Bilateral absent medial rectus with neurological abnormalities: A rare clinical presentation and its management

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Abstract:
We discuss here an interesting case of strabismus, due to bilateral absent medial rectus muscle associated with brain anomalies, and its surgical management.

Keywords:
Absent medial rectus, congenital cranial dysinnervation disorders, transposition

Introduction
Virtually all extraocular muscles have been described as being congenitally absent, and the management of such complex cases can be a challenge to the surgeons. We discuss here a case with bilateral absence of medial rectus (MR) associated with brain anomalies, which could be a variant of congenital cranial dysinnervation disorders (CCDDs). To our knowledge, only a few cases of absent MR have been reported in the literature up to date, and none of these showed any neurological associations, as seen in our case.

Case Report

A 1½-year-old child presented with a history of outward squinting of both eyes (BE) since birth. He was born from a second-degree consanguineous marriage following a forceps delivery. He had a stormy perinatal period with birth asphyxia, and a history of seizures at 3 months of age. No significant family history was present. On examination, the child had low set ears with obliquely placed palpebral fissures. He had a chin elevation and a right face turn, associated with high-frequency jerk nystagmus, and was strongly resisting occlusion to the left eye (LE). Both eyes were turned out with right eye more than left and both the eyes were not able to adduct even up to the midline. There was no ptosis, aberrant regeneration, or palpebral fissure changes. Anterior segment examination was unremarkable, except for the presence of microcornea in BE. Fundus was normal in BE. A magnetic resonance imaging was advised which revealed vermian hypoplasia with ballooning of the 4th ventricle, hypoplastic cerebellar hemispheres, moderate dilatation of lateral ventricles, dilated cortical sulci, and shallow orbits [Figure 1a-d]. A gross lateral deviation of the eyeball was seen, with inability to visualize the medial recti on both sides [Figure 1e]. Rest of the extraocular muscles (EOMs) had a normal course and thickness. The child was referred to a neurologist but lost to follow-up; only to return 10 years later, with constant outward deviation of RE more than LE. On examination during this visit, he had a right face turn with a slight chin-up
posture [Figure 2]. The best-corrected visual acuity was 6/18 and N6 BE. Worth’s four dot test showed right suppression for distance and near, and stereoaucity was poor measured with Randot stereotest. Prism bar cover test (PBCT) showed >90 prism diopters (PD) of exotropia. The ocular motility showed a limitation of adduction not even up to midline and an elevation limitation of −3 in BE [Figure 3]. A differential diagnosis of pupil-sparing third nerve palsy, a variant of CCDDs, or a wall-eyed bilateral internuclear ophthalmoplegia was made. A lateral rectus (LR) hemi-hang back and medial partial transposition of superior rectus (SR) and inferior rectus (IR) was planned in both the eyes to correct the abnormal head posture and for cosmesis.

A forced duction test done on the table revealed a tight LR and minimally positive for IR. Force generation test was not done. A 360° peritomy was performed, and exploration around the insertion of MR was made, but the muscle could not be identified. The LR muscle was found to be extremely tight and was hooked and isolated with great difficulty. Placement sutures were placed with 6-0 vicryl close to its insertion. The muscle was recessed using a hemi-hang back technique by taking sclera bites 10 mm (mm) behind the insertion site and leaving the cut insertion end, 5 mm behind the scleral bites (total hang back of 15 mm). SR was identified, isolated, and bluntly dissected with a small muscle hook into half, approximately 14–18 mm posteriorly from insertion. Two sutures of 6-0 vicryl was passed through the medial half of the SR close to its insertion, disinserted, and resutured back 5.5 mm behind the medial limbus at the apparent insertion site of MR. The half-width tendon was then anchored to the sclera 8 mm behind the new insertion with 5-0 mersilk; the same procedure was repeated for the IR muscle. In view of anticipated compromised blood supply, only a partial thickness muscle transposition was performed in BE sparing the anterior ciliary vessels. SR muscle was found to be normal in position and caliber.

