

### Breast cancer and genetics

If you have been newly diagnosed with breast cancer, it is not uncommon to worry about your loved ones. Many patients want to know about possible tests which could confirm an increased risk for breast cancer to their daughters, sisters, and even their sons or brothers.

Five to 10% of all breast cancers occur because of a gene mutation that allows cancers to happen more easily. Fortunately, most breast cancers do not occur because of any gene mutation. During our lifetime, our cells are continuously growing and making copies of themselves. Most breast cancers occur because of natural mistakes that happen during this process. Hormones can influence breast tissue growth and development and increased exposure to hormones can be associated with a higher chance of abnormal cells forming. The older we get, the more cells we've reproduced over the course of our lives. This means the longer we live, the greater the chance of mistakes happening. In other words, more often than not, having breast cancer has nothing to do with any problems in our genetic makeup, nor that of our relatives.

### What is genetic testing?

Genetic testing is performed as a blood test or from a sample of cells swabbed from the mouth along the inside of the cheek. All the cells in our bodies contain DNA within the genes that we inherit in equal amounts from our biological parents. Genetic testing analyzes our DNA pattern, comparing it to the typical pattern found in the majority of people to look for abnormalities called "mutations." Occasionally, an atypical pattern has been found within certain members of families that have had multiple or unusual cases of cancer. Research has shown that some of the genes with these abnormal patterns are in charge of regulating the body's "security systems" that detect unhealthy cells. If problems exist within these systems, abnormal growth and development can occur, which may result in cancer.

### When genetic testing is recommended

We typically recommend that a patient or family undergo evaluation for a possible genetic mutation if there is an unusual pattern of cancer in that family (see box below).

**The current recommendations to suggest that a patient or family be considered for genetic testing are the following:**

- ◆ Personal history of breast cancer
  - Particularly those:
    - Diagnosed younger than 50
      - Triple negative at any age
    - Personal history of two breast cancers
    - Known mutation within a family
    - 2 or more family members with a history of breast cancer
    - Any family member with a history of ovarian cancer
- ◆ Multiple related cancers within a family
- ◆ Ashkenazi Jewish heritage in breast cancer patients
- ◆ Male breast cancer in a family
- ◆ Breast cancer under the age of 45

It is best to have a patient who actually has or had cancer first undergo the tests to see if a genetic mutation is present. If not found in that patient, it would not be helpful to test any other relatives. If a gene mutation is found, that patient's relatives can then undergo testing to see if they share the mutation and might have a higher chance of cancer in the future.

## Test Results

There are three possible results from genetic testing for breast cancer: positive, negative, or indeterminate.

### Positive

Testing positive means that the genetic pattern found in your DNA is one we have seen in other families that makes cancer happen more easily. For you specifically, this may mean that we will monitor other organs for cancer, depending on the type of mutation present. Most commonly, we are concerned with an increased risk of ovarian cancer.

Having a genetic mutation can mean the chance of cancer happening again is greater. If you have not yet been treated for cancer, based on the possible risks, we may recommend being more aggressive during and after treatment. For example, we may want to perform prophylactic surgery to remove more healthy tissue than we originally planned or we may increase post-treatment monitoring. It does not change the prognosis or our ability to treat your cancer.

Testing positive for a gene mutation is a powerful piece of information for you and your family. It is not “bad” news. Rather, it is a gift of looking into the possible future and having the ability to prepare for it. It allows any of your relatives, male or female, the opportunity to decide if they want to undergo testing.

If you have tested positive, and your relatives want to know what this means to them, it is now very straightforward. It is a 50/50 chance that they share the gene mutation. If they are found to be gene positive and share a gene mutation with you, the first step is to make sure they are currently free of disease. If no disease is found, a plan for future screening, prophylactic surgery, or preventative medicine may be discussed to reduce their risk of ever having cancer themselves. If they test negative and do NOT share the mutation with you, then their risk falls to the level of an average person.

