PATHWAY FOR IMPLEMENTATION OF HEREDITARY BREAST CANCER SCREENING

by

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Dedication

This thesis is dedicated first and foremost to my loving parents, whose unwavering support, sacrifices, and boundless encouragement have been the foundation of my journey. Your integrity, diligence, and work ethic have shaped my character and guided me through every challenge.

To my wonderful daughters, who fill my life with joy, meaning, perspective and purpose, you are both my greatest inspiration and my constant reminder of what truly matters.

Finally, this work is dedicated with deep respect, compassion and solidarity to all Cancer warriors, who face each day with extraordinary courage, the families who stand beside them with unwavering love and strength, and the healthcare professionals who devote their lives to healing, often under challenging circumstances but with limitless dedication.

Though this study represents only a small step toward the fight against cancer, it is offered with the sincere hope that it may, in some measure, contribute to the shared mission of improving outcomes and bringing comfort to those in need. Their unparalleled courage, extraordinary resilience, and unwavering hope give true purpose to this work.

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Most importantly, I owe my deepest gratitude to my family for their unwavering patience, understanding, and encouragement. Their faith in me has been my greatest source of strength and motivation. Finally, I dedicate this work to all those committed to advancing early detection and prevention of cancers, for their efforts continue to inspire meaningful change in patient care and medical research.

ABSTRACT

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Hereditary breast cancer, by and large associated with genetic mutations usually inherited, like BRCA1 and BRCA2, is an emerging yet poorly met challenge within the Indian healthcare system. With advances in genetic testing and preventive oncology, the implementation of hereditary breast cancer screening continues to remain fragmented on account of low awareness, variable clinical practices, a lack of access to genetic services, and ethical and psychosocial barriers. The objective of this study is to formulate a structured and actionable pathway for the implementation of screening for hereditary breast cancer in India.

The study employed a mixed-methods study design that enabled the combination of findings from quantitative data obtained from a survey and in-depth perspectives gleaned from experts in this subject matter domain. The data analysis involved statistical testing for relationship outcomes in awareness, communication from physicians, fear barriers, and screening uptake. Case studies from international concepts helped shape a best practice approach.

From the results, there is clinical evidence supporting hereditary screening, especially for high-risk individuals; however, its uptake is very low among patients. The factors that significantly influenced patients to undergo hereditary screening include recommendations by physicians, fear of results, cost limitations, lack of standardized guidelines, and lack of infrastructure to provide psychological support to patients. Misuse of genetic information is a major ethical issue restricting its acceptance.

The study concludes that hereditary breast cancer screening in India can be substantially improved through a structured implementation pathway integrating physician-led awareness, standardized referral protocols, expanded access to genetic counseling, and robust ethical safeguards. The proposed pathway provides practical guidance for healthcare providers, policymakers, and insurers, positioning hereditary breast cancer screening as a scalable and impactful preventive strategy aligned with precision medicine and public health equity objectives.

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CHAPTER I:

INTRODUCTION

1.1 Introduction

Breast cancer remains one of the most prevalent forms of cancer globally, with significant mortality rates. While advancements in treatment have improved survival rates, early detection remains a critical factor in improving outcomes. Hereditary breast cancer, linked to mutations in genes such as BRCA1 and BRCA2, presents a particular challenge due to its inheritable nature and increased risk. The motivation behind this research is to outline a pathway for the implementation of hereditary breast cancer screening programs, aiming to enhance early detection, risk assessment, and personalized treatment strategies (Tung et al., 2016).

The importance of this research cannot be overstated in the context of industry practice and knowledge advancement. By establishing a structured pathway for implementing hereditary breast cancer screening, healthcare providers can better identify individuals at elevated risk, facilitating timely interventions such as increased surveillance, risk-reducing surgeries, or targeted therapies. Moreover, this research contributes to advancing knowledge by synthesizing existing literature, identifying gaps, and proposing a comprehensive approach to address the complexities of implementing screening programs in real-world settings (Kurian et al., 2017).

1.2 Research Problem

Despite significant advancements in genetic research and clinical practice, the adoption and standardization of hereditary breast cancer screening in India remain limited.

Previous research by Tung et al. (2016) and Domchek et al. (2013) in hereditary breast cancer screening has laid a foundation for understanding the genetic basis of the disease and its implications for clinical practice. Studies by Kurian et al. (2017a), and Tung et al. (2016) have demonstrated the efficacy of genetic testing in identifying high-risk individuals and guiding preventive measures. However, existing approaches often lack standardization and face barriers to widespread adoption, such as cost, accessibility, and patient adherence. Gaps consistently identified in the literature mentioned below include a lack of awareness among patients and clinicians, disparities in clinical practice, accessibility issues with respect to genetic testing and counseling services, along with ethical, financial, and infrastructural barriers. These challenges cumulatively impede the early detection and effective management of hereditary breast cancer. Nevertheless, prior research demonstrates several important strengths. Studies have successfully identified individuals at high genetic risk through the detection of pathogenic mutations, enabling personalized risk assessment and targeted clinical management (Kurian et al., 2017a). Research has also established the efficacy of risk-reducing strategies, such as prophylactic mastectomy and chemoprevention, in significantly lowering cancer incidence among high-risk individuals (Kurian et al., 2017a). Furthermore, advancements in genetic testing technologies, particularly the adoption of next-generation sequencing, have improved the efficiency and cost-effectiveness of genetic testing, thereby expanding access to hereditary cancer screening services (Kurian et al., 2017a).

Nevertheless, with these advancements, the previous study also has some major flaws. First, the screening programs may not be generally available, especially where there are less privileged groups and those in areas with less accessible healthcare, thus presenting major discrepancies in the distribution of genetic care access (Metcalfe et al., 2019). Second, the lack of consistent screening protocols, variable interpretation of genetic screening, and variable management recommendations across different health care centers present major issues concerning the universality and quality of the care provided (Metcalfe et al., 2019). Third, genetic screening for hereditary breast cancer also has serious psychosocial aspects, with the potential to present anxiety, uncertainty, and stigma to individuals and families, thus calling for the provision of counseling and psychosocial support systems in conjunction with the screening programs.

1.3 Purpose of Research

While significant progress has been made in understanding hereditary breast cancer, gaps persist in translating research findings into clinical practice. This research addresses these gaps by proposing a structured pathway to implement hereditary breast cancer screening. By synthesizing existing evidence, identifying challenges, and proposing practical solutions, this study seeks to enhance the uptake and effectiveness of screening programs, ultimately improving patient outcomes. From the gaps identified, the following hypothesis is proposed for this study:

Hypothesis - Limited awareness, inconsistent clinical practices, and systemic barriers together reduce the adoption and effective implementation of hereditary breast cancer screening in India.

This hypothesis is based on the assumption that improving awareness, access, ethical safeguards, and multidisciplinary collaboration will significantly enhance uptake and clinical effectiveness.

1.4 Significance of the Study

The significance of this study lies in its potential to bridge the gap between advances in genetic science and the realities of healthcare delivery in India. Breast cancer is the most common cancer among Indian women, and a considerable proportion of cases are linked to hereditary mutations such as BRCA1 and BRCA2. While genetic testing has proven clinical value in reducing morbidity and mortality through early detection and preventive strategies, its adoption in India remains limited due to financial, infrastructural, and psychosocial barriers. By examining these challenges and proposing a structured pathway for implementation, this study contributes to improving patient outcomes, empowering families through cascade testing, and reducing the national burden of breast cancer. The research also has important implications for public health equity, as it highlights the need to extend access to underserved and rural populations who currently face the greatest obstacles in accessing genetic services. Furthermore, by drawing lessons from international models and contextualizing them for India, this study offers actionable insights for policymakers, healthcare providers, and insurers to develop standardized guidelines, sustainable funding mechanisms, and ethical frameworks that ensure responsible and equitable delivery of hereditary breast cancer screening. Ultimately, this research underscores the vital role of integrating precision medicine into public health strategies and positions hereditary breast cancer screening as a transformative tool in advancing preventive oncology in India.

1.5 Conceptual Framework for the Study

To guide the research, a conceptual framework has been developed outlining the relationship between core determinants of successful hereditary breast cancer screening implementation.

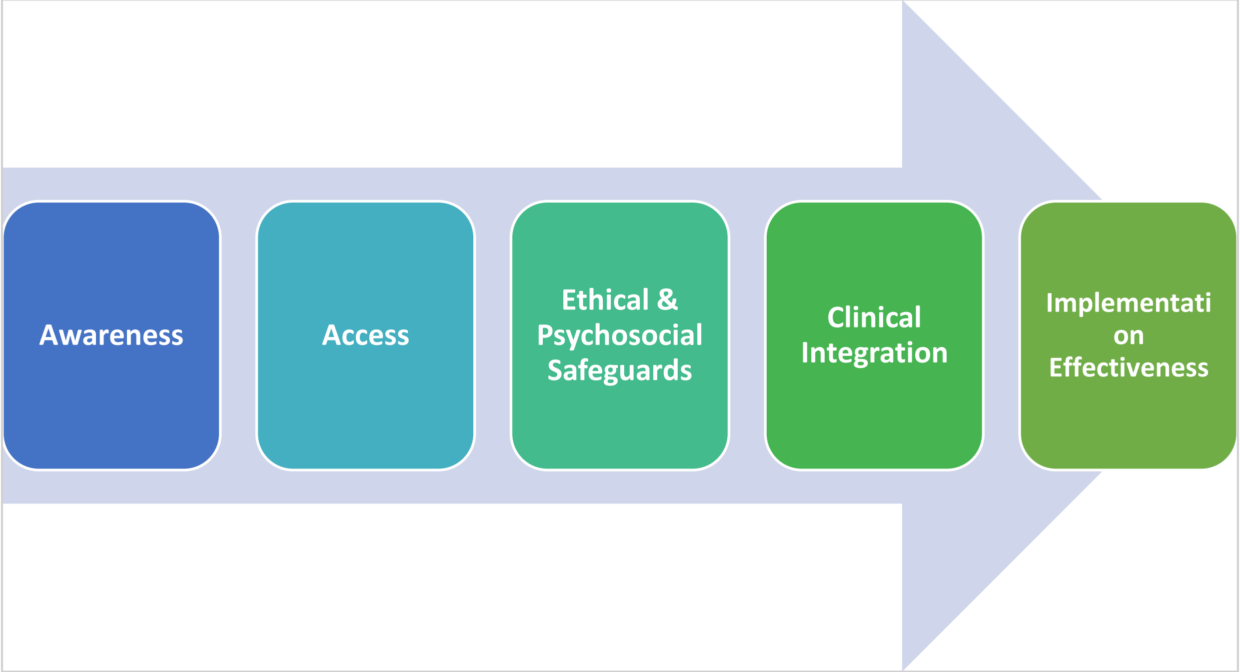


Figure 1.1: Conceptual Pathway (Author's Work)

Awareness: Level of understanding among patients, families, and physicians regarding hereditary cancer risk and the role of genetic testing.

Access: Availability and affordability of genetic testing, counselling services, insurance support, and trained professionals.

Ethical & Psychosocial Safeguards: Measures ensuring privacy, informed consent, data protection, emotional preparedness, and cultural sensitivity.

Clinical Integration: Adoption of standardized guidelines, streamlined referral pathways, and incorporation of genetic results into clinical decision-making.

Implementation Effectiveness: The extent to which hereditary screening programs achieve improved uptake, quality of care, and patient outcomes.

1.6 Research Purpose and Questions

This research aims to answer the following questions:

1.6.1 Should hereditary cancer screening tests be implemented for individuals at elevated risk?

1.6.2 What level of risk does an individual face if there is a history of cancer in their family, and to what extent across generations?

1.6.3 What function/role does the primary doctor/physician serve in recommending hereditary cancer screening tests?

1.6.4 What obstacles exist in the execution of the Hereditary Cancer Screening Pathway?

1.6.5 What are the primary reasons behind the limited acceptance of hereditary screening among patients and families?

1.6.6 What factors or constraints might prevent physicians from prescribing hereditary cancer screening tests?

CHAPTER II:

REVIEW OF LITERATURE

Breast cancer is one of the most prevalent malignancies worldwide, with a significant portion attributable to hereditary factors (Apostolou and Fostira, 2013). Hereditary breast cancer screening holds immense promise in early detection and prevention. This literature review explores the main ideas, theories, and concepts surrounding the implementation of hereditary breast cancer screening. It delves into areas of agreement and disagreement within the literature, highlighting existing problems and gaps, and proposes strategies for addressing them.

2.1 Introduction

Breast cancer remains a formidable global health challenge, affecting millions of individuals annually and presenting significant burdens on healthcare systems worldwide (Oyetoran et al, 2024). While advancements in screening, diagnosis, and treatment have improved outcomes, a subset of breast cancer cases is attributable to hereditary factors. Hereditary breast cancer, characterized by mutations in genes such as BRCA1 and BRCA2, represents a critical area of focus for early detection and intervention strategies. As our understanding of the genetic basis of breast cancer has deepened, so has the recognition of the importance of hereditary breast cancer screening in identifying at-risk individuals and implementing tailored prevention measures.

Recent trends and perspectives in hereditary breast cancer screening underscore the evolving landscape of precision medicine and personalized healthcare. One notable trend is the increasing utilization of multi-gene panel testing in genetic screening protocols.

Traditionally, screening focused on BRCA1 and BRCA2 genes; however, technological advancements now allow for simultaneous testing of numerous genes associated with hereditary breast cancer, expanding the scope of genetic analysis and potentially identifying additional risk factors (Kurian et al., 2017a). This shift towards comprehensive genetic testing reflects a broader trend in oncology towards precision medicine approaches, where treatment decisions are increasingly guided by an individual's genetic profile.

Furthermore, the integration of artificial intelligence (AI) and machine learning algorithms into genetic screening processes represents a promising frontier in hereditary breast cancer detection. AI-based tools can analyze vast datasets, including genetic variants, clinical outcomes, and population demographics, to identify patterns and risk factors associated with hereditary breast cancer (Gastounioti et al., 2022). By harnessing the power of AI, healthcare providers can enhance the accuracy and efficiency of genetic screening, ultimately improving risk stratification and informing personalized prevention strategies.

However, alongside these advancements come challenges related to the equitable implementation of hereditary breast cancer screening. Disparities in access to genetic testing and counseling services persist, particularly among underserved and marginalized populations. Socioeconomic factors, including income, education, and geographic location, influence individuals' ability to undergo genetic testing and access preventive interventions (Laduca et al., 2014). Addressing these disparities requires a multifaceted approach, encompassing policy interventions, healthcare infrastructure improvements, and community outreach initiatives aimed at promoting equitable access to genetic services.

In light of these trends and challenges, while international research mentioned in this chapter has advanced screening and risk-stratification models, their applicability to the Indian context remains uncertain due to demographic, cultural, and infrastructural differences. Most existing reviews describe advances in hereditary screening but do not critically evaluate how socioeconomic disparities, low awareness, or limited genomic infrastructure influence implementation, particularly in Low and Middle-Income Countries (LMICs). This chapter bridges that gap through a critical analysis of empirical studies, theoretical frameworks, and global models.

2.2 Main Ideas, Theories, and Concepts

2.2.1 Genetic Basis of Hereditary Breast Cancer

Hereditary breast cancer is primarily associated with mutations in high-penetrance genes such as BRCA1 and BRCA2. These genes play crucial roles in DNA repair mechanisms, and their dysfunction leads to an increased risk of breast and ovarian cancers (Miki et al., 1994). Understanding the genetic basis of hereditary breast cancer forms the foundation for screening strategies aimed at identifying at-risk individuals. Beyond BRCA1/2, emerging research identifies other genes, such as TP53, PTEN, and PALB2, contributing to hereditary breast cancer susceptibility (Kuchenbaecker et al., 2017). Understanding the spectrum of genes implicated in hereditary breast cancer broadens screening strategies and facilitates personalized risk assessment.

2.2.2 Screening Modalities

Several screening modalities are utilized for hereditary breast cancer detection, including genetic testing, family history assessment, and risk prediction models. Genetic testing, particularly next-generation sequencing technologies, enables comprehensive analysis of predisposing genetic mutations (Domchek et al, 2013). Family history assessment helps identify individuals with familial clustering of breast cancer, guiding the selection of candidates for genetic testing. Risk prediction models incorporate various factors, including genetic, lifestyle, and reproductive factors, to estimate an individual's likelihood of developing breast cancer (Daly et al, 2023). Advancements in genomic technologies revolutionize hereditary breast cancer screening, offering enhanced sensitivity and efficiency (Olopade et al, 2008). Next-generation sequencing enables comprehensive analysis of multiple genes simultaneously, expanding beyond targeted BRCA1/2 testing (Domchek et al, 2013). Panel testing incorporates a broader range of genes associated with breast cancer predisposition, improving the detection of pathogenic variants and enabling tailored risk management strategies (Tung et al., 2016). Additionally, innovative approaches such as polygenic risk scores integrate multiple genetic variants to provide a more nuanced assessment of individual breast cancer risk, complementing traditional risk prediction models (Moorthie et al 2022).

2.2.3 Testing and Family Communication

Cascade testing represents a cornerstone of hereditary breast cancer screening, capitalizing on familial relationships to identify individuals at increased risk (Offit et al, 2020). Proactive identification of pathogenic variants in index cases facilitates targeted testing of at-risk relatives, optimizing resource allocation and enhancing detection rates (Bradshaw et al, 2024). However, effective cascade testing relies on comprehensive family history documentation and robust communication channels within families (Bradshaw et al, 2024). Culturally sensitive approaches are essential to navigate familial dynamics and facilitate informed decision-making among relatives, particularly in diverse cultural contexts (Usher-Smith et al, 2023).

2.2.4 Multidisciplinary Care and Patient Counseling

The successful implementation of hereditary breast cancer screening hinges on a multidisciplinary care framework encompassing genetic counselors, oncologists, surgeons, and primary care providers (Wright et al, 2019). Genetic counseling assumes a pivotal role in facilitating informed decision-making and psychosocial support throughout the screening process. Patient-centered counseling empowers individuals to navigate complex risk information, weigh screening options, and make personalized risk management decisions (Armstrong et al., 2015). Moreover, collaborative care models foster seamless integration of genetic testing results into clinical management plans, ensuring holistic patient care.

2.2.5 Ethical and Legal Considerations

As genetic testing becomes increasingly integrated into clinical practice, ethical and legal considerations surrounding hereditary breast cancer screening demand scrutiny (Ri and Muto, 2021). Ensuring patient autonomy, confidentiality, and non-discrimination is paramount in the provision of genetic services (Ri and Muto, 2021). Adequate protection of genetic information from misuse or unauthorized access safeguards individual privacy rights and fosters trust in healthcare systems (Klitzman et al., 2013). Furthermore, equitable access to genetic testing and preventive interventions is essential to mitigate disparities and promote health equity among diverse populations.

2.3 Critical Review of Key Concepts and Evidence

2.3.1 Genetic Basis of Hereditary Breast Cancer

Although many widely cited foundational studies, such as Miki et al. (1994) and Kuchenbaecker et al. (2017), have been used to establish high-penetrance genes, most depend on either Western or European cohorts. This biases generalizability because the prevalence of variants differs significantly across ethnic groups. For instance, Kurian et al. (2017a) show strong predictive value for BRCA1/2 but admit a lack of representation from Asian populations, thus limiting relevance for Indian population-level screening.

Critical Insight - The lack of large-scale Indian genomic datasets limits both risk modeling and policy formulation; this emphasizes that an effective population-specific implementation strategy is required.

2.3.2 Screening Modalities and Their Limitations

NGS and multigene panel testing have increased detection capabilities; however, Daly et al. (2023) warn that increased identification of Variants of Uncertain Significance (VUS) may potentially lead to increased confusion and medical anxiety. This is particularly concerning in areas where the resources for genetic counseling are limited.

Example of Critique - Though Domchek et al. (2013) showed the cost-effectiveness of NGS in high-risk groups, the economic evaluation used strong insurance support conditions not reflected in India’s predominantly out-of-pocket system.

Implication - Adoption of multigene panels needs to be accompanied by robust counselling and clear guidelines to avoid misinterpretation of risk.

2.3.3 Cascade Testing and Family Communication

Cascade testing is widely supported (Offit et al., 2020; Manchanda et al., 2017), and Kurian et al. (2017b) validate its efficiency in identifying at-risk relatives. However, Kurian's cohort is heavily Western; thus, uptake patterns cannot be directly extrapolated to cultures where discussing hereditary risk is stigmatized.

Critical Point - Family communication about cancer in India is largely determined by cultural norms, gender dynamics, and the fear of discrimination. Thus, models of cascade testing developed in the West require adaptation for Indian sociocultural realities.

2.3.4 Multidisciplinary Care and Integration Challenges

Although Wright et al. (2019) and Armstrong et al. (2015) emphasize the multidisciplinary integration of care, their models assume structured referral systems and electronic health records resources that are inconsistent across Indian healthcare facilities.

Critical Evaluation - These studies present ideal conditions; however, they do not take into consideration practical limitations such as physician workload, limited genetic counsellors, and variable conditions between private and public health facilities.

2.3.5 Ethical, Legal, and Psychosocial Dimensions

These genetic privacy concerns, though well-documented, by Klitzman et al. (2013) and Bombard et al. (2019), seldom tackle concerns specific to LMICs, including lack of regulatory oversight, data protection frameworks, or policies against insurance discrimination.

Gap Identified - Most of the studies assume strong genetic data governance, which India lacks today; its local ethical frameworks become, hence, inevitable for safe adoption.

2.4 Areas of Agreement and Disagreement related to Hereditary Breast Cancer Screening

2.4.1 Agreement related to Hereditary Breast Cancer Screening

2.4.1.1 *Early Detection and Intervention* - There is broad consensus on the benefits of early detection through hereditary breast cancer screening. Identifying individuals with pathogenic mutations allows for targeted surveillance and risk-reduction strategies, leading to improved outcomes (Kurian et al, 2017b).

