

UNDERSTANDING  
Retinitis Pigmentosa



# Introduction

Many thanks to the patients and families affected with retinitis pigmentosa who provided valuable insight for the content and layout of this booklet.

Thanks also to the Kellogg Eye Center Inherited Retinal Dystrophy Clinic staff, Low Vision Clinic staff, and Marketing Department staff for making this booklet possible. Our research into inherited eye disease is generously funded by the Foundation Fighting Blindness, the Elmer and Sylvia Sramek Charitable Foundation, and the National Eye Institute/National Institutes of Health.

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*Published 2/2008*

*Funded through a grant from the  
FRIENDS of the University of Michigan Hospitals*

*Images on page 5 provided by the National Eye Institute,  
National Institutes of Health*

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## Learning about Retinitis Pigmentosa

It can be overwhelming to receive a diagnosis of a condition that will affect your vision profoundly. Because vision affects so many daily activities, many people find it difficult to adjust to even mild vision loss. Armed with information, patients and their families can face the future confident that they know what to expect and ready to explore all the possibilities that life will offer.

The purpose of this booklet is to provide information to individuals with retinitis pigmentosa (RP) and their families.

The first part of this booklet focuses on medical knowledge about RP. Later sections deal with support and daily living.

# Understanding Retinitis Pigmentosa

Retinitis pigmentosa affects 1 in 3500 people in the United States. RP is defined as an inherited retinal condition that gradually leads to visual field loss and retinal degeneration. Many conditions meet the definition of RP. This booklet describes general features common to most forms of RP.

The name “retinitis pigmentosa” refers to what the retina looks like in those with RP. The term “retinitis” was first used in the 1850’s because doctors thought the retina had an inflamed appearance. We now know that RP is not caused by infection, but is an inherited condition. When viewed with a specialized tool, the RP retina often shows clusters of pigment not seen in a normal retina. The word “pigmentosa” refers to the pigment deposits seen during the eye exam.

The term “retinitis pigmentosa” now refers to a group of eye conditions that affect the retina, or the layer of nerve cells at the back of the eye. There are two main cell types within the retina: rods and cones. Cone cells are present throughout the retina. The center of the retina (the macula) has the greatest amount of cones, and helps with central (reading) vision and color vision. Rod cells are present throughout the retina, except for the very center (fovea). Rods help with night vision and side vision. In RP, the rod cells and eventually the cone cells stop working, causing vision loss.

## **SIGNS AND SYMPTOMS OF RETINITIS PIGMENTOSA**

Retinitis pigmentosa is a progressive disease that mainly affects rod cells, the cells that help with peripheral (side) and night vision. Early symptoms of RP can vary from person to person, and typically include trouble seeing at night (night blindness) and/or loss of side vision. At first, people may not realize their peripheral or night vision is affected, since the loss is typically very gradual.

In early stages of RP, the quality of vision generally remains the same, while the field of vision is reduced. This produces “tunnel vision.” As RP progresses, visual clarity, or visual acuity, may also decrease. Both eyes are usually affected in a similar way.

There can be a very large range in the age of onset for RP. Some patients are diagnosed in early or middle childhood, while others are not affected until they are in early or late adulthood.

Understandably, individuals with a diagnosis of RP want to know exactly what will happen to their vision. Due to the slowly progressive nature of the disease, it is difficult for anyone to predict—including the doctors—exactly what vision will be like at a specific time in the future. There is wide variation, even among affected members of the same family. Retinitis pigmentosa can appear at an early age or later in life; some family members may be severely affected, while others will have milder forms of the disease.

Although patients are concerned about going completely blind from the condition, this is actually uncommon for people with RP. It is more likely that in the later stages of disease a patient would be considered “legally blind.” This is defined as having best corrected vision equal to or worse than 20/200 or less than 20 degrees on a visual field in both eyes. If legal blindness does occur, for most RP patients it is due to small visual fields, rather than blurry vision. The effect is like looking through a straw; the image is very limited, but it is still sharp and clear. It may take many years for vision to reach a stage where the person is considered “legally blind.”



