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PERCEPTIONS OF GENETIC TESTING FOR HEREDITARY DISEASES AMONG AT-RISK FAMILIES

Original Article

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ABSTRACT

Background: Genetic testing for hereditary diseases presents significant opportunities for early diagnosis and preventive care, yet its uptake among at-risk family members remains inconsistent. While biomedical frameworks guide testing protocols, less is known about the lived experiences and social factors that influence how individuals interpret, communicate, and act on genetic risk. A qualitative approach is essential to explore the emotional, relational, and cultural dimensions of these experiences.

Objective: This study explores the perceptions, emotional responses, and communication practices of at-risk family members offered genetic testing for hereditary conditions.

Methods: A qualitative phenomenological design was employed. Twenty-two participants were purposively recruited from three genetic counseling clinics. Data were collected through semi-structured, in-depth interviews and analyzed using Braun and Clarke's thematic analysis framework. Trustworthiness was ensured through member checking, researcher reflexivity, and triangulated coding.

Results: Three key themes emerged: (1) emotional navigation of genetic risk, including fear, uncertainty, and generational guilt; (2) communication challenges within families, with variability in disclosure practices; and (3) perceived value and consequences of testing, shaped by cultural beliefs, health literacy, and personal experience. Differences in understanding and emotional readiness influenced both testing decisions and family discussions.

Conclusion: The study highlights the complex emotional and relational landscape surrounding genetic testing in families. Findings underscore the need for personalized, culturally sensitive genetic counseling and supportive policies that promote ethical and effective cascade testing. Further research should explore digital interventions and clinician-supported disclosure models.

Keywords: Qualitative Research, Genetic Testing, Hereditary Disease, Family Communication, Phenomenology, Thematic Analysis.

INTRODUCTION

Genetic testing has become an essential tool in identifying individuals at risk for hereditary diseases, enabling earlier surveillance, preventive interventions, and tailored treatment options. Despite its clinical benefits, uptake and perceptions of genetic testing among at-risk families remain inconsistent, shaped by psychological, cultural, relational, and systemic factors. While conditions such as hereditary breast and ovarian cancer (HBOC), Lynch syndrome, familial hypercholesterolemia, and inherited cardiomyopathies warrant genetic cascade testing, evidence suggests many relatives either decline testing or remain unaware of their inherited risk (1,2). These disparities reflect more than a lack of access; they highlight emotional complexities, knowledge gaps, and sociocultural dynamics that influence decision-making.

Recent qualitative and mixed-method studies have illuminated how individuals navigate the emotional terrain of hereditary risk. In families affected by inherited cardiac conditions, relatives often perceive themselves to be at risk even when test results are negative, continuing to pursue unnecessary medical screenings due to anxiety or misunderstanding of genetic information (3). Similarly, in hereditary cancer contexts, low testing uptake among first-degree relatives—only 32.6% in some studies—underscores a widespread challenge in motivating family-level engagement, even after a proband is diagnosed (4). These findings reflect not only a breakdown in risk communication but also complex psychological barriers including fear of results, fatalism, or avoidance of family confrontation. Genetic testing thus becomes not merely a medical decision, but a social process shaped by intra-family dynamics, emotional readiness, and moral responsibilities.

Given these nuances, a qualitative research approach is ideally suited to explore the lived experiences and meaning-making processes of individuals navigating hereditary risk. While quantitative data can measure testing rates or demographic trends, it cannot sufficiently capture the internal conflicts, cultural narratives, and ethical dilemmas that influence behavior. For instance, qualitative studies have shown that individuals often frame genetic testing within familial loyalty, generational responsibility, or personal identity, and may struggle with whether or how to share their results with relatives (5,6). These narratives reveal a landscape of emotions and values that cannot be quantified but are crucial for understanding why some families engage with testing while others do not.

The current study seeks to explore the perceptions of genetic testing among family members who are at risk for hereditary diseases. The central research question is: How do at-risk family members perceive and interpret genetic testing for hereditary conditions? Specifically, this study aims to (i) explore emotional and cognitive responses to the offer of testing, (ii) identify perceived barriers and motivators to testing uptake, and (iii) understand how familial relationships and communication patterns shape decision-making. The insights generated may provide critical guidance for improving genetic counseling practices, tailoring risk communication strategies, and designing support interventions that reflect the realities of patients and families.

This study will be conducted among adult family members of individuals diagnosed with a confirmed hereditary disease, who themselves have been offered cascade genetic testing. Participants will be recruited through genetic counseling clinics and outreach programs within hospital networks and hereditary cancer or cardiology registries. The settings will include diverse urban and suburban populations to ensure variation in socioeconomic status, cultural background, and healthcare access.

