

Information about Genetic Testing

Why is genetic testing important?

There are ways to reduce your risk or overcome cancer in your lifetime. If a risk of cancer runs in your family, genetic testing may be an important step for you. Genetic testing can give you answers about your risk of inherited cancers by analyzing your genes for mutations responsible for inherited cancer risk. Results will help you make informed decisions about your options for early cancer detection and risk reduction.

If you've already been diagnosed with cancer, genetic testing can provide important information about your risk for a second cancer, which can help your doctor determine the most effective medical management strategy for you.

Genetic testing also provides information important to your family members, especially close relatives. In the case that you have inherited gene mutations that increase the risk for cancer, there's a chance that your parents, brothers and sisters or children might have these mutations, too.

Will my insurance cover the cost of genetic testing?

Most health insurances plans, including Medicare, do cover the cost of genetic testing depending on your medical history and/or family history of cancer.

If genetic testing is appropriate given your medical and family history, a blood sample may be submitted to the laboratory for testing. With your permission, the laboratory will contact your health insurance company on your behalf to determine insurance coverage before the testing begins. If your insurance company will pay 100% of the cost of testing, the laboratory will begin testing immediately. If you are responsible for any amount of the cost of genetic testing, the laboratory will contact you for approval BEFORE testing begins. This method insures you will know exactly how much you will have to pay for genetic testing before your test is performed. When the laboratory contacts you with your out of pocket cost, you will have the option to cancel the test at no cost.

More information about the genes for Breast and Ovarian Cancer, BRCA1 and BRCA2...

Ten percent of all breast and ovarian cancer cases are hereditary. In fact, as many as 1 in 500 people carry a mutation in either the *BRCA1* or the *BRCA2* genes. 1 in 40 people of Ashkenazi Jewish descent (Eastern European) are mutation carriers. Individuals who carry a mutation in one of these genes face a dramatically increased risk of both breast and ovarian cancer. Fortunately there are medical management strategies that can reduce these risks and genetic testing is available to accurately identify mutation carriers.

Individuals who carry a *BRCA1* or *BRCA2* mutation face increased risks for initial cancers, up to 87% for breast cancer and up to 44% for ovarian cancer. Mutation carriers who have already had breast cancer face increased risks for second cancers, up to 64% for another breast cancer and a 16% risk for ovarian cancer.

BRCA mutation carriers can reduce their risk and preempt cancer by altering their surveillance strategies.

Medical management options for *BRCA* mutation carriers include: increased surveillance, chemoprevention and prophylactic surgery. Self breast exams should begin between the ages of 18-21. Clinical breast exams, mammography, transvaginal ultrasound and CA-125 are recommended to begin between the ages of 25-35. Certain drugs such as tamoxifen and oral contraceptives have been associated with cancer risk reduction. Additionally, prophylactic mastectomy and oophorectomy also dramatically reduce a woman's cancer risk. Individuals who should be offered genetic risk assessment are those with a personal or family history of:

- Breast Cancer prior to age 50
- Ovarian Cancer at any age
- Both Breast and Ovarian Cancer
- Bilateral Breast Cancer at any age
- Family members with breast cancer before age 50 OR Ovarian cancer at any age
- Male Breast Cancer

Mutations in *BRCA1* and *BRCA2* are inherited in an autosomal dominant pattern. First degree relatives are at 50% risk to carry the same mutation. Once a mutation has been detected in a family, other family members can be tested for the specific family mutation. Those family members testing positive for the known mutation are managed more aggressively. By contrast, individuals who did not inherit the family mutation are not at increased risk for cancer and may adhere to general population screening guidelines.

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What are the possible test results?

Comprehensive Analysis

Comprehensive Analysis is a full-sequence and, in many cases, large rearrangement analysis of the gene or genes associated with a specific inherited cancer syndrome. Possible test results include:

- **Positive for a deleterious mutation** - Means that a mutation has been found that is believed to increase the risk of developing certain types of cancer.
- **Genetic change of uncertain significance** - Means that a gene alteration has been found but cancer risk has not yet been determined with certainty.
- **No mutation detected** -- No mutations found in the gene or genes. This does not mean you have no risk for cancer. You may still be at high risk for developing cancer because of gene abnormalities that cannot be detected by this test, or because of other gene mutations less commonly responsible for hereditary risk of these cancers