

## Minimally invasive endoscopic fenestration of a spinal arachnoid cyst in a child with tetrasomy 18p: illustrative case

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**BACKGROUND** Spinal arachnoid cysts (SAC) are rare, especially in children. Patients can be asymptomatic or present symptoms of spinal cord compression. In this latter case, surgery is indicated to relieve the compression. Different surgical techniques have been described to treat these cysts, endoscopic or endoscopy-assisted fenestration being the least invasive. Tetrasomy 18p describes the condition in which two copies of the short arms of chromosome 18 are present. It is an extremely rare pathology with a variable phenotype, including 100% of cases cognitive impairment and developmental delay. Different central nervous system (CNS) abnormalities have been found in these patients.

**OBSERVATIONS** The authors describe the case of a 3-year-old boy with a tetrasomy 18p and a wide spinal arachnoid cyst that received an endoscope-assisted treatment with a significantly improved motor outcome.

**LESSONS** Tetrasomy 18p is an extremely rare pathology and different CNS abnormalities have been described in association with, but to date spinal arachnoid cyst has never been reported. These children typically show global hypotonia and cognitive impairment. The authors recommend a thorough neurological assessment with cranio-spinal magnetic resonance imaging to rule out any possible malformation that could be improved by surgery.

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**KEYWORDS** tetrasomy 18p; spinal arachnoid cyst; children; endoscopy; intradural

Spinal arachnoid cysts (SAC) are rare and are reported to account for only 1% of all pediatric intraspinal lesions.<sup>1</sup> The etiology can be spontaneous, but few cases have been described secondary to trauma and intraspinal hemorrhage.<sup>2</sup> They can be found more commonly in the extradural space; however, they can be intradural or perineural as well and, in rare cases, intramedullary. The dorsal region is the most affected area, but no region is immune to the development of SAC.<sup>3</sup> Patients can be asymptomatic, but most present with symptoms of spinal cord compression affecting motor, sensory, and bladder functions. Once symptoms develop, surgery is indicated to relieve spinal compression. Different techniques have been described: laminotomy and microsurgical removal and/or fenestration of the cyst, endoscopic fenestration through a mini-invasive approach, and drainage of the cyst fluid in a different compartment.<sup>4-7</sup>

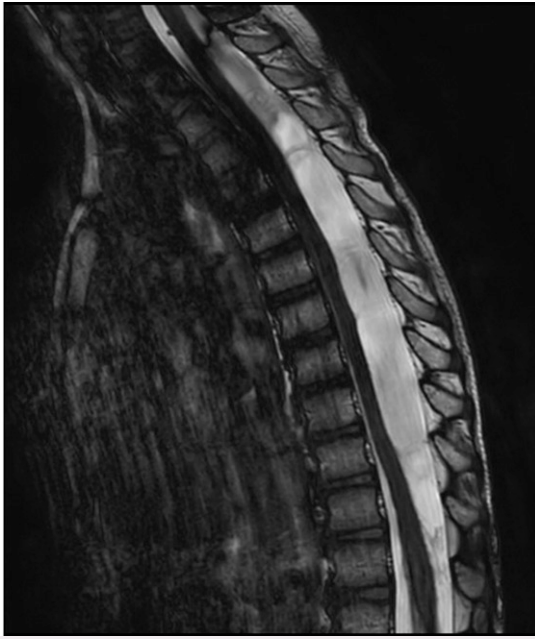
Tetrasomy 18p is exceedingly rare, occurring in approximately 1 in 625,000 births, with a prevalence of 1 case in 140,000 to 180,000 persons.<sup>8-10</sup> It is one of the most diagnosed isochromosomes.<sup>11</sup> The isochromosome is a supernumerary chromosome formed by two copies of the short arms of chromosome 18. The phenotype of tetrasomy 18 is variable, but it includes cognitive impairment and developmental delay in 100% of cases, thus making a thorough neurological assessment difficult. Patients with tetrasomy 18p can present with speech delay, strabismus, feeding problems, hypotonia/hypertonia, scoliosis/kyphosis, microcephaly, seizures, and hearing loss. The magnetic resonance imaging (MRI) abnormalities described to date are myelomeningocele, enlargement of the lateral ventricles, corpus callosum hypoplasia, a lipoma of the ambient cistern, dilatation of posterior fossa, and partial agenesis of cerebellar vermis.<sup>11,12</sup>

**ABBREVIATIONS** CNS = central nervous system; MRI = magnetic resonance imaging; SAC = spinal arachnoid cyst.

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**FIG. 1.** Preoperative sagittal T2 MRI showing the presence of a large dorsal arachnoid cyst extending from T4 to T12 with flow voids inside testifying the turbulence of the CSF inside the cyst.

