



Assessing the Role of Genetics in Breast Cancer Risk and Prevention Strategies

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Description

Breast cancer remains one of the most common cancers worldwide, with genetic factors playing a significant role in an individual's risk profile. While lifestyle and environmental factors certainly contribute to the chances of developing breast cancer, genetic history can significantly increase that risk. Over the past few decades, advances in genetic studies have identified several specific genes and inherited mutations associated with breast cancer, most notably *BRCA1* and *BRCA2*. Understanding the genetic component of breast cancer risk is essential for identifying high-risk individuals and developing effective prevention and early detection strategies. The majority of breast cancers occur sporadically, meaning there is no apparent family history. However, around 5%-10% of breast cancers are hereditary, often caused by mutations in genes that are passed from one generation to another.

The most widely studied genes associated with a heightened risk of breast cancer are *BRCA1* and *BRCA2*. Women with mutations in either of these genes have a 45%-65% lifetime risk of developing breast cancer, significantly higher than the average population risk of about 12%. In addition to BRCA mutations, other genes, including *TP53*, *PTEN*, *PALB2* and *CHEK2* have been linked to increased breast cancer risk. These genes play essential roles in DNA repair, cell cycle regulation and other processes that help prevent tumor formation. When mutations are present, the ability to repair damaged DNA is degenerated, allowing potentially cancerous cells to proliferate. While mutations in these genes are less common than BRCA mutations, they still account for a significant portion of hereditary breast cancers. Genetic testing for breast cancer risk assessment has become more accessible and widely available.

Testing primarily targets women with a strong family history of breast or ovarian cancer, especially those with multiple affected family members or early-onset cases. Identifying a mutation in high-risk genes allows individuals to make informed decisions about their health and implement strategies to reduce their cancer risk. Advancements in technology have also led to the development of multi-gene panels, which screen for mutations in multiple genes

simultaneously. These panels provide a larger scope of potential risk factors but also present difficulties, as not all identified mutations have well-understood implications for breast cancer risk. As a result, genetic analysis is an essential part of the testing process, helping individuals understand the significance of their results and the potential next steps. For individuals identified as having a high genetic risk of breast cancer, various prevention strategies are available, ranging from increased observation to preventive surgeries. Enhanced screening protocols are a first line of defense for individuals with a high genetic risk. These protocols typically involve starting screening at a younger age and utilizing more sensitive imaging techniques such as Magnetic Resonance Imaging (MRI) alongside mammography. Frequent screenings increase the likelihood of early detection, which is vital for improving treatment outcomes.

Certain medications, such as tamoxifen and raloxifene have been shown to reduce the risk of breast cancer in high-risk individuals by targeting estrogen receptors. This method is known as chemoprevention is often recommended for women who are at elevated risk due to genetic factors or other high-risk factors. However, these medications come with potential side effects and their use must be carefully weighed with the guidance of a healthcare professional. Some high-risk individuals choose to undergo preventive (prophylactic) surgeries to reduce their risk of developing breast cancer. The most common surgery is a bilateral mastectomy, which involves the removal of both breasts to drastically reduce the likelihood of cancer formation. Women with *BRCA1* or *BRCA2* mutations sometimes prefer for a prophylactic oophorectomy (removal of the ovaries), as these mutations increase the risk of ovarian cancer as well. While preventive surgeries significantly reduce cancer risk, they are life changing decisions and require thorough counseling.

Ongoing studies in the field of genetics and breast cancer continues to improve the understanding of cancer risk and prevention. Studies are increasingly focused on the genetic variants interact with environmental and lifestyle factors, providing a more thorough view of risk analysis. Polygenic risk scores, which assess an individual's genetic predisposition based on multiple low-risk variants, are being developed to provide specific risk estimations for individuals without known high-risk mutations.

Conclusion

Genetic factors play an essential role in breast cancer risk, particularly for individuals with specific gene mutations like *BRCA1* and *BRCA2*. Through genetic testing and risk assessment, high-risk individuals can identify their susceptibility and make informed decisions about prevention strategies. From increased surveillance and chemoprevention to preventive surgeries, there are various options available to manage and reduce the risk of breast cancer. As studies in genetics and cancer biology advances, along with the possibilities for specific prevention and treatment. For individuals with a genetic susceptibility to breast cancer, understanding their risk profile is essential for proactive health management. The continued study of genetic markers, combined with new technologies, potential to improve prevention strategies and outcomes for individuals worldwide.

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