ROSWELL PARK COMPREHENSIVE CANCER CENTER | PATIENT EDUCATION

Understanding Genetic Testing When You Have Cancer

Overview

Your doctor has advised you to consider genetic testing. This type of testing can provide major advantages to your treatment.

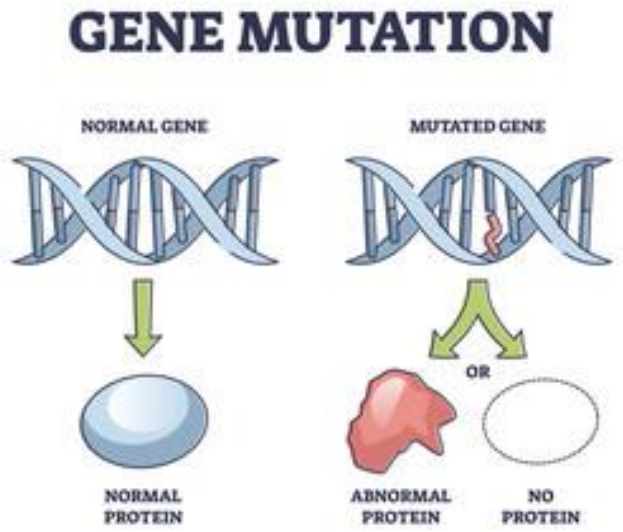
- ✓ A better understanding of why your cancer developed
- ✓ Improved knowledge of how to guide your treatment
- ✓ Identification of possible future cancer risks
- ✓ Information about cancer risks for your family members

Our genetic information, also known as DNA, is in every cell of our bodies, packaged into genes. Each gene has a specific job, such as helping to control cell growth. We have two copies of each gene: one from our mother and one from our father. A mutation, or error, in a gene *may* lead to cancer.

Most cancers are random. This means they are caused by multiple factors such as aging and environmental exposures. Only 5-10% of all cancers are due to hereditary factors such as genetic mutations passed down from parent to child. These mutations affect males and females equally, and they **influence** cancer risk. Influence means the risk is not 100%. Some people who have these gene mutations never develop cancer.

You may be a candidate for genetic testing if you or your family have 1 or more of the following risks.

- Multiple blood relatives who have had cancer. Although first degree relatives – parents, siblings, children – are most important, also consider cancers in grandparents, aunts, uncles and cousins too.
- Cancer diagnosed at a young age - generally before age 50.
- More than one primary cancer (not recurrence or metastases) diagnosed in a single family member.
- Rare cancers (ex: male breast, ovarian or pancreatic).
- Being of certain ancestral backgrounds (ex. Ashkenazi Jewish).



The most common hereditary cause for breast and ovarian cancers are mutations in the BRCA1 and BRCA2 genes. Mutations in these genes increase the risk for both men and women to develop certain types of cancer.

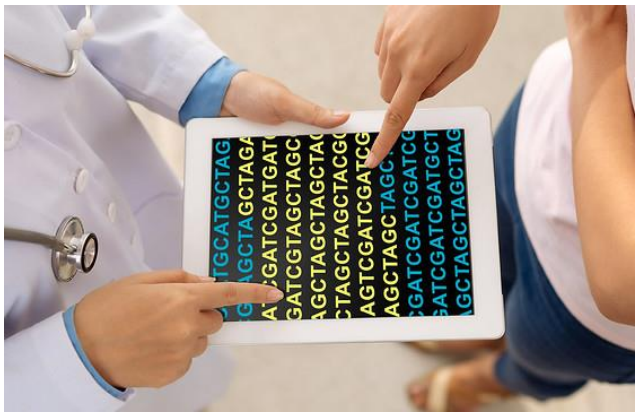
Women have an increased risk for breast and ovarian cancer, while men have an increased risk for breast and prostate cancer. Both men and women may also have higher risks for other cancers, such as melanoma and cancers of the pancreas and colon. There are specific recommendations for individuals with BRCA1 or BRCA 2 gene mutations for screening and risk reduction, so it is important to know if you have a mutation in one of these genes. Additionally, there may be specific treatments your doctor may prescribe to treat a cancer caused by a BRCA1 or 2 gene mutation, such as a medication called a PARP inhibitor.

Another condition associated with hereditary cancer risk is called Lynch syndrome (also known as Hereditary Nonpolyposis Colorectal Cancer or HNPCC). It is caused by a mutation in one of the mismatch repair genes (MLH1, MSH2, MSH6, PMS2, EPCAM). Men and women with Lynch syndrome have a higher risk of developing colon cancer, as well as cancers in the stomach, pancreas, kidneys, prostate, and other locations. Women have a higher risk for uterine and ovarian cancer.

Just like individuals with BRCA1 or 2 gene mutations, there are specific recommendations for screening and reducing risk for those with Lynch syndrome, so it is important to know if you have a mutation in one of these genes. Additionally, there may be specific treatments your doctor will prescribe to treat a cancer caused by Lynch syndrome.

In addition to BRCA1 and 2 and Lynch syndrome, there are other genes and conditions that can cause hereditary risk for cancer in families. Therefore, instead of just testing for a single condition or gene at a time, testing can be done through a panel. Panels test more than one gene or condition at the same time. These panels can test many genes related to cancer risk or they may test for just a few. Some panels only test genes that guide treatment or additional health screenings, while other larger panels may also include testing newly discovered genes that are not yet fully understood. There is no standard of who should have a panel test, or which panel to have. Your health care provider will discuss which genes you should consider testing for and the associated cancer risks and medical management options. They will also help you determine which panel options are best for you.

