Your next steps

Find a specialist
All patients should be seen in a clinical genetics department and/or cancer genetic unit for consultation.

Follow medical guidelines
A surveillance should be undertaken by a specialist according to the guidelines.

Monitor symptoms
Keep track of any new symptoms or health concerns and discuss it with your medical team.

Consider treatment options
Discuss with your medical team available screening, prevention & surgical options.

Stay updated on HBOC research
Clinical trials and research may provide additional opportunities for treatment and care.

Visit **www.genturis.eu** for medical guidelines and expert centres.

About ERN GENTURIS

ERN GENTURIS is a European expert network for hereditary tumour syndromes. While we do not offer direct treatment, we provide patients and healthcare professionals with trusted information, guidance, and connections to specialists.

More info and contact

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European Reference Network

Funded by the European Union





Patient information

Hereditary Breast and Ovarian Cancer Syndrome (HBOC): what this diagnosis means for your health and next steps

If you or a family member has been diagnosed with HBOC, this guide provides clear medical information and practical next steps.

European Reference Network
for rare or low prevalence complex diseases
Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

ERN GENTURIS
With every diagnosis we can help an entire family

What is Hereditary Breast and Ovarian Cancer Syndrome (HBOC)?

HBOC is caused by inherited pathogenic variants, also known as genetic changes, that mainly increase the risk of developing breast and ovarian cancer.The genes most commonly associated with HBOC are *BRCA1* and *BRCA2*. Other genes, such as *PALB2*, can also increase cancer risk.

Women with a *BRCA1* or *BRCA2* pathogenic variant have up to a 60–80% lifetime risk of breast cancer whereas the risk for an average woman is about 13%. The lifetime risk of ovarian cancer is up to 40% with *BRCA1* and 20% with *BRCA2*, whereas the risk for an average woman is about 1-2%.

Men can also be affected. *BRCA2* carriers have an increased risk of male breast cancer (up to 8%) and prostate cancer (up to 25%).

Having a pathogenic variant does not mean that you will definitely develop cancer. Knowing about it allows you and your family to access personalised screening and risk-reducing options.

How is HBOC inherited?

HBOC can be passed from a parent to a child. Each child of a person carrying a pathogenic variant has a 1 in 2 (50%) chance of inheriting the same variant. Both women and men can inherit and pass on HBOC-associated genetic changes.

What causes HBOC?

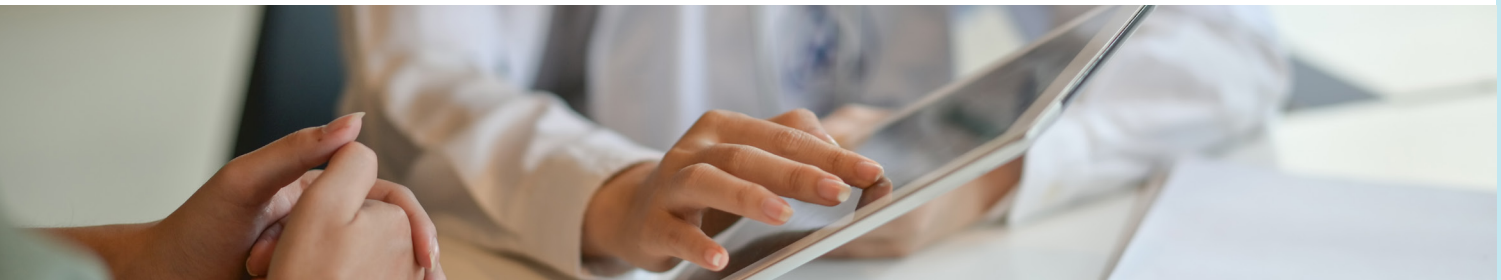
Most cases of HBOC are caused by pathogenic variants in the *BRCA1* and *BRCA2* genes. However, pathogenic variants in other genes can also increase the risk of breast and/or ovarian cancer. For breast cancer, these include *PALB2, CHEK2, ATM, BARD1, RAD51C, RAD51D, PTEN,* and *TP53*.

For ovarian cancer, these include *RAD51C, RAD51D, BRIP1*, and the genes related to Lynch syndrome (such as *MSH2, MSH6, MLH1,* and *PMS2*).

What are the surveillance options?

Surveillance depends on the gene involved and your personal and family history of cancer.

- *BRCA1*, *BRCA2* and *PALB2*: Breast screening usually starts at age 25–30. Preventive removal of both breasts can reduce breast cancer risk by more than 90%.
- In some EU countries, men with a *BRCA2* pathogenic variant are offered annual PSA (Prostate-Specific Antigen) measurements from the age of 40–45 years.
- Ovarian cancer: risk of ovarian cancer depends on the gene involved, family history and other risk factors. A specialized gynaecologic oncologist can inform you which preventive measures apply to you and the age to perform preventive surgery. There is currently no screening test proven to reliably detect ovarian cancer early or reduce deaths from the disease. Preventive removal of the ovaries and fallopian tubes (the tubes that connect the ovaries to the uterus) is the most effective way to reduce ovarian cancer risk, by about 90%.
- Other genes: Breast screening usually starts around age 40, or earlier depending on your family history and other risk factors.



How this diagnosis affects you?

Receiving a genetic diagnosis can raise many questions and requires ongoing medical follow-up and support. Here are some key points to consider:

How will Hereditary Breast and Ovarian Cancer Syndrome impact your health?

- The condition varies from person to person; regular medical follow up is important.
- You may need regular clinical examinations, MRI scans and mammograms over time.
- Since HBOC is genetic, family members should also consider genetic counselling and testing.

What medical steps should I take?

- **Step 1:** schedule an appointment with an expert centre.
- **Step 2:** discuss genetic testing and develop a personalised surveillance plan with your doctor.
- **Step 3:** discuss options for relatives, who may also benefit from genetic conselling and testing.

ERN GENTURIS provides information and resources to help you to understand this condition and guide your medical team toward the best possible treatment

More information on **HBOC syndrome**: