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Full-Endoscopic Decompression for Congenital Spinal Stenosis Caused by Diastrophic Dysplasia in a 13-year-old Pediatric Patient: A Case Report

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Introduction: Diastrophic dysplasia (DTD) is a rare disorder that affects the development of cartilage and bone. It is an autosomal recessive skeletal dysplasia that results in short stature with limb shortening, contractures of large joints, spinal deformities, cleft palate, clubfoot, cystic swelling of the external ear, and deformities of the hands. The spine frequently develops excessive lumbar lordosis, cervical and thoracolumbar kyphosis, and scoliosis. Spinal stenosis can also be found, but it is less commonly reported.

Case presentation: This study aims to present a case of congenital spinal stenosis caused by DTD causing conus medullaris syndrome in a pediatric patient, which was treated with a full endoscopic bilateral over-the-top interlaminar technique. The patient achieved complete symptomatic recovery without any perioperative complications from the procedure.

Conclusion: This is the first report of a full endoscopic decompression for spinal stenosis caused by DTD in the literature. Also, this is the first report of full endoscopic decompression for congenital spinal stenosis in a pediatric patient. Level of evidence IV; Case Series.

Keywords: Endoscopy; Spine; Low back pain; Spinal Stenosis; Osteochondrodysplasias;

INTRODUCTION

Diastrophic Dysplasia (DTD) is a rare autosomal recessive disorder of cartilage and bone development caused by pathogenic variants in the SLC26A2 gene encoding for a cell membrane sulfate/chloride antiporter crucial for sulfate uptake and Glycosaminoglycan (GAG) sulfation [1]. DTD is found in almost all populations with an estimated incidence of 1:100,000 [1,2] but it is widespread in the Finnish population, where the incidence can be as high as 1:33,000 due to the presence of a founder variant [1].

Clinical features of DTD include short stature with limb shortening, contractures of large joints, spinal deformities, cleft palate, clubfoot, cystic swelling of the external ear, and deformities of the hands [3]. The thumb is usually wide, hypermobile, with radial deviation, referred to as “hitchhiker's thumb.” The spine frequently develops excessive lumbar lordosis, thoracolumbar kyphosis, and scoliosis [2]. Scoliosis constitutes the most common spinal deformity, together with altered sagittal balance [4]. Cervical kyphosis is less common but may be a cause of severe neurological complications. The incidence of this deformity is up to 30%, and it is caused by hypoplasia of the vertebral bodies in the mid-cervical spine [6]. In a majority of DTD patients, the spinal deformity is not severe enough to require treatment [5]. Intelligence is normal [7]. Spinal stenosis can also be found, but it is less commonly reported [5, 8]. In young adulthood, progressive joint contractures and osteoarthritis result in mobility impairment [9]. Physical challenges, economic difficulties, and social adversities affect the quality of life [10].

This study aims to present a case of spinal stenosis caused by DTD in a pediatric patient, which was treated with

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a full endoscopic bilateral over-the-top interlaminar technique. This case is among the pioneers in the literature in the use of this technique in a pediatric patient with congenital spinal stenosis and, to our knowledge, is the first report of an endoscopic approach to a patient with DTD and spinal disease.

CASE REPORT

A 13-year-old boy, previously diagnosed with Diastrophic Dysplasia (DTD) by a medical geneticist, presented with thoracolumbar pain associated with paresthesia in both feet and the perineal region. Occasionally, he experienced a sensation of not being able to fully empty his bladder during urination (urinary retention). Although motor testing was otherwise normal, he reported subjective paraparesis, manifesting as fatigue in the lower limbs when walking long distances. The symptoms also worsened during prolonged sitting. During the neurologic examination, lower limb hyperreflexia was observed. These clinical findings were consistent with conus medullaris syndrome. The physical examination revealed clinical signs consistent with DTD, including short stature with normal head size, limb shortening, large joint contractures, and the characteristic "hitchhiker's thumb". The patient also had a positive family history: his parents were first-grade cousins, and his younger six-year-old sister had a similar congenital condition with spine deformity and was currently under diagnostic investigation.

The magnetic resonance image (MRI) revealed congenital spinal stenosis due to short pedicles associated with diffuse intervertebral disc bulging, causing spinal stenosis from T12 to L4 (Figure 1). The preoperative standing lateral x-ray showed a kyphotic thoracolumbar junction with a 23º Cobb angle from T9 to L2 (Figure 2A). The patient also exhibited other spinal anomalies, such as diffuse bone demineralization, L5 vertebral body sacralization, classified as Castellvi type IV [11], and spina bifida occulta in the lumbosacral segment.

