What can my family do with this information?

Inherited pathogenic variants are passed down from parent to child, which means if you have an identified pathogenic variant you may pass it onto your children. It also means that you received it from one of your parents, and that your siblings may also be carrying the same variant. **Identifying at-risk family members is one of the most important benefits of genetic testing, as it gives those who test positive the opportunity to make informed decisions on prevention strategies.** For those who test negative it can bring peace of mind.

Family members wishing to know their risk can be tested for the specific pathogenic variant you are carrying.

Genomic testing with Genomic Diagnostics

Genomic Diagnostics has several hereditary breast and ovarian cancer testing options, with results available within 4 weeks. We are also able to provide you with access to qualified genetic counsellors with expertise in counselling patients and families with inherited cancer.

Availability and cost of each of our testing options depends on whether your testing is being requested by a medical specialist or GP, and whether you are eligible for testing covered by Medicare.

1. BRAoVO™ Gene Panel: Testing for BRCA1, BRCA2 plus 11 additional genes (specialist or GP request*)
2. BRAoVO™ Plus Gene Panel: Testing for BRCA1, BRCA2 plus 16 additional genes (specialist request only)
3. Testing for BRCA1 & BRCA2 only (specialist request only)
4. Testing for familial variants (specialist request only)

All genes tested by Genomic Diagnostics have professional medical management guidelines to help your healthcare team develop the best medical care plan for your particular circumstances.

*Testing requested by GPs is only available with mandatory pre- and post-test genetic counselling and is not covered by Medicare.

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Understanding Hereditary Breast & Ovarian Cancer

Patient Guide

Hereditary Breast and Ovarian Cancer

Most cases of breast and ovarian cancer happen by chance. In some situations, however, individuals can have harmful changes (called pathogenic variants) in specific genes that convey an increased risk of developing breast (in both men and women) and ovarian cancers. These pathogenic variants are inherited (passed from parent to child) and can be detected by genomic testing.

Approximately 5 – 10% of breast cancers and at least 20% of ovarian cancers are due to inherited pathogenic variants. BRCA1 and BRCA2 are the most common genes associated with increased risk of breast and ovarian cancers, although there are additional high and moderate high risk genes associated with an increased lifetime risk of these and other cancers, such as prostate and pancreatic cancer.

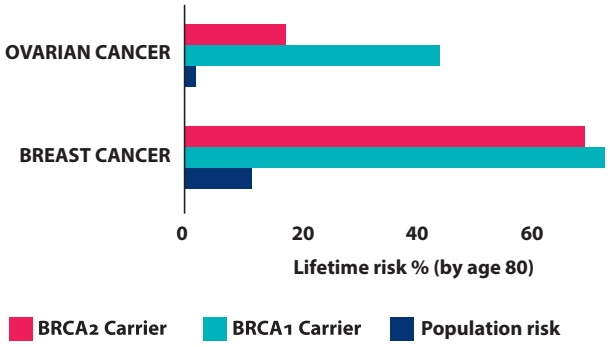

Figure 1: Lifetime risk of breast and ovarian cancer for BRCA gene carriers (adapted from eviQ.org.au)

What are the benefits of genomic testing?

Genomic testing in breast and ovarian cancer can be clinically useful in two main settings. Diagnostic testing is performed for individuals with a diagnosis or personal history of cancer, and predictive testing is performed in unaffected individuals to determine the future risk of cancer.

Awareness of inherited cancer risk can alter medical care for individuals and their families.

Detection of pathogenic variants in genes causing hereditary breast and ovarian cancer can help in the following ways:

- Provide specific information on prognosis and lifetime risk of cancer
- Direct the use of specific treatments
- Direct more frequent screening and consideration of prophylactic risk-reducing surgery, for example mastectomy, and medications
- Guide testing of at-risk family members
- Assist couples with reproductive decision-making
- Provide an explanation for personal or family history of cancer.

Who should consider genomic testing?

Not everyone with a personal or family history of breast and ovarian cancer will have an inherited pathogenic variant. There are clues, such as clinical features, tumour characteristics, ethnic background and family history that can help to identify individuals that may benefit from genomic testing.

Current Australian guidelines (https://www.eviq.org.au/cancer-genetics/) cover situations where genomic testing for hereditary breast and ovarian cancer is recommended. Your healthcare provider can help you understand your personal and family history and whether you might be a candidate for testing.

Genetic counselling

Genetic counselling is of benefit for all patients undergoing genomic testing for inherited pathogenic variants associated with cancer. It involves discussing benefits, limitations and the possible consequences of the genomic testing to be performed. Genetic counselling can be provided by your referring medical specialist or a qualified genetic counsellor and must be undertaken before your test is performed.

If you are found to have a pathogenic variant, you should also have post-test genetic counselling to fully understand what the results mean for you as well as any implications for family members.

Understanding your results

Once genomic testing has been completed, your medical specialist or genetic counsellor will receive a report describing what was found so that they can discuss it with you. Possible findings include:

Pathogenic variant found

Also referred to as a 'positive' result, this means that a gene change was identified that is known to increase your cancer risk. If you don't have a personal history of cancer it does not mean that you will definitely develop cancer. If you already have a cancer diagnosis, then it may influence your medical management. Information about the pathogenic variant should also be shared with your relatives who may want to be tested to understand their own cancer risk.

No pathogenic variants found

Also referred to as a 'negative' result, this indicates that you do not have a harmful change in any of the genes that were tested. It does not mean you will not get cancer, as there are many other factors that contribute to cancer risk. It also does not rule out pathogenic variants in other genes for which you have not been tested.

Variant of unknown significance (VUS) found

A gene change was identified however its impact on hereditary cancer risk is not yet understood. This is a common finding and does not change your ongoing medical care. Testing for this variant in unaffected at-risk relatives is not recommended.