Case report

CASE REPORT ON CONGENITAL FIBROSIS OF EXTRAOCULAR MUSCLES

ABSTRACT:-

BACKGROUND: Congenital fibrosis of extraocular muscle (CFEOM) is a rare congenital syndrome characterised by non-progressive unilateral or bilateral restrictive ocular motility with or without ptosis. It results from fibrosis of the extraocular muscle causing restricted ocular motility and optic nuclear dysplasia or hypoplasia. Patient can report with various presentations depending on different phenotypes of disease.

CASE PRESENTATION: A8-year-old boy came to CIMS eye opd with chief complaint given by informant (father) were unable to move eyeball along with watering in left eye since birth.

CONCLUSION: on thorough evaluation, it was observed patient had restricted ocular motility in all quadrants even with forced duction test with chin down position and hypermetropic refractive error, on MRI investigation showed thinning and fibrosis of extraocular muscles.

Keywords: Congenital fibrosis of extraocular muscle, dysplasia,hypoplasia, oculomotor nucleus

INTRODUCTION

Congenital fibrosis of the extraocular muscles (CFEOM) is diagnosed based on characteristic eye findings: congenital non-progressive ophthalmoplegia (inability to move the eyes) with or without ptosis (droopy eyelids) affecting part or all of the oculomotor nucleus and nerve (cranial nerve III) and its innervated muscles (superior, medial, and inferior recti, inferior oblique, and levatorpalpabrae superioris) and sometimes the trochlear and abducens nuclei and nerves (cranial nerves IV and VI) and their innervated muscles (superior oblique muscle and lateral rectus muscle, respectively) [5-8].Similar to Duane syndrome, it was assumed to be a developmental abnormality of the extra-ocular muscles for well over a century. It was only relatively recently that it was found to be a developmental abnormality of the oculomotor nucleus and nerve (Cranial Nerve III) [6].

CASE DESCRIPTION

An8-year-old boy visited CIMS eye OPD with chief complaint given by informant (father) were unable to move eyeball along with watering in left eye since birth. Sporadic presentation with no similar complaint among siblings. UCVA- (BE) 3/60, on cycloplegic correction- (BE) +5.00sph. BCVA- (BE) +4.00sph (6/60). Chin down position with no signs of ptosis.BE megalocornea 12.5*12.5 mm with intraocular pressure BE 17.3 mmhg. BE pupil were sluggishly reactive to light with BE fundus showed optic disc atrophy with pale neuroretinal rim, rest fundus within normal limits. On forced duction test it showed restricted

ocular motility in all quadrants. further MRI investigation revealed thinning and fibrosis of extraocular muscles.

Figure 1.a **Figure 1.b**

Figure 1.a showing mask like face with megalocornea

Figure 1.b showing chin down position.

