

Successful alignment following multiple surgeries in a child with severe esotropia and a congenital cranial dysinnervation disorder

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Abstract

Aim: To report the alignment, visual outcome and surgical strategy for severe bilateral congenital esotropia caused by a congenital cranial dysinnervation disorder (CCDD). This was successfully treated with a combination of interventions that included alternate occlusion, botulinum toxin, and conventional and augmented transposition procedures.

Methods: A girl presented at 2 months of age with a marked esotropia and severe bilateral impairment of abduction of both eyes, and an inability to fix with either eye in the primary position. Her investigation and treatment are described.

Results: The patient initially underwent alternate daily occlusion to prevent the development of amblyopia, followed by simultaneous bilateral medial rectus (MR) botulinum toxin and large bilateral MR recessions based on pre-operative forced duction testing (FDT). This operation was performed at 1 year of age with the aim of releasing the tight MR and reducing the esotropia. After initial improvement, the impairment of abduction gradually increased over the next month. She subsequently underwent sequential inferior oblique myectomy and augmented transposition procedures, which enabled her to fix in the primary position with either eye and with a small-angle esotropia.

Conclusion: Substantial improvement in the alignment of the eyes in a child with a complex congenital esotropia secondary to a CCDD is achievable.

Key words: Amblyopia, Botulinum toxin A, Congenital cranial dysinnervation syndrome, Congenital esotropia, Moebius syndrome, Muscle transposition surgery

Introduction

Congenital cranial dysinnervation disorders (CCDDs) are an uncommon, potentially visually disabling subgroup of strabismus conditions that 'arise from aberrant innervation of the ocular and facial musculature'. They generally have abnormal development of individual or multiple cranial nerve nuclei or their axonal connections.¹ The CCDDs share common clinical and/or pathophysiological findings including hypoplasia/

absence of one or more cranial nerves, synkinetic eye movements and restrictions or under-actions of one or more extraocular muscles,^{2,3} with genetic abnormalities in many.⁴ This group of strabismus disorders includes Duane's retraction syndrome (DRS), monocular elevation deficiency (MED), Moebius syndrome, congenital fibrosis syndrome of the extraocular muscles (CFEOMs),⁵ and some cases of congenital superior oblique palsy⁶ and of Brown's syndrome.⁷

These alignment abnormalities can compromise visual acuity and the field of binocular single vision, and result in persistently poor ocular alignment and potential secondary psychosocial symptoms. This case study describes the successful outcome in a child with severe bilateral congenital impairment of abduction with a management strategy that may be considered for similar motility scenarios.

Case report

Presentation

A 2-month-old girl was referred to the Ophthalmology Department from Neonatology at Sheffield Children's Hospital, with a severe esotropia and difficulties fixing and following since birth. She had bilateral talipes, mild scaphocephaly and there were concerns about her growth and development.

She was reviewed at the age of 3 months, when both eyes were found to be fixed in adduction, with the right eye being worse than the left, and both eyes were unable to move to the midline. Her fundus and media appeared normal. A cranial ultrasound scan was reported as normal, and a management decision was made to observe and commence daily alternate occlusion therapy to prevent amblyopia.

At the age of 7 months, visual acuity with both eyes open (BEO) was 6/48 with Cardiff Cards (CC) at 50 cm. The patient was adopting an abnormal head posture (AHP) in the form of a left to alternating marked face turn, with a marked right to alternating esotropia. Accurate measurement of the deviation was not possible, but the esotropia was estimated to be over 95^Δ (prism dioptres) on prism reflection testing (PRT) with split prisms. She was also suspected to have transient synkinetic vertical nystagmoid movements on attempted direct gaze. Daily alternate occlusion continued, and an MRI scan was performed to check for any significant brainstem or visual tract pathology. The brain MRI was reported as normal and the patient was scheduled to have surgery at the age of 1 year.

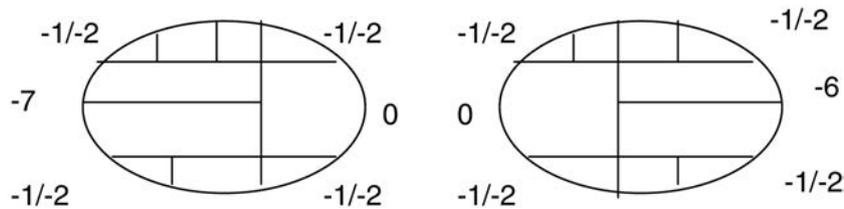


Fig. 1. Ocular motility on initial referral.



Fig. 2. Pre-operative photographs. (A) Without an abnormal head posture (AHP). (B) Using AHP face turn left to fix with the left eye. (C) Using AHP face turn right to fix with the right eye.



Fig. 3. Post-operative photographs. (A) Close-up photograph with mild AHP face turn left to fix with the left eye. (B) Using AHP face turn left to fix with the left eye. (C) Using AHP face turn right to fix with the right eye.

