidelines focusing on genetic testing were excluded from the analysis of the hereditary cancer management recommendations. One cancer guideline focusing on cancer communication was excluded from analysis of the genetic testing and hereditary cancer management recommendations; three cancer guidelines with a focus on advanced or metastatic breast cancer were excluded from analysis of the hereditary cancer management recommendations apart from ‘Risk of contralateral breast cancer and the timing, risks and benefits of contralateral mastectomy’ which was included in one such guideline; one cancer guideline with a focus on breast reconstruction was excluded from the hereditary cancer management recommendations about ovarian cancer.

To address the question, ‘Which recommendations were frequently and infrequently referred to by the genetics and cancer guidelines?’ the percentage of guidelines that referred to each recommendation was calculated.

To address the question, ‘Which recommendations were frequently and infrequently translated into expert opinion, health professionals’ communication and patients’ recall?’ further analysis was undertaken of the original data from the Delphi survey with expert health professionals (Study 2), transcripts of clinical consultations (Study 3), clinical protocols and patient leaflets (Study 4, stage II) and transcripts of patient interviews following pre- and post-test genetic counselling (Study 5 and Study 1). Where necessary the data from each study were grouped according to the recommendations and a mean
A score for each recommendation was calculated using the scoring system from the original study. The method for the original study and the procedure for secondary analysis of each dataset are summarised in Table 6.9.

**Table 6.9. Original studies and procedure for further analysis**

<table>
<thead>
<tr>
<th>Area of study</th>
<th>Data from original study</th>
<th>Procedure for Study 4III</th>
</tr>
</thead>
<tbody>
<tr>
<td>Guidelines</td>
<td>Study 4, stage I: Documentary analysis of clinical practice guidelines (unpublished)</td>
<td>Each recommendation counted once per guideline (exceptions made when recommendation not relevant for guideline)  Reference to recommendation =1 No reference to recommendation =0</td>
</tr>
<tr>
<td>Expert opinion</td>
<td>Study 2. Delphi survey with expert health professionals to identify judgements of the key messages required by patients about hereditary cancer (Jacobs et al, 2017)</td>
<td>Messages grouped under relevant recommendations Mean score for each recommendation per expert health professional Scored -2 not key to +2 key (as in Study 2)</td>
</tr>
<tr>
<td>Health professionals’ communication</td>
<td>Study 3. Content analysis of extent to which the key messages were communicated in post-test genetic counselling consultations (unpublished) Study 4, Stage II. Content analysis of extent to which the guideline recommendations were communicated in genetics clinical protocols and patient leaflets (unpublished)</td>
<td>Messages grouped under relevant recommendations Mean percentage of recommendations communicated per health professional Reference to message =1 No reference to message =0</td>
</tr>
<tr>
<td>Patients’ recall</td>
<td>Study 5. Transcripts of semi-structured interviews to investigate understanding and experiences of women with newly diagnosed breast cancer following pre-test genetic counselling (unpublished) Study 2. Transcripts of semi-structured interviews to investigate understanding and experiences of women with breast or ovarian cancer and a newly identified BRCA1/BRCA2 mutation (Jacobs et al, 2015)</td>
<td>Content analysis to identify references to genetic testing recommendations communicated per patient Scored as recalled where the main subject of the recommendation was mentioned: Reference to recommendation =1 No reference to recommendation =0 Content analysis to identify reference to the hereditary cancer management recommendations per patient Scored as for Study 5 (above)</td>
</tr>
</tbody>
</table>
6.4.3.2. Stage III: Analysis

The data from each dataset were transformed into a comparable score to enable analysis across the datasets in accordance with a mixed methods approach (Cresswell, 2011). This score is referred to as the ‘matrix score’. To calculate the matrix score, the difference between the highest and lowest scores in the range for each dataset was divided into five quintiles. Each quintile was labelled from 1 (lowest) to 5 (highest). The matrix scores for each dataset were documented onto a further matrix to enable analysis across the datasets. Calculations of the matrix scores for each dataset are shown in Table 6.10.

To address the question, ‘To what extent is the recommended information for patients with cancer referred to by the genetics and cancer guidelines?’ the proportion of recommendations (n) classed as referring to a recommendation was calculated for each guideline. These were then used to determine the mean (SD) proportion for each recommendation.

To address the questions, ‘Which recommendations were frequently and infrequently referred to by the genetics and cancer guidelines?’ and ‘Which recommendations were frequently and infrequently translated into expert opinion, health professionals’ communication and patients’ recall?’, the matrix scores were labelled as follows:

- Frequently referred to/translated (≥ 4);
- Intermittently referred to/translated (2.1 - 3.9)
- Infrequently referred to/translated (≤ 2).

The recommendations were grouped according to the matrix scores.
<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Expert opinion</th>
<th>Health professionals’ communication</th>
<th>Patients’ recall</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetics guidelines</td>
<td>Cancer guidelines</td>
<td>Expert opinion</td>
<td>Clinical consultations</td>
</tr>
<tr>
<td>Percentage guidelines referring to each recommendation (n=8 genetic testing/ n=5 hereditary cancer recommendations)</td>
<td>Percentage guidelines referring to each recommendation (n=8 genetic testing/ n=5 hereditary cancer recommendations)</td>
<td>Percentage guidelines referring to each recommendation (n=18 except for recommendations not relevant to the focus of the guideline)</td>
<td>Mean score per health professional (n=16)</td>
</tr>
<tr>
<td>Range: 20.00 – 100.00%</td>
<td>Range: 0.00 – 20.57%</td>
<td>Range: 0.00 – 20.57%</td>
<td>Score: +2 (a key message) to -2 (not a key message)</td>
</tr>
<tr>
<td>Difference: 80.00</td>
<td>Difference: 28.57</td>
<td>Difference: 1.33</td>
<td>Each quintile: 0.27</td>
</tr>
<tr>
<td>Each quintile: 16.00</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Matrix score</th>
<th>Range</th>
<th>Range</th>
<th>Range</th>
<th>Range</th>
<th>Range</th>
<th>Range</th>
<th>Range</th>
<th>Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>20.00 – 36.00</td>
<td>0 – 5.71</td>
<td>0.81 – 0.88</td>
<td>0.89 – 14.43</td>
<td>5.0 – 14.0</td>
<td>10.0 – 21.51</td>
<td>5.56 – 22.23</td>
<td>6.45 – 18.35</td>
</tr>
<tr>
<td>2</td>
<td>36.01 – 52.01</td>
<td>5.72 – 11.43</td>
<td>0.89 – 1.16</td>
<td>14.44 – 27.98</td>
<td>14.01 – 23.01</td>
<td>21.52 – 33.02</td>
<td>22.24 – 38.91</td>
<td>19.36 – 32.26</td>
</tr>
<tr>
<td>3</td>
<td>52.02 – 68.02</td>
<td>11.44 – 17.15</td>
<td>1.17 – 1.44</td>
<td>27.99 – 41.53</td>
<td>23.02 – 32.02</td>
<td>33.03 – 44.53</td>
<td>38.92 – 55.59</td>
<td>32.27 – 45.17</td>
</tr>
<tr>
<td>4</td>
<td>68.02 – 84.03</td>
<td>17.16 – 22.87</td>
<td>1.45 – 1.72</td>
<td>41.54 – 55.06</td>
<td>32.03 – 41.03</td>
<td>44.54 – 56.04</td>
<td>55.6 – 72.27</td>
<td>45.18 – 58.08</td>
</tr>
<tr>
<td>5</td>
<td>84.04 – 100.00</td>
<td>22.88 – 28.59</td>
<td>1.73 – 2.00</td>
<td>55.09 – 68.83</td>
<td>41.03 – 50.04</td>
<td>56.05 – 67.55</td>
<td>72.28 – 88.95</td>
<td>58.09 – 70.99</td>
</tr>
</tbody>
</table>
6.4.3.3. Stage III: Results

6.4.3.3.1. Proportion of recommendations for cancer patients referred to per guideline about genetic testing and hereditary cancer management

A total of 82 (20.97%) references were made to the recommendations for cancer patients. Of these, 59.6% (n = 64) were made by the genetics guidelines and 6.33% (n = 18) were made by the cancer guidelines.

On average, 57.81% (SD 27.5) of the genetic testing recommendations and 60.0% (SD 14.9) of the hereditary cancer management recommendations were referred to by the genetics guidelines.

On average, 1.39% (SD 4.04) of the genetic testing recommendations and 12.96% (SD 17.04) of the hereditary cancer management recommendations were referred to by the cancer guidelines.

The percentage of recommendations referred to by the genetics and cancer guidelines are tabulated in Table 6.11.

Table 6.11. Percentage (n) of recommendations for cancer patients referred to by genetics and cancer guidelines

<table>
<thead>
<tr>
<th>Recommendations relevant for cancer patients</th>
<th>Recommendations referred to % (n)</th>
<th>Recommendations not referred to % (n)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetics guidelines (n=109)</td>
<td>59.6 (64)</td>
<td>40.4 (45)</td>
</tr>
<tr>
<td>Genetic testing (n=64)</td>
<td>57.81 (37)</td>
<td>42.19 (27)</td>
</tr>
<tr>
<td>Hereditary cancer management (n=45)</td>
<td>60.0 (27)</td>
<td>40.0 (18)</td>
</tr>
<tr>
<td>Cancer guidelines (n=282)</td>
<td>6.32 (18)</td>
<td>93.68 (264)</td>
</tr>
<tr>
<td>Genetic testing (n=144)</td>
<td>1.39 (2)</td>
<td>98.61 (142)</td>
</tr>
<tr>
<td>Hereditary cancer management (n=138)</td>
<td>12.96 (16)</td>
<td>87.04 (122)</td>
</tr>
<tr>
<td>Total</td>
<td>20.97 (82)</td>
<td>79.03 (309)</td>
</tr>
</tbody>
</table>
6.4.3.3.2. Recommendations frequently and infrequently referred to by guidelines

The genetics and cancer guidelines frequently referred to one recommendation:
• The type and frequency of breast surveillance available according to age and risk.

The genetics and cancer guidelines infrequently referred to five recommendations:
• Autosomal dominant inheritance pattern,
• Possibility of hereditary breast and ovarian cancer and potential risk to relatives,
• Legal ethical and social implications of genetic testing,
• Informing at-risk relatives about the availability of genetic testing and/or surveillance,
• Risk of contralateral breast cancer and the timing, risks and benefits of contralateral mastectomy.

Two recommendations were frequently referred to by the genetics guidelines but infrequently referred to by the cancer guidelines:
• Reliability, limitations and informativeness of the genetic test,
• Potential risk and benefits of genetic testing for the individual and the family.

Table 6.12 shows the matrix score for each recommendation referred to by the genetics and cancer guidelines organised in descending order of mean. Table 6.13 shows the recommendations frequently and infrequently referred to by the genetics and cancer guidelines.
Table 6.12. Matrix scores for reference to recommendations in genetics and cancer guidelines

<table>
<thead>
<tr>
<th>Guideline recommendations</th>
<th>Guidelines</th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Genetics</td>
<td>Cancer</td>
<td>Mean</td>
<td></td>
</tr>
<tr>
<td>Risks, benefits and timing of bilateral salpingo-oophorectomy</td>
<td>5</td>
<td>3</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>Type and frequency of breast surveillance</td>
<td>4</td>
<td>4</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>Risks and benefits of risk-reducing mastectomy</td>
<td>5</td>
<td>3</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>Limitations and availability of ovarian surveillance</td>
<td>3</td>
<td>5</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>Reliability, limitations and informativeness of the genetic test</td>
<td>5</td>
<td>1</td>
<td>3</td>
<td></td>
</tr>
<tr>
<td>Potential risk and benefits of genetic testing</td>
<td>4</td>
<td>1</td>
<td>2.5</td>
<td></td>
</tr>
<tr>
<td>Options for prevention, early diagnosis and surveillance</td>
<td>3</td>
<td>1</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>Cancer risks, including penetrance and variable expression</td>
<td>3</td>
<td>1</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>Contralateral breast cancer and contralateral mastectomy</td>
<td>2</td>
<td>2</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>Possibility of hereditary breast and ovarian cancer and potential risk to relatives</td>
<td>2</td>
<td>2</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>Reconstruction to be discussed with specialist surgeon</td>
<td>3</td>
<td>1</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>Autosomal dominant inheritance pattern</td>
<td>2</td>
<td>1</td>
<td>1.5</td>
<td></td>
</tr>
<tr>
<td>Legal ethical and social implications of genetic testing</td>
<td>2</td>
<td>1</td>
<td>1.5</td>
<td></td>
</tr>
<tr>
<td>Risks, benefits and limitations of breast surveillance</td>
<td>2</td>
<td>1</td>
<td>1.5</td>
<td></td>
</tr>
<tr>
<td>Informing at-risk relatives about genetic testing/surveillance</td>
<td>2</td>
<td>1</td>
<td>1.5</td>
<td></td>
</tr>
<tr>
<td>Psychosocial and sexual consequences of breast surgery</td>
<td>3</td>
<td>3</td>
<td>3</td>
<td></td>
</tr>
</tbody>
</table>

1 Genetic testing recommendations
2 Hereditary cancer management recommendations
<table>
<thead>
<tr>
<th></th>
<th>Genetics guidelines</th>
<th>Cancer guidelines</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Frequently referred to</strong> ($\geq 4$)</td>
<td>Type and frequency of breast surveillance</td>
<td>Reliability, limitations and informativeness of the genetic test</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Potential risk and benefits of genetic testing for the individual and family</td>
</tr>
<tr>
<td><strong>Infrequently referred to</strong> ($\leq 2$)</td>
<td></td>
<td>Autosomal dominant inheritance pattern</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Possibility of hereditary breast and ovarian cancer and potential risk to relatives</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Legal ethical and social implications of genetic testing</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Informing at-risk relatives about the availability of genetic testing and/or surveillance</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Risk of contralateral breast cancer and the timing, risks and benefits of contralateral mastectomy</td>
</tr>
</tbody>
</table>

6.4.3.3.3. Guideline recommendations frequently and infrequently translated into expert opinion, health professionals’ communication and patients’ recall

Five *genetic testing* recommendations were *frequently* translated into expert opinion *and frequently* recalled by patients. The first four of the recommendations below were *frequently* communicated by health professionals. The recommendation about autosomal dominant inheritance pattern was *intermittently* communicated:

- Reliability, limitations and informativeness of the genetic test,
- Options for prevention, early diagnosis and surveillance,
- Potential risk and benefits of genetic testing for the individual and the family,
- Cancer risks, including penetrance and variable expression,
- Autosomal dominant inheritance pattern.
Three hereditary cancer management recommendations were frequently translated into expert opinion, intermittently communicated by health professionals and frequently recalled by patients:

- Risks, benefits and timing of bilateral salpingo-oophorectomy,
- Type and frequency of breast surveillance available according to age and risk,
- Risks and benefits of risk-reducing mastectomy.

One hereditary cancer management recommendation was infrequently translated into expert opinion, intermittently communicated by health professionals in protocols and leaflets and infrequently recalled by patients:

- Psychosocial and sexual consequences of breast surgery.

This recommendation was not judged to be a key message in Study 2 and therefore not assessed in clinical consultations.

Two hereditary cancer management recommendations were frequently translated into expert opinion, intermittently communicated by health professionals and infrequently recalled by patients:

- Risks, benefits and limitations of breast surveillance,
- Informing at-risk relatives about genetic testing and surveillance.

One hereditary cancer management recommendation was infrequently translated into expert opinion and infrequently communicated by health professionals but frequently recalled by patients:

- Risk of contralateral breast cancer and the timing, risks and benefits of contralateral mastectomy.

Table 6.14 shows the matrix score for each recommendation in descending order according to patients’ recall. Table 6.15 shows the recommendations frequently and infrequently translated into expert opinion and patients’ recall and the matrix scores for health professionals’ communication.
<table>
<thead>
<tr>
<th>Guideline recommendations</th>
<th>Expert opinion</th>
<th>Health professionals’ communication</th>
<th>Patients’ recall</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Consultations</td>
<td>Protocols</td>
<td>Leaflets</td>
</tr>
<tr>
<td>Reliability, limitations and informativeness of the genetic test</td>
<td>5</td>
<td>NA</td>
<td>4</td>
</tr>
<tr>
<td>Options for prevention, early diagnosis and surveillance</td>
<td>5</td>
<td>3</td>
<td>5</td>
</tr>
<tr>
<td>Potential risk and benefits of genetic testing</td>
<td>5</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>Cancer risks, including penetrance and variable expression</td>
<td>5</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>Autosomal dominant inheritance pattern</td>
<td>5</td>
<td>4</td>
<td>2</td>
</tr>
<tr>
<td>Risks, benefits and timing of bilateral salpingo-oophorectomy</td>
<td>5</td>
<td>3</td>
<td>3</td>
</tr>
<tr>
<td>Type and frequency of breast surveillance</td>
<td>5</td>
<td>5</td>
<td>2</td>
</tr>
<tr>
<td>Risks and benefits of risk reducing mastectomy</td>
<td>5</td>
<td>3</td>
<td>2</td>
</tr>
<tr>
<td>Contralateral breast cancer and contralateral mastectomy</td>
<td>1</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Possibility of hereditary breast and ovarian cancer and potential risk to relatives</td>
<td>1</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td>Limitations and availability of ovarian surveillance</td>
<td>5</td>
<td>4</td>
<td>2</td>
</tr>
<tr>
<td>Legal ethical and social implications of genetic testing</td>
<td>3</td>
<td>5</td>
<td>1</td>
</tr>
<tr>
<td>Risks, benefits and limitations of breast surveillance</td>
<td>4</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>Informing at risk relatives about genetic testing/surveillance</td>
<td>4</td>
<td>4</td>
<td>2</td>
</tr>
<tr>
<td>Reconstruction to be discussed with specialist surgeon</td>
<td>3</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>Psychosocial and sexual consequences of breast surgery</td>
<td>2</td>
<td>NA</td>
<td>2</td>
</tr>
</tbody>
</table>

1 Genetic testing recommendations  2 Hereditary cancer management recommendations
Table 6.15. Recommendations frequently and infrequently translated into expert opinion and patients’ recall, also showing health professionals communication

<table>
<thead>
<tr>
<th>Expert opinion</th>
<th>Patients’ recall</th>
<th>Frequently translated (≥ 4)</th>
<th>Infrequently translated (≤ 2)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Reliability, limitations and informativeness of the genetic test (<em>communicated 4.0</em>)</td>
<td>Risks, benefits and limitations of breast surveillance (<em>communicated 2.67</em>)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Options for prevention, early diagnosis and surveillance (<em>communicated 4.33</em>)</td>
<td>Informing at-risk relatives about the availability of genetic testing and/or surveillance (<em>communicated 2.33</em>)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Potential risk and benefits of genetic testing for the individual and the family (<em>communicated 4.0</em>)</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Cancer risks, including penetrance and variable expression (<em>communicated 4.0</em>)</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Autosomal dominant inheritance pattern (<em>communicated 3.33</em>)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Infrequently translated (≤ 2)</td>
<td>Risks, benefits and timing of bilateral salpingo-oophorectomy (<em>communicated 3.33</em>)</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Type and frequency of breast surveillance available according to age and risk (<em>communicated 3.33</em>)</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Risks and benefits of risk-reducing mastectomy (<em>communicated 3.0</em>)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Risk of contralateral breast cancer and the timing, risks and benefits of contralateral mastectomy (<em>communicated 1.00</em>)</td>
<td>Psychosocial and sexual consequences of breast surgery (<em>communicated 2.5</em>)</td>
<td></td>
</tr>
</tbody>
</table>
6.5. DISCUSSION

6.5.1. Guideline recommendations

Seventeen recommendations were identified from genetics and cancer guidelines about the information to be communicated to cancer patients. Of these, 15 recommendations were judged to be relevant for cancer patients and at-risk women, two recommendations were relevant for cancer patients but not for those at risk, including the risk and management of contralateral breast cancer. Recommended genetic testing information included the potential implications of testing and the risks and management options for carriers. Recommended hereditary cancer management information included the psychosocial consequences, availability, risks, benefits and limitations of surveillance and surgery. Fifteen recommendations were identified about the method of communicating this information to cancer patients. Of these, only four recommendations were judged to be relevant for cancer patients but not for those at risk. These recommendations were about the importance of ensuring continuity and avoiding unnecessary repeated assessments, involving relevant members of the multidisciplinary team, eligibility for genetic testing and the timing of offering genetic testing.

Only two of the information recommendations were specifically for women with cancer. Several studies have identified unmet information needs about preventative surgery, surveillance, chemoprevention, uncertainty about the future and dealing with the impact of a pathogenic variant on the family (Culver et al., 2011; Farrelly et al., 2013; Metcalfe et al., 2000). Two studies with women with newly diagnosed breast or ovarian cancer identified the need for information about the impact of testing for themselves and the available treatment options (Gleeson et al., 2013; Meiser, Gleeson, Watts, et al., 2012). In Study 2 of this programme of research, service users agreed that the impact of genetic testing on insurance, male inheritance and breast cancer risk, the importance of breast awareness and the impact of diet and lifestyle on cancer risk were key messages (Jacobs et al., 2017) although this information was not judged to be required by expert health professionals. Patients may need more specific cancer-focused information about the management of hereditary cancer than is currently recommended by guidelines. This information is particularly important for patients undergoing genetic testing shortly after diagnosis to inform treatment and surgical decisions.

The guideline recommendations about the method of communicating this information to cancer patients are somewhat generic. Previous studies investigating the method of communication have tended to focus on those at risk of cancer. These studies have consistently identified extensive provision of technical information, little engagement in
social or emotional issues and a focus on communication about biomedical rather than psychological and social issues (Aalfs, Oort, de Haes, Leschot, & Smets, 2006; L. Ellington et al., 2006; Ellington et al., 2005; Meiser, 2005; Meiser, Irle, Lobb, & Barlow-Stewart, 2008; Michie, Lester, Pinto, & Marteau, 2005; Pieterse, Van Dulmen, Ausems, Beemer, & Bensing, 2005; Roter et al., 2006). A growing number of cancer patients are being referred for genetic counselling, often at vulnerable stages in their diagnosis and treatment. Genetics health professionals may be unaware of the specific communication needs of cancer patients as these are not clearly addressed in genetics guidelines.

6.5.2. Clinical protocols and patient leaflets

More of the guideline recommendations in the clinical protocols and patient leaflets were judged to be relevant for cancer patients than not relevant. More of the guideline recommendations relevant for cancer patients were communicated in leaflets than in clinical protocols. Less than half of the recommendations judged to be relevant to communicate to cancer patients were communicated.

It is reassuring that more of the communicated recommendations were relevant for cancer patients than not relevant. This may in part be explained by the provision by several Centres of leaflets specifically for cancer patients about diagnostic genetic testing. Only two documents about hereditary cancer management focused on cancer patients despite the differences in risks, implications and outcomes of risk-reducing mastectomy for at-risk women and contralateral mastectomy for patients and at-risk women. This finding suggests that clinical protocols and patient leaflets used in genetics clinical practice focus on genetic testing. This is perhaps unsurprising given that current guidelines do not specifically address the information needs of women with cancer about hereditary cancer management.

More recommendations for women with cancer were communicated in patient leaflets than in clinical protocols. Protocols may be unnecessary if health professionals are already familiar with clinical practice guidelines. Apart from a few leaflets that were published on the Web, most Centres provided independently produced leaflets, highlighting the lack of leaflet standardisation. Patient leaflets can play an important role in enhancing and supplementing information provided by health professionals. Leaflets have been found to improve satisfaction (Mancini et al., 2006), improve recall (Sustersic et al., 2013), change behaviour (de Bont, Alink, Falkenberg, Dinant, & Cals, 2015; Sustersic et al., 2013) and influence decision-making (Mancini et al., 2006; Michie, Di
Lorenzo, Lane, Armstrong, & Sanderson, 2004). Patient leaflets appear to be widely used to communicate hereditary cancer information to patients with cancer; however, availability of leaflets is no guarantee of their content, quality or adherence to guideline recommendations.

Less than half of the genetic testing recommendations and less than a third of the hereditary cancer management recommendations judged to be relevant for cancer patients were communicated via the clinical protocols and patient leaflets. This finding suggests that clinical protocols and patient leaflets do not provide all the information recommended by clinical guidelines for cancer patients.

6.5.3. Translation of recommendations into practice
Approximately 60% of the recommendations for cancer patients were referred to by the genetics guidelines. The cancer guidelines did not refer to recommendations about genetic testing and rarely referred to information about hereditary cancer management. Recommendations about the potential implications of genetic testing and the risks and benefits of risk-reducing surgery were consistently reflected in expert opinion, communicated by health professionals and recalled by patients. It seems from this study that, although expert opinion considers information about sharing genetic risk and surveillance with at-risk relatives to be a key message, cancer patients do not recall the information. Three recommendations concerning the management of hereditary breast cancer were not consistently reflected in expert opinion, health professionals’ communication or patients’ recall: the risks, benefits and limitations of breast surveillance, the psychosocial and sexual consequences of breast surgery and the risks and management of contralateral breast cancer.

Given that until recently, cancer health professionals have had little direct involvement in communicating about genetic testing, it is not surprising that cancer guidelines make little reference to recommendations about hereditary cancer. This finding highlights the lack of specific guidance for oncology health professionals about the information to communicate to patients with cancer and the method of this communication. Research into the uptake and implementation of guidelines has consistently shown that the presence of guidelines does not guarantee their use in practice. Barriers can occur at the level of the individual health professional, within the profession or team, within the practice environment and at the patient level (Ferlie & Shortell, 2001). For oncology health professionals to effectively communicate about hereditary cancer requires a willingness to learn new skills and
knowledge. To facilitate effective communication about hereditary cancer with cancer patients, non-genetics health professionals need clear accessible guidelines and ongoing education and support.

Recommendations about inheritance, genetic testing and the availability of risk-reducing surgery were consistently reflected in expert opinion, clinical communication and patients’ recall. This finding concurs with previous research showing that, for the most part, genetics health professionals are good at communicating information about BRCA1 and BRCA2 (Butow & Lobb, 2004; Butow, Lobb, Meiser, Barratt, & Tucker, 2003). The accepted definitions of genetic counselling (ASHG, 1975; Resta et al., 2006) highlight the importance of communicating information about genetic testing and risk management. This finding suggests that current practice in communicating about genetic testing with cancer patients should inform new approaches to information delivery by non-genetics health professionals.

The recommendation that patients should inform at-risk relatives about the availability of genetic testing and/or breast surveillance was frequently reflected in expert opinion, intermittently communicated by health professionals and infrequently recalled by patients. Previous studies have found that genetics health professionals do generally actively encourage dissemination of information within families (Forrest, Delatycki, Curnow, Skene, & Aitken, 2010; Mendes, Paneque, Sousa, Clarke, & Sequeiros, 2016). The patients may have been given the information and not recalled it. Patients’ recall of medical information is affected by many factors, including large amounts of information (Cowan, 2005), poor understanding of the information, use of medical jargon (Roter et al., 2006), high and low states of anxiety (Kessels, 2003), lack of perceived relevance (Vos et al., 2011), emotional state (Landsbergen, Verhaak, Kraaimaat, & Hoogerbrugge, 2005), and the personal context of the information (Northouse and Northouse, 1998). This finding is concerning given the priority placed on family communication within genetic counselling. Measures to improve family communication are urgently required as genetic testing moves into mainstream oncology where at-risk relatives are less likely to be the focus of health professionals’ communication (Middleton, Hall, & Patch, 2015).

Recommended information about the risks, benefits and limitations of breast surveillance was frequently reflected in expert opinion, intermittently communicated by health professionals and infrequently recalled by patients. Genetics health professionals in clinical practice may consider this information to be outside of their remit and assume it will be discussed by breast health professionals. Patients may not
consider the information to be relevant if they have already decided to have a contralateral mastectomy. Information about breast surveillance is however relevant to patients and their relatives. In the UK, female BRCA1/BRCA2 carriers are offered annual mammography and Magnetic Resonance Imaging (MRI), depending on their age (National Institute for Health and Care Excellence, 2013). The risks and limitations of surveillance concern false-positive and false-negative results and over-diagnosis (Michell, 2012). To inform decisions about risk management options, patients need information about the limitations and harmful effects of surveillance, such as the possibility of recall, needle biopsy and the associated raised anxiety.

