

Hereditary Cancer Testing Considerations Before Testing



Sporadic vs Hereditary Cancer

In most cases, cancer happens by chance due to a combination of factors, such as age, lifestyle, environmental triggers, and multiple gene changes over time. These cancers are referred to as sporadic. However, about 5-10% of cancers are caused by a specific inherited genetic change (mutation). These cancers are referred to as hereditary cancer syndrome. All the cells in our bodies contain thousands of pairs of genes. Genes are the instructions that tell our bodies how to grow and function. We have two copies of each gene, as one copy is inherited from our mother and the other is inherited from our father. If there is a mutation or change in a gene, this too can be passed on. A person is born with the specific genetic change and is at an increased risk of developing certain cancers in adulthood. Families with hereditary cancer usually have more than one family member affected in more than one generation and the cancer diagnosis typically occurs at an earlier age, often before 50.

For females, the general population risk of developing sporadic breast cancer is 8-12%, or about 1 in 9, and the risk of developing ovarian cancer is 1.5%, or 1 in 70. Mutations in the BRCA1 and BRCA2 (BRCA1/2) genes are known to increase the lifetime risk of developing breast or ovarian cancer to approximately 85% and 40%, respectively. If a woman with a BRCA1/2 mutation has already been diagnosed with breast cancer, she has a 40-60% chance of developing a new breast cancer. Mutations in the BRCA1/2 genes are also associated with an increased risk of developing male breast and prostate cancers and, to a lesser degree, pancreatic cancer and melanoma.

Someone who has a mutation in one copy of a hereditary cancer gene (non-working) has a 50% DAD HAS ONE MOM HAS chance of passing on that mutation to each of **NON-WORKING TWO WORKING** his/her children. Individuals at increased risk GENE GENES of hereditary cancers should discuss cancer screening with their doctors. It is important to note that having a mutation in one copy of a WORKING GENE hereditary cancer genes does not mean that ☑ NON-WORKING GENE cancer will occur. 🗙 📀 CHILDREN HAVE TWO CHILDREN HAVE ONE WORKING GENES NON-WORKING GENE **IN 2 CHANCE (50%)** 1 IN 2 CHANCE (50%) THAT THE CHILD THAT THE CHILD WILL BE AT GENERAL WILL BE AT AN POPULATION RISK **INCREASED RISK OF** OR RISK BASED ON CANCER FAMILY HISTORY

Considerations Before Pursuing Hereditary Cancer Testing

- Genetic testing provides information that could lead to early detection of cancer and/or risk reducing options.
- Being aware of your genetic health can help educate other family members.
- Genetic testing can provide people with peace of mind. It can also cause emotional burdens (guilt, sadness, anxiety, anger), which can impact you and your family members.
- The Genetic Non-Discrimination Act (GNA) prohibits insurance companies from requesting the disclosure of genetic tests. It does not, however, prevent insurers from basing their decisions on an insured individual's current symptoms, diagnoses, and/or family history.

Possible Results of Genetic Testing for Hereditary Cancer Genes



Positive

A cancer-causing mutation has been identified. This individual has an increased risk for specific types of cancer. Different management options, such as screening, medications, and/or preventive surgeries, are available and referral to the appropriate specialists is suggested. Family members are at increased risk of carrying the same mutation.



True Negative

A previously-identified cancer-causing mutation in the family has not been identified in this individual. This individual's risk for cancer is not expected to be increased above the general population risk.



Uninformative Negative

A cancer-causing mutation has not been identified. If this individual has a personal and/or family history of cancer, the exact cause of the cancer remains unknown. Based on the family history assessment, this individual's risk of developing cancer may still be increased. If applicable, testing affected family members could be considered.



Variant of Unknown Significance (VUS)

A VUS indicates that the pathogenicity (whether a mutation causes a predisposition to cancer) of the identified variant cannot be established. Testing other family members may help clarify the clinical significance. Over time, variants may be reclassified as pathogenic or non-pathogenic. Based on the family history assessment, this individual's risk of developing cancer may still be increased.

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