An 'Atypical' Case of Vertical Retraction Syndrome in Association with Klippel-Feil Syndrome

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Abstract

A case is presented of a young child with an 'atypical' vertical retraction syndrome, demonstrated by limitation of ocular movement and lid retraction on depression without any associated globe retraction. The child has Goldenhar syndrome, including a Klippel-Feil anomaly, an external ear malformation and various other congenital anomalies. This case illustrates the wide range of associated anomalies that can present with problems such as Klippel-Feil disorder and Duane's retraction syndrome, suggesting a common teratogenic incident between the fourth and eighth weeks of gestation.

Key words:
Klippel-Feil syndrome, Goldenhar syndrome, Wildervanck syndrome, vertical retraction syndrome, Duane's retraction syndrome.

Klippel-Feil syndrome

Klippel-Feil syndrome was first reported by Klippel and Feil in 1912 [1,2]. The term is currently used to describe persons with congenital fusion of the cervical vertebrae, but this syndrome also has many associated signs. These include cervical problems such as a short neck, limited range of neck motion, low posterior hairline, webbing of the neck, elevation of the scapula, torticollis and scoliosis.[1-3] Both cranial and facial asymmetry may also occur, along with the facial anomalies of cleft lip and cleft palate; ocular anomalies such as coloboma and ptosis; and ear anomalies such as absence of the external auditory canal or ossicle malformation. There may be neurogenic problems such as deafness, facial nerve palsy, synkinesia, spasticity or flaccid paralysis and peripheral abnormalities of syndactyly or thumb hypoplasia. This syndrome may have multiple system involvement, with cardiovascular, renal, urogenital and pulmonary anomalies also reported.[1-3] The disorder is due to a failure of segmentation of somites of the cervical area during embryological development.[1,2]

The association of Klippel-Feil syndrome with Duane's retraction syndrome

With this wide range of associated problems there can be groupings of various anomalies, combining to form 'malformation syndromes'. Wildervanck syndrome (cervico-oculo-acoustic syndrome) describes a condition with the triad of Klippel-Feil anomaly, Duane's retraction syndrome and sensorineural deafness.[4,5] There have been various cases reported of Duane's retraction syndrome with Klippel-Feil anomaly[5-11] and various cases with Wildervanck syndrome have also been described.[5,6,12,13] Goldenhar syndrome (facio or oculo-auriculo-vertebral dysplasia) consists of a similar triad of signs including facial asymmetry, auricular malformations, epibulbar dermoids and vertebral anomalies.[3,5] Many cases of Goldenhar syndrome with Duane's retraction syndrome appear in the literature.[5,6,10,14-19] In a study of 186 cases with Duane's retraction syndrome, Pfaffenbach[5] reported that between 33% and 50% of the cases had one or more other congenital malformations.

Initially Duane's retraction syndrome was thought to be mechanical in origin, but the current theory is that in most instances it would be due to innervational anomalies, a misdirection of the nerve fibres during embryological development [8,12,20] or a spectrum of mechanical, anatomical and innervational factors.[21] Hoyt and Nachtigaller,[22] in a review of the literature, reported various anatomic anomalies, including absence of sixth nerve, extra branches of both the third and sixth nerves to other muscles and anastomoses of the third and sixth nerves.

Aetiology

Both Klippel-Feil disorder and Duane's retraction syndrome have been attributed to a disruption during embryological development.[2,5,7,8] The Klippel-Feil disorder shows a failure of segmentation of the somites of the cervical area. As the vertebrae of the neck are properly segmented in the Klippel-Feil Syndrome, then the teratogenic effect must occur later than the fourth week, when primary segmentation into the caudal and cephalic halves of the body has occurred.[1] At around the same time, the extraocular muscles are developing from three paired, separate masses of mesoderm, each associated with a cranial nerve. Initially, at 26 days, the four oculomotor muscles appear as one mass, then the lateral rectus develops at 27 days and the superior oblique at 29 days. The ocular motor nerves grow from the brain into the muscle masses slightly later, reaching the mesodermal connections at 31 days for the IIIrd and IVth nerves, and at 33 days for the VIth nerve. The muscle cone is in the posterior pole at 5 weeks and the muscles grow forwards towards the globe, fusing at their insertions near the end of the third month. The levator begins to separate from the superior rectus at around 45 to 55 days and is complete during the fourth month.[23,24]

The combination of malformations, ocular, acoustic, auricular, vertebral and other organ systems indicates a disturbance between the fourth and eighth weeks of gestation at the time of development of the ocular muscles and nerves, vertebral segmentation, external ear structures and the formation of organs.[5,6,22,25]

Case details

SM, a 10 month old boy, diagnosed with Goldenhar syndrome, has Klippel-Feil syndrome with the following associated signs:

