Klippel-Feil syndrome and supra-aortic arch anomaly: A case report

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Patients with Klippel-Feil syndrome have an increased incidence of supra-aortic arch anomalies. The present report presents the case of a

patient who, during evaluation of his carotid arteries for nonspecific symptoms, was found to have an anomaly of the supra-aortic branches and Klippel-Feil syndrome.

Key Words: *Cervical anomaly with Klippel-Feil syndrome*

Patients with Klippel-Feil syndrome (KFS) have an increased incidence of supra-aortic arch anomalies. We report a case of a patient who, during evaluation of his carotid arteries for nonspecific symptoms, was found to have an anomaly of the supra-aortic branches and KFS.

KFS includes any form of congenital cervical vertebral fusion anomaly. Several vertebral, neurological, cardiac and vascular anomalies are associated with the syndrome. The classic clinical triad includes a short neck, low hairline and limitation in movement of the neck. The triad is seen in only 40% to 50% of KFS patients (1,2).

CASE PRESENTATION

A 77-year-old man was referred for evaluation of his carotid arteries for the nonspecific symptoms of occipital headache, nausea and neck discomfort. On examination, he had a short neck and a marked limitation in the range of movements of his neck. His blood pressure was normal and equal in both arms.

A duplex scan of the neck suggested moderate disease in the right internal carotid artery (ICA), but the vertebral artery was larger than the ICA.

Computed tomography angiography of the neck was performed. There was fusion of the C2 through to the C5 anterior and posterior vertebral elements (Figure 1). There was also evidence of fusion of the posterior elements of C7 through to T2, suggesting a type 3 KFS (see Discussion). Cervical ribs were present, with bony contribution from C6 on the left and a rudimentary cervical rib at C6 on the right. There was no common carotid artery (CCA) on the right side, and the external carotid artery (ECA) and ICA originated from the brachiocephalic artery (Figures 2 and 3). The right vertebral artery was dominant and 35% larger than the right ICA. It had a retropharyngeal course up to the foramen transversarium of C4. The left CCA, ICA, ECA and the vertebral arteries were normal. Intracranially, there was a 1.5 mm aneurysm of the superior portion of the anterior communicating artery and a fusiform dilation of the cavernous carotid artery on the left.

Figure 1 Fusion of the anterior and posterior vertebral elements of vertebrae C2 through C5, suggesting Klippel-Feil syndrome. Note: The external carotid artery (ECA) is difficult to follow from its origin on maximum intensity projection images. ICA Internal carotid artery; Vert Vertebral artery

DISCUSSION

In 1912, Maurice Klippel and Andre Feil first described a patient who presented with a short neck, low posterior hairline and limited range of motion. KFS is defined as congenital fusion of two or more cervical vertebrae and is believed to result from faulty segmentation along the embryo's developing axis during weeks 3 to 8 of gestation. KFS is reported to be present in one of 42,000 individuals; 57% to 70% of all patients are female. Genetic analysis revealed an autosomal recessive trait for types 1 and 3, and an autosomal dominant trait for type 2. Three types of KFS have been described (3):

- Type 1: Cervical spine fusion in which elements of many vertebrae are incorporated into a single block.
- Type 2: Cervical spine fusion in which complete segmentation fails at only one or two cervical levels and may include an occipitoatlantal fusion.

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Figure 2) Imaging showing the right external carotid artery (ECA) and internal carotid artery (ICA) originating from the brachiocephalic artery. There is no common carotid artery on the right. The right vertebral artery (Vert) is dominant and retropharyngeal in its course.

Figure 3) Posterior oblique view showing the abnormal right carotid arteries. The left common carotid artery (CCA), external carotid artery (ECA), internal carotid artery (ICA) and the vertebral (Vert) arteries are normal. Note: The origin of the ECA is not well seen on maximum intensity projection images

- Type 3: Type 1 or 2 fusion, with coexisting segmentation errors in the lower dorsal or lumbar spine.

The classical clinical triad of KFS is a low neckline, short neck, and restriction of head and neck movements; it is only present in 40% to 50% of patients (2).

Different conditions can be seen with KFS, including skeletal system abnormalities (scoliosis and/or kyphosis [60%], Sprengel's deformity [30%] and torticollis), urinary system abnormalities (35%), loss of hearing (30%), facial asymmetry and flattening of the neck (20%), synkinesis or mirror movements (20%), and congenital heart disease (4% to 14%). Brain stem anomalies, congenital cervical stenosis, adrenal aplasia, ptosis, facial nerve paralysis, syndactylia and diffuse hypoplasia in the upper extremities may also be seen (2,4-7).

The bifurcation of the CCA is commonly between the C3 and C5 vertebral levels (Figure 4). Low bifurcations or separate origins of the carotid arteries are extremely rare, with only a few reported cases (8). The persistence of the ductus caroticus, associated with partial or complete involution of the third arch, increases the distance separating the CCA and ECA embryological origins, leading to a low bifurcation of the CCA, or separate origins of the ECA and ICA (9). An

Figure 4) Schematic representation of the carotid bifurcation development. **A** Normal development of the carotid bifurcation. The third arch (III) constitutes the proximal part of the carotid axis, which is divided into the common carotid artery (CCA) and proximal internal carotid artery (ICA), either by in situ sprouting of the ventral pharyngeal artery or by its migration from the aortic sac. The distal ICA is formed by the dorsal aorta distal to the ductus caroticus (DC). The DC is the segment of dorsal aorta located between the third and fourth (IV) aortic arches. Its regression at the 12 mm to 14 mm stage allows the carotid axis to take its definitive appearance. **B** Separate origins for the external carotid artery (ECA) and ICA from the aorta occur when the third aortic arch involutes instead of the DC (illustrated on the left side). The same process may occur on the right side, the separate origins of the ECA and ICA arising, in that case, from the innominate artery. **C** Low carotid bifurcations are formed when, in addition to the situation described in **B**, there is migration of the ECA origin toward the DC, and reconstitution of a short CCA. DA Ductus arteriosus; SCA Subclavian artery. Reproduced from references 9 and 11

abnormal origin of the left internal mammary artery during coronary artery bypass grafting has also been reported (10). A subclavian artery supply disruption sequence has been hypothesized to result in KFS on the basis of the theory that vascular disruption of the vertebral artery leads to ischemia during morphogenesis (9). Because of the complex vascular variants in this group of patients, investigative imaging of the aortic arch and supra-aortic vessels is recommended. These variants can cause diagnostic pitfalls by lying outside the region of interest in magnetic resonance angiography and can also render carotid endarterectomy challenging (10).

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