Original Research Paper

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LEBER HEREDITARY OPTIC NEUROPATHY IN A 12-YEAR-OLD FEMALE PRESENTING WITH BILATERAL VISION LOSS: A RARE CASE REPORT

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ABSTRACT **Background:** Leber Hereditary Optic Neuropathy (LHON) is a mitochondrial genetic disorder typically affecting young males, leading to acute or subacute painless central vision loss. Female and pediatric presentations are uncommon. **Case Presentation:** A 12-year-old female presented with progressive, painless diminution of vision in both eyes associated with colour vision deficiency. Fundus examination revealed bilateral hyperemic optic disc with peripapillary telangiectatic vessels. Optical coherence tomography (OCT) demonstrated retinal nerve fibre layer (RNFL) thickening in the acute phase. MRI brain and orbit were normal. Based on clinical findings, a diagnosis of LHON was made. **Conclusion:** LHON should be considered in paediatric patients with bilateral painless vision loss, even in females. Early diagnosis aids in counselling and genetic evaluation.

KEYWORDS : LHON, Mitochondrial Disorder, Optic Neuropathy, Paediatric, OCT

INTRODUCTION

Leber Hereditary Optic Neuropathy (LHON) is a maternally inherited mitochondrial disorder characterized by selective degeneration of retinal ganglion cells, leading to optic nerve atrophy. It most commonly affects young adult males, with peak onset in the second to third decade. Paediatric and female cases are rare, making diagnosis challenging. The disease is associated with point mutations in mitochondrial DNA, most commonly at positions 11778, 14484, and 3460.

CASE REPORT

A 12-year-old female presented to the ophthalmology outpatient department with complaints of gradual, painless blurring of vision in both eyes for 2 weeks. The visual loss initially involved one eye and subsequently progressed to the other eye.

There was no history of trauma, systemic illness, drug intake, or similar complaints in the family.

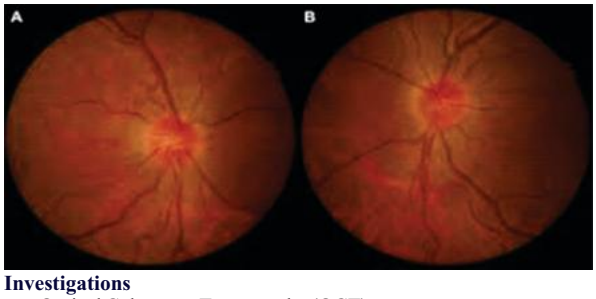
TABLE 1

OCULAR EXAMINATION	RIGHT EYE	LEFT EYE
VISUAL ACUITY	6/18 -> 6/9p	6/24->6/18p
AUTO REFRACTION	-2.00DC at 180 degree -> 6/18	-2.00DC at 180 degree -> 6/18
NEAR VISION	N6	N6
COLOUR VISION (Ishihara chart)	8/17 plates	8/17 plates
Amsler	Normal	Normal
IOP ON GAT (in straight gaze)	10 mmHG at 10:30am	12 mmHG AT 10:30am
ROPLAS	Negative	Negative
EXTRAOCULAR MOVEMENTS	Free, full and painless in all gazes	Free, full and painless in all gazes

TABLE 2

SLIT LAMP EXAMINATION	RIGHT EYE	LEFT EYE
EYELIDS	Normal	Normal
CONJUNCTIVA	Normal	Normal
CORNEA	Clear	Clear
ANTERIOR CHAMBER	Normal depth	Normal depth
IRIS	Normal in shape, colour and pattern	Normal in shape, colour and pattern
PUPIL No RAPD	3mm round, regular and reacting to light	3mm round, regular and reacting to light
LENS	Clear	Clear
FUNDUS (Dilated)	Media-clear Optic disc : Hyperemic, Pseudo edematous, Peripapillary telangiectasia C:DR – cannot be assessed A:V- Tortuosity Macula FR: Present Periphery-Normal	Media-clear Optic disc : Hyperemic, Pseudo edematous, Peripapillary telangiectasia C:DR – cannot be assessed A:V- 2:3, Tortuosity Macula FR: Present Periphery-Normal

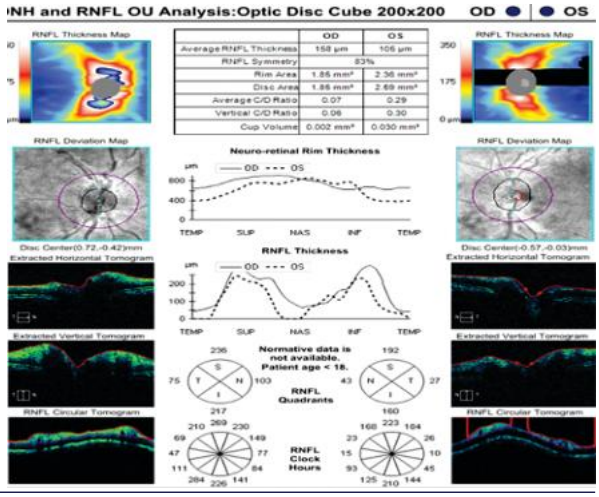
FIGURE 1



Investigations

- Optical Coherence Tomography (OCT): Increased RNFL thickness in the acute stage
- MRI Brain and Orbit: Normal study
- Blood investigations: Within normal limits
- Genetic testing: Not performed due to financial constraint.

FIGURE 2



Diagnosis
Based on clinical findings, a diagnosis of Leber Hereditary Optic Neuropathy (LHON) was made.

DISCUSSION
LHON is caused by mitochondrial DNA mutations affecting complex I of the electron transport chain, resulting in impaired ATP production and increased oxidative stress. This leads to selective damage to retinal ganglion cells.

The typical presentation involves:
- Painless, subacute central vision loss
- Sequential involvement of both eyes
- Central or centrocecal scotoma
- Colour vision defects

Fundus findings in early stages include:
- Hyperemic disc
- Peripapillary telangiectasia
- Pseudoedema without leakage

OCT initially shows RNFL thickening, followed by thinning in later stages due to optic atrophy.

Although LHON predominantly affects males, female cases can occur due to incomplete penetrance and environmental triggers.

Differential Diagnosis
- Optic neuritis
- Nutritional optic neuropathy
- Toxic optic neuropathy
- Compressive optic neuropathy

Management
There is no definitive cure. Management includes:
- Idebenone therapy (if available)
- Avoidance of risk factors (smoking, alcohol)
- Genetic counseling
- Visual rehabilitation

Prognosis varies depending on mutation type, with some spontaneous recovery reported.

CONCLUSION
LHON, though rare in pediatric females, should be considered in cases of bilateral painless vision loss with characteristic fundus findings. Early recognition is crucial for counseling, management, and genetic evaluation.

DECLARATION
- Patient consent: Obtained
- Conflict of interest: None
- Funding: None

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