myBRCA Test Considerations and Possible Outcomes

myBRCA is a screening test intended to inform you and your doctor about potentially increased risk levels for breast and ovarian cancer based on the presence of a BRCA mutation. Below we review questions that may be helpful when considering myBRCA, including possible test outcomes. Please note that any outcome - positive, negative or inconclusive - together with other genetic and non-genetic risk factors, should be further discussed with your doctor when making decisions about your healthcare.

If you have a personal and/or family history of breast and/or ovarian cancer, should you get screened for BRCA mutations?
Most healthcare professionals agree that in general, patients with a history of breast and/or ovarian cancer, or with a strong family history of breast and/or ovarian cancer, should be offered BRCA1 and BRCA2 genetic testing. In these situations, if the BRCA1/2 testing is negative, further genetic testing may be advised (i.e. myBRCA HiRisk). While mutations in the BRCA1 and BRCA2 genes are the most common causes of inherited breast and ovarian cancer, there are other genes which may be involved in these hereditary cancers. You should discuss your testing options further with your healthcare provider.

Should you undergo myBRCA screening if you have no known family members with cancer?
Recent studies show that some 50% of women with a BRCA mutation have no substantial family history of breast and ovarian cancer. This research suggests that 1 out of 2 women have no idea they are carrying harmful (cancer-causing) mutations. Our mission is to make BRCA1 and BRCA2 testing available to anyone who wishes to know. The decision of whether to screen for BRCA1 and BRCA2 mutations is ultimately yours. Some factors to consider are your age, how the results may impact your medical care, and how your results may impact your family members.

What if you test positive for a harmful mutation?
A positive result indicates that a harmful BRCA1 or BRCA2 mutation has been identified, which significantly increases your lifetime risk for breast, ovarian, and/or other cancers. For instance, a woman with a BRCA1/2 mutation may have an up to 80% chance of developing breast cancer and an up to 40% chance of developing ovarian cancer in her lifetime. Men with BRCA1/2 mutation have an up to 39% chance to develop prostate cancer and an up to 10% chance to develop male breast cancer in their lifetime.* There may also be increased risks for other cancer types for both men and women with a BRCA mutation.

It is important to note that carrying a harmful mutation in one of the BRCA genes does not mean that you will necessarily develop cancer. In fact, the risk for developing some cancers can actually be significantly reduced, if recommended medical management is followed.

It is strongly advised that all individuals who test positive for a harmful BRCA mutation discuss their own personal and family history with a genetic counselor or other qualified healthcare provider so that appropriate screening, treatment options and options for prophylactic surgeries can be discussed.

Additionally, because of the hereditary nature of BRCA mutations, there is a 50% (1 in 2) chance that first degree relatives (children, siblings and parents) can inherit the harmful mutation. Therefore, genetic testing for the identified BRCA mutation is also recommended for family members. Your physician or genetic counselor can help you identify which family members are at-risk and should consider testing.
What if you test negative?
A negative test result indicates that a BRCA mutation has not been identified. This can be more difficult to understand than a positive result. What the result means varies based on an individual’s personal medical history, family history of cancer, and if a mutation has been identified in a blood relative.

If a close (first-, second-, or third-degree) blood relative of the tested person is known to carry a harmful BRCA mutation, a negative test result is clear: it means that person does not carry the harmful mutation that is responsible for the cancer in the family, and thus cannot pass it on to their children. Such a test result is called a true negative. A person with such a test result is currently thought to have the same risk of BRCA associated cancer types as someone in the general population. A true negative result, however, does not eliminate the need for cancer screening and/or treatment. Your physician or genetic counselor will help you make appropriate decisions regarding your health and clinical needs.

A negative result is less clear if the tested person has a family history of cancer that suggests the possibility of having a harmful mutation, but gene testing identifies no such mutation in the family. The likelihood that genetic testing will miss a harmful mutation is very low, but it could happen. Moreover, scientists continue to discover new mutations and have not yet identified all potentially harmful ones. It is also possible that there is a mutation in an undiscovered gene. Therefore, in this scenario it is possible that a person with a “negative” test result may actually have an unknown harmful mutation or a mutation in an undiscovered gene.

A negative result is also unclear if the tested person has a family history suggestive of a harmful BRCA mutation, but the affected family member(s) have not undergone gene testing. In this situation, the underlying cause of the family members’ cancer is unknown, and it is difficult to interpret the negative myBRCA result in the person being tested. This person may still be at increased risk to develop breast and/or ovarian cancer due to harmful mutations in cancer predisposition genes not included on this test.

What if your results come back inconclusive?
You may learn that one or more variants of uncertain significance (VUS) were identified by this genetic test. A variant of uncertain significance (VUS) is a genetic change but the significance of this alteration remains unknown – usually because of limited or conflicting evidence in the medical literature. In the absence of further information, the interpretation of the variant is inconclusive and testing of other family members may be desired when possible to help determine the significance of the variant. Based on current medical literature, it is recommended that healthcare decisions should NOT be based on an “inconclusive” result. Your physician or genetic counselor will help you make appropriate decisions regarding your screening and/or treatment based on your personal and/or family history.