UPTAKE AND ACCEPTABILITY OF GENETIC COUNSELLING AMONGST BREAST CANCER PATIENTS AND THEIR FAMILIES IN THE UMGUNGUNDLOVU MUNICIPALITY IN 2004

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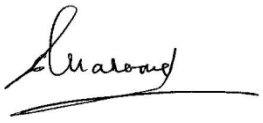

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ABSTRACT

Awareness of genetic counselling and risk assessment is imperative for women to seek out genetic services. The purpose of this study is to determine the level of genetic counselling and acceptability of genetic counselling by breast cancer survivors, at the time of diagnosis, and the female members of their families, after the diagnosis of the patient within their family, with a view to developing recommendations based on the findings of the study. Two questionnaires were administered to a conveniently selected sample of 48 women; 28 were breast cancer survivors from the Cancer Association of South Africa's breast cancer support group and 20 women from the general public. Interviews were then conducted with four randomly selected patients and their family members. A total of 12 participants were interviewed: four breast cancer survivors and eight family members, including two spouses and six children. During the interviews one breast cancer patient who was counselled by a psychologist indicated that she was not satisfied with the information she had been given and that further information on the genetics, recurrence, prognosis and family risk should have been provided as she considered this to be essential. The three patients who were not counselled were of the opinion that counselling on the genetics and risks of breast cancer would have been very useful. Four (50%) of the eight family members interviewed, indicated that they were not offered genetic counselling and would not be interested in knowing about the risks associated with breast cancer. Of the remaining four, two (25%) indicated that although they were not offered genetic counselling they would be interested in genetic counselling and learning about the risks associated with breast cancer as such information would be of great value. In terms of a breast cancer risk assessment, four of the eight family members (50%) indicated they were not interested with one further explaining that she might be interested at a later stage. The remaining four family members (50%) indicated their interest in attending a breast cancer risk assessment. In conclusion, educational interventions are required to increase awareness of genetic counselling and risk assessment.

DECLARATION

I, Bilqees Banoo Sayed, declare that:

(i) The research reported in this dissertation, except where otherwise indicated, is my original work.

(ii) This dissertation has not been submitted for any degree or examination at any other university.

(iii) This dissertation does not contain other persons' data, pictures, graphs or other information, unless specifically acknowledged as being sourced from other persons.

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DEDICATION

This study is dedicated to Mr Mehmood and Mrs Rooksana Sayed, my loving parents; Abdool Khalik Razak, my wonderful husband, and Sanáa and Rayhaan, my amazing children, for all their encouragement, patience and being my pillars of strength and inspiration.

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CHAPTER I: INTRODUCTION

1.1 Introduction

Cancer refers to any one of a large number of diseases characterized by the development of abnormal cells that divide uncontrollably and have the ability to infiltrate and destroy normal body tissue. Cancer also has the ability to spread throughout the body (Mayoclinic, 2014). The development of any form of cancer is a multi-step process characterized by genetic alterations that influence key cellular pathways involved in growth and development (Osborne *et al.*, 2004).

Cancer cells have five basic properties, namely their ability to

- grow uncontrollably,
- invade surrounding tissues,
- enter the bloodstream or other channels, such as lymph vessels, and travel to different locations in the body,
- establish secondary tumours in other organs, and
- produce substances that interfere with the control of various body functions, such as affecting nerves, muscles, and salt regulation (Buckman, 1997).

1.2 Epidemiology of cancer

According to the 2004 Global Burden of Disease, cancers ranked as the third-highest cause of mortality in men and women globally, accounting for 13.4% in men and 11.8% in women (WHO, 2008). Worldwide, there were 14.1 million new cancer cases, 8.2 million cancer deaths and 32.6 million people living with cancer (within a 5-year period of diagnosis) in 2012 (International Association for Research on Cancer [IARC], 2013). An estimated 57% (8 million) of the new cancer cases, 65% (5.3 million) of the cancer deaths and 48% (15.6 million) of the 5-year prevalent cancer cases occurred in the developing regions, which include south-eastern Asia, south-central Asia, eastern, western, northern and middle Africa, and central America (IARC, 2013).

Carcinoma of the lung (1.8 million, 13.0% of the total), breast (1.7 million, 11.9%), and colorectal cancer (1.4 million, 9.7%) were the most commonly diagnosed cancers

worldwide whilst cancers of the lung (1.6 million, 19.4% of the total), liver (0.8 million, 9.1%), and stomach (0.7 million, 8.8%) were the most common cause of mortality globally in 2011 (IARC, 2013).

Breast, colon or rectum, and lung cancer are most common in women whilst prostate, lung, and colon or rectum cancer are the most common cancers amongst men (Shibuya *et al.*, 2002).

In 2012, 1.7 million women were diagnosed with breast cancer, and 6.3 million women who had been diagnosed with breast cancer in the previous five years were still alive (IARC 2013). The incidence of breast cancer has increased by more than 20%, while mortality has increased by 14% since 2008 (IARC, 2013). Not only is breast cancer the most common cause of cancer death among women (522 000 deaths in 2012), it is also the most frequently diagnosed cancer among women in 140 of 184 countries worldwide, representing one in four of all cancers in women (IARC, 2013).

It is estimated that one in nine females will develop cancer of the breast in their lifetime; 80% will be post-menopausal, mostly aged between 60 and 64 (Ogden, 2004). In the United Kingdom (UK), breast cancer is the most common cancer, with approximately 49,560 women and 400 men being diagnosed with the disease annually (Cancer Research UK, 2012). Women in the UK have a one in 8 lifetime risk of developing breast cancer, with most women who develop breast cancer having been through menopause, and about 20% being under 50 years of age (Cancer Research UK, 2012).

Although prevalence of breast cancer is lower in South Africa than Europe (41 per 100 000 in South Africa versus 89.1 per 100 000 in the UK), the overall African mortality rates are among the highest in the world – a direct result of a lack of services, transport difficulties and the resultant late presentation. More people succumb to all cancers daily worldwide than the sum of HIV & AIDS, TB and malaria (Bateman, 2012).

The National Cancer Registry (2004) reported that breast cancer is the most common cancer within the South African female population. However, there are significant population differences, with cancer of the breast being the commonest cancer in Asian

(33.99%) and Coloured women (25.01%), but ranking second to cervical and basal cell carcinomas (BCC) in black (17.31%) and white women (19.56%), respectively (Mqoqi *et al.*, 2004).

By 2050, the incidence rates of breast cancer worldwide are conditionally predicted to reach six times the current levels (Bateman, 2012).

Although the incidence rates of breast cancer has increased, the improved screening and treatment, has provided women with a better chance of survival. In the United Kingdom, more than 90% of women with stage 1 breast cancer survived 5 years or more, and more than 85% survived over 10 years. More than 70% of women with stage 2 breast cancer survived more than 5 years, whilst more than 60% survived more than 10 years. There was a 50% 5-year survival rate and a 40% 10-year survival rate in women with stage 3 tumours (Cancer Research UK, 2012).

According to the American Cancer Society's 2011 report, the five-year relative survival rate for women with breast cancer is approximately 90%. Regarding breast cancer survival in Africa, the Zimbabwe Cancer registry indicated that survival of black (African) women diagnosed with breast cancer was 32,6% after five years, compared with 58,2% among white women in the same city (Harare) (Williams *et al.*, 2006). Similarly, Ghafoor *et al.* (2002) and Jemal *et al.* (2003) have shown that five-year survival rates amongst African-American females with breast cancer is lower (73 percent), as compared with white women (88 percent), at all stages of diagnosis.

In South Africa, delayed screening of women by overloaded and often understaffed public sector public health clinics does not increase the cancer detection rate of 1 in 26 women daily, nor improve on the current ratio of 1 in 7 dying of the disease (Bateman, 2012).

1.3 Statement of the problem

Genetic counselling is the process by which patients or relatives at risk of an inherited disorder are advised of the consequences and nature of the disorder, the probability of developing or transmitting it, and the options open to them in management and planning of families (Wikipedia, 2014). Genetic counselling will therefore provide both the patient and

the family with the necessary information and advice about their breast cancer. Genetic counselling will be suited to the individual needs of the patient and her family, thereby providing them with relevant information to make informed choices.

Considerable research exists on the steps in diagnosing breast cancer; that is, from the point of screening through to treatment of breast cancer. However, not much literature is available on genetic counselling and risk assessments for female family members of breast cancer patients.

1.4 Motivation for the study

Canadian, UK and USA-based research is available on genetic counselling and risk assessments for female family members of breast cancer patients but there is little reference to South African-based studies. It was therefore important to conduct a study on the need for genetic counselling and risk assessments for high risk women in South Africa, including women with a family history of the disease.

1.5 Purpose of the study

The purpose of the study was to determine the level of genetic counselling and acceptability of genetic counselling by breast cancer survivors, at the time of diagnosis, and the female members of their families, after the diagnosis of the patient in their family, with a view to making recommendations based on the findings.

1.6 Research objectives

In order to achieve the purpose, the objectives of the study were to:

- Determine the participants' socio-demographic profile.
- Ascertain the awareness and factors that affected the uptake of genetic counselling by breast cancer patients, their female family members and women in the general public in the Msunduzi Municipality in 2003.
- Assess the willingness of female children of a breast cancer patient to undergo genetic counselling or a risk assessment in the Msunduzi Municipality in 2003.

1.7 Significance of the study

The findings of this study should provide an analysis of acceptability and uptake of genetic counselling and stimulate further research in this field.

1.8 Assumptions underlying the study

Assumptions are basic principles that are assumed to be true based on logic and reason, without proof or verification (Brink *et al.*, 2006). Assumptions influence the logic of the study which leads to more rigorous study development. The assumptions underlying the study were as follows:

- Genetic counselling and risk assessments are offered for female offspring of breast cancer patients.
- If not, female offspring would be interested in consulting with a genetic counsellor and having a risk assessment conducted, to determine their risk of developing the disease.

1.9 Structure of the dissertation

The dissertation consists of six chapters. Chapter 1 introduces the study, including the epidemiology of cancer; statement of the problem; motivation for the study; purpose, objectives and significance of the study, and assumptions underlying the study.

Chapter 2 covers the literature review on gene mutations, genes that contribute to the development of breast cancer, biomarkers for breast cancer, risk factors associated with breast cancer, genetic counselling and testing, knowledge of and attitudes towards breast cancer screening and genetic testing, and the Health Belief Model of Health Promotion as a conceptual framework.

Chapter 3 describes the research design and methodology.

Chapter 4 presents the data analysis and interpretation, with results presented in tables and graphs.

Chapter 5 discusses the results of the study.

Chapter 6 provides a summary of the study and makes recommendations.

1.10 Summary

This chapter discussed the research problem, purpose, objectives and significance of the study; and the underlying assumptions of the study.

CHAPTER II: LITERATURE REVIEW

2.1 Introduction

This chapter covers the literature review conducted for the study. The literature review focused on breast cancer, awareness and factors that affect the uptake of genetic counselling and genetic counselling for family members of breast cancer patients. This included the risk factors associated with breast cancer; gene mutations; genes that contribute to the development of breast cancer; biomarkers for breast cancer; genetic counselling and testing; knowledge of and attitudes towards breast cancer screening and genetic testing; and the Health Belief Model of Health Promotion as a conceptual framework.

2.2 Purpose of the literature review

Significant progress has been made in diagnosing and treating breast cancer patients. Genetic counselling and risk assessments for female family members of breast cancer patients, as well as raising awareness and educating them on risk factors and primary prevention, do not play a prominent role in the South African health care system. The focus is only on the patient without consideration being given to women in her family who may be at risk of developing the disease. Accordingly, the researcher attempted to present the importance of genetic counselling and risk assessment as a primary means of prevention to females with a higher risk of developing cancer of the breast.

2.3 Scope of the literature review

The literature review focused on studies that examined the risk factors for breast cancer and the link to genetic markers; genes involved in breast cancer; diagnosis of breast cancer; treatment; uptake of genetic counselling and risk assessment, and patient knowledge. This included genes mutation; genes that contribute to the development of breast cancer; biomarkers for breast cancer; risk factors associated with breast cancer; genetic counselling and testing; knowledge of and attitudes towards breast cancer screening and genetic testing, and the health belief model of health promotion as a conceptual framework.

The review excluded studies on co-morbidity with breast cancer, such as HIV and AIDS and non-communicable diseases.

2.4 Sources of literature reviewed

The electronic databases *PubMed; Google Scholar; Google; Susan G. Komen; American Cancer Society* and the *National Health Laboratory Services* were used to search for information and studies on breast cancer. The researcher used the keywords and phrases: breast cancer; breast cancer risk factors; Breast Cancer gene 1(*BRCA1*); Breast Cancer gene 2 (*BRCA2*); *P53* tumour suppressor gene; *Bak* and *Bax* gene mutations; Human Epidermal growth factor receptor 1 (HER 1) and receptor 2 (HER 2) gene polymorphisms; biomarkers for breast cancer; knowledge of and attitudes toward breast cancer screening; genetic counselling in breast cancer; uptake of genetic counselling; barriers to genetic counselling, and breast cancer risk assessment.

2.5 Risk factors linked to breast cancer development

Cancer risk is inherited in a quantitative manner; consequently, numerous genetic and environmental factors selectively lead to the expression of the disease (Alberg & Helzlsouer, 1997; Smith *et al.*, 2001; Byers *et al.*, 2002). Established breast cancer risk factors include not only inherited susceptibility but also endogenous hormone levels, early age at first menstruation, menopause at a late age, giving birth to a first child at an older age, hormone replacement therapy, a high fat diet, alcohol consumption, smoking, stress and physical activity (Allan *et al.*, 1992, Alberg & Helzlsouer, 1997; Feigelson & Henderson, 2000; Sampson & Fenlon, 2002; Ogden, 2004).

The incidence of breast cancer in Brazil and trends amongst younger women has increased since 1980 (Carraro *et al.*, 2013). In the 25 to 29 year age range, there was an increase from 6.4 to 7.8 per 100,000 females; in the 30 to 34 year age range, incidence increased from 19 to 27.6 per 100,000 women (Ortega *et al.*, 2010). Risk factors for early development of breast cancer are still not well understood; however, a family history of cancer exists in 10 to 37% of all cases (Carraro *et al.*, 2013).

Age continues to be the most significant risk factor in developing breast cancer (Wang *et al.*, 2011). Estimates show that 20% of Americans will be older than the age of 65 by 2030. Approximately 50% of breast cancer patients are older than 65 years and approximately 35% are older than 70 years (Holmes & Muss, 2003; Society, 2003). However, most of the variation in breast cancer risk across populations and among individuals is due to environmental factors that are not inherited (Byers *et al.*, 2002).

a. Breast density

Mammographic breast density is probably the least valued and used risk factor when investigating cancer of the breast (Liotta *et al.*, 2001; Chung *et al.*, 2002; van de Vijver *et al.*, 2002). Breast density is inheritable, although the role of breast density to a greater risk is not dependent on the risk associated with *BRCA1* and *BRCA2* mutations (Bremer *et al.*, 2002). Bremer *et al.* (2002) reported that the average relative risk of breast cancer in women with greater breast density as compared with women with a lower breast density is 4. Genetic factors that determine breast cancer probably also play a role in breast cancer (van de Vijver *et al.*, 2002).

b. Genetic risk factors

Breast cancer can be inherited when there are cellular changes which make a female susceptible to developing the disease and these germ-line mutations account for the vast incidence of breast cancer, especially those women who develop the disease in both breasts at a young age (Joy *et al.*, 2005). Genetic inheritance and mutations within the *BRCA1* and *BRCA2* genes influences the risk of women contracting breast cancer (Smith *et al.*, 2001; Byers *et al.*, 2002). However, less than 10% of all breast cancer cases are considered to be a result of inherited mutations, *BRCA1* and *BRCA2*, which individually increase risk by a significant amount (Massoud & Gambhir, 2003).

Within the South African context approximately 5 – 10% of breast cancer cases are directly due to an inherited susceptibility (Schlebusch *et al.*, 2010). In South Africa, *BRCA1* and *BRCA2* disease-causing mutations are responsible for 19 and 47% of familial breast cancer respectively (Sluiter *et al.*, 2009).

Mutations that are common in the South African population are, the 3 mutations which make up >90% of *BRCA* mutations in Afrikaners – *BRCA1 1493delC*, *BRCA1 2760G>T (p.Glu881X)* and *BRCA2 8162delG*; the 3 mutations known to be common in those of Ashkenazi Jewish ancestry – *BRCA1 185delAG*, *BRCA1 1493insC* and *BRCA2 6174delT*; and a recently described mutation that has been found to be more common in people of Xhosa and mixed ancestry in the Western Cape (*BRCA2 5999del4*) was added to the panel of 'common mutations' in 2009 (Schoeman *et al.*, 2013).

c. Environmental risk factors

The majority of breast cancers result from an increase of cellular changes which take place over time; hence age is a significant factor because the longer a person lives, the more time there is for mutations to accumulate (Joy *et al.*, 2005). These types of changes are due to exposure to carcinogens from the external environment, or from extreme exposure to substances which promote breast cancer within the body, such as circulating hormones, or may even be due to random mutations that occur during cell division (Joy *et al.*, 2005).

d. Early onset of menstruation

Oestrogen is known to accelerate the development of half of all breast cancers, and the longer breast tissue is exposed to the natural oestrogens circulating in a woman's body, the greater her risk of breast cancer will be as she ages (Baron-Faust, 1995; Khan *et al.*, 1998; Haiman *et al.*, 2002; Ogden, 2004). Therefore, beginning menstruation before the age of twelve, increases a woman's risk by approximately one and a half times that of women who begin menstruating after the age of fifteen (Baron-Faust, 1995; Ogden, 2004).

e. Age at first pregnancy

Delaying childbirth until the late thirties or early forties may double or treble some women's risk for the development of breast cancer (Baron-Faust, 1995; Sampson & Fenlon, 2002) compared to women who give birth to their first child before the age of twenty (Baron-Faust, 1995; Buckman, 1997; Ebrahimi *et al.*, 2002). Scientists believe that the protective effect gained from early pregnancy may be due to the changes in breast cells during

pregnancy and lactation, where the lobules produce milk that is transported from the ducts to the nipple openings during breastfeeding (Ogden, 2004). Cells that produce milk are fully matured and are not as sensitive to DNA damage, resulting in a declined susceptibility to mutations in breast cells of females who gave birth at an early age (Clark *et al.*, 2005).

f. Late onset of menopause

The amount of exposure to natural oestrogen also affects women who are at the end of their reproductive cycle (Baron-Faust, 1995). Women who enter menopause after fifty-five years of age (Sampson & Fenlon, 2002) are at twice the risk of developing breast cancer compared with women whose menopause occurs before age forty-five (Baron-Faust, 1995). Furthermore, women with surgically removed ovaries prior to the natural age of menopause are at an even lower risk than women who enter an early natural menopause (Baron-Faust, 1995).

g. Use of hormone replacement therapy (HRT)

The use of hormone replacement therapy (HRT) by postmenopausal women is linked to a higher risk of developing breast cancer (Lawson *et al.*, 2001). HRT is usually given to older women when the breasts are less vulnerable to risk factors due to the slower rate of cell growth (Sampson & Fenlon, 2002). According to Sampson and Fenlon (2002), a woman's chances of developing breast cancer after five years of HRT is between 45 and 47 per 1000, and further rises to 51 per 1000 after ten years of using HRT.

h. Body mass index

In addition to the above risk factors, one that has received much attention is the relationship between body size (weight and height) and the risk of developing breast cancer (van den Brandt *et al.*, 2000). Body mass index, which is calculated as weight (kg)/height^2 (m^2), shows a positive link with postmenopausal breast cancer risk and an inverse association with developing premenopausal breast cancer (Hunter & Willett, 1993, Swanson *et al.*, 1996, Trentham-Dietz *et al.*, 1997). A positive link between breast cancer risk and adult height was seen mostly for postmenopausal women (Hunter & Willett, 1993; van den Brandt *et al.,* 2000).

