CENTOGENE
THE RARE DISEASE COMPANY

BRCA 1/2

Breast cancer testing

THINK ABOUT TOMORROW, TODAY

Patient information

GENETIC TESTING IS YOUR ALLY

Could you be at an increased risk of developing hereditary breast or ovarian cancer? What is the best way to find out and prepare yourself?

Genetic testing has become the safest, fastest and most reliable method to identify if you or anyone in your family has a genetic predisposition to developing certain cancers.

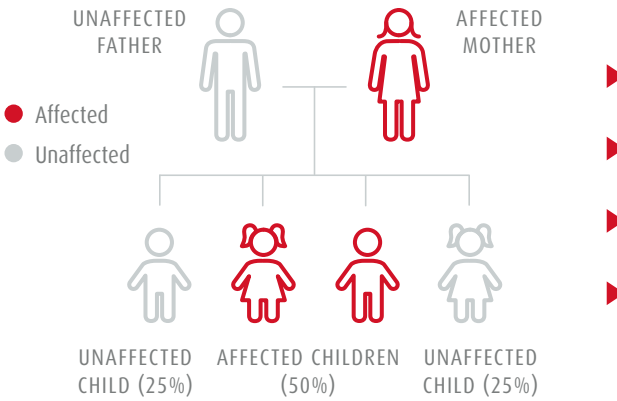
Breast and ovarian cancer

Breast cancer is the most common cancer in women and the **cause of 25% of all female cancers** diagnosed worldwide, with around **1.7 million new cases every year**[1]. Approximately 12% of women will develop breast cancer at some point in their lives, and 1% will develop ovarian cancer in their lifetime.

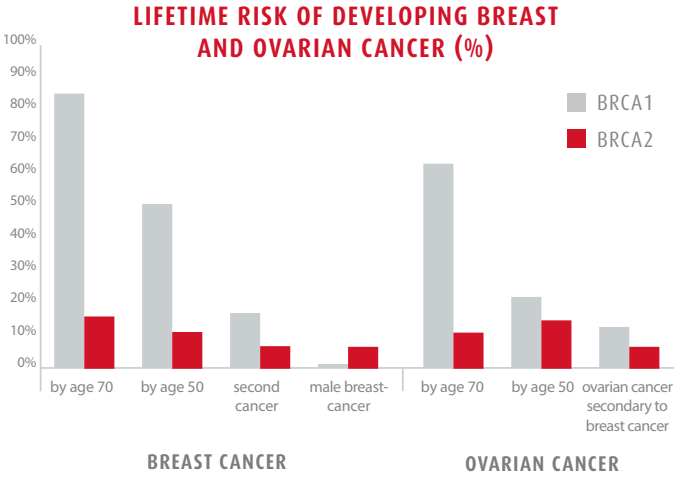
Despite the availability of excellent treatment options for both breast and ovarian cancer, these diseases still have high mortality rates, particularly where the disease is not diagnosed until a late stage. **Early detection is crucial to saving lives**.

BRCA1 and **BRCA2** genes have an important role in our cells, they are needed to **help to repair damage to our DNA** which occurs naturally over time. If you have a defective copy of either BRCA1 or BRCA2 then you have an increased predisposition to developing cancer, known as Hereditary Breast and Ovarian Cancer syndrome (HBOC).

What are the implications for family members?



Faulty BRCA1/2 genes increase the lifetime risk of developing cancer



In the general population, approximately 12% of women will develop breast cancer in their lifetime. In comparison, 55-65% of women carrying a BRCA1 mutation and ~45% of women carrying a BRCA2 mutation will develop breast cancer by age 70.

If you are diagnosed with **Hereditary Breast Ovarian Cancer syndrome**, in other words, if a mutation was found in your BRCA1 and 2 genes, as well as having an increased risk of developing cancer during your life, there is also 50% likelihood you will transmit this to your children.

Sharing any information you obtain will allow family members to decide individually how they want to proceed themselves. They might decide on genetic testing for themselves. If so, testing can be targeted to the specific mutation previously identified from you, saving time and cost.

What are the possible outcomes of the test?

A) POSITIVE

If the test identifies a disease mutation, then an increased predisposition to breast and/or ovarian cancer will be confirmed. This does not necessarily mean that you have cancer or that you will develop it. However, depending on the mutation, you will have an increased likelihood of developing cancer of between 46-87% over your lifetime.

B) NEGATIVE

If you have a personal of family history of breast cancer, but your BRCA 1/2 test is negative, testing for other breast cancer related genes might be still appropriate for you. Therefore, a negative test result should be discussed with your physician.

C) INCONCLUSIVE

Not all genetic variants in BRCA1/2 are disease causing. In some instances, the test may identify a variantion in BRCA1/2 which cannot be conclusively identified as disease causing. In such cases, CENTOGENE will update you if the classification of a variation changes regarding clinical consequence.

References

1. Ferlay J et al. GLOBOCAN 2012 v1.1, Cancer Incidence and Mortality Worldwide: IARC CancerBase No. 11 [Internet]. Lyon, France: International Agency for Research on Cancer; 2014. Available from: http://globocan.iarc.fr.
2. Tai, Y. C., Domchek, S., Parmigiani, G., & Chen, S. (2007). Breast cancer risk among male BRCA1 and BRCA2 mutation carriers. Journal of the National Cancer Institute, 99(23), 1811-1814.

When should you consider testing?

- If you have a family history of breast and/or ovarian cancer (1st-2nd degree relatives affected or diagnosed as carriers for breast and/or ovarian cancer). This is especially important when cases occur at a young age (before 50 years).

- If you have been diagnosed with breast and/or ovarian cancer, testing BRCA1/2 genes can inform you about the risk of developing a second cancer, the prognosis of your disease, the treatment options and if other members of your family should be tested.

Questions to ask your doctor:

- Your suitability for BRCA1/2 screening
- How is the test performed?
- How will you receive your test results?
- Availability of genetic counselling
- Implications of having and not having a mutation
- Lifestyle changes if you are found to carry a BRCA1/2 mutation
- Cost of testing

Myths about BRCA1/2 testing...

Genetic testing is very expensive.

Genetic testing prices have dropped significantly in recent years and it is now an affordable option for most patients. In addition, many private health insurance providers will cover the cost. It is recommended that you contact your insurance provider to confirm what is covered/not covered, or alternatively ask your doctor.

I have already been diagnosed with breast or ovarian cancer, so there is no reason for me to be tested for BRCA1 and 2 mutations.

If you have been diagnosed with breast or ovarian cancer, you are at greater risk of developing a second cancer if you carry a BRCA1 or BRCA2 disease-causing mutation. Depending on your family history, it may also be relevant to identify the presence of BRCA1/2 mutations to assess a predisposition to cancer in family members genetically related to you, in particular, your children.

Mutations can only be inherited through your mother.

No, you can inherit any breast/ovarian cancer mutations from your father and mother.

If I am a man, I do not need BRCA1/2 testing

Men also have breast tissue, just less of it. This tissue can be susceptible to breast cancer. The risk of developing breast cancer by 70 years if you are a man is 1.2% for BRCA1 mutation carriers and 6.8% for BRCA2 mutation carriers[2]. In addition, there is growing evidence to show that BRCA1/2 mutations can also predispose you to other types of cancer.

Please visit our website
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V1.1eng_February2018