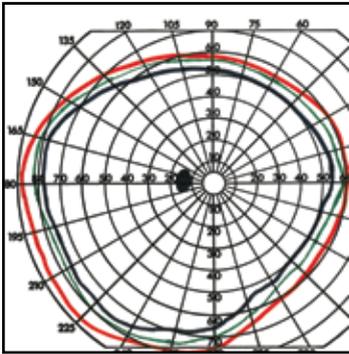
*Normal Vision*



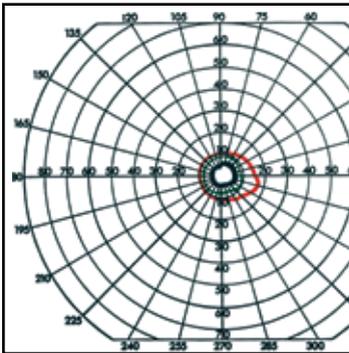
*Tunnel Vision*

## TESTING FOR RETINITIS PIGMENTOSA

You may wonder how RP is diagnosed, and why so many tests are needed to diagnose it properly. It is important to understand that a combination of many tests is often needed to separate RP from other retinal conditions. It is not uncommon for a person to visit several doctors before arriving at the diagnosis of RP. The tests also help your doctor understand how well your retina functions. Some of the tests are:



*Normal Visual Field*



*Retinitis Pigmentosa*

- **Visual Acuity Testing** — Visual acuity is another term for visual clarity. Most people are familiar with this test, in which they read letters from a chart while seated at a certain distance. A person with normal visual acuity is said to have 20/20 vision. A person with 20/40 vision can see at a distance of 20 feet what a person with “normal” vision can see at 40 feet.
- **Visual Field Testing** — This test measures a person’s field of vision. A light is brought in from the side on a screen, and slowly moved to the center of vision. Patients press a button as soon as they see the light. For individuals with RP, the field of vision gradually decreases over time. The area of vision will become smaller and smaller until it is like looking through a straw. At left you can compare the visual field of a person with healthy vision to that of a person with RP.

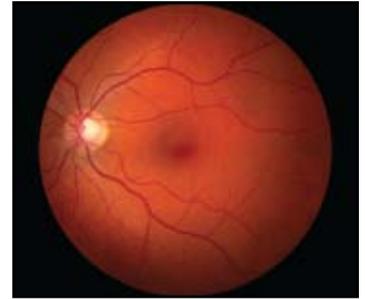
- **Electroretinogram (ERG)** — This tests rod and cone function, and is important for confirming a diagnosis of RP. In some cases, the ERG shows signs of RP even before the patient is aware of symptoms or before the doctor can see signs of RP in the retina. This specialized test is performed in only a small number of centers nationwide.

For ERG testing, a numbing drop is put in the eye and a special type of contact lens electrode is placed on the eye. Flashes of light are used to stimulate the retina. Electrodes measure the electrical response of the rods and cones to the flashing lights. This test is usually performed in a darkened room. The test is not painful, but some find it to be uncomfortable.

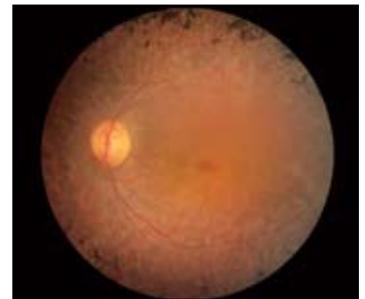
- **Fundus photographs** — Using a special camera, your doctor can photograph the fundus, or back of the eye. The testing is relatively fast, but requires that the eyes be dilated. The images at the right show the fundus images of a person with healthy vision and a person with RP.

- **Optical Coherence Tomography** — This test captures cross-sectional images of the retina. It measures the thickness of the retina and can identify retinal abnormalities. The device scans the retina surface with light to obtain images.

- **Fluorescein Angiogram** — This test may or may not be used at your visit. It involves a special dye (fluorescein) that allows your doctor to see the blood vessels at the back of the eye. The eyes are dilated and the dye is injected into a vein in the arm. A special camera records the dye as it passes through the blood vessels in the eye. The resulting photos allow your doctor to identify retinal problems.