The recommended information about the psychosocial and sexual consequences of breast surgery was infrequently reflected in expert opinion, intermittently communicated by health professionals and infrequently recalled by patients. This finding concurs with a review of the psychosocial issues experienced by young women with breast cancer. The review identified inadequate information provision about the impact of treatment and surgery on body image, sexual functioning, fertility and family (Ahmad, Fergus, & McCarthy, 2015). The extent to which experts, or opinion leaders, agree with evidence can influence the translation of evidence into practice (Grol & Grimshaw, 2003). The focus on biomedical information-giving within genetic counselling may have contributed to the infrequent translation of the recommendation into expert opinion (Biesecker & Peters, 2001; Meiser et al., 2008). Discussions about the psychosocial and sexual consequences of surgery can be sensitive, complex and time consuming. It can be challenging for genetics health professionals to provide sufficient information at the same time as attending to psychosocial needs (Hodgson & Weil, 2012a, 2012b). Psychosocial and sexual issues can be difficult to address in the written form, other than by providing access to contact numbers for support organisations (Lewis, Mehta, Kent, Skirton, & Coviello, 2007). Although possible barriers to information recall have been already been discussed, it is possible that patients did not recall the information because it was not communicated to them. For patients with cancer to make an informed decision about surgery to reduce the risk of contralateral breast cancer, information about the psychological and sexual consequences need to be provided.

The recommendation about quantification of the risk of contralateral breast cancer and the timing, risks and benefits of contralateral mastectomy was infrequently reflected in expert opinion and infrequently communicated by genetics health professionals but frequently recalled by patients. Contralateral mastectomy has been shown to be a cost-
effective risk management strategy for BRCA1/BRCA2 carriers (Boughey et al., 2016; Schrag, Kuntz, Garber & Weeks, 2000). Several studies have demonstrated a survival benefit (Heemskerk-Gerritsen, et al., 2015; Herrinton et al., 2005; Metcalfe et al., 2014). However, survival is affected by age at diagnosis, ER-status, the impact of adjuvant chemotherapy (Heemskerk-Gerritsen, Rookus et al., 2015) and the choice to have bilateral salpingo-oophorectomy (Metcalfe et al., 2015). Discussion about the risks and limitations of contralateral mastectomy is therefore an important element of decision-making (Lostumbo, Carbine Nora, & Wallace, 2010). Genetics health professionals may not have felt equipped to communicate the complexity of this information to patients or assumed that the discussion would take place elsewhere. Lack of skills or knowledge amongst health professionals has been identified as a barrier to the translation of guideline recommendations (Cabana et al., 1999). The frequency of contralateral mastectomy amongst cancer patients with a family history of breast cancer is higher than amongst those without, even when no pathogenic variant is identified (Davies et al., 2016). The rise in the frequency of contralateral mastectomy (Narod, 2014; Neuberger, MacNeill, Jeevan, van der Meulen, & Cromwell, 2013), suggests that this option is routinely discussed in the breast clinic. Patients’ recall of this information, even when genetics health professionals did not communicate it, may have been influenced by familiarity with the information from the breast clinic, anxiety about recurrence or social influences (Hawley, Jagsi, Morrow, & et al., 2014; Soran, Kamali Polat, Johnson, & McGuire, 2014)

These findings suggest that recommendations about genetic testing, and the availability of breast surveillance and risk-reducing surgery were consistently reflected in expert opinion, health professionals’ communication and patients’ recall. There were barriers to the translation of recommended information about family communication and the risks, benefits, limitations and consequences of surgical and surveillance options for patients with cancer.

6.6. LIMITATIONS

6.6.1. Stage I

Limitations to Stage I of this study concern the selection of guidelines written in English. By not including guidelines written in other languages, it is possible that some recommendations about genetic testing and hereditary cancer management may have been missed. Although national and international genetics guidelines were included, all
cancer guidelines were from the UK and Europe. Additional recommendations may have been made in cancer guidelines from other countries.

The documentary analysis method, used in Stage I and Stage II of this study, can provide insight into the implementation of guideline recommendations in practice, although it does not provide evidence of practice that actually takes place (Atkinson, 2011).

6.6.2. Stage II
There may be further relevant clinical protocols and patient leaflets that were not included in Stage II. It is recognised that these documents can only provide a snapshot of clinical communication. Because of the wide variety of clinical protocols and patient leaflets, it was necessary to organise them into those about genetic testing and those about hereditary cancer management for analysis. Grouping the protocols and leaflets in this way may not reflect their use in practice.

6.6.3. Stage III
The data used in Stage III were gathered during this PhD programme of research over several years. The data gathered for the study of health professionals’ communication in consultations (Study 4) and patients’ recall of hereditary cancer management information (Study 1) were gathered in 2006-2008, before the guidelines analysed for Stage I were published. However, as the recommendations identified are fairly broad, they were also present in the earlier version of the NICE guidelines for familial breast cancer (McIntosh, 2004; updated 2006) which was used in the UK at the time. It is not known if the recommendations were present in earlier versions of other guidelines reviewed.

Two of the guideline recommendations identified in Stage I of this study were not agreed as key messages by the expert health professionals in Study 2 and were not therefore included in the analysis of health professionals’ communication in consultations in Stage III. These were, ‘Reliability, limitations and informativeness of the genetic test’ and, ‘The psychosocial and sexual consequences of breast surgery’.

Recall of recommended information about the importance of sharing information about genetic testing and surveillance with family members was only assessed in pre-test counselling as this was classed as a genetic testing recommendation. This information may have been communicated and recalled following post-test genetic counselling.
The studies of recall used for Stage III of this study did not specifically assess information communicated in patient leaflets, although the patients may have received written information in addition to genetic counselling. The study of expert opinion (Study 3) and clinical communication in protocols and leaflets (Stage II of this study) involved participants from across the UK, the study of health professionals’ communication in consultations (Study 4) involved two genetics centres only and Study 5 involved one genetics centre.

6.7. CLINICAL IMPLICATIONS

For oncology health professionals to deliver genetic testing in mainstream oncology and for genetics health professionals to effectively communicate with newly diagnosed cancer patients, clear, accessible guidelines are needed about the information that should be communicated and the method of this communication. The guideline recommendations identified in this study provide a starting point for developing new guidelines for communicating with cancer patients about hereditary breast and ovarian cancer. Further research is needed to identify the communication needs of cancer patients who undergo genetic testing during cancer treatment or palliative care.

Given that leaflets are widely used to communicate information within Genetics Centres, it is important that these are targeted towards cancer patients. This does appear to be the case for leaflets about genetic testing. Leaflets about hereditary cancer management however are not focused on the needs of cancer patients. Standardisation of leaflets and the use of clinical protocols may help to improve the translation of guideline recommendations into health professionals’ communication and patients’ recall.

Recommendations about genetic testing and the availability of risk management options were consistently and frequently reflected in expert opinion, health professionals’ communication and patients’ recall. Existing genetic testing recommendations should inform future practice as genetic testing is integrated into mainstream oncology. Recommendations about other aspects of hereditary cancer management require further investigation, in particular the risks, benefits, limitations and consequences of risk-reducing breast surgery and surveillance for women with cancer. This study found that recommendations reflected in expert opinion were communicated in practice and recalled by patients. It is therefore important to ensure that expert genetics and oncology health professionals as well as service users with cancer are involved in developing any future
guidelines about communicating with cancer patients about hereditary cancer. Training for genetics health professionals in communicating with cancer patients as well as training for oncology health professionals in communicating about genetic testing may help to improve future translation of guidelines into practice.

6.8. CONCLUSIONS

National and international genetics guidelines broadly address the information needs of women with cancer. However, there is limited evidence of specific information to communicate to women who undergo genetic testing in order to make decisions about cancer management, or the method of this communication. Breast and ovarian cancer guidelines from the UK and Europe do not make recommendations about genetic testing and rarely refer to recommendations about hereditary cancer management. Genetics clinical protocols and patient leaflets do not provide all the information judged to be relevant for women with breast or ovarian cancer. Genetic testing recommendations were consistently reflected in expert opinion, clinical communication and patients’ recall. There were barriers to the translation of information about family communication between expert opinion and patients’ recall. The availability of breast surveillance and risk-reducing mastectomy were frequently reflected in expert opinion, communicated by health professionals and recalled by patients. However, the risks, benefits, limitations and consequences of these options for cancer patients were not. Information about all the available options is crucial for informed decision-making about breast cancer treatment and the management of future risk. To facilitate communication about hereditary cancer as genetic testing is integrated into mainstream oncology, guidelines need to focus on the communication needs of women with cancer, particularly those being tested in order to inform cancer management.
CHAPTER 7. STUDY 5: UNDERSTANDING AND EXPERIENCES OVER TIME OF PATIENTS WHO UNDERGO GENETIC COUNSELLING AND GENETIC TESTING FOR NEWLY DIAGNOSED BREAST CANCER - INTERPRETATIVE PHENOMENOLOGICAL ANALYSIS

7.1. INTRODUCTION

Patients who are newly diagnosed with breast cancer may be facing the most difficult news of their lives. In addition to learning about the diagnosis, undergoing cancer treatment and confronting their own mortality, approximately 10% of patients may be told that their cancer is, or could be, due to a genetic susceptibility. Such a prospect has an impact on the patient’s own cancer treatment and future cancer risks, and may also have a negative impact on the health of her biological relatives. Until recently, women have been offered genetic testing only after genetic counselling by a genetics specialist and following the completion of breast cancer treatment; such testing is often instigated by another family member. However, improved genetic testing techniques and advances in surgical and medical management of breast cancer have resulted in a lower threshold for genetic testing of cancer patients (Evans et al., 2011) and greater demand for testing to guide treatment (Tutt et al., 2009) and surgery (Domchek et al., 2010; Rebbeck, Kauff, & Domchek, 2009). Over the past five years, oncologists and breast surgeons have begun to refer patients for genetic testing shortly after cancer diagnosis. As demand grows, genetic testing is increasingly likely to be provided by oncology health professionals within cancer clinics with input from a genetics health professional only if a pathogenic variant is identified.

A fast track clinical cancer genetics referral pathway was set up at a UK Regional Genetics Centre in 2011 in response to the demand for genetic testing shortly after diagnosis. This pathway ensured rapid and seamless access to cancer genetics services for patients with newly diagnosed breast cancer where the cancer was potentially due to a pathogenic variant in the \textit{BRCA1}/\textit{BRCA2} genes. The pathway enabled genetic counselling and risk assessment, genetic testing where appropriate and multidisciplinary discussion about cancer management options for carriers to take place prior to definitive cancer surgery. Patients referred to the clinical genetics service prior to surgery entered the fast track referral pathway. Patients were referred immediately after diagnosis or during neo-adjuvant chemotherapy. All patients were triaged by a senior genetics health professional. Patients for whom genetic testing
might be suitable were offered a genetic counselling appointment within one week of referral. Depending on the urgency of the result, a rapid \textit{BRCA1}/\textit{BRCA2} genetic test was offered, with results available within three weeks, or a routine test, with results available within eight weeks. The genetic test result was given in a face-to-face appointment, by post or by telephone, according to the patient’s wishes. A follow-up multidisciplinary clinic appointment was arranged for \textit{BRCA}/\textit{BRCA2} carriers shortly after disclosure of the result. This longitudinal study (Study 5) was designed around the clinical pathway. The study used Interpretative Phenomenological Analysis to explore understanding and experience over time of genetic counselling and testing amongst patients with newly diagnosed breast cancer.

7.2. BACKGROUND

7.2.1. Psychological impact of genetic testing shortly after breast cancer diagnosis

Prior to the availability of targeted cancer treatment, patients and health professionals considered that genetic testing shortly after diagnosis would be overly stressful (Ardern-Jones, Kenen, & Eeles, 2005). Several more recent qualitative studies have investigated actual and hypothetical experiences of genetic testing close to diagnosis. These studies have found that, for the most part, patients and health professionals consider this type of genetic testing to be both acceptable and desirable for decision-making if treatment options are improved as a result (Lacour et al., 2008; Meiser, Gleeson, Kasparian, et al., 2012; Wevers et al., 2017; Zilliacus et al., 2012).

Despite the apparent acceptability of testing, studies examining the psychological impact of genetic testing shortly after cancer diagnosis show conflicting results. An early study found that patients diagnosed with breast cancer within one year prior to genetic testing were more anxious and reported greater levels of breast cancer specific distress than those tested over one year from diagnosis (Bonadona et al., 2002). However, long-term psychological distress amongst women approached about genetic testing at the start of adjuvant radiotherapy during the first year after diagnosis was found to be no greater than amongst patients who were not eligible for genetic counselling (Schlich-Bakker et al., 2008). A recent survey compared attitudes towards and experience of genetic testing amongst women tested shortly after diagnosis and those who received usual care. At six-month follow up, the women who received a genetic test result close to diagnosis were more likely to feel that the
result had influenced their treatment decisions, to have been more actively involved in decision-making and more satisfied with the timing of the testing (Wevers et al., 2017).

Patients’ understanding of the genetic test result however is unclear. A quantitative study found that although 48% of women with a BRCA1/BRCA2 mutation opted for a bilateral mastectomy as their definitive breast cancer treatment, 24% of those who received an uninformative result and 4% of those who declined testing also made this decision, suggesting that understanding amongst the women may be limited and that the approach of the surgeon may have a greater influence on surgical decision-making than the genetic test result (Schwartz et al., 2004). In a qualitative study with patients offered genetic testing shortly after diagnosis, women were satisfied with the timing of the genetic testing although they lacked understanding of the purpose and implications of the testing or the value of the genetics information in the context of their diagnosis (Vadaparampil, Quinn, Brzosowicz, & Miree, 2008).

There have been few studies of patients understanding and experiences of genetic testing shortly after diagnosis despite calls for further research in this area by the NICE guidelines for familial breast cancer (National Institute for Health and Care Excellence, 2013). A qualitative study that included patients tested before and after definitive surgery found that responses were similar regardless of the timing of testing, that few patients knew or understood that the genetic counselling could be separate to the decision to have a genetic test, and that many felt that the information had little utility in relation to their diagnosis (Vadaparampil et al., 2008). A recent qualitative study explored experiences of genetic testing shortly after diagnosis without genetic counselling. For these patients the experience was overwhelming, although this may in part be due to the lack of support throughout the process of genetic testing (Augestad, Høberg-Vetti, Bjorvatn, & Sekse, 2017).

**7.2.2. Perceived breast cancer risk and responses to genetic test results**

The relationship between perceived breast cancer risk and risk management behaviours amongst women without breast cancer has been widely investigated. Patients have a tendency to over-estimate their risk of breast cancer (Croyle & Lerman, 1999; Hopwood, 2000) and those with a high perceived breast cancer risk are more likely to pursue genetic testing or undergo prophylactic mastectomy than those who perceive their risk to be low (Croyle & Lerman, 1999; Katapodi, Lee, Facione, & Dodd,
2004). There have been few studies however of the impact of perceived risk on cancer patients. A systematic review looking at the uptake of genetic testing and the coping strategies of cancer patients found higher levels of anxiety amongst those who decline genetic testing than amongst those who receive a genetic test result, regardless of the result (Case, Andrews, Johnson, & Allard, 2005). A survey of women with breast cancer found that perceived risk influenced anxiety levels following \textit{BRCA1/BRCA2} results disclosure. Those who perceived their risk to be high prior to genetic testing and were found to have a pathogenic mutation expressed high levels of anxiety following the results disclosure. Conversely, carriers whose perceived prior probability of a mutation was low displayed lower levels of anxiety than those who received a VUS or uninformative result. Amongst the women who received a VUS result, those who had a higher perceived risk prior to testing had low anxiety levels following the result whereas those with a low perceived probability expressed a high level of anxiety on receipt of the result (Bredart et al., 2013). A recent quantitative study found that patients who did not expect a hereditary cause for their cancer, displayed higher perceived personal control once the result was disclosed than those who overestimated their prior risk (Bredart et al., 2016).

\section*{7.3. AIMS AND RESEARCH QUESTIONS}

This study aimed to explore understanding and experience over time of genetic counselling and testing amongst patients with newly diagnosed breast cancer.

The study addressed the following research questions:

1. How do women with newly diagnosed breast cancer understand and experience genetic counselling and genetic testing over time?
2. How do prior expectations about the cause of breast cancer impact on understanding and experience of the genetics information?
3. How do women who receive \textit{BRCA1/BRCA2} genetic test results during cancer treatment understand and experience interactions with health professionals over time?
7.4. METHODS

7.4.1. Interpretative Phenomenological Analysis

Interpretative Phenomenological Analysis (IPA) is a qualitative research approach that concerns detailed examination of lived experience in an attempt to understand how individuals perceive and make sense of what they are experiencing (Smith, Ni, & Muram, 2011). IPA is founded on the philosophical theories of phenomenology, hermeneutics and idiography (Smith, Flowers & Larkin 2009). A key element of the methodology is the concept of the ‘double hermeneutic’, whereby the researcher acknowledges that any analysis of the participant’s experience can only be an attempt to make sense of the participant making sense of their experience (Smith & Osborne, 2008).

IPA was selected for this study for several reasons. First the methodology is relevant to areas that are potentially sensitive and emotive as it focuses on the detailed and nuanced analysis of particular lived experiences using an iterative process. This approach is especially useful in areas where there is little previous research, as in the subject of this study. Second, IPA has been used to closely examine the lived experiences of participants in several other areas of genetics (Chapman & Smith, 2002; Macleod, Craufurd & Booth; Smith et al., 2006). Third, the double hermeneutic takes into account the role of the researcher in interpreting the participant’s experience. The concept of ‘bracketing’, drawn from the work of the phenomenological philosopher Husserl, is a central element of the approach, requiring the researcher to constantly reflect on how their own preconceptions might impact on the research and requiring the researcher to acknowledge and set aside preconceptions at all stages of the research process (Smith, Flowers & Larkin 2009). IPA therefore provided an approach and discipline that ‘fit’ well with the role of clinician-researcher with expertise in the field of study.

7.4.2. Design

This longitudinal study was designed to explore patients’ understanding and experiences at three time points: following genetics referral, following genetic counselling and genetic testing and following the genetic test result. Transcripts were subject to Interpretative Phenomenological Analysis, within cases, within groups and across groups.
7.4.3. Participants and recruitment

Purposive sampling methods were used to recruit patients with newly diagnosed breast cancer who were eligible for genetic testing for a BRCA1/BRCA2 mutation based on clinical criteria in order to guide cancer surgery or chemotherapy. The study selection criteria were designed to identify a homogenous sample in accordance with the methodology (Smith, Flowers & Larkin 2009). All participants were White British women aged between 25 and 70 years, who were newly diagnosed with breast cancer and had children or a close female relative under the age of age 50 and therefore eligible for early breast screening. To capture a range of expectations about the causes of breast cancer, recruitment included patients eligible for testing based on their diagnosis of cancer and a strong family history and those eligible based on their personal diagnosis of triple negative breast cancer.

Patients were excluded from the study if they were diagnosed with a second primary or metastatic breast cancer, had a major health condition which may impact on cancer treatment or genetic counselling, such as diabetes, asthma, other types of cancer or severe mental health problems and or if their lifestyle may impact on cancer treatment or genetic counselling, such as obesity, heavy smoking or addiction to drugs or alcohol.

Eligible participants were identified by the clinical genetics health professionals and informed about the study. Potential participants who verbally agreed to contact by the researcher were telephoned within 48 hours of the clinical contact to clarify that the research was separate from the clinical appointment, confirm eligibility, explain the study and answer study-related questions. Patients were informed that participation was entirely voluntary and they would be free to withdraw at any time without sanctions. For those who wished to take part, a research interview was arranged at the participant’s convenience.

Twenty participants were recruited to the study. Of these, two were unable to continue with the study due to the timing of their surgery; one was excluded because it became clear during the first interview that she had already had definitive breast cancer surgery; two were excluded because they were eligible for limited genetic testing based on their ethnicity and one was excluded because she had a life-threatening heart condition. Three further participants who received an uninformative result were interviewed at all three time points but were not selected for analysis as they were unaware of the significance of the family history and therefore had no prior
expectations about the cause of the breast cancer. Eleven participants were included in the analysis.

The mean age at diagnosis of the 11 participants selected for analysis was 45.4 years (range 32 to 67 years). Five participants had a rapid genetic test, receiving results within three weeks of genetic counselling. Six participants had a routine genetic test, receiving results within eight weeks of genetic counselling. The mean length of time between the genetics referral and the first interview was 4.36 days (range 2 to 12 days). The mean length of time between the genetic counselling consultation and the second interview was 18.45 days (range 0 to 67 days). For three participants, there were over five weeks between the consultation and interview 2 due to chemotherapy treatment. The mean length of time between receiving the genetic test result and the third interview was 8.45 days (range 0 to 37 days). Details of the participants selected for analysis and time frames are shown in Table 7.1.
<table>
<thead>
<tr>
<th>Participant</th>
<th>Age at diagnosis in years</th>
<th>At risk relatives</th>
<th>Eligibility for genetic testing</th>
<th>Number of days between genetics referral and interview 1</th>
<th>Number of days between genetic test and interview 2</th>
<th>Type of test</th>
<th>Number of days between genetic test result and interview 3</th>
<th>Genetic test result</th>
</tr>
</thead>
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<td>45</td>
<td>Children</td>
<td>Family history</td>
<td>3</td>
<td>0</td>
<td>Routine</td>
<td>5</td>
<td>Pathogenic variant</td>
</tr>
<tr>
<td>Nicola</td>
<td>32</td>
<td>Children</td>
<td>Family history</td>
<td>4</td>
<td>5</td>
<td>Routine</td>
<td>12</td>
<td>Pathogenic variant &amp; VUS</td>
</tr>
<tr>
<td>Susan</td>
<td>48</td>
<td>Female relative &lt;50</td>
<td>Personal diagnosis</td>
<td>4</td>
<td>67</td>
<td>Routine</td>
<td>37</td>
<td>VUS</td>
</tr>
<tr>
<td>Kate</td>
<td>35</td>
<td>Female relative &lt;50</td>
<td>Personal diagnosis</td>
<td>2</td>
<td>35</td>
<td>Routine</td>
<td>17</td>
<td>VUS</td>
</tr>
<tr>
<td>Angela</td>
<td>50</td>
<td>Children</td>
<td>Family history</td>
<td>3</td>
<td>3</td>
<td>Rapid</td>
<td>3</td>
<td>VUS</td>
</tr>
<tr>
<td>Fiona</td>
<td>49</td>
<td>Children</td>
<td>Family history</td>
<td>5</td>
<td>28</td>
<td>Rapid</td>
<td>5</td>
<td>No variant</td>
</tr>
<tr>
<td>Amanda</td>
<td>67</td>
<td>Children</td>
<td>Family history</td>
<td>3</td>
<td>4</td>
<td>Rapid</td>
<td>3</td>
<td>No variant</td>
</tr>
<tr>
<td>Sarah</td>
<td>35</td>
<td>Children</td>
<td>Family history</td>
<td>5</td>
<td>1</td>
<td>Rapid</td>
<td>0</td>
<td>No variant</td>
</tr>
<tr>
<td>Carla</td>
<td>45</td>
<td>Children</td>
<td>Personal diagnosis</td>
<td>12</td>
<td>49</td>
<td>Routine</td>
<td>1</td>
<td>No variant</td>
</tr>
<tr>
<td>Sophie</td>
<td>45</td>
<td>Children</td>
<td>Personal diagnosis</td>
<td>4</td>
<td>11</td>
<td>Routine</td>
<td>6</td>
<td>No variant</td>
</tr>
</tbody>
</table>
7.4.4. Procedure

7.4.4.1. Ethics

NHS Ethics approval was obtained for this study (12/LO/0243). Final versions of the study documents are shown in Appendix 1.7-1.8. The participant information sheet and consent forms were sent to potential participants at least 24 hours before the first interview and a copy filed in the clinical records. Participants were recruited to the study once the consent form had been signed. General Practitioners were informed of each patient’s participation in the study with the patient’s consent. Participants were advised to direct concerns or questions about their own genetics assessment or cancer management to the health professionals managing their care. All participants were provided with the contact numbers for their local breast care nurse and the clinical genetics department as well as the contact details of the researcher. Names were changed and any identifying words removed from the extracts of the interviews to protect participants’ identity.

7.4.4.2. Data collection

Data were collected between July 2012 and February 2014 using semi-structured research interviews. The goal of data collection in IPA is to adopt the best method for enabling participants to provide a rich, detailed, first person account of their experience. The semi-structured interview is considered the best way to collect data in IPA studies (Chapman & Smith, 2002) and aims to enable the participant to talk about their experiences in their own words.

An interview schedule for each time point was designed to help with preparation for the likely content of the interview and facilitate the participants to tell their story, openly and expansively. Each schedule was structured in such a way as to encourage the participant to begin with a descriptive account before moving to questions designed to elicit more analytical responses. Questions were open-ended. Prompts to encourage open discussion were planned in advance. Care was taken not to lead participants’ responses. The schedule was designed to be used flexibly, enabling the interview to be participant-led as far as possible. The schedules were reviewed by the research supervisor and the Study Interest Group which was set up at the start of the programme of research (see 1.8.5.4.). As a result of this review, amendments were made to the ordering of the questions and prompts were added. The first interview schedule was piloted on a female friend without a cancer family history or genetics experience. This process helped ensure the questions were
comprehensible and that the interview flowed. The rehearsal also increased interviewer familiarity with the order of the schedule. The interview schedules are shown in Appendix 2.5.

The first interview explored experiences leading up to and learning the breast cancer diagnosis, understanding of the cause of cancer and expectations of the genetics appointment. The second interview explored understanding and experiences of the genetic counselling and genetic testing. The third interview explored understanding and experiences of receiving the genetic test result, including the impact of the result on treatment, experiences of living with breast cancer in the knowledge of the genetic test result, understanding of the implications of the result and experiences of health professionals’ communication throughout the whole process.

As far as possible, interviews were conducted close to the patient’s experiences of breast cancer diagnosis, genetic counselling and testing and disclosure of the result. However, the timing was dependent on the wellness and convenience of the participant and the time frame of testing. Interviews were conducted in a quiet and private place of the participants’ choosing. Where participants chose to be interviewed in the genetics department prior to or after their genetic counselling appointment, sufficient time was allowed between the interview and the clinical appointment for the patient to have a break. Each interview was audio-recorded on two devices with the participant’s consent. The purpose of the study, the type of responses being sought and the opportunity to withdraw at any stage was explained before each interview. Following each interview, participants were given the opportunity to chat more generally and ask questions about the research or the process of genetic testing. Each interview lasted for 60 to 90 minutes.

The participants could not be grouped for analysis until the genetic test result was known. As a result the transcription and analysis was not undertaken until the third interviews were completed, avoiding latter interviews becoming increasingly deductive, a problem that can arise with longitudinal studies (Flowers, 2008). To enable the interview to focus on the individual’s experience, the interviewer listened to the audiotape of the previous interview on one occasion prior to the subsequent interview. Once all data were collected for each participant, the interviews were transcribed verbatim by a transcription service used within the NHS. The transcription was checked by the researcher for accuracy against the audiotape.
Once the result was known and all data had been collected, participants were stratified into four groups based on prior expectation of hereditary cancer and the match (congruence or incongruence) between prior expectation and the genetic test result (see Figure 7.1).