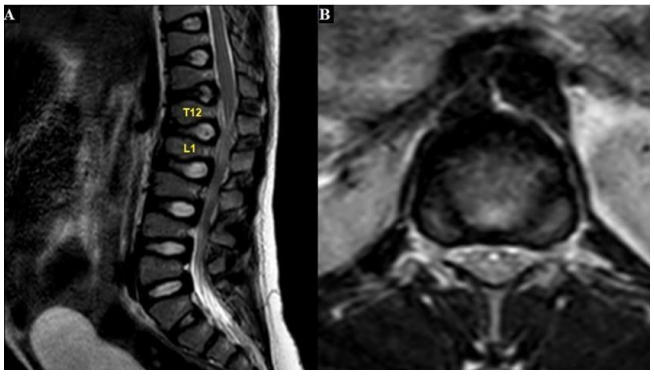

Figure 1: Magnetic Resonance Image of the thoracolumbar spine. (A) The sagittal plane image exhibits congenital spinal stenosis and diffuse disc bulging mainly between T12 and L4 vertebrae. The conus medullaris is observed in the T12-L1 transition. (B) Axial plane image demonstrates the T12-L1 transition where the compression is higher, due to the bony and soft tissue stenosis.

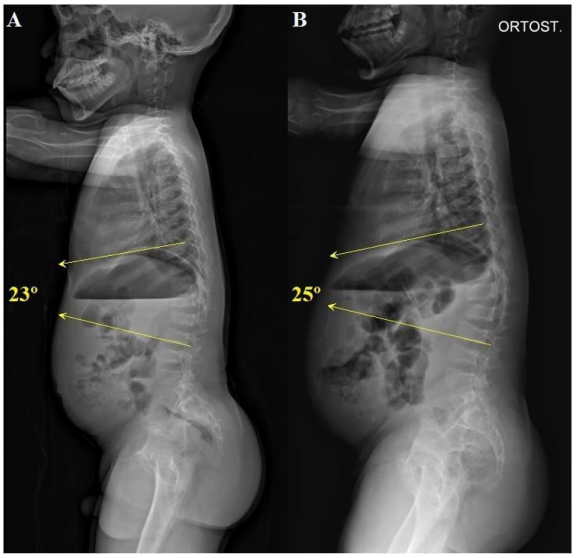

Figure 2: Standing full spine lateral x-rays. (A) The preoperative standing lateral x-ray showed a kyphotic thoracolumbar junction with a 23º Cobb angle from T9 to L2. (B) Two years later, the follow-up standing lateral x-ray demonstrated a mild increase of the T9-L2 Cobb angle to 25º.

Surgical procedure

Given the radiographic evidence of multilevel spinal stenosis, the first-line treatment would typically involve open decompression with instrumented arthrodesis. However, the patient's young age (13 years), underlying skeletal dysplasia, and preexisting short stature raised concerns about long-term complications, including hardware failure, growth restriction, and adjacent segment disease.

Additionally, the patient's signs and symptoms clearly suggested conus medullaris syndrome. Therefore, our team aimed to treat the most symptomatic level using the least invasive approach possible. Thus, full-endoscopic decompression was indicated via an interlaminar approach at the T12-L1 level, where the conus medullaris was identified on MRI.

The procedure was performed under general anesthesia with endotracheal intubation. The patient was placed on a radiolucent table in a prone position and the operating table was bent on the level of the lower lumbar spine to widen the interlaminar window. The midline was marked along the tips of the spinous process, and the correct level was confirmed with the C-arm X-ray fluoroscopy.

A 22G spinal needle was inserted into the interlaminar window laterally, and local anesthesia was injected into its

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pathway and along the area of the skin incision. A skin incision of 8 mm was made about 0.5 cm lateral to the midline. The needle was removed and a guide wire was inserted. The dilator was then inserted with its tip pointed at the lateral edge of the interlaminar window until the bony structure (facet joint) was felt.

A beveled working sheath was inserted through the dilator with its beveled opening toward the spinal canal. Subsequently, an endoscope was introduced through the working cannula with continuous normal saline drainage. Thereafter, the procedure was performed under direct visual control.

The interlaminar window and the ligamentum flavum were the first structures to be identified (Figure 3A). High-speed burrs were then used to remove the bone from the adjacent lamina, disconnecting the ligamentum flavum from its insertion (Figure 3B). In adult patients, bony hypertrophy of the facets caused by degenerative osteoarthritis may also be removed in this step, but it wasn't necessary in this case.

A Kerrison rongeur was further used to remove the laminae bone (laminotomy) partially and the yellow ligament bilaterally in a "cross-over" or "over-the-top" technique (Figure 3C) until the dural sac was fully exposed. A final inspection of the entire dural sac confirmed sufficient decompression (Figure 3D), and hemostasia was thoroughly done at this stage.

