



# Macular Society

Beating Macular Disease

Your guide to

# macular dystrophies



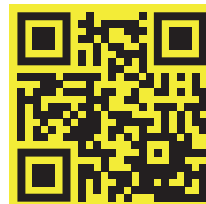
## Your guide to macular dystrophies

### 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 macular dystrophies provides information about several conditions, guidance on living with them, and places to find further support.

Being diagnosed with a macular dystrophy can be distressing and worrying, but with the right information and support people can cope very well.

The term ‘macular dystrophies’ covers a large number of rare, inherited conditions. They can appear in childhood but they are often not diagnosed until later in life. They are a result of faulty genes inherited from one or both parents.

Macular dystrophies cause loss of central vision as a result of damage to the macula, the most sensitive part of the retina. They are painless and do not lead to complete loss of sight, as a person’s peripheral (or side) vision is unaffected.

**Advice and Information Service 0300 3030 111**

## Your guide to macular dystrophies

### What is the macula?

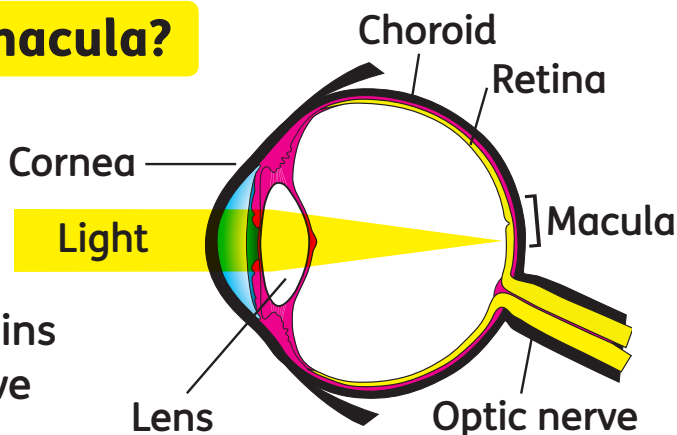
The macula is an area of the retina at the back of the eye.

The retina contains the light-sensitive cells which send

messages to the brain via the optic nerve.

Although it is only about the size of a grain of rice, the macula is responsible for all our central vision, most of our colour vision and the fine detail of what we see. If the macula is damaged, people find it hard to drive, read, watch TV or recognise faces.

In some dystrophies, the cells of the macula gradually die (atrophy). In others, tiny abnormal blood vessels grow into the macula. They are fragile so can leak and scar the macula. The medical term for this is choroidal neovascularisation (CNV). In the more common age-related macular degeneration (AMD), similar symptoms are sometimes referred to as 'dry' and 'wet' macular degeneration respectively.



## What are the symptoms?

People with macular dystrophies gradually lose part or all of their central vision. They may have problems identifying colours, dislike bright light or, in the early stages, find their night vision isn't very good. Children may find it hard to see the board at school. Some people see straight lines as bent, or see dark spots in the centre of their field of vision.

## Genetics

Macular dystrophies are caused by someone inheriting a faulty gene from one or both parents.

**Dominant forms** – these are caused by a faulty gene from one parent. The parent with the faulty gene will have the condition themselves and there is a 50:50 chance that they will pass the gene on to their child.

**Recessive forms** – these need a faulty gene from each parent. The parents probably won't have the condition themselves, although they both carry the faulty gene.

### How are macular dystrophies diagnosed?

Most people are diagnosed after visiting an optician who identifies a potential problem. The optician will refer the person to an ophthalmologist (specialist eye doctor) . Further tests to determine the specific dystrophy the person has might include:

#### 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.

## **Fluorescein angiography**

A dye called fluorescein is injected into the bloodstream via the patient's arm. This travels to the eye and highlights blood vessels in the retina. A photograph is taken of the back of the eye.

## **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 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 macular disease.

## **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.

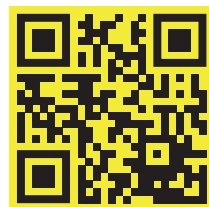
## Your guide to macular dystrophies

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.

### Genetic tests

A genetic test usually involves having a blood sample taken. DNA is extracted from blood cells and analysed in the laboratory. The analysis involves looking very carefully at the genetic code to try and identify the genetic alteration that is causing the condition. Many people have the genetic cause identified, although it's not always possible.

Scan this code to see our '**Genetics**' factsheet or visit [macularsociety.org/resources](http://macularsociety.org/resources)





## What are the main types of macular dystrophies?

### Stargardt disease (fundus flavimaculatus)

This is a recessive genetic condition caused by an alteration in a single gene. Also known as fundus flavimaculatus, it is the most common form of macular dystrophy.

Stargardt disease causes a wasting of an area of the retina which 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.

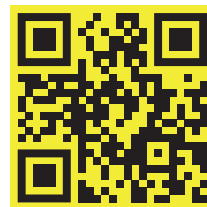
Stargardt disease is usually diagnosed in people under the age of twenty, when they first notice their vision getting worse. In the early stages, people may have good visual acuity, but have difficulty reading or seeing in dim light. Other common symptoms of Stargardt disease include blurriness and distortion of vision.

## Your guide to macular dystrophies

Children often first experience symptoms between the ages of 6 and 12 when 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.

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.

Scan this code to see our  
**'Stargardt disease'** leaflet  
or visit [macularsociety.org/resources](http://macularsociety.org/resources)



### **Best disease**

Best disease is a dominant form. In the early stages, pictures of the retina look like an egg yolk. Later they look like scrambled egg. There is also an accumulation of lipofuscin (a waste material).

