Rare Case of Pycnodysostosis: Case Report

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Introduction
It’s a rare disease of bone described under diastropic dwarfism. Inherited as autosomal recessive. Famous painter TOULOUSE LAUTREC had suffered from it. The term pycnodysostosis coined by Maroteaux and Lamy (1962)

Case Report
22 yr male patient resident of khammam referred to M.G.M Hospitals warangal in September 2016, with history of fall at home and fracture femur.

On Examination
LAB: HB 12%. WBC 5600. Serum Caicium, Phosphorous and ALP are normal.

Radiology
Generalized increase in bone density with cortical thickening but no obliteration of bone marrow. Open skull sutures and fontanellae. Notching of anterior vertebral bodies (spool shaped). Increased neck shaft angle (coxa plana). Aplasia of tufts of terminal phalanges of all the fingers.

Management
We operated the case by plating and above knee slab. Weight barring started at 2 months. No post op complications.

Differential Diagnosis
Osteopetrosis, Cleidocranial Dysostosis.

Discussion
Total 33 cases have been reported since 1923. Ours case adds one more. pycnodysostosis characterized by incread bone density because of cathepsin k deficiency in the osteoclaster cells. Which leads to failure of bone resorbtion especially type 1 collagen, which leads to characteristic features of this disease.
Summary

It’s a rare disease. Unlike in the literature our case does not have multiple fractures in childhood, rest all are like previous cases. This disease is not life threatening expect social disability because of short stature. From orthopedic side management is similar to other trauma cases expect practical problems while drilling the bone for screws while fixing the fractures.

References