Genetic Testing and Healthy Patients:
how to know if it is right for you

Breast Cancer and Genetics

Patients who have family members with breast cancer are frequently worried that they may share genetic traits that make them more susceptible to cancer. They have questions such as, "If my grandmother and great aunt had breast cancer – am I bound to get it too?" Many patients want to know about possible tests which could confirm an increased risk for breast cancer.

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, or that of our relatives.

What is genetic testing?

Genetic testing is performed as a blood test or from a sample of cells swabbed from inside the cheek. All cells within our body 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 of cells can occur, which may result in cancer.

When genetic testing is suggested or recommended

If you do have a pattern of cancer in your family that suggests that a genetic mutation may exist, we will want details about your family history and will want to know if it is possible to test some of your relatives who have actually had the cancer.

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

- Three or more first degree relatives with breast cancer
- Breast and ovarian cancer in a single patient
- Breast cancer and ovarian cancer in two first degree relatives
- Ashkenazi Jewish heritage in breast cancer patients
- Male breast cancer in a family
- Breast cancer under the age of 45
- Unusually aggressive types of cancer profiles, such as triple negative or basal type

Barber & Richardson, PC

275 Collier Rd, NW #470
Atlanta, GA 30309
404-351-1002
www.b-rsurgical.com

Acquiring Genetic Testing Results from Relatives with Cancer

It is important to note that testing a person who has had cancer is the BEST, MOST ACCURATE way to know if there is a gene mutation. Testing an unaffected relative is usually NOT the best place to start looking for a gene mutation.

If you CAN get results from relatives

If you have relatives who are survivors and are interested in testing or have been tested, then you are in the best position to benefit from this information. If your relative has tested negative for a mutation, testing yourself would not be helpful. If they have tested positive for a mutation that contributed to the formation of their cancer, you then have a choice to undergo testing yourself and see if you share the mutation with them or not …

… if you DO have a genetic mutation

You have been given a gift of information that many people are very grateful to have. Your worry about the future probably hasn't changed that much, but now we have justification to watch you more closely and to be more proactive with preventive measures.

It does not mean you WILL get cancer or that any future cancer can't be treated. It does mean that your risk is much higher than if you did not have the mutation. We make specific recommendations to patients, depending on the type of mutation they have, their age, and their individual health characteristics. If you have questions about testing positive for a gene mutation when you are a person with no evidence of cancer, we will be happy to provide detailed answers.

… if you DO NOT have a genetic mutation

Congratulations. It does not mean you can't or won't get cancer, but that if you do get cancer in the future, it will be because of chance and not because there is something in your makeup that allows cancer to happen more easily.

If you CANNOT get results from relatives

Your relatives who had or have cancer may be unable or unwilling to undergo testing. If a healthy person undergoes testing without having test results from a relative with we have no reference points, which can create a complicated picture with three possible outcomes:

Indeterminate	Positive	Negative
This means that the normal gene pattern is not present. However, the specific pattern that is present has not been detected in enough families with cancer for us to know for sure if that gene pattern adds to the risk for cancer. As we get more information on more families, we may find the significance of the pattern. A person with an indeterminate result would likely be watched closely, but surgery or preventative measures would only be offered as a case-by-case basis, depending on how concerned the patient is or how concerning the pattern is in the family.	This is the most straightforward of all outcomes. It leaves the least number of unanswered questions. If the person tests positive for the genetic mutation this means that there is an increased likelihood their relatives also have this same mutation. It will also allow other relatives to be tested to see if they have an increased risk.	This can be the most com-plicated and confusing of all three outcomes. It is also the most common. More often than not a healthy person with a family history of cancer will test negative for a gene mutation. *This outcome is discussed in more detail on the next page*

If a patient with a strong family history tests negative and we have no other relatives to reference the significance of this result, this could mean one of several things:

- There is a genetic mutation in a relative or family that we are unable to know about with any certainty. If we were in a position to know for sure that a relative had a mutation, then that the patient testing negative would have an average risk just like any other person without cancer in their family. However, without an additional test result from a relative with cancer to confirm this, we may never know.

- There are combinations of genes that may contribute to the formation of cancers in the patient's family. With multiple factors present, some family members would have a higher risk and others lower. We would not have a single test to declare the level of concern for the individual patient. In this case, we would be generally concerned about increased risk and we would watch out for that patient carefully. The type of imaging studies and plans for follow-up and screening would need to be tailored to the patient in this situation.

- There is a genetic mutation present; however, science may not have discovered this gene mutation or the tests currently available are not designed to detect this mutation. This patient may ultimately be found to be gene positive if they are re-tested in the future and it may be necessary to have patients or their family retested as new discoveries are made. We ask these patients to check back every 2 to 3 years to see if new developments have been made or if re-testing is indicated. If the patient or one of their family members develop a cancer, they can be reconsidered for testing at that point, which may put genetic testing results into better perspective.

The overall benefits of genetic testing

Obtaining information to review with a genetics 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 when you don't meet the recommendations

Sometimes currently healthy patients want to be considered for genetic testing even though they or their families may not meet the recommendations for testing. They have relatives with cancer or relatives that have survived or passed away as a result of cancer and they may be concerned about their own risks and want genetic testing. They may have a limited knowledge of their family history because of adoption or other circumstances, or they may want genetic testing for other reasons. For these patients there are some pros and cons associated with having genetic testing:

Reasons against testing	Reasons for testing
Insurance Concerns: 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. 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. ***Confusing results:*** Without significant personal or family history the likelihood of testing positive is incredibly low and 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.	***You do test positive:*** 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 you 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. ***Anxiety about health:*** 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.