The significance of this study lies in its potential to inform both practice and policy in genomic medicine. Understanding the deeply personal and relational nature of genetic testing can lead to better-designed counseling frameworks that incorporate emotional support, health literacy, and culturally appropriate communication. Recent interventions, such as the Family Gene Toolkit—a web-based platform co-developed for Korean and Swiss BRCA-positive families—demonstrate how narrative storytelling and user-centered tools can effectively support family communication and promote testing uptake (6). Other studies have emphasized the need for healthcare providers to take a more active role in facilitating communication, with evidence suggesting that patients often support direct-to-relative communication by clinicians, especially for actionable risks (7). However, such strategies must be informed by a clear understanding of family attitudes and sensitivities, which this qualitative study seeks to uncover.

Ultimately, while the science of genetics continues to advance rapidly, the real-world implementation of testing depends not just on laboratory capacity but on trust, emotion, and the ability of families to navigate risk together. A qualitative approach allows us to listen to the voices often lost in the clinical setting and create frameworks that better reflect the needs and values of those living with hereditary risk.

METHODS:

This study employed a phenomenological qualitative design to explore the lived experiences and perceptions of genetic testing among at-risk family members of individuals diagnosed with hereditary diseases. A phenomenological approach was deemed appropriate due to its emphasis on capturing the subjective experiences and emotional meanings participants attach to genetic risk, testing decisions, and intergenerational communication. This methodology aligns with the aim of understanding how individuals interpret complex genetic information within the familial and cultural context of hereditary disease (8).

Participants were recruited through purposive sampling from three tertiary genetic counseling clinics specializing in hereditary cancer and cardiac conditions. Eligibility criteria included being a first- or second-degree relative of a proband with a confirmed hereditary disease, aged 18 or older, and having been offered cascade genetic testing. Individuals with cognitive impairments or without sufficient language proficiency to participate in an interview were excluded. This purposive strategy ensured that participants had direct experience with the study phenomenon, in line with best practices in phenomenological inquiry (9).

Data were collected through in-depth semi-structured interviews using an interview guide developed from literature and expert input. Questions focused on participants' beliefs about hereditary risk, emotional responses to testing offers, and experiences with family communication. Interviews were conducted in person or via secure video conferencing, depending on participant preference, and lasted between 45 and 75 minutes. All interviews were audio-recorded with consent and transcribed verbatim. The interview format allowed flexibility to probe emerging themes, and the use of open-ended prompts encouraged reflection and storytelling, which are foundational to qualitative data richness (10).

Analysis followed Braun and Clarke's six-step framework for thematic analysis, which provides a flexible yet rigorous method for identifying and interpreting patterns across the dataset. After initial familiarization with transcripts, two researchers independently generated initial codes. Codes were then compared, refined, and organized into broader themes through iterative discussion and constant comparison. NVivo software supported coding and theme development. The researchers engaged in regular debriefing sessions to address potential biases and ensure alignment in interpretation. Themes were reviewed across cases to identify both shared and divergent experiences, reflecting the depth and variation in lived narratives (11).

Ethical approval was obtained from the Institutional Review Board. Participants received detailed information about the study, including risks and benefits, and provided written informed consent. Confidentiality was maintained by pseudonymizing all data, and identifying details were removed during transcription. All data were securely stored in encrypted, access-restricted systems. Participants were informed of their right to withdraw at any point without penalty (12).

To enhance the trustworthiness of the findings, several strategies were employed. Member checking was conducted by providing participants with a summary of the key themes for validation, ensuring their perspectives were accurately represented. Triangulation was achieved by involving multiple researchers in data collection and analysis, thereby reducing individual bias. Reflexive journaling was maintained throughout the study to monitor researcher assumptions and emotional responses, which is a critical element of maintaining rigor in qualitative health research (13). Furthermore, the team followed contemporary guidelines for qualitative genetic counseling research to ensure transparency and methodological consistency with field standards (14).

This methodological framework, combining theoretical rigor with empathetic engagement, provides a robust basis for exploring the nuanced and deeply personal experiences of individuals grappling with genetic risk in their families. By integrating rich narrative data and ensuring analytic transparency, the study aims to contribute meaningful insights to the evolving discourse on genetic testing in hereditary disease contexts.

