Fact Sheet

Leber’s Congenital Amaurosis

Compiled by Tanni L. Anthony

Information retrieved from: http://www.wonderbaby.org/articles/lca-faq.html#what-is-lca
and http://www.aapos.org/displaycommon.cfm?an=1&subarticlenbr=264

What is LCA? Leber’s Congenital Amaurosis (LCA) is a genetic disorder in which retinal dysfunction causes vision loss, often from birth. It is rare (3 cases per 100,000 births). About 10% of cases of congenital blindness or severely-reduced vision are caused by LCA. The extent of vision loss varies from person to person, but can be quite severe (with little to no light perception). LCA is named after the doctor who first described the condition, which is exists since birth and is usually hereditary. The amaurosis refers to a total loss of vision, especially when the vision loss does not result in any apparent change to the eye. LCA can also be seen as part of a system of disorders (e.g. neurologic dysfunction, kidney disease, and, rarely, chromosomal imbalance).

How is LCA Diagnosed? LCA is usually diagnosed by an ophthalmologist, based on the history and the physical findings. An electroretinogram (ERG) is often utilized to test the function of the rods and cones of the retina and is performed with special equipment. Blood tests for genetic changes can sometimes help make the diagnosis as well as identify carriers of the condition. The ERG (electroretinogram) tests the retina's response to light. The ERG involves placing contacts on the child's eyes that are hooked up to a machine by wires. Young children may be mildly or fully sedated for the exam. ERGs produce graphs that in normal eyes move up and down, while LCA eyes will produce comparatively flat graphs. The ERG cannot tell how much a person can or cannot see, it can only diagnose LCA. The second test is a genetic screen of the patient's blood that looks for the nine known genetic variations that cause LCA. This test involves drawing a blood sample. Some parents opt out of both tests and choose to diagnose their child based only on symptoms. This is always an option if the genetic test is too expensive (or turned up negative results) and the ERG seems too stressful for your child. Many doctors will provide a tentative diagnosis of LCA if the patient presents most of the common symptoms, even without testing.

What are the symptoms of LCA? In LCA, children are born with an absence of light-gathering cells (rods and cones) of the retina. A lack of visual attentiveness at and shortly after birth is the first sign of the disease. There are many different types of LCA and the disease will present differently in different patients. However, there are some basic symptoms doctors usually look for when diagnosing LCA:

- Severe vision impairment from birth
- Nystagmus (involuntary jerky rhythmic eye movement)
- A normal looking eye upon examination (though there may be some pigmentation on the retina)
- Extreme farsightedness
- Photophobia (sensitivity to light)
- Slow pupillary response to light
- Eye-pressing can be common with babies and children are visually impaired due to a retinal disorder. This can cause damage to the cornea and lens and may result in a loss of fatty tissue around the eye causing the eyes to look deep-set or sunken.

**How is LCA treated?** There is currently no treatment for LCA. It is primarily a genetic disorder and the most promising research right now involves gene discovery and gene therapy. Researchers have discovered nine genes that cause LCA so far, but they believe there are many more. To learn more about the known genes that cause LCA, visit the Foundation for Retinal Research web site at [http://tfrr.org/index.php?m=19&PHPSESSID=edaf92021de3b19214a5f501726a1f47#6](http://tfrr.org/index.php?m=19&PHPSESSID=edaf92021de3b19214a5f501726a1f47#6)

Routine examinations by an ophthalmologist are recommended to diagnose/treat other eye problems and prescribe glasses if necessary. Low vision aids may be utilized to maximize vision function. Educational programs and support agencies for the visually impaired (and family) are important. A certified teacher of students with visual impairments and a certified Orientation and Mobility Specialist will be key members of the educational team.

**Resources:**

The Foundation for Retinal Research funds LCA research. Their url is: [http://www.tfrr.org/index.php?m=19&PHPSESSID=7b15cac17c2c98e1a8954d64e0354faf](http://www.tfrr.org/index.php?m=19&PHPSESSID=7b15cac17c2c98e1a8954d64e0354faf)

Their *What is LCA* page is very informative.

**For more information about the CO Services for Children and Youth with Combined Vision and Hearing Loss Project and the project’s services specific to Colorado children who are deafblind, please contact:**

Tanni Anthony  
Phone: 303-866-6681  
Email: [anthony_t@cde.state.co.us](mailto:anthony_t@cde.state.co.us)

Gina Quintana  
Phone: 303-866-6605  
Email: [quintana_g@cde.state.co.us](mailto:quintana_g@cde.state.co.us)

Colorado Department of Education  
Fax: 303-866-6767  
Web Page Address: [http://www.cde.state.co.us/cdesped/Deafblind.asp](http://www.cde.state.co.us/cdesped/Deafblind.asp)

1560 Broadway, Suite 1175  
Denver, CO 80202

*Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 9/10*