Klippel Feil syndrome

Klippel Feil syndrome is a rare disease, initially reported in 1912 by Maurice Klippel and André Feil from France, characterized by the congenital fusion of any 2 of the 7 cervical vertebrae.

Congenital fusion of two or more cervical vertebrae. Ranges from the fusion of only the bodies (congenital block vertebrae) to fusion of the entire vertebrae (including posterior elements). Results from failure of normal segmentation of cervical somites between 3 and 8 weeks’ gestation. Involved vertebral bodies are often flattened, and associated disc spaces are absent or hypoplastic. Hemivertebrae may also occur. Neural foramina are smaller than normal and oval. Cervical stenosis is rare. The complete absence of the posterior elements with an enlarged foramen magnum and fixed hyperextension posture is called iniencephaly and is rare. The incidence of Klippel-Feil is unknown due to its rarity and the fact that it is frequently asymptomatic.

May occur in conjunction with other congenital cervical spine anomalies such as basilar impression and atlantooccipital fusion.

Classification

Samartzis classification

Type I Having a single congenitally fused cervical segment.

Type II Multiple noncontiguous, congenitally fused segments.

Type III Multiple contiguous, congenitally fused cervical segments

Type I: mass fusion of C-spine to upper T-spine

Type II: fusion of only 1 or 2 interspaces
Clinical features

Classic clinical triad (all 3 are present in < 50%):

1. low posterior hairline
2. shortened neck (brevicollis)
3. limitation of neck motion (may not be evident if < 3 vertebrae are fused, if fusion is limited only to the lower cervical levels, or if hypermobility of nonfused segments compensates). Limitation of movement is more common in rotation than flexion-extension or lateral bending

Other clinical associations include scoliosis in 60%, facial asymmetry, torticollis, webbing of the neck (called pterygium colli when severe), Sprengel’s deformity in 25-35% (raised scapula due to failure of the scapula to properly descend from its region of formation high in the neck to its normal position about the same time as the Klippel-Feil lesion occurs), synkinesis (mirror motions, primarily of hands but occasionally arms also), and less commonly facial nerve palsy, ptosis, cleft or high arched palate. Systemic congenital abnormalities may also occur, including genitourinary (the most frequent being a unilateral absence of a kidney), cardiopulmonary, CNS, and deafness in ≈ 30% (due to defective development of the osseous inner ear).

No neurologic symptoms have ever been directly attributed to the fused vertebrae, however, symptoms may occur from nonfused segments (less common in short-segment fusions) which may be hypermobile, possibly leading to instability or degenerative arthritic changes.

Treatment

Usually directed at detecting and managing the associated systemic anomalies. Patients should have a cardiac evaluation (EKG), CXR, and renal ultrasound. Serial examinations with lateral flexion-extension C-spine X-rays to monitor for instability. Occasionally, a judicious fusion of an unstable non-fused segment may be needed at the risk of further loss of mobility.

In patients with KFS with basilar invagination (BI), compression of the brainstem and upper cervical cord results in neurological deficits, and decompression and occipitocervical reconstruction are required. The highly varied anatomy of KFS makes a posterior occipitocervical fixation strategy challenging. For these patients, the transoral atlantoaxial reduction plate (TARP) operation is an optimal option to perform a direct anterior fixation to achieve stabilization.

Contraindications for participation in contact sports except associated with full ROM without occipitocervical anomalies, instability, disc disease absolute or degenerative changes.
Outcome

In the series Patel et al. the patients who pursued surgical treatment reported significantly more comorbidities (p = 0.02) and neurological symptoms (p = 0.01) than nonsurgically treated participants and were significantly older when pain worsened (p = 0.03), but there was no difference in levels of muscle, joint, or nerve pain (p = 0.32); headache/migraine pain (p = 0.35); total number of cervical fusions (p = 0.77); location of fusions; or age at pain onset (p = 0.16).

More than 90% of participants experienced pain. Participants with an increased number of overall cervical fusions or multilevel, contiguous fusions reported greater levels of muscle, joint, and nerve pain. Participants who pursued surgery had more comorbidities and neurological symptoms, such as balance and gait disturbances, but did not report more significant pain than nonsurgically treated participants ³.

Case series

Seventy-five patients (60 female, 14 male, and 1 unknown) were identified and classified as having the following types of Samartzis fusion: type I, n = 21 (28%); type II, n = 15 (20%); type III, n = 39 (52%). Seventy participants (93.3%) experienced pain associated with their KFS. The median age of patients at pain onset was 16.0 years (IQR 6.75-24.0 years), and the median age when pain worsened was 28.0 years (IQR 15.25-41.5 years). Muscle, joint, and nerve pain was primarily located in the shoulders/upper back (76%), neck (72%), and back of head (50.7%) and was characterized as tightness (73%), dull/aching (67%), and tingling/pins and needles (49%). Type III fusions were significantly associated with greater nerve pain (p = 0.02), headache/migraine pain (p = 0.02), and joint pain (p = 0.03) compared to other types of fusion. Patients with cervical fusions in the middle region (C2-6) tended to report greater muscle, joint, and nerve pain (p = 0.06). Participants rated the effectiveness of oral over-the-counter medications as 3 of 5 (IQR 1-3), oral prescribed medications as 3 of 5 (IQR 2-4), injections as 2 of 5 (IQR 1-4), and surgery as 3 of 5 (IQR 1-4), with 0 indicating the least pain relief and 5 the most pain relief. Participants who pursued surgical treatment reported significantly more comorbidities (p = 0.02) and neurological symptoms (p = 0.01) than nonsurgically treated participants and were significantly older when pain worsened (p = 0.03), but there was no difference in levels of muscle, joint, or nerve pain (p = 0.32); headache/migraine pain (p = 0.35); total number of cervical fusions (p = 0.77); location of fusions; or age at pain onset (p = 0.16).

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References


Operative Neurosurgery - https://operativeneurosurgery.com/