The Foundation Fighting Blindness

Guide to

Retinitis Pigmentosa and Related Conditions
Our Vision

To restore the gift of sight to people living with blindness.

Our Mission

The mission of The Foundation Fighting Blindness is to find the causes, treatments and ultimately the cures for retinitis pigmentosa, macular degeneration and related retinal diseases by the support and promotion of research and the development of public awareness.

What We Value

We value SIGHT, excellence in research, our partners in fighting blindness and the commitment to a cure.

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Introduction

The Foundation Fighting Blindness Guide to Retinitis Pigmentosa and Related Conditions is an update of Your Guide to RP: Information and Resources (1998, 2000). The information here is intended to be as reliable and relevant as possible. The diverse group of contributors include vision care professionals, service providers and members of the Retinal Degeneration (RD) Network. The RD Network is a group of volunteers affected by RP, either as a patient, family member or friend.

This booklet has been designed with low vision in mind, and it is the Network’s hope that individuals with RP, their family and friends, and any other concerned individual, can access its contents easily.

After reading The Foundation Fighting Blindness Guide to Retinitis Pigmentosa and Related Conditions, you will likely have a few questions of your own. This publication is not meant to replace discussions with your family doctor, eye doctor or other professionals involved in your vision care. Please also understand that in the rapidly advancing field of eye research, some information may change even before this handbook gets to you.

Remember, you are not just a ‘case’ of RP. You are a unique individual with specific attributes that are yours alone: genetic makeup, environmental influences and lived experience. Every person’s RP is different.
Historical Overview

The term retinitis pigmentosa (RP) was coined by Frans Cornelis Donders, a Dutch pioneer of vision and psychological sciences, in the middle of the 19th century. He used these words to describe the distinctive appearance of the retinas of a group of patients suffering from vision loss. The term “retinitis”, meaning inflammation of the retina, is somewhat misleading as RP is not an inflammation, but an inherited, slow-paced degeneration of the retina. The term “pigmentosa” refers to the characteristic pigment clumping seen on examination of the retina in many forms of RP.

Today, RP refers to a large group of inherited degenerative diseases of the retina. It has been found to occur in men and women of all races and ethnicities. It even occurs naturally in some animals.

RP is a relatively rare eye disorder. It is estimated that as many as one in 2,500 to 4,000 have some form of RP. There are good reasons why it has been difficult for scientists to calculate an accurate number - including the extreme variability of the disease, the large number of genes involved, the many types of RP and a lack of patient registries. Although RP had been recognized as an eye disease for many
decades, the first Canadian organization to commit to finding a cure was the RP Research Foundation of Canada which came to life in 1974. It was conceived by parents who had children diagnosed with various forms of RP.

**Research with a Goal**

As the Foundation evolved, it developed policies to ensure the integrity of the research it funds – it was to be relevant, worthwhile and have a high likelihood of success. A Scientific Advisory Board composed of first-rate scientists was assembled. Its membership has continually included elite researchers representing universities across Canada. As the availability of funds grew, a formal application process modeled on the Medical Research Council of Canada (currently the Canadian Institutes of Health Research) was instituted. Applications were peer-reviewed, evaluated, rated and ranked. The rigorous nature of the review process helped to ensure the reputation of the Foundation as it grew from a very small health charity into one that now funds more than $1 million for research projects each year.

Over the first 30 years of the Foundation’s history, much was learned about the complexity of vision, the retina and the genetics of eye diseases. In order to increase its public visibility and add momentum to its search for cures, the Foundation launched itself into the 21st century with a new name. As The Foundation Fighting Blindness (FFB), it took on the role of Canada’s leading health charity supporting retina research, with priorities that included finding the causes, treatments and cures of macular degeneration, retinitis pigmentosa and related diseases of the retina.
During the course of its evolution the FFB has witnessed significant advances in vision research, particularly in the area of genetics. The explosive growth of biotechnology and the almost unbelievable speed at which the Human Genome Project progressed has certainly helped in the identification of disease genes. In this fertile environment the FFB is committed to funding researchers who push the frontiers of known science forward so that treatments and cures will arrive as quickly as possible.

The ongoing success of the FFB rests squarely on the shoulders of its volunteers who raise funds for the organization. It is a diverse group that includes a robust and highly motivated Board of Directors, and individuals who host and work at fund raising events.

The goal of saving sight is shared by an increasing number of partners. The FFB would like to acknowledge the significant contribution of government and academic partners, corporations and individuals: without their support, we would not have been able to achieve our current level of success in forwarding retina research.
Acknowledgements

The FFB thanks the many individuals who contributed to *The Foundation Fighting Blindness Guide to Retinitis Pigmentosa and Related Conditions*. Volunteers are the backbone of the FFB, and it is their dedication that moves a project like this forward. But most of all, we are grateful for the fellowship, generosity and good humour provided by these members of the RP community. They truly shed a light of their own.

A debt of gratitude is owed to Dr. Alex V. Levin, MD, MHSc, FAAP, FAAO, FRCSC, who has made his medical expertise available and has a gift for making complicated information easy to understand. Dr. Levin is a paediatric ophthalmologist and full-time staff member at The Hospital for Sick Children in Toronto. He is a Professor in the Departments of Paediatrics, Genetics and Ophthalmology and Vision Sciences and Director of Postgraduate Bioethics Education at the University of Toronto and Co-Director of the Ocular Genetics Programme at The Hospital for Sick Children. He is a key Medical Advisor to the FFB.

Other important medical contributors are Dr. Elise Héon, MD, FRCSC, Dr. Nidhi Lodha, MD, Dr. Ian M. MacDonald, CM, MD, FRCSC, and Dr. Johanne Robitaille, MD, FRCSC.
Medical Aspects of Retinitis Pigmentosa

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The retina is the light-sensitive tissue that lines the inside of the back of your eye. The retina acts like film in a camera – it takes light from the world we view and turns it into electric signals that are sent to the brain where they are perceived as vision.

The choroid is the tissue that lies under the retina, between the retina and the outer white coat, or the sclera, of the eyeball. The choroid is one source of blood, bringing oxygen and nutrition, to the retina. The other source is the blood vessels of the retina.
Retinitis pigmentosa is a term used to describe the many forms of eye disease that affect the retina. RP is caused by abnormalities or mutations in specific genes. These genes make products – proteins - that are important for the very complex function of the retina. When a gene carries a genetic change or mutation, it either makes a protein that doesn't perform correctly or perhaps none at all. As a result, the retina begins to function poorly and degenerates, or dies. Depending on which gene is abnormal, many patterns of retinal abnormalities can be seen by an eye doctor.

Another key feature of RP is that it progresses. An affected individual may notice over time symptoms such as:

- difficulty seeing in dimly lit conditions;
- reduction or loss of night vision (nyctalopia);
- loss of peripheral or side vision that eventually progresses so that the patient can only see as if looking through a tiny a tunnel; and, usually lastly,
- blurring of central, or straight-ahead vision.

When examining the retina, an eye doctor may observe:

- attenuation (thinning) of the retinal blood vessel;
- atrophy (degeneration) of the retina or the underlying choroid tissue; and/or
- pigment clumping;
- pallor (whitening, weakness) of the optic nerve.
The results of an electroretinogram (ERG, see page 43) recording will be abnormal in RP. An ERG measures the amount of electrical activity generated by retinal cells. It is a measure of how well the retinal is functioning and whether certain parts of the retina work better than others.

Many people take part in diagnosing and caring for patients with RP: ophthalmologists (medical physicians and surgeons specializing in eye disease), optometrists (graduates of programmes specializing in diagnosis of eye disease and low vision management), opticians (makers of glasses), genetic counselors, social workers, low vision workers, nurses, teachers, visual electrophysiologists (who do special testing such as ERG, see page 43), ophthalmic photographers and imaging specialists, geneticists, ocular geneticists (ophthalmologists who specialize in RP and other genetic diseases of the eye), retina ophthalmologists, paediatric ophthalmologists, ophthalmic assistants and technicians (those who help ophthalmologists and optometrists with patient eye exams, such as performing vision tests or visual field tests), and the many support personnel (coordinators, administrative assistants) that help the eye care team function.
What Causes RP?

Every cell in the human body contains DNA. Our DNA is packaged into 23 pairs of strings called chromosomes for a total of 46 chromosomes in every cell. Each pair is made up of one chromosome from the person's mother and one from their father. The chromosomes are numbered one through 22. Each number represents one pair – for example, we have two copies of chromosome one, two copies of chromosome two and so on. The final pair are called the sex chromosomes. Females have two X chromosomes (XX) and males have one X and one Y chromosome (XY).

Our DNA contains the blueprint or recipe for everything about us: the colour of our skin, our height, and the function, shape, and size of every body part including the eyes. Each chromosome contains many genes, each of which makes a specific product – protein - that serves a specific function in our body. We have approximately 35,000 genes. Although every cell in our body has two copies of every gene, only certain genes are used in certain places whereas other genes may be used in more than one place. If a gene is only used in the eye, then an abnormality in that gene will only cause an eye problem – for example, RP. But if the gene is used in more than one place in addition to the eye, then an abnormality in the gene could cause an eye problem as well as other problems, such as hearing loss or extra fingers (see page 24).

It appears that all forms of RP are caused by defects in either one or both copies of a single gene. Usually only one gene is involved but in rare instances, more than one gene can be abnormal. Over 75 different genes may cause RP or similar disorders.
It is likely that hundreds of genes are in some way involved with allowing us to see. These genes may function in a number of ways. They may deliver the instructions to make the protein building blocks that form a retina, create the chemistry of turning light into electrical signals that go to the brain, keep retina cells healthy or construct the cells around the retina needed to help the retina stay alive.

A defect or mutation in any of these genes may cause malfunctions in retinal health, and therefore can lead to a breakdown of retinal function which results in the eye examination symptoms and findings that we call RP. Although gene testing is not available for many forms of RP, with recent advances in biotechnology, it is hoped that many more patients will have access to the knowledge of exactly which gene is causing their RP.

This picture shows the 46 chromosomes from a human cell. Each cell has two pairs of the 23 different chromosomes, except for males who have one X and one Y chromosome. Each chromosome contains thousands of genes which are too small to be visible.
A Word About Secondary RP

There are uncommon forms of RP called “secondary RP” which are not inherited. These forms of RP may be caused by an infection such as congenital rubella or German measles, vitamin malnutrition such as severe vitamin A deficiency, drug treatments, immune disorders, or injury to the retina from a physical trauma.

Secondary RP tends not to be progressive and is not transmitted to children via genes. Sometimes secondary RP can be treated, but this is not usually the case. Your eye doctor can distinguish secondary RP by carefully examining your retina for characteristic signs, asking specific questions about your medical and eye history, and learning about other things like your background, family history and diet. Blood tests may also be helpful in making the correct diagnosis.

How do you know what type of RP I have?

The different types of RP are often classified by inheritance patterns, age of onset, absence or presence of non-eye disease and retina appearance or ERG results. There may be overlap between each of these groups.
Types of Inheritance

Determining the type of inheritance for RP usually comes from taking a family history or family tree, also known as a pedigree, and analyzing the patterns by which the RP is transmitted to various family members. Occasionally, the eye examination or symptoms may also be useful.

The definitive answer comes from identification of the exact mutation in a specific gene. This is done by a blood test or a swab from the inside of the cheek inside the mouth. Once the gene defect is identified in a patient with RP, the same blood test can be done on other family members to see where the gene abnormality came from and how it is being passed or not passed from one person to their children.

