Incomplete split cord malformation presenting with unilateral vocal cord palsy and torticollis in a newborn

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ABSTRACT

SCMs involving the cervical cord is therefore largely unknown. In addition, the concomitant finding of brainstem involvement is presumably incompatible with life in the majority of patients, resulting in a paucity of data regarding this clinical scenario.

To report a rare case of SCM involving the brainstem, and discuss the clinical impact, diagnosis, and management.

INTRODUCTION & METHODS

Split cord malformation (SCM) is a rare abnormality of notochord development. The majority of cases occur in the thoracolumbar region,1 with just over thirty cases of cervical SCM reported.2 The clinical impact of SCMs involving the cervical cord is therefore largely unknown. In addition, the concomitant finding of brainstem involvement is presumably incompatible with life in the majority of patients, resulting in a paucity of data regarding this clinical scenario.

We present the first case, to our knowledge, of an incomplete cervical SCM involving the brainstem, and discuss the clinical impact, diagnosis, and management.

REVIEW OF CASE

Birth History:
• Born at 38 weeks with no perinatal complications.
• Parents noted apneic episodes, progressive lethargy, difficulty feeding and noisy breathing during sleep.
• Day seven of life: Patient became cyanotic and required cardiolunghropulmonary resuscitation by a family member.
• Transferred to our institution after a medically witnessed apneic episode associated with biphasic stridor.

Workup:
• On arrival, hemodynamically stable on continuous positive pressure support, with no obvious dysmorphic features.
• Isolated capillary hemangioma noted on arm, and unilateral left sided torticollis appreciated (Figure 1).
• Flexible endoscopy revealed left vocal cord paresis, which was confirmed during flexible state endoscopy, with an otherwise normal airway examination (Figure 2).
• Immature suck/swallow/breath sequence diagnosed via functional endoscopic examination of swallowing.
• Frank aspiration noted on video swallow and central apnea noted on polysomnogram.
• Chromosomal microarray normal, and infectious workup was negative.

Imaging Studies:
• Magnetic resonance imaging of the brain and spine revealed a 6 mm long cervical SCM with no evidence of syringohydromyelia or cord compression (Figure 3-4).
• Also notable was hypoplasia of the left dorsolateral medulla.

Management:
• No evidence of spinal cord tethering, and therefore conservative neurosurgical management was recommended.
• After demonstrating weight-gain, an improved suck/swallow/breath sequence, and a decreased number of apneic events over two weeks, patient was discharged on room air with overnight oximetry.

6 Week Follow Up:
• Decreased apneas on video swallow, but persistent laryngeal penetration on functional examination of swallowing.
• Unilateral vocal cord paresis and torticollis were unchanged, however her parents reported decreased apneas and noisy breathing.

REFERENCES


DISCUSSION & CONCLUSION

When used to categorize spinal malformations, Tortori-Donati et al describe open (OSD) or closed (CSD) spinal dysraphism, depending on whether the defect is covered by skin and/or bone, or is open to the external environment.1 Pang et al classically divided SCMs in two subtypes.2 In SCM Type 1, or diastematomyelia, two distinct hemisegments are separated by a bony septum, each contained within its own dural sheath.2 In contrast, Type II SCM, or diplomyelia, results in two hemisegments separated by a fibrous septum contained within a common dural sheath.2

Embryologically, in patients with SCM, the formation of the neural plate is interrupted by the presence of an anomalous communication between the notochord and the neurorachis, termed the accessory notochordal canal.4 The primitive streak tissue continues to develop around this canal and condenses to form an endodermalenchymal channel.2 If this tract develops into bone and cartilage, a Type I SCM will form, however, if the tract forms a fibrous septum, a Type II SCM results.2

The hemisegments grow caudally based on the development of their associated neural placode, found in all OSD and most CSD cases.1 Importantly, these placodes can be defined either as terminal or segmental, depending on the final location.1 Caudal to a segmental placode, the spinal cord appears normal both morphologically and structurally.1 Patients may also have an associated meningocele manqué, which refers to the formation of tethering meninges and associated meningocele manqué, which refers to the formation of tethering meninges and associated meningocele manqué, which refers to the formation of tethering meninges and surrounding tissues, and may cause tethered cord syndrome (TCS).

We hypothesize that the basic ontogenetic defect in our case resulted in malformation of the lateral midbrain and formation of a fibrous septum terminating in an aberrant segmental neural placode at the CI/C2 level, with a normal single cord caudal to this lesion. Thus suggesting an incomplete Type II cervical SCM possibly associated with an asymptomatic meningocele manqué.

Due to the rarity of cervical SCMs, their clinical impact is not well defined. Andro presented a case of cervical SCM with associated vertebral anomalies, resulting in surprisingly mild symptoms.2 In contrast, our patient has a subtle cervical SCM, which produced profound, life-threatening results. This suggests that the anatomic location of the derangement drives both clinical impact and prognosis.

Magnetic resonance imaging (MRI) offers the most detailed instrument for evaluating SCMs.6 The key radiologic findings in Type II SCMs are a single dural sac containing two hemisegments, best seen on T2-weighted or FIESTA weighted MRI, or the lack of a bony spur noted on T1-weighted imaging. In addition, evaluation of the thin fibrous septum may be possible with T2-weight or FIESTA weighted MRI. Notably, meningocele manqué, is extremely difficult to diagnose on MRI alone, and often relies on surgical evaluation for definitive diagnosis.

The unilateral vocal cord paresis and congenital torticollis noted in our patient likely resulted from involvement of the high cervical cord and ventrolateral caudal medulla. Vocal cord paralysis is the second most common cause of stridor in newborns, with an equal distribution between unilateral and bilateral paralysis.6 We hypothesize that the SCM and brainstem anomaly in our patient impacted the vagal nuclei resulting in unilateral cord paresis. While there are several surgical interventions available for vocal cord paralysis, including tracheostomy, arytenoidecotomy and cord mobilization, our patient remained hemodynamically stable and showed steady improvement with conservative management.

The rapid improvement of swallowing and aspiration during the first month of life is likely due to improved apnea of prematurity and her suck-swallow-breath reflex.6 The improved swallow study at day ten of life, in combination with her persistent cord weakness, supports this hypothesis, and she will be managed conservatively with compensatory feeding techniques.6

The torticollis noted in our patient is likely a direct complication of her high cervical SCM and brainstem hypoplasia and will be initially managed with physical therapy and stretching exercises.7

Management depends largely on neurologic sequelae, and is often dictated by the development of tethered cord syndrome (TCS). Surgical intervention to release the tethered segment is often recommended, however, surgery should be individualized to the patient and their risk of developing sequelae from the lesion. Our patient will be followed closely for signs of TCS and managed accordingly.