



Consent for Hereditary Cancer Genetic Testing for BRCA1 and BRCA2

Introduction. This form describes the benefits, risks, and limitations of genetic testing for inherited susceptibility to cancer. Please read this form carefully and discuss any questions that you have with your team of providers.

Purpose. This test analyzes a specific gene or genes for genetic changes called mutations. The gene(s) analyzed are associated with a specific hereditary cancer syndrome. This test will help determine if a person has a significantly increased risk of developing certain tumors due to a mutation(s) in a cancer-predisposing gene. Not everyone who has a gene mutation will develop cancer. The cancer risks associated with a *BRCA1* or *BRCA2* gene mutation are listed below. The percentages cited are estimates and may change as more research is done.

- ***BRCA1* gene alterations:** A woman with a *BRCA1* gene mutation has a 50-85% lifetime chance of developing breast cancer, and a 20-40% chance of developing ovarian cancer in her lifetime. There may also be a slightly increased chance of developing other cancers. In comparison, women in the general population have a 10-12% lifetime risk for breast cancer and 1-2% lifetime risk for ovarian cancer. Men who carry a *BRCA1* gene mutation may have a slightly increased risk for breast and prostate cancers, although these risks are not well defined.
- ***BRCA2* gene alterations:** A woman with a *BRCA2* gene mutation has a 50-85% lifetime chance of developing breast cancer, a 10-20% chance of developing ovarian cancer in her lifetime, and an increased chance of developing other forms of cancer, such as pancreatic cancer (though less is known about this). In comparison, women in the general population have a 10-12% lifetime risk for breast cancer and 1-2% lifetime risk for ovarian cancer. A man with a *BRCA2* gene mutation has a 6-10% lifetime risk of developing breast cancer and also has a slightly increased chance of developing other forms of cancer, including prostate and pancreatic cancers. The risk for a man to develop breast cancer in the general population is about 0.1% or 1 in a 1000.
- Both men and women with a *BRCA1* or *BRCA2* gene mutation have a 50% (1 in 2) chance of passing it on to each of their children. There is no evidence that being born with a *BRCA1* or *BRCA2* gene mutation increases the risk for childhood cancers.

Test Procedure. *Hartford Hospital* personnel will draw approximately 1 tube of blood and send it to Myriad Genetic Laboratories, Inc. The laboratory will analyze your *BRCA1* and *BRCA2* gene(s) to look for mutations. No other tests will be run on your sample without your written consent.

You are consenting to the following tests:

- Comprehensive BRACAnalysis - This test is a full sequence analysis + 5-site Rearrangement panel of the *BRCA1* and *BRCA2* genes
- BART – BRACAnalysis Rearrangement Test
- Single Site BRACAnalysis - If a *BRCA1* or *BRCA2* mutation has been previously found in your family, single-mutation analysis may be recommended to examine only the small portion of the DNA in which the familial mutation was previously identified
- Multisite3 BRACAnalysis -If you are of Ashkenazi Jewish ancestry, you may choose this test which tests for the three most common mutations found in persons of Ashkenazi Jewish ancestry
- Reflex BRACAnalysis - This test is for individuals whose results from Multisite3 BRACAnalysis are negative

_____ Patient initials

Test Results and Interpretation. Listed below are the possible results from testing:

- Positive – A mutation that is associated with an increased risk for hereditary cancer was identified. Knowing this information may help you and your doctor make more informed choices about your health care, such as screening, risk-reducing surgeries and preventive medication strategies.
- Negative – A mutation was not identified.
 - If you are the first person tested in your family, you still have at least the same risk of cancer as does a person in the general population. You may still be at greater than average risk for hereditary cancer due to a genetic predisposition that cannot be detected by this test, either in the gene(s) you were tested for or in another gene linked to hereditary cancer.
 - If you test negative for a mutation known to be in your family, you are considered to have the same risks as others in the general population.
 - None of the three specific mutations on Multisite3 Analysis were found. Since other mutations may exist elsewhere in the genes, persons with a negative result from this test may still be at high risk of breast and/or ovarian cancer due to other changes in the *BRCA1* and/or *BRCA2* genes or another gene not analyzed in this test.
- Uncertain – A genetic change (a Variant of Uncertain Significance, or VUS) was detected but it is not known if this change is linked to cancer risk. You still have at least the same risk of cancer as the general population, and may still be at greater than average risk due to a genetic predisposition that cannot be detected by this test.



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The genetic counselor will discuss the results with you and what they mean. Genetic test results have implications for blood relatives. If you choose, the genetic counselor will help you inform family members if a mutation is found.

Benefits. Your genetic test results may help you and your doctor make more informed choices about your health care, such as screening, risk-reducing surgeries and preventive medication strategies. The identification of gene mutation(s) in a family enables other blood relatives to determine whether or not they share the same hereditary cancer risks. If you test positive, you should discuss with your healthcare provider how hereditary cancer is inherited and learn about the chance your children and blood relatives may have inherited the same mutation(s) in the gene(s) tested. If you test negative for a known mutation in your family, you cannot pass on that mutation to your children. If you have had cancer or a pre-cancerous condition, you would still have some increased risk based on your own history. If you have not had cancer, you would still have the general population risk of cancer.

Risks. Learning that you have a genetic mutation may be upsetting and may increase feelings of depression, anxiety, and vulnerability. You could feel that you have to make difficult decisions about your medical care. Learning of a positive result may affect relationships with family members. News of an inconclusive result may increase feelings of distress. If more than one family member is tested, there is a possibility of learning sensitive information about your family, such as non-paternity or undisclosed adoption.

To address concerns regarding possible health insurance discrimination, most states and the federal government have enacted laws to prohibit genetic discrimination. Furthermore, broad federal legislation (HIPAA) prohibits unauthorized disclosure of confidential personal health information. However, for the most part, these laws do not prevent life and disability insurers from using genetic testing information in determining coverage. **It is our policy to include genetic information and test results into your medical record.**

Limitations. This test analyzes only the *BRCA1* and *BRCA2* genes. If no mutation is found, you may still be at risk for hereditary cancer due to a genetic predisposition that cannot be detected by this test, either in the gene(s) you were tested for or in another gene linked to hereditary cancer. Genetic testing clarifies cancer risks for only those cancers related to the genes analyzed.

Financial Responsibility. Genetic testing of appropriate individuals is typically reimbursed by health insurance. You are responsible for any cost of the genetic test not reimbursed by insurance, including applicable co-pays or deductibles.

Future Correspondence. Due to the dynamics of this field, there continues to be new information and data. It is recommended that you keep in contact with your healthcare provider, annually, to learn of any new developments in cancer genetics and to provide any updates to your personal or family history which may affect your cancer risks.

I confirm that the possible benefits, risks, limitations and costs of genetic testing for *BRCA1* and *BRCA2* have been explained to me. All my questions have been answered. I have read this consent form and will be given a copy for my records. My signature below indicates my willingness to have genetic testing for *BRCA1* and/or *BRCA2* gene mutation(s). I understand the confidentiality policy outlined in the consent form. This means that I recognize that genetic information and a copy of the test result will be included in my medical record.

Signature of Patient

Date

Time

Print name

I have explained the possible risks, limitations and costs as well as potential benefits of genetic testing for mutation(s) in *BRCA1* and *BRCA2* and have answered any questions regarding the test to the best of my ability.

Genetic Counselor/Clinician

Date

Time

_____ Patient initials