CASE REPORT

Pyknodysostosis: Report of a Rare Case and its Dental Management

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Abstract

Aim: This is a case report of a 16-year-old girl visiting MR Ambedkar Dental College and Hospital (Department of Pedodontics and Preventive Dentistry) for dental treatment.

Background: Osteopetrosis acroosteolytica or Toulouse-Lautrec syndrome or pyknodysostosis is a rare autosomal recessive bone dysplasia, characterized by osteosclerosis, and short stature. Montanari described a patient with an unusual variation of achondroplasia, which in retrospect was the first case of pyknodysostosis to be reported.[1]

Case description: A 16-year-old girl reported to the Department of Pediatric and Preventive Dentistry with a chief complaint of pain in the lower left back region of the jaw since past 2 weeks.

Conclusion: Pyknodysostosis is a rare condition that is diagnosed basically on its clinical and radiographic features.

Clinical significance: It is important to recognize these features so that correct diagnosis can be made. This allows the treatment and prevention of future complications and ensures a better quality of life to the patient.

Keywords: Pyknodysostosis, Rare, Short stature, Syndrome.

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Background

Osteopetrosis acroosteolytica or Toulouse-Lautrec syndrome or pyknodysostosis is a rare autosomal recessive bone dysplasia, characterized by osteosclerosis and short stature. In 1923, Montanari described a patient with an unusual variation of achondroplasia, which in retrospect was the first case of pyknodysostosis to be reported.[1] The term "pycnodysostosis" was first coined and described by Maroteux and Lamy in the year 1962, (Pycno—thick/dense, dysostosis—defective bone)[2] incidence of this osteosclerotic disorder is estimated to be 1.7 per million births.[3] The main features of patients with pyknodysostosis are short stature, acroosteolysis of distal phalanges, deformity of the skull, dysplasia of the clavicles, narrow and/or grooved high arched palate, midfacial hypoplasia, absence or hypopneumatization of the paranasal sinuses, generalized osteosclerosis and fragility of bone, dysplasia of terminal phalanges, elongation of coronoid apophyses, and mandibular condyles.[4,5] 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.[6,7]

This is a case report of a 16-year-old girl visiting the Department of Pedodontics for dental treatment.

Case Description

A 16-year-old girl reported to the Department of Pediatric and Preventive Dentistry with a chief complaint of pain in the lower left back region of the jaw since past 2 weeks. On general physical examination, she demonstrated short stature, frontal and parietal bossing, depressed nasal bridge, beaked nose, hypoplastic midface, palpable anterior and posterior fontanels. Stubby hands with wrinkled skin over the fingertips and crested nails. Patient gave a history of leg fracture thrice due to minor trauma. History of consanguinity was positive with parents. Presence of similar clinical features in maternal aunt (mother's sister) was also reported (Fig. 1).

Intraoral examination revealed presence of complete permanent dentition in both the arches, constriction of maxilla with grooved palate causing crowding, mandibular arch showed the presence of irregularly placed teeth. On clinical and radiographic examination over-retained 55 was observed, 37 showed dental caries involving pulp, 46 showed dental caries involving pulp with gross destruction of the clinical crown.

Hand wrist radiograph showed hypoplasia/absence of terminal tuft of the thumb and index finger, also known as acroosteolysis. Increased bone density was also noted. X-ray of the skull (water's view) showed non-pneumatized frontal sinuses, hypoplastic maxillary sinuses bilaterally, and hypoplastic mandible with obtuse

