RETINA UK
WORKING FOR PEOPLE WITH INHERITED SIGHT LOSS

A GUIDE TO CONDITIONS COVERED BY RETINA UK

The defining features of conditions covered by Retina UK are that:

- the cause of sight loss is retinal degeneration (cells in any part of the retina, including the macula, deteriorate and die)
- they have a direct genetic cause (they are caused by mutations in a particular part of a person's DNA).

The mutations cause failure of a particular structure or function within retinal cells, ultimately leading to degeneration of the cells and subsequent sight loss.

The mutations are almost always inherited from one or both parents, even in situations where both parents have healthy eyesight. In a tiny proportion of cases, the mutation appears for the first time in the affected person and is not carried by either parent.

Genetic retinal conditions usually cause vision problems that start in childhood or early to mid-adulthood.

WE ARE HERE FOR YOU

- Call: 01280 821334
- Email info@RetinaUK.org.uk
- Visit RetinaUK.org.uk

Vision problems starting later in life (50s or 60s) onwards are less likely to have a direct genetic cause, although this is possible. Most Retina UK conditions are progressive, becoming gradually worse over time, although a small number are “stationary”; they may be present at birth but do not progress.

Retina UK conditions include:

- Retinitis pigmentosa (RP), including X-linked RP
- Rod-cone dystrophy
- Cone-rod dystrophy
- Leber’s congenital amaurosis (LCA)
- Stargardt disease (a type of inherited juvenile macular degeneration)
- Choroideremia
- Achromatopsia
- Best disease (a type of inherited macular degeneration)

Plus other rarer conditions (details on our website) such as:

- Doyne honeycomb dystrophy
- Pattern dystrophy
- Familial exudative vitreoretinopathy
- Gyrate atrophy
- X-linked retinoschisis

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We also cover some inherited syndromes where one of the symptoms is retinal degeneration; these include:

- Usher syndrome
- Bardet-Beidl syndrome
- Alstrom syndrome
- Refsum syndrome

Plus other rarer syndromes (details on our website) such as:

- Stickler syndrome
- NARP
- Bassen-Kornzweig syndrome
- Pseudoxanthoma elasticum (PXE)
- Norrie disease

In these syndromes, the genetic fault causes problems within other systems in the body, as well as retinal degeneration.

Conditions outside of Retina UK's charitable objectives:

These are conditions that are not directly caused by a genetic fault and are not inherited:

- Age-related macular degeneration (AMD); this includes wet AMD, dry AMD and geographic atrophy. For support, signpost to the Macular Society. NB Inherited juvenile macular degeneration is within Retina UK's scope, so please clarify if necessary.

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- Central serous retinopathy, also known as Central serous chorioretinaopathy. For support, signpost to the RNIB and Moorfields Eye Hospital
- Diabetic retinopathy. For support, signpost to Diabetes UK.
- Leber's hereditary optic neuropathy (LHON) – this condition does have a genetic cause, however the optic nerve, rather than retina, is primarily affected. For support, signpost to LHON Society.
- Retinopathy of prematurity (ROP) – occurs as a result of premature birth. For support, signpost to Tommy's, the pregnancy & baby charity.
- Retinal vein occlusion – occurs because a blood vessel to the retina becomes blocked.
- Retinoblastoma – a kind of eye cancer that starts as a growth of cells in the retina. Support available from Cancer Research UK. This condition does have a genetic basis but is a type of eye of cancer,
- Retinal detachment – a serious eye condition where the retina, the light-sensitive tissue at the back of the eye, pulls away from its normal position, potentially leading to vision loss if not treated promptly. More information available from Guide Dogs.

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