Leber Hereditary Optic Neuropathy

What is Leber Hereditary Optic Neuropathy (LHON)?

Leber's hereditary optic neuropathy (LHON) is a genetic disease that primarily affects the optic nerves, leading to vision loss (typically in both eyes). In LHON, **one of the genes that controls energy production in the optic nerves is abnormal**, and over time the waste products of that energy production begin to damage the optic nerves. The abnormal gene is found in the DNA that exists in our mitochondria, the parts of our cells that control energy production, not in the DNA that makes up our chromosomes in the nucleus.

Over 50% of people with LHON experience sudden, painless, rapidly progressive visual loss in both eyes, whereas others experience rapidly progressive **painless vision loss in one eye, followed by similar vision loss in the other eye weeks or even months later**. The amount of vision loss usually is severe involving the central part of the visual field, but total blindness is very unusual. In many patients, the back of the eye, including the optic nerve head (the part of the optic nerve that can be seen) **can look completely normal at first**, which can make the diagnosis very challenging for doctors. Although the vision loss is typically permanent, there are some people who will recover some of their vision, depending on the specific genetic defect. Rarely, people with LHON will also have other problems, which may include heart rhythm problems, muscle spasms, or brain problems (encephalopathy).

There are many mitochondrial gene defects that can cause LHON, although most people (90%) have one of three mitochondrial DNA gene mutations, referred to as the 3 primary mutations.

Who Can Get LHON?

Although most people with LHON are between 15-35 years old, it can affect anyone of almost any age and although men are affected more often than women, women can definitely become affected. Some doctors believe that habits and activities that increase the body's energy and waste-removal needs (such as smoking, poor nutrition, heavy binge drinking of alcohol, etc.) can worsen or even trigger the onset of visual loss. Because LHON is a genetic disease, people with

family members who have unexplained vision loss may have the disease. Because mitochondrial DNA all comes from mothers, **LHON is passed down through mothers and men cannot pass the disease to their children**.

How is LHON Diagnosed?

Before genetic testing was available, doctors would diagnose people with LHON if someone had unexplained vision loss with normal testing (brain scans and lab tests) and the history of vision loss was very suspicious for LHON or there were family members with similar vision loss. There is now genetic testing available for LHON that can confirm the presence or absence of the three major gene defects from a blood sample.

How is LHON Treated?

Until very recently, there were very few treatments available for LHON. Researchers had looked at many different treatments, most notably with an antioxidant called idebenone (related to coenzyme Q) having some promising initial results.

Since 2015, gene therapy by an injection into the eye has been studied as a method of treating LHON in its first year of symptoms with potentially promising results. Research is ongoing.

What Should I Do About My Condition?

Your doctor will follow you closely while your vision is declining and may recommend that you take idebenone. Once your vision is stable, your doctor will begin to extend follow-up visits. Your doctor may recommend that you refrain from certain activities, such as smoking or heavy alcohol consumption.

Your doctor may recommend that you obtain an EKG (electrocardiogram), which looks at your heart rhythm to make sure that you do not have any abnormalities. This may be obtained through your primary care provider.

Your doctor may also talk with you about a **low vision evaluation** or visiting **visual rehabilitation** services. These centers use devices and lifestyle modifications to help maximize the vision you

still have. Although there may be some out-of-pocket expenses with these services, they may be able to help improve your quality of life during this difficult transition.

There are also support groups available for people with LHON. Although LHON is rare, you do not have to suffer alone. A link to one of the major support groups is listed below.

Other Resources

- **National Library of Medicine (patient-oriented article on LHON):**
  - https://ghr.nlm.nih.gov/condition/leber-hereditary-optic-neuropathy
- **GeneReviews (textbook article on LHON - very technical):**
  - http://www.ncbi.nlm.nih.gov/books/NBK1174/
- **LHON.org (support group):**
  - http://www.lhon.org

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