

Genetic testing for the 'Breast Cancer Gene'

Patient Information Sheet – May 2013

Angelina Jolie's recent decision to undergo a prophylactic double mastectomy has created increased community interest in genetic testing for breast cancer. This information sheet addresses some of the more common questions associated with BRCA1 and BRCA2 testing, and provides links to useful websites.

What is the 'breast cancer gene'?

There are two genes, where inherited faults (also known as 'mutations') are linked to hereditary breast and/or ovarian cancer. These two genes are called BRCA1 and BRCA2.

How do you test for BRCA gene faults?

Testing for BRCA1 or 2 mutations involves a blood test. DNA is extracted from a blood sample and the BRCA1 and BRCA2 genes are examined. However, testing is only performed if a patient has seen a clinical geneticist and a genetic counsellor.

What is a genetic counsellor and why is it necessary to consult one?

A genetic counsellor is a specially trained professional who understands the complexities associated with genetic testing. Discovering that you have a gene that predisposes you to a particular medical condition can be an emotionally confronting experience, and genetic counsellors have in-depth knowledge and experience to determine whether you are a suitable candidate for genetic testing. They can also provide you with an understanding of what to expect and how to handle these uncertainties.

A genetic counsellor will take an extensive family history, discuss what the BRCA test involves and the likelihood of a BRCA1 or BRCA2 mutation. They will also discuss which family member would be the most appropriate to be tested initially, and give counselling about what the results might be and their implications. The genetic counsellor will also obtain patient consent for testing.

If a person has a BRCA1 or BRCA2 gene fault, will they automatically go on to develop breast or ovarian cancer?

No. A patient who tests positive for a mutation in BRCA1 or BRCA2 is at *greater risk* of developing these cancers, but a positive result does not automatically mean that a patient will develop breast and/or ovarian cancer. Likewise, a negative result for BRCA gene testing doesn't guarantee that a patient will not develop breast or ovarian cancer.

Statistically, how many women in Australia get breast cancer?

Between one in eight to one in eleven women (or approximately 9-12.5% of women) will be diagnosed with breast cancer during their lifetime.

What proportion of women with breast cancer also have a BRCA gene fault?

Only 1-5% of women with breast cancer have a mutation in one of the BRCA genes. However, the likelihood of a BRCA1 or 2 mutation increases if there is a family history of breast or ovarian cancer, and can be up to 20% or more if the family history is very extensive.

How is the BRCA1 and BRCA2 gene linked to ovarian cancer?

Mutations or 'faults' in BRCA1 or BRCA2 are linked to an increased risk of ovarian cancer, as well as breast cancer. If there is an extensive family history of breast or ovarian cancer, then a referral to a clinical geneticist should be considered.

Is BRCA testing covered by Medicare?

BRCA testing is not covered by Medicare, which means that there is no rebate. The cost to private patients is about \$1650.

Useful websites

1. *Online risk-assessment tool for doctors for familial breast and ovarian cancers (Cancer Australia)* <http://canceraustralia.gov.au/clinical-best-practice/gynaecological-cancers/familial-risk-assessment-fra-boc>
2. *What is family cancer? (Educational resource on genetic testing, NSW Health)* <http://www.genetics.edu.au/Information/What-is%20Family-Cancer>
3. *What is BRCA1? (The Conversation)* <http://theconversation.com/angelina-jolie-has-had-a-double-mastectomy-so-what-is-brca1-14227>
4. *My Medical Choice – Angelina Jolie's article in The New York Times* http://www.nytimes.com/2013/05/14/opinion/my-medical-choice.html?_r=3&
5. *Pink Hope – Website for high risk breast and ovarian cancer community* <http://pinkhope.org.au>