

Pycnodysostosis

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Section: Paediatric radiology

Imaging Technique: MR

Case Type: Clinical Cases

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Patient: 13 years, female

Clinical History:

A thirteen-year-old girl presented in clinic with short stature failure to thrive and bowing of both legs. X-rays of the skull revealed widely open sutures and fontanelle, small facial bones with absent sinuses and an obtuse mandibular angle. The bones showed a uniformly increased density, and the medullary canal though distorted was still visible in all long bones.

Imaging Findings:

A thirteen-year-old girl presented in clinic with short stature failure to thrive and bowing of both legs. There was no history of any injury in the recent past and no significant family history. A full skeletal survey was done. X-rays of the skull revealed widely open sutures and fontanelle, small facial bones with absent sinuses and an obtuse mandibular angle. The outer third of clavicle was absent bilaterally. There was loss of terminal tufts of the phalanges of the hand, and in some fingers the entire terminal phalanx was absent. Both tibias were bowed with old fracture lines still visible. The bones showed a uniformly increased density, and the medullary canal though distorted was still visible in all long bones.

Discussion:

Pycnodysostosis is considered to be an autosomal recessive disease with an incidence of under one per million. The usual presentation is dwarfism and recurrent fractures following trivial trauma. The patient may suffer a fracture at any age. They are more common in the lower limb and can result in deformities though they heal in the usual time period. The fracture lines may remain visible for a long time after it has healed. Mental retardation has been seen in a few patients though no definite association has been identified. They are usually short-limbed dwarfs, with a relatively large head, open anterior fontanellae and frontal and parietal bossing. There is persistence of deciduous teeth and can give the appearance of a double row of teeth. Dental carries is a frequent associated finding. Eyes may be normal or more often have some degree of proptosis. Clavicles are present with partial aplasia of the acromial ends, though in some cases it may be completely absent. Varying degree of khyphosis, scoliosis or a combination of the two may be present. The hands and feet have short terminal phalanges and abnormalities of the distal radio ulnar joint have been reported. Lab investigations when done, have been normal. Criteria for diagnosis are increased bone density of the entire skeleton without complete obliteration of the medullary canal and characteristic changes in the skull hand and long bones.

Differential Diagnosis List: Pycnodysostosis

Final Diagnosis: Pycnodysostosis

References:

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Figure 1

a



Description: There is increased density of all bones through out. Though the medullary canal is distorted it can still be appreciated quite readily in the radius **Origin:**

b



Description: The tufts of the terminal phalanges are absent in the thumbs and most of the terminal phalanges themselves are missing in the other fingers. There is also increased bowing of the forearm bones. **Origin:**

Figure 2

a



Description: AP view reveals old healed transverse fracture of both tibiae. Transverse fractures are quite characteristic in patients. **Origin:**

b



Description: Lateral view shows bowing of both tibiae with old fracture lines still visible. The fracture lines can persist for several years as umbauzone line. **Origin:**

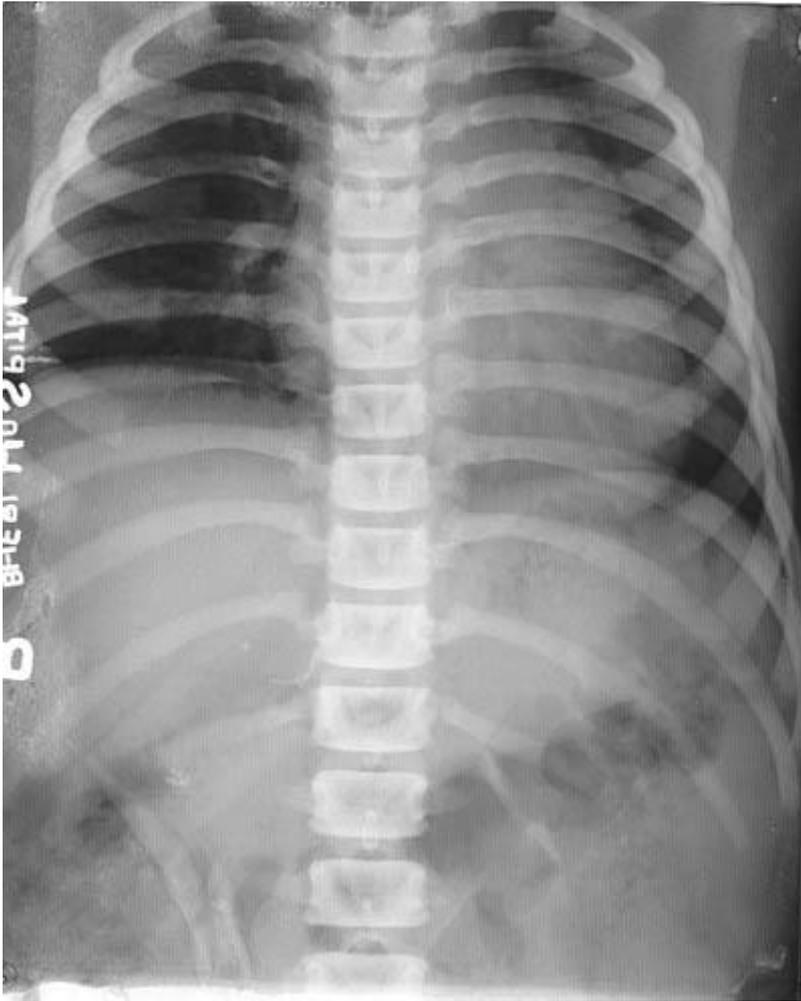
Figure 3

a



Description: Notching of the anterior vertebral bodies can be clearly visualised. There is also some loss of lumbar lordosis **Origin:**

b



Description: Origin:

Figure 4

a



Description: The acromial ends of both clavicles are absent. **Origin:**

Figure 5

a



Description: The suture lines are wide open; the facial bones are small, hypoplastic and non-pneumatized. A double row of teeth are visible. **Origin:**

b



Description: The lateral view shows the wide open fontanelles and sutures. **Origin:**

Figure 6

a



Description: The x-ray very clearly demonstrates the obtuse nature of the mandibular angle, which is almost 180 degrees. **Origin:**