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Pycnodysostosis with epilepsy- A rare case report.

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Abstract

Pycnodysostosis is a rare an autosomal recessive osteo chondrodysplasic characterized by osteosclerosis and short stature, maps to chromosome 1q21. Cathepsin K, a cysteine protease gene that is highly expressed in osteoclasts and is responsible for bone turnover. Due to mutation in Cathepsin K gene, patients bone become abnormally brittle and tend to suffer from numerous fractures. The estimated incidence of Pycnodysostosis is 1.7 per 1 million births. There is no gender predilection. Till now around 200 cases have been reported in the literature Pycnodysostosis with seizures is very rare. Till today only 3 case of Pycnodysostosis with seizures are reported. We present a case of 33-year-old patient who reported to our department with chief complaint of multiple decayed teeth and misalignment of teeth and rare presentation of Pycnodysostosis with epilepsy.

Keywords: Pycnodysostosis, cathepsin k gene, epilepsy, osteosclerosis, chromosome 1q21

Introduction

Pycnodysostosis is rare autosomal recessive genetic disorder. It is caused due to mutation in the gene Cathepsin K encoding cysteine protease.[1]. It was first described by Maroteaux and Lamy in 1962. [2,3] Other authors have named it the Toulouse-Lautrec syndrome, as Henri de Toulouse Lautrec the French painter is claimed to have suffered from the disorder [2].

This disorder manifests as generalized osteosclerosis of the skeleton due to decreased bone turnover.[4] The patient presents with multiple fractures of long bone due to increased bone density, and abnormally brittle bone

[5]. The estimated incidence of Pycnodysostosis is 1.7 per 1 million births. There is no gender predilection. Till now around 200 cases have been reported in the literature [6,7]. Diagnosis of Pycnodystosis is based on clinical and radiographic features [8]. We present a case of a 33-yearold male patient with the chief complaint of decayed teeth and misalignment of teeth, and a history of multiple fractures of long bones and epilepsy.

Case Report

A 33-year-old patient reported to the Department of Oral Medicine and Radiology with the chief complaint of multiple decayed teeth and misalignment of teeth. The patient's medical history revealed multiple fractures of long bones since he was 4 years old. He had fractures in his left tibia, right and left femur, and in right radius. The patient also gave a history of epilepsy since his childhood and was taking antiepileptic drugs gardenal sodium till 6-7 years of his age and then stopped the drug. He now sometimes experiences seizures. He experienced his last seizure 1 year back. But now he does not take medications for the same. His radius fracture was managed with parathyroid injection and was also given calcium tablets.

There was a positive history of the consanguineous marriage of his parents. He had three siblings. None of them were affected. A general examination of the patient revealed short stature (145cm) and proportionate dwarfism. [fig 1] Built was moderate. Fingers and toes are short [fig 2 and 3]. Various facial features were a retrognathic mandible [fig 4], a beak-shaped nose [fig 4], and bilateral proptosis of eyes [fig 5]. Vital signs were within normal limits.

A dental examination revealed crowding of teeth, multiple decayed teeth, a narrow and deep groove was seen on the palate [fig 6], bald tongue to some extent [fig 7].

Based on the history and clinical findings, a provisional diagnosis of dysplastic bone and a differential diagnosis of osteo genesis imperfecta, cleidocranial dysostosis, osteopetrosis, and pycnodysostosis was given.

The patient was advised for investigations like complete blood count, Serum Calcium -9.1mg/dl, Serum Phos phorus-4.1mg/dl, and Alkaline Phosphate-102u/I. All blood investigations were within normal limits. We per formed various radiological examinations of patients like Ortho pantomagram (OPG), Cone Beam Computed Tomography (CBCT), lateral, and front cephalogram.

OPG and CBCT revealed generalized increased bone density, osteosclerosis, obtuse mandibular angle [fig 8], elongation of the styloid process [fig 8], underdeveloped or hypopneumatized sphenoid, maxillary and frontal sinus [fig 9-12]. All fontanelles and cranial sutures were open [fig 13-17]

A fracture of the radius shaft 1/3rd region proximal to the implant was seen [fig 18]. Hand-wrist radiograph revealed mild acro-osteolysis of distal phalanges. [fig 19]

Based on clinical and radiological findings, a final diagnosis of pycnodysostosis with epilepsy was made.

