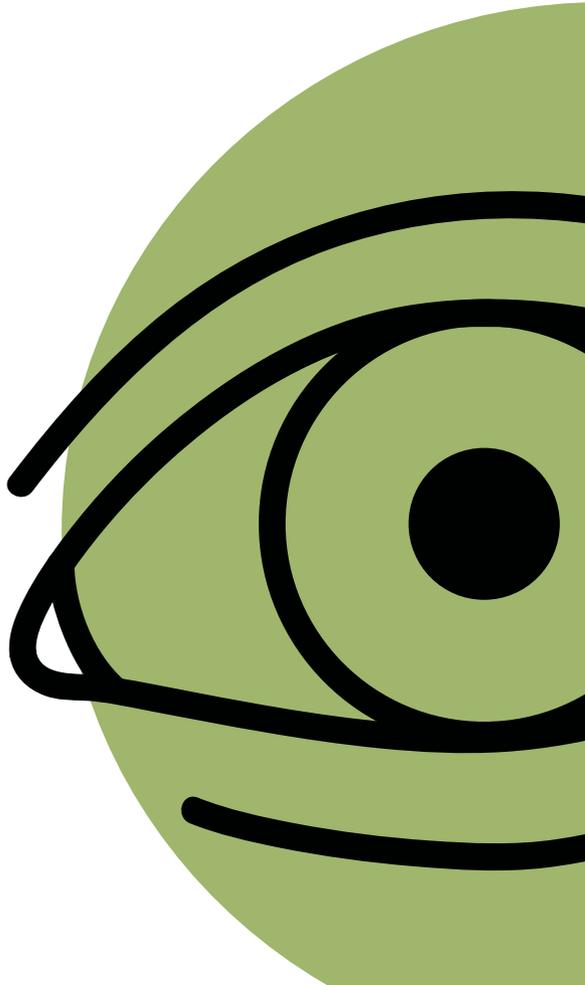




LHON
AWARENESS
DAY

LHON

**LEBER'S
HEREDITARY
OPTIC
NEUROPATHY**



international mito-patients

WHAT IS LHON?

LHON stands for Leber's Hereditary Optic Neuropathy or Leber Optic Atrophy.

LHON is a mitochondrial disease (mito) caused by a change in the function of mitochondria, which are the energy producing organelles.

In LHON the optic nerve is particularly vulnerable to mitochondrial defects because of the high-energy requirements and the need to keep the retina transparent to light.

LHON causes acute vision loss, occurring most commonly in adolescents or young adults. It is caused by mitochondrial DNA mutations and can only be inherited from the maternal line, unlike nuclear DNA

mutations. The mother can be unaware of transmitting the risk for such a disease as she frequently carries the mutation without being affected. There are three mitochondrial DNA mutations which are responsible for the majority of LHON cases (about 90%), however, more rare mutations have been discovered that also cause central vision loss – a typical hallmark of LHON.

LHON has a prevalence of 1/25.000 to 1/50.000 and it is more frequent in males.

WHAT ARE THE SYMPTOMS?

Patients typically notice vision loss in one eye, as if a small part of the picture had been cut out in the center. This usually bright spot, expands towards the edges leaving a small peripheral residue. At this point the other eye also starts to lose its central vision. This central hole is called "scotoma" and it may have different shapes, from a bright ball to a star. In the majority of cases the patient begins losing vision in the second eye a few weeks or months after initial onset, but a bilateral involvement can be also manifest since the beginning of the disease. Loss of vision in the second eye can lead to blindness, as the brain is not used to employing the peripheral visual residue.

The peripheral visual residue gives a low kind of vision, similar to a monitor where the pixels are no longer working and the image is composed of lots of little bright grey and white dots, this is sometimes known as "chess board vision".

HOW CAN OPHTHALMOLOGISTS HELP?

Unfortunately, LHON is often mistaken for Optic Neuritis and therefore Multiple Sclerosis, an ischemic lesion or even a brain tumor. It is important that eye doctors ask patients whether there is any family history of vision loss along the maternal line, and if it is accompanied by pain in the eye region,

which is usually absent in LHON. During an eye examination in the acute phase the back of the patient's eye in some cases may show typical signs of the disease (optic disc pseudoedema, hyperemia and microangiopathy) but in other cases it may look completely normal. Further investigations such as Optical Coherence Tomography (OCT), visual fields and electrophysiology (Pattern visual evoked potentials and Pattern Electroretinogram) may help to gain an accurate diagnosis. The OCT is the most reliable examination for quantitatively measuring the degree of damage of the optic nerve and its progression.

WHAT IS IT LIKE TO HAVE LHON?

Some patients describe it as being constantly blinded by the sun, day and night. LHON patients are unable to recognize faces, read books or street signs because of the constant bright light or large grey blind spot.

LHON, which can lead to legal blindness, is not characterized as the type of visual impairment mainly manifested in low light situations. Therefore, it must not be mistaken for Leber's Congenital Amaurosis, which is also called "darkness disease", a retinopathy not caused by a dysfunction of the mitochondrial metabolism.

Voice synthesizers and other technology on smart phones and computers can

significantly help legally blind people work, study and complete everyday activities.

IS THERE A CURE?

Currently, there is no definitive cure or way to stop the progression of the disease. High dosages of antioxidants, such as Idebenone, can help to stabilize the course of the disease and, if diagnosed early, can significantly lower the damage in both eyes, thus favoring vision improvement. Research into gene therapy, and other possible therapies is taking place around the world, with the hope that treatments and cures will be soon available, as currently under development.

As a means of prevention, it is strongly recommended to avoid smoking, as well as alcohol.

LHON AWARENESS DAY

The LHON Awareness Day is organized worldwide by International Mito Patients (IMP), the Federation of Mitochondrial Disease Patients Organizations, in order to raise awareness and understanding on Leber's Hereditary Optic Neuropathy (www.mitopatients.org). This year it occurs on the last day of the World Mitochondrial Diseases Week, which takes place every year during the third week of September with the aim to sensitize on mitochondrial diseases.

The LHON Awareness Day logo represents a rosemary branch, an evergreen plant, which grows around the world and has many beneficial health effects. Rosemary is the symbol of memory and energy, and its shape is similar to a mitochondrion. The color green represents mitochondrial diseases and so, whenever you see a rosemary branch, think of LHON and hope for a cure.

FURTHER INFORMATION

Visit the International Mito Patients (IMP) website to find your local mitochondrial disease organisation.



international mito-patients

International Mito Patients
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The LHON Awareness Day is supported by IMP.

PARTICIPATING COUNTRIES

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www.lhoncanada.ca



Germany

LHON-Deutschland.e.V
www.lhon-deutschland.de



Italy

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The Netherlands

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