

What is Stargardt disease?

Stargardt disease is the most common form of inherited macular degeneration. About 30,000 people in the U.S. have the condition. The progressive vision loss associated with Stargardt disease is caused by the death of photoreceptor cells in the central portion of the retina called the macula.

The retina is the thin, light-sensing tissue lining the back of the eye. Photoreceptor cells in the retina provide vision by converting light into electrical signals which the brain perceives as vision. The macula is responsible for central vision and perceiving details — for tasks like reading, watching television, and looking at faces.

Decreased central vision is a hallmark of Stargardt disease. Side vision is usually preserved. Stargardt disease typically develops during childhood and adolescence.

What are the symptoms?

The symptom that brings most people to an eye doctor is a change in central vision. A decrease in color perception can also occur. A doctor examining the retina of a person with Stargardt disease will see characteristic yellowish flecks, called lipofuscin, under the macula.

Lipofuscin is a fatty byproduct of normal retinal cell activity. In Stargardt disease, lipofuscin is not cleared properly from the retina and accumulates, leading to degeneration. The Foundation Fighting Blindness supports research to better understand lipofuscin buildup and ways to prevent it.

How quickly does vision fade?

The progression of symptoms in Stargardt disease varies from patient to patient. Visual acuity (the ability to distinguish details) may decrease slowly at first, accelerate, and then level off.

A study of 95 people with Stargardt disease showed that once a visual acuity of 20/40 is reached, there is often rapid progression of additional vision loss until it reaches 20/200. (Normal vision is 20/20. A person with 20/40 vision sees at 20 feet what someone with normal vision sees at 40 feet.) By age 50, approximately 50 percent of people in the study had visual acuities of 20/200 or worse.

Eventually, most people with Stargardt disease have a visual acuity in the range of 20/200 to 20/400.

Is it an inherited disease?

In about 95 percent of cases, Stargardt disease is inherited as an autosomal

STARGARDT DISEASE, Continued

recessive trait. It is inherited when both unaffected parents, called carriers, have one mutated copy of the Stargardt-disease gene (ABCA4) and one normal gene. Each child has a 25-percent chance of inheriting mutated copies of ABCA4 from both parents. Carrier parents are unaffected, because they have only one mutated copy of the gene.

Genetic counselors are an excellent resource for discussing inheritability, family planning, genetic testing, and other issues related to Stargardt disease.

What treatment is available?

While there are currently no treatments for Stargardt disease, the Foundation is supporting several promising avenues of research, including gene, stem-cell, and drug therapies.

UV-blocking sunglasses are generally recommended for outdoors. For people who already have significant vision loss, low vision aides are available.

For the latest research advances for Stargardt disease treatments, refer to the Foundation publication Stargardt Disease: Research Advances.

Are there any related diseases?

Stargardt disease is also known as Stargardt macular dystrophy or fundus flavimaculatus.

Low-vision resources and extensive information on research and clinical trials for Stargardt disease are available at:
www.FightingBlindness.org