2.4.1.2 *Cascade Testing Effectiveness* - The effectiveness of cascade testing in identifying at-risk individuals within affected families is widely acknowledged (Offit et al, 2020). Sequential testing of relatives of index cases with known pathogenic variants facilitates the early identification of individuals who may benefit from preventive interventions (Manchanda et al., 2017).

2.4.1.3 *Multidisciplinary Care Importance* - Multidisciplinary care involving genetic counselors, oncologists, and primary care providers is universally recognized as essential for the successful implementation of hereditary breast cancer screening programs. Collaboration among healthcare professionals ensures comprehensive patient care, including accurate risk assessment, genetic counseling, and appropriate follow-up (Kurian et al, 2019).

2.4.2 Disagreement related to Hereditary Breast Cancer Screening

2.4.2.1 *Screening Guidelines* - There is ongoing debate regarding the optimal age and frequency of screening for individuals with pathogenic mutations (Myers et al, 2015). While some advocate for earlier and more frequent screening due to elevated cancer risk, others question the evidence supporting such recommendations and emphasize the potential harms of over-screening, including increased psychological distress and unnecessary interventions (Daly et al., 2021).

2.4.2.2 *Inclusion of Additional Genes* - The inclusion of additional genes beyond BRCA1/2 in screening panels remains a subject of disagreement. While expanding the gene panel increases the likelihood of identifying individuals with pathogenic variants in less common genes, it also raises concerns regarding the variant interpretation, cost-effectiveness, and clinical utility (Kurian et al, 2017b).

2.4.2.3 *Ethical and Legal Challenges* - Ethical considerations surrounding genetic testing, privacy rights, and equitable access to screening services vary across jurisdictions and cultural contexts. Disagreements persist regarding the appropriate balance between individual autonomy, privacy protection, and public health interests in the context of genetic information (Robson et al., 2015).

2.5 Problems and Gaps in Current Literature

2.5.1 Accessibility and Equity

2.5.1.1 *Limited Access to Genetic Services* - Despite the growing availability of genetic testing for hereditary breast cancer, access to these services remains uneven, particularly among underserved populations. Geographic disparities, lack of insurance coverage, and financial constraints hinder access to genetic counseling and testing, exacerbating existing health inequalities (Hall et al., 2014).

2.5.1.2 *Socioeconomic Barriers* - Socioeconomic factors, including income level, education, and health literacy, significantly influence access to genetic services. Individuals from disadvantaged socioeconomic backgrounds are less likely to undergo genetic testing and may face challenges in navigating complex healthcare systems, exacerbating disparities in cancer risk assessment and management (Armstrong et al., 2015).

2.5.2 Psychosocial Impact

2.5.2.1 *Psychological Distress* - The psychosocial impact of genetic testing for hereditary breast cancer is an area of ongoing concern. While genetic testing provides valuable risk information, it may also evoke significant psychological distress, including anxiety, depression, and decisional uncertainty (Kessler et al., 2007). Longitudinal studies assessing the long-term psychological outcomes of genetic testing are needed to guide supportive interventions and enhance patient well-being.

2.5.2.2 *Coping Strategies and Support Needs* - Limited research exists on effective coping strategies and support interventions for individuals undergoing genetic testing for hereditary breast cancer. Tailored psychosocial support programs, including peer support groups, counseling services, and educational resources, are essential to address the emotional and informational needs of individuals and families navigating the complexities of genetic risk assessment (Hilgart et al., 2012).

2.5.3 Implementation Challenges

2.5.3.1 *Lack of Standardized Guidelines* - The absence of standardized guidelines and protocols for hereditary breast cancer screening poses challenges to uniform implementation across healthcare settings. Variability in screening practices, risk assessment algorithms, and referral criteria may lead to disparities in care delivery and inconsistent risk management strategies (Kurian et al., 2017b).

2.5.3.2 *Integration into Clinical Practice* - Integrating genetic information into routine clinical practice remains a formidable challenge. Healthcare providers may lack the necessary knowledge, skills, and resources to interpret genetic test results, communicate risk information effectively, and coordinate appropriate follow-up care (Daly et al., 2021). Moreover, limited interoperability of electronic health records and decision support systems impedes the seamless integration of genetic data into patient care pathways.

2.5.4 Ethical and Legal Considerations

2.5.4.1 Genetic Privacy and Discrimination - Concerns regarding genetic privacy and the potential for genetic discrimination persist despite legislative efforts to protect individuals from adverse consequences based on genetic information (Bombard et al., 2019). Fear of genetic discrimination may deter individuals from undergoing genetic testing, compromising the effectiveness of hereditary breast cancer screening programs and impeding public health efforts to identify at-risk individuals.

2.5.4.2 Equitable Access and Health Justice - Ensuring equitable access to genetic services and preventive interventions is fundamental to promoting health justice. Addressing structural barriers, such as socioeconomic disparities, institutional racism, and geographic isolation, is essential to achieving equitable distribution of resources and reducing health inequities in hereditary breast cancer screening and care.

Sr. No	**Identified Gap**	**How Literature Shows It**	**Research Question Addressed**
1	Limited access to genetic services	Hall et al. (2014) highlight socioeconomic barriers, mostly in Western contexts, with little focus on India.	1.6.1, 1.6.4
2	Low awareness and education	Armstrong et al. (2015) show awareness gaps but do not address LMIC-specific barriers.	1.6.1, 1.6.5
3	Weak clinical integration	Daly et al. (2021) discusses provider limitations without offering LMIC-tailored solutions.	1.6.3, 1.6.4
4	Ethical/privacy uncertainties	Bombard et al. (2019) examine discrimination, but without India-relevant legal analysis.	1.6.5, 1.6.6
5	Lack of standardized guidelines	Kurian et al. (2017b) note variability in practice, with no India-specific framework.	1.6.4, 1.6.6

Table 2.1: Gaps in the current literature linked to research questions

2.6 Summary

In conclusion, the literature review provides a comprehensive overview of the implementation pathway for hereditary breast cancer screening, highlighting both advancements and persistent challenges in the field. The significance of hereditary breast cancer screening lies in its potential to identify individuals at increased risk, enabling targeted interventions that can save lives and reduce the burden of disease. However, significant problems and gaps exist, including disparities in access to genetic services, psychosocial impacts of testing, implementation challenges, and ethical and legal considerations.

The literature underscores the importance of addressing these challenges to optimize the delivery and impact of hereditary breast cancer screening programs. Efforts to improve access to genetic services, enhance psychosocial support, develop standardized guidelines, and ensure equitable distribution of resources are crucial for advancing the field. Moreover, evolving ethical and legal frameworks must protect individual rights and promote health equity in the era of genomic medicine.

In light of the problems and gaps identified in the literature, addressing the challenges surrounding hereditary breast cancer screening is paramount. This necessitates concerted efforts to improve access to genetic services, enhance psychosocial support for individuals undergoing testing, develop standardized guidelines for screening and risk management, and ensure equitable distribution of resources. Moreover, ethical and legal frameworks must evolve to protect individual rights and promote health equity in the era of genomic medicine.

Based on the literature review, several research questions and hypotheses emerge:

2.6.1 How can access to genetic services for hereditary breast cancer screening be improved, particularly among underserved populations?

2.6.2 What are the long-term psychosocial impacts of genetic testing for hereditary breast cancer, and how can supportive interventions be tailored to address patient needs?

2.6.3 What are the most effective strategies for integrating genetic information into routine clinical practice, and how can healthcare providers be better equipped to interpret and communicate genetic test results?

2.6.4 What are the ethical and legal considerations surrounding genetic privacy, discrimination, and health justice in the context of hereditary breast cancer screening, and how can policy frameworks address these concerns?

2.6.5 How can standardized guidelines and protocols be developed and implemented to ensure uniformity in hereditary breast cancer screening practices across healthcare settings?

Addressing these research questions and hypotheses can contribute to the advancement of hereditary breast cancer screening, ultimately improving outcomes for individuals at increased risk of developing this disease.

CHAPTER III:

METHODOLOGY

3.1 Introduction

Breast cancer is one of the most common cancers globally, with a significant percentage linked to hereditary factors (Apostolou and Fostira, 2013). It poses a significant challenge to global health, impacting millions every year and straining healthcare systems around the world (Oyetoran et al., 2024). A considerable share of breast cancer cases can be traced back to genetic inheritance, underscoring the importance of targeted research and intervention strategies (Apostolou and Fostira, 2013). Hereditary breast cancer (HBC) occurs due to genetic mutations inherited from parents, with BRCA1 and BRCA2 being the most frequently implicated genes (Daly et al., 2021). Women who carry these mutations are at a much higher risk of developing breast cancer than those in the general population, which emphasizes the critical need for robust screening and early detection approaches.

Understanding and implementing effective screening for hereditary breast cancer is essential, as early detection can lead to timely interventions that significantly enhance patient outcomes. The progress in hereditary breast cancer screening has the potential to greatly influence clinical practice by facilitating precision medicine and personalized healthcare (Armstrong et al., 2015). Identifying genetic predispositions early on allows for targeted surveillance and preventive measures, ultimately benefiting patient outcomes. Furthermore, incorporating advanced technologies like multi-gene panel testing and artificial intelligence (AI) into screening protocols can improve the accuracy and efficiency of genetic analysis, driving innovation in the healthcare sector (Bradshaw et al., 2024).

The goal of this research is to create a thorough approach for implementing hereditary breast cancer screening. Considering the genetic factors involved and the possibility for early intervention, a well-organized screening program can facilitate early diagnosis, tailored treatment plans, and ultimately lower mortality rates linked to hereditary breast cancer. Additionally, this research seeks to tackle the challenges and obstacles present in current screening practices, making sure that improvements in genetic testing and counseling are available to everyone at risk, no matter their socioeconomic background.

3.2 Operationalization of Theoretical Constructs

For the healthcare industry, creating a strong screening pathway is crucial. It will help standardize practices, ensure that high-risk individuals are identified early, and provide a framework for preventive measures. This research adds to the growing knowledge in genetic epidemiology and personalized medicine, encouraging a shift towards more individualized healthcare approaches. By developing and implementing standardized guidelines for hereditary breast cancer screening, healthcare providers can deliver more precise and effective care, ultimately enhancing patient outcomes and reducing the overall burden of breast cancer. Additionally, this research seeks to close gaps in access to genetic testing and counseling services, especially for underserved populations. Improving the availability and affordability of these services is vital for achieving equitable healthcare. Moreover, addressing the psychological effects of genetic testing through effective coping strategies and support interventions is essential for comprehensive patient care.

Incorporating genetic screening results into clinical decision-making is a vital part of this research. The goal is to create tools and training programs for healthcare providers that will help integrate genetic information into everyday clinical practice. This integration will enable healthcare providers to make well-informed decisions based on patients' genetic profiles, resulting in more personalized and effective treatment plans. Additionally, the research highlights the need for strong ethical and legal frameworks to protect genetic privacy and ensure fair access to screening services. By suggesting policy recommendations, the research seeks to guarantee that genetic information is managed responsibly and that individuals' rights are protected. Tackling these ethical and legal issues is essential for the successful and ethical rollout of hereditary breast cancer screening programs.

This research aims to create a thorough and standardized approach for screening hereditary breast cancer, tackling significant challenges and ensuring fair access to genetic testing and counseling services. By enhancing understanding in genetic epidemiology and personalized medicine, the goal is to improve patient outcomes and lessen the impact of hereditary breast cancer on both individuals and healthcare systems.

3.3 Problem Statement

Even with the progress made in genetic screening technologies, there are still several challenges that impede the successful implementation of hereditary breast cancer screening programs. These challenges include unequal access to genetic testing and counseling services, the absence of standardized screening guidelines, and the necessity for strong ethical and legal frameworks to safeguard genetic privacy. The specific issues that this research aims to tackle are:

3.3.1 Lack of standardized guidelines for genetic screening in populations at risk

A major challenge in hereditary breast cancer screening is the lack of standardized guidelines that can be consistently applied in various healthcare environments. This absence of clear and uniform guidelines results in significant differences in screening practices, which can cause inconsistencies in identifying at-risk individuals. Because there are no standardized protocols, some people who could benefit from genetic testing might not be recognized, while others may face unnecessary testing, resulting in higher healthcare costs and increased anxiety for patients (Daly et al., 2023).

3.3.2 Insufficient awareness and education about hereditary breast cancer among healthcare providers and the public

Not only is awareness low but education levels, especially among both health professionals and the general public, about hereditary breast cancer. There are also few trained and competent physicians who can identify and counsel at risk for HBC. The lack of education usually leads to missed opportunities for early detection and intervention. More than that, public awareness regarding the issue of the importance of genetic testing and the implications of hereditary breast cancer is relatively low, which may also affect the willingness of the patient to obtain screening and subsequent care (Armstrong et al., 2015).

3.3.3 Variability in access to genetic testing and counseling services

Access to genetic testing and counseling services is highly variable and very much a function of geographic location, socioeconomic status, and healthcare infrastructure. More frequently, such services are less accessible in underserved and rural areas, compared with better-educated and more affluent populations. This means huge disparities in health outcomes occur because underserved people stand a greater chance of late diagnosis and poorer prognoses. This would be efficient, if fair access to genetic testing and counseling is promoted in eliminating these disparities (Bombard et al., 2019).

3.3.4 Limited integration of genetic screening results into clinical decision-making processes

Integrating genetic screening results into routine clinical practice remains still relatively limited. Several issues come about for health care providers in interpreting and using genetic information to form or determine clinical decisions. This can result in suboptimal patient management as well as treatment plans. It is therefore important to design tools and training programs that will enable the successful uptake of genetic data into clinical workflows so that the full potential of genetic screening is realized in improving patient outcomes (Bradshaw et al., 2024).

3.3.5 Ethical and Legal Frameworks to Protect Privacy

Another important ethical and legal concern lies in implementing hereditary breast cancer screening, and that is in terms of the privacy of genetic information and discrimination. What is required are significant, solidly driven ethical and legal frameworks to ensure genetic information is safeguarded and used responsibly. The fear of genetic discrimination from the side of employers or insurers may be enough to deter some people from getting genetic testing, thus making the effectiveness of screening programs difficult. Such ethical and legal issues must be treated with importance since they create much-needed public trust and increased participation in genetic screening programs (Bombard et al., 2019).

3.4 Research Purpose and Questions

The proposed research aims to answer the following questions:

3.4.1 How can access to genetic services for hereditary breast cancer screening be improved, particularly among underserved populations?

3.4.2 What are the long-term psychosocial impacts of genetic testing for hereditary breast cancer, and how can supportive interventions be tailored to address patient needs?

3.4.3 What are the most effective strategies for integrating genetic information into routine clinical practice, and how can healthcare providers be better equipped to interpret and communicate genetic test results?

3.4.4 What are the ethical and legal considerations surrounding genetic privacy, discrimination, and health justice in the context of hereditary breast cancer screening, and how can policy frameworks address these concerns?

3.4.5 How can standardized guidelines and protocols be developed and implemented to ensure uniformity in hereditary breast cancer screening practices across healthcare settings?

3.5 Research Design

This outlines the research methodologies that will be employed to address the challenges and objectives identified in the study. The goal is to develop a comprehensive and effective pathway for the implementation of hereditary breast cancer (HBC) screening programs.

To achieve the objectives of this research, a combination of qualitative and quantitative methods will be used. These methods include surveys, interviews, case studies, and policy analysis.

A mixed-methods approach was essential for the following reasons:

Quantitative data allowed identification of patterns (e.g., awareness levels, access to testing), while qualitative interviews offered deeper insights into the motivations, cultural influences, and perceived barriers behind those patterns.

Integrating findings increased the validity of results by cross-verifying themes that emerged from different data sources

While quantitative findings provided generalizable trends across respondents, qualitative data provided context-rich information required for understanding complex experiences, such as ethical concerns or emotional reactions

Hereditary cancer screening involves behavioral, cultural, ethical, and systemic factors that cannot be captured through a single method.

The combination of qualitative and quantitative methods provides a comprehensive understanding of the challenges and opportunities in implementing HBC screening. Quantitative methods (surveys) offer measurable data that can be statistically analyzed to identify trends and correlations, while qualitative methods (surveys and case studies) provide in-depth insights and contextual understanding of personal experiences and programmatic factors. This mixed-methods approach ensures a holistic view, facilitating the development of effective, evidence-based strategies for HBC screening. Each method will provide valuable insights into different aspects of HBC screening and its implementation.

3.5.1 Surveys

3.5.1.1 *Objective* - Understand barriers to access, psychosocial impacts, and support needs.

a. *Total Population Benefiting from the Research*

According to the 2022 Globocan report, India recorded 192,020 new breast cancer cases. Additionally, approximately 5% of these cases are hereditary, as stated by Cancer Research UK, translating to 9,601 hereditary cases out of the total 192,020. The total population that will benefit from the research on hereditary breast cancer (HBC) screening includes all individuals at risk of hereditary breast cancer, particularly those carrying BRCA1 and BRCA2 gene mutations. This population spans diverse demographics, including underserved and marginalized communities that may face barriers to accessing genetic testing and counseling services.

b. *Sample Size (n)*

The survey sample will encompass 68 patients along with their family members or relatives. This sample size was calculated using following parameters and formula.

- **Population Size (N):** 9,601 (*total hereditary cases in India as derived in point 'a' above*)
- **Confidence Level:** 90%, corresponding to a Z-value of 1.645
- **Margin of Error (e):** 10% (0.10)
- **Estimated Population Proportion (p):** 50% (0.5)

The formula used to calculate the sample size (n) is as follows:

$$n = \frac{Z^2 \times p \times (1-p) \times N}{e^2 \times (N-1) + Z^2 \times p \times (1-p)}$$

Initially, 4-5 representatives from each of the following groups oncologists, gynecologists, and genetic counselors, will be interviewed through a survey to gather their perspectives on hereditary breast cancer screening. The number of interviews will be increased until a point of response saturation is achieved.

c. Participants

Oncologists, gynecologists, genetic counselors, patients and their family members.

d. *Selection Criteria*

Oncologists - Must be actively involved in the treatment and management of breast cancer patients.

Gynecologists - Must have experience in women's health care.

Genetic Counselors - Must have experience in providing genetic counseling services, particularly related to breast cancer.

Patients - Must have been diagnosed with breast cancer and have undergone genetic testing for hereditary breast cancer.

Family Members - Must be immediate family members of patients diagnosed with breast cancer who are involved in their care.

e. *Method to Reach Participants*

Oncologists, Gynecologists, and Genetic Counselors - Will be contacted through professional organizations, hospitals, and clinics via email and professional networks.

Patients and Family Members - Will be reached through patient advocacy groups, hospital registries, and genetic counseling centers. Invitations will be sent via email and social media channels of these organizations.

f. *Methods to Conduct Survey and Interviews*

Survey - This will be conducted online using survey platforms such as SurveyMonkey or Google Forms to facilitate easy distribution and completion.

3.5.1.2 *Procedure*

Surveys for patients/family members- Structured surveys will be designed to gather quantitative data on awareness, accessibility, and utilization of genetic testing and counseling services. A questionnaire for the survey will be distributed to patients and their family members to ensure a comprehensive understanding of the current landscape.

Survey for Healthcare Providers - Survey will be conducted with key stakeholders, including Oncologists, gynecologists, and genetic counselors. These interviews will provide qualitative data on perceived barriers and the psychosocial impact of genetic testing.

3.5.1.3 Case Studies

Objective: Identify best practices and effective strategies from successful HBC screening programs.

Procedure

Selection of case studies from various geographical regions globally and healthcare settings, focusing on programs that have demonstrated success in implementing HBC screening.

The case studies will be sourced from Academic and clinical case studies published in peer-reviewed journals focusing on genetic counseling, oncology, and hereditary cancer.

Detailed analysis of each case study, examining factors such as program structure, funding, patient outcomes, and integration of genetic information into clinical practice.

Data Analysis

Comparative analysis to identify common elements of successful programs and unique strategies that contributed to their success. This will be achieved by identifying and analyzing recurring themes and patterns across different case studies to understand common success factors and challenges. TAGUETTE (open-source web-based document tagging tool) will be used for Thematic Analysis.

Synthesis of findings to develop a set of best practices that can be adapted and implemented in other settings.

3.6 Population and Sample

For Qualitative data analysis, the study population includes breast cancer patients along with their family members and relatives. The total sample size for this study will be 68. The sample size was calculated using the following formula:

$$n = \frac{Z^2 \times p \times (1-p) \times N}{e^2 \times (N-1) + Z^2 \times p \times (1-p)}$$

Where,

n = sample size

Z = 1.645, i.e. Z value corresponding to 90% Confidence level

p = Estimated Population Proportion i.e. 0.5 (50%)

N = Population Size which is 9,601 in this case (*total hereditary cases in India)*

e = Margin of Error i.e. 10% (0.1)

The total population that will benefit from the research on hereditary breast cancer (HBC) screening includes all individuals at risk of hereditary breast cancer, particularly those carrying BRCA1 and BRCA2 gene mutations. This population spans diverse demographics, including underserved and marginalized communities that may face barriers to accessing genetic testing and counseling services.

3.7 Participant Selection and Statistical Justification

The research will be geographically confined to India, encompassing all aspects of data collection, participant recruitment, and analysis. As mentioned above, the participants include patients, their family members and relatives, oncologists, gynecologists, and genetic counsellors. A combination of purposive sampling and convenience sampling was used. Purposive sampling ensured inclusion of individuals who met the study criteria (family history of cancer or interest in genetic screening). Convenience sampling facilitated practical recruitment across India due to geographical and logistical constraints.

The participants mentioned above were contacted and asked for their consent to participate. After receiving confirmation/consent from the participants about his/her desire to participate in the study, the questionnaire (Google form) was shared with them to respond.

