Putting A Spotlight On...

Leber Congenital Amaurosis

What is Leber Congenital Amaurosis?

- Leber Congenital Amaurosis is a rare eye disorder that is genetic. Infants affected by it are often blind at birth. LCA is usually inherited as an autosomal recessive genetic condition.

What are the signs/symptoms?

- Symptoms include:
  - Oculodigital reflex: children habitually press on their eyes
  - Eyes appear sunken or deep set
  - Keratoconus: cone shape to the front of the eye
  - Cataracts: clouding of the lens through which the light passes

What is Leber Congenital Amaurosis a result of?

- Leber Congenital Amaurosis is a result of mutations in at least 27 genes. Changes in genes account for 80-90% of cases of LCA.

What are the complications of Leber Congenital Amaurosis?

- Severe vision loss
- Strabismus
- Nystagmus
- Photophobia
- Some infants may exhibit hearing loss, intellectual disability, and/or developmental delay.

References:

- rarediseases.org/rare-diseases/leber-congenital-amaurosis/?filter=ovr-ds-resources
- www.fightingblindness.org/diseases/leber-congenital-amaurosis-lca#about-the-disease-85
- www.ucsfhealth.org/conditions/leber-congenital-amaurosis-lca/symptoms

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