

UNDERSTANDING  
Stargardt Disease



University of Michigan  
Kellogg Eye Center

# Introduction

Many thanks to the patients and families affected with Stargardt disease who provided valuable insight into the content and layout of this booklet.

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Authors:

**Amanda Openshaw, MS**

**Kari Branham, MS, CGC**

**John Heckenlively, MD**

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# Learning about Stargardt Disease

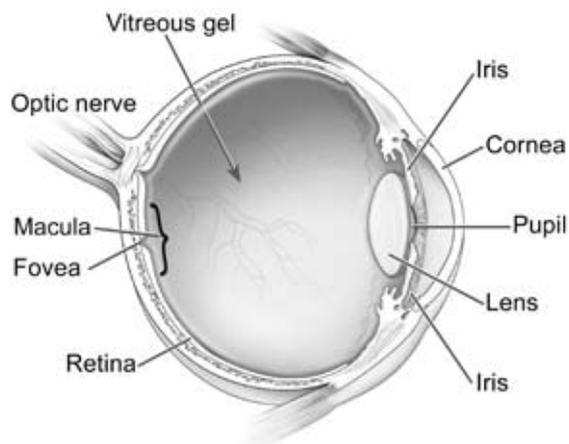
It can be overwhelming to receive a diagnosis of a condition that will affect your vision profoundly. Because vision affects so many daily activities, many people find it difficult to adjust to even mild vision loss. Armed with information, patients and their families can face the future confident that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

The purpose of this booklet is to provide that information to individuals with Stargardt disease and their families.

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

# Understanding Stargardt Disease

Stargardt disease affects 1 in 10,000 people in the United States. It was named for the German ophthalmologist Karl Stargardt, and may also be called fundus flavimaculatus. This condition affects the retina, which lines the back of the eye and is composed of many layers. One layer of the retina is the retinal pigment epithelium (RPE). In people with Stargardt disease the RPE collects a substance called lipofuscin, which can lead to vision problems. Vision loss in Stargardt disease is most intense in the macula, which is in the center of the retina. Stargardt disease is part of a group of diseases affecting the macular region of the retina, called macular degenerations. Stargardt disease is sometimes called a juvenile macular degeneration because it often appears at an early age.



## SIGNS AND SYMPTOMS OF STARGARDT DISEASE

Stargardt disease chiefly affects the macula, which controls central and detailed vision. A person with the condition may have reduced visual acuity, causing blurry vision. A first sign of Stargardt disease may be trouble reading. Blind spots can occur. At first, the size of the blind spot is small, but it may gradually increase in size. The bottom image on the left shows a person's vision in late stage Stargardt disease.



*Normal Vision*



*Stargardt (late stage)*

Central vision in those with Stargardt disease is typically decreased, but peripheral vision (side vision) usually remains stable. In late stages of disease, color vision may be affected. The changes tend to be similar in both eyes. Stargardt disease is often diagnosed in the teen years, although some cases may be diagnosed earlier or later in life.

Understandably, those with a diagnosis of Stargardt disease want to know exactly what will happen to their vision. There is wide variation from person to person in symptoms and in how quickly the disease progresses. These differences exist even among affected members of the same family. It is difficult for anyone to predict—including the doctors—exactly what vision will be like at a certain time in the future. Progression may be gradual over several years, or may occur in a shorter period of time. Stargardt disease can appear at an early age

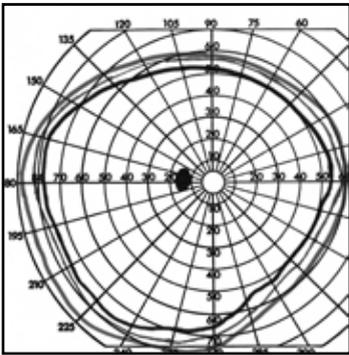
or later in life. Some family members may be severely affected, while others will have milder forms of the disease. Severity may be related to the type of gene change present.

Although patients are concerned about going completely blind from the condition, this is actually uncommon for people with Stargardt disease. It is more likely that a patient may become “legally blind.” This is defined as having best corrected vision equal to or worse than 20/200 in both eyes. For Stargardt patients, if legal blindness does occur, it is due to decreased central visual acuity. Side vision usually remains intact. Magnifiers may help with reading using side vision.

