Ophthalmology Research: An International Journal

Volume 19, Issue 3, Page 1-6, 2024; Article no.OR.115109
ISSN: 2321-7227

Congenital Fibrosis of Extraocular Muscles: A Case Report

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

Article Information

DOI: 10.9734/OR/2024/v19i3418

Open Peer Review History:
This journal follows the Advanced Open Peer Review policy. Identity of the Reviewers, Editor(s) and additional Reviewers, peer review comments, different versions of the manuscript, comments of the editors, etc are available here: https://www.sdiarticle5.com/review-history/115109

Case Report

Received: 01/02/2024
Accepted: 04/04/2024
Published: 11/04/2024

ABSTRACT

Unprogressive unilateral or bilateral limited ocular motility, with or without ptosis, is a hallmark of the uncommon congenital disease known as congenital fibrosis of extraocular muscle (CFEOM). Fibrosis of the extraocular muscle leads to optic nuclear dysplasia or hypoplasia and reduced ocular mobility, which in turn causes it. diverse phenotypes of illness might cause patients to report with diverse presentations.

Keywords: Congenital fibrosis of extraocular muscle; congenital syndrome; inherited strabismus syndromes.

1. INTRODUCTION

"One characteristic of numerous hereditary strabismus illnesses commonly known as CFEOM is congenital restrictive ophthalmoplegia, which limits globe movement in one or more fields of vision and affects extraocular muscles innervated by the CNIII and/or CNIV. Congenital Cranial Dysinnervation disorders (CCDDs) was the official nomenclature

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for the conditions affecting the innervation of muscles outside of the eye [1]. The CCDDs incorporate all of the CFEOM variants."

The patient with CFEOM who came to our clinic had a history of congenital watering, reduced ocular motility, and ptosis; we detail their clinical and neuro-radiological findings and evaluate the literature on the condition's genetics, treatment, and clinical aspects.

2. CASE PRESENTATION

An 8-year-old boy visited our department 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.

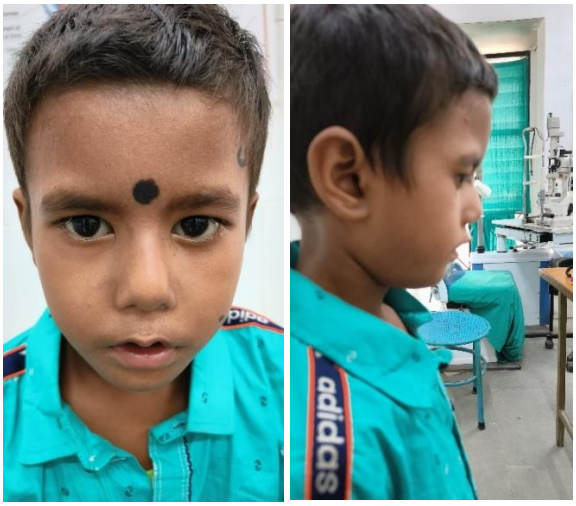

Fig. 1 a **Fig. 1 b**

Fig. 1a. Showing mask like face with megalocornea
Fig. 1b. Showing chin down position

"Abbreviations- UCVA- uncorrected visual acuity, BCVA- best corrected visual acuity, BE- both eyes"

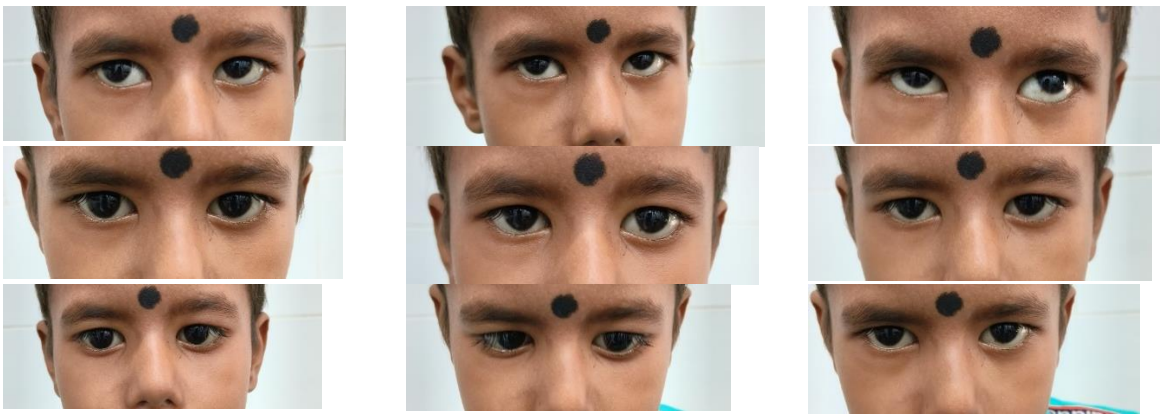

Fig. 2. Showing restricted ocular motility in all quadrants
Upper row- left to right- dextroelevation, elevation, levoelevation
Middle row- left to right- dextroversion, primary position, levoversion
Bottom row- left to right- dextrodepression, depression, levodepression

