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(CASE REPORT)

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MRI Findings in Leber's Hereditary Optic Neuropathy: A case report

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Abstract

Background: Leber's hereditary optic neuropathy (LHON) is a rare genetic condition which is mainly presented with painless, sub-acute central unilateral vision loss followed by contralateral vision loss after few weeks to months. Young adults are typically affected with men preponderance. Relatively, it is a common cause of blindness as a result of mutation in mitochondrial DNA (mtDNA).

Case report: We reported the case of two brothers, who had recent history of vision loss. Fundoscopy examination revealed bilateral optic nerve atrophy along with vessel narrowing and absent macular reflex. We performed MRI bilateral orbits with contrast which revealed abnormal signals and atrophy of the optic nerves suggesting optic neuritis.

Conclusion: Whenever young males develop bilateral sequential vision loss with a positive family history, the diagnosis of Leber's hereditary optic neuropathy should be suspected. Ophthalmological examination may reveal signs suggestive of Leber's hereditary optic neuropathy. MRI may demonstrate edema signals in both optic nerve along with thinning of nerve fibers suggesting nerve atrophy. MRI is considered an useful tool to show optic nerve abnormality and optic neuritis.

Keywords: LHON (Leber's hereditary optic neuropathy); MRI; Optic neuritis; Vision loss; Central scotoma.

1. Introduction

Leber's hereditary optic neuropathy (LHON) which was first described by Theodore Leber a German ophthalmologist, is a genetic disorder which cause focal degeneration of retinal ganglion cell in the retina and axonal degeneration along with demyelination of the central part of the optic nerve. It is clinically characterized by acute and subacute sequential and painless visual loss with large bilateral centrocecal scotoma. It is a rare disease prevalent in men more than women and affect young adults [1]. Its prognosis is very poor with end result of significant impaired vision.

In our case, two young brothers show symptoms compatible with LHON and thier MRI findings are discussed accordingly.

2. Case report

We reported the case of two brothers with family history of vision loss, who were referred from eye department with proper clinical history, examination and fundoscopy report. They had recent history of vision loss, however there was

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no history infection, muscle weakness or seizers. Fundoscopy examination revealed bilateral optic nerve atrophy along with vessel narrowing and absent macular reflex. We performed MRI bilateral orbits with contrast with a 1.5 tesla machine (TOSHIBA) and found that both optic nerves appeared swollen with diffuse T2 hyperintensity extended from optic disc up till optic canal (Fig. 1) along with post contrast enhancement (Fig. 2). Beyond this optic chiasma is thinned out. No mass lesion, nerve compression or displacement seen. However MRI brain was negative for demyelinating disease. Based on clinical, fundoscopy and MRI findings LHON was diagnosed which later confirmed by blood test.

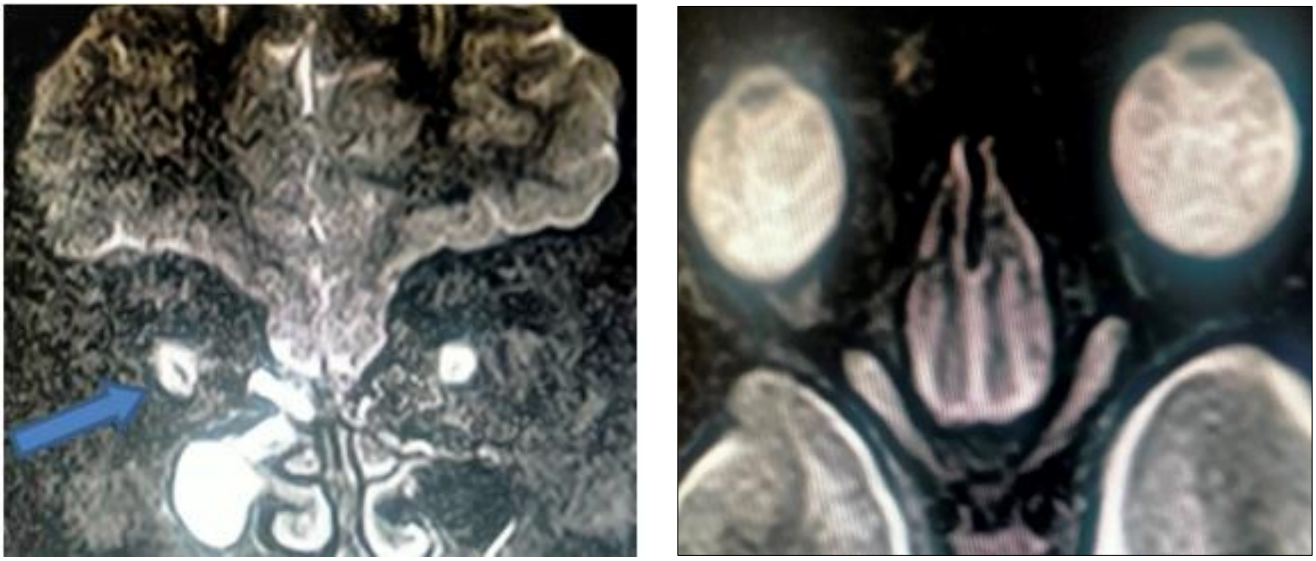

Figure 1 T2WI/FAT SAT (Axial and Coronal) show hyperintense signals in both optic nerve

