ovacome..
ovarian cancer

Genetic testing

Ovacome is a national charity providing support and information to anyone affected by ovarian cancer.

We run a free telephone and email support line and work to raise awareness and give a voice to all those affected by ovarian cancer.

This booklet is part of a series giving clinical information about ovarian cancer.

It is one of three booklets discussing genetics, genetic testing and reducing genetic risk.

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Genetic testing and ovarian cancer

There are many reasons why you may want to get a genetic test to find out about your risk of developing ovarian cancer.

You may have a history of ovarian and other cancers in your family. This might make you worried about your own risk and any increased risk to other family members. In this case you can ask your GP to refer you to the local genetic service for an assessment to see if you need a test for a changed gene. This type of testing is called predictive testing.

These tests can show if you have an increased risk of ovarian cancer due to a changed BRCA gene. This means you are BRCA1 or BRCA2 positive.

If you have recently been diagnosed with high grade serous epithelial ovarian cancer and want to find out if you carry a changed BRCA gene, or other gene change, you can have a blood test. This is called germline testing.

Your cancer tumour can be tested for changed genes while you are in hospital. This uses a sample taken from your tumour during surgery. This is called somatic testing.

Somatic testing of the cancer tumour can show changed genes that have caused a condition called HRD (homologous recombination deficiency).

The tests can also detect gene changes that show you have Lynch syndrome, and other genetic conditions that increase the risk of developing ovarian cancer and other cancers.

These tests are described in detail later in this booklet.

Inherited risk of ovarian cancer

You may know if there is a history of ovarian cancer and other cancers in your family. This means that you and others could have inherited, or passed on, an increased risk of developing ovarian cancer in the future.

This risk sometimes shows as a history of family members across generations developing cancers. These cancers can have a common genetic risk factor. For example, breast cancer, ovarian cancer, male breast cancer, prostate cancer and some pancreatic cancers in the same family can mean a changed BRCA gene is being inherited and passed on.

A pattern of colorectal cancers, ovarian cancers such as clear cell or endometrioid, womb cancer, ureter cancer and bladder cancer in your family could mean changed genes causing Lynch syndrome are present.

BRCA and risk

In the UK the risk of developing ovarian cancer is two per cent (2 in 100).

- Having a change in the BRCA1 gene is estimated to increase the risk of getting ovarian cancer, to 40 to 60 per cent (40 to 60 in 100) with risk increasing from age 40.
- A change in the BRCA2 gene is estimated to increase the risk to 10 to 30 per cent (10 to 30 in 100) from age mid to late 40s *(1)*.

Other genes PALB2, BRIP1, RAD51C and RAD51D can also develop changes that increase ovarian cancer risk.

Breast and other cancers

BRCA gene changes can increase the risk of breast cancer too. In the UK the lifetime risk (up to age 80) of breast cancer is 15 per cent (15 in 100).

A changed BRCA1 gene increases risk to 60-90 per cent (60 to 90 in 100).

A changed BRCA2 gene increases risk to 45-85 per cent (45 to 85 in 100) *(1)*.

BRCA gene changes can also increase the risk of prostate cancer and breast cancer in men, pancreatic and possibly other cancers.

Lynch syndrome and risk

Lynch syndrome can be caused by changes to five genes: MLH1, MSH2, MSH6, PMS2 and rarely EPCAM, that can increase the lifetime (up to age 75) risk of:

- Ovarian cancer to 17 per cent (17 in 100.)
- Colorectal cancer to 47 per cent (47 in 100.)
- Endometrial cancer to 49 per cent (49 in 100) *(2)*.

Predictive testing

If patterns of ovarian and other cancers can be seen in your family's medical history, you can ask your GP to refer you to the local NHS genetics service. They can assess your cancer risk. This should give more information about any increased cancer risks and the possible cause *(3)*.

You can be tested if a changed gene has already been found in one of your blood relatives. You can also be tested if there is a family history of ovarian cancer. This means that anyone with a first degree relative (mother, daughter or sister) with an ovarian cancer diagnosis, or a second degree relative (grandparent, grandchild, aunt, niece, or half-sister) with ovarian cancer, can be tested.

These relatives can be on your mother's side of the family or your father's side.

If you are from an Ashkenazi Jewish, Sephardi Jewish or Greenlander population your GP can refer you directly to a genetics service.

Using information about your family's cancer history, your risk of carrying a potentially cancer-causing gene change will be assessed based on your age and gender.

- For people born female this would be expected to rise from two per cent (2 in 100) at age 30 plus to 10 per cent or higher (10 in 100) aged 70 and over.
- For those born male the risk is expected to rise from six per cent (6 in 100) at age 30 plus to 10 per cent or higher at age 70 and over.

