Case Report

Nondecussating retinal-fugal fiber syndrome: Case report of a rare cause of congenital nystagmus

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Abstract

Congenital nystagmus is a condition presenting itself in neonatal period or early infancy and Persists throughout the life. The overall prevalence of nystagmus is reported to be between 2-3 cases per 10000 individuals. In majority of the cases the nystagmus is pendular or see saw. Though there are numerous causes of congenital nystagmus, congenital absence of optic chiasma is one of the rare causes. It is usually diagnosed on Magnetic resonance Imaging which is frequently done in infants presenting with nystagmus. We present here a case of 3 years old female child who presented to us with history of nystagmus since infancy and was found to be having congenital absence of optic chiasma on MRI imaging. **Case Report:** 3 years old female child was brought to OPD with the complaints of decreased vision in both eyes and abnormal eye movements. No significant family history. There was also a history of delayed developmental milestones in the form of delayed neck holding. On torchlight examination there was presence of pendular nystagmus. Slit lamp examination was in normal limits. Dilated Fundoscopy was normal. An MRI was ordered which showed absence of optic chiasma. In view of imaging findings, a diagnosis of congenital pendular nystagmus due to achiasma was made and bilateral Medial and lateral rectus recession surgery was done. Flash Visual evoked potential showed normal amplitudes with good reproducibility bilaterally whereas pattern Visual evoked potentials showed reduced amplitude with delayed latency for larger checker and extinguished peaks for smaller checker pattern in right eye and extinguished peaks for larger checker pattern in left eye. Patient was discharged with glasses prescription. **Conclusion:** Though rare, possibility of congenital achiasma remains undiagnosed unless MRI is advised.

Key Word: Congenital Nystagmus, Magnetic Resonance Imaging, Achiasma, Recession Surgery.

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INTRODUCTION

Nystagmus can be defined as regular and rhythmic, to-and-fro involuntary oscillatory eye movements. The overall prevalence of nystagmus is reported to be between 2-3 cases per 10000 individuals[1]. It can be divided into congenital or acquired nystagmus on the basis of age. It may be pendular or jerk nystagmus in pendular nystagmus, movements are of equal velocity in each direction. In jerk nystagmus the movements have a slow component in one direction and fast component in other direction. Acquired nystagmus usually present at any age after infancy. Infantile or congenital nystagmus presents in early infancy and persists throughout life. In many cases infants are brought with history of delayed developmental milestones. Congenital nystagmus can be divided into sensory and motor nystagmus[2]. Sensory nystagmus is usually associated with blindness due to defects or pathologies affecting afferent visual pathway including those of retina, optic nerve, chiasma, optic tract and cortical areas. Children loosing vision due to any reason before 6 months of age are likely to develop sensory nystagmus. Less common causes of sensory nystagmus include congenital defects such as absence of optic chiasma[3]. Motor nystagmus is due to pathologies involving efferent pathway of visual pathway and the most common type of motor nystagmus is idiopathic

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motor nystagmus. Many cases of congenital and infantile nystagmus are known to have familial pattern including X-linked, autosomal recessive and autosomal dominant patterns of transmission[4]. Congenital nystagmus usually presents within 6 months of age but in some cases it's noticed late by parents and hence the age of presentation varies. It usually present as symmetric, bilateral and conjugate eye movements which disappears during sleep. Sensory nystagmus due to congenital defects of visual pathway is an uncommon cause of sensory nystagmus presenting in early infancy. The conditions such as anopthalmos, albinism and septo-optic dysplasia may be associated with absence or hypoplastic optic chiasma[5]. Isolated absence of optic chiasma is one of the rare causes of congenital sensory nystagmus and very few cases of isolated absence of Optic chiasma have been reported in literature. Congenital absence of optic chiasma leading to sensory nystagmus is also known as Nondecussating retinal-fugal fiber syndrome in which the infant presents with nystagmus in early infancy. The diagnosis is usually confirmed on the basis of magnetic resonance imaging[6]. The management of patients with nystagmus depends upon type, severity and visual acuity of the patients. Medical management consists of correction of refractive error and amblyopia. GABA agonists or inhibitors of the excitatory neurotransmitter such as baclofen are used in adults but their use in children is controversial. Surgical management consists of Recession of rectus muscles and various studies have reported improvement in vision following surgery[7]. We are reporting here a case of congenital nystagmus due to isolated absence of optic chiasma because of its rarity in published literature sofar.

CASE REPORT

A 3 years old female child was referred to us in view of abnormal movements of eyes and decreased vision noticed by parents. There was no family history of nystagmus. Antenatal, natal and post natal history was uneventful. The baby was delivered by normal vaginal delivery to parents having first degree consanguinity. Baby cried immediately after birth. There were no immediate postnatal complications. There was no history of NICU admission. Developmental history revealed that there was delayed neck holding as well as delayed development of social smile. Gross motor and language development was normal. Past history showed presence of rapid eye movements since early infancy. On examination, vision was 20/200 in both eyes. There was 15-20 degree face turn. no chin lift. No facial asymmetry. Torchlight examination showed bilateral pendular nystagmus. Slit lamp examination of both the eyes was within normal range. Bilateral pupils were reacting to light. Dilated Fundoscopy showed OD was pink, CDR 0.2:1, macula fovea reflex present. There was no organomegaly and history of regression of milestones ruling out possibility of neurodegenerative disorders. In view of presence of nystagmus since early infancy a diagnosis of congenital nystagmus was made and neuroimaging was ordered. Magnetic resonance imaging showed volume loss in bilateral optic nerves with prominent peri-optic subarachnoid spaces. The nerves were seen extending beyond the orbital apex with relatively thinned out nerves seen converging anteriorly. There was a significant finding of absent optic chiasma on MRI. Features were consistent with congenital absence of achiasma.