We report the first case of a child with tetrasomy 18p and a wide dorsal intradural arachnoid cyst, who was treated at our institution with an endoscope-assisted technique that resulted in significant neurophysiological and clinical improvement after surgery.

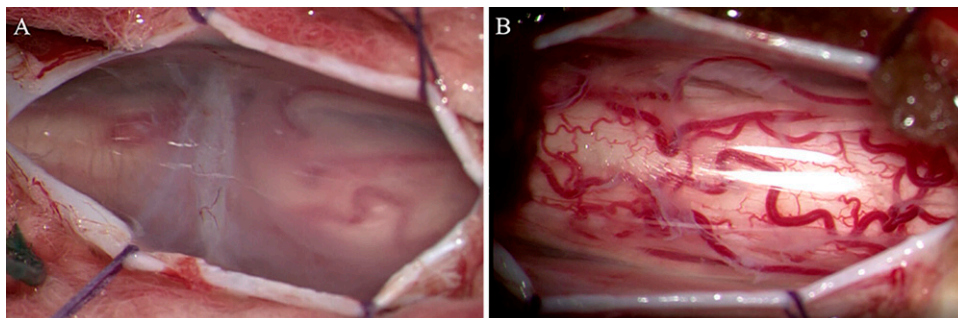
### Illustrative Case

A 3-year-old boy was referred to our institution due to the presence of a large dorsal arachnoid cyst at MRI. When he was 9 months old, he was diagnosed with tetrasomy 18p, following array comparative genomic hybridization (array-CGH). The child presented with generalized hypotonia and global developmental delay (Code: 315.8; ICD F88), specifically, the child showed 9 months language development, 7 months coordination, 6/7 months social, and 9 months postural abilities.<sup>13</sup> He performed psychomotor and speech therapy on a regular basis. Brain MRI was performed in the global assessment of the underlying pathology and showed hypoplasia of the corpus callosum,

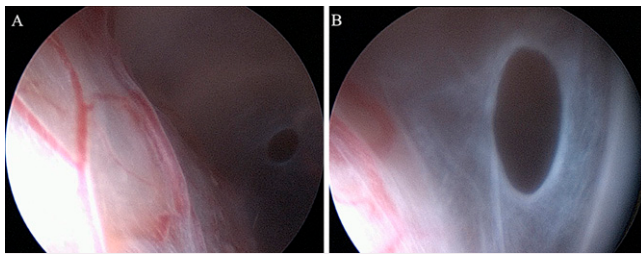
cerebellar tonsils extending 3 mm below the foramen magnum. The upper thoracic retromedullary spaces were noted to be extremely dilated, inducing significant anterior dislocation and compression of the spinal cord. Spinal MRI was therefore performed, which confirmed the presence of a large dorsal arachnoid cyst extending from T4 to T12 (Fig. 1). Somatosensory evoked potentials showed a decrease in amplitude of the left lower limb. A mini-invasive surgical approach was chosen to reduce the risk of spinal instability, considering the scarce compliance of the child to wear a corset and to practice physiotherapy. Gross motor function development was assessed preoperatively using the Gross Motor Function Measure, which is a four-point scale that consists of 88 items divided into five dimensions of gross motor function: A, lying and rolling; B, sitting; C, crawling and kneeling; D, standing; and E, walking, running, and jumping. The preoperative assessment confirmed a severe delay in motor development: dimension A, 96.1%; dimension B, 70%; dimension C, 45.2%; dimension D, 5.1%; and dimension E, 0%, thus giving a total result of 43.3% (16.8% of the objective).<sup>14</sup>

