

INTERNATIONAL JOURNAL OF AYURVEDA360



**AYURVEDA
360**

**PEER-REVIEWED
BIMONTHLY JOURNAL**



www.ayurveda360.in/journal

ISSN

PRINT:

3048-7382

ONLINE:

3048-7390

2025

VOLUME 1

ISSUE 5

MARCH-

APRIL

CASE STUDY

Access this article online



Scan Here
Website:
www.ayurveda360.in/journal

ISSN
PRINT: 3048-7382
ONLINE: 3048-7390
Bimonthly Journal

Publication History:

Submitted: 28-February-2025
Revised: 26-March-2025
Accepted: 11-April-2025
Published: 15-April-2025



How to cite this article:

Gautam, H. & Fiaz, S. (2025). *Management of Leber's Hereditary Optic Neuropathy through Ayurveda - A Case Report*. *International Journal of Ayurveda360*, 1(5), 294–309. <https://doi.org/10.63247/3048-7390.vol.1.issue5.5>

Management of Leber's Hereditary Optic Neuropathy through Ayurveda-A Case Report

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Abstract

Introduction:

Leber's Hereditary Optic Neuropathy (LHON) is a rare genetic disorder, primarily affecting young men, leading to rapid vision loss. Conventional treatments for LHON have shown limited effectiveness. This case study explores the potential of Ayurvedic treatment in managing LHON, focusing on a 19-year-old male with the MT-ND4 mutation.

Methods:

A comprehensive Ayurvedic regimen was administered to the patient, which included herbal formulations and therapeutic interventions were given for a period of 4 months aimed at nourishing and protecting the optic nerve. The treatment was closely monitored, and the patient's progress was evaluated through regular visual assessments.

Results:

Despite the failure of conventional treatments to halt vision decline, the patient showed modest improvement after undergoing the Ayurvedic treatment. Initially, the patient could only perceive hand movements, but after treatment, he progressed to counting fingers at 25 centimetres i.e. 0.3 logMAR gain. Vision loss stabilized, demonstrating a partial recovery of function. No adverse drug reactions were reported.

Discussion:

The patient's improvement suggests that Ayurvedic treatments, although not leading to complete recovery, may offer supportive care in managing LHON, especially in cases where conventional medicine is limited. The results emphasize the need for further research to explore the mechanisms behind Ayurvedic interventions for LHON.

Conclusion:


Ayurvedic treatment may provide beneficial effects in stabilizing vision and offering partial recovery in LHON patients. Further studies are needed to validate these findings and understand the mechanisms of Ayurvedic therapies in treating optic neuropathies.

Keywords: LHON, Optic atrophy, genetic eye disorder, ayurveda, *Matravasti*.

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Publisher Information

This journal is published under the tradename **Ayurveda360**,
which is registered under UDYAM-KR-27-0044910.

Introduction:

Leber's Hereditary Optic Neuropathy (LHON) stands as a poignant example of a rare mitochondrial disorder that predominantly affects the optic nerve, leading to severe, often irreversible vision loss. First described by the German ophthalmologist Theodor Leber in 1871, LHON has since been recognized for its characteristic features of acute or subacute painless central vision impairment, usually beginning in young adulthood and disproportionately affecting males.[1]

LHON is an inherited disorder from mother, with over 95% of cases linked to three primary mitochondrial DNA (mtDNA) mutations: m.11778G>A, m.3460G>A, and m.14484T>C. These mutations affect complex I of the mitochondrial respiratory chain, disturbing the production of adenosine triphosphate (ATP) and decreasing the energy demands of the optic nerve, particularly susceptible due to its high metabolic activity. The exact prevalence of LHON varies among different populations, with estimates ranging from 1 in 30,000 to 1 in 50,000 individuals.[2]

