



**Prevent
Blindness**

Our Vision Is Vision.

Stargardt Disease

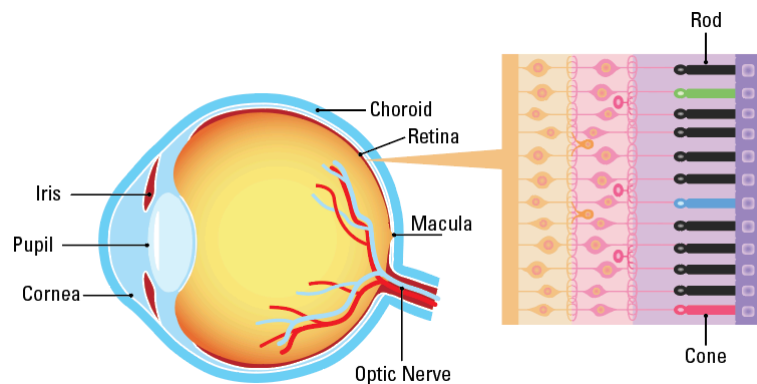
Factsheet 1 of 2 in a series. Find the rest of the series at PreventBlindness.org/living-with-stargardt



Photo credit: Partners for Pediatric Vision

What is Stargardt disease?

Stargardt disease, also called Stargardt macular dystrophy, causes vision loss due to a buildup of fatty material in the **macula**. The macula is a small area in the center of the **retina** that is responsible for sharp, straight-ahead vision. Stargardt disease is a rare eye genetic disease – part of a group of conditions called inherited retinal diseases (or IRDs), generally caused by a variation in one or more **genes**, causing the gene to not work properly. Stargardt disease affects about 1 in 10,000 people in the United States.¹



This disease typically causes central vision loss in both eyes during childhood or adolescence, but sometimes it occurs later in life. Central vision loss can cause difficulty recognizing faces and reading. Side (peripheral) vision is usually not affected, so it is rare for a person with Stargardt disease to lose all of their vision.

The rate of vision loss is different for each person. For some, Stargardt disease starts out slowly, then speeds up and levels off. At about 20/40 vision (meaning someone sees at 20 what a person with typical vision sees at 40 feet), vision loss can speed up. Vision may rapidly get worse until it reaches about 20/200.

After this point, vision usually stays about the same and cannot be corrected with eyeglasses, contact lenses or refractive surgery.

Stargardt Disease

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What are genes?



Cells are the basic building blocks of all living things.



Chromosomes are made up of **DNA**—the body's hereditary material.



A **gene** is a small section of DNA that contains the instructions for a specific molecule in the body, which is passed from parent to child.



The command center of each cell is called the **nucleus**, and it contains **chromosomes**.



Each gene contains the information required to build specific **proteins** needed in the body, for example, proteins build bones, determine eye color, allow muscles to move, control digestion, and keep your heart beating.

If there is a change in a gene's DNA sequence, it is called a **variant**. Not all variants cause disease. A disease-causing variant is called a **mutation**.

What causes Stargardt disease?

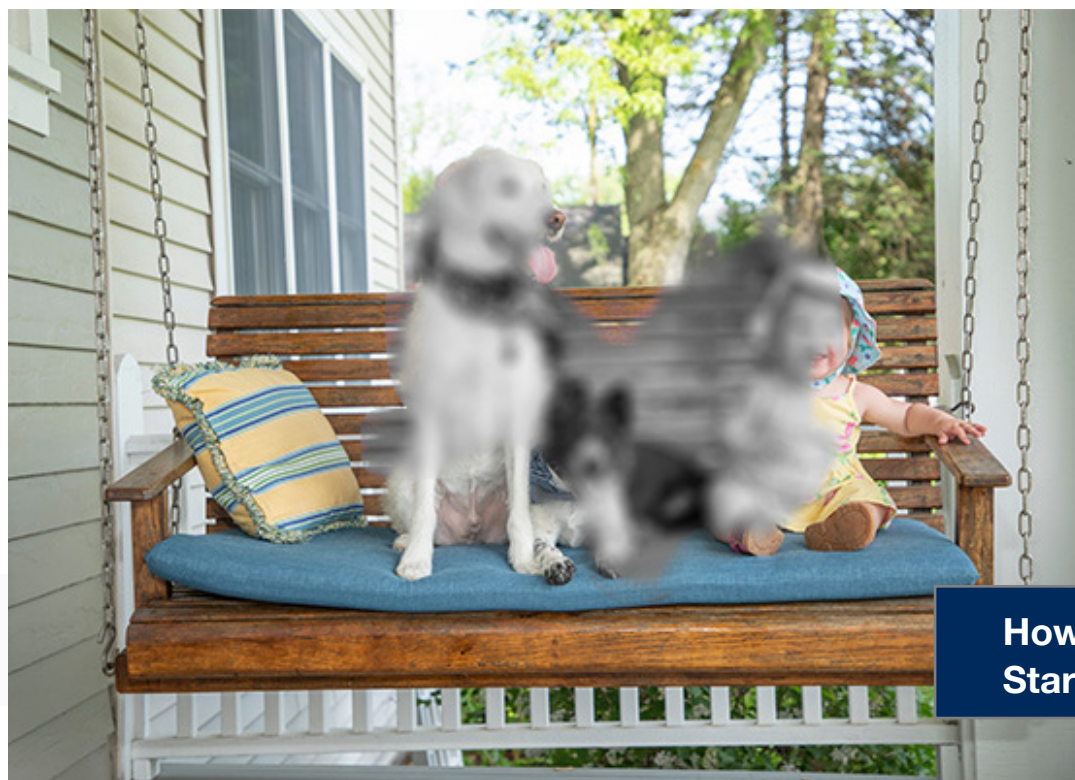
Stargardt disease is most commonly caused by a variant in the ABCA4 gene. The variant gene is what causes the build-up of extra fatty material in the retina.

