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Case Report

A case of coexistence of multiple vascular anomalies including the absence of a left internal carotid artery, a left vertebral artery arising from the subclavian artery in a high cervical location, and a bovine arch associated with vertebral fusion ☆,☆☆

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ABSTRACT

We report a rare 16-year-old male case of Klippel-Feil anomaly associated with fetal alcohol syndrome exhibiting complex congenital vascular anomalies. The congenital vascular anomalies observed were the absence of a left internal carotid artery, a left vertebral artery arising from the subclavian artery in a very high cervical location and a bovine arch. The vascular and vertebral anomalies were evaluated using CT and MRI before cervical surgery.

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Introduction

Klippel–Feil syndrome (KFS) is a congenital skeletal anomaly characterized by the presence of 2 or more fused cervical vertebrae [1,2]. KFS is a rare genetic disorder with 6 heterogenous phenotypes. Klippel–Feil anomaly (KFA) is defined as fused vertebrae anatomically. Fetal alcohol syndrome (FAS) results from intrauterine exposure to alcohol and is associated with multiple congenital anomalies, including cardiovascular and skeletal system anomalies, and KFA is one of the associated skeletal anomalies [3–7]. Furthermore, patients with KFS or FAS demonstrate additional developmental anomalies, including cardiovascular, urogenital, and musculoskeletal system anomalies [6–13].

A clinically important morphological variant of the cervical spine is the complete or partial fusion of 2 or more cervical vertebrae because of a defect in the formation or segmentation of the cervical spine. The most common symptoms in patients with KFA include pain and neurological manifestations, such as numbness and a decreased range of cervical motion. However, patients with KFA and cervical stenosis are at an increased risk of spinal cord injury after minor trauma because of hypermobility of various cervical segments [8,14].

Preoperative evaluation of the exact anatomy of fused vertebrae and supra-aortic vascular malformations is important because congenital vascular anomalies of the aorta, carotid arteries, subclavian artery, and vertebral artery are associated with KFS [9–14].

We report a case of KFA with multiple congenital cardiovascular anomalies. The complex anomalies of fused cervical vertebrae, absence of a left internal carotid artery, and a left vertebral artery arising from the subclavian artery in an extremely high cervical location could be evaluated using computed tomography and magnetic resonance imaging before surgery for spinal canal stenosis.

To the best of our knowledge, this was the first KFA case with complex vascular anomalies in a patient with FAS.

Case report

An 18-year-old male with short stature (151 cm) presented with a 1-year history of numbness in the left hand. The patient sometimes surfed. The patient was diagnosed with FAS and underwent surgical treatment for atrial septal defect, ventricular septal defect, and patent ductus arteriosus at the age of 1. Mild mental retardation and attention deficit–hyperactivity disorder are associated. The patient had congenital fusion of the bilateral radius and ulna. Abdominal ultrasonography revealed a horseshoe kidney. Plain radiography revealed fusion from C4 to Th2 with right to convex scoliosis (Fig. 1). Computed tomography angiography from the neck to the upper chest was performed. A fusion image of the cervical spine and arteries was obtained. A bovine arch with a thin left common carotid artery, absence of a left internal carotid artery, and a left vertebral artery arising from the subclavian artery in an extremely high cervical location were found (Fig. 2). The left middle cerebral artery was filled from the ticked left vertebral artery via the posterior communicating artery (Fig. 2). The vertebrae from C4 to Th2 were fused, and the lower transverse foramina were absent. The right second rib was hypoplastic. Both vertebral arteries flowed into the transverse foramina at the middle position of the fused vertebrae. Magnetic resonance imaging was performed and revealed fused C4–Th2 with scoliosis and spinal cord stenosis with high signal intensity at the C3/4 level, suspected for myelomalacia on T2-weighted imaging (Fig. 3). His hair line was normal, and a short neck was not present. The patient did not present with the characteristic facial features of FAS. No Sprengel deformity or omovertebral bone was observed.

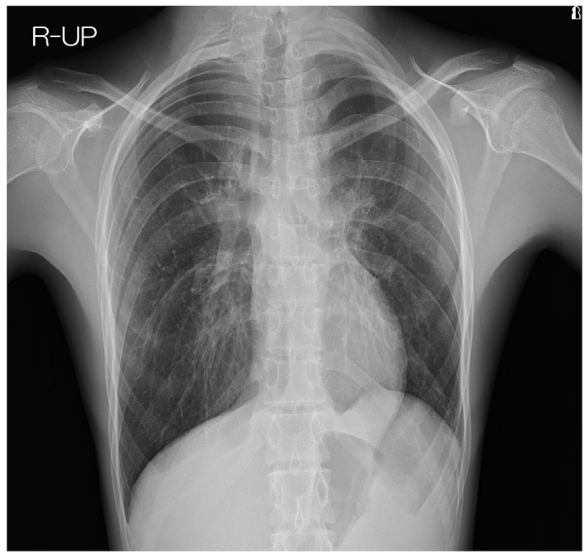

Fig. 1 – Plain radiograph reveals fusion from C4 to Th2 with right to convex scoliosis and a hypoplastic right second rib.