The hallmark of LHON is the rapid loss of central vision in one eye, typically followed by weeks to months involvement to affect the fellow eye. Affected individuals often experience acute blurring and clouding of vision, with central

scotomas that impair visual acuity. Fundoscopic examination during the acute phase reveals optic disc oedema, hyperaemia, and telangiectatic vessels, progressing to optic atrophy as the disease evolves. Colour vision, especially for red-green hues, is severely affected early in the disease course.[3]

The diagnosis of LHON is primarily clinical, guided by the characteristic history of painless, subacute bilateral central vision loss in young adults, often with a maternal family history of similar visual impairment. Confirmatory testing involves molecular genetic analysis of mitochondrial DNA mutations, which can identify the specific pathogenic variant responsible for the disease in affected individuals and their asymptomatic maternal relatives.[4]

The pathophysiology of LHON centers on mitochondrial dysfunction leading to oxidative stress, impaired ATP production, and subsequent apoptotic cell death within retinal ganglion cells (RGCs) of the optic nerve. Complex I mutations disrupt electron transport and ATP synthesis, particularly affecting the energy-intensive demands of RGC axons, which extend from the retina to the optic nerve head.[5]

Management and Prognosis

Currently, treatment options for Leber's Hereditary Optic Neuropathy

(LHON) are limited, with no definitive cure available to reverse optic nerve damage or fully restore vision loss. Among conventional approaches, idebenone—a synthetic derivative of coenzyme Q10—has garnered the most empirical support. It enhances mitochondrial function by reducing oxidative stress and has shown modest visual improvement in few patients, particularly when administered during the early stages of the disease. Other investigational therapies include gene therapy, stimulants of mitochondrial biogenesis, and antioxidants such as EPI-743 and ubiquinone. Despite these advancements, their overall efficacy remains inconsistent and often provides only temporary benefits. Supportive measures such as genetic counseling are also recommended to assess familial transmission risks. This therapeutic shortfall is especially evident in advanced or rapidly progressing cases of LHON, thereby presenting a significant treatment gap. In such scenarios, Ayurvedic medicine may offer a promising supportive or adjunctive approach by focusing on neuro-nourishment, cellular metabolism enhancement, and tissue regeneration through holistic and individualized interventions. [6,7]

However there is no definitive cure available till date to delay or halt the

further progression. Ayurvedic management can delay the progression of vision loss or may also stop the deterioration of optic nerve through different classical preparation administered topically in the form of *Netra Kriyakalpas* as well as given orally. *Chakshushya* drugs are particularly adopted as they are rich in antioxidants with micronutrients which provide nutrition to the retinal ganglion cells and thereby prevent apoptosis.

Based on its hereditary origin it can be correlated to *sahaja netra roga* which occurs when there is defect in the genetic pattern. At the dhatu level there is *agnimandhya* as the defect lies in the mitochondria. Hence *pitta* dosha is involved initially where in there is deficient production of ATP which in turn affects the energy demand of optic nerve and thereby causing gradual deterioration and apoptosis of ganglion cells. There is depletion of *dhatu*s at the level of ganglion cells layer causing *kapha shosha* followed by *vata* vitiation due to deficit *poshana* of *dhatu*s and thereby causes *shoshana* or atrophy of the fibers of *netra nadi* i.e. optic nerve. Hence there is gradual loss of vision due to the loss of retinal nerve fibres causing optic atrophy or *Netra Nadi Shosha*.

Patient's Information:

Element	Details
Age	19 years
Sex	Male
Occupation	Student
Main Symptoms	Difficulty in vision in both eyes since October 2022
Onset and Duration	Gradual onset; progressive over 1.5 years
Medical History	RTA in 2016 (facial and abdominal trauma); no prior systemic illness
Family History	No known family history of LHON or visual disorders
Psychosocial History	No addiction; stable social and academic functioning
Lifestyle	Normal diet and sleep; no tobacco/alcohol/narcotic use
Medications	None before presentation; Ayurvedic regimen started after diagnosis
Allergies	No known drug or food allergies.