*Normal Fundus*



*Retinitis Pigmentosa Fundus*

## **TREATMENT FOR RETINITIS PIGMENTOSA**

While there are no therapies today to cure RP, there are two important options for helping individuals with the disease. The first is to make the most of existing vision by using low vision therapy and aids. Second, it may be possible to slow further vision loss with the use of antioxidant vitamins. Meanwhile, research is ongoing. Scientists have effectively treated some animals with RP. Several treatment trials in humans are expected to begin in the near future.

Despite the lack of treatment for RP, general eye checkups are important because people with RP are still at risk for other kinds of eye problems that may affect anyone in the general population. Some may be treatable with surgery or medications. Regular visits to an RP specialist can also make you aware of current advances as we learn more about RP and treatments that may help you.

# Genetics of Retinitis Pigmentosa

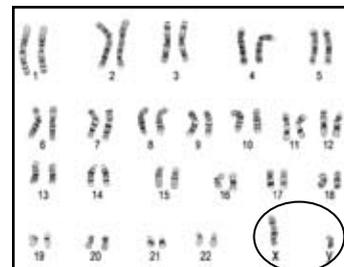
Retinitis pigmentosa is a condition that can be inherited in families, but may not be inherited the same way in every family. In order to understand the inheritance of RP, we should first briefly discuss genes and chromosomes.

## GENES AND CHROMOSOMES

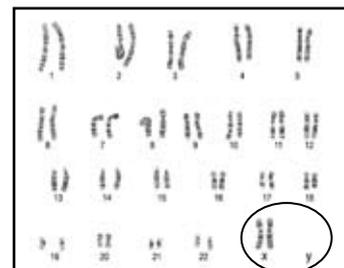
Each cell in our body contains the same set of genetic instructions that tells our body how to function. Half of the information comes from our mother, the other half from our father. The information is found on structures called chromosomes. Each cell has 23 pairs of chromosomes, or 46 total. Every chromosome has many genes. A gene contains specific instructions for a particular function in the body, like eye color. Since we have two copies of each chromosome, we have two copies of most genes.

The only chromosomes that don't always come in pairs are the sex chromosomes — X and Y. Males have an X and a Y chromosome, while females have two X chromosomes. Thus, females have two copies of genes on the X chromosome, while males have one.

Sometimes, a gene may not work properly because of a “typo” in the instructions for one or both copies of that gene. This “typo” is also referred to as a gene change or gene mutation. When the gene doesn't function as it typically should, it may lead to a genetic disease such as RP.



*Male Chromosomes*



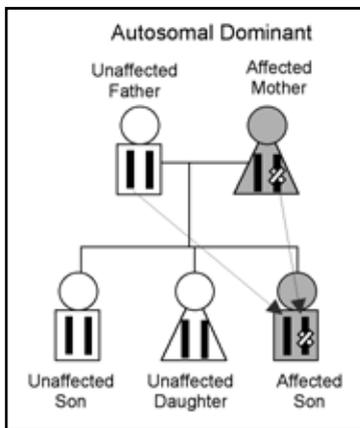
*Female Chromosomes*

## INHERITANCE OF RETINITIS PIGMENTOSA

Many mutations in many different genes are known to cause RP. These various mutations cause different forms of RP. A gene mutation can also be influenced by the environment, or interactions with other genes. This may explain why family members with RP can be affected differently even though each has the same mutation. Patients with questions about their personal form of RP should consult experienced health professionals, such as an ophthalmologist or genetic counselor, who know about genetics and RP.

Because there are different genes involved, there are different ways that RP can be inherited. The three primary patterns are: autosomal dominant, autosomal recessive, and X-linked recessive inheritance.

The diagrams of family trees (pedigrees) below show different ways that RP can run in families. A person with RP is filled in gray. The black rectangles represent chromosomes. Gene mutations are shown as a white X.



