

## Undifferentiated presentation of unilateral agenesis of a cervical pedicle and a contiguous vertebral hemangioma: illustrative case

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**BACKGROUND** Unilateral agenesis of a cervical pedicle is a known rare entity that has been well described over the past 70 years. It is usually an incidental or minimally symptomatic presentation with no significant clinical repercussion. No previous report has described concurrent non-osseous developmental abnormalities alongside this unique pathology.

**OBSERVATIONS** This case reported a cervical hemangioma with associated unilateral pedicle agenesis and an incidental finding of callosal dysgenesis and lipoma. The initial presentation consisted solely of persistent neck pain, with cervical radiography illustrating significant kyphotic deformity secondary to apparent anterolisthesis of C3-C4. The patient underwent a combined approach: anterior cervical corpectomy at C4-C5 with supplemental posterior fusion. The authors provided a review of the literature concerning developmental pedicle abnormalities and vertebral hemangioma. Pedicle agenesis is known to be associated with multiple pathologies, but the authors have not found evidence of a clinical paradigm consisting of a vertebral hemangioma in the presence of cervical pedicle agenesis, callosal dysgenesis, or callosal lipoma.

**LESSONS** Careful evaluation of radiographs with appropriate subsequent multimodal imaging is key to identifying unique pathologies in the spine that complement a patient's history and clinical findings. If multiple abnormalities are noted, a novel clinical etiology or syndrome must be considered.

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**KEYWORDS** cervical spine; pedicle agenesis; hemangioma; callosal dysgenesis; lipoma

Careful evaluation of radiographic imaging of the spine is of utmost importance in the assessment of both traumatic and atraumatic presentations. Multiple imaging modalities can provide valuable insight when unusual structural or morphological features are seen.<sup>1</sup> Unilateral absence of a cervical pedicle has been well described in the literature, with reports describing the abnormality in the cervical, thoracic, and lumbar spine. It is thought to be of congenital origin, although some cases conspicuously illustrate the cause to be from a pathological process such as metastases.<sup>2</sup> Despite current literature describing the congenital origin of pedicle agenesis, reports remain to only be describing concurrent non-osseous developmental abnormalities.

We describe here a distinct case demonstrating the absence of a pedicle with an associated contiguous vertebral hemangioma presenting with apparent instability and a kyphotic deformity. This presentation on its own would have been unusual; however, callosal dysgenesis and

lipoma were also identified. This indicates that there could be an underlying genetic association, or it is simply a coincidental finding. Additionally, we provide a review of the literature regarding the absence of a unilateral pedicle, both congenital and from acquired causes.

### Illustrative Case

A 14-year-old girl who had been healthy presented with neck pain that had persisted for 3 weeks. She had sought therapy with a physiotherapist with no resolution of her symptoms and increasing pain. At the time, she was not experiencing any neurological symptoms such as numbness, paresthesia, weakness, loss of dexterity, gait disturbance, or discoordination.

Eventually, she presented to the emergency department, where her neurological examination was unremarkable. She was sent for a cervical radiograph (Fig. 1), which depicted significant kyphotic deformity

**ABBREVIATIONS** CT = computed tomography; MRI = magnetic resonance imaging.

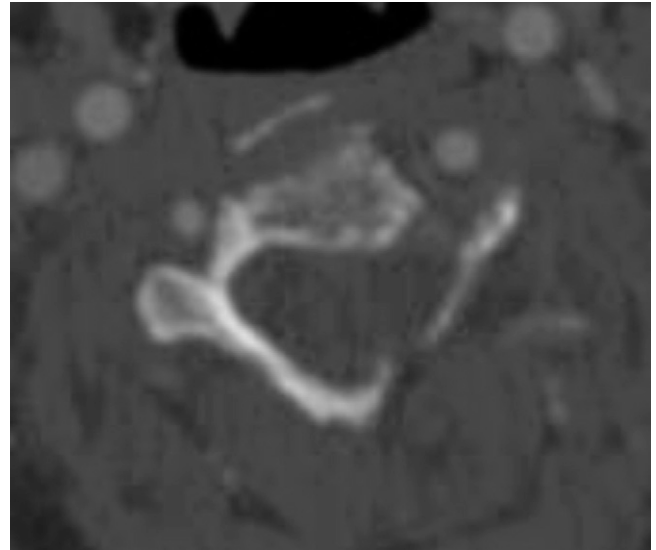
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**FIG. 1.** Lateral cervical spine radiograph showing focal kyphotic deformity at C4-C5 and abnormal lateral mass morphology.