### Negative

If a person with a concern for a possible mutation undergoes genetic testing and is found to be negative for any type of mutation, this is because of one of three possible reasons:

- 1) The person and their family members truly have no genetic mutation. This means that there is nothing in particular about their genetic makeup or genetic pattern that allows cancer to happen in them any more easily than in any other person.
- 2) Because we share so many different genes that combine to make us who we are, there may be several different genes that when combined allow cancers to happen more easily in certain patients. Some family members may have more of these genes and other family members may have less. At this time we do not have the technology to detect complicated combinations of genetic patterns. In this situation, we may have a high level of concern that a family has a higher risk of cancer because of existing cancer in that family. However, we would be unable to perform any tests to predict whether or not one person in the family has a higher risk or has a lower risk.
- 3) It may also be possible there is a distinct genetic mutation that could be detected, but we have not developed a way to detect it. In other words, the gene mutation may be there, but we just don't have a test for it yet.

### Indeterminate

The third possible outcome, an indeterminate result, means that the normal common configuration of the gene pattern is not present. However, the pattern that is present has not been detected in enough families with cancer for us to know for sure that that it adds to the risk for cancer or not. As we gather more information on more families, we may come to show that it is either significant or that it is a safe variation that does not increase that person's risk for cancer. This person would likely be watched closely, but surgery or preventative measures would be offered on a case-by- case basis, depending on how concerning the pattern is in the family and the level of concern of the patient.

## The overall benefits of genetic testing

If you and your doctor find a pattern in you or your family that is striking enough to suggest that a gene mutation could be responsible for your cancer, then it would be worthwhile for you to be tested. If a mutation is present it does not change what we think about your cancer or our ability to take care of you. However, the knowledge of a mutation gives us information as to why the cancer occurred and may give us insight into the future for your relatives.

Obtaining information to review with a genetic counselor can be a positive experience of reconnecting with your family. If you use this as a reason to talk with your relatives about your family history, you might learn things you did not know before.

When provided with accurate information, genetic counselors can be extremely helpful in deciding whether genetic testing would be in your best interest. If a genetic mutation known to be associated with increased risk of cancer is discovered, this can be a gift of information and advanced knowledge for you and your family.

**We are happy to discuss your personal and family history with you to see if you would be a good candidate to meet with one of our experienced genetic counselors.**

## Getting tested even though you don't meet the recommendations

Sometimes patients want to be considered for genetic testing even though they or their families may not meet the recommendations for testing. If you do NOT meet the requirements and decide to seek counseling or testing anyway there are some things to consider:

Reasons against testing	Reasons for testing
<p><b>Insurance Concerns:</b> Some people worry about being penalized on their health insurance policies from undergoing testing or having positive results in their family. Laws are in place to protect patients and their rights and only in a very few individual instances have there been difficulties with policies covering care for diseases or imaging plans for follow-up for genetic mutations. In fact, most patients find that additional testing or procedures are justified by the presence of the strong family history or genetic mutation.</p> <p>It is true that life insurance coverage or rates may sometimes be affected by the presence of a genetic mutation in a person or family; but the simple presence of cancer or concern for it in a family may also affect them. However, if you plan on obtaining a life insurance policy and are considering genetic testing or a thorough investigation into your family history, you may want to address your insurance first.</p> <p><b>Confusing results:</b> Without significant personal or family history, the likelihood of testing positive is incredibly low; it may be an unnecessary expense or results may cause confusion or unfounded concern. A negative result can give a person a false sense of security that they have a normal or lower risk than the relative who had cancer.</p>	<p><b>You do test positive:</b> Even if there is a low likelihood of being positive, if you are in the small percentage of people found to be positive, you have a chance to be proactive and make advanced decisions that may greatly impact your life and your family. It gives you the opportunity to be more proactive with surgery and surveillance, consider preventative medical therapy or change your personal habits.</p> <p><b>Anxiety about health:</b> If you have a great deal of anxiety about breast cancer and you test negative, while it does not mean you can't get breast cancer or that there is no inheritable increased risk for you, simply knowing that you do not have a documented gene mutation may give you peace of mind. Increased or person-specific screening plans may be indicated. Please go over this with your doctor if you feel this may apply to you.</p>