Surgery was performed under intraoperative monitoring. A T10–T11 laminotomy was performed to center the lower pole of the cyst. Laminae were not removed but elevated with a cranial pivot to preserve the posterior ligament complex; after dural opening, thick arachnoid membranes were carefully dissected from spinal cord and widely removed with microsurgical technique (Fig. 2). The fenestration of the cranial membranes was performed with endoscopic assistance (Fig. 3). At the end of surgery, CSF appeared to freely communicate on the surface of the spinal cord. Intraoperative neurophysiological monitoring was performed: somatosensory evoked potentials of the four limbs (with stimulation of bilateral median nerves and internal popliteal sciatic nerves and cortical registration), motor evoked potentials (registered at abductor pollicis brevis, rectus femoris, tibialis anterior, and abductor hallucis muscles), free-run electromyography and electroencephalography were recorded. Somatosensory evoked potentials showed significant improvement in the lower limbs, especially in the right leg at the end of the procedure (Fig. 4). The postoperative course was uneventful, and the early MRI showed reduction of the spinal cord compression and reappearance of the anterior CSF spaces (Fig. 5). The child was discharged on day 6 and admitted to our neurorehabilitation department for a period of intensive motor therapy.

The assessment of gross motor function was performed 5 months after surgery, revealing a discreet improvement: dimension A, 96%; B, 83%; C, 64%; D, 38%; E, 19%, giving a total result of 60% (29% of the objective).



**FIG. 2.** Intraoperative images of the microsurgical approach. **A:** The operative field after dural opening, showing thick arachnoid adhesions. **B:** At the end of the procedure, free CSF circulation on the surface of the spinal cord.



**FIG. 3.** Intraoperative endoscopic images showing a small fenestration performed in the cranial pole of the cyst (A) and the enlargement of the stoma (B), allowing free CSF circulation around the spinal cord.

## Discussion

### Observations

Regarding the etiology of spinal arachnoid cysts, Elsberg et al.<sup>15</sup> proposed in 1934 an origin from congenital diverticula or a congenital dural defect with herniation of the arachnoid. The congenital theory is supported by a few case reports of familial cases and the frequent association with neural tube defects.<sup>16–18</sup> Bond et al.<sup>19</sup> in 2012 published the largest series of pediatric spinal arachnoid cyst with 31 treated patients and stated that the 68% of them had a medical history of CNS abnormalities, thus adding support to the theory of congenital origin. Pathophysiology of arachnoid cyst of the spinal cord is not clear, but these malformative lesions have been related to traumatic events (fall on the back from a height, car accidents) and have been described in association with neural tube defects, that are also frequently found in patients with tetrasomy 18p. The patient described here fell frequently on his back according to his mother, probably as a consequence of his motor delay.

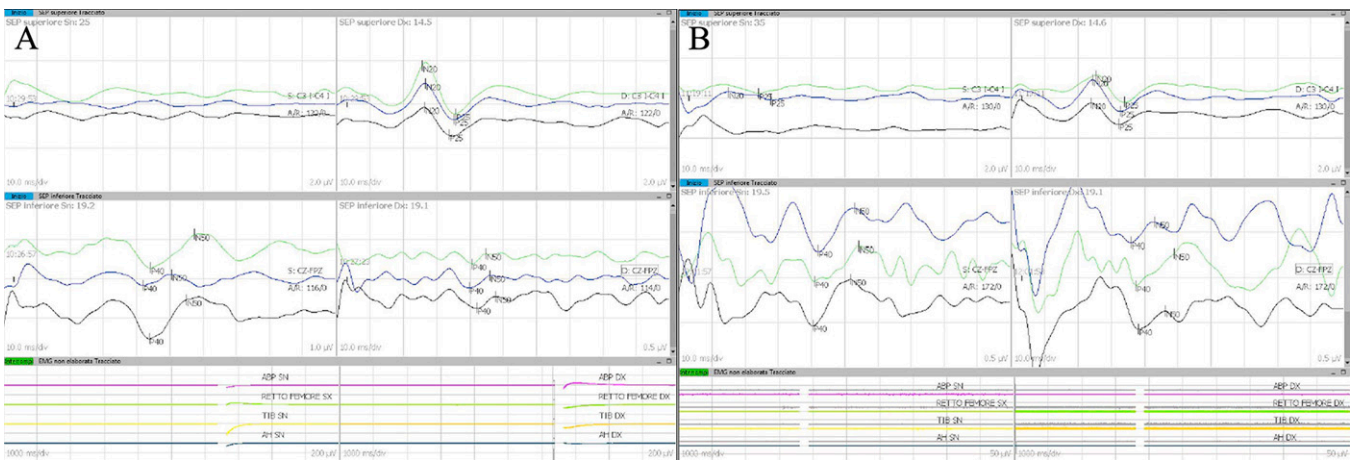
Once an arachnoid cyst is formed, its enlargement may depend on different factors: a ball-valve mechanism has been described in numerous cases.<sup>20,21</sup> Other authors propose the existence of an osmotic gradient between the cyst and the subarachnoid space, facilitating cyst expansion depending on the degeneration of cells of the arachnoid trabeculae.<sup>22</sup> Nevertheless, Sandberg et al.<sup>23</sup> demonstrated that the composition and osmolality of arachnoid cysts and CSF are quite similar.

