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Case Report Open Access

Spinal Cord Injury Secondary to Hereditary Spinal Arachnoid Cysts

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Abstract

Spinal arachnoid cysts are benign and uncommon in children. The etiology may be congenital or traumatic. Despite the development of magnetic resonance in recent years, its findings are generally accidental. The most common symptoms are pain and progressive paraparesis, asymmetrical. Its appearance may be part of lymphedema distichiasis syndrome to be motivated by mutations in the FOXC2 gene transmitted as an autosomal dominant.

A case of a girl with D3 ASIA C paraplegia is reported. The spine magnetic resonance revealed two posterior epidural spinal arachnoid cysts from D1-D2 to D8-D9 and from D12 to L2 and D6 and D8 myelopathy. Rehabilitation treatment program as well as surgical to evacuation of the cysts.

Reviewing the clinical history, by the father, grandmother and cousin had spinal arachnoid cysts and lymphedema distichiasis syndrome respectively. A genetic study of girl, her brother and father confirms the FOXC2 gene mutation.

Keywords: Spinal cord injuries; Hereditary spinal arachnoid cysts; Lymphedema distichiasis syndrome

Introduction

Spinal extradural arachnoid cysts are rare lesions in children1. Although their finding is generally accidental, because rarely cause a spinal compression, the incidence has increased due to the development of Magnetic Resonance Imaging (MRI) [1,2]. They usually are solitary lesions located in the dorsal region of the spinal cord [1,2]. Multiple Spinal Arachnoid Cysts (SAC) occur between the ages of 9 and 14 and their location varies from the posterolateral region of the cervical spinal cord to the sacral, displacing it to the front [1-6].

Nabors et al. described the classification of SAC dividing into 3 main categories [1-6].

1) Extradural cysts without nerve fibers (Type I). In turn subdivided into:

- Extradural arachnoid cysts (Type IA)
- Sacral meningoceles (Type IB)

2) Extradural cysts with nerve fibers (Type II)

3) Intradural cysts (Type III)

Although most cases are thought to be congenital, extradural SAC may be secondary to inflammation, trauma or iatrogenic [1,2,4].

The clinical presentation may also vary depending on the location and severity of spinal cord compression [3]. The cardinal symptom is spastic paraparesis [3]. Pain is rare in the thoracic cysts but not in cervical, lumbar and sacral [3]. The sensory loss is unusual even within its proprioception is most affected [3]. The detrusor-sphincter dysfunction is uncommon [3]. Nevertheless, some patients present with symptoms of acute compression [3].

Treatment of extradural SAC is complete resection with an excellent prognosis, regardless of the degree of the size of the cyst [1,2,4].

The appearance of these cysts may form part of Lymphedema Distichiasis Syndrome (LDS), rare and first described in 1964 by Falls and Kenesz, whose molecular basis is known, being due to mutations in the FOXC2 gene (16q24.3) transmitted as an Autosomal Dominant (AD) with high degree of penetrance and variable expressivity. We also found cases of spontaneous genetic mutation [5,7-10].

We report the case of a girl with D3 ASIA C paraplegia secondary to dorsal SAC. In her family inherited syndrome is diagnosed.

Case Report

12 years old girl with dorsal kyphosis. She has progressive difficulty walking with frequent falls. It makes a total column MRI describing two posterior SAC from D1-D2 to D8-D9 (16 cm cranio-caudal) and from D12 to L2 (7 cm cranio-caudal) with myelopathy D6 and D8.

Referred to the Spinal Cord Injury Unit, in the physical examination objective lymphedema of the lower limbs and symmetrical spastic D3 ASIA C paraplegia with severe proprioceptive affectation. Rehabilitation treatment programmed and after evaluation by the Neurosurgery Service, performing surgery: D5-D7 and D12-L1 laminectomy and evacuation of the two arachnoid cysts.