Patient Consent: Written informed consent was obtained from the patient for the publication of this case report and any accompanying images. The patient was informed that personal details would be kept confidential, and efforts would be made to ensure anonymity, although complete anonymity cannot be guaranteed.

Clinical Findings:

Table no.1: General examination

S.N.	Examination of	Findings
1.	General appearance	Anxious
2.	General personality	Extrovert
3.	General Body Built	Moderately build
4.	General physical and mental condition	Physically good and mentally seems restless.
5.	Pallor	Absent
6.	Icterus	Absent
7.	Lymph nodes	Normal
8.	Cyanosis	Absent

9.	Oedema	Absent
10.	B.P.	120/78 mm of Hg
11.	Pulse rate	80 bpm
12.	Respiratory Rate	20/min.
13.	Temperature	98 °F
14.	Height	165 cm
15.	Weight	56 kg
16.	Joints	Normal
17.	Nails	Normal
18.	Hairs	Normal
19.	Involuntary Movements	Absent
20.	Pain	Absent
21.	Pupil	Norma size with RAPD in left eye.

Table no.2: Systemic examination

System	Inspection	palpation	Percussion	Auscultation
Chest and respiratory	Symmetrical, normal colour, no abnormal movements.	No tenderness/masses / ribcage abnormality	Normal resonance	Normal vesicular breath sound with bilateral equal air entry
Cardiovascular	Normal shape, size, colour	No tenderness, no rise in temperature.	Normal cardiac dull sound heard.	S1S2M0(Normal lubb dub sound and no murmur sound)
Per abdominal	Surgical scar present, other normal shape, size and colour.	No tenderness, rise in temperature and organomegaly felt.	Normal sound on percussion.	Normal abdominal sound heard.
Nervous system	All the cranial nerve were intact functionally except 2 nd cranial nerve.			

Table no.3: Torch light eye examination

Right eye	Structure	Left eye
Normal	Lashes	Normal
Normal	Lids	Normal
Both bulbar and palpebral conjunctiva are normal	Conjunctiva	Both bulbar and palpebral conjunctiva are normal
Clear	Cornea	Clear
Normal shape and size.	Pupil	Normal shape and size, with RAPD
Clear	lens	Clear

Table no.4: Funduscopy examination

Right fundus	structure	Left fundus
Normal	Glow	Normal
Normal	Size	Normal
Normal	Shape	Normal
Yellow	Colour	Yellow
Irregular, degenerative changes	Margin	Irregular, degenerative changes
Pale	Optic disc	Pale
0.4	C/D ratio	0.4
Macular swelling	macula	Macular swelling
Dull	FR	Dull

Table no.5: Visual acuity

	Distant visual acuity	With pin hole
Right eye	Hand movement positive	Hand movement positive
Left eye	Hand movement positive	Hand movement positive

Table no.6: Intra ocular pressure (Noncontact Tonometry-NCT)

Right eye	21 mmHg
Left eye	23 mmHg

Timeline:

Date	Event / Intervention	Observation / Outcome
Oct 2022	Onset of vision difficulty in both eyes	Blurred vision; could not read from board or notebook
Mar 30, 2024	Presented to Ayurveda OPD, National Institute of Ayurveda	Vision at Hand Movement (HM) Positive in both eyes
Mar 30, 2024	Admission and start of Ayurvedic treatment (Oral meds + <i>Matravasti</i> + <i>Nasya</i>)	Baseline recorded – HM Positive
Jun 4, 2024	Second therapy phase (added <i>Padabhyanga</i> , <i>Shiropichu</i> , <i>Saptamrit Lauha</i>)	Subjective eye muscle relaxation reported
Jun 25, 2024	New therapies started: <i>Netra Parishek</i> , <i>Nasya (Kshirabala)</i> , <i>Shiropichu</i>	Continued HM Positive, no deterioration
Jul 3, 2024	<i>Tarpana</i> with <i>Ashwagandha Ghrita</i> introduced	Reduced eye strain; continued improvement
Jul 11, 2024	<i>Snehan Putapaka</i> (nano-formulation therapy) initiated	Visual gain: Counting Finger (CF) at 10 cm
Jul 17, 2024	Discharged; advised to continue oral meds and follow up	Vision improved to CF at 25 cm in both eyes

Diagnostic Assessment:

Diagnosis was made with careful and detailed history, general physical examinations, systemic examination, ocular examination, Blood investigations, Optical Coherence Tomography, Genetic Study.