Gene testing is becoming increasingly available but there are two obstacles to developing efficient gene testing: cost and knowledge. Governments and insurance companies are challenged by the increasing cost of new gene tests for all kinds of diseases. Additionally, we do not know many of the genes that cause RP. That means for many patients, blood tests can not yet identify the abnormal gene. Even when the defective gene for an individual’s RP is identified, the symptoms and severity may vary depending on what part of the gene has the mutation.

Another advantage of gene testing is that it allows researchers to understand the disease process, how the RP occurs, and as a result, possible avenues for eventual treatment. The discussion on the following pages is a brief outline of types of inheritance. It is strongly recommended that patients and families discuss their concerns about inheritance with a qualified healthcare professional such as an ophthalmologist, ocular geneticist, geneticist or genetic counselor.
**Sporadic RP (also known as RP Simplex)**

This is the most common form of RP. Many patients with RP are the first member of the family to be affected. This type of RP is genetic and therefore potentially heritable: meaning that the person with RP may still pass it on to their children or grandchildren. The person with sporadic RP may have gotten the RP from one or both parents who were carriers. Carriers have no symptoms of RP and usually have no idea they are carriers until someone in their family gets RP. For patients with sporadic RP, the inheritance category cannot be determined from the family tree since no other family members are known to have RP or carry an RP gene. To figure out the inheritance pattern, one must either use genetic blood testing, other clues from the history and examination, or wait to see if other family members eventually get RP.

**Autosomal Dominant**

Individuals who have autosomal dominant RP, whether they are male or female, have an abnormality in one copy of their defective gene. The other copy of that gene is normal. Since we only can pass one copy of each gene to a child, a person with autosomal dominant RP has a 50% chance with each pregnancy of having a child who will get RP. Males and females are affected equally because autosomal dominant RP does not involve genes on the X or Y chromosomes (sex chromosomes). The symptoms show a wide range of variability and severity but in general, patients with autosomal dominant RP are affected less severely – that is, they have later onset of symptoms and better retention of vision for more years than those with other types of RP. The severity of RP may vary between family members, but every person who has the abnormal copy of the defective gene usually shows some signs of RP eventually.
Autosomal Recessive (ARRP)

Both copies of the causative gene are defective in individuals with autosomal recessive RP. Usually, the person with RP got one defective copy from each parent. The parents are called carriers because they each have only one copy of the gene that is defective. Carriers of ARRP usually have no symptoms of RP, experience normal eye exams and have no idea that they carry an abnormal RP gene.

Every person in the world carries at least six to eight abnormal genes for autosomal recessive diseases. We never know which genes are abnormal unless we happen to have a child with a partner who by chance carries an abnormal copy of the very same gene.

If each of the partners gives their defective copy of the gene to a child, then the child has two defective copies and shows the disease, such as with RP. When two carriers conceive a child, there is a 25% chance with each pregnancy of having an affected child. Autosomal recessive diseases usually affect males and females equally.

Individuals with autosomal recessive RP rarely have affected children as they can only pass on one of their two abnormal copies of the gene. They would need to have a child with a partner who happened to carry an abnormality in the same gene. The chances are remote that they would marry a carrier of the same defective gene unless they marry a relative (consanguineous) or someone from the same small village or tightly knit ethnic group, as the chance of such a partner carrying the same gene abnormality is much higher. Autosomal recessive RP tends to be a more severe form of the disease.
X-linked Recessive

X-linked recessive RP tends to be a severe form of the disease, often affecting individuals during childhood. In general, this form of RP affects males but it is carried by females. It is caused by an abnormal gene on only one X chromosome. Since females have two X chromosomes, the one with the normal copy of the gene acts as a “backup” and the woman is a carrier, usually with no symptoms, although rarely this can occur. Sometimes, even in women carriers with no symptoms, doctors can detect that they are carriers by eye examination or ERG testing. Each time a female carrier of RP has a female child there is a 50% chance that the daughter will be a carrier, but no daughters of the carrier get RP. Each time a female carrier of RP has a male child there is a 50% chance that the son will get her defective X gene copy and therefore have RP. The sons and daughters who do not get the defective gene will always be normal, as will all of their descendents. A male who has X-linked recessive RP can not pass the disease on to any sons as sons get their Y chromosome from the father – in other words, they can’t get his defective X gene for RP. All of the affected father’s daughters will be carriers meaning that their male children could show RP. This sometimes gives the illusion that RP “skips generations”.

Autosomal Dominant  
Autosomal Recessive  
X-linked Recessive

square = males  circle = females  square filled = males affected w/ RP  circle filled = females affected w/ RP

Horizontal line connecting a male and female = marriage, Vertical lines = offspring
X-linked Dominant

This is a very rare form of RP in which women are affected. If a male inherited the defective gene, he would either die before birth or have a much more severe form of RP than the women in his family who have RP.

Mitochondrial

Mitochondria are the tiny parts of our cells that make energy for the cell to function. Each cell has many mitochondria and each mitochondria has its own DNA which is different than the 46 chromosomes found elsewhere in the cell. Mitochondrial RP is usually accompanied by other non-eye abnormalities. This form of RP, like the autosomal dominant and autosomal recessive forms, affects males and females equally. However, mitochondrial RP is only passed from mothers to their children. Affected men never pass this disease to their children.

Digenic and Multi-allelic

Some patients get their RP from abnormalities in more than one gene. In digenic RP two different genes are abnormal in the same individual. In multi-allelic - for example, tri-allelic RP, there may be two abnormalities in one gene plus another abnormality in one or more additional gene copies. Multi-allelic RP is associated with Bardet-Biedl syndrome (see page 25). Calculating the chance of having an affected child in these types of RP is much more complex and requires detailed counseling.
Age of Onset

Another way of categorizing RP type is based on the age that symptoms appear. This is called age of onset.

**Congenital RP**

There is a large group of RPs which are present at birth and are usually characterized by poor vision and virtually no recordable electrical response on the ERG test (*see page 43*). This group of disorders is often called Leber congenital amaurosis (LCA). Most of these patients have an autosomal recessive form of RP, although rarely autosomal dominant patterns have been described. Perhaps due to the severity of vision loss at birth, children do not experience as much progressive worsening with age as do children with other forms of RP. The retina examination can be quite variable, ranging from a normal appearance, despite very poor function, to a typical RP appearance (*see page 14*). LCA has been one area of genetic testing in which there have been many genes identified, thus opening up greater possibilities for exact diagnosis and perhaps someday treatment.

**Juvenile RP**

Juvenile RP simply refers to the onset of symptoms, abnormal ERG, and eye examination findings during childhood, but not as severe as congenital RP. All different inheritance forms can appear in this group.
Adult Onset RP

These are the most common forms of RP and involve all inheritance types. Even when patients do not recognize their RP symptoms until adulthood, there may have been earlier signs of this slowly progressive eye disease that escaped their attention. The ERG is often abnormal long before a patient knows they have RP.

RP with Other Disorders

Another way of categorizing different types of RP is by the presence or absence of associated non-eye findings. RP usually occurs in isolation with only the eyes being affected. The list of such disorders is far beyond the scope of this guide. Below is a sample of disorders in which RP appears along with other abnormalities. Most of these disorders are very rare and may require a specialist to be identified. Careful attention to other body problems is the key element in making these diagnoses.

Usher Syndrome

There are many different types of Usher syndrome, all of which share the presence of RP plus varying degrees of hearing loss and sometimes balance problems. There are also other forms of RP, such as congenital rubella syndrome (see page 17), that have RP and hearing loss but are not Usher syndrome. Many genes for Usher syndrome have been identified.
Bardet-Biedl Syndrome

Bardet-Biedl syndrome is a complex disorder which may involve many bodily functions. There are at least nine different types. Although RP is always present, other symptoms may include varying degrees of developmental delay, extra fingers or toes (polydactyly), obesity, hormone abnormalities and kidney abnormalities in addition to other potential body problems. Different affected family members may have different forms of the syndrome. At time of writing, eleven genes have been identified.

Alström Syndrome

These children often have kidney problems, aversion to sunlight, obesity, hearing loss and RP. At time of writing, one gene has been identified.

Mitochondrial Disorders

There are many different types of this disorder with a variety of non-eye problems which may or may not be present. These include short stature, diabetes, hearing loss, eye movement problems, heart problems and RP. These disorders go by a number of names, each of which identify a particular pattern of body problems: Kearn-Sayres syndrome, NARP, MELAS, MERRF and others. See also “Mitochondrial” on page 22. Genetic testing is available for many of these conditions.
Appearance of the Retina

The many types of RP are sometimes characterized by what the retina looks like during eye examination or by what the ERG shows. Here are a few examples:

**Paravenous** or **Pararterial RP** are identified by the way that pigmentary clumping lines up along retinal veins or arteries.

**Cone-Rod Dystrophy** is a form of RP where the cone cells of the retina are more affected than the rods. Cones are located mostly at the central part of the retina, an area used for “straight ahead” vision and seeing fine details. Therefore, these patients may experience blurring of straight ahead vision even before getting night blindness or peripheral vision loss.

**Rod-Cone Dystrophy** is the opposite of cone-rod dystrophy. In this case the rod cells are affected more severely and earlier. This would be a more “classic pattern” of RP. Since the rods deal with night vision and peripheral vision, patients have these problems first while their central vision is usually preserved longer.

**RP Inversus** is identified when the pigmentary clumping is more towards the central back of the retina, rather than at the retinal edges, as would usually be the case in RP.

**RP Sine Pigmenta** is a term sometimes used when patients have all of the usual symptoms, electroretinogram changes, and eye examination findings of RP with the exception of the pigmentary clumping, which appears to be absent or minimal.
Frequently Asked Questions

General Questions

What is the difference between retinitis pigmentosa, retinal dystrophy, retinal degeneration, macular dystrophy and macular degeneration?

The term retinal dystrophy refers to any form of progressive degeneration of the retina caused by a genetic defect. This means that the disease has been inherited and can possibly be passed on to children. All forms of RP are dystrophies. However, there are many other retinal dystrophies that are not RP – for example, Stargardt macular dystrophy, choroideremia and Best disease. There are hundreds of retinal dystrophies.

When a retinal dystrophy affects just the central part of the retina - the macula - it is called a macular dystrophy. RP is not a macular dystrophy because the entire retina is affected, although the macula may become affected.
Retinal degeneration describes the process of progressive loss of function of the retina. Any retinal disease that gets worse over time is called a degeneration. All forms of RP are retinal degenerations. However, there are forms of retinal degeneration which are neither RP nor a retinal dystrophy. These would be disorders in which the retina degenerates over time for reasons not related to gene defects. For example, a long-standing retinal detachment.

When just the central part of the retina degenerates it is called a macular degeneration. Almost all macular dystrophies cause macular degeneration. However, the term macular degeneration is most often used to describe a specific disease of older adults called Age Related Macular Degeneration (ARMD). In addition to its work with RP, the FFB is a major supporter of ARMD research.

Will I go blind?

Complete and total blindness (loss of perception of light) is extremely rare in patients with RP, and not all patients will suffer severe visual impairment. On average only one in 1,000 patients will go totally blind. Whether or not any given individual will become legally blind (see page 30) will depend on the type of RP. Some forms of RP are milder with vision preserved well into later life. Other forms of RP are more aggressive with legal blindness occurring in the teens or 20’s, if not earlier.
Is there any way of predicting the course of my RP?

Just as every individual is unique in their genetic make-up and the environment in which they live, the course of RP in any given individual will vary. However, we can get some clues to an individual’s progression based on family history, type of RP and genetic testing.

