Anomalous extraocular muscles in Crouzon syndrome with V-pattern exotropia

Seung Ah Chung, Seung Yeop Lee

Strabismus associated with Crouzon syndrome is common and often complex. V-pattern strabismus is most commonly reported in this condition and is mainly thought to be due to an excyclorotation of the orbits and rectus muscle pulleys. We report two cases of children with Crouzon syndrome and V-pattern exotropia who had rectus muscle heterotopy on orbital imaging and were also found intraoperatively to have anomalous extraocular muscles. At the time of surgery, bifid insertion, misdirection, and fibrosis of extraocular muscles were noted. This highlights the various causes of V-pattern strabismus associated with Crouzon syndrome, including dysmorphic orbits and extraocular muscle anomalies.

Key words: Crouzon syndrome, extraocular muscle anomalies, V-pattern strabismus

V-pattern horizontal strabismus has been reported as the most common deviation in Crouzon syndrome.[1-3] Various theories have been proposed as causes of the V-pattern.[2,4-6] It is most likely related to the excyclorotation of the rectus muscle pulleys based on the findings of orbital imaging.[1,2,5,6] In addition, anatomical abnormalities of extraocular muscles are relatively common in the patients with Crouzon syndrome and also considered as a contributing cause of strabismus.[3]

Access this article online

Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/ijo.IJO_1121_19

Department of Ophthalmology, Ajou University School of Medicine, Suwon, South Korea

Correspondence to: Dr. Seung Ah Chung, Ajou University Hospital, 164, World Cup-ro, Yeongtong-gu, Suwon - 16499, South Korea. E-mail: mingming8@naver.com

Received: 10-Jun-2019 Revision: 09-Sep-2019
Accepted: 22-Oct-2019 Published: 20-Apr-2020

Case Reports

Case 1

A 4-year-old boy with Crouzon syndrome and a confirmed heterozygous mutation in the fibroblast growth factor receptor 2 (FGFR-2) gene (c.1030G>C) was referred for ocular misalignment. He had undergone an expansion cranioplasty with trans-suture distraction osteogenesis at 3 years of age.

On presentation, his visual acuity was 20/30 in each eye, and +1.25 diopters of hyperopia in both eyes. Motility examination revealed bilateral inferior oblique overaction. He had an exotropia of 35 prism diopter (PD) and a left hypotropia of 20 PD in primary position. He showed a V-pattern exotropia with alternating hypertropia on lateral gazes. Fundus examination showed a prominent excyclotorsion in the left eye. A computerized tomography (CT) scan of the orbit confirmed the presence of all rectus muscles and superior oblique muscle and excyclorotation of the rectus muscles, more prominent in the left eye [Fig. 1].

A recession of lateral rectus muscle and inferior oblique myectomy in both eyes were planned via limbal incisions to correct the V-pattern exotropia, and a recession of left inferior rectus muscle was also planned to correct the left hypotropia. However, at the time of surgery, a bifid left lateral rectus muscle insertion was found and a thin left inferior rectus muscle was inserted abnormally anterior at 3.0 mm from the limbus with nasally slanting [Fig. 2]. We performed surgery as planned except the recession of left inferior rectus muscle. Three years postoperatively, he had a left hypotropia of 6 PD in primary position with mild left inferior oblique overaction.

Cite this article as: Chung SA, Lee SY. Anomalous extraocular muscles in Crouzon syndrome with V-pattern exotropia. Indian J Ophthalmol 2020;68:924-6.

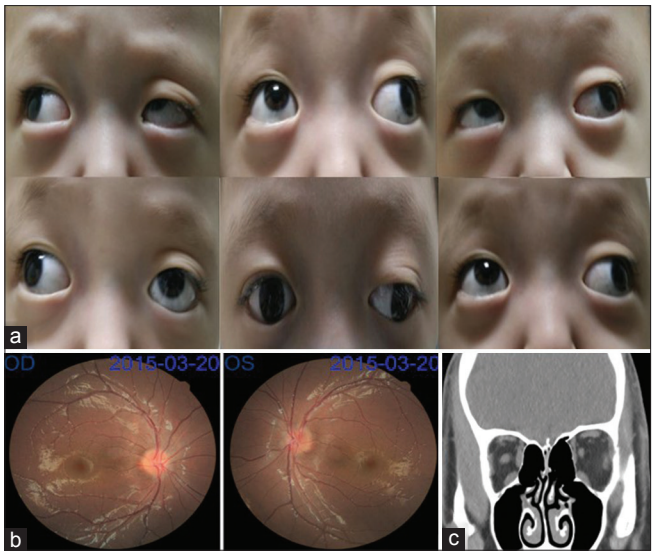

Figure 1: Gaze photography, fundus photography, and computerized tomography (CT) scan of the orbits for case 1. (a) Versions showing a left hypotropia and exotropia with bilateral inferior oblique overaction. Photographs of downgaze were not available due to patient's poor cooperation. (b) Fundus photographs showing excyclotorsion in the left eye. (c) CT scan showing excyclorotation of the rectus muscles in both eyes with the relative temporal displacement of the superior rectus muscle compared to the inferior rectus muscle and the relative inferior displacement of the lateral rectus muscle compared to the medial rectus muscle

