Letters to the Editor

Uncommon Manifestations of Klippel Feil Syndrome

Klippel Feil syndrome is characterized by congenital fusion of cervical vertebrae. We report a rare association of Klippel Feil syndrome with hypoplasia of depressor angularis oris muscle, triphalangeal thumb and distal limb abnormalities.

A male baby was delivered at term to 20 year old primigravida mother. The baby weighed 2.8 kg and cried immediately after birth. Examination revealed short neck, limited lateral flexion of neck along with low posterior hairline. Cleft palate and facial asymmetry in the form of hypoplasia / absence of depressor angularis muscle of left side of face were also present (*Fig. 1*). Other musculoskeletal abnormalities were absence of radius and first metacarpal bone, elongated-curved ulna and triphalangeal thumb on right side. Radiological examination revealed hemivertebra and fusion of lower cervical vertebrae. Echocardiography revealed peri-membranous ventricular septal defect.

The importance of recognizing Klippel Feil syndrome lies in the fact that there is strong association of this anomaly with other significant conditions like scoliosis (60%), genitourinary abnormalities (65%), Sprengel's deformity of the scapula (35%), cardiac abnormalities–most commonly VSD (15.35%), deafness (36%)(1).

Our case presented with short neck and restriction of neck movement. Associated features were cleft palate, hemivertebra and fusion of lower cervical vertebra with absence of radius, first metacarpal bone and right triphalangeal thumb along with peri-membranous VSD. Although musculoskeletal abnormalities are known to occur in this syndrome, there is no earlier documentation of associated hypoplasia of depressor angularis oris. Bimanual polydactyly of triphalangeal thumb and thenar hypoplasia(2) are also reported in this syndrome, but there is no earlier documentation of combined association of Klippel Feil syndrome with hypoplasia of depressor anguli oris muscle, triphalangeal thumb and distal limb abnormalities.

In a series of 50 subjects with unilateral hypoplasia of depressor angularis oris

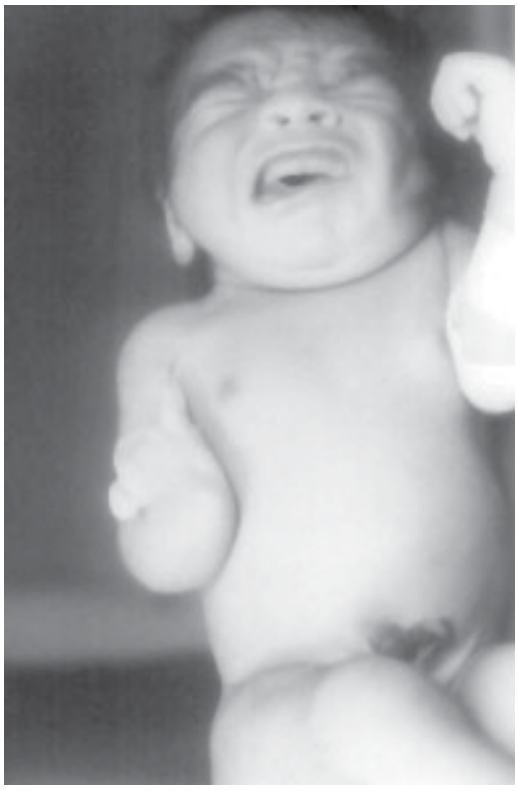

Fig. 1. Klippel Feil syndrome with hypoplasia of depressor angularis oris and distal limb abnormalities.

associated findings were anomalies of the head and neck (48%), heart (44%), skeleton (22%), genitourinary tract (24%), central nervous system (10%), gastrointestinal tract (6%), and miscellaneous minor anomalies (8%)(3). Additional findings that have been reported in association with congenital hypoplasia of depressor angularis oris are 4p deletion, Klinefelter syndrome, isolated CD4 deficiency and Treacher-Collins like facial appearance(4).

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Torsion of Vermiform Appendix

A 9-year-old boy presented to us with a 16-hour history of abdominal pain localized to right iliac fossa and repeated bouts of non-bilious vomiting. There was no previous history of abdominal pain. On examination, the patient was febrile (100º), had tachycardia (112/min) and tenderness and guarding in right iliac fossa. There was no rigidity; psoas and obturator tests and Rovsing's sign were negative. Rectal examination was inconclusive. A clinical diagnosis of acute appendicitis was made. Other than mild leucocytosis, pre-operative blood investiga-tions were essentially normal. At operation, 8 cm long retrocecal appendix was revealed that had torted 270º clockwise just distal to its base. The appendix was only minimally inflamed. There was no associated fecolith, adhesions, lipomas or mucocoele. A routine appendicectomy was performed. The post-operative period was uneventful. Histo-pathological evaluation revealed non-specific inflammation of the appendix.

Torsion of vermiform appendix is an extremely rare condition with only about 25 cases reported in world literature since its first description in 1918(1). The condition is pre-operatively indistinguishable from acute appendicitis and the diagnosis is usually made intra-operatively(2). The features that are commonly associated with torsion of appendix include long appendix and pelvic position of the appendix(1). The direction of rotation although variable, was more frequently counterclockwise(1). The site of the torsion is