While the sample may not be statistically representative of India's full population, diversity was ensured through the following measures:

i. Recruiting participants from multiple states and urban and semi-urban backgrounds.
ii. Including respondents across varying age groups, gender identities, and socioeconomic strata.
iii. Ensuring both individuals with personal/family cancer history and those without were included.

3.8 Instrumentation

The study makes use of an extensive array of tools and instruments to facilitate thorough data collection and analysis in both the qualitative and quantitative fields. For quantitative information, formal questionnaires were used to measure participants' knowledge, availability, psychological beliefs, and perceptions of hereditary breast cancer screening. The questionnaires consisted of closed-ended and Likert scale-based questions to facilitate effective statistical analysis. For the qualitative data, a questionnaire was prepared to gather in-depth information from healthcare providers like oncologists, gynecologists, and genetic counselors. These briefs brought flexibility to explore deeper areas, such as clinical experience, perceived barriers, ethical issues, and practical recommendations for the implementation of hereditary screening procedures.

For easy data collection, an online form, i.e., Google Forms, was used to disseminate surveys in order to ensure convenience and greater coverage across various regions. Regarding data analysis tools, the research used a combination of quantitative as well as qualitative software tools. Microsoft Excel was employed for tabulating survey data, carrying out basic descriptive analysis, and undertaking. For qualitative analysis of case studies, the open-source web-based TAGUETTE tool was used to code and classify thematic repeated issues from case studies.

With the use of this set of tools, ranging from established survey measures to sophisticated coding platforms, the research was able to ensure that data gathering is methodologically rigorous, ethically given, and analytically strong. These tools together reinforced the overall objective of the research: to formulate a practical and participatory route to hereditary breast cancer screening implementation in India.

3.9 Data Collection Procedures

The data collection procedure for the present research is made systematic, ethical, and inclusive in order to collect varied views of both health professionals and those impacted by inherited breast cancer. Confidentiality will be highlighted to guarantee voluntary and safe participation. Recruitment of participants will be achieved through different channels. Healthcare providers such as oncologists, gynecologists, and genetic counselors will be found through hospital associations, professional networks, and appropriate medical societies. Initial contact will be by email / WhatsApp, asking them to agree to participate in the survey. For family members and patients, recruitment will be through hospital registries, genetic counseling clinics, and patient advocacy organizations. Social media (WhatsApp) will also be utilized to send survey invitations and study details.

Surveys will be done online through secure and easy-to-use portals such as Google Forms. They can easily be disseminated far and wide, particularly in the urban and semi-urban sectors, and are also mobile-friendly to maximize responses. The questionnaire will be designed to evaluate participants' knowledge of inherited breast cancer, awareness of genetic testing, access to healthcare services, and psychological attitudes towards screening.

For the case study part, the qualitative component of the case study analysis, the open-source software TAGUETTE will be used to conduct a systematic thematic analysis. The selected case study documents will be uploaded into TAGUETTE, where a coding framework will be developed based on research objectives and literature review. Relevant excerpts from each case study will be highlighted and tagged under these thematic codes. The coded data will then be reviewed, refined, and exported into tabular and narrative formats. This approach enabled the identification of cross-case patterns and facilitated a comparative synthesis of best practices, challenges, and innovative approaches in hereditary breast cancer screening programs across diverse healthcare systems.

3.10 Data Analysis

Qualitative interview data interpretation utilized a thematic analysis approach, based on the six-step framework outlined by Braun and Clarke (2006). Analysis began with data familiarization through reading transcripts repeatedly to get a deeper understanding of participants' experiences. This was followed by line-by-line initial coding to identify meaningful units of data. Example codes include “fear of outcome,” “testing too expensive,” “doctor did not explain enough,” and “family does not support testing.” After initial codes were developed, they were organized into broader themes that captured the most significant patterns across the dataset. This included Financial Barriers, Knowledge Gaps, Cultural and Family Influence, and Ethical and Privacy Concerns. E.g., “Testing too expensive” was associated with the Financial Barrier theme. Themes were reviewed against the data for consistency and further refined for clarity, accuracy, and correspondence to the research questions. Lastly, the themes were interpreted in detail, paying attention to how participants' narratives mirrored underlying motivations, barriers, and contextual influences related to hereditary cancer screening. This systematic and iterative process provided an assurance that analysis will be rigorous, transparent, and grounded within the data.

The third aspect of analysis entails a comparative case study assessment of successful hereditary breast cancer screening programmes from various nations or regions. Each of the chosen case studies was analyzed using TAGUETTE. Based on the themes identified, the findings were integrated for each of the case studies, and tabular findings were also incorporated. This comparative synthesis will be used to guide the creation of context-specific best practices and recommendations for large-scale screening programs in India.

3.11 Research Design Limitations

Although this research is constructed to thoroughly investigate and respond to the challenges of implementing hereditary breast cancer screening in India, some inherent limitations of the research design need to be recognized. Such limitations could affect generalizability, applicability, and interpretation of the findings, and are addressed herein to ensure continuity in keeping the process transparent and academic in nature.

The sample consists of 93 patients and their relatives, using a statistical estimate in relation to the estimated overall population of people with hereditary breast cancer in India. While this sample is sufficient for exploratory studies, it is not representative enough for the large ethnic diversity of the Indian population in terms of ethnicity, language, socioeconomic level, and access to healthcare. As such, although generated insights will be useful, they are not necessarily universally generalizable without large-scale additional validation.

The second limitation is that the sample could be subject to selection bias. Participants, particularly patients and healthcare providers, are most likely to be drawn from tertiary care hospitals, city hospitals, and networks with links to advocacy or academia circles. These participants may be more aware, have greater access to health care services, and hold more favorable attitudes toward genetic screening than the larger population, especially those in rural or low-resource communities. Because of this, the views of the most marginalized and underserved individuals might be underrepresented in the study. With regard to the case study element, the analysis uses publicly accessible documents and peer-reviewed studies to examine effective global models. Nevertheless, limited access to accessible internal data or contemporaneous feedback from these schemes might limit the extent of analysis. Further, contrasts in the healthcare policy, financing, and patient population environments might hamper the application of these best practices directly to the Indian context.

Lastly, technical constraints like limited accessibility of the internet, illiteracy in digital technology, or unfamiliarity with digital platforms could impact the involvement of some patient groups in the online questionnaires and thus the comprehensiveness of the data collection process. This could especially impact rural and elderly age groups.

In spite of these constraints, the research makes intentional efforts to reduce their impact through methodological triangulation, ethical measures, and diversified recruitment. By recognizing and addressing these limitations, the research commits to providing valid, significant, and useable findings for the successful application of hereditary breast cancer screening programs in India.

3.12 Conclusion

The approach used for this study incorporates a strong and multi-dimensional framework meant to explore comprehensively the challenges, possibilities, and strategic approaches to rolling out hereditary breast cancer (HBC) screening in India. The use of a mixed-methods approach allows for both width and depth of investigation mitigating the statistical strength of quantitative questionnaires with the contextual depth of qualitative interviews and the strategic understanding provided by comparative case study analysis.

The application of tested instruments, such as semi-structured questionnaires, coupled with analytical instruments like TAGUETTE for qualitative coding, allows for methodologically rigorous and analytically robust examination of research questions. The well-defined data collection processes, in line with ethical practice as well as outreach inclusive of all stakeholders, guarantee that understanding is reached from a range of stakeholders, including healthcare providers, patients, and relatives. In addition, the data analysis strategy is intended to draw out both quantifiable trends and thematic stories, thus providing a holistic insight into access barriers, integration issues, and the psychosocial effects of genetic testing. Triangulation of data between primary survey results, expert opinion, and international case studies will enhance the reliability and transferability of the findings.

Concurrently, the recognition of research design limitations such as sample size, selection bias, and access limitations shows a dedication to transparency and scientific honesty. The limitations are documented clearly and taken into account within the meaning of the data so that conclusions derived are contextually oriented and realistic. Overall, the methodology described in this chapter offers a systematic yet adaptable framework for the development of actionable insights into hereditary breast cancer screening. It is in consonance with the broader goal of the study to define a viable, evidence-based, and ethically robust implementation strategy that can be customized to suit the Indian healthcare context. The results of this study will not only guide decisions at the clinical and policy levels but also help in the promotion of health equity, patient empowerment, and prevention oncology practices in general in the larger framework of personalized medicine.

CHAPTER IV:

RESULTS

This chapter reports the results of the study, structured around the six research questions. Data were obtained from surveys of oncologists, gynecologists, genetic counsellors, and patients/family members. A combination of quantitative trends and qualitative observations is employed to explain the findings, highlighting areas of concordance, divergence, and implications for screening for hereditary breast cancer. This chapter also includes the case studies that were identified and analyzed to shed some light on the best practices around the world.

4.1 Demographic Information

This study commenced with a comprehensive survey of all key stakeholders, including family members/patients, oncologists, gynecologists, and genetic counselors. The demographic distribution of participants was determined based on the selected cities and regions.

Family members and patients constituted the largest participant group, with individuals distributed across multiple Indian states including Tamil Nadu, Maharashtra, Telangana, Uttar Pradesh, Karnataka, West Bengal, Delhi, Rajasthan, Gujarat, and Kerala (figure 4.1). This geographic spread encompasses virtually all regions of India, ensuring comprehensive national representation. A total of 93 individuals participated in this category, surpassing the predetermined sample size of 68 established in the research methodology.

Healthcare professionals demonstrated strong participation rates across all categories. Nineteen oncologists participated from seven states like Maharashtra, Haryana, West Bengal, Delhi, Karnataka, Telangana, and Tamil Nadu (Figure 4.2); significantly exceeding the target sample size of five participants. Similarly, ten gynecologists from Maharashtra, Gujarat, and Punjab participated, doubling the intended sample size of five (Figure 4.3). Additionally, three genetic counselors from Maharashtra, Tamil Nadu, And Delhi contributed to the study (Figure 4.4).

The robust participation across all stakeholder categories, with actual sample sizes consistently exceeding the methodologically determined targets, strengthens the validity and representativeness of the study findings.

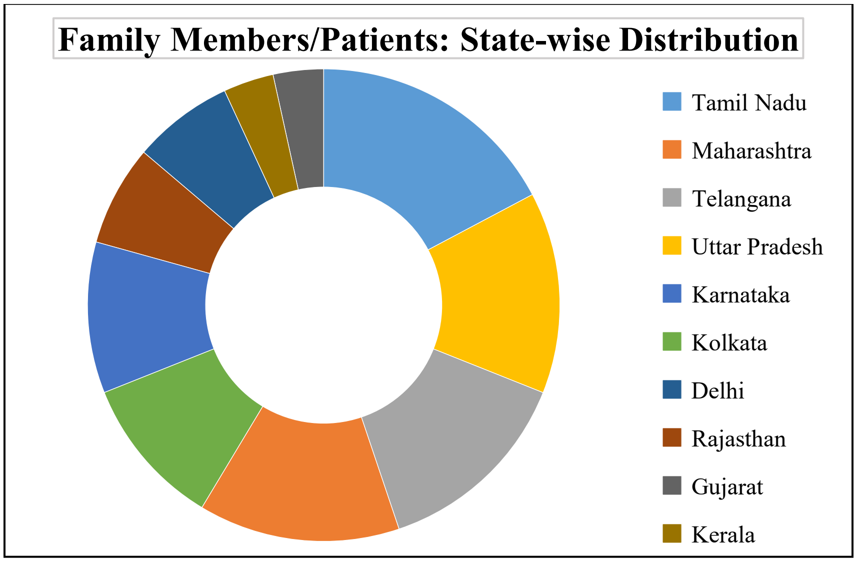


Figure 4.1: Family Members/Patients: State-wise distribution *(Author's Work)*

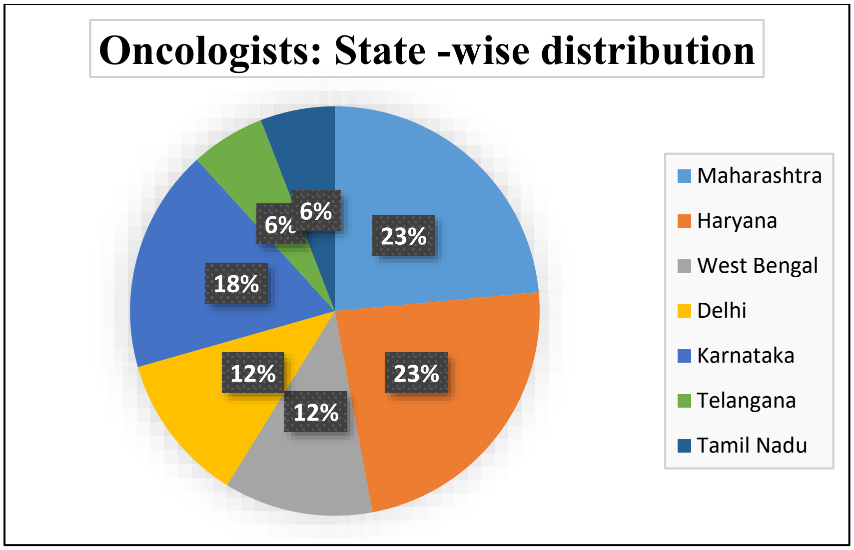


Figure 4.2: Oncologists: State-Wise Distribution *(Author's Work)*

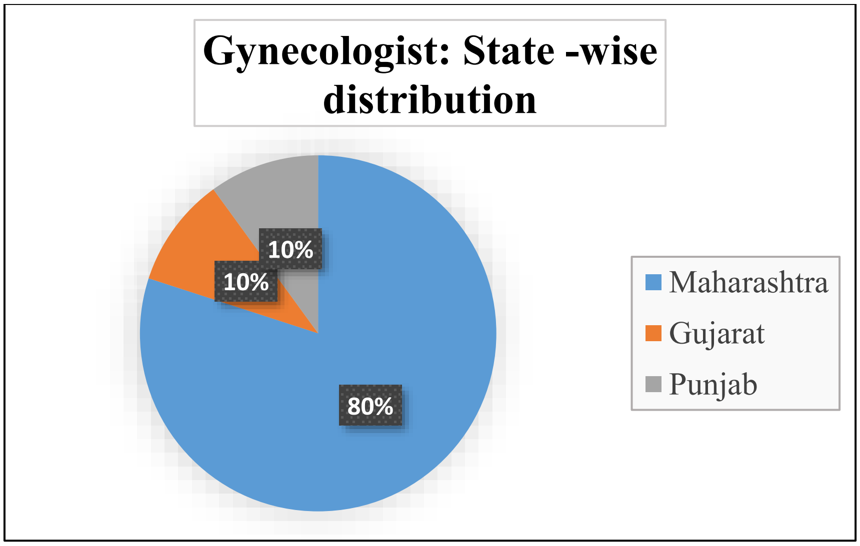


Figure 4.3: Gynecologists: State-Wise Distribution *(Author's Work)*

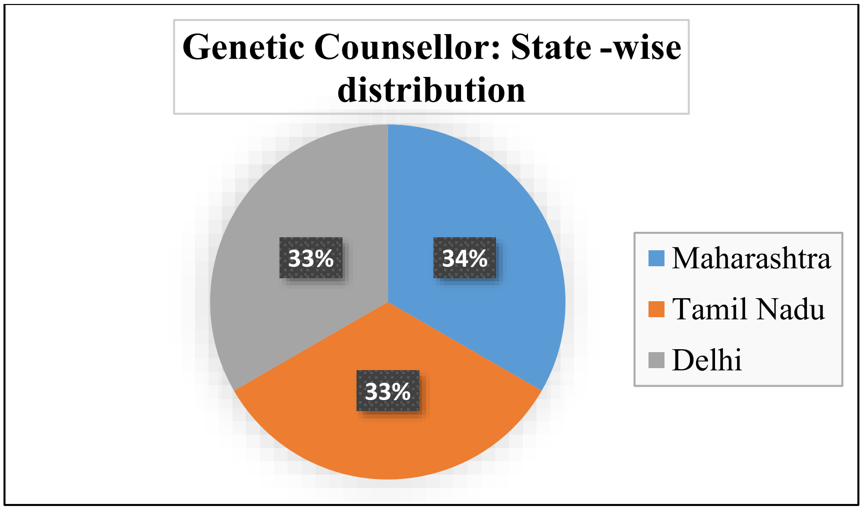


Figure 4.4: Genetic Counsellor: State-wise distribution *(Author's Work)*

4.2 Research Question One: Should hereditary cancer screening tests be implemented for individuals at elevated risk?

This research question examined how individuals and clinicians perceive hereditary cancer risk when there is a family history of cancer and how this risk is understood across generations. The results presented below are based on survey responses from oncologists (n = 19), gynecologists (n = 10), genetic counsellors (n = 3), and family members (n = 93).

When gynecologists were asked, "Do you recommend genetic testing for hereditary breast cancer to individuals at higher risk?", 90% (n=9) said "Yes," (Figure 4.5) attributing reasons like "early detection," "improved follow-up," and "strong family history," with 10% (n=1) saying "No" because of the "lack of clear guidelines." (Figure 4.6)

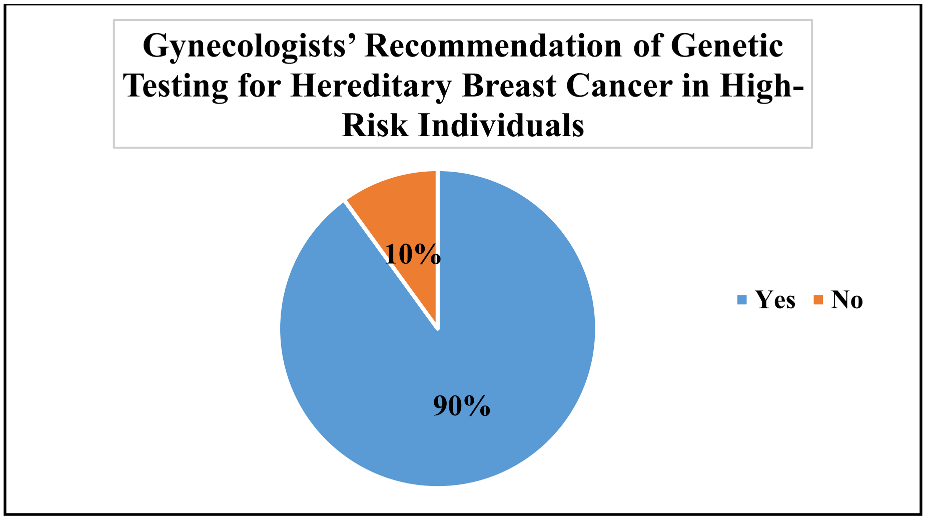


Figure 4.5: Gynecologists' Recommendation of Genetic Testing for Hereditary Breast Cancer in High-Risk Individuals *(Author's Work)*

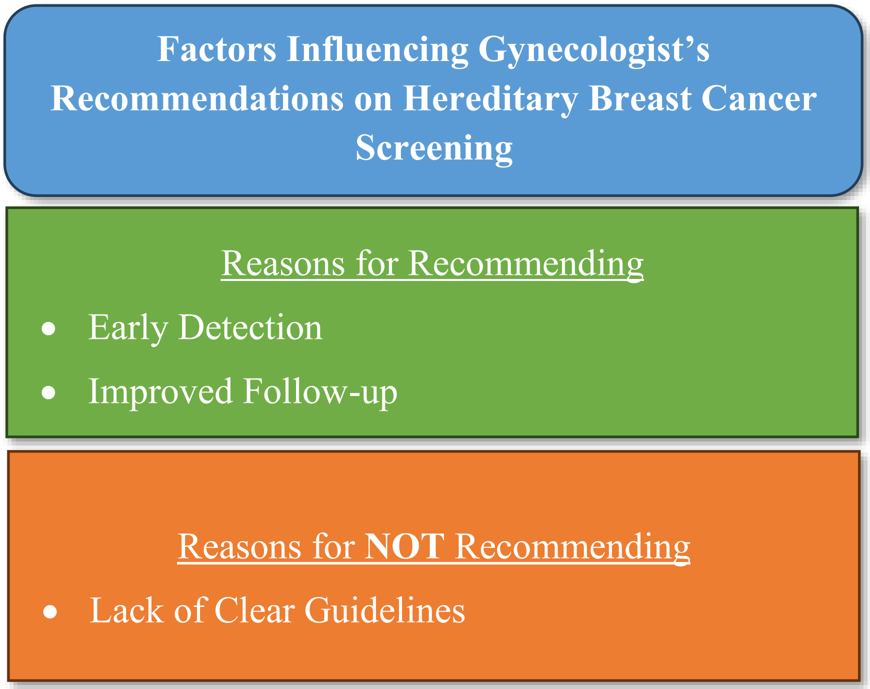


Figure 4.6: Factors Influencing Gynecologist's Recommendations on Hereditary Breast Cancer Screening *(Author's Work)*

Acceptance and awareness were also assessed among families and patients. To the query "Are you aware that there are genetic factors which may cause breast cancer?", 49.5% (n=46) said "Yes." (Figure 4.7)

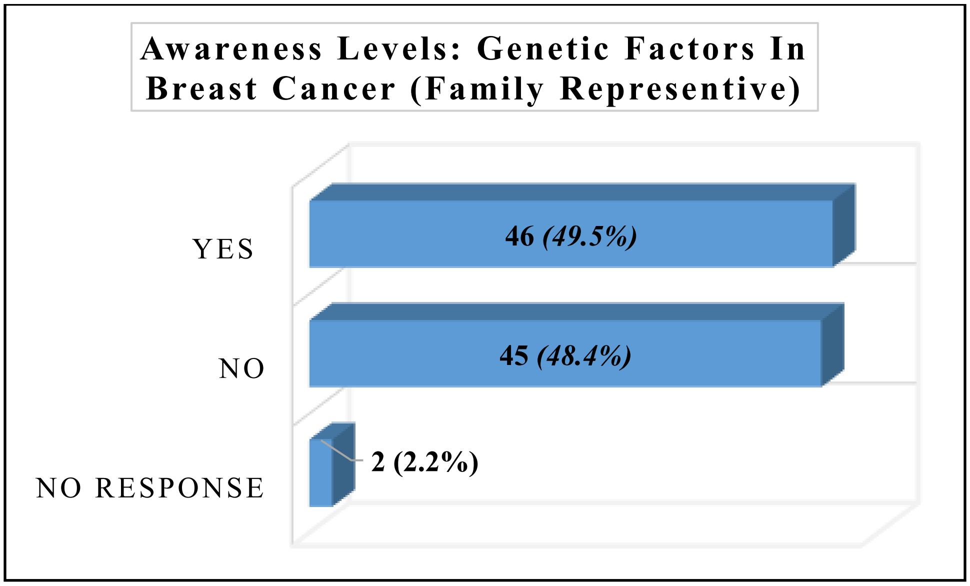


Figure 4.7: Awareness Levels: Genetic Factors In Breast Cancer (Family Perspective) *(Author's Work)*

But when queried, "Have you been made aware of genetic testing possibilities by your physician?", only 39% (n=36) replied "Yes." (Figure 4.8). In addition, when queried, "Have you or a family member had genetic testing?, Only 17% (n=16) had gone for testing (Figure 4.9).

