Breast cancer patients' experience of receiving pathogenic germline genetic results: A single centre experience in Oman.

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DECLARATION

I hereby declare that the work submitted here is the result of my own independent investigation. Where help was sought, it was acknowledged. I further declare that this work is submitted for the first time at this university towards a Magister degree in Genetic Counselling and that it has never been submitted to any other university for the purpose of obtaining a degree.

Signed by candidate

Signature

Date: 17 February 2025

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LIST OF ABBREVIATIONS

AYA	Adolescent and Young Adult
BRCA1	Breast Cancer 1
BRCA2	Breast Cancer 2
CT	Computed Tomography
DCU	Day Care Unite
EDTA	Ethylenediaminetetraacetic acid
GC	Genetic Counselling
GCC	Gulf Cooperation Council
HBOC	Hereditary Breast and Ovarian Cancer
HIS	Hospital Information System
IDC	Invasive Ductal Carcinoma
ILC	Invasive Lobular Carcinoma
LPV	Likely Pathogenic Variant
MDT	Multidisciplinary Team
MENA	Middle East and North Africa
MGT	Molecular Genetic Test
MHSO	Ministry of Health Sultanate of Oman
MLPA	Multiplex ligation-dependent probe amplification
MMedSc	Master in Medical Science
NCCN	National Comprehensive Cancer Network
OPD	Out Patient Department
PV	Pathogenic Variant
SQCCCRC	Sultan Qaboos Comprehensive Cancer Care and Research Centre
SQUH	Sultan Qaboos University Hospital
SSI	Semi-Structured Interview
TDM	Treatment Decision Making
UCT	University of Cape Town
UK	United Kingdom
USA	United States of America

ABSTRACT

Introduction: In Oman, little is known about breast cancer patients' experiences of receiving pathogenic germline genetic results. Receiving a positive germline genetic result can have a wide range of emotional and practical effects on breast cancer patients. This study explored the lived experiences of Omani women navigating a positive pathogenic germline genetic test result for hereditary breast cancer. Driven by the increasing prevalence of genetic testing and limited research on its psychosocial impact within this specific cultural context, the study aimed to understand how Omani women experience and make sense of a positive genetic test result.

Methods: This is a qualitative study based on an interpretive phenomenological approach. Semi-structured interviews were conducted with nine Omani women who had received a positive germline genetic result for breast cancer predisposition gene. Thematic analysis was employed to identify key themes emerging from these women's experiences.

Result and Discussion: Four themes emerged from the thematic analysis in this study. These included "Cancer journey", "Genetic testing motivation and expectations", "Receiving the positive result", and "Adapting to hereditary breast cancer diagnosis". The findings revealed the complex and multifaceted experience of breast cancer women with receiving positive germline genetic result. The perceived causation of their breast cancer, such as stress and pre-existing cultural beliefs, as well as their lived experiences during the cancer diagnosis and treatment, all influenced these women's response and understanding of their genetic test result. Emotional responses varied, ranging from anxiety and fear to relief and empowerment, highlighting the individual nature of this experience. Coping strategies were active coping such as leaving the matter to God's hand, increased surveillance and risk-reducing surgeries, engagement coping with family and friends for support, and meaning-focused coping, often grounded in religious and spiritual beliefs. Family dynamics and cultural norms played a crucial role in disclosure practices, with concerns about protecting family members from psychological stress having influenced their decisions about information sharing.

Conclusion: This research contributes valuable insights into the lived experiences of Omani women with hereditary breast cancer, highlighting the need for culturally sensitive and individualised support throughout the testing and decision-making process. The findings have implications for healthcare professionals, genetic counsellors, and policymakers, emphasizing the importance of providing comprehensive support that addresses the emotional, social, and cultural dimensions of hereditary cancer risk.

CHAPTER 1: LITERATURE OVERVIEW

International studies have shown that patients' responses to receiving positive genetic test results vary greatly, influenced by multiple factors. Some individuals experience significant psychological distress, including anxiety, depression and fear, upon receiving their results (Hartz et al., 2014). In contrast, other individuals demonstrate resilience and adaptation, viewing the information as empowering and motivating them to take proactive steps to manage their health risks (Francke et al., 2013). Therefore, understanding patients' experiences and influencing factors enables genetic professionals to provide tailored services and support throughout genetic counselling (GC) This allows them to address diverse responses, from significant emotional distress to resilience and adaptation. By recognising these factors, genetic professionals can offer personalised care and resources to help patients cope with their genetic findings and manage their health risks.

Given the limited understanding of how Omani breast cancer patients experience receiving pathogenic germline genetic results, this study aims to explore this topic among these women. This first chapter will present background literature on the subject and provide information on the local genetic counselling and testing services available to breast cancer patients at the Sultan Qaboos Comprehensive Cancer Care and Research Centre (SQCCCRC).

1.1 LITERATURE OVERVIEW

1.1.1 Inherited Breast Cancer

Cancer is a very common disease worldwide with more than 19.3 million new cases reported in 2022 (Chhikara, 2022). In Oman, 2307 cancer cases were reported in 2019 (Ministry of Health, Sultanate of Oman [MHSO], 2022). Of all cancers worldwide, breast cancer, at 25%, is reported to be the most prevalent cancer in females (Al-Azri et al., 2021). The incidence of breast cancer is increasing nationally and internationally with the incidence of breast cancer in the Arabian Gulf countries reported to account for 13%-35% of all cancer cases (Al-Azri et al., 2021). Although there are many known risk factors for breast cancer, including hormonal and lifestyle variables (multifactorial type), resulting from a combination of genetic and environmental influences (Pyeritz, Korf & Grody, 2019,p.323) Inheritance, due to germline pathogenic or likely pathogenic variation in high and moderate risk genes, could significantly increase the lifetime risk of developing breast cancer (Ngeow & Eng, 2014). The likelihood of developing breast cancer increases with age and the number of close relatives affected (Wendt & Margolin, 2019). Since the identification of the Breast Cancer 1 (*BRCA1*) and Breast Cancer 2 (*BRCA2*) genes in 1994 to 1995, many more genes have been identified. *BRCA1/2*, however, remain the most common cause, accounting for 20-30% of hereditary breast cancer cases (Pyeritz, Korf & Grody, 2019).

Breast cancer susceptibility genes and risk alleles are generally grouped into three risk groups, namely high, moderate and low penetrant genes. Several genes have been identified in each category. This study focused on women who had pathogenic and likely pathogenic variants on high and moderate risk genes.

- High-risk genes: Approximately 5 to 10% of breast cancer cases are believed to be caused by a pathogenic or likely pathogenic variant in a highly penetrant cancer predisposition gene (Allain, 2008). These genes are *BRCA1/2, CDH1, NF1, PTEN, TP53 and STK11* (Frey et al., 2017) with a breast cancer relative risk of greater than 4 (Wendt & Margolin, 2019).

- Moderate-risk variations: Carriers of pathogenic or likely pathogenic variants in these genes have an increased relative risk of 2-4 fold (Wendt & Margolin, 2019). Examples of these include *CHEK2, ATM, PALB2, MRN* complex (*RAD50, MRE11, NBN/NBS1)* (Frey et al., 2017).

- Low-risk variations: Causes a 1.5-fold increased risk of breast cancer (Wendt & Margolin, 2019). However, this group was not part of this study.

Genetic testing can reveal inherited mutations or variations in genes that increase an individual's risk of developing certain types of cancer, such as breast cancer. By understanding a patient's genetic profile, healthcare providers can better assess their cancer risk, recommend appropriate screening and prevention strategies, and provide more tailored and personalised care. Even though the high penetrance genes have the most clinical implications, they only account for a small number of inherited cancer cases. Specific cancer patterns and/or family clusters are likely due to moderate and low penetrance genes (Ngeow & Eng, 2014). Most of the high penetrance breast cancer genes, such as *BRCA1* and *BRCA2*, are associated with well-described genetic syndromes like Hereditary Breast and Ovarian Cancer syndrome (HBOC). The latter is characterised by an increased risk of breast, ovarian, prostate, and pancreatic cancers. Other high-risk genes like *TP53*, *PTEN*, and *CDH1* are associated with Li-Fraumeni syndrome, Cowden syndrome, and Hereditary Diffuse Gastric Cancer syndrome, respectively. Identifying these high-risk individuals and their families is essential for providing appropriate cancer screening, prevention strategies, and management of other associated cancer risks (Daly et al., 2021). Therefore, identifying these individuals and their families and providing them with genetic counselling and testing is essential for major decision-making, such as early screening and prophylactic surgeries (Beggs & Hodgson, 2009). According to the National Comprehensive Cancer Network (NCCN), moderate-risk gene carriers should be under regular surveillance. Currently, there is insufficient evidence to recommend prophylactic surgeries for them, but this option can be considered in the context of personal and family history (Daly et al., 2017). The clinical implications of low-penetrance genes are not yet fully understood. Nevertheless, research suggests that their effects can be cumulative and if an individual carries multiple copies of these low-risk alleles, and they may face a significantly increased risk of breast cancer. Polygenic risk scores, which is the combined effects of many low-risk genetic variants to estimate an individual's predisposition to breast cancer, can provide valuable understanding into an individual's susceptibility to breast cancer. However, these scores are not yet commonly used in clinical practice (Zhang et al., 2020).

1.1.2. Motivations and Barriers in Pursuing Genetic Counselling

Genetic counselling plays a crucial role in treating patients and their families holistically by addressing not only the genetic aspects of their condition but also considering their emotional, psychological, and social well-being. These sessions provide an opportunity for patients to understand their genetic test results, the potential risks for themselves and their family members, and the implications for future health management (Uhlmann, Schuette & Yashar, 2009, p.7). In addition, genetic counselling fosters a sense of trust and open communication between the patient and healthcare providers. By integrating both medical and emotional support, the session ensures that patients feel heard and understood, creating a space for patients to be completely honest and often for the first time voice things that they had not had the chance to do before (Uhlmann, Schuette & Yashar, 2009, p.10-11). Cancer patients who are referred for genetic counselling are counselled about hereditary cancers, inheritance patterns, cancer risks associated with these syndromes, available genetic testing options, and their implications. Cancer risk counselling is defined as "a communication process regarding an individual's possible increased risk of developing specific forms of cancer, which includes obtaining detailed family, medical, and lifestyle histories, documenting cancer-related diagnoses, constructing and analysing pedigrees, conducting risk assessment and counselling, and discussing options for early detection and prevention" (Coleman & Tsongalis, 2002:98).

A study by Morand et al. (2022) examined the motivations of adolescent and young adult cancer patients (ages 18-39) in the USA who sought genetic counselling and testing. Key motivations included understanding cancer risk, informing health decisions, benefiting family and gaining control. Barriers included logistical challenges, emotional concerns, and a perceived lack of immediate benefits. Interest in genetic counselling was also influenced by the time since diagnosis, with those further from their diagnosis showing more interest.

A study by Scott et al. (2020) in the USA found that breast cancer survivors with a high genetic risk are encouraged to pursue genetic testing by factors such as awareness of relatives' cancer risk, clinician recommendations and potential treatment implications. However, concerns about insurance, testing costs, and discrimination can deter them. In Australia, Quinlivan, Battikhi and Petersen, (2014) found that women knowledgeable about genetics and with positive views toward testing were more likely to agree to hypothetical testing. Previous experience with genetic testing also increased acceptance, while concerns about discrimination lowered it. Interestingly, factors like age, education and family cancer history did not significantly impact willingness to undergo testing.

1.1.3 Cancer Journey

Genetic testing and receiving a positive germline genetic result are parts of the cancer journey. Women frequently discussed their diagnostic journey and coping strategies leading up to genetic testing and the receipt of their positive genetic result. A study by Brédart et al.,(2013) explored the short-term emotional responses of women with breast cancer to *BRCA1/2* testing, considering their pre-existing perceptions about their genetic risk, and found that women with high expectations of carrying a *BRCA1/2* mutation, often shaped by information heard from healthcare providers or the media during their diagnosis or treatment period, tend to experience greater emotional distress after testing. Also, a study by Inan, Günüşen & Üstün, (2016) found that women after being diagnosed with breast cancer showed avoidance behaviours, they tried to avoid situations that were hard to cope with. Receiving a positive genetic test result can be a psychologically challenging experience (Qiu et al., 2016). Therefore, the above suggests that the way women react to their cancer diagnosis and treatment can influence their emotional experience of receiving genetic test results. This section explores the impact of the diagnosis and the perceptions of the cause of cancer.

1.1.3.1 The Impact of a Breast Cancer Diagnosis

Omani patients have been reported to face significant psychosocial challenges after a breast cancer diagnosis (Al-Azri et al. 2014). In this study, patients reported feeling shocked and overwhelmed by the prospect of death, leading to psychological distress, anxiety, depression, exhaustion, and self-blame. The diagnosis impacts not only the patients but also their families, causing sadness and anxiety among loved ones. Relationships, especially with spouses, can suffer, leading to sexual distancing, communication issues, and even relationship breakdowns. Additionally, societal stigma may lead to social distancing, with patients often seen as victims, leading to pity and discrimination. Some patients reported pressure from family and friends to pursue traditional treatments, such as "Wasam" (branding with a hot iron). Concerns about relapse, cancer spreading, and loss of femininity due to treatments were also expressed.

A study by Al-Azri et al. (2024) explored how breast cancer affects the social interactions of young Omani women. Interviews with eleven women under 45, 1-5 years post-diagnosis, revealed six key impacts. 1) Many women showed resilience through faith, 2) others struggled with self-confidence and withdrew socially, 3) interactions with children aimed to maintain normalcy, 4) spouses provided emotional support, though intimacy was often challenging, 5) family members offered essential support, and 6) stigma led some women to hide their diagnosis and limit social interactions.

International studies have also shown that breast cancer diagnosis significantly impacts patients. A study by Hegel et al. (2006) in the USA revealed that 41% of 236 newly diagnosed women experienced substantial distress, and 47% had at least one psychiatric disorder, such as major depression or post-traumatic stress disorder. These conditions were associated with notable functional impairments. Similarly, Inan et al. (2014) conducted research in Turkey with nine women diagnosed with breast cancer. The interviews revealed a mix of intense emotions—fear, sadness, anxiety, and despair—prompting the women to perceive their diagnosis as a traumatic event. This affected their emotional well-being and adherence to treatment, as they confronted uncertainty about their condition and treatment. While some coped by avoiding the issue, others showed resilience and hope amidst the challenges they faced.

A qualitative study by Iddrisu, Aziato and Dedey, (2020) examined the significant physical, body image, and emotional effects of breast cancer on young Ghanaian women. Through interviews with 12 participants, the study highlighted their experiences. Physically, the women dealt with weakness, limited mobility, and pain that disrupted their daily lives. Mastectomy treatment negatively impacted their body image, leading to feelings of unattractiveness and self-consciousness, with some resorting to makeshift prostheses. Chemotherapy-induced hair loss further affected their self-image. Emotionally, the diagnosis and treatment caused sadness, depression, anger and denial, with some feeling they had brought shame to their families due to the stigma surrounding breast cancer.

1.1.3.2 Breast Cancer Patients' Perceptions of Cancer Causes

A study by Thomson et al. (2014) explored beliefs about the causes of breast cancer among women in Canada. It compared responses from 1109 women with breast cancer and 1,633 without the disease. The findings revealed that women with breast cancer often attributed their illness to mental or emotional factors, as well as lifestyle and physiological factors. In contrast, women without breast cancer commonly believed the causes were familial or inherited factors, along with lifestyle and environmental factors.

Moreover, a qualitative study in Taiwan interviewed 29 breast cancer survivors to investigate their perceptions of the causes of their illness and the impact on their lifestyle changes. Most survivors attributed their cancer to "stress and emotions," leading them to prioritise stress reduction over adopting healthier habits like improved diet and exercise, even if it meant neglecting those habits. Similarly, a study on Bahraini women with breast cancer examined their quality of life after diagnosis (Jassim & Whitford, 2014). They found that many participants attributed their cancer to God's will, which provided comfort but also led to fatalism and lower engagement with medical treatment. Furthermore, they showed that some women held supernatural or traditional beliefs about their disease. This highlighted key differences from western studies, which included a strong emphasis on spiritual practices for comfort, the use of traditional clothing to hide physical changes, and significant family support (Jassim & Whitford, 2014).

A study by Beeken et al. (2016) investigated cancer survivors' perspectives on diet's influence on cancer development and recurrence. Through semi-structured interviews with 20 survivors, the research revealed that while participants viewed a healthy diet as important for overall health, they did not believe it was specifically crucial for cancer outcomes.

1.1.3.4. Cultural Attitudes Towards Cancer Disclosure in Oman

A study by Al-Bahri et al. (2019) investigated family involvement in treatment decisions for Omani women with breast cancer, surveying 79 women and their family members. It found that family members, particularly younger, male, employed, and close relatives, played a significant role in the decision-making process. The degree of involvement varied among families, highlighting the cultural importance of family support in healthcare decisions in Oman.

Furthermore, a narrative reflection of Al Balushi, (2021) explored how cancer diagnoses are often hidden from patients in Oman by family members and doctors typically at the insistence of family members who believe it protects the patients. Families feel responsible for shielding patients from distressing news, fearing that disclosing a diagnosis could bring shame or worsen the patient's condition. Open discussions about cancer are often seen as a loss of pride and dignity.

Similarly, a study in Saudi Arabia investigated cancer patients' and families' attitudes towards disclosing diagnoses. While most patients and family members preferred the diagnosis be told to the patient, some family members expressed concerns about the potential negative impact of disclosure on the patient's emotional wellbeing (Alzahrani et al., 2018).

1.1.4 Experience of Receiving a Pathogenic Genetic Result

Genetics professionals may often assume that because of the cancer history, patients will cope well with their positive cancer genetic status. According to Dorval et al., (2000), women who have cancer showed more distress and negative feelings receiving pathogenic results than those who had no personal history of cancer. In contrast, Croyle et al., (1997) reported that patients who showed more distress were the carriers with no cancer history rather than those who had already been diagnosed with cancer. Therefore, patients' experiences with pathogenic germline genetic testing results vary from one patient to another. A likely factor of distress is the long waiting time as Bartley et al. (2021) investigated cancer patients' experiences of uncertainty while awaiting genome sequencing results. The study involved cancer patients suspected of having hereditary cancer and utilized a mixed-methods approach. Although patients sought genome sequencing to alleviate uncertainty about their cancer risk, the waiting period introduced new uncertainties regarding the testing process and personal implications. Patients employed strategies such as seeking information, positive thinking, and social support to cope.

