A Case of Duane's Retraction Syndrome and Synergistic Convergence

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ABSTRACT

Duane's retraction syndrome (DRS) is a congenital eye movement disorder characterised by a limitation of horizontal gaze and narrowing of the palpebral fissure with globe retraction in adduction. Synergistic convergence, on the other hand, is a rare variant of DRS defined by simultaneous bilateral adduction on attempted lateral gaze. We report the case of an 11-year-old girl who presents with type III DRS of her right eye and synergistic convergence of her left eye.

Keywords: Duane's retraction syndrome, synergistic convergence

INTRODUCTION

Duane's retraction syndrome (DRS), a mechanical ocular motility disorder, was classified by Huber into three categories based on the disordered horizontal motility and its corresponding electromyographic findings.[1] Type I is characterised by limitation of abduction, type II by limitation of adduction and type III by limitation of both abduction and adduction. All three types of DRS are associated with narrowing of the palpebral fissure and globe retraction on adduction, as well as the possibility of up and down shoots on adduction. Evidence suggests that DRS is caused by a misinnervation of the lateral rectus muscle by branches of the third cranial nerve instead of the sixth cranial nerve.[1] This misinnervation explains the oculomotor synkinesis noted on clinical examination.

Synergistic convergence is defined as simultaneous adduction, with an absence of pupil miosis on lateral gaze.[2] The condition is also associated with changes to the palpebral fissure and globe retraction. In the clinical setting, synergistic convergence can easily be misdiagnosed as DRS without careful examination of the eye movements and associated clinical signs. This paper discusses a child who presented at the age of two with signs of bilateral ocular synkinesis who was later diagnosed at the age of eleven with DRS of one eye and synergistic convergence of the other eye.

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Accepted for publication: 26th March 2018

CASE REPORT

Initial presentation

A two-year-old girl presented to a tertiary hospital in Melbourne with a suspected ocular motility disorder. At this initial consultation, a bilateral limitation of abduction and adduction was noted in addition to narrowing of the palpebral fissures on adduction of either eye. On observation, it was noted that the patient had adopted a slight compensatory chin up head posture and cover testing revealed a small intermittent right esotropia in primary position at near and distance fixation. Unaided visual acuity was documented as 3/12 and 3/6 for her right and left eye respectively on Kay Picture testing.

The patient's birth and medical history indicated that she was diagnosed prenatally with oesophageal atresia with trachea-oesophageal fistula, which was surgically repaired one day post birth. She was also later diagnosed at the age of six years with congenital dextroscoliosis for which treatment was not prescribed. The family had no history of significant medical, neurologic, or ophthalmic disease.

At this time the patient was diagnosed with a bilateral type III DRS and moderate right strabismic amblyopia. She received occlusion therapy for amblyopia from two to three years of age.

Follow-up presentation

At the most recent ocular examination, at age 11, the patient demonstrated best-corrected vision of right 3/3.8 and left 3/3 (-2), on the LogMAR chart. Her full refractive correction was right eye +4.50 DS and left eye +3.50 DS. A small esophoria with rapid recovery was detected in primary position, tested at both near and distance fixation, with and without correction. There was no obvious compensatory head posture when examined at the most recent consultation. The patient was able to achieve 55" stereoacuity with the Frisby Stereotest and 200" with the Lang Stereotest, both assessed with best correction.

Initially on ocular movements, limitation of both abduction and adduction was noted for both the right and left eyes. Globe retraction and narrowing of the palpebral fissure was noted on attempted abduction and adduction of either eye, left eye more than right. However, on carefully observing left gaze, simultaneous bilateral adduction was observed; that is the left eye was noted to converge on attempted abduction. Vertical gaze appeared to be intact. Figure 1 shows the eye movements in nine positions of gaze and the video (https://www.youtube.com/watch?v=hO6OB47o3RM) the eye movements on right and left gaze.

Pupils were equally reactive to light and to an accommodative stimulus with no evidence of pupil miosis on attempted lateral gaze. No fundus or media abnormalities were noted on ocular examination. On the basis of the follow-up findings, the patient's diagnosis was reclassified as a right type III Duane's retraction syndrome and left synergistic convergence.

Informed consent was obtained from the patient for publication of this case report.

DISCUSSION

Synergistic convergence is a rare form of oculomotor synkinesis characterised by bilateral adduction on lateral gaze.[3,4] Whilst there are several documented cases of synergistic convergence, to the authors' knowledge this is the first patient reported to exhibit an isolated DRS of one eye and isolated synergistic convergence of the other eye.