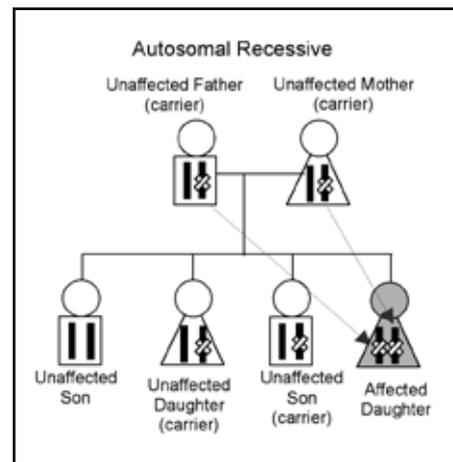
**Autosomal dominant** inheritance occurs when just one copy of a gene mutation causes RP. The mutation causes RP even when the second copy is normal. Some features of autosomal dominant inheritance are:

- The disease can be passed from one generation to the next: from parent to child to grandchild.
- Both males and females can be affected.
- When a person has a dominant form of RP, he or she has a 50% chance with each pregnancy of having a child affected with RP.
- About 15-25% of the people with RP have the autosomal dominant form.

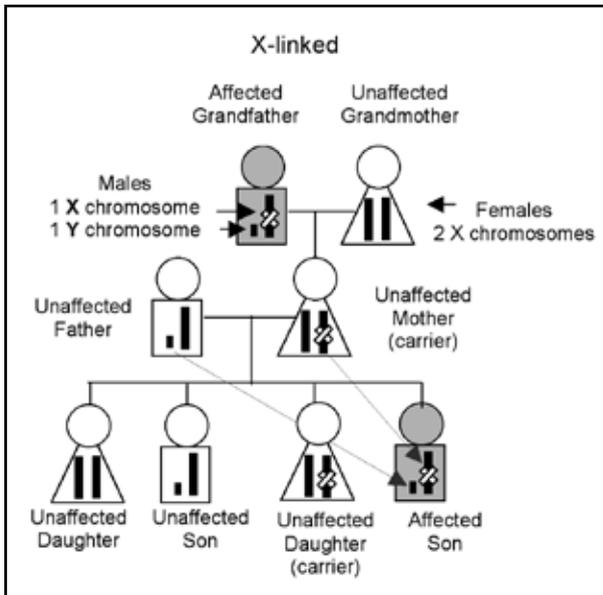
In families with autosomal dominant RP, not every person in the family is affected in the same way. Some may be affected so mildly that they are not even aware that they have signs of the disease. In rare cases, someone with a dominant mutation may not show any signs of the condition.

In **autosomal recessive** inheritance, on the other hand, a person develops the condition only when *both* copies of the gene don't work. That is, the gene from the mother and the gene from the father both have mutations. Some features of autosomal recessive inheritance are:

- Typically, only one generation is affected.
- Both males and females can be affected.
- Individuals with one normal gene and one mutated gene generally don't show symptoms of RP, and are called "carriers."
- When both parents are carriers, there is a 25% chance (with each pregnancy) the child will have RP.
- About 20-30% of individuals with RP have the autosomal recessive form.



In **X-linked RP**, the mutation is on the X chromosome. Males have an X and a Y chromosome and females have two X chromosomes. Because males have only one X chromosome, if they inherit an RP mutation on that chromosome, they will have RP. Females with one mutation and one regular copy of the gene typically do not show signs of RP. If they do, the symptoms are usually mild and tend to occur later in life. Some features of the X-linked form of inheritance are:



- Multiple generations of males are affected, connected through unaffected females. For example, an affected grandfather would have a daughter who is a carrier. She may have a son who is affected.
- X-linked RP is much more common in males than in females.
- When a male is affected, all of his daughters will be carriers and none of his sons will be affected.
- When a female is a carrier, each daughter has a 50% chance of being a carrier and each son has a 50% chance of being affected.
- About 10-15% of individuals with RP have the X-linked form.

## **UNCERTAIN INHERITANCE**

Often (40-50% of the time), a person diagnosed with RP has no other family members with the disease. There are several possible reasons for only finding one person with RP in the family:

- The RP mutation was a new event in that person.
- Other family members have the same mutation, but have not been diagnosed with RP. They may have a later age of onset, or have milder signs of the disease.
- The mutation has been in the family for a long time, but by chance, no other family members have been affected. For autosomal recessive RP, carriers may have been present in the mother's and father's side of the family for several generations, but a child won't develop RP unless both parents are carriers and both pass on a mutation to their child.

