

How to get genetic testing for inherited retinal diseases in QUEBEC



**FIGHTING
BLINDNESS
CANADA**

What are inherited retinal diseases?

An inherited retinal disease (IRD) is a disorder leading to vision loss that can be traced to a specific genetic cause. People with an IRD will have an eye condition that may run in the family. Symptoms of genetic eye diseases vary depending on which part of the eye is involved. Often, vision loss progresses over time. This diagnosis is usually made by your eye doctor.

What is genetic testing?

Genetic testing involves taking a sample of your DNA (from your blood) and analyzing your genes to find any changes that may cause your IRD and symptoms.

Why should I get genetic testing?

Genetic testing helps confirm your diagnosis. Knowing your genetic mutation may provide information about how your eye disease will progress and what may be done to help. Many new treatments being studied are gene specific. Knowing your gene mutation may improve your chance of being eligible for a clinical trial or gene-specific treatment.

Genetic testing can also tell you how the condition is passed through your family and if other family members are at risk of inheriting the condition.

Do I have to pay for this test?

Every Canadian provincial government covers genetic testing for IRDs. There are a few steps to this process which we have outlined on the next page.

What is a genetic counselor?

Genetic counselors are health professionals with training in medical genetics and counseling. They work with a healthcare team to provide patients and families with information about their genetic diseases. Patients and families may use this information to make informed decisions about their health.

Can I get genetic testing privately?

Access to private genetic testing should be discussed with your doctor or genetic counselor.

What do I do with my results?

Fighting Blindness Canada's IRD Patient Registry is a secure clinical database where information about your clinical and genetic information is collected. This is an opportunity to be connected with clinical trials that may be appropriate for you. Call or [email](#) the health information line to learn more.

Do you have questions? Our health information line is here to help.

healthinfo@fightingblindness.ca • 1.888.626.2995

1	After a diagnosis of a genetic eye disease talk to your eye care provider, family doctor or nurse practitioner about how to access genetic testing. You may be referred to a genetics clinic or your retinal specialist may guide you through this process.
2	If you are being referred to a genetic counselor, your healthcare provider can find a list of genetic centres near you using the Canadian Association of Genetic Counselors website. Make sure you are looking for “general genetics.”
3	The genetics clinic will look at your file, including your diagnosis, vision and family history. They may ask you, or your referring physician for more information before setting up an appointment. The wait time for an appointment may vary and may be up to one to two years. Testing for children and those in clinical trials is about 6 weeks. You will meet your counselor or discuss with your retinal specialist in person or virtually (i.e. on the phone).
4	It is important to determine if genetic testing is appropriate for you. To do this, the retinal specialist or genetic counselor will collect a full medical and family history which helps identify what genes should be tested. If your appointment is in an ophthalmology clinic, the physician may complete an eye exam.
5	If genetic testing is appropriate for you, the details of the test will be discussed with you. You will be asked to provide consent (usually in writing). Testing usually requires a small blood sample. Most genetic tests for eye conditions require approval from the Quebec Ministry of Health (RAMQ). RAMQ covers the cost of genetic testing if: <ul style="list-style-type: none">• The person’s health card is valid.• The testing is felt to be appropriate.• The person has not had the same or similar test before. Genetic testing results usually take about 6 weeks to come back.
6	When results are available it is important that your referring healthcare provider or genetic counsellor review the report with you. They will explain if and how these results may affect your eye and general health. You should request a copy of the results.
7	The next steps are: <ul style="list-style-type: none">• You may be directed towards relevant clinical trials or back to your eye care provider. Keep a copy of your genetic results and share it with your doctors in confidence.• Consider joining the Fighting Blindness Canada IRD patient registry: fightingblindness.ca/patient-registry