1.2.4.1. The Psychological and Behavioural Impact of Receiving Pathogenic Genetic Result

A study in Pennsylvania involving 60 individuals who received clinically actionable genetic test results found varied psychological responses to pathogenic genetic results. Positive reactions included gratitude, understanding of future health benefits, and a sense of support from the healthcare system and loved ones. Negative responses consisted of feelings of fear, guilt and worry about health implications for themselves and their families. Participants with negative results expressed surprise, shock and bitterness, describing their experience as being in a "twilight zone" (Baker et al., 2022). Another study in France examined the short-term psychological effects of *BRCA1/2* test results on 273 women with breast cancer. Those who tested positive reported low to moderate anxiety and depression (Brédart et al., 2013). Similarly, an Italian study found that a significant number of women (mean age 47.3) experienced emotional distress, including anxiety, depression, anger and feelings of vulnerability after receiving their results (Mella et al., 2017).

Furthermore, a mixed-methods study in China examined the quality of life and psychological state of breast cancer patients who underwent *BRCA1/2* genetic testing. Interviews with 13 mutation carriers revealed a mix of emotions; while many felt relief from understanding the cause of their cancer, others experienced fear, anxiety and guilt, particularly regarding their family's inherited risk. Following their test results, some women made lifestyle changes and sought more information about the implications for their health and that of their families, expressing a need for guidance on risk management and support for their loved ones.

A study in Germany involving 20 women with pathogenic variants in moderate-risk genes for hereditary breast cancer found that participants experienced a variety of emotions, including overwhelm, uncertainty, relief and empowerment (Stracke et al., 2022). Similarly, a mixed methods study in the USA with 505 women who received positive *BRCA1/2* results revealed concerns about passing on the mutation to their children, which significantly affected their reproductive decision-making. Additionally, the study highlighted that BRCA status can influence self-concept, particularly regarding perceived vulnerability and stigma (Hesse-Biber et al., 2022).

The timing of genetic test result disclosure significantly influences the surgical treatment decisions of breast cancer patients. A Canadian study found that women receiving *BRCA1/2 or PALB2* results before surgery were more likely to choose risk-reducing bilateral mastectomy, avoiding a second operation and harmful radiation therapy (Apostolova et al., 2024). Similarly, a study in the USA by Armstrong et al. (2021) revealed that many women received their genetic test results after initial surgery, indicating that receiving results before definitive surgery led to a higher likelihood of opting for bilateral mastectomy. No studies in the Omani or Arabic context were found hence this study was conducted.

1.2.4.2. Family Communication

Sharing genetic test results with family members is a complex and multifaceted process influenced by various factors (Abdel-Razeq et al., 2023). A study that examined the practices and attitudes of 169 Jordanian breast cancer patients with pathogenic germline variants regarding communicating their genetic test results to family members. This study revealed that while most opted to share their genetic results with first-degree female relatives, many of these relatives took no subsequent action. Barriers such as fear of positive results, financial limitations and social stigma significantly impacted family members' willingness to pursue genetic testing. In Japan, research indicated that all cancer patients shared their genetic findings with at least one family member, with a particular emphasis on first-degree relatives, including children and siblings (Fukuzaki et al., 2021). Meanwhile, an investigation among Italian women demonstrated high disclosure rates of hereditary cancer information, revealing that family dynamics and established communication styles heavily influenced the sharing process (Di Pietro et al., 2021).

Additionally, interviews conducted with women who had pathogenic variants showed that motivations for sharing their test results were often rooted in a sense of moral obligation and the desire to empower family members. However, these women faced challenges, including concerns about their family's reactions and the emotional ramifications of disclosing such information (Dean et al., 2021).

A study focusing on young Black women in the USA found that while most disclosed their BRCA genetic test results, those who tested positive were notably less likely to share the news with their daughters (Conley et al., 2020). Similarly, research on women with confirmed BRCA1/2 mutations showed that even though almost all shared their genetic risk with first-degree relatives, only about

20% of those relatives underwent genetic testing. Factors like emotional closeness and the woman's educational background influenced how the information was communicated (Silva et al., 2022).

1.2.4.3. Adaptation to Breast Cancer Diagnosis

Eventually, breast cancer patients learn to develop strategies to accept and cope with the psychosocial distress related to their diagnosis. They look for coping mechanisms based on cultural norms and religious beliefs by increasing spirituality, surrendering to God's will and focusing on essential requirements of life such as health and family closeness rather than focusing on wealth and social status (Al-Riyami, Abdulhadi & Al-Azri, 2020). In contrast, others reported that breast cancer diagnosis brought them closer to their families and spouses (Stracke et al., 2022).

A qualitative study in Ireland explored the coping needs of individuals with *BRCA1/2* alterations (Warner & Groarke, 2023), They identified two main themes: "Adjusting to a New Perspective", which addressed the range of emotions such as anxiety and relief, and "Making Sense of BRCA", focusing on finding meaning and hope. Another study in Portugal explored the complexities of how families adapt to hereditary cancer syndromes, revealing that families serve as the primary source of information and emotional support. It emphasised the importance of communication and a family-centred approach in coping with hereditary cancer challenges (Silva et al., 2022).

Furthermore, Jewett et al., (2022) found that a sense of connection was linked to better psychosocial outcomes in gynaecologic and breast cancer survivors. The researchers aimed to understand how feeling connected to others affected survivors' experiences of post-traumatic growth, meaning-making, peace, and spirituality. The results showed a strong positive association between a sense of connection, greater post-traumatic growth, and reduced loneliness. Additionally, Serna, Canut and Ortega, (2024) investigated the experiences of individuals with hereditary cancer syndromes, revealing that 22 interviewees from 10 European countries felt they received inadequate information on health behaviours related to cancer risk and sought more support from cancer professionals.

A study conducted in Germany explored the feelings and coping styles of women with moderate-risk breast cancer gene results. The coping styles were classified into three categories:

- Emotion-focused coping: Seeking support from family and friends, self-care, engaging in sports and nature, and turning to religion.
- Problem-focused coping: Making lifestyle changes, considering surgeries, and participating in surveillance programmes.
- Meaning-focused coping: Sharing personal experiences, recognising strengths, being aware of personal needs, and enjoying life. (Stracke et al., 2022)

1.2. Chapter Conclusion

The literature revealed limited information on the experiences of Omani women with breast cancer regarding the receipt of positive germline results for inherited breast cancer predisposition. The literature examined the challenges and considerations around genetic testing for hereditary breast cancer in the context of international studies. It studied breast cancer patients' responses to receiving positive genetic test results. Women with positive results may face additional psychological and practical difficulties, such as understanding the implications for their own and their family's health. The literature explored the diverse experiences and coping strategies of women as they navigate the breast cancer journey, from diagnosis through treatment and receiving positive cancer genetic results. In addition, the communication styles of hereditary breast cancer were explored.

CHAPTER 2: METHODOLOGY

The methodological framework used to investigate breast cancer patients' experience of receiving pathogenic germline genetic results in Oman is outlined in this chapter.

2.1 STUDY RATIONALE

Most of the studies that explore patients' experience of receiving pathogenic germline genetic results have been done in Western countries. Little is known in the Omani setting, but considering the cultural differences between the West and Oman. Omani patients might have different experiences than what is currently available in the literature. Further research is therefore warranted to gain a better understanding of Omani patients' experience of receiving positive cancer genetic testing results with the aim of improving service delivery and future patient care. The research question therefore is:

What is the Omani breast cancer patients' experience of receiving pathogenic germline genetic results?

2.2 AIM

This study aims to investigate breast cancer patients' experience of receiving pathogenic germline genetic results in a single cancer centre in Oman.

2.3 OBJECTIVES

The specific objectives are to explore the participants:

- Reasons and expectations for undergoing genetic testing.
- Reactions to receiving a pathogenic cancer predisposition result.
- Utilisation of the received pathogenic genetic result in their life.
- Family communication with regard to dissemination of results.
- Cultural practices regarding a cancer diagnosis and genetic testing.

2.4 STUDY DESIGN

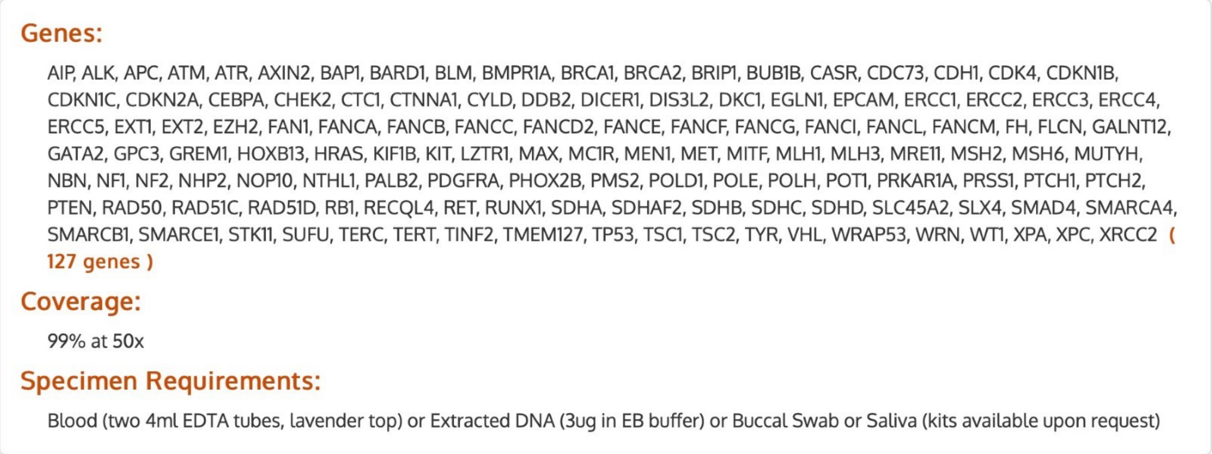
This study is based on an interpretive phenomenological philosophy, which assumes that humans construct meanings related to an experience and provides insight into the meaning of the phenomenon from the perspective of the participants (Scotland, 2012). As this study aims to understand the experience of women who received a positive germline genetic result, a phenomenological approach was deemed appropriate. The use of an interpretive phenomenological approach, enables a deeper and more detailed understanding of the research participants' lived experiences within their real-world contexts. This approach allows the researcher to explore the subjective meanings and perceptions that participants assign to the phenomenon being studied, rather than relying just on objective, measurable data. By adopting this methodology, the study can uncover rich insights that might not be accessible through quantitative techniques alone (Scotland, 2012).

2.4.1 Research Setting

Sultan Qaboos Comprehensive Cancer Care and Research Centre (SQCCCRC) provides comprehensive cancer care to adult patients with solid tumours. Currently, there are six different programmes, of which the breast cancer programme is the biggest programme in the centre.

The breast cancer programme at the SQCCCRC offers patient care through a multidisciplinary team (MDT). Each newly diagnosed breast cancer case is discussed during a weekly MDT meeting, and the course of action for diagnosis and treatment is decided. During these meetings, eligible patients are identified and referred for genetic counselling and testing based on several criteria. While the genetic community in the Middle East and North Africa (MENA) is currently developing more regionally specific guidelines for the Gulf Cooperation Council (GCC) countries, the international guidelines are currently used to refer and evaluate whether a hereditary cancer syndrome is present in a family. These criteria include: early age of onset for the specific cancer type, bilateral cancer, multi-focal tumours, presence of multiple cancer types in an individual, triple-negative breast cancer, , and significant family history of cancer, such as two or more first and/or second-degree relatives with the same common cancer, several first and/or second-degree relatives with a related type of cancer, two family members with the same rare cancer, and two family members with related rare cancers (Daly et al., 2020).

Since establishing the genetic clinic at SQCCCRC on September 1, 2021, 680 breast cancer patients have been referred to the GC and were counselled and offered genetic testing. In addition about three quarters of these cases were offered genetic counselling and in-house testing for only three genes which includes, *BRCA1/2* and *PALB2* in view of possible therapeutic benefits (Arun et al., 2024). At present only verified *BRCA1/2* and *PALB2* next generation sequencing is available in the molecular genetics laboratory at SQCCCRC but the analysis does not include Multiplex Ligation Dependent Probe Amplification (MLPA). Since 13.2% and 2.7% of pathogenic variants in *BRAC1* and *BRCA2* respectively (LaDuca et al., 2020) are deletions and duplications, MLPA should be performed for complete analysis. In addition, other high, moderate and low risk genes associated with hereditary breast cancer should also be evaluated. Therefore, all patients including those for possible therapeutic benefits are offered a comprehensive cancer panel through the American laboratory, Fulgent Genetics. Fulgent's comprehensive cancer panel includes sequencing and MLPA of 127 genes linked to hereditary cancers. Figure 1 below shows the list of the tested genes and the coverage. This comprehensive cancer panel includes potential genes that may have recently been found or for which further research is necessary, in addition to well-known genes that raise a person's risk of cancer (Fulgent, 2023).



Genes:

AIP, ALK, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, CTC1, CTNNA1, CYLD, DDB2, DICER1, DIS3L2, DKC1, EGLN1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, EZH2, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GALNT12, GATA2, GPC3, GREM1, HOXB13, HRAS, KIF1B, KIT, LZTR1, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NHP2, NOP10, NTHL1, PALB2, PDGFRA, PHOX2B, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PRSS1, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL4, RET, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC45A2, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TERC, TERT, TINF2, TMEM127, TP53, TSC1, TSC2, TYR, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2 (127 genes)

Coverage:

99% at 50x

Specimen Requirements:

Blood (two 4ml EDTA tubes, lavender top) or Extracted DNA (3ug in EB buffer) or Buccal Swab or Saliva (kits available upon request)

Figure 1. List of the genes tested in the full comprehensive cancer panel (Fulgent, 2023)

At SQCCCRC, genetic counselling is provided by certified genetic counsellors and/or clinical geneticists, who facilitate the informed genetic testing process. The consultation involves a detailed review of the patient's personal and family history, with a focus on cancer-related information. During the counselling session, each patient undergoes a risk assessment to determine their eligibility for genetic testing.

After consenting to the test, 10 mL (Ethylenediaminetetraacetic acid (EDTA) tubes) of blood is taken from a patient for the genetic test. When Fulgent laboratories result is back, another session is scheduled for result delivery.

2.4.2 Method: Semi-structured Interviews

The study data were collected using in-depth, face-to-face semi-structured interviews (SSI), in which a set of questions was prepared beforehand to guide the interviews. A semi-structured approach was chosen because SSI can be used when there is enough objective knowledge about a phenomenon but a lack of subjective knowledge (McIntosh & Morse, 2015).

The interview question guide was developed following the steps described in Naz et al. (2022). The interview guide attached in (appendix A) was developed based on these steps:

1. Defined the research question and aim.
2. Reviewed the existing literatures.
3. Identified the target population.
4. Developed the initial interview questions.
5. Organized the questions into a guide.
6. Pilot tested the interview guide.

2.4.3 Exploratory Interview (pilot study)

Four exploratory interviews were conducted before the actual interviews to validate and evaluate the interview content and process. These interviews also served as training for the researcher's interviewing skills and techniques (MacFarlane, Veach & LeRoy, 2014). Two interviews with staff from the genomic department at SQCCCRC were conducted under the direct supervision of one of the research supervisors. One of the staff members was a genetic counsellor in training in the department and the other was a coordinator in the centre. The latter two interviews were conducted with health professional students during a training rotation within the genetics department. These sessions were not directly supervised but discussed with the supervisors of this project. The whole process of the explorative interviews was evaluated and discussed with the study supervisors to make any necessary adjustments and improvements to the interview guide. No adjustments were made to the content of the interviews, such as the question guide or the information sheet; however, enhancements were made to the researcher’s interviewing skills, such as active listening, probing and asking follow-up questions during the interviews. Once the researcher's interview skills were sufficiently developed during the pilot interviews and the research proposal was accepted by the ethics committees of both the University of Cape Town (UCT) and SQCCCRC, then the actual interviews were conducted.

A pilot interview was conducted with an actual participant to gain further insight into the interview process and outcome, and was subsequently included in the data analysis as valuable information was obtained. The interview took place in Arabic and was then transcribed in Arabic and translated into English by the researcher.

2.4.4 Recruitment and Data Collection

While this study has effectively addressed the research question and objectives which were to investigate these women reasons for seeking testing

In this study, the sampling was purposive to provide rich and detailed information relevant to the research aim and objectives. Participants were Omani women diagnosed with breast cancer with identified pathogenic and likely pathogenic variants in high and moderate breast cancer genes (inclusion criteria). The breast cancer patients without an identified genetic aetiology to their diagnosis, and the non-Omani patients were excluded because they were not the targeted participants who would fulfil the aim of this study. All participants were recruited from a single centre in Oman, SQCCCRC. According to Coyne, (1997:629), "all sampling in qualitative research is purposive sampling. Thus, the sample is always intentionally selected according to the needs of the study".

During the recruitment, there were 31 eligible potential participants who were identified from the Genomic department patient’s database at SQCCCRC. All participants were invited telephonically to participate in the semi-structured interview. The coordinator of the genomic department at SQCCCRC contacted the proposed participants to invite them to participate in the study. The coordinator informed the patients about the research being conducted and notified them that their participation was entirely voluntary and if they did not participate, it would not affect the health services they were receiving from SQCCCRC. The coordinator's initial contact allowed the participants to freely ask questions and decline to participate without feeling pressured by the researcher (MacFarlane, Veach & LeRoy, 2014). The invitation script is attached in the appendix B, the researcher contacted those individuals who indicated interest in participating in the study.

For the patients who consented to participate in the study, the Hospital Information System (HIS) was used as a reference for patients' information such as demographic data, hospital appointments, and medical documents. Before the interviews, participants received a text message reminder, and a consent form (appendix C) and information sheet (appendix D) were attached. All interviews were conducted face-to-face during the participants' scheduled oncology visits and took place in a quiet and comfortable venue in SQCCCRC. The researcher conducted all nine interviews and made all the necessary arrangements. The interviews were carried out between March and October 2024, with durations ranging from 25 to 90 minutes. Each participant was interviewed only once.

Before the interviews, a good rapport was built with the interviewees to make them feel comfortable and safe sharing sensitive and personal details. The interviews started with a warm welcome, a thank you for participating and a short conversation off the research topic (Pietkiewicz & Smith, 2014). The interviews were in the local Omani dialects where the researcher spoke her own dialect and the interviewees spoke in their own dialects; however, this did not hinder understanding because the researcher is familiar with most of the local dialects in the country and there are no major differences in vocabulary but rather in pronunciation. The conversation between the participants and the researcher was an informal way to enhance more open and relaxed conversations to obtain an authentic and culturally richer experience (Swain & King, 2022).

The participants were asked open-ended questions derived from the research objectives; however, during the interviews, their precise phrasing and chronological sequence were not predetermined (Naz et al., 2022). The researcher allowed the participant's responses and the flow of the interview to determine the order in which the questions were asked during the interviews, which provided flexibility and a comfortable environment for the participants to express themselves. Follow-up questions and probes were used during the interviews to find deeper insights and understanding of their experiences. In addition, participants were allowed to ask questions and clarify concepts, which ensured accurate data collection (Alamri, 2019). The physical presence of the participants allowed the researcher to notice any discomfort in the respondents and reminded them of the option not to answer the question or stop the interview (McIntosh & Morse, 2015).