Figure 7.1. Stratification of participants into groups for analysis

<table>
<thead>
<tr>
<th>Prior expectation of inheritance</th>
<th>Pathogenic variant and/or VUS</th>
<th>No variant</th>
</tr>
</thead>
<tbody>
<tr>
<td>Expected hereditary cancer</td>
<td><em>Group 1 (congruence)</em></td>
<td><em>Group 3 (incongruence)</em></td>
</tr>
<tr>
<td></td>
<td>Tina</td>
<td>Fiona</td>
</tr>
<tr>
<td></td>
<td>Nicola</td>
<td>Amanda</td>
</tr>
<tr>
<td>Did not expect hereditary cancer</td>
<td><em>Group 2 (incongruence)</em></td>
<td><em>Group 4 (congruence)</em></td>
</tr>
<tr>
<td></td>
<td>Susan</td>
<td>Carla</td>
</tr>
<tr>
<td></td>
<td>Kate</td>
<td>Sophie</td>
</tr>
<tr>
<td></td>
<td>Angela</td>
<td>Andrea</td>
</tr>
</tbody>
</table>

7.4.5. Analysis

At all stages the analysis was supervised and audited by the study supervisor (JAS) to help test and develop the coherence and plausibility of the interpretation. The initial process of analysis involved immersion in the words of the participant through repeated listening to the audiotapes and reading through the transcripts. The next step was close line-by-line analysis of the transcript to identify the themes emerging from the data. In developing these emergent themes, areas of interest, commonality, individuality, convergence and divergence were noted. The aim of developing the emergent themes is to reduce the detail from the transcript and the notes into concise statements that capture the sense and complexity of the data. As far as possible the emergent themes reflected the words of the participant, keeping the analysis at this stage more descriptive than interpretative.

For each transcript, the emergent themes were listed onto a separate document. Duplicates were identified and similar emergent themes combined. Similar clusters of emergent themes were documented onto a table and all quotations from the transcripts relating to that theme were clustered together and their relevance double-checked. Once all the quotations were in the relevant groupings, the clusters of emergent themes were refined, re-labelled and documented onto a table, together
with two or three of the most relevant illustrative quotations. This process was systematically undertaken for the transcripts from each time point for one participant at a time and one group at a time.

To organise and analyse the themes from each time point for each participant and each group, the clustered themes were documented onto a matrix. A structure was developed from the data to illustrate the relationship between the themes across each time point for each group. This process was undertaken for one group at a time.

The final stage in the process involved analysis of the themes across the groups and across the time points. Convergences and divergences were identified between participants and between groups and the themes were re-clustered and re-worded to represent superordinate themes and subthemes. An overall matrix was drawn up for each superordinate theme showing the movement for each participant within each group. A narrative account of the superordinate themes and sub-themes illustrated with verbatim extracts from the participants' words is presented below.

7.5. RESULTS AND DISCUSSION

7.5.1. Presentation of the results and discussion

The results are presented as themes, subthemes and groups, drawing on quotations elucidated from participants. For each theme, the results and the discussion of the findings for each group are presented consecutively. This section is followed by a summary of the themes, explaining the factors affecting the clustering of the groups within each theme. Superordinate themes, sub themes and clustering of the groups are shown in Table 7.2.
<table>
<thead>
<tr>
<th>Group</th>
<th>Expectation of inheritance of cancer susceptibility</th>
<th>Genetic test result</th>
<th>Congruence between expectation and result</th>
<th>Theme A Impact of the genetics information on feelings about inheritance</th>
<th>Theme B Impact of the genetics information on feelings of responsibility for the family</th>
<th>Theme C Impact of the genetics information on feelings of certainty, confidence and concern in decisions about cancer surgery</th>
<th>Theme D Impact of interactions with health professionals on feelings of satisfaction and confidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Expected inheritance</td>
<td>Pathogenic variant</td>
<td>Congruence</td>
<td>Consistently feeling that the cancer is hereditary</td>
<td>Consistently feeling responsible for continuing the cancer legacy</td>
<td>Growing certainty and confidence in decisions about cancer surgery</td>
<td>Consistently feeling disaffected</td>
</tr>
<tr>
<td>2</td>
<td>Did not expect inheritance</td>
<td>VUS</td>
<td>Incongruence</td>
<td>Growing uncertainty about a potential hereditary cause for cancer</td>
<td>Growing relief from the responsibility for exposing the family to cancer</td>
<td>Growing uncertainty and concern in decisions about cancer surgery</td>
<td>Growing increasingly disaffected</td>
</tr>
<tr>
<td>3</td>
<td>Expected inheritance</td>
<td>No variant</td>
<td>Incongruence</td>
<td>Consistently feeling that the cancer is hereditary</td>
<td>Consistently feeling responsible for continuing the cancer legacy</td>
<td>Growing uncertainty and concern in decisions about cancer surgery</td>
<td>Consistently feeling confidence and satisfaction</td>
</tr>
<tr>
<td>4</td>
<td>Did not expect inheritance</td>
<td>No variant</td>
<td>Congruence</td>
<td>Growing uncertainty about a potential hereditary cause for cancer</td>
<td>Growing relief from the responsibility for exposing the family to cancer</td>
<td>Growing certainty and confidence in decisions about cancer surgery</td>
<td>Consistently feeling confidence and satisfaction</td>
</tr>
</tbody>
</table>
Four superordinate themes were identified. Sub themes were identified within each superordinate theme. The themes were labelled as follows:

- Theme A: Impact of the genetics information on feelings of certainty about inheritance;
- Theme B: Impact of the genetics information on feelings of responsibility for the family;
- Theme C: Impact of the genetics information on feelings of certainty, confidence and concern in decisions about cancer surgery;
- Theme D: Impact of interactions with health professionals on feelings of satisfaction.

For each group within each theme the expectation of the result, the actual result and congruence between the expectation of inheritance and the genetic test outcome is presented in the narrative as follows:

- Group 1 (expected inheritance, pathogenic variant: congruence);
- Group 2 (did not expect inheritance, VUS: incongruence);
- Group 3 (expected inheritance, no variant: incongruence);
- Group 4 (did not expect inheritance, no variant: congruence).

To document the change or consistency of experiences over time the number of the interview is shown at the end of each quotation as follows:

- T1 denotes responses at the first interview that took place at Time 1, following the genetics referral;
- T2 denotes responses at the second interview that took place at Time 2, following genetic counselling;
- T3 denotes responses at the third interview that took place at Time 3, following disclosure of the genetic test result.

The groups clustered differently for several of the themes and were influenced by different factors as follows:

- Themes A and B: Groups 1 and 3 versus Groups 2 and 4: influenced by prior views and family experiences
- Theme C: Groups 1 and 4 versus Groups 2 and 3: influenced by congruence between prior expectation of inheritance and the result.
- Theme D: Groups 1 and 2 versus Groups 3 and 4: influenced by the genetic test result.
7.5.2. Theme A: Impact of the genetics information on feelings about inheritance

7.5.2.1. Results (Theme A)

For 10 of the participants, the genetics information (referral, genetic counselling and result) impacted on feelings about inheritance. Those with a prior expectation of inheritance (Groups 1 and 3) consistently felt the cancer was due to a genetic cause. Those without a prior expectation of inheritance (Groups 2 and 4) felt growing uncertainty about a potential hereditary cause for the cancer.

7.5.2.1.1. Consistently feeling that the cancer is hereditary

For the participants in Groups 1 and 3 the genetics information made little difference to the way they felt about why they developed cancer. They were aware of their family histories, expected to develop cancer at some stage and attributed their cancer to a genetic cause, regardless of the genetic test result.

*Group 1 (expectation of inheritance, pathogenic variant: congruence)*

Although shocked at the diagnosis, both participants in Group 1 reflected at all time points on the inevitability of hereditary cancer.

Tina had no direct experience of breast cancer as none of her first-degree relatives were affected. However, once diagnosed, her response suggests prior awareness of the family history and a sense that the cancer has inevitably caught up with her:

‘Yeah Well I knew it. A lot of my family have had it so the chances of me getting it were quite big. I thought I was more my father’s side medically, obviously not. I’ve pulled in a few of my mother’s side …’ (T1)

However, although the susceptibility is inherited from her mother, Tina attributes the cancer to similarities with her father’s medical history and his uncertain heritage.

At time 3 Tina again emphasises her prior expectation of cancer and continues to express surprise at the likely maternal inheritance, despite the clear evidence of a strong maternal family history of cancer:

‘It (the genetic test result) wasn't a surprise. (…) The only thing is (…) my father being adopted we are presuming that the BRCA comes through my mother’s side because we are aware of that family history but it is not definite.’ (T3)
Tina’s consistent belief, or perhaps hope, that the predisposition has been inherited through her father may in part be due to the protectiveness she feels towards her mother.

Awareness of the family history left Nicola with a general feeling of cancer risk. She was not aware of the specific breast cancer risk prior to her diagnosis despite having previously had genetic counselling, possibly because her mother had fallopian tube cancer:

‘I kind of thought I would be more at risk of cancer because of my mum and (...) everybody that has had cancer which is why I made a decision to give up my smoking.’ (T1)

This feeling of risk of inheritance led Nicola to quit smoking but she was unconcerned about breast cancer, seemingly overly reassured by the earlier genetic counselling recommendation to commence breast screening at age 35.

At time 3, Nicola wryly reflects on the inevitability that she would develop cancer, incorrectly associating cervical smears with her mother’s fallopian tube cancer and the risk of ovarian cancer:

‘To be honest I have always had problems down there. I have always had to go back every six months for smear tests because they had come back wrong (...) So really to be honest did not surprise and obviously with my mum and fallopian.’ (T3)

Group 3 (expectation of inheritance, no variant: incongruence)

All three participants in Group 3 expected to have a hereditary predisposition to breast cancer, having lived through their mothers’ experiences of the disease. Although pleased to receive an uninformative genetic test result for their children, they all responded with restraint, suggesting that they continued to feel intuitively that the cancer was due to an inherited predisposition regardless of the genetic test result. Any relief they felt was tempered with an element of caution or, even disappointment, at the failure of the genetic test to explain the cause of the cancer.

In this extract, Fiona talks her physical and emotional likeness to her deceased mother:

‘I've spent years in the shadow of it (breast cancer) with my mum (...) I have become much more anxious like she used to be and (...) physically we are really similar.’ (T1)

At time 3, after receiving an uninformative genetic test result, Fiona experiences an
internal struggle between her feeling that the cancer, along with other characteristics, has been inherited from her mother and the evidence that no mutation has been identified:

‘I started thinking like it wasn't passed on from my Mum (…). No that's not true because she was never tested for it so there is nothing to say that she had the .... at all in fact she probably didn't so it's an unknown. (…) I feel that, I still have probably some kind of genetic propensity for cancer.’ (T3)

This extract suggests that Fiona continues to believe the cancer is hereditary despite the genetic test result.

Amanda too was prepared for a diagnosis, having experienced her mother’s cancer:

‘Because we had had breast cancer in the family with my mum, I think my sister and I had always been prepared that it was a sort of possibility.’

(Interview 1)

Yet when no mutation is found, she refers back to the uncertainty about her father’s side of the family, where testing of one of her paternal uncles some years earlier revealed a VUS:

‘Well certainly it would seem on my father’s side, and I think we may have (a genetic cause). (…) Aberrations on my father’s side were not actual faults so (…) maybe that is something that will come up in the future.’ (T3)

It seems that Amanda continues to feel that there is a genetic explanation for her cancer despite the result. When no mutation is identified in her sample she draws the conclusion that her own test has only looked at mother’s side of the family and that therefore the VUS on her father’s side might still may explain her cancer, even though no such VUS has been found in her own sample.

Sarah was also not surprised when she detected a breast lump:

‘I kind of expected that in my life I would be faced with breast cancer because it's quite common and because my mum's had it twice.’ (T1)

When no mutation is detected at time 3 Sarah is unconvinced:

‘I've always thought that there was some common genetic thing (…) It's good that these genes have been discounted I guess but I still don't know if it's anything to do with our genetic makeup or whether it's just you know a lifestyle thing, or just you know I'm one of those statistics that just would get it just by chance.’ (T3)

She appears to be disappointed that her own and her mother’s cancers cannot be explained and continues to feel that the cancer may be hereditary despite the result.
7.5.2.1.2. Growing uncertainty about a potential hereditary cause for cancer

For the participants in Groups 2 and 4 who had no prior expectation of inheritance, raising the question of a potential genetic predisposition appeared to lead to greater uncertainty about inheritance at time 3 than they felt at time 1.

**Group 2 (no expectation of inheritance, VUS: incongruence)**

For two of the participants in Group 2, uncertainty about inheritance once the result is known seems to be over and above any uncertainty about the meaning of the VUS result.

From the outset, Susan believes that there is ‘no increased risk in the family’ (T1) but also asserts in time 2 that she is ‘carrying on the same pathway’ as her mother. Once she knows the genetic test result, she continues to say that she did not think she had inherited the predisposition from her mother but feels an emotional connection through the shared breast cancer experience:

‘It is not that I feel like my mum gave me cancer, I don’t feel like I have inherited the cancer from her, (…) it sort of feels like, it is bringing me closer to knowing what she went through.’ (T3)

The genetic test result seems to have led to uncertainty about the connection between the cancers that she and her mother experienced. Whilst not believing that she has inherited a genetic predisposition, she feels the strength of the emotional connection with her mother through the shared cancer experience.

Kate had previously been assured that her mother’s breast cancer was the result of radiation treatment following Hodgkin’s Lymphoma. She and her mother were always skeptical and once Kate is diagnosed they become increasingly certain that there must be a genetic cause for the cancers:

‘It’s always been in the back of our minds a bit that whenever we’ve asked anybody they’ve always said it’s nothing to worry about. (…). It feels like there must be a connection’ (T1).

At time 2, having been assured again during genetic counselling that it is unlikely that her mother’s cancer is hereditary, she continues to feel ‘a logical, common sense, human kind of reaction which is my mum had it really young, I’m having it quite young, surely there must be a connection’. Once the genetic test result is known, despite assurances that no pathogenic variant has been detected, she is left with increased uncertainty about the link between the cancers in the family:

‘In the BRCA1 gene there is a slight mutation that (…) isn’t shown to have a strong link with breast cancer, but there might be in the future one day,’
leading us to understand that there is some kind of inherited cancer maybe, but equally it might not be.’ (T3)

Group 4 (no expectation of inheritance, no variant: congruence)

All the participants in Group 4 appeared to be more uncertain about inheritance of cancer predisposition at time 3 than they were at time 1. When the genetic test did not provide them with an explanation for the cancer, they sought an alternative explanation.

At time 1, Carla asserts that ‘there was never any expectation’ that she would develop breast cancer. However, by time 3 she is left with uncertainty:

‘To know that it is not ingrained in you, that there is something going in your DNA that is absolutely unavoidable. (...) I hope it is just the fact that I did smoke a long time ago, or the fact that I am quite a stressed person, or whatever it might be, whatever lifestyle factor, or it could be that nothing has caused it, it is just bad luck, um so I don’t know.’ (T3)

Although she says that she hopes that the cancer is due to her lifestyle, her use of the words ‘ingrained’ and ‘unavoidable’ hint at a wish to absolve herself from blame. In order to resolve this internal dilemma she opts for the fatalistic explanation that the cancer was ‘just bad luck’.

At time 1 Sophie explains that she had ‘always thought that (cancer) won’t be an issue.’ At time 3 however she comments that:

‘The likelihood is that it’s just one of these things and I have been unlucky, so if you try and look at it in that respect, I mean, is she (daughter) any more or less at risk than anybody else because I have had it and it’s not genetic? I don’t really know.’ (T3)

Like Carla, Sophie puts the cause of cancer down to having been ‘unlucky’. However, questions have been raised for her about what the possible inheritance might mean for her children.

For Andrea, the mention of genetic testing comes as ‘a complete shock’ and she rejects her husband’s suggestion that the cancer might be the result of stress:

‘I don’t believe it is stress. I think you have either got it in you or you haven’t. There is nothing you can do about it and I don’t believe it is anything to do with lifestyle, (...) unless, you know, it is smoking and heavy drinking.’ (T1)

Later, in time 3, Andrea says:

‘I have never believed it is because of stress or anything (...). I do believe a
lot of the genetics are hereditary, I know a lot of things can be hereditary but I think I am just not going to know.’ (T3)

There may be some self-preservation in Andrea’s insistence that the cancer is not due to stress, suggesting that she fears stress may indeed have been a contributory factor. In the absence of an acceptable and rational cause for the cancer, Andrea puts it down to ‘genetics’ about which, as the extract above demonstrates, she has limited understanding.

7.5.2.2. Discussion of groups (Theme A)

For both participants in Group 1, who expected and were found to have a pathogenic variant, knowledge of the family history and awareness of the cancer experiences of family members may have contributed to feelings of certainty about inheritance. It seems that their personal and family experiences of cancer and their interpretation of the family history led them to construct their own personal theories of inheritance (McAllister, 2003). This experience enabled them to explain their beliefs about the presence of a genetic predisposition to disease and to resign themselves to the inevitability of hereditary cancer.

The continued uncertainty about inheritance experienced by the participants in Group 3 is clinically correct, given the potential involvement of multiple genetic loci (see 1.4.1). These patients expected to have hereditary cancer but no variant was identified. However, the persistent feeling that the cancer was hereditary seemed to be related to the personal impact of the family experience, rather than the genetic information. Other studies have also reported a continued belief in a genetic cause for disease, even in the absence of a pathogenic variant (Hilgart, Coles, & Iredale, 2012; Michie, Smith, Senior, & Marteau, 2003). Cancer patients with a family history express greater concern about other genetic causes, the risk to children and the risk of breast cancer recurrence than those without a family history (Hamilton & Kopin, 2013). This finding is in line with the Common Sense Model of illness perceptions (CSM) (Leventhal H, 1980). When genes are already included in an individual’s representation of a health threat, for example due to their knowledge of their family history, receiving genetic risk information can have the effect of increasing the degree to which the threat is perceived as genetic and responses to that threat (Marteau & Weinman, 2006).

The participants in Group 2 did not expect to have hereditary cancer but were found to have a VUS. The uncertainty about the cause of the cancer experienced by the participants in Group 2 is perhaps not surprising given that a VUS result is inherently
uncertain. This finding is consistent with a study of the impact of prior probability on anxiety (Bredart et al., 2013), which found that women with a low perceived probability of inheritance displayed high anxiety, depression and intrusion on receiving a VUS result. Misunderstanding or misinterpretation may have contributed to the uncertainty for those in Group 2. Studies have repeatedly demonstrated poor understanding of a VUS result amongst patients (Hallowell et al., 2002; Richter, Graham, Haroun, Eisen, & Warner, 2012; Vos et al., 2008).

For the participants in Group 4, who did not expect a genetic predisposition and who received a result showing that no variant had been detected, there was evidence of growing uncertainty about inheritance and the cause of the cancer. The continued uncertainty about inheritance contradicts previous research which found that young women with breast cancer but without a family history who received an uninformative genetic test result regarded the cancer as a random event (Hamilton & Kopin, 2013). It seems that awareness of the limitations of genetic knowledge was raised for the women in Group 4 at time 2. It is possible that the short time frame between receiving the result and interview 3 revealed an immediate uncertain response that may not have persisted in the longer term.

7.5.3. Theme B: Impact of the genetics information on feelings of responsibility for the family

7.5.3.1. Results (Theme B)
Learning the genetics information impacted on feelings of responsibility for causing the family distress, raising the risk for family members and sharing difficult news amongst 10 of the participants. Regardless of the genetic test result, those that expected to have hereditary cancer (Groups 1 and 3) consistently experienced feelings of responsibility for continuing the cancer legacy, and those with no prior expectation of inheritance (Groups 2 and 4) experienced growing relief from the responsibility for exposing the family to cancer when no pathogenic variant was detected.

7.5.3.1.1. Consistently feeling responsible for continuing the cancer legacy
Regardless of the genetic test result, feelings of responsibility for continuing the cancer legacy persisted across all time points for both groups of participants.
**Group 1 (expectation of inheritance, pathogenic variant: congruence)**

For both participants in Group 1 there was an overwhelming sense of responsibility for protecting younger and older generations, both from distress caused as a result of their own cancer and the risk associated with the pathogenic mutation.

At time 1 Tina’s ability to provide for and protect the family is severely threatened by her diagnosis:

‘I’m the breadwinner and I’m self-employed (…) I have to do everything I can to make sure things are in place so that when I collapse and I’m rubbish that my family is supported.’ (T1)

Her need to protect the family, and indeed herself, from the genetic test result leads her to deny the significance for them. She does this in time 2 for her mother and sister and in time 3 for her son:

‘My mum is old, you know, if she gets ill, she is not going to survive it, so (…) she would not gain anything from knowing. My sister doesn’t really want to know, so that is her choice.’ (T2)

Nicola’s fierce sense of responsibility to protect her family is seen in her response to the genetics referral:

‘I was relieved but again not for me, for my daughter and also my younger sister, (…) it’s for the women in my family they need to know as much as I do.’ (T1)

Her ability to maintain the role of family protector is threatened by her debilitating chemotherapy:

‘I’ve sort of taken my mum’s place, you know in holding everybody else together and sorting them out so I thought maybe I need to take a little step back and let them do a little bit for me.’ (T2)

Despite her tentative suggestion at time 2 that she steps back from this role, by time 3 she has reverted to her usual position of protecting and nurturing the family:

‘I don’t try to worry them too much about things, I still keep things here. I had a little bit of a blow out last week and had a little bit of a wobble, but did it on my own and no one saw it. Because if I was to go they would all go.’ (T3)

**Group 3 (expectation of inheritance, no variant: incongruence)**

Two of the participants in Group 3 reflected on the cancer legacy, looking backwards at the painful experience for their mothers and forwards at the potential implications for their children and/or siblings. These participants continued to experience feelings
of responsibility for the family despite the genetic test result showing that no variant had been detected.

The experience of diagnosis leads Fiona to reflect on the pain of her mother’s experience and her own feelings of inadequacy at being unable to help her mother:

‘I saw her on the floor, panting like a dog and imagining somebody was coming and was at the door and sitting cowering in the corner of the car, and to help her was really difficult and she was angry and she was horrible to me (...) I couldn’t help her.’ (T1)

Her mother’s experience is also at the forefront of her mind as she acknowledges the potential impact of the cancer legacy in her siblings:

‘The three of us siblings, we are all carrying this experience that we had with my Mum.’ (T3)

Sarah constantly worries about the implications of both the diagnosis and the cancer legacy for her children and her mother. She refers to the shared experience with her mother at time 1:

‘The investigations I was having before made me think about me and mum and (...) what we’ve been through and seeing things from her perspective.’ (T1)

She worries about the implications of the genetic test for her mother. Here she is talking about the possible ovarian cancer risk associated with a pathogenic mutation:

‘It just worries me about my mum, (...) more than me just because she is in that age group.’ (T2)

Once she knows the result she is still left with a feeling of responsibility and concern for her daughters who are still young:

‘I started to wonder about my daughters, (...) what implication it has for them, (...) they probably would start to be monitored at forty, as opposed to thirty, so you start to worry if you know that’s okay or not.’ (T3)

7.5.3.1.2. Growing relief from the responsibility for exposing the family to cancer

Although several of the participants in Groups 2 and 4 had relatives with cancer, all (except Angela) had previously been reassured about the significance of the family history. Regardless of the genetic test result, feelings of responsibility for exposing the family to cancer persisted across times 1 and 2 but were relieved at time 3 for all the participants in these two groups.
Group 2 (no expectation of inheritance, VUS: incongruence)

For all the participants in Group 2 there is a sense of responsibility for the distress the information about the diagnosis and potential gene mutation would cause to relatives at Time 1. By Time 3, however the participants all experience a sense of relief of responsibility for the family, despite the abnormal genetic test result and the feelings of uncertainty about inheritance discussed in Theme A.

At time 1 Susan feels responsible for causing her sister worry:

‘I feel terrible (…) It's a bit irrational, but I think if I have got a genetic mutation I feel a bit responsible for her.’ (T1)

At time 2 she is ‘hugely relieved’ when her sister has a clear mammogram but the feeling of responsibility remains until she learns that no mutation has been detected in her own sample. At time 3 she expresses relief, although she acknowledges the uncertainty surrounding the VUS:

‘I'm just very, very, very thankful that she doesn't have to worry and whatever this genetic variant I have is (…) hopefully it doesn't mean that things are going to be very bad for either of us’ (T3)

For Kate, the feeling of responsibility is about causing distress to her family. She is acutely aware of her mother’s own cancer experience as she learns the diagnosis:

‘I kept thinking 'I'm so sorry', and feeling very upset for my mum, (…) knowing she’s going to have to relive it through me I found at that moment more difficult than the fact that it's happening to me’ (T1)

At time 3 she expresses relief at being able to share the good news with her family:

‘I just wanted to tell my mum and dad and my brother that this is okay’. (T3)

Angela also feels hugely responsible for the distress she will cause to the family by telling them about her diagnosis and the genetic test. In this extract from time 1 she worries about having to tell her sons about her own diagnosis just a year after their father’s cancer diagnosis:

‘I couldn’t tell them. (…) I didn't think I'd have to tell them for a second time that one of us has got cancer. Didn't want to do that.’ (T1)

She feels responsible, both for breaking the bad news and for putting them through the pain of cancer for a second time. In time 2 she talks again about not wanting to cause pain to her family by telling them about the genetic test:

‘You think how the hell am I going to tell people (…) I don’t want to be the one that is doing it.’ (T2)
Unlike Susan and Kate however, Angela chooses to hear only the good part of the genetic test result, completely overlooking the VUS result in her relief:

‘Lovely, fantastic, because of all the implications if it was positive for the family. (…) it was nice to be able to give my family some good news for a change.’ (T3)

Group 4 (no expectation of inheritance, no variant: congruence)
The participants in Group 4 felt heightened concern about exposing the family to cancer at time 2. For all three participants, there was a strong sense of relief from responsibility at being able to share the news that no mutation was found in their genetic test result.

For Carla at time 1 there is concern about distressing the family by telling them about her breast cancer diagnosis:

‘The worst thing is telling people and you just feel like, oh God I don't want to ruin another person’s day. (…) I'm upset, I'm devastated but I'm more devastated for the people around me really, you know for the impact it has on your family, your children, my mum.’ (T1)

Once she knows the result, she feels enormous relief of responsibility:

‘Not only are my kids having to go through me being ill but I could, (…) have passed a gene onto her (daughter), that would just be devastating. It was a sense of responsibility that, the fact that it had happened (…) to me first out of my immediate family, you know, you think, it was a feeling of a burden, it was a bit of a burden, (…) so to have that taken away.’ (T3)

The genetic test result appears to relieve Carla of responsibility, not only for the potential risk to her children, but also for being the first in the family to develop breast cancer thereby putting everyone at risk. Her repeated use of the word ‘burden’ suggests that this feeling of responsibility was at the forefront of her mind, and highlights the relief she feels when the ‘burden’ is lifted.

Sophie does not talk about the family at time 1 as the potential impact only begins to dawn on her at time 2 once she has been tested:

‘I've got a brother and a son and I suppose I didn't realise that it did have an implication for the males apart from passing on the gene if it was positive. (…) Obviously it’s got more consequences.’ (T2)

She expresses relief of responsibility for the family at time 3 saying:
I'm obviously very pleased that that's not something they are going to have - it's not a decision that they are going to have to be involved with.' (T3)

Andrea, whose father had died from an unrelated cancer, also feels responsible for putting the family through cancer again:

'We can't all go through this again (...). That was what my thought was. If I've got it we have all got to go through this again (...) me and my sister and brother and my mum’ (T1)

She refers to her feelings of responsibility at time 2, saying 'I feel I know how mum feels, if she has got the gene and passed it to us'. At time 3, when she knows the result she is relieved, both for herself and for her family that she ‘can tell them they don't have to be tested.'

7.5.3.2. Discussion of groups (Theme B)

The participants in Group 1, whose expectations of hereditary cancer were confirmed, regarded their matriarchal role as central to the functioning of the family, which may explain their strong sense of responsibility to be tested. Several studies have identified responsibility to provide genetic information for the family as a primary motivator for genetic testing amongst cancer patients tested before and after cancer treatment (Brandt, Hartmann, Ali, Tucci, & Gilman, 2002; Hallowell et al., 2003; Julian-Reynier et al., 1998; Zilliacus, 2012). There is a strong sense of obligation to undergo genetic testing for the benefit of other family members, in particular children and sisters, even at the expense of one’s own wishes (Dancyger, Smith, Jacobs, Wallace, & Michie, 2010). A qualitative study of women’s explanations for their decisions to have BRCA1/BRCA2 genetic testing found that notions of responsibility, caring and nurturing associated with womanhood and motherhood were intertwined with decision-making (Rowley, 2007).