Normally, when the retina processes light, there is leftover fatty material that gets taken away by different parts of the eye. With Stargardt disease, the fatty material builds up and damages the photoreceptors. **Photoreceptors (rods and cones)** are a special part of the retina used to process light and help us see. When our photoreceptors don't work well, there can be problems with vision.

The mutated ABCA4 gene must be passed down from both parents in order for it to cause eye problems.

Stargardt Disease

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How a person with Stargardt disease sees.

If only one parent has the mutated ABCA4 gene, then their child will be a carrier of the disease but won't have the disease themselves, and their vision will not be affected. Both parents must pass on the variant ABCA4 gene in order for the child to have vision loss due to Stargardt disease.

There are a few other genes in which a mutation can cause retinal disease that looks similar to Stargardt disease on eye examination and have similar symptoms. These genes include PRPH2, ELOVL4, and PROM1. Mutations in these genes are inherited differently. Depending on which gene is responsible for a person's eye condition, it may have different effects on disease

severity and different risk to other family members.

To learn more about how IRDs occur, visit: [PreventBlindness.org/inherited-retinal-diseases](https://www.preventblindness.org/inherited-retinal-diseases).

What symptoms might I have with Stargardt disease?

The most common symptom of Stargardt disease is a slow loss of central vision in both eyes. Some people lose their central vision more quickly than others. Rarely, a small number of people with Stargardt disease may also lose their side (peripheral) vision. It is unusual for someone to lose all vision due to Stargardt disease.

Stargardt Disease

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The age when blurry vision starts can predict how much vision will change as they age. For example, an individual with Stargardt disease who has blurry vision earlier in life will usually have worse vision than someone with Stargardt disease whose vision becomes blurry when they are older.

Other symptoms may include:

- Dark, gray, black, or hazy spots in the center of your vision
- Sensitivity to light
- Blurry vision that cannot be corrected with glasses
- Difficulty seeing small details
- Needing more time for your eyes to adjust between light and dark places
- Difficulty seeing in the dark or low light
- Color blindness
- Lack of depth perception
- Lack of contrast

You may not experience all the symptoms from the list above. Symptoms for each person will vary depending on your age, stage of the disease, and genetic variation.

How is Stargardt disease diagnosed?

Diagnosis of Stargardt disease may take longer than other, more commonly occurring vision conditions. The diagnosis typically starts during an eye examination with an eye doctor. If the eye doctor thinks that you have an IRD, such as Stargardt disease, they will ask you to see a retina specialist or an eye doctor more experienced in diagnosing and treating IRDs. The retina specialist will conduct further testing to confirm your diagnosis. They will conduct a very detailed examination including the steps listed below. The goals of the detailed examination are to:

- Establish the specific diagnosis.
- Get the treatment that is right for that diagnosis.
- Connect the patient to supportive services such as a genetic counselor (someone that specializes in the genetic testing process and educating patients) and vision rehabilitation.
- Educate other family members about their risk for the disease.
- Provide information to the patient about clinical trials and new therapies.

The initial and follow-up examinations may include the following:

- **Patient history:** Including current vision issues, patient medical history, current medications, and past medication use.