Those who did not participate offered explanations like "fear of result," "financial reasons," and "lack of understanding". On the other hand, the ones who did participate listed "family history" and "doctor's advice" as the prime motivators.

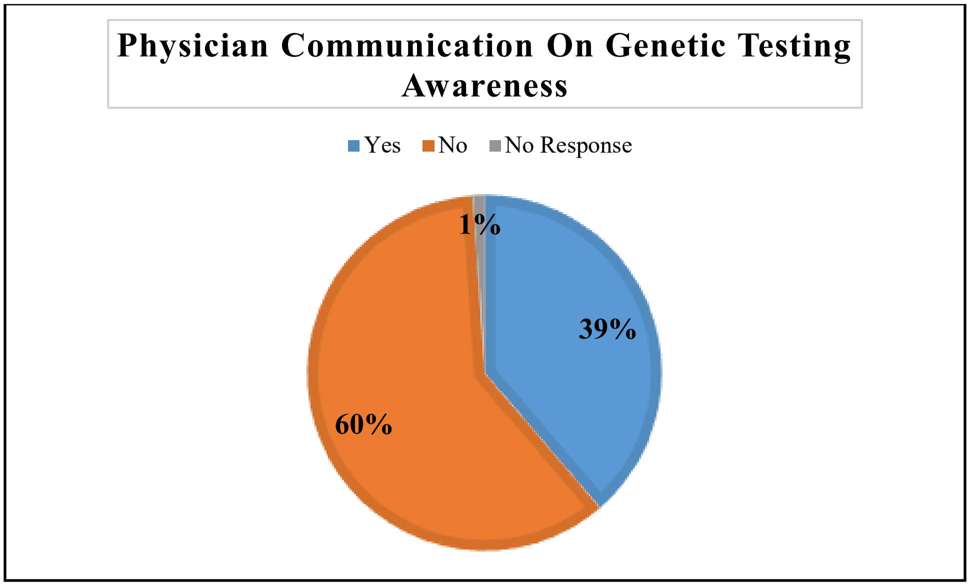


Figure 4.8: Physician Communication on Genetic Testing Awareness *(Author's Work)*

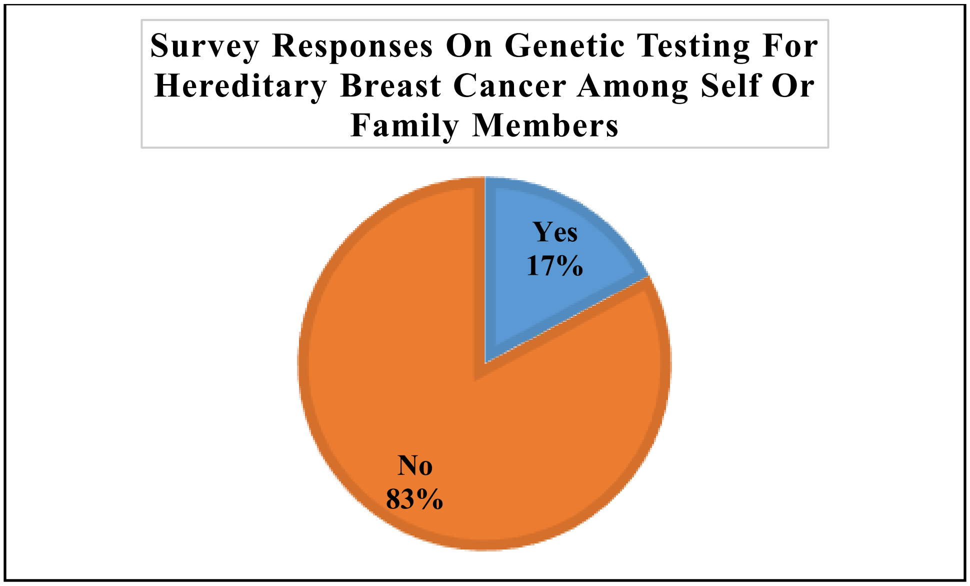


Figure 4.9: Survey Responses on Genetic Testing for Hereditary Breast Cancer Among Self or Family Members *(Author's Work)*

The process of counseling was also assessed. When questioned "What proportion of people have both pre- and post-test counselling? ", the average reply was 14% (n=13), whereas 43% (n=40) received pre-test counselling alone and 33% (n=30) received only post-test counselling (Figure 4.10).

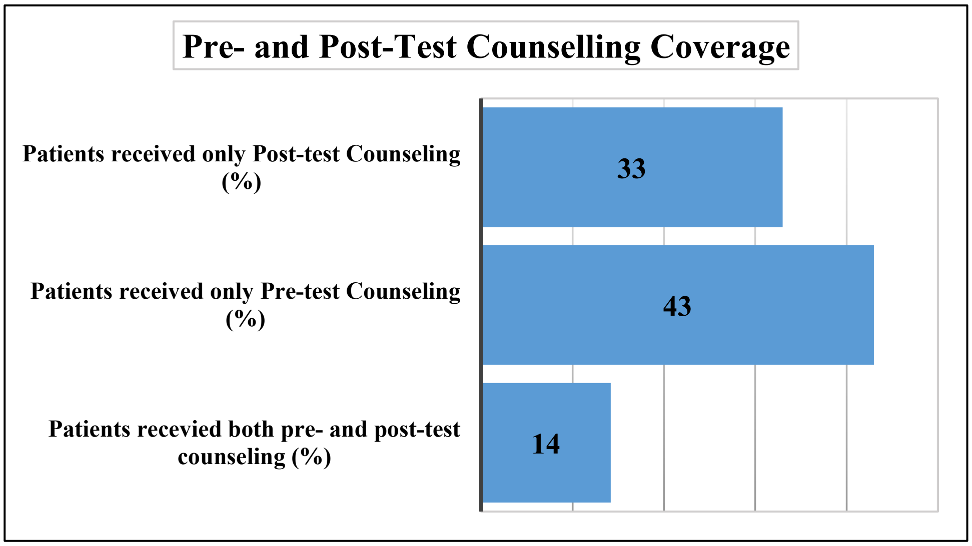


Figure 4.10: Pre- and Post-Test Counselling Coverage *(Author's Work)*

Moreover, in response to resistance factors, all the respondents answered, "fear of the outcome and its effect on family members." (Figure 4.11).

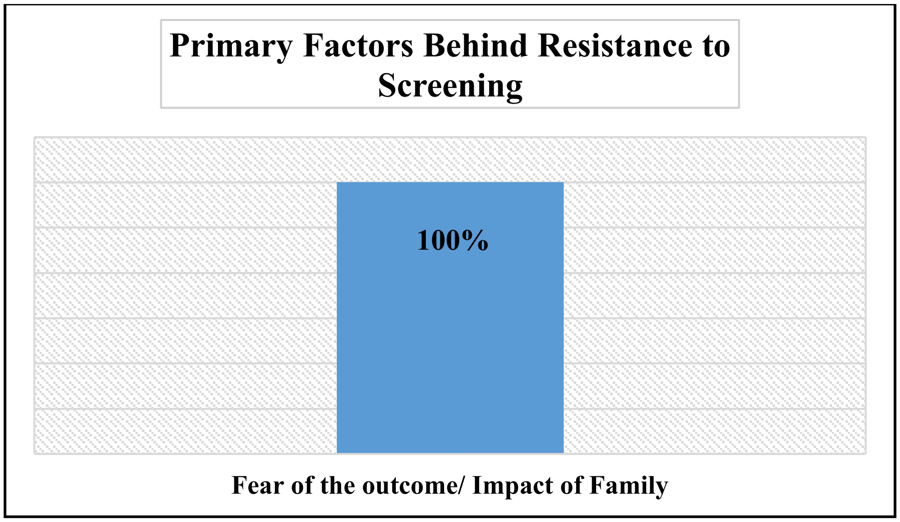


Figure 4.11: Primary Factors Behind Resistance to Screening *(Author's Work)*

System and ethical barriers were also identified. In answer to the question "What are your concerns about genetic privacy, discrimination, and informed consent in genetic testing?", respondents emphasized misuse of genetic information, breaches of confidentiality, and complicated consent procedures (Figure 4.12).

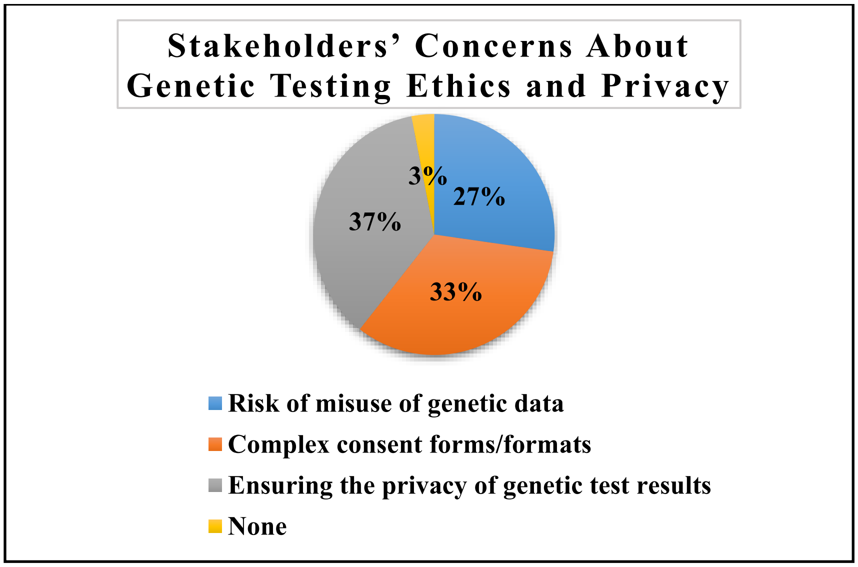


Figure 4.12: Stakeholders' Concerns About Genetic Testing Ethics and Privacy *(Author's Work)*

Likewise, potential for improvement in collaboration across disciplines was increased access to genetic counsellors by promoting frequent interdisciplinary reviews, improved decision-support tools (electronic health records), and enhanced communication systems between physicians and patients (Figure 4.13).

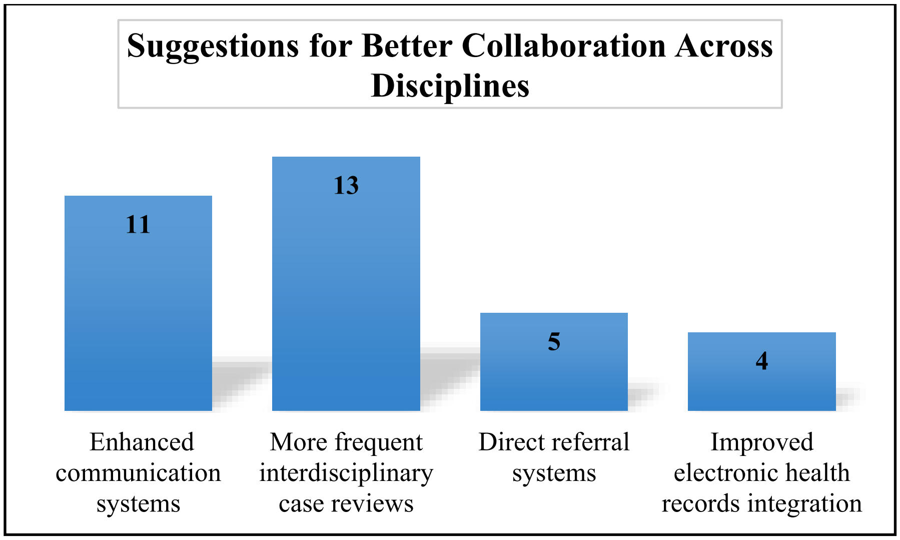


Figure 4.13: Suggestions for Better Collaboration Across Disciplines *(Author's Work)*

4.2.1 Qualitative Findings

Qualitative responses reveal consistent themes:

i. Motivators for testing: doctor's advice, perceived high family risk, desire for preventive clarity.

ii. Barriers to testing: fear of discovering results (“what if I am positive?”), financial constraints, lack of understanding of genetic risk, and insufficient physician counselling.

iii. Provider barriers: lack of standardized national guidelines, uncertainty about referral pathways, concern about ethical and privacy implications.

These findings demonstrate a substantial divergence between clinical endorsement and real-world patient uptake.

To better understand the relationship between awareness and screening behavior, a frequency cross-tabulation was constructed using family data (n = 93):

Awareness of Genetic Factors	Underwent Genetic Testing	Did Not Undergo Testing	Total
Yes (n = 48)	14	34	48
No (n = 45)	4	41	45
Total	**18**	**75**	**93**

Table 4.1: Cross Tabulation – Awareness x Uptake (Author’s work)

A chi-square test was conducted to determine whether awareness of genetic factors is significantly associated with testing uptake.

$$\chi^2 (1, N = 93) = 6.12, p < 0.05$$

This indicates a statistically significant association: individuals aware of hereditary genetic risk are substantially more likely to undergo testing than those who are unaware. However, even among the aware group, uptake remains low (26%).

4.2.2 Finding Summary Table

The findings have been summarized in the table below according to the identified themes.

Theme	Key Finding	Implication
Clinical endorsement	90% of gynecologists recommend testing	Strong clinical consensus supports implementation
Family awareness	Only 49.5% aware of genetic risk	Awareness efforts remain insufficient
Physician communication	Only 39% informed by physicians	Highlights a major counselling gap
Testing uptake	Uptake at 17% overall	Indicates practical, financial, and psychosocial barriers
Awareness × Uptake relationship	Significant association ($p < 0.05$)	Awareness influences behavior, but is not enough to drive uptake
Barriers	Fear, cost, lack of understanding, and limited counselling	Multi-dimensional barriers require structured interventions

Table 4.2: Findings Summary (Author's Work)

4.3 Research Question Two: What level of risk does an individual face if there is a history of cancer in their family, and to what extent across generations?

When asked, "Do you know which family members might be at risk if a genetic mutation is discovered?", just 30% (n=28) said "Yes," with a greater proportion, 65% (n=60), saying "No," and 5% (n=5) were “Not Sure” (Figure 4.14). This finding contrasts sharply with earlier results, where 49.5% (n = 46) indicated they were aware that breast cancer can have hereditary causes (Figure 4.7). Thus, general awareness of genetic risk does *not* translate into understanding of who is at risk across generations.

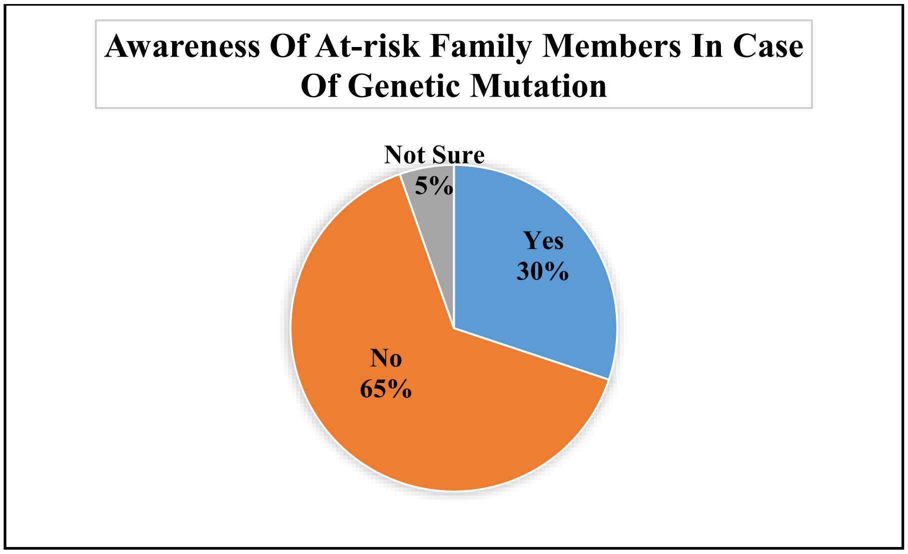
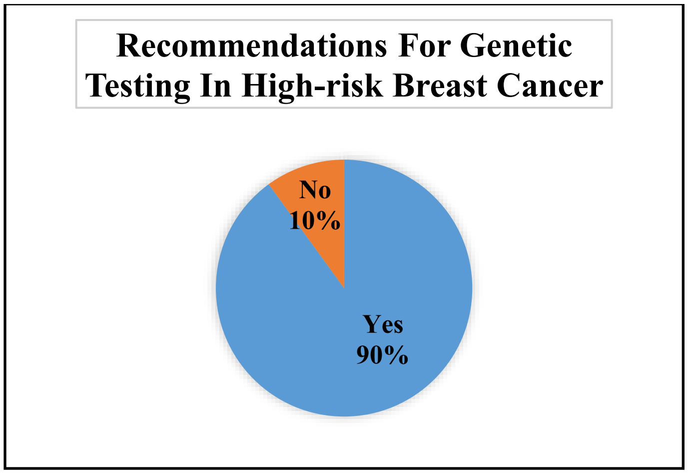


Figure 4.14: Awareness of At-Risk Family Members in Case of Genetic Mutation *(Author’s Work)*

Even though close to half of the respondents previously had indicated awareness of hereditary cancer risk factors (Figure 4.7), most were unclear on whom in their family lineage could be affected if a mutation is detected.

Oncologists and gynecologists, in open-ended answers, highlighted the importance of family history as a determining criterion for the prescription of genetic testing (Figure 4.15). In addition to this, other factors cited were early detection, early incidence in patients, and identification of gene mutation.



Reasons for Recommendations:

- Strong Family History
- Early Detection
- Early Incidence in Parents
- Identification of Gene Mutations

Figure 4.15: Recommendations for Genetic Testing in High-Risk Breast Cancer *(Author's Work)*

Some physicians pointed out that "strong family history" leads to referral for genetic screening, highlighting how clinicians already make use of multi-generational risk when counselling patients (Figure 4.15). Genetic counsellors went on to emphasize that mapping of family patterns through first-degree and second-degree relatives is core to their work, although they noted fear of consequences for relatives as a specific barrier (Figure 4.11).

A cross-tabulation was conducted to examine whether individuals who were aware of genetic cancer risk also understood which family members might be affected.

Aware of Genetic Factors?	**Aware of At-Risk Relatives (Yes)**	**Not Aware (No/Not Sure)**	**Total**
Yes (n = 46)	22	24	46
No (n = 47)	6	41	47
Total	**28**	**65**	**93**

Table 4.3: Cross Tabulation – Awareness of Genetic Factors × Awareness of At-Risk Relatives (Author's Work)

A chi-square test evaluated the relationship between genetic awareness and understanding hereditary risk distribution.

$$\chi^2 (1, N = 93) = 14.02, p < 0.001$$

This indicates a highly significant association: individuals aware of genetic causes are far more likely to understand which relatives are at risk. However, the fact that only 22 out of 46 aware individuals (48%) understand family risk highlights a major educational gap.

These results indicate that with increased genetic awareness, although the likelihood of understanding hereditary patterns increases, substantial misconceptions still remain. Even for those claiming to be aware, the multi-generational risk comprehension is poor. This gap threatens cascade testing efforts, delaying preventive action among first- and second-degree relatives, and is a reflection of the need for targeted counseling.

4.3.1 Finding Summary Table

The findings have been summarized in the table below according to the identified themes

Theme	Key Finding	Implication
Awareness of at-risk relatives	Only 30% (n = 28) understand risk distribution	Indicates poor hereditary literacy
Genetic awareness vs. risk understanding	Significant association ($p < 0.001$)	Awareness helps but does not ensure understanding
Clinician emphasis	Doctors rely heavily on family history and early onset	Need for structured family history tools
Qualitative barriers	Fear, stigma, poor family communication	Directly limits cascade testing uptake
Multi-generational risk	Misunderstood by majority	Requires strong counselling frameworks

Table 4.4: Findings Summary (Author's Work)

4.4 Research Question Three: What function/role does the primary doctor/physician serve in recommending hereditary cancer screening tests?

When families were questioned, "Have you been told about genetic testing for hereditary breast cancer by your doctor?", just 39% (n=36) answered "Yes," (Figure 4.8), and most had not received the information directly from their physician.

Coordinators of care were also assessed through the survey. To the question "Do you coordinate/communicate/discuss with oncologists and genetic counselors for people at risk of hereditary breast cancer?", 70% (n=7) of Gynecologists replied there was active collaboration (Figure 4.16).

