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Pycnodysostosis

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Pycnodysostosis is characterized by short stature, dysplasia of skull and outer end of clavicles, obtuse mandibular angle, partial or total aplasia of distal phalanges and generalized increased density of the skeleton(1). Seventy seven cases have been described in the world literature(2); from India 11 cases have been reported(3).

Case Report

An 11-year-old boy born of a non-consanguinous marriage was evaluated for short stature and inability to hear and speak from early life. On examination he was markedly stunted with parietal and frontal bossing, open anterior and posterior fontanelle and all sutures were palpable. He had dysmorphic facies with prominent eyes, malar and mandibular hypoplasia, high arched palate, macrognathia and malocclusion of teeth (*Fig. 1*). The eyes showed blue sclerae and the neck appeared short; the chest was pigeon shaped and showed milk lumbar lordosis. The upper and lower limbs were short, with short fingers, broad thumbs and hypoplastic and spoon shaped nails; skin over the fingers had excessive folds. The motor and sensory systems were normal.

X-ray of long bones showed uniform increase in density with thickened cortex and narrow medullary canals. Terminal phalanges of feet and hands showed acrosteolysis (*Fig. 2*). *X*-ray of chest showed increased density of ribs and hypoplastic clavicles. Radiograph of the spine showed increased density of vertebrae with normal disc spaces and the skull showed increase in the bone density with widely open fontanelle. The facial bones were small and hypoplastic and the mandibular angle was obtuse. His five siblings and parents had normal stature and no dysmorphic features.

Discussion

Pycnodysostosis is inherited as an autosomal recessive trait. Affected patients

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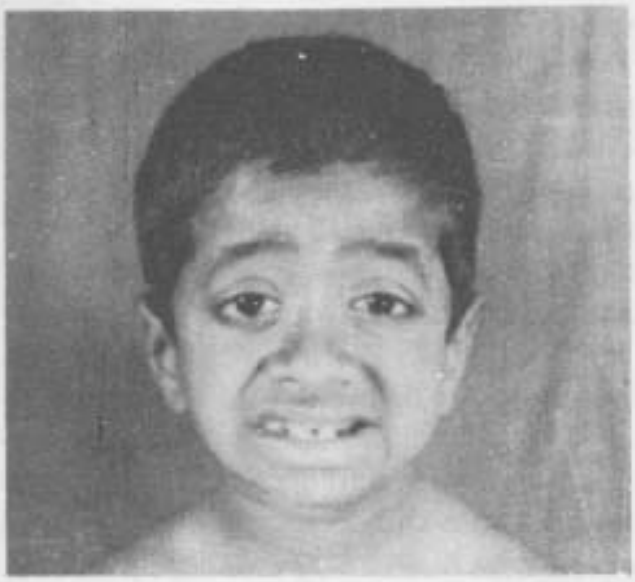

Fig. 1. Showing prominent eyes, malar and mandibular hypoplasia, macrognathia and malocclusion of teeth.

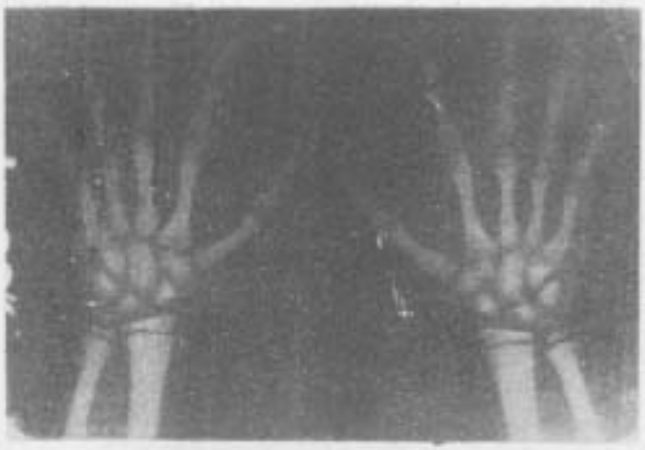

Fig. 2. Distal phalanges of hands are hypoplasuc and acrosteolysis is seen.

are stunted and have peculiar facies, consisting of parietal and frontal prominences, hypoplastic mandible, short fingers and toes. Mental retardation is reported in few cases. The fontanelle may remain open for years(4). Long bone deformities due to fractures may be present(5). They can also have hepatosplenomegaly, anemia and rickets with radiological signs, of pycnodysostosis(6,7). Repeated respiratory infections and right heart failure due to chronic upper airway obstruction, as a result of the micrognathic defect had been reported(8).

Radiological examination shows increased density of bone on the entire skeleton. The bones are dense with thin medullary cavity. Disappearance of the mandibular angle, aplasia of the terminal phalanges, clavicular hypoplasia and thin tubular long bones are characteristic findings(6). Break in the pars-inter-articularis of vertebrae has been reported(3).

In the differential diagnosis, osteopetrosis should be considered. In infancy, classical osteopetrosis is diagnosed by increased density of the long bones, progressive anemia; hepatosplenomegaly and neurological defects are constant findings in osteopetrosis. Clavicular and distal phalangeal dysplasia is absent(6).

Pycnodysostosis is compatible with normal life span. Pathological fractures and dental problems will need appropriate attention. There are many clinical, radiological and biochemical abnormalities, which are similar in pycnodysostosis and osteopetrosis. Pycnodysostosis has been considered to be a variant of osteopetrosis(6).

The present case had the characteristic clinical and radiological features of pycnodysostosis. Presence of deafness, was, however, an unusual manifestation. Occurrence of deafness is rarely reported in pycnodysostosis, history of deafness was also present(9).

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Sprengel's Deformity with Absent Ribs

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Sprengel's deformity is an uncommon congenital anomaly(1); but is one of the common anomalies of the shoulder(2,3). It consists of elevation and upward rotation of the scapula(4). It is more common in girls and on left side(1).

Sprengel's deformity is frequently associated with abnormalities of the cervical spine such as Klippel-Feil syndrome and congenital scoliosis, as well as cervical ribs and rib fusions(5). Other associated anomalies include cervical spina bifida, congenital kyphosis, syringomyelia, platybasia, situs inversus, mandibulofascial dysostosis, ipsilateral shortened humerus and clavicular anomalies and rarely renal anomalies(1,6,7). Hypoplasia and anomalies of the upper thoracic ribs are reported in Sprengel's deformity(4). There are no reports in literature describing absent ribs in patients with Sprengel's deformity. We report a case of Sprengel's deformity with absent ribs.

Case Report

The patient was a 2-month-old, Sikh, male child, product of a nonconsanguinous marriage after an uneventful 38 weeks gestation born to a primigravida mother. The parents have history of abnormal movements of the chest during respiration. There was no other associated complaint.

The examination revealed higher right shoulder and scapula than on the opposite side. On looking at from the rear, the inferior angle was displaced towards the spine. On attempted abduction of the arm, the lower angle of the right scapula did not rotate outwards as it should in normal. Kyphosis and scoliosis were present. On palpation right upper thoracic ribs were not felt. There was indrawing of the right upper chest wall during inspiration.

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