Surgical management of congenital thoracic kyphosis and multilevel bilateral thoracic pedicle aplasia: case report

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Pedicle aplasia is an uncommon congenital anomaly most frequently involving the absence of a single pedicle at a single vertebral level. Bilateral pedicle aplasia at multiple levels is exceedingly rare and has only been described once previously in the literature. While single-level pedicle aplasia is often asymptomatic and discovered incidentally, pedicle aplasia of multiple levels may produce severe spinal deformities and neurological deficits. Due to the rarity of this condition, optimal management remains uncertain. In this case report, the authors describe the surgical management of a healthy 9-year-old boy who presented with frequent falls, difficulty running, and severe thoracic kyphotic deformity and was found to have bilateral pedicle aplasia from T3 to T9. A review of the literature regarding pedicle aplasia is also presented.

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PEDICLE aplasia is an uncommon congenital anomaly of the spine that occurs most frequently in the cervical and lumbar spine^{4,5,13,18} and rarely in the thoracic spine.^{2,3,8,15,17,20} While the condition is frequently asymptomatic and discovered incidentally, patients may present with back pain or deficits secondary to spondylolisthesis, degenerative changes, or kyphosis.^{4,5,13} Existing case reports have primarily described pedicle aplasia that is limited to a single vertebra;^{4,13} there is only one prior case report in which the authors described the congenital absence of bilateral thoracic pedicles at multiple levels, resulting in kyphosis.¹⁵ Given the rarity of this condition, optimal management is uncertain; additionally, the feasibility of utilizing standard pedicle screw instrumentation systems in the absence of pedicles may be questioned. In this case report, we describe the presentation and surgical management of a pediatric patient with congenital absence of thoracic pedicles from T3 to T9 and resultant severe kyphosis of the thoracic spine. A review of the literature regarding pedicle aplasia and the principles of management are also presented.

Case Report

Clinical History and Presentation

A 9-year-old boy presented with complaints of mid-tho-

racic back pain, lower-extremity weakness, and leg pain for 6 weeks. The patient had been noted to have increasing difficulty running and more frequent falls. Bladder and bowel function were normal. He initially presented to a sports medicine clinic after the patient's mother had noticed that his upper back felt abnormal on palpation. On physical examination at our institution, he had obvious kyphosis that remained fixed on extension. Normal strength was exhibited throughout. The patient had patellar reflex of grade 3+ bilaterally; reflexes of the biceps and gastrocnemius muscles were normal. Sensation and proprioception were normal. The patient was noted to turn his toes slightly inward during ambulation and have poor coordination during running. The patient underwent formal pulmonary function testing, the results of which were normal. Of note, the patient's older sister had congenital scoliosis and underwent surgery during infancy.

Preoperative Imaging

Standing radiographs demonstrated regional T1–12 kyphosis of 39°, segmental T3–9 kyphosis of 58°, and focal T6–7 kyphosis of 81° (Fig. 1 right). There were no significant coronal plane abnormalities (Fig. 1 left). The preoperative T1–12 height was 17.7 cm. CT imaging showed absence of bilateral pedicles from T3 to T9, severe focal

ABBREVIATIONS VCR = vertebral column resection.

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FIG. 1. Preoperative radiographs demonstrating severe fixed focal kyphosis and absent pedicles at multiple levels. **Left:** Anteroposterior view demonstrating normal coronal plane alignment. **Right:** On the lateral view, pedicles cannot be visualized from T3 to T9. The patient had regional T1–12 kyphosis of 39°, segmental T3–9 kyphosis of 58°, and focal T6–7 kyphosis of 81°.

kyphosis at T6 and T7, and adjacent narrowing of the canal over the kyphotic apex (Fig. 2). MRI of the thoracic spine showed severe central stenosis at T6–7 with draping of the thoracic cord over the kyphotic apex; there was no T2 signal (Fig. 3). Based on radiography results, the patient had a Risser score of 1.

