Pyknodysostosis: a case report

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Introduction

Pyknodysostosis, also called Toulouse-Lautrec syndrome, was described in 1962 by Maroteaux & Lamy[1]. It is an autosomal recessive genetic disorder with an incidence of 1.7 per million[2]. We report a case of pyknodysostosis.

Case report

A ten-year-old girl presented with short stature and an ulcer over the right foot. There was a history of repeated fractures, early exfoliation of deciduous teeth and unerupted permanent teeth. Birth was not complicated. There was no history of delayed milestones, hearing and visual impairments or intellectual disabilities. There was no family history of bone disorders or short stature.

On examination, weight was 15kg, height was 95cm and both were below -3SD as per World Health Organisation growth charts. She had wide open anterior fontanelle and fronto-parietal bossing. There was poorly developed maxilla, hypoplastic jaw, nasal beaking, high arched palate and obtuse (increased) mandibular gonial angle with prognathism (Figure 1).

Her fingers were short, stubby and nails were flat, grooved and dysplastic. She had a healed fracture of the left leg with an ulcer on the right foot covered with a bandage (Figure 2).

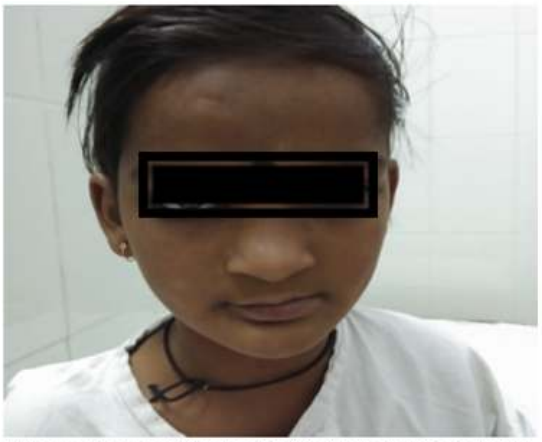

Figure 1: Face-Hypoplastic jaw, beaked nose
Permission given by parents to publish photograph

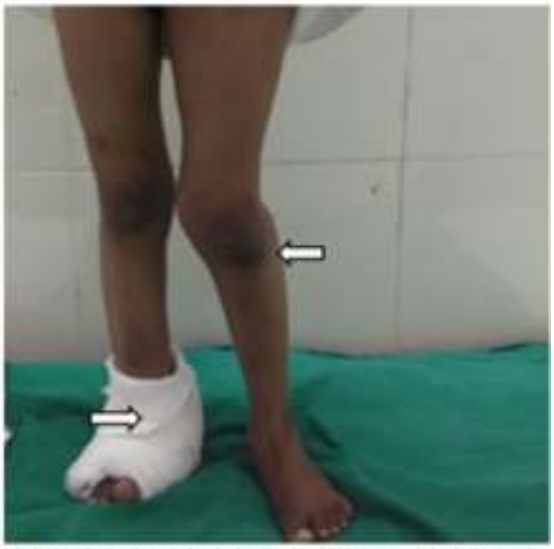

Figure 2: ***Healed fracture of left leg (marked with arrow) with ulcer on right foot covered with bandage (marked with arrow)***

Other system examination was normal. X-ray skull showed open cranial sutures, hypoplastic maxillae and mandible, obtuse (increased) mandibular gonial angle, with improperly developed dentition and multiple impacted permanent teeth (Figures 3).

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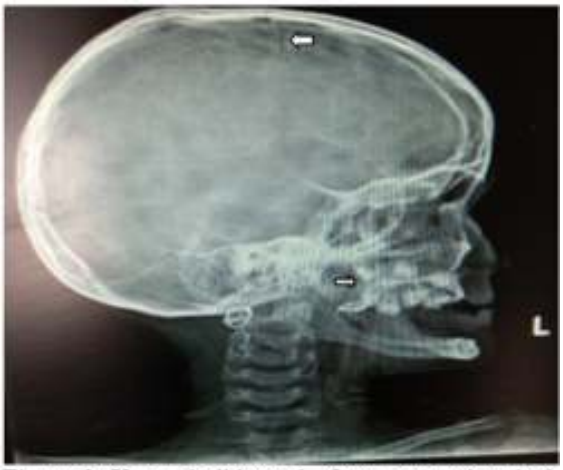

Figure 3: *X-ray skull lateral - Open suture (marked with arrow), obtuse gonial angle and multiple impacted permanent teeth (marked with arrow)*

X ray of both hands, wrists and lower end of radius and ulna suggested that cortices were thick and dense with preservation of the medulla and terminal phalanges were short and hypoplastic (Figure 4).

