Lions Eye Institute

FACT SHEET

What is inherited retinal disease?

Inherited retinal disease (IRD) refers to a range of conditions that collectively represent the leading cause of blindness in adults of working age.

IRDs occur because of an **abnormality in one or more of a person's genes** that are required for maintaining normal vision.

IRDs can commence at birth but are more commonly diagnosed in childhood. If left undiagnosed, they may cause severe irreversible sight damage. IRDs affect one in 4,000 people globally.

The most common IRD is **Stargardt disease**, which is a form of inherited juvenile macular degeneration. The disease causes damage to the macula, which is at the centre of the retina.

Another relatively common IRD is **Retinitis pigmentosa**, a condition that causes cells in the light-sensitive retina to degenerate slowly and progressively.

Other IRDs are:

- Usher syndrome
- Cone-rod dystrophy
- Batten disease
- Blue cone monochromacy
- Bietti crystalline dystrophy
- Bardet-Biedl syndrome
- Alport syndrome
- Leber congenital amaurosis.

What causes inherited retinal disease?

IRDs are genetic conditions caused by a mistake in one or more genes.

Currently there are approximately 250 genes known to cause IRDs. There are many more genes, however these are currently unknown and yet to be discovered. Some gene mutations that cause IRDs are more severe than others.

With an IRD, **cells in the retina do not work correctly**. The retina is the light-sensitive tissue at the back of the eye that sends information to the brain through the optic nerve, enabling people to see.

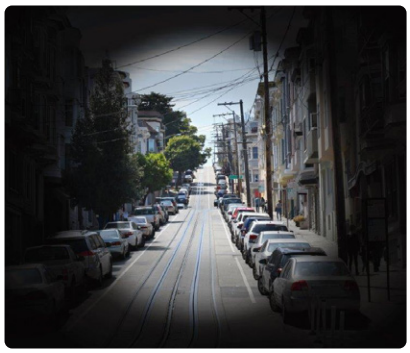
What are the symptoms of inherited retinal disease?

Some people with IRDs experience a gradual loss of vision, which may eventually lead to complete blindness. Others can be born with vision loss or experience it in early childhood.

How much sight a person loses relates to their age of onset.

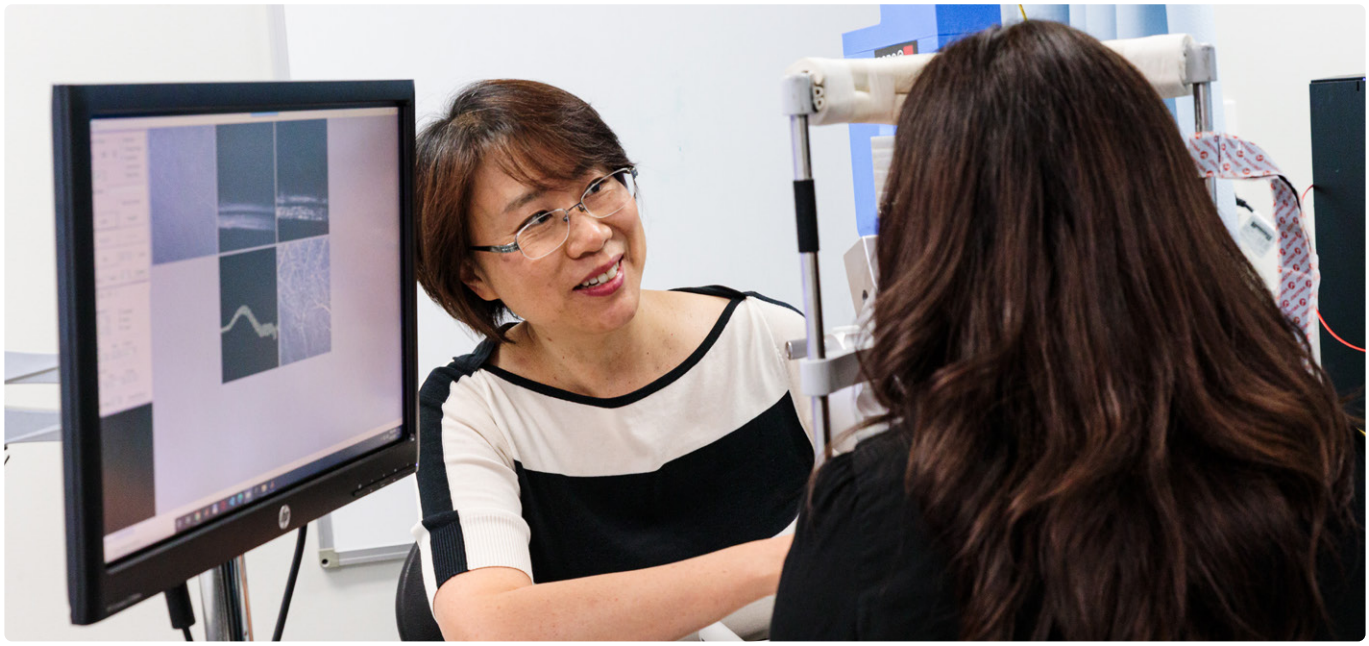
Typically, children suffer the worst impacts of IRDs, with some paediatric patients going almost completely blind.

Teenagers might have a gradual loss of central vision, while adults can lose patches of central vision.



Symptoms of IRD might include:

- Difficulty seeing in dark environments or in bright light
- Impaired peripheral vision
- Reduced central vision
- Not seeing colours or recognising differences between certain colours.



How is inherited retinal disease diagnosed?

As well as noting patient and family history, your ophthalmologist will undertake clinical eye examinations that are generally non-invasive.

They will perform eye image analysis, measuring retinal images to determine the rates of disease progression, and genetic analysis involving genetic testing to diagnose the cause of loss of sight.

Genetic testing is a type of medical test that identifies changes in chromosomes, genes or proteins. The results of a genetic test confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder.

Genetic counselling might be recommended, in order to help you understand the condition and what it might mean for families and individuals.

How is inherited retinal disease treated?

There is no cure for most IRDs, as too little is known about the genetic basis for these diseases. However, therapies such as gene therapy can be used to control the progress of disease.

A **crucial early step** is to collect information from you, for example analysing your genes and the changes in your eye using advanced imaging techniques.

Researchers and ophthalmologists are continually exploring gene therapies, stem cell technology and drug treatments that might prevent, slow or stop the damage that leads to vision loss and blindness.

Gene therapy aims to correct or compensate for the faulty gene. It might replace the faulty gene or add a new gene.

Need to know more?

Please contact the Lions Eye Institute to make an appointment with one of our ophthalmologists. Phone: (08) 9381 0777; email: carecentre@lei.org.au; or see our website: lei.org.au