Discussion Pycnodystosis is a rare autosomal recessive disorder characterized by generalized systemic osteo sclerosis due to decreased bone turnover. In this condition osteoclasts are defective which leads to impaired bone resorption and remodelling, which is essential for normal bone maintenance, both during growth and healing. This results in increased bone density and brittleness, which makes it susceptible to fracture. [9, 6] This disorder is usually diagnosed in childhood. However, it is sometimes diagnosed in latency as in our case. Life expectancy on this patient is normal. [6]

Condition such as cleidocranial dysostosis, osteo genesis imperfecta, and osteopetrosis should be considered for differential diagnosis. Cleidocranial dysostosis presents with open fontanelle and cranial sutures this condition is always affects clavicle and there is no increase in bone density. [8, 6]

In osteogenesis imperfecta type 1 collagen is affected. Patient has blue sclera and are prone to multiple fractures. But no increase in bone density [10, 6]

In osteopetrosis there is increase in bone density. Patient are more prone to fractures. fontanelle and cranial sutures are close. Average life expectancy in this patient is 11 years. [11, 6]

Pycnodysostosis with seizures is very rare. Only two case of pycnodysostosis with seizures have been documented to yet, and this is the third case. [12,13]

The main clinical maxillofacial features in the pycnodysostosis include a grooved palate, midfacial hypoplasia, mandibular hypoplasia, enamel hypo plasia, dental crowding, narrow palate, cross bite, class III skeletal, open bite and dental abnormalities.

The main radiographic maxillofacial features in the pycnodysostosis include obtuse mandibular angle, large head with frontal, parietal and occipital bossing, open soft cranial sutures and fontanelles, multiple impacted teeth, absence or hypo peumatization of the paranasal sinuses, Wormian bones, elongation of the coronoid process and the condyle, supernumerary teeth.[14] most of these features were evident in our case and epilepsy was added feature.

As there is no treatment for Pycnodystosis and aim of the management is usually to improve overall oral health of the patient. Precaution also needs to be taken as osteomyelitis is common complication in patient undergoing extraction.

Conclusion

Pycnodystosis as it is a rare condition should be diag nosed early for proper management of patient. Dentists should be aware of this condition and should do everything possible to make patients routine comfort able. We can only treat patient symptomatically.

Declaration of patient consent

The authors certify that they have obtained all ap propriate patient consent forms. In the form the patient (s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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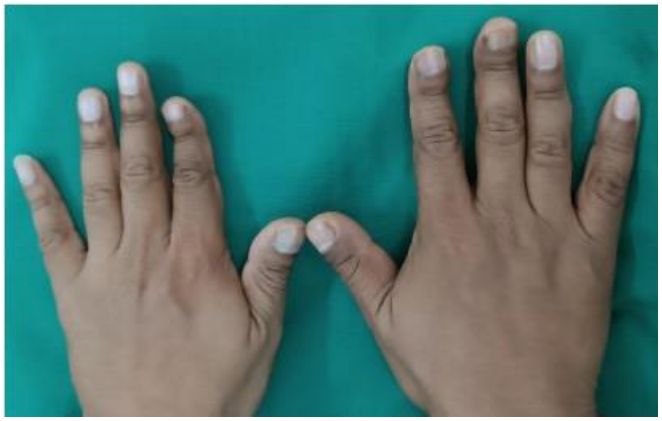
Legend Figures and Tables

Description (legends)
Stature is short with proportionate dwarfism
Phalanges of upper limb are short
Phalanges of upper limb are short
Retrognathic mandible, convex profile, beak shaped nose
Proptosis of eyes
crowding of teeth, multiple decayed teeth, a narrow and deep groove was seen on the palate
Baldness of tongue to some extent
Osteosclerosis seen in 15,16,17,25,26,27,36,37,46,47 Elongation of styloid process There is widening of the alveolar nerve canal

9	Mandibular angle is obtuse, Maxillary sinus is hypopneumatized Sphenoid sinus is hypopneumatized
10.	Maxillary sinus is hypopneumatized
11	Maxillary sinus is hypopneumatized
12	Frontal and Sphenoidal sinus is hypopneumatized
13.	Coronal and sagittal suture are open
14	Mastoid fontanelle is open
15	Posterior fontanelle is open
16	Lambdoid suture is open
17	Maxillary sinus is underdeveloped, Styloid process is elongated
18	Fracture of Radius shaft proximlL 1/3rd region proximal to previous implant
19	Mild acro-osteolysis of distal phalanges