If most affected individuals in a family maintain their vision to a late age, then it is reasonable to think that any given individual in that family might experience the same. However, this is not always the case and some individual family members may “defy the rule” set by their relatives.

Likewise, knowing the exact type of RP can help to identify forms of RP that progress more rapidly or more slowly. If we are able to identify the exact gene that is abnormal in a particular patient, as well as the exact mutation in that gene that is causing the RP, then one could compare any individual patient to other patients who have been found to have the exact same genetic mutation. However, even this method is not foolproof, as a given genetic mutation in one individual may have different effects than it does on another.
The term “legally blind” is basically an administrative term which defines a certain level of vision, below which an individual can access resources in the community, and other benefits that are designed to help individuals with visual impairment. It is a somewhat artificial designation that was created to do the greatest good for the most people with the optimum expenditure of resources.

Legal blindness is defined in Canada and many other countries as vision of 20/200 or worse in the better eye. An individual who sees 20/200 has to stand at 20 feet to see what a normal individual can see from 200 feet. The metric conversion is 6/60: a person has to stand at six metres to see what a normal person can see from 60 metres.

Another criterion for legal blindness has to do with peripheral vision. Even if a person’s straight ahead vision is better than 20/200 - even if it’s 20/20 - if the person loses enough peripheral vision so that they are seeing through a tunnel of less than 20 degrees, then they are declared legally blind.

Some patients do not exactly fit into either category, but may have a combination of visual field defects and a decrease in straight ahead vision which might allow the eye doctor to make a subjective designation of legal blindness.
Sometimes I see flashing white lights. What are they?

Flashing lights (photopsia) which occur spontaneously are often experienced in older individuals. As we age, the vitreous gel which fills our eyes begins to contract. As the gel contracts it pulls gently at the retina and stimulates it to cause the sensation of flashing lights. This is called a posterior vitreous detachment (PVD) and is seen in 70% of people by 70 years of age.

If a patient has RP, the contraction of the vitreous may occur earlier than normal. This might explain symptoms such as photopsia at a younger age. Your eye doctor can confirm this by examining your eyes and actually seeing the vitreous changes. This is usually a harmless phenomenon. However, if the lights seem to be increasing in frequency or number, are associated with the new onset of black floaters, or a sudden change in vision, then you should seek eye care urgently. These symptoms may be due to impending or present retinal detachment resulting from the contracting vitreous pulling too hard on the retina. This is a rare complication of PVD, even in individuals with normal eyes.

Does stress affect RP?

Many patients with RP report visual changes, often transient, when they are under stress. It is well known that stress can affect many physical body responses. All of us would like to minimize stress in our lives, although it is certainly impossible to eliminate it completely. There is no scientific evidence that shows that certain levels of stress cause an acceleration of the visual loss caused by RP. However, from what individuals with RP tell us, we can say that it is not unexpected if visual changes occur during high stress periods.
I heard that some patients with RP get swelling in their retina. What is this all about?

Patients with RP are prone to getting swelling in their macula, a condition known as **cystoid macular edema** (CME). The cause of the swelling is not known. Symptoms of CME may include visual loss or distortion of central vision (metamorphopsia). The decrease in vision occurs more rapidly than most patients with RP usually experience, and it affects straight ahead vision, not side or night vision.

An ophthalmologist can make the diagnosis of CME based on an eye exam although sometimes other tests, such as intravenous fluorescein angiogram (IVFA) or optical coherence tomography (OCT), may be necessary. For IVFA, vegetable dye is injected into a vein in the arm so it can flow to the eye via the bloodstream to enable special pictures to be taken of the eye, allowing the swelling to be better visualized. OCT is a type of photograph of the retina using a laser-like kind of light. OCT has the advantage of being painless. IVFA can sometimes cause an allergic reaction. Some patients with RP do not like IVFA because the bright light may leave them temporarily “blinded”. This usually reverts over an hour or two and causes no permanent visual loss.

The advantage of making the diagnosis of CME is that patients with RP, unlike other patients who may have CME, may respond to an oral medication, usually acetazolamide, also known by the name Diamox. This medicine can cause the swelling to go away and restore vision to the pre-swelling level, although it will not cure the RP. The treatment is only for this complication and not for the disease itself. In some patients, treatment does not work and permanent visual loss ensues. In a few instances, surgery may be recommended.
Will cataract surgery help me?

A cataract is an opacity, or clouding, in the lens of the eye. The lens sits directly behind the pupil. The pupil is the black hole in the centre of your iris. Light passes through the pupil and lens on its way to your retina. If there is an opacity in the centre of the lens of the eye, it may block some of the light as it gets to your retina. This causes a blurring of vision in addition to any blurring that the RP may be causing.

Cataracts which are not in the centre of the lens do not block vision and cause no harm to the eye. Removing these cataracts will not improve vision. However, if there is a central cataract, removing it may allow the retina to see to its best potential. If the retina is very sick (moderate or severely advanced RP), then the blurring caused by the cataract may be insignificant when compared to the blurring caused by the retina. In these situations cataract surgery is often not helpful.

Cataract surgery is particularly helpful when there is a central cataract blocking light getting to a retina which has a potential to see better than the cataract allows. Patients with RP often develop a small cataract at the centre of the back of the lens called a posterior subcapsular cataract (PSC). Particularly in patients who have very narrow central tunnels of vision, a very small PSC can cause even more disability than it might cause in a person with a healthy retina. This is because the cataract is blocking the tiny central tunnel of vision, whereas a healthy person could see around such a cataract. In these situations, removing the cataract earlier than one might remove it in an adult with a healthy retina may be useful.
Sometimes it is difficult to predict whether a patient with RP will benefit from cataract surgery. There are instruments such as potential acuity meters which use laser light passing through the cataract to help to determine what the potential for vision may be. This kind of laser light can pass through the cataract without being blurred and therefore depends only on how well the retina can see. This test, however, can be difficult for patients with RP to perform as it is done under low illumination and requires a field of vision that is big enough and vision that is good enough to see the laser light.

Like all forms of eye surgery, there are risks to cataract removal. One must weigh the potential risks versus the potential benefits in making a decision to proceed. These should be discussed with your cataract surgeon. In particular, patients with RP may have a higher risk of retinal swelling (cystoid macular edema or CME, see page 32) after cataract surgery. CME can happen in normal individuals, but may have a higher incidence in patients with RP. CME may lead to irreversible visual loss as a result of cataract surgery.

Will glasses help me see better?

Just like noses, hands and feet, eyeballs come in many different sizes even though these size differences are not large enough to be noticed by the casual observer. If an eyeball is not the “standard size” for normal vision (20/20), then glasses or contact lenses can be used to bend the light coming into our eyes so that it fits into our eyeballs more perfectly. They focus the light so that the picture we are seeing falls directly on our retina. This is a necessary step in transmitting the clearest message from the retina to the brain.
Individuals who are near-sighted (myopia), or farsighted (hyperopia) or astigmatic simply have eyes that are not of the standard shape or length. Therefore, the normal focusing of the eye does not allow the images we see to fall on the retina. Glasses help the eye to focus light in the right place. However, although glasses do give us our best vision, they do not fix anything that is wrong with the eye.

As an analogy, consider a camera. Let’s say you have just bought the best and most expensive lens that money can buy, and you have added it to the best camera available. What happens if you load the camera with bad film? Of course, you’ll have bad photographs when the film is developed.

A person with RP is like a camera with bad film. The retina in your eye is like film in a camera. If your retina is not working well, then glasses, even if they are perfect glasses which focus an image directly on the retina, are of little help. This is because the retina cannot send a clear, normal message to the brain, just like the bad film in the camera which will always give blurred pictures. Yet glasses will help some people with RP who are nearsighted, farsighted or astigmatic see the best that their retinas are able to see, even though that “best vision” may be much below normal. However, a low vision assessment may determine if special glasses can help for certain tasks like reading.

**Will my pregnancy impact my symptoms of RP?**

Women with RP who are pregnant may sometimes experience decreases in their vision due to swelling of the retina (*cystoid macular edema* or CME, see page 32) or perhaps hormonal factors. This may resolve after delivery of the baby, respond to certain medications, or at times may cause irreversible vision loss. There are no good studies that help us predict the risk of having this complication.
Do medications affect my vision? Is there any specific drug, vitamin, or food I should avoid?

Although there are many medications which can cause side effects involving the eyes in rare instances, very few are specifically not to be given to patients with RP. Patients with RP should bring their eye condition to the attention of their doctors before starting new medications.

There are no specific foods or vitamins that have been scientifically shown to have strong adverse effects on patients with RP. There is some weak evidence that a high dose of vitamin E could have a slightly detrimental effect, although this has been debated by various researchers, some of whom feel that vitamin E might even have a weak beneficial effect. There is not enough information or scientific data to make a recommendation for or against any particular substance, such as antioxidants, at this time.
Are there effective nutritional therapies for RP?

There have been many commercial companies, alternate caretakers (e.g. naturalists, homeopaths) and even the occasional physician who have recommended nutritional therapies for a wide range of disorders including RP. There is little scientific evidence at this time to support any of these treatments. Patients should be particularly wary of treatments which are proposed at high costs and with the suggestion that they can cure multiple unrelated diseases.

Multivitamin preparations are available specifically for patients with Age Related Macular Degeneration (ARMD, see page 28). These preparations have not been well tested for RP or related retinal or macular dystrophies.

As more genes which cause retinal dystrophies have been discovered, and the mechanisms by which they may be related to eye disease better understood, the potential for treatment increases. For example, one gene called ELOVL4 is involved with the way our body metabolizes fatty acids, an essential ingredient of our diet. Mutations in this gene may result in a retinal dystrophy (not RP) called Stargardt disease or other retinal diseases.

In September 2004, two articles appeared in the journal Archives of Ophthalmology. These publications represent the results of research done largely in the Boston area on patients over the age of 18 years and under the age of 55 years with RP. The lead investigator and author, Dr. Elliot Berson, who is a member of the Scientific Advisory Board of the Foundation Fighting Blindness in the United States, has interpreted these results to indicate that patients with RP should be taking both Vitamin A, and for certain periods during the course of Vitamin A treatment, also Docosahexaenoic Acid (DHA)
supplementations as well a diet rich in long chain-omega 3 fatty acids, found in fish such as salmon and tuna. Research from other centres has suggested that DHA may be useful in some cases. However, in this new study, when taken alone, DHA and omega 3 fatty acids had no significant effect on the progression of RP. The study did not look at patients who were only taking DHA and diets rich in omega 3 fatty acids without taking Vitamin A.

The second paper looked at subgroups of patients from the first paper and concluded that certain recommendations should be made regarding the diet, and DHA and Vitamin A intake for patients with RP. However, the FFB agrees with the National Institute of Health (NIH) that these findings should be interpreted with caution due to problems with the research methodology that makes it difficult to interpret the results accurately. In particular, the companion paper mixed small groups of patients with various kinds of RP making it difficult to say that there truly was an observed effect of the treatment as interpreted by the authors. RP is actually a collection of hundreds of different disorders based on different gene abnormalities and different mutations of any given gene.

It should also be strongly noted that the studies did not look at children under 18 years old, pregnant women, women who might get pregnant during the course of treatment, or patients who had RP with other body disorders other than deafness. As a result, these papers cannot be applied at all to these groups and in fact, the proposed treatments could be dangerous in these patients or even cause worsening of RP. Since they have not been tested, we don’t know the possible effects. In addition, Vitamin A can cause birth defects if taken by pregnant women. Excessive doses of vitamin A may be toxic for your eyes and body. At this time, the FFB does not recommend routine treatment with Vitamin A, DHA, or omega 3 fatty acids for patients with RP. We certainly
do support a well balanced diet including fish intake (especially fish rich in omega-3 such as salmon, tuna, mackerel, herring or sardines, one to two servings per week) in keeping with general health practices. We recommend that patients discuss these issues further with their health care providers.

