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Leber Congenital Amaurosis - A Case Report

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Abstract

Leber congenital amaurosis (LCA) is a congenital retinal dystrophy that results in severe vision loss in infancy associated with pigementary changes in later stages of life. We present to you a case of 9 month old male infant, who was referred for Ophthalmic evaluation as the child was
unable to fixate. Patient was case of global developmental delay with spastic cerebral palsy with seizure disorder secondary to hypoxic induced encephalopathy with hearing loss. On examination, patient doesn't follow light with either of eyes, pupil were central, circular, reactive to light. The lens were clear and there was torsional nystagmus. Rest anterior segment was within normal limit. Disc showed temporal pallor. Rest fundus was within normal limits. ERG
suggestive of bilateral marked diffuse reduction in photoreceptor functions. VEP shows poor response. Based on these investigations and our examinations, we came to diagnosis of LCA.

Keywords: Developmental delay, ERG, VEP

Introduction

A 9 month male infant was referred to Ophthalmic OPD for evaluation as infant was unable to fixate. Infant was as a case of developmental delay with history of spastic cerebral palsy with seizure disorder secondary to Hypoxic ischemic encephalopathy (HIE) with hearing loss.

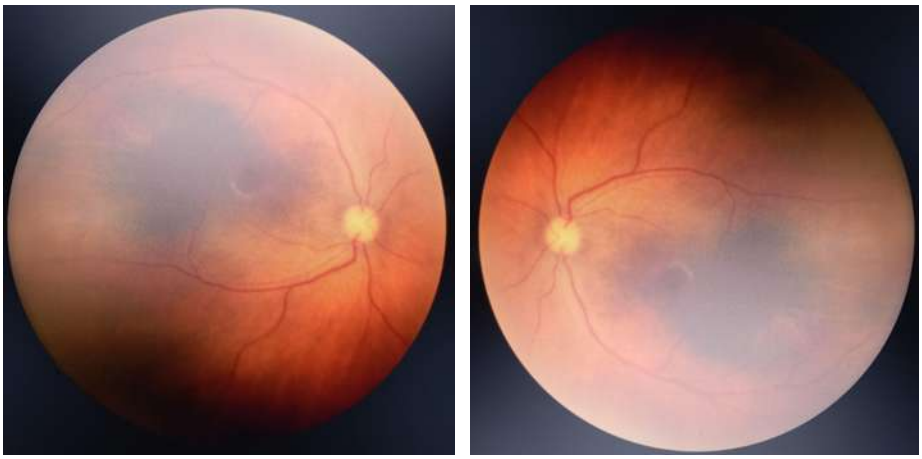
Birth History: Full Term Normal delivery, delayed crying at birth, NICU stay for 15 days from D1OL, H/o Oxygen administration for 15 days.

On Examination:

	OD	**OS**
Vision	Unable to follow and fixate light	Unable to follow and fixate light
Lids	Normal	Normal
Cornea	Clear	Clear

Conjunctiva	Clear	Clear
Anterior segment	WNL	WNL
Lens	Clear	Clear
Pupil	Central, circular, reactive to light	Central, circular, reactive to light
EOMS	Cannot be assessed at present, **Torsional Nystagmus** present	Cannot be assessed at present, **Torsional Nystagmus** present
Fundus	0.2 C/D ratio, wide disc margin, vertically oval, mild **temporal pallor** +, Macula WNL, FR+	0.2 C/D ratio, wide disc margin, vertically oval, mild **temporal pallor** +, Macula WNL, FR+

On evaluation, right sided head tilt was noted along with inability to hold head. Moderate



On evaluation, right sided head tilt was noted along with inability to hold head. Moderate hypermetropic refractive error was noted on retinoscopy. ERG was suggestive of Bilateral marked diffuse reduction in photoreceptor function in entire retina. VEP was suggestive of poor response. BERA was suggestive of normal hearing sensitivity. Sleep EEG was suggestive of

Epileptic encephalopathy and severe degree of physiological dysfunction.

MRI Brain (Plain) was suggestive of Peri-rolandiculegyria sequelae secondary to Hypoxic ischemic encephalopathy.

