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Pyknodysostosis: Report of A Rare Case

KEYWORDS

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ABSTRACT *Pyknodysostosis is a rare autosomal recessive sclerosing bone disorder, characterized by short stature, generalized diffuse osteosclerosis. Patient usually has a large head with separated sutures and open fontanelle. We report a 5 year old child with features pathognomic of Pyknodysostosis. The emphasis is mainly on the early diagnosis as it has an important role in the general health of such patients and prevention of complications.*

INTRODUCTION:

Pyknodysostosis is a rare autosomal recessive disorder. The main features of patients with pyknodysostosis are short stature, acro-osteolysis of distal phalanges, deformity of the skull, dysplasia of the clavicles, narrow and/or grooved palate, midfacial hypoplasia, absence or hypopneumatization of the paranasal sinuses, generalized osteosclerosis and fragility of bone [1,2]. Other features include wrinkled skin, finger and nail abnormalities, kyphosis and scoliosis, history of repeated chest infections, and sleep apnea. The intellectual and sexual development is usually normal in the patients[3,4].

This syndrome has been seen in many races and nationalities, including black, Arab and Caucasians[5]. Treatment is symptomatic and focused mainly on the management of dental problems and fractures. The prognosis is generally good, and patients typically reach heights of 130-150 cm[6]. This condition poses problems for good oral hygiene which leads to a high rate of caries and periodontal disease in these patients [7,8]. In some cases surgical intervention is required to correct malocclusion and esthetics in patients with dentofacial deformities caused by micrognathia and hypoplasia of maxilla [9,10,11].

CASE REPORT:

A 5 year old boy presented in paediatric opd of mmimsr, mullana with dysmorphic facies, short stature and failure to thrive. He is the first child of a non consanguineous couple and has one sister 1 nd half yr old. Child was born by full term vaginal delivery in hospital with no complications. Birth weight was normal and maternal antenatal history was uneventful. Child was fully immunised with normal developmetal milestones and intelligence. There was no history of frequent chest infections, fractures or trauma. On examination, vitals were normal, weight was 11kg(<3rd centile), height was 90cm(<3rd centile) and occipitofrontal head circumference was 47cm(<3rd centile). Upper segment was 50 cm and lower segment was 40cm with US:LS ratio of 1.2:1. (Fig. 1)

(Fig. 1: picture of patient showing short stature and dysmorphic features)

The child had frontal and parietal bossing. The sagittal, coronal and lambdoid sutures were separated and anterior and posterior fontanels were wide open. Dental examination revealed a narrow palate and overcrowding teeth. Extremities had short digits. There was no pallor, icterus, clubbing or lymphadenopathy. Lab investigations revealed normal complete blood count, serum calcium, liver function test, renal function test, thyroid function test, and an alkaline phosphatase of 233 (normal 179-416 IU/l).

The radiological findings were significant. X ray skull showed widened skull sutures and anterior fontanelle wide open with evidence of sutural diastasis(Fig. 2). Wormian bones were seen at lambdoid suture. Mandibular angle was absent and facial bones appeared smaller as compared to skull bones.

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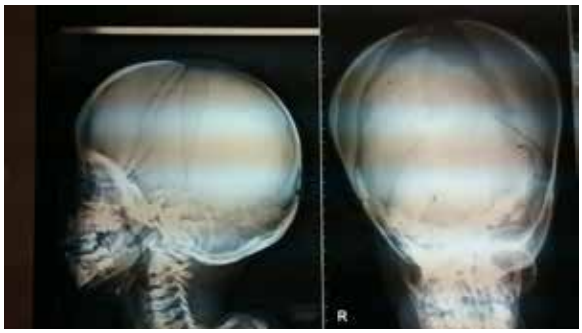
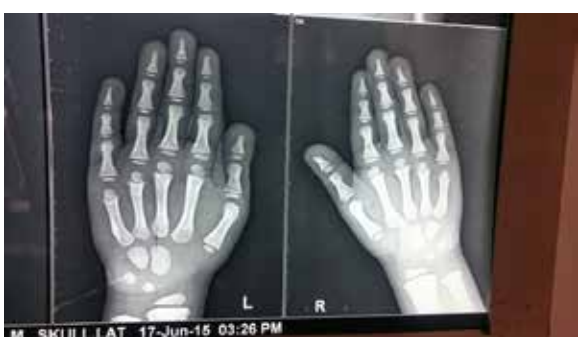