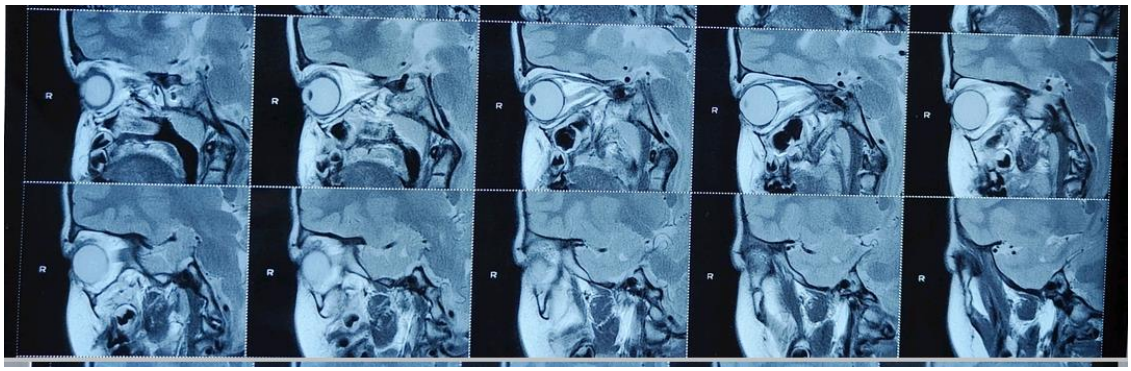

Fig. 3. Sagittal plane T2-weighted MRI image showing thinned out and ill-defined extraocular muscles with aberrant insertion

4. DISCUSSION

A uncommon congenital condition characterised by numerous limitations of extraocular muscles, CFEOM does not progress. Clinical features and genetics dictate its diagnosis and categorization. There are three distinct forms of CFEOM, distinguished by their respective clinical manifestations and genetic makeup [Table 1]. It is possible that bilateral CFEOM instances are very asymmetrical. Patients with CFEOM have a long list of systemic and ocular connections [Table 2]. It is important to distinguish CFEOM from other illnesses that may be similar to it [Table 3] [2]. Clinical history, results of a forced duction test, imaging studies, and genetic testing all come together to confirm a diagnosis of CFEOM. Optimal outcomes in planning and management can be achieved by using this strategy. However, genetic testing is crucial for diagnostic confirmation because several CFEOM groups have similar clinical symptoms.

Table 1. Classification of CFEOM [1,3,4,5,6]

Type 1	Type 2	Type 3 (A, B, C)
Orthoptics:		
• Each side experiencing ptosis • Reduced vision • Horizontal strabismus, a condition characterised by a narrowed field of vision, is prevalent, and its severity can vary. • Furthermore, pupils are often tiny and unresponsive. • Forced positive induction	• Each side experiencing ptosis • Anterior ocular tilt • The patient's ability to move their eyes laterally and vertically is severely limited, and they exhibit fluctuating abduction. • Irreactive, miotic eyes • Forced positive induction	• Classic symptoms of the illness are not present in all afflicted persons. They could have elevated eyes that aren't infraducted or that aren't below the midline. • One or both eyes might be impacted • Variability or absence of ptosis is possible. • Forced positive induction
Pathogenesis:		
Not present is the oculomotor nerve's upper division. "Abnormalities of the levator palpebrae superior and rectus superior"	"Problems with the innervated muscles and a complete lack of motor neurons in the oculomotor and trochlear nuclei"	"An abnormality in the development of the oculomotor nerve, with a preference for the superior branch over the inferior branch."
Genetics:		
Locus chromosome 12 Gene – KIF21A	Locus – chromosome 11 Gene – PHOX2A (ARIX/11q13)	A: Locus – Chromosome 16 "Gene – TUBB3"
Autosomal dominant	Autosomal recessive	"B: Locus chromosome" 12

Type 1	Type 2	Type 3 (A, B, C)
Fully penetrant		"Gene – KIF21A"
Variable expression		"C: Locus – Chromosome 13"
		"Gene – unknown"
		"Autosomal dominant"
		"Incomplete penetrance"
		"Variable expression"

Table 2. Ocular and systemic associations [5]

Ocular associations CFEOM
• "Refractive errors / amblyopia"
• "Neural misdirection – MG Phen., synergistic divergence / convergence"
• "Optic nerve dysplasia or hypoplasia"
• "Chorioretinal coloboma"
• "Microphthalmia"
• "Oculocutaneous albinism"
• "Marcus Gunn jaw – winking phenomenon"
Systemic associations CFEOM
• "Other cranial N anomalies – V, VII"
• "Facial dysmorphism"
• "Neurodevelopmental defects""

Table 3. Differential diagnoses of CFEOM [4]

Neurogenic
• "Congenital III nerve palsy"
• "Partial or complete VI nerve palsy"
• "Chronic progressive external ophthalmoplegia"
Restrictive
• "Brown's syndrome"
• "Orbital floor fracture"
• "Thyroid eye disease"
• "Double elevator palsy"
• "Möbius' syndrome"
• "Atypical Duane Syndrome"
"Myogenic with systemic involvement"
• Myasthenia gravis
• Kearns-Sayre Syndrome

"Neuroimaging has been suggested as a component of the examination of patients with CFEOM to rule out any orbital or brain sickness, since CFEOM is linked to neuro-radiological abnormalities.Hypoplasia of CN 3 on one or both sides has been shown by high-resolution magnetic resonance imaging in numerous patients with CFEOM.A neuropathic rather than a myopathic cause for CFEOM may be hinted at by CN 3 hypoplasia."