In the postoperative period, and due to the severe spasticity, it was decided to start drug treatment with oral baclofen at doses of 5 mg every 24 hours.

At that time, the family reports that the paternal grandmother was taking the same medication to also present SAC. Besides the father and paternal cousin also had asymptomatic SAC, lower extremity lymphedema and distichiasis, and our patient's brother had distichiasis.

Genetic testing confirming FOXC2 gene mutation on 16 chromosome in family members.

She continued physical and occupational therapy getting home walk with the help of an AFO in left leg and two crutches.

3 months later suffers a neurological worsening being unable to ambulate. Total column MRI is made reporting dural fistula and D3-D9 cyst recurrence. She is surgically reoperated removing the cyst and closing the fistulous communication.

After surgery, rehabilitation treatment is continued with full resolution of neurological deficit (D3 ASIA E paraplegia).

In subsequent revisions progression of kyphosis is seen for what is derived to Trauma Service initially decided expectantly observed but in the progression of angulation decides to intervene performing arthrodesis T1-L1. Today, the girl remains asymptomatic (Figures 1-3).

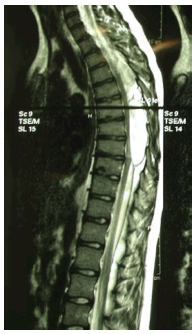

Figure 1: MRI describing posterior SAC.

Discussion

The SAC are a rare cause of spinal cord compression [3]. There are considered a diverticulum of arachnoid or dural sac [1,2,4]. As described above, Nabors et al simplified classification dividing in extradural and intradural, with and without the presence of neural tissue [1,2,4]. The extradural SAC are in communication with the subarachnoid space through a dural defect, which makes the content is cerebrospinal fluid (CSF) [1,2,4]. Type I cysts are found throughout the spinal canal, preferably at the thoracic level, while type II is more common sacral level [1,2,4,6]. Found predominantly in the posterolateral region of the spinal cord, moving above [1,2,4] Considering the anatomy of the thoracic spinal canal, this is relatively smaller in diameter than at other levels, for which the cysts in this area typically have greater clinical expression [1,2,4,6].

Predominate in males and the peak incidence occurs in the second decade of life for type I and adult life for type II [1,2,4].

Although the vast majority are asymptomatic, when given symptoms most characteristic is the pain at level of the lesion, in cervical, lumbar and sacral region cyst, and progressive spastic or flaccid paraplegia, often symmetrical; usually related to spinal cord compression [3]. Symptoms tend to fluctuate, with remissions and exacerbations3. Valsalva maneuvers could increase the volume of the cysts [3].

In our case, the most striking symptom was symmetric spastic paraplegia and severe impairment of proprioceptive sensitivity, made to orient diagnosis.

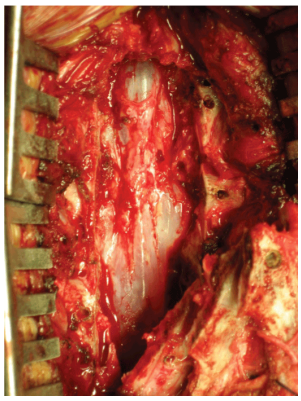

Figure 2: Macroscopic dorsal SAC image before opening the dura.

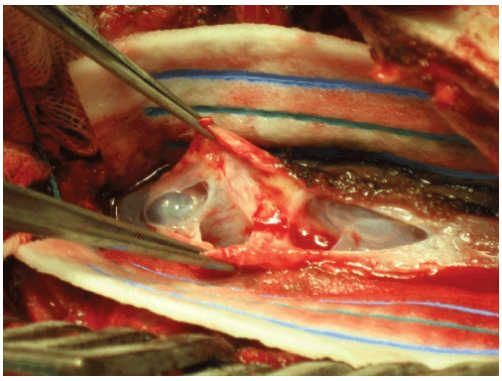

Figure 3: Macroscopic dorsal SAC image after opening the dura.