The operating sheath was removed, a single stitch was given to close the incision, and a sterile drape was applied. The patient reported complete relief from back pain the following day and was discharged on the third postoperative day.

At the 3-month follow-up post-operation, the patient referred relief of the subjective paraparesis previously reported, and didn't have perineal or lower limb paresthesias anymore, although the hyperreflexia persisted. He also didn't complain of any bladder symptoms.

At the 2-year follow-up clinical evaluation, he maintained symptomatic relief and underwent radiographic evaluation. The postoperative MRI (Figure 4) demonstrated decompression of the T12-L1 segment with preservation of the bony structures. Although congenital spinal stenosis remained, primarily in the lumbar segments, the patient exhibited no additional signs or symptoms. A postoperative standing X-ray (Figure 2B) revealed a mild increase in T9-L2 Cobb angle from 23º to 26º, which was not clinically relevant. No complications were observed until the last follow-up.

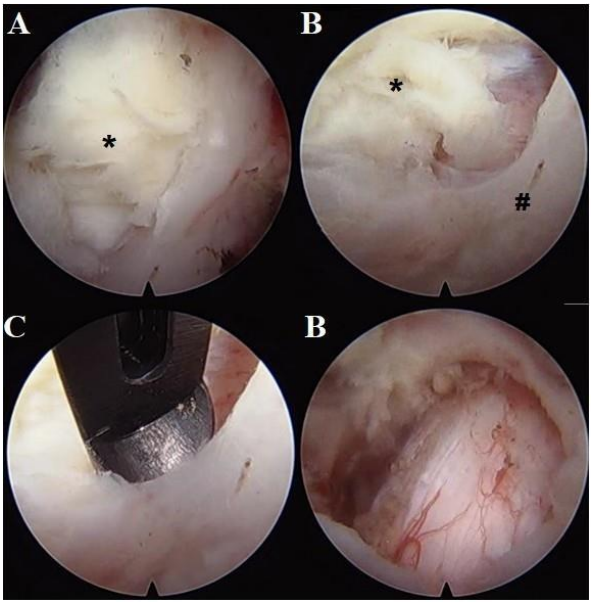

Figure 3: Intraoperative endoscopic images. (A) The interlaminar window and the ligamentum flavum (*) were the first structures to be identified. (B) High-speed burrs were then used to remove the bone from the adjacent lamina (#), disconnecting the ligamentum flavum from its insertion. (C) A Kerrison rongeur was further used to partially remove the laminae bone (laminotomy) and the yellow ligament bilaterally in a "cross-over" or "over-the-top" technique. (D) A final inspection of the entire dural sac confirmed sufficient decompression.

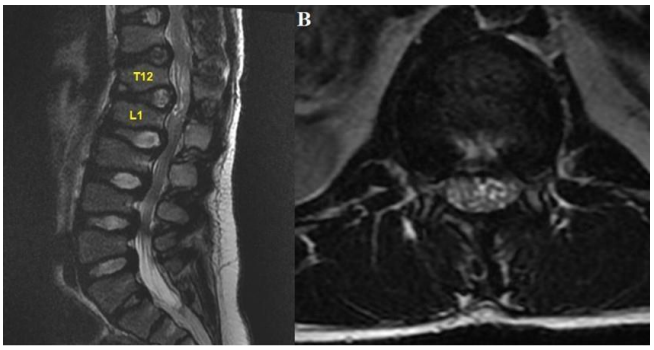

Figure 4: Postoperative 2-year follow-up MRI. The sagittal (A) and axial (B) images evidence the decompression of the T12-L1 segment with preservation of the bony structures. The lumbar spine still exhibits the congenital spinal stenosis, but no relevant deformity was observed, in spite of the preexisting thoracolumbar kyphosis.

DISCUSSION

Many genetic conditions impacting skeleton growth cause osseous spine abnormalities. Over 400 distinct disorders, called skeletal dysplasias, fall within this group [12]. Such abnormalities predispose patients to compression of neural elements, leading to myelopathy or radiculopathy at a relatively early age [8]. Few studies evaluated the outcomes of spine surgery amongst patients specifically with DTD. In this population, scoliosis constitutes the most common spinal deformity, along with altered sagittal balance [4]. Cervical kyphosis, while less common, can cause severe neurological complications and the incidence rates of

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this deformity can be as high as 30% [6]. Poussa et al. [5] studied 101 patients with DTD and spinal disease. The overall frequency of scoliosis was 37%, followed by cervical kyphosis, found in one-third of the patients. Only four patients had spinal stenosis in this series, of which two were operated on via posterior decompression. A recent study conducted by Shafi et al. [8] analyzed spine surgery outcomes for skeletally mature patients with skeletal dysplasia. The final case series included 19 (76%) patients with achondroplasia, 5 (20%) patients with spondyloepiphyseal dysplasia, and 1 patient (4%) with diastrophic dysplasia. The only patient with DTD presented with Lumbar spinal stenosis with bilateral lumbar radiculopathy and neurodeficit and was submitted to a posterior decompression from T12 to S1. After the surgery, the patient developed cauda equina syndrome and required a posterior revision decompression and fusion surgery from T7 to L2.

