Gloucestershire Hospitals
NHS Foundation Trust

Patient Information

BRCA1 & 2 genes and predictive testing

Introduction

This leaflet has been written so you will have information about what BRCA1 & BRCA2 predictive testing means; for you and your family. We have also included some resources that you may find useful.

What are genes?

Genes are the basic component of our heredity makeup and determine what traits we receive from each of our parents, such as hair and eye colour. Sometimes changes (alterations) occur in these genes and these can cause conditions or diseases such as cancer.

What does BRCA stand for?

BRCA is an abbreviation for **BR**east **CA**ncer gene.

BRCA1 and BRCA2 are tumour suppressor genes that help to protect us from developing cancers. When a tumor suppressor gene changes, uncontrolled cell growth may occur. Sometimes these genes alter or change and this increases the risk of developing some cancers.

What is the difference between BRCA1 & BRCA2?

BRCA1 is associated with the increased risk of breast cancer. BRCA2 increases the risk of breast, ovarian, prostate, pancreatic, gallbladder, bile duct and skin (melanoma) cancers.

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How are BRCA genes inherited?

The diagram below helps to explain how our genes come in pairs; one set from our mother and one set from our father. When children are conceived, the genes from both parents randomly pass on one of each pair. If a parent has an alteration in the BRCA1 or BRCA2 gene any of their children (boys as well as girls) have a one in two chance of inheriting it. This can be inherited from the mother or father. If the gene is not inherited it cannot be passed on.

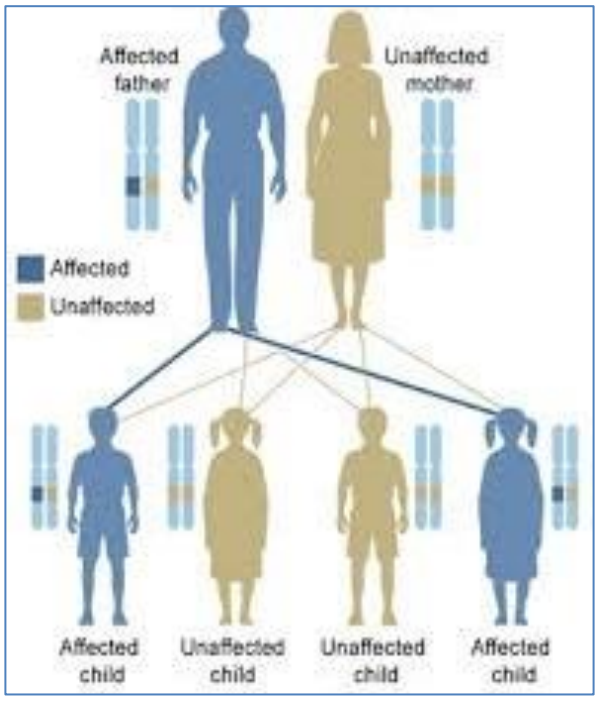

Figure 1: How we inherit our genes

Are there tests available to see if the cancer I or my family member have is related to BRCA1 or BRCA2?

At present current NICE guidelines state that there must be at least a 10% chance of having the BRCA gene before you can be offered a test. Testing is offered to families with a strong family history of breast, ovarian or prostate cancer.

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How do we get tested?

A blood sample is taken from the family member who has a diagnosis of breast, prostate or ovarian cancer. The sample will be sent to a specialist laboratory where the blood is checked to see if there are changes in the BRCA1 or 2 genes.

If a change in the genes is found, then other family members will be invited to be tested to see if they also have the change. This is called predictive testing.

Predictive testing means predicting your or a family member's risk of developing a cancer related to the BRCA gene.

Results of BRCA testing

If your or your family member's results show you have the BRCA1 or BRCA2 gene this may provide an explanation why a cancer has developed. The results can have implications for you and your family's health because of this. You will be offered a referral to the Clinical Genetics team to discuss your options.

Sometimes a genetic change is found which we are not certain of; this is called a Variant of Uncertain Significance (VUS). Because of the VUS result, testing will not be offered to other family members. However, more samples may be requested from you to gather further information.

What if my relative with breast, ovarian or prostate cancer is not available for testing? Can I still be tested?

When there is no affected family to test, the following may be offered:

a) To offer a test to a family member who has not had cancer. This may happen if a family history shows a significantly higher chance that there may be a BRCA1 or BRCA2 gene alteration. However, these results are more difficult to interpret; for example if no gene alteration is found then we cannot tell if a BRCA1 or BRCA2 alteration has been inherited or if there is another cause for the family history.

b) If your relative has died it may be possible to test a tumour sample that would have been taken from your relative during diagnostic tests. Such tests are more difficult than testing blood and it is possible the test will not work due to sample storage.

