Leber’s hereditary optic neuropathy (LHON)

LHON is a maternally-inherited form of vision loss

LHON is a rare disease causing progressive, central vision loss in both eyes.\textsuperscript{1-4} It is caused by dysfunction of the optic nerve that transmits visual information from the retina to the brain.\textsuperscript{6} LHON most commonly presents as painless loss of central vision in one eye, followed rapidly by loss of vision in the other eye within a few months.\textsuperscript{1-4} In some cases, both eyes will be affected from the onset.\textsuperscript{7}

Severe loss of central vision typically occurs within 1 year of the onset of symptoms.\textsuperscript{1,3,4}

LHON is caused by mutations in mitochondrial DNA

Mitochondria are organelles within cells that produce the energy vital for cell function. LHON is caused by mutations in mitochondrial DNA that encodes some proteins essential for energy production. The mutations change the protein so that it cannot function properly, and the mitochondria cannot produce energy as efficiently. Some cells such as the nerve cells at the back of the eye, known as retinal ganglion cells, need more energy than others. Retinal ganglion cells are strongly affected by reduced energy production and may stop working properly or even die.

Not all carriers of a LHON mutation will develop symptoms\textsuperscript{7}

Where there is one LHON patient, there is a family at risk:
Genetic carriers are people with LHON mutations who have no symptoms. Genetic counseling and lifestyle changes for carriers can reduce the risk of vision loss for family members who have normal vision.\textsuperscript{11,12}

Male carriers develop symptoms 5 times more often than female carriers\textsuperscript{3}

An estimated 140,000 people are affected by LHON worldwide\textsuperscript{8-10}
Risk factors for LHON carriers to develop vision loss

- Smoking
- High alcohol consumption
- Acute illness or psychological stress
- HIV medication
- Industrial toxins
- A lack of important vitamins or minerals

Getting to the right diagnosis

What are the most important symptoms you must tell your doctor about?
1. Painless, central vision loss
2. Family history of vision loss

Tests used for diagnosing LHON
1. Visual acuity test and standard eye tests
2. Genetic test for LHON mutations

Most people affected by LHON receive at least one misdiagnosis, the most common being multiple sclerosis.

60% of LHON patients have a family history of vision loss.

Visual acuity and standard eye tests
Simple blood test for the most common LHON mutations

Living with LHON

Those affected by LHON may face challenges such as:

- Lack of information about disease
- Difficulty in obtaining a correct diagnosis
- Few physicians with experience of LHON
- Acute depression, anxiety and stress
- Lack of access to quality healthcare
- Limited disease awareness among families, physicians and society as whole

Santhera’s work in rare diseases

For more than a decade, Santhera Pharmaceuticals has been committed to developing medicines to meet the needs of people living with mitochondrial disorders and other rare diseases. We continue to strive towards improved treatment options for people with LHON.

For more information about ongoing clinical trials, please visit www.clinicaltrials.gov (NCT02774005 and NCT02771379) or email ClinicalTrial_LEROS@Santhera.com.
For patient advocacy information, please contact patient.advocacy@santhera.com.

REFERENCES

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