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Case Report

SHORT NECK AND SPLIT BONES

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ABSTRACT
Short stature is one of the important reasons for referral in paediatric endocrine practice. It is essential to evaluate the causes of short stature and to exclude other associated anomalies and treat accordingly. Skeletal anomalies are important cause for short stature.

Keywords: *Scoliosis, Sprengle Deformity*

INTRODUCTION
Etiology of short stature is variable. Skeletal dysplasia causes short stature by defect in cartilage or bone component. Klippel feil syndrome is associated with short stature and is one of the rare causes for short stature.

CASES
A 12 year old boy referred to our endocrine out patient department for evaluation of short stature. He was born of non consanguinous parents had normal neonatal period. His milestones were normal. His academic performance is good. On examination his height is below 3 rd centile. Clinical examination revealed short neck with restricted neck movements. He had scoliosis and pectus carinatum. Neurological examination was normal. Biochemical investigation showed subclinical hypothyroidism and serum IGF-1 was normal. Inview of his skeletal deformity a skeletal survey was done. X ray cervical spine showed fusion of C6-C7 vertebra. Thoraco lumbar spine showed hemi vertebra. X ray Lumbo sacral spine was done in view of short neck, restricted neck movements and spinal deformity.

DISCUSSION
Klippel feil syndrome was described independently by Maurice klippel and Andre Feil in year 1912.It is characterized by fusion of cervical vertebra and hemi vertebra. It is described more common in females. The incident is about 0.2 to 0.7 per 10,000 population. The clinical features include Short Neck, Restricted Neck movements and low hair line.
There are three types of KFS described in literature
*KFS 1- Autosomal dominant caused by mutation in the GDF 6 gene on chromosome 8q+ 22 (Clarke *et al.*, 1995)
* KFS 2- AR caused by mutantion in MEO X 1 gene on ch 17q 21 (Fusion at C5- C6 value)
* KFS 3 - AD – GDF 3 g on chromosome 12p13
Feil classified the syndrome into 3 categories (Samartzis *et al.*, 2006)
Type I – Fusion of all cervical vertebrae.
Type II – Fusion of 1 or 2 vertebra.
Type III - Presence of thoracic and lumbar spine anomaly in association with type I or II.
In patients diagnosed with KFS, most commonly 50 % present with short neck, flexion deformity of neck and low hair line. Some patients may not have any symptoms and it may be detected incidentally. Presence of neurological symptoms is rarely seen in 20% of patients. In one of the largest case series of KFS, Heneinger *et al.*, described 50 patients (Samartzis, 2006). In that series 60% had scoliosis, 16 patients had renal anomaly which includes horse shoe kidney, agenesis and hydronephrosis. In the same series 7 patients had septal anomaly of heart.
Other anomalies described in KFS include sprengle deformity, cleft palate, hearing deficit, synkinesia, arterial anomalies.

Case Report

To the best of our knowledge, the L4 – L5 vertebrae fusion is very rare and has been reported in only one series.

This case report illustrates the importance of meticulous clinical examination in patients presenting with short stature to diagnose rarer causes such as klippel feil syndrome.

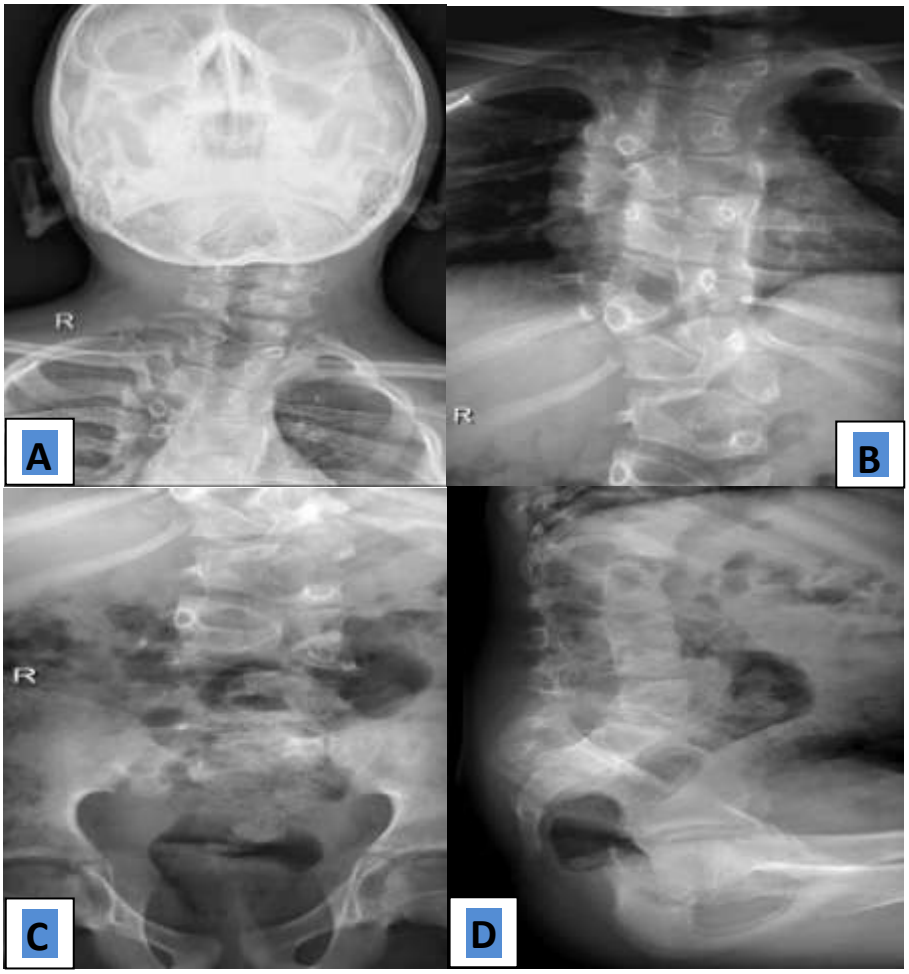

Figure A: Cervical vertebra fusion C5 – C6 with hemivertebra
Figure B: Hemivertebra in thoraco lumbar spine
Figure C: Hemivertebra in thoraco lumbar spine
Figure D: lumbar vertebra fusion – L4-L5

REFERENCES

Clarke RA, Singh S, McKenzie H, Kearsley JH and Yip MY (1995). Familial Klippel-Feil syndrome and paracentric inversion inv(8)(q22.2q23.3)**.** *The American Journal of Human Genetics* **57** 1364-1370. [PubMed: 8533765]

Samartzis D, Lubicky JP and Herman J (2006). Symptomatic cervical disc herniation in a pediatric Klippel-Feil patient: the risk of neural injury associated with extensive congenitally fused vertebrae and a hypermobile segment. *Spine* **31**(11) E335-8.

Samartzis DD, Herman J, Lubicky JP and *et al.*, (2006). Classification of congenitally fused cervical patterns in Klippel-Feil patients: epidemiology and role in the development of cervical spine-related symptoms. *Spine* **31**(21) E798-804.