For those in Group 3 in whom no variant was identified despite expectation, there was a sense of family solidarity that appeared to have a greater association with shared family cancer experience than shared genes. This finding is consistent with the work of the anthropologist Kaja Finkler who, in her study of the medicalisation of the family through genetic testing, points out ‘the distinction between experiencing oneself as a member of a “significant same” group that feels a sense of solidarity and relatedness associated with shared experiences from the beginning of life and experiencing oneself as a member of a family, or group, that shares DNA molecules.’ (Finkler, 2001, p.248).
None of the women in Group 2 or Group 4 had a prior expectation of cancer and all were surprised and concerned to learn about the potential wider impact of the test for the family. It seems that until the introduction at Time 2 of the concept of ‘genetic responsibility’ (Leefmann, Schaper, & Schicktanz, 2016) to enable family members and future generations to avoid inherited disease, participants without a prior expectation of inheritance were unaware of the conflicting responsibilities. For women with cancer, particularly those tested shortly after diagnosis when decisions about treatment and surgery are made, there is also a responsibility for managing their own cancer (Hallowell, Jacobs, Richards, & Gore, 2001). By making a decision that could potentially benefit their own treatment, patients have to balance the responsibility for exposing loved ones to cancer risk and difficult choices at the same time as protecting family members from distress and harm (Hallowell et al., 2003; Smith, Stephenson, Jacobs, & Quarrell, 2013). The tension between the responsibility for providing information that may be helpful for relatives and the responsibility for protecting loved ones from potential disease may have contributed to the strong feelings of responsibility expressed by the participants in Group 2 and Group 4.

The relief expressed for the family by the participants in Group 2 who were found to have a VUS contradicted their uncertainty about inheritance (Theme A). There are a number of possible explanations for this response, including lack of understanding or misinterpretation of the result (Hallowell et al., 2002), immediate relief that no pathogenic variant had been identified, confirmation of the prior view that the cancer was not hereditary or a combination of all three reasons.

7.5.4. Theme C: Impact of the genetics information on feelings of certainty, confidence and concern in decisions about cancer surgery

7.5.4.1. Results (Theme C)

Learning the genetics information had an impact on feelings of certainty, confidence and concern in decisions about cancer surgery for 10 of the participants. For those where there was congruence between the expectation of hereditary cancer and the genetic test results (Group 1 and Group 4), there was evidence of growing certainty and confidence in these decisions. For those whose expectations were incongruent
with their genetic test results (Group 2 and Group 3), there was growing uncertainty and concern about the decision not to have a double mastectomy.

7.5.4.1.1. Growing certainty and confidence in decisions about cancer surgery
The impact of the genetics information on feelings about cancer surgery was associated with the congruence between the prior expectation of the genetic test and the outcome of genetic testing, rather than the genetic test result itself.

Group 1 (expectation of inheritance, pathogenic mutation: congruence)
The participants in Group 1 were both clear from the outset that they wanted to have a double mastectomy. The genetics information further cemented this decision.

At time 1 and time 2, Tina talks about gathering the expert opinion of the genetics and surgical health professionals, even though her instinct is to go ahead with a double mastectomy. It is telling that in the extract below she accidentally uses the word ‘cemetery’ rather than ‘symmetry’, suggesting that her decision is influenced by the threat to survival as much as by the aesthetic reasons, even though it is these that she focuses on.

‘We’ll go and see the genetics because it might highlight whether we do a double mastectomy or you know (...) I come from a still life photography background (...) so there’s an element of cemetery, symmetry and balance in probably everything I do (...) I was like kind of seems a no brainer really.’ (T1)
Tina makes a pragmatic and proactive decision not to have a rapid genetic test that would have been available to her in order to guide surgical decision-making.

‘I had decided that actually I would have the double mastectomy because I would have to be having surgery for a reduction to balance me out anyway and because I had so much cancer in the family I just thought if I have got to have so much surgery anyway, why don’t I just have a double mastectomy, get that sorted straight away, that removes a large percentage of risk, therefore by doing that whether I am BRCA faulty or not will just enhance my decision rather than affecting it or swaying it’ (T3).
Tina’s decision to go ahead with a double mastectomy regardless of the result and against the advice of the surgeon, suggests that either that she is completely confident that the cancer is hereditary, or that were the genetic test result to show no abnormality she would not believe it.
Nicola takes a similarly pragmatic approach to the decision about surgery, having made up her mind about that she would want a double mastectomy before her genetics appointment:

‘I know (the genetic test) affects my surgery but (…) I had already decided I don’t want them anymore and obviously the ovaries (…) I don’t need them anymore, I’ve got my family that’s the way I look at it, I would rather be here than not here at all’ (T1).

At time 2 she emphasises that she sees the purpose of the genetic test as to persuade the surgeons that she needs a double mastectomy:

‘It’s not going to change that I’ve got cancer so the only thing that’s going to change for the surgeons’ view is my surgery. Whereas (…) I’ve already decided, that’s what I want to do anyway.’ (T2)

At time 3, once she knows the genetic test result, there is certainty and confidence in her rationale for why she would prefer to go ahead with having her ovaries removed without delay:

‘I obviously know that means a hysterectomy is on the cards. I have been told that they like to do it closer to the age of 40. I will not contest it but I would like it done. I have got my children. I do not plan on having any more children. So I obviously know that then that will have to be done. I would like it done sooner rather than later.’ (T3)

**Group 4 (no expectation of inheritance, no variant: congruence)**

For all three participants in Group 4 there was a sense of certainty and confidence about the progression of treatment once the threat of a double mastectomy had been eliminated by the genetic test result.

At time 1, Carla describes the genetic test as ‘another thing to go through’ and she is ‘fairly ambivalent about it’ (T1). At time 3 the genetic test result leads her to feel confident that she may no longer need to have radical surgery:

‘My initial reaction was, the very first time I had my diagnosis was I want a mastectomy immediately and now I am kind of thinking (…) I don’t want that at all (…) now I am feeling I might get away with it, I am kind of thinking maybe I will, you know, that is kind of in the back of my mind, maybe I will (…) get away with it and not have to do that if I keep getting good results.’ (T3)

Sophie too regards the genetic test as routine, saying at time 1 it is ‘just all part of it’. Once she knows the genetic test result she feels able to plan ahead:
‘I knew which way I was going to be going (with cancer surgery) and so it was good to actually have a plan’ (T3)

For Andrea at time 1 there is relief that the test has been offered and concern about having the test:

‘It does worry me a lot that if I have got, you know, the gene (…) It is horrible but I am really glad and I want things to move and carry on.’ (T1)

Andrea attributes the low feelings she experienced during her chemotherapy treatment in part to the extended wait for the genetic test results and the uncertainty involved. Once she learns the genetic test result, she expresses relief and hope as she gains certainty about her treatment:

‘I felt so rough after this chemo, really that was the worst one ever (…) and then waiting for the result it was all, it does get a bit much (…) I have got hope now and I have got things, you know, now I know that I am going to have surgery and once I have had the surgery that will be a big relief to know that it has gone.’ (T3)

7.5.4.1.2. Growing uncertainty and concern in decisions about cancer surgery

Where the genetic test result was contrary to expectation, the participants appeared to feel less certain and more concerned about the decision not to have a double mastectomy.

Group 2 (no expectation of inheritance, VUS: incongruence)

Two of the participants in Group 2 responded positively and with interest to the genetic test at time 1. However, once the VUS result was known, they both seemed unsure of about whether they should be having a double mastectomy or not.

At time 1, Susan is hopeful about the test saying, ‘if it helps them to understand my own treatment I would be very pleased.’ At time 3 however she is uncertain about what the result will mean for her treatment or who will make the decision about surgery:

‘I think it means that they (…) are probably not going to recommend any prophylactic surgery, but (the genetics health professional) sort of said probably, she didn't say they are definitely not going to, and I don't really know who makes that decision’. (T3)
Kate is unconcerned at the outset saying:

‘I feel really interested and curious (about genetics) … I don’t feel anxious … because it just doesn’t have implications for me.’ (T1)

At time 2, Kate is floored by the potential implications of the genetic test and is left feeling uncertain about what the result might mean for her treatment:

‘My understanding is that (the test) affects the operation in as far as there would be a discussion about whether I have (...) a double mastectomy in order to prevent the risk of it coming back but that’s all I’ve been told. I don’t know if it affects the other treatment or not.’ (T2)

At time 3, having learnt that a VUS has been detected in her sample, Kate questions the surgeons several times about what type of surgery she needs:

‘I said to (the breast surgeon) about this other mutation and he said, no, no evidence for surgery, and again I raised the question of having a double and he said, again, we only do that for proven medical reasons, we would only do that for your own mental well being, and that is again something that if you tell us that is important, we will do that for you. (...) What the plastic surgeon said to me is I can come back at any time and do something different.’ (T3)

Part of the reason for Kate’s continued uncertainty may be the mixed messages she is given by the surgeons who, on the one hand are telling her that she doesn’t need a double mastectomy and on the other are telling her she can have surgery if she wants to.

**Group 3 (expectation of inheritance, no variant: incongruence)**

All three participants in Group 3 had prepared themselves for a double mastectomy but the genetic test result indicated that such radical surgery would not be necessary.

At time 1, Fiona faces the decision about whether to have a lumpectomy or a bilateral mastectomy. She explains the importance of her breasts for her body image and sexuality and her sadness at the potential loss. In the extract below she tries to balance this loss with the risk associated with a genetic susceptibility:

‘I really like my breasts, (...) now and just as I found that, I’m going to lose them and I’m worried that I won’t have any sensation in them anymore. (...) They are saying that the genetic testing would have a lot to do with that (decision). It might point to them advising me to do that.’ (T1)
Fiona continues to feel uncertain about the surgical decision for several days after the genetic test result:

‘I stalled (the decision), I didn't immediately go "Right that's it that’s decided now". (…) Probably in the back of my mind I had decided I was only going to have the single mastectomy, but I didn't really, I didn't make my mind up immediately there and then.’ (T3)

Amanda had prepared herself for a double mastectomy:

‘With the genetics showing that we are more likely then we can make an informed decision about what needs to be done.’ (T1)

Her immediate concern on learning the genetic test result is that the cancer may have spread during the three weeks she has waited for the result. She appears to lack certainty about the surgical decision and there is a hint that she may have regrets about having had genetic testing in the extract below:

The breast surgeon said, ‘We don't need to do the double mastectomy. (…) If I had had to go ahead with the surgery not having the result I probably would have ended up having a double mastectomy (…) I suppose if it had been an aggressive sort it would have been necessary to do that and then I would have said "well OK, I have had to have a double mastectomy". (…) If I had been younger that could have been quite traumatic. At my age it is not such a problem.’ (T3)

At time 1 Sarah is confident that the genetic test would help with decision-making and expects to need a double mastectomy:

‘(The genetic test) will obviously affect the way I make a decision about my treatment and …there’s a positive to come out of it.’ (T1)

By time 3 however, she seems uncertain about what the right surgical decision is:

‘I’ll just have to mull over the decisions … make sure I’m making the right decision based on this (genetic test) result, depending on whether the consultant will advise me for less radical surgery … I’ll just have to see really.’ (T3)

A little later, as she reflects on what might have happened if a pathogenic variant had been found, there is a hint of regret that the test did not provide her with answers to help with decision-making and uncertainty about how to proceed:

‘I suppose if it had come back positive then on Monday it might have made decisions a bit easier because then you know you would know, I suppose it would help to map out your future a bit more because you could make a
7.5.4.2 Discussion of groups (Theme C)

Congruence between the prior expectation of inheritance and the result heightened certainty about how to proceed with surgery. Participants who expected hereditary cancer and received a pathogenic variant result (Group 1) appeared increasingly confident in the decision to have a double mastectomy. These participants also expressed the wish to proceed with bilateral salpingo-oophorectomy, even against the advice of their doctors.

Similar confidence in surgical decision-making was seen in the participants in Group 4. Their expectation that the cancer was not hereditary was confirmed by the genetic test result. Once the result was known, these participants felt confident in the decision not to have radical surgery and expressed relief at the ability to move on with the next stage in their treatment. A recent quantitative study found that patients who did not expect a hereditary cause for their cancer, displayed higher perceived personal control once the result was disclosed than those who overestimated their prior risk (Bredart et al., 2016).

For the participants in Group 2 who did not expect hereditary cancer but received a VUS result, the genetic test results were incongruent with prior expectations of inheritance. Several studies have highlighted the level of misunderstanding and misinterpretation amongst patients in whom no variant is detected (Hallowell et al., 2002; Maheu & Thorne, 2008; Richter et al., 2012; Vos et al., 2008). Primary care doctors and breast surgeons have also been shown to have poor understanding of the meaning of a VUS result (Eccles et al., 2015; Richter et al., 2012), which may have contributed to uncertainty about surgical decision-making amongst these patients.

Incongruence between the expectation of inheritance and the genetic test result also led to uncertainty and concern for those in Group 3. These patients expected hereditary cancer but were found not to have a variant. They had prepared themselves for a double mastectomy and when this was no longer required they are left unsure of how to proceed. Many patients who opt for a contralateral mastectomy do not have a pathogenic variant (Davies et al., 2016). As discussed in Theme A, the persistent feeling of hereditary cancer amongst cancer patients with a family history can lead to concerns about the risk of breast cancer recurrence (Hamilton & Kopin, 2013) and may explain the responses of the patients in Group 3 to the ‘normal’ genetic test result.
7.5.5. Theme D. Impact of interactions with health professionals on feelings of satisfaction and confidence

7.5.5.1. Results (Theme D)

For 10 of the participants, interactions with health professionals impacted on feelings of satisfaction and confidence about their experience. The participants who were found to have a pathogenic variant (Group 1) were consistently disaffected; those who were found to have a VUS (Group 2) grew increasingly disaffected and those who were found not to have a variant (Groups 3 and 4) consistently expressed feelings of confidence and satisfaction.

7.5.5.1.1. Consistently feeling disaffected

Group 1 (expectation of inheritance, pathogenic variant: congruence)

Both participants in Group 1 had previously had negative experiences with health professionals and, perhaps as because of this, had low expectations from the outset and remained disaffected throughout.

Tina is both challenging in and challenged by her interactions with breast health professionals. The interactions leave her feeling frustrated and disaffected. Her apparent satisfaction with her initial meeting with the breast surgeon is countered with assertiveness, perhaps to remind the surgeon that they are equal partners in decision-making:

‘(The breast surgeon) understood me, I understood him (…) we just didn’t mince words. It was what I wanted, I didn't want somebody to tiptoe around. I think you just go in with a certain confidence and (…) you've just got a job to do so its in, his professional opinion, he knows what he's doing, we'll go in shake hands, we say hi and I can look him in the eye.’ (T1)

Her frustration is evident at time 2 however when a biopsy result delays her surgery:

‘I phoned in and the biopsy was supposed to be ready (…) and then it is not going to happen, (…) I can’t afford not to work, and you know, the nurses are lovely, but it is like, you have got a job every single day of your life, when you only have jobs certain days of the week and they are non-changeable, that is how you earn your living’ (T2)

By time 3 she again expresses her dissatisfaction when a new breast surgeon questions her decision to have a bilateral mastectomy:
'It threw me when (the breast surgeon) shouldn't have done and he should have had a nurse in there which you can go back to (...). Because you never see the same consultant and you need to have one person who knows that history and it shouldn't be just patients it should be a nurse'. (T3)

Nicola is angry from the outset that genetic testing was not offered to her when her mother died. Her comments about the response of the oncology health professional to her anger at time 1 suggest that she has low expectations of the health professionals’ interactions:

‘The (breast) consultant just sat there really, he's a man isn't he, he doesn't know what to do and they see it every day don't they so I think he just sort of took a step back while I was going on’. (T1)

At time 3, her interaction with the oncologist about the implications of the genetic test result again leaves her feeling dissatisfied and unsupported:

‘(The oncologist) had not read it so she didn't know anything about it. (...) I am sure she is very good at her job but is not very talkative. She just said it into a corner... She just said, patient BRCA1 chosen bilateral mastectomy and that was it.’ (T3).

7.5.5.1.2. Growing increasingly disaffected

Group 2 (no expectation of inheritance, VUS: incongruence)

The participants in Group 2 were floored by the diagnosis and expressed complete confidence in the health professionals at time 1. By time 3, however, they were left feeling depersonalised, vulnerable and let down.

At time 1 Susan talks of handing over total responsibility to the oncology health professionals, even though this is out of character for her:

‘I've been very passive in some ways which is a bit unlike me. (...) I feel in good hands. I'm with people who really know more than I would find out for myself from the internet. (...) so I haven't really asked too many questions. (T1)

Following genetic counselling, she continues to feel reassured and confident. However, dissatisfaction with her interactions with the oncology team are becoming evident:
‘I found (the genetics health professional) very approachable and she had plenty of time on her hands, because it is so frantically busy downstairs (in oncology) (...) you always feel like you are holding the next person up half hour if you ask a question, because they are always running late.’ (T2)

This dissatisfaction is echoed at time 3 when her reflections on her experiences overall in genetics and breast surgery /oncology suggest feelings of abandonment and disaffection:

‘There is a little bit of a sense in this that there isn’t really anyone responsible for my care I suppose, you know I can go for my counselling but then she’s a counsellor, I see my Oncologist but they are really, really rushed. I see the nurses when I have my chemo but they are also quite rushed and you don’t see the same people all the time’ (T3).

Similarly, Kate is initially bolstered by the confidence of the breast surgeon:

‘(The breast surgeon) said (...) I can treat this, and it is treatable, and I have the confidence in it. I don’t know if he actually said that but he made me feel like, I felt confident in what he was telling me.’ (T1)

Kate remains satisfied with her interactions with the breast team but feels increasingly let down by the genetics staff:

‘I wasn’t sure that (the genetics health professional) appreciated what that meant in the context of everything I’ve been through (...) She was quite matter of fact (...) I felt like saying do you have any idea what that feels like, you’re facing that with all of this?’ (T2)

Kate appears to feel vulnerable, alarmed and unsupported by the clinical and impersonal nature of the genetics interaction. Her dissatisfaction continues into time 3 when her experience of receiving the result leaves her feeling let down and disappointed:

‘If (...) (genetics) could have made me an appointment for a time that I knew I would be well enough, (...) (it) would have empowered me in that process, because I felt quite disempowered.’ (T3)

At time 1, Angela reveres the genetics health professional so much that she worries that she is going to get into trouble for time wasting. She is reassured once they have met, describing her interaction as ‘absolutely brilliant’. However, at time 3, she fears she has been ‘forgotten’ by the breast team and, when the call about her genetic test result does not arrive on the day she expects it, she is left feeling worried, despondent and let down by health professionals in general:
‘I came home and I was really, really down that I hadn’t got the (result) (...) I didn’t want to talk to anybody (...) I just want to know.’ (T3)

7.5.5.1.3. Consistently feeling confidence and satisfaction

The participants in Groups 3 and 4 expressed consistent feelings of confidence and satisfaction in their healthcare interactions.

**Group 3 (expectation of inheritance, no variant: incongruence)**

For two of the participants in Group 3 there is a sense of self-preservation in the confidence and satisfaction they express. It seems as if by adopting the role of the ‘good’ patient they feel more confident in their relationships with health professionals.

Fiona consciously modifies her behaviour to please the staff:

‘I was laughing with all the nurses and I was being really cheerful and appreciative that they (the breast nurses) were there and they were doing the routine tests for me, getting the best out of them by being friendly’ (T1).

Her need to be liked is perhaps influenced by her mother’s negative experience which she alludes to in time 1, ‘I haven’t got a good memory of hospitals (...) they were being horrible to her’. In time 3 she continues to express confidence and satisfaction but also to worry about how she is coming across:

(The breast nurses and surgeons) ‘have been really great but I always feel aware that, you know how much time should I demand, am I pushing the boundaries, (...) I want to keep them sweet and I want them to keep on being helpful and liking me so I don’t want to be a complete pain in the butt, so I don’t know, maybe I am being too docile.’ (T3)

Her words suggest that she is fully aware of the power that the health professionals have and tries to be a ‘good patient’ in order to maintain her confidence and continue to receive good care.

As a former health professional, Amanda appears to manage her feelings about the health professionals by aligning herself with them. In time 1 her compliments about all the health professionals she has interacted with sound like a well-rehearsed platitude:

‘I’m getting all of this fantastic treatment you know I couldn’t be in better hands and it’s great that these people are taking the trouble to check things out.’ (T1)
Amanda is aware of the pressures on the health care team and arguably of the type of behaviour that is likely to be more popular with the health professionals. This may explain why in time 2 she blames herself rather than the genetics health professional when she doesn’t fully understand her chances of developing a further breast cancer:

‘I’m sure that’s not because (the genetics health professional) didn’t explain it, it’s probably because I just didn’t pick it up.’ (T2)

Her apparent confidence in health professional interactions is also evident in time 3 when she receives the genetic test result:

(The genetics health professional) ‘is a lovely lady, she is so positive, she is really young.’ (T3)

There is a sense for both participants in Group 3 that they are satisfied with the health professionals at all time points but that they have to work hard to maintain that level of satisfaction.

Group 4 (no expectation of inheritance, no variant: congruence)

When they are diagnosed, the participants in Group 4 placed their faith entirely on the health professionals, unquestioningly accepting the advice they were given. Although more confident in their own knowledge by time 3, they continued to express complete confidence and satisfaction in the health professionals.

In this extract, Carla is talking about the information she was given at the initial consultation. Whilst appreciating the breast surgeon’s approach she also emphasises her need to be able to put her trust in the health professional team:

‘The (breast) doctor was fantastic (…) it comes to a point where it’s all too much and it’s like, I don’t need to know all of that, I just need to know that you are going to make me better.’ (T1)

She hands over all responsibility as demonstrated by her comment at time 3 that she has not looked up information as she would normally:

‘I haven’t done the usual looking into it, but then the (genetics) consultation that I had up (…) was so thorough and she explained it so brilliantly, (…) so clearly and so comprehensively that actually I didn’t really have any outstanding questions anyway.’ (T3)
Sophie passively and unquestioningly trusts the health professionals to do the best for her:

‘I can only be led by my (oncology) clinicians and (breast) surgeons about my treatment and what is best for me.’ (T1)

Becoming informed by the genetic counselling is helpful for Sophie and she refers to her satisfaction with the communication at times 2 and 3:

‘I was feeling quite good about…speaking to somebody (in genetics) and getting the facts rather than getting myself stewed up and worrying about things unnecessarily.’ (T3)

Andrea too has absolute confidence in the oncology health professionals, seeing no choice but to accept the genetics referral even though she is afraid and anxious:

‘It does worry me a lot that if I have got, you know, the gene but (...) I am so glad that (the oncologist) did because I think if she hadn’t of done, if that had been missed ... I would never stop anything and not do anything, just, you go along with it because you have to, there is no other option is there?’ (T1)

She continues to trust implicitly in the health professionals at time 2 even as she worries about whether the cancer has spread during the wait for the genetic test result:

‘(The oncology health professionals) know what they are doing, they do, and I need to be guided, because I don’t know.’ (T2)

When reflecting on the whole process, her comments again suggest trust and satisfaction with the health professionals she has interacted with:

‘It has been very good from word go. It has been done very quick from the diagnosis and they have been ever so good at (hospital), really good, very helpful.’ (T3)

7.5.5.2. Discussion of groups (Theme D)

It is perhaps not surprising that those with a pathogenic variant or a VUS (Groups 1 and 2) were dissatisfied with the health professional interactions. The outcome of the genetic test would not have been what they had hoped for, even if, for those in Group 1, it was expected. Both participants in Group 1 were aware of the imbalance of power and knowledge that is implicit in the health professional-patient relationship (Mechanic & Meyer, 2000) and talked about needing to ‘fight’ for what they wanted.

Participants in Group 2 with no prior expectation of inheritance felt vulnerable and afraid at the outset, leaving them with a strong sense of trust and confidence in the
oncology health professionals who were caring for them. Satisfaction with health professionals seemed to diminish for those in Group 2 once the VUS result was known. For patients with a life-threatening disease such as cancer, there is little choice but to trust in those responsible for their care (Mechanic & Meyer, 2000; Salkeld, Solomon, Short, & Butow, 2004). Satisfaction with communication is increased amongst cancer patients where health professionals are trusted (Butow, Dowsett, Hagerty, & Tattersall, 2002; Eggly et al., 2006). The apparent loss of confidence in health professionals at time 3 by those in Group 2 appears to be due to the VUS genetic test result.

Both groups that received a no variant detected result (Group 3 and Group 4) maintained their confidence and satisfaction with the health professionals throughout the process. Those in Group 3 appeared to be aware of the importance of being a ‘good’ patient, attempting to be pleasant, uncomplaining and grateful to the health professionals. Their satisfaction with the health professionals’ interactions may have reflected their attempts not to complain.

Participants with no prior expectation of inheritance who received a result showing no variant had been detected (Group 4) experienced feelings of confidence in the ability to move forward with cancer treatment and relief of responsibility for the family, reflected in the sustained feeling of confidence and trust in the health professionals over time.

7.6. SUMMARY OF THEMES

Feelings of certainty about inheritance (Theme A) were affected by prior views and family experiences. Participants for whom genetic inheritance was part of their prior representation of disease (Groups 1 and 3) continued to feel that the cancer was hereditary, regardless of the genetic test result. Those who had no prior family experience of cancer (Groups 2 and 4) became uncertain about the possible cause for the cancer once the possibility of inheritance was raised, suggesting that the prior views had a greater influence on feelings about inheritance than the information gained during genetic counselling.

Feelings of responsibility for the family (Theme B) were also affected by prior views and family experiences. Amongst those who believed the cancer to be hereditary (Groups 1 and 3), feelings of responsibility for continuing the family cancer legacy persisted, even when no variant was identified. Amongst those who believed the
cancer not to be hereditary, genetic counselling raised feelings of responsibility for the family (Groups 2 and 4). When the result showed that no variant had been found, feelings of responsibility for the family were relieved, even when a VUS was identified.

Confidence and certainty about surgical decision-making appeared to be influenced by congruence or incongruence between prior expectations of inheritance and the genetic test result, rather than by the outcome of the genetic test (Theme C). Where the genetic test result confirmed prior expectations (Group 1 and Group 4), participants felt confident to make decisions and relieved at being able to move on with their treatment. Where there was incongruence between expectations of inheritance and the genetic test result (Group 2 and Group 3), participants were left with uncertainty and concern about the best way forward and questioned the decisions being made. Responses to incongruent genetic test results were emotional, nuanced and influenced by personal views and family experiences.

Confidence in health professionals and satisfaction with health professionals’ communication appeared to be related to the outcome of genetic testing, regardless of prior views and expectations (Theme D). Those without a pathogenic variant or VUS (Group 3 and Group 4) were satisfied and confident in genetics and oncology health professionals. Those with a pathogenic variant or VUS (Group 1 and Group 2) were dissatisfied. A previous study found that levels of trust in oncologists were highest amongst patients who had completed treatment compared with those in treatment (Taha, Matheson, Paquet, Verma, & Anisman, 2011). It seems that trust in health professionals may also be higher amongst patients who feel they have ‘completed’ their genetic testing rather than those left with an ambiguous or unwanted result.

7.7. CLINICAL IMPLICATIONS

As genetic testing becomes further integrated into mainstream oncology, pre-test information provision is likely to become more routine, possibly delivered via pamphlets or websites (Quinn et al., 2016; Sie et al., 2014), in group education sessions (Benusiglio et al., 2017; Calzone et al., 2005) or by oncology health professionals (George et al., 2016). Post-test genetic counselling to address the implications for the patient and the family and provide support will be important for all patients who are found to have a pathogenic variant. However, as this study
demonstrates, some patients may need additional time, counselling and support throughout the process of genetic testing.

For patients who do not expect a hereditary cause for their cancer and who are tested based on their personal diagnosis, communication of information within oncology may be sufficient. Low-key communication of minimal information may avoid raising concerns and uncertainty about inheritance and the implications for the family. If no variant is identified, post-test genetic counselling may well not be needed as these findings suggest that congruent results would enable patients to move on with their treatment without further concern. If a VUS is identified however, these patients may find it helpful to be referred for post-test genetic counselling to help them to understand the meaning of the result.

