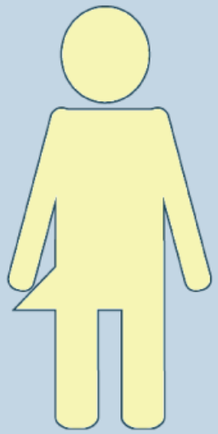
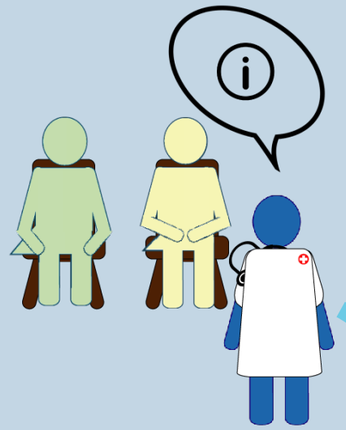
ERN GENTURIS (adult) patient journey: Hereditary Breast and Ovarian Cancer syndrome (HBOC)

Two genes are associated with the majority of HBOC families: *BRCA1* and *BRCA2*.
HBOC is characterised by an increased risk of breast, ovarian and pancreatic cancers in women and breast, prostate and pancreatic cancers in men.

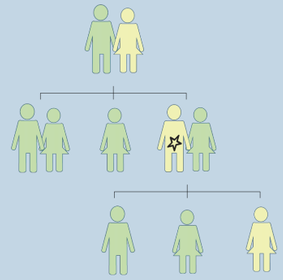
Affected person with unknown genetic status



Genetic counselling



Cascade testing (testing of blood relatives)

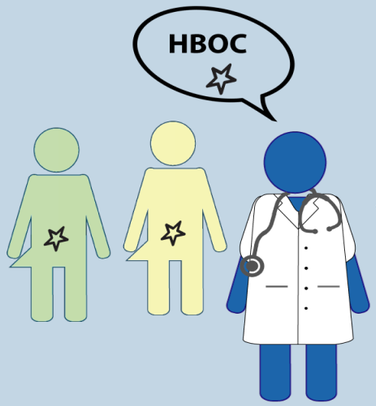


Surveillance

Eligible for germline genetic testing

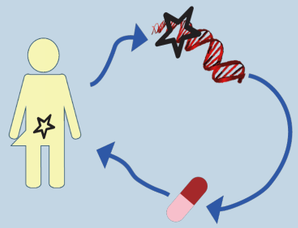
Germline genetic testing

Disclosure of the results

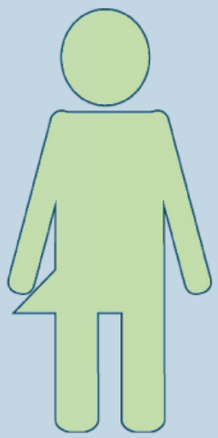


Cancer Diagnosis

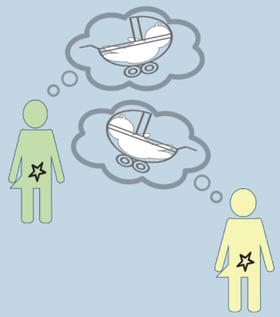
Targeted therapy



Unaffected person with unknown genetic status

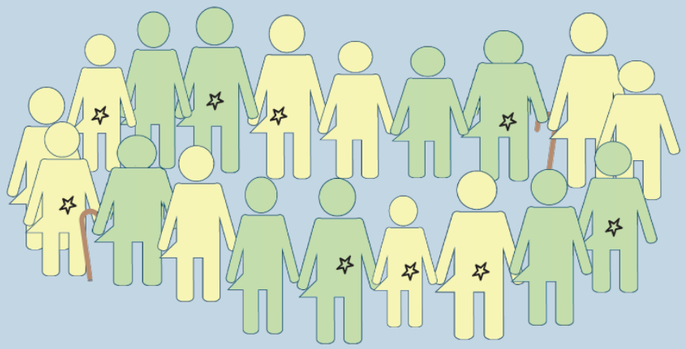


Reproductive decision-making



Risk reduction

Peer support and emotional support



European Reference Network

Genetic Tumour Risk Syndromes (ERN GENTURIS)

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Disclaimer: This patient journey is intended as a general overview of the clinical and diagnostic pathway for HBOC or its suspicion. It should not replace an individual clinical and genetic counselling at an expertise centre. It also does not intend to address all specific challenges of the complex condition of HBOC. Specific clinical guidelines, diagnostic criteria and nomenclature may change at short notice and therefore are only referred to in this patient journey.

Affected person with unknown genetic status

A person is already diagnosed with breast or ovarian (woman) or prostate (man) or pancreatic cancer but does not know their genetic status.

Unaffected person with unknown genetic status

A person is unaffected and does not know their genetic status.

Eligible for germline genetic testing

Eligibility criteria for germline genetic testing for HBOC in affected persons are often country-based according to:
- age at diagnosis
- family history
- hormone receptor and HER2 status, in particular triple negative breast cancer.

All patients with invasive epithelial ovarian cancer should be offered germline genetic testing.

Germline genetic testing should be performed as close as possible to the breast and/or ovarian cancer diagnosis (to be able to impact in surgical and oncological decision-making).

HBOC should be ruled out in an unaffected person if:
- a pathogenic variant in an HBOC gene within a family has been identified
- there is a strong family history and no option to test an affected family member.

