



Stargardt macular dystrophy is an inherited eye disorder, where people experience a progressive blurring of their central vision. Vision loss often begins in the teenage years. Approximately one in 10,000 Canadians has Stargardt disease.

The symptoms of Stargardt disease are similar to age-related macular degeneration, although the causes are different. Since the symptoms begin at a younger age, it is sometimes called a juvenile macular degeneration. The condition may also be called fundus flavimaculatus.

Early Symptoms

People with Stargardt disease are usually diagnosed before age 20 although a few may not notice any significant vision loss before the age of 30 or 40. Symptoms include a gradual loss in the person's ability to see fine detail and to distinguish faces and colours.

Medical Description

Stargardt disease most severely affects the macula, the central portion of the retina. In Stargardt disease, a substance called lipofuscin slowly builds-up amongst the retinal pigment epithelial (RPE) cells of the retina. RPE cells normally clear away waste products like lipofuscin, but in Stargardt disease they are overwhelmed. The RPE cells become increasingly unable to clear the lipofuscin and unable to support and nourish the light-sensing cells of the retina called photoreceptors, particularly the cone photoreceptors in the center of the

retina. Cone photoreceptors allow people to see fine details and colour.

An eye care professional, examining the retina of a person with Stargardt disease may see yellowish spots in, and under, the macula. These spots are deposits of lipofuscin.

Diagnosis

If changes in visual acuity (the clearness of vision) have been detected, and an ophthalmologist suspects Stargardt disease, these tests may clarify the diagnosis:

Fluorescein Angiogram uses a dye, called fluorescein, to examine the back of the eye. The dye is injected into the arm and a camera records as the dye passes through the blood vessels in the back of the eye. Patients with Stargardt disease usually show a "dark choroid effect," due to a buildup of lipofuscin under the retina, which makes the background of the retinal images appear darker.

Visual Field Testing allows the doctor to detect and monitor blind spots in vision, by having the patient record where they see (and don't see) flashes of light. This shows where and how vision is affected.

ERG (electroretinography) measures the electrical responses of the retina to light. During this test, a special contact lens is placed on the eye, and the eye is exposed to flashes of light. This helps distinguish Stargardt disease from other conditions.

These tests are usually sufficient to diagnose Stargardt disease, however genetic testing may be used to confirm the diagnosis. Genetic counselling may

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also help you and your family better understand the condition.

What to Expect

As Stargardt disease advances, a person's vision becomes less distinct, and a blind spot in the central vision may develop and grow. By mid-life, most people are legally blind due to central vision loss however peripheral vision is usually minimally affected and remains throughout life. Low vision aids can help maximize this vision. In the later stages of Stargardt disease, colour perception is often affected.

The Genetics of Stargardt Disease

Stargardt disease is a genetic disease caused by mutations in one large gene *ABCA4*. These mutations stop the *ABCA4* gene from producing a protein needed for vision and ultimately lead to the build-up of lipofuscin in the retina.

In most cases, the parents of people with Stargardt disease each had one damaged *ABCA4* gene. A child that inherits a damaged gene from each parent will be affected. This is autosomal recessive inheritance. Genes divide randomly, so about a quarter of children in these families will have Stargardt disease, but the chance of having an affected child is 25% for each pregnancy. However the risk of a person with Stargardt disease having an affected child is very low.

There is another condition called Stargardt-like disease. It is due to mutations in the *ELOVL4* gene. In this case, even one damaged gene is enough to cause the condition. This is called autosomal dominant inheritance.

Stargardt-like disease can occur in multiple generations of a family. Genetic testing and counselling can distinguish between these conditions.

Treatment

No treatments are currently approved to prevent or slow the vision loss associated with Stargardt disease. However, it is important to have regular eye exams even if your vision is not changing – to avoid serious but treatable complications that might further impair your vision, such as macular edema.

Research

Many research groups are working to develop treatments for Stargardt disease. Two types of treatment have already reached the stage of clinical trials.

Gene therapy refers to treatments that aim to place healthy genes into retinal cells, replacing the gene mutation. This might stop or slow the build-up of lipofuscin, preventing further vision loss. One pharmaceutical company, Oxford Biomedica, has begun safety testing in humans of a gene therapy called StarGen. Please see our gene therapy fact sheet for more information.

A second set of trials is exploring the **transplant of new RPE cells** derived from stem cells to help the retina clear the lipofuscin build-up. This might prevent or slow further vision loss. Early trials of this treatment have begun. Please see our stem cell fact sheet for details.

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Updated February 15 2013: Reviewed by *Dr. Nupura Bakshi, Mount Sinai Hospital and St. Michael's Hospital, University of Toronto*