Leber Congenital Amaurosis

WHAT IS LEBER CONGENITAL AMAUROSIS?

Leber congenital amaurosis (LCA) is a rare group of medical problems that causes legal blindness at birth or in the first few months of life. Children with LCA have eye shaking (roving nystagmus), are very sensitive to light, their pupils (dark spot in the center of the colored part of the eye) respond poorly to light, and they have abnormal results on tests of the retina (electroretinograms). Some children will poke their eyes with a finger as a way to try to stimulate their eye.

WHAT CAUSES LCA?

LCA is a type of inherited retinal dystrophy, which means that the retina does not work well due to a genetic mutation in the DNA that someone is born with. The retina is the inner part of the back of the eye that senses light. LCA is caused by recessive genetic mutations, which means that a child gets one mutation from each parent. So far, researchers have found more than 25 different genetic mutations that cause LCA.

HOW COMMON IS LCA?

LCA is estimated to affect people at a rate between 1:30,000 and 1:80,000. Approximately 20% of children in schools for the blind have LCA.

HOW IS LCA DIAGNOSED?

LCA can be diagnosed with the help of an eye exam from a pediatric ophthalmologist. At first, the retina looks normal when a child with LCA is young. Other signs on the eye exam like nystagmus, light sensitivity and poor pupil reaction may cause the ophthalmologist to think that LCA may be present. The eye exam alone is often not enough to diagnose LCA. Instead, LCA Is typically diagnosed with a specialized retina test called an electroretinogram. This test measures the electrical activity of the retina. Sometimes, a detailed family history of vision loss and genetic testing are helpful.

WHAT CAN SOMEONE WITH LCA SEE?

People with LCA have very poor vision from birth; however, people have different amounts of vision loss. Typically, vision is worse than 20/400 (the largest letter at the top of a traditional eye chart). One third of people with LCA are not able to see light.

ARE THERE OTHER MEDICAL PROBLEMS IN PEOPLE WITH LCA?

Most children with LCA are otherwise healthy. Brain, bone, muscle, heart, ear, and kidney problems have been reported in some people with LCA. Genetic testing and exams with a child's pediatrician can figure out if extra testing for medical problems is needed. Most children with LCA have normal intelligence.

WHAT TREATMENTS ARE THERE FOR LCA?

So far, there is no cure for most patients with LCA. The U.S. Food and Drug Administration approved gene therapy for patients with LCA due to the *RPE65* gene mutation in 2007; however, this mutation is not a very common cause of LCA.

Most current treatments for LCA try to help patients get the most out of their vision and help them get access to work and school-related services. Children sometimes benefit from glasses and low vision aids, such as magnifying glasses. Genetic counseling can sometimes be helpful to patients and their families.

For more information on LCA please see:

- Lighthouse Guild — Tele-support group for parents
- https://eyewiki.org/Leber_Congenital_Amaurosis - scientific information for families

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