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Head and Neck

Anomalous vertebral arteries in Klippel-Feil syndrome with occipitalized atlas: CT angiography

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ABSTRACT

Klippel-Feil syndrome is an uncommon anomaly that may be asymptomatic. Early clinical signs such as restricted neck motion or short neck can be subtle and incorrectly treated as spasms. High incidence of associated craniovertebral junction (CVJ) anomalies such as occipitalized atlas predisposes them to serious neurologic complications requiring invasive procedures and surgeries. However, these often have anomalous vertebral artery course which is more prone to injury during CVJ procedures, and also sparsely known in radiology literature. We demonstrate the importance of computed tomography angiography in preprocedural planning to avoid catastrophic injury to anomalous vertebral artery at CVJ in such case.

Introduction

Klippel-Feil syndrome (KFS), the congenital fusion of 1 or more cervical vertebrae, is associated with various skeletal anomalies, including craniovertebral junction (CVJ) anomalies. This may lead to progressive neurologic compromise, but it can be subtle or asymptomatic in early stages [1]. Thus, early detection by the radiologist could allow appropriate prevention and management plan, avoiding devastating consequences. Further, understanding of related vascular anomalies could help avoid injury to the potentially anomalous vertebral artery (VA) during procedures at the CVJ in such cases.

Case report

An 11-year-old boy presented with restricted range of neck motion. He was treated with antispasmodics but symptoms continued to worsen gradually. Plain radiographs of the neck revealed C2-C3 vertebral fusion. Computed tomography (CT) angiography (CTA) of the neck was performed after neurosurgery consult in anticipation of the upper cervical spine or CVJ surgery. Sagittal (Fig. 1A) and coronal (Fig. 1B) CTA images through the neck in bone window showed C2 through C3 cervical vertebral fusion (small white arrow) and occipitalization of the atlas vertebra (black arrow) associated with basilar

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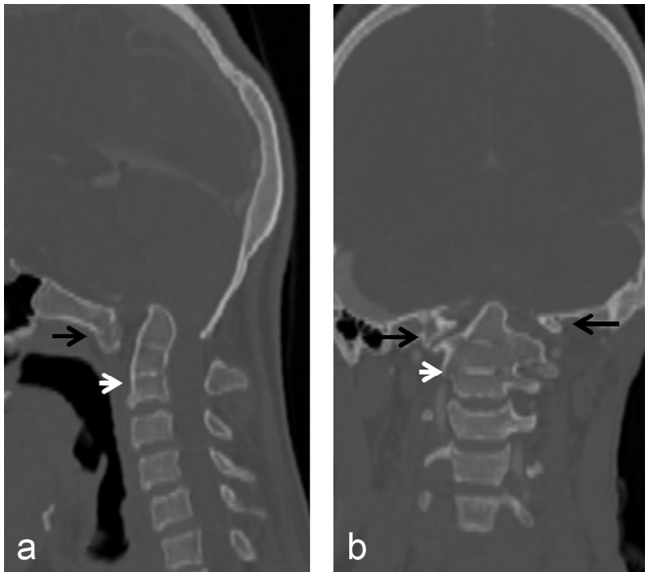

Fig. 1 – (A) Sagittal computed tomography angiography (CTA) image through the neck in bone window in an 11-year-old boy shows C2-C3 cervical vertebral fusion (small white arrow) consistent with Klippel-Feil syndrome (KFS). The C1 vertebra (atlas) is also fused with occiput, consistent with occipitalization of the atlas (C1) vertebra (black arrow). Tip of the dense process is just above the level of foramen magnum, consistent with basilar invagination. (B) Coronal CTA image through the neck in bone window in an 11-year-old boy shows C2 -C3 cervical vertebral fusion (small white arrow) consistent with KFS. The C1 vertebra (atlas) is also fused with occiput, consistent with occipitalization of the atlas (C1) vertebra (black arrow). Tip of the dense process is just above the level of foramen magnum, consistent with basilar invagination.

invagination. These features are consistent with KFS. Coronal CTA image in bone window (Fig. 2) showed the vertebral arteries passing through the corresponding bony canals between the fused Atlas vertebra and occiput. MRI of the spine, ultrasound of the abdomen, and echocardiography did not reveal any additional abnormalities.

Discussion

KFS is defined as the congenital fusion of 2 or more cervical vertebrae. It most frequently involves C2-C3 vertebrae. Exact incidence is still unknown, but it has been reported in up to 0.05% of live births [1]. It is considered a sporadic anomaly, but may present as part of other clinical syndromes. Exact etiology is unclear, but global fetal insult, vascular disruption, and different gene mutations have been suggested as the cause. The classical clinical triad of restricted neck motion, short neck, and low hairline is, in fact, seen in <50% of cases, some present with torticollis, or it may be incidental in many [1,2]. Our case presented with restricted cervical range of motion, which is the most consistent of the clinical triad [1,2]. The more extensive the fusion, the more likely is to exhibit 1 or more features of the clinical triad, in addition to developing myelopathic and radiculopathic symptoms [1].

Although our patient did not develop any neurologic symptoms yet, these can be an important consequence of KFS, as fusion of a cervical segment (such as C2-C3) will shift mobility to another segment (C1-C2) and result in hypermobility in the latter segment, increasing the risk of adjacent degenerative changes and disk herniation with its associated sequela [1,2]. This can be even more pronounced if associated with other anomalies at the CVJ, which in our case were occipitalization of the atlas (OA) (the fusion of the occiput with atlas vertebra) and basilar invagination (the upward displacement of vertebral elements into a normal foramen magnum).

