RETINA australia

Leber Congenital Amaurosis (LCA)

Leber Congenital Amaurosis (LCA) is a rare genetic eye disease that appears at birth or in the first few months of life. The extent of vision loss varies from person to person, but can be quite severe (with little to no light perception).

LCA is an umbrella term given to a group of diseases that are caused by mutations in at least 18 known genes. The gene mutations lead to failure in function of the photoreceptor cells (rod and cones cells that receive light), ultimately causing cell degeneration. Given the severity of the condition, it is one of the most extensively researched inherited retinal disorders, and a number of clinical trials have begun recently.

What are the symptoms of LCA?

There are many different types of LCA, and the disease can present differently in different children. However, there are some basic symptoms that are often associated with LCA. These include

- nystagmus (involuntary jerky rhythmic eye movements)
- Poor vision or blindness
- Strabismus (crossed eyes)
- photophobia (extreme sensitivity to light) and
- slow or no pupillary response to light – little or absence of visual responsiveness.

Eye-pressing and rubbing the eyes with a knuckle or finger can be common with babies and children who have very little vision. This can cause damage to the cornea (keratoconus) and lens and may result in a loss of fatty tissue around the eyes causing the eyes to look deep-set. The extent of degeneration depends on the type of LCA the child has and for some types of LCA the vision (or lack of vision) remains stable.

What is the cause of LCA and how is it inherited?

- LCA is a very rare condition which is estimated to affect around 1 in 80,000 in the population.
- Forms of LCA are caused by a mutation in one of a number of genes that are important for the development and function of the retina, resulting in vision loss or blindness.
- It is usually inherited in an autosomal recessive manner. However, there are rare incidences where the inheritance pattern may be autosomal dominant.

What treatments are available?

Currently, there is no cure for LCA.

However, for LCA caused by mutations in the RPE65 gene, there is now a gene therapy, Luxturna, that has been approved for clinical use by the US Food and Drug Administration (FDA, USA), National Institute for Health Care and Excellence (NICE, UK) and the Therapeutic Goods Administration (TGA) in Australia.

The RPE65 gene is involved in the production of a protein essential for the functioning of the retina. Luxturna delivers a functional copy of the RPE65 gene to the retina, restoring the production of the protein and improving vision. The treatment involves a one-time injection of the therapy into the affected eye, with benefits of lasting effects of improved vision.

It is important to note that Luxturna is only effective for LCA caused by RPE65 mutations and is not suitable for other forms of the disorder. Also, as with any medical treatment, there are potential risks and side effects associated with Luxturna, and it should only be administered by qualified healthcare professionals.

Treatments available for other forms of LCA that may help manage symptoms.

- Low-vision aids such as magnifying glasses, electronic reading devices, and braille
- Gene therapy, which involves replacing or correcting the defective genes responsible for the disorder
- Medications or supplements to address specific symptoms or nutritional deficiencies

It is important to note that not all treatments are suitable for all individuals with LCA, and treatment plans should be tailored to each person's unique needs and circumstances.