

Pyknodysostosis

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Pyknodysostosis is an autosomal recessive disorder of skeletal system characterized by short stature, generalized hyperostosis, hypoplastic or aplastic distal phalanges, skull dysplasia and an obtuse mandibular angle(1). Till 1988, 78 cases have been described in the world literature, out of which only 5 cases are reported from India(2). We report two siblings with pyknodysostosis, born to unrelated parents and who presented with bronchial asthma.

Care Reports

Case 1: A 10-year-old girl was referred for recurrent respiratory infections and not growing well since birth. She was the product of a non-consanguineous marriage, delivered at term through lower segment

cesarean section. She was fully vaccinated and attended school with normal scholastic performance. On examination her height was 112 cm (below 3rd percentile for her age), weight 18 kg, head circumference 55 cm, upper-lower segment ratio 1.2:1, and arm span 106 cm. The head was dolicocephalic with frontal and parietal bossing, sutures were palpable and a depression felt at the site of anterior fontanelle. The face was small and narrow with bilateral proptosis. The oral cavity showed double row of teeth, high arched palate, tonsils were normal in size but congested. The chest was barrel shaped with prominent sternum and the spine showed lumbar lordosis. The hands and feet and short terminal phalanges and koilonychia. The joint mobility was normal. Auscultation of chest revealed bilateral rhonchi.

Case 2: The brother of case 1 who was 6-years-old, and presented with recurrent respiratory tract infections and short stature. The height was 90 cm (below 3rd percentile), weight 13 kg and head circumference 50 cm, upper-lower segment ratio 1.3:1 and arm span 91.5. The facial and other features were similar to that of his sister.

Skeletal surveys showed similar radiological features in both cases. There was a generalized increase in bone density especially in the calvarium, long bones and phalanges with preservation of medullary cavity. The X-ray of skull showed sclerotic bones, widening of sutures, agenesis of frontal sinuses, widening of sella turcica with straightening of angle of mandible (*Fig. 1*). X-ray of the hands showed sclerotic bones, absent ungual tufts and terminal osteoacrololysis (*Fig. 2*). X-ray of both tibia and fibula showed dense osteosclerotic changes. The roentgenogram of the chest

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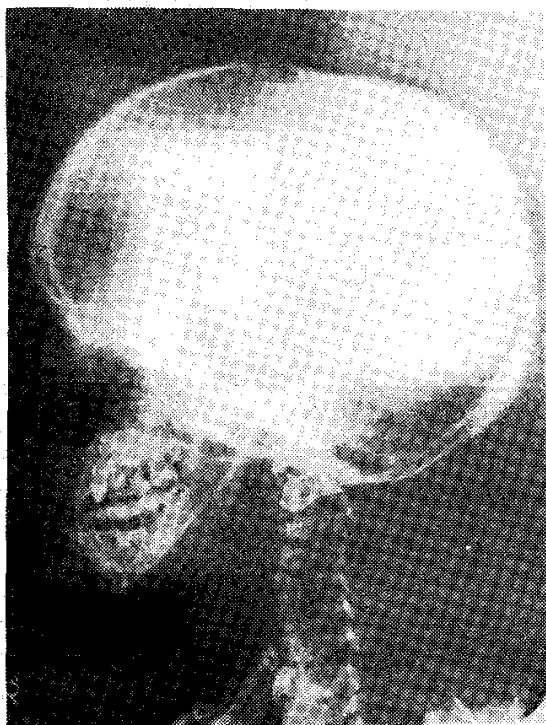


Fig. 1. X-ray skull showing sclerotic bones, widening of sutures, J shaped sellaturcica and straightening of angle of mandible.

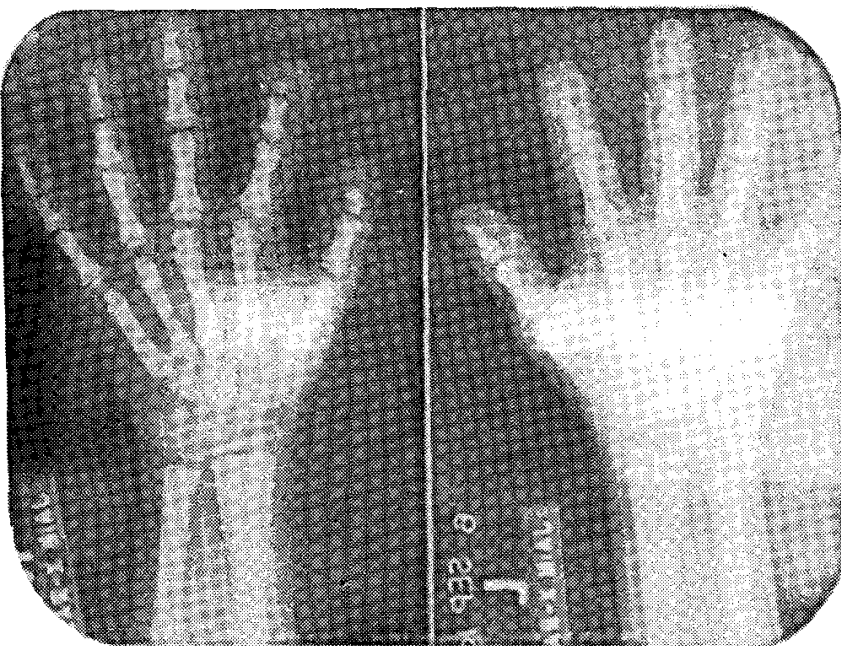


Fig. 2. X-ray of the hands showing sclerotic bones and terminal osteoacrolisis specially in IV phalange.

revealed osteosclerotic ribs, normal clavicles, hyperinflated lungs and normal cardiac outline.

On the basis of clinical and radiological features a diagnosis of pyknodysostosis was made. Both children were treated with antibiotics and bronchodilators.

Discussion

Pyknodysostosis commonly presents with history of repeated fractures sustained after trivial trauma(3). The cases reported from India did not present with history of fractures or recurrent respiratory tract infections(4). The condition should be differentiated from osteopetrosis, cleidocranial dysostosis and progressive diaphyseal dysplasia. Osteopetrosis can be recognized by the associated anemia, hepatosplenomegaly and absence of skull, mandibular and phalangeal involvement(1). Cleidocranial dysostosis while involving the cranium and clavicles, characteristically spares the mandible and phalanges; bone density is normal and height remains unaffected(5). Progressive diaphyseal dysplasia is predominantly a disorder of long bones—the density, width and the length being increased; skull and phalanges are, however, unremarkable(6). The tendency of spontaneous fractures in pyknodysostosis reduces with age and the family needs to be reassured regarding the nature of the disease(7).

The recurrent episodes of cough, rhonchi, hyperinflated lung fields and response to bronchodilators suggest that these children had bronchial asthma. In the available literature relationship of bronchial asthma and pyknodysostosis has not been mentioned. It is not known whether this condition is a predisposing factor for asthma or is a coincidence.

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Pyknodysostosis

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Pyknodysostosis 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-consanguineous 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

Pyknodysostosis is inherited as an autosomal recessive trait. Affected patients

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