The patient was suffering from mild radiculomyelopathy that was caused by the C3/4 instability associated with the congenital fusion of C4-T2. A posterior fusion surgery was performed by inserting lateral mass screws into C3 and lamina screws into the fused cervicothoracic mass, along with an iliac bone graft on the laminae wedged between the spinous processes of C3 and the fused lower cervical vertebrae. A cervical collar was applied for 1 month, and follow-up radiographs confirmed fusion.

Discussion

Congenital vertebral fusion (also known as KFA) is associated with KFS, FAS, Goldenhar syndrome, and Wildervanck syndrome [15]. Interruption of the early embryonic blood supply in the subclavian arteries, vertebral arteries, and/or their branches has been proposed as an etiology of congenital vertebral fusion [10,16]. Others consider aberrant differentiation of postotic neural crest cells to be a crucial factor for the development of vertebral abnormalities [17].

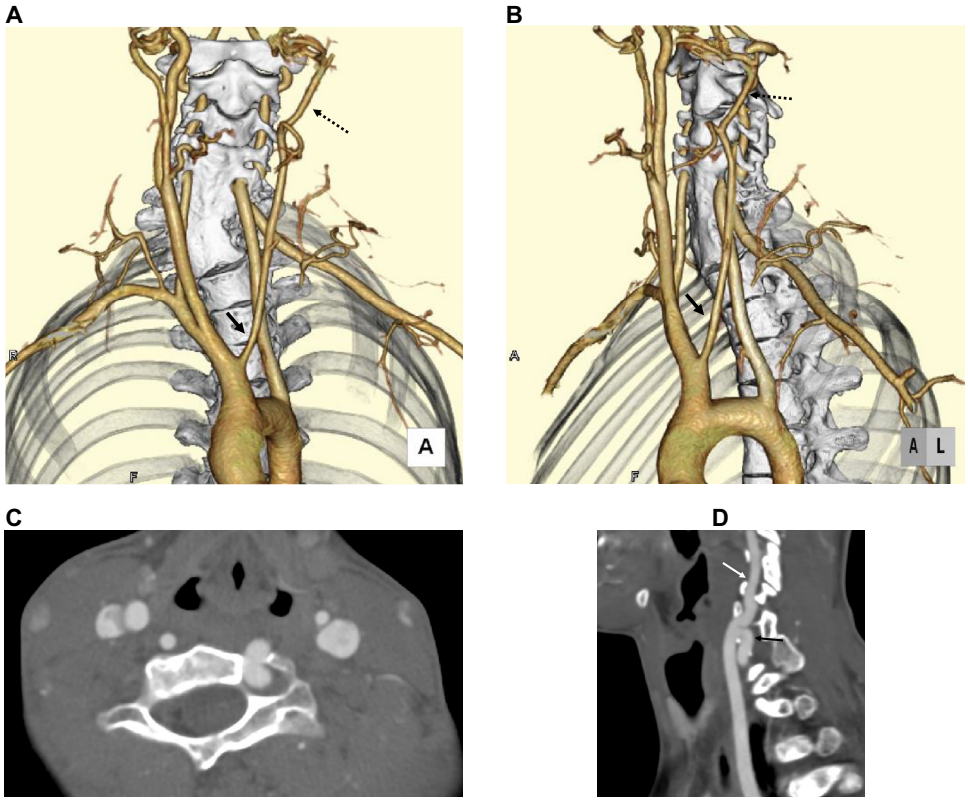

Fig. 2 – (A and B) Volume rendering images of fused CT angiography and bone (A. anterior image, B. left anterior oblique image) showing a bovine aortic arch with a thin left common carotid artery (black arrows), the absence of a left internal carotid artery, a left vertebral artery arising from the subclavian artery in an extremely high cervical location, and scoliosis with fused C4–Th2. The bilateral lower transverse foramina were absent. Dotted arrows indicate the left external carotid artery. Axial images (C) and covered multiplanar reconstruction (D) show that the left subclavian artery turns around and goes down within the transverse foramen (black arrow). White arrow shows a left vertebral artery.

