Testing of the genes responsible for familial breast or ovarian cancer is now rebated. Medicare rebates will improve access to genetic testing and it is important to understand the new criteria in order to best support your patients.

**Key points**

- BRCA1 and BRCA2 are the genes most commonly implicated, but familial mutations in other genes such as STK11, PTEN, CDH1, PALB2, and TP53 can also cause familial breast cancer. Mutations in any of these genes can also cause cancers other than breast and ovarian cancers in both men and women.
- There are a number of settings in which rebated testing of BRCA and other genes responsible for familial breast and ovarian cancer is available;
  - Testing an individual with advanced ovarian cancer
  - Testing an individual with breast or ovarian cancer and relevant family history
  - Testing an individual who does not have cancer but has family history
- Specific clinical requirements apply for Medicare eligibility.
- Medicare stipulates that requests for such testing can only be made by a medical specialist or consultant physician. General practitioners cannot order rebated BRCA gene testing.
- Testing can be performed for patients who do not meet the Medicare requirements, however BRCA gene testing will be privately billed. Sonic Genetics still requires that privately billed tests are requested by a medical specialist or consultant physician and that pre-test counselling with written consent has been completed.

**Testing an individual with advanced ovarian cancer**

Women with invasive ovarian cancer usually commence a platinum-containing chemotherapy drug. Most patients subsequently relapse. Of the women who relapse, approximately 15% have a familial mutation in the BRCA1 or BRCA2 gene. These women often respond to olaparib, a specific drug listed on the Pharmaceutical Benefits Scheme (PBS) for BRCA-related ovarian cancer. Olaparib is limited to women with evidence of a BRCA mutation.

**MBS Item 73295**

Detection of germline BRCA1 or BRCA2 gene mutations, in a patient with platinum-sensitive relapsed ovarian, fallopian tube or primary peritoneal cancer with high grade serous features or a high grade serous component, and who has responded to subsequent platinum-based chemotherapy, requested by a specialist or consultant physician, to determine whether the eligibility criteria for olaparib under the Pharmaceutical Benefits Scheme (PBS) are fulfilled.

This test should be requested by the specialist or consultant physician responsible for managing the patient’s chemotherapy. **GPs should refer a patient requesting such testing to the appropriate specialist.**

The primary purpose of this testing is to determine the choice of therapy for the patient being tested. Nonetheless, the identification of a familial mutation in a BRCA gene carries significant implications for both male and female relatives. For this reason, genetic counselling is strongly recommended for patients having this test.

The information in this leaflet may change as new decisions are made regarding testing and therapies. Please contact the laboratory for updated information or refer to the Medicare Benefits Schedule (MBS).
Testing an individual with breast or ovarian cancer and relevant family history

A proportion of patients with breast or ovarian cancer have a familial predisposition to develop these cancers. Overall, approximately 5% of patients with breast cancer and 15% of those with ovarian cancer have an inherited mutation in one of the BRCA genes. The probability that a patient has a mutation in one of the genes associated with familial cancer varies with the age and gender of the patient, the type of cancer, and the family history of cancers among relatives. Medicare rebates testing where patients have >10% risk of an identifiable mutation. Computerised tools and guidelines are available to assist clinicians to make an accurate assessment.

MBS Item 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.

The identification of such a mutation can provide important information to guide the current therapy of the patient, their risk of cancer in the future, and the risk of cancer among relatives. GPs should refer a patient requesting such testing to an appropriate specialist. Preliminary advice can be obtained through the links provided below.

Non-rebated testing of breast cancer genes

The requirements listed in this leaflet are stipulated by Medicare for rebated testing. Non-rebated testing is available for patients who are willing to cover the full cost of the test themselves. It is important that such patients seek advice from an experienced clinician to ensure that the test will be worthwhile.

Sonic Genetics will only accept requests for non-rebated testing of breast or ovarian cancer genes from a medical specialist or consultant physician, and requires signed confirmation that pre-test genetic counselling and written informed consent have been completed. The restriction on who can request the test applies even if the patient is willing to pay privately. This is because it is a complex test, with complex results and significant ramifications that require specialist follow-up.

Testing an individual who does not have cancer but has family history

If a mutation responsible for familial breast or ovarian cancer has been identified in the family, unaffected relatives may seek genetic testing to clarify their risk of developing cancer. The mutated gene is usually inherited in an autosomal dominant fashion i.e. there is a 50% chance of the mutation being present in a parent, sibling, or child of the person with the mutation. It is important to note that the risk of cancer is less than the risk of having the mutation, so some people with the mutation will not develop cancer.

MBS Item 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 received a service under item 73296.

This test provides valuable information regarding the person’s risk of cancer and whether strategies that could mitigate that risk are warranted. GPs should refer a patient requesting such testing to an appropriate specialist.

Pre-test genetic counselling to address the medical, psychological, financial and social implications of the test is recommended by international authorities and mandated by Australian regulators.

It is important to note that such testing is neither recommended nor rebated for people who do not have a documented mutation in their family. The presence of a family history of cancer does not, of itself, justify genetic testing. Patients who are concerned about their personal risk of developing cancer should be referred to a clinical geneticist, medical oncologist, or other medical specialist. Preliminary advice can be obtained through the links provided below.

For further information, including scientific and peer-reviewed publications, please refer to our website, www.sonicgenetics.com.au or call us on 1800 010 447

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• Cancer Australia: Tool for assessing risk of familial breast/ovarian cancer www.canceraustralia.gov.au (Keywords ‘Familial risk assessment FRA-BOC’)
• eviQ: Referral guidelines for breast cancer risk assessment and consideration of genetic testing www.eviq.org.au (Keywords ‘Breast risk guidelines’)
• Sonic Genetics: www.sonicgenetics.com.au (Keywords ‘BRCA’)

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