Devon in Sight Eye Condition Fact Sheet 26

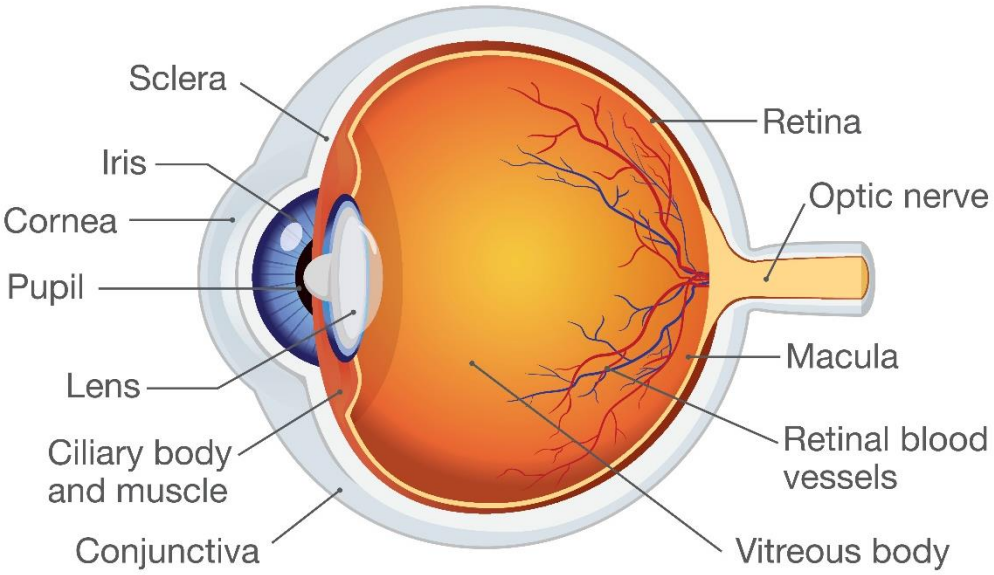
Stargadt Disease

Introduction

Stargardt disease is also known as Stargardt Macular Dystrophy, Juvenile Macular Dystrophy and Fundus Flavimaculatus.

Stargardt disease is an inherited eye condition that affects your macula which is the tiny central part of your retina, the light-sensitive layer at the back of your eye. It causes a reduction in your central detailed vision, which is the vision you use when looking directly at something. It doesn't usually affect your peripheral or side vision.

What is the macula?



Photograph: Visual representation of the anatomy of the eye

The macula is the small central area of your retina where the light entering your eye is focused. The retina is made up of cells called photoreceptors which are cells that are sensitive to light.

The macula is a specialised area that contains a high concentration of photoreceptor cells called cone cells. Cone cells work best in bright light and allow you to see fine detail for activities such as reading and watching television, as well as seeing colour. Therefore, the macula is very important and is responsible for:

- what you see straight in front of you.
- the vision you need for detailed activities such as reading and writing.
- your ability to appreciate colour.

Away from the central macula is the peripheral retina, made up of mostly the other type of photoreceptor called rod cells. Rod cells enable us to see in dim conditions and provide peripheral (side) vision outside of the main line of sight.

How does Stargardt disease affect your sight?

Stargardt disease is sometimes called a Juvenile Macular Dystrophy as it can first appear in childhood. However Startgardt disease can also begin later in adulthood.

At first Stargardt disease will make your vision unclear or blurry. Things may sometimes appear distorted or wavy. You can have problems with your central, detailed vision which can make activities such as reading and recognising faces difficult. Your colour perception may also be affected. If you've had Stargardt disease for a number of years then you may have a blank patch in the centre of your vision. This blank patch will not move and will always be in the very centre of your field of vision.

Stargardt disease doesn't usually affect other parts of your retina so your peripheral or side vision is not normally affected. Since you use your peripheral vision when you're moving around, most people with Stargardt disease can manage to continue getting out and about on their own.

Stargardt disease can also cause problems with light, such as glare and difficulties adapting to changing light conditions.

What causes Stargardt disease?

Stargardt disease is an inherited condition that is caused by a mistake in one of our genes. The faulty gene leads to a build-up of a waste product, lipofuscin, and this will eventually damage the cone cells in the macula.

When genes are identified they are given names usually made up of numbers and letters. The genes associated with Stargardts are ELOVL4, PROM1 and ABCA4. The gene responsible for most cases of Stargardt disease is ABCA4.

What is a “faulty” gene?

All genes come in pairs and you inherit one of each pair from your mother and one of each pair from your father. Your genes determine the many things which make you an individual such as hair or eye colour.

There are a number of ways a genetic condition can be passed through genes. If Stargardt disease is caused by a mutation in the ELOVL4 gene, then it is inherited in an autosomal dominant pattern. If the ABCA4 gene is involved then Stargardt disease is inherited as what is called a recessive condition.

Autosomal dominant condition

Dominant inheritance means that you inherit a disease from only one of your parents. The “faulty” gene that you inherit from one parent is the dominant one and over-rides the “normal” gene from your other parent. Usually the parent who has passed on the “faulty” gene will also have Stargardt disease.

When a parent with the dominant ELOVL4 Stargardt gene has a child with someone who does not carry the Stargardt gene, there is a 50 per cent chance that the faulty gene will be passed to a child. If a child does not inherit the faulty gene they cannot pass it on to their children.

A small number of cases of Stargardt disease are inherited in a dominant pattern.

