

**What is Leber congenital amaurosis?**

Leber congenital amaurosis (LCA) is an inherited retinal degenerative disease characterized by severe loss of vision at birth. A variety of other eye-related abnormalities, including roving eye movements, deep-set eyes, and sensitivity to bright light, also occur with this disease.

**What are the symptoms?**

Within an affected infant's first few months of life, parents usually notice a lack of visual responsiveness and roving eye movements, known as nystagmus. Eye examinations of infants with LCA can reveal normal-appearing retinas. However, an electroretinogram (ERG), which measures visual function, detects little if any activity in the retina. A low level of retinal activity, measured by an ERG, indicates very little visual function. ERG tests are essential to establishing a diagnosis of LCA.

By early adolescence, various changes in the retinas of patients with LCA become readily apparent. Blood vessels often become narrow and constricted. A variety of pigmentary (color) changes can also occur in the retinal pigment epithelium (RPE), the supportive tissue underlying the retina.

Although the appearance of the retina undergoes marked changes with age, the degree of vision loss usually remains fairly stable through young adult life. Visual acuity in patients with LCA is usually limited to the level of counting fingers or detecting hand motions or bright lights. Some patients are also extremely sensitive to light (photophobia). Patients with vision remaining are often extremely farsighted.

Many children with LCA habitually press their eyes with their fists or fingers. This habitual pressing on the eyes is known clinically as oculodigital reflex. The eyes of individuals with LCA can also appear sunken or deep set. Keratoconus (cone shape to the front of the eye) and cataracts (clouding of the lens, the clear, glass-like structure through which light passes) can occur with the disease.

In some cases, other body systems (e.g., kidneys) can be affected by the genetic defects that cause LCA.

**Is it an inherited disease?**

LCA is most often passed through families by the autosomal recessive pattern of inheritance. In this type of inheritance, both parents, called carriers, have one gene for the disease paired

## LEBER CONGENITAL AMAUROSIS, Continued

with one normal gene. Each of their children has a 25 percent chance (or 1 chance in 4) of inheriting the two LCA genes (one from each parent) needed to cause the disorder. Carriers are unaffected because they have only one copy of the gene.

### Are there any other related diseases?

Initially, LCA can be confused with congenital and hereditary optic atrophy, cortical blindness, congenital stationary night blindness, flecked retina syndrome, and achromatopsia. Although similarly named, LCA should not be confused with Leber optic atrophy. In addition, there are early onset syndromes that can cause visual impairment. A thorough ophthalmologic examination, including tests measuring retinal function and genetic testing, can provide a definitive diagnosis. Genetic counselors are excellent resources for discussing inheritability, family planning, genetic testing, and other related issues.

### What treatment is available?

Scientists have identified 24 genes that when mutated can each cause LCA. These genes account for about 75 percent of all cases of LCA. With this information, scientists are making excellent progress in developing treatments for saving and restoring vision.

Some individuals with LCA who have vision remaining may also benefit from the use of low-vision aids, including

electronic, computer-based, and optical aids. Orientation and mobility training, adaptive training skills, job placement, and income assistance are available through community resources.

The company Spark Therapeutics is seeking U.S. Food and Drug Administration approval for an RPE65 gene therapy that has restored vision in children and young adults with RPE65 mutations who were participating in a clinical trial. Early funding from the Foundation helped develop this treatment.

For the latest research advances for LCA, refer to the Foundation publication: [Leber Congenital Amaurosis: Research Advances](#).

**Low-vision resources and extensive information on research and clinical trials for Leber congenital amaurosis are available at [www.FightBlindness.org](http://www.FightBlindness.org).**