## **GENETIC TESTING FOR RP**

Testing a person's genes is an important part of diagnosing RP. At the Kellogg Eye Center you may be asked to give blood for genetic testing. Samples are sent either to a research laboratory or to a certified testing lab. Research laboratories do not charge a fee, but it will take months or even years to get a result. Certified labs charge a fee, but results are given in a shorter time. The lab chosen depends on the type of RP you have.

# Research into Retinitis Pigmentosa

## GENE IDENTIFICATION RESEARCH

There are over 55 RP-causing genes known. Fifty more genes cause other diseases with features similar to RP. More genes are being discovered all the time. Participating in RP research may help researchers find new RP-causing genes or develop a better understanding of this disease. This information can be used to produce better treatments and ultimately a cure for RP. Studies of many members of the same family may also be useful to identify RP-causing genes. Sometimes, if we know the mutation causing your RP, we can give you more information about disease progression or risks to family members.

## RESEARCH FOR NEW THERAPIES

Scientists believe that researching the genetic causes of RP will help in the development of future therapies. Research constantly produces new information, so it is difficult to report here on the “latest” findings. The best way to stay up to date is to ask your RP specialist about current research and clinical trials at your next visit. You can also find reliable information online at:

- National Eye Institute [www.nei.nih.gov](http://www.nei.nih.gov)
- U-M Kellogg Eye Center [www.kellogg.umich.edu](http://www.kellogg.umich.edu)
- Foundation Fighting Blindness [www.blindness.org](http://www.blindness.org)

# Help for People with Retinitis Pigmentosa

It may be very hard to deal with a diagnosis of RP. Many people have never heard of the disease and are unsure of how RP will affect them. It is important to realize that most of the time the disease progresses slowly. For the most part, you will not have to learn new skills overnight. People who have RP lead successful and full lives. There are people to help you along the way. Many kinds of services are available if and when they are needed.

## **LIFE WITH RP**

It is perfectly natural for someone with vision loss to feel anxious, fearful, angry, or unhappy. If your child has RP, it is also natural to have feelings of uncertainty, anxiety, and fear over what the future may hold for him or her. It is critical that you discuss these feelings with your medical team. Support groups can also be very helpful.

People with RP may feel helpless because there is currently no cure for the disease and very few treatment options. But if you are affected by RP, there are many ways to take an active role to manage your condition. This can help you live as independently as possible. The following pages offer suggestions and resources for living with RP.

## **LIVING WITH RP IN CHILDHOOD**

If you are a parent of a child with RP, it is important that you talk with your child about the condition. It is natural for parents to protect a child from knowing about the disease, but most often the child is aware of it and needs parental guidance. Children may be scared by having many

medical appointments and may be unsure of what to expect in the future. Your child should be encouraged to talk with you if he or she has any changes in vision or needs additional help at home or school.

Parents struggle to find a balance between allowing the child to explore his or her surroundings, and stepping in to help when needed. Remember, your child is an individual who happens to have vision loss. Your child can have a normal childhood and grow up to be independent and successful, like any other child. As a parent, your attitude will dramatically impact your child's views of living with RP and expectations for the future. It's important to maintain a sense of hope with your child, and to never give up. Open communication between parent and child will help.

While a positive attitude is crucial, it is also important to be realistic about the struggles for children and families dealing with low vision. Some parents find counseling for themselves, their child, or their entire family to be helpful. It can help the family maintain a sense of normalcy as they learn to adapt to the child's needs. It's also a good idea to introduce your child to several different peer groups besides the one at school. An extra set of peers in the community may help your child overcome difficulties with peers at school.

## **SUPPORT FROM SCHOOLS**

Your child's teachers and school officials can be important members of your support team as you all work to help your child succeed. School districts or counties may assign a special education counselor. This person may follow your child until high school graduation. You should contact your child's teachers and principal before the school year begins to discuss arrangements your child may require. A yearly

Individualized Education Plan (IEP) meeting is usually scheduled. This is a great way to stay in touch with school officials.