Obesity in premenopausal women is thought to protect women from developing breast cancer due to frequent anovulatory menstrual cycles, resulting in lower levels of estradiol and progesterone and decreased luteal phase progesterone levels in ovulatory cycles (Stoll, 1994). However, it is not clear as to the level of obesity that is needed to produce adequate anovulatory cycles to decrease the risk of developing breast cancer (Ursin *et al.*, 1994).

In postmenopausal women, it has been proposed that the distribution of fat is more predictive of breast cancer risk than body mass (Stoll, 1994). Considerable data indicates that obesity is linked to a higher risk of breast cancer in postmenopausal women who have not used HRT (National Cancer Institute, 2014).This may be because in postmenopausal women, the production of ovarian oestrogen is lessened, and oestrogen, which may assist the growth of tumours, comes about largely from the aromatization of androstenedione that occurs primarily in adipose tissue (van den Brandt *et al.*, 2000).

Obesity is constantly recognized as linked to cancer, where obesity contributes to a higher risk of cancer incidence and increases mortality as shown in three large population-based cohort studies (Calle *et al.*, 2003; Reeves *et al.*, 2007; Song *et al.*, 2008).

2.6 Gene mutation

Germline mutations are considered important contributors to disease-causing issues in the younger breast cancer patients globally (Carraro *et al.*, 2013).When cancer develops it is due to mutations in the body's cells. Young females have breast tissue that is sensitive to DNA damage due to cancer-related causes (Clark *et al.*, 2005). According to the National Cancer Institute (NCI), inheriting a mutation in either *BRCA1* or 2 not only increases a woman's chance of developing breast cancer, but these women also have an increased risk for the development of ovarian and other cancers (Zielinski, 2008).

2.7 Genes that contribute to the development of breast cancer

Genes that control normal cell division are called proto-oncogenes and are active where and when high rates of cell division are required (Lewis, 1994). Proto-oncogenes may be activated into oncogenes, cancer-causing genes, by point mutations, gene amplification or

gene translocation (Lewis, 1994) so that far more protein encoded by that gene is present, therefore enhancing its role (Osborne *et al.*, 2004).

Oncogene refers to those genes in which alterations or mutations cause a gain-of-function effect that contributes to the development of a malignant phenotype (Osborne *et al.*, 2004). However, full development of a cancerous state usually requires additional mutations, typically deletions or point mutations (Lewis, 1994), which affect genes normally involved in the restraint of cell growth (Snustad & Simmons, 2003). These mutations define a second class of cancer-related genes, the anti-oncogenes or recessive oncogenes (Lewis, 1994), better known as tumour suppressor genes (Snustad & Simmons, 2003). These genes lead to an inability to function which contributes to malignant phenotypes (Osborne *et al.*, 2004).

The tendency to develop certain types of cancer is known to run in families and to develop due to a combination of mutational events (Weaver, 1992). Abnormal genes are believed to account for 5 to 10% of all cancers of the breast (Ogden, 2004). There has been considerable progress in identifying these genes responsible for breast cancer (Cornelisse *et al.*, 1996). Two genes, *BRCA1* and *BRCA2*, are the most important tumour suppressor genes related to a higher risk of breast and ovarian cancers in families (Ogden, 2004).

BRCA1 and *BRCA2* are usually involved in regulating cell growth, but in some families the gene has been altered so that it no longer functions correctly (Sampson & Fenlon, 2002). Carraro *et al.* (2013) found that in 10 to 40% of cases an early onset of familial breast cancer was associated with mutations in *BRCA1* and *BRCA2*.

BRCA1 and *BRCA2* are the two most common genes associated with breast cancer and should women inherit a mutation in one of these genes their chances of developing cancer of the breast in their lifetime would be 36 to 85% (Zielinski, 2008). *BRCA1* and *BRCA2* are part of a group of genes known as tumour suppressor genes which assist in controlling cell division; *BRCA1* and *BRCA2* repair damaged DNA (Zielinski, 2008).

Regarding non-familial breast cancer, Akbari *et al.*, (2013) found that approximately 27% of unselected breast cancer cases in the Bahamian population can be attributed to a

BRCA1 or *BRCA2* mutation, a prevalence which far exceeds that of any other country. Other studies reported the frequency of *BRCA1* or *BRCA2* mutation among patients with an early onset of breast cancer, as ranging from 1 to 10% (Lallo *et al.*, 2006; Fackenthal & Olopade, 2007; Haffty *et al.*, 2009).

Only approximately 5% of breast cancers, and particularly those diagnosed in young women, are as a result of mutations in the *BRCA1* and *BRCA2* genes (Baron-Faust, 1995; Lakhani, 1999; Di Prospero *et al.*, 2001). Patients with mutations in the *BRCA1* and *BRCA2* genes have a 10 to 25% greater risk of developing breast cancer during their lifetime (Snustad & Simmons, 2003). However, it is not certain that a carrier of these mutations will develop the disease (Sampson & Fenlon, 2002; Ogden, 2004).

Human cancer cells with mutations in the *BRCA1* or *BRCA2* genes are hypersensitive to radiation and show chromosomal abnormalities (Zheng *et al.*, 2000). This reveals a potential role for both the *BRCA1* and *BRCA2* gene, in maintaining genetic stability by responding to DNA damage (Zheng *et al.*, 2000). Aneuploidy, chromosomal breaks and aberrant mitotic exchanges are all observed chromosomal abnormalities (Lee *et al.*, 1999).

a. *BRCA1* gene

Mutations in the *BRCA1* gene are associated with a large percentage of hereditary cases of breast cancer and are found in up to 40 to 50% of families with hereditary breast cancer (Zheng *et al.*, 2000). *BRCA1* is inherited as an autosomal dominant gene, meaning that a woman has a fifty-fifty chance of inheriting a flawed copy of the gene from a heterozygous parent and of developing breast cancer before age fifty (Baron-Faust, 1995). These women also have an 85% lifetime risk of developing the disease (Di Prospero *et al.*, 2001).

Tumours of breast cancer sufferers who carry the *BRCA1* mutant allele typically show continuous pushing margins of the tumour and lymphocytic infiltrate (Lakhani, 1999; Williams *et al.*, 2006). These features are part of the subset of characteristics that define medullary carcinoma (Lakhani, 1999). It is noteworthy that carriers of a mutated gene in familial breast cancer families cannot be phenotypically distinguished from non-carriers, as two mutational events are required to eliminate both functional copies of the gene within the cell (Yawitch, 2001). Due to this, as well as a high occurrence of sporadic breast

cancer in the general population, it has been difficult to conduct linkage studies to identify the gene responsible for the development of breast cancer in different families. A study of 23 extended families, including 146 cases of breast cancer, to map *BRCA1* by genetic linkage to the interval designated 17q 12-21 on the short arm of chromosome 17 (Hall *et al.*, 1990). However, there seems to be substantial variability in the ages at which breast cancer has been diagnosed in *BRCA1* mutation carriers (Rebbeck *et al.*, 1999). This could imply that germline mutations in *BRCA1* could also be a cause of the Mendelian pattern of breast cancer inheritance in certain families (Rebbeck *et al.*, 1999).

Steroid hormone pathways regulate *BRCA1* expression (Marks *et al.*, 1997). Rebbeck *et al.* (1999) therefore hypothesized that allelic variation in genes controlling hormonal signalling known to be involved in normal breast tissue development may be involved in modification of *BRCA1*-associated cancer risk. Such is AR, the androgen-receptor gene, which works as a ligand-dependent transcriptional activator in response to androgens (Rebbeck *et al.*, 1999). AR contains a highly polymorphic CAG trinucleotide repeat – AR-CAG, whose length is conversely linked with the amount of transcriptional activation exhibited by the AR (Chamberlain *et al.*, 1994). In order to effectively use strategies related to the prediction of risk or the prevention of cancer in *BRCA1* carriers, detailed knowledge of risk-modifying factors, as well as *BRCA1* mutation status, is required (Rebbeck *et al.*, 1999).

b. *BRCA2* gene

Tumours associated with mutations in the *BRCA2* gene are phenotypically similar to sporadic breast tumours, as they are usually of an intermediate grade, are frequently hormone receptor positive and the onset of breast cancer occurs at a later age than with females with a mutated *BRCA1* gene (Williams et al, 2006).

The *BRCA2* gene has been shown to be located on the short (q) arm of chromosome 13 (Baron-Faust, 1995; Osborne *et al.*, 2004). Although *BRCA1* and *BRCA2* are similar in respect of features, they have different structure (Osborne *et al.*, 2004). Over one hundred distinctive mutations of *BRCA2* have been depicted, with the majority causing premature truncation of the protein, as in the case of *BRCA1* (Osborne *et al.*, 2004). *BRCA1* encodes a phosphoprotein of 1863 amino acids (Miki *et al.*, 1994) characterised by two structural

motifs at each of its flanking termini (Zheng *et al.*, 2000). *BRCA*-associated ring domain or *BARD1* is a protein which interacts with *BRCA1*, and following a genotoxic insult, it has been shown that *BARD1*, together with *BRCA1* protein and Rad51, localise to areas of damaged DNA, and contributes to regulating transcription and repairing double-stranded DNA (Scully, 1997).

It is thought that the incidence of *BRCA1* and *BRCA2* heterozygotes is similar within the general population (Osborne *et al.*, 2004). In the Ashkenazi Jewish population, the mutation *6174delT* takes place at a rate of 1.5%; whilst the Icelandic population has the mutation *999del5*, which occurs at a rate of 0.5% (Osborne *et al.*, 2004). Mutations in the *BRCA2* gene are thought to account for about 35 percent of reported breast cancer cases in families (Wooster *et al.*, 1994).

c. *P53* Tumour suppressor gene

Germline mutations in approximately 10 genes that are linked to the repair of DNA have been shown to be linked with an inherited risk for cancer of the breast (Walsh *et al.*, 2006). The tumour suppressor gene, *TP53* plays an important part in the way cells respond to damaged DNA generating pathways involved in cell death, cell cycle arrest and the DNA repair mechanism to continue the cell's genomic integrity (Brosh & Rotter, 2009).

Germline mutations in *TP53* are rare and are linked to a higher risk of breast cancer in females (Lee *et al.*, 2012). *p53* tumour suppressor gene mutations are found on the long (p) arm of chromosome 17 (Osborne *et al.*, 2004), and are estimated to occur in approximately 20 to 30% of breast cancers (Hollstein *et al.*, 1991). Research on the prevalence of *TP53* germline mutations in families chosen by genetics clinics shows that many families with *TP53* mutations fulfil the criteria for either Li-Fraumeni syndrome (LFS), inherited in a dominant fashion (Shannon & Smith, 2003; Goldhirsch & Gelber, 2004). Li-Fraumeni-like or Chompret criteria are present when there is a family history of breast, bone or soft tissue sarcoma, brain tumours and adrenocortical carcinomas, which are LFS-linked cancers (Birch *et al.*, 2001; Walsh *et al.*, 2006; Gonzalez *et al.*, 2009; Ruijs *et al.*, 2010; Osborne *et al.*, 2004; Kruger & Apffelstaedt, 2007). Recent population-based studies discovered that women who had early-onset breast cancer before the age of 40, had *TP53* germline mutations (Mouchawar *et al.*, 2010; Lalloo *et al.*, 2006).

Carraro *et al.* (2013) found that the occurrence of germline *TP53* mutations, related to the early development of breast cancer, is linked to Li-Fraumeni Syndrome or Li-Fraumeni-like syndromes. These patients have an approximately 50% chance of developing cancer by the time they reach their forties and a 90% chance by their eightieth year (Malkin *et al.*, 1990; Li, 1990). A study conducted by Lee *et al.* (2012), found that in Asian areas, which are low-resourced and poor family history is reported, germline *TP53* mutations have mostly been found in women with breast cancer who have a family history of LFS-linked cancers.

Mutational inactivation of the *p53* gene that controls the G1/S cell cycle checkpoint may avoid ceasing growth, which is what normally happens to damaged DNA, as well as prevent *p53*-mediated cell death (Zheng *et al.*, 2000). This enables these cells to survive but with serious chromosomal damage (Zheng *et al.*, 2000) and may be the reason for the development of cancer in the breast in females with mutations in the *p53* gene.

2.8 Biomarkers for breast cancer

Biomarkers are being sought across a broad range of events in the development of cancer of the breast (table 2.1) with the clinical use of biomarkers of breast cancer being limited to prognosis, prediction of response to therapy and monitoring of diagnosed patients (Joy *et al.*, 2005).

Table 2-1: Biomarkers of events in the development of breast cancer: their potential uses and limitations (adapted from Joy et al., 2005)

Event	Potential use for biomarkers	Progress to date	Key limitations
Germ-line mutations	Risk indicator	Numerous mutations identified; genetic testing available for *BRCA1* and *BRCA2*	Account for only 10 % of breast cancers
Genetic polymorphisms	Risk indicator	Some candidate polymorphisms identified; thousands of SNPs have been mapped	Validation difficult due to genetic diversity among different ethnic populations and the need to measure cumulative effects of multiple SNPs
Somatic genetic alterations	Risk indicator; screening; diagnosis; prognosis	Loss of heterozygosity at several loci associated with premalignant disease, as well as early and late-stage breast cancer	Unknown which, if any, loss of heterozygosity events are specific to invasive or metastatic cancer
Epigenetic changes in breast cells	Risk indicator; screening; diagnosis; prognosis; therapeutic target	Research correlating methylation patterns at key loci with breast cancer presence and stage	Validation will require large-scale longitudinal studies and comprehensive cancer registry data
Altered gene expression in breast cells	Screening; diagnosis; prognosis; choosing therapy; monitoring outcome	Studies under way on several over-expressed and under-expressed genes in breast tumour tissue; estrogen receptor status predicts response to antiestrogen therapy	Validation will require large-scale longitudinal studies and comprehensive cancer registry data
Changes in protein signalling pathways in breast cells	Screening; diagnosis; prognosis; choosing therapy; monitoring outcome	Clinical trials underway in breast cancer patients before, during and after therapy	Population heterogeneity reduces sensitivity and complicates standardization; sampling involves

			microdissection
Changes in individual serum markers	Screening; diagnosis; prognosis; monitoring outcome	Preliminary findings indicate prognostic benefits of monitoring a mucin, CA 15-3, which has received FDA approval for the detection of recurrent breast cancer	Typically lack sensitivity for early malignancy and organ specificity; not elevated in all patients
Angiogenesis	Risk indicator; prognosis; choosing therapy	Research on several angiogenesis-related receptors being conducted to develop a possible treatment	Validation will require large-scale longitudinal studies. Main focus is currently on developing therapeutics
Invasion and metastasis	Prognosis	Candidate proteases and inhibitors have been identified; prognostic benefit of urokinase plasminogen activator for node-negative breast cancer confirmed in large prospective randomized trial	Lack of effective therapy for metastatic breast cancer

Most biomarkers are synthesized by normal as well as malignant tissues and are only rarely increased in premalignant or early stage disease (Joy *et al.*, 2005). Biomarker-based screening may prove to be a practical means for screening women who have a higher risk of developing breast cancer as such biomarker-based assays could detect groups of proliferating cells at a preclinical stage, as well as groups of cells that may never require treatment (Joy *et al.*, 2005).

2.9 Gene mutations in breast cancer

2.9.1 *Bak* and *Bax* gene mutations

Bak and *Bak* genes are the entry point to the mitochondrial passage of cell death or apoptosis (Kholoussi *et al.*, 2014). Cells twice as deficient in the two multi-domain proapoptotic *Bax* and *Bak* do not give off cytochrome c, thereby being resistant to all apoptotic stimuli that initiate the inherent pathway (Lindsten *et al.*, 2000; Wei *et al.*, 2001).

Gene transfer mediated increases in *Bak* protein levels boosts apoptosis induced by growth factor deprivation in breast cancer cells (Kholoussi *et al.*, 2014). *Bak*'s primary function is that of a promoter of apoptosis (Pataer *et al.*, 2000). Mutations occurring in the coding and promoter area of the *Bax* gene have been found to affect protein expression and the function of many cancers (Moshynska *et al.*, 2003). Koda *et al.* (2004) showed that changes in the expression of *Bak* coincide with the development and progression of breast cancer. Eguchi *et al.* (2000) found that *Bak* expression indicated a considerable increase at tumour stages T3 and T4. Kholoussi *et al.* (2014) achieved the same outcome where *Bak* mRNA levels showed considerable increases in the T3 and T4 tumour stages.

Kholoussi *et al.* (2014) also found that *Bak* expression does seem to lead to developing breast cancer and affects the progression of the disease. Furthermore, Kholoussi *et al.* (2014), *Bax* d and *Bax* δ could be used as a biomarker and risk factor for breast cancer. In addition, the G284A single nucleotide polymorphism (SNP) of *Bax* promoter has a considerable association in the breast cancer subject, which could suggest this SNP acts as a risk factor (Kholoussi *et al.*, 2014).

2.9.2 *HER1* and *HER2* polymorphisms

Mutation of genes and anomalies of signalling pathway of *HER1* and *HER2* have often been associated in carcinogenesis of many solid tumours (AbdRaboh *et al.*, 2013). Identification of the molecular characteristics of cancer of the breast allows accurate prediction of the path of the disease and its response to chemotherapy (Normanno *et al.*, 2005). The over-expression of *HER1* and *HER2* correlate with the poor prognosis in breast cancer (Uzan *et al.*, 2009). This over-expression was associated with gene amplification of

HER2 in approximately 10 to 25% of invasive breast cancer and an adverse consequence on the course of the disease (Menard *et al.*, 2001).

SNPs are the most common source of human genetic variation that contributes to susceptibility to malignant alterations (www.ncbi.nlm.nih.gov, 2011). According to Wang *et al.* (2007), an SNP at codon 497 of *HER1* gene causes the Arg (R) and Lys (K) substitution in the extracellular domain known as *HER1 R497K* has been identified in cancer of the breast. El-Mougy *et al.* (2012) indicated that an SNP at codon 655 of *HER2* shows an isoleucine-to-valine substitution in the trans-membrane domain which has been associated with the risk of breast cancer. AbdRaboh *et al.* (2013) found that *HER1497K* and *HER2655V* polymorphisms may be considered susceptibility genetic markers for the risk of breast cancer, but are not suitable to measure disease aggressiveness.