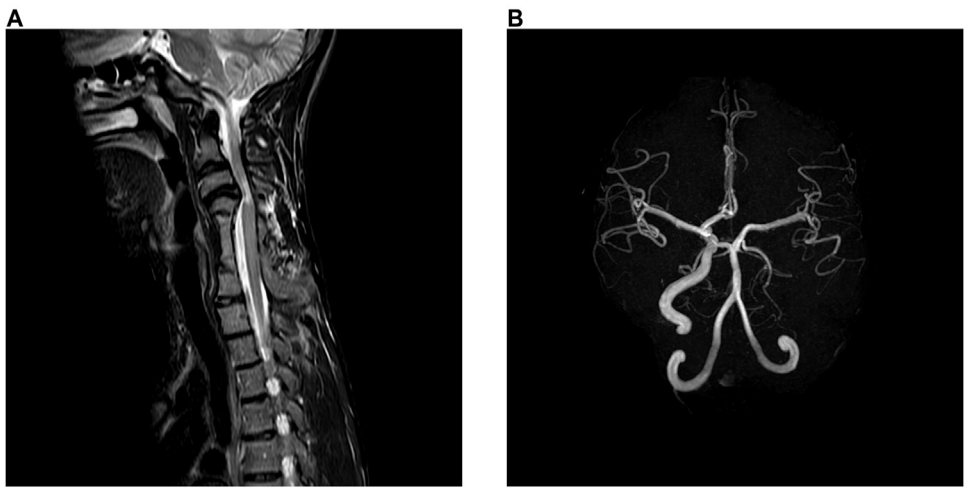

Fig. 3 – Magnetic resonance (MR) images of the cervical spine and brain angiography. (A) Sagittal T2WI reveals fused C4–Th2 and spinal cord stenosis with high signal intensity at the C3/4 level (arrow). (B) MR angiography reveals the absence of a left internal carotid artery. The left middle cerebral artery is inflow from the posterior communicating artery.

Congenital cardiovascular anomalies in FAS include ventricular and atrial septal defects. Congenital arch anomalies in KFA include mid-aortic syndromes, and aortic arch anomaly have not been reported [18]. Congenital vascular anomalies of the carotid, subclavian, and vertebral arteries are known to be associated with KFS because of embryology [10–13]. In KFA, congenital vascular anomalies are likely to occur as well as in KFS. However, congenital vascular anomalies that involve the carotid, vertebral, and subclavian arteries and aortic arch were not previously reported.

Our case had complex cardiovascular anomalies, including the absence of the left internal carotid artery, a left vertebral artery arising from the subclavian artery in an extremely high cervical location, and a bovine arch. Only 2 cases of KFS with a left vertebral artery arising from the subclavian artery in an extremely high cervical location have been reported [12,13]. In one of them, severe Sprengel deformity with omovertebral bone fused to the first block of the cervical arch was reported [16]. Another case with a Sprengel deformity combined with a left vertebral artery arising from the subclavian artery in an extremely high cervical location and bovine arch was also reported [13].

Two cases of KFS with the absence of an internal carotid artery have been reported [10,11]. The first case was associated with hypoplastic left common carotid and vertebral arteries, and the left middle cerebral artery flowed from the basilar artery [10]. The second case of postaxial polydactyly was associated with hypoplastic left common carotid and left vertebral arteries [11].

Individuals with KFA with few or mild extra-axial anomalies are asymptomatic during childhood [8,10,11]. Cardiovascular or renal anomalies may require intensive treatment. Cervical instability, occipitocervical anomalies, disk or joint degeneration, and scoliosis develop over time and lead to neurological complications [2,8,14]. Individuals with KFS and cervical stenosis may be at an increased risk of spinal cord injury after minor trauma because of hypermobility of various cervical segments [19,20]. Two KFS cases of acquired vertebral arterial dissection have been reported [19–21]. In such cases, anterior vertebral subluxation of C1 on C2 or fused vertebrae with stenotic transverse foramen is a significant risk factor for vascular accident. Furthermore, vascular anomalies of abnormal course contribute to vascular injury, such as dissection or pseudoaneurysm, and the surgical treatment for these anomalies is more complex. Understanding the correct anatomy of vascular and vertebral anomalies in patients with KFS is important. CT and/or MRI with angiography using volume rendering and multiplanar reconstruction images should be used for evaluation before spinal surgery.

Conclusion

We report a KFA case with complex congenital vascular anomalies including the absence of a left internal carotid artery, a left vertebral artery arising from the subclavian artery in a very high cervical location, and a bovine arch in patient with FAS. The correct anatomy of vascular and vertebral anomalies could be investigated using CT and MRI before surgery.

Patient consent

Informed consent for publication of their case was obtained from the patient.

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