- fusion of cervical vertebrae
- short webbed neck
- restricted neck motion
- right shoulder higher than left
- left facial hypoplasia, face and jaw
- left external ear malformation
- slight conduction loss in left ear
- hypertonia of upper limbs
- short left leg
- developmental delay
- poor growth
- pelvic kidney
- small muscular ventricular septal defect

Orthoptic assessment

On observation, his left palpebral fissure was wider than the right, with the appearance of left lid retraction. Orthoptic assessment showed the following results:

- Cover Test
  small L hypertropia
- Ocular Movements
  - limitation of left depression in all depressed positions of gaze, only minimal movement below the midline
  - widening of left palpebral fissure on depression
  - elevation of the left eye full, with minimal L/R in elevation
  - horizontal movements, full
  - no apparent globe retraction on depression or elevation

Vertical retraction syndrome

It appears that this child has some form of vertical retraction syndrome in association with the Klippel-Feil syndrome. Vertical retraction syndrome, a much rarer congenital condition than Duane's retraction syndrome, has been variously described in the literature as a limitation of either elevation or depression [26,27] or a limitation of both elevation and depression.[28,29] The retraction of the globe and narrowing of the palpebral fissure may not be as obvious as in Duane's retraction syndrome [28] and again there are opposing descriptions of the retraction, some authors stating that retraction may be seen on either elevation or depression,[27,28] others that retraction may only be evident on depression.[26,29] There may be either orthophoria, hypertropia, hypotropia or an associated horizontal deviation in the primary position.[21,26,28]

Two cases of bilateral vertical retraction syndrome in siblings were reported by

Khodadoust and von Noorden.[30] It was suggested in view of the positive results to forced duction tests, that both of these cases were due to structural rather than neurogenic anomalies, but as electromyography was not performed, paradoxical innervation could not be excluded as the primary cause. Other than a slight ptosis in one case, there were no detectable anomalies of the levator muscle, the changes in lid position occurring with retraction of the globe. The pathogenesis of vertical retraction syndrome is thought to be similar to that of Duane's retraction syndrome.[21]

'Atypical' Vertical Retraction Syndrome

Three cases have been reported of 'atypical' vertical retraction syndromes, the common feature of which appears to be lid retraction on depression.[31-33]

In one case [32] it appears that the levator fires simultaneously with both the superior oblique and the medial rectus, as there is lid retraction of the right eye on both laevodepression and laevoversion, with no muscle underactions. In a second case[33] there appears to be paradoxical innervation involving the levator and the inferior rectus as there is limitation of movement, lid retraction and slight proptosis of the left eye on depression and laevodepression. In a third case[31] the ocular movements appear to be a combination of the 'typical' vertical retraction syndrome (demonstrated by limited elevation of the right eye, limited adduction with downshoot and globe retraction, widening of the palpebral fissure on abduction and a positive forced duction test) and the 'atypical' syndrome which includes the characteristic of lid widening on depression. In this case, electromyographic results showed anomalous innervation of the vertical recti, but the authors were unable to explain the lid widening on depression in the presence of globe retraction.

It seems that the 'typical' vertical retraction syndrome involves some limitation of vertical movement associated with retraction of the globe and narrowing of the palpebral fissure, in a similar manner to the horizontal Duane's retraction syndrome. The two reported cases appear to have structural anomalies, evidenced by forced duction testing. The three reports of 'atypical' vertical retraction syndrome each appear to have the common factor of lid retraction on depression, indicating a paradoxical innervational anomaly between the levator and one or more of the extraocular muscles, and therefore may be somewhat different from the groups of vertical retraction syndrome or Duane's retraction syndrome.

Discussion and conclusion

The ocular motor results indicate that SM may have what others have termed an 'atypical' vertical retraction syndrome,[31-33] with anomalous innervation involving the levator and both the depressor muscles, as the limitation of depression is symmetrical across all depressed positions.

He has been given a diagnosis of Goldenhar syndrome, which includes Klippel-Feil disorder and the left external ear malformation, but there is no evidence of epibulbar dermoids. However, Baum[14] in a review of the literature, found that 24% of the reported cases with Goldenhar syndrome had no epibulbar dermoids. It could also be suggested that SM has an unusual variant of a Wildervanck syndrome, as he has Klippel-Feil disorder, some hearing loss and a variant of a retraction syndrome. It has been stated that a diagnosis of Wildervanck syndrome is still accepted in the absence of one of the three features.[5,6] DeRespinis[21] stated that Wildervanck and Goldenhar syndrome overlap, as can be seen with the multiple cases of Duane's retraction syndrome associated with Goldenhar syndrome.

SM presents as an infant with Goldenhar syndrome, an 'atypical' vertical retraction syndrome with anomalous innervation, a Klippel-Feil disorder and multiple congenital anomalies, presumably due to a common teratogenic effect between the fourth and eighth weeks of gestation. This association of anomalies, including skeletal, auricular, ocular, neural and multi-system involvement illustrates the importance of a general physical examination in each case of Duane's retraction syndrome as suggested by many other authors.[5,6,11,15,18]

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