RETINA australia

Cone-rod dystrophies

Cone-rod dystrophies refer to a group of inherited diseases that affect the photoreceptor (light sensing) cells that are responsible for capturing images from the visual field. These cells line the back of the eye in the region known as the retina. Cone cells are present throughout the retina, but are concentrated in the central region (the macula). They are useful for central (reading) vision. Rod cells are present throughout the retina except for the very centre and they help with night vision.

In contrast to typical retinitis pigmentosa (known as the rod-cone dystrophies), which results from the loss of rod cells and later the cone cells, cone-rod dystrophies reflect the opposite sequence of events, where cone cells are primarily first affected with later loss of rods.

What are the symptoms of Cone- Rod dystrophy?

The cone cells are initially involved, as previously mentioned, and difficulty with the clarity of vision, colour vision problems and light sensitivity can be some of the earliest symptoms experienced. This is followed by a progressive loss of rod cells, which leads to night blindness and loss of side vision.

The age of onset, progression and severity of cone-rod dystrophies can vary greatly from one person to another, even among individuals with the same type of cone-rod dystrophy. It is therefore very difficult to predict what an individual's vision will be like at a specific time in the future.

What is the Cause of Cone-rod dystrophy and how is it inherited?

Some forms of cone-rod dystrophy are inherited; other forms appear to occur spontaneously for no apparent reason (sporadically). Cone-rod dystrophies have many similarities to retinitis pigmentosa, and like RP they can follow varied inheritance patterns, including autosomal dominant, autosomal recessive, and X-linked.

What treatments are available?

Maximising an individual's remaining vision has remaining is a crucial first step to take. There are many new low vision aids, including telescopic and magnifying lenses, providing plenty of choice for users at all stages of sight loss. This technology has also removed many barriers to education and employment. There are, currently, no proven or effective cures for cone-rod dystrophies.

However, scientists have identified more than 20 genes that can have mutations which cause these conditions. It is likely that many more mutations in many more genes will be identified in the coming years. Many future treatments will rely on identification of these gene defects.

This is the reason why Retina Australia considers the Australian Inherited Retinal Disease Register and DNA Bank, designed to identify the genes responsible for inherited retinal degenerations in Australian patients, including those with cone-rod dystrophies, to be so important.

Despite the lack of current treatments for cone-rod dystrophies, general eye check-ups are important. People with these conditions are still at risk for other kinds of eye problems that can affect the general population and may be treatable. Patients with cone rod dystrophies tend to develop cataracts at an earlier age than the overall population.

Regular visits to your eye doctor can also make you aware of current advances as we learn more about these conditions. A small clinical Phase I/II pilot has recently (mid 2018) been concluded at the National Institutes of Health Medical Centre in Maryland, USA, involving the direct administration of Interferon Gamma-1bto the eye. No results are available as of yet.