Cone-rod dystrophies

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What are cone-rod dystrophies?

Cone-rod dystrophies refer to a group of inherited retinal diseases (IRDs) which cause damage to the photoreceptor cells. In contrast to typical retinitis pigmentosa (RP), which is also known as rod-cone dystrophy), cone-rod dystrophies cause loss of cone cells first, followed by rod cells. They cause central vision loss initially, but this usually extends to the periphery over time.

How are cone-rod dystrophies identified?

The appearance of a cone-rod dystrophy retina can vary significantly, depending on the causative gene mutation. General signs include a loss of photoreceptor cells on cross-sectional OCT scans (thinning of the retina), and atrophy of the retina. Definitive diagnosis requires genetic testing.

What are the symptoms of cone-rod dystrophies?

Blurred central 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 can vary greatly from one person to another, even among individuals with the same genetic mutation. 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 dystrophies and how are they inherited?

As in the case of rod-cone dystrophies (RP), the cone-rod dystrophies are highly variable in their presentation. Over 25 different genes have been identified and cone-rod IRDs have varied inheritance patterns, including autosomal dominant, autosomal recessive, and X-linked. Refer the Inheritance Patterns section of the website for more information.

What treatments are available?

There are currently no effective treatments. A number of emerging treatments are being developed that may assist in the future.

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