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An Ayurvedic Visual Rehabilitation In Lebers Congenital Amaurosis - Prospective Case Study

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ABSTRACT

Introduction: This is the case report of child who was diagnosed with LCA reported with complaints of large-amplitude, slow-frequency, roving nystagmus, frequent tendency to press on his eyes, enopthalmos, with completely normal ophthalmoscopic examination with normal appearing optic nerve and retina and has a non recordable ERG; considering this condition under the lines of *Vataja Nanatmaja Vyadhi* treatment was structured to render effective visual rehabilitation which showed marked results. **Methods:** 4 years old male child who was diagnosed with lebers congenital amaurosis was brought by his parents to Eye OPD, GAMC, Bengaluru. The presentation of this case includes bibliographic review of the subject, presentation of a clinical case and description of the importance of Ayurvedic prespective of handling of these patients. **Results:** The child showed improvement in fixing for light and also nystagmus showed marked improvement and there were considerable behavioural changes observed. **Discussion:** It is important to deepen the environment of the disease to know the possible implications in Ayurvedic management, recognize the magnitude of visual disability that our patient presents for the establishment of the treatment plan and provide an integral care of excellence in an interdisciplinary way in favor of visual rehabilitation of our patients and also help to restore quality of life with no potential risks of side effects.

Key words: *Leber's Congenital Amaurosis (LCA), Congenital Blindness, Janmabala Pravrutta Vyadhi.*

INTRODUCTION

Jataandya[1] is that which comes under the gamet of *Janmabala Pravrutta Vyadhi* that which is inherited from parents and is understood as *Shukrashonita Dusti* which can be considered while understanding lebers congenital amaurosis; Leber congenital amaurosis is a disorder of the photoreceptors and the retinal pigment epithelium in which photoreceptor function is extinguished. Leber congenital amaurosis,[2] a group of autosomal recessive retinal dystrophies, is the most common genetic cause of congenital visual impairment (Weleber et.al, 2006). Eight different gene mutations cause one third of cases. The common feature is disturbance of the retinal pigmentary epithelium; The clinical characteristics of Leber congenital amaurosis are moderate to severe visual impairment at or within a few months of birth, nystagmus, and sluggish pupillary responses. Additional features include symmetrical mid facial hypoplasia with enophthalmos and hypermetropic refractive errors. Although substantial variation between families exists, the phenotype is relatively constant within families. Ophthalmoscopy of the retina shows no abnormality in infancy and early childhood, but with time, progressive retinal stippling and pallor of the optic disk appear. Visual acuity is rarely better than 20/400.

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considering it to be a *Janmabala Pravrutta Vyadhi* and understanding the prognosis of the condition to be poor due to infantile onset progressive retinal degeneration which is not reversible and also considering the loss of structural and functional integrity of retina and ruling out the secondary systemic signs this case is been treated as per the treatment protocol of *Vataja Nanatmaja Timira*[3] and the baseline and follow-up improvement in his condition is been documented.

OBJECTIVES

1. To understand Leber congenital amaurosis in terms of Ayurvedic perspective for its management.
2. To study the effect of *Vatasya Upakrama* in the management of LCA.

MATERIALS AND METHODS

Assessment criteria

Subjective criteria

- Pupillary reaction
- Nistagmus / vague eye movements.
- Oculo-digital signs.
- Enopthalmos

CASE REPORT

Basic information of the patient

Age: 4 years

Sex: male

Religion: hindu

Socioeconomic status: middle class.

Father is currently working as a autodriver, mother has studied 10th standard, and she is house wife.

Chief complaints

Informant: Parents

- H/o rapid eye ball movements noticed at the age of 3months
- Not responding to verbal commands
- Unable to fix the eye (no eye contact)
- Deviation of both the eyes alternatively
- Frequent pressing and rubbing both the eyes since 6months of age.

Birth history: LSCS - term gestation

Family history: first degree consanguinity, mother is Rh Incompatable

History of present illnesses

The mother reports that the child was a term baby with 2,700gm born with no reported complications during birth, he has attained all motor milestones as per age with proper mental and social responding; at the age of 3months mother noticed that the child had rapid eye movements, and was unable to fix the eye to light response and would gaze at a different point and would frequently rub and press the eyes for the same above complaints they consulted Narayana Nethralaya where it was diagnosed to be (retinal dystrophy/ lebers congenital amaurosis) contributing to their consanguinity 1st degree marriage and the parents were advised to get the child for a regular follow-up and MRI scan was done at the age of 9 months which was normal, BE OCT showed signs of retinal dystrophy based on further clinical evaluation he was diagnosed with lebers congenital amaurosis and was advised +7d refraction for hypermetropia which is been changed to +4d since 1year and is been advised to encourage the child for visual rehabilitation and they approached us for further management.