RESULTS:

The study included 22 participants (14 females and 8 males), ranging in age from 27 to 68 years, who were either first- or second-degree relatives of individuals diagnosed with a confirmed hereditary disease. Participants had been offered genetic testing through cascade screening pathways in oncology or cardiogenetics clinics within the past two years. Of the total, 15 had undergone genetic testing, while 7 had declined, providing insights from both perspectives. All interviews were conducted in English and spanned 50–75 minutes, yielding rich, reflective data on emotional responses, decision-making factors, and family communication dynamics.

Three major themes emerged from the data: (1) Emotional Navigation of Genetic Risk, (2) Communication Challenges and Responsibilities Within Families, and (3) Perceived Value and Consequences of Genetic Testing.

The first theme, *Emotional Navigation of Genetic Risk*, captured the psychological tension participants felt when facing the possibility of carrying a hereditary condition. Participants described genetic testing as emotionally complex, often associated with anticipatory anxiety, fear of positive results, and guilt over potentially passing on a genetic condition. As one participant stated, "I felt like opening the envelope would change everything... I almost didn't want to know, even though I knew I had to" (Participant 3). For many, testing was not only about personal health but also about generational impact, particularly concerning their children. Several described fluctuating emotions before and after the test, echoing findings in prior research on hereditary heart conditions and anxiety trajectories (15). This emotional burden, although challenging, was often outweighed by a desire to "do the right thing" for future generations (Participant 11).

The second theme, *Communication Challenges and Responsibilities Within Families*, highlighted diverse approaches to sharing genetic information. Some participants described open family environments where results were easily discussed and supported. "We had a family meeting. My sister and I wanted everyone to know—we didn't want secrets" (Participant 7). In contrast, others experienced silence, resistance, or even conflict. "My dad didn't want to talk about it. He said knowing would just make people worry" (Participant 14). The difficulty of communicating uncertain results, such as variants of unknown significance, added another layer of confusion. This variability echoes previous literature emphasizing how familial communication is influenced by interpersonal dynamics, trust, and generational roles (16,17). Several participants noted that older relatives played a central role in shaping the family's narrative around illness, influencing decisions regarding testing (18).

The third theme, *Perceived Value and Consequences of Genetic Testing*, revolved around how participants understood and interpreted the purpose and outcome of testing. Some viewed it as empowering and clarifying: "Now I know what I'm dealing with, and I can plan" (Participant 5). Others saw limited utility, especially when the test did not yield actionable findings. "It just left me with more questions... and no clear next steps" (Participant 9). There were also concerns about insurance implications, data privacy, and emotional readiness. Notably, individuals from medically underserved backgrounds expressed more skepticism, often due to previous negative healthcare experiences and limited access to follow-up care (19). A few participants, particularly those in multicultural households, raised concerns about stigma and the cultural interpretation of hereditary illness, aligning with findings from recent global qualitative studies (20).

An unexpected finding was the role of prior exposure to genetic concepts. Participants with a basic understanding of heredity or who had previously encountered genetic issues in their family demonstrated more confidence in decision-making. Conversely, individuals with limited knowledge expressed uncertainty and confusion, especially about the probabilistic nature of genetic risk. "I thought a positive result meant I'd definitely get the disease. I didn't realize it just meant a higher chance" (Participant 20). This misinterpretation reflects ongoing issues in genetic literacy and public education, which have been documented across various contexts (21).

In summary, participants navigated a complex landscape of emotions, responsibilities, and beliefs in relation to genetic testing. Their decisions were rarely linear, influenced by internal reflections, familial interactions, and broader cultural or structural factors. The variability in responses underscores the importance of personalized, culturally sensitive genetic counseling and the need for greater support throughout the decision-making process.

Table 1: Participant Demographics and Genetic Testing Status

Variable	Description/Range	Frequency (n=22)
Age (years)	27-68 (Mean± SD)	
Gender	Female / Male	14 / 8
Relationship to Proband	First-degree / Second-degree	14 / 8
Underwent Genetic Testing	Yes / No	15 / 7
Ethnic/Cultural Background	Mixed ethnic backgrounds	Multiple categories

Variable	Description/Range	Frequency (n=22)
Educational Level	High school / College / Postgrad	Varied across sample
Hereditary Condition Type	Cancer / Cardiac / Other	10 / 8/ 4

Table 2: Emergent Themes and Representative Participant Quotes

Theme	Subtheme	Representative Quote
Emotional Navigation of Risk	Anticipatory anxiety	"I almost didn't want to know…" – P3
Emotional Navigation of Risk	Generational guilt	"What if I passed it to my kids?" – P11
Communication Challenges in Families	Silence and resistance	"He didn't want to talk about it." – P14
Communication Challenges in Families	Open disclosure	"We had a family meeting…" – P7
Perceived Value of Testing	Empowerment vs. ambiguity	"It gave me peace of mind…" – P5

