Klippel-Feil Syndrome and Sprengel Deformity Combined with an Intraspinal Course of the Left Subclavian Artery and a Bovine Aortic Arch Variant

F. Floemer, O. Magerkurth, C. Jauckus, J. Lütschg and J.F. Schneider

doi: https://doi.org/10.3174/ajnr.A0878
http://www.ajnr.org/content/29/2/306
Klippel-Feil syndrome and Sprengel Deformity Combined with an Intraspinal Course of the Left Subclavian Artery and a Bovine Aortic Arch Variant

SUMMARY: We present a case of Klippel-Feil syndrome and Sprengel deformity with a bovine aortic arch and an aberrant course of the left subclavian artery in a 14-year-old boy. CT and MR imaging of the neck and upper thorax demonstrated a cervical osseous segmentation anomaly, a left common carotid artery originating from the innominate artery, and a left subclavian artery coursing through the intraspinal space at the C6 through T1 level. Possible embryonic mechanisms and clinical significance of this variant are reviewed.

Discussion

In 1912, Maurice Klippel and Andre Feil first described a patient presenting an association of short neck, low posterior hairline, and limited range of motion. The complete clinical triad is seen in <50% of the cases, and many patients with KFS demonstrate additional developmental anomalies. Therefore, KFS is diagnosed by the presence of 2 or more nonsegmented cervical vertebrae alone.1

KFS is reported to be present in 1 of 42,000 individuals, and 57%–70% of all patients are female. Various classification schemes were developed. KFS was classically categorized into 3 groups, depending on the level and extent of vertebral fusion. Genetic analysis revealed an autosomal recessive trait for types I and III and an autosomal dominant trait for type II. Subsequent studies incorporated the mode of inheritance with the location of the most rostral vertebral fusion. This system includes 4 groups with different autosomal dominant or recessive or X-linked traits.

Anomalies of the aortic arch, carotid arteries, subclavian artery, vertebral artery, and a persistent trigeminal artery are known in association with KFS.2-6

SD has been found in 7%–42% of the patients with KFS. The scapula develops from the paraxial mesoderm and is, therefore, closely associated with the development of the cervical spine. Patients with KFS and SD show more congenital anomalies and cervical spine malformations than patients with only KFS.7

Cervical spine development starts with gastrulation in the fourteenth week by formation of 3 germ cell layers and the notochord. Between days 20–30, the paraxial mesoderm subdivides into somites, which divide into sclerotomes and der-
Momotomes. Sclerotomes resegment into a cranial and a caudal area, and in between forms the intervertebral disk. Fusion of the caudal section from a rostral somite with the cranial section of the corresponding caudal somite forms a vertebral body. Errors in segmentation may result in KFS. Vascular system development runs parallel to the development of the spinal cord. At days 21–29, thirty pairs of dorsal intersegmental arteries arise from the dorsal aortae and supply their corresponding somites. Between days 32–42, vertical and right-angle anastomoses develop between them and cross the vertebral bodies. The anastomoses serve as the origin of the vertebral arteries. During gestational days 37–42, the first 6 cervical intersegmental arteries regress, and only the seventh intersegmental arteries persist. The entire left and the greater part of the right subclavian artery originate from this seventh intersegmental artery.

Fetal vascular disruption disorders have been found to be responsible for various congenital anomalies, depending on the extent and timing of the disruptive event. 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. Subsequent structural anomalies will involve not only the definitive vascular pattern by itself but also the vasculature-dependent soft-tissue territories.8,9 Due to the heterogeneity in KFS, the exact influence of vascular and genetic factors remains unclear. Because of the complex vascular variants in this group of patients, prior imaging of the aortic arch and supra-aortic vessels is recommended.10

References