 Conjunctival closure was performed with 8-0 vicryl interrupted sutures. The same procedure was performed in the other eye. The inferior oblique muscle was noted to be inserted more posteriorly in BE.

On the 1st postoperative day, the alignment seemed to be good. On the 2nd postoperative day, PBCT showed a right esotropia of 20 PD with 12 PD of L/R with the child complaining of diplopia but significant improvement in head posture. His adduction and elevation improved to −2 BE. At 15 days and 1 month of follow-up, the child still had diplopia with 16 PD esotropia and 10 L/R for distance and near with minimal right head tilt. On the last follow-up 1 year postoperation, he had a significant improvement in the head posture with no diplopa.

On PBCT, he had a 20 PD esotropia with 15 PD right hypertropia [Figure 4].

**Discussion**

The EOMs are mesodermal in origin and start to develop at 3–4 weeks of gestation. The EOMs receive innervations from their respective nerves as early as 1 month of gestation.[1] Our case with bilateral absence of MR associated with brain anomalies could be a variant of CCDDs. The CCDDs represent static congenital abnormalities of eye movement with or without associated systemic abnormalities which may or may not be inherited, and there occurs some disturbance in the development of brainstem...
This group of disorders has received a lot of attention recently as there are several genes that cause individual clinical entities. Numerous causative genetic abnormalities have been identified which may involve exclusively the brainstem (ROBO3), the brainstem with both local and remote sequelae (HOXA1), the brainstem with predominantly lower motor neuron sequelae (PHOX2A), and lower motor neuron axonal guidance (KIF21A, TUBB3), sometimes with syndromic features (SALL4, CHN1, TUBB2B, and TUBB3). Every CCDDs gene characterized since 2002 has been associated with neuronal development at the nuclear, brainstem, or peripheral nerve level, supporting the hypothesis that CCDDs are neurogenic in origin.

In our case, a genetic analysis may help in identification of any gene abnormality. This may also support the hypothesis of a neurogenic origin of CCDDs, but the possibility of a syndrome complex also cannot be ruled out.

Again, the management of such complex cases is a real challenge. Partial- or full-tendon transposition procedures were introduced for the treatment of complex ocular motility abnormalities on the assumption that, they induced both, an active tonic force, by changing the duction force vectors and an increase in passive elastic forces, because of the longer route of the transposed muscles. A study by Snir et al. recommended combined medial vertical rectus muscle transposition with medial posterior fixation sutures for patients with adduction deficiency. Payse et al. described an augmented full-tendon transposition of the vertical rectus muscles to the MR muscle insertion in one patient with partial third nerve palsy. They noted a significant reduction in distance and near exo deviation from 60 to 70 PD preoperatively to 10 PD of intermittent exotropia at distance and 10–20 PD exotropia at near postoperatively. In our case, a LR recession alone would not have corrected the large exotropia, and a full-tendon transposition with LR recession had the possibility of producing anterior segment ischemia. Another modality is the injection of Botox to the tight LR with transposition, but this neurotoxin treatment cannot be expected to release the tightness associated with a chronic deviation caused by secondary structural changes in the muscle, conjunctiva, anterior Tenon’s capsule, and intermuscular membrane (posterior Tenon’s capsule). Botox alone cannot change the movement in the field of action of the missing muscle, and in addition, a repeat injection cannot be contemplated in this child, due to the complicacy of repeated general anesthesia. Hence, a partial-tendon transposition with posterior fixation was contemplated in our case along with LR recession. The child had diplopia and head posture in the first few postoperative visits which disappeared on the last visit, which could be attributed to a change in his vertical alignment. A similar case by Hill and Houtman et al. was described wherein they successfully treated the absent MR with vertical-tendon transposition.

We believe this case to be a variant of CCDDs or some syndrome complex requiring a genetic analysis. Surgical management of such complex cases could provide a cosmetic and psychological benefit to the patients.
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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Conflicts of interest
There are no conflicts of interest.

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