Operation: Vertebral Column Resection, Instrumentation, and Fusion

The patient was positioned prone. Somatosensory and motor evoked potentials were monitored throughout the procedure; at baseline, neuromonitoring indicated a decrease in the left lower-extremity signal. The posterior elements of the spine were exposed from T2 to T9; severe kyphosis was evident, as was congenital absence of the pedicles from T3 to T9. Under fluoroscopic guidance and using standard anatomical landmarks, pedicle screws were placed in the standard orientation into the vertebral bodies from T2 to T9. Following screw placement, the T7 ribs were exposed bilaterally and resected for bone grafting. A T5–8 laminectomy was performed. Subperiosteal dissection was continued anteriorly around the vertebral bodies of T6 and T7 until both were fully exposed. Following vertebral body exposure, temporizing rods were placed on the left side while T6 and T7 were resected via a costotransversectomy approach. Despite placement of a temporary rod on the left side, the spinal cord appeared to be under traction as the right-sided resection was being completed. However, there was a decrease in the neuromonitoring signals, requiring immediate place-

ment of a temporary rod on the right side and reduction and compression of the kyphosis. Neuromonitoring signals normalized, and the final rods were contoured and inserted. Next, an interbody cage was placed. Both rods were serially compressed to further kyphosis reduction. The lamina was decorticated, bone graft autograft as well as allograft was applied, and the wound was closed.

Complications and Reoperation

On postoperative day 4, the patient was noted to have inconsistent left lower-extremity weakness, which gradually improved without intervention. MRI at the time did not show acute spinal cord compression. Postoperative imaging was indicative of distal junctional failure and residual kyphosis acting as a lever arm, potentially producing intermittent traction on the spinal cord and explaining the inconsistent left lower-extremity weakness (Fig. 4). The patient was then brought back to the operating room for revision of his instrumentation, extension of distal fusion to T11, and further decompression from T6 to T9. Intraoperatively, placement in the prone position resulted in improvement in motor evoked potentials, and, at the end of the case, they further improved from his preoperative baseline. Postoperatively, his strength improved, and he was eventually discharged to a rehabilitation facility.

Postoperative Course

At the most recent follow-up of 27 months, the patient had normal neurological examination findings and

was able to participate in all regular activities. Follow-up imaging demonstrated solid fusion and improvement in thoracic kyphosis (Fig. 5). The patient's regional T1–12 kyphosis was 31°, T3–9 segmental kyphosis was 23°, focal kyphosis (inferior T5 and superior T8 [in between cage]) was 7°. The postoperative T1–12 height was 18.5 cm. The patient was kept in a brace postoperatively for 6 months, given that he had already experienced junctional failure immediately after surgery.

Discussion

Spinal pedicle aplasia is an uncommon congenital spinal anomaly that occurs due to retrosomatic cleft formation during embryonic development.^{4,13} Pedicle aplasia most frequently occurs in the cervical or lumbar spine, although thoracic pedicle aplasia has also been described.^{2–5,8,13,15,17,18,20} Most commonly, only one pedicle and one vertebral level are involved.^{13,20} Bilateral pedicle aplasia of multiple vertebrae appears to be exceedingly rare, with one prior case report in the literature.¹⁵

Patients with pedicle aplasia are frequently asymptomatic, in which case the anomaly is identified incidentally.^{8,10–12,21} The absent pedicle is typically evident on plain radiographs, although additional imaging with CT, myelography, or MR may be necessary, particularly in the case of a single missing pedicle, in order to rule out malignancy, vascular anomaly, or inflammatory disease.^{4,5,20} Images of pedicle aplasia typically demonstrate an abnormally enlarged neural foramen filled with fat and fibrous tissue.^{19,20} When a single pedicle is involved, the contralateral pedicle is often hypertrophied.²⁰ Pedicle aplasia is often associated with dysplasia or absence of other ipsilateral pillar or neural arch elements, including the lamina, spinous process, and transverse process.^{4,8,19} Pedicle aplasia is also frequently accompanied by hypoplasia of the articular facet and other facet joint abnormalities, which may result in biomechanical abnormalities due to compromised rotational and extensional stability.^{5,8,13,20}