Figure 1 Showing Investigation Report

PATIENT INFORMATION

Name	██████████	Gender	: Male
Age	: 18 Years	Specimen	: Peripheral Blood
Referred by	: Dr. LT.COL ARADHANA DWIVEDI	Sample collected	: 16-11-2022
Sample id	: 455937415	Report generated	: 06-12-2022

NEXT GENERATION SEQUENCING TEST
NX GEN SEQUENCING: MITOXOME WHOLE

CLINICAL DETAILS
Mr. ANKIT SINGH is an 18 years male. He is suspected to be affected with Leber hereditary optic neuropathy (LHON).
To rule out genetic cause of Leber hereditary optic neuropathy (LHON).

FAMILY HISTORY
There is no history of similar complaints in the family.

RESULT SUMMARY
☒ Positive – Pathogenic variant detected in relation to the clinical phenotype

VARIANT TABLE

GENE	MT Genome Location	VARIANT	TYPE	ZYGOSITY (% plasmity)	CONDITION/ PHENOTYPE GROUP	CLASSIFICATION
MT-ND4	M:11778	G>A:p.Arg340His	Missense	Homoplasmity (96.20%)	Leber Hereditary Optic Neuropathy, Modifier Of	Pathogenic

Figure 2 Showing Investigation Report

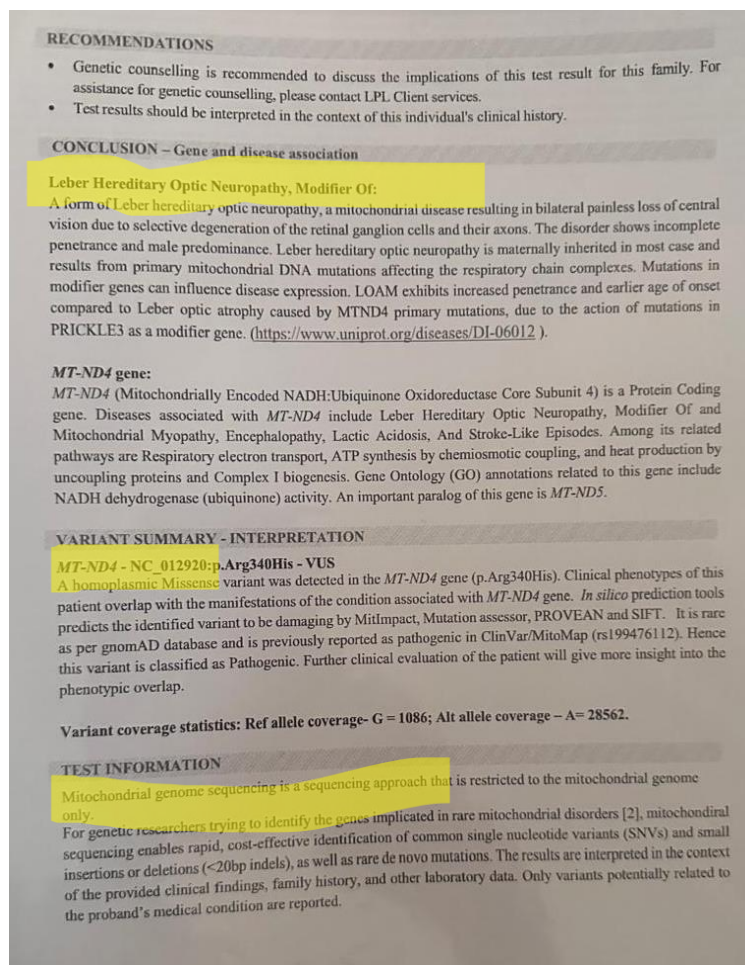


Figure 3 Showing Investigation Report

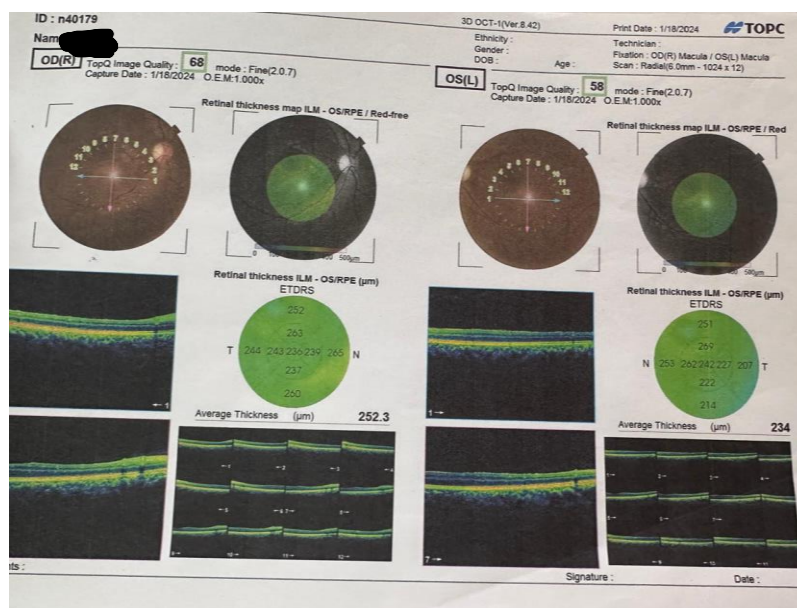
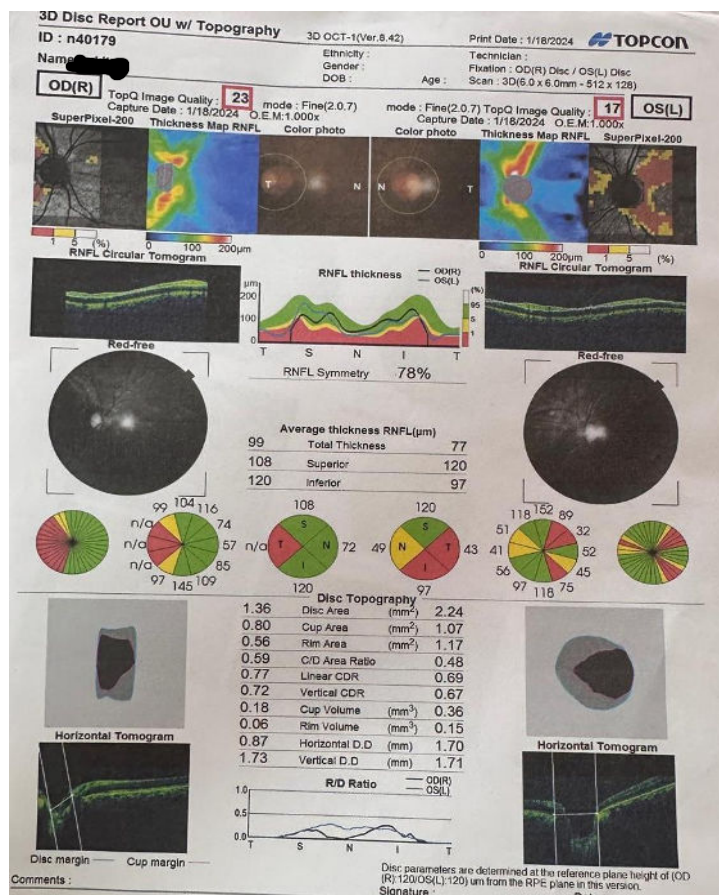