Fig 2: Xray skull showing wide open sutures with open fontanelle

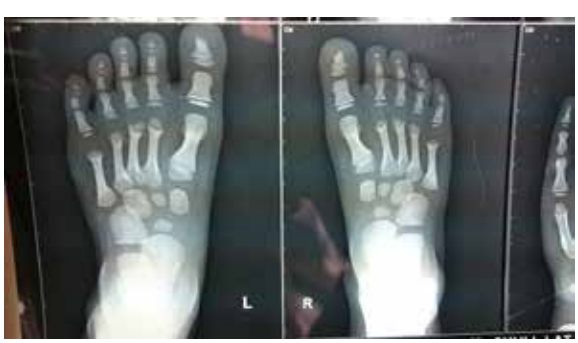
In xray chest the clavicles were normal. On lateral view of spine dorso lumbar vertebrae were spol shaped with anterior large defect.

In xray of pelvis, there is evidence of congenital coxa vara with decreased head and neck angle. Slightly increased density is seen at metaphyseal ends of both femurs. Medial cortices of both tibia is thickened. Epiphysis of lower end of tibia is impinging into the bony defect at lower end of tibia.

Xray of both hands and feet showed evidence of acro osteolysis (absorption of terminal phalanges)(Fig. 3A and 3B).



(Fig. 3A and 3B: Xray of both hands and feet showing acro osteolysis)



He was diagnosed as Pyknodysostosis based on characteristic clinical and radiological findings.The parents of the child were advised for CTSK gene mutation testing for confirmation of diagnosis of pyknodysostosis, however they refused for same in view of financial constraints.

DISCUSSION:

Pyknodysostosis is an autosomal recessive disease characterised by systemic osteosclerosis owing to decreased bone turnover[12]. During the 1990s, the genetic defect was located on chromosome 1q21 which led to specific genetic testing and accurate diagnosis[13]. A mutation in the gene that codes for the enzyme cathepsin K inhibits the normal functioning of osteoclasts[14]. Defective osteoclasts cause impaired bone resorbtion and remodelling, which is essential for normal bone maintenance, both during growth and healing[15]. Bones in affected individuals are therefore abnormally dense and brittle and easily fracture .

The main clinical maxillofacial features in the pyknodysostosis include a grooved palate, midfacial hypoplasia (observed in many-66.66% and 60.60% respectively), mandibular hypoplasia seen in 39.39%, dental crowding in 36.36%, narrow palate seen in 27.27%, cross bite in 27.27%, and dental abnormalities were observed in21.21%. The main radiographic maxillofacial features in the pyknodysostosis include obtuse mandibular angle (94.28%), large head with frontal, parietal and occiptal bossing (80%), open cranial sutures and fontanelles (77.14%), multiple overcrowded teeth (48.57%), absence or hypopeumatization of the paranasal sinuses (42.85%), wormian bones (20%) and the supernumerary teeth (5.71%) [16].

In our case study, the general features were mainly dysmorphic with short stature ,acro-osteolysis of distal phalanges, open fontanelle with wide open sutures, dental crowding, frontal and parietal bossing, pneumatization of paranasal sinuses which is the essential pathognomonic and similar findings have been reported by Pavani et al[17] and Quais et al[18]. According to some studies, intraoral features such as persistence of deciduous teeth with premature or delayed eruption of permanent teeth leading to crowding, enamel hypoplasia, extensive dental caries, narrowing of pulp cavities, shortness of tooth root, hypodontia or supernumerary, and hypercementosis are specific to pyknodysostosis[4,19,20]. Other reported abnormalities were midline antero-posterior ridge of the palate, high arched palate, macroglossia, and infection of unerupted permanent teeth follicles, leading to abscess formation. Also, there were reports which observed anomalies of various permanent teeth[3,4,19].

The differential diagnosis of pyknodysostosis is established with osteopetrosis, cleidocranial dysplasia and idiopathic acro-osteolysis.

The diagnosis of pyknodysostosis is primarily based on clinical features and radiographs; however a CTSK gene mutation analysis is the confirmatory test[18]. There is no specific treatment as known of date for this disorder and treatment is basically supportive. Since bone fractures are a primary threat to these patients, it is important that care is taken to prevent or minimize tendencies for fractures to occur. Such precautions include careful handling of an affected child, along with exercise and activities that are safe. Dental hygiene and regular dental checkups are especially helpful for affected individuals due to various dental anomalies.

CONCLUSION:

Pyknodysostosis is a rare condition that is diagnosed basically on its radiographic features and clinical features. The importance of recognition of these features in the diagnosis allows correct treatment and prevention of future complications and ensuring better quality of life to the patient.

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