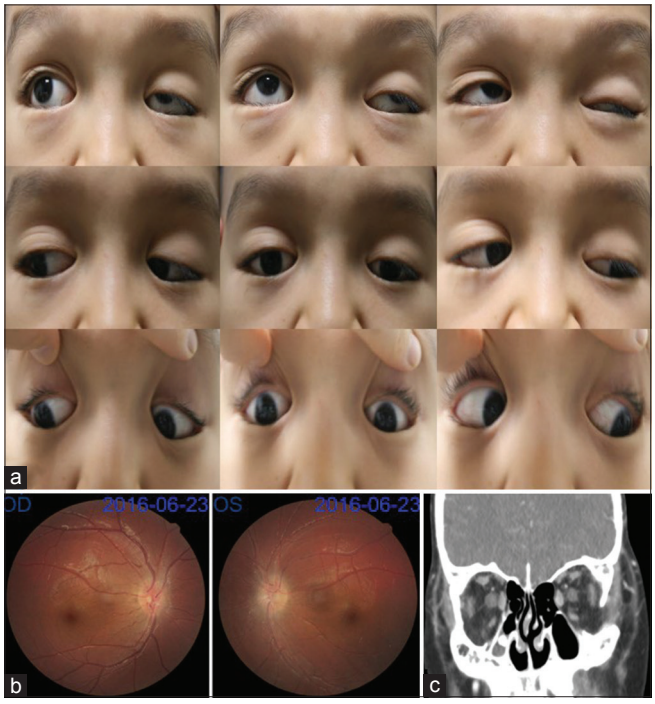

Figure 3: Gaze photography, fundus photography, and computerized tomography (CT) scan of the orbits for case 2. (a) Versions showing a left hypotropia and V-pattern exotropia with bilateral inferior oblique overaction. (b) Fundus photographs showing mild excyclotorsion in the left eye. (c) CT scan showing dysmorphic orbits and excyclorotation of the right orbit

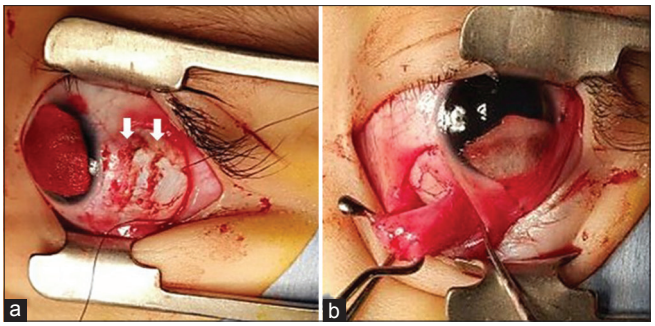

Figure 2: Intraoperative findings for case 1. (a) Bifid insertion of the left lateral rectus muscle (arrows). (b) The left inferior rectus muscle inserted abnormally anterior at 3.0 mm from the limbus and nasally misdirected

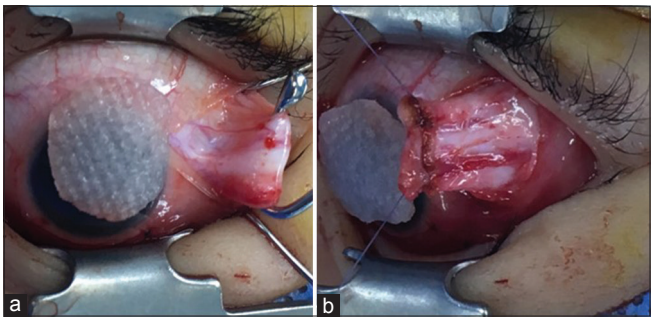

Figure 4: Intraoperative findings for case 2. (a and b) A fibrous and thick left lateral rectus muscle inserted abnormally anterior at 4.0 mm from the limbus

Case 2

A 7-year-old boy with familial Crouzon syndrome secondary to FGFR-2 gene mutation had difficulty with increased intracranial pressure and entropion of left lower eyelid in early childhood. He had undergone multiple expansion cranioplasties, a ventriculoperitoneal shunt, and multiple entropion corrections.

On presentation, his corrected visual acuity was 20/25 in the right eye and 20/40 in the left eye, with correction of mild myopic astigmatism. He had a history of papilledema, which had affected the left eye more prominently, consistent with mild gliosis on the left optic disc. He had a V-pattern exotropia of 25 PD in primary position, increased to 40 PD in upgaze and decreased to 20 PD in downgaze, associated with bilateral inferior oblique overaction. He also had a left hypotropia of 18 PD in primary position. Given no fundus excyclotorsion and mild inferior oblique overaction, bilateral inferior oblique myectomies were not planned. A CT scan of the orbit revealed that all rectus muscles and superior oblique muscle were present, but the rectus muscles were excyclorotated in the right eye [Fig. 3].

