



POSNA

The Core Curriculum

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 conitrial 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

1. Baba H, Maezawa Y, Furusawa N, Chen Q, Imura S, Tomita K. The cervical spine in the Klippel-Feil syndrome. A report of 57 cases. *International Orthopaedics* 1995;19(4):204-8.
2. Clarke RA, Catalan G, Diwan AD, Kearsley JH. Heterogeneity in Klippel-Feil syndrome: a new classification. *Pediatric Radiology* 1998;28(12):967-74.
3. David KM, Copp AJ, Stevens JM, Hayward RD, Crockard HA. Split cervical spinal cord with Klippel-Feil syndrome: seven cases. *Brain* 1996;119(Pt 6):1859-72.
4. Dubousset J. Torticollis in children caused by congenital anomalies of the atlas. *J Bone Joint Surg(Am)* 1986;68:178.
5. Herman MJ, Pizzutillo PD. Cervical spine disorders in children. *Orthopedic Clinics of North America* 1999;30(3):457-66, ix.
6. Macalister A. Notes on the development and variations of the atlas. *J Anat Physiol* 1983;27:519.
7. McGaughran JM, Kuna P, Das V. Audiological abnormalities in the Klippel-Feil syndrome. *Archives of Disease in Childhood* 1998;79(4):352-5.
8. Moore WB, Matthews TJ, Rabinowitz R. Genitourinary anomalies associated with Klippel-Feil syndrome. *J Bone Joint Surg(Am)* 1975;57:355.
9. Pizzutillo PD, Woods M, Nicholson L, MacEwen GD. Risk factors in Klippel-Feil syndrome. *Spine* 1994;19(18):2110-6.
10. Ritterbusch JF, McGinty LD, Spar J, Orrison WW. Magnetic resonance imaging for stenosis and subluxation in Klippel-Feil syndrome. *Spine* 1991;16(10 Suppl):S539-41.
11. Rouvreau P, Glorion C, Langlais J, Noury H, Pouliquen JC. Assessment and neurologic involvement of patients with cervical spine congenital synostosis as in Klippel-Feil syndrome: study of 19 cases. *Journal of Pediatric Orthopaedics. Part B* 1998;7(3):179-85.
12. Stark EW, Borton tE. Hearing loss and the Klippel-Feil syndrome. *Am J Dis Child* 1972;123:233.
13. Teodori JB, Painter MJ. Basilar impression in children. *Pediatrics* 1984;74:1097.
14. Theiss SM, Smith MD, Winter RB. The long-term follow-up of patients with Klippel-Feil syndrome and congenital scoliosis. *Spine* 1997;22(11):1219-22.
15. Thomsen MN, Schneider U, Weber M, Johannisson R, Niethard FU. Scoliosis and congenital anomalies associated with Klippel-Feil syndrome types I-III. *Spine* 1997;22(4):396-401.
16. Ulmer JL, Elster AD, Ginsberg LE, Williams DWd. Klippel-Feil syndrome: CT and MR of acquired and congenital abnormalities of cervical spine and cord. *Journal of Computer Assisted Tomography* 1993;17(2):215-24.
17. Van Kerckhoven MF, Fabry G. The Klippel-Feil syndrome: a constellation of deformities. *Acta Orthopaedica Belgica* 1989;55(2):107-18.