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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 pycnodysastosis 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

Patient general condition is good. Vitals stable. Non consanguineous marriage. Short stature(138 cm).weight 45 kg. No history of muitiple fracture in childhood .fingers are short(terminal phalanx). Anterior and posterior fontanellae are open. Over hanging of cutaneous wrinkles on the fingers. Prognathism present. High arched palate. Dental caries present. Nose is beaked. Secondary sexual characters are normal. No hepatosplenomegaly. No anaemia. No blue sclera. Histroy of fracture on opposite femure operated by plating 2yrs back.
LAB: HB 12%. WBC 5600. Serum Caicium, Phosphurous 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

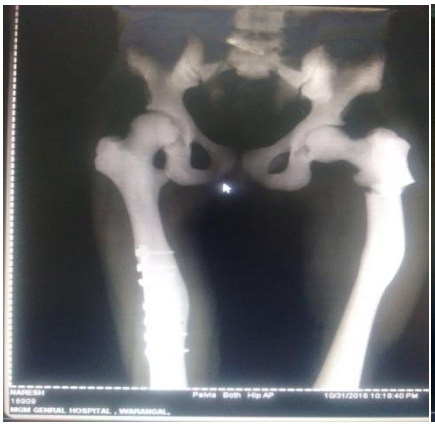
We operated the case by plating and above knee slab. Weight barring started at 2 months. No post op complications.

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 osteoclast cells. Which leads to failure of bone resorbtion especially type 1 collagen, which leads to characteristic features of this disease.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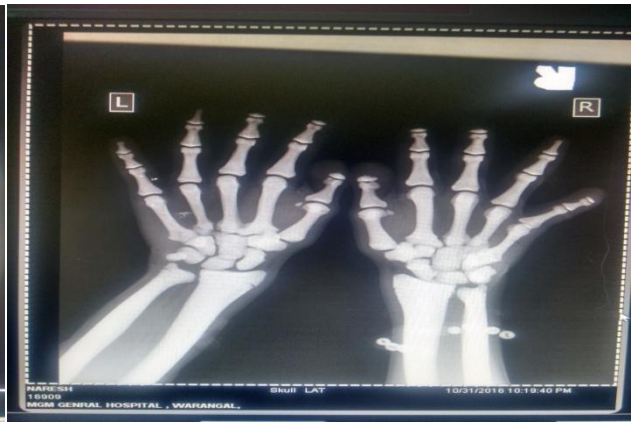
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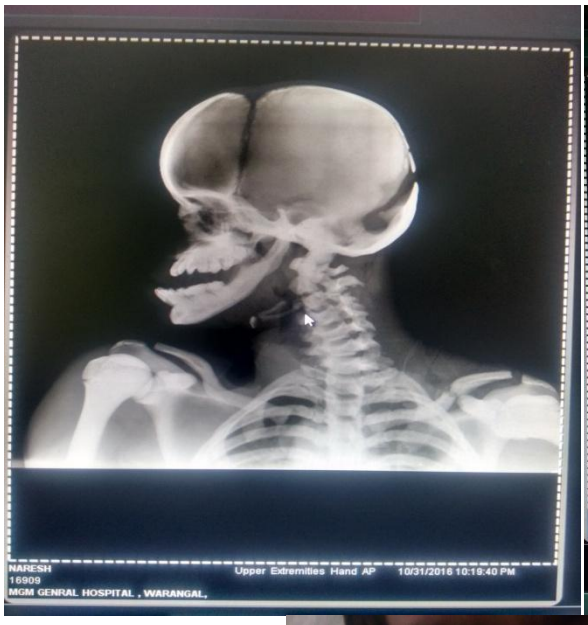
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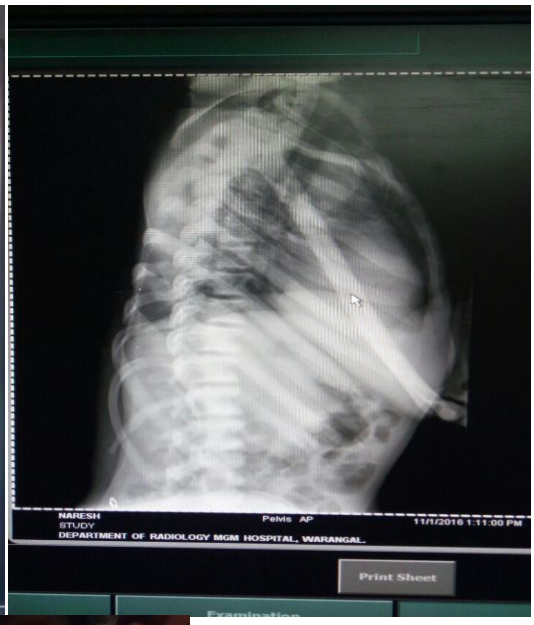
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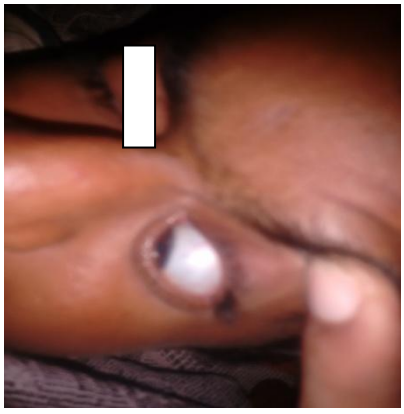

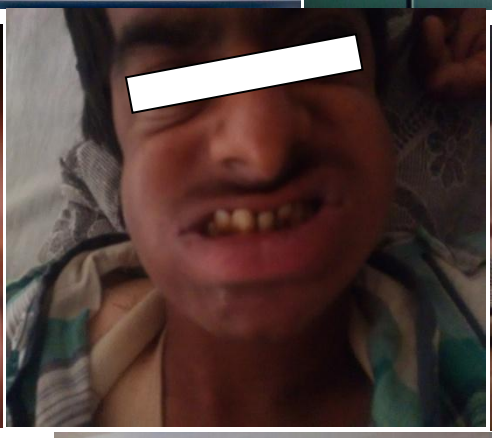

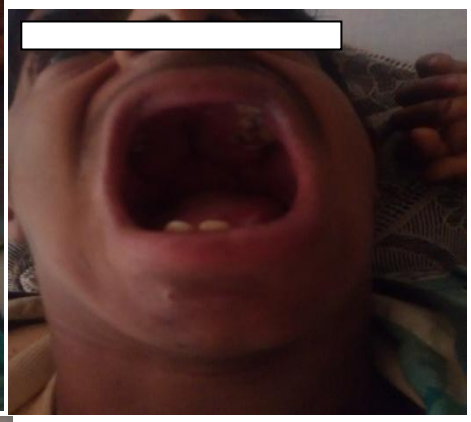

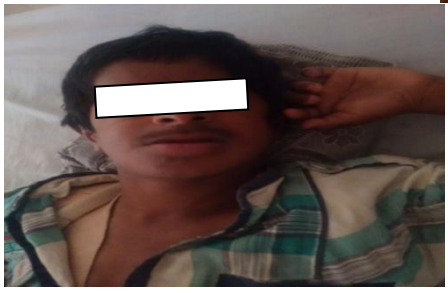

Summary

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.

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