Genetic counselling

Persons eligible for germline genetic testing and their family need thorough information before and after germline genetic testing regarding:
- clinical manifestations and natural history of HBOC
- process of genetic counselling and implications of germline genetic testing
- possible results of germline genetic testing
- legal, social, insurance and financial aspects of genetic results
- surveillance and risk reduction options
- reproductive decision-making
- emotional support including peer support: www.genturis.eu, section patient-area.

Germline genetic testing and disclosure of the results

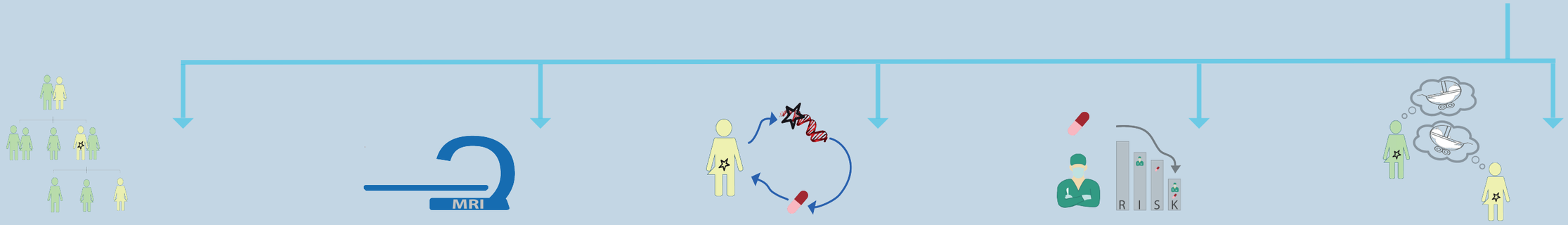
Individuals can find general information regarding germline genetic testing here:

https://www.coe.int/en/web/bioethics/information-brochure-on-genetic-tests-for-health-purposes

Germline genetic testing should at least consider HBOC-associated genes.

Rapid turnaround of germline genetic testing is recommended as diagnosis of HBOC could impact surgical and oncological decision making in a cancer patient. The turnaround of germline genetic testing should preferably be within 1 – 2 months, depending on urgency of medical implication.

Genetic counselling should accompany disclosure of the testing results.



Cascade testing (testing of blood relatives)

Cascade testing is the process of performing genetic counselling and germline genetic testing of blood relatives at risk for inheriting HBOC-related genes. The geneticist will determine for which family members germline genetic testing would be relevant and invite them for genetic counselling or provide a family letter that can be distributed by the index patient. **Testing of these family members and appropriate follow-up may save lives and improve quality of life.**
First degree relatives of carriers have 50% chance of inheriting the HBOC-associated gene (regardless of gender).
Second degree relatives have 25% chance of inheriting the HBOC-associated gene.
Note, pathogenic variants can be inherited from the mother as well as from the father.
Family members who decide to get germline genetic testing will be offered a test to look for the specific gene alteration present in the family.

Surveillance

There are specific surveillance programs for carriers of a pathogenic variant in an HBOC gene. These might differ between countries. Patients should ask their physicians or contact expert centres about recommendations in their country. Current recommendations regarding image-based surveillance at European level can be found on the ERN GENTURIS website (www.genturis.eu).

Patient-centric surveillance should be promoted by facilitating all check-ups on the same day.

Targeted therapy

For some hereditary cancers there are specific treatments in place (surgical or systemic therapies).

Oncologists should be aware of the possible impact of HBOC diagnoses on cancer treatment in patients with HBOC or contact expert centres regarding information on treatment.

The availability of targeted therapies might vary per country. Discuss with your physician if your genetics are suitable to consider targeted, surgical, or platinum-based therapies. This could also be relevant for other cancers in other organs than breast and ovaries, e.g. prostate or pancreatic cancer.

Risk reduction

In HBOC, risk-reducing surgery (like salpingo-oophorectomy or mastectomy) can be an option to reduce cancer risk. Patient's preference is key for an informed decision. Patients should ask their physicians about recommendations in their country. Current recommendations at European level can be found on the ERN GENTURIS website -care pathways (www.genturis.eu).

It is important to consider the management of side-effects after risk-reducing surgeries.

Reproductive decision-making

HBOC patients and carriers can pass on the pathogenic variant to their children. Even if a child is a HBOC carrier, the child is healthy at birth with regards to cancer. At adult age, an HBOC carrier has an increased risk of developing breast, ovarian, prostate or pancreatic cancer.

A referral for family planning and reproductive counselling should be given early in the patient journey.

There might be options for cryopreservation, prenatal testing and preimplantation genetic diagnostics in your country.

Financial resources for reproductive options in the public healthcare system and the legal framework differs per country.

Peer support and emotional support

Patients and their family might seek support repeatedly, at different timepoints. Psychological support by an expert for any HBOC-associated issue, if necessary, at any time of the patient's journey should be offered:
- -how to decide about genetic testing
- -how to inform family members on their genetic risk
- -in ongoing therapy
- -after being diagnosed with a pathogenic variant
- -how to cope with elevated cancer risk
- -how to deal with social consequences such as mortgage

Support can also be obtained via peers. Patient organisation can be found on:
https:/://www.genturis.eu/l=eng/patient-area/patient-associations.html

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