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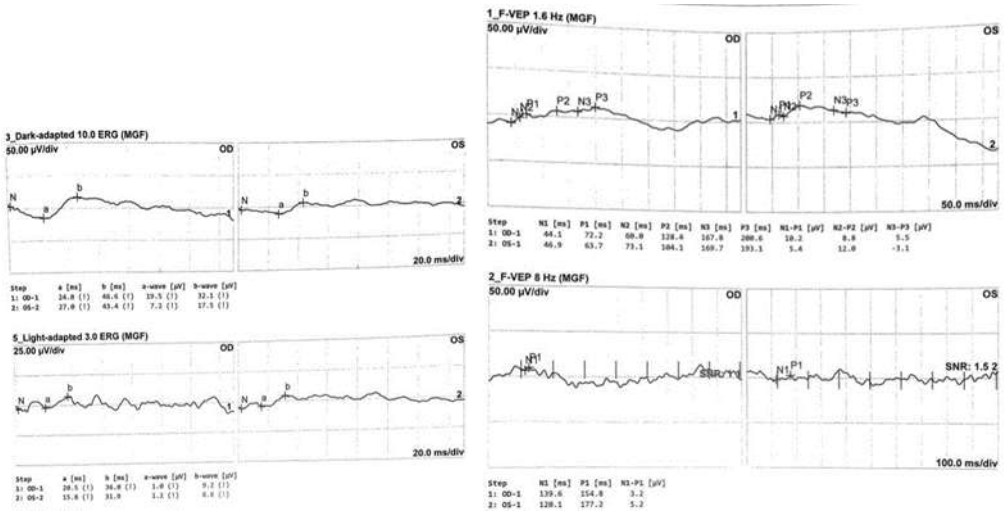

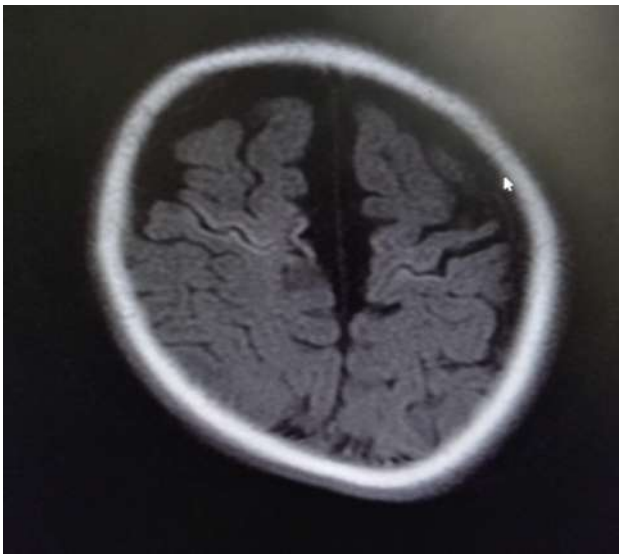

Based on detailed history, clinical evaluation and investigations diagnosis of LCA was made. Visual prognosis was explained to the patient's parents and the need for undergoing genetic testing and the recent advances in gene therapy and were asked to follow up after 6 months.

Discussion:

LCA refers to congenital retinal dystrophy that results in severe vision loss in infancy. It usually presents with nystagmus, sluggish or near absent pupillary response, high hyperopia. Estimated prevelance is 2-3 per 1,00,000 births(1). It is autosomal recessive inheritance(2). Various genes are affected like CEP 290, GUCY 2D, CRB1, RPE 65.

It is characterised by inability of eye to undergo phototransduction due to disruption of visual cycle. Various signs seen are "amaurotic" pupil, nystagmus, photophobia, hyperopia, oculo - digital sign.(3) It can also be associated with renal, vestibular and olfactory dysfunction.

Retina appears normal initially and later may show abnormalities like marbled fundus, choreoretinal degeneration, optic disc abnormality, coats like reaction. It is diagnosed clinically and based on

opthalmic history. ERG is suggestive of non recordable/ extinguished or severely reduced scotopic and photopic function. VEP shows variable response.(4)

Fundu Autofluorescence (FAF) can be done to measure lipofuscin accumulation in RPE which varies in different subtype of NCA. OCT shows retinal atrophy with bowing of macula.

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Precise diagnosis requires molecular genetic testing like DNA microarray, next generation sequencing, linkage analysis to identify specific genes affected in LCA.

Conclusion:

LCA should be considered in any infant presenting with inability to follow light especially in absence of pigmentary changes in eye. This case highlights the importance of thorough opthalmic evaluation in infants with developmental delay. Early detection is vital for supportive care, rehabilitation and genetic therapy consideration. Presence of torsional nystagmus, inability to fix and markedly reduced ERG response are suggestive of primary retinal dystrophy. Also recognition of existing comorbidities like spastic cerebral palsy, seizure disorder and hearing loss aid in comprehensive care of patient.

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