Breast Cancer: Genetic Testing

Cancer is a disease of the genes. Most cancers develop because of genetic damage or a change (mutation) that happens sometime during a person's lifetime. These are called sporadic cancers. This means they occur by chance. These types of mutations only affect the cells that grow from the original mutated cell.

Cancers that result from DNA damage that is passed on are called hereditary cancers. These cancers tend to run in families. They cause hereditary cancer syndromes. But only about 1 in 20 to 1 in 10 breast cancers are hereditary. If you may be at risk for a hereditary cancer syndrome, genetic testing can be helpful. It can be used to look for the gene changes linked to these syndromes.

Cancer and genes

The risk of developing cancer is higher in people who are born with certain gene changes (mutations) that are passed on from their parents. These gene changes are in every cell in their body. These changes are in areas of the DNA that affect cell division, cell death, and the repair of damaged DNA.

Genes are pieces of DNA. They determine how your body looks, grows, and works. But environmental factors affect the true outcome. For example, a person might have the genetic potential to be 6 feet tall. But if they had poor nutrition as a child, they may not reach that height. Or a person may have a genetic risk of developing breast cancer. But whether they develop it may be affected by things they have contact with during their lifetime.

Genes come in pairs. One pair is passed down (inherited) from your mother. The other pair comes from your father. Genetic risks and outcomes can come from either parent.

Several genetic defects can cause an increased risk for breast cancer. The most common defects are found on genes named BRCA1 (breast cancer gene 1) and BRCA2 (breast cancer gene 2). A person who has inherited a harmful mutation in BRCA1 or BRCA2 is at higher risk for breast cancer and some other types of cancer. But not all people with BRCA 1 or 2 mutations develop breast cancer. And not all people with breast cancer have these mutations. Environmental factors can affect a person’s cancer risk, just as they affect things such as height.

Common cancer syndromes that can be found with genetic testing

These are common gene mutations linked to cancer syndromes that can be found with genetic testing:

- **APC genemutations.** These can cause familial adenomatous polyposis (FAP). This syndrome causes colon polyps, colon and small intestine cancer, and cancers of the stomach, bone, skin, brain, and other tissues.
- **BRCA 1 and BRCA 2 gene mutations.** These are linked to hereditary breast and ovarian cancer syndrome. These mutations can put a person at an increased risk for breast cancer. They also raise the risk for ovarian, pancreatic, and prostate cancer.
- **MSH2, MLH1, MSH6, PMS1, and PMS2 gene changes.** These can cause Lynch Syndrome (hereditary nonpolyposis colorectal cancer). This is linked to many kinds of cancer. These include cancers that start in the colon, rectum, endometrium, ovary, kidney, ureters, pancreas, small intestine, liver and biliary tract, stomach, brain, and possibly breast.
- **PTEN gene changes.** These cause Cowden syndrome. This is linked to breast, thyroid, endometrium, ovarian, and other cancers.
- **TP53 and CHEK2 gene mutations.** These cause Li-Fraumeni syndrome. They are also linked to an increased risk for many cancer types, including breast cancer, sarcoma, leukemia, adrenal cancer, and brain tumors.

There are other rare gene mutations that can cause inherited breast cancer, too.

Deciding on genetic testing for cancer

Genetic testing for cancer should strongly be considered for a person if:

- They have personal or family history that suggests a hereditary cancer syndrome, such as:
    - Cancers have been found at unusually young ages
    - Several close relatives who have had the same kind of cancer
    - One person has had multiple types of cancer
    - Cancer has been diagnosed in both organs when organs are found in pairs, such as both breasts or both kidneys
    - There's evidence of other birth defects that are linked to certain cancer syndromes
    - Other family members have been tested and have genetic mutations that are linked to cancer
- A genetic test is available and has been proven to be accurate.
- The results of testing can help the person make decisions about medical care.

Anyone considering genetic testing should first talk with a healthcare provider about it. It's important to understand the limits of genetic testing and how the test results might be used. You also need to discuss the costs and find out if your health insurance will help pay for testing.

If you are considering genetic testing for cancer risks, you face many complex issues. If you test positive, you may face difficult decisions about treatments to prevent cancer. You may have increased fear and anxiety about developing cancer. You may have concerns about losing your health insurance coverage. If you test negative, you may also face difficult challenges, such as survivor guilt, if other family members have tested positive.

But if you are anxious or considering treatments, such as preventive surgery based on your family history, having the results may be very helpful. Because of these issues, you should seek genetic counseling before, during, and after any genetic testing. Remember, genetic testing cannot tell you everything about inherited cancers. A positive result does not mean you will develop cancer. In most cases, other factors are involved. In the same way, a negative result does not mean you won't get cancer. Genetic counselors can talk to you about what a test will or will not tell you. They can help you decide whether to get tested.

How is genetic testing done?

In most cases, a blood sample is needed to do genetic testing. Sometimes saliva, skin cells, or cells swabbed from the inside of the cheek can be used. The sample is sent to a lab where the testing is done. It usually takes a few weeks to get the results.

You will meet with the genetic counselor to go over the results and talk about what they mean for you and your family.

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