MRI is the best diagnostic method for its high accuracy for the exact location of these lesions, its extent and its relationship to the spinal cord [1,2,6]. Besides being a non-invasive method, allows us to visualize CSF flow characteristics, compressive myelopathy and associated injuries [1,2,6].

MRI revealed two extradural multiloculated SAC in the thoracic level D1-D9 and D12-L2 in our case. The spinal cord was compressed and displaced toward the front with hyperintense area in D6-D8. The two loculations were homogeneous and showed low signal intensity on T1-weighted images and high signal intensity on T2.

With respect to the treatment of these injuries most authors agree that it is a benign disease, only be surgically stopped symptomatic

lesions, whereas asymptomatic cysts can be managed conservatively with regular checks of MRI [1,2,6] (Figures 4,5).

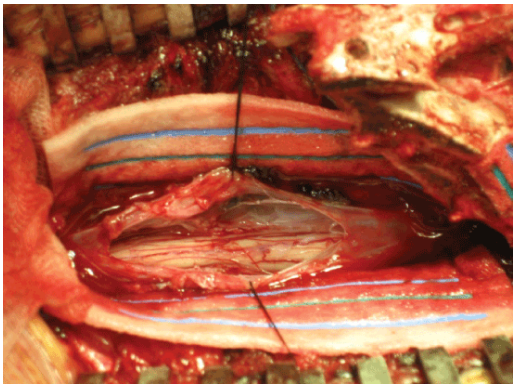

Figure 4: Macroscopic dorsal SAC image after opening the dura.

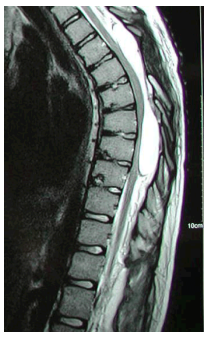

Figure 5: 3 months later MRI reporting D3-D9 cyst recurrence.

The goal of surgical treatment must be the neural decompression and cyst reproduction prevention, making complete removal of it and close the communication between the cyst and the subarachnoid space is the treatment of choice [1,2,4,6].

In our patient evacuated the two cysts with good clinical initial evolution, but a neurological worsening, which guided a radiologically confirmed recurrence, so the reoperation, in addition to the evacuation of the cyst proceeded to repair the observed fistula and to close the defect in the dura.

The appearance of SAC may be part of LDS, rare and due to mutations in the FOXC2 gene on chromosome 16 (16q24.3) [5,7-10]. Despite having found cases of spontaneous genetic mutation, most are AD transmitted with great degree of penetrance and variable expressivity [5,7-10].

Distichiasis is present at birth and affects both upper and lower eyelids, and lower extremity lymphedema, which occurs in most cases in the second decade of life, is located primarily below the knees, unilateral or bilateral [5,7-10]. Leg edema in years may precede the other [5,7-10].

Its phenotype is variable; there have been reports of mutations in some studied families, some only present lymphedema and others distichiasis [5,7-10]. Other malformations associated with this syndrome include early onset varicose veins, heart defects, unilateral or bilateral ptosis, ectropion in lower eyelid, cleft palate, pterygium colli, bifid uvula and widening of the spinal canal with extradural SAC associated with neurological disorders [5,7-10].

In our case, the patient had an incomplete lower extremity lymphedema syndrome and SAC with a hereditary paternal line; in all cases confirmed the genetic mutation.

Conclusion

The extradural type I SAC are located in the lower and middle thoracic region and dominate males. Although most are silent, which debuted, do so in the second decade of life with pain and flaccid or spastic progressive paraparesis, generally asymmetrical. Unlike the published cases, our patient was a girl and had a symmetrical spastic paraparesis.

Acquired lymphedema, distichiasis and SAC constitute a hereditary syndrome transmitted as an autosomal dominant, high penetrance and variable expressivity degree as seen in this case and her family.

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