For patients who do expect a hereditary cause for their cancer, minimal, non-personalised information provision is unlikely to be adequate, regardless of the eventual genetic test result. Exploring prior knowledge of a cancer family history, the cancer experiences of family members, prior views and expectations of inheritance may help to facilitate understanding, decision-making and adjustment. Pre-test assessment of these factors may help with identification of patients who might benefit from post-test support and counselling even if no variant is identified.

As an increasing number of patients undergo genetic testing to determine cancer management, it will be important to ensure that decisions about contralateral surgery and risk-reducing bilateral salpingo-oophorectomy are made on the basis of risk, rather than emotion. Oncology and genetics health professionals need to understand, interpret, explain and manage a VUS result. Clear reporting of VUS results from laboratories and the provision of education and support from genetics health professionals will be essential in facilitating appropriate surgical management of patients who do not have a pathogenic variant as well as those who do. These findings suggest that improvements are needed in the way in which oncology and genetics health professionals communicate about hereditary cancer with cancer patients. For oncology health professionals, finding a balance between preparing patients for the potential implications of testing for themselves and their families without causing undue alarm will be challenging, as will identifying those who would benefit from further counselling and support. Within genetic counselling, there needs to be greater focus on assessing and meeting the psychological and supportive needs of
newly diagnosed patients than on communicating biomedical education that is has become the dominant model within cancer genetics (Paul et al, 2015).

7.8. CONCLUSIONS

Amongst patients with newly diagnosed breast cancer undergoing genetic counselling and testing, prior views of inheritance and family experiences affected feelings of certainty about the cause of the cancer and feelings of responsibility for the family, regardless of the genetic test result. Congruence and incongruence between prior expectations of inheritance and the outcome of genetic testing affected feelings of certainty and confidence about the decision to have a contralateral risk-reducing mastectomy. The outcome of genetic testing affected feelings of confidence in genetics and oncology health professionals and satisfaction with health professionals' communication. Eliciting expectations of inheritance and prior views and experiences may help to identify patients who would benefit from specialist genetic counselling and support, regardless of the result.
8.1. INTRODUCTION

This thesis investigated health professionals’ communication with breast and ovarian cancer patients about genetic testing and hereditary cancer management. Discussions aim to facilitate decision-making amongst patients about whether to have a genetic test and how to manage existing and future cancer if a pathogenic variant is detected. Such decision-making requires the patient to be fully informed. For example, before a patient with breast cancer makes decisions about having a double mastectomy at the time of definitive cancer surgery, it is essential that she has access to information about the risks, consequences and limitations of surgery in the context of her diagnosis. It is also helpful for her to be aware of the potential psychosexual impact of surgery and the risks, benefits and limitations of breast surveillance.

The purpose of the research was to identify areas of good practice and areas for improvement to inform future practice, policy and research in this area as genetic testing shifts from the clinical genetics setting to mainstream oncology.

Gaps were discovered between the information that should be communicated according to guidelines and expert opinion, the information communicated by health professionals and recalled by patients and the understanding and experiences of patients with newly diagnosed breast cancer who undergo genetic testing shortly after diagnosis.

The findings were synthesised according to the objectives of the thesis into i) information that should be communicated and the method of communication, ii) information communicated by genetics health professionals, iii) information recalled by patients and their families and iv) patients’ experiences and understanding of communication. The limitations of the thesis and the implications of the findings for practice, policy and research are discussed.

8.2. SYNTHESIS OF FINDINGS

The key findings from each study were organised according to the thesis objectives. The main findings within each of these objectives are summarised below and shown in Table 8.1.
Table 8.1 Summary of thesis findings

<table>
<thead>
<tr>
<th>Study</th>
<th>Information to be communicated</th>
<th>Information communicated</th>
<th>Information recalled</th>
<th>Experience and understanding</th>
</tr>
</thead>
</table>
| Scoping review | • Need for treatment-related information for patients undergoing genetic testing shortly after diagnosis  
• Treatment-related information close to diagnosis. Family-related information at a later time  
• Communication via written/digital/group acceptable - no adverse effects | • Information about risks for patient infrequently communicated |                     | • Patients misunderstand/misinterpret VUS results  
• Overwhelming experience for patients when unprepared/unsupported |
| Study 1       |                                 |                          |                     | • Accuracy of recall low amongst patients and relatives  
• Recall about genetic testing more accurate than recall about hereditary cancer management |
| Study 2       | • High level of agreement  
• Need for genetic testing and hereditary cancer management information  
• No need for treatment-related information  
• Information required before genetic testing and repeated if a pathogenic variant is detected |                     |                     |                                      |
| Study 3       |                                 |                          | More information communicated about genetic testing than hereditary cancer management  
• Half of information |                                      |
Genetics guidelines mainly focused on recommendations for at-risk women. Cancer guidelines rarely referred to genetic testing or hereditary cancer management. Expert opinion about the information to be communicated reflected genetics guidelines. Health professionals frequently communicated information about genetic testing. However, information for cancer patients about hereditary cancer management was infrequently communicated. Patients recalled information about genetic testing, however information

<table>
<thead>
<tr>
<th>Study 4</th>
<th>Study 5</th>
</tr>
</thead>
</table>
| **Consistency between genetics clinical guidelines, expert health professionals and cancer patients about information to communicate**  
- Few recommendations in cancer guidelines  
- Genetics guidelines mainly relevant for cancer patients and at risk women  
- Some recommendations made by cancer guidelines not present in genetics guidelines | **Prior views/family experiences affected certainty about inheritance/feelings of responsibility for family**  
- Congruence between prior expectations of inheritance and result affected feelings of certainty/confidence about surgery  
- Confidence/satisfaction with communication affected by genetic test result |
| **Health professionals communicated less than half information needed**  
- More information communicated about genetic testing than hereditary cancer management  
- Irrelevant information communicated |  

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needed was communicated  
- Much of the communicated information was not relevant  
- Frequent recall of information about genetic testing following pre-test genetic counselling  
- Infrequent recall of information about hereditary cancer management following post-test counselling
about hereditary cancer management was infrequently recalled. Prior views, family experiences and congruence between expectations of inheritance and the genetic test result, influenced understanding amongst newly diagnosed breast cancer patients regardless of the information communicated during genetic counselling.

8.2.1. Information that should be communicated and the method of communication

The focus of the genetics guidelines was on at-risk individuals. Few references were made by genetics guidelines about the specific information needs of cancer patients or about the way health professionals should communicate with cancer patients (Study 4). The lack of reference to cancer patients may in part be due to the high level of professional and public awareness of hereditary breast cancer. The disclosure of a genetic predisposition to breast and ovarian cancer by Angelina Jolie in 2013 has resulted in steadily increasing demand for genetics referral from women with a family history of breast cancer, directly impacting on cancer genetics services worldwide (Troiano, Nante, & Cozzolino, 2017). Over recent years there has been a dramatic shift in the pattern of BRCA genetic testing between 2004 and 2014 from individuals with cancer to those without cancer has been highlighted in a recent US study which showed that the number of women without a personal history of breast or ovarian cancer undergoing BRCA1 testing increased from 24.3% to 61.5% (Guo et al., 2017).

Breast and ovarian cancer guidelines did make recommendations for communicating with cancer patients, however they rarely referred to hereditary cancer (Study 4). Much work has been undertaken on the field of oncology to improve health professionals’ communication with cancer patients (Cox, Jenkins, Catt, Langridge, & Fallowfield, 2006; Fallowfield & Jenkins, 1999). As a result, cancer guidelines frequently referred to the specific communication needs of cancer patients and oncology health professionals are required to undergo regular communication skills training in many Western countries (Moore, Rivera Mercado, Grez Artigues, & Lawrie, 2013; Stiefel et al., 2010). The absence of recommendations in cancer guidelines about hereditary cancer is most likely explained by the prevailing model of practice, whereby all genetic testing has been undertaken within specialist genetics services. Low levels of knowledge and confidence in understanding of genetics amongst non-genetics health professionals has consistently been identified as a major barrier to genetics referral (Alsop et al., 2012; Demsky et al., 2013; Mikat-Stevens, Larson, & Tarini, 2015; Skirton, O’Connor, & Humphreys, 2012).
Patients need information about genetic testing and hereditary cancer management to enable decision-making for themselves and to inform relevant family members (Study 2, Study 4, scoping review). The views of expert genetics and cancer health professionals and service users about the information patients need reflected the recommendations made by genetics guidelines (Study 2). The impact of guidelines on the perceived information needs of patients and health professionals is highlighted by the different information needs about the impact of genetic testing on treatment between UK and Australian participants. In Study 2, information about the impact of genetic testing on current and cancer treatment was not judged to be a key message. However, the scoping review identified a need for information about the potential impact of genetic testing on the risk of further cancers and surgical and treatment options in two Australian studies (Gleeson et al., 2013; Meiser et al., 2012). In the UK, NICE guidelines recommended against genetic testing shortly after diagnosis due to the lack of evidence of the psychological impact of such testing (National Institute for Health and Care Excellence, 2013). The Australian guidelines however did recommend genetic testing shortly after diagnosis and referred to options for contralateral mastectomy and chemotherapy options for BRCA carriers (https://guidelines.canceraustralia.gov.au/guidelines/gene_mutation/ch01s04.php. Accessed 30.12.17)

In summary, genetics and cancer guidelines made limited recommendations about information that should be communicated or the method of communication about hereditary cancer for cancer patients. The views of health professionals and patients about the information needs of patients reflected the guidelines.

8.2.2. Information communicated by health professionals

Health professionals frequently communicated information about the reliability, limitations and informativeness of the genetic test, options for prevention, early diagnosis and surveillance, the potential risk and benefits of genetic testing for the individual and the family and cancer risks, including penetrance and variable expression (Study 3, Study 4). Previous research has also found that genetics health professionals are good at communicating about genetic testing (Butow & Lobb, 2004; Butow, Lobb, Meiser, Barratt, & Tucker, 2003)

Study 3 found that significantly more genetic testing information was communicated than hereditary cancer management information in consultations. In clinical protocols and patient information leaflets a higher percentage of the information communicated
was about genetic testing than hereditary cancer management (Study 4). Information messages that were directly relevant to cancer patients were infrequently communicated in Studies 3 and 4, about the risk of contralateral breast cancer and the timing, risks and benefits of contralateral mastectomy and the benefit of testing for patients with cancer were infrequently communicated. Previous studies have identified that patients did not always receive information about the risk and management of future cancers (Butow & Lobb, 2004; Pieterse, van Dulmen, van Dijk, Bensing, & Ausems, 2006). However, as identified by the scoping review, few previous studies have examined the content and process of health professionals’ communication with cancer patients in as much detail as the studies in this thesis.

This thesis provides evidence that the verbal and written communication by genetics health professionals does not sufficiently meet the information needs of cancer patients as judged by expert health professionals and service users. To the author’s knowledge, this thesis provides the first evidence that hereditary cancer information is less well communicated by genetics health professionals than genetic testing information.

8.2.3. Information recalled by patients and relatives

Accuracy of recall of information amongst patients and relatives was lower for hereditary cancer management information than genetic testing information (Study 1). Previous studies have demonstrated an increase in patients’ knowledge following genetic counselling (Christie et al., 2012; Randall, et al., 2001; Scherr et al., 2016; Bredart et al., 2016; Benusiglio et al., 2017; Mancini et al., 2006; Quinn et al., 2016) However, to the author’s knowledge no previous studies have identified differences in recall between genetic testing information and hereditary cancer information.

This thesis found that following pre-test genetic counselling, patients recalled most of the information that was communicated about genetic testing. Information that was infrequently communicated about sharing genetic risk and surveillance advice with relatives and the legal and ethical implications of testing was also infrequently recalled (Study 4). Following post-test genetic counselling, more genetic testing information was recalled than hereditary cancer information (Study 1). The hereditary cancer information that was frequently communicated by health professionals was also frequently recalled by patients. However, patients infrequently recalled information about the risks, benefits and limitations of breast surveillance, advice to discuss breast reconstruction with a specialist surgeon and the psychological and sexual consequences of breast surgery
were infrequently recalled (Study 4). The information recalled by patients appears to reflect the information communicated by health professionals. Genetic testing information is arguably more straightforward and involves fewer numbers and less uncertainty than hereditary cancer management information. Information about inheritance and genes is routine for genetics health professionals to communicate and as a result may be delivered using a didactic teaching approach with the aid of charts and diagrams which can aid recall (Houts, Doak, Doak, & Loscalzo, 2006). Information about hereditary cancer management however is more personalised and nuanced, requiring the information to be put into context for the individual and family. Information about the limitations of screening or surgery for example may be considered negative and alarming.

Multiple factors affect the information recalled and retained following communication by health professionals, as highlighted by the studies identified in the scoping review (Hallowell et al., 2002; Maheu & Thorne, 2008; Vos, Gomez-Garcia, et al., 2012; Vos et al., 2008). To some extent at least, patients’ recall appears to be dependent on the information communicated.

This thesis provides evidence that, as judged by expert opinion, patients did not recall sufficient information to make informed decisions for themselves or to inform their relatives, especially about hereditary cancer management. To the author’s knowledge, this thesis provides the first evidence that hereditary cancer information is less well recalled than genetic testing information.

**8.2.4. Patients’ understanding and experiences of communication**

Feelings of certainty and concern in response to genetic testing shortly after diagnosis were influenced by prior views, family experiences and the match (congruence) between prior expectation of inheritance and the outcome of genetic testing, rather than information gained from genetic testing or genetic counselling. Patients’ confidence in health professionals and satisfaction with health professionals’ communication was influenced by the outcome of the genetic test.

Although several qualitative studies have identified feelings of confusion, uncertainty and anxiety following genetic testing (Maheu & Thorne, 2008; Hallowell et al., 2002; Vos et al., 2008), few previous studies have explored experiences of genetic counselling and genetic testing shortly after diagnosis. One study examined the experiences of patients tested without pre-test genetic counselling (Augestad, Høberg-Vetti, Bjorvatn, &
Sekse, 2017). The research presented in Study 5 provides the first evidence of the impact of genetic testing over time in patients with newly diagnosed breast cancer.

As for those tested after completing cancer treatment, understanding of the genetic test result was moderated and amplified by personal experiences of cancer (Hallowell, Foster, Eeles, Ardern-Jones, & Watson, 2004) and feelings about risk management were based on interpretation of the meaning of genetic test results rather than the information communicated during genetic counselling (Pieterse et al., 2011; Vos, Oosterwijk et al., 2012). Importantly, Study 5 also found that it was the congruence between the expectation of the result and the actual result rather than the result itself that had an impact on patients’ responses and feelings of confidence and certainty about decision-making. This finding is important for identifying patients who may need additional counselling and support regardless of their genetic test result.

The scoping review found that genetic counselling did not influence patients’ risk perception and that ambiguous results were misunderstood or misinterpreted (Hallowell et al., 2002; Maheu & Thorne, 2008; Richter, Graham, Haroun, Eisen, & Warner, 2012; van Dijk et al., 2004; Vos, Gomez-Garcia, et al., 2012; Vos et al., 2008).

8.3. LIMITATIONS OF THE THESIS

The limitations of this work have been addressed in the relevant chapters. The main limitations of the thesis involve selection of participants in population studies, developments in genetic testing and cancer management during the course of the thesis and changes in clinical practice during the course of the thesis.

This work only considers genetic testing of female patients with breast or ovarian cancer who underwent genetic testing for variants in the BRCA1/BRCA2 genes. These findings do not necessarily generalise to at-risk individuals, men with BRCA1/BRCA2-related cancer or patients tested for other breast and ovarian cancer pre-disposing genes.

Over the course of this work, there have been significant advances in research into targeted cancer treatment and techniques for genetic and genomic testing. Following the approval of PARP inhibitor olaparib in ovarian cancer patients and as a result, genetic testing of the BRCA1/BRCA2 genes is now being offered in mainstream
oncology to guide management (George et al., 2016; Percival et al., 2016). Within clinical genetics services, many patients are now tested for multiple intermediate penetrance breast and ovarian cancer genes in addition to the high risk \textit{BRCA1} and \textit{BRCA2} genes. However, \textit{BRCA1}/\textit{BRCA2} remain the most common breast and ovarian cancer predisposing genes. Whole genome testing is available in some areas in the research context. Testing of the whole genome is likely to be offered within clinical practice in the future which will result in further complexities that will need to be addressed (Davies, 2017). The method developed during this thesis could be applied to research into communication about other cancer genes and other types of hereditary cancer. However, the research reported in this thesis only investigated genetic counselling and testing of female cancer patients about variants in the \textit{BRCA1}/\textit{BRCA2} genes.

This thesis included primary and secondary data analysis. Some studies included data gathered in 2006-2008 as part of the Family Communication Study (section 1.8.5.5). There have been changes to hereditary breast and ovarian cancer guidelines and genetic counselling practice since that time.

\textbf{8.4. IMPLICATIONS FOR PRACTICE AND POLICY}

The findings from this thesis highlight the need for changes in six areas of practice and policy in communication about hereditary cancer to meet the communication needs of cancer patients: (i) guidelines should be updated so that they focus on the information and communication needs of cancer patients, including those undergoing genetic testing to inform management, (ii) the information leaflets used within clinical genetics departments should be standardised to reflect guidelines and include relevant information about genetic testing \textit{and} hereditary cancer management, (iii) health professionals should communicate the information patients need and avoid unnecessary information provision, (iv) close multidisciplinary working and streamlined pathways between oncology and genetics are needed to ensure access to genetic counselling, psychological support and specialist oncology and surgical expertise, (v) genetic counselling should focus less on biomedical information-giving and more on helping patients to understand the information in the context of their personal and family circumstances, providing psychosocial support and facilitating family communication and (vi) genetics and oncology health professionals would benefit from training in communicating with cancer patients about hereditary cancer.
i. **Guidelines:** Guidelines do not reflect practice for patients undergoing genetic testing shortly after diagnosis. This research found that guideline recommendations about genetic testing information were mostly translated into expert opinion, health professionals’ communication and patients’ recall. However, information about hereditary cancer management targeted at cancer patients was lacking in guidelines. Those hereditary cancer management recommendations that were made by guidelines were inconsistently translated into practice. The number of patients undergoing testing is likely to increase (Childers, Childers, Maggard-Gibbons, & Macinko, 2017). There is an urgent need to review and update guidelines so that they are relevant for cancer patients and accessible by oncology and genetics health professionals.

ii. **Patient information leaflets:** The information leaflets used in clinical practice should provide targeted information about hereditary cancer management as well as genetic testing. Leaflets vary in the quality and information they provide (Sustersic, Gauchet, Foote, & Bosson, 2017). However, the use of standardised leaflets has been shown to be effective in improving knowledge about hereditary cancer amongst cancer patients (Mancini et al., 2006). Study 1 found that accuracy of recall of information amongst relatives was improved when information was provided directly from genetics health professionals via several sources (Jacobs, Dancyger, Smith, & Michie, 2015). Providing high quality accurate information leaflets for patients is therefore important in order to inform patients and relatives.

iii. **Health professionals’ communication:** Genetics and oncology health professionals who communicate with cancer patients about hereditary cancer should ensure key messages about hereditary cancer management as well as genetic testing are communicated. Supplementary messages are only communicated when they are particularly relevant to the individual and there is the time to do so.

iv. **Co-ordinated multi-disciplinary approach:** To ensure seamless referral between oncology and genetics, streamlined patient pathways are needed. Although for many patients, pre-test information without genetic counselling may be sufficient or even preferable (Quinn et al., 2016; Sie et al., 2014), this research found that some patients may need enhanced genetic counselling and support even if no pathogenic variant is detected (Study 5). The short time frame between genetic
testing and cancer treatment will require close working and a coordinated multidisciplinary approach between oncology and genetics services. Prior to testing, oncology health professionals will need to identify patients who may benefit from enhanced genetic counselling and support by eliciting expectations of inheritance, prior views and family experiences. Following testing, genetics health professionals will need to ensure patients have access to expertise in cancer management, for example the opportunity to discuss breast reconstruction with an oncoplastic surgeon. The integration of genetics into mainstream medicine will require greater collaboration and flexibility across traditional disciplinary boundaries. Although several studies of clinical practice have found that multidisciplinary clinics increased patient satisfaction and improved decision-making, this model has not been widely adopted across the UK (Bancroft et al., 2010; Firth et al., 2011; Pichert et al., 2010)

Genetic counselling: This work provides evidence that the information communicated during genetic counselling had little impact on patients’ understanding or experiences and concurs with the findings of previous studies (Pieterse, Ausems, Spreeuwenberg, & van Dulmen, 2011; Vos, Oosterwijk, et al., 2012). Genetic counselling has been found to be provider driven (Meiser, Irle, Lobb, & Barlow-Stewart, 2008), verbally dominated by health professionals (Butow & Lobb, 2004; Ellington et al., 2005; Roter et al., 2006) and overly focused on the provision of biomedical information with little engagement in social or emotional issues (Aalfs, Oort, de Haes, Leschot, & Smets, 2006; Ellington et al., 2006; Ellington et al., 2005; Meiser, 2005; Meiser et al., 2008; Michie, Lester, Pinto, & Marteau, 2005; Pieterse, Van Dulmen, Ausems, Beemer, & Bensing, 2005; Roter et al., 2006). Providers’ communication behaviours have been shown to influence patient outcomes (Duric et al., 2003; Lobb et al., 2004; Michie, Axworthy, Weinman, & Marteau, 1996). Addressing emotional concerns increases patient satisfaction (Roter et al., 2006) and improves psychological adjustment and physical outcomes (Pennebaker, Barger, & Tiebout, 1989; Pennebaker, Hughes, & O’Heeron, 1987). Several authors have recommended that genetic counselling should progress from the educational biomedical model to a more psychosocial counselling approach (Austin, Semaka, & Hadjipavlou, 2014; Kessler, 1997; Veach, Bartels, & LeRoy, 2007; Weil, 2003). Pre-test communication is likely to become even more information-focused as genetic testing is undertaken within mainstream oncology.
The findings from this thesis suggest that post-test genetic counselling should focus on helping patients to understand the implications of hereditary cancer information in the context of their own risk, prognosis and treatment options and providing psychological support throughout the genetic testing process when needed. In addition, patients need help to understand the relevance of the information for relatives. The infrequent communication amongst health professionals of information for relatives (Study 4), the low levels of recall amongst patients and relatives, and the evidence that recall is improved when information is received via several sources (Study 1) suggest that genetics health professionals need to take a proactive approach to facilitating and supporting the dissemination of information within families. These findings suggest that a re-evaluation of the role and practice of genetic counselling is needed.

vi. **Health professionals’ education:** Increasingly genetics health professionals will be required to counsel patients at vulnerable stages in their treatment and diagnosis, and oncology health professionals will be required to communicate about genetics. Health professionals who counsel patients about genetic testing need to understand the relevant issues for cancer patients and their families, filter key information from the mass of available knowledge, interpret and explain genetic test results, provide non-directive counselling, manage the expectation of certainty and the reality of ambiguous genetic test results, facilitate informed decision-making, provide support to patients and assist with the dissemination of information to family members. In the UK, regular mandatory training in communication skills is a requirement of all oncology health professionals (Moore et al., 2013). However, oncology health professionals lack training and expertise in communicating about genetics. Although expert communication skills are central to the practice of genetic counselling (Resta et al., 2006), there is no mandatory requirement for genetics health professionals to undergo regular training in communication skills with cancer patients. To ensure patients’ communication needs are met, genetics and oncology health professionals may benefit from regular training in communication about hereditary cancer with cancer patients, and this should reflect changes in practice.
8.4. IMPLICATIONS FOR RESEARCH

Recommendations for future research have been made throughout this thesis. The studies in this thesis were hypothesis generating and included data gathered several years ago. These studies should therefore be repeated to find out whether the results are replicated. Replication studies would ideally involve primary collection of data about the same types of patient and health professional about the information communicated and recalled and patients’ experiences of health professionals’ communication.

Replication studies should cover all the breast and ovarian cancer genes that are now tested for, and communication by oncology as well as genetics health professionals. The methods used in this thesis could be replicated with other types of hereditary cancer, such as colorectal cancer, where little previous communication research has been undertaken.

Five areas for future research were identified from the findings of this thesis: (i) the information and support needs of patients undergoing genetic testing shortly after diagnosis, at metastatic relapse and in palliative care, (ii) communication about genetics by oncology health professionals, (iii) understanding and experiences of patients and health professionals about disclosure and implications of VUS results, (iv) standardisation of patient information leaflets and (v) measures to improve the translation of guideline recommendations.

i. **Information and support needs of patients:** Further research is needed to clarify the information and support needs of patients who undergo genetic testing shortly after diagnosis and those tested at other times at which the patient is likely to be vulnerable to emotional distress, such as during metastatic relapse or palliative care. Conflicting evidence about the need for information about the impact of genetic testing on treatment was highlighted by this thesis. No research was identified about communicating with patients with metastatic cancer about hereditary cancer and there is limited research about this communication during palliative care (Lillie, Clifford, & Metcalfe, 2011; Metcalfe, Pumphrey, & Clifford, 2010; Quillin, Bodurtha, Siminoff, & Smith, 2010).

ii. To identify the information and support needs of these patients, the Delphi survey (Study 2) should be replicated with a different group of genetics and cancer expert health professionals and patients who have undergone genetic testing shortly after breast and ovarian cancer diagnosis and those at other stages in the cancer trajectory.
iii. **Oncology health professionals’ communication about genetic testing and hereditary cancer management:** Research is needed to develop interventions to educate, facilitate and support oncology health professionals to communicate effectively about hereditary cancer. Once such interventions have been developed, further research should evaluate the effectiveness of this communication in comparison with usual care. The process and content of oncology health professionals’ communication along with patients’ understanding, recall and risk perception will need to be investigated. Further studies are needed to explore patients experiences of oncology health professionals’ communication about hereditary cancer. This research will need to address patients tested shortly after diagnosis and those tested at metastatic relapse and during palliative care as the information and support needs of these patients may differ.

iv. **Health professionals understanding and communication about VUS results:** This thesis identified confusion and uncertainty about VUS results and surgical decision-making amongst patients tested shortly after diagnosis. The increase in genetic testing of cancer patients will result in a greater number of patients receiving VUS results. Following these results, patients’ decision-making will be guided by genetics and oncology health professionals. Further research is needed to investigate health professionals’ understanding and experiences of communication about VUS results, both pre- and post-testing, and the interventions required to facilitate informed decision-making. Research is also needed to understand what is communicated by oncology and genetics health professionals to patients once a VUS is identified and the process of patients’ decision-making in the knowledge of a VUS.

v. **Patient information leaflets:** This research found that more information was communicated in patient information leaflets than in clinical protocols, suggesting that information leaflets are widely used within genetics. Further research is needed to examine the extent to which patients recall information communicated via information leaflets and the effect of standardising information leaflets on patients’ knowledge and understanding of the information.

vi. **Measures to improve the translation of guideline recommendations into practice:** The integration of genetics into mainstream medicine will require greater collaboration and flexibility across traditional disciplinary boundaries. Research
is needed to investigate the interface between disciplinary boundaries, develop measures to enhance the use of clinical guidelines, improve interdisciplinary working and provide patients and families with access to the information, counselling and support they need.

8.5. CONCLUSIONS

Guidelines do not sufficiently address the communication needs of cancer patients about genetic testing and hereditary cancer management. Genetics health professionals do not always communicate information that is necessary and frequently communicate information that is unnecessary. Genetics health professionals do communicate information about genetic testing and patients mostly recall this information. However, information about hereditary cancer management is less well communicated by health professionals and less frequently recalled by patients and their relatives. For patients with newly diagnosed breast cancer, understanding of hereditary cancer appears to be affected by prior views, family experiences, and the extent to which prior expectations of inheritance are consistent with the result of the genetic test, rather than information communicated during genetic counselling.