## **TESTING FOR STARGARDT DISEASE AND RELATED CONDITIONS**

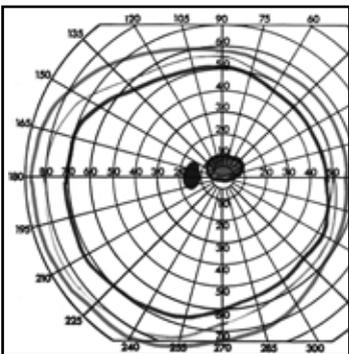
You may wonder how Stargardt disease is diagnosed, and why so many tests are needed to diagnose it properly. It is important to understand that a combination of many tests is often needed to separate Stargardt disease from other retinal conditions. It is not uncommon for a person to visit several doctors before one can confirm the diagnosis of Stargardt disease. The tests also help your doctor understand how well your retina functions. This can assist in planning the best aids and devices for your remaining vision. Some of the tests are:

- **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.



*Normal Visual Field*

- **Color Testing** — This measures a person’s color perception. A series of images composed of many small circles is presented to the patient. Someone with normal color vision can recognize numbers within the images. Individuals with Stargardt disease are typically able to perform color vision tests. In late-stage disease, patients may have some color vision loss.

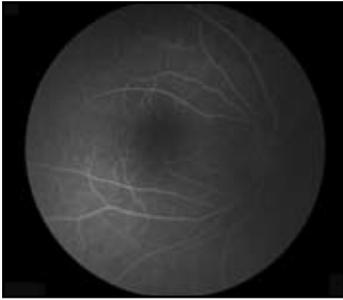


*Stargardt Visual Field*

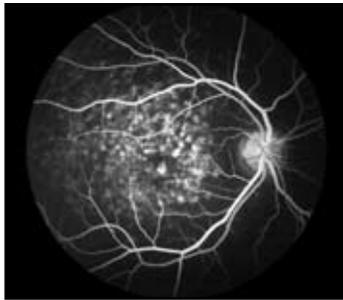
- **Visual Field Testing** — This test measures a person’s field of vision. A light is brought in from the side on a screen, and slowly moved to the center of vision. Patients press a button as soon as they see the light. For Stargardt disease patients there is seldom loss of side vision. Some patients may develop a blind spot in the center of vision. In the diagram at the left, the black circle in the center represents the blind spot.

■ **Electroretinogram (ERG)** — This tests rod and cone function. It is important for ruling out other diagnoses, like cone degeneration, which may appear similar to some cases of Stargardt disease. The test also measures retinal function. The ERG 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 recording contact lens 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 performed first in a dark room, then the lights are turned on and the eye re-tested. The test is not painful, but some find it to be uncomfortable. The ERG response of someone with Stargardt disease is typically normal or mildly abnormal. Some more severe cases can have bigger changes.



*Normal Fluorescein  
Angiogram*



*Stargardt Fluorescein  
Angiogram*

- **Fundus photographs** — Using a special camera, your doctor can photograph the retina fundus at the back of the eye. The testing is fairly quick, but requires that the eyes be dilated. Several types of changes can be seen in the retina of someone with Stargardt disease. Often, the ophthalmologist may see changes in the macula with yellow-white spots.
- **Fluorescein Angiogram** — This is one of the key diagnostic tests to confirm Stargardt disease. It involves a special dye (fluorescein) that allows your doctor to see the specific changes at the back of the eye associated with Stargardt disease. 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 back of the eye. The resulting photos allow your doctor to find retinal problems more easily. Patients with Stargardt disease usually show a “dark choroid effect.” This is common in Stargardt disease, and refers to darker than normal background under the retina.

## TREATMENT FOR STARGARDT DISEASE

While there are no therapies today to cure Stargardt disease, research has revealed clues about slowing its progression. Research animals with Stargardt disease build up abnormal proteins in their RPE layer under the rods and cones. These abnormal proteins are light-sensitive. It is likely that these proteins become more toxic when the patient is exposed to extreme sunlight. This suggests that sunlight and high energy light can worsen the disease. Patients with Stargardt disease should use UV-screening sunglasses when out in the direct sunlight. The use of brimmed hats or other shading devices is also warranted. Some evidence suggests that taking extra vitamin A, such as in capsules, may make things worse. Other antioxidant vitamins may slow the progression of disease. If the patient has poor vision, existing vision should be maximized by using low vision therapy and aids.

