HOW IS LEBER CONGENITAL AMAUROSIS INHERITED?

Mutations in one of more than two dozen genes can cause LCA. It is inherited when both parents have one mutated copy of the gene and a normal copy. They are unaffected carriers of LCA.

The child has a 25% chance of inheriting the two copies of the mutated gene (one from each parent) to cause the disease.

GENETIC TESTING
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.

WHAT TO EXPECT WITH LEBER CONGENITAL AMAUROSIS:

**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

To learn more about living with Leber congenital amaurosis, finding a specialist, genetic testing or clinical trials, visit FightingBlindness.org or call 888-332-3667.