Macular Society

Beating Macular Disease

Your guide to

Stargardt disease

QR codes

Throughout this leaflet you will see QR codes like the one below. If you have a smartphone or tablet with a QR code reader (free from your app store) and access to the internet, you can scan these codes to access other information on our website.

To download a QR reader onto your smartphone or tablet, simply visit your app store and search 'QR code reader'.

Have a practice run by scanning the code to the right. You will be directed to the home page of the Macular Society website.

If you don't have a QR code reader, you can access the same information using the web addresses instead.

The Macular Society is the national charity for anyone affected by macular conditions. This guide to Stargardt disease provides information about the condition, guidance on living with it, and places to find further support.

Being diagnosed with Stargardt disease can be distressing, but with the right information and support, people can cope very well.

It's important to remember that in the vast majority of cases, patients with Stargardt disease do not lose all their sight. Stargardt disease usually affects central vision only. Peripheral vision is not usually affected.

What is Stargardt disease?

Stargardt disease is a genetic condition caused by a tiny alteration in a single gene. Stargardt disease, also known as fundus flavimaculatus, is the most common form of juvenile macular dystrophy.

Stargardt causes a wasting of a central area of the retina called the macula. This area is eventually surrounded by a ring of white or yellow spots.

An important layer of the retina, the retinal pigment epithelium (RPE), is also affected by an accumulation of 'waste' material called lipofuscin.

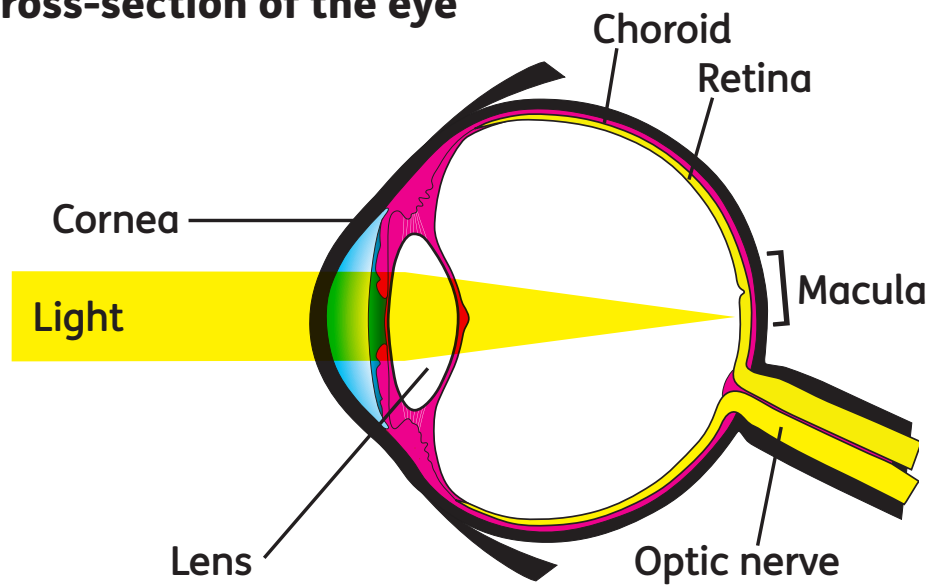
What is the macula?

The macula is part of the retina at the back of the eye. It is only about 5mm across but is responsible for our central vision, most of our colour vision and the fine detail of what we see.

The macula has a very high concentration of photoreceptor cells – the cells that detect light.

They send signals to the brain, which interprets them as images. The rest of the retina processes our peripheral, or side vision.

Cross-section of the eye



How is the disease inherited?

Genetic conditions are inherited as either a dominant or recessive form. Where a condition is caused by only one faulty gene from one parent it is called a dominant form. In these

cases the parent with the faulty gene will have the condition themselves and there is a 50% chance that they will pass the gene on to each child they have. This chance is the same for each child whether they are male or female and regardless of the birth order.

Other forms are called recessive. This is where the faulty gene is inherited from both parents. In a recessive condition the parents probably won't have the condition themselves although they both carry the faulty gene. With recessive inheritance, there is a 25% (1 in 4) chance of each child inheriting the condition. This chance is the same for each child whether they are male or female and regardless of the birth order.

Stargardt disease is most often inherited as the recessive form. The gene associated with this form of Stargardt disease is called ABCA4 on chromosome 1.

Alterations in another gene called ELOVL4 cause a less common dominant form of the disease.

How common is Stargardt disease?

Stargardt disease affects approximately one in 10,000 people. It affects both males and females.

What are the symptoms?

In the early stages, people may have good visual acuity, but may experience difficulty reading or seeing in dim light. Other common symptoms of Stargardt disease include blurriness and distortion of vision.