Despite the lack of treatment for Stargardt disease, general eye checkups are still important. People with Stargardt 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 ophthalmologist can also make you aware of current advances as we learn more about Stargardt disease and treatments that may help you.

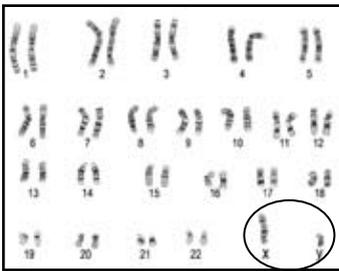
# Genetics of Stargardt Disease

Stargardt disease is a condition that can be inherited in families. Genes and chromosomes are the basis of inheritance.

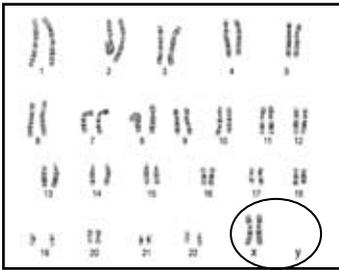
## GENES AND CHROMOSOMES

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.

Sometimes, a gene may not function well 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 like Stargardt disease.



*Male Chromosomes*



*Female Chromosomes*

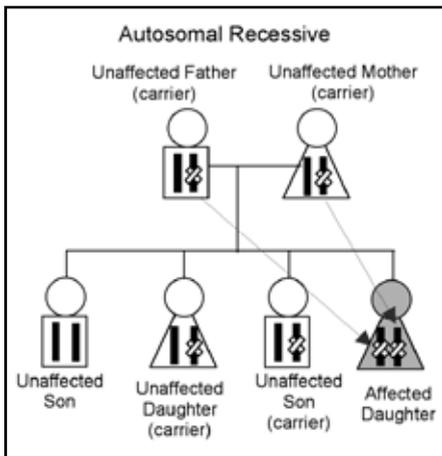
## INHERITANCE OF STARGARDT DISEASE

For most families with Stargardt disease, the inheritance pattern is autosomal recessive. Recently a small number of families were found to have an autosomal dominant pattern of inheritance. This form is called a “Stargardt-like” disease, and looks similar to the autosomal recessive form. These two forms are actually different diseases with different mechanisms. An accurate family tree or gene testing can help distinguish one from the other.

Autosomal recessive Stargardt disease is caused by mutations in a gene called *ABCA4*. A second gene called *ELOVL4* has been found to be the cause of the autosomal dominant form of Stargardt-like disease. Severity of disease may be related to how severely a gene change affects gene function. Sometimes, a gene is also influenced by other genes or the environment. These factors help explain why variation exists within the same family. Patients with questions about their personal form of Stargardt disease should be guided by experienced health professionals, such as an ophthalmologist or genetic counselor, who know about genetics and Stargardt disease.

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

In **autosomal recessive** inheritance, 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 recessive inheritance are:

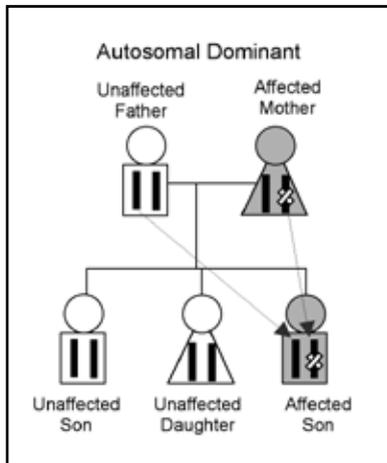


- Typically only one generation is affected.
- Both males and females can be affected.
- Individuals with one normal gene and one mutated gene generally don't show symptoms of Stargardt disease and are called "carriers."
- When both parents are carriers, there is a 25% chance (with each pregnancy) the child will have Stargardt disease.
- It is possible for only one person in a family to have Stargardt disease if that person's form is autosomal recessive.