Delivering relevant, clear and accessible information about genetic testing and hereditary cancer management via written, digital or group education is acceptable to many patients and may not require a routine pre-test genetic counselling appointment. Eliciting views about inheritance and the expectations of genetic testing from patients may help to identify those who need enhanced genetic counselling and support, regardless of genetic test results. Rather than primarily delivering biomedical information, genetic counselling should focus on facilitating personal understanding by helping patients to understand the information in the context of their personal and family circumstances, supporting shared decision-making, providing psychological support and enabling dissemination of information within the family.


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APPENDICES
Appendix 1:

Participant information sheets

and consent forms
Appendix 1. Patients’ participant information sheet (Family Communication Study)

OurRef:: Your
Ref:
Please quote reference on all correspondence

Date:  
Dictated:  

PARTICIPANT INFORMATION SHEET

Version: 1

Date: -

Study title

Communicating genetic information within families: The results consultation

Invitation

You are being invited to take part in a research study. Before you decide it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and discuss it with others if you wish. Ask us 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.

Thank you for reading this.

What is the purpose of the study?

The study will examine how genetic test information in the clinic is received by people and communicated through the family. We are interested in how people make sense of this information, who they decide to talk to about it and what they say to other family members.

Why have I been chosen?

We are inviting people intending to have the genetic test to look for a faulty breast/ovarian cancer gene to take part in the study.

Do I have to take part?

It is up to you to decide whether or not to take part. If you do decide to take part you will be given this information sheet to keep and be asked to sign a consent form. If you decide to take part you are still free to withdraw at any time and without giving a reason. A decision to withdraw at any time, or a decision not to take part, will not affect the standard of care you receive.
What will happen to me if I take part?

The first part of the study will examine what happens during the consultation process when the clinician gives a test result to the patient. For this phase, if you agree to take part, the researcher will have access to information about your genetic test, your results consultation may be tape-recorded and you may be interviewed by the researcher a few weeks later. This interview will also be tape recorded. The results consultation and the interview will be transcribed and analysed using standard procedures, and the tape will be destroyed after the project is finished.

Nothing else is required of you at this stage.

If you agree to take part in this first stage, we will explain about the subsequent stages of the research when you get your test result. You will be free to decide whether to continue being involved.

Will my taking part in this study be kept confidential?

All information which is collected about you during the course of the research will be kept strictly confidential. Any information about you which leaves the hospital/surgery will have your name and address removed so that you cannot be recognised from it.

What will happen to the results of the research study?

The results of the study will be written up for publication in medical and psychology journals. You will not be identified in any publication. If you are interested in reading the results of the study, you can contact the researcher or chief investigator up to 1 March 2009 (contact details below)

Who is organising and funding the research?

The research is organised through University College London. It is funded by a research grant from the Department of Health.

Who has reviewed the study?

The study has been reviewed by the London MREC Research Ethics Committee.

Contact for Further Information

If you would like more information, you can contact the main researcher on the project: [Contact Information]

What happens next?

If you agree to take part in the study or would like more information, in about a week, the researcher will phone you to talk about the project. You can ask her any questions you have.

If you decide to take part in the study, you will be given a copy of the information sheet and the signed consent form to keep.
Appendix 1.2. Patients' consent form (Family Communication Study)

CONSENT FORM

Title of Project:
Communicating genetic information within families: the results consultation

Name of Researcher: [Redacted]

1. I confirm that I have read and understand the information sheet dated 15/12/05 (version 1) for the above study and have had the opportunity to ask questions.

2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.

3. I agree to take part in the above study.

4. I understand that the researcher will have access to information about my genetic test for the purposes of this study.

5. I would like further information about the study before deciding whether or not to take part and agree that the researcher can contact me.

6. Please give contact details:
   daytime phone number
   evening phone number
   mobile phone number

7. I do not wish to take part in this study

Please initial box
PARTICIPANT INFORMATION SHEET & CONSENT FORM (Clinician)

Version: 1
Date: 15/12/05

Study title
Communicating genetic information within families: HBOC

What is the purpose of the study?
The study will examine how genetic test information in the clinic is received by patients and communicated through the family. We are interested in how patients make sense of this information, who they decide to talk to about it and what they say to other family members. In order to know what information they have received, we will tape the consultations of consenting participants.

Who has been chosen?
We are inviting all patients intending to have the genetic test to look for a faulty breast/ovarian cancer gene to take part in the study, and seeking their permission to audio tape the consultation in which they receive this test result.

Do I have to take part?
We are seeking your consent to tape the consultations of those patients who have consented to have their consultations tape recorded. It is up to you to decide whether or not to take part. If you do decide to take part you will be given this information sheet to keep and be asked to sign a consent form. If you decide to take part you are still free to withdraw at any time and without giving a reason.

What will happen if I take part?
We are asking you to recruit the patient to the study and switch on the tape recorder in order to audiotaape the consultation. The tape will be transcribed and analysed using standard procedures, and the tape will be destroyed after the project is finished.

Will my taking part in this study be kept confidential?
All information which is collected during the course of the research will be kept strictly confidential and anonymised so that neither you nor the patient can be recognised from it.
Appendix 1.4. Health professionals’ consent form (Family Communication Study)

Centre Number:  
Study Number:  
Clinician Identification Number for this study:

CONSENT FORM (Clinician)

Title of Project:
Communicating genetic information within families.

Name of Researcher:

Please initial box

1. I confirm that I have read and understand the information sheet dated ..............
   (version ............) for the above study and have had the opportunity to ask questions.

2. I understand that my participation is voluntary

3. I agree to take part in the above study.

_____________________________  ____________________________  ____________________________
Name of Clinician               Date                         Signature

_____________________________  ____________________________  ____________________________
Name of Person taking consent   Date                         Signature
(if different from researcher)

Contact details

1 for clinician; 1 for researcher;

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Appendix 1.5. Service users’ consent form (Study 2)

Centre Number: ____________________________
Study Number: ____________________________
Patient Identification Number for this study: ____________________________

CONSENT FORM

Title of study: 
Key messages about a BRCA1/2 gene fault for cancer patients

Name of Researcher: Ms Chris Jacobs

Please initial box

I confirm that I have read and understand the information sheet dated (version 1) for the above study, that I have had the opportunity to ask questions and that my questions have been answered.

I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.

I understand that other researchers and participants will have access to my anonymised judgements of key messages and comments

I agree to take part in the above study.

OR

I would like further information about the study before deciding whether or not to take part and agree that the researcher can contact me.

Please give contact details:
• Daytime phone number ____________________________
• Evening phone number ____________________________
• Mobile phone number ____________________________
• Email address ____________________________

Can we leave an answerphone message? Yes / No (please delete as appropriate)

OR

I do not wish to take part in this study

____________________ ____________________________
Name of Participant Date Signature

Ms Chris Jacobs ____________________________ ____________________________
Researcher Date Signature

Guy’s and St Thomas’ NHS Foundation Trust

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Appendix 1.6 Service users’ participant information sheet (Study 2)

You will be given a copy of this information sheet.

Title of Project: Key messages about BRCA1 and BRCA2 for women with breast or ovarian cancer

This study has been approved by the UCL Research Ethics Committee as Project ID Number: 4513/001

Name, Address and Contact Details of Investigators:

Researcher: Ms Chris Jacobs Consultant Genetic Counsellor, PhD student, NIHR Doctoral Research Fellow Department of Clinical, Educational and Health Psychology, UCL, London WC1E 7HB Email: Chris.Jacobs@gstt.nhs.uk

Supervisor: Professor Susan Michie Professor or Psychology Department of Clinical, Educational and Health Psychology, UCL, London WC1E 7HB Email: s.michie@ucl.ac.uk

We would like to invite you to participate in this research study. You should only participate if you want to; choosing not to take part will not disadvantage you in any way. Before you decide whether you want to take part, please read the following information carefully and discuss it with others if you wish. Ask us if there is anything that is not clear or you would like more information.

What is the purpose of the study?
The purpose of this study is to identify the key messages required by women with breast or ovarian cancer about BRCA1 and BRCA2 and when the key messages should be communicated. We are seeking the views of women who have had breast or ovarian cancer and have a cancer causing BRCA1 or BRCA2 gene fault, genetics clinicians and cancer clinicians.

Why have I been chosen?
You have been chosen to take part in the study because you have had breast or ovarian cancer and have a BRCA1 or BRCA2 gene fault.
Do I have to take part?
It is up to you to decide whether or not to take part. If you do decide to take part you will be given this information sheet to keep. You will not be asked to complete a consent form but returning the completed questionnaire will imply consent to participate.

What will happen to me if I take part?
If you take part in the study, you would be a member our expert panel of women with cancer who have a BRCA1/2 gene fault. We will ask you to review the information given to women with breast or ovarian cancer about a BRCA1/2 gene fault and to decide the information you consider to be key messages and the best time for the information to be communicated. You will be given some guidelines to help you with this. We will then circulate the anonymised responses of all the participants, to see if there is agreement. We may contact you up to three times about this. It may take you up to 45 minutes the first time you review the messages and up to 30 minutes after that. You would be able to do this in your own home. We may contact you again later to see if you would be willing to look at the judgements of key messages made by the expert clinicians' panel as well.

What will happen if taking part in this study raises questions or concerns about an aspect of my own health or management?
If you have questions or concerns about cancer risk or cancer risk management as a result of taking part in this study, we suggest that you contact the Breast Cancer Care helpline: 080 800 6000 in the first instance. If these questions are not answered by the helpline, you are advised to contact your local Regional Genetics Centre or your GP.

Will my taking part in this study be kept confidential?
All information collected during the course of the research will be confidential. Members of the research team will have access to your questionnaire responses. The responses of participants will be shared with other members of the expert panel but these will be summarised and anonymised.

It is up to you to decide whether to take part or not; choosing not to take part will not disadvantage you in any way. If you do decide to take part you are still free to withdraw at any time and without giving a reason.

All data will be collected and stored in accordance with the Data Protection Act 1998.

What will happen to the results of the research study?
The results of the study will form part of a PhD thesis. The findings of the study will be presented at academic and lay meetings and written up for publication in medical and psychology journals and. You will not be identified in any presentation or publication. A summary of the findings of the study will be sent to you by June 2016.
Appendix 1.7. Patients’ consent form (Study 5)

Guy’s and St Thomas’ NHS Foundation Trust

Centre Number: Study Number: Patient Identification Number for this study:

CONSENT FORM

Title of study: Communication about hereditary cancer: Exploring understanding and experiences of women who learn that their newly diagnosed breast cancer is, or could be, hereditary.

Name of Researcher: Ms Chris Jacobs

Please initial box

I confirm that I have read and understand the information sheet dated 02.02.12 (version 1) for the above study and that have had the opportunity to ask questions and my questions have been answered.

I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.

I understand that my interview will be audio tape-recorded.

I understand that my General Practitioner will be informed of my participation in the study.

I understand that relevant sections of my medical notes and data collected during the study, may be looked at by individuals from regulatory authorities or from the NHS Trust, where it is relevant to my taking part in this research. I give permission for these individuals to have access to my records.

I agree to take part in the above study.

I understand that the researcher will have access to information about my genetic test (if relevant) for the purposes of this study.

OR

I would like further information about the study before deciding whether or not to take part and agree that the researcher can contact me.

Please give contact details:

- Daytime phone number ........................................
- Evening phone number ........................................
- Mobile phone number ........................................
- Email address ..................................................


Can we leave an answerphone message? Yes / No (please delete as appropriate)

OR

I do not wish to take part in this study

__________________________  __________________________
Name of Patient              Date                      Signature

Ms Chris Jacobs
Researcher

__________________________  __________________________
Date                      Signature

(1 for Patient, 1 for Researcher, 1 to be kept with Genetics notes)
Appendix 1.8. Patients’ participant information sheet (Study 5)

Guy’s and St Thomas’
NHS Foundation Trust

PARTICIPANT INFORMATION SHEET

Study title
Communication about hereditary cancer: exploring understanding and experiences of women who learn that their newly diagnosed breast cancer is, or could be, hereditary.

Rationale for selection criteria for this study
This study is focusing on the experience and understanding of White British women who have been diagnosed with a new primary breast cancer between the ages of 35 and 55 and who have children or close female relatives. The reason for these criteria is to invite women to take part in the study who have similar life experiences, allowing us to focus on the specific issues surrounding learning about the possibility of hereditary cancer.

Invitation paragraph
You are being invited to take part in a research study. Before you decide whether or not to take part, it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and discuss it with others if you wish. Please ask the Researcher 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. Thank you for reading this Participant Information Sheet.

What is the purpose of the study?
The purpose of this study is to gain a deeper understanding of what it is like for women to learn that their newly diagnosed breast cancer is, or could be, hereditary. This is important as the possibility of a hereditary contribution to breast cancer (and referral to genetics services) is increasingly being discussed with women at the time of their diagnosis, yet little is known about what women understand about this or how the experience is for them.

Why have I been chosen?
We are inviting women to take part in the study who have recently been diagnosed with breast cancer and who are referred to the genetics department to discuss the possibility that their cancer could be hereditary.

Do I have to take part?
It is up to you to decide whether or not to take part. If you do decide to take part you will be given this information sheet to keep and be asked to sign a consent form. If you decide to take part you are still free to withdraw at any time and without giving a reason.

What will happen to me if I take part?
If you take part, the Researcher will agree a convenient time and place to interview you for about an hour on up to three occasions. The interview(s) will be audio tape-recorded and transcribed and then subjected to systematic analysis using a standard procedure.

The first interview will focus on your experience and understanding of being diagnosed with breast cancer. The second interview (if relevant) will be about your experience and understanding of the discussions in the genetics department and the third interview (if relevant) will be about how you feel once you know the outcome of your discussions in genetics.
If you have genetic testing as part of your clinical care, the Researcher will have access to this information after you have been informed of the result by your genetics clinician.

**Will my taking part in this study be kept confidential?**
All information which is collected about you during the course of the research will be kept strictly confidential. Any information about you which leaves the hospital will have your name and address removed so that you cannot be recognised from it. Other members of the research team will have access to the anonymised transcription of your interview. However, they will not have access to your personal details and they will respect your confidentiality at all times. In the event that you lose capacity to consent during the course of the study, no further interviews would be undertaken but the research team would retain any data already collected and continue to use this confidentially for the purposes of this study.

**What will happen if I raise questions or concerns about an aspect of my care with the Researcher?**
If you have questions or concerns about your care, the Researcher will encourage you to contact the relevant clinician. However, if you wish the Researcher to facilitate this contact, the Researcher will agree with you about how any contact will be made and this will be documented in your clinical genetics notes before any action is taken.

**What will happen to the results of the research study?**
The results of the study will be written up for publication in medical and psychology journals and will form part of a PhD thesis. You will not be identified in any publication. A summary of the findings of the study will be sent to you by June 2016.

**Who is organising and funding the research?**
The research is organised through Guy’s and St Thomas’ NHS Trust in conjunction with University College London and Birkbeck College. It is funded by a research grant from the National Institute of Health Research as part of a PhD project.

**Who has reviewed the study?**
The study has been reviewed by London Bromley Research Ethics Committee.

**Contact for Further Information**
If you wish to have more information, you can contact the researcher on the project: Ms Chris Jacobs (details below).

**What do I do next?**
You will either have been sent this sheet following a telephone call from the Researcher in which you agreed to consider taking part in the study, or because the genetics clinician who has arranged your genetics appointment thought you might be interested in this study.

The Researcher will contact you by telephone in the next few days to give you an opportunity to ask any further questions about the research. You can then decide whether or not you wish to take part. Alternatively you can ring or email the Researcher:

**Telephone:** 020 7188 1364 (Guy’s Hospital Clinical Genetics Department reception desk).
Please state that you are calling about the ‘Communication about hereditary cancer’ study and leave your name, telephone number and Genetics Reference Number and the Researcher will call you back. **Email:** Chris.Jacobs@gstt.nhs.uk **If you take part, you will be given a copy of the information sheet and a signed consent form to keep.**
Appendix 2:

Interview schedules and questionnaires
Appendix 2.1. Interview schedules for patients and relatives (Family Communication Study)

FCS Interview Schedule

A. Having the test and getting the result

Could you tell me how you came to have the genetic test? Why?
Note: how involved were family members in decision for this person to be tested? How much a family issue?

Could you tell me what you were told by the clinician when you received your genetic test result?
Prompt: try and get as much and detailed information about what exactly they remember

Can you remember what you were told about genetic testing at previous appointments?

How did you feel when you received your test result?
Prompt: Thoughts, feelings, reactions?

What did you expect your result to be?

What is your understanding of your risk of developing breast/ovarian/prostate cancer now?
Prompt: In comparison to general population

How do you feel about your risk now?

What is your understanding of your family’s risk of developing breast/ovarian/prostate cancer now?

How do you feel about your family’s risk now?

Has the test result affected your perception of your illness/condition?
Prompt: In particular, what do you see as the cause of your illness/condition?

Will the test result make any difference to what you do with regards to screening?

Did you tell any family member you were having the test done?

Did anyone come with you to get your test result?

Apart from the information you have been given at the clinic about BRCA1/2, have you found out anything more through other sources eg. television, newspaper, internet, charities etc.

B. Decision who to tell

Have you told any family member your test result?
Note: explain we mean biological relative

If no:
Do you intend to tell family members?
If not, why?
If yes, who? How have you decided who to tell?
What are you planning to tell them? When?
What do you think their reactions are likely to be?

If yes:
Who have you told? When did you tell them?
How did you decide who to tell?
   Prompt: What factors were important in making this decision?

Note: Make note of people told

C. The process of talking to each one

Out of the people who you have told, who do you think was the most important to tell? Why?

Note: Ask following for each family member, starting with person identified as most important to tell, then proceed with next important etc. focusing on up to three family members

Why did you decide to tell XXX?

How did you tell them?
   Prompt: face to face, email, telephone etc?

What did you say?
   Prompt: try and get as much and detailed information about what exactly they told XXX

How did you decide what to say?

Did you miss out or add bits? If so, why?

Do you think he/she understood what you were saying?
   Prompt: If not, why do you think this was?

How did he/she respond?
   Prompt: try and get as much detailed information about responses (what they said, emotional response)

Is there anyone you would definitely not tell? Why?

D. Reflections on the process

Is there anyone else you would like to tell or are planning to tell?

What do you think the genetic test result means for you and for your family?

F. Issues not yet identified

Is there anything else that you would like to add?
FCS Interview Schedule – Relatives

**Being told:**

Could you tell me how you came to know that [X] was having a genetic test?
*Prompt: Was this prior to them having the test, prior to receiving the result, or after receiving the result?*

How did [X] tell you her/his result?
*Prompt: was it in person, on the telephone, email etc? At a family event, in a group, on your own etc?*

Could you tell me what you were told by [X] when she/he spoke to you about her/his genetic test result?
*Prompt: try and get as much and detailed information about what exactly they remember, including content of information and the way it was conveyed*

Did you understand what [X] was saying?

How did you respond when you were told [X’s] result?
*Prompt: how did you feel, what did you say?*

Has this affected your relationship with [X] in any way?
*Prompt: brought you closer, raised difficulties?*

Why do you think [X] decided to tell you the test result?

Do you think [X] should have told you her/his test result?

How much did you know about genetic testing for BRCA1/2 before [X] spoke to you?

What, if any do you think the implications of this test result are for you?

For other members of your family?

**Telling others:**

Have you told any family member [X’s] result?

If no:
Do you intend to tell family members?
If not, why?
If yes, why? Who? How have you decided who to tell?
What are you planning to tell them? When?
What do you think their reactions are likely to be?
If yes:
Who have you told? When did you tell them?
How did you decide who to tell?
*Prompt: What factors were important in making this decision?*
What did you tell them?

How did they react?

Is there anyone you would definitely not tell? Why?

**Deciding what to do:**

Have you decided whether you would like to have a predictive genetic test?

What have you decided?

Why?

How did you decide?

Do you think that anything about how [X] told you or what she/he said influenced this decision?

Has knowing the test result influenced your behaviour in any other ways?
*Prompt: seeking increased screening, breast care etc.*

**Issues not yet identified:**

Is there anything else that you would like to add?

Demographics
Appendix 2.2. Service users’ preliminary questionnaire (Study 2)

Default Question Block

Key messages about BRCA1 and BRCA2 for women with breast or ovarian cancer: Questionnaire 1

Thank you for agreeing to take part in this study as a member of our expert panel of women with cancer and a BRCA1/2 gene fault.

The aims of this study
The study aims to answer two main questions:
- What are the key messages required by women with breast or ovarian cancer in order to enable them to understand the risks, implications and options for themselves and their relatives and to decide on a course of action that is appropriate for them?
- Should key messages be communicated to women with cancer before genetic testing or after a gene fault has been found?

What we are asking you to do
Please answer ALL the questions about your personal history of cancer and the genetic testing you have had, even if you are not sure. There is room for you to explain any of your answers if you need to.

What happens next
When we receive this questionnaire back from you we will send you another questionnaire which will ask you to look at a list of summarized statements of information given to women with cancer during genetic counselling after a BRCA1/2 fault has been found. We will ask you to decide if these are key messages or not and when the information should be communicated.

Contact details
If you have any questions about the study, please contact Chris.Jacobs@gett.nhs.uk and we will get back to you as soon as possible.

Many thanks for your help with this.

Have you ever had breast cancer?
- Yes
- No
- Not sure

If yes, how old were you when you were diagnosed with breast cancer?

Have you ever had ovarian cancer?
- Yes
- No
- Not sure

If yes, how old were you when you were diagnosed with ovarian cancer?

Have you had a genetic test for hereditary breast and ovarian cancer?
- Yes
- No
- Not sure

At what stage in your breast or ovarian cancer treatment did you have a genetic test?
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Which of these describes your genetic test result? Please tick all that apply

☐ A cancer causing gene fault (mutation) in BRCA1 or BRCA2 was found
☐ An Unclassified Variant (UV) or Variant of Unknown Significance (VUS) was found
☐ No BRCA1 or BRCA2 gene faults (mutations) were found
☐ Other (please explain):

☐ Not sure

Were you the first person in your family who was found to have a BRCA1/2 gene fault?

☐ Yes
☐ No
☐ Not sure

Where did you have genetic testing?

☐ England
☐ Scotland
☐ Northern Ireland
☐ Wales
☐ Other (please explain):

☐ Not sure

When did you receive your genetic test result?

☐ Before 2008
☐ Between 2008 and 2012
☐ 2013
☐ Not sure

How confident are you about your answers?

☐ Not at all confident
☐ Very confident
☐ Somewhat confident

How did you receive your genetic test result? (please tick all that apply)

☐ In a Genetics appointment
☐ By Telephone
☐ By post
☐ Other (please explain)


Page 2 of 3
Did you inform your at risk relatives about the gene fault?
- I informed all of my at risk relatives
- I informed most of my at risk relatives
- I did not inform my relatives
- Not sure

Please explain why you did not inform some or all of your relatives

What year were you born?

How would you describe your ethnic group?

What is the highest level of education you have completed?
- No qualifications
- GCSE or equivalent
- A level or equivalent
- First degree BA/ BSc etc
- Masters degree
- PhD
- Not sure
Appendix 2.3. Health professionals’ preliminary questionnaire (Study 2)

Questionnaire 1 for expert clinicians’ panel

Q1 Key messages about BRCA1 and BRCA2 for women with breast or ovarian cancer:

Thank you for agreeing to take part in this study as a member of our expert clinicians’ panel. The study aims to answer two main questions:

What are the key messages required by women with breast or ovarian cancer in order to enable them to understand the risks, implications and options for themselves and their relatives in relation to BRCA1/2 and to decide on a course of action that is appropriate for them?

Should key messages be communicated to women with cancer before genetic testing or after a gene mutation has been identified?

What we are asking you to do now: Please answer the questions about your role and experience with women with breast or ovarian cancer and a BRCA1 or BRCA2 gene mutation.

What happens next: When we receive this questionnaire back from you we will send you another questionnaire which will ask you to look at a list of summarised statements of information that were taken from transcripts of genetic counselling sessions with women with cancer after a BRCA1/2 mutation has been identified. We will ask you to decide if these are key messages or not and when the information should be communicated i.e. before genetic testing or after a mutation has been identified.

Contact details: If have any questions about the study, please contact Chris.Jacobs@gstt.nhs.uk and we will get back to you as soon as possible. Many thanks for your help with this.
Q1 What is your profession? (please tick the most appropriate answer)
- Doctor
- Nurse
- Genetic Counsellor
- Other (please explain) ________________

Q2 What is your professional title? (if Dr is selected)
- Consultant
- Junior doctor
- Medical student
- Other (please explain) ________________

Q3 What is your professional title? (if nurse is selected)
- Consultant Nurse
- Advanced Nurse Practitioner
- Clinical Nurse Specialist
- Registered Nurse
- Student Nurse
- Other (please explain) ________________

Q4 What is your professional title? (if genetic counsellor is selected)
- Consultant Genetic Counsellor
- Genetic Counsellor Manager
- Principal/ Lead Genetic Counsellor
- Registered Genetic Counsellor
- Trainee Genetic Counsellor
- Student Genetic Counsellor
- Other (please explain) ________________

Q5 What is your main area of professional expertise?
- Oncology
- Breast surgery/ care
- Genetics
- Gynaecological surgery/ care
- Other (please explain) ________________

Q6 Do you currently practice within the NHS?
- Yes
- No

Q7 Which of the following statements best describes your experience with patients who have/ may have a BRCA1/2 mutation?
- I do not have any experience with patients who have/ may have a BRCA1/2 mutation
- I refer patients to to discuss BRCA1/2
- I manage/ care for patients who have/ may have a BRCA1/2 gene mutation
- I counsel patients about BRCA1/2
- Other (please explain) ________________

Q8 How often do you see patients who have, or may have, a BRCA1/2 gene mutation?
- Never
- Less than Once a Month
- Once a Month
- 2-3 Times a Month
- Once a Week
- 2-3 Times a Week
- Daily

Q9 What year were you born?
Appendix 2.4. Online questionnaire round 1 (Study 2)

Default Question Block

Key messages about BRCA1 and BRCA2 for women with breast or ovarian cancer: Questionnaire 2

Thank you for returning Questionnaire 1.

What we are asking you to do now
In Questionnaire 2 you will find a list of summarised statements of information that have been taken from transcripts of genetic counselling sessions with women with breast or ovarian cancer after a BRCA1 or BRCA2 gene fault was identified. This was part of an earlier research study.

We are asking you to decide if each of these statements is a key message or not and when you think the information should be communicated to women with breast or ovarian cancer (ie before testing, once a gene fault has been found, at both times or at a different time). Please answer ALL the questions, even if you are not sure.

Finally, please tell us if there is any additional information that you consider to be a key message and when you think this should be communicated to women with breast or ovarian cancer about BRCA1 or BRCA2.

As you do this, please keep in mind the following definition of a key message in this context which is derived from the goals of genetic counselling. You will find this definition at the top each page of the questionnaire. The examples are at the bottom of each page:

A key message is information required by the individual with cancer in order to understand the risks, implications and options for themselves and their relatives in relation to BRCA1/2 and to decide on a course of action that is appropriate for them.

If an individual did not accurately receive this information they would be less likely to:
- Inform ‘at risk’ relatives OR
- Seek/ have genetic counselling/ genetic testing OR
- Take measures to detect cancer early, such as breast awareness or surveillance OR
- Take measures (appropriate to the prognosis) to reduce the risk of cancer such as surgery OR
- Seek other medical advice or information related to the risks, such as contraception, reproductive options or Hormone Replacement Therapy (HRT)

What will happen next
Once we have received all the responses back from the expert panel, we will take out any statements where most participants agree. We will then send you the remaining statements again and remind you of how you rated these. We will also circulate a summary of how the rest of the panel rated these statements and any anonymised comments. We will ask you if you wish to change your mind having seen the responses of the whole panel. We may contact you on up to two further occasions about this. We may also contact you again later to see if you would be willing to look at the judgements of key messages made by other expert panels.

Contact details
If you have any questions about the study, please contact Chris.Jacobs@astt.nhs.uk with your contact details and we will get back to you as soon as possible.

Many thanks for your help with this.

Inheritance

The children and brothers and sisters of a person with a BRCA1 or BRCA2 gene fault each have a 50% risk of inheriting the gene fault.