**Are there alternative treatments or medicines available?**

There are many “alternative treatments” that have been offered to patients with RP from countries around the world and also from health centres in North America. There is currently no evidence that any of these treatments can cure RP. Be sure to discuss such treatments with your ophthalmologist or eye geneticist to see if there is scientific evidence to support their use before spending money and taking unknown risks. We all want to find the cure for RP, but not at the expense of our patients’ health and vision.

**What about alcohol and smoking?**

Recent research suggests that smoking can be damaging to the retina and may worsen RP. It is unknown whether alcohol is good or bad for the vision of individuals with RP.
Maintaining Eye Health

**Is there anything I can do to maintain the vision I have?**

There are no known environmental factors or activities which have been shown to have a direct effect on the progression of RP. Common sense tells us that eating a well balanced diet, and avoiding fad diets, smoking, and other “bad lifestyle habits” might affect our general health, which in turn could conceivably have an effect on our eyes. There is no scientific evidence regarding the effects of these factors specifically on RP.

A popular myth is that one can “overuse” your eyes. There is no evidence suggesting that excessive use of the eyes, whether it is for reading, television or computer work, has any adverse effect on patients experiencing visual loss from RP. There is some evidence that ultraviolet (UV) rays have an effect in causing a progression of either cataracts, Age Related Macular Degeneration (*ARMD*, see page 28), and possibly RP. Certainly, wearing sunglasses can not be harmful and may even help. Most inexpensive sunglasses give almost the same protection from UV light as the most expensive brands.

**How often should I visit my ophthalmologist?**

The frequency of visits must be determined individually for each patient based on the type of RP, age of the patient, progression of symptoms, other coincidental eye findings such as glaucoma, and the physician’s practice. Intermittent contact with a specialist in RP every two to four years might be useful in keeping updated on progress and new developments in the field. Otherwise, after the diagnosis of RP has been confirmed and your questions answered, your progress can be monitored annually by a community general ophthalmologist or
optometrist. If you hear or read about something new, feel comfortable bringing this to the attention of your eye doctor.

**Should I see a retinal specialist or will a regular ophthalmologist be adequate for my needs?**

There are two kinds of retinal specialists: surgical retinal ophthalmologists and medical retinal ophthalmologists. Medical retinal ophthalmologists and eye geneticists (ocular geneticists) are the two types of ophthalmologists who usually specialize in eye diseases such as RP. Ocular geneticists are often also paediatric ophthalmologists who devote part of their practice to seeing adults with genetic eye disease such as RP. These individuals are likely to be up-to-date with regards to ongoing research projects and new developments in the field. They may or may not provide routine care, such as eye check-ups and the prescription of glasses.

Some local ophthalmologists will also be comfortable in taking care of patients with RP and attending to their special needs, including arranging special tests (e.g. visual fields, ERG, see page 43) and treating problems such as retinal swelling. However, most local ophthalmologists are more equipped to provide your routine care (e.g. check-ups, eyeglass prescriptions, treatment of other eye diseases such as glaucoma), and may wish to refer you to a medical retinal ophthalmologist or eye geneticist for concerns or problems specifically related to your RP.

There are many more ophthalmologists than eye geneticists. Therefore, it may be wise and more convenient to make your regular visits to your local ophthalmologist with visits every two to four years, or as needed or recommended, to the eye geneticist or medical retinal ophthalmologist.
Why have appointments with eye doctors if they can’t do anything to improve my sight?

Your community eye doctor has many jobs in addition to caring for your RP. Until there is a cure for RP your eye doctor will continue to make sure you are seeing the best you can. Your ophthalmologist will ensure you have the proper glasses prescription, screen you for related or unrelated eye problems such as glaucoma, make sure that you are not developing new eye problems related to your RP such as CME (see page 32), and along with your retina specialist or eye geneticist, keep you current on significant research and eventually on treatments when they become available.

Your eye doctor will also attend to other needs related to your visual loss, such as registration with the CNIB. Regular visits will help you to avoid problems before they happen, allow detection of new problems earlier, and keep you abreast of the latest RP news.

Will laser eye surgery help me?

There are two kinds of laser eye surgery that one hears about frequently. Lasers can be used to help some patients with bleeding in their retina due to Age Related Macular Degeneration (ARMD, see page 28). More often we hear about lasers to help people stop wearing glasses. This procedure uses a laser to cut the front surface of the eye so that it can focus the light itself rather than using glasses or contact lenses. This is not a treatment for RP and there is no laser treatment for RP.
Diagnostic Tests

What is a visual field test?

Because patients with RP often lose their peripheral vision, a visual field test can be performed to assess how much has been lost. There are two general ways in which this test can be performed. In both types of field tests, lights are presented briefly to the patient, whose chin is resting on the inside of a large, upright white bowl while looking straight ahead. The patient must indicate whether or not the light was seen and when.

The Goldmann visual field test is a manual test. A technician presents each light individually, often interacting with the patient as the test proceeds. The Humphrey field test is a computer-automated field test.

Although each doctor may have a preference as to what test they use to measure the visual field of a patient with RP, at our centre we find the Goldmann test to be most useful.

Visual field tests are painless and do not involve eye drops. However, patients sometimes find them time-consuming and frustrating. Remember, the technician or computer is able to judge how well you’re doing so that your eye doctor can assess for the reliability of the test in giving you feedback about your results. These tests have no effect on the eyes.

What is an ERG?

ERG stands for electroretinogram. It is an eye test used to measure the electrical activity in the retina. The retina is the light sensitive lining inside the eye which makes a picture of what we see, like the film in a camera. The ERG is used to
test how well the retina is working when a disease of the retina is known or suspected. Sometimes an ERG is performed on the relatives of people who have a retinal disorder. Most hospitals and eye doctors do not do this specialized testing. It is usually done in a designated centre, often at a university hospital.

Some patients find the test uncomfortable although it is not painful. A large contact lens is placed on the eye. This special lens has an elevated edge which also keeps the eyelids apart. Flashes of light of various colours and intensity are used to test the retina. One or both eyes may be tested. The test usually starts with a period of approximately one half hour when both eyes are covered by black patches to keep out all light. The test is then performed in a dark room. If a child is not able to sit still for this test, it can be done while the child is asleep under anesthesia or with a sleeping medicine (chloral hydrate) in a clinic setting.

**What is focal ERG, pattern ERG and multi-focal ERG?**

These tests are very similar to a regular ERG, but there are a few differences. They measure the electrical activity present in centre of the retina, called the macula. The macula is the part of the retina which lets us see straight ahead, as well as fine detail and colours. The tests differ in the kind of light that is presented to the eye, which in turn allows the eye doctors to understand which parts of the retina are working better than others. These tests are usually performed in a university hospital setting.

**What is an EOG?**

EOG stands for electro-oculogram. It is a test used to measure the electrical response of the layer of cells on which the retina sits: the retinal pigmented epithelium (RPE). The EOG is used to test how well the cells under the retina are working when certain diseases are known or suspected. Sometimes an
EOG is performed on the relatives of people who have a retinal disorder. Examples of eye disease in which the EOG is often abnormal include Best disease, Stargardt disease and RP.

Most hospitals and eye doctors do not do this specialized testing. It is done usually in a designated centre, often a university hospital.

**What is a VEP?**

VEP stands for visual evoked potential. It is a test used to measure electrical activity of the vision pathways, in particular the nerves which carry the message from the eyeball to the brain, similar to the way ECGs (electrocardiograms) are used to measure the heart’s activity. By analyzing the electrical signals recorded from the vision centres in the brain, we can find out how well the visual system is working. A VEP is a way of telling if the vision messages are getting from the eye to the brain properly.

Most hospitals and eye doctors do not do this specialized testing. It is done usually in a designated centre, often a university hospital.

**What is OCT?**

OCT stands for optical coherence tomography. This is a highly magnified photograph of the retina. The picture is taken using a harmless laser light. It is painless and takes a few minutes. It is particularly useful in detecting retinal swelling or other abnormalities (*cystoid macular edema or CME, see page 32*).

**What is autofluorescence testing?**

This is another type of retinal photograph using painless and harmless
laser light. It is used specifically to detect abnormal waste products that can build up under the retina if it is not working properly.

**What is genetic testing?**

Genetic testing aims at identifying the many genetic changes that can cause RP. Results from genetic testing may be inconclusive as not all the genes have been identified yet. In order to have genetic testing done, one must donate a blood sample from which DNA will be extracted. The ophthalmologist will help choose which are the best genes to start testing based on your eye examination.

If a doctor is able to determine which exact mutation is causing the RP, it is more likely that some prediction of the course and progress of the disease can be made. It would also provide the doctor with important information for providing genetic counseling to the patient. In the future, gene therapy will be available but will require knowledge of the specific gene causing RP in a patient.

**What is a diagnostic chip?**

RP is caused by genetic abnormalities. Detecting which gene has the mutation that causes the RP in a given patient is sometimes possible. Diagnostic chips, also known as microarrays, are a new technology that allow a laboratory to look for many different possible gene abnormalities at one time rather than the more expensive and time-consuming way of testing one gene at a time. There are many types of diagnostic screening chips and it is a rapidly evolving area of biotechnology. The FFB is supporting the development of screening chips for specific forms of RP.
Why are there so many people in the room while I am being examined?

Most likely, you are describing a situation where your eyes are being examined in an academic, university health science centre. In these situations, there may be medical students (learning to be doctors), residents (medical doctors training to be ophthalmologists), or fellows (ophthalmologists learning a specific subspecialty such as retina, paediatric ophthalmology or eye genetics).

Teaching hospitals have three goals: clinical care, education and research. Therefore, in these centres, there may be several individuals, such as these students, who are trying to learn about RP so that they can deliver proper care to others. It is important to remember that the physician who is taking care of you learned in the same fashion as the students who are now in the room. If we don’t allow these individuals to have an educational experience that involves your clinical care, then we won’t have enough doctors who can take care of patients with RP (and we already have too few!).

Keep in mind, that although RP may seem common in your eye doctor’s office - particularly if your eye doctor is a specialist in RP - it is relatively uncommon when compared to other conditions such as crossed eyes, cataracts or glaucoma. Therefore, you and your eyes represent a special and unique opportunity to teach individuals, such as these students, about RP. However, it is always your choice as to who may or may not be in the room, and how many times you may or may not want to be examined. If you feel uncomfortable in any way, you should discuss this with your eye doctor. In addition, we strongly recommend that you ask who is in the room, particularly if your eyesight is not good enough to allow you to identify all the individuals. Don’t be embarrassed. It’s your right to know.
Scientists funded by the FFB have been responsible for many internationally celebrated advances in eye research.

RP research is currently taking many forms at centres throughout the world. Not every centre does research in every area. Although many of the ideas discussed on the following pages are promising, we are still a bit away from offering treatment to patients. You can always ask your ophthalmologist or eye geneticist about ongoing research and whether there are projects that would accept you as a patient. However, one should always be cautious about making vision worse while trying to make it better.

Clinical research is done to understand the effect of disease on patients. As has been the case for many years, eye doctors are always trying to learn more about the different types of RP, the course of visual loss, possible environmental risk factors, the influence of RP on lifestyle, and other factors which impact on the daily lives of individuals with RP. This research is so named because it directly involves patients or their medical records.
Genetic Research

Identifying genes which cause RP has been a major thrust of research over the last few years, and has yielded the discovery of several genes, although there are still many to be discovered. By identifying the genes that cause RP, we can begin to understand why it occurs. This is a critical step in planning possible treatments.

