Your Guide to a Diagnosis of Retinitis Pigmentosa

Sunnybrook Health Sciences Centre
Ophthalmology
Created By: Dr. Brian Ballios, Dr. Radha Kohly and Dr. Austin Pereira

What is Retinitis Pigmentosa?

Retinitis Pigmentosa (RP) is an **inherited retinal disease** (IRD) that results in loss of the specialized cells (**photoreceptors**) that detect light in the back of the eye. The rod photoreceptors, which are responsible for night and side vision, are first affected by RP. Cone photoreceptors, responsible for central and daytime vision, can be affected later in the disease.

RP is a clinical diagnosis, made by your IRD specialist, with the help of specific tests. For example, an **electroretinogram (ERG)** may be used to measure how well the photoreceptors are working. Once a diagnosis has been made, genetic testing helps determine what gene is mutated. RP can be inherited in a variety of ways in a family. A **genetic counsellor** can then help determine the risk to relatives and future generations.

Challenges with Vision

Loss of peripheral and night-time vision are typically the first symptoms experienced in RP. However, in advanced disease, some patients may have difficulty with their central vision and light sensitivity as well. **Legal blindness** is defined as a visual acuity less than 20/200 or a visual field narrower than 20 degrees.

Medical Treatments

To date, **Luxturna (voretigene neparvovec-rzyl)** is the only Health Canada approved treatment for the specific genetic form of RP, called RPE65. To be considered for treatment, patients must undergo genetic testing to confirm an RPE65 gene mutation. Research and clinical trials are ongoing for dozens of other RP-associated genes, and new treatments will be coming. RP patients may also develop retinal swelling (**edema**), which can sometimes be treated with drops or oral medications.

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Clinical Trials

Research is being conducted to increase the number of available treatments for patients with RP. Depending on your type of RP, you may be eligible to be enrolled in research studies and clinical trials for **new treatment modalities.** We advise you to speak to your IRD specialist about which studies you may be eligible for. We also strongly advise being cautious of information or treatment options found online as they may not be supported by reliable data. Before participating in any trials, please discuss this with your IRD specialist.
https://clinicaltrials.gov
https://www.fightingblindness.ca/resources/everything-you-need-to-know-about-clinical-trials/

Genetic Testing

A key factor in the diagnosis of RP is genetic testing. Your genetic code (DNA) instructs your body to make proteins important for vision. Mis-spellings (**mutations)** in specific sequences of DNA are the basis of RP.

Not every person with RP has the same disease experience. RP has been linked to mutations in over 70 genes. Genetic testing is crucial to help **diagnose** RP, to determine the risk of **inheritance** for other family members and may help with family planning. Genetic testing can indicate whether you may be eligible for specific clinical trials and/or potential new therapies.

The **cost of genetic testing is covered** by the Ontario Health Insurance Program (OHIP). If appropriate, your physician will apply for testing, or engage a genetic counsellor or clinical genetics team to assist. Genetic testing requires a sample of DNA from saliva or blood.

Even though our genetic knowledge is growing, up to 30% of people with genetic testing will have a “negative result,” meaning that we are not yet able to identify the mutation causing their RP.
https://www.fightingblindness.ca/resources/genetic-testing-for-inherited-retinal-diseases/

Driving and RP

Patients with RP may find driving **more difficult,** and at some point, may no longer meet the standards set by the **Ministry of Transportation (MTO).** According to **Ontario law,** it is the legal responsibility of your doctor to report vision failing to meet the minimum standard to the MTO.

Vision Waiver Program

The vision waiver program is available for class G drivers in Ontario who do not meet the **visual field requirement** set out by the MTO. Your ophthalmologist or IRD specialist can help to complete the vision waiver program forms. An occupational therapist can also help assess any challenges affecting your ability to drive.
For more information:
https://files.ontario.ca/mto-fact-sheet-class-g-vision-waiver-en-2022-03-29.pdf

Resources

Your IRD specialist will implement a **multi-disciplinary approach** to manage your vision issues. Several resources used in this approach are listed below:

Canadian National Institute for the Blind (CNIB)
CNIB is aimed to empower those impacted by low vision. This includes providing low-vision aids, connecting with support groups, and registration for clinical trials.

Fighting Blindness Canada (FBC)
FBC is Canada's largest health charity foundation. It leads in the funding of promising research for treatments and cures for eye diseases such as RP.

Mental Health and Wellness
Living with RP is challenging for both patients and their families. Psychiatrists or counsellors are available for support.

Occupational Health Resources
To help mitigate the impact of low vision at work or around the home, occupational therapists can be engaged to help optimize your daily functioning.

Disability Tax Benefits Information:
https://www.canada.ca/en/revenue-agency/services/tax/individuals/segments/tax-credits-deductions-persons-disabilities.html