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<thead>
<tr>
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The children and brothers and sisters of a person with a BRCA1 or BRCA2 gene fault each have a 50% risk of inheriting the gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing | Once a gene fault has been identified | Not sure or don't have a view | Both before genetic testing AND once a gene fault has been identified | Another occasion (please explain)
Please add any comments on the reason for your decision or the wording of the statement.

<table>
<thead>
<tr>
<th>Identifying which side of the family is at risk of cancer is important.</th>
</tr>
</thead>
<tbody>
<tr>
<td>NOT a key message</td>
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When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.

A BRCA1 or BRCA2 gene fault does not 'skip' a generation.

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Please add any comments on the reason for your decision or the wording of the statement.

A BRCA1 or BRCA2 gene fault may explain the family history of cancer.
A BRCA1 or BRCA2 gene fault may explain the family history of cancer.

When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.

Please add any additional key messages about inheritance (according to the above definition of a key message).

BRCA1/2 genes

The fault is in the (BRCA1 or BRCA2) gene (ie specifying the name of the gene involved).

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The fault is in the (BRCA1 or BRCA2) gene (ie specifying the name of the gene involved).

When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.

Not everyone who inherits a BRCA1 or BRCA2 gene fault will develop cancer.

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Please add any comments on the reason for your decision or the wording of the statement.

The BRCA1 and BRCA2 genes have a role in protecting a woman from breast and ovarian cancer.

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Please add any comments on the reason for your decision or the wording of the statement.

Please add any additional key messages about the BRCA1 or BRCA2 genes (according to the above definition of a key message).

Genetic counselling/ testing

Predictive genetic testing is available for relatives once a BRCA1 or BRCA2 gene fault has been identified.

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Predictive genetic testing is available for relatives once a BRCA1 or BRCA2 gene fault has been identified.

When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.

People who have a negative BRCA1 or BRCA2 predictive test are still at risk of cancer.

- NOT a key message
- Probably NOT a key message
- Not sure or don't have a view
- Probably IS a key message
- A KEY MESSAGE

People who have a negative BRCA1 or BRCA2 predictive test are still at risk of cancer.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don't have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Female relatives who are at 50% risk of inheriting a gene fault and do not want to have a genetic test can have the same screening as women who have inherited the gene fault.

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- Probably NOT a key message
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- Probably IS a key message
- A KEY MESSAGE

Female relatives who are at 50% risk of inheriting a gene fault and do not want to have a genetic test can have the same screening as women who have inherited the gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don't have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

It is important to inform all relatives who are at risk that genetic testing is available.

- NOT a key message
- Probably NOT a key message
- Not sure or don't have a view
- Probably IS a key message
- A KEY MESSAGE
It is important to inform all relatives who are at risk that genetic testing is available.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing
Once a gene fault has been identified
Not sure or don't have a view
Before genetic testing AND once a gene fault has been identified
Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Predictive genetic testing is not recommended at a young age (before the age of 18).

NOT a key message
Probably NOT a key message
Not sure or don't have a view
Probably IS a key message
A KEY MESSAGE

Predictive genetic testing is not recommended at a young age (before the age of 18).

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing
Once a gene fault has been identified
Not sure or don't have a view
Before genetic testing AND once a gene fault has been identified
Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Before having a genetic test it is important to discuss the implications and possible outcomes

NOT a key message
Probably NOT a key message
Not sure or don't have a view
 Probably IS a key message
A KEY MESSAGE

Before having a genetic test it is important to discuss the implications and possible outcomes

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing
Once a gene fault has been identified
Not sure or don't have a view
Before genetic testing AND once a gene fault has been identified
Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.
Once a gene fault has been found in a family it is up to each individual to decide if they want a genetic test or not. Some relatives may decide not to be tested.

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When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.

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Genetic test results will not affect insurance.

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Please add any comments on the reason for your decision or the wording of the statement.

Please add any additional key messages about genetic counselling/testing (according to the above definition of a key message).
Cancer risks for women with cancer

Women with breast cancer who have a BRCA1 or BRCA2 gene fault are at increased risk of developing further primary breast cancers.

NOT a key message
Probably NOT a key message
Not sure or don’t have a view
Probably IS a key message
A KEY MESSAGE

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing
Once a gene fault has been identified
Not sure or don’t have a view
Before genetic testing AND once a gene fault has been identified
Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Women with ovarian cancer who have a BRCA1 or BRCA2 gene fault are at increased risk of developing breast cancer.

NOT a key message
 Probably NOT a key message
 Not sure or don’t have a view
 Probably IS a key message
 A KEY MESSAGE

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing
Once a gene fault has been identified
Not sure or don’t have a view
Before genetic testing AND once a gene fault has been identified
Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

It’s best to wait a while after cancer treatment before having risk reducing surgery.

NOT a key message
 Probably NOT a key message
 Not sure or don’t have a view
 Probably IS a key message
 A KEY MESSAGE

When do you think this information should be given to a woman with breast or ovarian cancer?

Once a gene fault has been
Before genetic testing AND once
Another occasion (please explain)
### Before genetic testing
- Identification (ID)
- Not sure or don’t have a view
- A gene fault has been identified
- A key message

#### Please add any comments on the reason for your decision or the wording of the statement.

**Most BRCA1 related breast cancer is ER-ve and so does not respond to Tamoxifen treatment.**

- NOT a key message
- Probably NOT a key message
- Not sure or don’t have a view
- Probably IS a key message

#### When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don’t have a view
- Before genetic testing AND once a gene fault has been identified

#### Please add any comments on the reason for your decision or the wording of the statement.

**Women with breast cancer who have a BRCA1 or BRCA2 gene fault are at increased risk of ovarian cancer.**

- NOT a key message
- Probably NOT a key message
- Not sure or don’t have a view
- Probably IS a key message

#### When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don’t have a view
- Before genetic testing AND once a gene fault has been identified

#### Please add any comments on the reason for your decision or the wording of the statement.

**Much is still unknown about the risks associated with a BRCA1 or BRCA2 gene fault.**

- NOT a key message
- Probably NOT a key message
- Not sure or don’t have a view
- Probably IS a key message

---


Page 9 of 23
Much is still unknown about the risks associated with a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing  
Once a gene fault has been identified  
Not sure or don't have a view  
Before genetic testing AND once a gene fault has been identified  
Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Treatment trials may be available for women with cancer who have a BRCA1 or BRCA2 gene fault.

NOT a key message  
Probably NOT a key message  
Not sure or don't have a view  
Probably IS a key message  
A KEY MESSAGE

Treatment trials may be available for women with cancer who have a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing  
Once a gene fault has been identified  
Not sure or don't have a view  
Before genetic testing AND once a gene fault has been identified  
Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Chemotherapy may still be required if a woman with cancer and a BRCA1 or BRCA2 gene fault were to develop further cancers.

NOT a key message  
Probably NOT a key message  
Not sure or don't have a view  
Probably IS a key message  
A KEY MESSAGE

Chemotherapy may still be required if a woman with cancer and a BRCA1 or BRCA2 gene fault were to develop further cancers.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing  
Once a gene fault has been identified  
Not sure or don't have a view  
Before genetic testing AND once a gene fault has been identified  
Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.
Further cancers would be no more difficult to treat because of a BRCA1 or BRCA2 gene fault.

NOT a key message   Probably NOT a key message  Not sure or don’t have a view  Probably IS a key message   A KEY MESSAGE

Further cancers would be no more difficult to treat because of a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing   Once a gene fault has been identified  Not sure or don’t have a view  Before genetic testing AND once a gene fault has been identified  Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

A BRCA1 or BRCA2 gene fault does not increase the risk of cancer recurrence or metastases (secondaries).

NOT a key message   Probably NOT a key message  Not sure or don’t have a view  Probably IS a key message   A KEY MESSAGE

A BRCA1 or BRCA2 gene fault does not increase the risk of cancer recurrence or metastases (secondaries).

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing   Once a gene fault has been identified  Not sure or don’t have a view  Before genetic testing AND once a gene fault has been identified  Another occasion (please explain)

Please add comments on the reason for your decision or the wording of the statement.

Please add any additional key messages about cancer risk to women with cancer (according to the above definition of a key message).

Cancer risks for women and men without cancer

Breast cancer risk is increased for women without cancer who have a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don't have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Breast cancer risk starts to increase from age 30 years in women who have a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don't have a view
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- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Ovarian cancer risk is increased for women without cancer who have a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don't have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.
Please add any comments on the reason for your decision or the wording of the statement.

Ovarian cancer risk starts to increase at age 40 to 45 years in women who have a BRCA1 or BRCA2 gene fault.

- NOT a key message
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- A KEY MESSAGE

Ovarian cancer risk starts to increase at age 40 to 45 years in women who have a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
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- Not sure or don’t have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Prostate cancer risk is increased for men who have a BRCA1 or BRCA2 gene fault.

- NOT a key message
- Probably NOT a key message
- Not sure or don’t have a view
- Probably IS a key message
- A KEY MESSAGE

Prostate cancer risk is increased for men who have a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don’t have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Male breast cancer risk is not increased by a BRCA1 gene fault.

- NOT a key message
- Probably NOT a key message
- Not sure or don’t have a view
- Probably IS a key message
- A KEY MESSAGE
Male breast cancer risk is **not** increased by a BRCA1 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don’t have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Male breast cancer risk is **increased** by a BRCA2 gene fault.

- **NOT** a key message
- Probably NOT a key message
- Not sure or don’t have a view
- Probably IS a key message
- A KEY MESSAGE

Male breast cancer risk is **increased** by a BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don’t have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Pancreatic cancer risk is **not** increased by a BRCA1 gene fault.

- **NOT** a key message
- Probably NOT a key message
- Not sure or don’t have a view
- Probably IS a key message
- A KEY MESSAGE

Pancreatic cancer risk is **not** increased by a BRCA1 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
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- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.
Pancreatic cancer risk is increased by a BRCA2 gene fault.

NOT a key message  Probably NOT a key message  Not sure or don't have a view  Probably IS a key message  A KEY MESSAGE

Pancreatic cancer risk is increased by a BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing  Once a gene fault has been identified  Not sure or don't have a view  Before genetic testing AND once a gene fault has been identified  Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Please add any additional key messages about cancer risk to women and men without cancer (according to the above definition of a key message).

Cancer risk management options

Diet and lifestyle can help to protect from cancer.

NOT a key message  Probably NOT a key message  Not sure or don't have a view  Probably IS a key message  A KEY MESSAGE

Diet and lifestyle can help to protect from cancer.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing  Once a gene fault has been identified  Not sure or don't have a view  Before genetic testing AND once a gene fault has been identified  Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.
Risk Reducing Mastectomy (surgery to remove the breasts in order to reduce the risk of cancer) is an option for women who have a BRCA1 or BRCA2 gene fault.

NOT a key message  Probably NOT a key message  Not sure or don’t have a view  Probably IS a key message  A KEY MESSAGE

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing  Once a gene fault has been identified  Not sure or don’t have a view  Before genetic testing AND once a gene fault has been identified  Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Breast reconstruction is an option after Risk Reducing Mastectomy.

NOT a key message  Probably NOT a key message  Not sure or don’t have a view  Probably IS a key message  A KEY MESSAGE

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing  Once a gene fault has been identified  Not sure or don’t have a view  Before genetic testing AND once a gene fault has been identified  Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Breast reconstruction can achieve good cosmetic results

NOT a key message  Probably NOT a key message  Not sure or don’t have a view  Probably IS a key message  A KEY MESSAGE

When do you think this information should be given to a woman with breast or ovarian cancer?
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Please add any comments on the reason for your decision or the wording of the statement.

**Risk Reducing Mastectomy reduces the risk of breast cancer (but a small risk of breast cancer remains).**

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**Risk Reducing Mastectomy reduces the risk of breast cancer (but a small risk of breast cancer remains).**

When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.

**Risk Reducing Mastectomy can be offered at a young age.**

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**Risk Reducing Mastectomy can be offered at a young age.**

When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.

Enhanced breast screening is available for women who have a **BRCA1 or BRCA2 gene fault**.
Enhanced breast screening is available for women who have a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

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There are limitations to breast screening.

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<th>Not sure or don't have a view</th>
<th>Probably IS a key message</th>
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</tr>
</thead>
</table>

There are limitations to breast screening.

When do you think this information should be given to a woman with breast or ovarian cancer?

<table>
<thead>
<tr>
<th>Before genetic testing</th>
<th>Once a gene fault has been identified</th>
<th>Not sure or don't have a view</th>
<th>Before genetic testing AND once a gene fault has been identified</th>
<th>Another occasion (please explain)</th>
</tr>
</thead>
</table>

Please add any comments on the reason for your decision or the wording of the statement.

---

Breast awareness is important.

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Breast awareness is important.

When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.
There is no ovarian screening available on the NHS.

There is no ovarian screening available on the NHS.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing

Once a gene fault has been identified

Not sure or don't have a view

Before genetic testing AND once a gene fault has been identified

Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Risk Reducing Bilateral Salpingo-Oophorectomy (surgery to remove the ovaries and fallopian tubes in order to reduce the risk of cancer) is an option for women who have a BRCA1 or BRCA2 gene fault.

Risk Reducing Bilateral Salpingo-Oophorectomy (surgery to remove the ovaries and fallopian tubes in order to reduce the risk of cancer) is an option for women who have a BRCA1 or BRCA2 gene fault.

When do you think this information should be given to a woman with breast or ovarian cancer?

Before genetic testing

Once a gene fault has been identified

Not sure or don't have a view

Before genetic testing AND once a gene fault has been identified

Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Bilateral Salpingo-Oophorectomy reduces the ovarian cancer risk (but a small risk of primary peritoneal cancer remains).
When do you think this information should be given to a woman with breast or ovarian cancer?

<table>
<thead>
<tr>
<th>Before genetic testing</th>
<th>Once a gene fault has been identified</th>
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<th>Another occasion (please explain)</th>
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Please add any comments on the reason for your decision or the wording of the statement.

Bilateral Salpingo-Oophorectomy before the natural menopause reduces a woman's risk of breast cancer.

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Bilateral Salpingo-Oophorectomy before the natural menopause reduces a woman's risk of breast cancer.

When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.

Bilateral Salpingo-Oophorectomy causes surgical menopause.

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Bilateral Salpingo-Oophorectomy causes surgical menopause.

When do you think this information should be given to a woman with breast or ovarian cancer?

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Please add any comments on the reason for your decision or the wording of the statement.
### Appendix

**After surgical menopause Hormone Replacement Therapy (HRT) may be prescribed until the age of 50 (unless a woman has had ER+ breast cancer).**

<table>
<thead>
<tr>
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**When do you think this information should be given to a woman with breast or ovarian cancer?**

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Please add any comments on the reason for your decision or the wording of the statement.

---

**There is no prostate screening available on the NHS.**

<table>
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Please add any comments on the reason for your decision or the wording of the statement.

---

**There are tests that can be done before or during pregnancy to reduce the chance of passing a faulty BRCA1 or BRCA2 gene onto future children.**

<table>
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**When do you think this information should be given to a woman with breast or ovarian cancer?**

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Please add any comments on the reason for your decision or the wording of the statement.

Taking the Oral Contraceptive Pill and having children protect a woman from ovarian cancer.

- NOT a key message
- Probably NOT a key message
- Not sure or don't have a view
- Probably IS a key message
- A KEY MESSAGE

Taking the Oral Contraceptive Pill and having children protect a woman from ovarian cancer.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don't have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Discussion with other specialists in the multidisciplinary team may be helpful.

- NOT a key message
- Probably NOT a key message
- Not sure or don't have a view
- Probably IS a key message
- A KEY MESSAGE

Discussion with other specialists in the multidisciplinary team may be helpful.

When do you think this information should be given to a woman with breast or ovarian cancer?

- Before genetic testing
- Once a gene fault has been identified
- Not sure or don't have a view
- Before genetic testing AND once a gene fault has been identified
- Another occasion (please explain)

Please add any comments on the reason for your decision or the wording of the statement.

Please add any additional key messages about managing cancer risks (according to the above definition of a key message).
If an individual did not accurately receive this information, they would be less likely to: inform ‘at risk’ relatives; OR seek/have genetic counselling; genetic testing; OR take measures to detect cancer early, e.g. breast assessment or surveillance; OR take measures (appropriate to the prognosis) to reduce the risk of cancer, e.g. surgery; OR seek other medical advice or information related to the risks, e.g. contraception, reproductive options or Hormone Replacement Therapy (HRT).
Appendix 2.5 Semi-structured schedules for interviews 1, 2 and 3 (Study 5)

Schedule Interview 1
Before the genetics appointment

I would like to ask you some questions about your experience of being diagnosed with breast cancer, starting with what happened in your most recent appointment when the diagnosis was made and going back to what led up to that point. I’m interested in how you were feeling and what it was like for you, rather than the steps that took place.

Understanding and experience of being diagnosed with breast cancer
1. Can you begin by telling me what you recall about your most recent consultation in the Breast Unit?
   Prompts:
   • What did the doctor tell you about your diagnosis and treatment?
   • How were you told about the diagnosis?
   • How did you react?
   • How did you feel?

Understanding and experience of the lead up to the diagnosis
2. Are you able to tell me what led you to be seen in the Breast Unit?
   Prompts:
   • Were you having regular breast screening? If yes, why?
   • (If found a breast lump) What was going through your mind when you first found the lump in your breast?
   • How did the GP let you know that would need to be referred to the Breast Unit?
   • How did you react?
   • How did you feel?

Present experiences
3. How are you feeling now?
   Prompts:
   • How have you got through the last few weeks?
   • What has been important to you since the diagnosis?
   • (If information seeking) Where have you gone to for information?
   • Who have you turned to for support?

Understanding about why breast cancer developed and family/ personal explanations/ risk
4. Why do you think you got breast cancer?
   Prompts:
   • How did you feel about the chance that you would get breast cancer before you were diagnosed?
   • If felt at higher risk, why?
   • If has a family history, why do you think members of your family have had breast cancer?

Feelings about the experience of genetics being raised at time of diagnosis
5. Why do you think you have been referred to the genetics clinic?
   Prompts
   • Can you recall what the doctor said to you about genetics?
   • How did you react?
   • How did you feel?
   • How did you feel about the way the doctor talked with you about genetics/ the timing of the doctor mentioning genetics to you?
Expectations about genetics

6. What does being referred to ‘genetics’ mean to you?
Prompts:
• What do you think will happen in your genetics appointment?
• What do you understand about genetics/ hereditary cancer?
• What difference, if any, do you think the genetics appointment will make to your treatment/ surgery/ screening/ cancer risk/ family?

Schedule Interview 2
Following genetic counseling appointment

I would like to ask you some questions about your understanding and experiences of the genetics appointment. I’m interested in how you were feeling and what it was like for you, rather than the steps that took place.

Experience of the genetic counselling
1. Can you begin by telling me what you recall about your genetic counseling appointment?
Prompts:
• What did the genetic counselor/ doctor say to you?
• How did you feel?
• How did you react?
• Was there anything unexpected?
• How did you feel?
• How did you react?

2. How did you feel about the way the genetic counselor/ doctor talked with you about genetics/ cancer?
Prompts:
• How did you feel?
• How did you react?

Understanding of the genetic counselling
3. What did you understand from the appointment?
Prompts:
• What do you understand to be the cause of your cancer?
• What does this mean for you/ your family?
• What difference, if any, do you think the genetics appointment will make to your treatment/ surgery/ screening/ cancer risk/ family?
• How do you feel about that?

Understanding and experience before the appointment
4. Are you able to tell me how you felt before the appointment?
Prompts:
• What were you expecting?
• How did you feel?

Present experiences
5. How are you feeling now?
Prompts:
• How did you feel when you came out of the appointment?
• How do you feel now?
• What has been important to you since the appointment?
• (If information seeking) Where have you gone to for information?
• Who have you turned to for support?

**Future**

6. **What do you expect will happen next?**
Prompts:
• How do you feel about what will happen next?

---

**Schedule Interview 3**

**Following the genetic test result and decisions regarding treatment**

I would like to ask you some questions about your understanding and experiences now that you have the result of your genetic test and a treatment plan. I'm interested in how you were feeling and what it was like for you, rather than the steps that took place.

**Understanding and experience about the genetic test result**

1. **Can you begin by telling me about your genetic test result?**
Prompts:
• How do you feel about the result?
• What do you understand by the result?
• What do you understand about the implications of this result for yourself (current treatment/ future risks)?
• How do you feel about that?
• What do you understand about the implications of this result for your family/ children?
• How do you feel about that?
• (If information seeking) Where have you gone to for information?
• Who have you turned to for support?

**Understanding and experience of receiving the result**

**Can you tell me about the experience of receiving the genetic test result?**
Prompts:
• How were you given the result?
• What did the genetic counselor/ doctor tell you about your result?
• How did you feel about the way the genetic counselor/ doctor gave you the result?
• How did you feel when you received the result?
• How did you react?

**Experiences leading up to the result**

**How was it for you during the time between the genetics appointment and receiving the result?**
Prompts:
• How did you feel during that waiting time?
• How did you get through this period?
• Who did you turned to for support?
• (If information seeking) Where did you go to for information?

**Understanding and experience of treatment plan in the context of the genetic test result**

**Can you tell me what you recall about your consultation with the Breast Team/ multidisciplinary team after you received your genetic test result?**
Prompts:
• Can you recall what the doctor/ other clinicians said to you about the result? (impact on treatment/ future risks/ family)
• What treatment plan has been made for you?
• How did you feel about the way the doctor/ other clinicians talked with you about genetics/ cancer?
• What did you understand from that/ those appointment(s)?
• How did you feel during the discussions?
• How did you react during the discussions?

Present experiences
How have you been feeling since the decisions were made about your treatment?
Prompts:
• How do you feel about the decisions that have been made? (mastectomy/ lumpectomy)
• (If information seeking) Where have you gone to for information?
• Who have you turned to for support?
• What is important for you now?
• What do you expect to happen next?
• How do you feel about what will happen next?

The future
How do you feel about the future now that you know this information?
Prompts:
• How do you feel about the future for yourself?
• How do you feel about the future for your family?
• What is important for you now?

Reflections on the whole experience
How do you feel about having learnt that the cancers in your family are/ could be your hereditary so close to your diagnosis?
Prompts:
• How would you describe the whole experience?
• How has the whole experience made you feel?
• If you were talking to a woman who is about to go through all of this what would you say to her?
• Is there anything else you would like to share with me about the whole experience?
Appendix 3:

Other study documents
### Coding scheme for identifying key messages

<table>
<thead>
<tr>
<th>Generic Knowledge</th>
<th>Testing information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>5</strong> The fault is in the (BRCA1 or BRCA2) gene (ie specifying the name of the gene involved).</td>
<td>There are three possible results of a BRCA1 or BRCA2 genetic test for a person with cancer: a cancer-causing gene fault is found which means that a predictive (targeted) genetic test is available for 'blood relatives'; no cancer-causing gene fault is found, meaning that it is very unlikely that the cancer is due to a BRCA1 or BRCA2 gene fault and a predictive genetic test is not available for relatives; a gene change is found that may or may not cause cancer. This is called a variant of unknown significance (VUS) or an unclassified variant (UV). This result means that a predictive (targeted) genetic test is not available for relatives.</td>
</tr>
<tr>
<td><strong>7</strong> BRCA1 and BRCA2 genes are DNA damage repair genes. A fault in one of these genes results in a loss of their function and increases the risk of breast, ovarian and prostate cancer.</td>
<td></td>
</tr>
<tr>
<td><strong>21</strong> In the future it is likely that research will lead to greater understanding about the role of BRCA1/2 genes, their interaction with other genes and the role of lifestyle factors in the development of cancer.</td>
<td></td>
</tr>
<tr>
<td><strong>1</strong> The children of a person with a BRCA1 or BRCA2 gene fault each have a 50% (1 in 2) risk of inheriting the gene fault.</td>
<td>Predictive (targeted) genetic testing for a BRCA1 or BRCA2 gene fault is not generally offered before the age of 18.</td>
</tr>
<tr>
<td><strong>35</strong> Diet and lifestyle can make a difference to the risk of cancer generally but the impact is likely to be small compared with the risk associated with the BRCA1 or BRCA2 gene fault.</td>
<td>Predictive (targeted) genetic testing is available for relatives once a BRCA1 or BRCA2 gene fault has been identified. This will show whether or not the person has inherited the known faulty gene, and so predicts whether they might be at risk (this is called a predictive test).</td>
</tr>
<tr>
<td><strong>3</strong> A BRCA1 or BRCA2 gene fault does not 'skip' a generation</td>
<td>Some people feel guilty about the possibility that their children and grandchildren may inherit the faulty gene from them.</td>
</tr>
<tr>
<td><strong>6</strong> If a person inherits a BRCA1 or BRCA2 gene fault their risks of breast/ovarian/prostate cancer will be significantly increased but it is not inevitable that they will develop cancer.</td>
<td>If a person does not inherit a known BRCA1 or BRCA2 gene fault, their risks of breast/ovarian/prostate cancer will be similar to other people in the general population.</td>
</tr>
<tr>
<td><strong>56</strong> In the general population approximately 1 woman in 2 develops breast cancer in her lifetime and 1 woman in 50 develops ovarian cancer. Most breast and ovarian cancer occurs after the age of 50 and is not due to a faulty BRCA1 or BRCA 2 gene.</td>
<td>Genetic testing can lead to complex emotions which may be unexpected, like shock, fear, sadness and upset, especially close to the result. The health care team can provide information about the support that is available to help with this.</td>
</tr>
<tr>
<td><strong>54</strong> Both men and women can inherit a BRCA1 or BRCA2 gene fault. Therefore, if either parent carries a gene fault, each child will have a 50% (1 in 2) chance of inheriting it from them.</td>
<td>Once a gene fault has been found in a family it is up to each individual to decide if they want a genetic test or not. Some relatives may decide not to be tested.</td>
</tr>
<tr>
<td><strong>4</strong> It is possible that breast/ovarian/prostate cancers in the family can be explained by a BRCA1 or BRCA2 gene fault.</td>
<td>It is important to try and inform all your relatives who are at risk that genetic testing is available.</td>
</tr>
<tr>
<td><strong>2</strong> Identifying which side of the family is at risk of hereditary cancer is important.</td>
<td>Before having a genetic test it is important to discuss the implications and possible outcomes</td>
</tr>
<tr>
<td><strong>58</strong> Cancer risks may vary for each individual with a gene fault depending on genetic, environmental and lifestyle factors as well as personal and family history of cancer.</td>
<td>A personal diagnosis or family history of cancer may affect insurance. There is however currently an agreement between the British Government and the Association of British insurers which means that people who have had a predictive (targeted) genetic test are not required to disclose the results in order to obtain insurance. There are financial limits to these policies. The agreement is in place until 2017 and it is likely to be extended but this cannot be guaranteed.</td>
</tr>
<tr>
<td><strong>65</strong> Unsolicited information that is not on the above list</td>
<td></td>
</tr>
<tr>
<td><strong>66</strong> Information that is not on the above list but that is given in response to patient’s questions or required to explain patient’s specific circumstances</td>
<td></td>
</tr>
<tr>
<td>Consequences and risks</td>
<td>Specific to individuals with cancer</td>
</tr>
<tr>
<td>------------------------</td>
<td>-------------------------------------</td>
</tr>
<tr>
<td>16 Women with breast cancer who have a BRCA1 or BRCA2 gene fault are at increased risk of developing further primary breast cancers</td>
<td>26 Breast cancer risk is increased for women without cancer who have a BRCA1 or BRCA2 gene fault.</td>
</tr>
<tr>
<td>17 Women who have/have had ovarian/ tubal cancer and have a BRCA1 or BRCA2 gene fault are at increased risk of developing breast cancer.</td>
<td>27 For women with a fault in BRCA1 or BRCA2, the risk of developing breast cancer between age 25 and 30 may be higher than for women in the general population, but most of the risk occurs after the age of 30.</td>
</tr>
<tr>
<td>62 For a woman who has had breast or ovarian cancer, a Risk Reducing or Contralateral Mastectomy will reduce the risk of a future new primary breast cancer but will not reduce the risk of metastases from the initial cancer.</td>
<td>28 For women who have a BRCA1 or BRCA2 gene fault, ovarian cancer (including fallopian tube and primary peritoneal cancer) risk is increased.</td>
</tr>
<tr>
<td>20 Women with breast cancer who have a BRCA1 or BRCA2 gene fault are at increased risk of ovarian/tubal cancer.</td>
<td>29 For women with a fault in BRCA1, the risk of developing ovarian/ tubal cancer before age 40 may be increased but most of the risk occurs after the age of 40. For women with a fault in BRCA2, most of the risk occurs after the age of 45.</td>
</tr>
<tr>
<td>57 Genetic testing may provide helpful risk and management information for women with cancer, as well as for relatives who have not had cancer.</td>
<td>34 A BRCA2 gene fault may slightly increase the risk of other cancers, such as pancreatic, gall bladder and bile duct cancer. However the risks are small and there is no screening available.</td>
</tr>
<tr>
<td>25 A BRCA1 or BRCA2 gene fault does not increase the risk of cancer recurrence or metastases (secondaries).</td>
<td>33 Pancreatic cancer risk is not increased by a BRCA1 gene fault.</td>
</tr>
<tr>
<td></td>
<td>61 Identifying a faulty gene can help people to be more aware of the symptoms of cancer.</td>
</tr>
<tr>
<td>Risks to men</td>
<td></td>
</tr>
<tr>
<td>30 Prostate cancer risk is increased for men who have a BRCA1 or BRCA2 gene fault. The risk for men with BRCA2 fault is higher than for men with a BRCA1 fault.</td>
<td>44 Ovarian screening has not yet been shown to be effective. Therefore no NHS ovarian screening programme is available. However, women who have symptoms such as fatigue, bloating, loss of appetite or unexplained weight loss are advised to see their General Practitioner.</td>
</tr>
<tr>
<td>31 Male breast cancer risk is not increased by a BRCA1 gene fault.</td>
<td></td>
</tr>
<tr>
<td>32 The breast cancer risk for men with a BRCA2 fault is 5-10% throughout their lifetime. Men are advised to check their chest and underarms for changes such as lumps, nipple discharge or skin changes and report any changes promptly to their GP but no regular breast screening is recommended.</td>
<td>53 Once a BRCA1 or BRCA2 gene fault has been identified, it may be helpful to have a discussion with other specialists in the multidisciplinary team (e.g. oncoplastic breast surgeon, gynaecologist and/or clinical psychologist) in order to understand all the options available.</td>
</tr>
</tbody>
</table>
Appendix 3.2. Coding guidelines: Identifying recommendations from the genetics and cancer guidelines (Study 4)

