A rare case of congenital fibrosis of extra ocular muscles with Kallmann syndrome

Raman Yenugandula, Supraja K Ramavath, Krishna Kishore, Darakshan Qureishi

Key words: Kallmann Syndrome (KS), Congenital fibrosis of extraocular muscles (CFEOM), Krimsky's Prism bar cover test (KPBCT), Extraocular movements (EOM), Visually handicapped certificate(VHC), Exotropia

Access this article online	
Quick Response Code:	Website: www.ijo.in
	DOI: 10.4103/ijo.IJO_2130_21

Osmania Medical College, Sarojini Devi Eye Hospital, Hyderabad, Telangana, India

Correspondence to: Dr. Raman Yenugandula, Professor & HOD of Department of Pediatric Ophthalmology Strabismus and Neurophthalmology, Sarojini Devi Eye Hospital / Osmania Medical College, Mehdipatnam, Hyderabad - 500 028, Telangana, India . E-mail: drraman33@gmail.com

Received: 24-Aug-2021 Revision: 16-Oct-2021
Accepted: 18-Feb-2022 Published: 30-Jun-2022

Congenital fibrosis of extra ocular muscle (CFEOM) is a congenital nonprogressive, familial/sporadic condition involving fibrosis of EOM with ptosis, chin elevation, and deviation of eyes,[1] presenting a rare association of Kallmann syndrome (KS).

A 29-year-old man, K/C/O CFEOM with KS, visited OPD for VHC. VA RE: HM, LE: 6/36 -2dsph/-1cyl X 180° 6/12 A/S normal. Fundus RE – pathological myopia, [Fig. 1a] LE – normal [Fig.1b]. BE ptosis covering more than half the pupil [Fig. 2b]. H/O squint correction LE 20 years back and RE 9 years back for correction of large exotropia [Fig. 2a]. RE - Rt beat jerky nystagmus. Worth 4 dot test. RE suppression. KPBCT: RE neutralized >60 Base-in PD. EOM restricted in all gazes. Systemic evaluation revealed that he lacked secondary sexual characteristics: gynecomastia small genitalia [Fig. 3a–c]. H/o absence of libido, erectile dysfunction anosmia, and it was found that he is K/C/O KS.MRI brain with contrast showed elongated configuration of right globe with normal pituitary gland FSH and LH-low. S. Testosterone level was low - 1.38 ng/dL (normal: 270–1070 ng/dL). S. Prolactin was elevated. 2D ECHO and ECG: normal. Karyotyping 46XY, diagnosis made on clinical basis.[2]

Cite this article as: Yenugandula R, Ramavath SK, Kishore K, Qureishi D. A rare case of congenital fibrosis of extra ocular muscles with Kallmann syndrome. Indian J Ophthalmol 2022;70:2746-8.

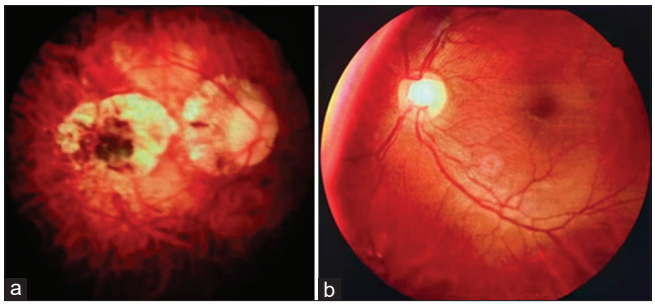

Figure 1: On fundus examination (a) pathological myopia (b) within normal limits

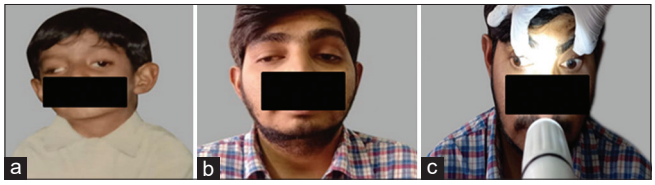

Figure 2: (a) RE 450 Exotropia in childhood (b) CHP prominent chin elevation (c) RE residual 45° exotropia and 10° hypotropia; LE residual 15° hypotropia

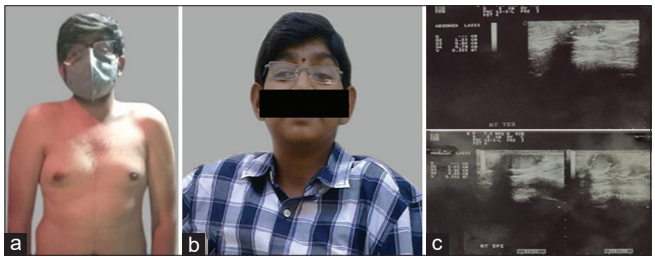

Figure 3: (a) Gynecomastia (b) absence of beard in puberty (c) USG scrotum shows small testes

Discussion

CFEOM is often associated with CNS, CVS, and other systemic abnormalities,[3] but association with KS is a rare entity. This case signifies the importance of systemic evaluation. The patient was advised bilateral IR recession for hypotropia to correct the chin elevation [Fig. 2b and c], but he denied surgery as he already underwent squint correction twice but there is recurrence.[4,5] He needs follow-up with an endocrinologist and inj. testosterone I/M every 3 weeks and oral testosterone.

This case exemplifies the need for systemic evaluation in the case of CFEOM.[3] KS is a hypogonadotropic hypogonadism condition. Along with hypotropia correction, we have to support them psychologically and socially for their wellbeing by rehabilitation.

Crutch glasses are advised for ptosis to prevent lagophthalmos. Appropriate refractive correction is required every 6 months.

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.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Whitman MC, Jurgens JA, Hunter DG, Engle EC. Congenital Fibrosis of the Extraocular Muscles Overview. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1348/.
2. Reardon W. Kallamann syndrome presenting as congenital ptosis in brothers. Clin Dysmorphol 2007;16:207-8. doi: 10.1097/MCD.0b013e3280b10beb.
3. Flaherty MP, Grattan-Smith P, Steinberg A, Jamieson R, Engle EC, Congenital fibrosis of the extraocular muscles associated with cortical dysplasia and maldevelopment of the basal ganglia. Ophthalmology 2001;108:1313-22.
4. Okita Y, Kimura A, Okamoto M, Mimura O, Gomi F. Surgical management of pediatric patients with congenital fibrosis of the extraocular muscles. Jpn J Ophthalmol 2020;64:86-92.
5. Heidary G, Mackinnon S, Elliott A, Barry BJ, Engle EC, Hunter DG. Outcomes of strabismus surgery in genetically confirmed congenital fibrosis of the extraocular muscles. J AAPOS 2019;23:253e1-6.