Information for non-genetic specialists considering ordering diagnostic *BRCA1* and *BRCA2* gene testing

Who can order the test?

Diagnostic *BRCA1* and *BRCA2* gene testing for cancer-affected patients can be ordered by their treating specialist. General practitioners cannot order Medicare rebated *BRCA* gene testing.

It is the recommendation of Genetic Health Queensland (GHQ) that specialists who do not feel comfortable discussing genetic testing with their patients and interpreting genetic test results access further education prior to ordering *BRCA* gene testing.

Health professionals involved in the care of cancer patients can register for online training modules at: www.mainstreamgenetictesting.com.au.

Genetic Health Queensland (GHQ) can be contacted to discuss alternative training options.

Predictive *BRCA* gene testing (i.e. testing for a pathogenic variant* already identified in the patient's family) should be arranged by a genetic or family cancer service.

*For the purposes of this document a "pathogenic variant" refers to a class 5 pathogenic variant or a class 4 likely pathogenic variant. These variants are referred to as "mutations" in some pathology reports and communications from family cancer and cancer genetic services.

Who pays for the test?

Medicare

A patient may be eligible for *BRCA1* and *BRCA2* gene testing under one of the following Medicare item numbers: 73295, 73296 and 73297. More information about MBS items can be found on the MBS Online webpage at: www.mbsonline.gov.au.

Which patients are eligible under MBS item numbers 73295, 73296 and 73297?

Item number 73295

Women diagnosed with relapsed platinum-sensitive high-grade serous ovarian, fallopian tube or primary peritoneal cancer who have responded to subsequent platinum-based chemotherapy.

Item Number 73296

An individual diagnosed with breast or ovarian cancer who has a ≥ 10% likelihood of having a pathogenic mutation in *BRCA1* or *BRCA2*. This likelihood must be calculated using a validated algorithm. Accepted algorithms are listed as weblinks below:

- Manchester Score
- BRCAPro
- BOADICEA

NB: Of these options, the Manchester score is most easily adapted to a busy cancer outpatient setting.

Individuals who are eligible for testing under this item number may have testing of *BRCA1* or *BRCA2* only, or the item number can be used when *BRCA1* and *BRCA2* are tested as part of a gene panel containing one of more of the following additional genes; *STK11, PTEN, CDH1, PALB2* or *TP53*.

Item Number 73297

Individuals who are the biological relative of a patient who has had a pathogenic variant identified in *BRCA1* or *BRCA2* or one or more of the following genes; *STK11, PTEN, CDH1, PALB2* or *TP53* and has not previously received a service under item 73296.

Who can request *BRCA1* and *BRCA2* genetic testing using item numbers 73295, 73296 and 73297?

BRCA1 or *BRCA2* genetic testing under item numbers 73295, 73296 and 73297 must be requested by the patient's treating specialist or consultant physician. (This item number cannot be used by a general practitioner).

Which laboratories can be used for Medicare rebatable *BRCA* gene testing?

In order to be eligible for the Medicare rebate, testing needs to be performed by an Australian laboratory with NATA accreditation for both sequencing and copy number variant analysis.

Patients who do not qualify for the medicare rebate:

If a patient does not meet Medicare criteria for *BRCA* gene testing but fulfils eviQ criteria, publicly funded gene testing will be offered by GHQ to referred patients.

Patients referred to GHQ who do not fulfil either Medicare or eviQ criteria have the option of self-funded gene testing after genetic counselling.

If a patient does not meet Medicare criteria for *BRCA* gene testing and the test is ordered by a cancer specialist external to GHQ, the test will either be funded by the cancer service (pre-approval of the director of the service recommended) or the patient.

In some instances, *BRCA* gene testing may be available through research studies or industry sponsored programs. Please note that not all research studies use NATA accredited laboratories.

What does *BRCA1* and *BRCA2* genetic testing involve?

BRCA1 and *BRCA2* genetic testing is performed on a blood sample (2 x 4ml EDTA). If ordered through Pathology Queensland, this testing can be requested using a Pathology Queensland request form. The results of *BRCA1* and *BRCA2* genetic testing are typically available in 6-10 weeks. Once the result is available, it is returned to the requesting health professional. This health professional is then responsible for ensuring the result is returned to the patient. It is important that the ordering health professional is able to interpret the result and organise the necessary follow-up for the patient.

What resources are available for patients?

Resources are available to assist patients in making a decision regarding *BRCA1* and *BRCA2* genetic testing. Some of these resources are available through the NSW Centre for Genetics Education and include topics such as:

- Understanding genetic tests for breast and ovarian cancer that runs in the family
- Understanding genetic testing after a diagnosis of ovarian cancer
- Making a decision about treatment-focused genetic testing

For patients who are concerned about the implications of genetic testing on insurance policies further information can be found on this fact-sheet.

If a patient requires further discussion, or should they have questions outside the cancer service's area of expertise, a referral to GHQ is recommended.

Is consent required for *BRCA1* and *BRCA2* genetic testing?

It is important that there is documented informed consent, ideally written, prior to any genetic testing. Patients should be informed that the results of genetic testing may assist in deciding their current or future management. Additionally, they should be aware that the results of genetic testing can have implications for their biological relatives. It is important that patients understand that genetic testing is voluntary and that they can decline or defer testing. For patients requiring an interpreter for consent, a relative should **not** be used for this purpose. Potential outcomes of the testing should be discussed (see below).

GHQ recommends that a copy of the consent form be forwarded to the laboratory with the pathology request form as well as kept in the patient's medical record. If the test is not ordered through Pathology Queensland, the testing laboratory should be contacted for their requirements regarding consent.

What are the possible outcomes of *BRCA1* and *BRCA2* genetic testing?

There are three potential outcomes to genetic testing:

- No reportable variants identified
- A variant of uncertain significance (class 3 variant, sometimes called an unclassified variant) identified
- A pathogenic (class 5) or likely pathogenic (class 4) variant identified

It is very important that the ordering specialist is familiar with the potential outcomes of genetic testing and their implications for the patient and their family.

Panel gene testing

The ordering and interpretation of the results of multi-gene panels requires an additional level of expertise over and above that required for *BRCA1* and *BRCA2* gene testing. GHQ can be contacted with any questions regarding this.

Who should be referred to Genetic Health Queensland?

It is recommended that the following patients who have been offered gene testing by a non-cancer genetics specialist be referred:

- Requires genetic counselling beyond what is able to be provided by the cancer service
- A pathogenic or likely pathogenic variant identified
- A variant of uncertain significance identified
- No reportable variants identified but a significant family history of breast and/or ovarian cancer or, in the case of ovarian cancer patients, a family history of Lynch syndrome associated cancers (such as colorectal or endometrial cancer)
- A patient with loss of mismatch repair proteins on immunohistochemistry

In addition, any patient with a *BRCA* gene pathogenic or likely pathogenic variant identified on somatic tumour testing should be referred to GHQ.

GHQ will **not** automatically contact any patient who has not been referred to the service, irrespective of the outcome of their genetic testing.

For further information about referrals to Genetic Health Queensland please see the GHQ webpage at: www.health.qld.gov.au/ghq or the Metro North Hospital and Health Service refer your patient webpage.

Who can I contact?

Should you have any questions or concerns you can contact Genetic Health Queensland and request to speak with the on call Genetic Counsellor or Registrar. The contact details for Genetic Health Queensland can be found below:

Genetic Health Queensland

Level 6, Block 7

Royal Brisbane and Women's Hospital

Ph: (07) 3646 1686

Email: ghq@health.qld.gov.au