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.

Over 50% of people with LHON experience sudden bilateral, painless, rapidly progressive visual loss, 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, 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 or even all of their vision. Some people with LHON will also have other problems, which may include heart rhythm problems, muscle spasms, or brain problems (encephalopathy). There are also some associations between LHON and multiple sclerosis.

There are many genes defects that can cause LHON, though most people only have one gene mutation.

Who can get LHON?

Although most people with LHON are between 20-50 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, drinking alcohol, etc.) may 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. It is passed down through mothers, so 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 a genetic test available for LHON that can confirm the presence or absence of the three major gene types from a blood sample. The genetic test can be expensive; you may need to talk with your insurance company to see if they may be able to help cover part of the cost.

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. A large study looking at brimonidine, a glaucoma medication, did not show any significant improvement in outcome.

However, since 2015, gene therapy by injecting medication into the eye has been studied as a method of treating LHON in its first few months of symptoms with somewhat promising results.

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 heavy exercise, smoking, or alcohol consumption, to reduce your body’s energy production needs.

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):**

- **GeneReviews (textbook article on LHON - very technical):**

- **LHON.org (support group):**
  - [http://www.lhon.org](http://www.lhon.org)