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RNIB, supporting people with sightloss

Registered charity number 226227 (England and Wales) and SC039316 (Scotland)

Eye condition fact sheet: Stargardt macular dystrophy

# Stargardt disease

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

## The macula

The macula is found at the centre of your retina and is where the incoming rays of light are focused. This area has a concentration of cone cells which are the cells that sense light.

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
* and your ability to appreciate colour.

## How does Stargardt disease affect your sight

Stargardt disease is sometimes called a juvenile macular dystrophy as it can first appear between the ages of 10 to 20. However, Startgardt disease can also begin later than this.

At first Stargardt disease will make your vision unclear and then sometimes distorted or blurred. You may have problems with your central, detailed vision and possibly colour perception. If you have 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 does not usually affect other parts of your retina so does not normally affect peripheral or side vision. Since you use your peripheral vision when you are moving around, most people with Stargardt disease can manage to keep getting out and about on their own.

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

## How does Stargardt disease affect the eye?

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

In Stargardt disease there are yellowish flecks which often surround your macula. These are very characteristic of Stargardt disease. As your macula becomes damaged there is also an oval lesion which is often referred to as “beaten bronze” in appearance within your macular area. This tends to change over time and reflects the damage to your macula.

Sometimes people have just these flecks without the macular lesion. In the past people who just had the yellow flecks used to be diagnosed with fundus flavimaculatus. However, researchers believe that these two problems, the macular lesion and the yellow flecks, are different expressions of the same genetic problem.

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

## What causes Stargardt disease?

It had been recognised for a long time that Stargardt disease is an inherited condition that is caused by a mistake in one of our genes. Recently, the gene responsible for most cases of Stargardt disease has been identified. When genes are identified they are given names usually made up of numbers and letters. The gene responsible for most cases of Stargardt disease is named ABCA4.

### What are genes?

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 to individuals. Stargardt disease is inherited as what is called a recessive condition.

### Recessive genetic conditions

In a recessive condition, like Stargardt disease, an individual inherits two faulty copies of a gene – one from each parent. Therefore both parents have to carry a faulty copy. Since a parent carries one normal and one faulty copy they are termed carriers of the faulty gene. For parents who are not affected by the condition, the normal copy compensates for the presence of the single faulty copy. If you inherit both faulty copies of the gene, then you will develop the genetic condition, in this case Stargardt disease.

Stargardt disease has an autosomal recessive pattern of inheritance in almost all cases. When two people who carry a faulty ABCA4 gene, both pass their faulty copy to one of their children then that child will have Stargardt disease. Most people who have Stargardt disease have parents who don’t have the condition. This means that Stargardt disease often occurs in families that have no history of the disease in the past.

It is highly unlikely that if you have Stargardt disease that you will have a child who is affected by the condition as your partner will be unlikely to be a carrier of the “faulty” gene. Therefore most people with Stargardt disease have children with normal vision who are carriers of a faulty ABCA4 gene.

For more information on genetics and how Stargardt disease is passed through families you may want to speak to your GP about a referral to a Genetic Unit. You may also wish to contact the Genetic Alliance for more information. Their address is listed below.

## Are there any treatments for Stargardt disease?

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

Researchers have reported that exposure to ultraviolet (UV) light may theoretically cause further retinal damage. Therefore it may be sensible to wear sunglasses with UV protection, that conform to European Standard, and a hat with a wide brim to protect you from the sun’s damaging ultraviolet rays.

## 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. All these feelings are natural.

Some people may want to talk over some of these feelings with someone outside their circle of friends or family. RNIB can help, with our telephone Helpline and our Emotional Support Service. Your GP or social worker may also be able to help you find a counsellor if you think this would help you.

The Macular Disease Society has local groups which meet throughout the country and also offer a telephone counselling service. Sometimes it can help to talk about your feelings or share your experience with people who may have had similar experiences.

### Help to see things better

Stargardt disease can cause severe problems with your central vision. However, most people with the condition retain important aspects of their vision; there are a lot of things you can do to make the most of this remaining vision. This may mean making things bigger, using brighter lighting, or using colour to make things easier to see.

Ask your ophthalmologist, optometrist or GP about low vision aids, like a magnifier, and ask for a referral to your local low vision service. 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.

Local social services should also be able to offer you information on staying safe in your home and getting out and about safely. They should also be able to offer you some practical mobility training to give you more confidence when you are out.

Our Helpline can also give you information about the low vision services available, schooling, work and employment, and our website offers lots of practical information about adapting to changes in your vision and products that make everyday tasks easier.

### What next

#### Talk to someone

The RNIB Helpline is your direct line to the support, advice and products you need. We’ll help you to find out what’s available in your area and beyond, both from RNIB and other organisations.

Whether you want to know more about your eye condition, buy a product from our shop, join our library, find out about possible benefit entitlements, be put in touch with a trained counsellor, or make a general enquiry, we’re only a call away.

RNIB Helpline
t: **0303 123 9999**
**helpline@rnib.org.uk**

We are ready to answer your call Monday to Friday 8.45am to 5.30pm. Outside these times leave us a message and we’ll get back to you as soon as possible.

Unfortunately, we can only answer email enquiries from the UK.

#### RNIB discussion forums

Join our discussion forums – share your ideas with others and have your say about the issues that affect you. Visit rnib.org.uk/discussions.

#### Get information

RNIB’s guide to adapting to sight loss (rnib.org.uk/adapting) offers ideas for people with sight problems on making life easier and ways of getting the most out of life.

You may also find the following sections of our website useful:

* living ([rnib.org.uk/education](http://www.rnib.org.uk/education))
* working ([rnib.org.uk/employment](http://www.rnib.org.uk/employment)).

## Useful contacts

### The Macular Society

PO Box 1870

Andover

SP10 9AD

t: 0300 30 30 111
info@macularsociety.org

The Macular Disease Society is a self help group for individuals who are affected by eye conditions relating to the macula. The society is a source of information and support.

### Genetic Alliance UK

Unit 4D, Leroy House

436 Essex Road

London

N1 3QP

t: 0207 704 3141

contactus@geneticalliance.org.uk

Genetic Alliance UK is a national alliance of patient organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders.

### LOOK

Queen Alexandra College
Court Oak Road
Harbourne
Birmingham
B17 9TG
t: 0121 428 5038

LOOK is an organization, which seeks to identify sight-related support groups and put parents in contact with relevant services.

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