In addition, identifying genes, and abnormalities or mutations within these genes allows for better sub-classification of the different forms of RP. This may also help give individuals an idea of what the future course of their visual loss might be. Identifying an abnormal gene also allows individuals to make reproductive choices if desired, as the recurrence rates, carriers, and even affected individuals with no symptoms, could be identified.

As gene therapy becomes available in the future - and it is already being done with some good results in animals - it will be essential to know what gene abnormality is causing a patient’s RP before they would be able to have gene therapy.

Treatment Research

There are many different approaches that are being researched in the hope of either restoring visual function to patients with visual loss secondary to RP or preventing or halting further visual loss from occurring. At the time of this writing, there is no treatment for RP and the few experiments being conducted in humans are reserved for those with very sick or completely blind eyes. However, more human treatment experiments will be coming in the near future.
Stem Cell Research

Stem cells are special cells in our body which have the potential to become many different kinds of cells. They are like babies who can grow up into different kinds of people based on how they are raised. Some stem cells can grow into new retina cells. One possible avenue for treating retinal diseases like RP would be to inject stem cells under the sick retina and hope that new healthy retina cells would grow and maybe even restore vision.

While stem cell research may give rise to treatments for a variety of chronic diseases and injuries, it is still a very new field. Scientists first have to learn the basics of how stem cells work before they can apply that knowledge to specific diseases, and in turn, develop treatments. As a result, stem cell therapies for RP will probably not be available for several years.

The FFB has an ongoing commitment to fund stem cell research and has supported scientists who are leaders in this field.

Gene Therapy

If the gene which is causing the RP in an individual is known, then one might approach treatment by: fixing the gene, replacing the gene, turning on compensatory genes, turning off contributory genes, or replacing the gene function.

One of the obstacles that still remains regarding gene therapy is trying to localize the effect to the eyeball without affecting other genes in the body. In addition, if a new gene or medication were to be delivered directly to the eye, it would have to be done in a way that would not harm the eye.
Gene therapy has been successful in some non-eye diseases and a clinical trial is being initiated in the United States for one very specific form of childhood RP. It is yet to be seen if this treatment will be effective in humans, although it has been shown to restore vision in one breed of dog affected by this form of RP. The first phase of this trial will indicate whether it is possible to safely deliver the new gene to the retina of a human eye. Only after it has been proven to be a safe treatment will the researchers test the gene therapy to see if it actually restores vision. Gene therapy has reversed ERG results and vision problems in several forms of genetic retinal disease in mice and other small animals, so there is much promise for humans in the future.

Artificial Vision

While at least one group has taken on the daunting task of trying to build a complete artificial eyeball (bionic eye), a more likely possibility for restoring some vision would be the implant of an electronic receptor computer chip onto the retina. The “chip” would send visual messages to the brain just like the retina does. Some experiments have been done in humans and even totally blind eyes have regained some ability to see large coloured shapes or lights. Real functional vision has yet to be achieved. In addition, one must be certain that the chip will not damage the retina to which it is attached.

Retinal Transplantation

Another approach to the treatment of patients with RP would be to give them a new retina. As the technology to peel off a retina from a donor eye and give it to the patient is not yet available, researchers are trying to inject cells, which
have the potential to grow a new retina \textit{(such as stem cells, see page 50)}, under the sick retina of a patient with RP. This research looks very promising although the benefit for patients with RP has yet to be shown. We know that if the patient does not reject the donor cells, it is possible for these cells to survive. What is unknown is whether they can actually form functional retinal cells that will restore vision.

In addition, injections under the retina may make the patient’s condition worse. Several laboratories around the world are trying to overcome these hurdles.

\textbf{Eyeball Transplantation}

Today, when a patient has an “eye transplant,” they actually only receive the cornea, the crystalline dome which covers the outside surface of the eye or for certain special operations, a piece of the white of the donor eye \textit{(sclera)}. Cornea transplant \textit{(penetrating keratoplasty)} can restore vision to patients with scars on the front surface of their eye but will do nothing for a patient whose problem lies in their retina.

Some laboratories are working on the possibility of transplanting an entire eyeball from a donor to a patient. Although this has never been tried on human beings, there have been some early successes in animals, such as rabbits and rats, that have indicated that it may at least be possible to get a donor eyeball to integrate into a new site. Much research needs to be done to know whether this can be done in humans and whether it will restore functional vision.
A major obstacle is whether we can find a way to make the optic nerve of the donor grow back to the optic nerve of the patient. Without that connection, messages from the new eyeball could not get to the brain. Some researchers are also working on artificial connections, such as by using wire.

Can I be part of a research project?

The term “research project” is very non-specific. There are literally hundreds of RP research projects going on around the world. One must carefully assess the risks of new research versus the potential benefits. In addition, one must be sure to enroll in a research project which is pertinent to your form of RP. If, for example, you have congenital RP (Leber congenital amaurosis), you might not be a candidate for a study looking for patients with adult onset RP.

Being part of a research project also requires informed consent. This means that it is very important that you understand exactly what you are getting involved with and what the potential for benefits or harm may be. Your RP specialist should know whether or not you are a candidate for a research project. Almost all RP specialists also collaborate with other specialists around the world who are doing research in various areas, as no single researcher can research everything!

The FFB is currently supporting the development of a central registry for patients with RP. This will allow researchers to better understand the clinical aspects of the disease and also offer research opportunities to specific groups of patients as it becomes available. The FFB attempts to stay informed about ongoing research projects that may help patients. Please contact the FFB if you have any questions.
Patients diagnosed with RP usually have many questions, but often don’t know where to start. Here is a list we put together that covers some of the basics, but you may have more you want to know. Do not be afraid to ask questions. If your doctor seems too busy and rushed to answer your questions, ask if you can submit them in writing before or after your appointment, or ask to book a separate appointment specifically to get your questions answered. Your doctor works for you so it is important that you get the information you need. Another strategy is to consult with your eye doctor’s support personnel or other resources in the office that might be able to help you get the time and answers you need.

Doctor, can you tell me…

• What is the name of my eye condition? Can you write that down?

• If I have no family history of this disease, how did I get it?

• Should other members of my family be checked to see if they have this eye disease?
Patients diagnosed with RP usually have many questions, but often don’t know where to start. Here is a list we put together that covers some of the basics, but you may have more you want to know. Do not be afraid to ask questions. If your doctor seems too busy and rushed to answer your questions, ask if you can submit them in writing before or after your appointment, or ask to book a separate appointment specifically to get your questions answered. Your doctor works for you so it is important that you get the information you need. Another strategy is to consult with your eye doctor’s support personnel or other resources in the office that might be able to help you get the time and answers you need.

- What is genetic counseling? Is it something I should look into? Can you refer me to a genetic counselor? Is the cost of this service covered by our healthcare system (or insurance where applicable)?
- What is my visual acuity (central vision)?
- What is my visual field (side vision)?
- Am I likely to lose more vision? If so, when?
- Are there any signs or symptoms I should watch for? What should I do if these occur?
- Are there any other tests that I should have?
- Are my medications affecting my vision?
- How do I see the best I can? Are there any types of glasses or lenses that might help?
- I’d like to see a retina specialist or eye geneticist. Can you refer me to someone who specializes in RP?
- Do I need a low vision evaluation? Where would I go for that?
- Can you recommend someone to talk to who has this condition as well?
- Do you have any patient information I can take home with me?
- Can you refer me to any organizations that provide services for this condition?
- Is there a central registry for people with this eye disease? How do I get in touch?
Adapting to Change

Attitudes ................................................................. pg.58
Managing Symptoms of Retinitis Pigmentosa ............. pg.60
In any society there are stigmas and taboos linked to disabilities, but you should not feel ashamed or guilty that you have vision loss from a genetic eye disease such as RP.

Rather, you must be your own advocate. It is important for you to take action and find out how you can best help yourself. Learn more about your eye condition – talk to your doctor or ophthalmologist; go to a support organization; and search on the internet.

Although the internet is a great source of information, remember that it is not regulated and it may be difficult to know if the information you read is accurate. Be sure to print the articles and information and bring it to your eye doctor appointment.

Adapting to changes in vision may present ongoing challenges for people living with RP. It is amazing what a little imagination and good old-fashioned common sense can do when it comes to overcoming obstacles. Brainstorm with friends and family, and access the resources offered by organizations dedicated to helping people with vision problems. In Canada, examples of such organizations include the FFB, the Canadian National Institute for the Blind (CNIB), and local organizations such as BALANCE.
Encourage your family members and friends to become involved in raising awareness about low vision and blindness. The FFB supports several fundraising activities throughout the year, the most well known being the coast-to-coast motorcycle rally, Ride for Sight. The FFB also hosts a conference, Vision Quest, every other year. If these events are not available in your community, consider creating a fundraising event and make it a family affair! You can also volunteer to assist others who are newly diagnosed - this may help you to adjust to your own vision loss.

Become familiar and comfortable with your limitations. Be creative and hopeful. When you need help from others, be specific: tell them where you need help and how they can assist. For example, crossing streets, finding the right bus, looking for specific items in the store, etc.

Connect with other people and support groups. If one is not available in your area you could start your own group. Talking and listening to others will give you more ideas on how to cope with vision loss. Contact the FFB to find a support network closest to your home.
The visual impairment caused by RP occurs in varying degrees in each individual. In general, action should be taken when any of the symptoms affect your daily activities. If this happens, the first step is to identify the cause of the symptoms with the help of a qualified vision care professional.

A regular eye examination – usually once a year - by an eye doctor, can help determine the medical cause of the vision problem. Depending on the cause and how much the symptom interferes with your activities, you may or may not need referral to a specialist in RP. It may be helpful to have a low vision assessment to help you learn how to get the most out of the vision you have.

Your ophthalmologist can help direct you to local services, as these differ greatly from region to region. There are many high- and low-tech devices specially designed to maximize vision. Exploring these options, as well as adapting your working, learning and living environments may greatly improve your quality of life.

The following is a list of symptoms frequently experienced by individuals with RP. The common causes are listed below them, along with what can be done to reduce the impact on your sight. The options listed are not exhaustive. Keep in mind that how each symptom is managed needs to be tailored individually.
Night Blindness

**Cause**

RP affects mainly the rod cells of the retina. These cells are important for night vision. Night blindness progresses slowly and gradually over time.

**What can I do?**

- Use good lighting source (*See section on glare on page 64*)
- Keep flashlight handy, use nightlights in house
- Low vision aids: Night vision aid, pocket scope, filters to decrease glare and improve contrast vision
- Wide angle mobility light (WAML)

Loss of Peripheral or Side Vision

**Cause**

Peripheral or side vision relies on the proper function of rod cells in the retina. RP causes the slow and gradual loss of rod function. The term **tunnel vision** refers to the total loss of peripheral vision, with only central vision remaining.

**What can I do?**

- Practise scanning skills.
- Field-enhancing devices work for some people, but not everyone.
• Mobility training.
• If there is a severe loss of side vision, adopt the use of a white cane to avoid personal injury. Consider using a guide dog.
• Special prism glasses have been used for some RP patients with poor side vision. The glasses widen the vision field, but they may be difficult to get used to.

Loss of Reading or Central Vision

Causes

People with RP may experience a decrease in central vision for a number of reasons. Sometimes, people with RP have reduced vision that may be caused by a problem that is correctable, such as retinal swelling (cystoid macular edema or CME, see page 32).