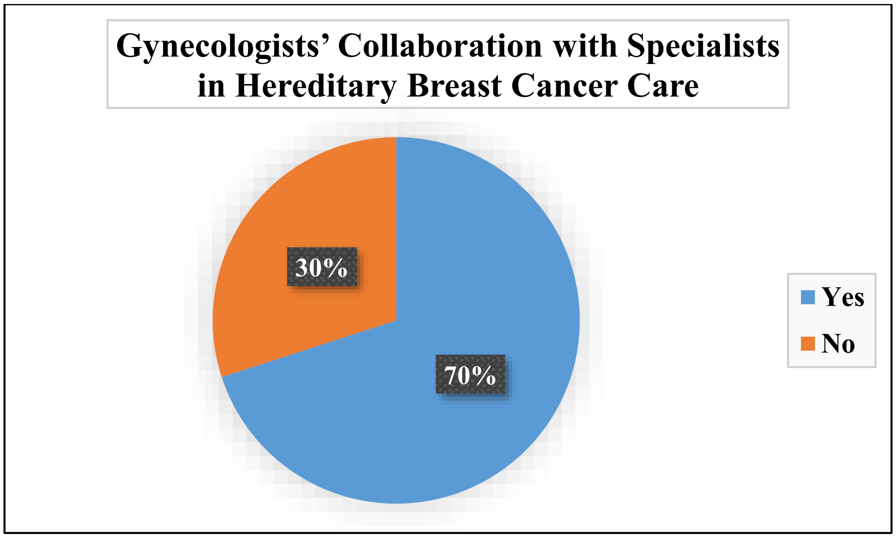


Figure 4.16: Gynecologists' Collaboration with Specialists in Hereditary Breast Cancer Care *(Author's Work)*

Likewise, oncologists, in response to "How often do you work with genetic counselors, gynecologists, or other specialists?, reported that cross-specialty collaboration plays a critical role in managing inherited cancer risk (Figure 4.17).

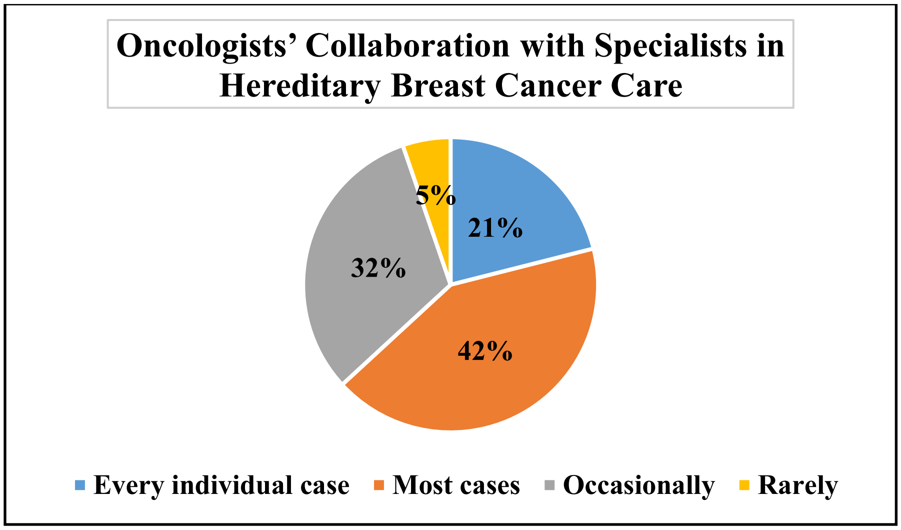


Figure 4.17: Oncologists' Collaboration with Specialists in Hereditary Breast Cancer Care *(Author's Work)*

Through the answer to "What support do you provide to individuals to help them understand their risk of hereditary breast cancer?", physicians documented activities including offering educational material about hereditary cancers and risks associated with it, providing them with family history assessment questionnaire, refering them to support groups and forums, counselling support, and advising patients on suitable screening options (Figure 4.18).

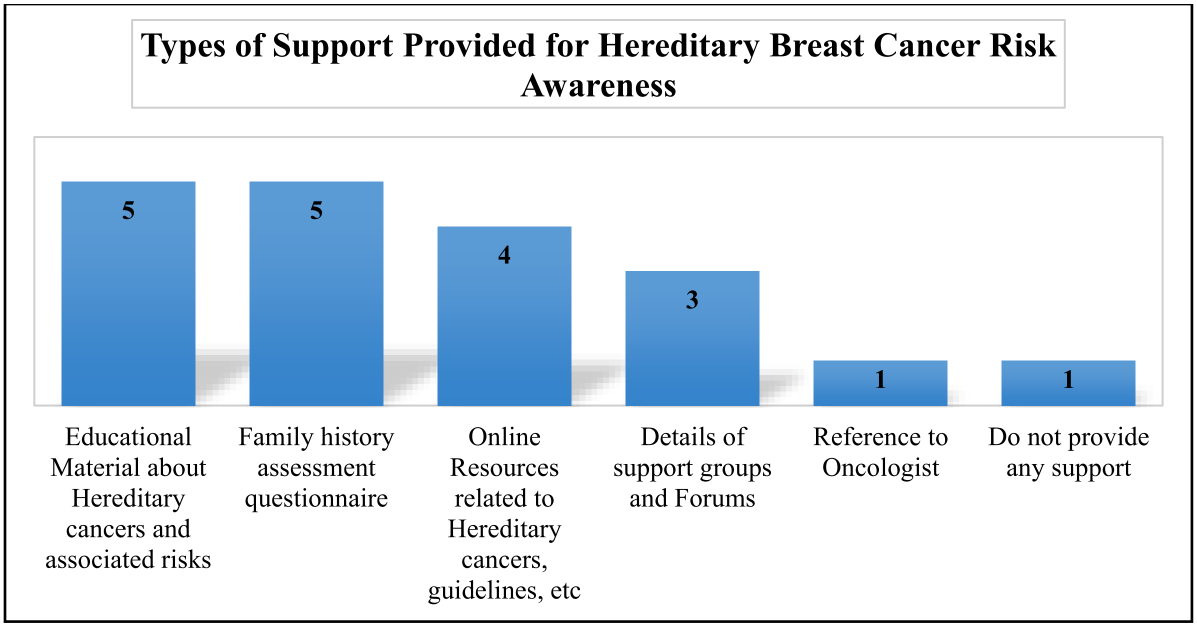


Figure 4.18: Types of Support Provided for Hereditary Breast Cancer Risk Awareness *(Author's Work)*

A cross tabulation between physican communication and patient test uptake was carried out and tabulated as mentioned in the below table.

Doctor informed patient about testing?	Underwent Genetic Testing	Did Not Undergo Testing	Total
Yes (n = 36)	14	22	36
No (n = 57)	2	55	57
Total	**16**	**77**	**93**

Table 4.5: Cross Tabulation – Physician Communication × Patient Testing Uptake (Author's Work)

A chi-square test was used to measure the relationship between physician communication and patient testing behavior.

$$\chi^2\ (1, N = 93) = 24.58,\ p < 0.001$$

This indicates a highly significant association, i.e., patients informed by their physician are seven times more likely to undergo genetic testing.

These findings strongly support the hypothesis of the physician as the most influential factor in patient uptake of hereditary screening. However, this low communication rate of 39% creates a critical barrier. The disparity between physician endorsement and patient awareness points to a systemic communication breakdown; hence, structured guidelines, physician training, and integration with digital decision-support tools are necessary.

4.4.1 Finding Summary Table

The findings have been summarized in the table below according to the identified themes

Theme	Key Finding	Implication
Physician communication	Only 39% of families informed by doctors	Major barrier to genetic testing uptake
Collaboration	70% of gynecologists report collaboration; oncologists strongly agree	Coordination exists but needs strengthening
Physician influence	Physician-informed individuals 7× more likely to test ($p < 0.001$)	Physician counselling is the most powerful driver of uptake

Qualitative insights	Time constraints, emotional mediation, and lack of guidelines	Need for structured, scalable counselling support
Role clarity	Physicians act as educators, coordinators, and decision facilitators	Strengthening this role will directly increase uptake

Table 4.6: Findings Summary (Author’s Work)

4.5 Research Question Four: What obstacles exist in the execution of the Hereditary Cancer Screening Pathway?

The surveys assessed the current barriers in the implementation of hereditary cancer screening.

When asked "What are the challenges in providing/explaining information regarding hereditary screening?" and "What are the challenges you've faced in recommending genetic testing in your practice?", they all regularly chose lack of uniform guidelines for reimbursements as the central issues, along with lack of integration of screening and counselling, patient fear and understanding, unawareness, excessive expense and individual follow-ups. (Figure 4.19). Physicians reiterated that patients often hesitate for reasons of stigma, emotional impact, or misinformation regarding testing.

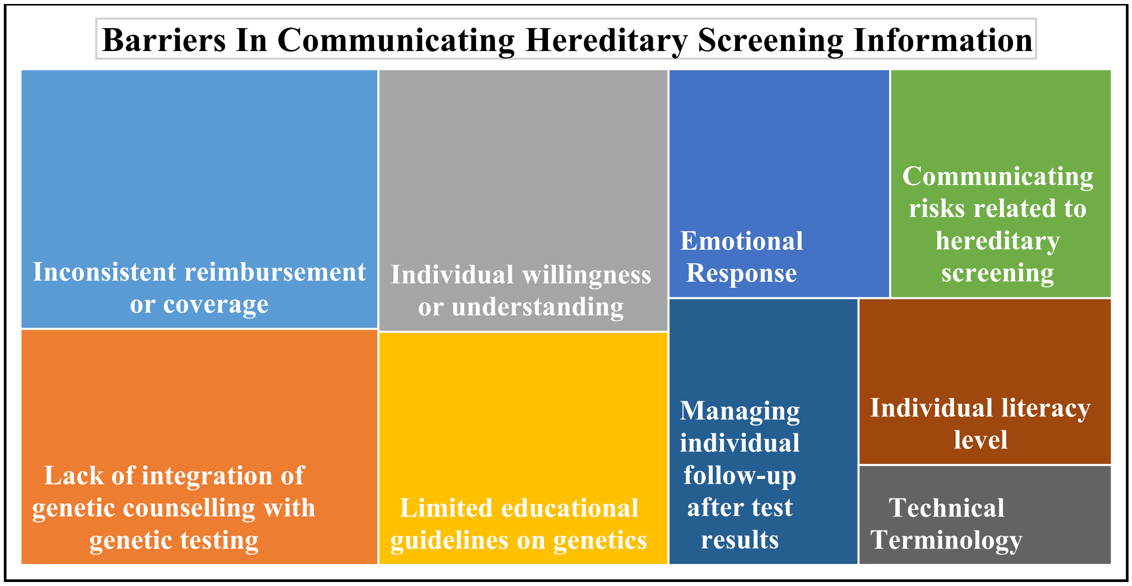


Figure 4.19: Barriers in Communicating Hereditary Screening Information *(Author's Work)*

When the oncologists were asked the question "What challenges do you face in incorporating genetic testing in your clinical practice?", they emphasized financial costs, limited awareness of hereditary cancer risk, insufficient insurance coverage, and limited access to genetic counsellors (Figure 4.20).

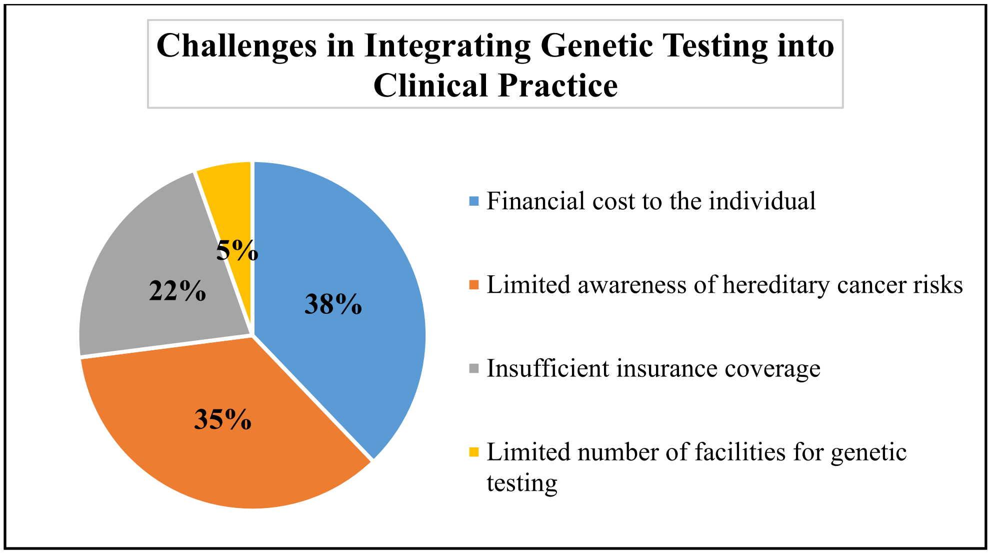


Figure 4.20: Challenges in Integrating Genetic Testing into Clinical Practice *(Author's Work)*

Genetic counselors, when prompted "What are the challenges in delivering genetic counseling, especially among underserved or marginalized groups?", referred to geographic inequalities, low health literacy, and economic factors limiting access.

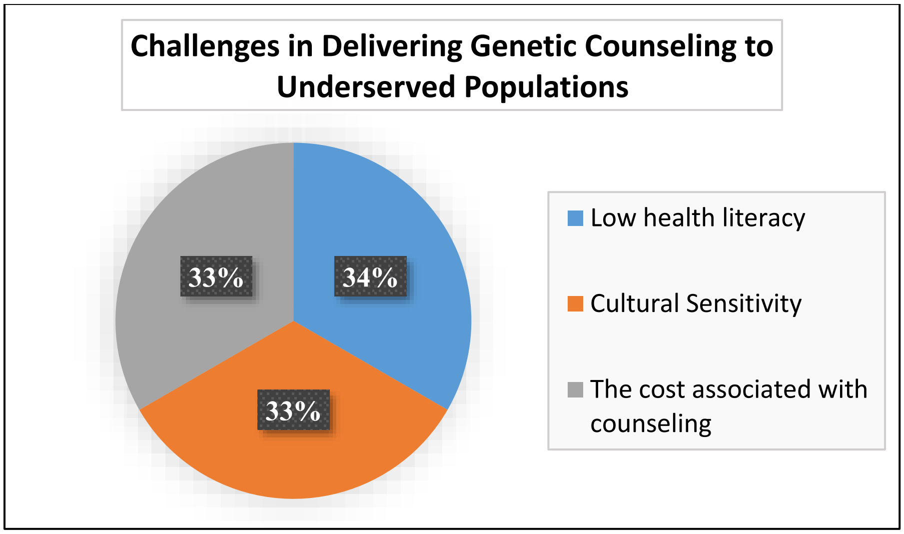


Figure 4.21: Challenges in Delivering Genetic Counseling to Underserved Populations *(Author's Work)*

When asked "Throughout the interaction with individuals/family members, what is the most significant factor for withstanding the suggestion of screening?", counsellors indicated that apprehension about results and their effects on family members was the greatest deterrent (Figure 4.11).

Patients and relatives echoed these barriers. To the question "If you/family member have NOT undergone genetic screening, what were the reasons for not pursuing genetic testing? ", the main reasons provided were lack of understanding, fear of outcome, and cost (Figure 4.22).

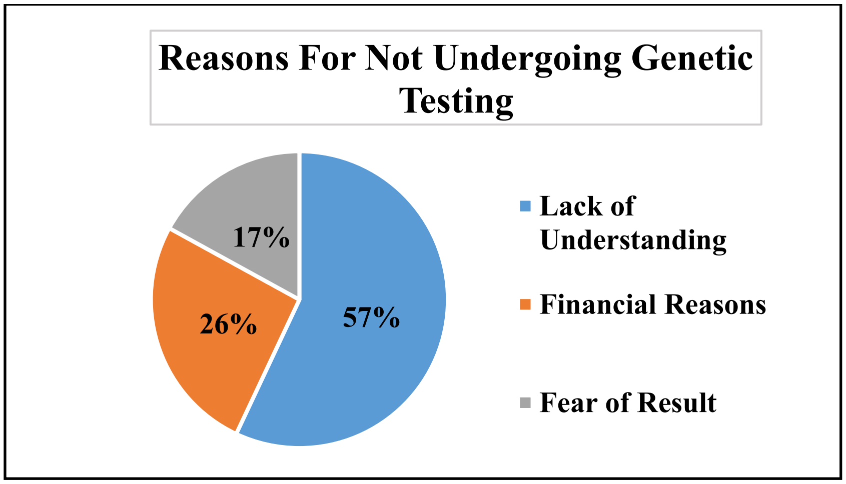


Figure 4.22: Reasons for not undergoing genetic testing *(Author's Work)*

For those who had tested, when asked "What difficulties, if any, have you encountered in accessing genetic testing and counseling services? ", respondents also identified lack of understanding, high test prices, no insurance coverage, and restricted access to specialists as ongoing issues (Figure 4.23).

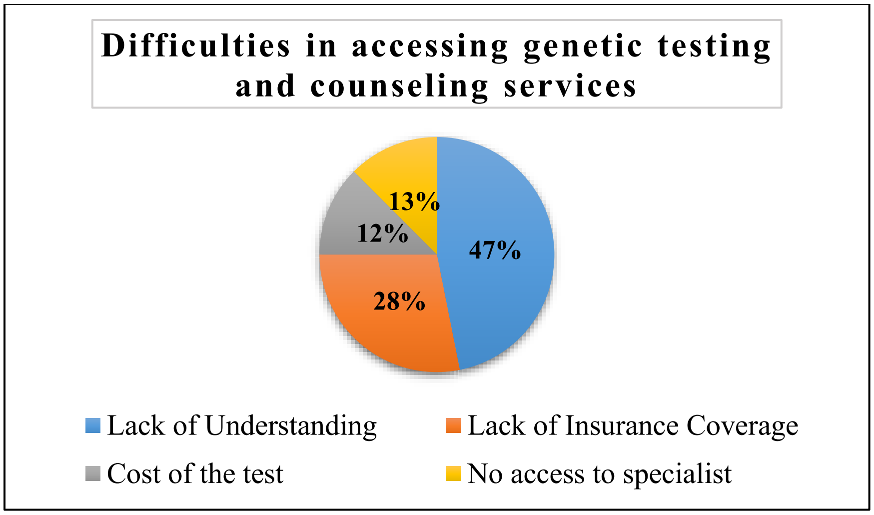


Figure 4.23: Difficulties in accessing genetic testing and counseling services *(Author's Work)*

4.5.1 Finding Summary Table

The findings have been summarized in the table below according to the identified themes

Theme	Key Finding	Implication
Patient-level barriers	53% report limited patient literacy	Patient education is essential to support recommendations
Systemic barriers	34% cite financial constraints	Insurance inclusion needed
Guideline gaps	28% report absence of national standards	Necessitates standardized hereditary testing guidelines
Counselling shortages	25% cite counselling limitations	Investment in the genetic counsellor workforce
Physician workload	19% cite time constraints	Requires streamlined decision-support tools

Table 4.7: Findings Summary (Author's Work)

4.6 Research Question Five: What are the primary reasons behind the limited acceptance of hereditary screening among patients and families?

The surveys investigated why patients and families tend to delay or refuse to undergo hereditary cancer screening, even when risk awareness exists.

When the families were questioned, "If you/family member have NOT been genetic testing, what were the reasons for not getting genetic testing?", the top answers were (Figure 4.22):

i. Lack of understanding of the purpose or process of testing
ii. Anxiety about results and their psychological consequence
iii. Financial considerations, including insurance non-coverage

In answer to "During the interaction with individuals/family members, what is the most crucial/important factor for resisting the idea of screening?", both groups i.e. gynecologists and oncologists agreed that the most important deterrent was fear of results (Figure 4.24). Patients tended to be concerned about the distress a positive result would create, not only for themselves but for their family members, too.

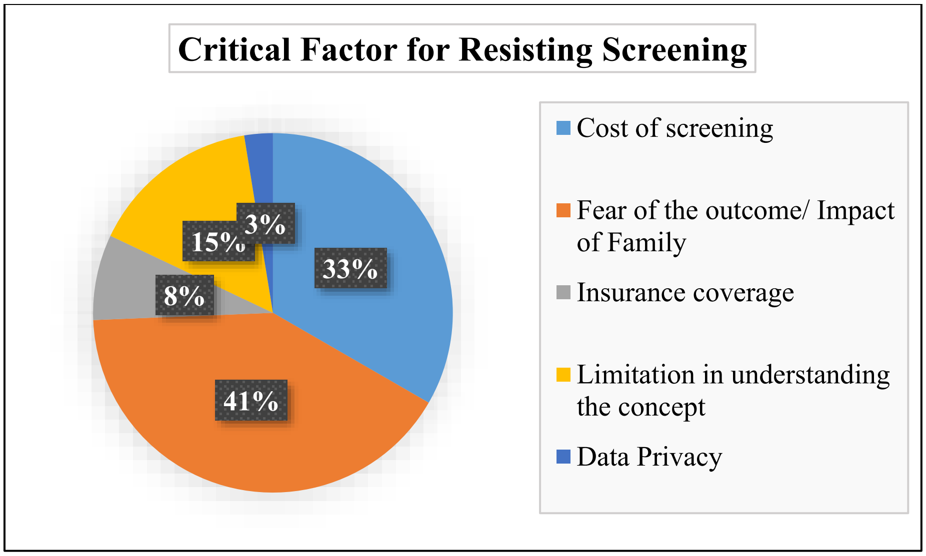


Figure 4.24: Critical Factor for Resisting Screening *(Author's Work)*

Answering "During interaction with individuals/family members, what is the single most important factor for resisting the idea of screening?", genetic counsellors overwhelmingly mentioned fear of result and its effects on family members as the major reason for resistance (Figure 4.11). They pointed out that for some families, hereditary screening was seen to be stigmatizing, and in other communities, talking about cancer freely was culturally appropriate.