8-15 and 18-19	Osteosclerosis

Figure 1:



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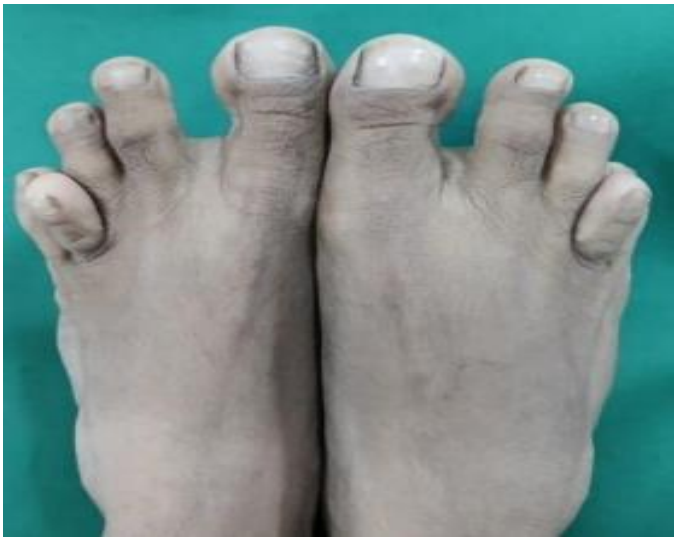

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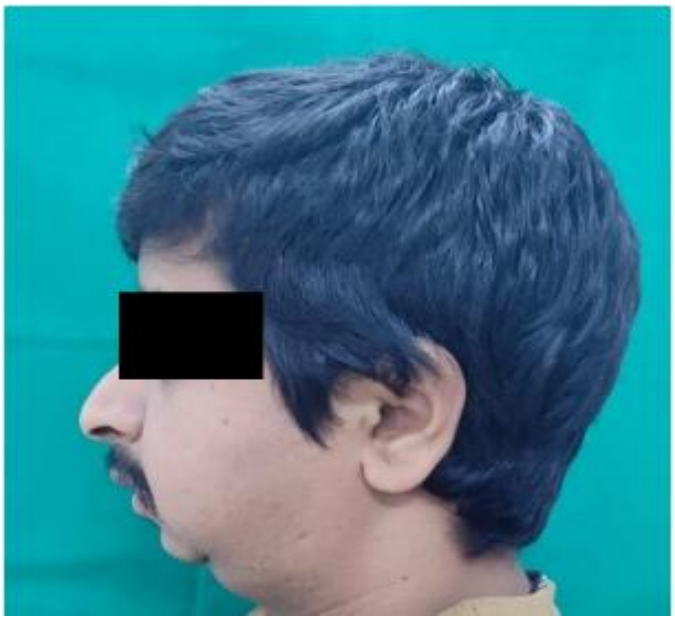

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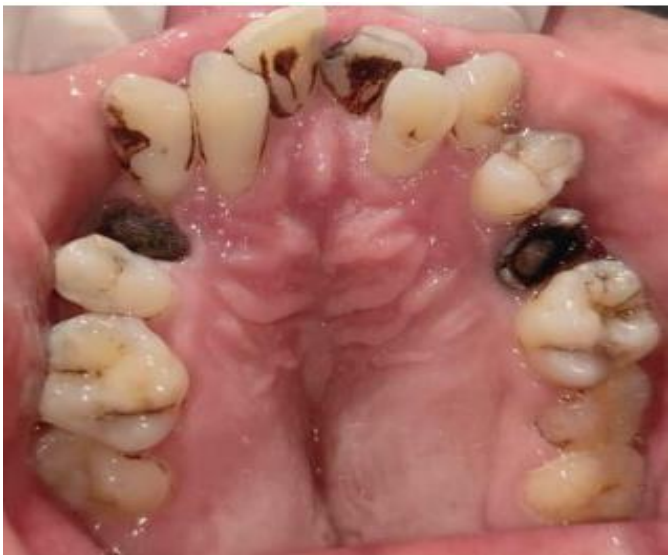
Figure 5:



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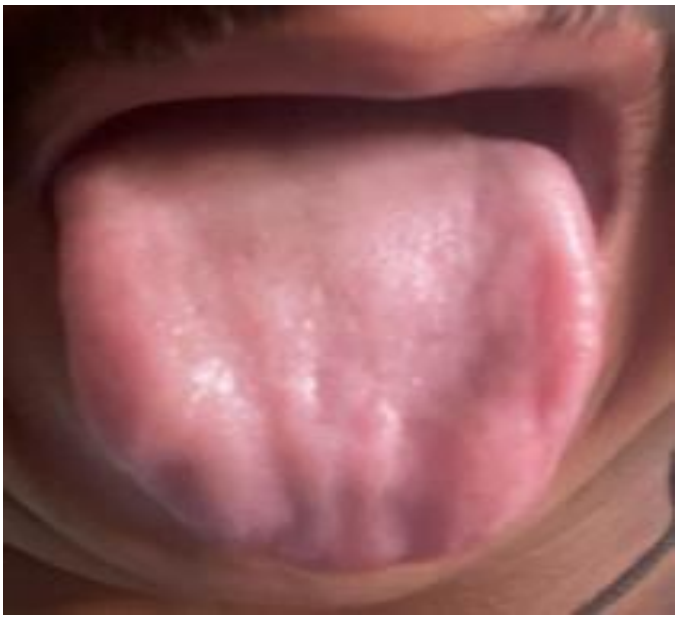

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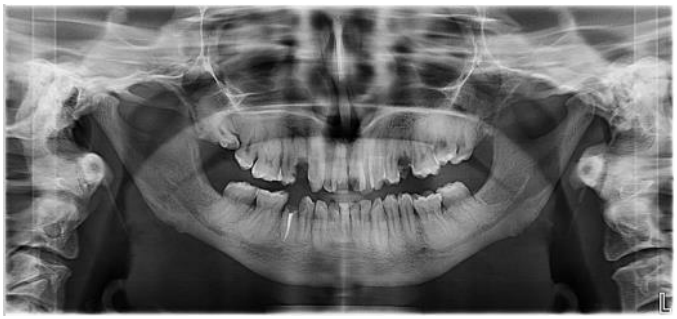

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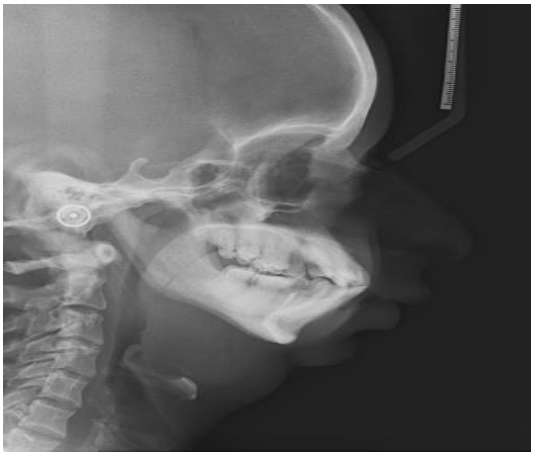

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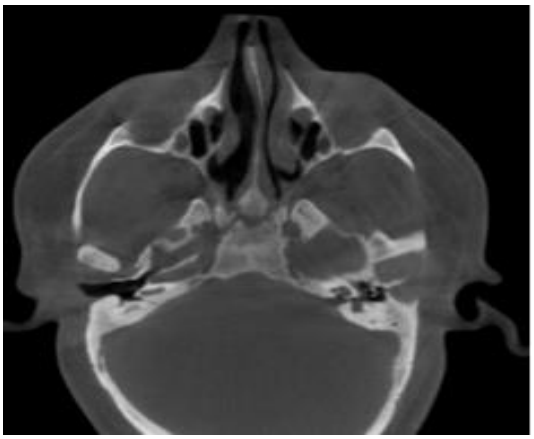

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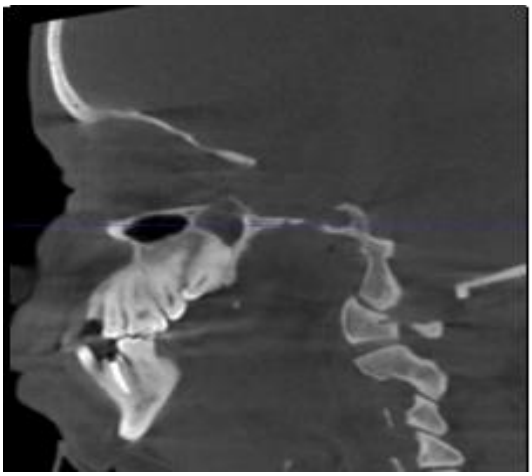