Polymorphisms probably affect the risk of developing breast cancer by a small percentage, but their impact on breast cancer risk may be significantly higher than that of the relatively rare *BRCA* mutations (Fabian *et al.*, 1996), and the combined effect of several polymorphisms on the risk of breast cancer could be substantial (Joy *et al.*, 2005).

Further research on genetic polymorphisms that influence breast cancer susceptibility is still required prior to it being useful in classifying women into risk groups for breast cancer (Fabian *et al.*, 1996).

2.10 Genetic counselling and testing

Genetic education and counselling includes identifying the most informative person in the family to test, which may be an affected family member rather than the individual seeking genetic services (National Cancer Institute, 2014). In addition, counselling includes discussing the limitations of the test, all potential test outcomes, and the consequences of identifying a variant of unknown clinical significance (Riley *et al.*, 2012).

The aim of genetic counselling is to educate people about their risk and encourage individuals who have an increased risk of developing the disease, to develop a management plan (Watson *et al.*, 1999). However, it is unclear whether or not genetic counselling helps to allay the fears of these women (Watson *et al.*, 1999). Women at risk

of developing hereditary breast cancer feel more emotionally burdened (Lloyd *et al.*, 1996). Furthermore, it is unclear whether or not women understand the information provided to them or are even able to use it to the benefit of their mental or physical health (Watson *et al.*, 1999).

Doctors are crucial in identifying women with an increased risk of developing cancer of the breast (de Bock *et al.*, 2001). Providing genetic advice about cancer of the breast at the primary health care (PHC) level is doubtful, as women display poor compliance with the advice provided, poor compliance by the doctors themselves with advice given by the clinical geneticist, as well as the fact that no evidence is available which shows that surveillance is effective in females under the age of 50 (de Bock *et al.*, 2001).

Offering genetic testing is recommended when a risk assessment suggests the presence of an inherited cancer syndrome for which specific genes have been identified (National Cancer Institute, 2014).

The American Society of Clinical Oncology (ASCO) (2003) and Robson *et al.*, (2010) propose that genetic testing be offered when the following apply:

- The individual has a personal or family history suggestive of a genetic cancer susceptibility syndrome;
- The results of the test can be interpreted;
- Testing will influence medical management.

However, no such study has been conducted in the South African context. Women are now more accepting and open to being educated on their health. The situation may therefore be quite different in the South African context, and a much higher compliance may be achieved. Moreover, extensive attention is given to breast cancer in South Africa, as the media encourages women to better understand their bodies.

Genetic counselling provides individuals with information that either tells them what their risk is of developing the disease per year or by a certain age (Watson *et al.*, 1999). In order to provide a useful service, it is imperative that people understand and utilize the information provided to them, as lack of understanding would not enable them to make

proper decisions about management of their health, as well as cause more concern over their health (Watson *et al.*, 1999).

Genetic testing for mutations in cancer susceptibility genes in children is particularly complex. While both parents (Bradbury *et al.*, 2010) and providers (O'Neill *et al.*, 2010) may request or recommend testing for minor children, many experts recommend that unless there is evidence that the test result will influence the medical management of the child or adolescent, genetic testing should be deferred until legal adulthood (age 18 years or older) because of concerns about autonomy, potential discrimination and possible psychosocial effects (Wertz *et al.*, 1994; Nelson *et al.*, 2001).

In South Africa, diagnostic *BRCA* testing has been available since 2005, and has been facilitated by the identification of common mutations in the *BRCA1* and 2 genes in both Afrikaner (Reeves *et al.*, 2004; van der Merwe and van Rensburg, 2009) and other local populations (van der Merwe *et al.*, 2012). This has allowed genetic counselling and testing to be offered, but attempts to establish programmes for genetic counselling and testing to stratify patients for possible genetic testing are difficult to implement in the South African environment and have faced many challenges, including, complexity of the risk assessment and testing approach; limited financial and human resources; limited community knowledge of breast cancer or of the possibility of a familial cancer; and difficulty accessing old hospital records of family members (Schoeman *et al.*, 2013).

2.11 Uptake of genetic counselling and testing

Genetic testing entails educating women on their risk, personalized genetic information and providing them with recommendations for continuous risk management, which includes regular screening, chemoprevention and prophylactic surgical interventions (Bouchard *et al.*, 2004). Genetic testing benefits women who have already had breast cancer as well as unaffected women within affected families (Sherman *et al.*, 2014). Women who have already been diagnosed with breast cancer and found to be *BRCA1*/2 carriers may consider prophylactic strategies to decrease their risk of developing a secondary breast cancer (Miller *et al.*, 2006). Genetic risk information to unaffected women may assist in clarifying their risk status, reduce medical uncertainty, and aid with decisions

about risk management (Patenaude, 2005).

Genetic counselling and *BRCA1* and *BRCA2* gene testing are regularly offered in a clinical setting yet no information is available on the proportion of breast cancer patients who have a family history, undergoing genetic counselling (Ayme *et al.*, 2014). Despite the value of genetic testing and genetic counselling for high risk breast cancer survivors, not much information on their uptake is available (Hamann *et al.*, 2013).

Hamann *et al.* (2013) found that 30.8% of the respondent survivors reported having seen a genetic counsellor in the period after their breast cancer diagnosis whilst 33.6% indicated that they had a genetic test done. Ayme *et al.* (2014) found a significant increase in the utilization of genetic counselling over time and further that patients who had a high familial risk had genetic counselling more often than those with an average familial risk.

The most common reasons for first degree relatives of breast cancer patients wanting genetic testing was to learn about their children's risk, to increase the use of cancer screening tests, and to take better care of themselves (Lerman *et al.*, 1995). Lerman *et al.* (1995) found that women with a less formal education were motivated by decisions of childbearing and future planning compared to women who had an education beyond high school.

2.12 Knowledge of and attitudes towards breast cancer screening and genetic testing

In a study of 314 Trinidadian women, 40 years and older, to determine their breast cancer knowledge, attitudes and practices, Gosein *et al.* (2014) found that women who had a higher level of education had a greater knowledge about the benefits of detecting breast cancer at an early stage. In addition, they could further understand that an abnormal mammography result did not necessarily mean they had breast cancer. However, incorrect beliefs, especially that compression of the breast causes cancer, were found among women with the least amount of education (Gosein *et al.*, 2014).

An analysis of knowledge and attitudinal factors amongst African Americans associated with the uptake of genetic testing found positive expectations about the benefits of genetic testing, although participants' knowledge on breast cancer genetics and the availability of genetic testing was relatively low (Halbert *et al.*, 2005).

It is important that culturally sensitive awareness campaigns take place to educate women who are hesitant about breast cancer screening due to perceived pain (Gosein *et al.*, 2014). Charles *et al.* (2006) found that African American women who received culturally modified genetic counselling were more likely to report lesser cancer-related worries compared to women who received standard counselling.

Patenaude *et al.* (2013) conducted a study on daughters of *BRCA1*/2 mutation carriers, aged 18 to 24 years, on their understanding of their 50% chance of being a mutation carrier, their options for risk reduction or management; the extent and nature of their cancer-related distress, and the effect of their mothers' mutation status on their future plans. The results showed that the daughters' genetic knowledge is suboptimal with gaps and misconceptions being common. More than one-third of the daughters reported high levels of distress and indicated that the disclosure of their mothers' genetic information was concern for their future, especially in terms of childbearing (Patenaude *et al.*, 2013). Targeted professional attention to this high-risk group is crucial to inform the daughters of *BRCA* mutation carriers and encourage screening by the age of 25, as this could improve survival, and psycho-education could reduce their distress related to cancer (Patenaude *et al.,* 2013).

Parsa *et al.* (2008) found that the most common reasons women provided for not wanting to go for a breast cancer screening were lack of knowledge, being too busy, feeling embarrassed, fear of a cancer diagnosis, the cost of screening, and thinking it is not necessary. The study used 425 high school teachers; of the participants, 18 had a postgraduate degree; 376 had a degree; 23 had a diploma; and the remaining 8 had other qualifications. Parsa *et al.* (2008) concluded that the participants' knowledge about breast cancer was inadequate; very few performed breast self-examinations, and most did not know how to correctly and routinely perform breast self-examinations. This study yet again

shows the importance of educating women; whether educated or not, many women still require further information to assist them with taking important health decisions.

2.13 Risk assessment

In order to ensure that they provide appropriate information to women about increased breast cancer risks, doctors need to understand what modifiable and non-modifiable factors contribute to these risks (Steiner *et al.*, 2008). This will also provide women with the opportunity to better understand and participate more actively in their health and well being (Steiner *et al.*, 2008). Risk assessment will most probably remain a crucial component of early detection for breast cancer (Joy *et al.*, 2005).

The aim of improving risk assessment is to classify strategies for the detection of breast cancer in order to increase survival and decrease cost and complications in high- and low-risk women, respectively (Flacke *et al.*, 2001).

There are two types of risk assessment: firstly, what a woman's chances are of developing breast cancer over a specified time, which includes her lifetime, and secondly, the chances of having a mutation in one of the high-risk genes such as *BRCA1* or *BRCA2* (Gareth *et al.*, 2007).

Tools have been developed and are available to determine a woman's risk. For example, the Tyrer-Cuzick model, which assesses breast cancer risk over time (Gareth *et al.*, 2007) and the Gail model. Gail *et al.* (1989) used data from the Breast Cancer Detection Demonstration Project to develop a model to determine the absolute risk of breast cancer for women in a given age interval.

The Gail model considers, age of the person, age at menarche, age first live birth, number of previous benign breast biopsy examinations, and number of first-degree relatives with breast cancer, in assessing the risk of breast cancer (Chen *et al.*, 2006; Reddy Challa *et al.*, 2013). The Tyrer-Cuzick model takes into consideration age, body mass index, age at menarche, age at first live birth, age at menopause, hormone replacement therapy use, breast biopsies and family history (first-degree relatives, second-degree relatives, age at onset of breast cancer, bilateral breast cancer and ovarian cancer) in assessing the risk of

breast cancer (Reddy Challa et *et al.*, 2013). In a validation process, Amir *et al.* (2003) found that the Tyrer-Cuzick model performed by far the best at breast cancer risk estimation.

2.14 Barriers to genetic risk assessment

There are specific cognitive and affective factors that influence an African American woman's interest in and her decision to undergo a genetic risk assessment (Sherman *et al.*, 2014. These factors include their awareness of their risk in developing breast cancer, the level to which she approves specific limitations of undergoing genetic testing, her philosophical beliefs and temporal orientation, as well as her cancer-related distress. Furthermore, lack of knowledge and/or negative attitudes regarding genetics and genetics research, as well as concerns about the potential for racial discrimination were found to be barriers to genetic counselling and testing (Simon & Petrucelli, 2009).

In a review focused primarily on systematic factors affecting uptake among minority populations, black, Hispanic, and Asian-Americans, Forman and Hall (2009) highlighted barriers such as time limitations, geographic barriers and cost, access to specialist services, as well as limitations of the current genetic modelling technologies.

Comparisons of African-American and Caucasian women, found considerable differences regarding their knowledge about the genetics of breast cancer (Hughes *et al.*, 1997; Donovan & Tucker, 2000;); perceptions of risk (Donovan & Tucker, 2000); support for the benefits and limitations of undergoing counselling and testing (Hughes *et al.*, 1997; Donovan & Tucker, 2000; Thompson *et al.*, 2003), and ability to cope with emotional distress associated with genetic testing (Donovan & Tucker, 2000). According to Sherman *et al.* (2014), this emphasises the need for targeted interventions to assist with decisions about participating in genetic counselling and testing that should be tailored to the specific cognitive-affective profile of an African-American female.

2.15 Health Belief Model of Health Promotion as a conceptual framework

The Health Belief Model is the most commonly used theory in health promotion and education with the underlying concept being that an individual's personal beliefs or perceptions about a disease determine an individual's health behaviour (www.jblearning.com). Four perceptions are the main constructs of the model (www.jblearning.com):

- Perceived seriousness: This relates to how seriously the individual perceives the disease to be.
- Perceived susceptibility: Individuals' perceived risk or susceptibility to developing a disease and the role this plays in their adopting a healthier lifestyle – the greater they perceive their risk to be, the greater the changes of behaviour to decrease their risk.
- Perceived benefits: How individuals view the usefulness of a new behaviour in decreasing their risk of developing a disease – people adopt new behaviours if they believe the new behaviour will decrease their chances of developing the disease.
- Perceived barriers: Individuals evaluate the barriers in the way of adopting a new behaviour – this is the most significant construct in determining behaviour change.

In addition to the original four constructs, the following were included:

- Modifying variables: how individuals' personal factors affect whether or not they will adopt the new behaviour;
- Cues to action: factors that will initiate a change to behaviour; and
- Self-efficacy: individuals' belief in their ability to do something (www.jblearning.com).

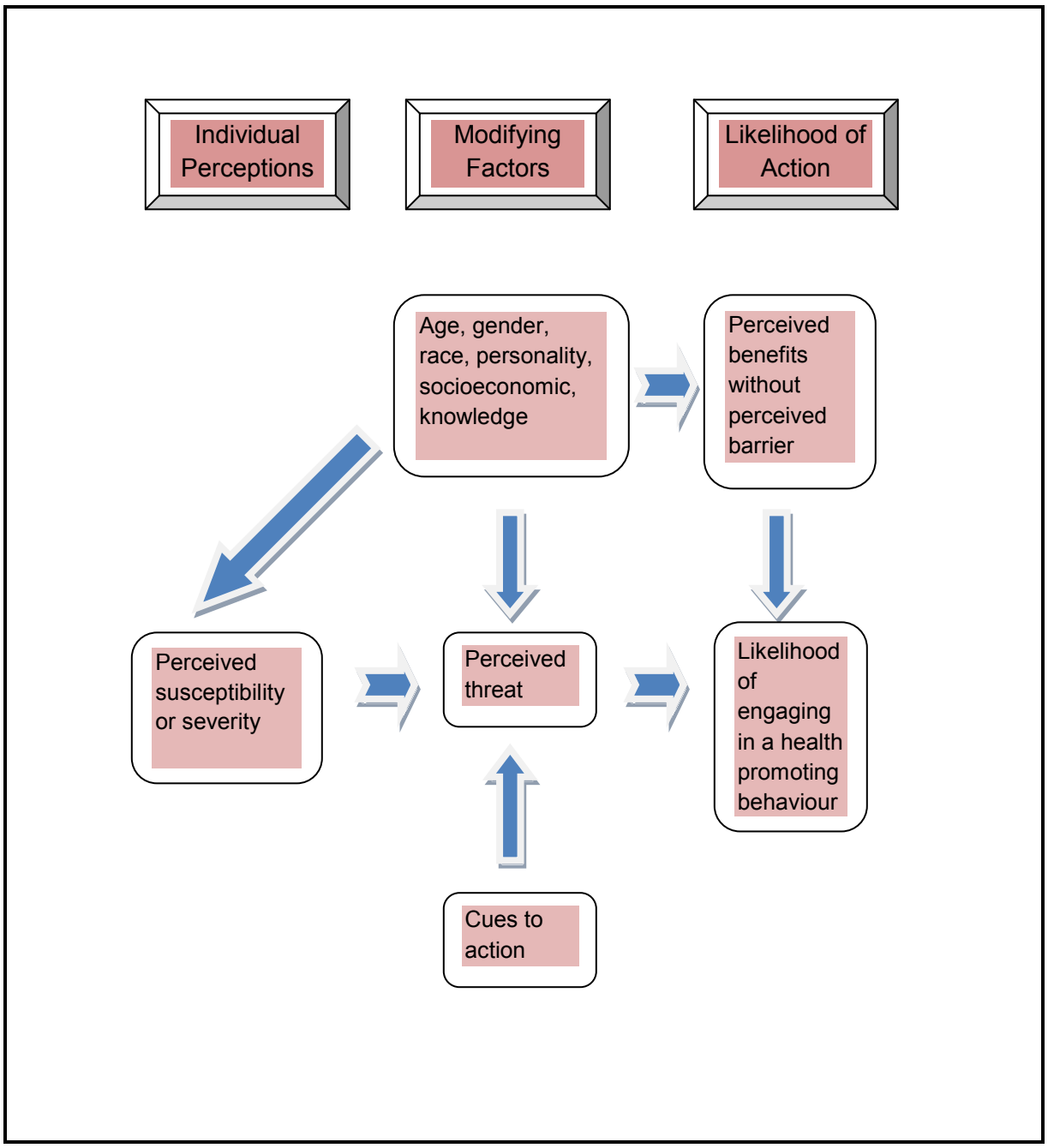

Figure 2.1: Health Belief Model of Health Promotion

Adapted from ***www.jblearning.com***; ***http://en.wikipedia.org***

Parsa *et al.* (2008) found that women with a perceived susceptibility or seriousness of breast cancer are more likely to participate in breast cancer screening, but must also view the benefits to screening greater than the barriers.

It is imperative to understand how South African women perceive screening and assessments for breast cancer. Currently such information is not available; however, should studies be conducted to determine this, it will assist with the development of health education programmes with culturally appropriate information and also provide the basis for the establishment of screening services in rural areas. In order to decrease mortality due to breast cancer, health care professionals need to understand what influences women's behaviours for breast cancer screening and assessment. Health care professionals must further educate and inform women about the need and benefits of screening and assessments in order to achieve the goal of early detection in South Africa.

2.16 Summary

Breast cancer not only affects the individual who has been diagnosed, but is also a life-altering experience for the patient's or survivor's family. It is therefore essential that breast cancer patients and their families be provided with the necessary counselling to help them cope with this stressful situation. Since breast cancer can be hereditary, family members of patients or survivors of breast cancer should be provided with genetic counselling to determine their risk of developing the disease. In addition, more focus must be placed on raising awareness amongst women on the importance of breast self-examination, clinical breast examination, and mammography. Early detection is the key to saving numerous lives, especially since breast cancer is a disease that can be cured if detected and treated early.

CHAPTER III: METHODS

3.1 Introduction

This chapter describes the research design and methodology of the study including the location (setting), study population, sample and sampling, data collection, management and analysis, and validity and reliability.

3.2 Research design

The study was a descriptive cross-sectional study and utilised quantitative and qualitative methods of data collection.

3.3 Study location

The study was conducted in the Msunduzi Local Municipality in KwaZulu-Natal. Msunduzi Municipality, which is commonly known as Pietermaritzburg, is located along the N3 and is considered an industrial as well as an agro-industrial corridor (IDP, 2011- 2016). Pietermaritzburg is the second largest city in KwaZulu-Natal and a contributor to 80% of the GDP by nine of the largest cities in South Africa (IDP, 2011-2016). Pietermaritzburg is the capital city of KwaZulu-Natal and the main economic hub with the Umgungundlovu District (IDP, 2011-2016).