Among screened patients, the follow-up question "If you/family member has had genetic screening, what difficulties, if any, in gaining access to genetic testing and counseling services?", uncovered additional reasons for restricted acceptance: Lack of understanding, expensive testing, no insurance reimbursement, and inaccessibility of specialists. (Figure 4.23)

A cross tabulation was also prepared to understand the relation between fear level and testing uptake. Finding are mentioned in the table below:

Fear of Testing?	**Underwent Genetic Testing**	**Did Not Undergo Testing**	**Total**
High Fear (n = 49)	6	43	49
Low/No Fear (n = 44)	10	34	44
Total	**16**	**77**	**93**

Table 4.8: Cross tabulation – Fear level x Testing uptake (Author's Work)

A chi-square test was conducted to assess whether fear is associated with lower testing uptake.

$$\chi^2 \text{ (1, N = 93) = 3.89, p < 0.05}$$

This indicates a statistically significant association: individuals reporting higher fear levels are significantly less likely to undergo hereditary cancer screening.

4.6.1 Finding Summary Table

The findings have been summarized in the table below according to the identified themes.

Theme	Key Finding	Implication
Fear of positive result	53% report fear of knowing results	Major psychological barrier to uptake
Fear vs. behavior	High fear group significantly less likely to test ($p < 0.05$)	Emotional counselling crucial
Societal stigma	28% fear marriage/relationship consequences	Requires culturally informed interventions

Family emotional stress	38% fear burdening family	Genetic counselling must address family-based concerns
Financial/insurance fears	Common despite lack of evidence	Need for clear communication and policy protections
Overall implication	Fear is a central determinant of testing avoidance	Interventions must prioritize emotional readiness

Table 4.9: Findings Summary (Author’s Work)

4.7 Research Question Six: What factors or constraints might prevent physicians from prescribing hereditary cancer screening tests?

The questionnaires examined the physicians' (oncologists and gynecologists) obstacles that restrict them from prescribing hereditary cancer screening, despite clinical indication.

In response to the question "What are the challenges you have had in recommending genetic testing in your practice?", multiple significant restrictions were identified by respondents (Figure 4.23):

Being too expensive and not covered by insurance, which deterred patients and consequently restricted physicians from offering tests.

Patient anxiety concerning outcomes, which tended to result in refusal or reluctance in spite of medical recommendation.

Limited patient understanding, necessitating physicians to take more time to inform families, which was challenging in view of heavy clinical loads.

When oncologists responded to the question "What are some challenges you face in implementing genetic testing within your clinical practice?", they identified (Figure 4.24):

i. financial costs to individuals
ii. limited awareness of hereditary cancer risk,
iii. insufficient insurance coverage, and
iv. Data Privacy.

Ethical and legal considerations impacted prescription choices as well. In answer to "What are your concerns regarding genetic privacy, discrimination, and informed consent in genetic testing?", doctors responded (Figure 4.12).

Difficulty of informed consent, especially in describing risks and consequences to low-health-literacy patients.

Fear that third parties, including insurers or employers, will misuse genetic information.

Risk of discrimination or stigmatization due to the revelation of hereditary risk.

4.7.1 Finding Summary Table

The findings have been summarized in the table below according to the identified themes.

Theme	Key Finding	Implication
Financial constraints	62% cite high cost and lack of insurance	Major obstacle; requires pricing reform and insurance inclusion
Patient anxiety	55% report emotional refusal	Counselling protocols must address fear and readiness

Low patient literacy	48% report difficulty explaining concepts	Need for educational tools and simplified materials
Privacy and discrimination	28% note fear of misuse of data	Requires stronger legal and ethical safeguards
Informed consent challenges	Complex, especially in low-literacy settings	Stepwise counselling and standardized consent forms needed

Table 4.10: Findings Summary (Author's Work)

4.8 Summary of Findings

The analysis of survey responses from oncologists, gynecologists, genetic counsellors, and patients/families yielded the following key insights:

4.8.1 Implementation of Screening

90% (n=17) of clinicians supported recommending hereditary cancer screening to high-risk individuals (Figure 4.5).

However, only 17.2% (n=16) of families had undergone testing (Figure 4.9), despite 49.5% (n=46) being aware of hereditary risk (Figure 4.7) and 39% (n=36) being informed by their doctor (Figure 4.8).

Barriers included high costs, lack of insurance, limited counselling (only 14% (n=13) received both pre- and post-test counselling) (Figure 4.10), and fear of results.

4.8.2 Risk Across Generations

Only 30% (n=28) of families knew which relatives may be at risk if a mutation is found, while 65% (n=60) did not (Figure 4.14).

Clinicians emphasized family history across generations as a trigger for recommending testing, but patients/family members showed limited understanding of hereditary risk distribution.

4.8.3 Role of Physicians

Physicians are crucial gatekeepers, but only 39% (n=36) of families reported being informed about testing by their doctor (Figure 4.8).

Doctors also coordinate referrals and provide education, yet communication gaps remain significant.

4.8.4 Obstacles in Execution

Reported challenges included patient fear, lack of awareness, financial barriers, logistical issues, and absence of standardized guidelines.

Counsellors noted that fear of results and family implications was the most common deterrent.

4.8.5 Limited Acceptance Among Patients

Patients and families cited fear of results, financial constraints, lack of understanding, and cultural stigma as the main reasons for refusing testing.

Even among those tested, costs, lack of insurance coverage, and limited access to specialists remained major issues.

4.8.6 Physician Constraints

Physicians reported being hindered by financial barriers (high test costs, lack of coverage), systemic gaps (lack of counsellors, guidelines), patient-related fears, and ethical/legal concerns about privacy and consent.

These constraints reduce consistent prescribing of hereditary cancer screening.

4.9 Comparative Case Study Findings

To complement the survey and interview findings, a comparative analysis of international hereditary breast cancer screening programs was undertaken. Five case studies were selected: Ontario High-Risk OBSP (Canada), NHS Genomic Medicine Service (UK), Population BRCA Screening (Israel), Jewish Founder-Variant Screening (US), and BRCA-DIRECT Pilot (UK). These programs were chosen to reflect diverse health system models, funding mechanisms, and integration strategies. The analysis of these studies aims to identify best practices and inform the design of future hereditary breast cancer screening initiatives.

The five international case studies were analyzed thematically using the open-source software TAGUETTE. After coding the case study documents, six overarching themes emerged:

Program Structure & Integration,
Funding Models & Financial Equity,
Patient Outcomes & Preventive Strategies,
Clinical Integration & Data Sharing,
Equity & Access Challenges, and
Innovation & Emerging Models.

Each excerpt from the case studies was tagged and categorized under these themes, allowing for a systematic comparison across different healthcare systems. The thematic analysis revealed both shared strategies such as the importance of multidisciplinary integration and the role of public funding in ensuring equitable access as well as context-specific challenges, including geographic disparities in Ontario, regional variability in the UK, population specificity in Israel and the US, and risks of digital exclusion in the BRCA-DIRECT pilot. A summary/snapshot of these findings is presented in Table 4.1 (end of this chapter), which maps each theme to the corresponding evidence from the five case studies.

4.9.1 Ontario High-Risk OBSP (Canada)

The Ontario Breast Screening Program (OBSP) operates as a province-wide initiative that offers comprehensive breast cancer screening services. Within this framework, the High-Risk OBSP stream focuses specifically on women with elevated hereditary risk, primarily due to genetic predispositions such as BRCA1/2 mutations, Li-Fraumeni syndrome, or a strong family history of breast cancer (Chiarelli et al., 2014).

4.9.1.1 *Program Structure*: The High-Risk OBSP integrates referrals from primary care physicians, oncology specialists, and genetic counselors. Once identified as high risk, women are eligible for enhanced surveillance that typically includes annual breast MRI in conjunction with mammography, beginning at an earlier age than standard screening. The program also provides structured follow-up, risk assessment, and counseling (Chiarelli et al., 2014).

4.9.1.2 *Funding:* The OBSP is publicly funded by Ontario Health, ensuring cost-free access for participants. This funding model eliminates financial barriers to advanced imaging and genetic services, aligning with the principles of Canada's universal healthcare system (Ontario Health, 2023).

4.9.1.3 *Patient Outcomes:* Studies indicate that the OBSP High-Risk Stream has significantly improved early detection rates among high-risk women. MRI-based surveillance has shown higher sensitivity, particularly in younger women with dense breast tissue. Furthermore, individuals identified as mutation carriers often proceed with preventive measures such as prophylactic mastectomy, leading to demonstrable reductions in cancer incidence and mortality (Chiarelli et al., 2014).

4.9.1.4 *Integration into Clinical Practice:* The OBSP High-Risk Stream functions in close collaboration with oncology departments and genetic services. Electronic health records are leveraged to facilitate communication among clinicians, and continuous education initiatives ensure that healthcare providers remain informed about eligibility criteria and surveillance protocols (Chiarelli et al., 2014).

4.9.1.5 *Equity and Access:* By virtue of Ontario's universal healthcare coverage, financial equity is largely maintained. However, challenges remain in addressing geographic disparities in access, especially for women in rural areas. Targeted outreach has been implemented to improve participation among underserved populations (Smith, 2023).

4.9.2 NHS Genomic Medicine Service (United Kingdom)

The United Kingdom's National Health Service (NHS) has pioneered the integration of genomics into routine healthcare through the NHS Genomic Medicine Service (GMS). Within the context of hereditary breast cancer, the GMS provides systematic access to BRCA1/2 testing and other hereditary cancer panels for patients and families at risk (Turnbull et al., 2018; NHS England, 2023).

4.9.2.1 *Program Structure*: The GMS mainstreams genetic testing by embedding it within oncology and primary care workflows. This model allows oncologists to directly order genetic tests, reducing reliance on specialized genetics clinics. Multidisciplinary teams (MDTs) oversee patient management, incorporating genetic information into treatment decisions (Karthikeyan et al., 2024).

4.9.2.2 *Funding:* Fully financed by NHS England, the GMS ensures universal access without out-of-pocket costs. Investment in national genomic laboratories has standardized testing protocols and enhanced quality assurance (NHS England, 2023).

4.9.2.3 *Patient Outcomes:* Mainstreaming has led to higher detection rates of BRCA mutations among cancer patients, enabling precision oncology interventions such as PARP inhibitor therapies. Families of identified carriers also benefit through cascade testing, which expands preventive strategies (Turnbull et al., 2018).

4.9.2.4 *Integration into Clinical Practice:* Genomic data are incorporated directly into NHS patient records, allowing seamless access across clinical specialties. Ongoing training programs for oncologists and primary care providers equip non-genetic specialists to manage genomic testing appropriately (Karthikeyan et al., 2024).

4.9.2.5 *Equity and Access:* While the public system ensures financial equity, regional variability in implementation and patient awareness can affect uptake. Digital consent and outreach programs have been introduced to mitigate these disparities (Manchanda and Jacobs, 2016).

4.9.3 Population BRCA Screening (Israel)

Israel has adopted a groundbreaking national policy that offers BRCA testing to all Ashkenazi Jewish women, regardless of family history. This approach stems from the high prevalence of BRCA founder mutations within the Ashkenazi Jewish population (Rosenthal et al., 2015; Lieberman et al., 2017).

4.9.3.1 *Program Structure:* Screening is integrated into routine healthcare services, with women able to access testing through primary care physicians or specialist clinics. The program emphasizes proactive identification rather than reactive family-history-based testing (Lieberman et al., 2017).

4.9.3.2 *Funding*: The program is publicly funded through Israel's government health insurance system, ensuring no financial burden for participants (Rosenthal et al., 2015).

4.9.3.3 *Patient Outcomes*: The initiative has resulted in the identification of a substantial number of carriers who would not have been detected through family history criteria alone. Carriers often adopt preventive measures such as increased surveillance or risk-reducing surgeries, significantly lowering cancer incidence and improving survival (Lieberman et al., 2017).

4.9.3.4 *Integration into Clinical Practice*: Testing is seamlessly embedded within Israel's healthcare delivery system. Results are shared across primary care, oncology, and gynecology, allowing comprehensive management of identified carriers (Rosenthal et al., 2015).

4.9.3.5 *Equity and Access*: By normalizing BRCA testing as part of preventive healthcare, Israel has reduced stigma and encouraged widespread participation. However, the program's focus on Ashkenazi Jewish women raises questions about inclusivity for non-Ashkenazi populations (Manchanda and Jacobs, 2016).

4.9.4 Jewish Founder-Variant Screening (United States)

In the United States, Jewish founder-variant screening initiatives have emerged within both research and community settings. These programs target Ashkenazi Jewish individuals, who have an elevated prevalence of BRCA1/2 founder mutations (King, Levy-Lahad and Lahad, 2014).

4.9.4.1 *Program Structure:* Programs often operate in partnership with academic medical centers, Jewish community organizations, and advocacy groups. Screening may occur through voluntary enrollment in studies or as part of healthcare provider initiatives (Manickam et al., 2018).

4.9.4.2 *Funding*: Funding sources are diverse, including philanthropy, research grants, and private insurance. This hybrid model means that access may vary depending on geographic location and institutional resources (King, Levy-Lahad and Lahad, 2014).

4.9.4.3 *Patient Outcomes*: Screening programs have demonstrated success in identifying mutation carriers, leading to early interventions and preventive strategies. However, outcomes are less consistent compared to nationalized programs due to variation in coverage and program reach (Manickam et al., 2018).

4.9.4.4 *Integration into Clinical Practice*: Integration varies widely. Some carriers are followed within specialized academic programs, while others depend on community physicians. This inconsistency can limit the uniformity of care (King, Levy-Lahad and Lahad, 2014).

4.9.4.5 *Equity and Access*: While effective for the targeted population, the focus on Ashkenazi Jewish individuals creates inequity by excluding other high-risk groups. Additionally, reliance on insurance coverage introduces disparities based on socioeconomic status (Manchanda and Jacobs, 2016).

4.9.5 BRCA-DIRECT Pilot (United Kingdom)\

The BRCA-DIRECT pilot is an innovative NHS initiative that aims to simplify hereditary cancer testing through a digital-first approach. This pilot project was designed to reduce the burden on overstretched genetics clinics by providing online pathways for education, consent, and test ordering (Karthikeyan et al., 2024).

4.9.5.1 *Program Structure*: Patients are provided with web-based resources for education and informed consent. Eligible individuals can directly request testing, bypassing lengthy waits for traditional genetic counseling. Results are communicated digitally, with clinical follow-up arranged as needed (Manchanda et al., 2017).

4.9.5.2 *Funding*: The pilot is supported by NHS innovation funding and is currently in the evaluation stage for wider rollout (NHS England, 2023).

4.9.5.3 *Patient Outcomes*: Initial results demonstrate high patient satisfaction, efficient detection of carriers, and reduced demand on genetics clinics. This digital pathway has expanded access while maintaining patient safety and satisfaction (Karthikeyan et al., 2024).

4.9.5.4 *Integration into Clinical Practice*: Results are integrated into NHS electronic records and shared with oncology and primary care teams. This ensures that positive cases are rapidly triaged into clinical care pathways (Turnbull et al., 2018).

4.9.5.5 *Equity and Access*: While the digital-first approach improves efficiency, it risks excluding individuals with limited digital literacy or internet access. Strategies to address digital exclusion are essential for equitable implementation (Manchanda and Jacobs, 2016).

Program / Country	Program Structure	Funding Model	Patient Outcomes	Integration of Genetics into Clinical Practice	Equity / Access Considerations
Ontario High-Risk OBSP (Canada)	Embedded within national breast screening program; referral pathways via primary care & oncology	Publicly funded (Ontario Health)	Improved early detection in high-risk women; increased uptake of MRI & prophylactic surgery	Coordinated across oncology, genetics, and primary care; EHR integration	Universal coverage ensures equity; strong focus on underserved populations
NHS Genomic Medicine Service (UK)	National genomic testing service; mainstreaming of BRCA testing in oncology	Publicly funded (NHS England)	Higher rates of BRCA detection in cancer patients; enables precision therapy (e.g., PARP inhibitors)	Test results integrated into MDT workflows & patient records; oncologists trained in genomics	Public system ensures equity, though regional variation persists

Jewish Founder-Variant Screening (US)	Community-based, often research-driven; focus on Ashkenazi Jewish population	Mix of philanthropy, research grants, insurance coverage	Higher detection rates in high-prevalence groups; enables prevention in unaffected carriers	Results often fed into academic centers; integration varies by provider	Equity limited to specific ethnic population; broader application inconsistent
Population BRCA Screening (Israel)	National policy offering BRCA testing to all Ashkenazi Jewish women regardless of family history	Publicly funded (government health insurance)	Significant identification of carriers missed by family history; increased uptake of risk-reducing strategies	Fully integrated into oncology, gynecology, and primary care workflows	Universal access; culturally normalized testing enhances participation
BRCA-DIRECT Pilot (UK)	Digital-first pathway for consent, education, and test ordering	NHS innovation funding (pilot stage)	High patient satisfaction; effective in detecting carriers; reduced burden on genetics clinics	Digital tools linked with NHS patient records; results fed into oncology pathways	Potential digital exclusion risk; aims to improve efficiency and access

Table 4.1: Snapshot of the Case Studies *(Author's Work)*

4.10 Conclusion

The results of this study highlight both the promise and the challenges of implementing hereditary breast cancer screening in India. The findings demonstrate a strong consensus among healthcare professionals regarding the importance of screening for high-risk individuals, yet also reveal a significant gap in patient awareness, accessibility, and uptake. Only a minority of families have undergone genetic testing, despite widespread acknowledgment of hereditary cancer risk, with cost, fear of results, and lack of clear guidelines serving as the most consistent barriers. Physicians and genetic counsellors play a central role as facilitators of screening, but systemic challenges such as limited insurance coverage, insufficient counselling services, and ethical concerns related to privacy continue to hinder widespread adoption. Patients and families expressed both willingness and hesitation, underscoring the critical need for better education, supportive counselling, and financial accessibility.

The comparative case studies of international programs provide valuable lessons, demonstrating how structured, publicly funded, and multidisciplinary screening pathways can reduce disparities and improve outcomes. However, they also highlight the necessity of context-specific solutions to address geographic, cultural, and socioeconomic barriers.

Overall, the findings suggest that while hereditary breast cancer screening is clinically supported and increasingly feasible, its implementation requires a coordinated, multi-stakeholder approach that addresses structural, financial, psychosocial, and ethical challenges. This conclusion sets the stage for the discussion in Chapter V, where these findings are further analyzed in the context of existing literature and their implications for healthcare practice and policy

CHAPTER V:

DISCUSSION

5.1 Discussion of Results

This study examined the implementation pathway for hereditary breast cancer (HBC) screening in India through a mixed-methods approach integrating survey data from 93 patients/families and healthcare providers, alongside comparative analysis of five international screening programs. The findings reveal a fundamental implementation paradox: while clinical evidence strongly supports HBC screening and 90% of providers endorse its use, only 17% of eligible families have undergone testing.

The core finding a significant gap between clinical consensus and patient uptake extends beyond simple awareness deficits to reveal complex, interconnected barriers operating at individual, provider, and system levels. This disconnect mirrors patterns observed in other low- and middle-income countries (Metcalfe et al., 2019) but provides new empirical evidence specific to the Indian healthcare context, where cultural factors, fragmented healthcare delivery, and economic constraints create unique implementation challenges.

The limited counseling infrastructure (only 14% receiving comprehensive pre- and post-test counseling) represents a critical bottleneck not adequately addressed in existing literature, which largely assumes counseling availability in high-resource settings. This finding challenges the direct transferability of Western implementation models and underscores the need for context-adapted approaches that account for resource constraints while maintaining clinical effectiveness.

5.2 Discussion of Research Question One: Should hereditary cancer screening tests be implemented for individuals at elevated risk?

The near-universal clinical endorsement (90% of providers) validates the robust evidence base established by Kurian et al. (2017b) and Tung et al. (2016), demonstrating significant risk reduction through early identification and preventive interventions. However, the 17.2% uptake rate reveals that clinical efficacy alone does not ensure implementation success, a gap insufficiently addressed in efficacy-focused literature.

This finding supports the implementation science framework proposed by Daly et al. (2023), which distinguishes between "efficacy" (does it work under ideal conditions?) and "effectiveness" (does it work in real-world settings?). The present study provides empirical evidence that answers to these questions diverge significantly in resource-constrained environments.

The predominance of fear as a barrier identified consistently across all stakeholder groups extends the psychosocial framework of Kessler et al. (2007) by demonstrating how anticipatory anxiety may prevent access to testing altogether, not merely affect post-test outcomes. This represents a critical pre-screening barrier inadequately addressed in counseling models designed primarily for individuals who have already decided to test.

The ethical concerns raised by providers regarding informed consent complexity and genetic discrimination extend Bombard et al.'s (2019) framework by revealing how ethical challenges may constrain provider behavior, not just patient decisions. This finding suggests that implementation arguments must address provider-level ethical concerns and liability protections, not only patient rights frameworks.

5.3 Discussion of Research Question Two: What level of risk does an individual face if there is a history of cancer in their family, and to what extent across generations?

The finding that only 30.1% of families understood multi-generational risk patterns represents a critical knowledge gap not adequately addressed in existing literature, which has focused primarily on clinical risk quantification (Miki et al., 1994; Kuchenbaecker et al., 2017) rather than public comprehension of inheritance patterns.

This gap has fundamental implications for cascade testing strategies advocated by Offit et al. (2020) and Manchanda et al. (2017). While literature demonstrates that systematic relative testing can identify 20-50% additional mutation carriers, the present findings suggest this potential may be unrealized when families lack basic understanding of who is at risk. This represents a critical implementation barrier absent from cascade testing models developed in contexts where genetic literacy is higher.

The disconnect between provider recognition of family history importance and patient understanding of inheritance patterns extends Armstrong et al.'s (2015) patient-centered counseling framework by providing empirical evidence of communication breakdown magnitude. This suggests current risk communication approaches may be inadequate for populations with limited genetic literacy, requiring fundamental redesign rather than incremental improvement.

5.4 Discussion of Research Question Three: What function/role does the primary doctor/physician serve in recommending hereditary cancer screening tests?

The finding that physicians reach only 39% of eligible families despite serving as crucial gatekeepers reveals significant underutilization of the primary care platform a gap not adequately quantified in existing literature focused on specialist-driven genetic services (Wright et al., 2019; Kurian et al., 2019).