Children often first experience symptoms between the ages of 6 and 12. They may begin to find it difficult to adapt from dark to light or light to dark surroundings (known as 'dark-adaptation'). However, some people do not have any symptoms until adulthood. Stargardt disease is usually diagnosed in people under the age of 20. There is a late onset form of the disease which can begin in people over the age of 50.

How quickly does vision deteriorate?

The progression of Stargardt disease varies. Visual acuity (the ability to distinguish details and shape) may decrease slowly at first.

Once visual acuity of 6/12 is reached, there is often rapid vision loss until it reaches 6/60. ('Normal' vision is 6/6. A person with 6/12 vision sees at 6 metres what someone with 'normal' vision sees at 12 metres.)

Eventually, almost everyone with Stargardt disease has a visual acuity in the range of 6/60 to 6/120. The vision loss is not correctable with prescription eyeglasses, contact lenses, or surgery.

How is Stargardt disease diagnosed?

An eye care professional can diagnose Stargardt disease by examining the retina. Lipofuscin deposits can be seen as yellowish flecks in the macula.

The flecks are irregular in shape and usually extend outward from the macula in a ring. The number, size, colour and appearance of these flecks varies from person to person. A standard eye chart and other tests may be used to assess symptoms of vision loss in Stargardt disease, including:

Visual field testing
Visual field testing assesses the full horizontal and vertical range and sensitivity of a person's vision, and detects blind spots (scotomas) which could be a sign of eye disease. There are several types of test but they are not painful or invasive.

The most common type of visual field test is often seen in local opticians. The patient sits at the machine with their chin on a rest and a patch

over one eye. They have a button which they are asked to press when they see one or more flashing lights. This process results in a map of the person's visual field, and can point to areas of the retina where there is vision loss.

Electroretinography (ERG)
ERG measures electrical signals produced by the retina following flashes of light. The test uses electrodes placed on the cheek under each eye. The patient looks at black and white checks moving across a TV screen and a lamp that flashes light into the eye three times per second.

The electrical responses are viewed and recorded on a monitor. Abnormal patterns of light response suggest the presence of Stargardt disease or other diseases that involve retinal degeneration.

Optical coherence tomography (OCT)
OCT is a scanning device that works a little like ultrasound. Ultrasound captures images by bouncing sound waves off living tissues; OCT does it with light waves.

The patient places his or her head on a chin rest and invisible, near-infrared light is focused on the retina. Cross-sectional pictures of the retina are analysed for any abnormalities which could indicate retinal degeneration. OCT is sometimes combined with infrared scanning laser ophthalmoscopy (ISLO) to provide additional surface images of the retina.

Autofluorescence imaging

Your eyes may be photographed with a special camera that measures the fluorescence of cells in the back of your eye. A specific pattern of autofluorescence imaging may be seen in patients with Stargardt disease.

Genetic testing

As part of the diagnosis, you may be invited to take a genetic test to learn more about your particular condition, and enable research scientists to learn more about the condition in general. You are not obliged to take a genetic test, but some people find it helpful to know more. In some cases, the tests don't always return conclusive answers.

Some people may find it helpful to talk with a genetic counsellor first. Usually part of the research team, the genetic counsellor will help you and your family understand how the condition has been inherited, and the implications for other family members and future generations. They will enable you to make informed decisions about testing and support you through the process.

Ask your consultant for more information about genetic testing.

Emotional support

Being diagnosed can be stressful and highly emotional. You may benefit from working with a professional counsellor to understand your situation and find ways to manage your feelings. Our free telephone counselling service is provided by accredited counsellors who have extensive experience of working with people with sight loss. Call 0300 3030 111 for a referral.

Medical treatments

There is currently no treatment for Stargardt disease. However, there is a wide range of support available as outlined below to help people with the condition as well as their family and friends. If you experience any sudden changes in vision seek urgent medical advice from an eye hospital.

Research

There is a great deal of research going on into potential treatments. We are finding out more about genetics and gene-based therapies are being developed. Stem cell research is also encouraging.

What can I do to look after my eyes?

It is thought that people with Stargardt disease cannot process vitamin A properly so it is important to avoid excess vitamin A. Also avoid bright light as it may accelerate the progress of the disease.

- Visit your optician at least every two years for a general eye test.
- Don't smoke.
- Maintain a healthy weight and blood pressure.
- Wear lenses which block UV light, particularly in bright sunlight. Blue block filters also reduce glare.
- Wear a hat with a wide brim or visor to shade eyes from direct sunlight.
- Limit alcohol intake to recommended levels.
- Eat lots of fruit and green, leafy vegetables.

Scan this code to see our **'Protecting your eyes'** leaflet or visit **macularsociety.org/resources**

What support is available?

Emotional, practical and financial information and support can help you stay independent.

