Klippel-Feil syndrome/congenital malformations of the cervical spine

Objectives

1. List the clinical features (triad of Feil) noted in children with Klippel-Feil syndrome
2. Discuss workup and management of a child recently diagnosed as having Klippel-Feil syndrome
3. Describe spinal anomalies associated with Klippel-Feil syndrome
4. Discuss the differential diagnosis and diagnostic approach of a child with a short neck and limited motion

Discussion points

1. What type of imaging studies are indicated for evaluation of children with a short neck and limited rotation?
2. What is the etiology of Klippel-Feil syndrome? Should children with Klippel-Feil syndrome play sports?
3. What are indications for surgical intervention in patients with a short neck secondary to Klippel-Feil? To basilar impression? To hemivertebra of C1?

Discussion

A basic understanding of the clinical features of upper cervical spine anomalies are an essential component of the knowledge base for orthopaedists; even though management is generally performed at specialized centers. The prototype of upper cervical spine anomalies is the Klippel-Feil syndrome. This has a characteristic clinical triad of short neck, low hairline, and restricted neck mobility. Klippel-Feil syndrome has traditionally been classified morphologically, however it can be expected that newer classifications based on the genetic locus responsible will be refined. Other system anomalies are common and important. The majority will have hearing deficits. Urinary tract anomalies are important, and ultrasonography is indicated in all children with Klippel-Feil syndrome. Upper cervical spine abnormalities portend more central nervous system anomalies and potential difficulties with instability. MRI imaging has aided in appreciating the incidence and character of contralateral nervous system anomalies. Cardiopulmonary and hand anomalies are also frequent components of the Klippel-Feil syndrome.

Basilar impression and congenital anomalies of C1 can also present with a short neck and limited motion. The exact nature of the anomaly is often difficult to ascertain by plane radiography; CT and MR imaging are essentially routinely indicated as often a combined orthopaedic and neurosurgical approach is indicated. Proprioceptive and cerebellar symptoms are suggestive of basilar impression. Occipitocervical arthrodesis with or without neurosurgical decompression is virtually always necessary for anomalies of C1 and basilar impression.
References