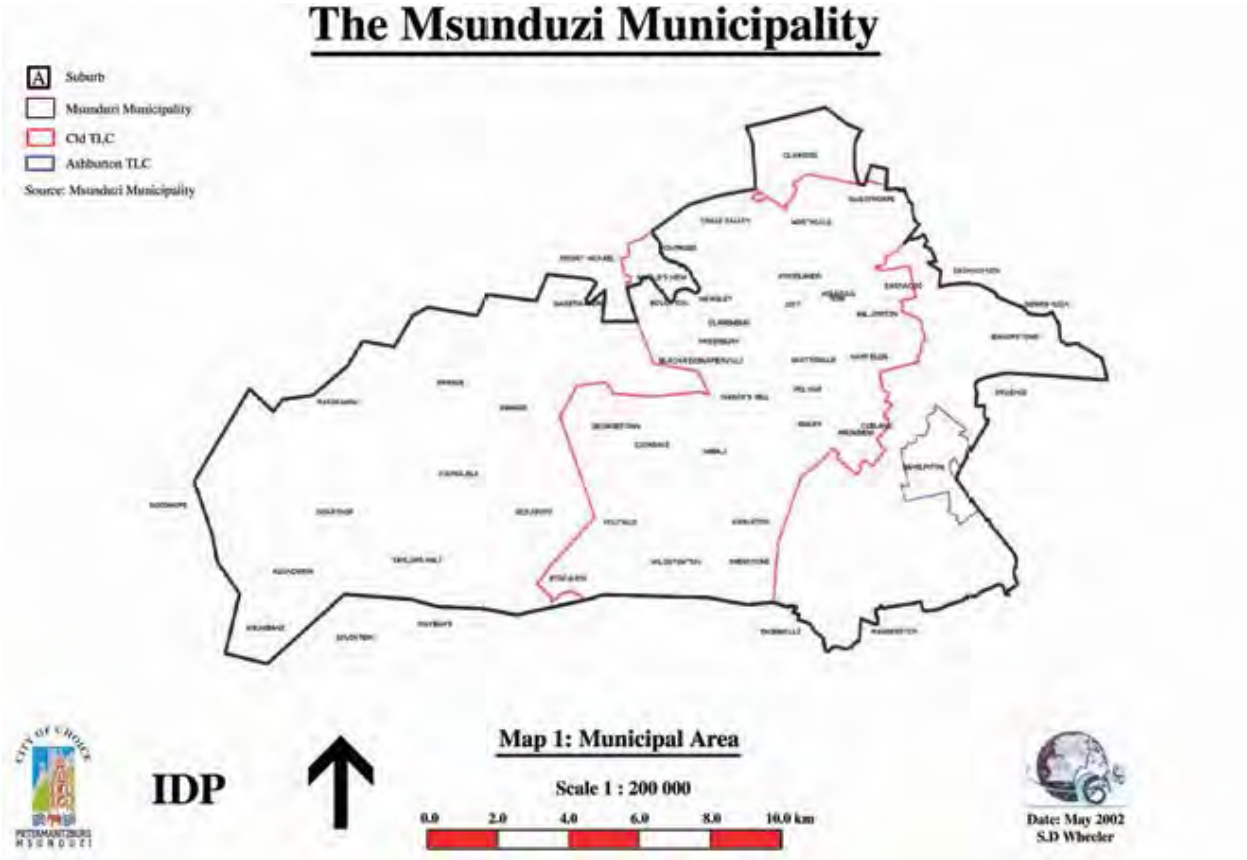

Figure 3.1 Map of Msunduzi Municipality

Source: Pietermaritzburg Msunduzi Municipality, Integrated Development Plan (IDP) (2011-2016)

3.4 Target population

All females, and their spouses, from the Msunduzi Local Municipality who were diagnosed with, as well as survivors of breast cancer, in 2003.

3.5 Study population

The study population consisted of female breast cancer survivors who attended the Pietermaritzburg Cancer Association of South Africa (CANSA) support group, their spouses and children, as well as a group of women from the general public.

3.6 Inclusion criteria

The main criteria for inclusion in the study were that participants:

- Were female breast cancer survivors from the Pietermaritzburg CANSA support group;
- had biological children,
- were above the age of twenty from the general public.

In addition, the women

- had to be from various racial groups on the CANSA support group database;
- could be from rural or urban areas;
- had to reside within a 40km radius of Pietermaritzburg region in KwaZulu-Natal, and
- need not have a spouse (as older participants may have lost their spouses).

3.7 Exclusion criteria

Females below the age of 20 and above the age of 80 in the general public were excluded from participation.

3.8 Sample size

There was no sample size calculation done. All breast cancer patients enrolled within the Cancer Association of South Africa's breast cancer support group, as well as selected family members were enrolled in the study. A convenient number of women from the general public were also enrolled in the study. A total of fifty-six participants were enrolled for the study.

3.9 Selection of sample

The researcher was advised, in 2003, that the Cancer Association of South Africa was running a support group for all breast cancer patients and would therefore be the ideal source from which to gather information on patients with breast cancer. The CANSA breast cancer support group did not have any black or coloured females. All patients requested to participate in the study consented to their participation.

Twenty-eight female breast cancer patients enrolled with the Pietermaritzburg CANSA support group; four families of breast cancer survivors, including two spouses and six children, and twenty females aged between 20 and 70 from the general public were selected as the sample for the study.

3.10 Sampling technique

Purposive and convenient sampling methods were utilized. Purposive sampling was used as it allowed ease of access to a number of breast cancer patients in a localized area. Cost considerations influenced the choice of convenient sampling as all costs for travel were incurred by the researcher.

All breast cancer survivors who were part of the Pietermaritzburg CANSA support group, and agreed to participate, were included in the study. Twenty-eight breast cancer survivors consented to participate and filled in the self-administered questionnaire.

Four families of the twenty-eight breast cancer survivors were then randomly selected to participate in a semi-structured interview with the researcher, during which time the patient was interviewed separately from her spouse and children.

Women from the general public who worked at the South African Police Services (SAPS) Head Office in Pietermaritzburg, third- and fourth-year University of KwaZulu-Natal female students, and home executives in the Northdale area in the Msunduzi local municipality were approached at meetings and home visits to participate in the study.

3.11 Data collection

Quantitative and qualitative data was collected using self-administered questionnaires and semi-structured interviews.

3.12 Data collection instruments

Data was collected by means of two research instruments, namely self-administered questionnaires and semi-structured interviews.

- *Self-administered questionnaires*. These were administered to breast cancer patients and women of the general public. The questionnaire contained multiple choice questions, which included questions on age at menarche, age at birth of first live child, number of previous benign breast biopsy examinations and number of first-degree relatives with breast cancer, as well as open-ended questions. The Gail Model (Euhus, 2001) was used as a guide in developing the multiple choice questions for the questionnaire. The open-ended questions were designed to obtain further information from the multiple choice questions.

- *Semi-structured interviews*. These were conducted with breast cancer patients and their family members. The questions were developed based on the information received from the self-administered patient questionnaires. The semi-structured interviews therefore served as a means of clarifying and expanding on the data collected from the questionnaires.

The utilization of these two data-collection methods prompted the development of four research instruments. Each instrument was developed for one of the specific target groups. Out of the four measuring instruments developed, instruments I, III, and IV, consisted of sub-sections, for easy categorisation and analysis of data (table 3.1).

Table 3.1 Data-collection instruments designed for the study

Instrument	Measurement
Instrument I (Annexure 1)	Self-administered questionnaire for breast cancer patients.
Instrument II (Annexure 2)	Self-administered questionnaire for women from the general public.
Instrument III (Annexure 3)	Semi-structured interview with breast cancer patients.
Instrument IV (Annexure 4)	Semi-structured interview with family members of breast cancer patients.

Instrument I: In an attempt to determine whether women seek genetic counselling regarding a predisposition to breast cancer, the patients were questioned about any advice offered to them regarding the potential risk factors of breast cancer before being diagnosed. Finally, patients were asked if they would have attended a breast cancer risk-

assessment counselling session, to provide them with information about their breast cancer risks, screening options, as well as an option to undergo gene testing for hereditary cancer. The questionnaire also probed the breast cancer patients' knowledge of reconstructive surgery, the success rate of reconstructive surgery, and any type of family counselling offered to their family. The latter question was composed to determine if any counselling was offered to family members of breast cancer patients to assist them in coping emotionally and mentally with the patients' diagnosis.

Instrument II: The questionnaire sought to determine the level of knowledge regarding breast cancer; whether these women conducted clinical or self-breast examinations as a precautionary measure and whether they would be interested in seeking genetic counselling and testing services.

Instrument III: The semi-structured interviews focused mainly on the emotional status of the patient; the need to obtain more information, either medical or genetic in nature, and the need to obtain genetic counselling. Questions regarding counselling were asked at four stages: after diagnosis, before treatment, after treatment, and family attitudes. Questions on counselling sought answers on whether the doctor had suggested they visit or consult a counsellor, type of counsellor visited, and information obtained from the counsellor.

Instrument IV: The questions in these interviews were designed to determine family members' knowledge about breast cancer at the time of diagnosis, and whether they attempted to obtain additional information on breast cancer. Family members were also probed about genetic counselling and risks associated with breast cancer. They were further asked about the type of information they would have appreciated and considered useful from a counsellor.

3.13 Data collection techniques

3.13.1 Questionnaires

The questionnaires were administered in two stages. The questionnaire for the women of the general public was administered two weeks after instrument I.

Instrument II was administered two weeks later as it provided the researcher with an opportunity to identify candidates from the general public to be included in the study and to obtain the necessary consent from these women.

3.13.2 Semi-structured interviews

Semi-structured interviews were scheduled with the breast cancer patients, their spouses and their children, so as to obtain additional information that had not been captured through the administration of the questionnaires.

The selection of breast cancer patients for the semi-structured interviews was such that one woman was selected from the age groups: 30-40, 41-50, 51-60 and 61-70. This ensured that women were married and also had offspring. Four patients were chosen at random from the respondents of the initial questionnaire (instrument I). The chosen patients were then contacted and asked to voluntarily participate in a semi-structured interview. The patients were also informed that their participation would eventually require the participation of their spouse as well as their children in a separate semi-structured interview. It was therefore suggested to them that they discuss this participation in the semi-structured interview with their family, prior to their agreement of participation. The patients were given a period of one week to discuss their family's participation in the interviews and were contacted to determine what decision had been taken. The patients were then asked to advise the researcher of the time, date and venue that would be most suitable for themselves, their spouses, and their children to participate in the semi-structured interviews. The interviews were scheduled at times which participants had requested. This ensured the interviews did not interfere with work, school or university hours. No compensation was provided to participants.

After verbal consent had been obtained from all the participants, final preparations for the interviews were made. The participants were contacted and the dates, times and venues were discussed and agreed upon. All the participants indicated that they felt more comfortable being interviewed at their homes. In instances where the children resided outside Pietermaritzburg, the semi-structured interviews were conducted telephonically. Prior to commencement of each of the semi-structured interviews, breast cancer patients, their spouses, and their children were advised of the purpose of the interview, that all

information provided by them would be treated with the strictest of confidence and that their participation in the interview was voluntary.

At the beginning of each semi-structured interview the breast cancer patient and her spouse and children were informed that each interview would be conducted separately so as to avoid any bias of information obtained, and invasion of privacy. The semi-structured interviews were conducted on an informal basis at the patient's home and this ensured a friendly and relaxed atmosphere.

Of the 12 semi-structured interviews conducted, ten were face-to-face interviews at the patients' home and two were telephonic, as the children resided outside of KwaZulu Natal. All semi-structured interviews were conducted in English. Notes were taken by the researcher during each interview. A time period of one hour was allocated for each semi-structured interview.

3.14 Design and development of the research instruments

The research instruments were designed, after which additional improvements to the original instruments were made in order to develop refined instruments, which ultimately resulted in the final instruments used in the study.

3.15 Pilot study

A pre-test of the instruments was conducted at the University of Kwa-Zulu Natal prior to commencement of the study. This involved twenty-three participants, consisting of breast cancer survivors, as well as students from the University. The pre-test assisted with rectifying ambiguities and grammar in the instruments and resulted in the instruments being amended accordingly. The pre-test resulted in data-collection instruments that were understood by both the researcher and the sampled respondents, providing confidence and satisfaction that the instruments met the acceptable standards of reliability and validity. Results from the pilot interviews were not included in the final results.

3.16 Reliability and validity

The quality of research is determined by its validity and reliability (De Vos *et al.*, 2005). Reliability refers to the extent to which results can provide accurate representation of the total population over time while validity determines whether the research instrument properly measures what it is intended to measure. Pre-testing the instruments in the pilot study resulted in clarification and ensured acceptable standards of validity and reliability.

3.17 Data management

All data received at each stage of the study was captured electronically within a week of receipt and hard copies filed as a back-up. At the end of data capturing, data verification was done by the researcher to avoid any duplication or double entry of data captured.

3.18 Data analysis

Descriptive analysis was conducted on the data. The data was summarised. Continuous numerical data was summarised using measures of central tendency such as means and medians. Categorical data were summarised in proportions and displayed graphically in pie charts or bar graphs.

The raw data obtained from instrument III and IV was organised into conceptual categories, and themes or concepts were created to analyse the data.

3.19 Application and variables measured

3.19.1 Instrument I: Self-administered questionnaire for breast cancer patients

The questionnaire measured the following variables:

- biographical data: age, race, weight and height of the patient
- data on lifestyle before diagnosis: smoking habits, alcohol intake per day, number of servings of vegetables per day, and amount of radiation they had been exposed to in the past
- breast cancer diagnosis: uptake of genetic counselling or risk assessment

3.19.2 Instrument II: Self-administered questionnaire for women from the general public

The questionnaire measured the following variables:

- age
- racial group
- family history of breast cancer
- lifestyle
- knowledge of breast cancer
- uptake of genetic counselling or risk assessment

3.19.3 Instrument III: Semi-structured interview with breast cancer patient

The interview covered:

- provision of counselling
- risk calculations for family members

3.19.4 Instrument IV: Semi-structured interview with family members of breast cancer patients

The interview covered:

- children's and spouse's understanding of breast cancer
- views about breast cancer
- information on breast cancer obtained after diagnosis
- uptake of genetic counselling or risk assessment by the children of breast cancer survivors
- knowledge of breast cancer at the time of diagnosis
- knowledge of associated risks
- methods of primary prevention and early detection of breast cancer.

3.20 Quality assurance

The researcher monitored all the processes involved in planning and conducting this study. The researcher took full responsibility in developing and conducting the research, but with appropriate supervision at all times.

Procedures and methods followed were fully documented. All processes were transparent in obtaining and capturing of the data obtained and all research data obtained from the questionnaires and semi-structured interviews confidentially filed.

3.21 Researcher's role in the study

The researcher's role was to facilitate, as an "outside agent", the administering of the questionnaires and conducting of the semi-structured interviews without any bias, and to further capture and report all results as reported by participants in the study.

3.22 Ethics and informed consent

Ethical approval was waivered by the Dean of Science and Agriculture, Professor D. Jaganyi, due to the fact that when this study was conducted, the amendment to the National Health Act (Act 61 of 2003) was not affected. Hence, obtaining ethical approval from an Ethics Committee was, at the time of this study, not required.

A covering letter accompanied each questionnaire, indicating the purpose of the study, that participation was voluntary and assuring the participant of confidentiality. Prior to each interview, the researcher explained the purpose of the study, the voluntary basis of participation and the assurance of confidentiality. Informed verbal consent to participate was received from each participant before commencing the interview.

3.23 Data collection

Table 3.2 indicates the data-collection methods for the objectives of the study.

Table 3.2: Data-collection methods for the objectives

Objective	Data-collection tool	Sources	Data analysis
To determine biographical, medical and lifestyle profiles of study participants	Instrument I	Surviving breast cancer patients who had undergone a mastectomy, but not necessarily reconstructive surgery	Descriptive analysis: summary of numerical data presented by tables and graphs
To identify the factors that affect the uptake of genetic counselling by breast cancer patients and women in the general public	Instrument I, II and III	Breast cancer patients, women from the general public (control group), as well as female family members of breast cancer patients.	Summary of categorical data. Qualitative analysis – thematic analysis
To assess the willingness of female children of a breast cancer patient to undergo genetic counselling	Instrument IV	Female family members of breast cancer patients	Summary of categorical data. Qualitative analysis – thematic analysis

3.24 Summary

This chapter discussed the research design and methodology, including the development, pre-test, reliability and validity of the data-collection instruments, data management and analysis, and ethical considerations. The whole process afforded the researcher the opportunity to collect quality data from all the participants specific to the study.

CHAPTER IV: RESULTS

4.1 Introduction

This chapter describes the interpretation and explanation of the findings of the study in line with the research questions and objectives of the study. The results are discussed under the following subtopics: socio demographic profile of the study population, racial profile of the study population; risk factors for developing breast cancer; surviving breast cancer patient therapy; genetic counselling - awareness and practice of breast cancer survivors and women in the general public; and genetic counselling of female children of breast cancer patients.

4.2 Results

4.2.1 Biographical, medical and lifestyle profiles

4.2.1.1 Profile of study participants

Twenty-eight female breast cancer patients and twenty females from the general public responded to questionnaires. Four breast cancer patients; two spouses and six children were interviewed (table 4.1)

Table 4.1: Frequency distribution of respondents for each of the instruments (2003)

Study population	Instrument	Number responded	Percentage
Breast cancer patients	I (4 of these patients also answered Instrument III)	28	50%
Women from the general public	II	20	35.7%
Children	IV	6	10.7%
Spouses	IV	2	3.6%
Total		**56**	**100%**

4.2.1.2 Participants' age profile

Patients diagnosed with breast cancer were aged between 34 and 75 years of age, and the women from the general public were aged between 21 and 70 years. The majority (n=43 or 89.6%) of the respondents were 30 to 70 years old (table 4.2).

Table 4.2: Frequency distribution of participants' age groups (2003)

Age group	Breast cancer patients number/ percentage	General population number/ percentage	Total sample number/ percentage
21-30	Nil (0%)	5 (25%)	5 (10.4%)
31-40	4 (14.3%)	9 (45%)	13 (27.1%)
41-50	5 (17.9%)	5 (25%)	10 (20.8%)
51-60	4 (14.3%)	Nil (0%)	4 (8.3%)
61-70	7 (25%)	1 (5%)	8 (16.7%)
71-80	8 (28.6%)	Nil (0%)	8 (16.7%)
Total	**28 (100%)**	**20 (100%)**	**48 (100%)**

4.2.1.3 Patients' age at first breast cancer diagnosis

Of the respondents, 35.7% (n=10) were in the 61 to 70 year age group when diagnosed with breast cancer; 25% (n=7) were in the 41 to 50 year age group; 21.4% (n=6) were in the 51-60 year age group; 14.3% (n=4) were in the 30 to 40 year age group, and only 3.6% (n=1) in the 71-80 year age group (table 4.3).

Table 4.3: Frequency distribution of age at first diagnosis (2003)

Age group	Number of patients	Percentage
30-40	4	14.3%
41-50	7	25%
51-60	6	21.4%
61-70	10	35.7%
71-80	1	3.6%
Total	**28**	**100%**

4.2.1.4 Participants' gender profile

Of the study population participants, 94.6% (n=53) were female and only 5.4% (n=3) were males (figure 4.1). The female participants included working class women (n=34), females who were studying (n=5), as well as home executives (n=14). Of the three male respondents, two were spouses and one was the biological son of the breast cancer patient. The spouses (n=2) were employed and the son (n=1) is a student.

