

Selective aplasia of global fibres of all extraocular muscles in congenital fibrosis of extraocular muscles (CFEOM): a rare presentation

Manu Saini,¹ Pradeep Sharma,¹ Nripen Gaur,¹ Jagjeet Singh²

¹Department of Ophthalmology, Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, Delhi, India

²Department of Radiology, All India Institute of Medical Sciences, New Delhi, Delhi, India

Correspondence to

Professor Pradeep Sharma, drpsharma57@yahoo.com

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DESCRIPTION

A 15-year-old boy presented in our strabismus clinic with complaints of bilateral ptosis and limitation of ocular movements since birth. He had a positive family history of consanguinity and similar ocular movement abnormalities in his three siblings. There was no history of systemic illness. Physical examination showed normal growth parameters without craniofacial dysmorphism except blepharoptosis and lagophthalmos in both eyes, more marked in the right eye. Right eye best-corrected visual acuity (BCVA) was 6/12 and left eye BCVA was 6/9. Slit-lamp biomicroscopy of right eye showed exposure keratopathy, rest was unremarkable. Fundus examination was within normal limits, with no pigmentary retinopathy or optic atrophy changes. There was almost total external ophthalmoplegia, with normal reacting pupils to both direct and consensual light reflex in each eye. The patient preferred fixation with the left eye as his right eye had severe ptosis covering the pupillary area. The patient had chin up and 15° right head tilt position with 30 prism dioptre (PD) exotropia and 14 PD hypertropia in the right eye in primary position. Right eye medial rectus resection with downshift to inferior rectus was planned to improve exotropia and

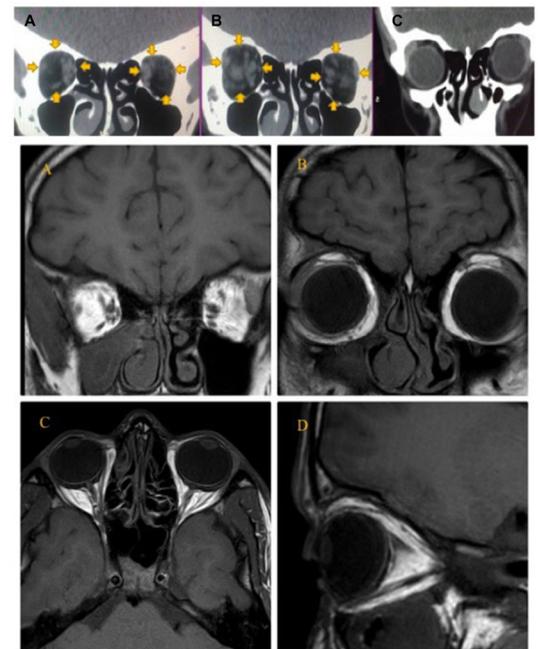


Figure 2 Non-contrast CT coronal view of subject showing origin of extraocular muscles (EOMs) at the orbital apex in (A), thinning of muscles at equator in (B) (demonstrated by arrowheads) and absence of EOMs beyond the equator in (C). Additional T1-weighted MRI collaborating CT findings showing origin (A), EOM thinning (B) and absent bilaterally beyond the equator in axial view (C) and sagittal view (D).

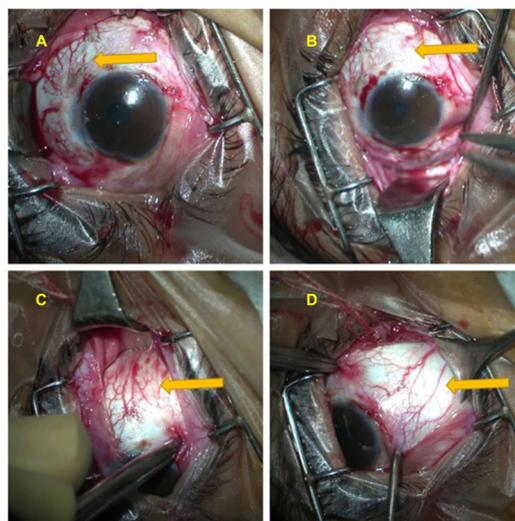


Figure 1 Surgeon's view, eyelid retractor was used to explore sclera following limbal peritomy. Intraoperative photograph shows absence of inferior rectus (A), lateral rectus (B), medial rectus (C) and superior rectus extraocular muscles (D) with intact ciliary vessels at their usual insertion sites.

hypertropia in primary gaze. Intraoperatively, the medial quadrant was explored to hook the medial rectus muscle but no muscle fibres were found. Further dissection was carried out to explore the other three recti and the oblique muscles. On careful exploration of scleral surface, no muscle fibres were identified, except for the presence of the anterior ciliary vessels at the insertion site of muscles, suggesting the absence of all extraocular muscles (figure 1). CT imaging and MRI disclosed the presence of extraocular muscles at the orbital apex that were abnormally thin and cord-like structure until the equator and missing beyond that. This was seen in both the eyes (figure 2).

Congenital absence of extraocular muscles (EOMs) is often associated with craniosynostosis.¹ EOM volume reduction secondary to denervation has been noted in congenital fibrosis of extraocular muscles (CFEOM), but total absence of muscle fibres anterior to equator (probably only the global



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fibres) has not been documented in CFEOM. MRI of the anterior tendinous part of extraocular muscles may not corroborate with the intraoperative findings in such cases as has been reported in the literature before by Singh *et al.*² They have reported bilateral medial rectus aplasia in three members of a single family. MRI of these cases revealed mild hypoplasia of medial rectus. However, intraoperative findings revealed empty muscle sheath in all these cases.

Selective absence of global layer of all EOMs connote ocular insult at neuromuscular interaction level during embryological development,³ suggesting separate developmental possibility of orbital and global layer fibres of the muscles. To the best of our knowledge, there has been no published literature describing

selective absence of global layer of all EOMs in CFEOM. Separate developmental possibility of orbital and global layer fibres has been suggested as also documented earlier.

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Learning points

- ▶ Recognition of such anomalies is crucial in avoiding unwanted outcomes of surgery.
- ▶ Orbital imaging should be performed to confirm the clinical suspicion of absent or anomalous extraocular muscles.

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