Children may try to hide struggles they experience, and can sometimes behave differently at home compared to school. Parents need to be alert for signs their child is struggling and needs help. Frequent contact with your child's teachers and counselors can make you aware of issues your child might not bring up at home.

Your child's teachers or school counselor may not know about RP. A booklet like this one may be useful to help them understand your child's condition. They may also find information from your child's eye doctor helpful in understanding your child's condition and needs (e.g., limitations in visual acuity). Some children might benefit from better lighting. Others may need extra time for tests. Some children may benefit from other low vision aids in school.

## **MAKING THE TRANSITION FROM HOME TO COLLEGE**

Groups such as the Michigan Commission for the Blind can help you manage requests for services from the state after high school graduation. Other states have similar organizations.

Most colleges and universities have services for students with special needs like low vision. These include help registering for classes, obtaining books, making testing arrangements, finding a reader, transportation, etc. Many schools have programs that provide low vision students with equipment to record lectures. There may also be orientation workshops for students to familiarize themselves with campus before classes begin. Taking advantage of services requires planning ahead (as when ordering large-print textbooks), but there is

no reason a person with low vision can't be successful at any university, or in any field of study.

Teenagers with low vision can become discouraged because of the social issues they face. They may feel different because they can't drive or enjoy certain typical teenage activities. Many young (and older!) adults forget how much they can do. While those with low vision do experience some limitations, most agree that their quality of life is largely dependent on attitude, not vision.

Many services are available to college students with visual impairment, but the student has to seek them out. Schools usually have an office for students with disabilities. As children enter the teenage and college years and become more independent, parents can help them learn to advocate for their own needs.

## **LIFE WITH RP AS AN ADULT**

Most adults describe living a full and interesting life despite the practical and social problems that can arise because of RP. Although many individuals with RP are diagnosed in childhood, some may not be diagnosed until they are adults. Others might not have a need for low vision adaptation until they are adults with established careers. When this happens, they may feel as though they can no longer perform their job functions. However, individuals with RP can and do have a wide variety of jobs, from computer programmer to musician, and often find it unnecessary to change careers because of RP. Some adults may need help in the workplace at first, perhaps through on-the-job training or a change in the worksite setting. In Michigan, the Commission for the Blind offers services to help adults with low vision adapt to work settings and to live independently.

In the later stages of disease, some people with RP may need to apply for disability insurance benefits. This can be done by contacting your local state government social security office.

Many successful adults with low vision and the specialists who work with them stress the importance of focusing on what you can do, and maintaining a positive attitude. One woman with low vision has described it this way: "If you were on the freeway and it was closed, you would find another way to get where you're going. You wouldn't just give up and stop moving." All individuals, including those who happen to have low vision, have unique gifts and talents that make them valuable members of society.

## **AMERICANS WITH DISABILITIES ACT**

The Americans with Disabilities Act gives civil rights protections to individuals with disabilities, such as low vision due to RP. Its policies prevent employment discrimination and allow equal access to education, healthcare, and public transportation. If you have low vision, you should understand your rights under this act. For more information, you can call 800-514-0301 or visit [www.ada.gov](http://www.ada.gov).

## **SENIOR SERVICES**

Managing low vision after retirement is typically not different from managing the condition as a younger adult. Challenges can arise that would affect anyone at this stage of life. For example, people with RP may still be at risk for other age-related eye conditions. It is important to continue regular checkups with your eye doctor and primary care physician. Individuals with RP of any age may find support groups helpful.

## Talking to Others about Retinitis Pigmentosa

While it may seem difficult at first for you to understand your condition, it can be especially difficult to discuss your condition with others. Unlike other “disabilities,” a person with low vision often shows no outward features of the problem. Others may not be aware of your visual impairment. Many people feel awkward about discussing their visual needs. They don’t realize that others are probably eager to help in any way they can, but may not know how to approach you.

In deciding how and when to talk to other people about your condition, you and/or your child should find the right balance for you. Individuals with low vision should not feel pressured to educate everyone they encounter. Still, they can be advocates for themselves by sharing their needs with those who can help. Some find open and honest conversations to be most helpful. Others prefer to share written materials, then answer questions as needed. Several groups, such as the Foundation Fighting Blindness ([blindness.org](http://blindness.org)), can provide helpful information about your condition and can be accessed online.