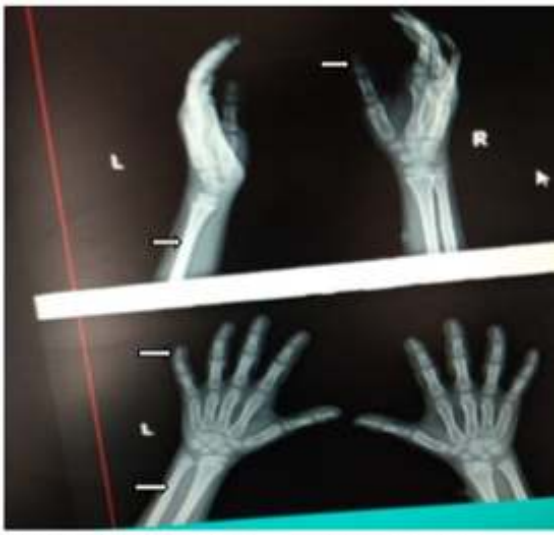

Figure 4: *X-ray of both hands, wrist, lower end of radius and ulna showing thick, dense cortex with preserved medulla (marked with arrow) and short hypoplastic terminal phalanges (marked with arrow)*

X ray chest and spine was suggestive of vertebral body sclerosis (Figure 5).

Complete blood count, erythrocyte sedimentation rate, peripheral smear, serum calcium, kidney function test, liver function test and thyroid function test reports were normal.

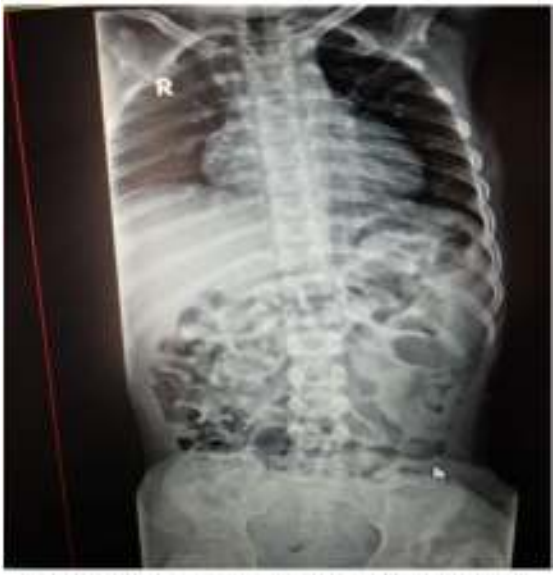

Figure 5: *X-ray spine- Vertebral body sclerosis*

Discussion

Pyknodysostosis is caused by dysfunction of osteoclasts resulting in osteosclerosis. It is due to defect in cathepsin K (CTSK) gene located on chromosome 1q21, which encodes a lysosomal protease enzyme, predominantly found in osteoclasts that absorb bone tissue during growth and healing[2]. It results in defective bone resorption and new bone formation leading to abnormally dense and brittle bones[3]. The classical features are short stature, open fontanelles, straightened mandible, maxillary hypoplasia and high susceptibility to fractures. Radiographs show acro-osteolysis with sclerosis of the terminal phalanges, obtuse angle of mandible, osteosclerosis with narrowed medullary cavities, increased opacity of long bones, spine and base of skull and facial bone hypoplasia or dysplasia which may be partial or total. Crowding of the teeth and multiple impacted supernumerary teeth are the prominent intraoral features[3,4,5].

Osteopetrosis and cleidocranial dysplasia are differential diagnoses of pyknodysostosis. Osteopetrosis is characterized by diffuse osteosclerosis involving medullary bone, involvement of cranial nerves, stunted growth but no maxillary hypoplasia, normal cranial sutures and gonial angle. Cleidocranial dysplasia shows clavicles which are aplastic or hypoplastic but no diffuse osteosclerosis and long bone involvement and normal height. Diagnosis of pyknodysostosis is primarily based on clinical features and x-rays, but the CTSK gene mutation analysis is confirmatory[2]. It was not done in our patient due to economic constraints. Treatment is only supportive. Dental treatment must be planned carefully because of the risk of complications such as jaw fracture and osteomyelitis, which are more common in adults.

Planned tooth extraction or dental implants can be done. No protocol is available for orthodontic treatment because the low remodeling capacity of the bone puts the patient at high risk of orthodontic failure[6-10]. Intelligence and life expectancy are normal[9,10].

Early diagnosis of pyknodysostosis can give a better quality life to the patient with timely, specialized dental care, treatment of bone fractures and craniofacial surgeries. Our patient had the classical features of pyknodysostosis. Growth hormone therapy for short stature in pyknodysostosis and enzyme inhibitors for treatment require clinical trials.

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