Your guide to juvenile macular dystrophies
The Macular Society is a national charity for anyone affected by macular conditions. This guide to juvenile macular dystrophies (JMD) provides information about the condition, guidance on living with it, and where 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 ‘juvenile 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.

Juvenile macular dystrophy causes loss of central vision as a result of damage to the macula, the most sensitive part of the retina. It is painless. It doesn’t lead to complete loss of sight as a person’s peripheral, or side vision, is unaffected.

Helpline 0300 3030 111
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 and 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.
What are the symptoms?

People with JMD 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 dark spots appear in the centre of their field of vision.

Genetics

Macular dystrophies are caused by someone inheriting a faulty gene from either one or both parents. Some are caused by only one faulty gene from one parent, called a dominant form. In these cases 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. Other forms are called recessive. These need a faulty gene from each parent. In a recessive condition the parents probably won’t have the condition themselves although they both carry the faulty gene.
How are juvenile dystrophies diagnosed?

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

**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.

**Electrodiagnostics**
Tiny electrodes are applied to the eye’s surface. The electrical responses of the retina and optic nerve to different visual stimuli are recorded.

**Optical coherence tomography (OCT)**
A non-invasive 3D scan of the retina and macula.

**Genetic tests**
Genetic tests can sometimes be used to identify specific conditions.
What are the main types of juvenile macular dystrophy?

Stargardt’s Macular Dystrophy – Fundus Flavimaculatus
Stargardt’s is probably the most common form of JMD. It is a recessive form. It is thought that people with Stargardt’s 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.

Stargardt’s 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.

Best’s Disease
Best’s is a dominant form. In the early stages pictures of the retina look like an egg yolk and later look like scrambled egg. There is also an accumulation of lipofuscin.
Head injuries may make Best’s worse. Sensible protective measures include wearing a helmet for cycling and avoiding contact sports.

**Sorsby’s Dystrophy**
This is very rare. People normally develop it in their 20s or 30s, usually in both eyes. Sorsby’s is inherited from a dominant gene and can be diagnosed with a genetic test. Often a person with Sorsby’s also develops CNV. They may have sudden distorted or loss of vision. If this happens urgent medical attention at a hospital eye unit is vital, as the sight loss may be slowed if treated quickly.

**Pattern Dystrophy**
This is another 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. Age of onset and severity of the sight loss varies and it can be inherited in many ways.

Doyne’s Honeycomb Dystrophy
This is a dominant condition which can be diagnosed with a genetic test. Small spots called drusen appear which gradually form a honeycomb pattern of damage to the retina.

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. Rapid involuntary eye movements called nystagmus may develop in those who develop the disease early. Central vision may become affected later.
PXE
Pseudoxanthoma Elasticum or ‘PXE’ is a disease affecting many parts of the body. The skin loses some of its elasticity and 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. Symptoms may be distorted or sudden loss of vision. If this happens 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 or visit www.pxe.org.uk.
Medical treatments

There are no treatments for most dystrophies yet. However, if you develop CNV (abnormal blood vessels) in your eye you may be able to have injections to stop their growth. 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 which genes are responsible for dystrophies and gene-based therapies are being developed. Stem cell research is also encouraging. Currently, trials are testing stem cell therapy in patients with Startgardt’s and AMD.
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 and blue 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 a diet low in saturated fats and rich in omega 3 fatty acids (e.g. oily fish, walnuts).

• Eat lots of fruit and green, leafy vegetables.
What other support can I get?

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.

• Registration – registering as visually impaired could help you access practical help and benefits.

• Low vision aids and lighting – 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.

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

• Genetic counselling – understanding the genetics of JMD can be emotional and complex. Genetic counselling before and after genetic testing can help you and your family understand how the condition was inherited, the implications for other family members and future generations. Referrals can be made via your consultant ophthalmologist or GP.

• Emotional support – being told you have a macular condition can be difficult for the person and their family. You may feel many emotions including sadness and anger. Our experienced helpline staff can offer information and advice. We also offer free telephone counselling by qualified counsellors for anyone affected by macular disease. Our telephone befriending service can also provide regular social calls from someone who understands.
• Support groups – many people benefit from meeting others in a similar situation. We have groups across the country. They are mostly run by people with the condition. Some are specifically for younger people. They meet informally, usually at weekends or during the evening. Come along and meet the experts on living with macular disease!

We are here for anyone affected by macular disease. This can be for anyone with a macular condition or anyone caring for or supporting someone with a macular condition.

For more information about any of our services call our Helpline

0300 3030 111

Monday – Friday
9am – 5pm

help@macularsociety.org
Join us

Our members help us:
• campaign for better care as the voice of people with macular conditions;
• expand our services to others so that no one has to face macular disease alone;
• fund important research to find a cure to overcome macular disease.

With your help we can continue to provide help for today and bring hope for tomorrow.

For information about becoming a member or regular supporter, or to simply join:
• visit www.macularsociety.org
• call 01264 350 551.