The interviews were voice recorded by the researcher's password-secured device. Recording the interviews allowed the production of accurate transcripts in which the contextual meaning was preserved, as well as essential details (Rutakumwa et al., 2020). Interviews were recorded with the participants' permission before the interviews; see the consent form attached in the Appendix C.

2.4.5 Data Analysis

The audio recordings were manually transcribed verbatim in Arabic by the researcher soon after the interviews. The researcher then translated the transcripts into English. The translations were checked by another staff member from the genomics department at SQCCCRC to ensure no meaning was lost during the translation process. It is worth mentioning that the researcher is a genetic counsellor in training and has three years of experience translating from Arabic to English in the genetic counselling settings as part of her daily duties, and the researcher is from the same culture as the participants. Al-Amer et al. (2016) reported that in cross-language studies, where the data must be translated from its source language (Arabic) to the targeted language (English), the reliability of the translation process is essential for the integrity of the results, as inaccuracies can significantly compromise the study outcomes. Al-Amer et al. (2016) suggested that the person who should conduct the interview has to be an "insider” and understands the language and the culture.

Systematic thematic analysis was used to analyse the data. Thematic analysis is a technique used to find and interpret patterns and themes in the data. It is essential that the researcher keep their own biases in check when interpreting the data (Naeem et al., 2023). In this study, the entire analysis process, coding and identifying the themes, was discussed and checked with the supervisors. The role of the supervisors was to provide external validation and to ensure the objectivity of the analysis, thereby minimising personal biases during data analysis.

The researcher adopted both inductive and deductive approaches in data analysis. The analysis began with an inductive approach, where the researcher read the transcripts line by line to drive the initial codes. The first two interviews were entirely inductively coded. By the third interview, the researcher included deductive analysis. The deductive approach was used after some of the categories and themes started to develop to help with labelling concepts and categorising them into correct themes and categories. The research objectives and the interview questions played a crucial role in creating a starting a framework of codes and themes from the existing literature on the phenomenon under inquiry (Azungah, 2018). Later, all interviews were analysed using both approaches. The statements that directly fitted into one of the codes in the codebook were coded deductively. The statements that did not quite fit into the coding list were not forced into one of the codes but rather have been coded inductively (Naeem et al., 2023).

The researcher was the coder, and the supervisors were the co-coders of the data. Manual coding and computer software like NVIVO were used for organisation of the coding in this study. The researcher coded first and then the supervisors provided feedback and suggestion on the created codes and themes. The thematic analysis steps below were followed to analyse the data (Braun & Clarke, 2006):

- Familiarised with the data: transcribed and translated them, read them repeatedly, and then wrote down initial ideas.
- Generated initial codes: identified and coded relevant features from the data relating to the phenomenon.
- Searched for themes: gathered the codes into possible themes and collected information related to each theme.

- Reviewed themes: themes were refined. For example, created new themes, combined themes, and collapsed others until satisfied with all themes.
- Defined and named themes: continued refining and providing suitable names for each theme.
- Produced the report: linked the complete themes to the literature and the research question.

Eleven participants were invited up until data saturation was reached; two declined to participate; one declined because she was already part of another ongoing study at SQCCCRC, and the other did not provide a reason for not participating. Data saturation was attained by the time of analysing the seventh interview. No new codes or themes were emerging and the data collected was sufficient to fully address the study's objectives (Braun & Clarke, 2021). Two further interviews were conducted to ensure that data saturation was reached. A total of nine participants were interviewed, and the obtained information started to reaffirm previously identified codes and categories, indicating that further data collection is unlikely to substantially change or add to the current thematic framework.

2.5 ETHICAL CONSIDERATIONS

This research adhered to the Declaration of Helsinki. Informed consent was obtained, and participants' autonomy was respected. The informed consent form is attached in Appendix C.

The study has ethical clearance of Sultan Qaboos Comprehensive Cancer Care and Research Centre (SQCCCRC) (CCCRC-37-2023) The approval letter is attached in Appendix E. And it has ethical clearance from the Human Research Ethics Committee (HREC) of the University of Cape Town was also obtained under reference 822/2023, which is attached in Appendix F.

2.5.1 Privacy, Confidentiality and Consent

Privacy and confidentiality were assured by not sharing or revealing any identifying information; the participants were given a code name such as (P1, P2…). Only the main researcher had full access to the data; no third party had access, as the researcher did all the transcription. The supervisors had access to the data anonymously and without any identifiable information. All research data were stored on the researcher's password-protected devices, and only the researcher could access them. A folder was created specially to store the research data, which will be destroyed after the completion of the study (Mohd Arifin, 2018). In the consent form, the participant was informed that their participation was entirely voluntary, and they were assured that no discrimination would be taken against them if they declined to participate or withdrew from the study. They were also reassured that they did not have to answer any question they felt uncomfortable with. The researcher did not approach the patients directly; the centres' staff approached them to minimise risk of coercion. It was illustrated that there was no financial gain from participating in the study. Also, permission to record the session was obtained. Before signing the consent form, the participants were allowed to ask questions to clear up any uncertainties (Shokrollahi, 2010).

2.5.2 Foreseeable Risks and Benefits for Participants

There were no major potential risks to the participants in this study. Participants were informed that if the questions provoked emotions during the course of the interview, they were allowed to stop the interview or refrain from answering the questions without any consequences or effect on the health services received. It was a measure to ensure participants' comfort and wellbeing throughout the study (Moriña, 2021). In addition, participants would have been referred to a genetic counsellor or psychologist or to a suitable health care provider if serious emotional distress was recognised, the researcher would have arranged all the counselling services, and participants would have been informed about it. However, this did not happen during the data collection process. There was no direct benefit gained by participating in this study as it would not change their current treatment plan. Benefit is for future service.

2.6 RESEARCH OUTPUT

This study is submitted as the mini dissertation component of the MMedSc in genetic counselling degree at the UCT and submitted to SQCCCRC as it is done on the Omani population. Through this study, we have gained a deeper understanding of the Omani population's perception of cancer genetic testing and the psychological and behavioural impact of receiving a pathogenic germline genetic result. The outcomes of this study can be used to enhance service delivery, leading to improved post-disclosure referrals and better family communication and testing. Furthermore, as SQCCCRC is a research centre, this study can serve as a valuable reference for researchers, oncologists and genetic counsellors in Oman, the Gulf, and Arabian countries, where people share the same characteristics and cultural values.

CHAPTER 3: RESULTS AND DISCUSSION

3.1. INTRODUCTION

In this chapter, the findings of the thematic analysis of the interviews are presented alongside a discussion highlighting how the identified themes and sub-themes contribute to the understanding of the participants' experience with receiving pathogenic germline genetic results.

Table 1 is a summary of the themes that emerged from the data.

Table 1: Themes and their sub-themes

Theme	**Sub-Theme**
Cancer Journey	Cancer Causation Beliefs Diagnostic Journey Treatment Journey
Genetic Testing Motivation and Expectations	Rationale for Undergoing Genetic Testing Anticipation of Result
Receiving the Positive Result	Reaction Upon Receiving the Result Time Receiving the Result Family Communication Up-Take of Genetic Testing by Family Members
Adapting to Hereditary Breast Cancer Diagnosis	Active Coping Engagement Coping Meaning Focused Coping

Each theme and its sub-themes will be presented with quotes from the participants as evidence followed by a discussion. After each quote, the participant number, gene they were diagnosed with and its pathogenicity will be presented e.g. P2, *BRCA2* PV

3.2. PARTICIPANT DEMOGRAPHICS

Table 2 provides a summary of the participants' demographic information. Apart from one patient who was in her later 20s, all of the patients were in their 40s to late 50s, all but one had a higher education qualification, four were employed, eight were married and all had children except for one participant who did not had children yet.

Table 2: Participants' demographic information

Participant unique code	**Current age**	**Marital status**	**Number of children**	**Education**	**Employment status**
P1	44	Married	Have children (number unknown)	University -PhD	Employed
P2	26	Married	0	University - undergraduate	Unemployed
P3	42	Married	5	University - undergraduate	Employed
P4	40	Divorced, Re-married	4	University - undergraduate	Unemployed
P5	45	Married	5	High School	Unemployed
P6	44	Single	0	University -PhD	Employed
P7	46	Married	3	High School	Unemployed
P8	51	Married	6	University - undergraduate	Employed
P9	58	Married	7	Primary School	Unemployed

Table 3 provides a summary of the participants' cancer and genetic test information. Apart from three participants who had additional cancer diagnoses, the majority of patients had unilateral breast cancer, with invasive ductal carcinoma being the most common subtype. Four participants had metastatic disease. The participants were known with pathogenic variants in *BRCA2*, *BRCA1*, *PALB2* and *BRIP1*, with *BRCA2* being the most common, present in six of the participants. Four participants had total hysterectomy with bilateral salpingo-oophorectomy, and five participants were under Screening management.

Table 3: Participants' cancer and genetic test information

Participant unique code	**Cancer Type**	**Uni-lateral/ bilateral**	**TNM stage**	**ER**	**PR**	**Her2**	**Type**	**MGT result**	**Screening management/ Prophylactic surgery**
P1	Breast cancer	Unilateral	pT1cN1Mx	Positive	Positive	Negative	IDC, Grade 1	*BRCA2* (Pathogenic)	Total hysterectomy with bilateral salpingo-oophorectomy
P2	Breast cancer & Thyroid Papillary Carcinoma	Unilateral	cT2N1Mx	Positive	Negative	Positive	IDC, Grade 2	*BRIP1* (Likely Pathogenic)	Screening management
P3	Breast cancer	Unilateral	pT1cN1M0	Positive	Positive	Negative	ILC, Grade 2	*BRCA2* (Pathogenic)	Screening management
P4	Breast cancer	Unilateral	cT2N0M0	Negative	Negative	Positive	IDC, Grade 3	*BRCA2* (Pathogenic)	Screening management
P5	Breast cancer +Thyroid cancer	Unilateral	pT2N1M0	Positive	Positive	Negative	IDC, Grade 3	*PALB2* (Likely Pathogenic)	Bilateral nipple sparing mastectomy Total hysterectomy with bilateral salpingo-oophorectomy
P6	Breast cancer	Unilateral	pT1pN0M0	Positive	Positive	Positive	IDC, Grade 2	*BRCA2* (Pathogenic)	Screening management
P7	Breast cancer	Unilateral	cT3N1Mx	Positive	Positive	Negative	IDC, Grade 2	*BRCA2* (Pathogenic)	Screening management
P8	Breast cancer	Unilateral	CT4N3b M1	Negative	Negative	Positive	IDC, Grade 2	*BRCA2* (Pathogenic)	Total hysterectomy with bilateral salpingo-oophorectomy
P9	Breast cancer +Ovarian cancer	Unilateral	PT3N1M0	Negative	Negative	Unknown	Unknown	*BRCA1* (Pathogenic)	Bilateral nipple sparing mastectomy Total hysterectomy with bilateral salpingo-oophorectomy

A summary of the timeline from when the participants was diagnosed with cancer to their pre-test consultation, result consultation and the interview date can be viewed in Table 4. The time between result consultation and the research interview ranged from a few weeks to two years, with an average of one year and two months. For two participants, the exact duration was unknown.

Table 4: Timeline of the cancer diagnosis and genetic consultations in relation to the research interviews

Participants	**Breast cancer diagnosis**	**Pre-test consultation**	**Result consultation**	**Research interview date**
P1	Unknown	Unknown	January 2022	06/03/2024
P2	March 2022	September 2022	October 2022	17/04/2024
P3	Unknown	Feb 2024	May 2024	13/05/2024
P4	March 2023	May 2023	August 2023	04/08/2024
P5	November 2021	Unknown	October 2022	07/08/2024
P6	June 2022	October 2022	March 2023	02/10/2024
P7	July 2022	August 2022	March 2023	03/10/2024
P8	December 2023	Feb 2024	June 2024	22/12/2024
P9	2000	August 2024	November 2024	26/12/2024

3.3. THEME 1: CANCER JOURNEY

The participants' cancer journey was an integral part of understanding their experience of receiving a pathogenic germline genetic result. The "Cancer Journey" incorporates various stages of their journey. It describes their life before their cancer diagnosis, receiving a diagnosis of breast cancer, the treatments that followed the diagnosis, how they navigated their healthcare through their treatments and the cultural practices related to a cancer diagnosis.

3.3.1. Sub-Theme 1.1: Cancer Causation Beliefs

Life Stressors

The participants described that they were living a normal life without major health issues before their diagnosis. However, they described that they had faced a number of life stressors which they believed contributed to them developing breast cancer. They described facing work-related stress, such as the demanding nature of their work, including getting late-night calls, long commutes between their work and family residence, and working after hours from home. Apart from the work-life balance contributing to life stressors these women reported managing household duties and raising children with little help from their family.

Some women also seemed to face significant financial pressures. They described having insufficient household incomes, leading them to work in jobs at shopping centres and seek assistance from charities to meet their families' needs. P4 for example, prior to re-marriage, she shared facing a lifelong challenge since she was first married at a very young age (at 16 years old), and later she had to go through a divorce and dealt with court cases. She faced financial strains and as a single mother she had to provide accommodation for her children because her family could not support them. She was forced to work in shops and ask charities for help to support her children. Going through these life stressors made her feel depressed and she said,

> *"I have been exhausted raising my children, spending (time and money) on them, providing for them, dealing with court cases (for divorce) and such. Imagine, I was dependent on what people give me, you know, my family's financial situation was bad, I rented a place alone because they (the family) don't have a place for me and my kids, my only option was the charity team. I have worked in shops and such just to make ends meet. It's feels like this by itself causes depression and illness (she means cancer). After you were married when you were young, at 16 years old, it's very difficult".* -P4, *BRCA2* PV

P4 even after receiving her positive MGT result, she still believed that the lived stressors had contributed to her breast cancer diagnosis alongside the genetic factor. She stated,

> *"I feel that the stress and the life that I have been living, they are what made me sick…They are what cause illnesses…I mean the stresses that I went through, the things that happened to me, and the genetic factor. I feel like they all came together at once."* - P4, *BRCA2* PV

A further example can be seen by P3, who described an increase in life stressors, such as increased financial and family pressures, which caused her discomfort and anxiety after previously living a comfortable life.

> *"You know, I mean, until yesterday I was saying, up to 2020, I was very mentally comfortable at home with my family, in my job and all. Since 2020, I started facing financial and family pressures, I don't know how, Subhan Allah (Glory be to God), everything changed for me. It increased with my pregnancy with (daughter name), more and more. This caused me a lot (referring to stress), as you can say, something inside me is not comfortable. It kept increasing from 2020 increased in 2021. With time my anxiety increased"*- P3, *BRCA2* PV

When P3 was asked, post-receiving the positive genetic result, about her perception of what caused her breast cancer, she said she thought it was both factors, the genetic predisposition and the stressors that had contributed to her diagnosis of breast cancer.

> *"So, I say it's both, psychological* (due to lived stress) *and genetics, I'm not telling you one side without the other, I'm telling you both sides together."*- P3*, BRCA2* PV

Another participant, a doctor by profession, described her work-related stress that her job was highly stressful and demanding in terms of doing late nights calls, driving a long distance to work and being responsible for an entire clinic in the morning. She stated that,

> *"Of course, our work is stressful, doing calls being called at 01:00 to go and drive for about 80 kilos (kilometres in distance) in the night. So yeah, it's really stressful. And in morning time, you are managing the whole clinic". –* P1, *BRCA2* PV

P1 further added that due to the stress she decided to retire from her work she said, *"I decided to also to retire in order to take my time,you know, our work is stressful being a docto r"*-P1, *BRCA2* PV, and when asked by the interviewer "if genetic test result has to do with her decision of retirnment, she responded that,

> *"Maybe everything…, Being in a chemo journey. Sorry, the cancer journey.*
> *It's not something like light. The surgery itself, the chemo, the radio.*
> *It's of course a stress. I've been in a stressful situation."*-P1*, BRCA2* PV

All of these stressors had an emotional toll on these women, who reported feeling negative emotions such as anxiety, severe stress, and depression. Even after learning about their genetic predisposition, they still firmly believed that these life stressors had contributed to their breast cancer diagnosis together with genetic predisposition.

The above findings are consistent with what breast cancer patients who did not had genetic testing previously reported in the literature. A recent study in Taiwan that interviewed 29 breast cancer patients found that most participants thought that their "stress and emotions" were major contributing factors to them developing breast cancer (Niu et al., 2022). Thomson et al., 2014, who interviewed 1,109 women diagnosed with breast cancer and 1,633 women without breast cancer from Alberta, Canada. These women were asked about their breast cancer causation beliefs, they found that many women, both with and without breast cancer, linked the disease to stress or emotional factors. However, women without breast cancer diagnosis more frequently reported inhereited factor as a possible cause. Another study conducted in the United States reported similar findings. Researchers surveyed 2,117 cancer survivors, including 416 with breast cancer, 165 with prostate cancer, and 89 with colorectal cancer, about their beliefs regarding the causes of cancer. Among the reported beliefs were genetic factors and stress/emotional factors, which is consistent with the findings of this study (Wold et al., 2005).

Myths and beliefs

Many participants shared myths and beliefs within the Omani population related to what causes cancer. Many expressed that Hasad (evil eye or malevolent envy) is a common belief within their family and community as a cause of cancer. P5 noted that her aunt, who also had cancer strongly, believed that cancer is caused by Hasad. She went further to say that her sister confirmed this by linking certain happy events in P5's life, such as buying a new expensive car and moving to a new house, to cause people to be jealous of her good fortune. She did not share these feeling and informed her family not to put these thoughts in her mind because she believes her cancer is from Allah (God) and she accepts that everything is from God. In view of that she stated,

> *"My aunt, who got cancer before me, says that cancer mostly happens due to the evil eye, and my sister also insists on this. She says, "I'm sure of it" Before I knew about my breast cancer diagnosis, my husband got me an expensive car, and there were people who commented on it…during the thyroid cancer diagnosis, I was moving to a new house. My sister says it's not normal when you moved to a new house, you get thyroid cancer, and now a car and you got this (breast cancer). I told her do not put these ideas in my mind, I do not like to put these things in my mind, I accept anything from Allah…"*- P5, *PALP2* LPV

Conversely to P5, P7 personally believed that her cancer was caused by the Hasad, even after knowing about her genetic predisposition. She stated,

> *" Mostly, Hasad (Evil eye) of people, especially when breastfeeding my children in front of (other) women… to be honest, this is my belief."*- P7, *BRAC2* PV

These findings indicate that beliefs about cancer being caused by supernatural or spiritual forces, such as the "evil eye" or black magic, are deeply rooted in the Omani culture. These findings align with previous research on Omani and Bahraini breast cancer patients. A mixed-method study on Omani women aged 20-50 investigating breast cancer awareness and found that a significant number of Omani women believe in the "evil eye" as a risk factor for breast cancer (Niu et al., 2022) Similarly, a study interviewing 12 Bahraini women aged 39-68 identified the "evil eye" as one of the most common perceived causes of cancer (Jassim & Whitford, 2014).