The most common symptom reported by patients with single-level pedicle aplasia is back pain responsive to conservative management, although patients may develop neurological deficits due to spondylolisthesis or degenerative changes.^{13,14} Lederman and Kaufman reviewed 9 patients with thoracic pedicle aplasia or hypoplasia, of whom 2 patients reported nonspecific low-back pain and none of whom required surgical intervention.⁸ Patients with unilateral lumbosacral pedicle aplasia have been reported to develop spondylolisthesis, likely secondary to facet joint abnormalities, resulting in instability; these patients present with refractory low-back pain and variable lower-extremity deficits.^{13,14} Degenerative changes on the contralateral side have also been reported to result in a contralateral radiculopathy at the level of the pedicle aplasia.^{5,14} In these cases, it was thought that the absent pedicle and articular facets resulted in instability and overload of the contralateral pedicle, resulting in degenerative changes and radicular pain. Patients with intractable back pain or neurological deficits require surgical intervention.⁴

One prior case report exists in which pedicle aplasia of multiple thoracic vertebrae resulting in severe flexible

FIG. 2. Preoperative CT scans demonstrating multilevel bilateral pedicle aplasia. Sagittal images (*left*) demonstrating severe focal kyphosis at T6 and T7 with narrowing of the canal from T5 to T8. Axial images (*right*) at the levels of T3 (**A**), T7 (**B**), and T8 (**C**), demonstrating absence of pedicles.

thoracic kyphosis is described.¹⁵ Rauzzino et al. described a patient with bilateral aplasia of the T4–8 pedicles, resulting in 64° of kyphosis. The patient had back pain, decreased sensation below T6, and asymmetrical weakness in the lower extremities. The patient underwent a posterior laminectomy from T4 to T8 with arthrodesis from T2 to T12 with a hook claw construct and autologous bone

FIG. 3. Preoperative images demonstrating bilateral, multilevel pedicle aplasia with severe kyphosis and draping of cord across kyphotic apex. **A:** Sagittal T2-weighted MR image demonstrating severe kyphotic deformity with severe canal stenosis resulting in spinal cord compression and spinal cord flattening over the kyphotic apex of T6–7. **B–D:** Sagittal CT images demonstrating absence of pedicles from T3 to T9.

grafting. Postoperatively, the kyphosis corrected to 22°. At the 1-year follow-up, the patient had complete back pain resolution with full return of strength and sensation.

The case described by Rauzzino et al. bears significant resemblance to our case. These cases illustrate the unique clinical considerations posed by multilevel, bilateral involvement as opposed to the more common aplasia of a single pedicle at a single level. While the majority of patients with single-level pedicle aplasia are asymptomatic or present with low-back pain that responds well to con-

servative management, both patients with multilevel thoracic involvement developed significant thoracic kyphosis and lower-extremity weakness requiring surgical intervention. In contrast, only one documented single-level pedicle aplasia case in the literature was associated with scoliosis, and in that case the scoliotic apex was 6 levels above the absent pedicle.¹⁶ While patients with unilateral pedicle aplasia often have a normal if not enlarged spinal canal and thus typically do not require decompression,⁸ both patients with bilateral involvement had clinically sig-

FIG. 4. Radiographs obtained immediately postoperatively, demonstrating distal junctional failure and residual kyphosis. **Left:** Anteroposterior view demonstrating normal coronal balance. **Right:** Lateral view demonstrating ongoing kyphosis from a lever arm effect.

FIG. 5. Follow-up imaging after revision and extension of the distal construct, demonstrating extension of the instrumentation distally to T11. **Left:** Lateral view demonstrating significant improvement in thoracic kyphosis. Regional T1–12 kyphosis was 31°, T3–9 segmental kyphosis was 23°, and focal kyphosis (inferior T5 and superior T8 [in between cage]) was 7°. **Right:** Anteroposterior view demonstrating good coronal plane balance with intact hardware.

nificant stenosis at the level of the missing pedicles and benefited from decompression. Multilevel pedicle aplasia thus appears to be clinically distinct from single-level disease.

