

Stargardt macular degeneration

Description

Stargardt macular degeneration is a genetic eye disorder that causes progressive vision loss. This disorder affects the retina, the specialized light-sensitive tissue that lines the back of the eye. Specifically, Stargardt macular degeneration affects a small area near the center of the retina called the macula. The macula is responsible for the type of vision needed for detailed tasks such as reading, driving, and recognizing faces. In most people with Stargardt macular degeneration, a fatty yellow pigment called lipofuscin builds up in cells underlying the macula. Over time, the abnormal accumulation of this substance can damage cells that are critical for clear vision. People with Stargardt macular degeneration have problems with night vision that can make it difficult to navigate in low light. Some affected individuals also have impaired color vision. The signs and symptoms of Stargardt macular degeneration typically appear in late childhood to early adulthood and worsen over time.

Frequency

Stargardt macular degeneration is the most common form of juvenile macular degeneration, the signs and symptoms of which begin in childhood. The estimated prevalence of Stargardt macular degeneration is about 1 in 6,500 individuals.

Causes

In most cases, Stargardt macular degeneration is caused by variants (also called mutations) in the *ABCA4* gene. Less often, variants in the *ELOVL4* gene cause this condition. The *ABCA4* and *ELOVL4* genes provide instructions for making proteins that are found in light-sensing cells (photoreceptors) in the retina.

The *ABCA4* protein transports potentially toxic substances out of photoreceptors. These substances form after phototransduction, the process by which light entering the eye is converted into electrical signals that are transmitted to the brain. Variants in the *ABCA4* gene prevent the *ABCA4* protein from removing toxic byproducts from photoreceptors. These toxic substances build up and form lipofuscin in the photoreceptors and the surrounding cells of the retina, eventually causing cell death. Loss of cells in the retina causes the progressive vision loss that is characteristic of Stargardt macular degeneration.

The ELOVL4 protein plays a role in making a group of fats called very long-chain fatty acids. The ELOVL4 protein is primarily found in the retina, but is also expressed in the brain and skin. The function of very long-chain fatty acids within the retina is unknown. Variants in the *ELOVL4* gene lead to the formation of ELOVL4 protein clumps (aggregates) that build up and may interfere with retinal cell functions, ultimately leading to cell death.

[Learn more about the genes associated with Stargardt macular degeneration](#)

- ABCA4
- ELOVL4

Inheritance

Stargardt macular degeneration can have different inheritance patterns.

When variants in the *ABCA4* gene cause this condition, it is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell must have a variant to cause the disorder. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

When this condition is caused by variants in the *ELOVL4* gene, it is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Juvenile macular degeneration
- Macular dystrophy with flecks, type 1
- Stargardt disease
- STGD

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Severe early-childhood-onset retinal dystrophy (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1855465/>)
- Genetic Testing Registry: Stargardt Disease 3 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1838644/>)

[Genetic and Rare Diseases Information Center](#)

- Stargardt disease (<https://rarediseases.info.nih.gov/diseases/181/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Stargardt macular degeneration%22](https://clinicaltrials.gov/search?cond=%22Stargardt+macular+degeneration%22))

Catalog of Genes and Diseases from OMIM

- STARGARDT DISEASE 3; STGD3 (<https://omim.org/entry/600110>)
- STARGARDT DISEASE 1; STGD1 (<https://omim.org/entry/248200>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Macular+Degeneration%5BAJR%5D%29+AND+%28%28stargardt+macular+degeneration%5BTIAB%5D%29+OR+%28stargardt+disease%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

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