Leber congenital amaurosis (LCA) is a group of inherited retinal diseases characterized by severe impaired vision or blindness at birth. Some retinal experts consider LCA to be a severe form of retinitis pigmentosa (RP). The condition is caused by degeneration and/or dysfunction of photoreceptors.

The retina is a thin piece of tissue lining the back of the eye. The optic nerve sends electrical signals to the back of the brain to create the images we see.

Photoreceptors are cells in the retina that capture light and convert it into electrical signals.

Diagnosis

- Often with an affected infant, parents notice a lack of visual responsiveness and roving eye movements, known as nystagmus.
- Eye examinations of infants with LCA sometimes reveal normal-appearing retinas. In other cases, several abnormalities are observed.
- An electroretinogram (ERG) test measures retinal function and is often essential to establishing a diagnosis of LCA.
- A genetic test can often provide a definitive diagnosis.

Symptoms

- Oculodigital reflex: Children habitually press on their eyes.
- Eyes can appear sunken or deep set.
- Keratoconus: Cone shape to the front of the eye.
- Cataracts: Clouding of the lens through which light passes.

Genetic testing helps with attaining an accurate diagnosis. A patient and family with a genetic diagnosis are in a better position to understand which emerging treatment approaches and clinical trials are most appropriate for them.