In our report, the patient suffered from congenital spinal stenosis, which is a relatively uncommon presentation in DTD patients, according to the literature [2, 5, 6, 8]. Surgical treatment for spinal stenosis typically involves decompression of soft tissues and dissection/debulking of the intervertebral disc, as well as the removal of bony structures. However, over time, patients may experience facet joint and spinal destabilization, which could necessitate additional surgeries and result in a prolonged recovery period [13]. Performing surgery on individuals with skeletal dysplasias presents a significant challenge due to the high rates of associated complications. Atypical collagen leads to friable connective tissues, predisposing patients to dural tears and wound healing complications. At the same time, unfamiliar anatomy may complicate the surgical approach or instrumentation, increasing the risk of neurologic injury. Additionally, aberrant anatomy (e.g. short pedicles) increases the risk of disease progression [8]. Given the high complication rates of spine surgery in patients with skeletal dysplasias previously reported [8], we aimed to perform a minimally invasive procedure to decompress the spinal canal, with the minimum tissue trauma along the surgical access pathway, minimized epidural scarring, short hospital stay, and rapid rehabilitation. Thus, we performed a full-endoscopic bilateral over the top decompression of the canal, targeting the most symptomatic segment, where the conus medullaris was identified on MRI. Multilevel decompression was avoided due to the risk of further instability and the potential need for extensive instrumented arthrodesis. With this approach, the trade-off was a theoretical risk of future reoperation at different and/or adjacent levels. In up to two years of follow-up, the patient maintained the symptomatic relief and the images did not show deformity progression. Although spinal stenosis radiographically persisted in the lumbar segment, it alone does not constitute an indication for surgery, as patients often remain asymptomatic for years.

As technology advanced, full endoscopic decompression of the spinal canal has become possible, and the paradigm of spinal endoscopy is shifting from the treatment of soft disc herniation to that of lumbar spinal stenosis. Lee et al. [14] conducted a meta-analysis of five studies involving 156 patients with lumbar spinal stenosis treated with a full-endoscopic interlaminar approach. The overall mean difference of patients with Oswestry Disability Index (ODI) and Visual Analogue Scales (VAS) scores for leg and back pain exceeded the criteria for minimal clinically important difference with statistically significance, concluding that successful clinical outcomes can be achieved with full-endoscopic decompression for lumbar central spinal stenosis.

Regarding the pediatric population, there have been no published series of patients with congenital spinal stenosis treated using a full-endoscopic approach. On the other hand, the percutaneous endoscopic discectomy performed in this population is well described and has been evaluated in several studies with short-term success rates of 91% to 100% [15]. To our knowledge, only three patients have undergone open surgery to treat spinal stenosis caused by DTD, one in the Shafi et al series[8] and two in the Poussa et al series.[5]

CONCLUSION

This is the first report of a full endoscopic decompression for spinal stenosis caused by DTD in the literature. Also, this is the first report of a full endoscopic decompression for congenital spinal stenosis in a pediatric patient. The procedure was performed with safety and it was effective, but further studies are necessary to establish its effectiveness in larger series concerning this population and over longer periods.

DISCLOSURES

Ethical approval

This study was performed in line with the principles of the Declaration of Helsinki. Ethics Committee approval was not required for this work, as it solely involves a case description without any patient intervention. However, we have obtained written consent from the responsible party for the patient's case report to be published in the journal

Consent to participate

The patients gave consent to use their information and images for research purposes. *Consent for publication*

The patient gave consent to use his information and images for publication.

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Conflict of interest

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

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Artificial intelligence

The authors affirm that no artificial intelligence tools were used in the writing, editing, or content generation of this manuscript. All work was conducted manually, based on thorough research and academic expertise.

CONTRIBUTIONS

-**Emilson José de Souza Camapum**: Conceptualization, Formal Analysis, Writing – original draft
-**Benicio Otton de Lima**: Methodology
-**Flavio Leão Lima**: Methodology
-Tiago Medeiros Nobre: Formal Analysis
-Amauri Araújo Godinho Junior: Writing – review & editing
-Baldomero Pinto Soares: Conceptualization, Methodology, Writing – review & editingWriting – review & editing

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