Genetic testing for women diagnosed with ovarian cancer
Frequently Asked Questions (FAQs)

Ovarian cancer can sometimes be caused by one or more faulty genes. Genetic testing looks for faulty genes that are known to cause ovarian cancer. Knowing whether a woman has a faulty gene can help in making decisions about which treatments to use. Information from genetic testing can also help to assess and manage the woman’s risk of developing other cancers. If a faulty gene is found, this information can also help manage the risk of ovarian cancer and other cancers in family members.

What is ovarian cancer?
Ovarian cancer occurs when abnormal cells in the ovary, fallopian tube or peritoneum grow in an uncontrolled way. Most ovarian cancers start in the epithelial cells, which form the outer layer of tissue around the ovary. The full name for this type of cancer is epithelial ovarian cancer, but it is usually referred to as just ovarian cancer.

Which faulty genes are associated with ovarian cancer?
Ovarian cancer can be caused by faults in BRCA1 and BRCA2 genes. Faults in BRCA1 and BRCA2 genes are found in one in seven women with ovarian cancer in Australia. Faulty genes may be inherited from the mother’s or father’s side of the family.

Less commonly, ovarian cancer can be caused by faults in genes involved in Lynch syndrome. Lynch syndrome is an inherited condition associated with increased risk of endometrial, colorectal, ovarian and other cancers.

Some other gene faults have been found in women with ovarian cancer. However, the risk associated with these gene faults is not yet known.

What is the difference between assessment of genetic risk (genetic assessment) and genetic testing?
Genetic assessment is a way of finding out whether someone may be at increased risk of having one or more faulty genes associated with a particular disease or condition. It involves asking questions about things like family history and personal history of different diseases or conditions, and considers the person’s age and other characteristics. All women diagnosed with ovarian cancer should be offered genetic assessment.

Genetic testing involves testing a sample of blood or tumour tissue in a laboratory to look for genetic changes that are known to be associated with a particular disease.

A woman with ovarian cancer should be offered genetic testing if genetic assessment suggests that she may be at increased risk of having one or more faulty genes. It is best practice that genetic testing is ordered by a health professional experienced in interpreting the results.

When should a woman with ovarian cancer be offered genetic testing?
A woman with ovarian cancer should be offered genetic testing for a faulty BRCA1 or BRCA2 gene if:

- she has Grade 2 or 3 invasive non-mucinous ovarian cancer diagnosed at 70 years or younger; or
- she has invasive non-mucinous ovarian cancer and a personal history of breast cancer (regardless of age); or
- she has invasive non-mucinous ovarian cancer and a family history of breast or ovarian cancer (regardless of age); or
- she is from a population known to have a higher risk of faulty BRCA1 or BRCA2 gene (such as Ashkenazi Jewish women); or
- her ovarian cancer has come back after treatment with a platinum-based treatment, and meets the Medicare Benefits Schedule (MBS) criteria for treatment with a Poly ADP-ribose polymerase (PARP) inhibitor.

For a woman with ovarian cancer, her risk of having a BRCA1 or BRCA2 mutation can be assessed, based on her family and personal history. If her risk of having a faulty BRCA1 or BRCA2 gene is more than 10%, she should be offered genetic testing.

A woman with ovarian cancer should be offered testing for Lynch syndrome gene faults if she has a personal or family history of endometrial, colorectal or other cancer associated with Lynch syndrome.
How might having a faulty gene affect treatment for ovarian cancer?

Some treatments for ovarian cancer are known to be more effective in women with a faulty BRCA1 or BRCA2 gene. Knowing whether a woman has a faulty BRCA1 or BRCA2 gene can help in choosing the most appropriate treatment.

Women with ovarian cancer who have a faulty BRCA1 or BRCA2 gene:

- are more likely to respond to platinum-based chemotherapy
- are more likely to benefit from treatment with a PARP inhibitor if ovarian cancer comes back after treatment with platinum-based chemotherapy.

How might having a faulty gene affect surveillance and risk reduction for other cancers?

Women with a faulty BRCA1 or BRCA2 gene are at increased risk of developing breast cancer. If a woman has a faulty BRCA1 or BRCA2 gene, she may choose to have more regular tests to look for signs of breast cancer (such as mammography, ultrasound or magnetic resonance imaging (MRI)). Strategies to reduce the risk of breast cancer may be considered, including mastectomy or use of medicines known to reduce the risk of breast cancer.

Women with Lynch syndrome are at increased risk of developing colorectal cancer, endometrial cancer and some other cancers. If a woman has Lynch syndrome, she may be advised to have regular surveillance and / or screening tests to look for signs of cancer.

What are the implications of finding a faulty gene for family members?

If a woman with ovarian cancer has a faulty gene known to increase the risk of ovarian cancer or other cancers, relevant family members can be offered genetic counselling and, where appropriate, have genetic testing themselves.

If a faulty gene is found, risk-management options may be considered. These may include more frequent testing to look for signs of cancer, or strategies to reduce the risk of cancer.

If a faulty gene is not found, the family member(s) can usually be reassured that they are not at increased risk of ovarian cancer or other cancers associated with the gene fault.

Where can I find more information about genetic assessment and testing?

If you are concerned about your familial risk of breast and ovarian cancer, or have questions about genetic testing, speak to a member of your health care team. For more information, see the Cancer Australia website.

Find out more about Ovarian Cancer

ovarian-cancer.canceraustralia.gov.au

Cancer Australia aims to improve outcomes for women diagnosed with gynaecological cancers in Australia through the translation of evidence, development of new resources, investigation of innovative and sustainable models of care and the continued investment in priority areas of gynaecological cancer research.