In addition to Hasad, the women also mentioned specific dietary factors which are culturally believed to be potential causes of cancer. Some mentioned instant noodles, fast food, soda drinks, and red meat. However, a prominent cultural belief was centred on sugar, as shared by P6:

> *"Culture mostly concentrates on that it is due to sugar. They (the society) keep saying sugar, sugar, sugar to the degree that we get scared of the word "Sugar". I searched why sugar would cause cancer despite the dietitian here saying no, it does not".*
>
> *- P6, BRAC2* PV

In the same line as P6, P8 also reported that consuming sugar and meat is culturally believed to cause cancer, and she thinks it is a good thing to reduce the amount. However, she did not believe these to be the main cause of her cancer due to focusing on what her doctors informed her versus cultural beliefs.

> *"They mostly believe it's due to sugar; they say to stay away from sugar and meat, but doctors don't tell me this; I mostly follow what the doctors tell me. Yes, reduce the amount of sugar and meat, but they are not the causative".* - P8*, BRAC2* PV

The fear and anxiety surrounding sugar, as expressed by P6, suggest that these beliefs can potentially influence the dietary choices and overall well-being of breast cancer patients. These findings are consistent with previous local literature on Omani women (Al-Khasawneh et al., 2016) and international studies on adult cancer patients in the United Kingdoms (UK) and breast cancer patients in Australia that all reported similar beliefs linking certain dietary factors, such as fast food, sugary food, soda, and red meat, to cancer risk (Beeken et al., 2016, Thomson et al., 2014).

In addition to dietary factors, P1 mentioned that there are specific rumours circulating in her community about certain types of bras and deodorants potentially causing cancer. She expressed that she was cautious about these potential breast cancer risk factors, even if they were based on rumours. She explained,

> *"There are some rumours about even the kind of bra could cause cancer, like if it's having, if it's metallic. I am avoiding all this, even deodorant. I'm searching for the healthy ones, or I don't use it directly (on my skin), even from before (breast cancer diagnosis)".* - P1 *BRAC2* PV

This quote illustrates how anxieties about cancer can extend to everyday objects which may reflect broader cultural anxieties about chemicals and environmental toxins, as reported by González et al. (2014).

3.3.2. Sub-Theme 1.2: Diagnostic Journey

The diagnosis journey was the beginning of a new phase in these women's lives. This section explores the initial symptoms which alarmed them in seeking a diagnosis, and their reaction upon receiving a breast cancer diagnosis. The diagnostic journey provides significant insights into the challenges the women faced in obtaining a diagnosis.

Initial Symptoms and Seeking a Diagnosis

Initially, they reported facing a range of symptoms that caused them to seek medical care. These symptoms were described as feeling a sting or a pain in the breast, feeling a lump or swelling or something like a ball moving or a hard texture in the breast. Additionally, they reported experiencing fever, bloody discharge from the nipple, breast inflammation, and fluid-filled cysts. The women identified alarming breast related symptoms during a variety of means, such as during performing breast self-examination, feeling a lump while watching television, or breastfeeding their babies, going for investigation for breast unrelated symptoms, and having a mammogram upon a friend's advice. For example, one participant described her initial symptoms as having a painful tumour she said:

> *"I had a tumour; it was painful, and it gave me fever and I couldn't sleep at night. It was filled with fluid (the cysts)"*- P9*, BRCA1* PV

Other women did not have any symptoms, and their tumours were found through screening. Participant six, a nurse, discovered her tumour while performing regular breast self-examination, she said, *"I do self-examination for myself. It matters a lot to me, female body structure, so I was checking, and suddenly I discovered that I have a hard texture in my right breast; it was not showing in the appearance, no redness, no swelling, nothing, just hard touch"* - P6*, BRCA2* PV

Similarly, P8's breast tumour was found through a mammogram that she only had because a friend suggested it while she participated in a study on uterine cancer. In the study hospital she asked the doctor to schedule her an appointment for a mammogram.

"In (name of governmental) Polyclinic, they did a study to screen for uterine cancer because of its high incidence among women…one of my friends advised me to get a mammogram, May Allah reward her… (then) I asked the doctor to make an appointment for me (for mammogram), and she referred me to (name of secondary care governmental) Hospital… so I did the scan, and it was found I have this (tumour)" - P8, *BRCA2* PV

For some participants confirming the diagnosis was a quick process. As reported by one of the participants who immediately after her initial symptom of feeling a lump contacted her doctor at the private hospital who took immediate action.

"I immediately called the doctor at (name of private) hospital and made an appointment. When I got there, Subhan Allah (Glory to God), the doctor was not on duty; there was an Indian doctor instead, but she said she would see me. Mashallah, she was knowledgeable. As soon as she saw me, she immediately sent me for scanning and gave me a high probability that this might be it (cancer)-P5, BALB2 LPV

However, for the majority of participants obtaining a definitive diagnosis wasn't straightforward. These women reported facing many obstacles such as visiting multiple hospitals, undergoing a series of tests and check-ups, lengthy waiting times for appointments, and misdiagnosis or dismissal of their symptoms by healthcare providers before receiving clarity about their symptoms which lead to a delay in diagnosis, and therefore negative psychological consequences.

As one of the participants expressed that she had suffered until she got a diagnosis, she went to multiple hospitals, she went to primary, secondary, and tertiary governmental hospitals, she even went to private hospitals to find an answer for her symptoms (what was it?). But unfortunately, her symptoms got dismissed and on top of this in one of the tertiary care doctors humiliated her when she was insisting something is wrong. All of that left her with no option but to travel abroad to India to seek a diagnosis.

> *"… I got very scared, and I went to a private hospital (hospital name), they said as long as it doesn't cause you pain, then there's no problem… At the (name of a tertiary care governmental) Hospital I went to one of the specialists. I wanted to be reassured because there was something inside me that was not comfortable. I wasn't comfortable, especially when I used to read and I can see that there was something abnormal going on, but the doctor told me there was nothing. I told him doctor, there is discharge that is close to blood, I told him, is it not possible to remove this gland? I swear to my lord I asked him this, he told me you are diagnosing yourself. You are trying to be a doctor? he had a female doctor and a trainee nurse with him. Imagine, I went in, after Allah, I wanted a doctor to reassure me and ease my mind, to give me something, a diagnosis so I can be at ease, No, "he wiped the floor using me" (it is a local saying means treated her so bad) and I left crying, … After two weeks, I travelled to India…The doctor just with the hand (clinical examination) told me this is cancer"*- P3, *BRCA2* PV

Participants also reported receiving an initial misdiagnosis, which necessitated repeated investigations and procedures. As shown in P1's experience. She went for investigations with different specialist who reassured her it was a benign bump. It was only after a Computed Tomography (CT) scan for abdominal pain that it revealed a breast tumour. She said,

> *"You know, like for my diagnosis, I felt auxiliary lymph node like nine months ahead of that (the diagnosis). And I've been to a surgeon, and I 've been to breast radiologist. Everything was done, they reassured me it's benign…but nine months after that I was doing just incidentally for my abdominal pain, a CT which showed a lesion in the breast. And then I go for investigations again"*- P1, *BRCA2* PV

The findings above show the varied ways in which these women discovered their breast cancer symptoms and upon them sought medical care which suggest that Omani women are aware of the alarming signs of breast cancer. These findings are in contrast to what was previously found in the Omani population where 369 women were surveyed and it was found that only 21% demonstrated good knowledge, while 59% had average and 19% had poor knowledge (Renganathan et al., 2014).

The findings of the study also show the complicated and often challenging experiences breast cancer patients' encounter when seeking a diagnosis. Consistent with existing research, many participants reported facing many obstacles, including repeated visits to multiple healthcare providers, going through many tests and procedures, and facing dismissals or misdiagnoses of their symptoms. Similar experiences have been documented in studies conducted in Singapore, where 36 breast cancer patients were interviewed by Ng et al., (2020), and in Tanzania, where 12 women were interviewed to explore factors contributing to delayed diagnosis (Sakafu et al., 2022). A study in Oman by Al-Azri, (2016) found that diagnostic delays can occur at multiple levels, such as patient delays in recognising symptoms and seeking help, or at the level of primary and secondary healthcare where doctors fail to recognise symptoms and provide timely referrals. These challenges and obstacles have negative psychological consequences for these women. This is problematic given that unaddressed psychological distress in newly diagnosed breast cancer patients can persist for many years and adversely impact the rest of their cancer journey, treatment outcomes, and quality of life (Hegel et al., 2006).

Reaction to Diagnosis

Once a diagnosis was made, the study findings revealed a range of emotions that these women experienced. The women described feeling afraid, anxious, uncertain, fearful, and even despair, particularly those who did not expect the diagnosis or had limited prior exposure to cancer. As seen in P9, who has been diagnosed with two primary cancers.

> *"To be honest, I was afraid because back then (2000) cancer was not that common, not that many people had cancer. There is fear when someone shock you and informs you that you have cancer, and we will remove the breast".* - P9*, BRCA1* PV

In contrast, she shared that upon receiving her ovarian cancer diagnosis in 2024 she did not feel scared or anxious nor did she wonder how the treatment was going to be because she already survived cancer before.

> *"It's fine, I wasn't afraid or got nervous or anything or wonder what chemo is and what it's going to do because I am used to it"*- P9*, BRCA1* PV

P5 also had a previous history of cancer, she had thyroid cancer prior to her breast cancer diagnosis. She showed mixed feelings. Initially there was a sense of acceptance and a desire to confirm her diagnosis in a governmental hospital, then when her diagnosis was confirmed, she cried and wished God had "written this disease in" for her at a much later stage of her life like in her 50s. But at the end she accepted her diagnosis by saying Alhamdulillah (Praise be to God). When her diagnosis was confirmed, her response was,

> *"My brother-in-law was giving me tissues, saying, "Just tears coming down, there's no reaction, nothing. So, I said, Alhamdulillah* (Praise be to God)*. It's just that I don't like hospitals, I don't like anything called waiting'."* She further said: *"I consider myself young, if only God had written for me a little delay, like at the age 50…I was in my forties, just turned 40"* - P5, *PALB2* LPV

Many participants felt despair and reported worrying about their children and grandchildren as a first reaction towards their diagnosis. As noted by P7 who said that her instant reaction was thinking and worrying about who will take care of her children at this very young age while she is sick. She said:

> *"When they told me about the tumour, I immediately thought about my children; I cried, wondering how I would be strong for my children, as they were still very young".*
> - P7*, BRCA2* PV

Breast cancer diagnosis is not just a turning point in the life of the women, but it also affected other family members. Receiving a diagnosis was as much a shock to the family as it was to the woman. One participant described in particular how her diagnosis affected her husband. His reaction was one of shock and disbelief.

"My poor husband was standing, unable to sit". - P5, *PALB2* LPV

These findings align with the existing research on the psychological toll of a breast cancer diagnosis, which has documented high rates of distress, anxiety and depression among newly diagnosed breast cancer patients. These experiences were the same irrespective of age, ethnicity or stage of cancer diagnosis (Ng et al., 2020). Participants expressed feelings of fear about hospitalisation and death, and uncertainty related to the side effects of treatment and concerns about who will take care of their children. These experiences were described in qualitative focused group studies of young adult cancer, with 75% female and 25% male patients. The participants had a variety of cancer types, most had breasts cancer (around 38%), followed by colon cancer (around 13%), and acute myeloid leukaemia (13%). The mean age of the participants was 33 years old (Victorson et al., 2019) and in another qualitative study in Turkey on breast cancer patients, the mean age of participants was 42.11 years (Inan, Günüşen & Üstün, 2016). In both studies the mean ages of participants are similar to the age range of our participants.

The practice of Non-disclosure of Cancer Diagnosis

The practice of non-disclosure of a cancer diagnosis was prevalent, where family members often shield the patient's cancer diagnosis or patients shield their own diagnosis from family members and the community. The practice of non-disclosure has also been observed in the communication of positive genetic test results, as discussed in Section 3.5.3 under Sub-theme 3.3: Family Communication. This is due to reasons such as protecting loved ones from the psychological distress of knowing the cancer diagnosis and avoiding social stress from constant advice, recommendations, and inquiries.

It was found that the cancer diagnosis was kept a secret. Either the woman did not tell others or the family did not tell her during the initial diagnosis. P4 hid the diagnosis from her mother to protect her. Her mother had breast cancer and was undergoing treatment at the same time and in the same hospital.

> *"My mother is here (in the cancer centre) too; we have the same appointments. Imagine today is my appointment, and tomorrow is my mother's appointment. My son will bring her to the appointment, but she does not know that I am sick like her. We had our operation at the same time, side by side in rooms, next to each other, but she didn't know that I had the same illness. I decided not to tell her; she has enough to deal with... I mean, she is old, and I can handle it by myself. So, no one knows except my children and my husband".* - P4*, BRCA 2* PV

However, she did not just keep her diagnosis a secret from her mother but also from her extended family and friends. This was to protect herself from people constantly asking about her health and treatment and giving her advice, she stated,

> *"No one knows about my diagnosis; if they knew, everyone would give me something (advices or treatment remedy), so I didn't talk about it."*- P4*, BRCA2* PV

In contrast to P4, P8's entire neighbourhood knew about her cancer diagnosis resulting in frequent visits which left her with no time for rest.

> *"When I started taking the treatment, we stopped allowing visits because I was exhausted. In our hometown, people visit a lot, and you cannot rest; that's why we stopped allowing visitors"*
> - P8*, BRCA2* PV

P8's response provides clear reasoning why some Omani women like P4 choose not to disclose their cancer diagnosis, especially to their extended family and community. It also highlights the potential social pressures and expectations that can accompany a cancer diagnosis within the Omani community. This finding has been previously reported in a cross-sectional survey study conducted in Saudi Arabia (Alzahrani et al., 2018). The study aimed to investigate the preferences and attitudes of 581 cancer patients and their family members towards the disclosure of a cancer diagnosis. Participants included 304 cancer patients and 277 family members, with breast cancer being the most prevalent cancer type. The study identified several reasons why patients may choose not to disclose their cancer diagnosis, such as cultural norms and expectations, believing that sharing the information would not change the outcome and may only cause unnecessary worry, and that non-disclosure allows patients to avoid constant reminders of their illness and maintain a more positive outlook.

Keeping the diagnosis secret from the participant was also described. P3, who travelled to India for a diagnosis and part of her treatment, described how her family members concealed her diagnosis from her when she was first diagnosed in India. She said,

> *"But truly, they hid from me and told me it was a benign tumour. They even brought the doctor for me, and he (the doctor) told me there was nothing, it was in the early stages, and everything was fine. And I was like, why chemotherapy if it's benign? they never told me until I came back from India in October (that when) I had a translator on my phone, so I translated the papers (the medical reports), and I saw my diagnosis that it was a malignant tumour".* - P3*, BRCA 2* PV

Even though she was suspicious she came to accept what had happened and was even grateful that they intended to protect her in the beginning. She accepted that they wanted to protect her from the psychological burden and later agreed with their decision.

> *"He (her brother) said, and if you knew, what would you do? Alhamdulillah, you have passed a stage; you could have collapsed there in a foreign land (India), and your children are small, and you left them (at home in Oman). And if we told you that it's malignant, that would have increased what you were going through. They saw my situation. That's why they did not share it with me."* She added, *"So, then I said Subhanallah (Glory to God), God knows what is best for me, mashallah (What God has willed, here it used to express appreciation) on them (her siblings)".* - P3*, BRCA 2* PV

This acceptance highlights the cultural acceptance of practices, where family members assume responsibility for managing information and making decisions on behalf of their loved ones, even about health matters. These findings are consistent with previous research on disclosure practices in Oman and other conservative societies. For example, 21 Omani women were interviewed in a study by (Al-Riyami, Abdulhadi and Al-Azri, 2020) and the majority of women reported reluctance to discuss their breast cancer diagnosis with family or friends, among the reasons was to protect the family members from the emotional impact of knowing about their cancer. Only a small number of women felt comfortable opening up about their diagnosis, but even then, they were cautious about providing too many details. Based on the experiences of a practicing radiation oncologist in Oman, (Balushi, 2020), a cancer diagnosis may be concealed not only from older family members, but also from younger adults. The author illuminate that you cannot predicate or assume who is going to be responsible for caring for the ill individual in the family. The same oncologist also noted that patients themselves do not always welcome being informed of their cancer diagnosis.

3.3.3. Sub-Theme 1.3: Treatment Journey

Following a diagnosis, patients begin the treatment journey. All the participants underwent genetic testing during their cancer treatment, which is a process with its own unique medical, physical, emotional, familial, and social challenges. P6 describes this journey as being personal and unique to each patient.,

> *"Discovering, I mean you discover what happens to you along the journey...every patient has their own experience… Even though millions of cancer patients share their experience with others, the emotions that they feel every day and the complications they go through are very unique, meaning every patient has their own experience".* - P6*, BRCA2* PV

Where to Get the Treatment

The majority of participants received their treatment in Oman; however, some reported traveling abroad to undergo specific treatments or procedures. Others sought second opinions or were referred by the government to receive treatments that were unavailable in Oman at the time. Services which were not available in Oman at that time. P9 for example had radiology treatment in India, a service which was not available in Oman in 2000.

"At that time laser (radiology) was not available here in Oman, so they (the government hospital) send me abroad to Mumbai, India for three months for laser (Radiology) treatment". - P9*, BRCA1* PV

Even with medical advancements of healthcare here in Oman, some participants still chose to get treatment abroad like P1 who went to Thailand for her breast operation.

"After I was diagnosed with breast cancer, I travelled to Thailand for the operation"
- P1*, BRCA2* PV

The decision of seeking medical care abroad was not always an easy one to make, as P3 expresses the difficult dilemma she faced regarding treatment abroad. While she was not opposed to the idea of traveling overseas, the impact on her young children and the lack of clear information from the medical professionals in Oman forced here to make this decision.

"I was not against the idea of travelling and getting my treatment abroad, but the thing was that I have two small children, and the other kids are in school in September, it is a three-month long treatment, from 22/06 to 3/09 were the dates. I was not against traveling and getting my treatment, but at the same time here in Oman they didn't tell me anything, so I was between a rock and a hard place"-P3, BRCA2 PV

The above findings show that the main motives for seeking healthcare abroad were for getting certain treatments and procedures, such as radiation therapy, that were not available in Oman at the time, as well as getting a second opinion from physicians at international cancer centres. This was previously reported in a study by (Al Shaaibi and Burney, 2019) which aimed to investigate the prevalence and reasons behind medical tourism among cancer patients in Oman. Sixty-four cancer patients were interviewed, and it was found that medical tourism is quite common among

Omani cancer patients, to seek treatment opinions and obtaining care that was not available locally. However, not all participants felt it necessary to travel abroad for treatment. This is evident in P6's response where she said preferred to have her treatment in Oman even though she was offered sponsored healthcare abroad that. She elaborated,

"Everything was done here in Oman…Even though they gave me the option to go abroad and get treated, sponsored by my institution, I told them that the treatments are available here. I know the doctors, and I trust them; my family's support is the most important thing. Why go overseas? Just accompanied by one member of my family, while here I have all of them, surrounded by all of them"- P6*, BRCA2* PV

Similarly, P9 who are already in treatment at SQCCCRC who had an offer from her husband to take her abroad for treatment, she expressed her trust in the quality of care she's receiving and her decision to continue treatment at the centre in Oman. She commented that,

"My husband told me 'I will not leave you here; I will take you abroad for chemo session.' I said, 'No, the centre is good, and the doctors are good; they reassured you, and I will continue my treatment here.'" - P9*, BRCA1* PV

The decision of where to receive treatment was not always limited to Oman or abroad. Omani patients sometimes had to choose between different hospitals within Oman as well. As P8 stated, she chose to receive her treatment at SQCCCRC. She valued its recognition and reputation as a research centre and its ability to provide better cancer care. She expressed that she does not regret her decision as she felt psychologically at ease while receiving her cancer treatment there.