A recession of left lateral rectus muscle and a recession of left inferior rectus muscle were performed via limbal incisions. At the time of surgery, a thick and fibrous lateral rectus muscle was noted to be inserted anterior at 4.0 mm from the limbus [Fig. 4]. Prior to dissection, the forced duction testing did not reveal any restriction of adduction in the left eye. Two years

postoperatively, he had a left exotropia of 12 PD in primary position with mild left inferior oblique overaction.

Discussion

Patients with Crouzon syndrome commonly have a characteristic V-pattern strabismus, accompanied by a prominent overaction of the inferior oblique muscles.[1-6] The excyclorotation of the orbits and rectus muscle pulleys secondary to the fused coronal sutures may be mainly responsible for the V-pattern strabismus in these patients.[2,5,6] Weiss *et al*. reported that the magnitude of V-pattern exotropia simulated by pulley displacements using a computer program was highly correlated with those observed in patients with Crouzon syndrome.[5] In addition, Dagi *et al.* reported that the magnitude of excyclorotation of the orbits on imaging was clinically correlated with severity of V-pattern in syndromic craniosynsotosis.[6] Thus, preoperative orbital imaging for extraocular muscles is crucial in surgical planning in these patients.

However, Greenberg and Pollard suggested a limited exploration of all extraocular muscles because anomalies of extraocular muscles still play an important role in strabismus of craniosynostosis, but may not be recognized on preoperative orbital imaging.[7] It is well known that high incidence of extraocular muscle anomalies including bifid muscles, fused muscles, abnormal muscle insertions, and fibrous bands in these patients.[3,4,7] The patient of case 1 had a left hypotropia paradoxically with apparent overaction of the left inferior oblique muscle, one of the elevators. It could be related with intraoperative findings of abnormal anterior insertion of the left inferior rectus muscle, which can act as a stronger depressor than usual.

The cause of these extraocular muscle anomalies is not well known. Miller suggested that the dysmorphic orbit could restrict a development of adjacent structures including extraocular muscles.[8] Khan *et al*. proposed that abnormal expressions of FGFR-2 in the extraocular muscles may be responsible for anomalies of extraocular muscle structure in patients with syndromic craniosynostosis secondary to FGFR-2 gene mutations.[9] The patient of case 2 had a whitish and inelastic lateral rectus muscle with rich fibrous tissue. It could be related with the fact that FGFR-2 mutation leads to the excessive production of the fibrous tissue.[10]

This case report highlights the multifactorial causes of V-pattern strabismus in patients with Crouzon syndrome, abnormal morphology of extraocular muscles as well as abnormal location. The extraocular muscle imaging prior to strabismus surgery and handling unexpected intraoperative findings of extraocular muscle should be important in these patients.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Rosenberg JB, Tepper OM, Medow NB. Strabismus in craniosynostosis. J Pediatr Ophthalmol Strabismus 2013;50:140-8.
2. Ron Y, Dagi LR. The etiology of V pattern strabismus in patients with craniosynostosis. Int Ophthalmol Clin 2008;48:215-23.
3. Qiao T, Wang G, Xiong J, Luo W, Chen J. Surgical treatment of V-pattern exotropia in Crouzon syndrome. J Pediatr Ophthalmol Strabismus 2015;52:299-304.
4. Coats DK, Paysse EA, Stager DR. Surgical management of V-pattern strabismus and oblique dysfunction in craniofacial dysostosis. J AAPOS 2000;4:338-42.
5. Weiss AH, Phillips J, Kelly JP. Crouzon syndrome: relationship of rectus muscle pulley location to pattern strabismus. Invest Ophthalmol Vis Sci 2014;55:310-7.
6. Dagi LR, MacKinnon S, Zurakowski D, Prabhu SP. Rectus muscle excyclorotation and V-pattern strabismus: A quantitative appraisal of clinical relevance in syndromic craniosynostosis. Br J Ophthalmol 2017;101:1560-5.
7. Greenberg MF, Pollard ZF. Absence of multiple extraocular muscles in craniosynostosis. J AAPOS 1998;2:307-9.
8. Miller MT. Ocular abnormalities in craniofacial malformations. Int Ophthalmol Clin 1984;24:143-63.
9. Khan SH, Britto JA, Evans RD, Nischal KK. Expression of FGFR-2 and FGFR-3 in the normal human fetal orbit. Br J Ophthalmol 2005;89:1643-5.
10. Steinberger D, Reinhartz T, Unsöld R, Müller U. FGFR2 mutation in clinically nonclassifiable autosomal dominant craniosynostosis with pronounced phenotypic variation. Am J Med Genet 1996;66:81-6.