BRCA1 and BRCA2 Gene Testing

Patient Information Sheet (PIS-1)

This information sheet is for patients with cancer who are being offered a BRCA1 and BRCA2 gene test by the cancer team. Your cancer team includes your breast surgeon and oncology team. A familial cancer genetic team is also part of the cancer team and would be available for further consultation if required.

Most of the time, cancer occurs by chance. In a small group of people, breast cancer (about 4%, or 1 in 25 patients) and ovarian cancer (about 15%, or 1 in 7 patients) occur due to a significant change (also known as a pathogenic variant) in their BRCA1 or BRCA2 gene. A gene is a unit or structure in the cells of our body that could be passed from a parent to a child. These gene changes also increase the risks of breast, ovarian, pancreatic and prostate cancer.

Significant changes in the BRCA1 or BRCA2 gene occur more frequently in certain groups of people. These include people with breast and/or ovarian cancer (usually developed at a younger age), people with certain types of cancer, and those with a strong family history of breast and/or ovarian cancer.

It is essential to check if your cancer is related to a significant BRCA1 or BRCA2 gene change. This information could help you and your doctor decide about your cancer treatment and measures to reduce your future risk of cancer.

The test could also inform your relatives about their cancer risks, as these significant changes in the BRCA1 or BRCA2 genes could run in the family.

1. **Why am I being offered this test?**

This genetic test looks for changes in the BRCA1 and BRCA2 genes. You are being offered this test because you have been diagnosed with cancer and have fulfilled the criteria for genetic testing. To ensure the test is cost-effective, the following criteria will help narrow the chance of having a positive BRCA result to 1 in 10 rather than 1 in 25 in unselected breast cancer patients.

You have fulfilled one or more of the following criteria from the Malaysia Clinical Practice Guidelines 2019 testing criteria:

1. Developed a type of ovarian cancer known as epithelial non-mucinous ovarian cancer.
2. Diagnosed with breast cancer when you are 45 years old or younger.
3. Diagnosed with breast cancer on both sides at the same time or at different times, when you are 60 years old or younger.
4. Diagnosed with triple-negative breast cancer when you are 60 years old or younger; triple-negative breast cancer is a rare type of breast cancer where the cancer cells have no receptors (types of proteins) attached to them.
5. Men who developed breast cancer at any age
6. Developed breast cancer, and your parent, sibling or child meets any of the above criteria.

(Adapted from Kemp et al, JAMA Network 2019)

2. **What are the benefits of having this test for me?**

This test could provide more information about your cancer. Knowing if you have a significant change in the BRCA1 or BRCA2 gene would help the cancer team recommend suitable treatments. For example, they would recommend the most suitable type of chemotherapy and targeted drugs or surgery for you. The test result would also provide information about your future risk of developing another cancer.

This information could better inform you of your future plans, such as marriage and reproductive planning. You may also consider genetic testing while you are pregnant or before pregnancy.

3. **How is the test done?**

After discussing with your cancer specialist, you would need to provide a blood sample or a buccal swab (cells from your inner cheek) or saliva sample to the lab. The lab test would be done, and the report would be given and explained to you by your cancer specialist.

4. **How long does it take for me to get my results?**

The results will usually be ready within 4-6 weeks.

5. **Who will inform me of the results?**

The cancer team would schedule an appointment with you to discuss the results and address any questions you have. A copy of the gene test results would be given to you. Please make sure you have an appointment date.

Appointment date: .....
Venue: .....

6. **Which lab will do the test?**

The cancer team would inform you which lab your blood or buccal swab (cells from your inner cheek) sample would be sent to.

Lab: ……

7. a) What will happen if "NO significant change in BRCA1 or BRCA2 gene is found"?

Most women do not carry a significant change in the BRCA1 or BRCA2 gene (in this case, 9 out of 10 people tested will not have a significant BRCA gene change), so this is the most likely result most of the time. This means that your cancer is unlikely due to changes in the BRCA1 or BRCA2 gene. This information helps inform the cancer team about your cancer management decisions.

