Pycnodysostosis: Clinicoradiographic Report of a Rare Case

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Abstract

Pycnodysostosis is an uncommon autosomal recessive sclerosing bone disorder which is characterized by short stature and generalized diffuse osteosclerosis. Patients usually have a large head with separated sutures, open fontanels, aplasia of frontal sinuses, obtuse mandibular gonial angle, and acroosteolysis of the distal phalanges. This case report showed a 25-year-old female with features pathognomonic of pycnodysostosis. 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.

Keywords: *Acroosteolysis, obtuse mandibular gonial angle, osteomyelitis, osteosclerosis, pycnodysostosis*

Introduction

Pycnodysostosis was documented as far back as 1962 by Maroteaux and Lamy and is also known as Toulouse–Lautrec syndrome. It is a rare autosomal recessive disorder with a prevalence rate of 1–1.7 in one million.[1] Pycnodysostosis is defined by a short stature, acroosteolysis (resorption of the terminal phalanges), generalized osteosclerosis with fragile bones, open skull sutures, and dysplasia of the clavicle. Minor trauma can also give rise to fractures in such patients.[2]

Case Report

A 25-year-old female measuring 140 cm in height and weight 38 kg reported to the oral medicine and radiology department, with a chief complaint of swelling in and pus discharge from the left mandibular body region since 15 days [Figure 1].

The patient had a history of bilateral femur fractures 1 year ago, which was preceded by only minimal traumatic event. This was treated surgically with bone plates and screws [Figure 2]. Her medical history was insignificant with no history of parental consanguinity. Patient was fully immunized with normal developmental milestones and intelligence.

General physical examination revealed dysmorphic features such as short stature, frontal and bilateral parietal bossing and hypoplastic midface, short, broad, spoon-shaped, stubby digits, and dysplastic grooved nails, toes showing sandal gap deformity [Figures 3 and 4]. On extraoral examination, swelling and pus discharge in relation to the left mandibular body. Dental examination revealed inflamed gingiva with pus discharge in the left lower posterior edentulous region. On hard-tissue examination, high-arched narrow grooved palate, microdontia, crowding, missing teeth, and multiple carious teeth were reported [Figure 5].

The above-mentioned clinical findings led us to a provisional diagnosis of chronic suppurative osteomyelitis. Differential diagnosis was based on general examination, intraoral features, and patient history and following were suspected, osteopetrosis, pycnodysostosis, and cleidocranial dysostosis. The patient was subjected to further investigations.

Panoramic radiograph (orthopantomogram) showed both maxillary third molars to be microdontic, maxillary right lateral incisor was palatally malposed, maxillary left lateral incisor was missing, and impacted maxillary left second molar. Notably, bilateral sequestra presented in relation to mandibular body region, presence of obtuse mandibular angles, resorption of posterior borders of rami, elongation of the condylar apophyses giving mild beaked shape condylar appearance, partially pneumatized maxillary sinuses, generalized osteosclerosis of maxilla, and mandible

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with fracture of inferior border of mandible in the left body region [Figure 6].

Hand–wrist radiograph showing acroosteolysis of distal phalanges and osteosclerosis [Figure 7].

Skull and facial plain radiographs and computed tomography scan showed a brachycephaly, with persistent open fontanels and wide sutures, thick calvarium, generalized osteosclerosis of skull and facial bones,

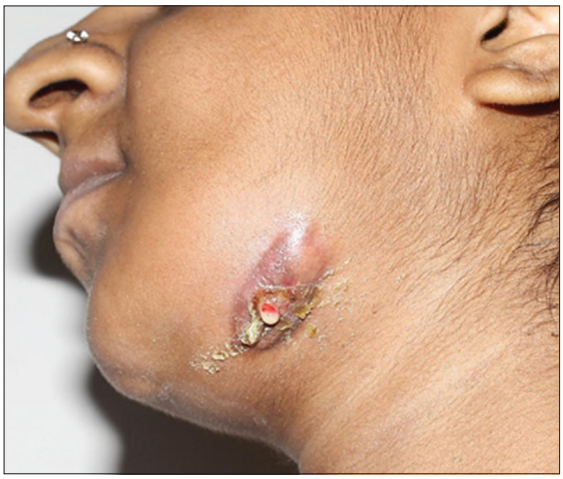

Figure 1: Swelling and pus discharge from the left submandibular body region

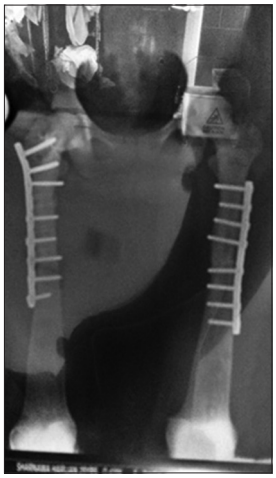

Figure 2: Pelvic and long bone X-ray showing bilateral fractures of femurs treated with bone plates and screws