If a BRCA1 or BRCA2 change is found will a cancer develop?

We are not sure why some people with the gene alteration develop cancers and others don't. Lifestyle may have some impact, but it is important to remember that even if you or someone in your family develops cancer there is a high probability it is usually curable if diagnosed and treated early.

Implications of BRCA1 & 2 testing for you/your family

There are many emotions that may be involved if you or your family member are told you carry the genetic alteration. These emotions can involve shock, anxiety, anger or even guilt that you may have passed the gene alteration on to children.

Your keyworker/clinician can provide you with a letter that you can give to your family with details of how to access testing.

At present, genetic testing does not cause problems when applying for life insurance.

Will having the BRCA1 or 2 gene affect the cancer treatment my family member or I may get?

Yes, it will allow your oncologist to introduce new medications called PARP inhibitors (Poly ADP Ribose Polymerase). PARP inhibitors appear to improve survival and reduce the risk of recurrence for women diagnosed with breast or ovarian cancers related to the BRCA1 or BRCA2 gene alterations.

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What screening is available if I or my family have the gene alteration?

Women

MRI (Magnetic Resonance Imaging) breast screening is offered to women aged 30 to 49 years who have the BRCA1 or BRCA2 gene alteration and to women who have a 50% chance of having the gene.

Mammography: Women with the BRCA1 or BRCA2 genetic alteration will be offered a yearly mammogram from the age of 40 to 69 years. As with MRIs, women with a 50% chance of the gene alteration are also offered screening. After the age of 70 years, women can request a mammogram every 3 years by contacting their breast unit.

Ovarian screening: At present there is not enough evidence to suggest screening for ovarian cancer saves lives, so for now, it is not currently offered by the NHS. Screening is however available privately, please ask your genetic counselor for information. This involves an ultrasound scan of your lower abdomen and a blood test to look for cancer markers in your blood.

Men

There is at present no national screening for prostate cancer in the UK. There is however a blood test called PSA (Prostate-Specific Antigen) to help detect cancer. This is available to all men aged 50 years or over. If men are found to have the BRCA2 gene alteration it may be appropriate to be referred to a urologist to discuss further screening such as an MRI scan. With BRCA1 gene alteration the risk of developing a prostate cancer is low therefore screening is not offered at present.

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Risk reducing surgery

It may be suggested that you or family members consider 'Risk reducing surgery'. This can reduce the risks of certain cancers developing. These surgeries include:

- Breast surgery (risk reducing bilateral mastectomy): This is surgery to remove healthy breast tissue to prevent cancer developing. Research shows that this type of surgery will reduce the chances of developing a breast cancer by 90 to 99%. This is a major operation and serious consideration is needed not only due to the physical aspect of surgery but the psychological impact this may have on you/your family members.
- Removal of ovaries and fallopian tubes: By having the ovaries and fallopian tubes removed the chances of developing ovarian cancer is reduced by 95%. Occasionally this surgery can reduce the chance of a breast cancer if the ovaries are removed before menopause. If you/your family member has not already been through the menopause it will start immediately after this surgery. It may be appropriate to discuss any symptoms or concerns with your GP. HRT is not always recommended for women who have had breast cancer.

Are there any risk reducing medications?

There are certain medications that have been shown to reduce the risk of breast cancer. Tamoxifen is offered to women who have not reached the menopause. Women who have already gone through the menopause may be offered raloxifene and anastrazole. Side effects for these medications will be discussed with you.

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What if I/my partner carries the BRCA1 or BRCA2 alteration but we want to start a family?

A genetic councilor can discuss your options but it should not stop you planning a pregnancy. Most individuals opt to have children in the normal/usual way, however, if the BRCA1 or BRCA2 gene alteration is found in you/your partner you may have the option of having a Pre-Implantation Genetic Diagnosis (PGD). PGD means you would need to start fertility treatment called In Vitro Fertilization (IVF). PGD allows the embryos to be screened and the ones which do not have the BRCA alteration to be implanted back into the womb.

Contact information

Consultant: _____

Specialist Nurse: _____

Genetic Counsellor: _____

Useful websites

Cancer Research UK
Website: www.cancerresearchuk.org

The Eve Appeal
Website: www.eveappeal.org.uk

Macmillan Cancer Support
Website: www.macmillan.org.uk

Ovarian Cancer Action
Website: www.ovarian.org.uk

Target Ovarian Cancer
Website: www.targetovariancancer.org.uk

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Your appointments

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