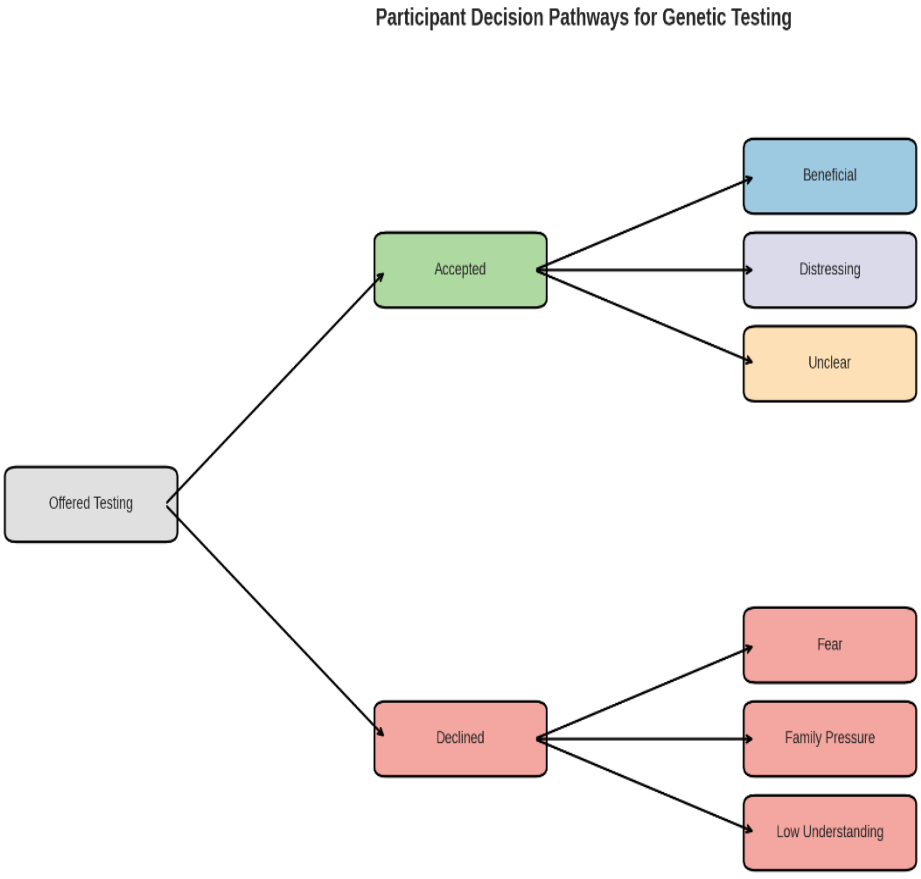

Figure 1 Participants Decision Pathways for Genetics Testing

DISCUSSION:

The findings of this study reveal the deeply personal and relational nature of how individuals perceive and respond to genetic testing for hereditary diseases. Participants' emotional responses, communication practices within families, and interpretation of testing outcomes underscore the complexity of decisions surrounding genetic risk. These results align with the study's objective to explore family members' attitudes, beliefs, and concerns, and highlight the need for more individualized, context-aware genetic counseling approaches. The emotional ambivalence described—ranging from anxiety to empowerment—reflects the psychological weight that many carry when engaging with their genetic identity and familial responsibility.

These insights resonate with recent literature showing that genetic information, while clinically valuable, is often filtered through personal experiences, cultural frames, and prior encounters with disease. Machirori et al. (2021) described this interpretive process as "moving within a landscape of knowledge," emphasizing how familial experience with disease shapes the meaning of genetic results far beyond the biomedical lens (22). Similarly, Daykin et al. (2023) found that in families with multiple genetic risks, risk perception and memory of genetic information were inconsistent, even after counseling—supporting our finding that prior knowledge and emotional readiness shape testing decisions and recall (23). The significance of emotional context, especially in vulnerable populations, was further emphasized in Bhasin et al. (2022), who reported that understanding of genetic test results was significantly influenced by education and language barriers, with many patients misinterpreting genetic risk as a deterministic diagnosis rather than a probability (24).

The variability in how participants communicated results to family members—ranging from open sharing to intentional silence—adds complexity to established views of genetic responsibility. Harrison et al. (2023) found that although many patients feel a moral obligation to inform relatives, low health literacy, family conflict, and privacy concerns often hinder disclosure (25). Our findings align with this, particularly in families where older members resisted discussing risk or downplayed its significance. Moreover, legal and ethical ambiguities regarding whether clinicians should disclose results directly to at-risk relatives further complicate the issue, as highlighted by Gilbar and Barnoy (2020), who noted tension between patient confidentiality and familial benefit in clinical practice (26). These unresolved ethical concerns point to the necessity for clearer guidelines and stronger clinician-family communication frameworks.