Stargardt Disease

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- **Family history:** Constructing a family medical history to show the genetic relationships and medical disorders that occur in a family. From the family medical history, patterns of familial disorders and how diseases were passed down may emerge. This leads doctors to a clear diagnosis of the genetic disorder and allows assessments for family members who may also be at risk for the disorder.
- **Imaging:** The eye doctor will conduct a series of imaging tests to view different parts of the eye, including:
 - **Retinal photographs:** A photo taken of the inside surface of the eye.
 - **Optical Coherence Tomography (OCT):** A test that takes cross-sectional pictures of the retina. This allows your ophthalmologist to map and measure the thickness of the retina.
 - **Fundus autofluorescence:** A test used to create a density map of the layers of the retina.
 - **Infrared autofluorescence:** A test that provides information on the distribution of pigment in the retina.
 - **Fluorescein angiography:** A test that uses a fluorescent dye injected in the vein of the arm or hand and pictures taken of the retina through dilated pupils.

Clinical eye examination: The eye examination includes:

- **Dilation:** Eye drops are put into the eye to widen the opening on the front of the eye, called the pupil. This allows the eye doctor to examine the health of the retina.
- **Visual Acuity:** A measure of the ability to see detail up close and in the distance.
- **Color vision testing:** Stargardt disease can cause color blindness, so your eye doctor may also test your color vision.
- **Slit lamp:** A non-invasive procedure that uses a microscope and bright light to look at different parts of the eye.
- **Indirect ophthalmoscopy:** A non-invasive tool worn by the eye doctor that provides magnification, allowing them to examine the back of the eye.
- **Visual Field Testing:** A test that measures central and side or peripheral vision.
- **Electroretinography (ERG):** A test to measure the electrical response of the eye's light-sensitive cells, called rods and cones. During the test, the eye doctor will place drops into your eyes, so you will not have any discomfort during the test.

Stargardt Disease

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Why is Genetic Testing Important?

Identifying the genetic cause of disease is an important part of care for patients with Inherited Retinal Diseases (IRDs). Many times, the exact type of IRD a person has can be difficult to determine based only on tests conducted in the eye doctor's office. Results from genetic testing can lead to an accurate diagnosis if there is a positive finding. Having the genetic diagnosis may help to:

- Identify potential treatment options or clinical trial options for patients.
- Inform them about the potential risk of disease to other family members.
- Identify the potential risk to other organs in the patient's body that may be affected.
- In the case of infants and young children, genetic testing will identify those children who are at risk of other health problems and who will benefit from early diagnosis and therapy.
- Inform a person about what will happen to their vision over time.

A health care provider will order the genetic test, collect the sample, and review the results with the patient. Ideally, the health care provider would include a genetic counselor to guide the patient and their family through the results of the genetic testing, discuss the impact on other family members, and assist couples who have questions around family planning decisions. It may be helpful to have genetic testing done if you are interested in joining a clinical trial, or if it has been several years since your last genetic test was completed.

- **Genetic testing:** Patients may meet with a genetic counselor (someone that specializes in the genetic testing process and educating patients) to determine the testing approach that is best based on the other test results. Patients are often asked to provide a blood or saliva sample that will be sent to a lab for analysis. The genetic test confirms or disputes the diagnosis of a specific IRD, so that correct information is provided to the patient and family. Genetic testing is often a requirement for participation in IRD-related clinical research trials.

What is the treatment for Stargardt disease?

There is currently no FDA-approved treatment for Stargardt disease, however, there are several clinical trials in process that may lead to treatment options. Continue to see your eye doctor every 1-2 years or as recommended. It is important to check your eyes regularly as other eye health conditions can occur.

Stargardt Disease

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“[Stargardt is] progressive. That means you know that things are going to get challenging, but with the right mindset you will be able to overcome adversity.”

– Charlie C., motivational speaker and Stargardt patient

If at any time you feel there has been a change in your vision, you should see an eye doctor and have a dilated eye examination. It is also important to have a current record of an eye exam if you would like to join a clinical trial.

There are steps you can take now to protect your eyes and slow vision loss:

- Do not smoke and try to avoid secondhand smoke.
- Wear a hat and sunglasses to protect your eyes from sunlight when you go outside.
- Learn about how vitamin A affects your health. **Do not take dietary supplements with vitamin A.**
- Get **protective sport eyeglasses** ([PreventBlindness.org/tips-for-buying-sports-eye-protectors](https://www.preventblindness.org/tips-for-buying-sports-eye-protectors)) when playing sports or working in or around potentially dangerous conditions. This includes work in manufacturing, construction, landscaping, auto repair, plumbing, woodworking, agriculture, mining, metal fabrication and health care.
- Ask your eye doctor about participating in a **clinical trial** ([PreventBlindness.org/clinical-trials-for-eye-diseases-and-vision](https://www.preventblindness.org/clinical-trials-for-eye-diseases-and-vision)).