The Genetic Testing Process at Roswell Park



Once you or your health care provider identify that you are a candidate for genetic testing you will either:

- be referred to speak with a genetic counselor to discuss your options for genetic testing and the impact of such for you and your family - OR -
- you may be offered the opportunity to pursue genetic testing directly through your home clinic, in conjunction with a future appointment, where you would watch a patient education video (instead of meeting with a genetic counselor)

- If you decide to pursue genetic testing, you will sign a consent form and lab test request form, and have your blood drawn
- Genetic test results take approximately 3 weeks to reach your health care provider, who will then share this information with you and discuss how it impacts you and your family.

Importantly, genetic testing is voluntary. If you are not interested in pursuing testing at this time, you can discuss it further with your doctor, or, again, request to meet with a genetic counselor for further discussion. Meeting with a genetic counselor does not obligate you to pursuing genetic testing.

Insurance Coverage

Genetic testing is typically covered by insurance when people meet certain criteria and when test results will have an impact on your medical care. Based on your personal and/or family history of cancer, health care providers can generally determine the likelihood that genetic testing will be covered by insurance. The laboratory performing the test will help verify your insurance coverage and typically will contact you directly if you are expected to have to pay more than $100 out-of-pocket for your testing. The lab typically offers discounted rates and payment plan options for those who need such.

Test Results

It is important to have a conversation with your doctor or genetic counselor about your genetic test result, as this information means different things for you and your family. Your test result may provide powerful medical management information, however it may also cause feelings of anxiety or guilt.

There are 3 possible results when having genetic testing.

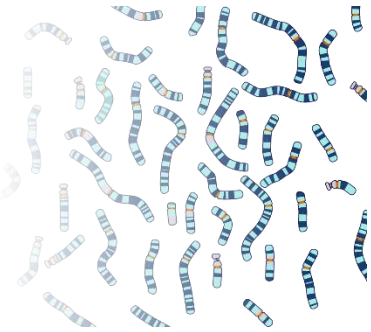
1. **Positive**: A positive test result means a mutation was found in one of the genes tested. Individuals with a mutation are at higher risk for cancer, and their screening and medical management should be altered with this in mind. If you are currently being treated for cancer, you may be a candidate for certain targeted treatment approaches. Your health care provider will discuss these options with you.

   This is also important information for your family. Again, we have two copies of every gene, and it is

random which copy is passed from parent to each of their children. Therefore, each of your brothers, sisters, and children have up to a 50% chance of also having this same genetic change. We cannot control what genetic information we inherited from our parents, nor what our children inherit from us. Genetic testing is an appropriate consideration for your relatives to help them better understand their risk for cancer and approaches to their screening and medical care.

2. **Negative:** A negative test result means that no mutation was found. It is possible that there is not a genetic cause to the cancer in you or your family. It is important that you understand that a negative genetic test result does not entirely rule out an increased risk for cancer. Perhaps we don't have the right testing available yet to identify the possible genetic cause for the cancers in your family. With a negative genetic test result, you and your family's cancer screening should be based on your personal and/or family history of cancer.

   If you have negative genetic testing, testing of these genes is not indicated for your children (based on your personal and/or family history of cancer). Testing for other family members, however, may still be indicated based on their personal and family history of cancer. A genetic counselor can perform a risk assessment based on your family history and make recommendations.



3. **Variant of Unknown Significance (VUS)**: Most of the time, testing gives a clear positive or negative result, but sometimes results are inconclusive. This result is called a *variant of unknown significance.* This means a change was found in one of the genes tested, but there isn't enough known about this exact change to say if it is associated with cancer risk, or if it is benign (not associated with a cancer risk). Variants are treated as a negative result until more information is found. When the laboratory collects enough data about the variant, they send the information to your health care provider, who will then share such with you. Most variants are ultimately found to be benign changes.

Anyone can be found to have a variant, or more than one variant, when they have genetic testing. Medical management should not be changed because a variant was found, and in most cases other family members are not recommended to have testing for a variant.

Legal Considerations

When considering genetic testing, it's important to know your legal protections. HIPAA is a law that protects your medical information from being shared. Your genetic information is held at an even higher level of protection than your other medical records. Your genetic information is only given to the doctors who need it in order to care for you – and it is not shared freely.

The Genetics Information Nondiscrimination Act of 2008, also known as GINA, guarantees that if you have group health insurance, you will receive the same health insurance coverage for the same cost as everyone at your place of employment. There are some exceptions to this law, so please ask your health care provider if you have questions.

Be aware that other coverage, such as individual health insurance plans, life insurance, long term care insurance, and disability insurance, have no limitations on what can be asked on an insurance application or on how much an individual will be charged. Getting a genetic test result, just like changes in your health, does not impact existing policies already in place.

Questions

If you have questions and/or are interested in pursuing genetic testing you should discuss such with your health care provider. You can also request to be referred to speak with a genetic counselor, or you can call 1-800-ROSWELL to set up an appointment yourself.

Want to learn more about genetics and genetic counseling?

Click on the link or type in the URL provided.

- Genetic Counseling - Roswell Park Patient Education (video on youtube)

  URL:https://www.youtube.com/playlist?list=PLFHGdK7nQxlO6bvN3hr67nFhJMjjFbVPL

- National Library of Medicine (NLM), Medline Plus Genetic Disorders

  URL: https://medlineplus.gov/geneticdisorders.html

- NLM, MedlinePlus: How are genetic screening tests different from genetic diagnostic tests?

  URL:https://medlineplus.gov/genetics/understanding/testing/differenttests

- National Human Genome Research Institute

  URL: https://www.genome.gov

- NLM, National Center for Biotechnology Information: Genetic Testing Registry

  URL: https://www.ncbi.nlm.nih.gov/gtr/