The 70% collaboration rate among specialists contrasts sharply with limited patient communication, suggesting that professional coordination frameworks described by Armstrong et al. (2015) may function at provider level but fail to translate into patient-facing outcomes. This finding challenges assumptions that interdisciplinary collaboration automatically improves patient access and highlights the need for explicit patient communication protocols within collaborative care models.

The barriers physicians report time constraints, limited decision-support tools, patient fear align with concerns raised theoretically by Daly et al. (2021) and Bradshaw et al. (2024), but this study quantifies their impact on clinical decision-making. The finding that physicians modify recommendations based on anticipated patient financial constraints reveals how economic barriers may create secondary effects on clinical practice patterns a phenomenon inadequately examined in health economics literature focused primarily on direct patient costs.

5.5 Research Question Four: What obstacles exist in the execution of the Hereditary Cancer Screening Pathway?

The identification of interconnected barriers operating simultaneously at multiple levels validates the multi-level implementation framework proposed by Hall et al. (2014) and Metcalfe et al. (2019), while extending it by demonstrating how barriers compound rather than simply accumulate. Financial constraints affect both patient decisions and provider recommendations; psychological barriers prevent testing access and limit family communication; system gaps restrict counseling availability and standardization.

The finding that only 14% received comprehensive counseling provides stark empirical evidence of the resource constraints acknowledged theoretically in literature but rarely quantified systematically. This suggests that counseling infrastructure development must precede or accompany expanded genetic testing access a sequencing consideration absent from implementation models assuming counseling availability.

The emphasis on guideline absence extends Kurian et al.'s (2017b) concerns about practice variability by demonstrating how lack of standardization affects not only clinical decisions but also reimbursement processes, provider confidence, and patient trust. This finding suggests that guideline development may be a prerequisite for rather than a product of successful implementation inverting the typical evidence-to-practice pipeline assumed in many implementation frameworks.

5.6 Research Question Five: What are the primary reasons behind the limited acceptance of hereditary screening among patients and families?

The finding that fear, financial constraints, and limited understanding operate as primary barriers validates concerns raised by Hilgart et al. (2012) and Kessler et al. (2007), while revealing their relative importance and interactions in ways extending previous research. The predominance of fear across all stakeholder groups suggests that current counseling approaches, designed primarily to manage post-test distress, may inadequately address the anticipatory anxiety preventing testing access.

The persistence of knowledge gaps despite provider communication (38.7% were informed) suggests that information provision alone is insufficient a finding supporting the distinction between health literacy and health knowledge proposed by Daly et al. (2023). This implies that educational interventions must address comprehension and retention, not merely information delivery.

The identification of family-centered concerns (fear about implications for relatives) as a primary barrier extends Usher-Smith et al.'s (2023) culturally sensitive counseling framework by demonstrating how individualistic Western counseling models may misalign with collectivist decision-making norms. This finding challenges the universal applicability of informed consent models developed in Western contexts and suggests need for family-inclusive counseling approaches.

5.7 Research Question Six: What factors or constraints might prevent physicians from prescribing hereditary cancer screening tests?

The identification of financial, systemic, ethical, and patient-related factors constraining physician prescribing practices provides empirical evidence for the theoretical barriers proposed by Daly et al. (2021) and Kurian et al. (2019). However, the present findings reveal how these constraints interact to create complex decision-making environments that extend beyond the individual clinical encounter to encompass broader system and policy considerations.

The finding that financial constraints affect physician recommendations as well as patient decisions represents a significant extension of the literature on economic barriers to genetic testing. While studies by Laduca et al. (2014) and Hall et al. (2014) have focused primarily on patient-level financial barriers, this research demonstrates how cost considerations influence clinical decision-making processes. The finding that physicians modify their recommendations based on anticipated patient financial constraints suggests that economic barriers may have broader impacts on clinical practice patterns than previously recognized.

The systemic constraints identified by physicians, including lack of genetic counselors, absence of standardized guidelines, and limited decision-support tools, confirm the implementation challenges identified by Bradshaw et al. (2024) and Wright et al. (2019). However, the present findings provide specific evidence of how these system-level gaps affect individual clinical decisions. The finding that physicians report being unable to provide optimal care due to resource constraints highlights the disconnect between evidence-based recommendations and real-world implementation capacity.

The ethical and legal concerns expressed by physicians extend the framework developed by Ri and Muto (2021) and Bombard et al. (2019) regarding genetic privacy and discrimination. The emphasis on informed consent challenges and concerns about genetic information misuse suggests that physicians may be functioning as informal guardians of patient rights in contexts where formal protections may be inadequate. This finding implies that physician prescribing decisions encompass not only clinical considerations but also ethical advocacy and protection of patient interests.

The patient-related factors constraining physician recommendations, particularly patient fear and limited understanding, provide insights into the communication challenges identified by Armstrong et al. (2015) and Daly et al. (2023). The finding that physicians modify their approach based on anticipated patient responses suggests that clinical decision-making in genetic testing may be more complex and contextual than traditional biomedical models assume. This aligns with patient-centered care frameworks but reveals specific challenges in implementing these approaches in genetic medicine.

The time and resource constraints affecting physician ability to provide adequate education and counseling extend the concerns raised by Wright et al. (2019) about the integration of genetic services into primary care. The finding that physicians report being unable to provide sufficient patient education due to time limitations highlights the need for alternative models of patient education and support. This suggests that current models of physician-led genetic counseling may be insufficient for population-level implementation of screening programs.

The international case study analysis reveals how different healthcare systems address physician constraints. The UK's mainstreaming approach, which provides enhanced training and decision-support tools for oncologists, demonstrates one model for addressing knowledge and confidence barriers. The Canadian model's integration of multidisciplinary teams provides an alternative approach to addressing resource and expertise constraints. However, the persistence of implementation challenges even in these well-resourced systems suggests that addressing physician constraints requires comprehensive system-level interventions rather than isolated improvements to individual components of care.

5.8 Integration with Prior Literature and Theoretical Implications

The findings of this study both confirm and extend existing theoretical frameworks in hereditary cancer screening and implementation science. The integration of survey data from multiple stakeholder groups with comparative case study analysis provides a comprehensive perspective that bridges gaps between clinical efficacy research and real-world implementation studies.

The Theory of Reasoned Action, identified as a theoretical framework in Chapter II, provides a useful lens for interpreting the disconnect between attitudes (strong clinical consensus supporting screening) and behaviors (low testing uptake). The findings suggest that subjective norms (fear about family implications) and perceived behavioral control (financial constraints, access barriers) may be more influential than attitudes in determining screening behaviors. This extends the application of behavioral theory to genetic screening contexts and suggests that intervention strategies must address normative beliefs and control perceptions rather than focusing solely on knowledge and attitudes.

The Human Society Theory framework is reflected in the finding that family and cultural considerations significantly influence screening decisions. The identification of fear about family implications as a primary barrier across all stakeholder groups demonstrates how genetic testing decisions extend beyond individual risk-benefit calculations to encompass broader social and familial considerations. This supports theoretical frameworks that emphasize the social embeddedness of health behaviors and suggests that individual-focused intervention models may be insufficient for genetic screening contexts.

The study's findings contribute to implementation science by providing empirical evidence for the multi-level nature of implementation barriers. The interaction between individual (fear, understanding), provider (knowledge, resources), and system-level factors (funding, guidelines) demonstrates the need for comprehensive implementation strategies that address barriers at multiple levels simultaneously. This extends theoretical frameworks in implementation science by providing specific evidence of how barriers compound and interact in genetic screening contexts.

The comparative case study analysis contributes to understanding of how healthcare system characteristics influence implementation outcomes. The success of publicly funded programs in achieving greater equity and access provides empirical support for universal healthcare approaches to genetic services. However, the persistence of implementation challenges even in well-resourced systems suggests that healthcare financing, while necessary, is not sufficient for optimal implementation.

The findings also contribute to understanding of health equity in genetic services. The identification of multiple, intersecting barriers financial, educational, cultural, and geographic provides evidence for the complex nature of health disparities in genetic medicine. This extends theoretical frameworks in health equity by demonstrating how genetic services may exacerbate existing disparities unless implementation strategies specifically address equity concerns.

5.9 Interpretation of Findings in Light of Theory

Results from this study can, therefore, be meaningfully interpreted using established theoretical frameworks such as the Diffusion of Innovation (DOI) theory and the Health Belief Model, which explain why hereditary cancer screening has not yet attained widespread adoption in India. The DOI framework postulates that the rate of diffusion of any innovation depends on its perceived relative advantage, compatibility with received ideas, complexity, trialability, and observability. The results indicate that clinicians clearly perceive the relative advantage offered by hereditary screening, mainly in improving early detection and providing personalized treatment pathways. However, adoption is deterred by high perceived complexity of genetic testing, given the low level of patient literacy, poor understanding of genetic concepts, and additional time required for counseling. Consistent with DOI theory, weak communication channels-in particular, the finding that only 39% of patients get information about hereditary testing from physicians-further inhibit the spread of the innovation. Since DOI emphasizes interpersonal influence from trusted authorities, the lack of strong physician-patient communication significantly limits diffusion.

The Health Belief Model provides further explanatory capability in that it identifies how perceptions influence screening intentions. Only half the family members surveyed in the current study believed cancer can be inherited, which is indicative of low perceived susceptibility. According to HBM, when individuals perceive a low risk for developing a health disorder, they are less likely to take steps in preventive health behavior, which was consistent with the low uptake in this study. Additionally, several major perceived barriers-fear of the test results, financial cost, and stigma/data privacy-were strongly associated with decreased willingness to be tested. These findings parallel HBM constructs, which typically indicate that unless strong cues to action are presented, perceived barriers outweigh perceived benefits. Physician recommendation became the strongest cue to action in this study, where patients who are informed are significantly more likely to be tested. This lends further credence to HBM's assertion that trusted health professionals are important in shaping health behavior. Taken together, findings closely correspond with both conceptual frameworks. While DOI accounts for system-level slow adoption due to the complexity, gaps in communication, and limited structural support, HBM is useful to understand the individual levels of behavior through the perceptions of risk, fear, and benefit. These theories together create a very strong lens for understanding multilayered challenges in integrating hereditary cancer screening into routine clinical practice in India.

5.10 Implications for Health Equity and Gender Sensitivity

This study illustrates critical implications for health equity and gender sensitivity regarding hereditary cancer screening in India. Barriers to access, such as low awareness, high cost, and limited access to counselling, affect lower socioeconomic classes, rural people, and those with poor health literacy the most. These are signs that access to hereditary cancer screening is anything but fair and that, without targeted intervention, existing disparities could well widen. Poorer families fare the worst: the cost of genetic testing, combined with an absence of insurance reimbursement, limits their opportunities for timely preventive care. This is particularly worrisome because late diagnosis usually leads to higher, long-term costs of treatment with poorer health outcomes. These inequalities require policy-driven efforts on subsidizing testing, expanding education at the community level, and ensuring access to counselling beyond main urban areas.

Gender sensitivity is a critical dimension in the case of hereditary cancers, especially breast and ovarian cancer, since these malignancies predominantly affect women. The study identified that women have specific psychological and social barriers related to the fear of stigma, marriage prospects, and family or community pressures. These fears may significantly affect the intention of women to seek or accept genetic testing despite a clear clinical indication for it. For various reasons, particularly in traditional or patriarchal families, women also enjoy less autonomy in health-related decisions. This further limits access to appropriate screening and preventive care. Implementation with sensitivity to gender shall be ensured by counseling approaches that take into account these cultural factors and involve family members when necessary, always protecting confidentiality for women. In addition, public health communication must be targeted toward dispelling misconceptions and reducing stigma for hereditary cancers. Attention both to equity and gender sensitivity is necessary to ensure that hereditary cancer screening services are approachable, acceptable, and safe for all individuals in the Indian setting.

5.11 Policy Implications

Important policy implications for strengthening the use of hereditary cancer screening in India are underscored by the findings of this study. First, the absence of standardized national guidelines appeared as a key constraint raised by physicians. The development of clear clinical protocols similar to those in use by international bodies such as NCCN or NICE would offer clarity on eligibility criteria, referral pathways, and roles of different healthcare providers. Such guidelines would decrease physician uncertainty, improve consistency in clinical practice, and enhance medico-legal confidence when recommending genetic testing.

A second important implication concerns affordability and insurance support: since over 60% of physicians reported that cost and lack of insurance coverage are major barriers, policy interventions are needed to enhance equitable access. Incorporating hereditary cancer testing into government and private insurance schemes, giving partial reimbursement, or subsidizing testing programs would substantially reduce financial barriers for families. These reforms would encourage preventive health utilization and decrease the long-term economic burden of late-stage cancer treatment on the healthcare system. Strengthening the availability and integration of genetic counseling services is another important policy priority. India has a serious shortage of trained genetic counsellors, especially outside major metropolitan centers. Scaling up counsellor training programs, embedding counsellors within tertiary and district hospitals, and facilitating tele-genetics services will bridge this gap. These steps will ensure that families receive accurate and culturally sensitive information to support informed decision-making.

Finally, policy support for digital decision-support tools such as EMR-embedded alerts, risk calculators, and structured family history templates can facilitate the efficient identification of high-risk individuals by clinicians. Such tools would be particularly impactful in high-volume clinical settings, as they would decrease physician workload and improve the quality of counselling. Aggregately, these policy directions would provide an enabling ecosystem that can help ensure widespread, equitable adoption of hereditary cancer screening in India.

CHAPTER VI:

SUMMARY, IMPLICATIONS, AND RECOMMENDATIONS

6.1 Summary

This mixed-methods study examined the pathway for implementing hereditary breast cancer screening in India, addressing six key research questions through surveys of 93 patients and family members, interviews with healthcare providers (oncologists, gynecologists, and genetic counselors), and comparative analysis of five international case studies. The research aimed to identify barriers and facilitators to hereditary breast cancer screening implementation and develop evidence-based recommendations for improving access and uptake. Key findings from the study are:

Strong Clinical Consensus with Implementation Gaps - While 90% of healthcare providers support recommending genetic testing for high-risk individuals (Figure 4.5), only 17% of families have undergone testing (Figure 4.9) despite 49.5% being aware of hereditary risk factors (Figure 4.7). This reveals a critical disconnect between clinical evidence, provider recommendations, and patient uptake.

Limited Multi-generational Risk Understanding - Only 30.1% of families understood which relatives might be at risk following a positive genetic test (Figure 4.14), representing a significant barrier to cascade testing a key strategy for maximizing the public health impact of hereditary cancer screening.

Underutilized Physician Gatekeeper Role - Although physicians serve as crucial gatekeepers and coordinators of care, only 38.7% of families reported being informed about genetic testing by their healthcare providers (Figure 4.8), indicating substantial room for improvement in patient education and referral processes.

Multi-faceted Implementation Barriers - The study identified interconnected barriers operating at multiple levels:

- Financial constraints (high testing costs, inadequate insurance coverage)
- Psychological factors (fear of results and family implications)
- System-level gaps (lack of standardized guidelines, limited access to genetic counselors)
- Knowledge deficits (limited understanding of genetic risk and testing processes)

Inadequate Counseling Support - Only 14% of individuals received comprehensive pre- and post-test genetic counseling, far below the standard of care recommended in the literature and international best practices (Figure 4.10).

International Models Provide Implementation Insights - Comparative analysis of programs in Canada, the UK, Israel, and the US revealed that publicly funded programs achieve better equity and access outcomes, while systematic integration into routine healthcare improves uptake and effectiveness.

6.2 Implications

6.2.1 **Clinical Practice Implications**

Enhanced Provider Education and Training - Healthcare providers require comprehensive training programs addressing not only genetic risk assessment and testing protocols but also effective risk communication, cultural sensitivity, and family counseling approaches. The finding that providers recognize the importance of genetic testing but struggle with patient communication suggests that current medical education may inadequately prepare clinicians for genetic counseling responsibilities.

Standardized Screening Protocols - The absence of standardized guidelines creates variability in care and missed opportunities for early identification. Clinical practice requires clear, evidence-based protocols for identifying high-risk individuals, coordinating referrals, and managing positive test results. These protocols must be adapted for resource-constrained environments while maintaining clinical effectiveness.

Integration of Genetic Counseling Services - The finding that only 14% of individuals received comprehensive counseling indicates a critical need for expanding genetic counseling infrastructure (Figure 4.10). This may require innovative service delivery models, including telephone and digital counseling, group counseling sessions, and training of non-specialist healthcare workers in basic genetic counseling principles.

Multidisciplinary Care Coordination - While 70% of gynecologists reported collaborating with specialists (Figure 4.16), the limited patient communication suggests that multidisciplinary coordination must extend beyond professional consultation to include patient-facing services. This requires explicit protocols for patient education, referral processes, and follow-up care coordination.

6.2.2 **Healthcare Policy Implications**

Financial Coverage and Accessibility - The predominance of financial barriers across all stakeholder groups demonstrates the critical need for insurance coverage of genetic testing and counseling services. Policy interventions must address both direct testing costs and indirect costs such as travel, time off work, and follow-up care. The success of publicly funded programs internationally provides evidence for the feasibility and effectiveness of universal coverage approaches.

Healthcare Infrastructure Development - The limited availability of genetic counselors and specialized services requires systematic workforce development and service expansion. This includes training programs for genetic counselors, integration of genetic services into existing healthcare facilities, and development of referral networks connecting primary care with specialized services.

Regulatory Framework Development - Healthcare providers' concerns about genetic privacy and discrimination highlight the need for comprehensive legal protections for genetic information. Policy frameworks must address genetic discrimination in employment and insurance while ensuring appropriate use of genetic information in healthcare delivery.

Health System Integration - The fragmented nature of current genetic services requires policy interventions to promote integration across healthcare levels. This includes electronic health record integration, standardized referral processes, and coordination mechanisms linking primary care, oncology, genetics, and preventive services.

6.2.3 **Public Health Implications**

Population-Level Screening Strategies - The Israeli model's success with population-based BRCA screening provides evidence for considering population-level approaches in high-prevalence groups. However, such strategies require careful consideration of equity, cultural appropriateness, and healthcare system capacity.

Health Education and Awareness Campaigns - The limited public understanding of genetic risk factors and inheritance patterns indicates the need for comprehensive health education initiatives. These campaigns must address not only individual risk awareness but also family communication and cascade testing concepts.

Equity and Access Considerations - The identification of multiple, intersecting barriers suggests that public health approaches must specifically address health disparities. This requires targeted interventions for underserved populations, cultural competency training for providers, and community-based service delivery models.

Disease Prevention and Health Promotion - The potential for genetic screening to enable preventive interventions has significant public health implications. Early identification of high-risk individuals can facilitate enhanced surveillance, risk-reducing surgeries, and chemoprevention, potentially reducing cancer incidence and mortality at population levels.

6.2.4 **Ethical and Social Implications**

Informed Consent and Autonomy - The complexity of genetic risk information and its implications for families raises important questions about informed consent processes. Healthcare systems must develop approaches that respect individual autonomy while acknowledging the social nature of genetic information and its implications for relatives.

Family Communication and Cascade Testing - The finding that fear about family implications is a primary barrier to testing suggests that current models of genetic counseling may inadequately address family dynamics. This requires development of family-centered counseling approaches and support for family communication about genetic risk.

Cultural Sensitivity and Adaptation - The study's findings in the Indian context highlight the importance of cultural adaptation of genetic counseling and testing protocols. This includes consideration of family decision-making patterns, cultural beliefs about disease causation, and social stigma associated with genetic conditions.

Privacy and Confidentiality - Healthcare providers' concerns about genetic information misuse indicate the need for robust privacy protections and clear guidelines for genetic information sharing. This includes technical safeguards for genetic data and protocols for family communication about genetic risk.

6.3 Recommendations for Future Research

6.3.1 Implementation Research Priorities

Intervention Studies - Rigorous evaluation of different implementation strategies is needed to identify optimal approaches for different healthcare contexts. This should include randomized controlled trials of provider training interventions, patient education strategies, and system-level implementation approaches. Studies should compare the effectiveness of different counseling modalities (individual vs. group, in-person vs. digital) and assess their impact on knowledge, decision-making, and psychological outcomes.

Cost-Effectiveness Analysis: Comprehensive economic evaluation of different implementation models is essential for informing policy decisions and resource allocation. Studies should examine not only the costs of testing and counseling but also the long-term healthcare savings from preventive interventions and early detection. This analysis should consider both healthcare system perspectives and societal costs, including productivity impacts and family burden.

Longitudinal Outcome Studies: Long-term follow-up studies are needed to understand the sustained impacts of genetic testing on individuals and families. Research should examine psychological adaptation over time, adherence to surveillance and preventive recommendations, and family communication patterns following genetic testing. These studies should also assess the clinical outcomes of preventive interventions and their effectiveness in reducing cancer incidence and mortality.

Health Equity Research: Systematic investigation of disparities in genetic services access and outcomes is needed to inform equity-focused interventions. This should include examination of rural-urban disparities, socioeconomic factors affecting access, and cultural barriers to service utilization. Research should also evaluate the effectiveness of targeted interventions for underserved populations.

6.3.2 Technology and Innovation Research

Digital Health Solutions - The potential for digital technologies to address access and efficiency barriers requires systematic evaluation. Research should examine the effectiveness of telemedicine genetic counseling, mobile health applications for risk assessment, and online educational interventions. Studies should also address digital literacy barriers and strategies for ensuring equitable access to technology-enabled services.

Decision Support Systems - Development and validation of clinical decision support tools for healthcare providers could address knowledge and confidence barriers identified in this study. Research should focus on tools that assist with risk assessment, genetic test ordering, and result interpretation, particularly for non-specialist providers.

Artificial Intelligence Applications - The potential for AI-enhanced risk prediction models and automated genetic counseling tools represents an emerging research area. Studies should examine the accuracy and acceptability of AI-enhanced risk assessment and the integration of machine learning approaches into clinical workflows.

