Cancer Genetic Testing (BRCA1 & BRCA2) in a primary care setting

Genes can have variations (also known as mutations) that increase the risk of certain cancers. The most well-known and frequent of these for breast cancer are the genes BRCA1 and BRCA2[2].

Increased public awareness and the new Medicare rebate rules have resulted in increased enquiries from patients about these tests.

What testing options are available:

In Australia, subsidised testing for cancer genetic testing is available through private specialists for patients meeting Medicare eligibility criteria, or in state hospital familial cancer clinics. The Medicare rebate is available to breast and ovarian cancer patients who have a proven high family risk, and testing can only be ordered by a specialist. The new rebates also allow any relative of a patient with a known pathogenic variant (mutation) to access testing for that specific variant.

As can be seen from the MBS descriptor below the key requirements for patient access are:

1. They must have diagnosed Breast or Ovarian cancer
2. The test must be ordered by a specialist or consultant physician and
3. The patient must have a greater than 10% familial risk as calculated using a quantitative algorithm.

MBS Item Descriptor – 73296

Characterisation of germline gene mutations, requested by a specialist or consultant physician, including copy number variation in BRCA1 and BRCA2 genes and one or more of the following genes STK11, PTEN, CDH1, PALB2 or TP53 in a patient with breast or ovarian cancer for whom clinical and family history criteria, as assessed by the specialist or consultant physician who requests the service using a quantitative algorithm, place the patient at >10% risk of having a pathogenic mutation identified in one or more of the genes specified above.

For family members of a patient with a previously identified pathogenic cancer risk variant (mutation), the key elements for access are:

1. The patient must have a biological relative with an identified pathogenic DNA variant in one of the listed genes
2. The test can be ordered only by a specialist or consultant physician

MBS Item Descriptor – 73297

Characterisation of germline gene mutations, requested by a specialist or consultant physician, including copy number variation in BRCA1 and BRCA2 genes and one or more of the following genes STK11, PTEN, CDH1, PALB2 or TP53 in a patient who is a biological relative of a patient who has had a pathogenic mutation identified in one or more of the genes specified above, and has not previously recieved a service under item 73296.

In public hospital funded familiar cancer clinics, many patients may not qualify for free access due to strict testing eligibility criteria. Recent reports have also shown that current familial risk assessments to determine test access will exclude some patients with pathogenic variants[3].

Genomic Diagnostics provides testing for Medicare rebated tests ordered by specialists but we also provide access to rapid turnaround (3-4 weeks) testing for private fee-paying patients who may not meet these strict eligibility criteria. The two recommended tests for genomic testing relevant to Breast or Ovarian cancer are shown below. General Practitioners should seek specialist advice on which option is most suitable for their patient.

Panel Test	Genes Tested
Comprehensive BRCA1 and BRCA2	BRCA1 and BRCA2 including deletion/duplications
BRaOVO	ATM, BRCA1, BRCA2, CHEK2, CDH1, PALB2, PTEN, STK11 and TP53 including deletion/ duplications

Table 1: Cancer risk panel tests from Genomic Diagnostics

Dorevitch PATHOLOGY

The role of genetic counselling:

Genetic counselling is essential when testing for cancer risk genes. It is important that patients have a clear understanding of the clinical implications of the results and any limitations of the test. Additionally, the implications for insurance and for other family members should also be discussed prior to testing.

Genomic Diagnostics requires that prior to testing from a referral, we have confirmation that the patient has had appropriate genetic counselling. In **Table 2** we have listed a range of private genetic counselling services in Australia that are able to provide counselling to facilitate this process.

Service	Location	Face to Face consults	Tele-Health consults	Website	Contact
Genetic Counselling Australia	National	No	Yes	geneticcounsellingaustralia.com.au	1300 100 422
Sydney Cancer Genetics	Sydney	Yes	Yes	sydneycancergenetics.com.au	02 9304 0438
The Genetic Clinic	Sydney	Yes	No	thegeneticclinic.com.au	0402 083 119
Canberra Genetic Counselling	Canberra	Yes	Yes	cwhealth.com.au/genetic- counselling	02 6162 0582
Dr Manju Salaria	Melbourne	Yes	Yes	Werribee & Clayton consulting rooms	03 9908 2998 or 03 9013 9795
SilesHealth	Melbourne	Yes	Yes	sileshealth.com.au	03 9328 4403
Brisbane Genetics	Brisbane	Yes	Yes	brisbanegenetics.com.au	07 3217 8244
Medical Genetics Australia	Brisbane/ Gold Coast	Yes	Yes	medgen.com.au	1300 651 741
WOMEN Centre	Perth	Yes	Yes	womencentre.com.au	08 9468 5188
Adelaide Oncology	Adelaide	Yes	Yes	aoah.com.au	08 8100 5333
Wellend Health	Adelaide	Yes	Yes	wellendhealth.com	1300 652 028

Table 2: Private Genetic Counselling Services in Australia

It is also possible to access publicly funded services for genetic counselling based on access criteria. A comprehensive list of public services across Australia is maintained by NSW Health and can be found at: www.genetics.edu.au/genetic-services/general-genetics-clinics.

Genomic Diagnostics Testing Process:

1. Patient sees General Practitioner and is interested in getting cancer risk testing performed.
2. Patient is referred to genetic counselling service for pre-test counselling.
3. Patient confirms interest in testing post counselling – referrer completes request and consent forms. These are available on request or from our website.
4. Patient calls 1800 822 999 to pay for testing, attends pathology collection site and sample is collected.
5. Results are returned to clinician through their preferred method.

About Genomic Diagnostics

Genomic Diagnostics is the genomic testing arm of SDS Pathology representing Laverty Pathology in NSW, Dorevitch Pathology in Victoria, QML in Queensland, TML in Tasmania, Western Diagnostic Pathology in Western Australia and Abbott Pathology in South Australia. We specialise in genetic testing and with 20 years experience in inherited cancer analysis we are your reliable, local partner*.

*Genomic Diagnostics does not share or sell de-identified information to third parties[4]

References:

1. Romero-Laorden, N & Castro, E. Inherited mutations in DNA repair genes and cancer risk. Curr Probl Cancer. 2017; 41:251-264
2. Petrucelli, N & Feldman, G. Hereditary breast and ovarian cancer due to mutations in BRCA1 and BRCA2. Genetics in Medicine. 2010; 12:245-259
3. Grinedal, E. et.al. Current guidelines for BRCA testing of breast cancer patients are insufficient to detect all mutation carriers. BMC Cancer. 2017; 17:438
4. www.genomicdiagnostics.com.au/privacy/

Dorevitch PATHOLOGY

Genomic Diagnostics

Specialist Diagnostic Services Pty Ltd ABN 84 007 190 043 APA trading as Dorevitch Pathology DP0862 29.03.18