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Pycnodysostosis*

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Pycnodysostosis is the name coined by Maroteaux and Lamy[1,2,3] in 1962 for a condition which they recognized to be a new and distinct entity. The main clinical features of this entity are shortness of stature and a tendency for multiple fractures. However, the landmark of the syndrome is its radiological appearance which consists of a combination of osteopetrosis and some findings of cleidocranial dysostosis. All the bones of the skeleton reveal a striking sclerosis and it is from this dense appearance of the osseous system that the condition derives its name ('pyknos' in Greek meaning 'dense'). The additional presence of specific skeletal anomalies and its distinct clinical picture justify the consideration of it as a clinical entity, separate from osteopetrosis.

There are reports of 33 cases in the literature fulfilling the criteria for the diagnosis of pycnodysostosis.[4,5,6,7,8,9,10] The following is the report of another case, the first report from Turkey to the best of our knowledge, and the discussion of the various aspects of this interesting disease.

Case Report

(A. A. Hosp. No. 328878) This 9 year old girl was brought to the Hacettepe Children's Hospital Out-patient Clinic because of shortness of stature. Being the first offspring of an unrelated and healthy couple she had a normal development up until two years of age. Thereafter, her parents noted her to be stunted in her growth. She has a 7 year-old brother who is in good health and has developed normally.

Physical examination revealed her to be an alert child with normal mental development. She weighed 14.5 kgs. and had a height of 97 cms. The forehead was prominent with an exophtalmic appearance of the

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eyes. The latter was the result of a receding upper jaw and hypoplastic zygomatic bones (Figures 1 and 2). There were multiple caries in the irregular and malformed teeth. Palpation of the cranial vault revealed the sutures to be open. There was an upper dorsal kyphotic deformity of the spine. The fingers and toes were short, the wide nails appearing abnormally hard and spoon shaped. (Figure 3)

Laboratory tests, including serum determinations for calcium, phosphorus and alkaline phosphatase were within normal limits. A bone marrow aspiration demonsrated the presence of hypocellular and otherwise unremarkable marrow elements.

Roentgenological examination of the skull showed the mandible to be extremely hypoplastic with loss of the mandibular angle (Figure 4). The maxillae were equally underdeveloped with no identifiable sinus cavity. The sutures and fontanels were open. The vertebrae and the long bones of the extremities were very sclerotic with a failure of remodeling of the distal femora resulting in a flask shaped enlargement in the same (Figure 5). In addition there were multiple congenital ano-

