

BRCA TESTING

Germline mutations are mutations that occur in every cell in your body and have the possibility to be passed down to your children. Germline variants in *BRCA1* or *BRCA2* are inherited in an autosomal dominant manner. The offspring of an individual identified as having a *BRCA1* or *BRCA2* germline pathogenic variant have a 50% chance of inheriting the variant from that parent.



What is *BRCA1* & *BRCA2* Testing?

- Around 5 – 10 % of breast cancers result from a mutation in the *BRCA1* and *BRCA2* genes. *BRCA* mutations increase the risk of developing breast and ovarian cancer, and patients with *BRCA* mutations tend to develop breast cancers at a younger age.
- Mutations in the *BRCA* genes can be sporadic, but they are often germline, meaning they are in all cells in your body and may be passed down to your children. Testing can be performed on a buccal swab sample which may indicate if there is a germline mutation in the *BRCA* gene. If the mutation is germline, family members can be tested to see if they have inherited the mutation, allowing earlier detection and prevention.

Why is it important to have genomic testing if I have a family history of cancer?

Testing for Inherited cancer syndromes informs clinical decision making and may assist in the prevention of adverse health outcomes. *BRCA1* and *BRCA2* are part of complexes involved in DNA repair using homologous recombination. Women with an abnormal *BRCA1* or *BRCA2* gene have up to an 80% risk of developing breast cancer by age 90 and up to 55% risk of developing ovarian cancer. In addition to breast cancer, mutations in the *BRCA1/2* gene also increase the risk of ovarian, fallopian tube, pancreatic cancer, gastric cancer and prostate cancers.

If there is a family history of breast cancer, other members of your family can undergo genomics testing to see if they are a carrier of germline *BRCA* mutations. If they are identified as a carrier of the mutations, this can enable them to:

- Start cancer screening tests earlier
- Get screened for that type of cancer more often
- Get screening tests that are used only for people known to be at increased cancer risk
- Watch yourself closely for signs or symptoms of that kind of cancer
- Learn about options to help reduce the risk of certain types of cancer, such as drugs or surgery

How do I organise Testing?