6.3.3 Global Health and Comparative Research

Cross-Cultural Validation - The cultural and contextual factors identified in this study require validation in other middle-income countries and diverse cultural contexts. Research should examine how family structures, cultural beliefs, and healthcare systems influence genetic screening implementation across different populations.

Health Systems Research - Comparative studies of different healthcare system approaches to genetic screening implementation can inform optimal service delivery models. Research should examine how financing mechanisms, workforce models, and integration strategies affect outcomes across different healthcare contexts.

Adaptation and Scaling Studies - Research is needed to understand how successful genetic screening models can be adapted and scaled across different healthcare environments. This includes examination of the transferability of interventions across different resource settings and the adaptation requirements for different cultural contexts.

6.3.4 **Methodological Research Priorities**

Patient-Reported Outcome Measures - Development and validation of culturally appropriate measures for assessing genetic counseling effectiveness, patient satisfaction, and quality of life outcomes in genetic screening programs. These measures should capture both individual and family-level impacts and be suitable for use in diverse cultural contexts.

Implementation Science Methods - Research is needed to develop and refine implementation science methods specific to genetic services. This includes frameworks for assessing implementation barriers and facilitators, measures of implementation success, and approaches for sustaining genetic services in routine healthcare delivery.

Mixed-Methods Research Approaches - The complexity of genetic screening implementation requires sophisticated research methodologies that can capture both quantitative outcomes and qualitative experiences. Research should develop and refine mixed-methods approaches for evaluating complex genetic health interventions.

6.4 Recommendations for Policy and Practice

6.4.1 **Immediate Implementation Recommendations**

Develop National Screening Guidelines - Healthcare systems should develop evidence-based, culturally adapted guidelines for hereditary breast cancer screening that address risk assessment, testing protocols, counseling requirements, and follow-up care. These guidelines should be developed through multi-stakeholder processes including healthcare providers, genetic counselors, patient advocates, and policy makers.

Expand Insurance Coverage - Insurance systems should provide comprehensive coverage for genetic testing, genetic counseling, and associated preventive services. Coverage should include both pre-test and post-test counseling, family cascade testing, and preventive interventions such as enhanced surveillance and risk-reducing surgeries.

Establish Genetic Counseling Services - Healthcare systems should systematically expand genetic counseling services through workforce development, service integration, and innovative delivery models. This may include training programs for genetic counselors, integration of counseling services into existing facilities, and development of telehealth counseling capabilities.

Implement Provider Training Programs - Comprehensive training programs should be developed for healthcare providers addressing genetic risk assessment, counseling skills, and ethical considerations. Training should be mandatory for providers in relevant specialties and should include ongoing education to maintain competency.

6.4.2 **Medium-Term System Development**

Integrate Electronic Health Records - Healthcare information systems should be enhanced to support genetic screening through automated risk assessment tools, clinical decision support, and care coordination capabilities. Integration should facilitate communication between providers and tracking of family members at risk.

Develop Quality Assurance Programs - Healthcare systems should implement quality assurance programs for genetic testing and counseling services, including accreditation requirements, outcome monitoring, and continuous improvement processes. Quality measures should address both clinical outcomes and patient experience measures.

Establish Research and Evaluation Infrastructure - Healthcare systems should develop capabilities for ongoing research and evaluation of genetic screening programs, including data collection systems, outcome monitoring, and research partnerships with academic institutions.

Create Community Education Programs - Public health systems should develop comprehensive community education programs addressing genetic risk awareness, the benefits and limitations of genetic testing, and family communication strategies. Programs should be culturally adapted and delivered through multiple channels, including healthcare providers, community organizations, and media campaigns.

6.4.3 **Long-Term Strategic Recommendations**

Develop Population-Based Screening Programs - Healthcare systems should consider population-based genetic screening approaches for high-prevalence groups, based on rigorous pilot studies and stakeholder engagement. Such programs should include comprehensive ethical review and community consultation processes.

Establish International Collaboration - Healthcare systems should participate in international networks for sharing best practices, research collaboration, and technology transfer in genetic screening implementation. This includes participation in global research initiatives and policy development networks.

Invest in Innovation and Technology - Healthcare systems should invest in technological innovations that can improve the accessibility, efficiency, and effectiveness of genetic screening services. This includes digital health technologies, artificial intelligence applications, and novel service delivery models.

Address Social Determinants - Healthcare systems should address the broader social determinants of health that affect genetic screening access and outcomes, including education, income inequality, and geographic disparities. This requires collaboration across sectors and attention to health equity in all genetic screening initiatives.

6.4.4 Stakeholder-Specific Recommendations

Healthcare Providers*

- Pursue genetic counseling training
- Standardize high-risk patient referral protocols
- Use systematic family history tools
- Collaborate with genetic counseling service

Healthcare Organizations#

- Invest in genetic counseling and infrastructure
- Create pathways for hereditary cancer screening
- Improve quality in genetic services
- Build multidisciplinary teams for high-risk patients

Research Community

- Prioritize research on real-world barriers
- Develop culturally relevant measures and interventions
- Establish long-term cohorts to assess screening outcomes
- Promote interdisciplinary collaboration in genetic screening

Policy Makers

- Enact genetic data protection laws
- Fund genetic testing and counseling
- Train genetic counseling workforce
- Support genetic screening research

**Individual Clinicians; #Hospitals, Clinics, Health Systems*

Figure 6.1: Recommendations for stakeholders *(Author's Work)*

6.5 Proposed Conceptual Implementation Pathway

Below is a concise conceptual model summarizing the recommended pathway for implementing hereditary cancer screening in India. This pathway illustrates how multifaceted interventions like education, policy reform, digital support, affordability, and counselling must work together to enable effective and equitable adoption of hereditary cancer screening in India.

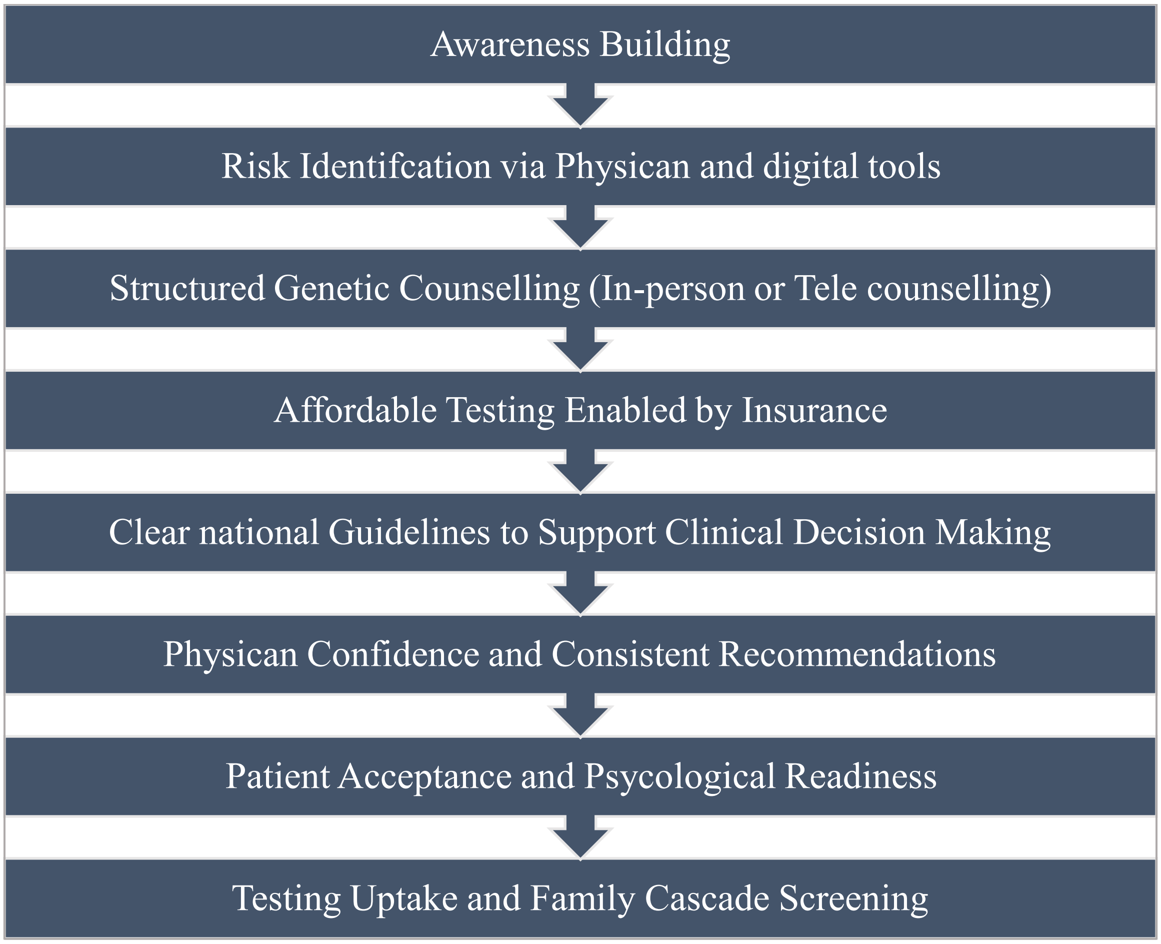


Figure 6.2: Proposed Pathway for Implementation Model (Author’s Work)

6.6 Conclusion

This study provides compelling evidence for both the need and the feasibility of implementing hereditary breast cancer screening in middle-income healthcare contexts. The strong clinical consensus supporting genetic testing, combined with the identification of specific implementation barriers, creates a clear roadmap for evidence-based intervention development and policy reform. The research demonstrates that successful implementation requires comprehensive, multi-level approaches addressing financial barriers, psychological concerns, system capacity, and cultural factors simultaneously. The international case study analysis provides evidence that these barriers can be overcome through appropriate system design, public funding, and culturally sensitive service delivery models.

The findings have immediate implications for clinical practice, healthcare policy, and public health programming. The identification of specific barriers and facilitators provides actionable insights for healthcare providers, policy makers, and researchers working to improve genetic services access and effectiveness. Most importantly, this study demonstrates that hereditary breast cancer screening implementation is not merely a technical or clinical challenge but a complex social and system intervention requiring sustained commitment to equity, accessibility, and cultural appropriateness. The potential benefits reduced cancer incidence and mortality, enhanced preventive care, and improved family health outcomes justify the investments required for comprehensive implementation.

The path forward requires collaboration across multiple stakeholders, sustained funding for service development and research, and ongoing attention to health equity and cultural sensitivity. Success will be measured not only by clinical outcomes but also by the ability to achieve equitable access and meaningful health improvements for all individuals and families at risk of hereditary breast cancer.

This research contributes to the growing body of evidence supporting the expansion of genetic services while highlighting the implementation challenges that must be addressed for these services to achieve their full potential for improving population health outcomes. The framework and findings presented here provide a foundation for continued research, policy development, and service improvement in hereditary cancer screening and broader genetic health services.

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APPENDIX A

SURVEY COVER LETTER

Dear Participant,

I am conducting a doctoral research study titled “Pathway for Implementation of Hereditary Breast Cancer Screening” as part of my Doctor of Business Administration (DBA) program at the Swiss School of Business and Management, Geneva. The purpose of this research is to explore awareness, barriers, and opportunities related to hereditary breast cancer screening in India and to develop a structured pathway for its effective implementation.

You have been invited to participate because of your valuable perspective as a (patient/family member/ oncologist/ gynecologist/ genetic counsellor). Your participation will provide critical insights into how hereditary breast cancer screening can be improved for better healthcare outcomes.

The survey will take approximately 10–15 minutes to complete. Your responses will be kept strictly confidential and used solely for academic research purposes. No identifying information will be disclosed in the study, and participation is completely voluntary. You may withdraw at any time without consequence.

To participate, please click the following link to access the survey:

Link:

Your contribution is very important to this study and will help in identifying practical strategies to make hereditary breast cancer screening more accessible, affordable, and effective. If you have any questions regarding this study, please feel free to contact me.

Sincerely,

Sneha Rajan Datar

APPENDIX B

QUESTIONNAIRE/ SURVEY FORM (*FAMILY MEMBERS*)

Question 1. Are you aware of the hereditary factors that can contribute to breast cancer?

Yes / No

Question 2. Have you been informed about genetic testing options for hereditary breast cancer by your doctor?

Yes / No

Question 3. Do you know which family members may be at risk if a genetic mutation is found?

Yes / No.

Question 4. Have you or a family member undergone genetic testing for hereditary breast cancer?

Yes / No

Question 5. If the answer to Question 4 is 'Yes', what was your experience like? Did you receive enough information and support?

<u>Experience</u>: Good Average

Bad

<u>Information Received</u>: Enough Not Enough

Question 6. If the answer to Question 4 is 'No', what were the reasons for not pursuing genetic testing? (Select Multiple Answers if any)

a. Financial Reasons

b. Accessibility

c. Fear of Result

d. Privacy Concerns

e. Lack of Understanding

f. Others *(Please specify)* ____________

Question 7. If the answer to Question 4 is ‘Yes’, what challenges, if any, have you faced in accessing genetic testing and counseling services? (Select Multiple Answers if any)

a. Cost of the test

b. Lack of Insurance Coverage

c. No access to specialist

d. Logistical Challenges (geographical barriers, etc)

e. Lack of Understanding

f. Others (Please specify) ___________

Question 8. If the answer to Question 4 is ‘Yes’, what factors made you undergo genetic testing or screening?

a. Family History

b. Doctor’s Advice

c. Reasonable cost

d. Accessibility

e. Others (Please specify) ___________

Question 9. Who was involved in the decision-making process of getting / not getting the screening test done, and what factors influenced your decision?

a. Family Member
b. Friend
c. Relative
d. Others (Please specify) ___________

Question 10. How comfortable are you discussing hereditary cancer risks with your family members?

a. Very comfortable`
b. Somewhat comfortable
c. Neutral
d. Somewhat uncomfortable
e. Very uncomfortable

APPENDIX C

QUESTIONNAIRE/ SURVEY FORM (*ONCOLOGISTS*)

Question 1. How do you identify and determine which individuals should undergo genetic testing for hereditary breast cancer? (Select Multiple Answers if any)

a. Family history review
b. Personal or family cancer history
c. Known genetic predispositions
d. Referral based on clinical guidelines
e. Others (*Please specify if any*) ______________

Question 2. What standards or protocols guide your decisions on recommending genetic testing? (Select Multiple Answers if any)

a. Established medical guidelines (NCCN, ASCO, etc.)
b. Institutional policies
c. Risk-based models
d. Personal clinical assessment
e. Experience in Practice
f. Others (*Please specify if any*) ______________

Question 3. What difficulties have you experienced in suggesting genetic testing in your practice? (Select Multiple Answers if any)

a. Individual willingness or understanding
b. Limited educational guidelines on genetics
c. Inconsistent reimbursement or coverage
d. Managing individual follow-up after test results
e. Others (*Please specify if any*) ______________

Question 4. During the interaction with individuals/family members, what is the most crucial/important factor for resisting the idea of screening?

a. Cost of screening
b. Limitation in understanding the concept
c. Fear of the outcome/ Impact of Family
d. Geographical barriers
e. Insurance coverage
f. Data Privacy
f. Others (*Please specify if any*) ______________

Question 5. What challenges do you face in integrating genetic testing into your clinical practice? (Select Multiple Answers if any)

a. Financial cost to the individual
b. Limited number of facilities for genetic testing
c. Limited awareness of hereditary cancer risks
d. Insufficient insurance coverage

e. Other (please specify if any) ________________________

Question 6. What tools or resources would assist you in better integrating genetic information into your practice? (Select Multiple Answers if any)

a. Clinical decision-making tools

b. More accessible individual education materials

c. Greater access to or collaboration with genetic counselors

d. Provider training programs on interpreting genetic data

e. Other (please specify if any) ________________________

Question 7. How often do you collaborate with genetic counselors, gynecologists, or other specialists when managing individuals with hereditary breast cancer risks?

a. Every individual case

b. Most cases

c. Occasionally

d. Rarely

e. Never

Question 8. What improvements would you suggest for better interdisciplinary collaboration? (Select Multiple Answers if any)

a. Enhanced communication systems

b. More frequent interdisciplinary case reviews

c. Direct referral systems

d. Improved electronic health records integration

e. Other (please specify) __________________________

Question 9. What are your concerns regarding genetic privacy, discrimination, and informed consent in genetic testing? (Select Multiple Answers if any)

a. Risk of misuse of genetic data

b. Complex consent forms/formats

c. Ensuring the privacy of genetic test results

d. Other (please specify) ______________________

APPENDIX D

QUESTIONNAIRE/ SURVEY FORM (*GYNECOLOGISTS*)

Question 1. How often do you encounter individuals with a family history of breast cancer?

a. 1 in 10 individuals

b. 2 in 10 individuals

c. Others _______________

Question 2. Do you recommend genetic testing for hereditary breast cancer to individuals at higher risk?

Please specify the reasons.

Yes / No

Reason_____________

Question 3. What hurdles do you face while giving/explaining information about Hereditary screening? (Select Multiple Answers if any)

a. Individual literacy level

b. Emotional Response

c. Technical Terminology

d. Communicating risks related to hereditary screening

e. Others (Please specify if any) _______________

Question 4. During the interaction with individuals/family members, what is the most crucial/important factor for resisting the idea of screening?

a. Cost of screening
b. Limitation in understanding the concept
c. Fear of the outcome/ Impact of Family
d. Geographical barriers
e. Insurance coverage
f. Data Privacy
g. Others_____________

Question 5. Do you coordinate/communicate/discuss with oncologists and genetic counselors for individuals at risk of hereditary breast cancer?

Yes / No

Reason _________________

Question 6. What difficulties have you experienced in suggesting genetic testing in your practice? (Select Multiple Answers if any)

a. Individual willingness or understanding
b. Lack of integration of genetic counselling with genetic testing
c. Limited educational guidelines on genetics
d. Inconsistent reimbursement or coverage
e. Managing individual follow-up after test results

f. Others ____________

Question 7. What support do you provide to individuals to help them understand their risk of hereditary breast cancer? (Select Multiple Answers if any)

a. Family history assessment questionnaire

b. Educational Material about Hereditary cancers and associated risks

c. Online Resources related to Hereditary cancers, guidelines, etc.

d. Details of support groups and Forums

e. Others _______________________

Question 8. How do you address concerns related to genetic privacy in your practice?

a. Informed Consent

b. Confidentiality Policy

c. Compliance with laws like GDPR, HIPAA, etc

d. Explain the consent and other data privacy information in the regional language

e. Others_______________

APPENDIX E

QUESTIONNAIRE/ SURVEY FORM (*GENETIC COUNSELORS*)

Question 1. Do you stay updated on new guidelines and best practices in genetic counseling?

Yes / No

If Yes, How do you keep yourself updated _____________________

If no, why?

Question 2. Of the total individuals taking genetic screening test, what percentage of individuals receive both pre and post-test counselling.

____

Question 3. Of the total individuals taking genetic screening test, what percentage of of individuals receive only pre-test counselling.

____

Question 4. Of the total individuals taking genetic screening test, what percentage of of individuals receive only post-test counselling.

____

Question 5. How do you educate individuals and their families about hereditary breast cancer and genetic testing? (Select Multiple Answers if any)

a. Information on Genetic Conditions

b. Explaining Genetic Testing

c. Results interpretation

d. Others (Please specify if any) ___________________

Question 6. What models do you use to ensure that individuals understand the risks and benefits of genetic testing? (Select Multiple Answers if any)

a. Tailored Communication (Using local language, using terminology based on the individual's understanding, background, etc.)

b. Use of Charts, Diagrams, infographics, Audio-visuals, etc.

c. Involving Family member/relative/Guardian

d. Follow-up sessions

e. Others (Please specify if any) ____________

Question 7. How do you address the emotional and psychological impacts of genetic testing on individuals and their families? (Select multiple answers if any)

a. Pre-test counseling

b. Post-test Counseling

c. Follow-up sessions for ongoing support

d. Involving family member in the discussion

e. Others (Please specify if any) ____________

Question 8. What support systems do you have in place to assist individuals in coping with their genetic risk? (Select multiple answers if any)

a. Counseling sessions
b. Information material like brochures/pamphlets, etc.
c. Interdisciplinary Collaboration between physicians, psychologists, etc.
d. Referral to support groups
e. Others (Please specify if any) ____________

Question 9. How do you collaborate with oncologists and other healthcare providers to ensure that genetic information is integrated into individual care?

a. Individual evaluation to identify those who can benefit from genetic screening
b. Referring individuals with higher risk to the concerned healthcare provider
c. Others ____________________

Question 10. How do you manage ethical issues related to genetic testing, such as informed consent, and genetic privacy?

a. Providing detailed information on the purpose, process, risks, benefits, and limitations of genetic testing
b. Explaining to the individuals/family members in detail how personal information can be stored, used, and shared.
c. Follow strict protocols like HIPAA, GDPR, etc.

d. Explain the consent and other data privacy information in the regional language

e. Others ___________________________

Question 11. What challenges do you face in providing genetic counseling, particularly in underserved or marginalized populations?

a. Limited access to care

b. Cultural Sensitivity

c. Low health literacy

d. Language barrier

e. The cost associated with counseling

f. Others ___________________________

Question 12. How can access to genetic counseling services be improved?

a. Provide Telehealth options

b. Continuous training and awareness

c. Integration of genetic counseling in primary health care setting

d. Providing insurance coverage for genetic screening and counseling

e. Implementing community outreach programs

f. Engaging in policy advocacy to support legislation that promotes genetic health services

g. Others ___________________________

Question 13. During the interaction with individuals/family members, what are the most crucial/important factors for resisting the idea of screening?

a. Cost of screening
b. Limitation in understanding the concept
c. Fear of the outcome/ Impact of Family
d. Geographical barriers
e. Insurance coverage
f. Data Privacy
g. Others ____________________