<table>
<thead>
<tr>
<th>Genetics guidelines for women with or at risk of hereditary breast and ovarian cancer about genetic risk assessment, genetic testing and risk management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Identifying recommendations about what should be communicated about genetic risk assessment, genetic testing and risk management to women with or at risk of hereditary breast and ovarian cancer:</td>
</tr>
<tr>
<td>• Recommendations that directly refer to information that should be discussed with individuals with or at risk of hereditary breast and ovarian cancer should be coded as 'what'. For example, 'Discussion of the probability of finding a mutation, the implications for the individual and the family, and the implications of either a variant of uncertain significance or a null result (no mutation found)' or 'Breastfeeding should be encouraged'.</td>
</tr>
<tr>
<td>• Recommendations that refer to an action to be taken by the health professional and infer information that should be communicated to the woman with or at risk of should be coded as 'what'. For example, 'Women at increased risk of breast cancer should be breast aware in line with Department of Health advice for all women' or 'Annual screening MRI should be commenced from the age of 25 with the addition of annual mammography from the age of 30'.</td>
</tr>
<tr>
<td>• Recommendations that management could be 'considered' or should not be offered imply that the patient does not require information unless specifically stated and should be coded as 'not applicable' (N/A). For example, the following recommendation would not be required: 'Contralateral RRM among patients with a previous breast cancer diagnosis can be considered', whereas this one would: 'Before RRSO, 6-monthly, trans-vaginal ultrasound and measures of serum CA125 may be considered from the age of 30; however, the limited value of these tools as an effective screening measure should be communicated to individuals'.</td>
</tr>
<tr>
<td>Identifying recommendations about how communication about genetic risk, testing or risk management should take place with women with or at risk of hereditary breast and ovarian cancer:</td>
</tr>
<tr>
<td>• Recommendations that refer to the communication style or approach or action that should be taken by the health professional when communicating about hereditary breast and ovarian cancer should be coded as 'how'. For example, 'People should be sent a written summary of their consultation in a specialist genetic clinic, which includes their personal risk information' or 'Predictive genetic testing should not be offered without adequate genetic counselling.'</td>
</tr>
<tr>
<td>N.B. Some recommendations include aspects of 'what' and 'how' and should be coded as both, for example 'Women considering risk-reducing bilateral oophorectomy should be informed of possible psychosocial and sexual consequences of the procedure (what) and have the opportunity to discuss these issues (how)' or 'Follow-up counselling (how) outlining options for screening for early detection, risk-reducing measures and issues pertaining to fertility in women who have not completed their family (what) is fundamental'.</td>
</tr>
<tr>
<td>Recommendations from the genetics guidelines about how communication should take place with women with cancer about hereditary breast and ovarian cancer will be coded separately to recommendations made by the cancer guidelines about how communication should take place about cancer generally</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Cancer guidelines for women with breast or ovarian cancer about cancer management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Identifying recommendations about what should be communicated about hereditary breast and ovarian cancer to women with cancer:</td>
</tr>
<tr>
<td>• Recommendations that directly refer to information that should be discussed with individuals with breast or ovarian cancer about hereditary cancer should be coded as 'what'.</td>
</tr>
<tr>
<td>• Recommendations that refer to an action to be taken by the health professional and infer information that should be communicated to the woman with breast or ovarian cancer about hereditary cancer should be coded as 'what'.</td>
</tr>
<tr>
<td>• Recommendations that management could be 'considered' or should not be offered imply that the patient does not require information unless specifically stated and should not be coded as 'not applicable' (N/A)</td>
</tr>
<tr>
<td>Identifying recommendations about how communication about cancer should take place with women with breast or ovarian cancer:</td>
</tr>
<tr>
<td>• Recommendations that refer to the communication style or approach or action that should be taken by the health professional when communicating about cancer diagnosis, surgery and treatment should be coded as 'how'. For example, 'Assess the patient's individual preference for how much they wish to be involved in decision making. Reassess this as circumstances change' or 'All young women should be referred for special counselling/consultation if interested in fertility preservation prior to commencement of any therapy.'</td>
</tr>
</tbody>
</table>
### Appendix 3.3. Coding guidelines: identifying information and method of communication for cancer patients (Study 4)

**What should be communicated**

i. The recommendation is present in the cancer and the genetics guidelines or in the cancer guidelines only;

ii. The recommendation was agreed a key message for women with breast or ovarian cancer by expert health professionals and/ or service users in Study 2, unless it was also judged as not key by the other group

iii. The recommendation does not meet criteria A or B.

<table>
<thead>
<tr>
<th>Recommendation from guidelines</th>
<th>Criteria for selection</th>
<th>Guideline number</th>
<th>Agreement as key/ not key message by health professionals and/ or service users in Study 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Possibility of HBOC and potential risk to relatives.</td>
<td>i.</td>
<td>Genetics: 1, 3, 8 Cancer: 14, 21</td>
<td></td>
</tr>
<tr>
<td>Potential risk and benefits of genetic testing specific to the type of test being offered, including the implications for the individual and the family and the purpose of testing.</td>
<td>ii.</td>
<td>Genetics: 1, 3, 4, 5, 6, 8</td>
<td>Agreed as key by health professionals and service users</td>
</tr>
<tr>
<td>Potential cancer risks, including penetrance and variable expression.</td>
<td>ii.</td>
<td>Genetics: 1, 3, 4, 6, 7</td>
<td>Agreed as key by health professionals and service users</td>
</tr>
<tr>
<td>Potential options for prevention, early diagnosis and surveillance.</td>
<td>ii.</td>
<td>Genetics: 1, 3, 4, 5, 6</td>
<td>Agreed as key by health professionals and service users</td>
</tr>
<tr>
<td>Autosomal dominant inheritance pattern.</td>
<td>ii.</td>
<td>Genetics: 1, 3, 6, 7</td>
<td>Agreed as key by health professionals and service users</td>
</tr>
<tr>
<td>Reliability, limitations and informativeness of the genetic test, the probability of finding a mutation, the possibility of a VUS and the likely timescale of results. (Where panel testing is offered - potential for incidental and secondary germline information specific to the test being offered, the relevance and potential benefits and the limitations for the individual and family including the possibility and implications of incidental findings.)</td>
<td>ii.</td>
<td>Genetics: 1, 2, 3, 4, 6, 7, 8</td>
<td>Agreed as key by health professionals only</td>
</tr>
<tr>
<td>Legal ethical and social implications of genetic testing including the right to decide not to know.</td>
<td>ii.</td>
<td>Genetics: 3, 6, 8</td>
<td>Agreed as key by health professionals and service users</td>
</tr>
<tr>
<td>Informing at risk relatives about the availability of genetic testing and/ or surveillance.</td>
<td>ii.</td>
<td>Genetics: 2, 3, 6, 7</td>
<td>Agreed as key by health professionals and service users</td>
</tr>
<tr>
<td>Ovarian symptom awareness.</td>
<td>i. ii.</td>
<td>Genetics: 7 Cancer: 19</td>
<td>Agreed as key by health professionals and service users</td>
</tr>
<tr>
<td>Type and frequency of breast surveillance available according to age and risk.</td>
<td>i. ii.</td>
<td>Genetics: 1, 2, 4, 7, Cancer: 21, 27, 28</td>
<td>Agreed as key by health professionals and service users</td>
</tr>
<tr>
<td>Topic</td>
<td>Section</td>
<td>Genetics</td>
<td>Agreed as key by health professionals and service users</td>
</tr>
<tr>
<td>----------------------------------------------------------------------</td>
<td>---------</td>
<td>----------</td>
<td>---------------------------------------------------------</td>
</tr>
<tr>
<td>Risks, benefits and limitations of breast surveillance (including written information). (For TP53 carriers with breast cancer - the risks of malignancy associated with radiation).</td>
<td>ii.</td>
<td>Genetics: 1, 9</td>
<td>Agree as key by health professionals and service users</td>
</tr>
<tr>
<td>Limitations and availability of ovarian surveillance.</td>
<td>i. ii.</td>
<td>Genetics: 2, 4, 7 Cancer: 16, 19, 28</td>
<td>Agree as key by health professionals and service users</td>
</tr>
<tr>
<td>Risks and benefits of risk reducing mastectomy, including possible outcomes, risk reduction, the possibility of cancer being diagnosed histologically, possibility of immediate or delayed reconstruction and differences in the look and feel of the reconstructed breast.</td>
<td>i. ii.</td>
<td>Genetics: 1, 2, 4, 7, 9 Cancer: 17, 21</td>
<td>Agree as key by health professionals and service users</td>
</tr>
<tr>
<td>Clear quantification of the risk of contralateral breast cancer and the timing, risks and benefits of contralateral mastectomy, including relief of anxiety about developing breast cancer, the likely prognosis of breast cancer and the risk of distal recurrence of their previous breast cancer. (For women who have breast conserving treatment, adjuvant endocrine therapy should be used when appropriate based on hormone receptor status to reduce the risk of ipsilateral and contralateral events.)</td>
<td>i. ii.</td>
<td>Genetics: 1, 9 Cancer: 17, 26</td>
<td>Agree as key by health professionals and service users</td>
</tr>
<tr>
<td>Options, risks and benefits of immediate and delayed reconstruction should be discussed with a member of a surgical team with specialist skills in oncoplastic surgery or breast reconstruction</td>
<td>ii.</td>
<td>Genetics: 1, 2, 4</td>
<td>Agree as key by health professionals and service users</td>
</tr>
<tr>
<td>Psychosocial and sexual consequences of surgery, including adjustment to the reconstructed breast and changed body image, loss of sensation, emotional well-being and quality of life and details of local and national support services.</td>
<td>i.</td>
<td>Genetics: 1, 2, 7 Cancer: 17, 20</td>
<td></td>
</tr>
<tr>
<td>Risks, benefits and timing of bilateral salpingo-oophorectomy including early menopause, ovarian and breast cancer risk reduction and fertility issues.</td>
<td>ii.</td>
<td>Genetics: 1, 2, 4, 7, 9 Cancer: 16, 28</td>
<td>Agree as key by health professionals and service users</td>
</tr>
<tr>
<td>What to expect from risk assessment and genetic counselling.</td>
<td>iii.</td>
<td>Genetics: 1</td>
<td>Agree as not key by health professionals and service users</td>
</tr>
<tr>
<td>Personal genetic risk assessment the uncertainties of risk estimation and advice to seek re-assessment if the family history changes or symptoms develop.</td>
<td>iii.</td>
<td>Genetics: 1</td>
<td>Agree as not key by health professionals and service users</td>
</tr>
<tr>
<td>Cancer in the general population (including breast and ovarian cancer and age as a risk factor).</td>
<td>iii.</td>
<td>Genetics: 1</td>
<td></td>
</tr>
<tr>
<td>Potential: reproductive choices for carriers</td>
<td>iii.</td>
<td>Genetics: 1, 2, 3, 4, 6, 7</td>
<td></td>
</tr>
<tr>
<td>Privacy, confidentiality and possible consequences related to disclosure of the result.</td>
<td>iii.</td>
<td>Genetics: 3, 6</td>
<td></td>
</tr>
<tr>
<td>Psychosocial consequences of genetic testing for the individual and family, including information about support groups and voluntary organisations.</td>
<td>iii.</td>
<td>Genetics: 3, 4, 6, 8</td>
<td>Agree as not key by health professionals and service users</td>
</tr>
</tbody>
</table>
### Modifiable hormonal and reproductive risk factors, including OCP, HRT and breast feeding.

- **Criteria for selection:** iii.
- **Guideline no.:** Genetics: 1, 2, 4
- **Genetics/ Cancer guideline recommendations from which the synthesised recommendations are drawn:**

### Modifiable lifestyle risk factors, including alcohol, smoking, being overweight and exercise.

- **Criteria for selection:** iii.
- **Guideline no.:** Genetics: 1, 2
- **Genetics/ Cancer guideline recommendations from which the synthesised recommendations are drawn:**

### Breast awareness and clinical breast examination.

- **Criteria for selection:** iii.
- **Guideline no.:** Genetics: 1, 2, 4, 7
- **Genetics/ Cancer guideline recommendations from which the synthesised recommendations are drawn:**

### Limitations and availability of surveillance for other BRCA1/2-related cancers.

- **Criteria for selection:** iii.
- **Guideline no.:** Genetics: 2, 4
- **Genetics/ Cancer guideline recommendations from which the synthesised recommendations are drawn:**

### Symptom awareness and screening for male carriers.

- **Criteria for selection:** iii.
- **Guideline no.:** Genetics: 2, 4, 7
- **Genetics/ Cancer guideline recommendations from which the synthesised recommendations are drawn:**

### Psychosocial and sexual consequences of risk reducing bilateral salpingo-oophorectomy.

- **Criteria for selection:** iii.
- **Guideline no.:** Genetics: 1, 2, 7
- **Genetics/ Cancer guideline recommendations from which the synthesised recommendations are drawn:**

### Options, risks and benefits of PGD and prenatal testing options for BRCA1/2 carriers.

- **Criteria for selection:** iii.
- **Guideline no.:** Genetics: 2, 7
- **Genetics/ Cancer guideline recommendations from which the synthesised recommendations are drawn:**

### Risks and benefits of OCP and HRT use for women at high risk of HBOC.

- **Criteria for selection:** iii.
- **Guideline no.:** Genetics: 1
- **Genetics/ Cancer guideline recommendations from which the synthesised recommendations are drawn:**

### How communication should take place

i. The recommendation is in the cancer guidelines;

ii. The recommendation is about how genetic testing should be offered to women with breast or ovarian cancer and those at risk of HBOC;

iii. The recommendation does not meet criteria A or B.

<table>
<thead>
<tr>
<th>Recommendation</th>
<th>Criteria for selection</th>
<th>Guideline no.</th>
<th>Genetics/ Cancer guideline recommendations from which the synthesised recommendations are drawn</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ensure information is of evidence-based clear and consistent and does not contradict messages from other service providers.</td>
<td>i.</td>
<td>Genetics: 1</td>
<td>Ensure standard information is evidence-based and does not contradict messages from other service providers. Ensure information (written and verbal) is full, clear, accurate, objective, consistent, evidence-based, up to date and tailored to the individual - mindful of changing needs over time.</td>
</tr>
<tr>
<td>Ensure information in a suitable format that is accessible, understandable, culturally appropriate and tailored to the individual.</td>
<td>i.</td>
<td>Genetics: 1, 3, 6, 10, 11, 12, 13, 14, 15, 17, 18, 20, 25, 27, 28 Cancer: 10, 11, 12, 14, 18, 22, 27, 28</td>
<td>Provide information that is tailored to the individual in a choice of format and appropriate language. Ensure information is in a suitable format, comprehensible, culturally appropriate and accessible to people with additional needs.</td>
</tr>
<tr>
<td>Provide written information to support discussions.</td>
<td>i.</td>
<td>Genetics: 1, 3, 8 Cancer: 11, 12, 17, 19</td>
<td>Provide written information to support discussions. Provide patients with a permanent record of consultations.</td>
</tr>
<tr>
<td>Provide help with navigating and understanding and information and disseminating genetic information to relatives.</td>
<td>i. ii.</td>
<td>Genetics: 3 Cancer: 11, 18, 24</td>
<td>Provide assistance with informing relatives. Provide help with navigating and understanding and disseminating information.</td>
</tr>
</tbody>
</table>

---

**Appendix**

371
<table>
<thead>
<tr>
<th>Topic</th>
<th>Genomics/Genetics</th>
<th>Cancer</th>
<th>Remarks</th>
</tr>
</thead>
<tbody>
<tr>
<td>Facilitate shared decision-making and informed consent.</td>
<td>i.</td>
<td>Genetics: 1, 3, 6, 10, 11, 12, 13, 14, 15, 17, 19, 20, 22</td>
<td>Facilitate shared decision-making and informed consent according to capacity.</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Facilitate shared decision-making, involving families and carers with the patient's agreement.</td>
</tr>
<tr>
<td>Provide adequate time for decision-making and respect the decisions made.</td>
<td>i.</td>
<td>Genetics: 1, 3, 10, 11, 17, 18, 20, 28</td>
<td>Provide adequate time for decision-making and respect decisions made.</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Provide adequate time for the patient to ask questions, have them answered and to reflect before making decisions.</td>
</tr>
<tr>
<td>Offer timely psychological and social support and further counselling to patients concerned about genetic risk or risk reducing surgery. Provide the opportunity to talk to women who have undergone the procedure. Where appropriate offer support to carers and families.</td>
<td>i. ii.</td>
<td>Genetics: 1, 3, 2, 8, 10, 11, 12, 14, 16, 17, 18, 20, 22, 23, 24</td>
<td>Offer women concerned about genetic risk access to psychological and/or social support and counselling.</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Offer women considering risk reducing surgery counselling and access to additional psychological support, including the opportunity to talk to women who have undergone the procedure.</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Provide timely access to expert psychosocial support for patients, families and carers including specialist referral where required.</td>
</tr>
<tr>
<td>Ensure continuity, avoid unnecessary repeated assessments from different health professionals aiming to elicit similar information and provide contact details for a named point of contact in genetics and oncology.</td>
<td>i. ii.</td>
<td>Genetics: 1, 2, 3, 9, 10, 11, 12, 14, 15, 17, 18, 19</td>
<td>Involve of all members of the multidisciplinary team and provide a designated genetics contact.</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Ensure continuity of care and avoid unnecessary repeated assessments from different professionals aiming to elicit similar information.</td>
</tr>
<tr>
<td>Involve relevant members of the multidisciplinary team, provide timely referral for expert discussion about surgical options, and offer a follow up plan and second opinion where required.</td>
<td>i.</td>
<td>Genetics: 1, 2, 3, 10, 11, 12, 14, 18, 24, 26, 27, 28</td>
<td>Offer women considering risk reducing mastectomy referral to oncoplastic surgeons to discuss options, including immediate and delayed reconstruction.</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Offer a follow up plan and provide further consultations and a second opinion as required.</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Facilitate access to specialist consultation and multidisciplinary input when required.</td>
</tr>
<tr>
<td>Ensure health professionals are appropriately trained in genetic risk assessment, genetic counselling, genetic testing, psychosocial needs assessment.</td>
<td>i. ii.</td>
<td>Genetics: 1, 2, 3, 4, 5, 6, 7, 8, 10, 11, 12, 14, 18, 20, 22</td>
<td>Offer cancer risk assessment and genetic counselling prior to genetic testing if appropriate by an appropriately trained health professional with experience and expertise in cancer genetics.</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Ensure all health care personnel are trained and competent in psychosocial needs assessment and communication skills. Offer genetic risk assessment, genetic counselling and genetic testing by an appropriately trained health professional regardless of the time frame for testing.</td>
</tr>
<tr>
<td>Where possible offer genetic testing to individuals with breast or ovarian cancer and</td>
<td>i. ii.</td>
<td>Genetics: 1, 5, 10, 11, 12, 14, 18, 20, 22</td>
<td>Where possible offer genetic testing to individuals with breast or ovarian cancer and at least a 10% probability of a genetic disorder.</td>
</tr>
</tbody>
</table>
| at least a 10% probability of a pathogenic variant before testing unaffected relatives. | Cancer: 21 | pathogenic variant before testing unaffected relatives.  
Where possible offer genetic testing to individuals with breast or ovarian cancer and at least a 10% probability of a pathogenic variant before testing unaffected relatives. |
<table>
<thead>
<tr>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Offer genetic testing during initial cancer management to inform decision-making and treatment. If women are not ready to consider testing at diagnosis, offer again at follow up.</td>
<td>i. ii. Genetics: 1, 4, 9, Cancer: 22, 28</td>
<td>Offer genetic testing during initial cancer management to inform decision-making and treatment. If women are not ready to consider testing at diagnosis, offer again at follow up. Offer genetic counselling before the start of treatment where the result may impact on treatment decisions or clinical trials. If women are not ready to consider testing at diagnosis, offer again at follow up.</td>
</tr>
<tr>
<td>Establish an environment conducive to good communication, including honesty and transparency, confidentiality and privacy and respect for the views of the individual.</td>
<td>i. Cancer: 10, 11, 17, 18, 20, 22, 23, 28</td>
<td></td>
</tr>
<tr>
<td>Assess risk using a validated model and present personal risk assessment in several ways to facilitate understanding.</td>
<td>ii. Genetics: 1, 3, 4</td>
<td></td>
</tr>
<tr>
<td>Provide interpretation of genetic test results by a trained health professional with experience and expertise in cancer genetics.</td>
<td>ii. Genetics: 6, 7, 8</td>
<td></td>
</tr>
<tr>
<td>Offer genetic testing in specialist genetic clinics to a person with no personal history of breast or ovarian cancer if their combined BRCA1 and BRCA2 mutation carrier probability is 10% or more and an affected relative is unavailable for testing</td>
<td>iii. Genetics: 1</td>
<td></td>
</tr>
<tr>
<td>Offer predictive genetic testing to adults after pre-test genetic counselling and with informed consent</td>
<td>iii. Genetics: 1, 2, 6, 7</td>
<td></td>
</tr>
</tbody>
</table>
### Appendix 3.4. Coding guidelines: identifying references to guideline recommendations in clinical protocols and patient leaflets (Study 4)

<table>
<thead>
<tr>
<th>Recommendation no.</th>
<th>The recommendation is applicable to documents that address:</th>
<th>The recommendation is accepted as communicated when there is reference to:</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Genetic testing prior to the test being undertaken.</td>
<td>The percentage of breast or ovarian cancer that is hereditary AND/ OR eligibility criteria for genetic testing. May include genes involved.</td>
</tr>
<tr>
<td>2</td>
<td>The implications of genetic testing prior to the test being undertaken. Not applicable to documents that only address testing criteria.</td>
<td>The potential risks or benefits of genetic testing for the individual AND/ OR the family OR a discussion has taken/ will take place about the risks and benefits of testing. ‘BRCA gene testing discussed’ is not acceptable.</td>
</tr>
<tr>
<td>3</td>
<td>Increased cancer risks associated with a mutation. May include penetrance AND/ OR variable expression. Note ‘BRCA gene testing discussed’ is not acceptable.</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>Screening AND/ OR risk reducing surgery. Note ‘BRCA gene testing discussed’ is not acceptable</td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>Inheritance. Note ‘BRCA gene testing discussed’ is not acceptable</td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>Reliability, limitations (may include VUS) AND/ OR informativeness of the test. Note timescale of testing alone is not acceptable.</td>
<td></td>
</tr>
<tr>
<td>7</td>
<td>Genetic testing prior to the test being undertaken</td>
<td>Legal, ethical AND/OR social implications of genetic testing OR the right not to know. Note at pre-test genetic counselling this will apply to women with cancer considering diagnostic genetic testing. At post-test genetic counselling this will apply to information for relatives eligible for predictive testing.</td>
</tr>
<tr>
<td>8</td>
<td>Genetic testing prior to the test being undertaken</td>
<td>Informing relatives about the availability of screening AND/ OR genetic testing. Note this may apply to pre- and post-test genetic counselling.</td>
</tr>
<tr>
<td>9</td>
<td>Breast cancer risk and management.</td>
<td>The type, frequency AND/ OR age of starting screening</td>
</tr>
<tr>
<td>10</td>
<td>Applicable to documents referred to in 9 (i.e. addressing breast screening).</td>
<td>The risks AND/ OR limitations of screening even if the benefits are not referred to</td>
</tr>
<tr>
<td>11</td>
<td>Ovarian cancer risk and management.</td>
<td>The limitations of ovarian screening AND/OR the availability of screening</td>
</tr>
<tr>
<td>12</td>
<td>Breast cancer risk and management</td>
<td>The risks AND/ OR benefits of surgery or it is implied that a discussion has taken/ will take place.</td>
</tr>
<tr>
<td>13</td>
<td>Applicable to documents referred to 12 (i.e. addressing breast cancer risk management).</td>
<td>The risk of contralateral breast cancer AND/ OR the risks of contralateral mastectomy</td>
</tr>
<tr>
<td>14</td>
<td>Surgical management of breast cancer risk</td>
<td>The option, risk AND/ OR benefit of reconstruction or referral to a specialist surgeon.</td>
</tr>
<tr>
<td>15</td>
<td>Risk reducing mastectomy or psychological impact of breast surgery</td>
<td>The psychosocial AND/ OR sexual consequences AND details of support services. Note ‘Complex issues’ or reference to websites without further information is not acceptable.</td>
</tr>
<tr>
<td>16</td>
<td>Applicable to documents referred to in 11 (i.e. addressing ovarian cancer risk management)</td>
<td>Risks, benefits AND/ OR timing of RRBSO.</td>
</tr>
<tr>
<td>Reason for rejection</td>
<td>Rejected guidelines</td>
<td></td>
</tr>
<tr>
<td>-------------------------------------------------------------------------------------</td>
<td>--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------</td>
<td></td>
</tr>
<tr>
<td>Recommendations from an international expert panel on the use of neoadjuvant (primary) systemic treatment of operable breast cancer: new perspectives (Kaufmann et al., 2007)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Superseded/rescinded/ out of date with no new guidance available (n=5)</td>
<td>ESMO BRCA in Breast cancer (Balmaña et al., 2011)</td>
<td>ESMO Advanced breast cancer 2 (Cardoso et al., 2014)</td>
</tr>
<tr>
<td>---</td>
<td>---</td>
<td>---</td>
</tr>
<tr>
<td></td>
<td>NHS London Psychological support for people living with cancer - commissioning guidance for cancer care</td>
<td></td>
</tr>
</tbody>
</table>
References for rejected guidelines


References for rejected guidelines continued


Appendix 4:

Peer-reviewed publications