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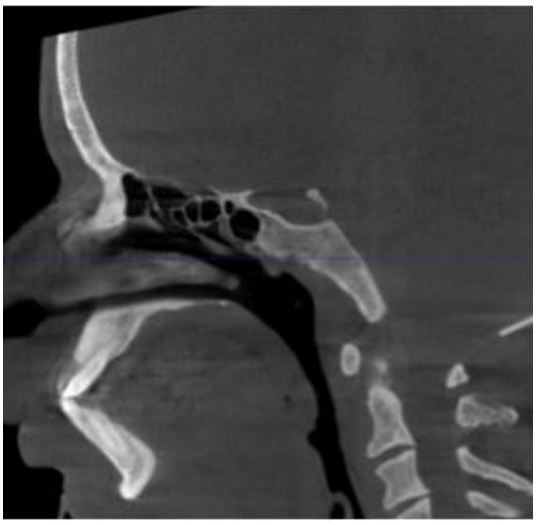

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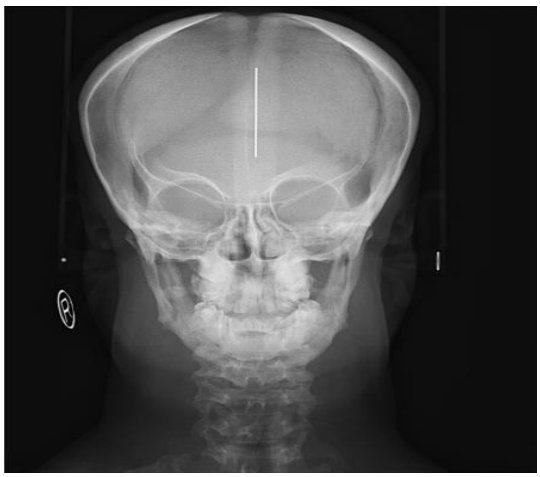

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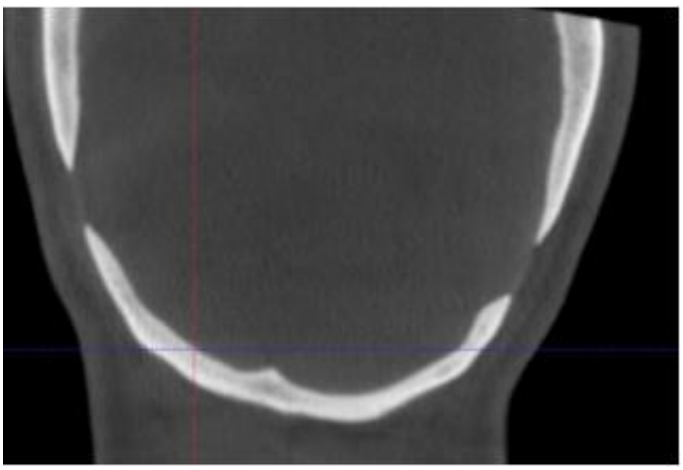

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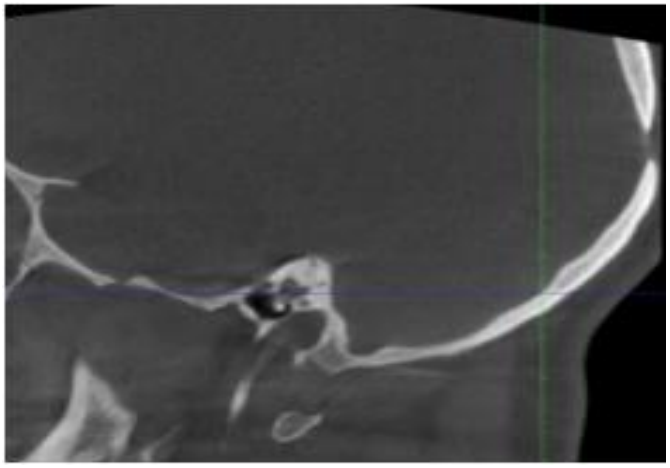