Find more information about Stargardt disease.

- **National Eye Institute – Stargardt Disease:** [NEI.NIH.gov/learn-about-eye-health/eye-conditions-and-diseases/stargardt-disease](https://www.nei.nih.gov/learn-about-eye-health/eye-conditions-and-diseases/stargardt-disease)
- **Foundation Fighting Blindness** [FightingBlindness.org/diseases/stargardt-disease](https://www.fightingblindness.org/diseases/stargardt-disease)
- **Your North Starg:** [YourNorthStarg.com](https://www.yournorthstarg.com)

Stargardt Disease

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Resources for those with Stargardt Disease:

Vision rehabilitation center: (LowVision.PreventBlindness.org/finding-low-vision-rehabilitation-services) Find a vision rehabilitation center so you can learn how to maximize the use of the vision do have to help you work, do daily activities, and do the things you enjoy.

Living Well with Low Vision: Prevent Blindness offers resources and help finding low vision clinics near you and clinical trials for individuals with vision loss on the Living Well with Low Vision website: (LowVision.PreventBlindness.org).

Blind Life: Blind Life website (TheBlindLife.net) and YouTube (YouTube.com/c/theblindlife) channel provides videos, tips, and insight for those living with vision loss.

Thriving Blind Academy: Thriving Blind Academy (ThrivingBlindAcademy.org) is dedicated to promoting literacy, employment and financial awareness in the blind community. Free and paid programs are available for people of all ages.

Hadley: Hadley (HadleyHelps.org) offers practical help, connection and support free of charge to anyone with a visual impairment, their families and professionals supporting them.

Guide Dogs of America: Many patients with Stargardt disease and vision loss benefit from use of a guide dog. A guide dog is a specially trained service animal that helps an individual with vision loss or blindness. Information on guide dog services in the United States can be found at GuideDogsofAmerica.org, and GuideDog.org.

Stargardt Disease Masterclass taught by Bradford Manning, Founder of Two Blind Brothers: Didit.org/courses/bradford-manning-teaches-stargardt-disease

Participate in a support group for patients with Stargardt disease or connect to an online community.

- Global Genes: GlobalGenes.org
- MD Support: MDSupport.org
- Eye2Eye: Eye2Eye (SHP.Rutgers.edu/psychiatric-rehabilitation/eye2eye) is a free, phone-based, peer support program, designed to assist adults who are blind or visually impaired and their families.

Questions to ask your doctor:

- How do I manage living with Stargardt disease?
- Can you suggest a vision rehabilitation center in my area?
- How often do you recommend getting an eye exam?
- Are there foods with vitamin A I should avoid?
- Am I eligible to take part in a clinical trial?
- Should I speak to a genetic counselor and get genetic testing?
- Are there eyeglasses, contact lenses, or a combination of the two that can help me see a little clearer?
- Can you suggest things that will help me cope with my vision loss?

Complete genetic testing.

Learn more about IRDs and benefits of genetic testing at [FightingBlindness.org/genetic-testing](https://fightingblindness.org/genetic-testing)

Find a provider to help you access genetic testing: The Foundation Fighting Blindness's list of Retina Doctors ([FightingBlindness.org/retinal-specialists](https://fightingblindness.org/retinal-specialists)) makes it easy to find a genetic testing specialist or health care provider in your area of the United States who can discuss your testing options and assist you in learning more about your IRD.

Stay up to date on clinical trials.

Stay up to date on clinical trials. Learn more about clinical trials for Stargardt disease at [ClinicalTrials.gov](https://clinicaltrials.gov)

- Learn about current clinical trials for Stargardt disease: [FightingBlindness.org/research/stargardt-disease-research-advances-6](https://fightingblindness.org/research/stargardt-disease-research-advances-6).
- Search studies in the U.S. that are currently active or recruiting people aged birth to 65+ who have Stargardt disease: [ClinicalTrials.gov/ct2/results?cond=Stargardt+Disease&country=US&Search=Apply&recrs=a&recrs=d&age_v=&age=0&age=1&age=2&gndr=&type=&rslt=](https://clinicaltrials.gov/ct2/results?cond=Stargardt+Disease&country=US&Search=Apply&recrs=a&recrs=d&age_v=&age=0&age=1&age=2&gndr=&type=&rslt=)

Continued in Factsheet 2: Living With Stargardt Disease.

Reference:

1. Farnoodian M., et al (2022). Cell-autonomous lipid-handling defects in Stargardt iPSC-derived retinal pigment epithelium cells. *Stem Cell Reports*, 17(11) 2438-2450. <https://doi.org/10.1016/j.stemcr.2022.10.001>

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