Retinitis pigmentosa (RP) is a genetic condition that slowly damages the retina. The condition progresses throughout a person’s life, affecting 1:3500 Canadians.

**Early Symptoms**
RP is usually diagnosed in childhood or adolescence, although some people have no recognized symptoms until their adult years. The most common early symptom is difficulty seeing at night and adapting to dim light conditions. This is called nyctalopia (night blindness). People also begin to lose peripheral vision quite early in the disease.

**Medical Description**
RP occurs because the light-sensing retinal cells, called photoreceptors, are slowing damaged due to an inherited genetic mutation. Many different mutations can cause RP.

There are two types of photoreceptors: rod cells and cone cells. Rod photoreceptors are responsible for peripheral vision and night vision; cone photoreceptors are responsible for central vision and for seeing fine detail and colours. Night blindness occurs early in RP because the mutations that cause RP damage the rod cells first.

Over time, as more rod photoreceptors are lost, cell death also occurs amongst the cone cells. This is not well understood, but cone cell loss seems to be triggered by the death of rod cells. When cones die, central vision and visual acuity are lost.

An eye professional, examining the retina of a person with RP, usually sees a mottled pattern in the retina. This is due to clumps of dark pigment that accumulate as the retina is damaged. They give the disease its name. Other typical findings of a retinal examination include thinning of blood vessels and a pale, waxy appearance of the optic nerve; all of these are most pronounced in late-stage RP.

In most cases, the symptoms of RP are restricted to the eye; however,
in 20-30% of cases, RP can be part of a disease syndrome that affects other parts of the body. The most common examples are Usher syndrome and Bardet-Biedl syndrome. More than 30 rare inherited conditions can include RP-type vision loss.

**Diagnosis**

An ophthalmologist may suspect RP on the basis of a person’s symptoms and the findings of a simple eye examination. Two tests are used to clarify the diagnosis:

**ERG (electroretinography)** measures the electrical responses of the retina to light. It can measure the responses of both rod and cone photoreceptors. Although both rods and cones may be affected in people with RP, the most marked changes early in disease are in the rod cells; this characteristic pattern helps diagnose the condition. To have an ERG test, you will be asked to stay in a darkened room for 30 minutes, and drops will be put into the eye or eyes being tested. For the test itself, a special contact lens or gold-foil electrode is placed on the eye or lower eyelid, and the eye is exposed to flashes of light.

**Visual Field Testing** detects, measures, and monitors blind spots in vision. In this test, you look into a device that emits flashes of light and indicate which ones you see. The flashes that you see (or don’t) are recorded. This gives a measure of how much of your vision is affected.

In addition to these tests, genetic counselling is also an important part of the diagnostic process.

**The Genetics of RP**

RP is a genetic condition caused by mutations that adversely affect the retina’s light-sensing photoreceptors. It can be inherited in several different inheritance patterns: autosomal dominant, autosomal recessive, or x-linked recessive. A genetic counsellor can talk with you about your family history and determine which of these patterns is associated with your vision loss. With this information, the counsellor may be able to tell you more about how your condition will progress, and give you and your family information about the risks of vision loss for other family members.

In some cases, people with RP may also have genetic testing done from a blood sample. This type of testing may determine the specific mutations that cause a person’s disease.

Typically, each person with RP only has damage in one pair of genes. Scientists have now identified more than 50 genes that can have mutations that cause RP. It is likely that mutations in more than 100 different genes will eventually be identified. Because so many RP-causing gene mutations are still unknown, there is about a 50:50 chance that genetic testing will
provide a definitive result. Given your family history and the inheritance pattern of your RP, your genetic counsellor will be able to advise you about the likelihood that a genetic test will provide a definitive result. See our genetic testing fact sheet for more details.

Different genetic mutations can damage the retina or impair its function in different ways; for example, some mutations affect how the retina processes nutrients, while others damage the photoreceptors. It’s important to identify the specific gene and mutation, because many treatments being developed for RP will be specific for particular genetic types.

**What to Expect**

As RP progresses, peripheral vision is slowly lost, resulting in ‘tunnel vision.’ By age 40, many people with RP are legally blind, with a severely constricted field of vision, although many may retain the ability to read and recognize faces. Uncomfortable sensitivity to light and glare is common, as is photopsia (seeing flashes of light or shimmering).

RP also causes a loss of visual acuity (the ability to see clearly), but the onset is more variable. Some patients retain normal visual acuity, even when their vision is reduced to a small central island; others lose acuity much earlier in the course of disease. Eventually, however most people with RP will begin to lose central vision and some will lose all light perception.

**Treatment**

No treatments are currently approved to prevent or slow the vision loss associated with RP. In spite of this, it is important to have regular eye exams even if your vision is not changing – to avoid serious but treatable complications that might further impair your vision, such as cataracts and macular edema.

**Research**

Many research groups are working to develop treatments and cures for RP. Experimental treatments can be divided into three broad categories:

- Protective Therapies
- Corrective Therapies
- Sight-Restoring Therapies

**Protective therapies** aim to stop (or at least slow) the damage caused by genetic mutations. Often protective therapies are not specific to one mutation, but may benefit people with many types of RP. These include treatments to stop the process of photoreceptor death (apoptosis), as well as cell-derived therapies that aim to help photoreceptors survive. One example of this latter type, called NT-501, is being developed by Neurotech Pharmaceuticals. A small capsule of live cells, which have been genetically modified to
release growth factors that can protect the photoreceptors, is implanted into the eye through a small incision. A trial of this therapy is currently underway at the University of California, San Francisco. Another protective therapy being tested is a drug that can be taken orally, valproic acid, which may be useful for some forms of autosomal dominant RP.

Some protective therapies aim specifically to prevent the death of cone cells in RP – and thus, the loss of central vision – in later stages of the disease. See our fact sheet, to learn more about cone protective therapies.

**Corrective therapies** aim to reverse the underlying genetic defect that causes vision loss. If these therapies are successful they might prevent a person who is treated when first diagnosed, from ever developing vision loss. Corrective therapies might also help slow the disease in people whose vision has already been affected, especially in the earlier stages. The corrective therapies being developed now are specific to certain forms of recessively inherited RP. Gene therapies, which replace a non-functioning gene, are one type of corrective therapy. Clinical trials of gene therapies for several types of RP have begun, and the results so far are encouraging. Agents that replace the lost function, such as a drug being developed by Canadian company QLT Inc for RP due to mutations in RPE65 and LRAT, also may be corrective.

**Sight-restoring therapies** are also a growing area of research success. These therapies are intended for people who have already lost all, or much, of their vision. Stem cell therapies aim to replace the retina’s lost photoreceptors. There are promising early results with stem cell trials involving other retinal degenerative diseases; trials with RP are on the horizon. Retinal prosthetics, such as the Argus II or “Bionic Eye,” use computer technology to generate vision. The Foundation Fighting Blindness is helping to support the first Canadian trial of the Argus II. Drug and gene therapies are also being developed that may give non-photoreceptor nerve cells in the retina the capacity to sense light.

Thanks to our generous donors, we are funding groundbreaking research in these areas. Visit our website at [www.ffb.ca](http://www.ffb.ca) to learn more.

*Updated November 7, 2014:* Updated by Dr. Mary Sunderland, Senior Manager, Research & Education, Foundation Fighting Blindness. Initially reviewed by Dr. Jane Green, Ocular Genetics Research, Professor, Memorial University, St. John’s, Newfoundland & Labrador, and Dr. Bill Stell, The Foundation Fighting Blindness Expert Scientific Advisor and Professor of Cell Biology & Anatomy at the University of Calgary.