

Cancer is a leading cause of death in Americans, second only to heart disease. In women, breast cancer is by far the most common cancer, and represents the second most frequent cause of cancer-related death.

While only 5 to 10 percent of breast cancer is influenced by an inherited susceptibility factor, knowing whether this is the case can be helpful to individuals with a personal or family history of breast cancer.

What is an inherited cancer susceptibility factor?

These are genetic alterations that increase the risk of developing a certain type (or types) of cancer. They are usually transmitted from parent to child. Someone with an inherited cancer susceptibility factor is at increased risk for cancer; however, not everyone with a susceptibility factor will ultimately develop cancer.

Even though most cancers are not related to hereditary genetic factors, in certain cases the genes we inherit from our parents play a role in cancer development. The BRCA1 and BRCA2 genes are the most common inherited susceptibility factors for breast and ovarian cancer.

There are several other genes that account for increased risk for colon and uterine cancer, pancreatic cancer and melanoma, colon cancer associated with multiple polyps, and several other genetic conditions in which multiple malignancies are present.

Who should be concerned about a family history of cancer?

Many people have at least one family member with a cancer diagnosis. However, in general, individuals with an inherited cancer susceptibility factor tend to develop cancers at relatively young ages, may have two or more cancers, and often have several relatives with the same type (or types) of cancer. Primary care physicians are often able to determine whether a genetic consultation, for a formal risk assessment and consideration of genetic testing, is warranted. A genetic consultation may provide additional reassurance if a physician is uncertain whether genetic testing will be of benefit.



How does a geneticist evaluate hereditary cancer risk?

The geneticist obtains a detailed medical history, including information about a suspected or proven cancer diagnosis. A complete family tree is constructed, and pertinent medical records are reviewed. The consultation conducted by our team of a specially trained genetic counselor and a physician-geneticist takes approximately one hour.

Three important questions are addressed during our evaluation. First, is a hereditary factor a possible explanation for the cancer diagnoses in the family? Second, what is the likelihood that a factor will be identified? Finally (and sometimes most importantly), who is the ideal person in the family for testing or further evaluation?

Is genetic testing difficult?

The actual sample collection process is very straightforward; after appropriate written informed consent is obtained, a small blood sample is drawn and sent to the appropriate clinical laboratory. The laboratory performs highly sophisticated genetic analysis on the DNA extracted from the blood specimen. Depending on the specific testing requested, results can be available in as little as two weeks. A follow up visit is always arranged to discuss the result and clinical implications for both the individual who was tested and their family members.

Testing for inherited cancer susceptibility is well-established, and most health insurance plans cover the testing, similarly to other medically necessary laboratory tests. Both federal and state regulations provide protection against discrimination based on results of genetic testing. Test results, both positive and negative, provide valuable information for optimal cancer treatment, surveillance, and prevention.

Can a non-geneticist physician deal with the genetic issues?

To some extent, yes. However, genetics is a rapidly changing and highly specialized field. Genetics professionals are trained in genetic theory and practice, in order to understand and apply the latest technological advances for their patients. Geneticists have statistical data, probability models, and valuable insights that aid patients in considering the pros and cons of genetic testing. A geneticist's expertise is particularly helpful in interpreting and explaining genetic test results.





The Genetics Center is a full service genetics facility combining clinical genetics, laboratory genetics, and genetic counseling in a comfortable private office setting. Established in 1991, The Genetics Center offers a full range of genetic services, including prenatal genetics, prenatal diagnosis by amniocentesis, genetic risk evaluation for cancer and other inherited conditions, and medical evaluations for adults or children who have, or may have, genetic disorders. The Genetics Center participates in many managed care plans, and our services are typically covered by health insurance.

Terminology

BRCA1 & BRCA2 - Two genes related to hereditary breast and ovarian cancer susceptibility.

DNA - The molecule in our cells that contains the instructions for how our bodies grow and develop. In humans, DNA is bundled into 23 pairs of microscopic structures called chromosomes. The nucleus of each cell contains a complete set of these instructions.

Familial - A trait or condition found in multiple individuals in a family. This may be due to a single or several genes, environmental influences, or a complex combination of these factors.

Genes - Segments of DNA responsible for our unique and common characteristics. There are an estimated 20-30,000 genes in humans, each encoding a protein that is necessary for controlling cellular functions. With few exceptions, we have two copies of each gene; one inherited from each parent.

Genetic consultation - A specialist evaluation by a geneticist to assess genetic issues. Details of the evaluation are summarized in a written report to the referring physician.

Genetic risk assessment - Determining the chance for having a genetic condition or variation. Information used for this may include family history, personal medical history, lifestyle factors, and other genetic test results.

Mutation - A variation in the code of a particular gene that may affect the structure or function of the protein determined by the gene. A "deleterious" mutation in a cancer susceptibility gene can increase the risk for certain cancers.

Presymptomatic genetic testing - DNA analysis to determine whether a particular condition may develop in the future. For example, testing someone without a cancer diagnosis for a BRCA gene mutation to see if they are at increased risk for breast and ovarian cancer.

Genetics & Cancer



the Genetics Center, inc.

48 Route 25A, Suite 205
Smithtown, New York 11787
631-862-3620
www.thegeneticscenter.com

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*Straightforward answers
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