Unusual Variants of Spina Bifida - Four Additional Cases

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Abstract

Four different atypical cases of spina bifida are reported from a tertiary institution in Kenya. These include a myelomeningocele associated with a vestigial appendage, a paravertebral myelomeningocele, a tandem lesion, and a hamartoma within a myelomeningocele. A discussion follows on the pathogenesis of human tails, multi-site spinal dysraphism, paravertebral spina bifida and hamartoma association.

Index Words: Spina bifida, human tail, tandem lesions, hamartoma.

Introduction

Congenital craniospinal malformations, dysraphic malformations or neural tube defects (NTDs) can be classified in two general classes: aperta or cystica (open) and occult forms. Aperta lesions are the result of a localized failure of the primary neurulation and are represented by defects such as anencephaly, myelomeningocele, and meningocele[1].

Occult lesions might be more difficult to diagnose as their anatomic defect is more subtle and patients may not have neurological impairment. Spinal lipoma, split cord malformations, neurenteric cysts, dermal sinus tracts, myelocystocele, thickened filum terminale, and caudal agenesis are some examples[2].

The usual forms of spina bifida have been well described in the medical literature. However, atypical lesions are rare and still not well understood. We report four cases of atypical spina bifida: a human tail, a tandem lesion, a case of paravertebral spinal dysraphism, and a myelomingocele associated with a hamartoma.

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Case 1

A one day-old baby girl presented with a lumbosacral skin appendage. The term baby was born by spontaneous vaginal delivery (SVD) following an uncomplicated pregnancy. Birth weight was 3.0 kg and physical exam revealed a fluid-filled tail-like cutaneous structure measuring 15 cm x 7 cm in the lumbosacral area (Fig. 1). A lumbar bony defect was palpable through his skin. Neurological exam revealed normal tone and strength in the lower extremities and a left brachial plexus injury. Operative findings included a lumbar meningocele in conjunction with the skin appendage. The skin appendage was resected and the meningocele closed. Physiotherapy was started

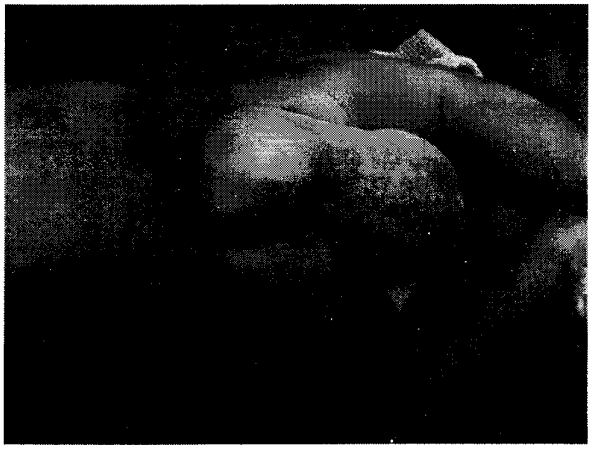

Fig. 1: Meningocele associated with vestigial tail

for the brachial plexus injury. Routine post-operative bladder evaluation revealed a neurogenic bladder which prompted the initiation of clean intermittent catheterization (CIC) by the parent. The patient was discharged on the seventh post-operative day and made an uneventful recovery.

Case 2

A healthy seven week-old baby girl was admitted to our service with two congenital spinal lesions in the thoracic and sacral region (T5 & L1)

Fig. 2: Tandem spina bifida lesions

(Fig. 2). Physical exam was otherwise normal and the patient had normal lower extremity motion and no evidence of hydrocephalus. Operative findings showed a sacral meningocele and a thoracic superficial spina bifida occulta lesion. The sacral lesion was repaired with a standard dural closure, and the thoracic lesion repaired with subcutaneous closure only. Post-operative bladder evaluation was normal. The patient was discharged home on the fourth post-operative day following an uneventful recovery.

Case 3

A 1 month-old baby was brought to our emergency with a large flank mass since birth. The baby was born at term after an uncomplicated SVD. Physical examination revealed a soft skin-covered mass 15x10cm seemingly originating from the right

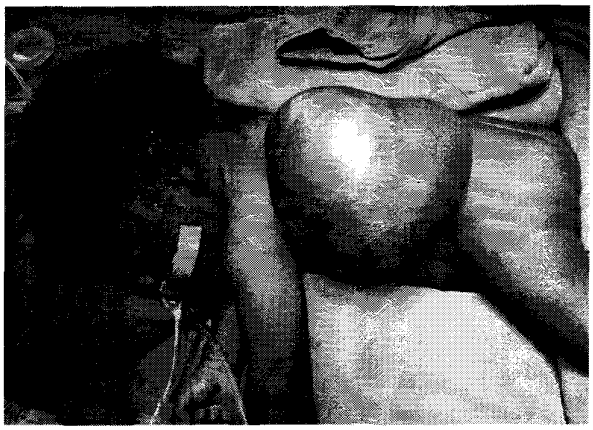

Fig. 3: Paravertebral myelomeningocele

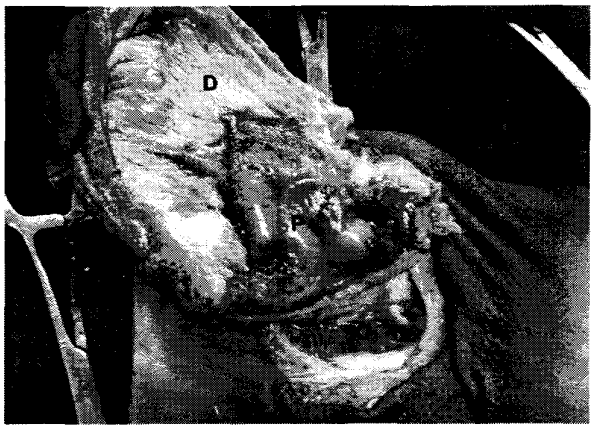

Fig. 4: Paravertebral myelomeningocele - intra-operative view. P = placode; D = dura; O = origin of sac from paravertebral site

lumbar paravertebral area (Fig. 3). There was also dextroscoliosis and a left inguinal hernia, but normal lower extremity function. The initial differential diagnosis included a NTD, lymphangioma and teratoma. At operation the mass was found to be CSF-filled and originate from the immediate paravertebral area. The sac contained neural tissue, with the placode presenting as a sliding component of the dural sac (Fig. 4). Dural closure was obtained with inversion of the placode, and the inguinal hernia repaired. The child had pneumonia post-operatively as well as a contained CSF leak which resolved after one puncture. He remains in follow-up for his scoliosis.