If you meet these criteria, then you will be tested.

These criteria are set in NICE guideline Ovarian cancer: Identifying and managing familial and genetic risk (NG241) March 2024.

You can see it here:
https://www.nice.org.uk/guidance/NG241/chapter/recommendations

The NICE guideline also applies to Wales.

Scotland uses existing guidance here:
https://www.sign.ac.uk/media/2010/sign135_oct2022.pdf

Northern Ireland is deciding whether to implement the NICE guidance see more information here: https://online.hscni.net/our-work/nice/

Predictive testing might show that you have increased risk, but it doesn't mean that you have ovarian or other cancers, or that you will definitely get cancer.

Should I be tested?

Genetic testing is a very personal choice. It is a decision you can take after you have had genetic counselling and have a full understanding of what the test is for. You need to think about how the results might affect you and your family, what your feelings might be, the decisions you will need to make, and how you will cope with this.

Pros

- If you are found to have a changed gene, you can plan to reduce your risk of developing cancer.
- You can have regular screening, preventative treatments and make changes in your life which might lower your risk.
- Knowing your result might reduce the anxiety of not knowing.

But:

- Some tests might not have a clear result.
- A positive test result could lead to more stress and anxiety.
- Should you tell your family the result?
- How will they react?

Sue says: "I wasn't offered genetic testing, I requested it because my maternal cousin was diagnosed with breast cancer at the same time I was diagnosed with ovarian cancer. She had the test for BRCA gene mutations, tested positive and informed the rest of the family and sent a copy of a letter confirming it to support anyone else's request for testing."

What happens when you are tested?

Testing for a changed BRCA gene

You can be tested if you have a relative who has been tested and found to have a changed BRCA gene. Or you may have a first or second degree relative who has been diagnosed with ovarian cancer.

There may be strong history of breast cancer and other BRCA related cancers in your family. Prostate cancer, pancreatic cancer and male breast cancer can also be related to changed BRCA genes.

If you are a member of an at-risk population (Ashkenazi Jewish, Seraphic Jewish or Greenlander) your GP can refer you directly to the genetics service for the test, which is usually a blood test.

Your results may take two to four weeks.

You may also be tested for changes in PALB2, RAD51C, RAD51D, ATM, CHEK2 genes and maybe others, as well as the BRCA genes. Testing a range of genes is called panel testing.

Testing for Lynch syndrome

If you are concerned that your family may have Lynch syndrome, tell your GP if:

- Three or more blood relatives have had Lynch related cancers, such as colorectal, endometrial (womb), small bowel, bladder, and types of ovarian cancer such as clear cell and endometrioid.
- They were in more than two generations of your family.
- At least one was aged under 50 when diagnosed.
- One of these is a first degree relative of the other two (parent, child, or sibling).

The Lynch syndrome predictive test looks for changes which affect genes' abilities to repair cells. The gene MSH6 is involved with Lynch syndrome and changes in this gene increase risks of gynaecological cancers, including ovarian cancer and endometrial cancer *(2)*.

Hilary says: "I was diagnosed with ovarian cancer in April 2006 at the age of 48…my mother was a long-term survivor of four primary cancers: endometrial cancer aged 46, ovarian cancer at 58, colon cancer at 65 and finally breast cancer when she was 83.

"I went for genetic testing, which showed that we are affected by Lynch syndrome".

Before any of this testing takes place, you should be offered specialist genetic counselling to make sure you fully understand what the results may mean and how this could affect you and family members.

When you are ready to go ahead you will be asked to sign a consent form.

The test will usually take a blood sample. It will look for BRCA gene changes. If you are being tested for Lynch syndrome it will look for changes in MLH1, MSH2, MSH6 and PMS2 genes.

The test may also include MLPA (Multiplex ligation-dependent probe amplification), a further process to find gene abnormalities.

These test results can take four to eight weeks or longer.

What will the results of your predictive test mean?

If the predictive test is positive, that means you have been found to have a changed gene which increases the risk of developing cancer.

Remember that there are other factors that affect cancer risk, such as your medical history, the environment you live in, the lifestyle choices you make. You can change some of these.

If the test found you have a changed gene, this means there is a 50 per cent (50 in 100) chance that you will pass this on to your children. There is also a 50 per cent chance that each of your siblings has the same gene change.

The results, and their meaning to you and your family, will be discussed with you by the staff at the genetic centre. You should be able to speak to a genetic counsellor too.

If your result is negative, this means that no changed genes were found. This means your risks of developing cancer are the same as the general population. In the case of ovarian cancer that means two per cent (2 in 100).

It also means that you will not pass on to your children a changed gene that can increase their cancer risk.

Home testing

Tests to see if you carry a changed gene can also be ordered privately online or by post. They are designed to be used at home and then sent away for analysis.

