

A photograph of a female doctor in a white lab coat sitting on a white bench, holding a clipboard and a pen, and talking to a pregnant woman. The woman is wearing a light blue long-sleeved shirt and blue jeans. They are in a clinical setting with a white wall and a potted plant in the background.

CBCN Advocacy Guide: Accessing Genetic Testing in Canada

What is Genetic Testing

Genetic testing is a medical test that assesses changes in an individual's chromosomes, genes, or proteins to determine whether they have a genetic condition, may develop a genetic condition, or may pass on a genetic disorder. For those who already have a diagnosis for a certain disease or conditions, genetic testing can help to make better-informed treatment decisions. For those who have not been diagnosed but whose test results show mutations, **preventive treatment** options can be looked into to address their higher risk of developing the condition they test positive for.

What Does Genetic Testing Involve?

Genetic testing is done by taking a sample of blood, saliva, hair, skin, or other tissue then sending the sample to a laboratory that analyzes the sample. The results are then sent back to the healthcare professional who requested the test. Genetic testing involves two steps: genetic risk assessment and genetic counselling.

Genetic Risk Assessment

A genetic risk assessment, which is done by a doctor, nurse, or genetic counsellor, looks at your likelihood of developing a certain disease based on family history and other factors. A risk assessment discusses and includes the following ^[1]:

- Why you want to have genetic testing done?
- Your family and medical history
- Lifestyle factors
- Physical exam

Genetic Counselling

A genetic counsellor identifies those at risk with hereditary genetic conditions, provides information and supportive counselling, reviews testing options and connects patients/families with the right community resources.

Breast Cancer, Treatment Decisions and Genetic Testing

Certain mutations in an individual's genes, such as the BRCA genes, have been linked to breast cancer. For people diagnosed with breast cancer, genetic testing can be used to determine which treatment or surgery you may respond to better or benefit from more. For example, research has found that those with BRCA-mutations respond better to some treatments and therapies over others. Because individuals with BRCA-mutations have a higher risk of recurrence, when it comes to **breast surgery**, a bilateral mastectomy may be recommended over a lumpectomy. Similarly, **studies** have shown that HR-negative BRCA1 mutated breast cancers benefit more from platinum agents such as cisplatin rather than taxanes. There are also new treatments emerging for this patient population with research showing that some patients with BRCA mutation may also benefit from specific treatments like PARP inhibitors.

When to Get Genetic Testing Done

For more accurate test results, it is generally recommended for the family member that is already diagnosed to be the first person to get genetic testing done. Those whose genetic testing results are positive for hereditary mutations may then want to inform other family members that they get tested.

If you have a risk of developing cancer due to your personal and/or family history, your doctor may recommend genetic testing. Some personal and family history that may suggest a hereditary cancer syndrome include ^[2]:

- Being diagnosed with cancer at a young age
- Being diagnosed with cancer in both organs, for organs that come in a set of pairs (eg. lungs, breasts)
- Multiple cancers in one individual
- Ethnicity
- Birth defects known to be associated with inherited cancer syndromes
- Rare cancer (for example, **male breast cancer**)
- Multiple first-degree relatives with the same type of cancer
- Multiple family members diagnosed with cancer

^[1] Genetic testing. *Canadian Cancer Society*.

<https://www.cancer.ca/en/cancer-information/cancer-101/what-is-cancer/genes-and-cancer/genetic-testing/?region=on>

^[2] Genetic Testing for Inherited Cancer Susceptibility Syndromes. *National Cancer Institute*.

<https://www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet>

Advocating to Get Genetic Testing Done

Because publicly-funded genetic testing varies across the country and is also restricted to certain high-risk individuals, you may want to advocate for more equitable access to this testing. This is especially relevant if you live in a province or territory that does not cover genetic testing or if you do not meet the specific eligibility criteria. Below we discuss avenues and actions you can take to advocate to get access to hereditary genetic testing. Before taking these steps, it is important to first discuss with your healthcare team to confirm that genetic testing would be beneficial to you.

Contact Your Provincial MPP/MLA

Depending on where you live, you can reach out to your MPP (Member of Provincial Parliament) or MLA (Member of Legislative Assembly) and detail your situation, whether it is that genetic testing is not covered in your province/territory or that you do not fit the eligibility criteria. This is also a way to explore whether policy changes need to be made to ensure that other breast cancer patients who may need to access genetic testing can do so.

You can [find the contact information](#) for your elected official using your home address. In your correspondence to them, be sure to include what the importance of having access to genetic testing would mean to you.

Contact Your Ministry of Health

The Ministry of Health where you live is responsible for setting healthcare policies in your province. You can reach out to them to voice your concerns regarding access to genetic testing. If you reach out to your MPP/MLA, they can also reach out to your provincial Ministry of Health on your behalf. You can find the contact information for your Ministry of Health [here](#). In your correspondence to them, be sure to include what the importance of having access to genetic testing would mean to you.

You can also contact the Minister of Health of Canada and voice your concerns about not being able to access genetic testing and why it is important for yourself and other breast cancer patients to be able to access it. The contact information for the Minister of Health of Canada is:

Minister of Health
House of Commons
Ottawa, Ontario K1A 0A6
hccminister.ministresc@canada.ca

Contact Your Private Insurance Company

If you have private insurance, then you may be able to have your genetic testing covered if you are able to find a private clinic that offers this type of test. The first step would be to check what your current coverage is, or to call them directly to see if whether it can be covered. You can find a list of questions to ask your private insurance company regarding coverage as well as questions to ask if genetic testing is not covered [here](#). If you end up in a situation where your insurance does not cover genetic testing, you can take [these steps](#) to appeal.

Pay Out-of-Pocket

If you don't wish to contact a government official, don't have private insurance, or if your private insurance company is unable to cover these tests, the option to pay out-of-pocket is available. Simply visit the official website of the companies that offer genetic testing to see if you can buy it directly through them. Be sure to read through their terms and conditions and well as what the test results can and cannot guarantee. It is also advisable to speak to your doctor prior to buying any tests to ensure that you are getting the appropriate test for yourself which will help to ensure that the test results will be valuable to you.