All of the causes of vision loss listed below are slow and gradual. Some causes are correctable and some are not, and more than one cause may be present at the same time. It can be difficult to distinguish between the correctable and non-correctable causes of the vision loss.

1) Dysfunction of cone cells. Central or reading vision relies on healthy cones in a specialized area of the retina called the macula. The extent to which central vision is affected in those who have RP varies much from one person to another.
2) **Cataracts** *(see page 33)* may develop at an early age in people who have RP – for example, in their 30’s or 40’s. Cataracts result from the clouding of the lens of the eye and prevent images from reaching the retina properly.

3) **Refractive errors** or the need for glasses are common in all people. Blurring may result if these errors are not properly corrected.

4) **Macular edema,** or the thickening of the retina in the macula, causes blurring of vision from swelling to the portion of the retina responsible for reading vision.

**What can I do?**

- A regular annual eye examination by an eye doctor will help determine how your vision is doing. If you experience a more sudden loss of vision, see your eye doctor urgently.
- Glasses may be required and prescribed
- Cataracts may be surgically removed in some cases *(see page 33)*
- Macular edema sometimes responds well to medication *(see page 32)*
- Low vision assessment for loss of vision due to RP damage *(see page 43)*
- Optical aids such as a telescope or magnifiers
- Non-optical aids: Proper lighting *(see section on glare on page 64)*, large print typewriter, large-size font on computers and Braille
- Electronic/High Tech: CCTV, reading machines, talking computers/calculators, scanners and software

**If you experience sudden loss of vision, you should contact your eye doctor immediately.**
Glare and Light Sensitivity

Causes

Glare or light sensitivity may be associated with low vision or with cataracts. Sometimes patients with RP have sensitivity to lights because their retinal cells are not working properly.

What can I do?

• An eye examination will determine the cause of the problem. A cataract may be removed surgically (see page 33).
• Tinted lenses may be helpful for both indoor and outdoor use. Amber or reddish brown are most popular, but the colour and darkness (how much light goes through the lens) depend on their use and an individual’s preference. The colour and darkness of the most suitable lenses may change over time. Corning lens are particularly designed to filter the kind of light that causes light sensitivity (photophobia) in patients with RP. Ask your eye doctor or optician for more information if you suffer from photophobia.
• Reduce light coming from sides by using either a broad-brimmed hat, visor or side shields on glasses. Some glasses have a rim on the top as well as side shields.
• Adapt lighting sources. Avoid or modify conditions contributing to glare such as direct light through optical devices, fluorescent lights, daylight room windows, computer or CCTV monitors, glossy books and magazines, and glossy surfaces. You may want to use a rheostat on lights, a spring-balanced lamp, different lamp and lighting systems for specific tasks, coloured acetates, and magnifiers with lights.
Reduced Contrast Sensitivity

Cause

Difficulty seeing contours of objects when placed against similarly coloured backgrounds is a common problem in people with low vision and retinal diseases.

What can I do?

Your eye doctor may be able to measure your contrast sensitivity with this special chart

- **Tinted lenses** may improve contrast and help in various environments. The tinted lens may improve your ability to distinguish a sidewalk curb or the edges of concrete steps.

- Whenever possible, choose a sharply contrasting background to view an object. For instance, use white plates on a dark tablecloth, pour milk into a dark cup and use black ink on white paper.

*Pelli-Robinson Contrast Sensitivity Chart*
Variable Visual Function

Cause

A common complaint in people with eye problems is fluctuations in vision. Changing lighting conditions such as cloudy or sunny days, and the presence of many large windows in a room such as a classroom, can alter how well anyone functions visually. Even stress may play a role (see page 31). Variability in symptoms does not mean that your condition is necessarily getting worse. It is just a part of having RP.

What can I do?

• Identify what worsens and improves your vision. Change the environment to accommodate your needs whenever possible. For example, put curtains, blinds or shutters on windows, or change the wattage/strength of light bulbs.
• Keep tinted glasses or a flashlight handy and consider tinted glasses for indoor use.
What are the best conditions for reading?

In a recent study, a group of researchers (Drs. Elise Héon, Carol Westall, Michael Brent and Nidhi Lodha) working at The Hospital for Sick Children in Toronto found that the reading ease of individuals affected by RP and other eye conditions causing low vision can be greatly increased by modifying lighting conditions.

Depending on the individual visual condition, lighting may affect visual acuity, visual efficiency and the field of vision available for the task. The research shows that by assessing the individual's lighting needs, it is possible to couple visual aids and correction strategies to specific personal tasks such as reading bills and writing cheques. The study was using strong halogen lights up to 400 watts. Results showed an improvement of about four to five lines on an average reading chart. You might want to try this with the help of your optometrist or low vision specialist.

Go to your local hardware store and ask about halogen bulbs and light fixtures. A halogen bulb emits a bright, white light and generates a lot of heat. They are not like regular, incandescent light bulbs and do not screw into a regular lamp socket. Other options are the new, low-energy compact fluorescent light bulbs. These can cast a lot of light, draw little energy and can be used in a regular light fixture.
Getting Where You Want to Go

RP Thoughts from Donna Green, President, FFB

“I thought about using a white cane for almost 20 years. The minute I lost my driver’s license and was diagnosed as being legally blind, I began to use it. I use my cane in crowded situations where it is really difficult but not impossible to get around – like parties, busy and dark environments, unfamiliar places, public transportation, etc.

“The bottom line is that I started to use my cane when getting around became exhausting and dangerous. When I reach AO - apology overload - and I find myself apologizing too many times in a day for bumping into people and things, I know it’s time to pull out the cane!”

When do people start to use a white cane?

According to an expert in orientation and mobility from the CNIB, there is no hard and fast rule. However, if you are experiencing problems with travel at night, in crowds, and in unfamiliar places, you may benefit from the use of a white cane.

Many people with RP are reluctant to use a white cane. However, once caning techniques are mastered, most users feel a sense of accomplishment and a renewed sense of independence. Mobility training takes time, patience and practice - start early! It may be stressful at first, but it hurts less than bumping into things!
Am I eligible for a guide dog?

According to the CNIB, if you are reliant on a white cane for both day and night travel, you may be eligible to apply for a guide dog. You must first complete a mobility training course and learn to rely on sound and tactile cues. It has been found that if you use your own vision too much, the dog’s skills will not be practiced, and as a service animal it may become undependable.

Who determines whether or not I can drive a car?

The Ontario Highway Traffic Act states the minimum visual standards to operate a motor vehicle. These recommendations were developed jointly by the Canadian Ophthalmologic Society and the Canadian Association of Optometrists. The criteria include colour perception, depth perception, visual acuity and horizontal fields.

In Ontario, an applicant for or a holder of a G, GI, G2, L, M, M1, M2 or R driver’s license must have:

1. In the case of a person whose vision in one eye is better than in the other, visual acuity in the better eye that is not poorer than 20/50 (corrected) as measured by Snellen Rating; and

2. A horizontal visual field of at least 120 degrees as measured by a specific type of Goldmann visual field test (see page 43) done with both eyes open.
In most provinces, regulations dictate that your physician must notify the ministry responsible for transportation if your vision doesn’t meet the minimum standards. Doctors do not like doing this but they are required to do so by law. If you feel your doctor has reported in error, you can seek a second opinion or file an appeal with your province’s ministry responsible for transportation.

Giving up a driver’s license is a very difficult loss. Some people with RP resign their license only after they’ve been in a motor vehicle accident. Others don’t see their ophthalmologists because they’re afraid to learn they don’t have enough vision to drive legally.

Use common sense, check the regulations in your province and discuss your options with your doctor and family. Driving with a visual impairment is driving impaired – putting your safety, and that of others, at risk.

Let’s Go Out

People with RP are out and about, just like anyone else. Here are some practical tips to keep you sailing smoothly through the day.

• Be the first to extend your hand for a warm handshake. Let the person you are greeting find your hand.
• When arranging to meet someone, state the exact location where you will be.

• Ask your friend or colleague to watch out for you, and ask what colour or clothing they’ll be wearing.

**Compare these directions:**

• “Let’s meet at the shopping mall.”
• “Let’s meet inside the doors to the north entrance of the shopping mall. I’ll be wearing my blue jacket.”

• Shopping centres can pose challenges for those with RP. Large glass doors, store windows and mirrors can be confusing. Pause frequently to orient yourself. Despite the well-lit interiors, these types of environments may warrant the use of a white cane for testing doorways and floor displays.

• At home, teach children at an early age not to lie on the floor or leave articles lying around. Make it a practice to scan each room before entering. The same habit holds true if you’ve dropped something – step back to the doorway and scan the floor from a distance.

“I’ve always found it amusing that people with RP have little difficulty finding each other when they’ve made arrangements to get together. We tend to give highly specific and detailed directions. Who else would mention potential hazards such as glass doors, steps and crowds? We always seem to manage to find a well-lit, quiet corner with a clear view!”
An important part of staying healthy, both physically and mentally, is keeping active. If you have RP you may notice changes in hand-eye coordination and the ability to maintain balance. These are normal consequences of losing the perceptual cues offered by vision. Therefore, you may benefit from exercises and games that train these skills and heighten your body awareness.

Regardless, the best activity for you will be one that you enjoy. Choose your activities according to your individual preference and abilities. Don’t sit on the sidelines, there’s always something you can do!

What sports should I play?

The CNIB suggests activities that don’t require quick, visual movements - for instance, cycling and goal ball. There are sports clubs across Canada that accommodate the needs of people with all degrees of vision loss. Ask at your local office of the CNIB or the Canadian Council of the Blind (CCB) to find out what’s offered in your area. There are no specific activities which are known to make RP worse. Engage in whatever activities you can comfortably do and enjoy. Charles, a member of the RD Network, is an ardent outdoors man despite his substantial vision loss. He recommends camping and hiking as rewarding activities.
“Make sure you mark your trail and are aware of where you are at all times. Take the time to notice your surroundings so you won’t get lost,” says Charles.

Some Sporty Ideas

- In-line skating on a quiet street
- Hike on a conservation area path
- Go to the gym and lift weights or take aerobics classes
- Learn Tai Chi or yoga
- Take tap, ballet, jazz or belly dancing lessons
- Go fishing or rowing
- Bowling, curling, or golfing with friends
- Join a runners’ club
- Rent a tandem bicycle with your partner

There are lots of things to do, and many of them can be performed with your eyes shut! Staying strong will give you confidence and a sense of freedom.
Will I have to give up board games and playing cards?

Many games can be modified or are available in a large print version. If you play a game like chess, you may want to keep to the traditional ivory and ebony pieces and use a board that has high contrast squares. Some players like the small travel size version so they can see the entire board at once. Card players can buy large print playing cards from the CNIB, or use poker cards, which have much larger markings than regular ones.

There is an old myth that using your eyes more makes them worse. Reading and computer use do not adversely affect RP. Some patients with RP may blink less when engaging in these activities which can cause a dry sensation of the eyes or blurring. Over-the-counter artificial tears can be used to avoid this problem, or just try to blink more. Remember to discuss these symptoms with your eye doctor.
Finding What to Wear

Many people have difficulty distinguishing dark colours, especially black and navy blue. If you are a person with RP, the challenge is even greater, especially in dim light. It may be worth the effort to devise a personal colour tracking system to organize clothing and wardrobe items.