**FIG. 2.** Axial CT of cervical spine region showing the absence of the C4 pedicle, with a dysplastic lateral mass.

secondary to apparent anterolisthesis of C3-C4. This warranted further investigation; thus, computed tomography (CT) and magnetic resonance imaging (MRI) of the brain and cervical spine were ordered.

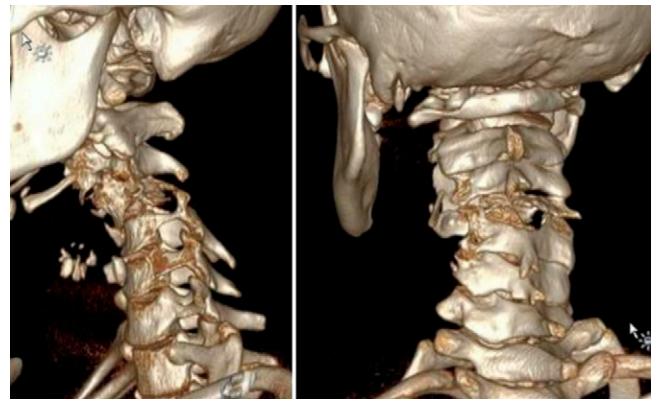
CT and angiography of the cervical spine revealed agenesis of the left C4 pedicle with significant thinning and elongation of the left C3 and C5 pedicle (Fig. 2). MRI of the cervical spine revealed a hyperintense signal located within the vertebral bodies of C3 and C4 with extraosseous extension, suggestive of a hemangioma. CT volume rendering revealed abnormal thinning of the cortical bone and a left lateral mass around C3-C4 (Fig. 3). Additionally, MRI of the brain showed callosal dysgenesis with the presence of an adjacent lipoma (Fig. 4).

Based on our analysis, the left pedicle and lateral mass appearance at both levels were suggestive of a developmental abnormality rather than erosion from the contiguous hemangioma. Additionally, the findings noted above within the cervical spine were likely contributing to the C3-C4 anterolisthesis, which could be seen with apparent thinning of the pedicle and lateral mass. This, coupled with agenesis of the pedicle on the left side, was likely contributory to the pathological process underlying the kyphotic deformity. The simultaneous presentation of callosal dysgenesis and lipoma, vertebral hemangioma, and absence of the C4 cervical pedicle did not seem to fit a clinical paradigm. The coincidental nature of the cerebral and vertebral findings was likely; however, an underlying genetic etiology was also possible.

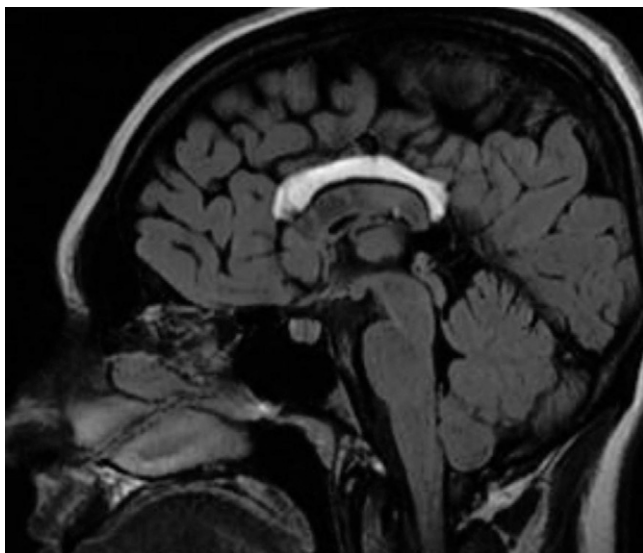
The patient received correction of the deformities and stabilization of the C3-C6 segment of the cervical spine. We elected for an anterior C4 and C5 corpectomy, with vertebral replacement using a fibular strut allograft to correct the acute kyphosis with underweighted traction using Gardner Wells tongs. The procedure was accomplished with

neuromonitoring using both somatosensory and motor evoked potentials. She was stable throughout the period of traction and surgery. The patient was then turned prone and received placement of posterior instrumentation. Bilateral mass screws and an interconnecting rod from C3 to C6 were inserted. The lateral masses of C4 were avoided bilaterally, considering the malformed facet. Tissue from the corpectomy was sent to pathology, which confirmed the presence of a hemangioma. Postoperative radiography illustrated satisfactory reduction in cervical kyphosis as well as adequate instrumentation placement.

