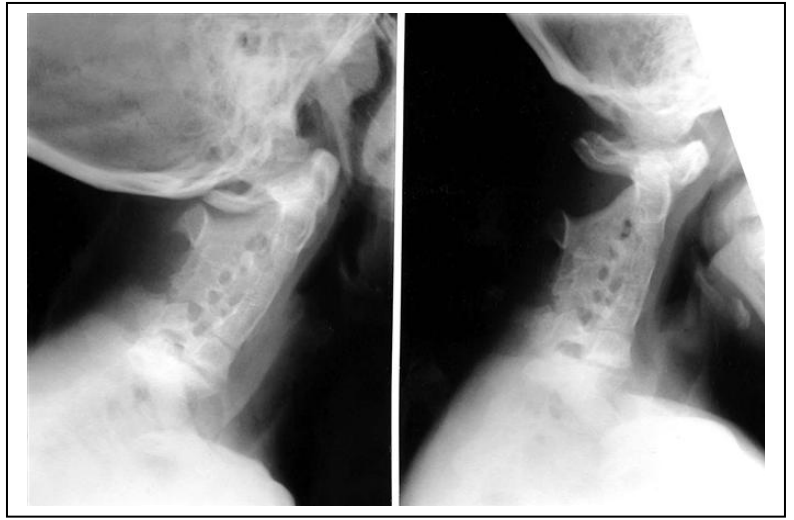


SEND Bulletin NO. 16



Klippel Feil Syndrome

Klippel-Feil Syndrome is a rare disorder characterised by the congenital fusion of two or more cervical (neck) vertebrae. It is caused by a failure in the normal segmentation or division of the cervical vertebrae during the early weeks of foetal development. The fused vertebrae can cause nerve damage and pain in the head, neck, or back. Associated abnormalities may include *scoliosis* (curvature of the spine), *spina bifida* (a birth defect of the spine), cleft palate, respiratory problems, and heart malformations. Other features may include joint pain; anomalies of the head and face, skeleton, sex organs, muscles, brain and spinal cord, arms, legs, and fingers; and difficulties hearing. Most cases are sporadic (happen on their own) but mutations in the GDF6 (growth differentiation factor 6) or GDF3 (growth differentiation factor 3) genes can cause the disorder. These genes make proteins that are involved in bone development and segmentation of the vertebrae.

Treatment

Treatment for Klippel-Feil Syndrome is symptomatic and may include surgery to relieve cervical or craniocervical instability and constriction of the spinal cord, and to correct scoliosis. Physical therapy may also be useful.

Key characteristics

Children with Klippel Feil Syndrome may:

- have a short neck
- have a low hairline at the back of the head
- have restricted mobility of the upper spine

- will need to avoid activities that can potentially injure the neck i.e. contact sports

Support strategies

You may need to:

- have surgery to relieve cervical or craniocervical instability and constriction of the spinal cord to correct scoliosis
- adapt the curriculum to ensure that the child is included as much as possible without risk of further injury
- receive regular physiotherapy from an OT

Support agencies

- <https://rarediseases.org/rare-diseases/klippel-feil-syndrome/>
- <https://contact.org.uk/advice-and-support/health-medical-information/conditions/k/klippel-feil-syndrome/>
- <https://www.ninds.nih.gov/Disorders/All-Disorders/Klippel-Feil-Syndrome-Information-Page>

If you have any interesting information or resources which you would like to share with your colleagues via this fortnightly bulletin, please e-mail them to me:

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Many thanks

Anne