Treatment for CFEOM is challenging. It is important to rectify any refractive defect and amblyopia. Some individuals with CFEOM adopt very forward or backward chin postures, which can lead to eccentric seeing through corrective lenses and less than ideal refractive correction. This might be the root cause of our patients' impaired eyesight. Refraction varies significantly after extraocular muscle surgery because the muscles' push on the globe changes in both magnitude and direction [7].

Strabismus and ptosis are difficult surgical conditions to treat in CFEOM. Before attempting to treat ptosis, strabismus surgery is always considered. Both the patient and their parents need to know what to expect from strabismus surgery in order for it to be a successful procedure. There may be signs of quite big recessions (12mm) in the muscles that are afflicted. Because of the risk of exacerbating the enophthalmos, CCDDs typically refrain from resecting the extraocular muscles [2] Both before and during strabismus surgery, a forced duction test should be performed. Regarding ptosis surgery, it is recommended to undercorrect the condition since there is no Bell's phenomenon and the danger of exposure keratopathy. In order to avoid deprivation amblyopia, accurate ptosis should straighten the visual axis, and reduce or eliminate head position to some extent [8-10].

CONCLUSION

My patient was diagnosed with type 3 CFEOM and presented with several related variables, such as bilateral megalocornea measuring 12.5*12.5mm, optic atrophy accompanied by a pale optic disc, intraocular pressure of 17.3 mm Hg in both eyes, and reduced ocular movement in both quadrants. The patient's primary care included corrective eyewear, monitoring for glaucoma, and eventual surgical surgery. We will also do a mitochondrial test. To stay up with the related variations of CFEOM, constant reporting of instances is necessary, along with enhanced genetic profiling.

CONSENT

All necessary patient permission documents have been received, as the authors attest. This form confirms that the patient or patients have granted their permission for the journal to publish their photos and other clinical data. Although every attempt will be taken to ensure the patients' confidentiality, they are aware that their names and initials will not be published.
Ethical Approval:

As per international standards or university standards written ethical approval has been collected and preserved by the author(s).

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

REFERENCES

1. Gutowski NJ, Bosley TM, Engle EC. 110th ENMC International Workshop: the congenital cranial dysinnervation disorders (CCDDs). Naarden, The Netherlands, 25-27 October, 2002. None Neuromuscul Disord. 2003;13:573–8.
2. Mohamad S, Jaafar MD, Traboulsi EI. Clinical strabismus management, principles and surgical techniques. In: Rosenbaum AL, Santiago AP, editors. Congenital fibrosis of the extraocular muscles. Saunders Company; 1999. ISBN: 0-7216-7673-1 WB.
3. Flaherty MP, Grattan-Smith P, Steinberg A, Jamieson R, Engle EC. Congenital fibrosis of the extraocular muscles associated with cortical dysplasia and mal development of the basal ganglia. *Ophthalmology.* 2001;108: 1313–22.
4. Traboulsi EI. Congenital abnormalities of cranial nerve development: Overview, molecular mechanisms, and further evidence of heterogeneity and complexity of syndromes with congenital limitations of eye movements. *Trans Am Ophthalmol Soc.* 2004;102:373–89.
5. Doherty EJ, Macy ME, Wang SM, Dykeman CP, Melanson MT, Engle EC. CFEOM3: A new extraocular congenital fibrosis syndrome that maps to 16q24.2-q24 3. Invest Ophthalmol Vis Sci. 1999;40:1687–94.
6. OMIM. Online Mendelian inheritance in Man. Fibrosis of extraocular Muscles, Congenital, 1; CFEOM1. Johns Hopkins University.

   Available:http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=135700
7. Ansons AM, Davis H. 3rd ed. Diagnosis and management of ocular motility disorders; p. 438. Chapter 21, ISBN-10:0-632-04798-4; 2010.
8. Liu C, Ohri R, Frongia G, Collin R. Surgical correction of ptosis in ocular fibrosis syndrome. *Br J Ophthalmol.* 1994;78: 271–4.

9. Khan AO, Khalil DS, Al Sharif LJ, Al-Ghadhfan FE, Al Tassan NA. Germline mosaicism for KIF21A mutation (p.R954L) mimicking recessive inheritance for congenital fibrosis of the extraocular muscles. Ophthalmology. 2010;117:154–8.

10. Lim KH, Engle EC, Demer JL. Abnormalities of the oculomotor nerve in congenital fibrosis of the extraocular muscles and congenital oculomotor palsy. Invest Ophthalmol Vis Sci. 2007;48: 1601–6.

Peer-review history:
The peer review history for this paper can be accessed here:
https://www.sdiarticle5.com/review-history/115109