Symptomatic patients with pedicle aplasia may undergo either anterior or posterior instrumentation and fusion.^{4,14} Hsieh et al. noted that pedicle aplasia increases the difficulties associated with pedicle screw placement and solely placed percutaneous pedicle screws in normal pedicles;⁴ similarly, in the patient described by Rauzzino et al., hook claw instrumentation was placed.¹⁵ However, Kaito et al. noted that pedicle screws may be placed at the site of pedicle aplasia directly into the vertebral body where the pedicle would otherwise be located.⁵ Our case differs tremendously from these prior reports as our patient underwent a 2-level vertebral column resection (VCR) for deformity correction and had 7 levels with absent pedicles bilaterally. Long-segment solid fixation was necessary, prohibiting the skipping of the aplastic levels or strictly utilizing a hook construct. It is our experience that pedicle screws can be placed in a normal orientation into the vertebral body even in the absence of pedicles, and in this case screws were placed uneventfully at multiple levels with congenitally absent pedicles. Fluoroscopic guidance was sufficient for accurate placement of screws, and there were no complications or issues related to inappropriate pedicle screw trajectory. Our experience and that of Kaito et al. indicate that instrumentation and fusion using pedicle screws is safe and feasible in patients with pedicle aplasia, even at multiple levels.

A distinguishing factor between this case and the previously published case of multilevel thoracic pedicle aplasia

was the presence of a fixed kyphotic deformity and extent of surgical correction required. In the case published by Rauzzino et al., the kyphosis was flexible, allowing for adequate deformity correction without osteotomy.¹⁵ In our case, VCR was necessary; the patient ultimately did well, and significant deformity correction was achieved. VCR is a formidable and inherently destabilizing surgical intervention that requires long-construct instrumentation, typically through anterior column (via cage) reconstruction and multilevel posterior fixation to restore stability.⁹ The integrity of the posterior screw construct is of particular importance after multilevel VCR.⁷ Our patient did require extension of instrumentation due to distal junctional failure causing intermittent cord traction and secondary myelopathy. Fortunately, following his revision, the patient did very well with excellent recovery of lower-extremity strength and sensation.

One important consideration in long-construct fusions in the skeletally immature pediatric population is the potential for impaired thoracic growth and resultant long-term respiratory problems.¹ Early fusion of the thoracic spine can reduce final thoracic height, producing a long-term reduction in pulmonary function. Fusion of over 8 thoracic vertebrae early in life may produce a decrease in final forced vital capacity of over 50% and significantly increase the risk of restrictive lung disease.^{1,6} Authors have indicated that a final thoracic spine height of over 18 cm is necessary to allow for adequate respiratory function.⁶ In this case, while the potential sequelae associated with fusion of the thoracic spine were a serious concern, the patient's clinical presentation (severe, rigid kyphosis with symptomatic neurological deficits) necessitated a degree of deformity correction that was thought to only be feasible via VCR, which inherently requires instrumentation and fusion to restore stability. Fortunately, the preoperative T1–12 height was almost 18 cm, and the postoperative T1–12 spine height was 18.5 cm. However, the patient will require close monitoring and long-term pulmonary follow-up.

Pedicle aplasia is an uncommon congenital anomaly that is most frequently asymptomatic. Multilevel bilateral pedicle aplasia is an exceedingly rare entity that may cause severe deformity, back pain, and neurological deficits. In symptomatic patients with multilevel bilateral pedicle aplasia, deformity correction and spinal decompression may be necessary, with the potential for improvement in or reversal of neurological deficits. Image-guided screw instrumentation remains a viable option, despite the absence of pedicles with direct insertion into the vertebral bodies.

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Disclosures

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Author Contributions

Conception and design: Lau, Sun. Acquisition of data: Lau, Policy. Analysis and interpretation of data: Martin. Drafting the article: Lau, Dalle Ore. Critically revising the article: Lau, Dalle Ore, Sun. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: Lau.

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