• Take advantage of the expertise of sales clerks and don’t be afraid to mention you have difficulty distinguishing colours.
• Install lights in your closets. For example, lights on a pressure switch that will turn on when the door is opened, and turned off when the door is closed.
• Use track lights in your bedroom – point one directly to the closet.
• Buy liquid label maker that is raised when it dries.
• Purchase textured blue socks and plain knit black socks.
• Put a white mat in the bottom of your closet, to help you find your shoes.
• Check for assistive devices at your local CNIB, such as a portable colour reader that recognizes colours when pressed against fabric.
Remember safety first. If you are feeling uncomfortable with cooking, ask for assistance from friends, family or a low vision worker who may be able to give you ideas, like those above, to ensure your safety.

Use the Principles of High Contrast

• Raised lettering on plastic measuring spoons and cups can be enhanced with a black permanent marker.
• Make sure ingredients contrast against the interior colour of mixing bowls.
• Have both light and dark preparation areas and cutting boards.
• A light-coloured meal will show up nicely on a dark plate, whereas coffee is more visible in a cup with a light interior.

Light it Up!

• Under-the-counter lights.
• Interior cupboard lights and/or white interiors.
• Track lighting.
• Drawers for storage in place of cupboards.
Keeping Track

• Keep tall items and glassware pushed to the back of the counter where you can’t knock them over.
• When cooking or baking, place all ingredients on a single tray where they can be easily retrieved.
• Plastic bounces better than glass.
• Before you purchase any appliance, make sure you can read the displays easily.
• Electronic touch pads are sometimes difficult to see, particularly in dim light.
Reading and Communication

• Check your local library for large-print books.

• Computer screens and software can be adjusted to provide a wide range of type sizes and shapes, and different levels of contrast.

• Get a supply of primary school pencils, available at stationery stores and shops specializing in educational materials. These pencils are brightly coloured and make thick marks which are easily seen.

• Use white or light-coloured notepaper.

• Before you purchase a cell phone, make sure it has features that will work with you and not against you. Make sure you can see all the features on the keypad and that you alter the display to read it easily.

• If a printed newspaper doesn’t provide enough contrast in its print, take advantage of online newspaper services.

• There are some pretty nifty illuminated magnifiers available that can be stowed in a pocket or purse. They’re great for reading menus in dark places or the program in a theatre.
Let’s Talk About It

Your visual impairment may be a sensitive issue to discuss with others. Confusion may stem from the discrepancy between the idea of “visual impairment” and the fact that many people with RP show no obvious symptoms of vision loss. A good start is to learn about your RP. Being “comfortably informed” may help put you at ease when discussing the issue with others.

Often, someone will not ask you how you feel about RP – most of the time, they’ll want to know how it affects your sight and your ability to do things.

To help avoid isolation, remember you’re not the only one with RP. Reach out to other RP patients and supporters through the internet, mailing lists and chat lines.

Family dynamics are complex and unique, so if you have concerns about talking with your family, discuss them with your doctor or spiritual advisor, or ask for a referral to a counselor. A qualified professional may provide invaluable support and guidelines, particularly if you are feeling vulnerable.

Informing your family about a diagnosis of RP does not have to be difficult or uncomfortable. It is likely that family members will take their cue from you, so if you feel confident and relaxed, the chances are that they will respond in a similar way.
Learn to speak up! Let your partner know when you’re okay, as well as when you’re in need. Don’t leave your partner playing the guessing game - that only leads to frustration on both sides.

Advice to Spouses and Family Members

• Try attending support group meetings with your spouse, but only if you are comfortable with the environment.
• Go to low vision appointments so that you understand the visual limits of your partner. Don’t be afraid to ask the eye doctor questions.
• Work with your partner to develop a “good sight” plan for your home that addresses lighting and safety issues.
• Don’t be overprotective. Your RP partner still needs to be as independent as possible, and should let you know when assistance is necessary.
• Be supportive when the inevitable minor accidents occur.
• Don’t change the location or position of familiar objects around the home.
• Remember that humour should still be an essential element in your relationship.
• Encourage participation in familiar activities and keep your social contacts.
Children and RP

Introduction ................................................................. pg. 82
Children at School ......................................................... pg. 84
Children are remarkably adaptive and those with vision abnormalities may seem to play like any other child with normal sight for the first few years of life. When put together in the same play area, it’s often difficult to tell which child has the severe visual problem. It is not until school begins and there are more visual demands on a child that a deficit becomes more apparent.

If RP is present at birth (Leber congenital amaurosis, see page 23) there may be a more severe visual deficit and some symptoms may be more obvious. This might include eyes that appear to shake (nystagmus) or wander around without any true sense of fixation. Some children may seem to poke at their eyes (oculodigital sign) or be attracted to bright lights. Others have an aversion to bright lights. Others will have slightly better vision. This wide variability reflects the many kinds of RP that can be present at birth.

In children who develop RP at a young age, the first sign may be not seeing in dim light. This may show itself as a fear of the dark, a desire to sleep with a nightlight or obvious groping around when it is dark. Yet, most children show no visual abnormalities in an eye examination, particularly in the first five or six years of life.
Children with RP may tilt their heads in unusual positions or put their face close to books and papers when they read. A symptom in the school environment might be a desire to sit close to the blackboard. These adaptations should not be discouraged. Children with more severe problems may benefit from a low vision assessment by a specialist trained in this area once they get into grade one or two.

The Children’s Research Fund was created in 2000. The purpose of this special fund is to provide financial support, through the FFB, to researchers seeking to cure or prevent childhood blindness, including RP.
It is normal for parents to be concerned about their child’s education. It is at school that children learn many of the social and academic skills that are fundamental for their future success. Because RP is rare, many teachers are unfamiliar with its symptoms and its impact on sight. Those responsible for a child’s education should be made aware of any special needs, including those that involve vision loss.

The following suggestions come from parents and educators who have experience with children in the school environment. Think of this list as a starting point or as a guideline for you to develop to fit your own child and situation. Each school board provides different services and you may have to adapt your actions to suit the resources available.

It is highly recommended that you communicate personally with teachers and caregivers. If you can, provide them with written information about RP. We strongly suggest that you have them contact the FFB and/or the CNIB for supplemental information.
Make sure that all supervisors (e.g., school ground monitors) and adults in a position of responsibility are aware of your child’s visual limits. It is recommended that you maintain an open and ongoing dialogue with school staff so your child is ready for the changing demands of the school environment. You’ll want to make sure that accommodations are in place for everything from fire drills to class trips.

Ask your doctor to write a letter to the school explaining RP and your child’s vision and eye problems.

**In the Appendix at the back of this guide (see page 104) you will find a sample of a note that you might send along to a teacher or caregiver to get a conversation rolling. It can easily be appended to specific information relating to your child’s eye condition.**
Other Resources

A Network of Friends ................................................................. pg. 88
Key Organizations and Websites ................................................ pg. 90
Government Services ................................................................. pg. 91
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Canadian National Institute for the Blind (CNIB)

The Canadian National Institute for the Blind (CNIB) provides services to visually impaired and blind individuals. CNIB services are available, free of charge, to anyone who has difficulties as a result of vision loss.

Some of their basic programs include:

- Counseling and Referral
- Sight Enhancement (Vision Rehabilitation)
- Orientation and Mobility
- Technical Aids
- CNIB Library for the Blind
- Volunteer Services

Many CNIB centres offer additional services that serve the interests and needs of their consumers. Make sure you ask your local office to find out what services can be made available to you.

The knowledge, experience and understanding of the staff and volunteers can make the CNIB an invaluable resource. Many of the staff have experienced vision loss themselves, and will be sensitive to your issues and concerns.
The Ocular Genetics Programme at The Hospital for Sick Children

The Ocular Genetics Programme at The Hospital for Sick Children has a multi-disciplinary team of healthcare providers dedicated to the treatment of children and adults with genetic eye disease. Clinics are held every week at the hospital in Toronto.

The two major goals of the team are:

• To provide holistic care to individuals with genetic eye disease including attention to their psychosocial, environmental, rehabilitative, and counseling needs, while minimizing the number of visits necessary to obtain all appropriate examinations, diagnostic testing and interventions.
• To offer the opportunity to patients with genetic eye disease to actively participate in research endeavours and to apply the results of that research directly back to patient care.

Other university centres in the major Canadian cities are similarly involved in clinical care and research regarding RP. The FFB can help you identify eye doctors closest to your home who specialize in RP.
Key Organizations and Websites

The Foundation Fighting Blindness: www.ffb.ca

The Ride for Sight: www.rideforsight.com

Vision Health Research Council: www.vhrc.net

Canadian National Institute for the Blind: www.cnib.ca

The Hospital for Sick Children: www.sickkids.ca

Retina International: www.retina-international.com

National Coalition for Vision Health: www.visionhealth.ca

Canadian Ophthalmological Society: www.eyesite.ca

Royal National Institute for the Blind (UK): www.rnib.org.uk

PubMed
* Search for medical articles from around the world on virtually any topic.

Online Mendelian Inheritance in Man
* Search for medical genetic information.
The Government of Canada has programs that assist people who are legally blind, no matter what the cause of this disability. For this reason a medical doctor must complete a referral form to ensure your eligibility. Provincial governments also have programs which vary from province to province. The CNIB is a good place to begin an inquiry about these programs, but they do not administer them.

Be persistent in your inquiries and remember to ask if there are restrictions, or if there are time limits on any service.

Find out what services are available in your area, and get involved in your local government and community.
Books

*Do You Remember the Color Blue?*
*The Questions Children Ask About Blindness*
Alexander, Sally Hobart
Puffin, 2002

*Mom Can’t See Me*
Alexander, Sally Hobart
Simon & Schuster, 1990

*Taking Hold: My Journey Into Blindness*
Alexander, Sally Hobart

*Amazing Grace: Autobiography of a Survivor*
Halloran, Grace
North Star Publications, 1993

*Retinitis Pigmentosa*
Heckenlively, John R
Lippincott Williams & Wilkins, 1997

*On Sight and Insight*
Hull, John
Oneworld Publications, 1997
*Touching the Rock: An Experience of Blindness*
Hull, John
Vintage, 1992

*Slackjaw*
Knipfel, Jim
Berkley Publishing Group, 2000

*Helen Keller*
Markham, Lois
Franklin Watts, 1993

*The Mystery of the Eye and the Shadow of Blindness*
Michalko, Rod
University of Toronto Press, 1998

*The Two-in-One: Walking with Smokie, Walking with Blindness (Animals, Culture, and Society)*
Michalko, Rod
Temple University Press, 1999

*No Finish Line*
Runyan, Marla
Berkley Trade, 2002

*Self-Esteem and Adjusting with Blindness: The Process of Responding to Life’s Demands.*
Tuttle, Dean W.
Charles C. Thomas, 2004
Glossary of Terms
When we see, light passes through the cornea, pupil, and lens to the retina, which lines the inner surface of the eye. The cornea and lens focus the light on the retina (sometimes with the help of glasses or contact lenses). The retina then translates the picture into an electric message which is sent along the optic nerve to the brain where it is translated into the picture we perceive.

**accommodation**  
The ability of the eye to change its focus from distant to near objects; process achieved by the lens changing its shape.

**anterior chamber**  
The space in front of the iris and behind the cornea.

**aqueous humor, aqueous fluid (A-kwe-us)**  
Clear, watery fluid that flows between and nourishes the lens and the cornea; secreted by the ciliary processes behind the iris and flows through the pupil from behind into the front of the eye.

**astigmatism**  
A condition in which the surface of the cornea is not perfectly round; causes a blurred image to be received by the retina.

**blind spot (scotoma)**  
Any gap in vision where there is no vision. Caused by a defect in retina function. Everyone also has a normal blind spot caused by the optic nerve which we do not perceive.