Case 4

A 2 week-old baby boy was referred from a

Fig. 5: Meningocele associated with hamartoma (H).

district hospital with multiple congenital abnormalities. The maternal history was uneventful and the baby born at term by uncomplicated SVD. Physical exam showed hydrocephalus, clef lip, a large thoracic myelomeningocele adjacent to a dorsal midline soft mass, left lumbar hernia, and flaccid lower extremities with bilateral club feet. Abdominal ultrasound showed normal kidneys. A lumbar x-ray demonstrated thoracic lordosis with an ectopic bone. Operative findings revealed a thoracic bony diastomyelia-like growth proximal to the myelomeningocele lesion, which merged into an irregular soft tissue mass (Fig. 5). The mass was excised, dura closed, and skin defect closed with a latissimus muscle flap. On the second post-operative day the patient developed aspiration pneumonia which progressed despite treatment to a fatal cardiorespiratory arrest two days later. Pathological examination concluded that the specimen contained bone, cartilage, muscle and fat in keeping with the diagnosis of hamartoma.

Discussion

As per Frank et al[3], there were 59 cases of human tail or lumbosacrococcygeal appendage reported in the literature from 1960 to 1997. During the fifth and sixth weeks of gestation, the embryo develops a caudal appendage containing a vertebrated portion that will retract during the seventh and the eighth week of gestation and a nonvertebrated portion which will undergo phagocytosis (? apoptosis) at the end of the eighth week.

In 1994, Dao and Netsky suggested a distinction between true tails and pseudotails[4]. A true tail is usually considered as a remnant of the unvertebrated mesodermal portion of the embryonic appendage. A pseudotail is described as an abnormal extension of coccygeal vertebrae, lipoma, teratoma, chondromegaly and glioma[5]. However, this classification is not widely accepted as it encounters problems in its clinical usefulness.

In addition, with more detailed studies available, more cases are now labeled as pseudotails associated with spinal dysraphism. In the review by Frank et al[3], this association accounted for 49% of the cases. Therefore, they proposed a new classification based on the tail being associated with spinal dysraphism or not, based on the finding that the occurrence of a tethered cord syndrome was the key prognostic factor. The authors even suggested that a pseudotail has no relationship to the true tail and would be the result of an abnormal tissue incidentally placed in the lumbosacroccygeal area[3]. Neurulation is the process that allows the neural plate to become the neural tube. The neural plate initially forms a groove, which then fuses to become a tube. It was originally thought that the neural groove was initiating its fusion from an upper-dorsal location and extending simultaneously towards its rostral and caudal ends. Hence, a rostral defect would lead to anencephaly and a caudal defect to spinal dysraphism. This theory was questioned with the multi-site neural tube defects reported in the medical literature.

In 1993, Allen Van published his theory on the evidence for multi-site closure of the neural tube in humans. He demonstrated that there are four sites of closure in mice, and probably five in humans. He also advocated for classification of NTDs according to their closure site defect as many syndromes could be explained by the combinations found in these

defect locations[6].

Juriloff and Harris also argued that most NTDs reflect a breakdown of the neural fold elevation in systematically specific zones that would prevent a normal closure[7]. They demonstrated that the specificity of NTD types is caused by distinct mutations in mice. If the problem lies in the neural tube elevation process, we have yet to define the molecular mechanisms that allow these disruptions. Without scientific evidence, one may question whether paravertebral NTDs are not caused by unilateral defects in the neural tube elevation process. Rare cases of thoracic and lumbar lateral meningoceles have been reported[8], often associated with neurofibromatosis[9]. Any paravertebral cutaneous lesion should also be suspected in patients with lateral dermal sinus tracts[10].

Hamartomas are focal malformations composed of heterogenic well-developed tissues such as bone, cartilage, fat, muscle, nervous and fibrous tissue. Although uncommon, congenital midline hamartomas of the spine are a recognized clinical entity. They are distinct from spinal dysraphism as they do not result from of a neurological malformation. However, their close relationship to the spinal cord may lead to insidious neurological impairments from mass effect or tethered cord. Hamartomas are clearly divergent from teratomas as they do not own the potential for uncontrolled cell proliferation[11]. There are a few reports of hamartomas in association with meningoceles[12]. In our series, we present a case of a hamartoma associated with a myelomeningocele, which, to our knowledge, is the first case of this particular association in the literature.

We like to conclude that despite their rarity, a wide variety of atypical spina bifida lesions exist. Human tails have a high correlation with spinal dysraphism and should prompt thorough imaging studies. Multi-site lesions have been linked to specific genetic mutations, and are the basis for the popular multi-site neural tube closure theory for the pathogenesis of NTDs. Although rare and poorly understood, paravertebral spina bifida appears to be frequently associated to neurofibromatosis. Hamartoma is a relatively benign clinical entity which hitherto has not been reported in association with a myelomeningocele.

Awareness of these unusual forms of NTDs can help in their correct diagnosis and appropriate treatment.

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