"The doctor gave me the option of referring me to the (name of governmental) Hospital or the cancer centre (SQCCCRC). Of course, this is better (The centre) even if it is far but better. It is an advanced research centre, so I chose here, and Alhamdulillah (the praise to God), it was a good choice. Psychologically, I mean, when I enter the centre, I feel comfortable; it's different from the other hospitals; you don't feel depressed when entering it. Here, it relieves your stress" -P8*, BRCA2* PV

Participant three also described that she feels comfortable when she enters the centre. Despite having to undergo surgery she had a positive experience and, she did not feel she wanted to leave the centre.

"Alhamdulillah (Praise be to God), you know it was my first time entering a hospital and feeling comfortable in a way I can't imagine, even when I was admitted and had the operation, I told them for the first time I feel like I don't want to leave a hospital" - P3*, BRCA2* PV

Impact of Treatment

Participants discussed the significant physical and emotional side effects they experienced from cancer treatments such as chemotherapy, radiation, and surgery. Many participants highlighted the distressing impact of chemotherapy-induced hair loss, on their self-image and psychological well-being as illustrated by P5 who felt it made her less beautiful.

"I was a bit affected when I lost my hair, it affected me a bit, affected me not in terms of anything, just beauty; I mean, my appearance changed". - P5*, PALB2* LPV

Another woman expressed that losing her hair was the hardest thing she faced during her entire cancer journey, she said,

"If you ask me what the most challenging thing during my cancer journey was, it is losing my hair". - P6*, BRCA2* PV

Participant five reported that the physical changes following her cancer treatment and the prophylactic surgeries which she had due to her positive genetic result caused physiological changes in her body and affected intimacy,

"I mean after the treatment, after chemotherapy, and the prophylactic surgeries, there are many physiological things changed in my body…There is dryness and a lack of desire".
- P5*, PALB2* LPV

The changes in sexual function impacted her relationship with her husband. as he could not comprehend that her treatments could cause sexual dysfunction.

> *"…The problem is that my husband is a bit traditional, He's young, we are the same age, he's just a year older than me, but when you see him, he has old-fashioned ideas. He does not understand that things get changed, he says it's only in your head. There are things I am not comfortable with, and I am having hard time to convince him".* - P5, *PALB2* LPV

Apart from the physical changes which led to concerns about body image and intimacy the women reported being physically and emotionally tired from chemotherapy which led to negatively impacting their psychological well-being and consequently influenced their treatment decisions and family relationships as P7 described:

> *"I was pregnant, and I took two cycles of chemo, and I got tired, I got into a terrible psychological state, and I could not even come to the hospital at all. I refused to take the third chemotherapy session, and I stopped the entire treatment… I was exhausted to the extent that I did not want to come to this hospital. So, Subhan Allah (Glory to God), I stopped the treatment for two years."* - P7, *BRCA2* PV

Participant five also shared that the chemotherapy period was psychologically hard, but in her case, it impacted her relationships with her older children at home. She expressed:

> *"But the chemotherapy period was tiring for me; it tired me at home psychologically with my children. I have older kids; my daughter is in university, and my son has just graduated, so it was tiring for me with them"*- P5, *PALB2* LPV

Participant four experienced an additional layer of distress and anxiety as her child was diagnosed with cancer while she was undergoing her own breast cancer treatment. She shared that before she could complete her radiotherapy sessions, her younger daughter was diagnosed with leukaemia. This meant that she had to navigate her daughter's treatment journey, including hospital admissions and appointments, which made it difficult for her to continue with her own medical appointments. She expressed,

> *"Before I could finish the radiation treatment, my younger daughter got affected.... In the blood, and she's being treated at the Hospital.... It's what made me distressed and anxious. Even my own follow up appointments, I don't feel like going.... when she is admitted in the hospital, I stay with her. Sometimes, I have to leave her to come here for my appointments so, it is a bit difficult.... Very difficult, she's young, I can't leave her alone with anyone".* - P4*, BRCA2* PV

The findings above highlight the significant psychological and physical toll that cancer treatment takes on breast cancer patients. They also reveal the challenges of discussing sensitive topics like sexual health, especially within relationships where one partner may not fully grasp or acknowledge the physical and emotional changes triggered by breast cancer treatment. These challenges profoundly influenced their treatment decisions, family relationships, body image, and sexuality. Similar concerns have been reported in prior local studies by (Al-Riyami, Abdulhadi & Al-Azri, 2020) on breast cancer patients, such as women experiencing physical changes from hair loss and surgeries that led to dissatisfaction with their appearance and a loss of femininity. These changes subsequently impacted their sexual relationships and intimacy. An international study in Ghana involving 12 breast cancer patients aged 15-59 also found that these patients felt physically and emotionally drained from the side effects of treatment (Iddrisu, Asiata & Dedey, 2020).

Some participants also reported limiting their social interactions due to concerns about contracting illnesses from others while their immune systems were compromised from cancer treatments. As P9 explained.

> *"I don't go out, I don't socialise with people…because you know sometime people are sick and I am returning from hospital, I have to look after myself, I don't know what they have, what disease"* - P9*, BRCA1 PV*

One participant noted that her family had become overprotective, restricting visitors even against her wishes. She wanted people to visit and support her. She shared that,

> *"When I was hospitalized, my mother called my sister to say, 'Don't let anyone in,' and my sister did what my mother told her. I told her, now in your WhatsApp status, take a picture or just write 'Alhamdulillah,' or anything that says my sister is in the hospital…I wanted someone to visit me; I'm not cut off from a tree (an expression that means I am not alone, I belong to a family or a tribe); I have family. So, I sent a message to my brothers' (WhatsApp) group, saying, Visits are welcome from this hour to that hour"* - P5, *PALB2* LPV

This participant viewed social interaction as a valuable source of support during her treatment journey, in contrast to P9 who limited social contact for safety reasons. The findings from this study align with a recent investigation conducted at the SQCCCRC, where 11 Omani breast cancer patients aged 24-44 years were interviewed. Some of these participants reported maintaining their social interactions to avoid feeling isolated, while others initially withdrew due to being physically and mentally fatigued from the treatment, which differs from P9's reason of limiting social interaction for safety concerns (Al-Azri et al., 2024).

While patients experience the physical, social and emotional effects of treatment, their families also experience significant challenges. P3 who had to travel abroad to receive a diagnosis also received part of her treatment in India. She reported that her sister and husband described how distressed her children were in her absence, especially the youngest child.

> *"...And he (her husband) said your kids, especially the two little ones, their eyes were looking for you, every time they hear a sound, they look around to find you. And my sister said "one day she came and said (name of her little boy) was looking at her and started to cry, just looked and cried.... so, my sister said that night at 1 AM we were going from house to house, the little one got a very high fever and we took him to the hospital and he was crying, and we were unable to do anything for him."* - P3, *BRCA 2* PV

The above statement shows the significant emotional distress P3's family experienced due to her absence to get treatment abroad.

Traditional Medicine and Cultural Practices

Many participants reported that alongside the conventional medicine they sought treatment from traditional medicine, and cultural practices.

Women reported turning to religious practices involving Quranic treatment, where patients would recite verses from the Holy Quran upon themselves or have others read the verses over them. This is based on the belief that the Quran holds divine power and blessings capable of bringing about physical and spiritual well-being. For instance, P7 shared that she had stopped her conventional medical treatment due to being in a poor psychological state and sought Quranic treatment. Although the tumour actually shrank, the disease had metastasized to her lungs and liver. She described,

> *"During the two years during which I stopped taking the medical treatment, I continued treatment with the Quran. The tumour started to shrink, and I even registered for Quranic treatment courses for cancer. The tumour shrank, and only a small part remained, like the palm of a hand. But Subhanallah, two knots formed here, and these knots grew larger and larger; they did not resolve, and I started experiencing shortness of breath. Because of the breast cancer, they found knots in my liver and lungs"* – P7, *BRCA2* PV

While initially, P7 observed a positive effect from Quranic treatment subsequently, her health declined, and later she was diagnosed with metastasis to liver and lungs, indicating the importance of conventional medical treatment for cancer and not solely depending on Quranic treatment for cancer. The above findings were previously reported in literature among Muslim cancer patients in Malaysia who reported seeking Islamic healing as a complementary treatment to conventional medicine (Suhami, Muhamad & Krauss, 2015). The findings prompt the importance of understanding the cultural significance of such practices, even if they may seem unusual or unpleasant from an outside perspective. Further research could explore the prevalence of such practices, the reasons behind their continued use and perceived benefits, and the potential health consequences.

The participants also described using different herbal remedies during their cancer treatment journey. For instance, P9 described a traditional herbal remedy, *AL-Mughalan*, found in Oman's mountainous regions. She believes this herbal remedy is very beneficial and highlights the use of traditional medicine alongside, or sometimes in place of, conventional treatments. She explains,

> *"This herb is called (AL-Mughalan), it is not available in coastal regions, it's only available in mountain areas, places like Liwa, Shinas, it's abundantly available after rain. So, we take the leaves, and we wash and clean it, then we grind it and put salt on it. Then we soaked in water and rinsed it. Then we put lemon on it to remove the bitterness because it's very bitter. We store it in the fridge and drink one cup in the morning and one cup in the evening (before Maghrib prayer). Alhamdullah (Praise be to God) it is very beneficial."*- P9*, BRCA1* PV

Participant six also described taking herbal remedy provided by her mother. She took it out of respect for her mother's efforts after she made sure that all ingredients are natural. The remedy included turmeric, myrrh (a plant), and other unidentified ingredients, which she took on an empty stomach each morning.

> *"Herbs. I took it because my mother brought it. I did not want to break her; she worked very hard for me, especially when I made sure that all the ingredients were natural and no harm could be caused. So, I took it, and it was very bitter. You cannot imagine how bitter it was… As far as I remember, it contained turmeric, myrrh (a plant), and other unknown ingredients. I had to take it on an empty stomach, and I was taking it every morning."* - P6*, BRCA2* PV

This response highlights the significant influence of family, particularly elderly family members such as mothers, on Omani breast cancer patients' decisions to use traditional remedies and medicines. Patients often feel compelled to honour their family's wishes and use these traditional practices, even when they may have reservations, out of respect and to avoid upsetting their loved ones. The findings show that family dynamics and cultural traditions significantly influence the treatment approaches of Omani breast cancer patients, beyond just individual preferences. It also shows the significant influence of family, particularly elderly family members such as mothers, on the decisions to use traditional remedies and medicines. This was previously reported in Al-Bahri et al.’s (2018), a study on Omani breast cancer women, to evaluate the extent of family involvement in the treatment decision making (TDM). It was found family members played a significant role in the TDM process for Omani women with breast cancer.

In addition to herbal remedies, participants mentioned other traditional practices such as burning the tumour or the skin with hot iron rod (Wasam) and drinking camel urine, some of which may be considered unconventional or even unacceptable by those outside the cultural context. P4 four described one such remedy which is out there in the society and people used as remedy to cure cancer. She said,

> *"It's disgusting, like camel milk mixed with urine. Or camel urine and such… They drink it as a remedy for these diseases (cancers)"* - P4*, BRCA2* PV

Another traditional practice mentioned by most participants is "wasam,", which a traditional cauterisation technique and involves burning the skin or the tumour with a hot iron rod. Participant one noted that people in her community do not refuse modern medicine but some still would go for wasam. She said,

> *"The medical treatments, and like nobody having objection, but you know, like they will go for 'wasam'... they will go. This (wasam) is like usual here, and it is still happening. I can say it's still happening. I heard many of them."*- P1*, BRCA2* PV

This quote suggests that "wasam", continues to be practised within the community, often in conjunction with conventional medical treatments. This was previously reported in Al-Azri et al., (2014) who interviewed 19 Omani women diagnosed with breast cancer, and some participants reported feeling pressured by family or community members to try traditional treatments, including "wasam".

The above quotes show the various traditional remedies and common cultural practices within the Omani community regarding cancer treatment which prompts the importance of understanding their cultural significance, even if they may seem unusual or unpleasant from an outside perspective.

Another participant offered a more balanced perspective regarding this, suggesting integrating religious practices with medical treatment rather than completely substituting conventional medicine with traditional. She stated:

> *"Even if you treat yourself with the Quran and go to a Sheikh (a religious man that reads verses from the Quran on a person in an attempt to heal him), you shouldn't stop the medical treatment".* - P4*, BRCA2* PV

This quote reflects a practical way of acknowledging the potential comfort and support received from religious practices; at the same time, it recognised the importance of conventional medicine in treating breast cancer.

3.4 THEME 2: GENETIC TESTING MOTIVATIONS AND EXPECTATIONS

The cancer diagnosis starts a complex journey as discussed in theme one, it not only includes physical and emotional challenges but also a search for an understanding the origin of the disease. For some, this search leads to genetic testing, particularly when there are indications for a hereditary component. This theme explores patients' motivations and expectations surrounding genetic testing, revealing the diverse perspectives and experiences that shape this stage of these women's journey.

Most participants in this study were unfamiliar with genetic counselling services, and self-referral was uncommon. They typically became aware of these services only after being referred as illustrated by P7's statement when asked about what prompted her to seek genetic counselling. She stated, *"They, the doctors (who referred her). I did not know about the availability of genetic testing until the doctor told me".* - P7, *BRCA2* PV

3.4.1 Sub-Theme 2.1: Rationale for Undergoing Genetic Testing

P8'srationale for undergoing genetic testing was that when a doctor from the genetic department approached her and counselled her on the benefits of genetic testing, it sounded like a good idea, so she decided to proceed with the testing. As P8 stated:

"She came to me, someone from here; I don't know her name, she is from the genetic department here. She counselled me on genetic testing, so I said it's good, and I did the test, that's it".
- P8, *BRCA2* PV

P6, for example, expressed positive feelings regarding access to this service. She expressed gratitude and happiness regarding availability of genetic testing, and she underwent testing to find answers about the cause of her breast cancer. She stated:

"I am actually very happy; when they first told me, they would do the genetic testing for me, I was very happy that we have this service available here…It is better that a person knows than not knowing why they got this disease. I mean, a person would think, why me? Why did I get this disease? What do I have done?"- P6, *BRCA2* PV

Similarly, P1 consented to genetic testing due to the lack of a clear explanation for her young age of breast cancer diagnosis and her awareness of the growing recognition of genetics in understanding disease aetiology. P1 stated:

"The early age of my diagnosis and absence of any clear reason for it, so I consented to the genetic, and I know many of things like we know or we don't know it's going to be explained by genetic at the end in our medical field, most of our diseases". - P1, *BRCA2* PV

The findings suggest that various factors motivate breast cancer patients to pursue genetic testing, such as the desire to understand or find an explanation for the young age of breast cancer diagnosis, awareness of the availability of genetic testing services, and healthcare providers' recommendations. These findings are consistent with a study in the USA where 30 adult cancer patients aged 18-39 years were interviewed. These women reported that they consented to genetic testing to understand the reason behind their young age of onset of cancer. In contrast, others expressed that simply being aware of the availability of the service prompted them to undergo testing (Morand et al., 2022). In another study in the United Kingdom (UK) and the USA, 1322 breast cancer patients were surveyed about the factors that encouraged them to undergo genetic testing; one of the most prevalent reasons was the doctor's recommendation (Scott et al., 2020).

P5 said that she agreed to the genetic testing out of respect, trust, and appreciation for the late Sultan's vision of building the cancer centre (SQCCCRC). She viewed refusing to sign consent forms as being disrespectful to his efforts. She stated:

> *"Yes, it wasn't of my own will; it became my will later (because), honestly, I was signing off on everything; I had no problem with anything related to the hospital... If he (Qaboos, the late Sultan of Oman), may he rest in peace, did all this for us, and I refuse a paper (consent form), that would be very bad of me. I mean, he didn't do any department unless it was in the interest of the people".* - P5*, PALB2* LPV.

Her narrative reveals how cultural context, trust and respect for authority can influence healthcare decisions, even taking priority over initial personal preferences. In Oman in some scenarios, patients willingly delegate high stakes medical decisions to doctors or authorities, trusting their judgment over their own. This is viewed as part of a culturally rooted reallocation of social roles during illness (Al-Azri, 2019). In a study by Coulombe and Laberge, (2024), where the factors that influence patients' decision-making and consenting to genetic testing were investigated, it was found that not only the knowledge provided during the genetic counselling sessions influenced patients' decisions but rather there were other factors such as patients' values and contextual factors that also played a role. Similar findings were reported in a study in a UK focus group with clinical geneticists, and in-depth interviews with patients. They showed that patients' decisions to consent to genetic testing are socially contextualised beyond just information giving (Samuel et al., 2017).

3.4.2 Sub-Theme 2.2 Anticipation of Result

Some participants expected a positive genetic test result. For instance, P6, who had a strong family history of cancer, anticipated a positive result based on her family's history. She stated,

> *"I expected it…The family history, my family tree has a lot of cancer positives, and of course, nobody did the genetic testing because all the cancer in the family happened in days when there was no genetic testing available. But I was feeling that I would be positive."*- P6*, BRCA2* PV

Interestingly, her strong expectation of a positive result led her to proactively inform her family, especially her parents, about the possibility of them being called for genetic testing even before receiving her own test results.

> *"I told my family inshallah (meaning) they would call me for genetic testing result, and after my result, they would call you for genetic testing. I was preparing my family, especially my parents…"*
>
> - P6*, BRCA 2* PV

P3 also expected a positive result based on her family history. However, she expressed that she had not fully grasped the concept of inherited risk or genes at that time.