Our patient recovered well postoperatively, with expected postoperative pain that resolved at the 6-week follow-up visit. She was discharged with an aspen collar and continued to use it when upright. A follow-up radiograph was obtained at 6 weeks (Fig. 5), demonstrating persistent stability. The patient reported significant reduction of arm and neck pain. Considering the benign nature of the histopathological diagnosis of hemangioma, no further oncological therapy was warranted. The aim of the surgery was twofold: restoration of alignment and stabilization of the targeted segment in the cervical spine, both of which were achieved.



**FIG. 3.** Three-dimensional cervical spine CT reformats showing the absence of the left C4 pedicle as well as thinning and elongation of the C5 pedicle (left) and the dysplastic lateral mass involving the C4 and C5 levels (right).



**FIG. 4.** Sagittal T1-weighted MRI illustrating midline structures with notable callosal dysgenesis and an adjacent callosal lipoma.

## Discussion

### Observations

The first report of unilateral absence of a cervical pedicle was by Hadley et al. in 1946, in which he described three cases of unilateral congenitally absent pedicles in the subaxial cervical spine.<sup>3</sup> This abnormality has also been recognized in a medieval skeleton, demonstrating its presence centuries ago.<sup>4</sup> After reviewing the literature, we found reports of more than 70 cases of cervical absence



**FIG. 5.** Standing cervical spine radiograph at the 6-week follow-up.

of a pedicle, most of which involved the lower subaxial spine and were unilateral. Congenital absence of a pedicle has been described in the thoracic and lumbar spine and may be associated with coronal dislocation.<sup>5</sup> Absence of a pedicle in the lumbar spine has also been reported in conjunction with spondylolisthesis.<sup>6</sup>

Congenital absence of a pedicle is a rare entity, usually described and seen as asymptomatic or found incidentally with radiographs after trauma.<sup>7</sup> Dynamic instability and spondylolysis have been described with this entity as well as myelopathy requiring intervention.<sup>8,9</sup> Many cases are reported in association with other findings such as spina bifida, dysplastic facet, and conjoined nerve roots.<sup>10–13</sup> The morphological findings with this entity are characteristic and have been described to demonstrate a triad of an enlarged foramen and dorsally displaced lateral mass.<sup>7</sup> Furthermore, the absence of a cervical pedicle has been reported with spondylolysis and has been misdiagnosed as facet dislocation.<sup>14,15</sup> Song et al.<sup>15</sup> described three cases of absence of a cervical pedicle, two of which were initially thought to represent facet dislocation and one of which was associated with spondylolisthesis.

The osseous spine develops from three mirrored pairs of chondrification centers, corresponding to the formation of the vertebral body, pedicle and lateral mass, and the lamina with the corresponding spinous process.<sup>16</sup> The pathophysiology is described as a failure of normal development of a chondrification center during the embryological formation of the spine. This in turn leads to the absence of a pedicle and abnormal development of the lateral mass.<sup>17</sup> Being congenital but not hereditary, it is thought to be a consequence of aberrancy in development but is not necessarily genetic. Although there have been cases of pedicle dysplasia throughout the spine reported in association with neurofibromatosis type 1, some have been associated with scoliosis.<sup>18</sup> Mandell reported on three distinct cases associated with neurofibromatosis type 1 with unilateral absence, bilateral aplasia, and hypoplasia, which is thought to arise as a consequence of mesodermal dysplasia.<sup>19</sup> Idiopathic scoliosis on its own has also been associated with abnormal pedicle morphology.<sup>20</sup> Sarwahi et al. have described a higher incidence of abnormal pedicles in patients with idiopathic scoliosis.<sup>20</sup>

There have been prior reports of erosion of the pedicle and posterior elements in the cervical spine from vascular causes.<sup>21</sup> Taguchi et al. have reported on a case with unilateral pedicle aplasia that presented with an epidural hematoma.<sup>22</sup> However, in their reported case, there was no associated vascular anomaly. There have also been cases with erosion from metastases resembling aplasia of the pedicles.<sup>23</sup> Hemangiomas have also been described with osseous destruction. In a series of seven cases, Gao et al. report the presence of an epidural extension of spinal hemangiomas.<sup>24</sup> There were no cases in which the cervical spine was involved, and in one case the epidural extension was seen to be a consequence of osseous erosion. The appearance, however, seems to be distinct from our case, in which the cortical borders were expanded and the relationship between the posterior and anterior elements of the spine was preserved.

