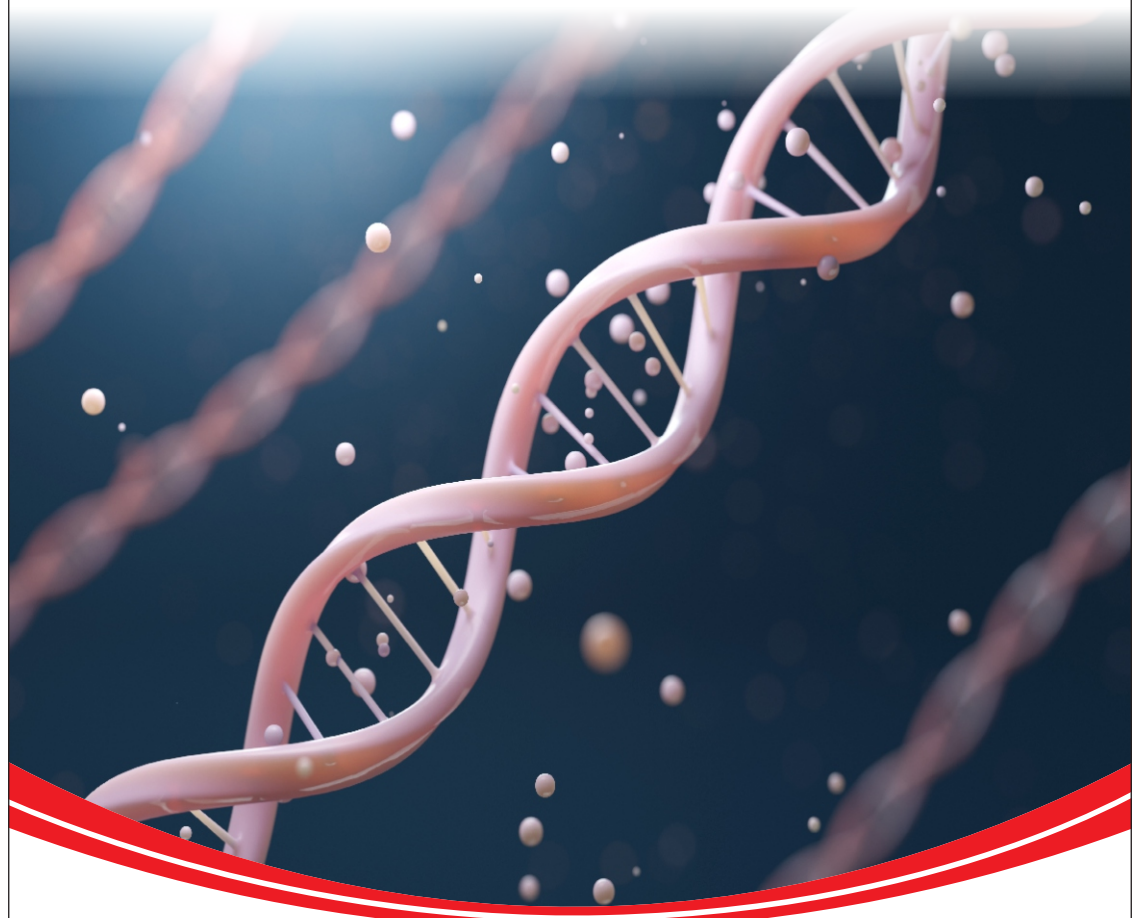
آغا خان یونیورسٹی ہسپتال
The Aga Khan University Hospital

Genetic Risk Factors for Hereditary Breast Cancer

Information for Patients/Guardians



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Approximately 1 in 8 women face the risk of developing breast cancer during their lifetime.

- Most individuals (60-85%) diagnosed with breast cancer have no identifiable genetic risk factors and are considered to have “sporadic” cancer.
- Around 10-30 % may have a family history of cancer.
- Only 5-10% have an inherited genetic mutation that can be identified by genetic testing. BRCA1 and BRCA2 are the most well-known genes linked to hereditary breast cancer, but as our understanding of genetics improves, we are discovering more genes that contribute to this risk.

Does it make a difference if I have sporadic, familial, or hereditary breast cancer?

Yes, your immediate treatment and follow-up plan, as well as the plan for some of your family members, may change if you are found to have hereditary breast cancer.

What factors increase my risk of hereditary breast cancer?

If you have been diagnosed with breast cancer before age 45 or have a “triple negative” breast cancer before age 60, you may have a higher chance of carrying a genetic mutation. The following factors may also increase your risk of hereditary breast cancer, even if you have not been diagnosed:

1. A close relative on either your mother's or father's side (including grandmothers, mother, aunts, sisters) with breast cancer who has also been found to have an abnormal gene on a genetic test.
2. A close relative who was diagnosed with breast cancer before age 45.
3. Multiple cancers include breast, ovarian, pancreatic, uterine, colorectal, or metastatic prostate cancer on one side of the family.
4. Multiple close relatives on either side are diagnosed with breast cancer.
5. A close relative with multiple cancers such as breast and ovarian.
6. A close male relative with breast cancer.

How do I find out if I have hereditary breast cancer?

- If you are considered at risk, you will be referred to a geneticist or genetic counsellor for a pre-test genetic counselling session. This session will involve discussing your medical and family history in detail to determine if you are a suitable candidate for genetic testing.

- If testing is offered and you agree to pursue the test, a blood sample will need to be given at the AKUH lab. The results of the test are reported in 2-3 weeks.
- The results will be discussed with you in a post-test counselling session by your geneticist or genetic counsellor. You will receive your genetic test results and be advised on how the results may impact your breast cancer management if you are affected. If you do not have breast cancer but underwent testing due to a high-risk family history, your test results may guide future surgical or preventive options and surveillance.

What can the genetic test results show?

The genetic test results may have one or more of the following three possibilities:

1. **Negative Result:** No abnormality or change in the genes.
2. **Positive Result:** A change or abnormality in a gene has been identified that is known to increase the risk of cancer.
3. **Variant of Uncertain Significance (VUS):** A change has been identified, but it is unclear whether it increases the risk of cancer. A VUS result may not require a change in your treatment plan. However, it's important to follow up with your genetic counsellor every year, as the interpretation of this result might change in the future.

How will a genetic test help me and my family?

- If there is a genetic cause to your breast cancer, it can help us develop a more personalised treatment plan for your current condition and guide preventive strategies for the future.
- This may include measures to detect cancers as early as possible or reduce the risk of other cancers that may be associated with the particular genetic change.
- If you are found to have a genetic change but have no personal history of cancer, you can take appropriate measures to reduce your risk of developing cancer or have closer monitoring for future cancers.
- Your test results may even have important implications for your immediate family and close relatives. Timely identification of these family members through genetic testing can allow them to take preventive measures, such as opting for risk-reducing surgery and/or a tailored surveillance plan.

The Aga Khan University Hospital

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