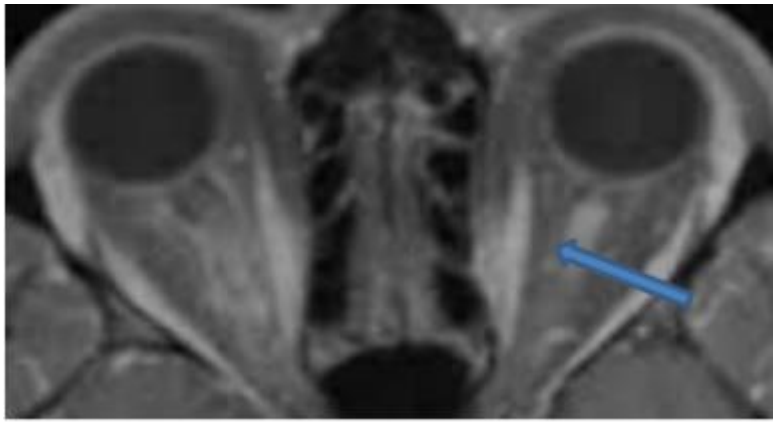

Figure 2 T1WI/FAT SAT post contrast image shows enhancement of optic nerves bilaterally

3. Discussion

LHON is a rare disease which is inherited as autosomally dominant, recessive or X-linked [2]. It is prevalent in men more than women and affect young adults resulting in vision loss due to loss of retinal ganglion cells and axon of optic nerve.

In 50% of patients the disease has maternal inheritance, while in other 50% individuals it is sporadic.[3,4]

In Leber's hereditary optic neuropathy, males in their second and third decade of life suffer acute sequential vision loss in 78% of cases while they are suffered simultaneously in 22% of cases.[3]

In initial stages, the fundus examination reveals papilledema and peripapillary microangioapthy, leading to leading to atrophy of the nerve fiber layer of the retina and optic nerve atrophy along with centrocecal scotoma.[5] however, the

ophthalmologic changes in the presymptomatic stage include microangiopathy, retinal nerve fiber layer swelling and tortuosity of small vessel.

Clinically, it is characterized by acute and subacute sequential and painless visual loss with large bilateral centrocecal scotoma, optic atrophy and dyschromatopsia.

In the acute phase of LHON, the first symptoms are usually sudden painless blurring and clouding of vision, which begin in one eye or both eyes simultaneously. If one eye is affected by the disease process, the other eye will usually start losing vision within the period of several weeks to months. Although, most of the people are severely impaired by 3 or 4 months, the rate of progression may vary from days to years.

Over time, the optic nerve atrophy will result in worsening of vision in both eyes in the form of loss of visual acuity and color blindness.[6]

Multiple sclerosis and other demyelinating diseases cause a rather uncommon comorbidity in patients with Leber's hereditary optic neuropathy. Sometimes clinical features in Leber's hereditary optic neuropathy are not distinguishable phenotypically from multiple sclerosis, and mutations in LHON are considered as a risk factor in the pathophysiology of multiple sclerosis.[7]

Currently, mitochondrial DNA analysis is recommended in all young male patients diagnosed with multiple sclerosis who have initial neuro-ophthalmological manifestations, peripapillary microangiopathy, bilateral vision loss and a positive family history.[8]

Although the mtDNA mutation is necessary for the development of LHON but it is not sufficient. Other, still poorly defined factors including genetic or environmental may be implicated. Family history is not clear in about 40% patients affected with LHON. [9]

Smoking is established as the most common factor as well, which demonstrates the strong influence of environmental risk factors. [10]

The prognosis for vision recovery depends on the type of mutation.[3, 11]

4. Conclusion

Whenever young males develop bilateral sequential vision loss with a positive family history, the diagnosis of Leber's hereditary optic neuropathy should be suspected. Ophthalmological examination may reveal signs suggestive of Leber's hereditary optic neuropathy

In Leber's hereditary optic neuropathy patients the most frequent mutation of mDNA is hhe mutation at the 11778 DNA locus. However, these mutations are not sufficient alone to cause loss of sight. Secondary factors likely contributed in conversion of LHON to the symptomatic disease.

In contrast, patients with clinically isolated acute optic neuritis in which MRI shows multifocal white matter lesions, there may be minor and no characteristic alterations in a few cases with LHON. In our case increased optic nerve signal and thinning of its fibers were evident. This data suggest the presence of optic nerve swelling which can resolve subsequently. Utilizing STIR sequences, Kermode et al have recently reported the similar findings to our own results in a small number of LHON patient. Our results show same abnormality of the optic nerve found in the study by Kermode et al. In conditions, using classical FAST and spin echo sequences, MRI does not show typical changes in LHON patients. However, it can be considered as an usefull tool together with family data to differentiate optic neuritis.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of ethical approval

For conducting this study approval from the ethical committee, Fatima Jinnah Medical university/Sir Ganga Ram hospital-Lahore has been taken.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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