## WORKING WITH A LOW VISION SPECIALIST

Many individuals gain a great deal of confidence by working with a low vision specialist. These specialists can help you achieve independence, teaching you how to manage daily activities and make the most of your available vision. In addition, they can recommend assistive devices that will magnify or otherwise enhance your range of vision. Research shows that those who receive training from a low vision specialist are far more likely to use their devices properly and to benefit from them down the road. Low vision specialists can also put you in contact with rehabilitation services or other specialists.

## LOW VISION EXAMINATIONS

To determine the extent of your useful vision you will need to have your eyes examined. Exams for low vision may differ from typical eye exams. During a low vision exam, your low vision provider may administer the following tests:

- Refraction — to assess your vision, determine whether glasses would be helpful, and provide the prescription for your glasses
- Visual field — to assess your peripheral vision
- Ocular motility — to assess how well your eyes move

Because low vision exams may involve many tests, they are often more time-consuming than the standard examination. For instance, refraction may be done through a telescope or trial lens so you can judge which lens is best. Based on the results of the low vision exam, the low vision specialist will make suggestions for optical aids or techniques for adjusting to daily living activities. It may take several visits to learn how to use the device well. Some eye centers, such as the Kellogg Eye Center, have specialists who will travel to your home. They can help adapt your home setting to low vision needs.

## **LOW VISION AIDS**

Some people may benefit from using special lighting to complete their daily tasks. There are also devices and programs that make it easier to use computers. Some computer-based programs can act as magnifiers, or even convert text to audio formats. Patients with very late-stage disease are often helped by learning how to use a cane to find objects or curbs in their path when walking. They may also find a guide dog helpful. Some patients ask if it is necessary to learn Braille. Only a small number of patients in very late stages of RP need Braille for reading.

## **LOCATING LOW VISION PROVIDERS**

Most larger eye centers, including the Kellogg Eye Center, have low vision clinics. Your local ophthalmologist or RP specialist should be able to give you a list of providers in your area.

## **LOW VISION SUPPORT GROUPS**

Some people may be interested in meeting with others affected by vision loss. Meeting with others facing vision loss can provide useful support and let you know that you are not alone. Topics such as anger and fear about dealing with low vision are often discussed. There are more than 100 low vision support groups in Michigan. The Retina Clinic at the Kellogg Eye Center can give you a listing of these support groups. Another organization, the National Association for Parents of Children with Visual Impairments (or regional branches of this organization), can also offer support. Visit [www.napvi.org](http://www.napvi.org).

Online chat rooms and message boards can also be helpful. Some organizations, such as the Foundation Fighting Blindness ([www.blindness.org](http://www.blindness.org)) have online message boards for individuals with RP. However, if you have factual questions about your condition it is always best to talk to your ophthalmologist.

## Vision Insurance

It is possible to obtain vision insurance to help with the costs of exams, glasses, and other assistive devices. Several government and volunteer organizations offer free or low-cost equipment to those with low vision. Check with your retina specialist for suggestions.

## Resources

Below are a few organizations that can provide information about RP and about living with low vision. These groups may also put you in contact with others in your area with RP. The Retina Clinic at the Kellogg Eye Center has an extensive listing of resources for individuals with low vision. Please contact us if you would like more resources.

- U-M Kellogg Eye Center  
[www.kellogg.umich.edu](http://www.kellogg.umich.edu)
- Foundation Fighting Blindness  
[www.blindness.org](http://www.blindness.org)
- National Eye Institute  
[www.nei.nih.gov](http://www.nei.nih.gov)
- American Foundation for the Blind  
[www.afb.org](http://www.afb.org)
- British RP Society  
[www.brps.org.uk](http://www.brps.org.uk)
- Foundation Fighting Blindness – Canada  
[www.ffb.ca](http://www.ffb.ca)
- Lighthouse International  
[www.lighthouse.org](http://www.lighthouse.org)
- National Association for Parents of Children with Visual Impairments  
[www.napvi.org](http://www.napvi.org)



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