The main characteristic which facilitated the redefinition of this patient's diagnosis from DRS to synergistic convergence of the left eye was the bilateral adduction on left gaze. It is likely that the left eye movements were misinterpreted as DRS due to the similar clinical signs noted between the two conditions and the presence of right DRS. At the follow-up visit, substituted convergence was more easily excluded as a diagnosis given the absence of pupillary changes on attempted abduction. Substituted convergence is associated with bilateral adduction on lateral gaze and is distinguished from synergistic convergence by pupil miosis on attempted lateral gaze.[2] It has also been linked to patients who have cerebellar, posterior fossa or pontine lesions.[4]

Given the lack of pupillary involvement noted in synergistic convergence, it is suggested that it is most likely caused by peripheral mechanisms.[4] Peripheral causes noted to be

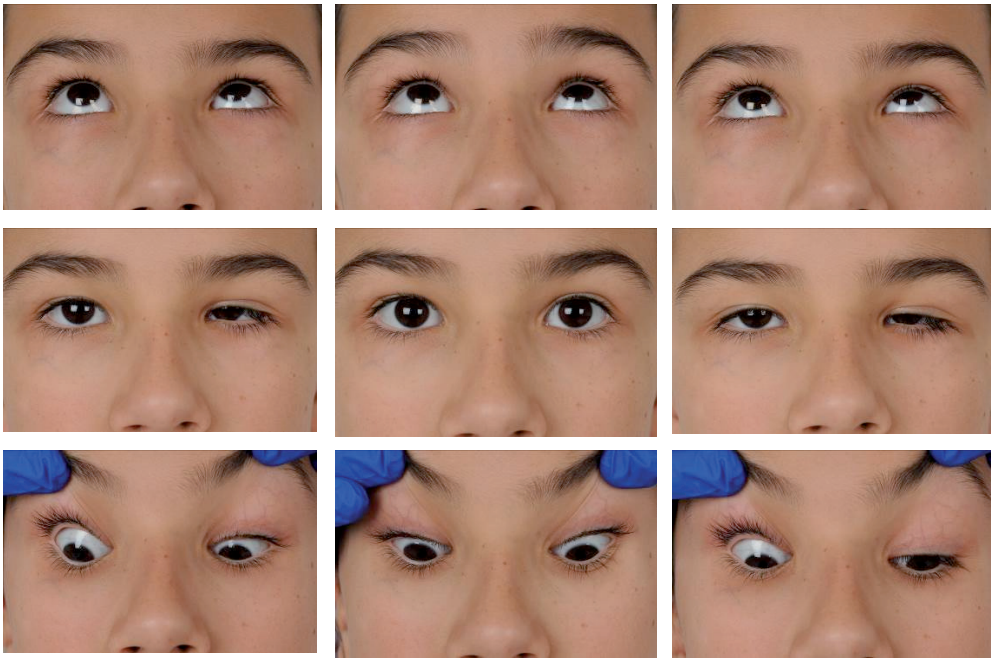

Figure 1. Eye movements in nine positions of gaze.

associated with the condition include congenital fibrosis of extra-ocular muscles (CFEOM), congenital cranial dysinnervation syndrome and ocular misinnervation.[5] Central causes, which are rarely involved, include progressive scoliosis and brainstem dysplasia.[5] According to Jain et al,[5] horizontal gaze palsy and progressive scoliosis (HGPPS) also shares a previously recognised association. In our patient's case, the presence of scoliosis and synergistic convergence could potentially be linked to an underlying congenital disorder of the pons, specifically congenital cleavage of the pons or brainstem dysfunction.[5,6] However, magnetic resonance imaging (MRI) was not performed so the presence of any cortical changes cannot be confirmed.

More recent research also suggests genetic mutations linked to synergistic convergence. For instance, a homozygous mutation in the gene ROB03 has been linked to progressive scoliosis and horizontal gaze palsies, which may suggest a similar mutation in this patient.[7] However, significant phenotypic variability can exist among individuals with identical genetic mutations, suggesting that other mechanisms could be involved in the clinical presentation.[8]

Finally, Pieh et al[3] have proposed that isolated synergistic convergence is associated with aberrant nerve sprouting during embryogenesis. When considering its counterpart, synergistic divergence, the suspected cause would be aberrant innervation of the oculomotor nerve fibres to the lateral rectus.[9,10] In this case, an assumed miswiring of the abducens motor neurons to the medial rectus muscle is more likely to be the cause. This is demonstrated by the ocular movements of the patient's left eye; adduction on attempted left lateral gaze, globe retraction and narrowing of the palpebral fissure on attempted abduction and adduction. However, as this is an exceptionally rare condition with variable associations, the current literature is unlikely to offer a definitive etiopathogensis.[2] This patient would require further testing to identify the most likely cause of the ocular motility disorder, such as MRI of the brain and orbits and genetic testing. In this case, it is noteworthy that the underlying cause of this ocular motility disorder is unlikely to influence the ongoing management of this patient's ocular condition. In primary position the patient is orthotropic and demonstrates binocular single vision. She also manages the limited eye movements in lateral gaze through the use of head movement and as such maintains good cosmesis, despite the ocular motility disorder.

CONCLUSION

This case study suggests that Duane's retraction syndrome and synergistic convergence can be diagnosed in a single patient. In the presence of DRS, it may be difficult to observe the nuanced differences between the two conditions, however careful inspection of the eye movements, the palpebral fissures, and pupillary reactions can distinguish synergistic convergence from DRS and substituted convergence. Whilst synergistic convergence is rare, clinicians should be mindful that a patient who presents with bilateral oculomotor synkinesis can have a principal diagnosis that differs between the two eyes.

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