BRCA testing FREE for high risk patients

From 1st November, funded BRCA testing will be available for high risk patients without having to queue up in the public system or having to travel long distances to a public familial cancer clinic. This is great news!!

Two new MBS item numbers have been created: 73296 and 73297. This means that high risk patients can have genetic testing of BRCA1 and BRCA2 (as well as 5 other genes associated with breast cancer risk) and the test will be fully funded by Medicare.

So who qualifies?
In all cases, the test must be ordered by a specialist or consultant physician.

As for the previously approved item number 73295, "Appropriate genetic counselling should be provided to the patient either by the specialist treating practitioner, a genetic counselling service or a clinical geneticist on referral".

Item number 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."

Plain English Explanation for Patients: The item number 73297 allows funded predictive testing. That is, if a mutation has already been identified then a man or woman who is related by blood (ie a biological relative such as a parent, sibling or even cousin) can to have funded testing. Predictive testing is a "yes or no" kind of test: either the mutation (change in the DNA code that is known to increase cancer risk) is there or it's not.

Predictive testing is very important. If you do not carry the mutation responsible for the cancers in your family, you are usually regarded as being at average risk. That means you can follow population based screening. Also, if you do not carry the mutation you cannot pass it on to your children. This is very powerful information for people who have lost many loved ones to cancer!

If you do carry a mutation in a gene that increases cancer risk, there are many things you could choose to do to reduce that risk. For example, a woman who carries a BRCA mutation may elect to have bilateral mastectomies (reducing her risk to almost zero) or she may starting breast screening earlier, at age 30. In addition, she would have funded access to more sensitive screening modalities such as breast MRI.

Many people are aware that mutations in the BRCA genes increase breast cancer risk (that is where gene name comes from after all: BReast CAncer genes). However, they are less aware of the increased risk of ovarian cancer, prostate cancer and, in some families, pancreatic cancer. If you don't know you are at risk, you can't take steps to reduce or eliminate that risk.

We all carry 2 copies of our genes (with the exception of men, who only have one copy of the X chromosome, and so only one copy of those genes). We get one copy from our mother (via the egg) and one copy from our father (via the sperm). That means that there is a 50% chance of a person who carries a mutation, whether male or female, passing the mutation to their son or daughter. Some families, if they know they are carriers of a high risk mutation, may use preimplantation genetic diagnosis (PGD) in the setting of in vitro fertilisation (IVF) to prevent the mutation being passed on to future offspring.

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Hereditary Health and Hope

Item number 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."

Plain English Explanation for Patients: The item number 73296 applies to:

1. a man or woman who has had breast cancer or a woman who has had ovarian cancer where:
   - The breast cancer was "triple negative" and occurred at or before age 40.
   - The ovarian cancer was "invasive non mucinous" and occurred at or before 70 OR at any age if there's a family history of either breast or ovarian cancer
2. a patient comes from a population where a common founder mutation exists and they have a personal or family history of breast and/or ovarian cancer (eg Ashkenazi Jewish)
3. a man or woman who has had breast cancer or a woman who has had ovarian cancer where a validated risk prediction model (eg Manchester Score, BOADICEA, BRCAPRO) places the patient at greater than 10% risk of having a pathogenic (cancer causing) mutation.

If no mutation can be detected to explain the cancers in the individual or family, or if a genetic change of uncertain clinical significance called "a variant" is found, family members may still be at increased risk. That's why appropriate pre and post test genetic counselling is so important.

Which genes?

In addition to the BRCA1 and BRCA2 genes, the item number allows testing of 5 other genes known to increase breast cancer risk as well as certain other cancers. These genes are STK11, PTEN, CDH1, PALB2 and TP53 (See our website www.SydneyCancerGenetics.com.au for information about these genes and the cancer syndromes they are associated with)

Why now?

The discussion around greater access to funded testing has been going on for years, driven in part by long queues in the public system and in part by the falling costs of genetic testing making more widespread testing more economical (for a full discussion of the issues involved, see the public summary document submitted by the Royal College of Pathologist of Australasia and published by the Medical Services Advisory Committee, the body that creates Medicare item numbers:
http://www.msac.gov.au/internet/msac/publishing.nsf/content/D3E96917F7B2253BCA25801000123C2E/$File/PSD_1411.1.pdf)

What to do?

You can assess your patient's risk by having them complete a family history questionnaire (see http://www.sydneycancergenetics.com.au/services/) to discuss with you.

At Sydney Cancer Genetics we will access your patient's risk (including using the validated risk prediction models such as the Manchester Score, BOADICEA, BRCAPRO), provide genetic counselling and organise genetic testing (including self-funded testing if appropriate the patient is high risk but not eligible) as well as interpret the results and provide risk management advice. To make an appointment, you can send a referral to us by fax (02) 9304 0468 or email info@SydneyCancerGenetics.com.au or call us on (02) 9304 0438. (Yes, our numbers have changed, due to the NBN)

Sydney Cancer Genetics offers bulk billed testing throughout rural and regional Australia as well as face to face appointments in our rooms in Ultimo and Wahroonga. Dr High is a Genetic Oncologist. She can order the Medicare Funded test and also provide the appropriate genetic counselling. Alternatively, you could refer your patient to your nearest public familial cancer clinic (see www.SydneyCancerGenetics.com.au/links for a list).