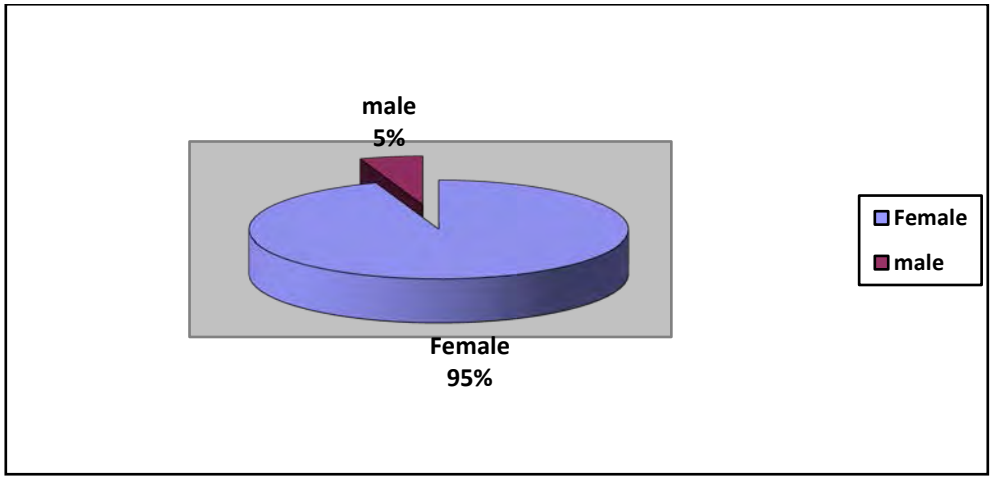

Figure 4.1: Participants' gender distribution (2003)

4.2.1.5 Participants' racial profile

Breast cancer patients from two of the four major South African racial groups, namely Indian and White, responded to instrument I; whilst Black, Indian and White women from the general public responded to instrument II. One Indian and one White spouse were interviewed, and of the six children interviewed, four were white females and two Indian females (figure 4.2).

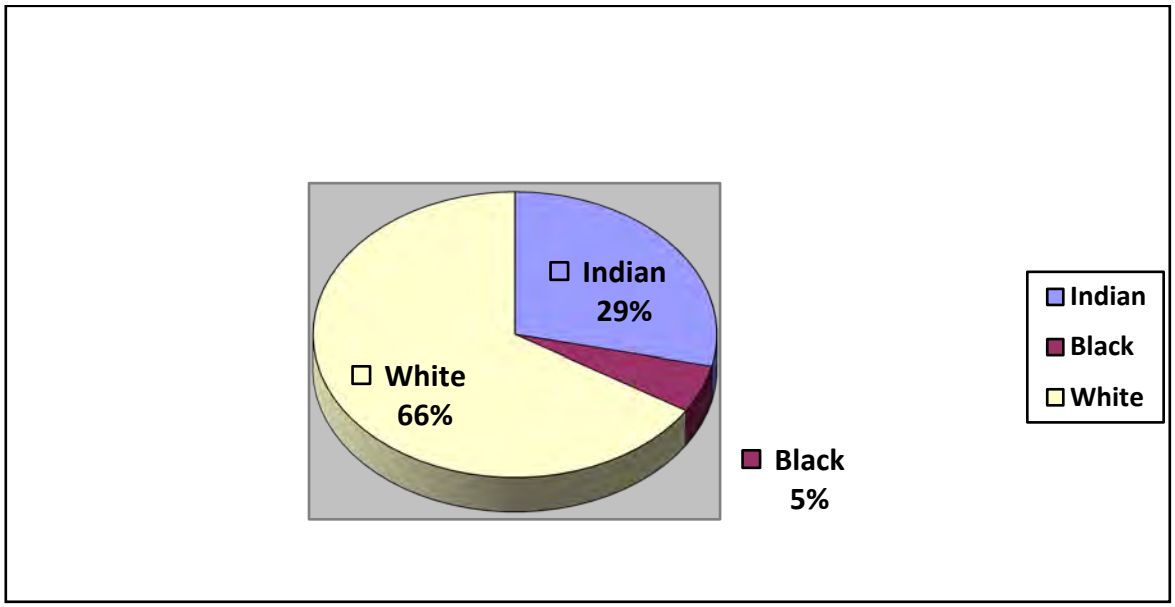

Figure 4.2: Participants' racial profile

4.2.2 Risk factors which may contribute to the development of breast cancer

4.2.2.1 Patients perspective

Of the twenty-eight breast cancer patients, 28.5% (n=8) noted that only one of the factors listed affected them in some way, while 3.5% (n=1) noted that none of the factors listed affected her in any way.

Ten of the twenty-eight breast cancer patients, 35.7% (n=10) noted that two of the seven factors affected them as follows: 50% (n=5) indicated early menstruation and early live birth; 20% (n=2) indicated early menstruation and alcohol; 10% (n=1) indicated smoking and early live birth; 10% (n=1) indicated alcohol and radiation, and 10% (n=1) indicated early menstruation and oestrogen contraceptives. Twenty three (82%) of the patients identified early menstruation as a factor that could probably have contributed to the development of their breast cancer (table 4.4).

Table 4.4: Frequency distribution of risk factors which may have contributed to the development of breast cancer according to breast cancer patients (2003)

Risk factors	Breast cancer patients number	Percentage
Alcohol	8	11.9%
Smoking	3	4.5%
Radiation	2	3%
Early menstruation (<15 years of age)	23	34.3%
Early live birth (<24 years of age)	16	23.9%
Late live birth (>25 years of age)	11	16.4%
Oestrogen contraceptives	4	6%

4.2.2.2 General public perspective

Of the twenty respondents from the general population, 10% (n=2) noted that none of the factors listed affected them in any way and 25% (n=5) noted that only one of the factors listed affected them in some way. Ten respondents (50%) noted that two of the seven factors affected them as follows: 80% (n=8) indicated early menstruation and early live birth, and 20% (n=2) indicated early menstruation and oestrogen contraceptives.

Three respondents, (15%), noted that three of the seven factors affected them as follows: 66.7% (n=2) indicated early live birth, early menstruation and oestrogen contraceptives, and 33.3% (n=1) indicated smoking, early live birth and early menstruation.

Fifteen of the twenty respondents from the general public, 75% noted early menstruation as a factor that could probably contribute to them developing breast cancer (table 4.5).

Table 4.5: Frequency distribution of risk factors which may contribute to the development of breast cancer according to women from the general public (2003)

Risk factors	General population number/ percentage	Total sample number/ percentage
Alcohol	Nil	0%
Smoking	1	2.9%
Radiation	Nil	0%
Early menstruation (<15 years of age)	15	44.1%
Early live birth (<24 years of age)	14	41.1%
Late live birth (>25 years of age)	Nil	0%
Oestrogen contraceptives	4	11.8%

4.2.3 Common symptoms, survival and therapy for breast cancer patients

4.2.3.1 Common symptoms before breast cancer diagnosis

The majority of the patients, (67.9%; n=19) experienced a lump or thickening in the breast or armpit, which led to a positive diagnosis of breast cancer while 14.3% (n=4) experienced none. One patient, (3.6%) had a combination of two symptoms, namely a lump or thickening in the breast or armpit as well as changes around the nipple.

A combination of three symptoms was experienced by 7.1% (n=2) of the patients; one patient experienced a lump or thickening in the breast or armpit, together with changes around the nipple and changes in size or shape of the breast, and the other patient experienced changes in the skin and nipple, as well as changes in the size or shape of the breast.

Table 4.6: Frequency distribution of patients' most common symptoms before diagnosis (2003)

Symptoms	Number of patients	Percentage
Lump or thickening in the breast or armpit	19	67.9%
Changes in the skin – dimpling, puckering or redness	3	10.7%
Changes in the nipple – direction of the nipple or an unusual discharge	3	10.7%
Changes around the nipple – unusual rash or sore area	2	7.1%
Changes in the size or shape of the breast	2	7.1%
None	4	14.3%

4.2.3.2 Years of survival after breast cancer diagnosis

The majority of patients, 57.1% (n =16), had survived between 0 and 5 years after being diagnosed with breast cancer, whilst 28.6% had survived for between 6 and 10 years and 14.3% of patients had survived between 11 and 15 years after the diagnosis. At the time of the study four patients were between the ages of 30 and 40, seven patients were between the ages of 41 and 50, seven patients were between the ages of 51 and 60, nine patients were between the ages of 61 and 70 and only one patient was 77 at the time of diagnosis (table 4.7).

Table 4.7: Frequency distribution of years of survival after breast cancer diagnosis (2003)

Number of years	Number of patients	Percentage
0-5	16	57.1%
6-10	8	28.6%
11-15	4	14.3%

4.2.3.3 Surviving breast cancer patient therapy

Only one (3.6%) of the 28 patients underwent chemotherapy alone, whilst 12 (42.9%) of the 28 patients did not have chemotherapy or radiation therapy and opted for surgery. Seven (25%) of the 28 patients underwent chemotherapy and surgery, whilst eight (28.6%) patients opted to have chemotherapy, radiation therapy as well as surgery (table 4.8).

Table 4.8: Frequency distribution of type of therapy breast cancer patients underwent (2003)

Type of therapy	Number of patients	Percentage
Chemotherapy	1	3.6%
Surgery	12	42.9%
Chemotherapy + Surgery	7	25%
Chemotherapy + Radiation therapy + Surgery	8	28.6%
Total	**28**	**100%**

4.2.4 Knowledge about breast cancer and provision of counselling

4.2.4.1 Level of knowledge amongst participants about breast cancer

4.2.4.1.1 Knowledge of breast cancer

Nine patients (32%) indicated they were unsure about the causes of their breast cancer; five patients (18%) attributed the cause of their breast cancer to hormone replacement therapy; three patients (10.7%) attributed the cause of their breast cancer to stress; two patients each (7%) to a sports injury and hereditary; and one patient each to smoking, insufficient exercise, environmental pollution, rapid cell change and post natal depression. One patient attributed the cause of their breast cancer to two causes (stress and heredity); and a further patient listed four causes (stress, hormone replacement therapy, environmental pollution and insufficient exercise) as reasons for the development of her breast cancer.

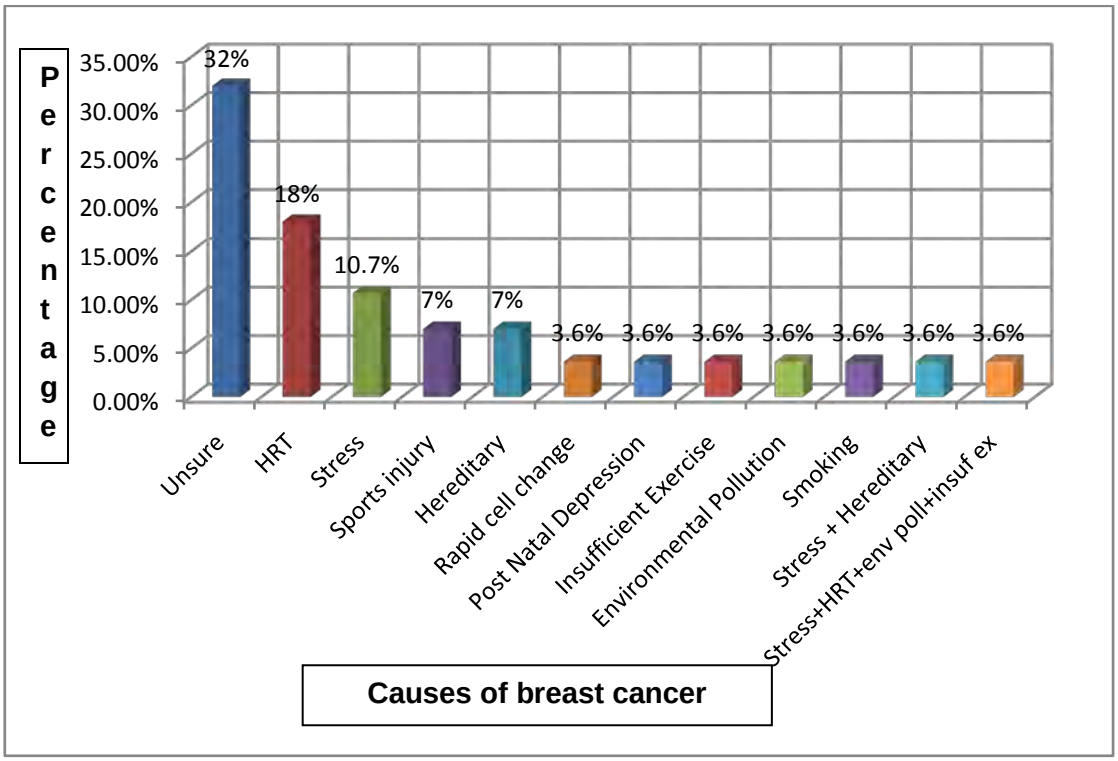

Figure 4.3: Most likely causes of breast cancer according to breast cancer patient in the Msunduzi Municipality (2003)

4.2.4.1.2 Participants' interest in attending a risk assessment session

Twenty-one study participants (43.8%, n=21) indicated they were not interested in attending a risk assessment session; 35.4% n=17 showed their interest in attending such a session; whilst 20.8% n=10 indicated they were unsure (table 4.9).

Within the breast cancer patients eleven of the twenty eight breast cancer patients (39.3%, n=11) indicated that they would have been interested in sending their daughters for a risk assessment after their diagnosis, twelve patients (42.3%, n=12) indicated that neither them nor any of their immediate family members were interested, and 17.9% (n=5) were unsure about having a risk assessment done, either for themselves or their family.

In the general population 45% (n=9) of respondents indicated that they would not be interested in attending a risk assessment session. One of these nine respondents who indicated that they would not like to have a breast cancer risk assessment done, explained

that whilst she may not be interested at present, should the need arise for such an assessment in future, she will definitely go. Thirty percent of respondents (n=6) indicated their interest in attending a risk assessment session whilst 25% (n=5) of respondents said they were unsure. Twenty percent of the participants (n=1) who indicated she was unsure, further clarified that she would need to know what type of service a genetic counsellor offered prior to going to one.

Table 4.9: Frequency distribution of study participants' interest in attending a risk assessment session (2003)

Risk assessment session	Breast cancer patients Number / percentage	General population Number / percentage	Total sample Number / percentage	General population Number / percentage
Attended	Nil	Nil	Nil	Nil (0%)
Interested in attending	11 (39.3%)	6 (30%)	17 (35.4%)	17 (35.4%)
Not interested in attending (after diagnosis)	12 (42.9%)	9 (45%)	21 (43.8%)	21 (43.8%)
Unsure	5 (17.9%)	5 (25%)	10 (20.8%)	10 (20.8%)
Total	**28**	**20**	**48**	**100%**

4.2.4.1.3 Advice on breast cancer risk factors

Fourteen (50%) people had received advice on risk factors for breast cancer. Of these, nine (64.3%) indicated that they had received this advice through the media; either through reading magazine articles, pamphlets or through advertisements. Four of the fourteen (28.6%) patients indicated that they had received advice on breast cancer risk factors from their general practitioner (GP) or gynaecologist. Only one (7.1%) patient indicated that she had received advice from her doctor as well as through the media (table 4.10).

Table 4.10: Frequency distribution of patients who were either advised or not advised about breast cancer risk factors prior to diagnosis (2003)

Advice on risk factors	Number of patients	Percentage	Advised by
Advised prior to diagnosis	14	50%	Media (64.3%) GP/Gynaecologist (28.6%) GP and media (7.1%)
Not advised prior to diagnosis	14	50%	

4.2.4.1.4 Advice on factors that reduce the risk of recurrence of breast cancer

Seventeen patients (60.7%) were advised on factors that reduce the risk of developing breast cancer. Of the four patients in the 30 to 40 year age group, only one patient indicated that she was offered advice; four of the five patients in the 41 to 50 year age group, three patients each in the 51 to 60 and 61 to 70 year age group and six patients in the 71 to 80 year age group indicated that they were offered advice (figure 4.4).

These figures indicate that 80 percent of the 41 to 50, and 75 percent each in the 51 to 60 and 71 to 80 year age groups were offered advice. Only 42 and 25 percent of the 61 to 70 and 30 to 40 year age groups, respectively, were advised on these factors.

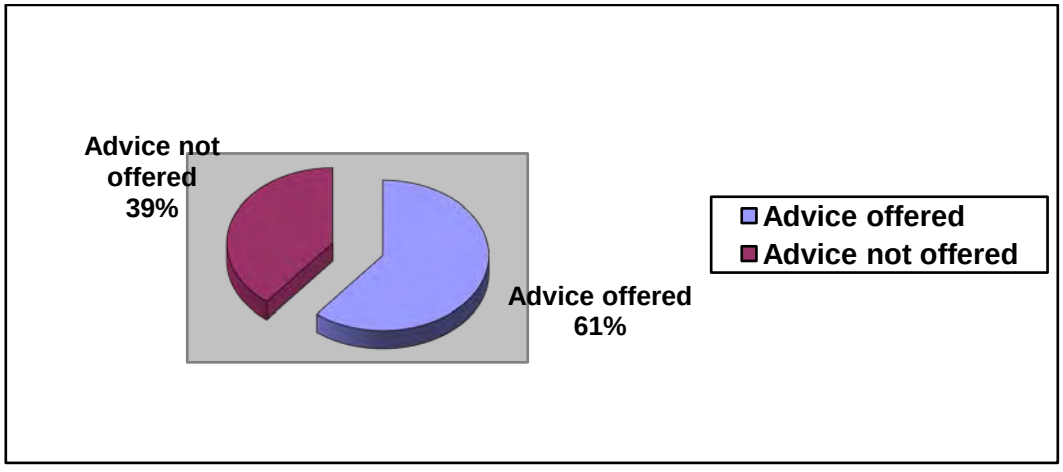

Figure 4.4: Advice received on factors that reduce the risk of recurrence of breast cancer (2003)

4.2.4.1.5 Advisor on reduction of risk factors related to breast cancer recurrence

Eleven patients (39.3%) indicated they had not received any advice on reduction of risk factors related to the recurrence of breast cancer. Eight patients (25%) indicated that they had been advised by a medical doctor alone; six (21.4%) patients indicated that they had been advised by a person not listed in Table 4.11; three (10.7%) patients indicated that they were each advised by a medical doctor and a person not listed; medical doctor and medical nurse; and medical doctor and a social worker. One (3.6%) patient indicated that she had been advised by a medical doctor, social worker and a dietician advised them; another patient (3.6%) indicated that a psychologist, medical doctor, medical nurse and a dietician advised her; and one patient (3.6%) indicated that a psychologist alone advised her. None of the 28 patients were counselled by a genetic counsellor.

Only one of the seven patients indicated that they were advised by various speakers who were called to present during support group meetings. The other six patients did not indicate who they had been advised by.