Initial pre-operative findings

Pre-operatively, the visual acuity was 6/30+1 in the right eye and 6/24 in the left eye (CC at 1 m). Ocular motility testing and restrictions are shown in Fig. 1. Measurement of the esotropia by PRT with split prisms revealed it to be approximately $120\text{--}130^\Delta$. The extent of the strabismus can be seen in Fig. 2.

First surgery intra-operative findings

At surgery forced duction testing (FDT) revealed a -4 to -5 restriction of the medial recti (MR) of both eyes. The vertical recti were not restricted. Both MR were injected with 4 units of Botox A (botulinum toxin), and recessed, being reattached directly to the sclera 12 mm from the limbus. FDT revealed no restriction of abduction following recession surgery.

Post-operative assessment

Post-operatively (2 days) the patient had a marked esotropia with a slight alternating hypertropia, but the angle of the esotropia had reduced to $70\text{--}80^\Delta$. This can be seen in Fig. 3.

The limitation of abduction improved to -4 to -5 in the right eye and -5 in the left eye. Ocular motility testing revealed a -1 under-action of adduction of both eyes, no evidence of anomalous innervation of either lateral rectus, an updrift on the left eye only, and it was queried whether elevation and depression were full.

One month after surgery the impaired abduction increased and was recorded as -5 to -6 in either eye. There was also an approximate -1 bilateral under-action of elevation and depression with bilateral updrifts on adduction of either eye.

Second surgery

The patient underwent a full tendon-width horizontal transposition of the vertical recti coupled with Foster sutures to the right eye and an ipsilateral inferior oblique myectomy, which was performed adjacent to the temporal border of the inferior rectus muscle at 14 months of age. At surgery the FDT to abduction and adduction were free bilaterally. The aim was to improve abduction, reduce the updrift on adduction, and allow the patient to fix in the primary position. Bilateral MR Botox



Fig. 4. A photograph taken in the primary position, after full tendon-width horizontal transposition of the vertical recti, coupled with Foster sutures to the right eye and ipsilateral inferior oblique myectomy.

injections were performed to temporarily weaken adduction to initially help maximise the transposition effect.

Post-operative findings after second surgery

Post-operatively the patient was able to fix in the primary position with the right eye and had a moderate left esotropia, with the left eye unable to take up central fixation. Ocular motility testing showed a -3.5 under-action of abduction and -1 under-action of elevation in the right eye, a bilateral -2 under-action of depression, and a $-5/-6$ under-action of abduction with an updrift on adduction of the left eye.

Post-operatively prism reflection measurement revealed a $45-50^\Delta$ esotropia. Fig. 4 is a post-operative photograph.

Third surgery

As the outcome for the right eye was successful, an identical operation was performed on the left eye 2 months later.

Post-operative assessment

Following left eye surgery, the patient had a slight left esotropia and a slight left hypertropia. This measured 10^Δ of esotropia and $8-10^\Delta$ hypertropia. Ocular motility had improved to a -2 under-action of abduction, -0.5 under-action of elevation and a -1 under-action of adduction in the right eye and a -3 under-action of abduction and a -0.5 under-action of adduction of the left eye. This resulted in a good cosmetic and functional result as it eliminated the abnormal head posture and substantially improved the patient's binocular field of vision. This can be seen in the final post-operative photograph (Fig. 5).

Long-term outcome

It is now 3 years since the patient's last operation and the primary position angle has fluctuated from a minimal esotropia to minimal exotropia and then back to minimal esotropia. However, the under-action of abduction has remained stable at -2 and the under-action of adduction has remained stable at -1 . The vertical deviation has changed slightly as the patient has developed asymmetrical, generally asymptomatic intermittent dissociated



Fig. 5. Post-operative photograph in the primary position after the final surgical procedure.

vertical deviation (DVD), larger in the right eye. Compliance with alternate daily occlusion has been variable. She developed mild left amblyopia, which was successfully treated with right part-time total occlusion. At the last follow-up visit the patient was still undergoing maintenance right part-time total occlusion.

Currently, she is under the care of a paediatric neurologist for stable mild facial weakness, neck flexion weakness, marked weakness of the lower and upper limbs due to an unclassified myopathy (confirmed by skeletal muscle biopsy) causing incomplete muscle dysgenesis and multiple contractures of the lower limbs. This is being managed by the orthopaedic team.

Discussion

This 3-year-old girl presented at 2 months of age with an inability to fix in the primary position due to marked bilateral limitation of abduction. The differential diagnosis included bilateral sixth cranial nerve palsy, infantile esotropia, congenital adherence syndrome, general fibrosis syndrome, or CCDDs such as Moebius syndrome, bilateral Duane's retraction syndrome (DRS) or bilateral radial ray Duane's retraction syndrome.