Best disease is also known as early-onset vitelliform macular dystrophy. It usually appears

in childhood, but the onset of symptoms and the severity of vision loss vary widely. The adult-onset form of Best disease begins later, usually in mid-adulthood, and tends to cause vision loss that worsens slowly over time. The two forms of Best disease each have characteristic changes in the macula that can be detected during an eye examination.

Over time, the abnormal accumulation of lipofuscin can damage cells that are critical for clear central vision. As a result, people with this disorder often lose their central vision, and their eyesight may become blurry or distorted. It does not affect side (peripheral) vision or the ability to see at night.

Head injuries may make Best disease worse. Sensible protective measures include wearing a helmet for cycling and avoiding contact sports.

### **Sorsby fundus dystrophy**

This is very rare. People normally develop it in their 20s or 30s, usually in both eyes.

## Your guide to macular dystrophies

Sorsby dystrophy is inherited from a dominant gene and can be diagnosed with a genetic test. Often a person with Sorsby dystrophy also develops CNV. They may have a sudden distortion or loss of vision. If this happens, **it's vital to get urgent medical attention at a hospital eye unit**, as the sight loss may be slowed if treated quickly.

### Pattern dystrophy

This is a dominant form, usually occurring later in life and sometimes confused with AMD. People with pattern dystrophies may show different patterns of damage in each eye, which may change over time. Most people keep good vision in at least one eye.

### Bull's eye maculopathy

This describes a number of different conditions in which there is a ring of pale-looking damage around a darker area of the macula. The macula can often appear to have circular bands of different shades of pink and orange. Age of onset and severity of sight loss varies, and it can be inherited in many ways.

## **Doyne honeycomb dystrophy**

This is a dominant condition. Small spots called drusen appear which gradually form a honeycomb pattern of damage to the retina. Typically, people with this condition do not have symptoms until they are aged between 30 and 40 years old. Early visual symptoms may include decreased visual acuity, problems seeing colour, eye discomfort in bright light and distorted vision.

## **Cone dystrophy**

There are various ways in which cone dystrophy is inherited but it is most commonly described as 'sporadic' (no recognised pattern of inheritance). People with the condition tend to be light-sensitive and have problems distinguishing colours from the early stages of the disease.

People who develop the disease early may also experience rapid involuntary eye movements called nystagmus. Central vision may become affected later.



## Your guide to macular dystrophies

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The symptoms of cone dystrophy vary from one person to another, even among individuals with the same form of the disorder. The age of onset, specific symptoms, severity, and progression (if any) can vary greatly.

### **PXE**

Pseudoxanthoma elasticum or 'PXE' is a disease affecting many parts of the body. Fibres in the skin, eyes and blood vessels lose their elasticity. When this happens behind the eye, the retina can stretch and become brittle, causing cracks. Blood vessels grow through the cracks and leak, causing scarring of the macula. If you experience sudden distortion or loss of vision, get urgent attention at an eye hospital. Treatment might stop the blood vessels growing.

PXE patients also need to be assessed by a heart specialist (cardiologist) for potential heart problems.

**PXE Society: Elspeth Lax on 01628 476 687, email [pxeeurope@aol.com](mailto:pxeeurope@aol.com) or visit [pxe.org.uk](http://pxe.org.uk).**

## Medical treatments

There are no treatments for most dystrophies. However, if you develop CNV (abnormal blood vessels) you may be able to have injections to stop their growth and preserve the sight you have left. 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 the faults in the genes which are responsible for dystrophies and researchers are looking at the potential of drugs, stem cell therapy and gene therapy as new treatments which could slow the progress of the disease or even provide a cure.



### What can I do to look after my eyes?

- 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, 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](http://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

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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](https://www.gov.uk/access-to-work)



In Northern Ireland go to [nidirect.gov.uk](https://www.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](https://www.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 between 16 and 64



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you could get financial support by claiming Personal Independence Payment (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.

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](https://www.gov.uk/pip)  
[gov.uk/carers-allowance](https://www.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](http://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.

The Macular Society has produced resources to help teachers make lessons accessible for visually impaired students.

Scan this code to download  
the resources or visit  
[macularsociety.org/teaching](http://macularsociety.org/teaching)



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



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### How we help

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.

Our research programme is focused on finding new treatments and a cure to Beat Macular Disease forever. To support people affected by macular disease now, the Macular Society provides a range of support, information and services:

**The Advice and Information Service (0300 3030 111)** is available Monday to Friday, 9am to 5pm. Alternatively, you can email [help@macularsociety.org](mailto:help@macularsociety.org)

## **Counselling**

Coming to terms with a diagnosis of macular disease can be emotional and complex.

The Macular Society offers a free telephone counselling service delivered by professional registered counsellors who all have extensive experience of working with people with sight loss. Over a series of weekly sessions, a counsellor may be able to help you adjust to your situation and manage the emotions you may experience.

Please call our Advice and Information Service to request a referral.

Our **website** provides a wide range of information and resources for people affected by macular disease. You can also find out more about the services we offer. Visit it at **macularsociety.org**

Our network of over **400 Macular Support Groups** stretches across the UK. Each one offers practical and emotional support for people with macular disease, from those living with it today.

## 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](https://macularsociety.org/donate) or call **01264 350 551**.

## Macular Society

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