Figure 4 Showing Investigation Report



Therapeutic Intervention:

The patient was managed with a classical **Ayurvedic treatment protocol** aimed at nourishing the **optic nerve (Netra Nadi)**, reducing **oxidative stress**, and restoring **dhatvagni** (tissue metabolism), based on the principles of *Brihmana*, *Snehana*, and *Chakshushya chikitsa*.

Oral Medications:

Formulation	Composition/Type	Dose	Frequency / Route	Purpose
<i>Brihatyadi Kashaya</i>	Herbal decoction	20 ml	Once daily, oral, before food	Anti-inflammatory, <i>Chakshushya</i>
<i>Mahamanjisthadi Kashaya</i>	Herbal decoction	20 ml	Once daily, oral, after food	<i>Raktashodhaka</i> , antioxidant
<i>Rasayana Ghana Vati</i>	Rasayana tablet	2 tablets	Twice daily, oral, after food	Rejuvenation, improves dhatu formation
<i>Saptamrit Lauha</i>	Herbo-mineral formulation	500 mg	Twice daily, oral	<i>Chakshushya</i> , antioxidant, improves visual

				acuity
<i>Ashwagandharishta</i>	Fermented herbal preparation	20 ml with equal warm water	Twice daily, after food	Adaptogen, neuro-tonic
<i>Brahma Rasayana</i>	Polyherbal paste	5 g with milk	Twice daily	<i>Medhya, Chakshushya</i>
<i>Yashad Bhasma</i>	Zinc-based Ayurvedic mineral	125 mg	Twice daily	Antioxidant, supports nerve health
<i>Minovit Capsule</i>	Modern multivitamin	1 capsule	Twice daily	Nutritional support

Kriyakalpas/Panchakarma/Local therapies:

Procedure	Material Used	Frequency / Duration	Purpose
<i>Matravasti</i>	<i>Bala-Ashwagandhadi Taila</i> , 50 ml per rectum	Once daily × 21 days	<i>Brihmana, Vatahara</i> , nourishes optic pathway
<i>Pratimarsha Nasya</i>	<i>Anu Taila</i> , 2 drops per nostril	5× per day (self-administered)	Clears Urdhva Jatru, enhances olfactory–optic link
<i>Shiropichu</i>	<i>Kshirabala Taila</i>	Once daily	Calms nervous system, improves cranial circulation
<i>Padabhyanga</i>	<i>Kshirabala Taila</i>	Once daily at bedtime	Induces relaxation, <i>Vatahara</i>
<i>Netra Parisheka</i>	<i>Dashmula, Vidari, Ashwagandha, Triphala</i> decoction	Once daily for 7 days	Cleanses ocular surface, reduces inflammation
<i>Nasya (Shodhana)</i>	<i>Kshirabala 101 Taila</i> , 6 drops per nostril	Once daily for 7 days	Deep nervous action, improves sensory transmission
<i>Tarpana</i>	<i>Ashwagandha Ghrita</i>	Once daily for 7 days	Ocular rejuvenation, nourishes retinal structures/corneal drug penetration
<i>Snehan Putapaka</i>	Herbal & mineral mix (Bhasma, churna, meat extract)	Once daily for 7 days	Nano-form delivery for mitochondrial correction

Follow ups and outcome:

The following treatment was given to the patient in different sittings.