OA per se has been reported in about 0.08%-3% of the general population [3] and is in up to 30%-50% cases of KFS [1]. The foramen magnum in cases of OA is often found to be severely impaired, especially when associated with basilar invagination; this is associated with increased risk of compressive brainstem and cord injury. Chiari malformation is also a common associated anomaly with OA due to small posterior cranial fossa [3]. Although our patent did not have any of these complications, it is important to recognize these as future risks and take appropriate preventive interventions. KFS is often associated with other skeletal and organ anomalies as well. Thoracic scoliosis is the most common in these cases, which sometimes occur as a compensation for the cervical scoliosis. Sprengel deformity, characterized by congenital elevation of the scapula, in addition to renal and cardiac anomalies, is also commonly seen. Other anomalies include cleft palate, craniofacial anomalies, abnormal ears, spinal dysraphism, and syringomyelia [1,4].

Plain radiograph of the neck, in both lateral and anteroposterior views, is the initial modality for diagnosis [1]. This method can show varying degrees of cervical vertebral fusion in many cases. In some, this method also shows high riding scapula and rib anomalies, which were not seen in our case. Further workup is variable, but when CVJ anomaly is in question

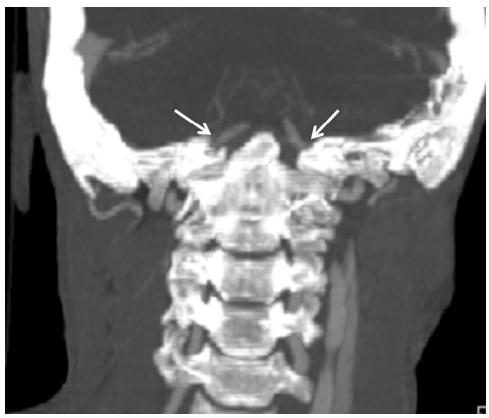

Fig. 2 – Coronal maximum intensity projection computed tomography angiogram image in bone window shows both the vertebral arteries coming out of the respective bony canal through the fused/occipitalized bone (white arrows) between the fused Atlas (C1) vertebra and occiput, before their intracranial course.

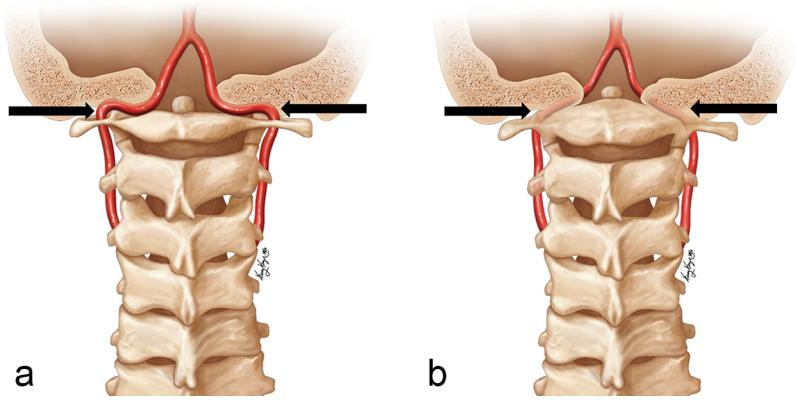

Fig. 3 – (A) A coronal illustration of the computed tomography angiography (CTA) of the neck-craniovertebral junction in a normal patient demonstrates the normal course (arrows) of the V3 segment of vertebral artery (VA) above the posterior arch of atlas. (B) A coronal illustration of the CTA of the neck-craniovertebral junction in a patient with occipitalized atlas demonstrates the abnormal course (arrows) of V3 VA through the respective bony canals within the fused bone between the atlas and occiput.

as in our case, CTA is an important imaging modality for evaluation. CT scan with 3-dimensional reconstruction is useful for confirming and finely delineating the skeletal deformities [5]. CT revealed atlanto-occipital fusion in our case, altering the management all together. Using angiogram protocol during CT scan is helpful to detect associated VA course which can be anomalous in these cases. Magnetic resonance imaging of the spine is important to detect or exclude other craniospinal anomalies such as syringomyelia, diastematomyelia, spinal canal changes, and Chiari malformations [5]. Finally, to complete the workup, ultrasound of the abdomen is performed to rule out renal anomalies, and echocardiography to exclude cardiac anomalies, most commonly ventricular septal defect [5].

In our case, radiologic features of C2-C3 fusion with associated OA was diagnostic of KFS, and this was an isolated abnormality. KFS can also be seen as part of other entities such as fetal alcohol syndrome, Goldenhar syndrome, and anomalies of the extremities. These syndromes are more of a clinical diagnosis, whereas KFS is a radiologic diagnosis. Other differential diagnosis includes ankylosing spondylitis, juvenile rheumatoid arthritis, and Wildervanck syndrome. Ankylosing spondylitis usually has associated sacroiliac joint erosions and ankylosis. Juvenile rheumatoid arthritis causes inflammatory arthropathy of the peripheral joints, commonly affecting the hands, in addition to cervical spine involvement. Wildervanck syndrome is very rare, and it includes KFS as part of its triad with deafness and eye motion abnormalities.

Treatment is usually variable depending on the presentation, set of deformities, location, and severity. Surgery is usually contemplated in cases with spinal instability or failure of conservative management [4,6,7]. Nevertheless, before attempting any surgical procedure on cases with cervical vertebra or CVJ anomalies, defining the course of VA is important. With OA for example, VA is almost always associated with abnormal course, with 78.4% traversing through a bony canal between the fused atlas and occiput as seen in our case (Fig. 3), and 20.3% VAs passing below C1. Operating without knowledge of such abnormal course could have devastating consequences if injury to VA occurs during the procedure [8]. In conclusion, KFS is often associated with CVJ anomalies such as OA, which, in turn, can be associated with anomalous VA course. Hence, CTA is useful to prevent potential catastrophic VA injury during procedures at CVJ in such cases.

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