



Newsletter

BRCA TESTING FOR HEREDITARY BREAST AND OVARIAN CANCER

Compiled by: Lancet Laboratories South Africa

What is hereditary or Familial Cancer?

Cancer occurs when cells in your body start to grow out of control. These cancer cells can multiply very quickly, cause growth or tumors, invade surrounding tissues, and even spread. (metastasize) to other areas of the body.

Cancer is a complex disease that can be caused by several different factors. A few types of cancer run in families and this is called Hereditary or Familial Cancer. Hereditary cancers have been linked to changes in genes, this is referred to as mutations that can be passed from parents to their children.

All genes come in pairs, and your cells need both copies to work normally. One copy of each gene is inherited from your Mother, and the other copy from your Father. When you inherit one Gene with a mutation, it increases the risk of you having to develop that cancer, but it does not necessarily mean that you will develop Cancer. The other copy of a Gene without the mutation can keep your cells from growing out of control. But when the normal copy of the Gene is damaged by other factors such as: chemicals, radiation, or smoking, as the risk of developing cancer increases further. Many of the factors that can damage these Genes are still unknown.

What are Hereditary Breast & Ovarian Cancer Syndrome?

Hereditary breast and Ovarian cancer syndrome are a type of Familial Cancer and is linked to mutations in two Genes called BRCA1 and BRCA2. BRCA1 and BRCA2 are known as "Tumor Suppressor Genes" since they prevent cells from growing too rapidly or in an uncontrolled way.

Thousands of mutations have been found in these genes, but only some have been linked to an increased risk of cancer. Inheriting one of these mutations in BRCA1 or BRCA2 increases your risk of developing Breast, Ovarian and certain other types of Cancer. Not all cases of breast or ovarian cancer are associated with mutations in BRCA1 and BRCA2. In fact, only 5 - 10% of Breast cancer cases and 10 - 15% of Ovarian cancer cases are due to mutations in BRCA1 and BRCA2.



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Should I be tested for a BRCA gene mutation?

A Genetic risk assessment and potential BRCA Gene mutation testing is recommended if you have any of the following:

- A personal or family history of Breast or Ovarian Cancer,
- If you were younger than 50 years of age when Breast Cancer was diagnosed,
- If you have had Breast Cancer diagnosed more than once. (In the same breast or in both breasts.)
- If you are a man and diagnosed with Breast cancer at any age.
- If you have a close relative with a known BRCA1 or BRCA2 mutation.
- If your personal and family history is suggestive of a BRCA gene mutation, a blood sample will be taken for genetic testing of the BRCA1 and BRCA2 genes.
- If a specific BRCA gene fault is known in your family, at risk relatives can be tested for that specific mutation.

Understanding your test results

It is strongly advised that you should have genetic counselling before and after testing for any cancer-related gene mutations. This will help you to understand which specific blood test is needed based on your personal and family history, how the test is done, what the test results mean, and help you to set up a management plan or protocol based on your results.

What does a positive BRCA gene mutation result mean?

If a known Cancer-associated mutation is found in a BRCA Gene, you have an increased risk of developing Cancer. (See Table 1) It does NOT mean that you will get Cancer. No test can tell with absolute certainty that you will develop Cancer. Additional Breast Cancer preventive strategies, such as: enhanced screening tests, medication and surgery should be discussed with your Genetic counsellor and Healthcare provider.

	Breast Cancer	Ovarian Cancer
Risk for the general population	11%	1-2%
Risk for BRCA1 mutation	60-80%	60-80%
Risk for BRCA2 mutation	60-80%	10-30%

Table 1. Risk of developing Breast or Ovarian cancer for women with BRCA mutations. Having a BRCA mutation does mean that you can pass the mutation on to your children. Each child has a 50% chance of inheriting their parent's BRCA gene fault. Your siblings may also have inherited the BRCA gene fault.

Recommended testing

- How many people carry a BRCA gene mutation?
- About 1 in 300 people carry a BRCA1 mutation.
- About 1 in 800 people carry a BRCA2 mutation.
- BRCA1 and BRCA2 gene mutations are found more commonly in certain ethnic groups.

