STARGARDT DYSTROPHY

Division of Human Genetics
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WHAT IS STARGARDT DYSTROPHY?

Stargardt dystrophy [STGD] is the most common form of inherited juvenile onset macular degeneration. The progressive vision loss associated with STGD is caused by the death of photoreceptor cells in the central portion of the retina called the macula.

The retina is the delicate light-sensing tissue lining the back inside wall of the eye. Photoreceptor cells in the retina provide vision by conveying information from the visual field to the brain. The macula is responsible for sharp central vision — for tasks like reading, watching television, and looking at faces.

Decreased central vision is a hallmark of STGD. Side vision (often referred to as peripheral vision) is usually preserved. STGD typically develops during childhood and adolescence. Also involved in STGD is a region beneath the macula called the retinal pigment epithelium [RPE].

WHAT ARE THE SYMPTOMS?

The symptom that brings most people to an eye specialist is a change in central vision. A doctor looking at the retina of a person with STGD will see characteristic yellowish flecks in and under the macula. The flecks might extend outward in a ring-like fashion. The flecks are deposits of Lipofuscin, a fatty by-product of normal cell activity. In Stargardt disease, Lipofuscin accumulates abnormally. The Lipofuscin build-up is thought to occur because of the inability of the RPE to get rid of excess waste products.

A decrease in colour perception may also occur in STGD. This is because photoreceptor cells involved in colour perception are concentrated in the macula.

HOW QUICKLY DOES VISION FADE?

The progression of symptoms in STGD is variable. Visual acuity (the ability to distinguish details and shape) may decrease slowly at first, accelerate, and then level off.

A study of 95 people with STGD 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% of people in the study had visual acuities of 20/200 or worse.

Eventually, almost everyone with Stargardt disease has a visual acuity in the range of 20/200 to 20/400. The vision loss is not correctable with prescription eyeglasses, contact lenses, or refractive surgery, but adaptive devices and high magnification lenses can make a huge difference. Adaptive computer programmes and electronic devices can increase access to communication. A low-vision therapist or access

technology expert should be consulted. People with STGD do not usually require mobility assistance as their peripheral vision usually remains unaffected.

IS IT AN INHERITED DISEASE?

STGD is almost always inherited as an autosomal recessive trait. It is inherited when both parents, called carriers, have one changed (mutated) copy of the disease-causing gene paired with one normal copy of the disease associated gene. Each child has a 25% chance of inheriting the two copies of the STGD causing gene (one from each parent). Carrier parents are unaffected because they have only one copy of the faulty gene. In 1997, researchers in the USA found an important disease-causing gene for STGD, ABCA4 (also referred to as the ABCR gene), which normally causes the production of a protein involved in the visual cycle. Lipofuscin build-up appears to be related to a mutation in this gene, and the resulting production of a dysfunctional protein. To date more than 600 different mutations (changes) in the ABCA4 gene have been found. Of these, seven mutations have been found that are quite common in the South African Afrikaner population. It is important to note that STGD, and ABCA4 mutations have been found in all our population groups.

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Genetic testing to identify the exact ABCA4 gene mutation is available through Retina South Africa. At-risk family members and spouses may also be tested for carrier status.

Genetic counsellors are an excellent resource for discussing inheritability, genetic risk assessment and management, family planning, career choices and other issues related to living with STGD and genetic counselling is a pre-requisite for genetic testing.

WHAT TREATMENT IS AVAILABLE?

There are currently no treatments for STGD, but two significant clinical trials have begun. One study is to replace the entire ABCA4 gene using a lenti virus for the delivery of the normal copy of the ABCA4 gene. The second is using stem cells to grow new RPE cells and then transplant them into the retina of both STGD and Age Related Macular Degeneration patients.

A healthy lifestyle and diet may help to slow the rate of degeneration. Leafy green vegetables are high in lutein - a structural component of the macular. The inclusion of all vegetables and fruit will help to combat the oxidative stress that occurs in retinal degeneration.

A special anti-oxidant formula containing Lutein, Zeaxanthin, Alpha Lipoic acid and L- Glutathione should be considered. It is very important that individuals with STGD must not take any supplements containing Vitamin A or beta carotene as this may speed up the build-up of Lipofuscin. Please discuss all supplementation with your eye specialist. UV blocking sunglasses are strongly recommended for outdoors.

ARE THERE ANY RELATED DISEASES?

STGD may be known as Stargardt disease, Stargardt Macular dystrophy or Fundus Flavimaculatus. In addition to recessive STGD, there are other rarer forms inherited as dominant rather than recessive traits. Cone and Cone Rod dystrophy may also sometimes be diagnosed as STGD, especially in the early stages of the condition and may also be associated with ABCA4 gene mutations (changes).

The information in this article is offered only as a general description. It is not intended as a substitute for a visit to your Eye Specialist. Even for patients with a known retinal condition regular eye tests are recommended.

Who do I contact for more information regarding testing?

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