Table 4.11: Frequency distribution depicting advisor on the reduction of risk factors related to the recurrence of breast cancer (2003)

Advisor	Number of patients	Percentage
Medical doctor	7	25%
Other	6	21.4%
Medical doctor + other	1	3.6%
Medical doctor + medical nurse	1	3.6%
Medical doctor + social worker	1	3.6%
Medical doctor + social worker + dietician	1	3.6%
Psychologist + medical doctor + medical nurse + dietician	1	3.6%
Psychologist	1	3.6%
None	9	32.1%
Genetic counsellor	Nil	0%
Total	**28**	100%

4.2.4.1.6 Link between advice on risk of recurrence and patient's opinion on causes of their breast cancer

Of the eleven patients recorded as not being advised about the risk for reducing breast cancer, seven were unsure as to the causes of their breast cancer. Of the remaining four patients who were not advised about the risk for reducing breast cancer, one patient indicated that she thought it was due to a sport injury but was not quite sure; another indicated that she thought it is hereditary as a number of her relatives have had breast cancer, the third patient indicated that she was told that her cells were changing rapidly and the fourth patient indicated that she thought it was due to her post-natal depression when she had a very stressful life (Table 4.12).

Table 4.12: Frequency distribution of link between advice on breast cancer risk factors and patients' opinion as to the causes of their breast cancer (2003)

Causes according to patient	Number of patients	Number / percentage of patients to whom Advice on risk factors offered	Number / percentage of patients to whom advice on risk factors was not offered	Total number / percentage
Stress	6	6		6 (18.8%)
Post-natal depression	1		1	1 (3.1%)
Rapid cell change	1		1	1 (3.1%)
Hereditary	3	2	1	3 (9.4%)
Hormone replacement therapy	6	6		6 (18.8%)
Environmental pollution	2	2		2 (6.3%)
Insufficient exercise	1	1		1 (3.1%)
Smoking	1	1		1 (3.1%)
Sport injury	2	1	1	2 (6.3%)
Unsure	9	2	7	9 (28.1%)

4.2.4.2 Genetic counselling and risk assessment

4.2.4.2.1 Counselling opportunities provided for families of breast cancer patients

Only five of the twenty-eight (17.9%) patient's families were given the opportunity to be counselled after the diagnosis. Two of the five patients were between the ages of 41 and 50 and one patient each in the 51 to 60, 61 to 70 and 71 to 80 age groups (figure 4.5).

Only two of the five patients indicated who counselled their families; one patient indicated that a medical doctor had counselled her family and the other indicated that her oncologist had counselled her family. Both these patients were in the 41 to 50 year age group.

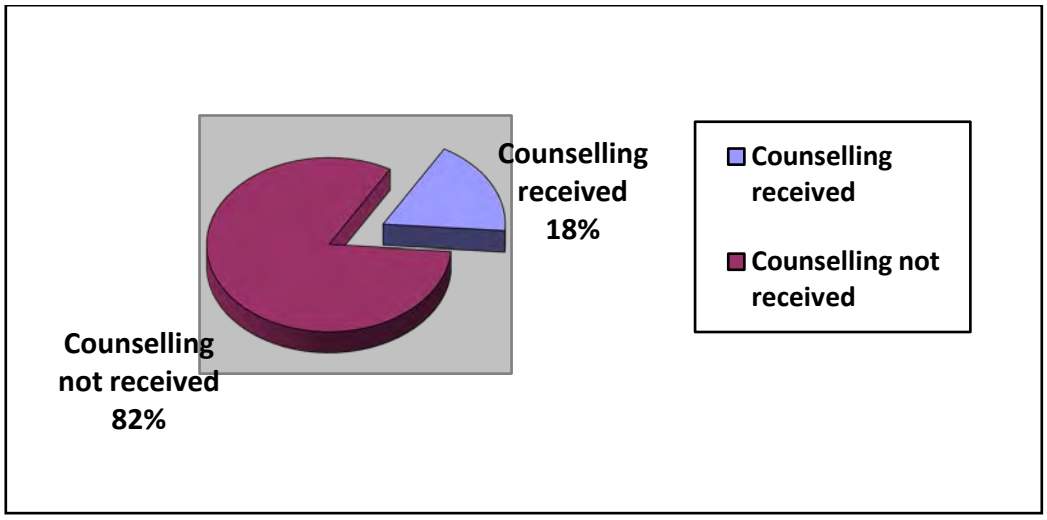

Figure 4.5: Counselling opportunities provided to families of breast cancer patients (2003)

4.2.4.2.2 Provision of counselling

At the time of diagnosis only one patient was counselled and three were not. Although counselling was provided to one patient at the time of diagnosis, she had indicated that no counselling was given on the genetics and risks involved with breast cancer. She further indicated that she was not satisfied with the information the psychologist had offered, and considered information on the genetics, recurrence, prognosis and family risk to be essential information that should have been provided.

The three patients who were not provided with counselling at the time of diagnosis mentioned that even though they had not been counselled they would have found counselling on the genetics and risks of breast cancer very useful. Of these three patients one patient mentioned that she was quite comfortable with her doctor, one patient said that she would have liked to consult with an oncologist and the other patient said that she would have liked to meet with a person who went through a similar experience as she did.

During the period before surgery two of the four patients indicated they had been counselled, one by a plastic surgeon and one by a surgeon. The patient counselled by the surgeon was presented with two options, to have a mastectomy or, to continue with chemotherapy and radiation therapy. The patient counselled by the plastic surgeon was given information on reconstructive surgery.

Of the two patients who were not counselled, one said that she would not have wanted to consult with a counsellor and the other patient indicated that although she did not consider counselling, she thought that family counselling for trauma would have assisted in easing the pain. She further felt that the views of an oncologist, as well as information on the advantages and disadvantages of lumpectomy should have been provided. In addition, the patient indicated that by her not being provided with this information she felt the surgeon had denied her of her right to information.

During the period after surgery only one patient was counselled by the Reach for Recovery breast cancer support group. This is the patient who indicated before surgery that she did not want to be counselled and mentioned that she found it “helpful to talk to people who have been through the same situation”.

One of the three patients who were not counselled after surgery reiterated that although she did not have any counselling, she thought that family counselling would have helped. The other two patients stated that even though they were counselled before surgery, they were of the opinion that counselling provided by an appropriate counsellor rather than a surgeon and plastic surgeon would have helped them cope better with their mastectomy (Table 4.13).

Table 4.13: Provision of counselling to the patient at various stages (2003)

Stages at which counselling took place	Provided with counselling	Not provided with counselling	Type of counsellor
At diagnosis	1 (25%)	3 (75%)	Psychologist
Before surgery	2 (50%)	2 (50%)	Surgeon; Plastic Surgeon
After surgery	1 (25%)	3 (75%)	Reach for Recovery

4.2.5 Genetic counselling, risks, risk assessment, awareness and education for family members of breast cancer patients

4.2.5.1 Genetic counselling, risks and risk assessment

Six of the eight respondents indicated they had not been for genetic counselling and were not counselled on the risks associated with the development of breast cancer. Of the six, two respondents indicated that although they did not receive genetic counselling or information on the risks associated with breast cancer, they were of the opinion that such information would have been very valuable.

The remaining two respondents indicated that they had been for genetic counselling and have been informed about the risks associated with the development of breast cancer. This assisted them in understanding how they could be affected by breast cancer.

In terms of the risk assessment, four of the eight respondents indicated that they were not interested in attending a breast cancer risk assessment. However, one of these four respondents indicated that she might consider a risk assessment at a later stage. The remaining four respondents showed their interest in attending a risk assessment.

4.2.5.2 Precautionary measures, creating awareness and educating family members on breast cancer

Five of the respondents were females and two of the five indicated that they have not taken any precautionary measures. One of the three remaining females responded saying that she is now consuming more soya and will not take any hormone pills; the second and

third indicated that they were now going for regular check-ups, whilst the third respondent further added that she was also going for regular mammograms.

In terms of creating awareness, seven of the eight respondents were of the opinion that enough was being done. However, in terms of educating families of breast cancer patients, half of the respondents (four) believed that enough was being done whilst the other half thought that not enough was being done to educate family members of breast cancer patients (Table 4.14).

Table 4.14: Precautionary measures, education and raising awareness about breast cancer

Respondent	Precautionary measures taken by female offspring	Thoughts about creating awareness on breast cancer	Thoughts about educating family members of patients
1		Not enough being done	Not enough being done
2	Yes	Enough being done	Enough being done
3		Enough being done	Enough being done
4	Yes	Enough being done	Enough being done
5		Enough being done	Not enough being done
6	No	Enough being done	Not enough being done
7	No	Enough being done	Enough being done
6	No	Enough being done	Not enough being done

4.3 Summary

This chapter provides a detailed overview of the results of the study indicating that there is a lack of education and awareness about genetic counselling and risk assessment. Women from the general public may have basic knowledge about breast cancer but do not know about the genetic services available. Likewise, there is very little, if any, emphasis placed on the families of breast cancer patients. Appropriate counselling to assist them with understanding and accepting the diagnosis as well as the provision of appropriate information on the genetics of breast cancer, genetic counselling availability and risk assessment is not offered to all families.

CHAPTER V: DISCUSSION

5.1 Introduction

This chapter presents the data interpretation and explains the findings in line with the research questions and objectives of the study while at the same time making comparisons with findings of similar studies that have been conducted in other settings. The results are discussed under the following subtopics: biographical, medical and lifestyle profiles of the study participants; knowledge, awareness and practice; and genetic counselling, risk assessment, awareness and education for family members of breast cancer patients.

5.2 Findings

The key findings of this study show that participants identified that early menstruation (<15 years of age), early first pregnancy, before the age of 24 years were noted as major risk factors for the development of breast cancer. A lump or thickening in the breast or armpit prior to diagnosis was the most common symptom or sign of breast cancer, with surgery being the most common modality of treatment.

Of the eight family members interviewed, four family members (50%) indicated that they were not offered genetic counselling and would not be interested in knowing about the risks associated with breast cancer. Of the remaining four, two (25%) indicated that although they were not offered genetic counselling they would be interested in genetic counselling and learning about the risks associated with breast cancer as such information would be of great value.

In terms of a breast cancer risk assessment, four of the eight family members (50%) indicated they were not interested, with one further explaining that she might be interested at a later stage. The remaining four family members (50%) indicated their interest in attending a breast cancer risk assessment.

Five of the eight family member (62.5%) interviewed were females. Three females (60%) indicated they were taking precautionary measures by going for regular check-ups and mammograms, consuming more soya and not taking any precautionary measures.

Seven of the eight family members (87.5%) indicated that they thought enough was being done to create awareness on breast cancer; whilst four family members (50%) were of the opinion that not enough was being done to educate family members of breast cancer patients.

The study found that the participant breast cancer patients were not informed about the availability of genetic counselling and testing and neither were their family members. Besides genetic counselling, family members, together with the breast cancer patient, were not appropriately counselled or provided with the option of having a risk assessment done. The health care professionals did not seem to be providing relevant information to the patients and their family, which would assist both the patient and the family in making informed decisions about prevention and early detection for their children.

5.2.1 Biographical, medical and lifestyle profiles

5.2.1.1 Participants' age and racial profile

Approximately 54% (n=15) of the breast surviving patients were between 51 and 80 years of age. Anecdotal evidence provided by the chairperson of the support group indicated that a number of young female breast cancer survivors were working women and mothers. Their career and family did not allow them sufficient time to be involved in the cancer support group. However, older women had more time available and attendance at the support group sessions provided them with a sense of fulfilment to assist other female breast cancer patients cope with their breast cancer.

The majority of patients in this study were White, whilst two were Indian and none were Black. This was in keeping with the available information in the National Cancer Registry (2014) on the incidence of breast cancers amongst the various population groups in South Africa.

According to the National Cancer Registry Report (2014), breast cancer was the most commonly diagnosed cancer in all females, accounting for 21.02% of all cancers diagnosed with an age-specific incidence rate of newly diagnosed cases in Whites being 54.74 per 100 000, and 15.87 per 100 000 for Black females.

5.2.1.2 Age at first breast cancer diagnosis

According to Buckman (1997), a woman's chances of developing breast cancer increases with age; it is rare before the thirties, begins to increase in incidence in the forties, and becomes more common in the fifties and over. The findings of this study, which found that 60.7% (n=17) were over the age of 50 at the time of being diagnosed with breast cancer; concur with Forbes (1997) who found that developing breast cancer increased with age, thereby making age the most important risk factor for breast cancer.

However, even though a woman's chances of developing breast cancer increases with age, the impact of developing breast cancer at a younger age is probably more difficult for a younger woman to accept and deal with than women over the age of 50. Avis *et al.* (2005) found that younger breast cancer survivors are at risk for impaired quality of life for several years after diagnosis and may need interventions that specifically target their needs related to menopausal symptoms and problems with relationships and body image.

5.2.1.3 Risk factors which may contribute to the development of breast cancer according to participants

The breast cancer patients noted a variety of risk factors that may have contributed to them developing breast cancer. Of the twenty-eight patients, 82% (n=23) noted early menstruation (<15 years of age) and 57% (n=16) noted early live birth (<24 years of age) as the main risk factors to the development of breast cancer. Of the twenty women from the general population, 75% (n=15) noted early menstruation and 41.1% (n=14) indicated early live birth (<24 years of age) as a factor that could probably contribute to their development of breast cancer. This contrasted with Kelsey *et al.* (1993) who found that later age at first birth was associated with increased risk of breast cancer.

Of the 28 breast cancer patients, 29% (n=8) indicated that alcohol may have contributed to their development of breast cancer, whilst 10.7% (n=3) indicated that they were of the opinion that smoking may have contributed to their development of breast cancer. Brown *et al.* (2009) found that low alcohol intake is not related to increased breast cancer risk in Asian-American women and that neither alcohol nor cigarette use contributed to the elevated risks in Asian-American women. Over the years smoking has been postulated to increase the risk of breast cancer (Brown *et al.*, 2009); however, whilst many epidemiological studies have not supported an overall association, there are still unanswered questions regarding the influence of early initiation and long duration (Terry, 2002; Ahern *et al.*, 2009).

Interestingly, none of the patients noted family history as a risk factor which may have contributed to the development of their breast cancer. None of the women from the general public had identified family history as a risk factor.

5.2.1.4 Most common symptoms before breast cancer diagnosis

Aiello *et al.* (2004) found that the presence of a lump was associated with a two- to three-fold greater risk of breast cancer with and without the presence of any other symptoms in postmenopausal women. In our study 67% (n=19) indicated that they had a lump or thickening in the breast or armpit, which led to a positive diagnosis of breast cancer. Aiello *et al.* (2004), therefore suggest that a reported lump must be fully evaluated and reassure physicians that careful observation of women with nipple discharge and pain may be justified.

5.2.1.5 Years of survival after breast cancer diagnosis

The National Cancer Institute estimates that approximately 2.6 million US women with a history of breast cancer were alive in January 2008, more than half of whom were diagnosed less than 10 years earlier (American Cancer Society, 2011). Allen *et al.* (2009) attribute the rising numbers of women surviving and living with breast cancer for longer periods of time to improved methods of early detection and treatment. Burstein and Winer (2000) indicate that ninety-one percent of women diagnosed annually with breast cancer

will survive more than five years. The results of this study show that 42% (n=12) have survived between 6 and 15 years after being diagnosed with breast cancer.

5.2.1.6 Advice on the advantages and disadvantages of breast cancer therapy

The patient and physician often decide on an appropriate treatment option together for the patient after taking into consideration a number of factors, such as the biological characteristics and stage of the cancer, the patient's age and preferences, as well as the risks and benefits related to each treatment protocol (American Cancer Society, 2011). However, most breast cancer patients will undergo surgery which is often combined with other methods of treatment such as radiation therapy, hormone therapy, chemotherapy and/or targeted therapy (American Cancer Society, 2011).

A woman who chooses lumpectomy and radiation will have the same expected long-term survival as if she had chosen mastectomy; however, there is a higher risk of local recurrence (cancer returning to the breast) with lumpectomy (Jatoi & Proschan, 2005). The manner in which radiation therapy is given is dependent on the type, stage and the area in which the tumour is located (American Cancer Society, 2011). Accurate targeting of radiation therapy has radically increased in the past few years resulting in fewer side effects and a reduction in treatment time (Beitsch *et al.*, 2011). Women who have metastatic breast cancer and who may not qualify for surgery due to the wide spread of the cancer, would qualify for systemic therapy as their foremost treatment option (American Cancer Society, 2011).

In a study on breast cancer patients 70 years and older, Wang *et al.* (2011) found that surgery was performed in the majority of these patients, with approximately 50% undergoing lumpectomy and 50% undergoing a mastectomy. Adjuvant therapies were frequently excluded, with only hormonal therapy being the most commonly used. The overall 5-year survival rate was significantly worse in patients 80 years and older (Wang *et al.*, 2011).

In this study 57% (n=16) of the breast cancer survivors chose to have a mastectomy after being diagnosed with breast cancer. Only one patient had bilateral breast cancer, resulting in the patient having a total mastectomy on the right side and a radical mastectomy on the left side. Paci *et al.* (2002) reported that there had been a change in rates of radical surgery and incidence of breast cancer since the introduction of the Florence mammographic screening programme. The rates of breast conserving surgery in women aged 50-69 were 1.18 per thousand in 1990 and increased to 1.87 per thousand in 1996, whilst the rates of mastectomy decreased from 1.08 to 0.62 per thousand from 1990 to 1996 (Paci *et al.*,2002). The researcher is of the opinion that such a screening programme may be beneficial to the South African population, particularly since breast cancer was the leading cancer in females in South Africa in 1999, with 19.36% of females being diagnosed with the disease (Mqoqi *et al.*, 2004).

5.2.2 Knowledge about breast cancer and provision of counselling

5.2.2.1 Cause of breast cancer according to patient

According to Chalmers *et al.* (1996), women may develop perceptions of vulnerability from a lived experience of cancer and through strong identification with an affected or deceased mother or sister. This would cause them to judge an experience that is cognitively available as more likely to occur, and beliefs about the frequency of lethal events may lead to an overestimation of risk of disease occurrence or of the seriousness of risk (Hopwood, 2000). In keeping with this, Offit and Brown (1994) found that women who had strong family histories might admit to being at an increased risk, but often thought in non-Mendelian terms and were more influenced by their particular familial experience of the condition. Of the breast cancer patients, 21.4% (n=6) listed stress as the most likely cause for the development of their breast cancer; a further 21.4% (n=6) listed hormone replacement therapy.

Of the patients, 7.1% (n=2), whose sisters had developed breast cancer, considered their breast cancer as hereditary. Koehly *et al.* (2008) reported that significant within-family correlation was found for breast cancer risk and worry, suggesting that sisters perceived a shared threat for breast cancer.

5.2.2.2 Participants' interest in attending a risk assessment session

A Canadian study conducted by Bottorff *et al.* (2002) found that the two most frequent reasons women gave for being interested in genetic testing for breast cancer risk were "curiosity" and "to warn family." Other frequently reported reasons were "to take preventive action", "to achieve peace of mind" and "to reduce worry" (Bottorff *et al.*, 2002).

This study has found that 39.3% (n=11) indicated their interest in sending their daughters for a risk assessment.

