Klippel-Feil Syndrome-A Consortium of Anomalies: A Review

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Abstract

Klippel-Feil syndrome is a bone disorder characterized by the abnormal fusion of two or more cervical vertebrae, which is present from birth. It is an autosomal dominant congenital defect. It is characterized by the classical triad-short webbed neck, low posterior hairline, restricted head and neck movements and commonly associated with a tuft of hair at lumbosacral region and torticollis (abnormal neck position). Many aspects of this syndrome are surfacing and it is not clear whether it is a separate entity or if it is a part of congenital spinal deformities. Only if the link is found between the genetic etiology and phenotypical pathoanatomy, the heterogeneity of the syndrome can be understood. This disease is linked to so many other syndromes and diseases, and the clinician must be well aware of the variations, and common associations in order to avoid a misdiagnosis.

Keywords

Klippel-Feil syndrome, vertebral synostosis, cervical vertebrae fusion, congenital vertebral anomaly, brevicollis, block vertebrae.

Klippel-Feil syndrome (KFS) is a condition affecting the development of the bones in the spine. People with KFS are born with abnormal fusion of at least two spinal bones (vertebrae) in the neck. Common features may include a short neck, low hairline at the back of the head, and restricted movement of the upper spine. Some people with KFS have no symptoms. Others may have frequent headaches, back and neck pain, and other nerve issues. People with KFS are at risk for severe spinal injury. KFS can occur along with other types of birth defects, and sometimes KFS occurs as a feature of another dis... References References. Frikha R. Klippel-Feil syndrome: a review of the literature. Clin Dysmorphol. 2020; 29(1):35-37. https://pubmed.ncbi.nlm.nih.gov/31577545. Klippel-Feil syndrome is a complex heterogeneous entity that results in cervical vertebral fusion. Two or more non-segmented cervical vertebrae are usually sufficient for diagnosis. Epidemiology There is a recognized female predominance 1. Klipp... Diagnostic importance of 3D CT images in Klippel-Feil Syndrome with multiple skeletal anomalies: a case report. Korean J Radiol. 2006;6(4): 278-81. Free text at pubmed - Published citation. 3. Ulmer JL, Eister AD, Ginsberg LE et-al. Klippel-Feil syndrome: CT and MR of acquired and congenital abnormalities of cervical spine and cord. J Comput Assist Tomogr. 17 (2): 215-24. - Published citation. 4. Karasick D, Schweitzer ME, Vaccaro AR. The traumatized cervical spine in Klippel-Feil syndrome: imaging features. AJR Am J Roentgenol. Klippel-Feil Syndrome. NORD gratefully acknowledges Raymond A. Clarke, PhD, Associate Professor, School of Psychiatry, University of New South Wales, Sydney, Australia, for assistance in the preparation of this report. Synonyms of Klippel-Feil Syndrome. cervical vertebral fusion. congenital cervical synostosis. isolated Klippel-Feil syndrome. Klippel-Feil anomaly. KFS. Subdivisions of Klippel-Feil Syndrome. Klippel-Feil syndrome, type I. KFS may occur as an isolated abnormality (Klippel-Feil anomaly) or as a syndrome with associated anomalies. In many individuals with KFS, the condition appears to occur randomly for unknown reasons (sporadically). In other cases, KFS may be inherited as an autosomal dominant or autosomal recessive trait.