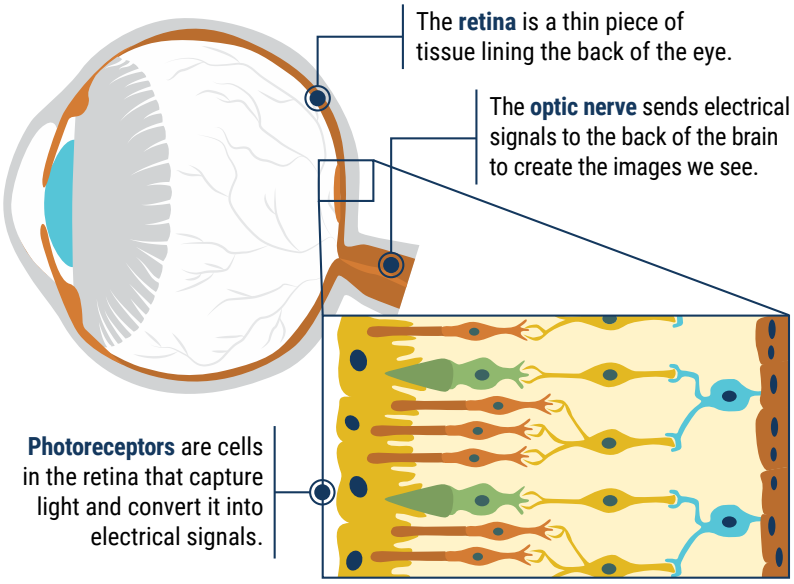
WHAT YOU SHOULD KNOW ABOUT

LEBER CONGENITAL AMAUROSIS

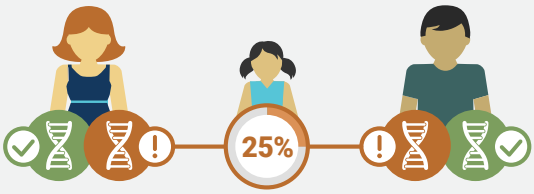
WHAT IS LEBER CONGENITAL AMAUROSIS (LCA)?

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.



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

FOUNDATION FIGHTING BLINDNESS

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**.