Bottorff *et al.* (2002) further showed that women expressed interest in genetic testing for breast cancer. The percentage of women interested in genetic testing is estimated to range from 43 to 89%, with interest being higher in younger women and women with a diagnosis of breast cancer (Bottorff *et al.*, 2002). The current study concurs with this in that 30% (n=6) aged between 21 and 40 years indicated interest in having breast cancer risk assessment.

5.2.2.3 Advice on breast cancer risk factors prior to breast cancer diagnosis

In a study amongst UK women, Grunfeld *et al.* (2005) found that although women had a good understanding of certain aspects of breast cancer, there were variations in knowledge of risk and the various symptoms related to breast cancer. Older women were particularly poor at identifying symptoms of breast cancer, risk factors associated with breast cancer and their personal risk of developing the disease. The inferior knowledge of symptoms and risks among older women could probably clarify the strong correlation between older age and delay in seeking healthcare (Grunfeld *et al.*, 2005).

5.2.2.4 Advice and advisor on the reduction of risk factors related to the recurrence of breast cancer

According to Manuel *et al.* (2007), younger women show greater psychological morbidity than older women after a breast cancer diagnosis. Manuel *et al.* (2007) suggest that clinicians should identify patients' particular stressors and help with advice on coping techniques targeting particular concerns, one being social support, which is helpful in dealing with anger or depression.

One of the coping techniques is providing the opportunity for counselling. In this study only 18% of the target group had the opportunity for family counselling. Every patient should be offered the opportunity to undergo counselling, whether it is for herself or her family. It should be the responsibility of the doctor who diagnosis her breast cancer to provide her with this information. However, this information about counselling should be suggested and not forced upon the patient. By equipping the patient with relevant information at the time of diagnosis, enables the patient to be knowledgeable about the services available to her and her family. Ultimately though, it must be the decision of the patient as to whether her family or the patient should attend counselling and at a time when she considers it appropriate.

From the data, very little if any emphasis is placed on the well-being of a patient's family. A diagnosis of breast cancer not only has a negative impact on the patient, but impacts on her entire family. Allowing a patient's family to be involved, with the consent of the patient, from the moment of diagnosis, would, in most cases, create a supportive environment for the patient. This would lead to the patient being able to share her fears and frustrations with her family instead of having to deal with the situation on her own. Moreover, this could have a positive impact on the patient's well-being as a supportive and caring environment will allow her to cope better with her diagnosis, as well as speed up the recovery process.

5.2.2.5 Link between advice on risk of recurrence and patients' opinion on causes of their breast cancer

Graham *et al.* (2002) state that the relation between stressful life experiences and the onset of breast cancer has been the subject of considerable research, much of which has been characterised by weak design. However, a meta-analysis conducted by Petticrew *et al.* (1999) concluded that the few well-designed studies that have been carried out have failed to find a link between stressful life experiences and the onset of breast cancer (Graham *et al.*, 2002).

Research has shown that increased levels of physical activity decrease the risk of developing breast cancer in the general population by between 20 and 40% (Monninkhof *et al.*, 2007; Lahmann *et al.*, 2007; Sprague *et al.*, 2008). There is a greater association in

postmenopausal women, although Maruti *et al.* (2008), Suzuki *et al.* (2008) and Howard *et al.* (2009) maintain that increased physical activity may decrease the risk for premenopausal breast cancer as well. Due to physical activity being one of the few modifiable risk factors, it may provide a target to add to breast cancer prevention in *BRCA1*/2 mutation carriers (Pijpe *et al.*, 2009). However, the results of this study have shown only one breast cancer survivor stating insufficient exercise as a probable cause of her developing breast cancer.

5.2.2.6 Provision of counselling for the patient at the time of diagnosis

The patients experienced various emotions ranging from “numbness” and “denial” to “devastation” and “shock.” Since the sample group was so small, it was not possible for the researcher to classify the range of emotions under a specific type of emotion. However, it can be said that all the patients went through a number of different and varying degrees of emotions. According to Avis et al (2005), preparing younger women for the impact of breast cancer may also prove beneficial. However, in our setting with cancer affecting all age groups, it is imperative that counselling be provided across all ages.

Tessaro *et al.* (1997) and Mouchawar *et al.* (1999) found that women with and without a diagnosis of breast cancer were reported to have poor or limited knowledge of the availability of genetic testing for breast cancer risk, the information provided by testing, and the implications of testing. Bottorff *et al.* (2002) indicate that in clinical settings, knowledge of genetic testing for breast cancer risk and its limitations may reduce the interest in testing in high-risk individuals, although a similar association has not been observed amongst women in the general public.

Henselmans *et al.* (2009) point out that when women are diagnosed with breast cancer, they are confronted with different stressors, such as surgery, the spread of the cancer, side effects and recurrence, throughout the course of the illness. It is important that in the face of these stressors, women are able to exercise personal control, which is the belief that life is not ruled by fate but that a person is able to influence the important events or situations in their life (Henselmans *et al.*, 2009). This belief is related to a variety of positive outcomes such as a lower risk of disease (Bosma *et al.*, 2005), successfully adjusting to the illness (Helgeson *et al.*, 2004) and survival (Surtees *et al.*, 2006).

The patients' responses in this study show that women have very different needs at the time of diagnosis. Whilst some patients felt more comfortable confiding in a normal general practitioner (GP), others wanted more information and would therefore feel more comfortable talking to a specialist. Furthermore, some patients did not want to be overburdened by medical knowledge and would therefore opt to talk to a layperson.

In addition, from the data gathered from the breast cancer survivors, the researcher noticed that information on genetics, recurrence, prognosis and family risk appeared to be more important to the women with teenage daughters. This emphasised that women would want to know whether their daughters were also at risk of developing breast cancer.

Nationally, there are approximately six genetic services in South Africa. However, these services are not specific to cancer genetics. At the time of the study no genetic services were available specifically for breast cancer and the only genetic service available for KwaZulu-Natal was based in Durban. In terms of referral, once a patient was diagnosed with breast cancer, either at Northdale or Edendale Hospitals, the patients were then referred for treatment to Grey's Hospital, which is a tertiary hospital in Pietermaritzburg. However, even at Grey's hospital no genetic counselling services were available for the patient or her family.

5.2.2.7 Provision of counselling for the patient after surgery

The findings of this study indicate that counselling of women who have undergone a mastectomy is imperative in helping them deal with and accept their change in body configuration. All four patients indicated that counselling either helped or would have helped them to cope better.

Hopwood (2000) noted that women who are at risk of developing breast cancer are likely to differ in the type and amount of information they need, as well as in their preferences for involvement in decision-making processes.

Many doctors and surgeons, however, do not properly think through the needs of breast cancer patients. De Bock *et al.* (2001) found that in 30% of individual consultations,

general practitioners did not follow the advice of the clinical geneticist. The researcher is of the opinion that health professionals need to realise that a diagnosis of breast cancer affects not only the patient, but the family as well. This aspect should be seriously considered, as family will be many of these women's support system through this traumatic period. Women who do not undergo a mastectomy and instead undergo chemotherapy or radiation therapy, depend greatly on the support of their family. If appropriate information and counselling is not offered, it leads to patients' harbouring bitter emotions, as is evident from patients' responses.

The four patients interviewed indicated that prior to being diagnosed with breast cancer, their knowledge about breast cancer was minimal in that they knew it was a cancer that affected females, but were not familiar with the risk factors associated with breast cancer, the advantages and disadvantages of the various therapies, nor did they know about genetic counselling or risk assessment. However, since being diagnosed, they had been self-educated about risk factors and the various therapies for breast cancer, yet only one respondent (25%) indicated that she had later learnt about genetic counselling, which she had sent both her daughters for. All four patients attributed their increase in knowledge to reading books and magazines, talking to other female breast cancer survivors, and through attending support groups. This further proves the need for education and counselling for women diagnosed with breast cancer. The healing process takes a long time and counselling provides the necessary support through this trying time.

5.2.3 Genetic counselling, risk assessment, awareness and education for family members of breast cancer patients

5.2.3.1 Genetic counselling and risk assessment for family members of breast cancer patients

Hopwood (2000) points out that identification of breast cancer predisposing genes has created a demand for personalized risk information in families with a family history of cancer, prompting the development of services to respond to this need, and genetic risk counselling for women with histories of familial breast cancer is widely supported (Hopwood, 2000).

The findings of this study indicate that 40% (n=2) of the female respondents were offered this type of counselling and were not interested. Of the remaining 60% (n=3), 20% (n=1) indicated that although not counselled, genetic counselling would have been of benefit, while 40% (n=2) indicated that they were counselled, with one participant indicating that it helped her understand how breast cancer could affect her.

Brandt *et al.* (2002) and van Asperen *et al.* (2002) stress that the reason many women at increased breast cancer risk apply for genetic counselling and DNA testing is to reduce uncertainty, and the need for information on surveillance and surgery. This means that many counselees expect to receive a clear positive or negative result (Press *et al.*, 2001; Frost *et al.*, 2004). However, Vos *et al.* (2008) emphasised that approximately 90% of the results do not provide certainty and the communicated cancer risks and risk management options remain solely based on family history.

According to Evans *et al.* (1993) and Cull *et al.* (1999), very few women had an accurate view of their chances of developing breast cancer before they underwent genetic risk counselling, whilst the majority of women tended to either over- or underestimate their risks. Furthermore, women in the USA and Canada appeared to overestimate their risk (Lerman *et al.*, 1994; Lerman *et al.*, 1995; Smith *et al.*, 1996) to a greater extent than women in the UK, who were more likely to underestimate their risk (Evans *et al.*, 1993; Cull *et al.*, 1999). Even though genetic risk counselling has been shown to significantly improve the accuracy of risk perception (Evans *et al.*, 1993; Cull *et al.*, 1999; Watson *et al.*, 1999), it has been found that up to 30% of UK women and 66% of US women continue to report exaggerated risks of cancer.

According to Pijpe *et al.* (2009), *BRCA1* and *BRCA2* mutation carriers have a high lifetime risk of developing breast cancer. Clarke *et al.* (2008) found that some participants in their study struggled with their children's decision on genetic testing, whilst others, although recognising the need that their child would ultimately have to be tested, were concerned for the consequences or costs that might ensue if the result was positive. According to Smith *et al.* (2008), information provided by genetic testing is highly beneficial, but may also be the cause of psychological distress or discomfort due to the risk of a serious illness being revealed.

5.2.3.2 Precautionary measures, creating awareness and educating family members on breast cancer

Tryggvadottir *et al.* (2006) suggest that it is possible that the incidence of breast cancer diagnosis according to a woman's age is highly influenced by the amount of awareness she has been subjected to, as well as screening. This would be in keeping with the fact that when a woman is diagnosed with breast cancer, her family becomes more aware of the disease therefore taking further precautionary measures to ensure either prevention or early detection of the disease.

Silberman (2014) states that, in terms of raising awareness, enough is being done. Children from elementary through to high school are aware about breast cancer and are able to explain that particular bracelets they wear are to show their support of breast cancer (Silberman, 2014). In terms of educating family members, Warner *et al.* (2003) points out that education about risk of developing breast cancer is extremely important for women with a family history of breast cancer, as this may assist in reducing their anxiety as well as the avoidance of unnecessarily being transferred to high-risk clinics for further investigations which are not required.

This study found that 60% (n=3) of the female respondents are taking precautionary measures by either following a healthy eating plan or going for regular check-ups. The majority of the respondents (88%; n=7) were of the opinion that enough is being done to create awareness on breast cancer.

5.3 Limitations of the study

Although due diligence was maintained in order to ensure the integrity of the study the following limitations of the study were identified:

5.3.1 Sample size

The sample of this study is very limited with only fifty-six participants and therefore not large enough to generalise across South Africa.

5.3.2 Selection bias

The study was conducted in an urban area, which resulted in mainly White and Indian respondents. The non-recruitment of Coloured and limited participation of Black females in the study is considered a limitation. All respondents were literate with at minimum, a Grade 10 pass. The respondents were therefore literate and able to understand and respond to questions. Respondents from the general public were purposely selected to ensure that responses to questionnaires will be easily obtained and participants could be easily followed-up. In addition, the involvement of respondents from the CANSA support group only, for breast cancer survivor respondents was also limiting in terms of racial representivity.

5.3.3 Information bias

The richness of the information collected and analysed was limited by the design of the questionnaire; non-explanation to participants about the concepts of genetic counselling and risk assessment. This may have been one of the reasons many women from the general public indicated their disinterest or uncertainty in attending a risk assessment session.

5.3.4 Age of data

Data for this study was collected more than 10 years ago. During this time, advances in medical and social contexts made, would result in the findings of a similar study today, differing to the findings of the current study.

5.4 Summary

This chapter discussed the key findings of the study in relationship to international studies. A comparison with the literature reviewed indicated that while significant effort has been placed on creating awareness on breast cancer, there is a deficiency in terms of educating family members about risks of breast cancer as well as genetic counselling and risk assessment for women at high risk. This lack of knowledge has a negative impact on the use of these services and ultimately impacts on early detection. Without early detection and appropriate management of breast cancer, mortality due to breast cancer will continue to rise.

CHAPTER VI: CONCLUSIONS AND RECOMMENDATIONS

6.1 Introduction

This chapter summarises the study and makes recommendations to strengthen the uptake for genetic counselling and assessment for breast cancer in women of all races in South Africa and for further research.

6.2 Conclusion

Strengthening the health care system through increasing the knowledge of health professionals on genetic counselling and risk assessment will contribute to the uptake of these services. Presently, neither breast cancer patients and their families, nor women from the general public are aware of the availability of such services.

This study has found that only 25% of family members went for genetic counselling, which has resulted in them taking the necessary precautionary measures and understanding their risks involved in the development of breast cancer. A further 25% of respondents indicated that although they were never offered this type of counselling, they would have considered this beneficial in further understanding their risks in developing breast cancer, as well as obtaining a better understanding of what breast cancer is.

In addition, the lack of educating patients and their families on the availability of genetic counselling has negatively impacted many patients and families, as they were not provided with the opportunity to attend genetic counselling sessions. This has impacted on the uptake of genetic counselling services within the families of breast cancer patients.

6.3 Recommendations

Based on the findings of the study, the researcher makes the following recommendations structured according to the Health Belief Model.

6.3.1 Modifying factors

a. Knowledge

Education and awareness raising programmes on the importance of breast cancer screening and risk assessments for women at high risk should be made available. This will provide women with the appropriate knowledge and information to make the necessary decisions for prevention, early detection, treatment and survival.

b. Race and socioeconomic factors

Due to the cultural diversity of the South African population, education and awareness raising programmes should be culturally appropriate. Furthermore, the level of literacy varies drastically and this should be taken into consideration when these programmes are developed so as to ensure all women in South Africa are provided with the same information, relevant to their level of education and understanding.

6.3.2 Cues to action

a. Policy

An integrated strategy for the early detection and treatment of breast cancer should be developed. The strategy must focus on education about breast cancer, strengthening methods for early detection and treatment, genetic counselling and testing, as well as psychosocial support for both patients and families.

Strengthening the provision of genetic counselling and risk assessment services to high-risk breast cancer survivors and their female family members is essential. This will enable women to understand their risks and to be identified in the early stages of breast cancer. Early identification and treatment will decrease the mortality rate due to breast cancer in South Africa.

It is important that not only survivors but health professionals also understand its importance. Hence, enhanced information should be provided to health professionals about the benefits associated with genetic counselling and risk assessments.

Family members should also be counselled to help them cope emotionally and mentally with the diagnosis of the patient and to also understand how they can provide the patient with the necessary support throughout the treatment process.

Health care providers must take the opportunity at facilities to conduct health talks that are both culturally and linguistically appropriate, to patients and explain what breast self-examination is and when and how it should be conducted. In addition, health care providers must explain the services available for detection of breast cancer and what this service entails. If genetic counselling services are offered by a clinic, women at risk should be afforded the opportunity to attend the genetic unit to further understand what it is about.

A multi-sectoral team should be available to offer every patient the opportunity to undergo counselling, whether for herself or her family. It should be the responsibility of the doctor who diagnoses her breast cancer to provide her with this information. However, this information about counselling should be suggested and not imposed upon the patient. Equipping the patient with relevant information at the time of diagnosis enables her to be knowledgeable about the services available to her and her family. Ultimately though, it must be the decision of the patient as to whether she or her family should attend counselling and at a time that she considers appropriate.

Patients should speak to a genetic counsellor, psychologist, medical doctor, medical nurse, social worker, or any other person they would feel most comfortable with.

There is an urgent need for improved training of health professionals on breast cancer epidemiology, risk factors and psycho-social counselling of patients.

6.3.3 Likelihood of action

a. Perceived benefits without perceived barrier

Female family members of breast cancer patients should be educated about their increased risk and advised about screening and risk assessment services they should utilize. Information must be provided as clearly as possible to assist with decisions on screening and risk assessment.

A diagnosis of breast cancer has a negative impact not only on the patient, but on her entire family. Allowing a patient's family to be involved, with the consent of the patient, from the moment of diagnosis, would, in most cases, create a supportive environment for the patient. This would lead to the patient being able to share her fears and frustrations with her family instead of having to deal with the situation on her own. Moreover, this could have a positive impact on the patient's well-being, as a supportive and caring environment will allow her to cope better with her diagnosis, as well as speed up the recovery process.

Every patient should be offered the opportunity to undergo counselling, whether for herself or her family. It should be the responsibility of the doctor who diagnoses her breast cancer to provide her with this information. However, this information about counselling should be suggested and not imposed. By equipping the patient with relevant information at the time of diagnosis, enables the patient to be knowledgeable about the services available to her and her family. Ultimately though, it must be the patient's decision on whether she or her family should attend counselling and at a time that she considers appropriate.

6.3.4 Other recommendations

Education programmes targeting men should be developed to encourage men to screen for breast cancer and make them aware of the fact that breast cancer does not only affect women. These education programmes should include the positive medical experiences men have had and how early detection contributes to saving a life.

Further research should be conducted on the following topics:

- An investigation in the South African context on ways in which to overcome the barriers to genetic counselling and risk assessment to increase the use of genetic services
- An exploration of men's perceptions of breast cancer and its detection
- Investigation of culturally appropriate strategies to overcome barriers to the early detection and treatment of breast cancer
- Health care professionals' perceptions of the role and involvement of family members in breast cancer patient support and rehabilitation

6.4 Summary

Genetic counselling and testing is not well-known amongst the general population, breast cancer survivors or their family members. In order to ensure genetic counselling and risk assessment services are strengthened and utilized, education and awareness is required. Education by health care professionals, provision of information, education and communication material on breast cancer and breast cancer services will further contribute to an increase in the number of people utilizing the service, early detection of the disease, as well as a reduction in mortality due to breast cancer.

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ANNEXURES

Annexure 1: Self-administered questionnaire for breast cancer patients

SELF-ADMINISTERED QUESTIONNAIRE FOR BREAST CANCER PATIENTS

1. What is your age?

_____

2. Please indicate your race:

- ☐ Black
- ☐ White
- ☐ Indian
- ☐ Coloured

3. Is your ethnicity mostly Jewish?

- ☐ Yes
- ☐ No

4. What is your height and weight?

_____

5. Is your ethnicity mostly Jewish?

- ☐ Yes
- ☐ No

6. Do you usually drink one or more servings of alcohol per day?

- ☐ Yes
- ☐ No

7. Do you eat three or more servings of vegetables per day?

(One serving is about one cup of raw leafy greens or half a cup of other vegetables, raw or cooked).