The diagnosis of a CCDD was made through the signs of nystagmus movements on attempted elevation, pre-operative FDT findings and the persistent but improved under-action of abduction after performing surgery to recess both medial recti. Initially, the findings were compatible with severe bilateral DRS type I or bilateral radial ray Duane's retraction syndrome. However, as the patient aged and became more co-operative a partial facial weakness started to become more apparent, raising the diagnosis of Moebius syndrome, especially in view of the lower limb abnormalities, a non-diagnostic skeletal muscle biopsy and very large angle esotropia. The 'classic clinical findings include: evidence of sixth and seventh cranial nerve involvement, often with associated malformations of limbs, craniofacial structures, and other cranial nerves'.⁸ This fitted with the presentation being from birth, as Moebius syndrome results from 'a cascade of secondary events after an embryonic insult from heterogeneous causes'.⁸ The signs of co-contraction of the extraocular muscles in this case can also be recognised in Moebius syndrome, where some forms result from developmental dysinnervation.² However, the patient also has abnormal muscles in her lower limbs with an 'unidentified myopathy'.

The literature describing the management outcomes of this condition, especially when there is a very large-angle complex congenital esotropia, is limited. Therefore, the surgical plan was based on the patient's deviation, ocular motility and FDT during surgery. The aim was to functionally and cosmetically reduce the AHP and the primary position deviation. According to Laby,⁹ no single surgical approach is successful in all patients and in many cases surgery will achieve optimal alignment in the primary position with a manifest esotropia persisting on lateral gaze. He suggests that a FDT must be performed on each eye to identify any tight muscles, with a staged surgical approach to maintain maximal control of each abducting procedure. After a case study of 3 patients with bilateral Moebius syndrome, Sun and Gole¹⁰ suggested medial rectus weakening by either chemodenervation or surgical recession for patients with medial rectus muscle contraction. However, in bilateral cases this is not as efficient an abducting procedure as augmented vertical rectus transpositions. Therefore, they suggested a staged medial rectus recession followed by an augmented vertical rectus transposition in cases with medial rectus recession, and an augmented vertical rectus transposition in cases without tight medial recti.

At the time of the initial surgical approach (bilateral MR Botox A and recession with reattachment 12 mm from the limbus), FDT was essential in view of the marked esotropia to identify any tight muscles.⁹ This revealed very tight medial recti muscles. This is not unusual in Moebius syndrome, in which fibrotic muscles are often found.³ In view of the marked esotropia the decision to perform therapeutic and diagnostic simultaneous bilateral medial rectus botulinum toxin injections and recessions was based on the presumed diagnosis of a CCDD. This initially resulted in a large correction of the esotropia from 120–130^Δ to 70–80^Δ and reduced the AHP, but confirmed a persistent innervational underaction of abduction bilaterally. Subsequently, the AHP, deviation and limitation of abduction increased post-operatively, consistent at least in part with the declining effect of Botox A on the medial recti with time. Therefore, further surgery was necessary to enhance the tonic abduction of each eye.

In the absence of obvious anomalous co-contraction of the lateral recti, transposition procedures were selected. This was done using the O'Connor approach, which involves a full-width transposition of the inferior rectus and superior rectus muscles adjacent to the lateral rectus muscle. Whilst this transposition procedure may be considered the least suitable of all transposition procedures, as it increases the risk of anterior segment ischaemia, reports have suggested this procedure produces a greater abducting effect than the Jensen procedure.^{11,12} It was augmented further by lateral rectus posterior fixation sutures. The addition of such lateral fixation sutures to full vertical recti muscle transpositions has been shown by Foster¹³ to improve the tonic abducting force, without compromising adduction in patients with lateral rectus palsy and type I Duane's syndrome. In selected patients with Duane's syndrome it has been reported to eliminate

the face turn in up to 80%.¹³ The fixation suture also stabilises the lateral rectus muscle and the risk of induced vertical deviation is uncommon.¹³ These procedures improved abduction without limiting adduction further, and in our patient resulted in a further reduction in the primary position esotropia from 45–50^Δ to 8–10^Δ. The horizontal deviation has remained stable to date. However, the patient has since developed DVD. This is common in patients with early-onset esotropia,¹⁴ and is likely to be a result of the age of onset of the deviation rather than being a feature of Moebius plus syndrome.

Conclusion

Following this three-step staged surgical plan the outcome in our patient with a complex, severe esotropia (restrictive and innervational) was functionally and cosmetically successful. There was a huge decrease in the size of the complex congenital esotropia from 120–130^Δ to 8–10^Δ, with satisfactory balanced horizontal deviations. Therefore, we would recommend this approach in other similar cases of large-angle complex esotropia without anomalous lateral rectus co-contraction. We advise that substantial improvement in the alignment of eyes with complex congenital esotropia is achievable in selected patients with CCDDs.

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