> *"I have an experience, you know, that my father, may Allah have mercy on him, died of cancer, and my cousin, a young girl of 28, died of the same thing, breast cancer, you know, it's something like you can say, almost expected to be genetic…At the same time, I didn't keep in mind that it could be transmitted to my family or that I or anyone from my family has an inherited gene. As I told you, I wasn't grasping the picture at that time".*
>
> - P3*, BRCA2* PV

P8 shared that she did not expect a genetic cause for her cancer because there was only one isolated cancer in her family. As she stated,

> *"I expected it to be something sporadic that got removed, and that's it; it will not return… I completely excluded the hereditary component because there is no family history, except my maternal cousin had uterine cancer, and she removed her uterus in India"*
>
> - P8*, BRCA2* PV

The current study highlights that some participants expected a positive genetic test result based on their strong family history of cancer. In contrast, others did not expect a genetic link to their cancer diagnosis, particularly if their family history was less significant. This aligns with previous research indicating that the perceived risk of hereditary cancer, based on family history, is a key motivator for pursuing genetic testing based on a suspected genetic link to their disease (Quinlivan, Battikhi & Petersen, 2014; Morand, 2021).

For P2, although she initially believed her breast cancer was due to environmental factors because she didn't have a family history, the extended waiting period made her think she would be positive.

> *"I thought it might be due to environmental factors because no one in my family had it…but I waited a lot until the result came out; I was a little afraid that it would come out positive"*- P2*, BRIP1* LPV

P6 described being told the testing would take a long time and how she coped with the uncertainty by trying to forget about it.

> *"They (the genetic healthcare providers) told me from the beginning that it would take a long time for the test results to come out because they take the test abroad (so at the time of waiting) try to forget about it." "*- P6*, BRCA2* PV

Individual coping strategies may affect how patients manage their emotions during the waiting period. A study of 30 breast and ovarian cancer patients investigating the impact of *BRCA1* and

BRCA2 mutation testing revealed that the waiting period did not provoke anxiety (Hallowell et al., 2002). A mixed-method study in Australia exploring cancer patients' experiences of uncertainty while awaiting genome sequencing results found that patients employed various coping strategies, such as avoidance, to buffer the emotional impact, similar to P6's approach of trying to forget about the genetic test (Bartley et al., 2021).

Overall, the findings of this sub-theme provided insights into the motivations and expectations of patients undergoing genetic counselling and testing. Suggesting that patients' reasons for seeking genetic testing are multi-faceted, involving factors like personal and family histories, recommendations from healthcare providers, and a desire for an answer for the cause of cancer.

3.5 THEME 3: RECEIVING THE POSITIVE RESULT

The lived experience of receiving a diagnosis of cancer, going through treatments, and then realising the risk of hereditary cancer, subsequently led the women to undergo genetic testing which then revealed a positive result. Upon receiving a positive result, it emerged that these women experienced an initial emotional response, followed by immediate concerns about their families also being at risk. They felt compelled to share the results with their family members who were also potentially at risk.

The majority of the women received their genetic test results face-to-face from a genetic professional during a scheduled appointment at SQCCCRC, he only exception was P1, who received her result from a nurse while she was abroad for cancer treatment. She did not specify the method used, such as phone, video call, or email. However, she later expressed dissatisfaction with how her result was communicated and recommended a different approach for other patients. She said,

> *"I was actually abroad...the concerned nurse or the assigned nurse, she communicated with me and she sent the result...It was the way they communicated when they disclosed to me..., so making the experience a little bit different for other individual people."-P1, BRCA2 PV*

While it is not clear what the preference would have been of the current study participants, as this was not explored, it would be interesting to find out in the future what best suits this particular community.

Previous literature supports that face-to-face result disclosure is still the preferred by many genetic healthcare providers and preferred by patients as well, especially when disclosing or receiving positive genetic results. In a study in the USA by Allison et al., (2022) which used a survey-based approach to explore the willingness and preferences for result delivery via telemedicine compared to face to face, they found that despite the increased uptake of telemedicine, many participants about 74.1 % still preferred in-person counselling, because it allowed patients to receive in-person emotional support and directly interact and ask questions with the counsellor. Another study by Beri et al., (2019) in the USA examined patient preferences for in-person versus telephone disclosure of cancer genetic test results. The study found that while telephone disclosure is a viable option for many, a significant portion of patients (18%) preferred in-person communication. The reasons for this preference quoted in the study were in relation to individuals undergoing multi-gene panel testing, having higher anxiety levels and cancer related distress.

Conversely, research by Baumanis et al., (2009), which investigated genetic counsellor and patient satisfaction with telephone disclosure of *BRCA1/2* genetic test results, and it was found that patients were equally satisfied with receiving their results over the phone or in person. Another study by Bradbury et al., (2018), in which researchers at multiple locations randomly assigned participants to either receive their genetic test results via telephone or in person. The goal was to determine if telephone disclosure was no worse than in-person disclosure. The study concluded that telephone disclosure of germline cancer genetic test results is a reasonable alternative to in-person disclosure for interested patients, especially after in-person pretest counseling, which is contrary to P1's experience. It is unclear if P1's experience is unique as her experience is unique in this study and it is difficult to comment further on this. The same research also suggested that allowing patients to choose how they receive their results is preferable, which in turn may increase patient's satisfaction. It would be interesting to ask this question of future individuals undergoing genetic testing and result delivery.

3.5.1 Sub-theme 3.1: Reaction Upon Receiving the Result

Participants described a range of reactions upon learning about their pathogenic genetic variant, including feelings of uncertainty, disbelief, hopelessness, vulnerability, anger, fear, anxiety, and sadness. The emotional responses to a positive result were diverse and intense. P6 described being so emotionally overwhelmed on the day of receiving her result that it initially interfered with her ability to comprehend the information. She said,

"On the day that I received the... I was so emotional my emotions intervened in the result"
- P6*,* *BRCA2* PV

A common emotion that many of these women experienced was feeling sad and they described crying at the time of learning their positive genetic test result. P2 described that receiving a positive genetic result was difficult, especially since it was unexpected news.

"Honestly, it was difficult. I felt sadness and experienced difficult emotions. I didn't expect it to be a genetic condition, and I cried". - P2*,* *BRIP1* LPV

Similarly, P7 also described feeling sad upon receiving her result but her sadness was related to the pattern continuing in her family.

"I was sad because it's hard, to be honest, to be hereditary each time a new diagnosis happens in the family". - P7*,* *BRCA2* PV

P8 shared being shocked upon learning about her *BRCA2* pathogenic variant and the implication for further prophylactic measures due to her elevated recurrence risk and felt it added an additional burden to her journey.

"But of course, I was shocked when I learned it is hereditary. I have BRCA2, and there are other prophylactic measures related to this because my chance of recurrence is higher than ordinary

> *people… It's not easy to be diagnosed with cancer, and in the same year, you learn it's hereditary, and there is a chance for the disease to come back; it's not easy at all".*
> -P8*, BRCA2* PV

These findings highlight the intense psychological burden and distress that accompanies positive genetic test results, especially in the immediate period after disclosure. The available literature suggests that receiving a positive genetic test result for *BRCA1/2* pathogenic variant can elicit a variety of emotional responses in individuals, including shock, anxiety, fear, sadness, uncertainty, and concern about the implications for themselves and their family members Mella et al. (2017) carried out a study aimed at assessing the emotional state of Italian women who underwent genetic testing for *BRCA1/2* pathogenic variant one month after receiving their results. Also in a mixed methods study by (Bradbury et al., 2018), the aim was to understand the quality of life and psychological state of Chinese breast cancer patients who underwent *BRCA1/2* genetic testing. These women reported experiencing negative feelings upon receiving their *BRCA* positive result. The study concluded that genetic testing itself does not cause long-term psychosocial effects, but carriers may experience initial negative emotions upon learning their results.

In contrast to the initial saddened and shocked reactions reported by some participants, others described more neutral or even grateful, hopeful or accepting responses upon receiving their positive genetic test results. For instance, P3 explained that when the counsellor informed her that her cancer was hereditary, she was not shocked or surprised, as she had anticipated the possibility of a hereditary condition. In fact, she responded by saying "Alhamdulillah (Praise be to God)", expressing gratitude. As she recounted:

> *"When (the counsellor) informed me she said, I have unhappy news for you, that the disease is inherited. I mean, I wasn't shocked, and at the same time, as you can say, I wasn't sad or upset… I said, Alhamdulillah. The counsellor asked me, "Weren't you shocked? I said, I was not shocked, I don't know why, is it because I expected it?"*- P3*, BRCA2* PV

Similarly, P5 reported a neutral response to the news of her positive genetic test result for the *PALB2* variant. She reported:

"It was fine, I don't know; maybe because at that time I had just finished chemotherapy, so the feeling was neutral It was neutral". - P5*, PALB2* LPV

The findings suggest that women's emotional experiences upon learning of a positive genetic result can vary, and not all individuals will necessarily have an overwhelmingly negative reaction. This aligns with the literature where Stracke et al., (2022), interviewed 11 women carrying pathogenic variants in *CHEK2, ATM*, or *PALB2* and found that patients receiving positive genetic results experience a diverse range of emotional responses, both positive and negative. In the current study the patients' initial emotional responses also seemed to correlate with their expectations of what the result would be. This was reported in (Brédart et al., 2013) which examined the psychological impact of *BRCA1/2* test results in women with breast cancer, specifically considering their perceived probability of a positive result. They found that the pre-existing beliefs and expectations played a role in how individuals processed and reacted to their genetic information. Additionally, another study explored the role of expectations in shaping the emotional responses of 331 participants without a cancer diagnosis when receiving genetic results related to their cancer risk. The results showed that participants felt relieved when the results matched their expectations, but were surprised and anxious when the results exceeded their expectations (Hilgart et al., 2010).

Beyond the immediate emotional responses, fear of disease recurrence emerged as a significant concern. For instance, P5 vividly expressed that her biggest worry after receiving the positive genetic test result was the increased risk of the disease returning, and she kept thinking about the chemotherapy period, which she did not want to revisit. She expressed:

"The only thing that scared me more was the possibility of it returning. So, what kept crossing my mind was the chemotherapy period". - P5*, PALB2* LPV

In addition to personal concerns, many women expressed worries about passing the gene to their children or felt responsible for their family members who were now also at risk. As with P2, who This was concerned about the possibility of passing the gene to her future children.

"Later on, I started having thoughts like maybe I would pass this on to my children and things like that, which made it difficult". - P2*, BRIP1* LPV

Similarly, P6, a single woman, said her deepest fear after knowing her genetic predisposition was the wellbeing of her family members.

"My biggest fear was for my family, to be honest, in that what if one of them is carrying the gene and, therefore, susceptible to cancer, all types of cancer."- P6*, BRCA2* PV

These findings highlight how a positive genetic test result can cause ongoing concerns about the implications of one's genetic predisposition. This includes fears of disease recurrence and worries about the impact on their families. These concerns are consistent with previous research. A study that aimed to assess the level of fear of cancer recurrence in breast cancer survivors with a *BRCA1/2* mutation found that 70.8% of the 89 women surveyed exhibited significant clinical levels of fear of reoccurrence (Michel et al., 2024). Additionally, a mixed-methods study in the USA explored the impact of *BRCA* status on reproductive decision-making (Hesse-Biber et al., 2022). The researchers found that many women expressed concerns about the possibility of passing on a pathogenic *BRCA* variant to their offspring.

In addition to showing intense emotional reaction upon receiving the result, some of the women also had intense cognitive responses. For example, P1, who is a doctor, initially doubted the accuracy of the test and the laboratory, because she was told during the pre-counselling session that it was going to be a quick test by using a kit. She stated,

"I felt like, is this really true? Always we are questioning ourselves. Is it (the genetic test result) really true? Is the lab they dealt with was accurate, because they told me like a quick sample they are doing there. It was a kit. They have a kit so, they did it. I felt maybe I need to repeat it somewhere else, because with the absence of family history, all these questions would come to my mind…and I started reading, actually, you know, I am a doctor, but I am not in the field of oncology, so I am not reading much about genetic. Maybe I hear about it, but I never been

through what is its consequences and what is the effect of the gene. So, at the time, those few days (post result disclosure). I was continuously reading about its effect". - P1*, BRCA2* PV

The initial denial period can also be related to the unexpected nature of the result by P1 who described being surprised by the positive genetic test result because she did not expect her cancer to be hereditary.

"I was surprised when the doctor told me it was hereditary. It was not expected at all".

- P1*, BRCA2* PV

The above aligns with the first stage of the Kübler-Ross model: denial, which is characterized by disbelief and a refusal to accept the reality of a situation. It is a temporary defense mechanism, leading to gradually process the difficult information (Ali & Bellcross, 2020, pp 83)

Later these women experienced positive or negative emotions for example, P4 who regretted undergoing genetic testing she said,

"If I could go back in time, I would tell them I don't want to do the test. It's better to leave things like that". -P4, BRCA2 PV

In contrast to P4 who regretted undergoing genetic testing, the majority of women in the study described having a more positive perspective on their genetic test results. P1, for instance, described how the knowledge about her hereditary predisposition made difficult decisions such as prophylactic surgeries easier. She stated,

"Knowing about the genetic, let me more accepting whatever subsequent procedures they did for me as prevention. Even for me family members because one of my cousins, the daughter of my aunt who passed away (of cancer), she also removed her ovaries because after she knew that she's also BRCA. So, it makes things more understandable, more absorbable, when you know that there is a reason behind it". - P1*, BRCA* PV

Similarly, P6 expressed the positive impact of genetic testing on her family members who tested positive. She expressed feeling happy and reassured about her father's health in particular.

> *"Look, the most person that I am happy that got tested and he is a carrier is my father, you know he is old, and you know old people are high risk for prostate and such things, and now he is following up with the urinary tract clinic, so I got reassured that my father at this age is under trusted hands being followed up, God forbid if anything happens to the prostate or the urinary tract, they would know"*- P6*, BRAC 2* PV

3.5.2. Sub-theme 3.2. Time Receiving the Result

Participants in this study received their genetic test results at varying stages of their cancer journey, including during active cancer treatment or in the post-treatment remission period. The timing of when individuals received their pathogenic genetic result appeared to influence their emotional responses and ability to cope with this sensitive information.

P4, who received her positive genetic test result at a time when she was about to complete her treatment, shared that she initially felt scared upon being informed that certain other organs were at an increased risk of cancer as well. This was particularly difficult news to receive when she was in a poor psychological state due to the impact of her cancer treatment journey. The fear manifested as hypervigilance, with her constantly questioning and overthinking every physical symptom she got, wondering if it could be a sign of the cancer returning.

> *"To be honest, they scared me. The doctor told me the result is that the disease could come back to me in specific places, and I was just about to complete my treatment and psychologically was little tired. In the middle of the session I went out, I told her I don't want to continue. Two of my kids were with me, and I told them to leave not to stay as well. I left and went to my chemotherapy session…I feel like I'm traumatised…I think about it when I have abdominal pain…in every moment I would think what if I have the disease? That is what makes you overthink, you know".* - P4*, BRCA2* PV

P4's comments highlight a significant factor that may influence patients' reactions upon receiving their pathogenic genetic test result which was the timing of receiving the result. As P6 illustrated that she found it beneficial to receive her *BRCA2* positive result after undergoing chemotherapy and adapting to her breast cancer diagnosis. She even suggested that this approach might be a better strategy for other patients as well. She said,

> *"I also told the doctor that it was good that you did the genetic test for me after I have adapted to the disease and after I understood the disease, and after I took the chemotherapy and went through all the side effects of the chemotherapy. So, the genetic result was a little bit light on me, okay I was shocked that I am positive for BRCA2, but the intensity of the shock was less when I thought about it and compared whether I would want to receive it before or after treatment I prefer that, I even told her that you should do the genetic test for patient after they went through chemo and the side effects of it".*- P6*, BRCA2* PV

In contrast, P3 expressed regret about the timing of her genetic test results. She said if she had known she was *BRCA2* positive before her surgery, she would have gone for a double mastectomy (removal of both breasts) as her doctors would have recommended because why undergo two separate surgeries if she could have done one. She said,

> *"It was after 13th of May, because on 13th of May was the operation, they (the doctors) told me if they had known earlier, they would have removed both breasts together, One operation instead of two separate ones… If it's beneficial for me, why not have one operation instead of two".* -P3, *BRCA2* PV

Receiving distressing genetic news while one is already physically and emotionally tired from cancer treatment can cause negative response upon result disclosure. This was reported in previous research by Brédart et al. (2013) where 243 breast cancer patients were surveyed, and it was found that women who received their genetic results during active cancer treatment showed higher levels of anxiety compared to survivors who had completed their treatment.

The timing of receiving genetic test results can also impact some patients' decision-making about their current treatment plan and their willingness to undergo risk-reducing surgeries. This is consistent with previous literature. In a study of 287 women with *BRCA1/2* or *PALB2* gene mutations, researchers found that women who were aware of their genetic status before breast cancer surgery were nearly nine times more likely to choose a double mastectomy during their initial surgery, compared to those who did not know their genetic status prior to surgery (Apostolova et al., 2024). Similarly, a study of 1209 newly diagnosed breast cancer patients found that nearly half of the participants did not receive their *BRCA* test results before undergoing surgery, which hindered their ability to make fully informed decisions about their surgical treatment options (Armstrong et al., 2021).

As shown, it is important to acknowledge that there may not be a universally "good" or "optimal" time to disclose positive genetic test results, regardless of the patient's stage of life or cancer journey. Each individual is unique, with different needs and readiness to cope with this information. Therefore, it is crucial to have open conversations with patients about the timing and their readiness to receive the genetic test result, preferably during a pre-counselling consultation to allow for proper preparation and exploration of the most suitable approach to support the patient in managing the impact of a positive genetic finding.

3.5.3 Sub-theme 3.3: Family Communication

This sub-theme explores the dynamics of sharing genetic information within families, the reasoning behind informing family members, and the uptake of genetic testing by family members.

Participants felt a strong sense of responsibility to inform their at-risk relatives about their positive genetic results. For instance, P8 shared that she had communicated her positive genetic result to her siblings, children, and husband, as her parents were no longer alive. She felt a sense of responsibility to inform them.

"Of course, I have to tell them I told them to get tested, I texted them on WhatsApp because this is a responsibility, and I have to tell them whether to test or not it is something that concerns them"- P8*, BRCA2* PV

P5 also shared her positive genetic test result with her siblings, as she believed it would be beneficial for them to also undergo genetic testing. To facilitate this, she provided them with the necessary contact information to arrange their own testing appointments.

"I said it was a good thing, and I would inform my siblings to do the testing. I gave them the contact number, and they arranged their own appointments for genetic testing"- P7*, BRCA2* PV

P2 explained that she informed her parents about her positive genetic test result and encouraged them to undergo genetic testing to know from which parent this gene was inherited. This was to help them understand their own risks and facilitate early detection and active management of any potential health concerns. Her statement reflects this,

"First of all, my husband was with me, so he knew about the result. Then I informed my parents so that they could undergo the test as well, in case the condition came from either my mother or my father. It was better for them to get tested, and if there was anything, God forbid, it would be known from the beginning"- P2*, BRIP1* LPV

While many women felt a responsibility to share their genetic test results with family members, some took a more selective approach to disclosure, often influenced by concerns about the emotional well-being of their relatives. For instance, P5 chose to inform her older paternal cousins about her positive genetic result but withheld this information from her mother due to concerns for her mother's health.

