CASE REPORTS

KLIPPEL-FEIL SYNDROME*

S. K. DIKSHT, S. P. AGARWAL, R. C. GUPTA AND R. S. SINGH

Varanasi

Klippel and Feil in 1912 described a complete clinical picture of the syndrome in a 46-year-old tailor having an unusually short neck with gross restriction of movements. The entire spinal column consisted of only twelve clearly distinguishable vertebrae but its uppermost part was an osseous mass evidently formed by the fusion of several vertebral elements. A distinct atlas and axis could not be found. The first 4 pairs of ribs were partially fused at their origin. Since then a number of cases have been reported from other countries. There have been only a few reports of this syndrome from India (Dayal 1959, Basu 1961, Khatua 1965 and Jain et al. 1965). We are reporting two cases of this syndrome; one presented with quadriparesis and another with restrictive movements of the neck and right shoulder joint along with congenital heart disease.

Report of Cases

Case I (Hospital No. 2000/67)

G., a 12-year-old Hindu male child was admitted on 4-3-67 to the Children’s Hospital, College of Medical Sciences, Banaras Hindu University, with complaints of difficulty in walking and weakness in both upper limbs for the previous 20 days. As stated by the child, he was alright 20 days previously when he found some difficulty in handling a glass of water with one hand. For this he used to provide support with the other hand. For the same duration he had also developed difficulty in walking.

Prenatal, natal and postnatal histories were normal. The family consisted of 5 members including the father, mother, one brother and one sister besides the patient. All were normal.

The patient was of a thin built, with a short neck. He weighed 26.5 Kg. All other systems were normal excepting the nervous system. The higher cerebral functions, cranial nerves, pupils and fundi were normal. There was no neck rigidity. The power was reduced in all limbs, with increase in tone, normal coordination, no abnormal movements, exaggerated reflexes, normal superficial reflexes and extensor plantars bilaterally. There was some wasting in the upper limbs (Fig. 1). Sensation was normal.

Investigations. The routine tests on the blood were normal. The C.S.F. was traumatic. V.D.R.L. was negative.
X-rays of the cervical spine revealed fusion of the first and second, and of 5th and 6th cervical vertebrae with their spinal processes. Spina bifida and hemivertebrae between the 5th and the 7th cervical vertebrae and bilateral cervical ribs were present. There was also a forward spondylolisthesis of the 1st degree, of C7 over D1.

Case 2 (Hospital No. 2732/67)

Baby, a 1½-month-old female child, was admitted on 28-3-67 for complaints of diarrhoea since birth, inability to move the right arm at the shoulder joint, asymmetry of the face on crying, keeping the neck turned to the left side and inability to move it towards the right side since birth.

Prenatal and natal histories were essentially normal. The baby was the product of a full-term uneventful normal delivery conducted at home. There was no history of malformation on either the maternal or the paternal side. The patient had 3 elder brothers alive and healthy. One brother had died during delivery due to head arrest after breach presentation.

An apparently well-looking, thin child, was seen lying with her neck rotated towards the left side. Weight was 3.75 Kg., height 20" and head circumference 14.2", temperature 99°F, pulse 120 minute, respiration 30 per minute.

The neck was short with a low hairline. A band of skin and connective tissue running from the right shoulder to the right mastoid region was present. The right scapula was raised compared to the left.

Active movements were not possible at the right shoulder joint although the right arm could be abducted passively up to 60°. A faint right posterior axillary fold was present. The latissimus dorsi muscle was inserted more anteriorly than normal. The trapezius was short and taut.

The cardiovascular system examination revealed a grade III pan-systolic murmur in the fourth left intercostal space just lateral to the sternum with a fan-shaped radiation. On neurological examination, evidence of right infranuclear facial palsy was present. The other systems were normal.

Investigations. The total W. B. C. count was 11,000/cu.mm., polys were 28%, lymphos 68%, eosinophils 4%, hemoglobin 13.5 G.%. X-ray of the chest and E.C.G. was normal. X-ray of the cervical region, A. P. and lateral views showed the Klippel-Feil as well as Sprengel's deformity.

Discussion

The Klippel-Feil syndrome is a rare congenital abnormality. Its rarity may be due to its being symptomless in many instances. Slight abnormalities may go unrecognized and may be detected only by a chance X-ray of the cervical spine. Both the sexes are equally affected.

The syndrome results from a widespread failure of segmentation of the mesodermal somites which normally should occur from the 3rd to the 8th weeks of intrauterine life. Although Canetti found a familial incidence in some cases, its hereditary and familial nature has not yet been definitely established. Jarcho and Levin (1938) described the syndrome in an American Negro brother and sister, whose mother had a defective 5th cervical lamina. Jarcho (1965) described a family in which the Klippel-Feil anomaly had occurred in more than one person.