History of past illness

No history of seizures / any other complaints after birth

No H/O neurodevelopmental delay, mental disability and systemic abnormalities.

Treatment history

The child has been advised with visual rehabilitation

Family history

1st degree consanguineous marriage

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Birth history

Natal: term (36 weeks) LSCS (due to prev LSCS) was done. Birth weight was 2.7 kg.

History of immunization

Proper for age

Personal history

Aharaja

Diet is dominant in *Madhura Rasa* (sweet diet).

Personal history showed that the child did not attend any school, with daily personal hygiene, and oral hygiene once a day.

Viharaja

Nature of activity - child is very talkative with good memory and good social responses was reported with frequent falls due to poor vision. Sleep was disturbed (2-3 h/day, 6-7 h/night).

Examination

Vitals were normal. Cardiovascular system, respiratory system and per abdomen examinations had shown no deformity. *Prakṛti* (constitution) was *Vatadhikapitta*.

Central nervous system examination

No signs of Mental retardation and other neurological disturbances.

Ocular examination

	Right eye	Left eye
Eye ball	Enopthalmos - present ALT ESOMETROPIA +	Enopthalmos - present ALT ESOMETROPIA +
Cornea	Keratoconus +	Keratoconus +
Pupillary reflex	Severe photophobia + Does not fix to light ? Menace Reflex Absent papillary reflexes	Severe photophobia + Does not fix to light ? Menace Reflex Absent papillary reflexes
Nystagmus	Roving in all positions of gaze	Roving in all positions of gaze
Anterior chamber	Shallow - small eye High hypermetropia	Shallow - small eye High hypermetropia
Franceschetti Oculodigital Sign	Present - frequent eye poking +	Present - frequent eye poking +
Lens	Clear	Clear
Fundus	Media - clear Disc - 0.3 A:V - 2:3 Foveal reflex- Blunted with absence of Peri and Para Foveal Reflxes ONH- Mild Pallor in Temporal Rim NRR - healthy- Mid Zone Fine Grey pigmentary changes	Media - clear Disc - 0.3 A:V - 2:3 Foveal reflex- present ONH- Mild Pallor in Temporal Rim NRR - healthy - no pigmentary changes
Refraction	+4 D	+4 D

Investigations

ERG - No response

MRI Brain - Normal study

BERA for hearing - Normal

Diagnosis : Lebers Congenital Amaurosis

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Probable pathophysiology and its management

Roga Prakruti	***Samprapti Ghataka***	***Samprapti Vigatana***
Dosha	*Vata*	*Matra Basti* - 20 ml - *Ksheera Bala Taila*[6] for 3 sittings with the gap of 7 days
Dushya	*Rasa, Rakta*	*Rakta Prasadana - Shiro Pichu*
Agni	*Mandya*	Improved by *Deepana, Pachana*
Srotas	*Raktavaha* - In *Netra Bhaga*	*Ushiradi Anjana*[4]
Sroto Dusti	Sanga - Improper Gene coding leading to progressive destruction of Retinal Pigments - *Beeja Dusti*	*Basti*
Udbhava Stana	*Pakvashaya (Vata Vyadhi)*	*Basti*
Vyaktastana	*Netra*	*Chakshusya - Shatavari Amalaka Payasa* [5]
Roga	*Jata Andya - Vataja Nanatmaja Vikara*	*Vatasya Upakrama - Snigdha Virechana, Basti, Brihmananga Snehapana.*
Upadrava	*Timira*	*Pushpa Bhandana*
Sadhya-Asadhyata	*Yapya*	*Vatasya Upakrama - Snigdha Virechana, Basti, Brihmananga Snehapana* Treatment Protocol of - 48 days

Treatment protocol

	Treatment	**Duration**	**Justification**
Deepana, Pachana	*Hingwastaka Churna* 3gm	3 days	To correct the mitrochondrial metabolism
Snigdha Virechana	*Gandharva Hastyadi Taila* - 5ml with 10ml milk	3 days	*Vatanulomana*
Matra Basti	With *Ksheera Bala Taila* 20ml	7 days - 3 sittings with interval of 7 days	It stimulates nerve impulses - stimulates photoreceptive activity and helps in strengthening ocular muscles - arrests further enopthalmos
Netra Kriya Kalpa	*Ushiradi Anjana* 1-0-1	48 days	Promotes microcirculation there by targets the pigmentary changes in retina
	Pushpa Bandhana - 20min	48 days	Targets nystagmus
Shamana	*Shatavari Amalaka* granules 5gm BD	48 days	Rich in Vitamin - A, stregthens visual pigments.
	Maha Triphaladi Gritha -	48 days	*Brihmana* and *Vataja Timirahara*