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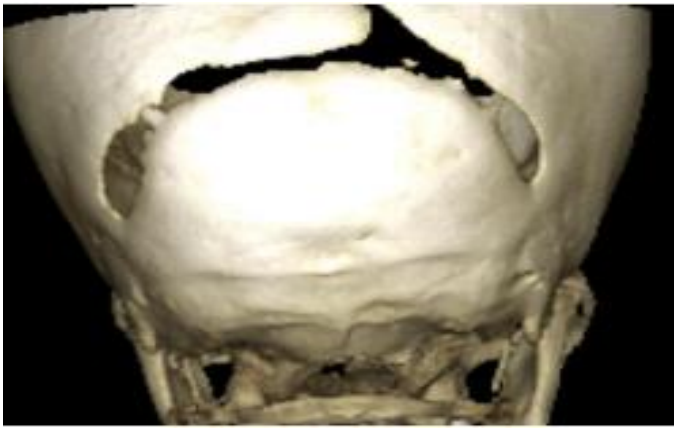

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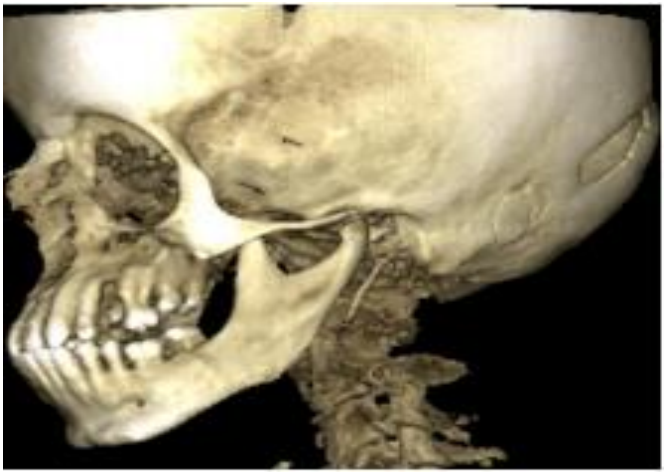

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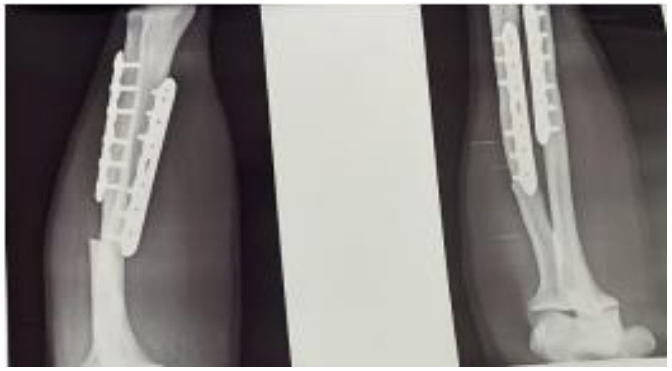

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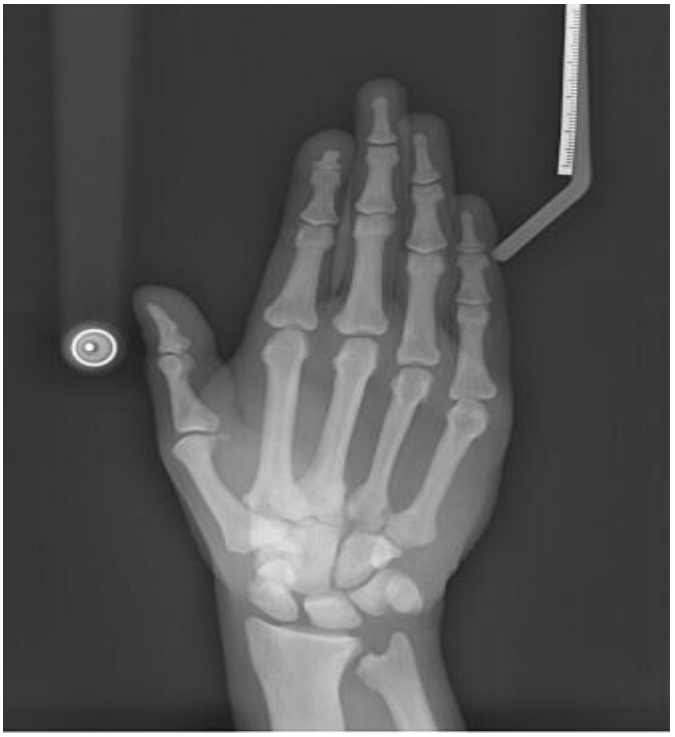

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