Some gene changes may not be detected in the current laboratory testing. Rarely, significant changes in other genes could be linked to breast or ovarian cancer. The cancer team may have reasons to believe other genetic causes might be involved in causing your cancer, for example, if you have a strong family history of cancer. The cancer team may refer you to the genetic clinic in that case. The genetic specialist doctor would discuss whether you should have further genetic tests.

b) What will happen if a "BRCA1 or BRCA2 pathogenic variant (significant change)" is found?

The cancer team would discuss what this result means for you. They would discuss the significance of this result for your current treatments, your future risk of cancer, the options for future cancer screening and the measures to reduce your future cancer risks.

You may be referred to a breast surgeon, gynaecologist, or genetics specialist, depending on your needs. A genetics specialist would further discuss what this test result means for you and your relatives. They would discuss your risks of developing another cancer, your options for cancer screening and measures to reduce your cancer risk in the future. They would also be able to provide psychosocial support.

A standard referral letter would be prepared for your relatives so they could self-refer to the genetic services. They could make an appointment for genetic counselling to learn more about their future risks of cancer and consider genetic testing to check if they have inherited a significant change in the BRCA1 or BRCA2 gene.

c) What will happen if the test result is unclear?

Sometimes a gene change is found in your sample, known as a "variant of unknown significance". This means that we found a gene change that has not been listed as a significant cause of cancer by experts. Further assessments may be needed to decide if it is linked to your cancer.

This result is not helpful in your current cancer management decisions. The cancer team would refer you to the genetic clinic. The genetic specialist would explain the result in more detail. They would also discuss with you whether further genetic testing would be helpful to check if the gene change is linked to your cancer.

8. How is my personal data handled?

All your personal data would be held under the Personal Data Protection Act 2010 provisions. Your personal data and test results will be securely stored. Only the hospital staff and a few others involved in reporting the results would have access to your data. They are all trained in data handling and protection.

9. Does the test have implications for my family?

No significant change in the BRCA1 or BRCA2 gene would be found in most people. This means that your cancer is unlikely due to a BRCA1 or BRCA2 gene change. This test result does not have any potential implications for your family. The cancer risk of your relatives is the same as that of the general population.

If your test result is positive (you carry a significant gene change), some of your relatives might also have inherited the gene change. They may wish to make an appointment with a genetic specialist to discuss whether or not to have the genetic test.

10. Does the test have implications for my insurance and employment?

In the case where you have life insurance, critical illness insurance or income protection insurance after the gene test is done, you would need to disclose your test result as well as the information about your cancer diagnosis.

Your gene test result is unlikely to affect your existing insurance coverage. If you have the gene test

after your insurance policies are in force, you DO NOT need to disclose your test result.

However, your relatives would need to provide information to the insurance company about your cancer diagnosis and the test result when asked about their family history if they are aware of it.

If the test result is normal, this may be taken into consideration by some insurance companies when handling the unfavourable impact of the family history on the policy. Therefore, relatives may choose to provide information about the normal test result to insurance companies.

You need to be aware that the Genetic Information Non-discrimination Act (GINA) is not available in Malaysia and some parts of the world.

11. **Do I have to take the gene test?**

No, it is your decision whether or not to take this test. Your decision would NOT affect the standard of care the hospital or the staff provides.

12. **What if I am not sure whether or not I want to have the gene test?**

We would suggest you further discuss this with the specialists in the cancer team or genetic team to obtain more information before making a decision.

13. **What will happen next if I agree to take the test?**

If you agree to take the test, you must sign a consent form after the doctor has explained the test in more detail. You need to provide a blood or buccal sample (cells from your inner cheek) for the gene test.

If you have any questions, please contact ……
(E.g. UMMC Breast Cancer Resource Centre Whatsapp number)