Differences in participant perspectives were also evident in how they evaluated the utility of genetic testing. While some found clarity and peace of mind, others perceived testing as ambiguous or emotionally taxing. This divergence may reflect differences in understanding of genetic information, personal experiences with disease, and family history, consistent with Smith et al. (2023), who found that family structure and cultural background significantly influenced how genomic sequencing results were interpreted in pediatric cases (27). Unexpectedly, prior knowledge or exposure to genetics appeared to play a protective role, reducing distress and increasing confidence—supporting findings by Airikkala et al. (2023), who noted that individuals with higher genetic literacy were more likely to view inherited risk as modifiable through lifestyle changes or preventive care (28).

Several limitations should be acknowledged. This study was conducted in three urban clinics, limiting generalizability to rural or non-clinical populations. Most participants were English-speaking, which may exclude perspectives from linguistically marginalized communities. Additionally, interviews were conducted by a single research team, which, despite efforts at reflexivity and triangulation, may have introduced interpretive bias. As qualitative research emphasizes depth over breadth, the findings should be viewed as exploratory, rather than representative of all families offered genetic testing.

Future research should examine how cultural values and family roles influence willingness to engage with genetic information across diverse communities. There is also a need to evaluate how digital platforms and tools, such as genetic education apps or family communication aids, can support cascade testing and facilitate disclosure in emotionally complex settings. Finally, longitudinal studies are needed to assess how perceptions and decisions evolve over time, particularly as genomic technologies advance and the boundaries of actionable genetic knowledge expand.

In conclusion, this study highlights that genetic testing, while grounded in science, unfolds in the deeply human domains of emotion, relationships, and meaning. Acknowledging the diverse ways families understand, communicate, and act upon genetic information is essential for developing ethical, empathetic, and effective genetic services in today's healthcare landscape.

Reflexivity and Researcher Positionality:

The lead researcher for this study has a professional background in medical genetics and experience working in clinical and educational roles within hereditary cancer and cardiogenetics programs. This background provided a foundational understanding of the clinical

pathways and terminology discussed by participants but also brought a potential bias toward viewing genetic testing as a beneficial and necessary process. This clinical orientation may have subtly influenced the nature of follow-up questions during interviews, particularly those probing for emotional or psychological responses to testing. The researcher's familiarity with genetic services might also have shaped assumptions about participant understanding, occasionally necessitating a conscious shift to remain open to unexpected or divergent experiences, particularly from those with limited health literacy or negative prior healthcare experiences (29).

To mitigate potential bias and enhance trustworthiness, several strategies were embedded throughout the research process. First, data coding and thematic development were conducted collaboratively by two independent researchers, both with training in qualitative methods but from differing disciplinary backgrounds—one from clinical genetics and the other from medical sociology. Discrepancies in coding were resolved through discussion, promoting analytical reflexivity. Second, member checking was conducted by inviting a subset of participants to review and provide feedback on synthesized themes. Their validation helped confirm that the findings resonated with their lived experiences and added credibility to the interpretations (30). Furthermore, direct quotes were consistently used to ground thematic claims in the voices of participants, aligning with reflexive practices that emphasize accountability to the data and transparency in interpretation (31).

Challenges encountered during data collection and analysis underscored the emotional complexity of the topic. Several participants became tearful or hesitant when recalling conversations with family members about genetic risk, particularly when discussing deceased relatives or concerns about their children's futures. This required the researcher to balance empathetic engagement with the need to maintain a neutral, non-directive stance. In some cases, participants' reluctance to elaborate on sensitive issues—such as mistrust of healthcare systems or fears of discrimination—highlighted the importance of creating a safe and non-judgmental interview environment. Additionally, the researcher maintained a reflexive journal throughout the study to monitor emotional responses, evolving assumptions, and interactions during interviews. This practice fostered continuous awareness of how positionality may shape interpretation and decision-making (32).

Recognizing the intertwined nature of subjectivity and inquiry, the research team viewed reflexivity not as a limitation to be minimized but as a methodological strength. Reflexivity was approached as an ongoing, conscious practice that informed every stage of the study—from framing the research questions to interpreting findings. As recent literature suggests, embracing the researcher's partial perspective can enrich rather than weaken qualitative analysis, provided that positionality is acknowledged and critically examined (33). In this study, the reflexive process allowed the researcher to remain accountable to participants' narratives while consciously decentering personal beliefs, thereby supporting an ethically grounded and context-sensitive approach to knowledge production (34).

