SHORT NECK AND SPLIT BONES

*Suresh P Rajasekaran, Senthil k Rajasekaran and Vijayalakshmi Balachandran
Department of Endocrinology, PSG hospitals, Peelamedu, Coimbatore-0422-2570170 -5354
*Author for Correspondence

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.
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.

**REFERENCES**