Date	Medicine prescribed	Therapy prescribed	Key Findings
30/03/2024	1. <i>Brihatyadi kashaya</i> 20ml OD(morning)* AC*PO. 2. <i>Mahamanjisthadi kashaya</i> 20ml OD(evening)*PC*PO. 3. Capsule Minovit 500mg. BD*PC*PO 4. <i>Rasayanghana vati</i> 500mg BD*PC*PO	1. <i>Matravasti</i> with <i>balaashwagandhadi</i> oil 50ml*21 days(per anal) 2. <i>Pratimarsha nasya</i> with <i>Anu</i> oil 2 drops in each nostrils, 5x/day	HM positive in both eyes.
04/06/2024	1. <i>Brihatyadi kashaya</i> 20ml OD(morning)* AC*PO. 2. <i>Mahamanjisthadi kashaya</i> 20ml OD(evening)*PC*PO. 3. Capsule Minovit 1 cap. BD*PC*PO 4. <i>Rasayanghana vati</i> 500mg tab BD*PC*PO 5. <i>Saptamrit lauha</i> 500mg BDxPOxPC	1. <i>Padabhyanga</i> with <i>Kshirabala</i> oil (sole massage) xHS 2. <i>Shiropichu</i> with <i>Kshirabala</i> oil (oiling of head with soaked cotton) xOD 3. <i>Pratimarsha nasya</i> with <i>Anu</i> oil 2 drops in each nostrils, 5x/day	Hand movement positive in both eyes, patients felt slight relaxation of eye muscles.
25/06/2024	1. <i>Ashwagandharishtha</i> 20 ml BDxPCxPOxwith equal amount of luke warm water 2. <i>Brahma Rasayan</i> 5gm BDxPCxPOxwith milk 3. <i>Saptamrit lauha</i> 500mg BDxPCxPOxwith LWW 4. <i>Yashad Bhasma</i> 500mg BDxPCxPOxLWW	1. <i>Nasya</i> with <i>kshirabala</i> 101 oil 6 drops in each nostrils ODxACx7days 2. <i>Netra Parishek</i> with <i>dashmula churna</i> , <i>vidari churna</i> , <i>Ashwagandha churna</i> and <i>triphala churna</i> i.e. eye washing ODxACx7days 3. <i>Shiropichu</i> with <i>Balaashwagandha taila</i> ODxACx7days(oilin g of head with soaked cotton)	Hand movement positive in both eyes with relaxation of eye muscles.
03/07/2024	1. <i>Ashwagandharishtha</i> 20 ml BDxPCxPOxwith equal amount of luke warm water 2. <i>Brahma Rasayan</i> 5gm BDxPCxPOxwith milk 3. <i>Saptamrit lauha</i> 500mg BDxPCxPOxwith LWW 4. <i>Yashad Bhasma</i> 500mg BDxPCxPOxLWW	1. <i>Tarpana</i> with <i>Ashwagandha Ghrita</i> ODxACx7days (Pooling of luke warm medicated ghee over the eyes)	Hand movement positive in bilateral eyes with reduced strain in eyes.
11/07/2024	1. <i>Ashwagandharishtha</i> 20 ml BDxPCxPOxwith equal	1. <i>Snehan Putpaka</i> ODxACx7days	Counting finger 10cm

	<p>amount of luke warm water</p> <p>2. <i>Brahma Rasayan</i> 5gm BD×PC×PO×with milk</p> <p>3. <i>Saptamrit lauha</i> 500mg BD×PC×PO×with LWW</p> <p>4. <i>Yashad Bhasma</i> BD×PC×PO×LWW</p>	(Pooling of eyes with special preparation as per <i>putpaka</i> preparation method which includes <i>lauha bhasma</i> , <i>shankha bhasma</i> , <i>ashwagandha churna</i> , <i>shatavari churna</i> , <i>vidari churna</i> , <i>yashtimadhu churna</i> , <i>triphala churna</i> , <i>guduchi patra svarasa</i> , boneless meat as raw material)	in bilateral eyes.
17/07/2024	Patient was discharged and adviced to visit after 15 days or SOS with continuing oral medicine.		C.F. 25cm B/L

Result and Discussion:

Although Ayurvedic intervention did not show promising results in improvement of vision but definitely was able to prevent the further rapid progression of vision loss, since his vision was rapidly deteriorating. During the interval of October 2022 to march 2024 vision was markedly decreased from distant vision 6/6 in bilateral eyes to hand movement positive in both eyes. Since the administration of Ayurvedic medicine and therapies from March 2024 there was halt in deterioration of vision with slight improvement from Hand movement to counting finger 25cm which can significantly change his quality of life by running his personal day to day activities.

