

Rare Disease Day 2023: GenSight Biologics Raises Awareness of Leber Hereditary Optic Neuropathy (LHON)

- Leber Hereditary Optic Neuropathy (LHON) is a rare disease predominantly affecting people aged 15 to 35 year that causes irreversible and severe vision loss, resulting in blindness
- Patients, caregivers and society as a whole bear the burden of LHON
- Lack of awareness of LHON leads to misdiagnoses and imposes a psychological toll on patients and their families
- Launch of a European LHON awareness campaign in close collaboration with LHON experts

Paris, France, February 28, 2023, 7:30 am CET – GenSight Biologics (Euronext: SIGHT, ISIN: FR0013183985, PEA-PME eligible), a biopharma company focused on developing and commercializing innovative gene therapies for retinal neurodegenerative diseases and central nervous system disorders, highlights its commitment to fight rare neurodegenerative diseases by raising awareness of Leber Hereditary Optic Neuropathy (LHON) at Rare Disease Day through the launch of a European awareness campaign in Q1 2023, in close collaboration with LHON experts.

Rare Disease Day is a globally coordinated movement aiming to raise awareness for the 300 million people living with a rare disease worldwide. GenSight Biologics is pleased to join the movement and take this opportunity to raise awareness for LHON, a rare disease with significant unmet need.

Leber Hereditary Optic Neuropathy (LHON): a rare, devastating disease that causes irreversible and severe vision loss, resulting in blindness

LHON is a relatively unknown disease that strikes people in the prime of life. It is characterized by rapid, usually irreversible, painless, and severe vision loss¹⁻⁴.

This condition is thought to affect from one out of 30,000 to one out of 50,000 people in Europe¹⁻³. Though women are also affected by the condition, LHON most often impacts young men². Onset typically occurs between ages 15 and 35, but the disease has been observed in patients from 2 to 87 years of age¹.

Patients typically lose central vision in one eye and within a few weeks or months, vision begins to degrade in the second eye⁵. Visual acuity eventually stabilizes at a very low visual acuity¹, the natural progression of the disease leads to legal blindness⁶.

Patients, caregivers and society as a whole bear the burden of LHON

Patients are affected physically but also socially and mentally^{7,8}. The deterioration of central vision severely impedes the ability to carry out day-to-day activities such as reading, writing and cooking. Social interactions become more difficult, and individual pastimes become more challenging to pursue. For many

patients, the loss of autonomy is an emotional blow. Taking a walk, become nearly impossible ⁷⁻⁹ for example. Patients also report feeling psychological impacts from the disease.

In an online survey, **nearly 22% of LHON patients reported thoughts of suicide** after experiencing bilateral vision loss ¹⁰.

“I was told that I had LHON, that it was a genetic disease transmitted by the mother, that it was rare and that I would lose my sight in three months. Everything is slipping away: I won't be able to drive, I won't be able to see my wife's face, I won't be able to distinguish colors or what I eat”.
LHON patient

Though often forgotten, caregivers may also find themselves affected by LHON as well⁹. They might find their lives upended as a result of the demands of the disease, and caregivers report feeling compelled to put careers on hold or to reorganize their lives in order to accompany the patient day in and day out ⁹.

“I've needed to be part time really, to have the time and emotional energy to deal with things that suddenly crop up and paperwork and dealing with meetings and so on,” parent of a LHON patient.

LHON's impact is felt beyond the immediate social circle of the patient. Other costs associated with the condition include increased medical costs due to repeat hospital visits and health care administration, as well as the cost of low-vision aids, home-based care, and modifications to a patient's home ¹¹.

Estimates of the annual productivity loss from legal blindness run to \$411 billion⁸.

Lack of awareness of LHON leads to misdiagnoses and imposes a psychological toll on patients and their families

LHON, like many rare diseases, is often overlooked and diagnosis delayed because it can be confused with more well-known conditions like optic neuritis (multiple sclerosis) and retinopathy^{12,13}. It is a psychological tool for the patients and their loved ones who are often sent to various physicians before to finally obtain a definitive diagnosis thanks to genetic testing. Interventions that support psychological health, reinforce coping mechanisms, and change illness perceptions are critical to patients with LHON ⁹. Additionally, intervention to educate patients on visual aid technologies may help improve their psychological well-being, and local patient groups and charities can also provide support to those with visual impairment ^{14,15}.

Launch of an awareness campaign towards ophthalmologists and neurologists in Europe in close collaboration with LHON experts

As a healthcare company, GenSight has a responsibility that goes far beyond giving access to breakthrough solutions to patients. We are committed to actively support and help to improve the care of patients suffering from rare ophthalmic diseases, such as LHON. The Company has therefore developed, in close collaboration with LHON experts, an awareness campaign to educate and increase the understanding of this devastating disease. The ultimate goal: accelerate time to diagnosis.

The campaign will be launched in the top 4 EU countries and UK in Q1 2023 by the GenSight local teams in the countries and is addressed to ophthalmologists and neurologists.

Any person suspecting to be suffering from LHON should contact his health care professional and seek support from the local patient groups.

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About GenSight Biologics

GenSight Biologics S.A. is a clinical-stage biopharma company focused on developing and commercializing innovative gene therapies for retinal neurodegenerative diseases and central nervous system disorders. GenSight Biologics' pipeline leverages two core technology platforms, the Mitochondrial Targeting Sequence (MTS) and optogenetics, to help preserve or restore vision in patients suffering from blinding retinal diseases.