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	10gm with milk at bed time		

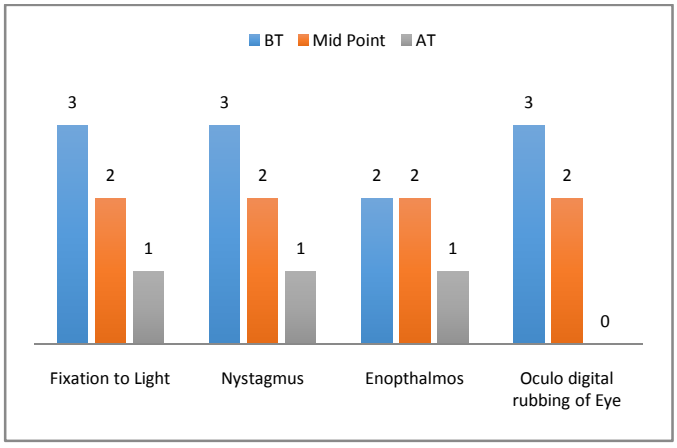
RESULTS

1. Grades of Nystagmus

Grade 1: Present only when looking in the direction of the quick component.
Grade 2 : Also present when looking straight ahead.
Grade 3 : present when looking in the direction of the quick component, when looking straight ahead and when looking in the direction of the slow component.

Grade 1 : <2mm -
Grade 2 : 2mm-3mm
Grade 3 : >3mm

1. There was marked improvement in nystagmus from grade 3 - grade 1.
2. The child was able to follow light due to improved fixation.
3. Frequent rubbing of eyes gradually came down.
4. Enopthalmos - Mainted at grade 2 and improved to grade1.
5. Photophobia reduced to marked extent.
6. There were no further changes found on fundus examination



DISCUSSION

In this clinical trial the treatment protocol was designed based on the understanding of the disease and also to address the non-availability of treatment in the contemporary science upart from the only hope of genetic counseling and genetic therapy which is not into practice yet in this era. *Basti* with *Vatahara Taila* advised as the line of treatment in the classics with intention to alleviate *Vata*. Is with a Probable mode of action: The main causative factor is considered to be *Vata* and best therapy is supposed to be *Basti Chikitsa*. This is most probably due to its controlling and regulating mechanism over the nervous system. *Basti Dravya* may enter minute channels of body and tissues get proper nourishment. Drugs administered high in the rectum are usually carried directly to the liver and thus, are subject to metabolism. Drugs administered low in the rectum are drained systemically by the inferior and middle rectal veins before passing through the liver. Rectal pH may also influence drug uptake by altering the amount of drug that is ionized. The greater lipid solubility of no ionized drugs enhances their movement across biological membrane. The pH of the rectal Vault in the children ranges from 7.2 to 12.2. This pH ranges favors absorption of the drug.; *Anjana* which was administered in the form of drops here is to enhance the microcirulation to the posterior segment of the eye and the *Brihmanaga Sneha* with *Shatavari* and *Amalaki* rich in vitamin A and C is a right combination to improve the functional capacity of the degenerating photoreceptors and to repair progressive destructive changes at the layer of retinal pigmental epithelium; with this approach as a *Samprapti Vigatana Chikitsa* the results observed is quite satisfactory thereby proving this protocol to be a effective visual-rehabilitation in this condition.

CONCLUSION

In this patient, the overall effect was found near 10-15% improvement in subjective parameters. As this disorder is incurable, this percentage of improvement also helps the patient to improve the quality of life (QOL). Treatment of this kind of condition is

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important and in that, if we are able to make small improvements in an earlier age, then it will reflect as a major benefit in later age in the form of developing skills. Previously, it was believed that neurons do not repair or rejuvenate after any injury, but the new concept of neuroplasticity says that CNS have the ability to repair their neurons by axonal sprouting to take over the function of damaged neurons. This improvement in patients also supports the concept of Neuroplasticity. Going by the results of this case study, we can conclude that Ayurvedic treatment protocol along with appropriate internal medication can do a lot for the improvement in QOL.

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