

Retinal Dystrophy

Information Guide

What is Retinal Dystrophy?

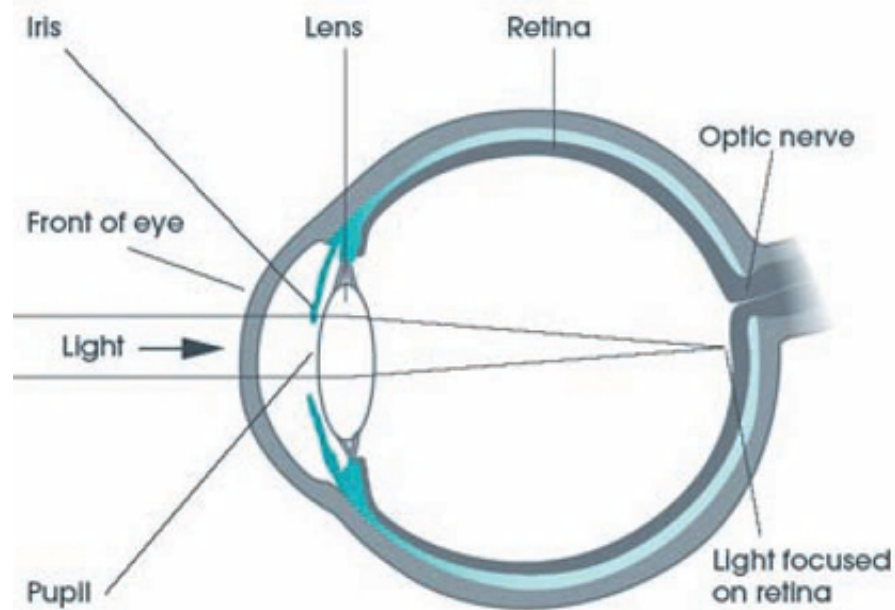
- A retinal dystrophy is a condition associated with reduced or deteriorating vision in both eyes.
- Retinal Dystrophy is not one single condition but the general name given to a wide range of eye conditions (for example, Retinitis Pigmentosa). 'Dystrophy' means a condition that a person is born with, 'retinal' means relating to the retina – the light sensitive film at the back of the eye.
- Most retinal dystrophies are genetic. This means they are caused by a mistake in a person's genes and both eyes are likely to be affected.
- Often the gene mistake has been passed (inherited) from one or both parents. There are a number of different ways in which this can happen (see 'Inheritance' section). Sometimes a new gene mistake occurs for the first time in an individual within a family but can then be passed on to future generations.
- Some retinal dystrophies are diagnosed early in life but sometimes symptoms do not develop until adulthood.
- Occasionally a retinal dystrophy can also be part of a pattern of particular problems which affect other parts of the body as well as the eyes ('Syndromic retinal dystrophies').

How does the eye work?

The eye is made up of 3 parts:

- A light focussing part at the front of the eye (cornea and lens).
- A light sensitive film at the back of the eye (retina).
- A large collection of communication wires to the brain (optic nerve).

Structure of the eye



The Retina

The retina has two types of light sensitive cells called Rods and Cones.

Rods are good for detecting:

- Things that move
- In the dark
- In black and white
- Less detail
- Mostly outer vision (peripheral)

Rods help us to see the surrounding bits of vision and help us to walk around without bumping into things, especially in the dark.

Cones are good for detecting:

- Things that are not moving
- In daylight
- In colour
- In fine detail
- Mostly central vision

Cones help us to see the central bit of vision that we use for reading, looking at photographs and recognising faces.

Retinal Dystrophy occurs in a wide range of eye conditions and can be divided into two main groups.