Hemangiomas are benign hamartomatous aggregates of sinusoidal vessels with associated bony expansion and erosion.<sup>25</sup> They are a common incidental finding, with a reported prevalence of 10%–30% of the population, and are most commonly seen in the lower thoracic spine.<sup>26,27</sup> A characteristic polka dot appearance on CT is usually seen representing sclerosis of trabeculae, seen on MR as hyperintense on T1 and T2 sequences.<sup>27</sup> These lesions are rarely symptomatic and usually do not warrant any treatment.

However, occasionally they may present with pain or neurological compromise from the expansion of vertebral cortex, bone erosion, and epidural extension.<sup>28</sup> With significant bone erosion, pathological fractures may also occur.<sup>29</sup> These lesions are usually asymptomatic and require treatment. There are reported treatments with radiotherapy or embolization when these lesions are symptomatic or warrant treatment.<sup>30</sup>

Vertebral hemangiomas are a common finding; however, they are less often found in the cervical spine, as in our case. Although associated with erosion and expansion of bone, we have not found an association with complete erosion of a pedicle. The presence of the hemangioma, although unlikely, may have contributed to abnormal development of the spine. This would theoretically occur if the lesion was present early during the development of the spine; however, this has not been previously reported.

Our case represents a unique conglomeration of developmental anomalies. In isolation, these findings may be noted as incidental aberrancies in development. The combination of these abnormalities has not been described previously; a unifying syndrome is possible but none has been previously described. The underlying pathophysiology as well as the progressive nature of our patient's deformity are also interesting. There have been multiple previous reports of the absence of a cervical pedicle as shown above, often found incidentally with no associated clinical manifestation. However, in our case, the presence of a contiguous hemangioma involving the vertebral body and lateral mass seems to contribute to the instability and progressive anterolisthesis. The morphological appearance of the cervical spine in our patient alludes to the possibility of unilateral agenesis of a cervical pedicle rather than erosion caused by the hemangioma. This is seen by the complete absence of the left C4 pedicle, elongation and thinning of the left C3 and C5 pedicle, and abnormal morphology of the facet. There are also features of osseous erosion from the contiguous hemangioma, such as cortical thinning of the associated vertebral body as well as extraosseous extension.

The incidental finding of callosal dysgenesis and lipoma can be associated with certain pathologies, such as spinal dysraphism,<sup>31,32</sup> but we have not recognized an association after reviewing the literature with pedicle agenesis or vertebral hemangioma. However, an unrecognized association between these anomalies is possible. Morphological differences in spinal anatomy can create a challenge for the determination of pathology, the decision on the best course of treatment, and surgical planning. Recognition of instability and careful assessment of the osseous anatomy were vital in our case, which was possible with the use of multiple imaging modalities in addition to three-dimensional reconstruction.

## Lessons

Meticulous inspection of radiographic imaging as well as use of multiple imaging modalities are important in determining morphological, structural, and mechanical pathology in the spine. We describe a unique case with multiple distinct anomalies within the neurocranium and cervical spine. Demonstrated in our case is the concomitant presentation of a focal kyphotic deformity with unilateral absence of a cervical pedicle in association with a contiguous hemangioma as well as an incidental finding of callosal dysgenesis and lipoma. After a review of the literature, we found no prior report of these findings in conjunction. If multiple abnormalities are noted, a unifying clinical etiology or syndrome must be considered but may

not be described previously. Although evidence remains preliminary, physicians must be vigilant of this possible etiology. Lastly, further research is warranted to confirm the observed association.

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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: Alsuwaihel. Acquisition of data: Alsuwaihel. Analysis and interpretation of data: El-Mughayyar. Drafting the article: all authors. Critically revising the article: El-Mughayyar, MacLennan, Attabib. Reviewed submitted version of manuscript: El-Mughayyar, MacLennan, Attabib. Approved the final version of the manuscript on behalf of all authors: El-Mughayyar. Administrative/technical/material support: El-Mughayyar. Study supervision: El-Mughayyar, Attabib.

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