

UNDERSTANDING GENETIC TESTING IN CANCER CARE



In this series, we break down common types of genetic testing. This information is meant to support learning and conversation, not to replace medical advice.

WHAT IS GENETIC TESTING?

Genetic testing looks at DNA to find changes (variants) that may affect how cancer starts, grows or responds to treatment.

EXAMPLE:

A tumour is tested to identify DNA changes that may help doctors choose treatment.

WHEN IS IT USED?

- At diagnosis
- When planning treatment
- If cancer changes or returns

WHY IT MATTERS:

Genetic testing helps doctors and patients choose treatments that are more personalized, which may be more effective.

WHAT SAMPLES ARE USED?



Blood or saliva



Tumour tissue from biopsy or surgery



WHY GENETIC TEST RESULTS MATTER FOR TREATMENT

Genetic test results can help your care team:

- Identify targeted therapies
- Determine eligibility for clinical trials
- Predict response to certain treatments

Genetic Test

Result

Personalised Treatment

Talk to your healthcare team about how genetic testing can impact your care.

TYPES OF GENETIC TESTING:

GERMLINE VS. SOMATIC TESTING

GERMLINE TESTING

Germline testing looks for DNA changes a person is born with and that are present in every cell in their body.

WHEN IS IT USED?

- To assess inherited cancer risk
- To guide family planning
- To identify the best screening approaches
- To guide treatment decisions

WHAT SAMPLES ARE USED?

Blood or saliva



EXAMPLE: Changes in genes like BRCA1 or BRCA2 that increase risk of breast and ovarian cancer.

WHY IT MATTERS:

May provide information about the risk of specific cancers for you and your family members

SOMATIC TESTING

Somatic testing looks for DNA changes that happen only in cancer cells.

WHEN IS IT USED?

- To identify mutations in the tumor that can guide targeted therapy or clinical trial eligibility
- To identify mutations that could help to diagnose the tumour

WHAT SAMPLES ARE USED?

Tumour tissue from biopsy or surgery



EXAMPLE: Changes in the EGFR gene that are associated with a tumour cell being sensitive to an EGFR inhibitor drug.

WHY IT MATTERS:

Helps doctors identify personalised treatment based on the tumor's unique genetic profile

WHO DOES IT?

GERMLINE TESTING

Clinical Geneticist or Oncologist (some cases)

Testing is performed in an accredited provincial laboratory by licensed genetic technologists

Test results are interpreted by trained genome analysts

The report is signed out by a certified molecular genetics scientist

Results are explained with support from a genetic counsellor

ORDERS THE TEST

WHERE IS THE TESTING DONE?

INTERPRETS THE TEST RESULTS

SIGNS THE REPORT

REVIEWS THE RESULTS WITH YOU

SOMATIC TESTING

Oncologist or Pathologist

Testing is performed in an accredited provincial laboratory by licensed genetic technologists

Test results are interpreted by trained genome analysts

The report is signed out by either a certified molecular genetics scientist or a molecular pathologist

Results are reviewed with the patient by the medical oncologist

SINGLE-GENE TESTING

Single-gene testing looks for changes in one specific gene known to be linked to cancer or treatment options.

WHEN IS IT USED?

- When a specific gene is closely linked to treatment
- When a fast, focused answer is needed

EXAMPLE: Testing the EGFR gene in lung cancer to see if a targeted drug may help.

WHY IT MATTERS:

It can quickly show whether a targeted treatment is likely to be effective.

WHAT SAMPLES ARE USED?



Tumour tissue from biopsy or surgery

MULTI-GENE TESTING

Multi-gene panel testing looks for changes in several genes at once to find changes that may affect cancer or treatment response.

WHEN IS IT USED?

- When more information is needed to guide treatment
- When multiple genes may be involved in the cancer

WHY IT MATTERS:

Testing multiple genes at once provides more information from a single test, reducing the need for extra procedures.

EXAMPLE:

A tumour test checks dozens of genes that may guide therapy decisions.

WHAT SAMPLES ARE USED?



Tumour tissue from biopsy or surgery

RNA SEQUENCING

RNA sequencing measures which genes are active in a cell and how they are functioning.

WHY IT MATTERS:

It shows how genetic changes affect cancer behavior, helping guide treatment decisions.

WHEN IS IT USED?

- When doctors need more information on cancer genes
- When DNA testing does not provide enough detail

WHAT SAMPLES ARE USED?

Tumour tissue from biopsy or surgery



EXAMPLE:

A tumour test checks the activity of dozens of genes that may guide therapy decisions.



WHOLE EXOME SEQUENCING

Whole-exome sequencing examines all parts of genes that produce proteins, where most disease-causing changes happen.

WHEN IS IT USED?

- When previous tests give unclear results
- In specialised care or research settings

WHAT SAMPLES ARE USED?



Blood or saliva



Tumour tissue from biopsy or surgery

EXAMPLE: Used when standard tests do not explain the behaviour of a patient's cancer.

WHY IT MATTERS:

It can identify rare changes that may guide treatment decisions or access to clinical trials.

WHOLE GENOME SEQUENCING

Whole-genome sequencing looks at all of a person's DNA, including areas outside of protein-coding genes.

WHY IT MATTERS:

It provides the most comprehensive view of genetic changes, helping doctors understand cancer better and plan personalised treatment.

EXAMPLE:

Used in complex or rare cancers to provide a complete genetic picture.

WHEN IS IT USED?

- In research studies
- In rare or difficult-to-treat cases

WHAT SAMPLES ARE USED?



Blood or saliva



Tumour tissue from biopsy or surgery

QUESTIONS TO ASK YOUR HEALTHCARE TEAM ABOUT GENETIC TESTING

1. Which type of test is right for me?
2. How will results affect my treatment?
3. Could results impact my family?



Being informed is the first step toward personalised care



Want to learn more?

Explore OMPRN's Sporadic vs Familial vs Hereditary Cancer infographic and our interview with a genetic counsellor for more information on genetics and family history. We also have our enhanced glossary for any terms you would like explained and defined.