Neck Pain Found to Have Klippel-Feil Syndrome: A Case Report

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PATIENT PRESENTATION

- 19-year-old female with history of hypermobile Ehlers-Danlos Syndrome presenting with severe back and arm discomfort
- On exam, patient had symptoms consistent with right upper extremity radiculopathy

WORKUP

- Imaging revealed congenital fusion of C2-3 and C5-7 consistent with diagnosis of Klippel-Feil syndrome
- Imaging also revealed scoliosis in the cervical and thoracic spine as well as multilevel foraminal stenosis worse at C5-6, C6-7, C7-T1 on the right

MANAGEMENT AND OUTCOMES

- Conservative measures failed to help the patient improve
- Underwent multilevel cervical hemilaminotomies and foraminotomies with neurosurgery
- Admitted to acute inpatient rehabilitation where patient recovered some function in her right upper extremity as well as a decrease in pain
- As an outpatient, she continues to have improved function, however, continues to have myofascial pain and benefits from paraspinal muscle trigger point injections

FIGURES

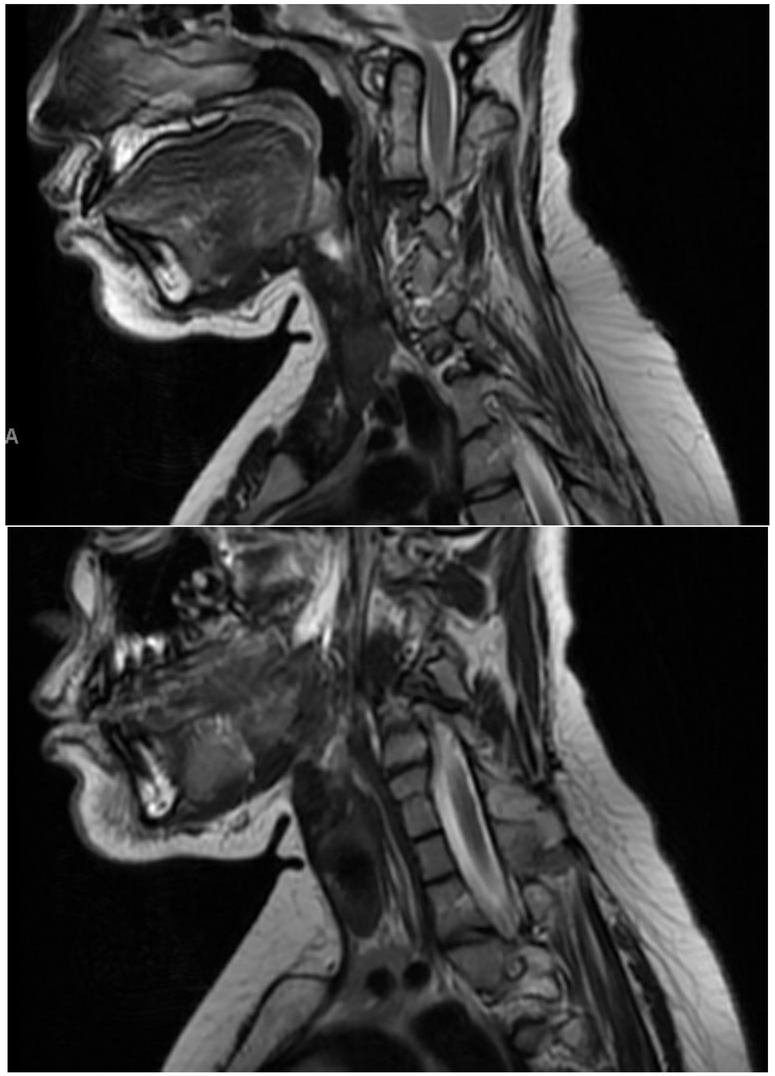

Figure 1: MRI of the cervical spine, T2-weighted imaging showing multilevel vertebral fusion at C2-3 with encroachment on the R C8 nerve root (Top) and additional segmental anomalies between C4 and T1 (bottom).

DISCUSSION

- Klippel-Feil syndrome is defined by an abnormal fusion of the cervical vertebrae[1]
- Occurs in 1 in 40,000 to 42,000 people worldwide[2]
- May have a genetic component and can be inherited in an autosomal dominant pattern[1]
- Patients typically present with a short neck with limited range of motion[1]
- Associated symptoms include headaches and pain, both of which our patient had
- Additional deformities, such as a cleft palate, Sprengel deformity, or internal organs (such as the heart, lungs, or urinary tract), may be present[1]
- Treatment is symptomatic, but may require surgery due to progression of the illness, pain, weakness, or instability, as in the case of our patient[2]

CONCLUSIONS/TAKE HOME POINTS

- Klippel-Feil syndrome is a rare disorder marked by fusion of one or more cervical vertebrae[1,2]
- Treatment is symptomatic but may be operative in certain instances[2]
- Careful monitoring of pediatric patients for having a short neck, short stature, and a low hairline on the neck in the clinical setting can lead to early detection of the syndrome[1,2]

REFERENCES

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2. Georgiev GP, Groudeva V. Klippel-Feil Syndrome with Sprengel Deformity. J Radiol Case Rep. 2019 May 31;13(5):24-29. doi: 10.3941/jrcr.v13i5.3565. PMID: 31558956; PMCID: PMC6742447.