Figure 3: Characteristic craniofacial features-short stature, frontal bossing, midface hypoplasia, beaked nose

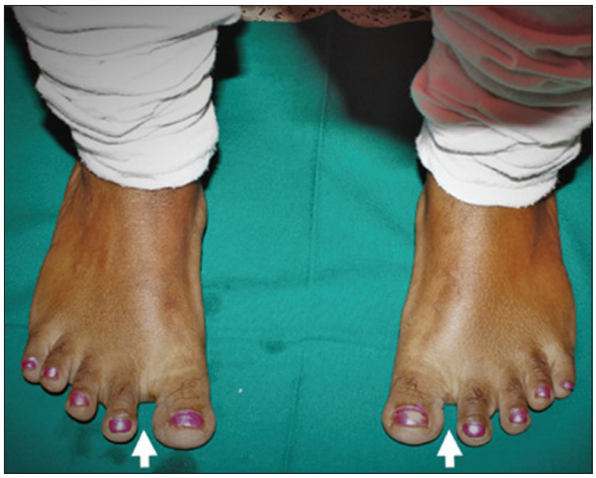

Figure 4: Toes photograph showing sandal gap deformity (white arrows) and short-broad digits

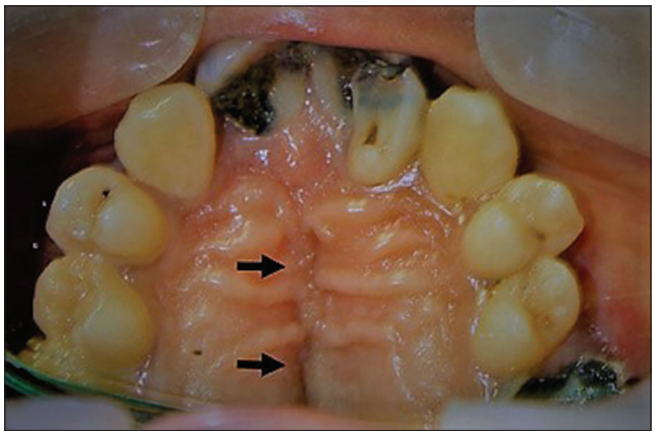

Figure 5: Maxillary arch showing deep grooved narrow palate (black arrow) and upper anterior crowding

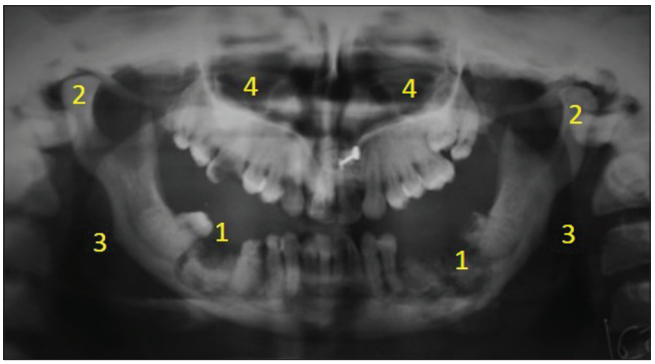

Figure 6: Orthopantomograph showing osteomyelitis (1) bilaterally in the mandibular body region with pathological fracture at inferior border in the left side, elongation of the condyles apophyses (2), an obtuse mandibular angle (3), partially pneumatized maxillary sinuses, generalized osteosclerosis of maxilla and mandible (4), partially pneumatized maxillary sinuses

and abnormal articulation between anterior arch of first cervical vertebrae (C1) to occipital condyle on the right side. The maxilla was hypoplastic with nonpneumatization of the frontal sinuses and depressed nasal bridge. Underdevelopment of the mandible was noticed with an obtuse jaw angle [Figures 8-10].

All laboratory investigations were normal, including liver and renal function tests, serum electrolytes, serum calcium, and phosphate. Alkaline phosphatase level was found to be within normal limits.

The characteristic clinicoradiographic features led us to a probable diagnosis of pycnodysostosis with suppurative osteomyelitis of mandible.

In the above case, the left body of the mandible showed a pathologic fracture; hence, sequestrectomy and saucerization were carried out along with plating along the inferior cortex of the mandible. Similar treatment is planned for treating the osteomyelitic changes seen on the right side.