Isolated retinal dystrophies:

- Early onset retinal dystrophies, for example, Leber Congenital Amaurosis
- Rod-Cone Dystrophies, where the rods are first part of the retina to be affected, for example, Retinitis Pigmentosa
- Cone-Rod Dystrophies where the cones are the first part of the retina to be affected.
- Macular dystrophies, such as Stargardts disease, Best's disease

Syndromic retinal dystrophies, for example:

- Usher Syndrome
- Batten Disease
- Bardet-Biedl

Inheritance

Retinal dystrophies can be passed on within families by one of several different patterns of inheritance. These are known as Dominant, Recessive and X-linked. There are separate leaflets to explain each of these in detail, once you have been told which the likely pattern is in your particular condition/family.

Treatment

There is currently no treatment available to cure retinal dystrophy or halt its progress. It is the result of incorrect instructions being passed to the body's chemistry by changes or mistakes in individual genes which are present in every cell in the body. Some of the genes known to cause retinal dystrophy have now been identified and research in this area is very encouraging. A number of gene therapy trials are underway but it is still likely to be many years before this translates into possible treatment options.

Where a faulty gene can be identified it may be possible to offer genetic testing to other family members to see whether they have inherited the same change.

Further Support

Adjusting to a diagnosis of retinal dystrophy can be difficult but a number of organisations offer support/practical assistance including:

- RNIB: Helpline 0303 123 9999 e-mail: helpline@rnib.org.uk
- RP Fighting Blindness: Helpline 0845 123 2354, website: www.brps.org.uk
- Action for Blind People: Helpline 0800 915 4666
- Henshaws Society for Blind People 88-92 Talbot Rd
Manchester M16 0GS
Tel 0161 872 1234, website: www.henshaws.org.uk
- Local Low Vision Aid clinics
- Social Services Sensory Impairment Teams

Zero tolerance Policy

We are committed to the well-being and safety of our patients and or our staff. Please treat other patients and staff with the courtesy and respect that you expect to receive. Verbal abuse, harassment and physical violence are unacceptable and will lead to prosecutions.

No Smoking Policy

The NHS has the responsibility for the nation's health.

Protect yourself, patients, visitors and staff by adhering to our no smoking policy. Smoking is not permitted within any of our hospital buildings or grounds.

The Manchester Stop Smoking Service can be contacted on Tel: (0161) 205 5998 (www.stopsmokingmanchester.co.uk).

Suggestions, concerns and complaints

If you wish to make a comment, have a concern or want to complain, it is best in the first instance to speak to the manager of the ward or department involved.

The Trust has a Patient Advice and Liaison Service (PALS) who can be contacted on (0161) 276 8686 and via e-mail: pals@cmft.nhs.uk. They will help you if have a concern, want advice or wish to make a comment or complaint.

Information leaflets about the service are readily available throughout the Trust. Please ask any member of staff for a copy.

Translation and Interpretation Service

Do you have difficulty speaking or understanding English ?

আপনি কি ইংরেজীতে বুঝতে কিংবা বুঝাতে পেরেছেন ? (BENGALI)

क्या आपको अंग्रेजी बोलने या समझने में कठिनाई है ? (HINDI)

તમે ભાષા કારણે વાતચીત કરવામાં મુશ્કેલી આવે છે ? (GUJARATI)

ਕਿ ਤੁਹਾਨੂੰ ਅੰਗ੍ਰੇਜ਼ੀ ਬੋਲਣ ਜਾਂ ਸਮਝਣ ਵਿਚ ਦਿਕਤ ਹੈ ? (PUNJABI)

Miyey ku adagtahay inaad ku hadasho Ingriisida aad sahamto (SOMALI)

هل لديك مشاكل في فهم اوالتكلم باللغة الانجليزية ؟ (ARABIC)

你有困難講英語或明白英語嗎？ (CANTONESE)

کیا آپ کو انگریزی سمجھنے اور سمجھانے میں دقت پیش آتی ہے؟ (URDU)

☎ Telephone 0161 276 6202 / 6342

Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio, British Sign Language video/DVD) or in another language.

For more information

If you need more advice about any aspect of retinal dystrophy, you are welcome to contact:

Regional Genetics Service
Saint Mary's Hospital (Sixth Floor)
Oxford Road
Manchester
M13 9WL

Tel: 0161 276 6505
www.cmft.nhs.uk