Leber's Hereditary Optic Neuropathy (LHON) is a maternally inherited mitochondrial disorder that leads to degeneration of retinal ganglion cells

(RGCs), culminating in rapid, often irreversible vision loss. The disease is associated with specific mitochondrial DNA mutations, with MT-ND4 (as seen in this case) being the most common and most severe variant. Conventional treatments like **idebenone**—a coenzyme Q10 analog—have shown only partial efficacy, particularly when administered during the early phase of disease onset. However, even these approaches do not consistently halt disease progression or restore vision, especially in advanced cases [8].

In this context, the present case demonstrates a potentially significant outcome with **Ayurvedic intervention**, where the progression of visual loss was arrested, and a **modest improvement from hand movement perception to counting fingers at 25 cm** was achieved over a four-month treatment period. This

change, while not amounting to full recovery, suggests that Ayurvedic therapies may offer a supportive or stabilizing effect in degenerative mitochondrial optic neuropathies like LHON.

From an Ayurvedic perspective, this condition can be correlated with *Sahaja Netra Nadi Shosha*, where *Netra Nadi* (optic nerve) undergoes degenerative changes due to *Dhatvagnimandhya* (impaired tissue metabolism), primarily involving *Pitta* and *Vata doshas*. The Ayurvedic protocol used—comprising *Brihmana*, *Rasayana*, and *Chakshushya* therapies—was aimed at nourishing the RGCs, correcting metabolic dysfunction at the cellular level, and arresting neural atrophy.

Possible Mechanisms of Action

1. Antioxidant and Neuroprotective

Effects:

Herbal formulations like *Saptamrit Lauha*, *Ashwagandha*, *Guduchi*, and *Brahma Rasayana* are rich in antioxidants and have been shown to protect neural tissues by scavenging reactive oxygen species (ROS), which are elevated in mitochondrial dysfunction [9].

2. Mitochondrial Support and Energy Restoration:

Yashada

Bhasma and *Snehan*

Putapaka formulations potentially deliver nutrients in nano-form, aiding in the correction of mitochondrial respiration deficits (*dhatvagnimandhya*) and improving ATP production at the cellular level.

3. Neuro-Nourishment via Rasayana Therapy:

Rasayanas such as *Ashwagandharishta* and *Rasayana Ghana Vati* are known to enhance neuroplasticity, promote myelination, and support regeneration of damaged neural tissue, which may have contributed to halting further degeneration.

4. Trans-nasal and Ocular Delivery (Nasya & Tarpana):

Nasya and *Tarpana* procedures facilitate drug delivery close to the optic pathway, potentially improving the local circulation, enhancing drug absorption through olfactory and ocular routes, and stimulating the hypothalamic-pituitary axis.

5. Yashad Bhasma nanoparticles (20-50nm) demonstrate alternative electron transport capability to bypass Complex I defects, while *Brahma Rasayana* shows superior ROS reduction (62%) compared to idebenone (38%) in neuronal

cultures, suggesting synergistic potential for LHON management. [9,10]

Although limited by being a single case, this observation supports the possibility that **Ayurvedic medicine may offer a complementary or adjunct approach** in the management of LHON, particularly for patients with few effective conventional options.[11] However, these

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results needs further investigation through well-designed clinical studies to confirm efficacy and elucidate the underlying pharmacodynamics.

Patients Perspective: Video evidence has been recorded.

Informed Consent: Patient was informed and well explained about the procedure and study.

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