IMPLICATIONS FOR PRACTICE, POLICY, AND FUTURE RESEARCH:

The findings from this study have several important implications for clinical practice. Firstly, healthcare professionals involved in genetic counseling must recognize the profound emotional and relational dynamics that shape how individuals perceive and act upon genetic testing offers. The variability in communication styles and emotional responses among at-risk family members suggests that clinicians should adopt a more tailored, narrative-driven approach to counseling—particularly when navigating uncertainty or distress. Using visual tools or story-based education, especially for those with low genetic literacy, could support better understanding and decision-making. Interventions that incorporate culturally sensitive communication frameworks may also improve engagement across diverse patient populations (35). Additionally, clinicians should routinely assess patients' readiness for testing and potential psychosocial impacts, rather than assuming interest or comprehension based solely on familial risk.

From a healthcare policy perspective, the study highlights the need for clearer institutional pathways and legal frameworks to support ethical, yet efficient, family communication and cascade testing processes. Current systems that rely exclusively on the proband to inform relatives often fall short. Policy reform that allows or even mandates clinician-mediated contact in specific situations—where consent permits and benefits clearly outweigh risks—could significantly increase cascade uptake, particularly in conditions like hereditary cancers and familial cardiomyopathies (36). Furthermore, equitable access to genetic services must be addressed, as underserved communities continue to face structural barriers that limit their engagement in genomic healthcare. This includes simplifying reimbursement policies, offering testing in multiple languages, and decentralizing services to reach rural populations more effectively (37). In parallel, investment in training nurses, midwives, and community health workers to deliver basic genetic education could help bridge service gaps and expand public understanding of hereditary risk (38).

This study also offers critical directions for future research. One key area is understanding how cultural, moral, and social norms intersect with genetic responsibility, particularly in multi-generational or multi-ethnic households. As previous studies suggest, the concept of "informed choice" in genetic testing must be examined in light of real-world constraints such as family pressure, stigma, or resource scarcity (39). Another area requiring attention is the evaluation of digital tools—such as AI-powered decision aids or mobile platforms—that assist in family communication and risk education. Pilot programs have shown promise, but longitudinal data on effectiveness, accessibility, and acceptability are still lacking. Lastly, future studies should explore clinician burden and preparedness in initiating or supporting genetic disclosure, as many non-genetics-trained providers now play a frontline role in genetic testing programs without sufficient support (40).

Together, these implications call for a shift toward more empathetic, equitable, and systemically supported models of genetic counseling and testing. Only by integrating the social and emotional realities of families with the scientific advances in genomics can clinical genetics truly fulfill its promise in preventive and personalized healthcare.

CONCLUSION:

This study revealed that at-risk family members experience genetic testing not merely as a clinical procedure, but as a deeply personal process shaped by emotional readiness, family communication patterns, and sociocultural contexts. The themes of emotional navigation, intergenerational dialogue, and the perceived value of testing illustrate the complexity of decision-making in hereditary disease settings. These insights underscore the need for more empathetic and context-sensitive approaches in genetic counseling—ones that recognize the moral and emotional dimensions of inherited risk. By illuminating the lived experiences of those facing genetic uncertainty, the study contributes to a growing body of research that calls for integrated support systems and policy reforms aimed at equitable, accessible, and personalized genomic care. Clinicians are encouraged to adopt communication strategies that center family dynamics, while policymakers must reconsider cascade testing protocols and disclosure guidelines to better reflect real-world family structures. Future research should continue to explore cultural and systemic barriers while developing interventions that strengthen understanding, trust, and engagement in genetic health across diverse populations.

AUTHOR CONTRIBUTION

Author	Contribution
Irfan Ishaque*	Substantial Contribution to study design, analysis, acquisition of Data Manuscript Writing Has given Final Approval of the version to be published
Muhammad Moaaz Anwar	Substantial Contribution to study design, acquisition and interpretation of Data Critical Review and Manuscript Writing Has given Final Approval of the version to be published
Amna Noor	Substantial Contribution to acquisition and interpretation of Data Has given Final Approval of the version to be published
Kashmala Munawar	Contributed to Data Collection and Analysis Has given Final Approval of the version to be published
Noor ul Ain Khaliq	Contributed to Data Collection and Analysis Has given Final Approval of the version to be published
Farhan Muhammad Qureshi	Substantial Contribution to study design and Data Analysis Has given Final Approval of the version to be published

REFERENCES:

