Understanding Ovarian Cancer Tumour Genetic Testing: Information for Patients

Genetics and Cancer

Our bodies are made up of many cells. Genes in our cells provide instructions to our body about how to grow, develop, and stay healthy. Most cells in our body contain our full genetic code, about 20, 000 different genes. Cancer can develop if one of our cells contains a mutation (harmful change) in certain genes.

What is tumour genetic testing?

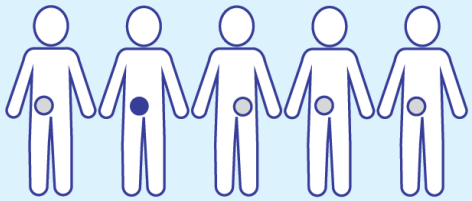
We use tumour genetic testing to see if there are mutations in the *BRCA1* or *BRCA2* genes of your tumour cells that may have led to cancer developing. This genetic testing does not look at genes in your blood or other cells in your body.

Why is tumour genetic testing done?

The purpose of tumour genetic testing is to help decide what type of treatment could work best for you. For example, a person with ovarian cancer may benefit from treatment with a type of medication called "PARP-inhibitors" if their tumour has a *BRCA1* or *BRCA2* gene mutation.

Tumour genetic testing may be ordered if you have a high-grade serous ovarian cancer. About 1 in 5, or 20%, of people with this type of ovarian cancer have a *BRCA1* or *BRCA2* mutation. Your doctor will talk with you about this test if it is important for you.

People with high-grade serous ovarian cancer have a 1 in 5, or 20%, chance of having a *BRCA1* or *BRCA2* mutation in their tumour.



1 in 5

What if my tumour genetic test result shows that I have a *BRCA1* or *BRCA2* mutation?

When a test result shows you have a *BRCA1* or *BRCA2* mutation, it is called a "positive" tumour genetic test result. About 1 in 5, or 20%, of people with high-grade serous ovarian cancer will have a positive result. The care team will talk with you about any changes they want to make to your treatment based on this test result.

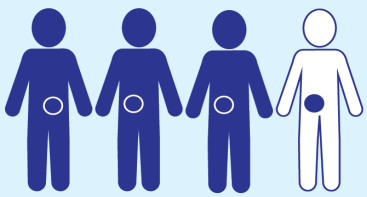
Gene mutations in tumour cells can arise in different ways.

- For some people, those gene mutations are **acquired** – they develop by chance in a cell that was healthy. The mutation is only in the tumour.
- For other people, the gene mutation is **inherited** from a parent, and is present in the tumour and all other cells of the body (including eggs and sperm). This is also called a germline mutation.

Whether aquired or inherited, a *BRCA1* or *BRCA2* mutation can lead to cancer.

About 3 in 4, or 75%, of people with a positive ovarian tumour genetic test have inherited a *BRCA1* or *BRCA2* mutation. Those people with an inherited mutation will benefit from further discussion about what this means for them and for their family.

About 3 in 4, or 75%, of people with a positive ovarian tumour genetic test will have a *BRCA1* or *BRCA2* inherited genetic mutation.



3 in 4

How will I know if a positive tumour test is due to an inherited or acquired mutation?

To know whether a tumour *BRCA1* or *BRCA2* mutation is inherited or acquired we must compare your tumour genetic test result to results of a genetic test for the same genes in cells outside of the tumour (usually blood). If you have high-grade serous ovarian cancer, you will be offered a tumour genetic test, as well as a blood genetic test to look for inherited mutations.

The blood genetic test looks for inherited mutations in the *BRCA1* and *BRCA2* genes, as well as other genes that can be linked to inherited ovarian cancer. The results together will help us understand if a mutation is inherited or acquired.

What is the difference between an acquired mutation and an inherited mutation?

Aquired gene mutation	Inherited gene mutation
The mutation is only in your cancer cells. It occurred by chance in the gene of one of your cells and cancer developed. 	This gene mutation is in all of your cells because it is part of your body's full genetic code. This includes the cancer cells. It was likely inherited from one of your parents. 
• You may be eligible for treatment with PARP inhibitors. • You likely do not have increased genetic risk to develop other cancers. • Your relatives are not at risk to carry the mutation.	• You may be eligible for treatment with PARP inhibitors. • You may have increased risk for other cancers linked to the inherited gene mutation. • Your relatives may also carry the mutation. All first-degree relatives have a 1 in 2, or 50%, chance of having the same gene mutation and associated increased cancer risks. • The gene mutation can affect what cancer screening you or your family may need.

For details, see the "Hereditary Cancer and Genetic Testing After a Cancer Diagnosis" booklet (go to https://www.albertahealthservices.ca/cancer/Page16322.aspx and look in the 'Tests' section).

Hereditary Cancer Clinic

Alberta Cancer Patient Education

Alberta Health Services
CancerControl Alberta

Procedures, Infusions, Tests | Tumour Genetic Testing | PROV | 2020 | CPE-L0058