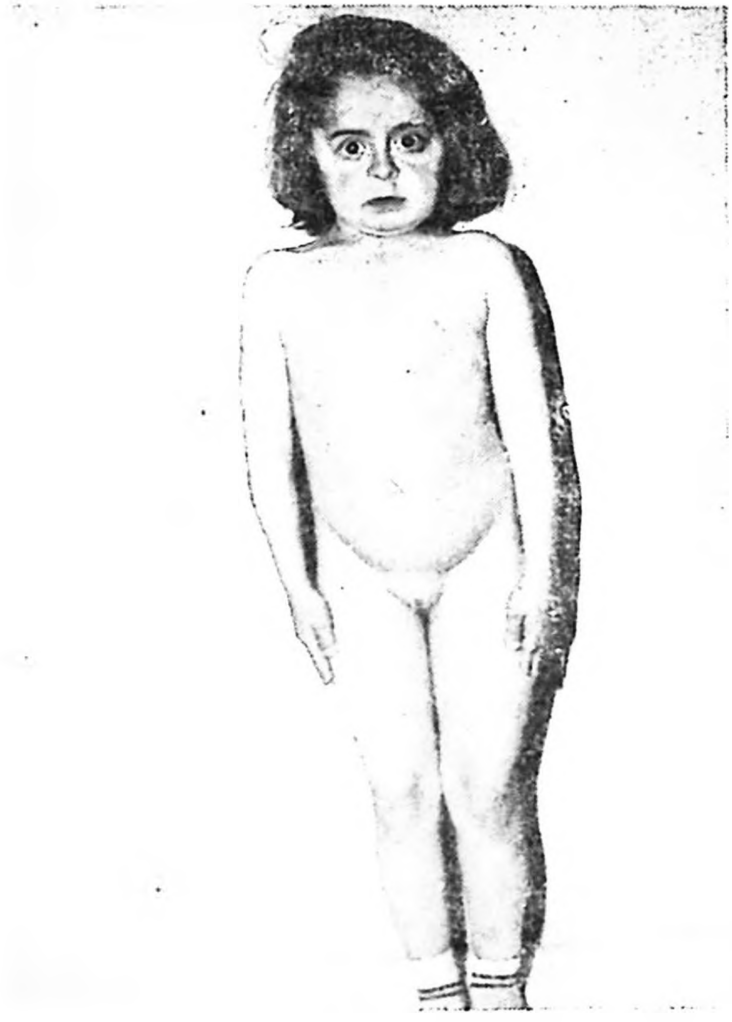

Figure 1

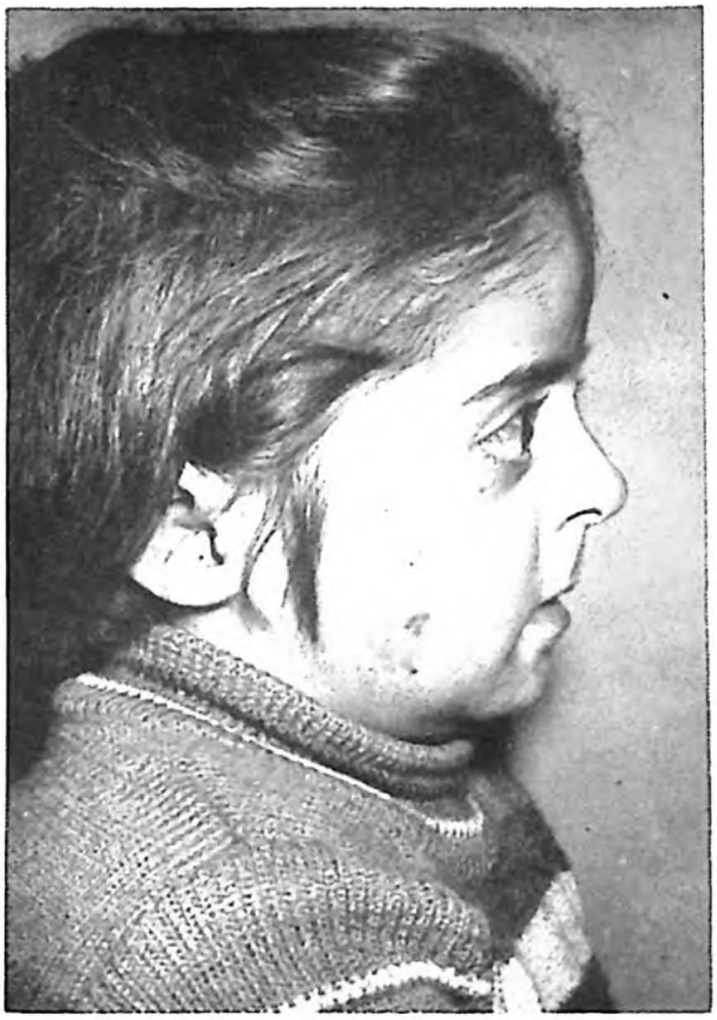

Figure 2

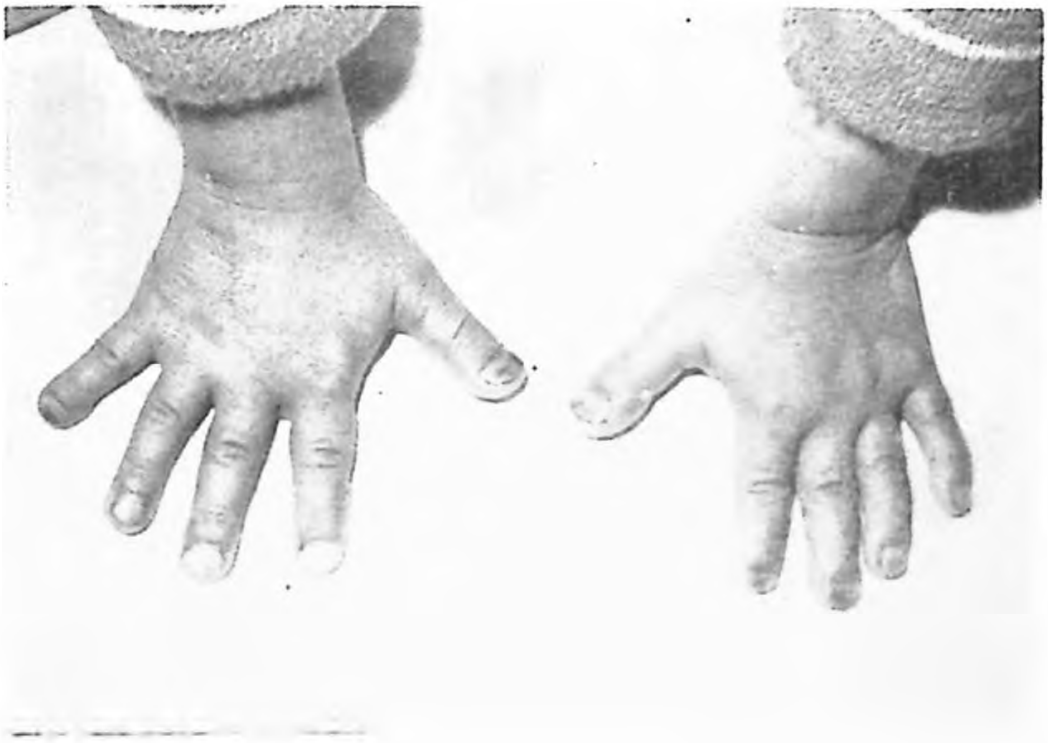

Figure 3

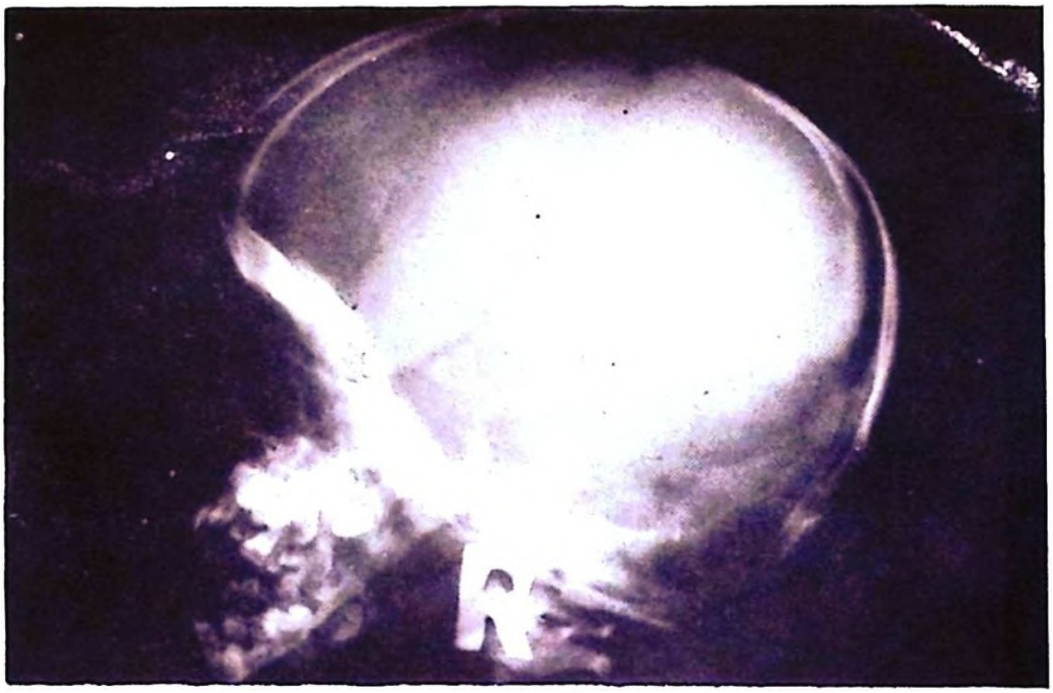

Figure 4

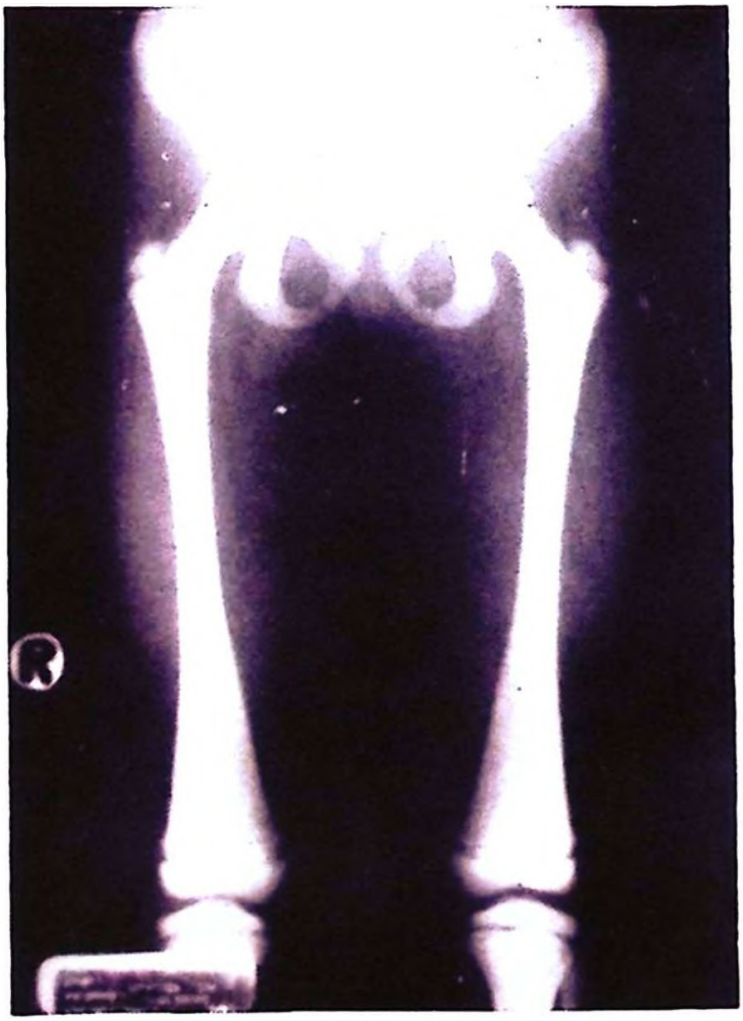

Figure 5

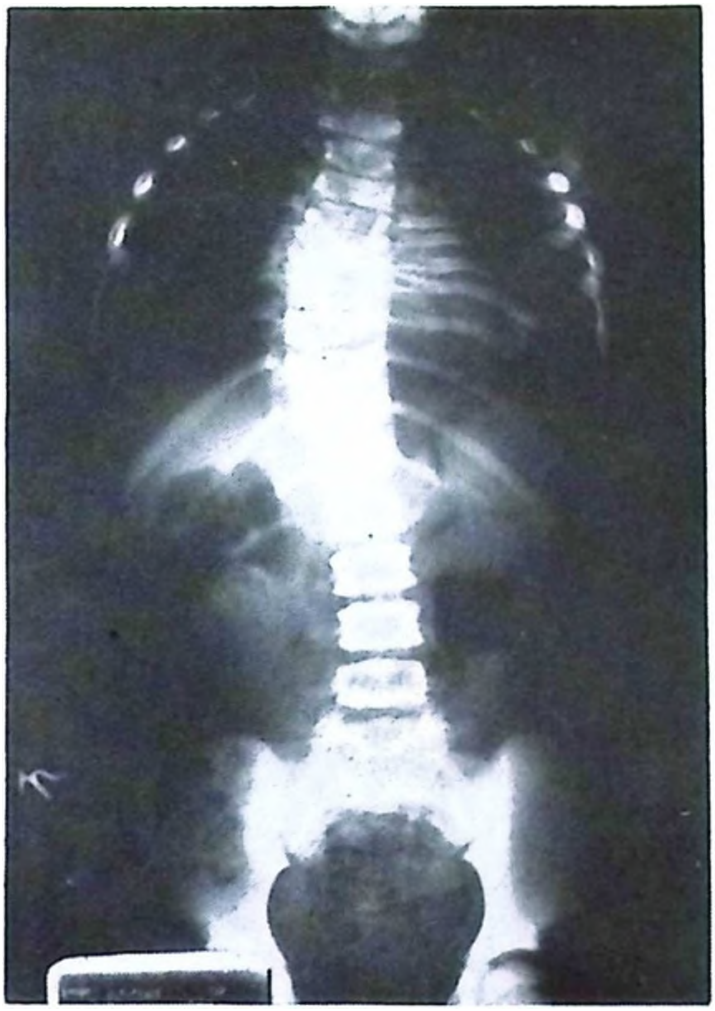

Figure 6

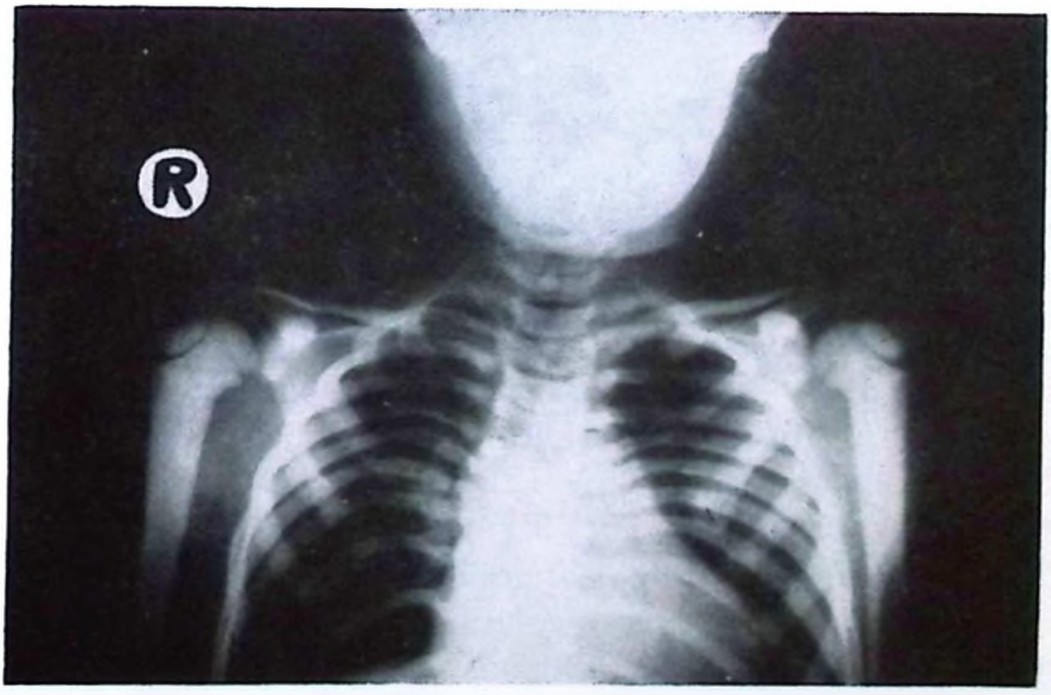

Figure 7

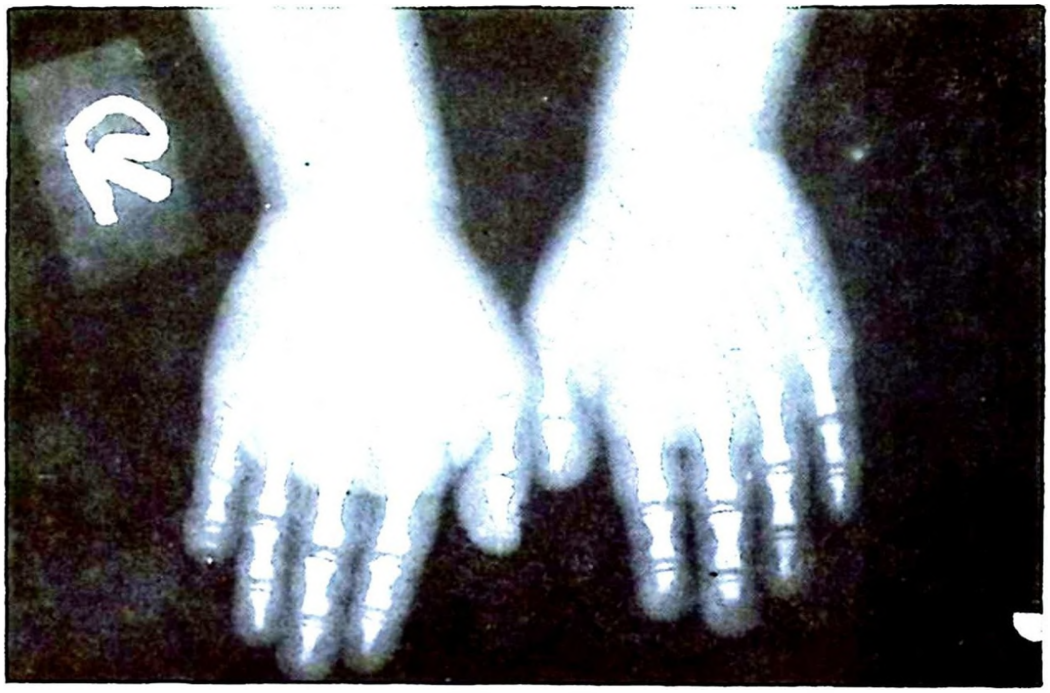

Figure 8

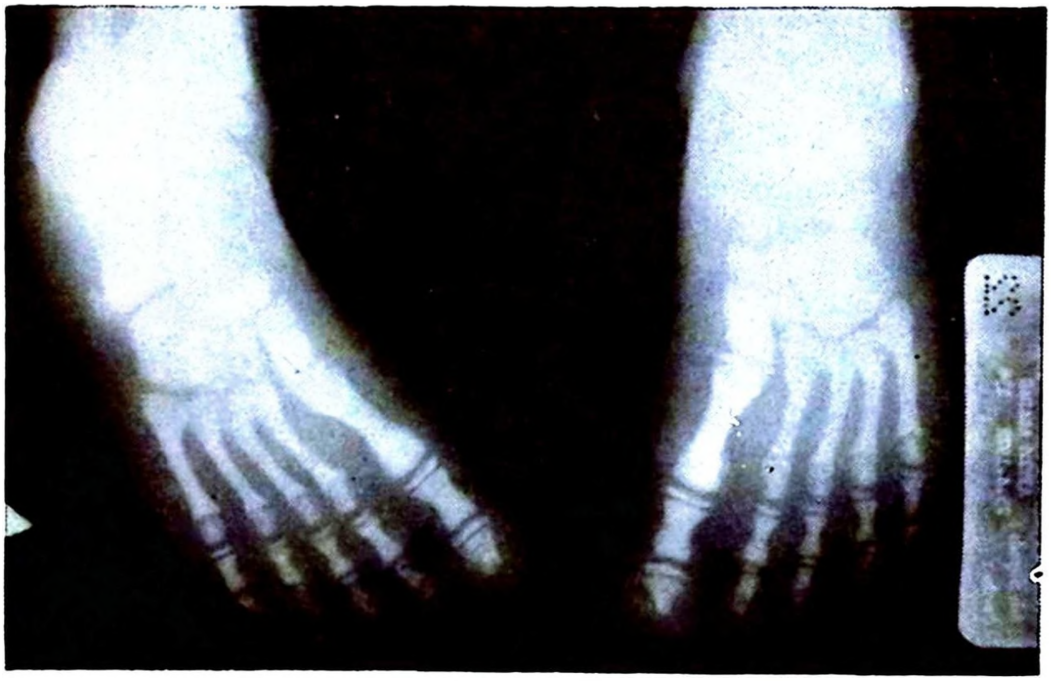

Figure 9

malies of the mid-dorsal vertebrae with fusion between the posterior elements of the D6 to D9 vertebrae (Figure 6). There was symmetrical pencil-like tapering of both clavicles at their distal ends, the distal metaphyses being completely unossified for a couple of centimeters near the acromio-clavicular joint (Figure 7). The X-rays of the hands showed tapering of the distal phalanges in addition to the osteosclerosis. There was some degree of acroosteolysis in all of the ungual tufts (Figure 8). Radiograms of the feet demonstrated the presence of identical changes in the toes (Figure 9).

Discussion

The salient clinical features of patients with pycnodysostosis consist of shortness and a peculiar facial appearance. These patients reach a height of only 1.35-1.5 metres in their adulthood.[8] The hypoplasia of the facial bones causes a rather flat appearance of the face and relative bossing of the frontal bone. These features were present in our patient. There usually is a history of easy fragility of the bones although this is not as striking as in osteogenesis imperfecta and may be altogether absent. Our patient like the patients of Shuler[8] and Blasilhas no complaints of fractures. The blood chemistry in these patients are normal and their fractures heal in a normal fashion. The mental development of these patients is normal. Girls and boys are affected equally.[4] The presence of this condition in more than one member of a family indicates its genetic nature. The mode of inheritance appears to be autosomal recessive.[1]

The radiological examination of the skeletal system reveals generalized osteopetrosis. Bone biopsies in these patients have shown abnormally dense cortical bone with very small haversian canals, thus explaining this radiological appearance.[5,8] However this is primordial bone with cellular disorganization and a failure of remodeling and therefore highly brittle, not withstanding its dense appearance. The tubular bones have thin shafts with wide metaphyses.

The diagnostic radiological findings are mainly confined to the skull. The mandible and the maxillae are very hypoplastic with loss of the mandibular angle and undevelopment of the paranasal sinuses. The teeth are irregular with multiple caries. The cranial sutures fail to close and the fontanels remain open.

There may be tapering or complete absence of the distal clavicles as seen in our patient. The vertebrae show the striking sclerosis of osteopetrosis. None of the reported cases in the literature had a congenital vertebral anomaly similar to our patient. This may be an incidental finding in our case or if future reports provide more examples of this type of anomaly, may turn out to be an occasional accompaniment of this syndrome.

The fingers and toes are short. There is tapering of the distal phalanges with varying degrees of retardation in the development of the ungual tufts. This finding was very striking in our patient with a small proximal ossicle representing the whole of a distal phalanx.

Almost the only disease to be seriously considered in the differential diagnosis is classical osteopetrosis or Albers-Schönberg disease.[11] The

radiological findings in the facial bones, cranial vault and the fingers of patients with pycnodysostosis are not seen in osteopetrosis. In addition, the hypoplastic anemia, cranial nerve compression complications and the alternating zones of greater and lesser densities of the long bone metaphyses on radiograms, all frequently present in osteopetrosis are not seen in pycnodysostosis.

The bones may be very dense in idiopathic hypercalcemia and vitamin D intoxication but the above-mentioned specific radiological changes are not seen in these disorders, and the correct diagnosis can be established without much difficulty in a patient with pycnodysostosis.

Summary

A nine year-old girl with pynodysostosis is presented. She is stunted in her growth and radiological examinations reveal severe hypoplasia of her facial bones with her fontanels and sutures remaining open. There is hypoplasia of distal clavicles and ungual tufts of the fingers and toes.

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