Work

The government **Access to Work** scheme provides you and your employer with advice and support with any extra costs which may arise because of your sight loss.

Employers must make certain changes (known as 'reasonable adjustments') to make sure that people with disabilities are not substantially disadvantaged when doing their job. These could include changing working hours or providing equipment. If the help you need at work isn't covered by your employer making reasonable adjustments, you may be able to get help from Access to Work.

You need to have a paid job, or be about to start or return to one. You'll be offered support based on your needs, which may include a grant to help cover the costs of practical support in the workplace.

An Access to Work grant can pay for:

- Specialist equipment, adaptations or support worker services to help you do things like answer the phone or go to meetings
- Help getting to and from work.

You might not get a grant if you already get certain benefits. The money doesn't have to be paid back and won't affect your other benefits.

Scan this code for more information or go to **gov.uk/access-to-work**

In Northern Ireland go to **nidirect.gov.uk**

Blind in Business offers help and support with finding work, the interview process and obtaining equipment. It also supports employers in hiring and working with people with visual impairments.

Scan this code for more information or go to **blindinbusiness.org.uk**

Personal Independence Payment (PIP)
You may be able to get help with some of the extra costs caused by long term ill-health or disability. If you're aged 16 to 64 you could get financial support by claiming PIP.

The amount you get depends on how your condition affects you, not the condition itself. The amount payable is based on difficulty with daily living tasks such as preparing food and reading and mobility difficulties.

For further information on PIP please visit our website or contact our Advice and Information Service.

You'll be assessed by a health professional to work out the level of help you can get. Your rate will be regularly reviewed to make sure you're getting the right support.

If applicable, your carer could get Carer's Allowance if you have substantial caring needs.

For more information go to **gov.uk/pip** or **gov.uk/carers-allowance**

Registering as visually impaired
Registering as visually impaired could help you access practical help and benefits. To be registered, you must have your sight examined by a hospital consultant ophthalmologist.

Scan this code to see our **'Registering as sight impaired'** leaflet or visit **macularsociety.org/resources**

Low vision aids, lighting and technology
A wide range of low vision equipment is available,

from magnifiers and navigation aids to computer software. Good lighting is also very important.

To find the best advice and equipment, ask your GP, consultant or social services department for a low vision assessment.

Scan this code to see our **'Low vision aids'**, **'Lighting'** and **'Using technology'** leaflets or visit **macularsociety.org/resources**

Skills for Seeing

We offer free one-to-one training in practical techniques that can help people with central vision loss to make the most of their remaining vision, especially if both eyes are affected. Skills for Seeing training can help with activities such as reading and watching TV, and with recognising faces.

Scan this code to see our **'Skills for Seeing'** leaflet or visit **macularsociety.org/resources**

Support in Education

Children with sight loss are eligible for special educational needs (SEN) support in school. This can provide your child with the adaptations, equipment and support they need to fully access the curriculum.

In higher education, the Disabled Students' Allowance (DSA) can fund equipment and support.

Beating macular disease

Macular disease is the biggest cause of sight loss in the UK, with around 300 people diagnosed every day.

The Macular Society is the only charity determined to beat the fear and isolation of macular disease with world-class research, and the best advice and support.

To support people affected by macular disease now, the Macular Society provides a range of support, information and services. These include:

- The **Advice and Information Service (0300 3030 111).** Available Monday to Friday, 9am to 5pm. Alternatively, you can email help@macularsociety.org

- Our website at **macularsociety.org**, which provides a wide range of information and resources for people affected by macular disease.

Macular Society Working Age & Young People's (WAYP) Services:

- **WAYP Macular Support Groups** meet in a relaxed and social environment, sharing experience, information and informal support.
- **WAYP Facebook Group** provides a closed, supportive forum where you can share your experience and learn from others.
- **Parents' Forum**, for parents who have a macular condition, or whose children do, includes a regular catch-up by telephone conference call.
- **WAYP Advice Service** offers one to one advice including benefits and employment rights.
- Free, confidential **Counselling Service**, which offers support over the phone from one of our trained counsellors. Call the Advice and Information Service for more information and to arrange a referral.
- **Telephone Befriending Service** pairs you up for regular telephone calls with another person

who knows what it is like to live with the condition. Calls can be about anything, and provide friendly support.

- **Skills for Seeing training** can help you with techniques to make the most of your functional vision.

To find out more about any of the ways we can help, please contact the **Advice and Information Service** on **0300 3030 111.**

Beating macular disease:

- Providing the best advice and information on living with macular disease.
- Local Support Groups helping you to beat the isolation of macular disease – with support and companionship.
- Research focused on finding a cure to Beat Macular Disease forever.

We rely on donations to fund support services and research to find a cure. To donate please go to **macularsociety.org/donate** or call **01264 350 551**.

Macular Society

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