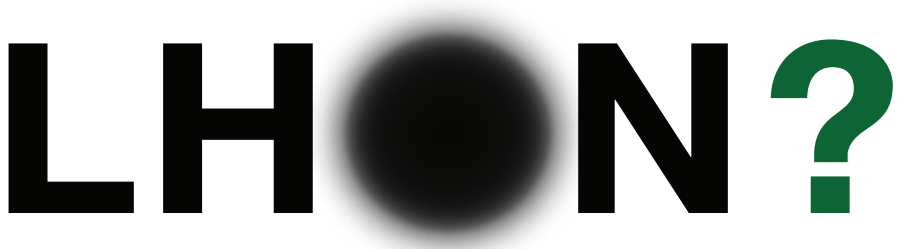
COULD IT BE



Leber's **H**ereditary **O**ptic **N**europathy

LHON causes rapid, painless, central loss of vision, typically leading to blindness[1,2]

WARNING SIGNS *for* SUSPICION *of* LHON[1-4,10]

- **Rapid, painless, central loss of vision**
- One eye affected initially, followed by the second eye within weeks to months
- Family history of loss of vision or LHON*
- Most often central or centrocaecal scotoma
- The pupillary light reflex is relatively preserved despite the severity of optic atrophy[6]
- Microangiopathy, telangiectatic vessels and hyperemia of the optic disc in the acute stage
- Optic disc pseudo-oedema and retinal nerve fibre layer thickening in the first months after disease onset, followed by progressive thinning[7**]
- Early and persistent ganglion cell inner plexiform layer thinning detectable before onset of visual loss[8]
- Non-responder to glucocorticoids

* Family history of LHON may be absent in up to 40% of cases[5] ** Retina can lack typical features of LHON in 20–40% of cases[9]

LHON typically presents in males aged 15-35, but can occur in either sex at any age

References: 1. Yu-Wai-Man P, et al. Prog Retin Eye Res. 2011; 30:81-114. **2.** Mascialino B, et al. Eur J Ophthalmol. 2012; 22:461-465. **3.** Newman NJ, Nat Rev Neurol. 2012; 8:545-556. **4.** Fraser JA, et al. Surv Ophthalmol. 2010; 55:299-334. **5.** Yu-Wai-Man P et al, J Med Genet. 2002 Mar;39(3):162-9. **6.** Moura AM, Invest Ophthalmol Vis Sci. 2013; 54(7): 4471–4477. **7.** Barboni P et al. Ophthalmology. 2010;117(3):623-7. **8.** Balducci N, et al., Br J Ophthalmol, 2016-09, Vol.100 (9), p.1232-1237. **9.** Yu-Wai-Man P et al. Eye. 2014 28(5):521-37. **10.** Carelli V et al. J Neuroophthalmol. 2017;37(4):37181.

315-2021-MARK October 2021

Chiesi Pharma AB Klara Norra kyrkogata 34, 111 22 Stockholm,chiesipharma.se | chiesipharma.dk I chiesi.fi | chiesi.no

Chiesi
Global Rare Diseases