Stargardt's Disease
(Fundus Flavimaculatus)

While macular degeneration generally is associated with aging eyes, an inherited form known as Stargardt's disease can affect children and young adults.

Stargardt's disease — also called fundus flavimaculatus or Stargardt's macular dystrophy (SMD) — affects approximately one in 10,000 people and is characterized by central vision loss early in life. (Some researchers believe a distinction should be made between Stargardt's disease and fundus flavimaculatus, because they say each describes a different variant of the eye disease.)

Stargardt's generally refers to a group of inherited diseases causing light-sensitive cells in the inner back of the eye (retina) to deteriorate, particularly in the area of the macula where fine focusing occurs. Central vision loss also occurs, while peripheral vision usually is retained.

Stargardt's disease is diagnosed by the presence of small, yellowish spots of deteriorating tissue (drusen) sloughed off from the colored or outer covering of the retina (retinal pigment epithelium). Progressive vision loss eventually leads to blindness in most cases.

What Causes Stargardt's Disease?

Stargardt's is an inherited disease passed along to children when both parents carry mutations of a gene associated with vitamin A processing in the eye. Parents can carry recessive genetic traits responsible for Stargardt's, even though they themselves may not have the disease.

Researchers have found that about 5 percent of the human population carry gene mutations causing inherited retinal diseases such as Stargardt's and retinitis pigmentosa.*

How Fast Does Stargardt's Progress?

Vision loss from Stargardt's generally begins to show up within the first 20 years of a young person's life, particularly in early childhood.

But it's difficult to pinpoint exactly when retinal damage will occur or how fast it will progress, because variations can occur even among family members with similar inherited tendencies.

For example, one study reported in the May 2006 issue of American Journal of Ophthalmology noted the case of three siblings, two of whom showed signs of retinal disease in early childhood. But the third sibling was affected beginning at around age 19.

In some cases, a person with Stargardt's (particularly the fundus flavimaculatus version of the disease) may reach middle age before vision problems are noticed.

The reference book Ophthalmology notes that vision loss from Stargardt's as measured on a standard eye chart can range between 20/50 and 20/200. (In the United States, legal blindness is defined as visual acuity of 20/200 or worse while wearing corrective lenses.) Those who have the fundus flavimaculatus form of the disease, however, are likely to experience even more severe vision loss.
Symptoms of Stargardt's disease can include blurry or distorted vision, inability to see in low lighting and difficulty recognizing familiar faces. In late stages of Stargardt's, color vision also may be lost.

**Can Stargardt's Disease Be Prevented or Treated?**

Some research indicates that exposure to bright light may play a role in triggering the retinal damage that occurs with Stargardt's. While there is no known treatment for Stargardt's disease at this time, people with the condition often are advised to wear eyeglasses or sunglasses that block 100 percent of UV light to reduce the possibility of additional eye damage caused by the sun.

If you have Stargardt's, your eye doctor also may suggest that you wear eyeglasses with specially tinted lenses to block certain wavelengths of light.

Retinal pigment epithelial (RPE) stem cells such as these are being used in human clinical trials to regenerate and support function of the eye's light-sensitive cells damaged by Stargardt's disease. (Image: Advanced Cell Technology)

One company developing a treatment for Stargardt's disease is Advanced Cell Technology (ACT). In March 2010, the company received an orphan drug designation from the U.S. FDA for a stem cell treatment designed to protect and regenerate photoreceptors in the retina that are damaged by eye diseases like Stargardt's.

The stem cells have been tested in animal models of eye disease, chief scientific officer Robert Lanza, MD, said in a company statement released in November 2010.

"In rats, we've seen 100 percent improvement in visual performance over untreated animals without any adverse effects," Lanza said. "Our studies showed that the cells were capable of extensive rescue of photoreceptors in animals that otherwise would have gone blind. Near-normal function was also achieved in a mouse model of Stargardt's disease. We hope to see a similar benefit in patients with various forms of macular degeneration."

In July 2012, ACT provided an update of its Phase I/II clinical trial to evaluate the safety and tolerability of the treatment. That month, the fourth patient in the study received an intraocular injection of 100,000 human embryonic stem cell-derived retinal pigment epithelial (RPE) cells. The first three patients in the study received injections of 50,000 cells.

A total of 12 Stargardt's patients will participate in the study, with cohorts of three patients each in an ascending dosage format. Results of the study will be released when all participants have been followed for a period of 12 months after receiving the injections.

Studies suggest aggregation or "clumping" of vitamin A in the retina may be associated both with Stargardt's disease and age-related macular degeneration (AMD). These clumpy deposits are known as "vitamin A dimers."

In studies conducted at Columbia University Medical Center (New York) in 2011, researchers created an altered form of vitamin A that appears to slow the formation of vitamin A dimers in the eye when given to mice with the same genetic defect as humans with Stargardt's disease.

Also, the modified vitamin A behaved exactly as normal vitamin A does in all other aspects, making it an attractive potential therapy for preventing blindness in humans, the researchers said.

The research team was led by led by Ilyas Washington, PhD, a professor in the department of ophthalmology at Columbia's Harkness Eye Institute.
Dr. Washington's lab has been awarded a $1.25 million grant from the National Eye Institute to further investigate the link between vitamin A dimers and various retinal degenerations, which could result in new approaches to treat these diseases.

Alkeus Pharmaceuticals (Cambridge, Mass.) has licensed from Columbia certain patents relating to Dr. Washington's discoveries and intends to launch clinical trials for the treatment of Stargardt's disease and AMD in the near future.

**Coping With Stargardt's Disease**

The American Macular Degeneration Foundation recommends that people with Stargardt's or a history of the eye disease in close family members obtain genetic counseling before starting their own families.

Because vision loss often appears in young children with Stargardt's, low vision counseling from your eye doctor is essential to make sure that classroom learning is not hampered. For example, a child with Stargardt's may need to use large print books and special devices that magnify print. [Read more about low vision devices.]