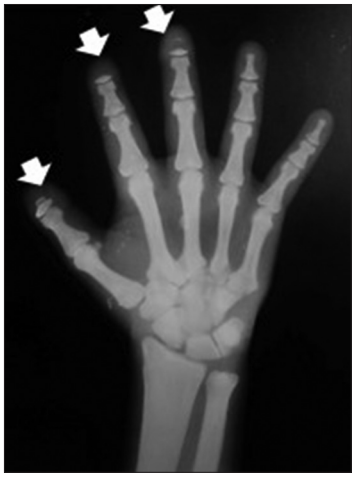

Figure 7: Hand–wrist radiograph showing acroosteolysis of distal phalanges (white arrow)

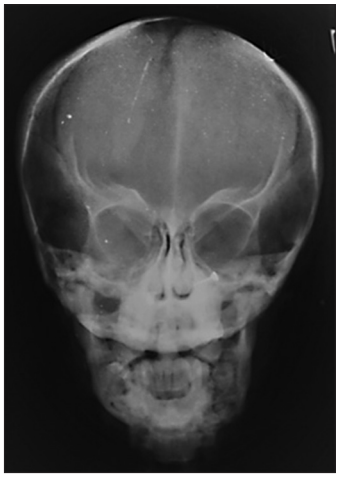

Figure 9: Posteroanterior skull view showing brachycephaly, open anterior fontanel, hypoplasia of maxilla and mandible, nonpneumatized sinuses, thick calvarium, generalized osteosclerosis of facial bones

Discussion

Pycnodysostosis is a rare autosomal recessive sclerosing bone disorder which is characterized by short stature and generalized diffuse osteosclerosis with fragile bones. Pycnodysostosis word was derived from the Greek terms "pucnos" (dense), "dys" (defective), and "ostosis" (bone condition).[2] The estimated prevalence of pycnodysostosis is 1–1.7 per million and has been reported worldwide with minimal difference affecting all races regardless of age and equal sex. The disorder is caused by a homozygous or compound heterozygous mutation in cathepsin K (CTSK) located on chromosome 1q21, which is a lysosomal cysteine protease that is highly expressed in osteoclasts. CTSK is involved in the degradation of bone matrix proteins, type I and type II collagen, osteopontin, and osteonectin at a low pH. Defective osteoclasts lead to impairment of resorption and remodeling of the bone, which inhibits normal growth and healing of the bone. Therefore, patients are prone to pathologic fractures owing to abnormally dense and brittle bones in affected individuals. Characteristically, patients

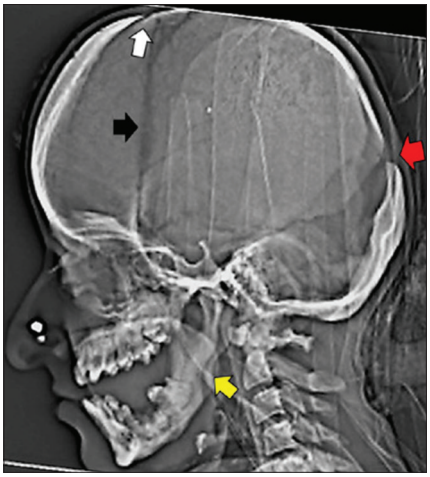

Figure 8: Lateral skull radiograph showing open fontanels (anterior = white arrow, posterior = red arrow), wide sutures (black arrow), obtuse mandibular angle (yellow arrow), anterior and posterior open bite, generalize osteosclerosis, and nonpneumatized sinuses

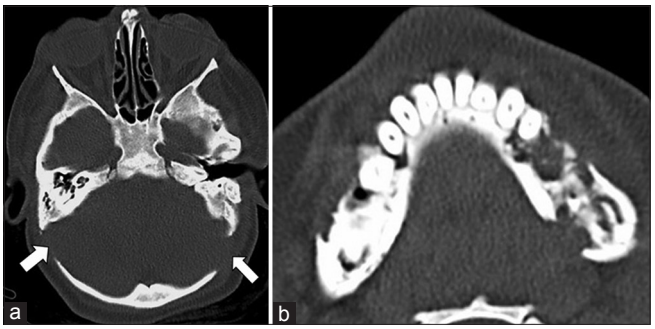

Figure 10: (a): Computed tomography scan axial view: open bilateral posterolateral fontanels (white arrows) and generalized increased bone density. (b) Computed tomography scan of mandible showing large ground glass attenuation in bilaterally body regions

show normal hematopoietic function as the medullary cavity is unaffected.[1-3]

