Leber Hereditary Optic Neuropathy

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Bita SHALBAFAN MD

Leber Hereditary Optic Neuropathy (LHON) is the commonest primary mitochondrial disease associated with bilateral optic neuropathies. It is recognized as the most frequent cause of isolated blindness in young men. Maternally-transmitted LHON pedigrees show incomplete penetrance with a male preponderance for visual loss. Age of onset is variable ranging from the first to eighth decade.

Visual loss associated with LHON is characteristically subacute, central and painless. Typically visual loss occurs in isolation. Visual field defects consistent with central or cecocentral defects are typical. Colour vision dysfunction may be an early and severe feature of LHON-associated visual loss. Pupillary function is relatively preserved. Fundoscopic changes are characterized by peripapillary telangiectatic microangiopathy or circumpapillary nerve fiber layer swelling (pseudooedema) in the early stages ,later followed by non-specific optic atrophy with nerve fiber layer drop out, especially in the papillomacular bundle.

Although LHON is typically associated with isolated visual loss, neurological and multisystem organ involvement has been recognized in association with a multiple-sclerosis-like illness (usually in women), dystonia, myelopathy, neuropathy, severe encephalomyelopathy and cerebellar ataxia.

One of three mtDNA point mutations account for up to 95% of known cases of LHON: m.3460G>A, m.11778G>A, m.14484T>C all of which occur in mitochondrial-encoded structural subunits of complex I. Younger age-at-onset (less than 15 years) and mutation type appear to dictate visual outcome; patients with the m.14484T>C MTND6 mutation have a better visual prognosis with 60% attaining some visual improvement compared to only 5% of those harbouring the m.11778G>A MTND4 gene mutation.

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Neurologist, Specialist on Neurometabolic Disorders, Social Security Organization, Tehran, Iran
Corresponding Author:
Shalbafan B. MD
North Kargar Ave, Nosrat St, Sara Medical building complex, Tehran, Iran
Tel: +98 9161119656
Email: shalbafan.b@gmail.com