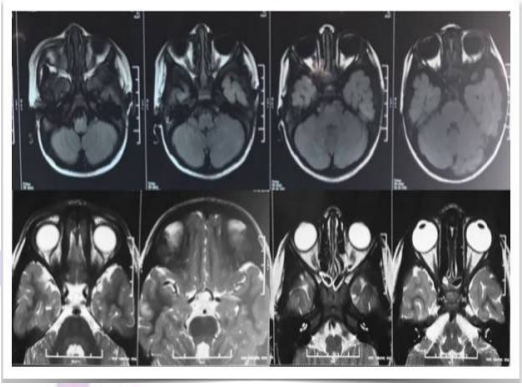

Figure 1: MRI Coronal T1 and T2 Weighted Images showing absence of optic chiasma. Also note normal optic nerves on both sides.

The final diagnosis of nondecussating retinal-fugal fiber syndrome was made on the basis of absent optic chiasma on neuroimaging along with presence of nystagmus since early infancy. Since there was considerable diminution of vision, surgery was planned. Bilateral medial and lateral rectus recession surgery in both eyes was done under GA. Post-surgery Flash Visual evoked potential showed normal amplitudes with good reproducibility bilaterally whereas pattern Visual evoked potentials showed reduced amplitude with delayed latency for larger checker and extinguished peaks for smaller checker pattern in right eye and extinguished peaks for larger checker pattern in left eye. Patient was finally discharged with glasses prescription which improved to 20/125 in both eyes with ocular motility full and free in all directions.

DISCUSSION

Abnormalities of eye movements are one of the common causes for which ophthalmic consultations are sought in adults but they are relatively uncommon in children. In pediatric age group particularly in infancy presence of nystagmus may be an important sign of structural anomalies involving central nervous system or visual pathway[8]. It is interesting to know that the first reported

case of congenitally absent optic chiasma was reported not in human being but in a sheep dog and was reported by Williams *et al* in 1990[9]. Later Apkarian *et al* reported cases of 2 children who had unusual visual pathway malformation in which nasal-retinal cortical projections, unable to decussate due to the inborn absence of an optic chiasma, erroneously route ipsilaterally to visual projection targets. The authors named it Nondecussating retinal-fugal fibre syndrome(NRFFS)[10]. There are very few case reports of congenital nystagmus due to isolated absence of optic chiasma. First case report of its association with vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal defects, and limb defects (VACTERL) was reported by Prakash S *et al* who reported a case of 29 year old male with VACTERL anomalies who presented with headache, photophobia, and worsening nystagmus[11]. Neuroimaging showed a thin remnant of the optic chiasm but normal optic nerves. Functional MRI during monocular visual stimulation showed non-crossing of the visual evoked responses in the occipital cortex, confirming achiasma. Leitch *et al* reported another case of achiasma with midline craniofacial defect who had a nasoethmoidal encephalocele and see-saw nystagmus[12]. Magnetic resonance imaging is the most common imaging investigation done for the confirmation of the diagnosis in these cases. Isolated achiasma by definition shows absence of optic chiasma with normal midline structures and rest of the optic pathway. It is important to look for achiasma on thin section and there are chances of missing this on MRI if only thick sections are evaluated. Advanced Imaging such as functional MRI can help in assessing non-crossing of retinal axonal fibers at the optic chiasm. Moreover it has high spatial resolution which permits the investigation of anatomic and functional visual pathway organization in humans with achiasma. After the improvements in imaging technologies many cases of optic achiasma are being diagnosed who had earlier been erroneously labeled to be having idiopathicnystagmus[13]. The management of nystagmus due to optic chiasma depends upon many factors including age of the patient, severity of nystagmus and visual acuity. The refractive errors and amblyopia needs to be corrected. The Pharmacologically useful agents for adult patients with nystagmus are primarily GABA agonists or inhibitors of the excitatory neurotransmitter system but their utility in infants is controversial[14]. Extraocular muscles recession surgery is found to be effective in treating congenital nystagmus. Hertle RW conducted a study of 5 children with congenital nystagmus to determine the effectiveness of horizontal rectus tenotomy in changing the nystagmus. Simple tenotomy of all four horizontal recti with reattachment at the original insertion was accomplished. Search-coil and infrared eye movement recordings and clinical examinations were performed before and 1, 6, 26, and 52 weeks after surgery. The authors concluded that tenotomy causes significant improvements in nystagmus foveation measure and measured visual function[15].

CONCLUSION

Congenital nystagmus due to absence of optic chiasma also known as Nondecussating retinal-fugal fibre syndrome is rare but important cause of congenital nystagmus and its possibility must be considered in any infant presenting with nystagmus. Early diagnosis and appropriate surgical intervention is found to be associated with better visual outcome in these patients.

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