Autosomal recessive condition

Most cases of Stargardt are inherited in a recessive pattern. A recessive condition can only be inherited when you get two faulty genes, one from each parent. Therefore, both parents have to carry a faulty gene. If your parents have one normal gene and one faulty gene, they are carriers of Stargardt and their sight won't be affected. This is because the "normal" gene compensates for the "faulty" gene. If both your parents pass on their faulty ABCA4 gene, then you have two abnormal genes, and will inherit Stargardt disease.

Most people who have Stargardt disease have parents who don't have the condition. This means that Stargardt often occurs in families that have no history of the disease in the past.

For more information on how Stargardt disease may be passed through your family, you may want to speak to your GP about a referral to a clinical geneticist (a doctor who specialises in genetic conditions). They can discuss your situation with you and your family in more detail. You may also wish to contact the Genetic Alliance for more information on genetics.

How does Stargardt disease affect your eye?

Stargardt disease causes changes to the appearance of the macula area of your retina. When the ophthalmologist (hospital eye doctor) looks into your eye to examine your retina they may notice differences which can help them to diagnose the condition:

- Yellowish flecks which surround your macula are very characteristic of Stargardt disease. These yellow flecks are lipofuscin which is a by-product of cell activity.
- As the disease progresses, an oval lesion can be seen which is often referred to as "beaten bronze" in appearance within your macular area.

Sometimes people have just the flecks without the macular lesion, and in the past, these people may have been diagnosed with an eye condition called fundus flavimaculatus. However, researchers now believe that these two problems, the macular lesion and the yellow flecks, are part of the same genetic problem, but are just expressed in different ways.

It would seem that some people with fundus flavimaculatus can develop more severe sight problems than people with more classic forms of Stargardt disease.

Are there any treatments for Stargardt disease?

Unfortunately at the moment there is no treatment for Stargardt disease. Research and developments into gene therapy and stem cells is very active and it's hoped that this may lead to treatments becoming available at some stage in the future.

Gene therapy aims to replace the faulty gene such as ABCA4 within the affected retinal cells with a new gene that works properly. The "normal" gene is injected into the eye of the person with Stargardt disease. The hope is that the affected cells then begin to work correctly thereby stopping the progression of the disease.

Stem cells are cells that can divide many times and can replace damaged or missing cells in different organs and tissues of the body. If stem cells can be turned into the specialised retinal cells, it may be possible to replace the cells that have been damaged in Stargardt disease.

Another area of research is looking at medications that can reduce the amount of vitamin A by-product that builds up in the eyes of those with Stargardt disease. If you have Stargardt disease you aren't able to clear vitamin A by-product in your eye due to the faulty gene. These by-products collect in the macula and this affects how well the cells work, leading to your sight being affected.

Researchers have reported that exposure to ultraviolet (UV) light may theoretically cause further retinal damage. Therefore protecting your eyes from UV and blue light with sunglasses that have 100 per cent UV filtering is important. Wraparound styles provide protection from light coming in from the sides and tops. We have more information on choosing sunglasses.

There is much that can be done to help make the most of your sight. This includes visual aids and adaptations around the home, adaptations and specialised software for computers and at work, and training from social services.

Coping

Being diagnosed with an eye condition can be very upsetting. You may find that you are worried about the future and how you will manage with a change in your vision. Devon in Sight's Community Support Workers are here to support you.

Help to see things better

Stargardt disease can cause severe problems with your central vision. However, most people with the condition retain their peripheral vision, so won't lose their sight completely.

There are lots of things that you can do to make the most of the vision you have. This may mean making things bigger or smaller, using brighter lighting or using colour to make things easier to see. A low vision assessment can explore how to make the most of your sight. Your GP, optometrist or ophthalmologist can refer you to your local low vision service for an assessment.

You should also ask whether you are eligible to register as sight impaired (partially sighted) or severely sight impaired (blind). Registration can act as your passport to expert help and sometimes to financial concessions. Even if you aren't registered a lot of this support is still available to you.

At Devon in Sight we will offer you information, advice and guidance on your sight condition and staying safe in your home and getting out and about safely.

Information, advice and support

Macular Society is a self-help group for individuals who are affected by eye conditions relating to the macula.

Website: https://www.macularsociety.org/

Genetic Alliance Website: https://geneticalliance.org.uk/

What to do next?

If you are experiencing any of the above symptoms or your sight loss deteriorates, please contact your GP/Optician/Accident & Emergency Department at your local Hospital.

If you need any further advice call our Helpline: 01392 876 666

Devon in Sight is a member of the **Helpline Partnership Community.**

Credit

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Further Support Available from Devon in Sight

- **Information, Advice and Guidance**
- **Demonstrations of Low Vision Aids & Daily Living Equipment**
- **Assistive Technology Solutions**
- **Our holistic needs assessment (Sight Loss MOT)**
- **Training for people with central vision loss (Skills for Seeing)**

Information Disclaimer

Devon in Sight is committed to producing Information, Advice & Guidance Resources that are clear, accurate, evidence-based, up-to-date and easy to use. We have a robust information production process based on best practice to ensure that information we produce meets the needs of people who are blind or partially sighted and their family and friends.

Devon in Sight is not a medical organisation; therefore we can only provide general information that is not intended to be a substitute for a proper medical assessment. Our information is not intended to be used

for individual cases. If you have a specific question about your eye condition, we recommend that you consult an eye care professional.

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Our Information, Advice & Guidance Resources were correct at the time of writing. However, due to research and medical advances, the content may not be completely up to date.

Devon in Sight Contact Information

Devon in Sight
Your local sight loss charity

Helpline 01392 876 666
www.devoninsight.org.uk
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Devon in Sight is a Registered Charity No 1140978

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