I didn't tell my mom, but I informed my older paternal cousins. My mom was present (at the family gathering in which she informed her cousins), but I didn't tell her… My mom has diabetes and now she's starting to have high blood pressure. My mom is the kind who gets very scared, even of a fever. Just the thought of illness makes her anxious, so you see her always in the

> *hospital, it's like her second home. She goes to the hospital for anything; she regularly does her check-ups continuously from a long time ago, even before I had any illnesses.*"- P5*, PALB2* LPV

P4 also selectively informed certain members of her family about the genetic test results. She only told her children, as she expected the rest of her family members would refuse to get tested even if she encouraged them to do so. As she explained,

> *Only my children. The others don't know…the problem is that they won't accept it, even if I tell them I have done a test, you go get tested as well, most of the family members won't agree to get tested.*"- P4, *BRCA2* PV

Participants also adopted various strategies for sharing their genetic test results with their families. While some directly informed their relatives, others enlisted the help of family members to coordinate the dissemination of the information. For instance, after receiving her positive *BRCA2* result, P1 informed her older brother and entrusted him with the responsibility of notifying other family members and arranging predictive genetic testing appointments for them. She said,

> *"The one who was most important was my elderly brother, because we took it from there. I handed the issue to him, to whom to talk first. So, he was like arranging for everybody the genetics appointment"*- P1, *BRCA2* PV

The findings show that all women shared their genetic test results with at least one family member, for example, parents, siblings, children, and cousins. The results were mostly shared with first-degree relatives, with some women also informing their second-degree relatives like their close cousins. The findings are consistent with previous research, such as a cross-sectional study by Abdel-Razeq et al., (2023) of adult breast cancer patients with confirmed positive breast cancer susceptibility gene variants treated at a cancer centre in Jordan. That study explored how these patients communicated their genetic test results within their families across generations and assessed their attitudes towards genetic testing. The study found that while most participants shared their results with first-degree relatives, communication with more distant relatives was less common. Another study by Fukuzaki et al., (2021), which investigated the extent to which

Japanese cancer patients share their germline genetic test results, found that all participants shared their results with at least one relative, with sharing rates of 85.7% for first-degree relatives and 10% for second-degree relatives. The sharing rate with first-degree relatives was comparable to rates in Europe and the USA. In a study on 252 Italian women, it was found that probands (individuals who undergo genetic testing) most frequently shared results with first-degree relatives, particularly siblings and children. It also highlighted that pre-existing family relationships and communication patterns played a significant role in whether women disclosed their HBOC genetic test results (Pietro et al., 2020). In a study that examined how 149 Black women with breast cancer aged 50 or younger shared their *BRCA1/2* genetic test results with family members, it showed that the majority (77%) disclosed their results to at least one family member, mostly a first degree relative. They also found that those who tested positive were significantly less likely to disclose results to their daughters to protect them from psychological distress (Conley et al., 2020).

The above findings show that among many factors regarding motivation to share their genetic test result to their family members, one is related to feeling a sense of responsibility to protect high risk family members. This is consistent with a previous study by Dean et al. (2021) where 62 women were interviewed to explore their motivations and challenges of sharing their positive genetic test results with family members. The study found that motivators for sharing results included a desire to enable family members to take steps to protect their health, as well as a sense of moral obligation to inform at-risk family members and to empower them to understand their cancer risk.

The challenges of disclosing results in this study however, included concerns for family members' reactions and the potential emotional impact of the result on them. The practice of non-disclosure, particularly towards older people such as parents, is common in Oman and other Middle Eastern countries. In this study it was mixed some women have informed their parents and some did not inform them. This was previously reported by a study in Saudi Arabia, which found that the practice of non-disclosure of health information to parents might be a widely accepted practice in some parts of the Middle East. The non-disclosure of sensitive health information to older people, such as parents, is often done with the intention of protecting them from emotional distress (Alzahrani et al., 2018).

A study in the United Arab Emirates involving 52 residents and recent graduates in nine focus groups revealed that non-disclosure of medical information in serious illnesses is common in UAE hospitals. Key barriers identified were family objections, deficits in medical training, and inconsistently implemented institutional guidelines (Ibrahim & Harhara, 2022).

3.5.4 Sub-Theme 3.4: Uptake of Genetic Testing by Family Members

When genetic test results were shared, participants reported varying levels of uptake of genetic testing among their family members. Some described a high degree of engagement, with most of their first-degree relatives who were eligible for testing undergoing predictive genetic counselling and testing. For instance, P3 explained that most of her siblings had already taken the test, and the remaining few were planning to do so.

> *"Same thing with my siblings, they accepted this and most of them came for testing, only three of them left, will come after Eid to do the test. Even my mom, she was not convinced to do the test then I said that we are not saying you have this for 100 percent, and that it's only a study. After that she got convinced and did the test, and all my siblings got the test".* - P3, *BRCA2* PV

P3, however, faced difficulties in convincing her husband, who is also her first degree relative from the side of the family with a cancer history, to undergo genetic testing.

> "*I told my husband to go and get the test because he is my relative. He said tell them my husband passed away. I told him no they want you... He said no, I swear I will not go. I told him you go and get tested and at the end everything is in the hands of God, at least you would know. He is an over-thinker".* - P3, *BRCA2* PV

P9 shared that one of her daughter's already got tested and received her result which was negative. The other two daughters recently got tested and were still waiting for their genetic test result. She said,

> *"They tested (daughter's name), and yesterday they tested two of my daughters. Now the girls are waiting for their result. One of the girls' results is out and they said she does not have the mutation, all is good".* - P9*, BRCA1* PV

In contrast, other participants reported limited interest in testing among their family members. For example, P4 reported that only two of her siblings had the genetic test, on the same day of her result disclosure but that the rest were not interested. P4 stated:

> *"They (her son and her daughter) continued the (her result disclosure) session and did the genetic testing. My sister, who lives in (name of the region), is the only one who said she wanted to be tested; the rest refused".* - P4 *BRCA2* PV

While P2 reported that none of her family members came for predictive genetic testing for the identified variant. Similarly, P8 said her family members did not want genetic testing in the fear that they would overthink the risks for the rest of their life. Fortunately, they at least chose regular screening which include mammograms.

> *"But they (my sisters) said they didn't want to be tested. They only had the mammogram because they didn't want to overthink it their whole lives".* - P8*, BRCA2* PV

The above shows that the uptake of genetic testing among family members of positive genetic result is highly variable, with some families demonstrating a high degree of testing uptake and others showing low uptake. These findings were previously reported in literatures of family members' uptake of genetic testing. Low uptake of predictive testing has been shown by where one of the aims was to explore the uptake of predictive testing by family members of a group of 87 Asian (Chinese, Indian, Malay) breast and ovarian cancer patients with *BRCA1/2* pathogenic variants. It was found that 62.0% of eligible first-degree and second-degree relatives were notified by the proband about the test result, while only 11.5% of eligible first-degree and second-degree relatives underwent genetic testing. Additionally, first-degree relatives were more likely to have been informed and tested compared to second-degree relatives, and sisters were more likely to have been tested compared to brothers. Additionally, a study in Turkey by Seven et al., (2022) examined how women with *BRCA1/2* mutations share their genetic risk information with family members. The study involved 92 women and their 417 first-degree relatives, and found that almost all probands (94.5%) informed at least one relative about their positive result, but that only 19.9% of eligible relatives pursued genetic testing.

3.6 THEME 4: ADAPTING TO HEREDITARY BREAST CANCER DIAGNOSIS

Eventually, the women developed coping strategies and made adjustments as they adapted to their hereditary breast cancer diagnosis and positive genetic test result. They had to navigate the complex emotional, psychological, and lifestyle changes that came with this life-altering information. Because the cancer diagnosis is an integral part of their journey in receiving a positive genetic result, the adaptation process cannot be viewed separately. However, there are certain adjustments that are specifically related to the genetic result. This theme will explore these strategies.

3.6.1. Sub-Theme 4.1: Active Coping

Participants reported various emotional-based coping strategies when faced with their cancer and positive genetic test results. For example, P2 explained that she has not thought much about her positive genetic test result for a long time. She has left the matter in God's hands and no longer dwells on it.

"Honestly, I haven't thought about the result in a long time... I left it to God. I don't think about it, honestly". - P2, *PRAB1* LPV

P2s response indicate that she is using selectivity and religious coping strategies to cope with her positive genetic result as was previously reported in literature on Omani women which reported that Omani women most often use religious and spiritual coping strategies when faced with cancer diagnosis (Al-Riyami, Abdulhadi & Al-Azri, 2020).

Some participants made significant lifestyle changes to manage the stress and emotional burden associated with their hereditary breast cancer diagnosis. These changes often involved prioritising self-care, reducing stress, and focusing on overall well-being. For example, P1 decided to retire from her career and create a less stressful environment and a slower-paced lifestyle. She said,

> *“I decided to also to retire in order to take my time. You know, our work is stressful being a doctor. So, like to take my time in going to a gym, take the life in a rest mode slowly”.*
> - P1*, BRCA2* PV

Similarly, P2 reported adopting an active lifestyle through exercising and eating healthily to improve her quality of life.

> *"It has improved the quality of my life through exercise and healthy eating, I have been exercising more, I joined a gym. I wanted to protect myself from diseases and improve my fitness”. –* P2*, BRIP1* LPV

Others, like P3, who had just completed treatment explained that she is planning to prioritise her well-being as she used to deprive herself of many things. She reported,

> *“Like I told you, taking care of myself, going out with my friends. I used to deprive myself of many things”. -* P3*, BRCA2* PV

These lifestyle changes demonstrate a problem-focused coping approach, where participants actively take steps to manage the stress and health risks associated with their hereditary cancer diagnosis (Stracke et al., 2022) By prioritizing self-care, reducing stress, and improving overall well-being, the participant took concrete actions to mitigate the challenges they face. This aligns with research (Stracke et al., 2022) highlighting the importance of adopting healthy behaviours and lifestyle modifications as effective coping strategies for individuals dealing with hereditary cancer syndromes.

The majority of participants shared that they have regular medical follow-up post receiving their positive genetic result. P5 for example, emphasized her commitment to ongoing management.

"I even was traveling and cut my trip short to come back quickly for the appointments (follow ups). I didn't want to delay them, and it didn't occur to me to postpone them".
- P5, *PALB2* LPV

P1 described being placed in a screening programme with a gynaecologist and ultimately undergoing a prophylactic total hysterectomy.

"They (the doctors) put me, of course on screening for the ovarian with the gynae. So, I 've been with them and then eventually I ended up doing oophorectomy and hysterectomy as a prophylactic". -P1, BRCA2 PV

P8, who was interviewed during her admission for prophylactic oophorectomy surgery, shared that although she was not 100% convinced to undergo surgery, her persuasion and her desire to avoid reliving the distress of previous cancer treatment and chemotherapy were key factors in her decision-making process.

"I am not 100% convinced, 90%, and also my husband convinced me to do the surgery; it is better than being anxious, and I don't want to go back to the same suffering that I have been through during the treatment, during chemotherapy". - P8, *BRCA2* PV

The above responses demonstrate the proactive steps that many participants took to manage their health following a positive genetic test result. This included regular medical follow-ups and preventative surgeries. Such active problem-focused coping strategies have been widely recognized in the literature as effective means of managing the distress and health risks associated with a hereditary cancer syndrome. A German study that involved interviews with 11 women who carry a pathogenic variant in moderate risk genes reported that participants were in surveillance programmes and were willing to undergo (risk-reducing) surgeries (Stracke et al., 2022). Similarly, in a study in Ireland to understand the lived experiences and coping strategies of individuals with a *BRCA1/2* variant, interviewed 18 participants and it was found that some participants underwent risk-reducing mastectomies and/or risk-reducing salpingo-oophorectomies. The study highlights the emotional impact of these decisions, with some women expressing relief and a sense of control, while others experienced anxiety and uncertainty (Warner and Groarke, 2023).

P6 reported a proactive approach to managing her emotional well-being by seeking psychological support from a professional psychologist throughout her journey, including after receiving her genetic test result,

"I am fine because all the emotions that I am talking about now; I have already spoken to her (the psychologist) about them. I also met her after receiving my genetic result". - P6*, BRCA2* PV

This highlights the important role of professional psychological support in helping individuals cope with the emotional challenges associated with hereditary breast cancer. Consistent with the findings of Warner & Groarke, (2023), individuals with a *BRCA1/2* PV or LPV require specialized psychological support to assist them in navigating their situation.

3.6.2. Sub-Theme 4.2: Engagement Coping

The support from family members also proved to be a significant source of emotional strength and resilience among these women. P9 clearly expressed the enduring impact of her family's support. She expressed,

"My husband, my children's (both daughters and sons) They all supported me in a way that I will never forget, and they are still supporting me". - P9*, BRCA1* PV

Similar P2 shared that even before receiving the genetic test result, her husband was incredibly supportive, and he stood by her throughout the entire treatment process. She said,

> *"Well, even before the test result, he was supportive, and he stayed with me throughout the treatment process. Even after we found out it was hereditary, nothing changed. If anything, it grew stronger."*- P2*, PRIP1 LPV*

The above findings indicate that the support from family members appears to be a critical factor that helped the women cope with the challenges of a hereditary cancer diagnosis. This finding aligns with broader research emphasising the importance of social and family support in coping with hereditary cancer syndromes. For example, a study that interviewed 13 women with hereditary cancers, including breast and ovarian cancer, found that families are the primary source of support, enhancing individuals' well-being and resilience (Silva et al., 2022). Similarly, a study by Al-Azri, et al. (2014) on Omani breast cancer patients identified that Omani women use various coping strategies to deal with their cancer diagnosis, including avoidance, spiritual and religious beliefs and practices, as well as family support,. This is confirmed by another study on Omani breast cancer patients by Al-Riyami, Abdulhadi and Al-Azri, (2020) which found that Omani breast cancer patients cope through cultural, religious and family resources.

When P6 learned that one of her friends also had a pathogenic variant in *BRCA1*, and her friend's risk was higher than her own, she felt a sense of shared experience and reframed her own situation. She expressed that they (referring to her fiend) support each other because they were going through a similar situation. She said:

> *"After I knew that I was BRCA2 positive, one of my friends told me that she was BRCA1, and when she shared her experience of genetic testing, I felt like, oh, there are people out there whose risk is higher than me; her mother passed away of cancer, so we support each other".*
>
> \- P6*, BRCA2* PV

The above indicates that connecting with others facing similar challenges can be comforting. Hearing about the experiences of others, especially those facing greater difficulties, can provide valuable perspective and help individuals view their own situation in a new light. Previous research supports this idea. For instance, a study in the USA used cross-sectional data from two ongoing cohort studies of gynaecologic and breast cancer survivors at the University of Minnesota, finding that patients who knew someone in a similar life situation felt a strong sense of connection and community (Jewett et al., 2022). Additionally, a study that interviewed 22 patients with hereditary cancer syndromes, including hereditary breast and ovarian cancer, from 10 European countries, reported that connecting with others in patient support groups provided a sense of validation and reassurance. Participants felt more confident in the information they received, believing it was vetted by knowledgeable individuals. Most importantly, these groups offered a space to feel understood and connect with others who shared their concerns (Serna, Lluch-Canut & Fernández-Ortega, 2024).

3.6.3. Sub-theme 4.3: Meaning Focused Coping

The majority of the women reported finding comfort and meaning in their spiritual or religious beliefs when confronting a hereditary cancer diagnosis and positive genetic test result. For instance, P8 accepted her fate and destiny when faced with the genetic test result. She expressed that resisting the reality would be useless, and instead emphasized the importance of adapting to her circumstances.

> *"What can I do if I don't accept this result? Humans should accept what is in their fate and destiny and try to adapt to it."*- P8*,* *BRCA2* PV

For some participants, spiritual coping involved accepting their genetic test result as part of a divine plan. As a result, P3 was not overtly anxious, as she believed there were medical options available, and that God would continue to support her. As she expressed,

> *"The reassurance is there because it is God's will. Why am I the only one among my siblings? Why couldn't it be one of my siblings? Why me if it was genetics? I mean it's God's will the Almighty, and I don't get anxious about it, like this will happen, that would not happen. There is a prophylactic option, and the Lord Almighty is here, you know".* - P3*,* *BRCA2* PV

P6 also found comfort and meaning in her religious beliefs, which helped her to accept the diagnosis and genetic test result and its implications for her health. She believed that everything is part of His divine plan. She said,

> *"Everything is from God in the first place; I believe everything is from God and that God chooses everything for humans, whether it is bad in their point of view or good. It is God's provision; everything is a provision from God; my belief in this helped a lot with me accepting my cancer diagnosis".* - P6*, BRCA2* PV

These findings suggest that religion and spirituality serve as pivotal coping mechanisms for Omani women grappling with breast cancer and hereditary genetic test results. As previously mentioned, previous studies on Omani breast cancer patients, such as those by Al-Azri et al., (2014) and Al-Riyami, Abdulhadi and Al-Azri, (2020), have similarly observed that religious beliefs and practices are among the most prevalent and powerful coping strategies employed by these women.

CHAPTER 4: CONCLUSION, RECCOMENDATIONS AND LIMITATIONS

4.1. CONCLUSION

This research explored the lived experiences of Omani women navigating a positive pathogenic germline genetic test result for hereditary breast cancer, focusing on four key themes: cancer journey, genetic testing motivations and expectations, receiving the positive result, and adapting to a hereditary breast cancer diagnosis.

While this study has effectively addressed the research question and objectives which were to investigate these women's reasons for seeking testing, their reactions to receiving a positive result, how they use this information in their lives, and how they communicate the results to their families. The research also considers the influence of cultural practices surrounding cancer diagnoses and genetic testing. Due to the fact that genetic testing and receiving positive result were part of the cancer journey, it added a new layer of meaning to their diagnosis, connecting their cancer experience to potential hereditary implications. Therefore, throughout the interviews the women often focused on their cancer experience rather than solely discussing the genetic aspect as per the research focus and question. The women's cancer journey was detailed so extensively that it became a major theme in this study. The finding suggests that the cancer journey holds such importance for these women that it became intertwined with their experience of receiving a positive genetic result. It is not a surprise that women who had gone through the genetic counselling process and who were then interviewed about their experience of this by a genetic counselling colleague would talk a lot about their physical, emotional, psychological, social, and spiritual aspects of life before, during and after cancer.

The findings revealed that perceived beliefs about the breast cancer causes, which often linked with cultural and familial narratives, shaped women's overall experience. The responses to the positive genetic test result, ranged from anxiety and fear to relief and empowerment. Coping mechanisms varied, including active coping like leaving the matter to Allah (God), increased surveillance, risk-reducing surgeries and lifestyle changes, engagement coping which includes seeking emotional support from family and friends and meaning-focused coping often rooted in religious and spiritual beliefs. Furthermore, family dynamics and cultural norms played an important role in disclosure practices, with concerns about protecting family members from the psychological distress, influencing decisions about information sharing.