Evangelou et al.<sup>24</sup> assumed that many of the spinal arachnoid cysts are not congenital, but adhesions that transform into pseudocystic vent mechanism. Their theory is supported by the intraoperative finding of multiple arachnoid layers and adhesions, together with the observation that even a mild or minor trauma can lead to a dramatic deterioration of the neurological function.

Surgery is indicated whenever symptoms from compression of the spinal cord and/or nerve roots are present. Different techniques have been proposed. Historically, exposure of the cyst through a multilevel laminectomy followed by microsurgical resection of the cyst wall has been advocated as a surgical approach that provides satisfactory clinical results and a low risk of recurrent cyst formation.<sup>25,26</sup> However, tight adhesion of the cyst wall to the pia mater of the spinal cord can make radical resection challenging. Moreover, in case of large arachnoid cysts extending over multiple vertebral segments, the invasiveness of radical exposure can be an issue.<sup>25,27</sup> Endo et al.<sup>5</sup> in 2010 described their endoscope-assisted technique, by which they treated six patients limiting the approach to a two- or three-level hemilaminectomy or laminectomy. In two cases they could not reach the edge of the cyst wall because the operative field was not wide enough. Few cases have been reported of spontaneous disappearance of an intradural spinal arachnoid cyst, however these cysts showed recurrence in all cases until surgery was performed.<sup>4</sup>

### Lessons

We found it relevant to describe this case because of its association with tetrasomy 18p. This genetical condition is systematically associated with hypotonia and motor delay. Diagnostic workup could be limited to brain MRI, that could fail to show low-lying spinal arachnoid cysts, that by the mean of spinal cord compression could be responsible (as in our case) for significant motor delay. Surgical decompression and intensive postoperative neurorehabilitation, although not completely resolving the clinical deficit, may allow significant improvement both in the motor function and in the quality of life. Minimally invasive approach to the dorsal spine was chosen because of the high risk of postoperative deformity and endoscopic support was helpful to improve fenestrations of the cyst wall, thus avoiding more invasive laminotomy for full exposure of the cyst.



**FIG. 4.** Intraoperative neurophysiological recordings of somatosensory evoked potentials. A: Baseline of the four limbs. B: Final registration at the end of surgery showing significant improvement in the lower limbs' potentials, especially in the right leg.





**FIG. 5.** Twenty-four-hour postoperative MRI showing the D10–D11 approach, the reduction of the spinal cord compression, and better visualization of the anterior CSF spaces.

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## Disclosures

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

## Author Contributions

Conception and design: all authors. Acquisition of data: Imperato, MA Cinalli, Servodio Iammarrone. Analysis and interpretation of data: MA Cinalli, Servodio Iammarrone, Ruggiero, G Cinalli. Drafting the article: Imperato, MA Cinalli. Critically revising the article: Imperato, G Cinalli. Reviewed submitted version of manuscript: Imperato, Ruggiero. Approved the final version of the manuscript on behalf of all authors: Imperato. Administrative/technical/material support: MA Cinalli. Study supervision: Ruggiero.

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