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My46 Trait Profile

Stargardt disease

Other Names: Juvenile Onset Macular Degeneration, Stargardt Macular Dystrophy

Stargardt disease is a genetic condition that is characterized by central vision loss early in life. This vision loss is not correctable with glasses. It is caused by mutations in the genes ABCA4 and ELOVL4, each of which makes a protein found in the eye.

Characteristics of Stargardt disease

Stargardt disease is the most common form of inherited juvenile macular degeneration (i.e., deterioration of the macula – the center part of the back of the eye). It affects about 1 in 8,000 to 10,000 individuals. The progressive central vision loss is due to problems in the cone cells in the back of the eye (retina). Cone cells are the cells responsible for central vision, color perception and perception to detail. If cone cells are not working properly, it can be difficult to read, recognize faces, and distinguish colors. People with Stargardt disease can have difficulty with all of these activities. The age of onset of symptoms can be highly variable, but generally individuals notice changes to their vision somewhere between late childhood and early adulthood.

There are two types of Stargardt disease: Stargardt type1 (STGD1) and Stargardt type 3 (STGD3). Usually an ophthalmologist who is familiar with the two types can identify which type an individual has after an examination. Information about family history can also help the healthcare provider understand which type of SD the individual has. STGD1 is much more common and people generally have no family history of Stargardt disease.

Diagnosis/Testing

Ophthalmologists can usually diagnose Stargardt disease during an ophthalmic examination using a device called a slit lamp. Often, a fatty yellow pigment called lipofuscin builds up in the macula, which can be seen on exam. In addition to ophthalmic exam, fluorescein angiography plays an important role in establishing the diagnosis. This test uses a dye that is injected into a patients arm to show how blood flows in the retina.

STGD1 is caused by a change or mutation in a gene called ABCA4. STGD3 is less common and is caused by mutations in the ELOVL4 gene. The ABCA4 and ELOVL4 genes make proteins that are found primarily in the retina. Mutations in these two genes do not allow the protein to function properly, and causes progressive vision loss.

Management/Surveillance

Management of Stargardt disease includes annual exams to evaluate changes in vision as well as an evaluation of overall eye health. Annual eye exams can also inform individuals of ongoing clinical studies that may be available for them to participate in.

It is not recommended for individuals with STGD1 to use any sort of vitamin A supplementation. This is because studies have shown that vitamin A can damage the retina of individuals with STGD1. For individuals with STGD3, fish oil supplementation may help slow the progression of the disease.

Low vision aids may also be helpful for individuals with Stargardt disease. There are low vision specialists affiliated with many major eye centers who can help assess the best tools based on the symptoms. Individuals with Stargardt disease can also discuss educational support plans with their local school or contact a licensed social worker for assistance.

Mode of inheritance

Stargardt disease may be inherited in one of two patterns of inheritance: autosomal recessive and autosomal dominant.

Autosomal recessive inheritance:

Stargardt disease type 1 (STGD1) is inherited in an autosomal recessive pattern. This means that an individual has to inherit two ABCA4 mutations (i.e., one from each parent) to be affected with STGD1. If both parents are carriers of a ABCA4 mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with STGD1. In about 30% of individuals with a clinical diagnosis of STGD1, only one disease causing mutation is found.

Autosomal dominant inheritance:

Stargardt disease type 3 (STGD3) is inherited in an autosomal dominant pattern. This means inheriting one ELOVL4 mutation is enough for an individual to be affected and show signs of Stargardt disease. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on the pattern of inheritance.

Autosomal recessive inheritance:

Parents of a child with STGD1 are carriers. If a sibling of a child with STGD1 is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier.

Autosomal dominant inheritance:

Since STGD3 is autosomal dominant, the risk to family members depends on whether or not the individual with STGD3 has an affected parent. If a parent also has the condition, the risk of having a child with STGD3 is 50% with each pregnancy. If a parent does not have the condition, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Genetics Home Reference: Stargardt disease
http://ghr.nlm.nih.gov/condition/stargardt-macular-degeneration
Foundation Fighting Blindness
http://www.blindness.org/
Kellogg Eye Center (Understanding Stargardt Disease, Information Book)
http://www.kellogg.umich.edu/patientcare/downloads/Understand-Stargardt.pdf
National Association for Parents of Children with Visual Impairments
http://www.napvi.org

References

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Koenekoop, RK. (2003)."The gene for Stargardt disease, ABCA4, is a major retinal gene: a mini-review." Ophthalmic Genetics 24(2): 75-80.

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