This study directly addresses the research question of how Omani women experience and make sense of a positive germline genetic test result, highlighting the complex interplay of personal, familial, cultural, and healthcare factors that shape this experience. These findings have significant implications for healthcare professionals, genetic counsellors, and policymakers. They underscore the need for culturally sensitive and individualized support for women navigating hereditary cancer risk, emphasizing the importance of addressing not only the medical aspects but also the emotional, social, and cultural dimensions of this experience. The study highlights the important role that genetic counsellors can play in managing patient expectations and emotions throughout the testing process. Patients may have different levels of understanding about genetics and the potential implications of test results, understanding the need for thorough pre-test counselling. Genetic Counsellors are ideally placed to attend to the medical, psychological, familial can cultural aspects the women with a risk of an inherited breast cancer syndrome is faced with.

The findings also highlight the need for accessible and comprehensive genetic counselling services that address patients' pre-existing beliefs, provide tailored support throughout the testing and decision-making process, and acknowledge the influence of family dynamics and cultural norms on disclosure practices. Furthermore, the study emphasizes the importance of providing high-quality cancer care within Oman to improve the overall treatment experience and outcomes for Omani patients, which in turn can positively influence their experience of receiving and adapting to a positive germline genetic result.

4.2 LIMITATIONS

This study acknowledges several limitations. The sample size was relatively small and focused on a specific cultural context, which may limit how well the findings apply to other populations.

However, this study provided valuable information for health care professionals in the management of women at risk of an inherited breast cancer syndrome in Oman, and possibly other Arabic populations.

This study was the first qualitative research conducted by the researcher; therefore, the data collection and analysis might be not to standard. During interviews researcher might have framed the interview questions in a way that encouraged them to discuss their cancer experiences more extensively

It is acknowledged that the study relied on self-reported data collected through interviews, which can be affected by recall bias. Participants may have remembered events or experiences differently from how they actually occurred, or they may have presented themselves in a more positive way. As a qualitative study, the interpretation of data is inherently subjective.

The researcher tried to ensure rigour through using established qualitative analysis methods. The research was aware that her own perspectives and experiences could have had an influence on the identification and interpretation of codes and themes, however caution was taken by involving the supervisors in an independent data analysis and reaching agreement on the final themes. Finally, time constraints during data collection and analysis may have limited the depth of exploration into certain aspects of the women's experiences.

4.3 RECOMMENDATIONS

Further future research could explore the cultural beliefs and practices related to cancer causes and treatment, including why they are still used, their perceived benefits, and any health effects. Future studies could also look at the long-term psychological and social impacts of a positive genetic test result, assess different support programmes, and explore how healthcare providers can help with informed decision-making. We also need to better understand the cultural nuances around hereditary cancer risk and sharing information.

Note: Grammarly (Writing assistant software tool) and professional editing assistance were used while writing this dissertation to refine language, check meaning, and grammar.

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APPENDICES

Appendix A: Semi-Structured Interview Guide

The questions will explore in depth the breast cancer patients' experience with receiving pathogenic germline genetic results in a single centre in Oman.

1. Tell me about the day you got your genetic result / Tell me about your experience when you first received your genetic result.

2. What were your reasoning and expectations for undergoing genetic testing?

3. What does a positive genetic result mean to you?

4. How did you use the genetic result?

5. Did you communicate your genetic result with your family? With whom? And why?

6. Is there a specific practice/belief regarding cancer diagnosis in your family/ culture?

Appendix B: Telephonic Script

Good morning. This is (the coordinator's name); I am calling from Sultan Qaboos

Comprehensive Cancer Care and Research Centre. I am calling to inform you about research taking place in the centre. This research aims to study Omani Breast Cancer Patients' experience of receiving pathogenic germline genetic results. The information we get from this study will be used to improve future services provided by the centre. Also, it can be used as a work base for other researchers.

Participation in this study is entirely voluntary, and if you do not participate, it will not affect the health services you receive from SQCCCRC.

If you are interested in participating, the researcher will contact you to provide additional information and details about the research.

Appendix C: Consent Form

Consent form

Breast cancer patients' experience with receiving pathogenic germline genetic results: single centre experience in Oman

I, -- agree to participate in this study.

I give permission to the researcher to access my Hospital Information System (HIS) personal file, to access the demographical data, the hospital appointments and the medical documents.

I understand the purpose of the study is to explore breast cancer patients’ experience of receiving a pathogenic genetic result.

I understand that my participation in this study is completely voluntary, and I can withdraw from the study at any time without facing any discrimination against me, and it will not affect the health services I am receiving from Sultan Qaboos Comprehensive Cancer Care and Research Centre (SQCCCRC).

I understand that I have the choice to refuse to answer any questions that may be uncomfortable to me without any consequences.

I understand the interview will be audio-recorded for research purposes and stored in the researcher’s password-protected device. And will be deleted after the research is completed.

I understand that all information will remain confidential, only be accessed by the researcher, used for research purposes only, and will be discarded when the research is completed.

I understand that I might not receive direct benefits from this study and that there will not be any financial gain from participating in this study.

I understand that part of my interview will be quoted in the dissertation and presentation posters.

Note: If it is identified that the participants require further discussion or counselling regarding their diagnosis, they will be referred to the relevant services.

Participant first name, surname : --
Participant signature: ---

Participant contact details: phone number: --------------------------- email: ------------------------
Researcher signature: -- date: ------------------------------

Appendix D: Information Sheet for Research Participants

Dear Participants,

You are invited to participate in a study investigating breast cancer patients’ experience of receiving pathogenic genetic results in Oman.

This study will be part of a master’s thesis in genetic counselling at UCT. The study has ethics clearness from both the University of Cape Town (UCT) and Sultan Qaboos Comprehensive Cancer Care and Research Centre (SQCCCRC).

This study will help genetic health professionals to understand patients’ experiences according to patients’ perspectives to improve future health services and care and to be more aware of patients' experiences during service delivery.

The study will involve you being interviewed by me. The interviews will be conducted at Sultan Qaboos Comprehensive Cancer Care and Research Centre (SQCCCRC).. It is estimated that the interview will take about 1-1:30 hours. The interviews will be audio-recorded and stored in the researcher's password protected device; only the researcher can access it. The obtained information will be appropriately discarded after the study is completed.

Participation in this study is voluntary, and participants may withdraw from the study at any time without any consequences. You will not receive direct benefits from participating in this study, but the research will assist in improving the service provided

For any further inquiries.

Please contact Ms. Amira AL Balushi at +96891997073 or albami001@myuct.ac.za or A/Prof. Tina-Marié Wessels at Tina.wessels@uct.ac.za or Dr. Chantel van Wyk at c.wyk@cccrc.gov.om

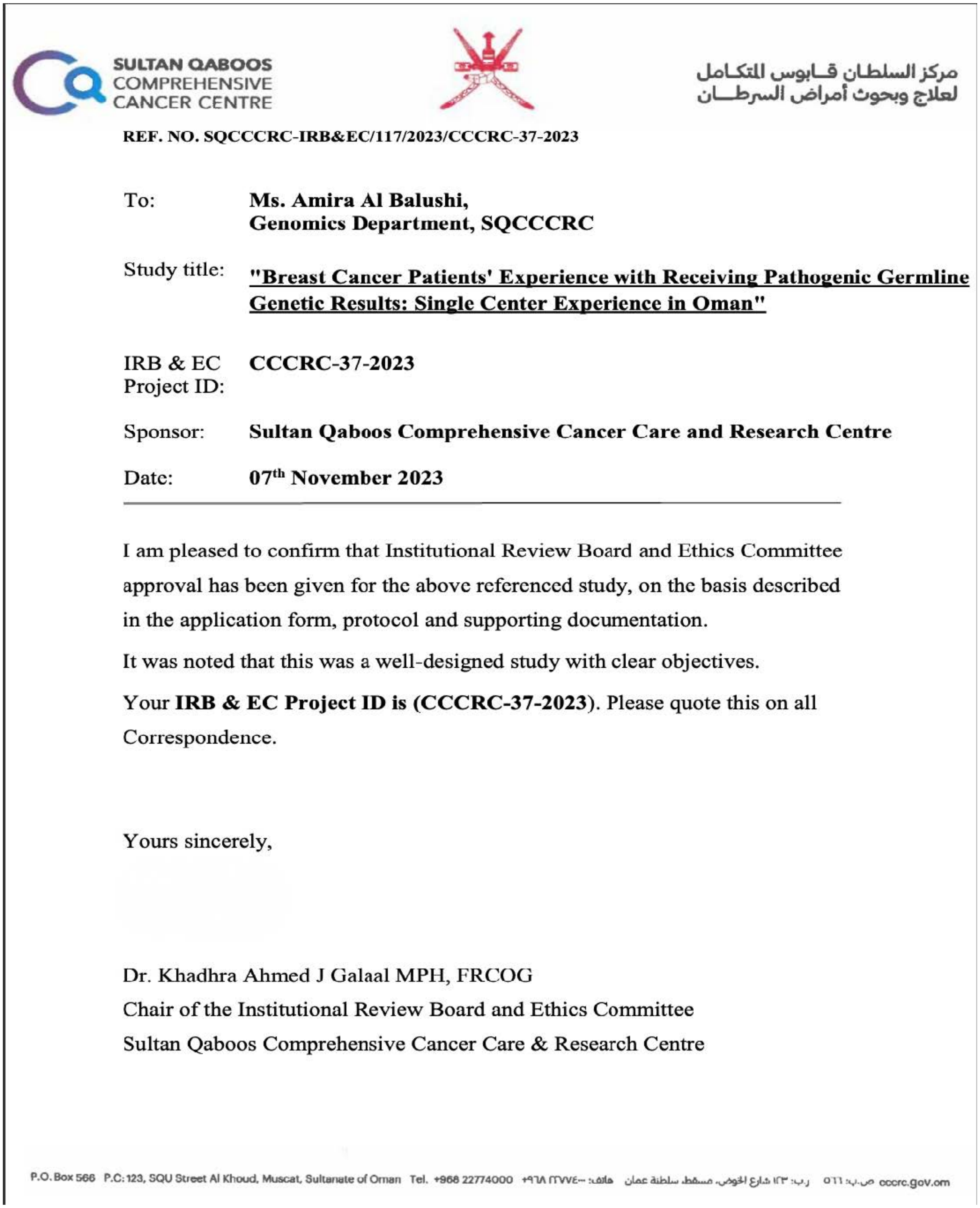
or Prof Marc Blockman mac.blockman@uct.ac.za

Kindly note that a consent form must be signed before the interview. Please note the attached consent form.

Kind Regards,

Amira AL Balushi

Appendix E: SQCCCRC Ethical Approval



SULTAN QABOOS
COMPREHENSIVE
CANCER CENTRE

مركز السلطان قابوس المتكامل
لعلاج وبحوث أمراض السرطان

REF. NO. SQCCCRC-IRB&EC/117/2023/CCCRC-37-2023

To:	**Ms. Amira Al Balushi,** **Genomics Department, SQCCCRC**
Study title:	**"Breast Cancer Patients' Experience with Receiving Pathogenic Germline Genetic Results: Single Center Experience in Oman"**
IRB & EC Project ID:	**CCCRC-37-2023**
Sponsor:	**Sultan Qaboos Comprehensive Cancer Care and Research Centre**
Date:	**07th November 2023**

I am pleased to confirm that Institutional Review Board and Ethics Committee approval has been given for the above referenced study, on the basis described in the application form, protocol and supporting documentation.

It was noted that this was a well-designed study with clear objectives.

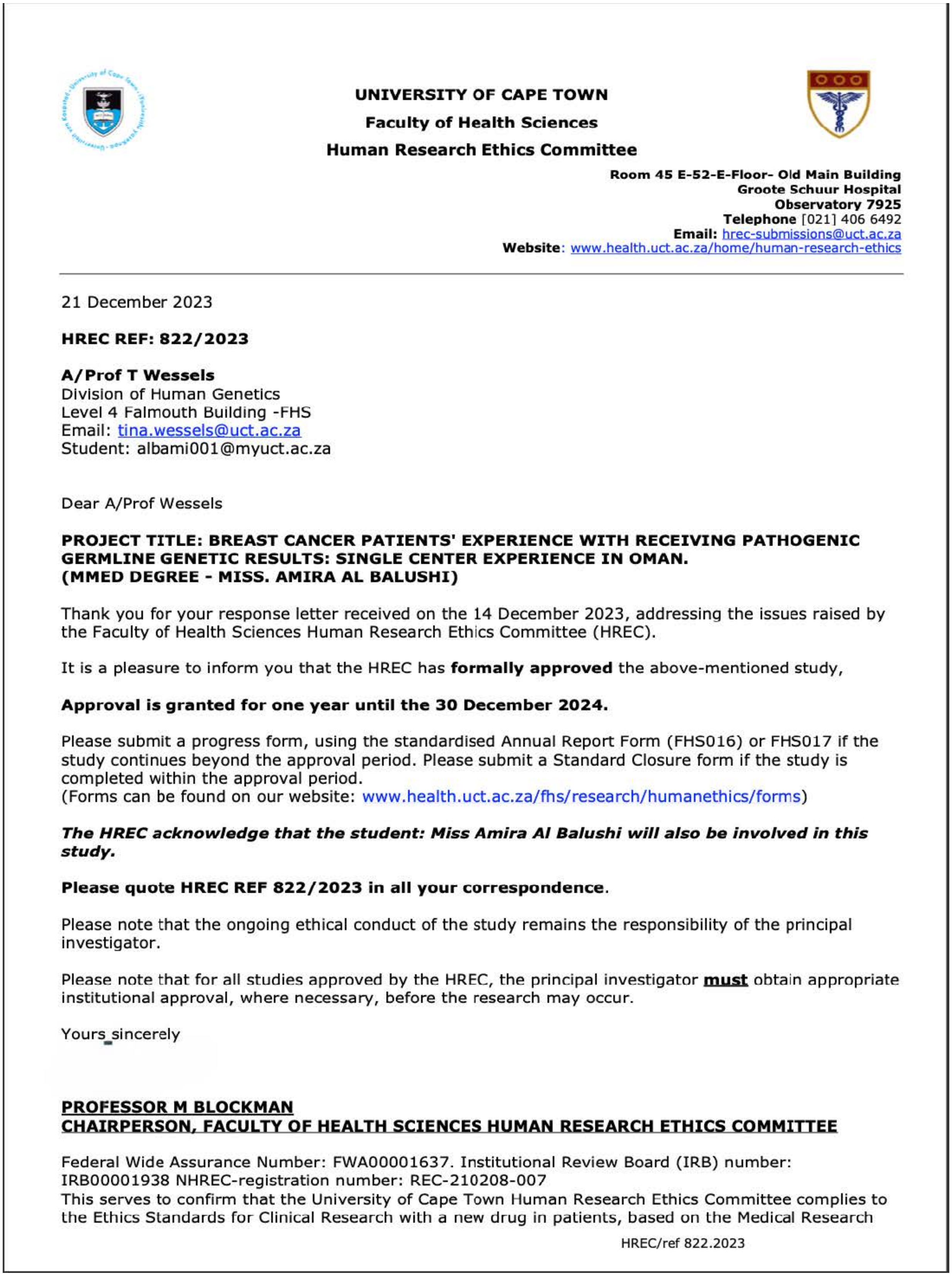
Your **IRB & EC Project ID is (CCCRC-37-2023)**. Please quote this on all Correspondence.

Yours sincerely,

Dr. Khadhra Ahmed J Galaal MPH, FRCOG
Chair of the Institutional Review Board and Ethics Committee
Sultan Qaboos Comprehensive Cancer Care & Research Centre

P.O. Box 566 P.C: 123, SQU Street Al Khoud, Muscat, Sultanate of Oman Tel. +968 22774000 هاتف: ‎+٩٦٨ ٢٢٧٧٤٠٠٠ سلطنة عمان مسقط، شارع الخوض، ر.ب: ١٢٣ ص.ب: ٥٦٦ cccrc.gov.om

Appendix F: HREC Approval



UNIVERSITY OF CAPE TOWN
Faculty of Health Sciences
Human Research Ethics Committee

Room 45 E-52-E-Floor- Old Main Building
Groote Schuur Hospital
Observatory 7925
Telephone [021] 406 6492
Email: hrec-submissions@uct.ac.za
Website: www.health.uct.ac.za/home/human-research-ethics

21 December 2023

HREC REF: 822/2023

A/Prof T Wessels
Division of Human Genetics
Level 4 Falmouth Building -FHS
Email: tina.wessels@uct.ac.za
Student: albami001@myuct.ac.za

Dear A/Prof Wessels

PROJECT TITLE: BREAST CANCER PATIENTS' EXPERIENCE WITH RECEIVING PATHOGENIC GERMLINE GENETIC RESULTS: SINGLE CENTER EXPERIENCE IN OMAN.
(MMED DEGREE - MISS. AMIRA AL BALUSHI)

Thank you for your response letter received on the 14 December 2023, addressing the issues raised by the Faculty of Health Sciences Human Research Ethics Committee (HREC).

It is a pleasure to inform you that the HREC has **formally approved** the above-mentioned study,

Approval is granted for one year until the 30 December 2024.

Please submit a progress form, using the standardised Annual Report Form (FHS016) or FHS017 if the study continues beyond the approval period. Please submit a Standard Closure form if the study is completed within the approval period.
(Forms can be found on our website: www.health.uct.ac.za/fhs/research/humanethics/forms)

The HREC acknowledge that the student: Miss Amira Al Balushi will also be involved in this study.

Please quote HREC REF 822/2023 in all your correspondence.

Please note that the ongoing ethical conduct of the study remains the responsibility of the principal investigator.

Please note that for all studies approved by the HREC, the principal investigator **must** obtain appropriate institutional approval, where necessary, before the research may occur.

Yours sincerely

PROFESSOR M BLOCKMAN
CHAIRPERSON, FACULTY OF HEALTH SCIENCES HUMAN RESEARCH ETHICS COMMITTEE

Federal Wide Assurance Number: FWA00001637. Institutional Review Board (IRB) number: IRB00001938 NHREC-registration number: REC-210208-007
This serves to confirm that the University of Cape Town Human Research Ethics Committee complies to the Ethics Standards for Clinical Research with a new drug in patients, based on the Medical Research

Council (MRC-SA), Food and Drug Administration (FDA-USA), International Council for Harmonisation of Technical Requirements for Pharmaceuticals for Human Use: Good Clinical Practice (ICH GCP), South African Good Clinical Practice Guidelines (DoH 2020), based on the Association of the British Pharmaceutical Industry Guidelines (ABPI), and Declaration of Helsinki (2013) guidelines. The Human Research Ethics Committee granting this approval is in compliance with the ICH Harmonised Tripartite Guidelines E6: Note for Guidance on Good Clinical Practice (CPMP/ICH/135/95) and FDA Code Federal Regulation Part 50, 56 and 312.