The major maxillofacial clinical features are the presence of groove in midpalate (66.66%), midfacial hypoplasia (60.60%), mandibular hypoplasia (39.39%), dental crowding (36.36%), narrow palate (27.27%), cross bite (27.27%), and dental abnormalities in 21.21% of the reported cases. The radiographic maxillofacial characteristics are obtuse mandibular angle (94.28%), large head with bossing of frontal, parietal and occipital regions (80%), open skull sutures and fontanels (77.14%), overcrowded teeth (48.57%), aplasia or hypopeumatization of the paranasal sinuses (42.85%), Wormian bones (20%), and the supernumerary teeth (5.71%). The clinical and radiographic features of the case being reported here were similar to those reported by other authors.[1-4]

Other exceptional and rare findings of this case were sandal gap deformity which was reported by Mujawar *et al*.[2] and vertebral segmentation anomalies such as abnormal articulation of C1 to occipital condyle have been observed.[5]

This condition is usually diagnosed at a young age due to the typical clinical appearance with dwarfism and dysmorphic facies. However, in some cases (as seen in the present case), it may sometimes be diagnosed at a later age as a result of complications such as fractures and infections due to decrease bone vascularity and increase in bone density. In such patients, cognitive functioning and life expectancy are unaffected.[2]

Differential diagnosis of pycnodysostosis is osteopetrosis, cleidocranial dysplasia, idiopathic acroosteolysis, and osteogenesis imperfecta. Pycnodysostosis is a specific form of osteopetrosis and affected patients have osteosclerosis related to decreased bone resorption. In osteopetrosis, there are alterations in hematopoietic features due to the absence of the bone marrow and paralysis of cranial nerves, deafness, or pain. However, the presence of acroosteolysis together with osteosclerosis is a highly indicative feature of pycnodysostosis. Cleidocranial dysplasia may appear similar to pycnodysostosis with agenesis or aplasia of clavicles and alterations bone turnover rate; however, it is autosomal dominant inheritance and shows normal bone density. Patients with idiopathic acroosteolysis are characterized by hypotelorism, exophthalmos, and an upturned nose. However, the mandible has acute angle and normal bone density. In osteogenesis imperfecta, fractures are multiple and more severe than in pycnodysostosis and have characteristics features of choanal atresia and blue sclera.[6,7]

Pycnodysostosis is usually diagnosed based on clinical and radiographs features. A CTSK gene mutation analysis is the gold standard confirmatory test but it was not done in our case because of its high cost.[6]

Osteomyelitis is the most serious complication of pycnodysostosis; in our case patient presented with bilateral osteomyelitis of the mandible which has not been reported so far. Previously mentioned, inappropriate remodeling of bone by dysfunctional osteoclasts, while bone formation continues normally. This leads to brittle bones and decreased vascularization which will continue to worsen with age. The increased susceptibility to osteomyelitis with age can be attributed to the increased endosteum bone production, which gradually eliminates the medullary spaces in the jaws and compromises vascularization and the local immune defenses. In adults, osteomyelitis with pycnodysostosis is common but in children, it is uncommon. Obstructive sleep apnea (OSA) has been reported in young children with pycnodysostosis often with fatal outcomes. OSA is defined as the cessation of respiratory air flow for longer than 10 s more than thirty times in a 7 h period. The craniofacial abnormalities in pycnodysostosis decrease the upper airway space and increase the risk of respiratory insufficiency.[8]

The treatment for pycnodysostosis is basically supportive. To prevent oral complications, maintenance of proper oral hygiene and regular dental visits is very important. Tooth extraction and other surgeries should be done carefully to prevent postextraction osteomyelitis. One of the problems for such patients is maintenance of oral hygiene due to dental crowding. Although some authors argue that the lack of bone remodeling would impede satisfactory results, early orthodontic treatment along with planned sequenced extractions would be more recommendable.[2] Atraumatic tooth extraction with proper asepsis should be carrying out in such patients to prevent the risk of fracture, especially in the mandible. In addition, the greater bone density increases the probability. The prognosis of pycnodysostosis is good and no other serious systemic complications have been reported.[2,9]

Conclusion

Pycnodysostosis is a rare syndrome that is diagnosed based on its clinical and radiographic features. Pycnodysostosis includes group of disorders that requires a multispecialty approach. The importance of early recognition of clinical features in the diagnosis allows correct treatment planning and reduces the chance of complications in future, thus ensuring a better quality of life to the patient. Underdiagnosis or misdiagnosis of patients is a major concern; therefore, awareness about pycnodysostosis is a must.

Declaration of patient consent

The authors certify that they have obtained all appropriate 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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Conflicts of interest

There are no conflicts of interest.

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