Images in Clinical Practice

Cleidocranial Dysplasia

A full term male baby weighing 2.4 Kg was delivered by normal vaginal delivery. On examination the shoulders could be approximated in the midline (*Fig. 1*). The fontanelle were large and sutural diastesis was present. Radiograph of the chest showed partial absence of the clavicles, characteristic of Cleidocranial dysplasia (CCD)(*Fig. 2*).

CCD is an autosomal-dominant bone disorder, caused by heterozygous mutations in runt related gene (RUNX2/polyomavirus enhancer binding protein 2A(PEBP2A)/core binding factor A1). The pathogenesis of CCD may be related to inability of the truncated RUNX2 protein to interact with the signal transducers of transforming growth factor B (Smad) and induce osteoblast like phenotype in myeloblasts. The disease is characterized by generalized skeletal dysplasia especially abnormal clavicles, patent sutures and fontanelles, supernumerary teeth, short stature, and a variety of other skeletal changes. Typically the clavicles can be approximated in the midline. Complete absence of the clavicles is rare and usually only the acromial end is absent. The skull is brachycephalic with bossing of frontal, parietal and occipital bones, late closure of the

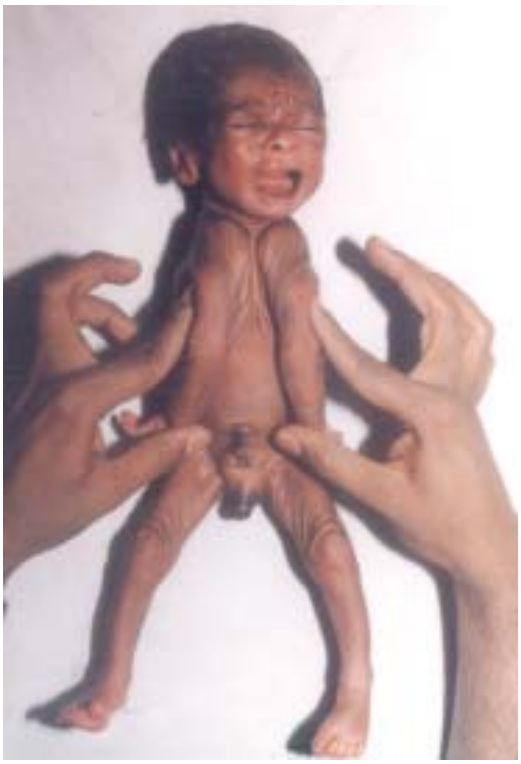

Fig. 1. Shoulders approximate in midline in a case of cleidocranial dysplasia.

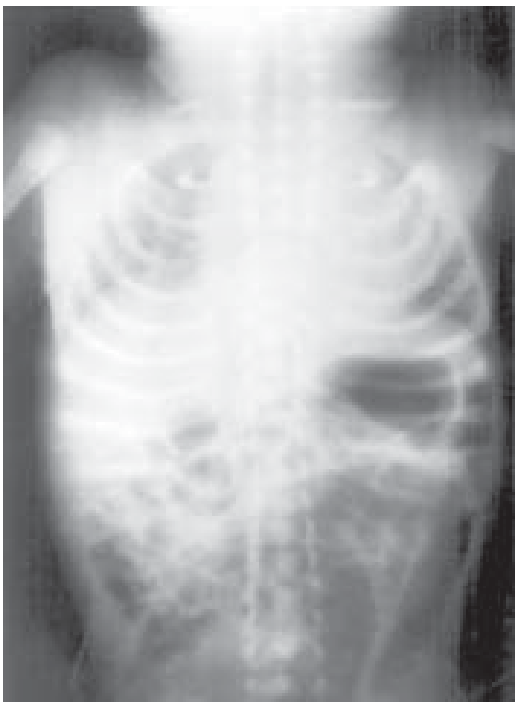

Fig. 2. Radiograph of chest showing partial absence of clavicles.

fontanelle and mineralization of sutures. Wormian bones are present. The mandible is prognathic as a result of hypoplasia of the maxilla and other facial bones. Skeletal defects in the hands and pelvic bones have also been described. The permanent teeth erupt late and are often abnormal with numerous supernumemary teeth. The patients may also have conductive deafness. The course is usually uncomplicated except for dislocations, especially of the shoulders, and dental anomalies that require therapy.

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Necrotizing Fascitis

A 6-year-old, male child was admitted with history of minor penetrating injury with nail on anteromedial aspect of left thigh 7 days prior to admission. Three days later, he developed fever, discoloration of skin and swelling of left thigh. On examination child was toxic, drowsy, febrile with poor respiratory effort and shock. Local examination of left thigh revealed 8×10 cm necrotic area with blue-black discoloration with marked swelling of thigh and leg with no palpable crepitus (*Fig. 1*). Investigation revealed multi-organ dysfunction. A diagnosis of necrotizing fascitis was made. The child was provided intensive treatment with inotropes, antibiotics and mechanical ventilation and extensive surgical debridment. Child deteriorated over next 5 days and expired.

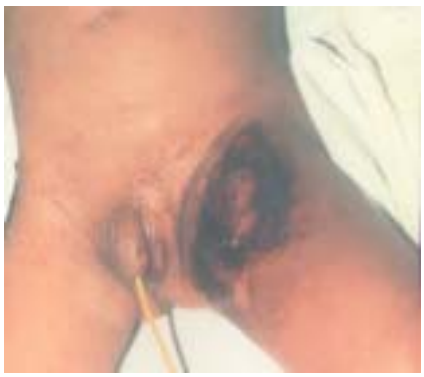

Fig. 1. Necrotizing fascitis of left thigh.

Necrotizing fascitis is characterized by involvement of soft tissue of deep dermis, adipose tissue and subcutaneous fascia. Hallmark of the disease is extensive areas of both necrosis and cellulitis. Portal of entry of organisms is usually through a cut or surgical wound. Common organisms implicated are b-hemolytic group A streptococci, Pseudomonas and anerobes. The disease has a varied spectrum ranging from simple cellulitis to septic shock and multiorgan dysfunction. Over 50% mortality has been reported. Management of necrotizing fascitis includes early surgical debridment. Antibiotics alone do not improve outcome. Amputation can be done in difficult cases. Role of hyperbaric oxygen is doubtful.

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