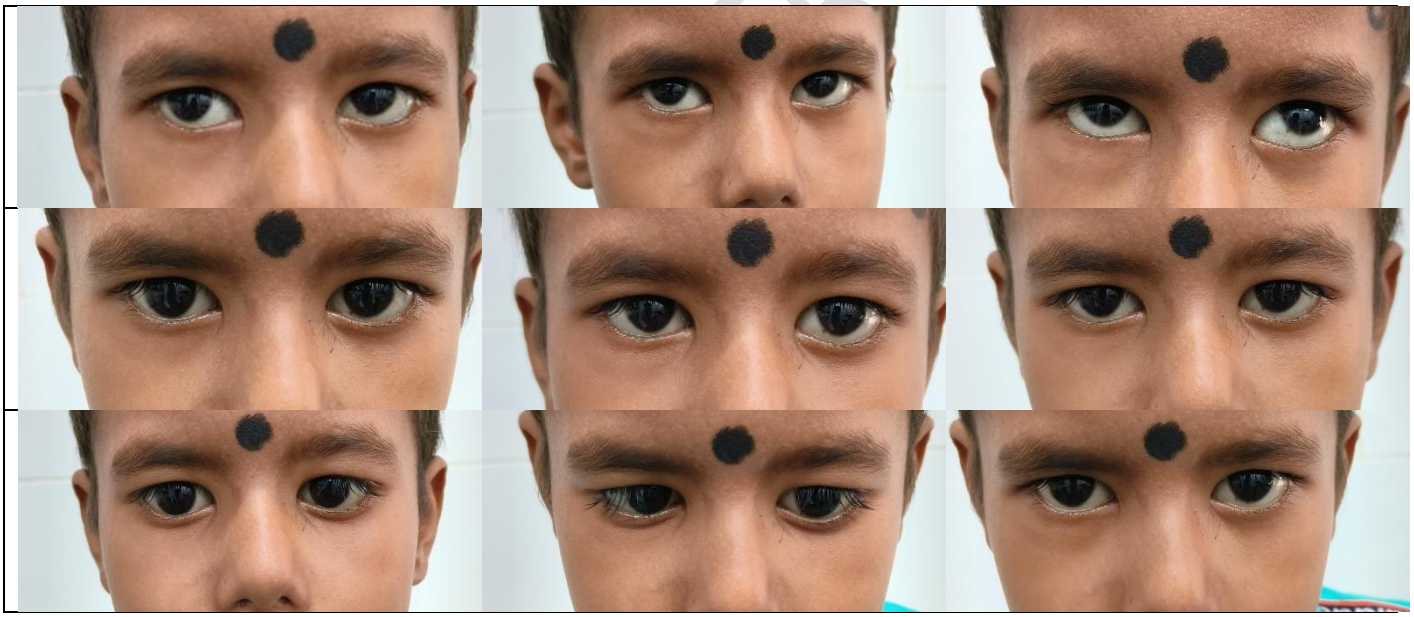

FIGURE 2: showing restricted ocular motility in all quadrants

Bottom row- left to right- dextrodepression, depression, levodepression

Middle row- left to right- dextroversion, primary position, levoversion

Upper row- left to right- dextroelevation, elevation, levoelevation

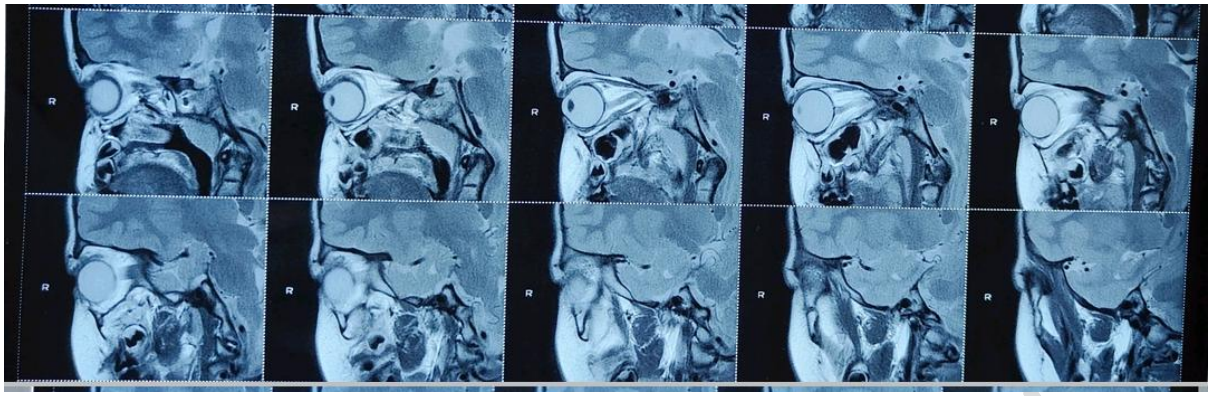

FIGURE 3: MRI showing thinned out and ill-defined extraocular muscles with aberrant insertion.

DISCUSSION:

- Congenital fibrosis of extraocular muscles presents with associated syndromes
- It is classified based upon clinical characteristics, forced duction test, radiological investigation and genetics.
- CFEOM 1- b/lptosis, hypotropia, restricted up gaze, horizontal strabismus, miotic pupil and positive forced duction test.
- CFEOM 2- b/l ptosis, exotropia, severe restriction of horizontal and vertical gaze, miotic poorly reacting pupil, positive forced duction test.
- CFEOM 3- atypical presentations
- Such as KearnsSayre syndrome, myotonic dystrophy, chronic progressive external ophthalmoplegia, oculopharyngeal muscular dystrophy.
- Primary management with refractive correction is done with surgical intervention to be persuaded in near future.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials

will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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