**cataract**  
A clouding of the lens resulting in a dimming or distortion of vision.
choroid (*KOR-oyd*)
The layer filled with blood vessels that lies under and nourishes the retina. It is part of the uveal tract.

ciliary muscles
Muscles that enable the lens to change shape for focusing.

cones, cone cells
One type of specialized light-sensitive cells (photoreceptors) in the retina that provide sharp central vision and color vision. Also see rods.

conjunctiva (*KAHN-junk-TY-vuh*)
The thin, moist tissue (membrane) that lines the inner surfaces of the eyelids and the outer surface of the white of the eye (sclera).

cornea
The outer, transparent, dome-like structure that covers the iris, pupil, and anterior chamber; also a part of the eye’s focusing system.

dilation
A process by which the pupil is temporarily enlarged using eye drops (mydriatics); allows the eye care specialist to view the fundus.

fovea (*FOE – v-ah*) centralis
An area in the centre of the macula that allows the clearest vision; it is made up of cone cells.
fundus
The interior lining of the eyeball, including the retina, optic nerve and macula; portion of the inner eye that can be seen during an examination by looking through the pupil.

hyperopia (farsightedness, long-sightedness)
An eye that is shorter than average can not focus the image on the retina resulting in blurry images, especially for up close vision.

intraocular pressure (IOP)
Pressure of the fluid inside the eye.

iris
The coloured ring of tissue suspended behind the cornea and immediately in front of the lens; regulates the amount of light entering eye by adjusting the size of the pupil.

legal blindness in Canada
(1) visual acuity of 20/200  better eye with corrective lenses (20/200 means that the patient can see at 20 feet what a person with normal vision can see at 200 feet) or, (2) visual field restricted to 20 degrees of central vision (tunnel vision) in the better eye.

lens
The transparent, focusing structure that lies just behind the pupil.

macula (MAK-yoo-luh)
The small, sensitive area of the central retina that provides straight ahead vision for fine work and reading.
myopia (nearsightedness, short-sightedness)
An eye that is longer than average can not focus the image on the retina resulting in blurry images, especially for distance vision.

nerve fibres (axons)
The cells that extend from the retina through the optic nerve to the brain; carry the electrical message of vision from the eye to the brain.

optic cup
The white, depression in the centre of the optic disc.

optic disc (optic nerve head)
The circular area where the optic nerve connects to the back part of the retina.

optic nerve
The optic nerve is a bundle of nerve fibers, about the diameter of a pencil, which passes from the back of the eyeball to the brain carrying the message of vision from the retina.

peripheral vision
Side vision; ability to see objects and movement outside of the direct straight ahead line of vision.

photoreceptor cells
Specialized cells in the retina that transform light into electrical signals that can be sent to the brain. See rods and cones.
**posterior chamber**
The space between the back of the iris and the front face of the retina; filled with the vitreous gel.

**pupil**
The adjustable opening at the centre of the iris that allows varying amounts of light to enter the eye.

**pupillary response**
The constriction and dilation of the pupil caused by changes in light or accommodation.

**refraction**
The bending of light rays as they pass through the cornea, lens and fluids of the eye. The light is focused on the light-sensing tissue of the retina.

**refractive error**
Glasses prescription, amount of nearsightedness, farsightedness or astigmatism.

**retina**
The light-sensitive layer of tissue that lines the inside back of the eyeball. It contains the rods and cones which receive light and convert it to electrical signals for transmission via the optic nerve to the brain.

**retinal pigment epithelium (ep-ih-THEE-lee-um)**
The pigmented cell layer just under the retina that helps to remove the waste products of retinal cells.
rods, rod cells
One type of specialized light-sensitive cells (photoreceptors) in the retina that provide side vision and the ability to see objects in dim light (night vision).
Also see cones.

sclera (SKLER-uh)
The tough, white, outer layer (coat) of the eyeball; with the cornea, it protects the entire eyeball.

uvea (YOO-vee-uh) uveal tract
The middle layer (coat) of the eyeball, consisting of the choroid in the back of the eye and the ciliary body and iris in the front of the eye.

visual acuity
The ability to distinguish details and shapes of objects; also called central vision. Usually measured by eye chart.

visual field
The area or extent of space visible to an eye when looking straight ahead.
There is a central visual field that is directly in front of us, the target at which we are looking, and a peripheral visual field that we perceive as our “side vision”. The fields of each eye partly overlap.

vitreous (VIT-ree-us)
The transparent, colorless gel in front of the retina; fills the posterior chamber.
Appendix

Children and RP:
Overview of RP for Teachers and Caregivers....................... pg. 104
Retinitis pigmentosa (RP) is a group of rare but very serious eye conditions which are commonly diagnosed in the pre-teen and teenage years. RP causes deterioration of the retina. This is the tissue that lines the back, inside of the eye. As RP progresses it impairs the function of the retina and causes permanent loss of sight. Prescription eyeglasses do not correct the problem. There are currently no treatments or cures for this group of eye diseases.

The symptoms of RP vary from person to person, with the most common being an inability to see in dim light, night blindness and progressive loss of peripheral vision. Students with RP may have special needs and concerns in the classroom. As an educator you can minimize the effects of this visual disorder by adopting some relatively easy and inexpensive accommodations.

The following are tips collected from experienced teachers, parents and students affected by RP. Please keep in mind that the symptoms of the disease are unique to each individual. While many students may benefit from these suggestions, others may have enough sight that visual aids and modifications are not yet needed. It is recommended that you ask the student and/or parents about the timely and appropriate use of any accommodation.
Easy Ways to Help a Student with Low Vision

Adopt a **flexible seating arrangement** that lets the student sit near the blackboard when necessary, but also allows for a seat at the back of the classroom if a wider ‘view’ is needed.

If needed, allow **more time** to complete tasks and assignments, as well as tests and examinations. This may be helpful if the student has a field of vision so narrow he/she cannot easily scan the page. Check to see if the student needs time to rest their eyes as a result of vision fatigue.

**How a child with RP sees a page using their “tunnel vision”**

Although visual acuity is often unimpaired in pre-college years, **large print** (word processors and/or black felt tipped markers) may be helpful in some cases. **High contrast type**, such as black ink on white paper is easier to read for some children.

**High contrast print is easier for patients with RP to see**
Use **soft chalk**, preferably on a black, rather than a green board. Remember that high contrast printing is easier for everyone to read. If you use a white board, make sure to use heavier, darker coloured markers.

**Digital recordings** of books are now commonly available, especially through organizations like the Canadian National Institute for the Blind and its Digital Library. Character recognition software can translate the printed word into digital format. Once it is in digital format, it can be enlarged or read aloud using voice software.

Become familiar with the various **visual aids** that may benefit a low vision student. An aid can be something as simple as writing paper with thick lines or as sophisticated as a CCTV (closed circuit television) which greatly increases the size of print. A low vision rehabilitation specialist may be an appropriate resource to approach for assistance.

If you are working with younger children, make use of the reading-listening centres available. With these the headsets are attached to the same tape recorder.

For the younger child, **games can be adapted** so that everyone can participate. For example, beeping balls are available so that auditory as well as visual cues are enlisted in play.

**Rules to games can be modified.** Some accommodations work well for all students. Make an effort to create a level playing field for participants of all abilities.

**Keep walkways and corridors clear of low obstacles.** Cluttered environments can be hazardous. Make sure cupboard doors are closed.
Develop a “guidance system” to manage movement in the classroom when the usual classroom seating plan is not being used (e.g., an agreed upon orientation point).

Be especially mindful when classmates are seated on the floor. Devise a cooperative way of guiding the low vision student to a place that avoids both tripping and “squashing.” Everyone should know how to make room (e.g., tuck in legs) for their classmates.

If the student has a restricted visual field, agree on “attention-getting” signals. These could be tactile (tap on the shoulder) or audible/verbal signals. Be sensitive to issues that may surround inappropriate touching.

Field Trips and Offsite Activities

The rules that apply within the school and classroom are very much in effect when visiting new environments. A site visit before a field trip or sporting event would be ideal to assess its vision friendliness. Dim lighting, poorly marked stairways, tone-on-tone colour schemes and eccentric floor plans may present challenges for the RP student. A buddy system is always a good idea, but carrying a portable source of illumination like a flashlight might also prove helpful, especially in dark theatres.
Sports and Physical Education

The vision loss caused by RP does not in and of itself cause poor coordination and balance problems (notable exception being people having Usher syndrome type 1 which involves profound deafness at birth). Most aspects of athletics are unaffected by RP, and it is not unusual for RP students to excel at sports as often as any other student.

Unfortunately, some will avoid sports as the expectations set for them are unrealistic. Some of the things to watch for include: tripping or stumbling on uneven surfaces; losing sight of the ball when playing catch; accidentally running into other players and tripping over obstacles or field markers.

Fast-moving team sports that rely on peripheral vision may present extreme challenges for students with RP and may be very frustrating to master. Performance of skills during daylight hours may be much easier than during twilight or in dimly lit circumstances.

Compensating for Lighting Conditions

One of the common consequences of RP is a limited ability to adapt to different lighting conditions, e.g., from low to bright light and vice versa. Some will not be able to adapt to darkness at all, while others find bright daylight painful. Abrupt changes in lighting can be very uncomfortable and it will take longer for the RP student’s eyes to adjust.
Find out what conditions are optimum, and work to eliminate the worst case lighting environments. Following are some situations to keep in mind.

Work with the student to find the **best classroom lighting**. Think in terms of both seeing across the classroom (listening, watching videos) and working on tasks at the desktop (reading, writing). It may or may not be near natural daylight. A desk lamp with task lighting may be useful.

Remember **to turn on the lights immediately after a video** or slide presentation in a darkened room. Do not conduct lessons in semidarkness and expect an RP student to participate comfortably. Consider options for note taking: a student partner, a tape recorder or a small task light.

Make sure that **common areas** like stairwells, washrooms, corridors, assembly halls, backstage areas, gymnasium, locker rooms and connecting hallways are properly illuminated. Do not expect independent or confident mobility of an RP student in dark or dimly lit areas.

Students with RP may face great challenges when dealing with dusk and night time situations. Changing seasons bring different lighting conditions. Be prepared to **accommodate travel** when lighting is low. For instance, make sure buses are parked in areas with bright street lights and that walkways are illuminated. Alternatively, make sure the student has a guide to assist with their travel.
Interpersonal Communications

Acknowledge that the student has special needs, but avoid treating the student as ‘special’. Students want to be a member of their peer group, ‘Just like anyone else’.

An ongoing feedback loop between the student and teacher is essential. Encourage your student to voice concerns and to let you know what is working well.

Students with RP have voiced concerns about being perceived as ‘snobs’ because they do not acknowledge a gesture or other visual signal of friendship. Resolving problems like these rests with the student and his friends, but as an educator, you can promote an atmosphere of acceptance in the classroom.

Doing a class presentation on RP may take away the stigma and fear other students have about their classmate’s vision problems. Always consult with the student and parents before arranging this, as some may not be ready to do so.

Disclosure and Overcoming Prejudice

Respect the student’s decision to discuss RP with curious classmates. Both students and adults alike may mistakenly think that a student with RP is ‘faking’ or exaggerating the visual disability. RP is confusing and defies the general preconceptions of blindness or visual impairment. Why can someone read a book, but can’t find a light switch? Remember that resolving these conflicts can be difficult for the affected student as well as his friends. Chances are that other students will be accepting of the visual impairment if they understand the nature of the problem.
Career guidance is important for all students, and those with RP should be encouraged to strive to reach their full potential. With advances in technology today, there are few obstacles that cannot be overcome.
Disclaimer

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