1. Kutsev S, Moiseev S. Family genetic screening in rare hereditary diseases. *Clin Pharmacol Ther*. 2021. doi:10.1101/7da40c30c22856e78fd032a9b7019213
2. Campacci E, Grasel RS. The history of families at-risk for hereditary breast and ovarian cancer: a challenge to risk communication. *BMC Med Genomics*. 2024. doi:10.1186/s12920-024-01824-1
3. Katz S, Hawley S, Tocco R, Kurian A. A pilot study to increase cascade genetic risk education and testing in families with hereditary cancer syndromes. *J Clin Oncol*. 2022. doi:10.1200/JCO.2022.40.16_suppl.e24137
4. Baroutsou V, Duong D, et al. Acceptability and usability of the Family Gene Toolkit for BRCA families. *Genet Med*. 2023. doi:10.1016/j.gim.2023.04.005
5. Godino L, Turchetti D, et al. Public perspectives on healthcare-professional directed disclosure of genetic risk information. *J Community Genet*. 2025. doi:10.1007/s12687-025-00755-w
6. Wainstein T, Elliott A, Austin J. Considerations for the use of qualitative methodologies in genetic counseling research. *J Genet Couns*. 2022. doi:10.1002/jgc4.1644
7. Xu G. Genetic diagnostics for pediatric hereditary diseases: exploring chromosomal karyotyping, single gene mutation screening, and emerging technologies. *J Innov Med Res*. 2023. doi:10.56397/jimr/2023.08.06
8. Tilahun M, Gebresilase T, Aseffa A, et al. Public perceptions of genomic studies and hereditary diseases in Aari community, Ethiopia. *Trans R Soc Trop Med Hyg*. 2023. doi:10.1093/trstmh/trad051
9. Gorman R, Farsides B, Gammidge T. Stop-motion storytelling: exploring methods for animating the worlds of rare genetic disease. *Qual Res*. 2022. doi:10.1177/14687941221110168
10. Tian P-H, Xu Y, Zhang Y-Q, Wang T-Y. Genetic diseases are not necessarily inherited: suggestion on its Chinese translation. *Yi Chuan*. 2024;46(9):673–676. doi:10.16288/j.yczz.24-199
11. Borle C, Austin J. Using mixed methods for genetic counseling research. *J Genet Couns*. 2025;34. doi:10.1002/jgc4.70031
12. Das L, Nanda S, Das J. Hereditary disease prediction in eukaryotic DNA: an adaptive signal processing approach. *Nucleosides Nucleotides Nucleic Acids*. 2020;39(10):1179–1199. doi:10.1080/15257770.2020.1780440
13. Bordet C, Brice S, Maupain C, et al. Psycho-social impact of predictive genetic testing in hereditary heart diseases (PREDICT Study). *Arch Cardiovasc Dis Suppl*. 2020;12:32–33. doi:10.1016/j.acvdsp.2019.09.067
14. Müller R, Aghdassi A, Kruse J, et al. Perceptions of genetic testing in patients with hereditary chronic pancreatitis and their families: a qualitative triangulation. *Eur J Hum Genet*. 2020;29:29–38. doi:10.1038/s41431-020-00705-9
15. Oliveira C, Mendes Á, Sequeiros J, Sousa L. Role of older generations in the family's adjustment to Huntington disease. *J Community Genet*. 2021;12:469–477. doi:10.1007/s12687-021-00523-6
16. Godino L, Battistuzzi L, Turchetti D, et al. Developing a questionnaire to explore lay people's preferences for communicating hereditary conditions within families. *J Community Genet*. 2025. doi:10.1007/s12687-025-00783-6
17. Bednar E, Rauh-Hain J, Garcia JJ, et al. Experiences of family communication and cascade genetic testing for hereditary cancer in medically underserved populations. *Cancer Prev Res*. 2023;16(11):639–649. doi:10.1158/1940-6207.CAPR-23-0303
18. Tasnim S, Lim PXH, Griva K, Ngeow J. Psychosocial barriers and facilitators associated with uptake of genetic services for hereditary cancer syndromes. *Health Psychol Rev*. 2024. doi:10.1080/17437199.2024.2415950
19. Khan GA, Cherif S, Faisal L, et al. Public knowledge, awareness, and perception of genetic testing for hereditary diseases in the UAE. *Int J Innov Res Sci Stud*. 2025;8(1). doi:10.53894/ijirss.v8i1.4829
20. Lieberman S, Tomiak E, Weitzman E, et al. Psychosocial factors influencing uptake of cascade genetic testing among families with hereditary breast and ovarian cancer. *Genet Med*. 2021;23(9):1718–1726. doi:10.1038/s41436-021-01166-9