- ☐ Yes
- ☐ No

8. Were you exposed to significant radiation in the past?

- ☐ Yes
- ☐ No

9. How old were you when you first menstruated?

- ☐ Younger than 15
- ☐ Older than 15

10. Are you currently taking birth control pills?

- ☐ Yes
- ☐ No

11. How many children have you given birth to?

- [ ] None
- [ ] One
- [ ] Two or more

12. What was your age at first live birth?

- [ ] Unknown
- [ ] No births
- [ ] <20
- [ ] 20 to 24
- [ ] 25 to 30
- [ ] >30

13. Are you menopausal?

- [ ] Yes
- [ ] No

14. Have you ever had a hysterectomy (removal of the uterus)?

(Having a hysterectomy does not increase your risk of getting breast cancer).

- [ ] Yes
- [ ] No

15. Do you examine your breasts monthly?

- [ ] Yes
- [ ] No

16. Have you ever had any type of cancer, except for non-melanoma skin cancer?

- [ ] Yes
- [ ] No

17. Have you ever conducted any of the below-listed examinations?

- [ ] Breast self-examination
- [ ] Clinical breast examination
- [ ] Both

18. About yourself. What about you?

- [ ] Never had breast disease
- [ ] Have had previous lumps or cysts
- [ ] Have had previous breast cancer

19. Has your sister ever had breast cancer?

- [ ] Yes
- [ ] No

20. Has you mother ever had breast cancer?

- ☐ Yes
- ☐ No

21. Have you ever had a breast biopsy?

- ☐ Yes
- ☐ No

22. How many breast biopsies, positive or negative, have you had?

- ☐ 1
- ☐ >1

23. Have you had at least one breast biopsy with atypical hyperplasia (a precancerous condition)?

- ☐ Yes
- ☐ No
- ☐ Unknown

24. Before being diagnosed with breast cancer, were you ever advised about the potential risk factors for breast cancer?

- ☐ Yes
- ☐ No

If yes, by whom were you advised?

_____

25. Before being diagnosed, did you have any of these signs or symptoms? Please tick the appropriate block.

- ☐ A lump or thickening in the breast or armpit
- ☐ Changes in the skin – dimpling, puckering or redness
- ☐ Changes in the nipple – direction of the nipple or an unusual discharge
- ☐ Changes around the nipple – unusual rash or sore area
- ☐ Changes in the size or shape of the breast

26. Do you take oestrogen-containing oral contraceptives?

- ☐ Yes
- ☐ No

27. Were you given the option to have a breast cancer risk-assessment counselling session?

(In this type of counseling session patients learn more about their cancer risks, screening options, and gene testing for hereditary cancer).

- ☐ Yes
- ☐ No

If yes, please answer the following:

(i) Who, or what agency, did the assessment?

_____

(ii) What formula was used to calculate the risk?

_____

_____

_____

28. If you answered no to the above question, would you be interested in attending a breast cancer risk-assessment session, if not for yourself, then for family and friends?

- ☐ Yes
- ☐ No

AFTER DIAGNOSIS

1. What method of detection was used to diagnose you as a breast cancer patient?

- ☐ Mammography
- ☐ Biopsy
- ☐ Clinical breast examination
- ☐ Breast self examination

2. At which stage of breast cancer were you when you were first diagnosed?

- ☐ Stage 0
- ☐ Stage 1
- ☐ Stage 2
- ☐ Stage 3
- ☐ Stage 4
- ☐ Recurrent
- ☐ Unknown

3. At what age were you first diagnosed with breast cancer?

_____

4. After being diagnosed with breast cancer, what type of therapy were you given? Tick the appropriate box(es)

- ☐ Chemotherapy
- ☐ Radiation Therapy
- ☐ Surgery

5. Were you advised about the advantages and disadvantages of the different types of therapy?

- ☐ Yes
- ☐ No

6. If you were given either chemotherapy and/or radiation therapy, please state what the side effects of either treatment was.

Chemotherapy:

_____

Radiation Therapy:

_____

7. If you opted for surgery, what type of surgery did you undergo?

☐ ***Total Mastectomy:***

an operation to remove the breast or as much of the breast as possible; some lymph nodes under the arm are also removed.

☐ ***Breast Conserving Therapy or Lumpectomy:***

an operation to remove the cancer and not the breast

☐ ***Modified Radical Mastectomy:***

removal of the whole breast, most of the lymph nodes under the arm and often the lining over the chest muscles.

☐ ***Radical Mastectomy:***

removal of the breast, both chest muscles, all of the lymph nodes under the arm, and some additional fat and skin.

8. Only answer if you have undergone breast surgery

After having had breast surgery, how often are you required to go for check-ups?

---

9. After being diagnosed with breast cancer, is there any specific diet you have been encouraged to follow?

☐ Yes

☐ No

10. How many times a week do you eat red meat?

☐ None

☐ 1 to 3

☐ 4 or more

11. After being diagnosed with breast cancer, is there a specific physical/exercise programme you were encouraged to follow?

- ☐ Yes
- ☐ No

12. How often do you exercise aerobically (e.g. brisk walking or jogging)?

- ☐ Never
- ☐ Less than once per week
- ☐ Once or twice per week
- ☐ At least three to four times per week

13. How long do you exercise for each time?

- ☐ None to 15 minutes
- ☐ 16 to 30 minutes
- ☐ More than 30 minutes

14. Are you taking Tamoxifen?

(Tamoxifen is a medicine that can reduce the risk of developing breast cancer in high risk women. Its benefit to normal risk women is unknown).

---

15. Within the past year have you been examined by a physician and have had a negative mammogram?

- ☐ Yes
- ☐ No

16. After being diagnosed with breast cancer, have you ever been advised about factors which reduce the risk of developing breast cancer? (Factors such as limiting the use of hormone replacement therapy, avoiding obesity, staying physically active).

	Yes
	No

17. Were you given the opportunity to speak to someone who would explain breast cancer? Tick the appropriate box.

	Genetic counselor
	Psychologist
	Medical Doctor
	Medical Nurse
	Social worker
	Dietician
	Other

18. What, in your opinion, were some of the risk factors that affected you?

Risk factors	
Alcohol	
Smoking	
Radiation	
Early menstruation (<15 years of age)	
Early live birth (<24 years of age)	
Late live birth (>25 years of age)	
Oestrogen contraceptives	
Family history	

19. What do you think was the cause of your cancer?

_____

20. Do you know of any causes of cancer in general?

_____

21. Is having the option of reconstructive surgery important to you?

_____

22. Have you had reconstructive surgery?

	Yes
	No

23. If you have undergone reconstructive surgery, was it successful?

☐	Yes
☐	No

24. If you answered no to the above, what is/was the reason?

_____

_____

_____

_____

_____

25. Was your family given the opportunity to undergo counselling?

☐	Yes
☐	No

THANK YOU FOR THE TIME TAKEN TO COMPLETE THIS QUESTIONNAIRE

Annexure 2: Self-administered questionnaire for women from the general public

SELF-ADMINISTERED QUESTIONNAIRE FOR WOMEN FROM THE GENERAL PUBLIC

1. What is your age?

_____

2. Please indicate your race:

- ☐ Black
- ☐ White
- ☐ Indian
- ☐ Coloured

3. Is your ethnicity mostly Jewish?

- ☐ Yes
- ☐ No

4. What is your height and weight?

_____

5. Do you smoke?

- ☐ Yes
- ☐ No

6. Do you usually drink one or more servings of alcohol per day?

- ☐ Yes
- ☐ No

7. Do you eat three or more servings of vegetables per day?

(One serving is about one cup of raw leafy greens or half a cup of other vegetables, raw or cooked).

- ☐ Yes
- ☐ No

8. Were you exposed to significant radiation in the past?

- ☐ Yes
- ☐ No

9. How old were you when you first menstruated?

- ☐ Younger than 15
- ☐ Older than 15

10. Are you currently taking birth control pills?

- ☐ Yes
- ☐ No

11. How many children have you given birth to?

- [ ] None
- [ ] One
- [ ] Two or more

12. What was your age at first live birth?

- [ ] Unknown
- [ ] No births
- [ ] <20
- [ ] 20 to 24
- [ ] 25 to 30
- [ ] >30

13. Do you examine your breasts monthly?

- [ ] Yes
- [ ] No

14. Have you ever had any type of cancer, except for non-melanoma skin cancer?

- [ ] Yes
- [ ] No

15. What, in your opinion, are some of the risk factors for breast cancer that affects you?

Risk factors	
Alcohol	
Smoking	
Radiation	
Early menstruation (<15 years of age)	
Early live birth (<24 years of age)	
Late live birth (>25 years of age)	
Oestrogen contraceptives	
Family history	

16. Have you ever conducted any of the below-listed examinations?

- ☐ Breast self-examination
- ☐ Clinical breast examination
- ☐ Both

17. About yourself. What about you?

- ☐ Never had breast disease
- ☐ Have had previous lumps or cysts
- ☐ Have had previous breast cancer

18. Has your sister ever had breast cancer?

- ☐ Yes
- ☐ No

19. Has you mother ever had breast cancer?

- ☐ Yes
- ☐ No

20. Have you ever had a breast biopsy?

- ☐ Yes
- ☐ No

21. How many breast biopsies, positive or negative, have you had?

- ☐ 1
- ☐ >1

22. Have you had at least one breast biopsy with atypical hyperplasia (a precancerous condition)?

- ☐ Yes
- ☐ No
- ☐ Unknown

Only answer questions 23 and 24 if you have been diagnosed with breast cancer

23. Before being diagnosed with breast cancer, were you ever advised about the potential risk factors for breast cancer?

- ☐ Yes
- ☐ No

If yes, by whom were you advised?

_____

24. Before being diagnosed, did you have any of these signs or symptoms? Please tick the appropriate block.

☐	A lump or thickening in the breast or armpit
☐	Changes in the skin – dimpling, puckering or redness
☐	Changes in the nipple – direction of the nipple or an unusual discharge
☐	Changes around the nipple – unusual rash or sore area
☐	Changes in the size or shape of the breast

25. Do you take oestrogen-containing oral contraceptives?

☐	Yes
☐	No

26. If given the opportunity, would you be interested in consulting with a genetic counsellor to assess your risk of developing breast cancer? Any other reasons

_____

_____

_____

THANK YOU FOR THE TIME TAKEN TO COMPLETE THIS QUESTIONNAIRE

Annexure 3: Semi-structured interview with breast cancer patients

SEMI-STRUCTURED INTERVIEW WITH BREAST CANCER PATIENTS

Interview for breast cancer patients

The following questions were asked to breast cancer patients under the headings of:

- ✓ pre-interview orientation,
- ✓ period until diagnosis,
- ✓ period surrounding the mastectomy,
- ✓ after mastectomy, and
- ✓ family/friends attitudes

A. Pre-interview orientation

1. At the beginning of the interview the patient should state her name, age, race, and age at first diagnosis (AAFD).

Name:
Age:
Race:
AAFD:

2. Have any of your family members been diagnosed with cancer of any kind?

Yes	No

3. If yes, what type of cancer?

_____

4. What is their relationship to you?

Family member's name	Relationship to you	Type of cancer

B. Period until diagnosis

1. What were your earliest signs/symptoms that prompted you to see a doctor?

2. How long did you wait from this point to seeing a doctor? (Time response)

3. How did you feel when you were first informed that you have breast cancer?

_____
_____
_____
_____
_____

4. What method of detection was used to diagnose your breast cancer?

_____
_____

5. What information did your doctor provide you with after diagnosis?

_____
_____
_____
_____
_____

Description of disease	Yes	No
Prognosis	Yes	No
Chance of reoccurrence	Yes	No
How it may affect family members	Yes	No
Consult with a genetic counsellor	Yes	No

Other: _____
_____

6. How did you feel about the information?

_____
_____
_____
_____
_____

7. Were you happy with the information?

Yes	No

8. Did you understand the information that was given to you?

Yes	No

9. In your opinion, was the information that was given at that time enough?

Yes	No

10.1 If counsellor was suggested:

10.1.1 Did you visit a counsellor?

Yes	No

10.1.2 What type of counsellor did you visit?

10.1.3 What type of information did you obtain from the counsellor?

_____
_____
_____
_____

10.1.4 Would you have liked to be counselled about the genetics and risks involved with breast cancer?

Yes	No

10.2 If no counselling was suggested:

10.2.1 Would you have liked to visit a counsellor?

Yes	No

10.2.2 What type of counsellor would you have liked to visit?

10.2.3 Would you have liked to be counselled about the genetics and risks involved with breast cancer?

Yes	No

11. Would you have liked to know more about the disease, breast cancer, such as the genetics, the reoccurrence, the prognosis and your family's risk of getting breast cancer?

Genetics	Yes	No
Reoccurrence	Yes	No
Prognosis	Yes	No
Family Risk	Yes	No

C. Period surrounding the mastectomy

1. How long after diagnosis did you have a mastectomy? (Time response)

2. Did you receive counselling regarding a mastectomy before the mastectomy was performed?

Yes	No

3.1 If yes

3.1.1 What type of counsellor did you visit?

3.1.2 What type of information did you obtain from the counsellor?

_____
_____
_____
_____

3.1.3 Would you have liked to know more?

Yes	No

3.1.4 If yes, what additional information would you have liked?

_____
_____
_____
_____

3.2 If no counselling was received before your mastectomy:

3.2.1 Would you have liked to visit a counsellor?

Yes	No

3.2.2 What type of counsellor would you have liked to visit?

3.2.3 What type of information would you have liked to obtain?

_____
_____
_____
_____

D. After mastectomy

1. How long has it been since you've had a mastectomy?
_____

2. How did you feel immediately after the mastectomy, when you came to your senses?
_____
_____
_____
_____

3. How do you feel now? (A while after the mastectomy)
_____
_____
_____
_____

4. Explain your emotions after realizing that you had undergone a mastectomy.
_____
_____
_____
_____

5. Did you receive counselling after your mastectomy?

Yes	No

6.1 If yes:

6.1.1 What type of counsellor did you visit?

6.1.2 Was the counselling of any help emotionally?

Yes	No

Explain:_____
_____
_____
_____

6.1.3 Would you have liked further counselling?

Yes	No

6.2 If no counselling was received:

6.2.1 Would you have liked to visit a counsellor?

Yes	No

6.2.2 Do you think a counsellor would have helped you cope better with your mastectomy?

Yes	No

7. Did you accept the change in your body's configuration immediately?

Yes	No

8. If not, have you now accepted it?

Yes	No

9. If yes, how long did it take you to accept it? (Time response)

10. Do you have a change of feelings now, about the information provided at diagnosis?

Yes	No

11. If yes, what influenced the change in your feelings?

_____
_____
_____
_____

12. Do you now feel the information you were given at your first diagnosis was sufficient?

Yes	No

13. If not, what other information do you feel should have been given to you to help you cope emotionally and mentally?

_____
_____
_____
_____

14. Do you now feel you are wiser than when you were first diagnosed with breast cancer?

Yes	No

15. If yes, what has helped you become wiser?

Counselling	Yes	No
Support Groups	Yes	No
Reading	Yes	No

Other:_____
_____

E. Family/friends attitudes

1. Did you inform family members and friends of your diagnosis?

Yes	No

2. If yes, when did you inform them? (Time response)

3. From your perspective, how did the knowledge of your diagnosis affect them?

_____
_____
_____
_____

4. How did your spouse feel about your change in body configuration?

_____
_____
_____
_____

5. How did your family feel about your physical change?

_____
_____
_____
_____

6. Were any of your family members given the opportunity to be counselled?

Yes	No

7. If yes:
   7.1 What type of counsellor did you visit?

   7.2 Was the counselling of any help?

Yes	No

   7.3 Was your family counselled on the genetics of the disease and the risks to them as family members?

Yes	No

   7.4 If not, do you think such information is important and relevant?

Yes	No

8. If no counselling was obtained:
   8.1 Would you have liked to visit a counsellor?

Yes	No

   8.2 Do you think it is important for family members to be counselled?

Yes	No

8.3 What type of counsellor would you have liked to visit?

8.4 Do you think a counsellor would have helped your family cope better with your breast cancer?

Yes	No

8.5 What type of information would you have liked the counsellor to give your family during counselling?

_____
_____
_____
_____

9. Was any risk calculations made for family members?

Yes	No

10. Would you be interested in knowing the risks of your family members to breast cancer?

Yes	No

11. Do you think enough is being done about:

11.1 Creating an awareness of breast cancer?

Yes	No

11.2 Educating people as to what exactly breast cancer is?

Yes	No

Annexure 4: Semi-structured interview with family members of breast cancer patients

SEMI-STRUCTURED INTERVIEW WITH FAMILY MEMBERS OF BREAST CANCER PATIENTS

At the beginning of the interview family members should state their names, age, sex, and relationship to patient.

Name	Age	Sex	Relationship to patient

1. What is your understanding of breast cancer and how do you view it?

_____
_____
_____
_____
_____
_____
_____
_____

2. Did the patient mention any suspicions before diagnosis?

Yes No

3. If yes, what made her suspicious?

_____
_____
_____
_____

4. When were you informed about her diagnosis? (Immediately/weeks/months/etc)

_____

5. At the time of diagnosis, did you have any understanding of what cancer was?

Yes	No

6. After the diagnosis, did you gather more information about breast cancer?

Yes	No

7. If yes, how did you get it and what is it?

_____
_____
_____
_____

8. Do you feel that you now have a better understanding and knowledge about breast cancer, or, would you like to learn more? (If they would like to learn more) What more would you like to learn about?

_____
_____
_____
_____

9. Were you offered any counselling to help you cope with your family member's diagnosis and mastectomy?

Yes	No

10.1 If yes:

10.1.1 What type of counsellor did you visit?

10.1.2 Was the counselling of any help?

Yes	No

10.1.3 What type of information did you obtain from the counsellor?

_____
_____
_____
_____

10.1.4 Were you counselled about the genetics and risks involved with breast cancer?

Yes	No

10.1.5 If not, would you have liked to be counselled on the genetics and risks involved with breast cancer?

Yes	No

10.2 If no counselling was obtained:

10.2.1 Would you have liked to be counselled as a family?

Yes	No

10.2.2 What type of counsellor would you have liked to visit?

10.2.3 What type of information would you have liked?

_____
_____
_____
_____

10.2.4 Would you have liked to be counselled on the genetics and risks of breast cancer?

Yes	No

11. Would you like to undergo a breast cancer risk assessment should you be given the opportunity?

Yes	No

12. Do you fear getting the disease?

_____

13. What do you fear the most?

_____
_____
_____
_____

14. As a female offspring of a woman with breast cancer, what precautionary measures have you undertaken to ensure you do not fall victim to breast cancer?

_____
_____
_____
_____

15. Do you think enough is being done about:

15.1 Creating an awareness of breast cancer?

Yes	No

15.2 Educating family members of breast cancer patients as to what exactly breast cancer is?

Yes	No