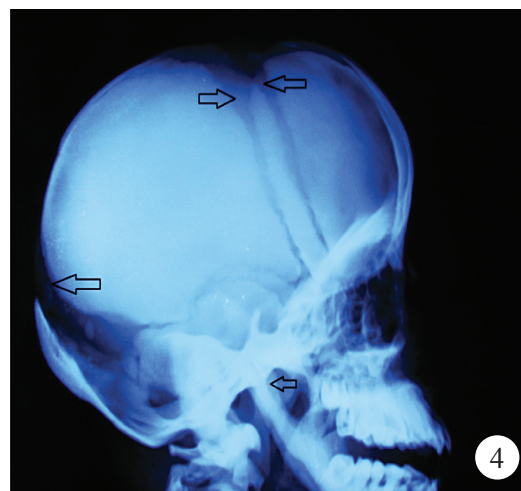
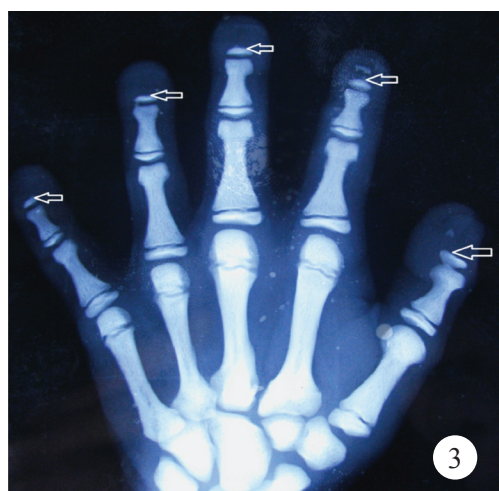
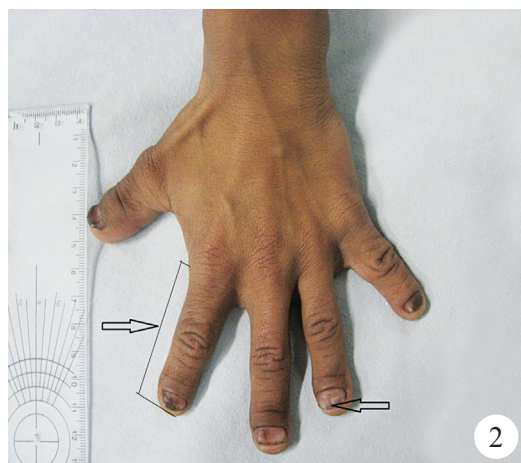
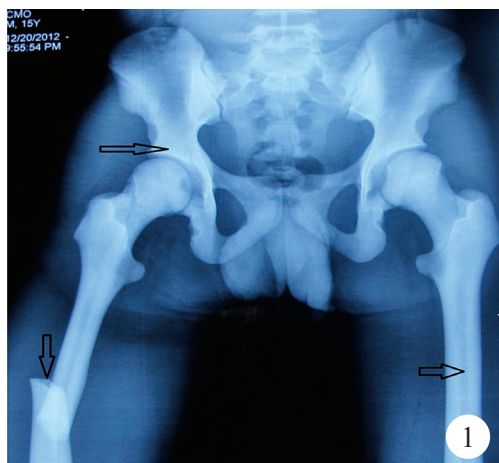


## Clinical Images

### A case of pycnodysostosis presented with pathological femoral shaft fracture



**Fig. 1.** X-ray both hips with thigh showing sclerotic bones, pathological fracture in shaft femur and narrow medullary canal (arrows). **Fig. 2.** Small fingers and grooved nails (arrow). **Fig. 3.** X-ray of hand showing hypoplastic distal phalynx (arrows). **Fig. 4.** X-ray of skull showing open anterior and posterior fontanelle and obtuse angle of mandible (arrows).

Pycnodysostosis is an autosomal recessive lysosomal storage disorder of bone in which the enzyme cathepsin K is mutated which maps on chromosome 1q21 causing osteoclast dysfunction. A 15-year old male patient of 140 cm height presented

to the Department of Orthopaedics, Maulana Azad Medical College, New Delhi, India, in November 2012 with pathological fracture in shaft of femur (Fig. 1). On the basis of clinical and radiological findings, he was diagnosed as a case of pycnodysostosis. The patient

had history of recurrent fractures with normal bone healing, open fontanelles, short stature, short finger tips, flat and grooved nails (Fig. 2), micrognathia, and irregular teeth with hypodontia. The patient had normal intramedullary haematopoiesis and no hepatosplenomegaly. Radiological evaluation of patient revealed generalized osteopetrosis, short distal phalanges (Fig. 3), obtuse angle of mandible and open fontanelles (Fig. 4). He was managed conservatively

by upper tibial skeletal traction and Thomas knee splint and showed normal bone healing pattern.

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