



The Importance of Cascade Genetic Testing in Hereditary Breast Cancer

Matthew Warner*

Department of Medical Oncology, Gastrointestinal Cancer Center, Boston, Massachusetts, USA

INTRODUCTION

Breast cancer is one of the most common cancers worldwide, and a significant number of cases are hereditary, often resulting from mutations in specific genes like *BRCA1*, *BRCA2*, and others associated with heightened cancer risks. Identifying and managing hereditary breast cancer is not only critical for the individual diagnosed but also has implications for their biological relatives. Cascade genetic testing is a proactive approach that leverages genetic testing results from an affected family member to identify at-risk relatives. This targeted approach enables families to understand their cancer risk, pursue early detection, and adopt preventive measures. In this article, we will examine the importance of cascade genetic testing in hereditary breast cancer, the process involved, and its potential to improve health outcomes across generations.

DESCRIPTION

Understanding hereditary breast cancer

Hereditary breast cancer is often linked to inherited mutations in specific genes that significantly increase the risk of developing breast and other cancers. The most well-known of these genes are *BRCA1* and *BRCA2*. A woman with a *BRCA1* or *BRCA2* mutation faces a lifetime risk of breast cancer between 45-87%, substantially higher than the average population risk of about 13%. Men with these mutations also have an increased risk of breast cancer, and both genders face heightened risks of other cancers, including ovarian and prostate cancer. Other genes, such as *PALB2*, *TP53*, and *CHEK2*, are also associated with hereditary breast cancer and contribute to a family history of the disease.

What is cascade genetic testing?

Cascade genetic testing is a process in which genetic testing information from an individual diagnosed with a hereditary cancer syndrome is used to systematically identify and test family members who may carry the same mutation. This "cascade" effect begins with a relative diagnosed with cancer and known to

carry a genetic mutation, termed the "proband." Genetic testing is then offered to first-degree relatives (parents, siblings, and children), and if any are found to carry the mutation, the offer is extended to their relatives, creating a ripple effect across generations and branches of the family tree.

By testing relatives, especially those at the highest risk of carrying the mutation, cascade testing allows family members to take preventive measures and prioritize early detection, significantly altering the health outcomes for those at high risk.

Early detection and prevention: Cascade genetic testing plays a crucial role in allowing at-risk individuals to make informed health decisions. For individuals who test positive for a breast cancer-linked mutation, regular screenings, such as mammograms and MRIs, are often recommended at younger ages and more frequently than the general population. Preventive options, such as prophylactic mastectomy and oophorectomy, may also be considered to reduce cancer risk.

Cost-effectiveness and targeted testing: Since genetic testing can be expensive and may not be universally accessible, cascade testing offers a cost-effective solution. Testing high-risk family members rather than screening the general population focuses resources on those who are most likely to benefit, maximizing the cost-effectiveness of genetic services.

Empowerment through knowledge: Knowing one's genetic risk can be empowering. Cascade testing provides family members with critical information about their cancer risks, enabling them to take proactive steps to protect their health. For instance, knowing their *BRCA1* or *BRCA2* mutation status allows individuals to weigh options, such as lifestyle changes, enhanced surveillance, or preventive surgeries. Informed individuals often experience increased peace of mind and a sense of control over their health.

Reducing cancer-related mortality: Cascade genetic testing enables early detection and prevention of cancer, which can directly impact mortality rates in families with hereditary cancer syndromes. Studies show that individuals with hereditary cancer mutations who undergo regular surveillance and take preventive

Correspondence to: Matthew Warner, Department of Medical Oncology, Gastrointestinal Cancer Center, Boston, Massachusetts, USA; E-mail: mwarner08656@gmail.com

Received: 30-Oct-2024, Manuscript No. HGCR-24-27357; **Editor assigned:** 01-Nov-2024, PreQC No. HGCR-24-27357 (PQ); **Reviewed:** 15-Nov-2024, QC No. HGCR-24-27357; **Revised:** 14-Jun-2025, Manuscript No. HGCR-24-27357 (R); **Published:** 21-Jun-2025, DOI: 10.35248/2161-1041.25.14.308

Citation: Warner M (2025) The Importance of Cascade Genetic Testing in Hereditary Breast Cancer. *Hereditary Genet.* 14:308.

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measures have significantly lower cancer-related mortality compared to those who do not know their genetic status.

Challenges in implementing cascade genetic testing

Despite its benefits, several challenges can impact the effectiveness of cascade genetic testing. Privacy concerns, varying family dynamics, and differences in attitudes toward genetic testing can pose obstacles. Some family members may be unwilling to undergo testing, while others may struggle to inform relatives due to family disagreements or geographic distances. Additionally, genetic counseling resources may be limited, and some individuals may lack access to genetic services.

Addressing these challenges requires a supportive approach, often involving genetic counselors who are trained to navigate sensitive family dynamics and provide education on the value of cascade testing. For cascade testing to be effective, it is

important to offer adequate psychosocial support, making the testing process as accessible and comprehensible as possible.

CONCLUSION

Cascade genetic testing in hereditary breast cancer is a powerful tool for extending prevention and early detection to family members who may be at risk. By using the knowledge gained from one family member's genetic test, cascade testing identifies high-risk individuals, empowering them to make informed choices and take preventive actions. While challenges exist in implementing cascade testing, its potential to save lives by reducing cancer incidence and mortality makes it an invaluable component of hereditary cancer management. With the continued advancement of genetic counseling services and accessibility, cascade genetic testing can serve as a proactive, family-centered approach to combating hereditary breast cancer.