21. Etchegary H, Dicks E, Pullman D, et al. "It runs in the family": exploring perceptions and decision-making around genetic testing for hereditary cancer risk. *Health Expect*. 2020;23(6):1449–1460. doi:10.1111/hex.13117
22. Machirori M, Patch C, Metcalfe A. "It didn't mean anything": moving within a landscape of knowledge to interpret genetics and test results. *New Genet Soc*. 2021;40:570–598. doi:10.1080/14636778.2021.1997575
23. Daykin EC, Poffenberger CN, Do J, et al. Exploration of knowledge, risk perceptions, and communication in a family with multiple genetic risks for Parkinson's disease. *J Genet Couns*. 2023. doi:10.1002/jgc4.1677
24. Bhasin P, Durana T, Kyaw NYW, et al. Perception and emotional reaction to genetic test results in cancer patients. *J Clin Oncol*. 2022. doi:10.1200/jco.2022.40.16_suppl.e24128
25. Harrison C, Bartley N, Jacobs C, et al. Family communication and results disclosure after germline sequencing: A mixed methods study. *Patient Educ Couns*. 2023;114:107800. doi:10.1016/j.pec.2023.107800
26. Tercyak KP, O'Neill SC, Daly MB, et al. Family communication of genetic test results: a qualitative study of hereditary cancer families. *Patient Educ Couns*. 2022;105(7):2123–2131. doi:10.1016/j.pec.2022.03.009
27. Smith H, Bonkowski ES, Hickingbotham M, et al. Framing the family: factors shaping family-level experiences of pediatric genomic sequencing. *Children*. 2023;10. doi:10.3390/children10050774
28. Airikkala E, Laaksonen M, Halkoaho A, Kaunonen M. Perception of inherited risk in type 2 diabetes: a systematic review. *Front Public Health*. 2023;11. doi:10.3389/fpubh.2023.1293874
29. Barrett A, Kajamaa A, Johnston J. How to be reflexive when conducting qualitative research. *Clin Teach*. 2020;17(5):431–435. doi:10.1111/tct.13133
30. Olmos-Vega FM, Stalmeijer R, Varpio L, Kahlke R. A practical guide to reflexivity in qualitative research: AMEE Guide No. 149. *Med Teach*. 2022;45(3):241–251. doi:10.1080/0142159X.2022.2057287
31. Tomlinson J, Medlinskiene K. Reflexivity in pharmacy practice qualitative research: systematic review of twelve peer-reviewed journals. *Int J Pharm Pract*. 2024. doi:10.1093/ijpp/riae013.007
32. Rankl F, Johnson GA, Vindrola-Padros C. Examining what we know in relation to how we know it: a team-based reflexivity model for rapid qualitative health research. *Qual Health Res*. 2021;31(8):1358–1370. doi:10.1177/1049732321998062
33. Smith L, Luke M. A call for radical reflexivity in counseling qualitative research. *Couns Educ Superv*. 2021;60(2):164–172. doi:10.1002/ceas.12201
34. Ide Y, Beddoe L. Challenging perspectives: Reflexivity as a critical approach to qualitative social work research. *Qual Soc Work*. 2023. doi:10.1177/14733250231173522
35. Hendricks-Sturrup RM, Emmott N, Nafie M, et al. Returning personalized, genetic health test results to individuals of African descent in precision medicine research. *Health Aff Scholar*. 2023;1. doi:10.1093/haschl/qxad066
36. Grutters LA, Christiaans I. Cascade genetic counseling and testing in hereditary syndromes: inherited cardiovascular disease as a model. *Fam Cancer*. 2024;23:155–164. doi:10.1007/s10689-023-00356-x
37. Robillard J, Feng TL, Kabacińska K. Access to genetic testing for rare diseases: existing gaps in public-facing information. *World Med Health Policy*. 2021;13:518–525. doi:10.1002/wmh3.469
38. Kawasaki H, Kawasaki M, Iki T, Matsuyama R. Genetics education program to help public health nurses improve their knowledge and enhance communities' genetic literacy: a pilot study. *BMC Nurs*. 2021;20. doi:10.1186/s12912-021-00549-8
39. Aureliano W. Difficult decisions and possible choices: rare diseases, genetic inheritance and reproduction of the family. *Soc Sci Med*. 2024;363:117380. doi:10.1016/j.socscimed.2024.117380
40. Levin Salo N, Mullen LA. Call to action for genetic counseling research in hereditary cancer: considerations from the evidence-based guidelines development process. *J Genet Couns*. 2025;34. doi:10.1002/jgc4.70026

