



Stargardt Disease

WHAT IS STARGARDT DISEASE?

Stargardt Disease is the most common inherited (runs in families) eye condition that affects the central retina and vision in children. The retina is the inner lining of the back wall of the eye. In order for us to see, light comes into the eye, gets picked up by the retina and sent to the brain where the brain turns the light into a picture. Photoreceptors are a specialized part of the retina used to help process light and help us see. In Stargardt Disease the photoreceptors are not working well in the area of the retina called the macula. The macula is the part of the retina that helps with fine detailed vision in the center of where you focus. This type of retina disease is called a macular dystrophy.

WHAT CAUSES STARGARDT?

In Stargardt Disease there is commonly a mutation in the ABCA4 gene. This abnormal gene is usually passed down from both parents in order for it to cause eye problems. If someone only has one gene that is not normal, then they will be a carrier of the disease but they won't have the disease themselves, and the vision will be normal. Both those with Stargardt Disease and those who are carriers can pass the gene and its eye problems on to their children.

The abnormal gene causes a build-up of leftover material in the retina. Normally, when the retina processes light there is left over material that usually gets taken away by different parts of the eye. In Stargardt Disease, the extra material builds up and damages the photoreceptors (mentioned above). When the photoreceptors don't work well, there can be problems with vision.

WHO GETS STARGARDT?

Stargardt Disease affects about 1 in 10,000 people. It affects males and females equally.

WHAT ARE THE SYMPTOMS OF STARGARDT?

Patients may present with blurry vision in both eyes, light sensitivity, and they may start to have trouble seeing colors. They may have problems reading, recognizing faces, or seeing small details. Children with this condition may have trouble adjusting to light, moving from a sun-lit room into a dark room.



Problems with vision tend to start in early childhood or as a teenager, although some people develop vision problems later in life.

The vision may worsen slowly but can then change more quickly until it is 20/200 (the line just below the big 'E' at the top of a classic vision chart). Once the vision gets really blurry, it usually does not change.

Even though the central vision is blurry, the side or peripheral vision is not as blurry. People with Stargardt tend to keep very good side vision throughout their life.

The age when blurry vision starts tends to predict how bad the final vision will be, this means that people with Stargardt who have blurry vision earlier in life tend to do worse than those who get blurry vision when they are older.

HOW IS STARGARDT DISEASE DIAGNOSED?

An ophthalmologist can generally diagnose Stargardt Disease with a full eye exam and some extra testing. During the eye exam, the pupils (dark spot in the center of the colored part of the eye) will be dilated (made larger) with special eye drops. Then, as part of the eye exam, the retina will be looked at. In Stargardt the macula can look normal early on. Later, yellow flecks may be seen in the retina. These yellow flecks are the build up of extra material when the retina processes light, as mentioned above.

Different tests may also be used to make the diagnosis of Stargardt Disease. An OCT is a special photo of the eye which shows the different parts of the retina and can show if the photoreceptors are not healthy. Sometimes a fluorescein angiogram can help show signs of Stargardt Disease. In this test, dye is injected into a vein in the arm or taken by mouth and photographs are taken as the dye runs through the arteries and veins in the retina. The dye is blocked in the macula in Stargardt Disease, which helps in making the diagnosis. The OCT test may be repeated over time to see if the eye disease is getting worse.

Genetic testing can also help diagnose Stargardt Disease and understand if it will affect other family members. **HOW IS STARGARDT DISEASE TREATED?**

Unfortunately, there is no cure for Stargardt Disease. However, there are things we can do to try and protect the eyes. Avoiding direct sunlight by wearing ultraviolet-blocking sunglasses can help with light sensitivity and protect the eyes. People with Stargardt Disease should not smoke cigarettes or be around cigarette smoke as it may make the eye problems worse. Also, studies suggest that Vitamin A supplements should be avoided as this can make the disease worse.



There is ongoing research looking for gene therapies to help those with Stargardt disease.

WHERE CAN I FIND MORE INFORMATION ABOUT STARGARDT DISEASE?

- Support groups:
 - <https://globalgenes.org>
 - <https://mdsupport.org>
- Other scientific information:
https://eyewiki.org/Stargardt_Disease/Fundus_Flavimaculatus

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