Often, a person diagnosed with Stargardt disease has no other family members with the disease. There are several possible reasons for finding only one person with Stargardt disease in the family:

- The Stargardt disease mutations were new events in that person.
- Other family members have the same mutations, but have not been diagnosed with Stargardt disease. 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 Stargardt disease, carriers may be present in the mother's and father's side of the family for several generations, but a child won't develop Stargardt disease unless both parents are carriers and both pass on a mutation to their child.

In a few rare families, an **autosomal dominant** form of Stargardt-like disease has been seen. Autosomal dominant inheritance occurs when just one copy of a gene mutation causes Stargardt disease. The mutation causes Stargardt 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 Stargardt disease, he or she has a 50% chance with each pregnancy of having a child affected with Stargardt disease.

In some of the families with autosomal dominant Stargardt-like disease, not every person in the family is affected in the same way. Some may be diagnosed in childhood, while others might not be diagnosed until later in life.

## **GENETIC TESTING FOR STARGARDT DISEASE**

Testing a person's DNA is an important part of diagnosing Stargardt disease. When genetic testing is performed, genetic information is obtained from a sample of blood. The gene for Stargardt disease can then be examined to see if "typos" or mutations can be found that are responsible for the disease. Genetic testing for both the dominant and recessive forms of Stargardt disease is available. Mutations may not be found in every person tested, even if the person has clinical signs of Stargardt disease. It is possible that there are other genes for Stargardt disease that have not yet been found.

# Research into Stargardt Disease

## GENETIC RESEARCH

There are many Stargardt disease-causing mutations known in the *ABCA4* gene, and more are being discovered all the time. Participating in Stargardt disease research may help researchers find new Stargardt disease-causing genes or develop a better understanding of the disease. This information can be used to produce better treatments and ultimately a cure for Stargardt disease. Studies of many members of the same family may also be useful to identify Stargardt-causing genes. Sometimes, if we know the mutation causing your Stargardt disease, 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 Stargardt 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 Stargardt 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 Stargardt Disease

It may be very hard to deal with a diagnosis of Stargardt disease. Many people have never heard of the disease and are unsure how Stargardt disease 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 Stargardt disease lead successful and full lives. There are people to help you along the way. Below are suggestions for those desiring help with low vision adaptation, if and when they are needed.

## **LIFE WITH STARGARDT DISEASE**

It is perfectly natural for someone with vision loss to feel anxious, fearful, angry, or unhappy. If your child has Stargardt disease, it is also natural to have feelings of uncertainty and anxiety 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 Stargardt disease may be discouraged because there is currently no cure for the disease and very few treatment options. But if you or a family member is affected by Stargardt disease, there are many ways to take an active role in managing your

condition and live as independently as possible. The following pages offer several suggestions and sources of information for living with Stargardt disease.

## **LIVING WITH STARGARDT DISEASE IN CHILDHOOD**

If you are a parent of a child with Stargardt disease, 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 extra 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 greatly impact your child's views of living with Stargardt disease and expectations for the future. It's important to maintain a sense of hope with your child, and never to 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 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 meeting (IEP) 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 Stargardt disease. 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, or homework printed in larger text. 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 groups.

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 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 begin to become more independent, parents can help them learn to advocate for their own needs.

## **LIFE WITH STARGARDT DISEASE AS AN ADULT**

Most adults describe living a full and interesting life despite the practical and social problems that can arise because of Stargardt disease. Although many individuals with Stargardt disease 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 Stargardt disease can and do have a wide variety of jobs, from computer programmer to musician, and often find it unnecessary to change careers because of Stargardt disease. 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 later stages of disease, some people with Stargardt disease 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 Stargardt disease. 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 just as they can for anyone at this stage of life. For example, people with Stargardt disease may still be at risk for age-related eye conditions like glaucoma. It is important to continue regular checkups with your eye doctor and primary care physician. Individuals with Stargardt disease of any age may find support groups helpful.

## Talking to Others about Stargardt Disease

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.

# Low Vision Clinics

## 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 standard exams. 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 very small number of patients in very late stages of Stargardt 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 retina 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 resources for individuals with Stargardt. 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 Stargardt disease and about living with low vision. These groups may also put you in contact with others in your area with Stargardt disease. 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.

- 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)
- 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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