These tests are supplied by private companies, not the NHS, so if you want to use them you will have to pay. These tests are not designed to look for specific gene changes that affect your family.

They work by analysing DNA using saliva or a cheek swab that you have collected at home. The sample is then mailed to a private laboratory, and you will receive the results online or by phone or post.

These tests are not intended to diagnose a medical condition. They will look for some of the gene changes that can increase your risk of developing certain conditions. These tests should not be used as an alternative to seeing your local genetics service, and they are not intended for use with children.

Tests that include the Color or Invitae test will be accepted by the NHS.

The cheaper tests may only test a few out of thousands of BRCA mutations and may not include genetic counselling as part of their service.

If you or a family member think there would be benefit from gene testing, the best advice is to see your GP.

Germline testing

This is the test that is carried out in hospital, when you have been diagnosed with ovarian cancer.

It is usually a blood test, but it may use a saliva sample.

NICE guidelines say that everyone with a diagnosis of epithelial ovarian cancer should be offered a genetic test. This includes people diagnosed with subtypes, such as mucinous epithelial ovarian cancer.

Staff should offer you a test for a changed gene, which is usually a test for a BRCA gene change and a Lynch syndrome gene change.

It is up to you to choose if you want to have a germline test. If you are not ready you can wait and make the decision later. If you decide to be tested, you will be asked to sign a consent form.

The results will probably take about six weeks.

Finding out your BRCA result can be important when you make treatment choices. If you are BRCA positive, you are likely to have wider treatment choices and access to certain targeted therapies. These are drugs you can take to control the cancer and prolong remissions.

You can learn more about targeted therapies here:
https://www.ovacome.org.uk/targeted-therapies-booklet

Somatic (tumour) testing for HRD and Lynch syndrome

Somatic testing means using a sample from your ovarian cancer tumour. It looks for gene changes that have affected how the tumour grows. These changes are called HRD (homologous recombination deficiency).

This information can help to find out which treatments will work best for you.

HRD can arise by chance. If this has occurred it cannot be inherited or passed on, so it does not affect other family members. However, it can also be due to inherited gene changes, so you may be offered germline testing too.

NICE (National Institute for Health and Care Excellence) recommends that patients who are newly diagnosed with colorectal or womb cancer should have their tumour assessed to see if it has signs of Lynch syndrome. If it does, then they should be offered germline testing to see if they carry a gene change associated with Lynch syndrome.

The results of your germline and somatic tests

If your result is negative, and you are found not to have a changed gene this is likely to mean your cancer has arisen by chance.

Sometimes the test result can be uncertain, or there is not a result at all. Then you may be offered a repeat test or a further test to find out more.

Information about your result will be confidential. Only the staff in the genetic service or your hospital team, and the lab staff who carried out your test, will know your identity.

Lynne says: "The shock came when I found out I was BRCA 2 positive. I felt guilty that I may have passed the mutation to my children. And then possibly onto my beloved grandchildren. I talked to them about that possibility. I felt that they should be tested but it had to be their decision.

"The genetic service offered counselling for me and them which my daughter took up. Both my son and daughter decided they would be tested. They both were OK which was a great relief."

Talking to your family

You don't have to tell your family about your test results if you don't want to.

If you decide to speak with counsellors at the genetic service, or in hospital, they will talk to you about how your results may affect you and your family.

It could be helpful to tell family members that you are going to have a genetic test. They too may be aware of a family history of cancer, and wondering what this could mean.

If your results show that you do carry a changed gene, other family members may want to know this so they can find out if they too carry a gene change. They might want to take steps to reduce their increased risk of ovarian or other cancers. They may make the choice not to explore this further or know their own personal risk. It is an individual decision.

Younger family members might want to change their plans about when to have children. They may decide to have their families earlier, and then, if necessary, have risk reducing surgery.

These are complex issues, and some decisions may carry their own risks. Family members may need support from specialist genetic counsellors and other professionals to reduce their risks, and progress in their lives.

References

1. Royal Marsden Hospital. A beginner's guide to BRCA1 and BRCA2
2. Royal Marsden Hospital. A beginner's guide to Lynch syndrome
3. NICE Guideline Ovarian cancer: Identifying and managing familial and genetic risk (NG 241)
4. The role of BRCA testing in hereditary pancreatic and prostate families. Cancer Prevention Hereditary Genetics and Epidemiology May 2019

We welcome your feedback on this booklet, email ovacome@ovacome.org.uk or call us on 0800 008 7054. Calls are free. If you would like to discuss anything about ovarian cancer, please phone our free support line on 0800 008 7054 Monday to Friday between 10am and 5pm.

You can also visit our website at www.ovacome.org.uk

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