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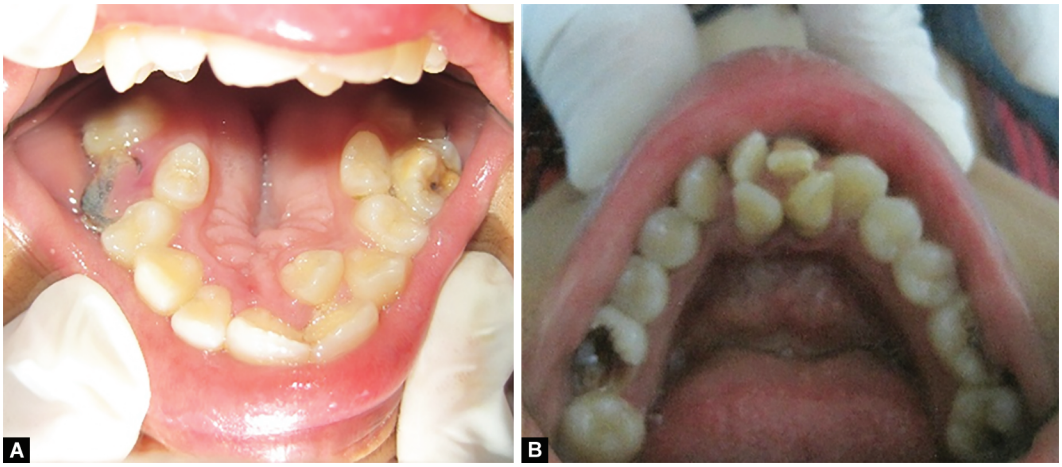


Figs 1A and B: Intraoral images: (A) Upper arch showing grooved palate; (B) Lower arch showing crowding with lower anteriors

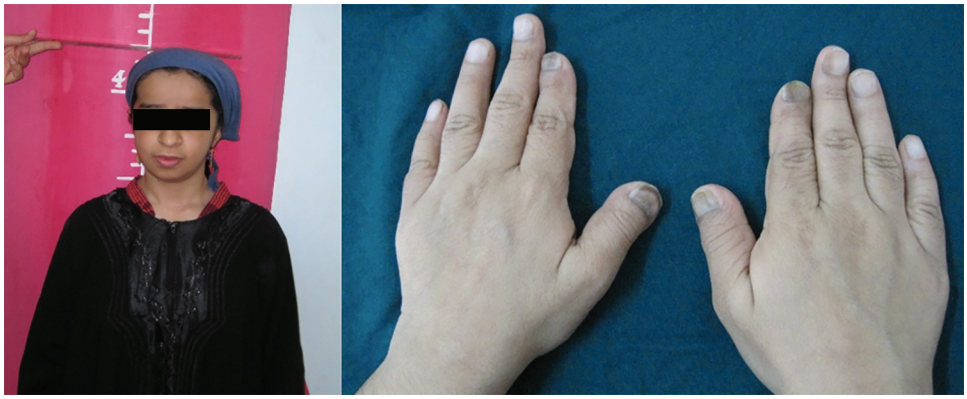


Fig. 2: Clinical features showing short stature and wrinkled skin over fingers

mandibular angle. Orthopantomogram and lateral cephalogram showed elongation of condyle and hypoplasia of coronoid process. Aforementioned clinical and radiographic findings were suggestive of pyknodysostosis (Fig. 2).

Dental Management

Endodontic therapy was planned for 37 extractions w.r.t. 55 and 46 under local anesthesia. After obtaining the physician's consent and complete blood investigations, dental treatment was started. As the patient presented with restricted mouth opening performing root canal treatment on 37 was difficult. The tooth also presented, with root dilacerations and aberrant canals, one mesial canal placed exactly in the center of the access cavity and two distal canals. Extra precautions were taken to avoid fracture of the mandible, and any untoward results, as osteomyelitis is a common complication after extraction in cases with pyknodysostosis. While performing extraction with 46 to avoid any kind of trauma to the already brittle mandible, two roots w.r.t. 46 were split with the help of straight handpiece at the furcation area, and both the root pieces were removed separately. Extraction was carried out under complete antibiotic coverage and calcium supplements. No complications were experienced during or posttreatment as the wound healed uneventfully (Fig. 3).

Discussion

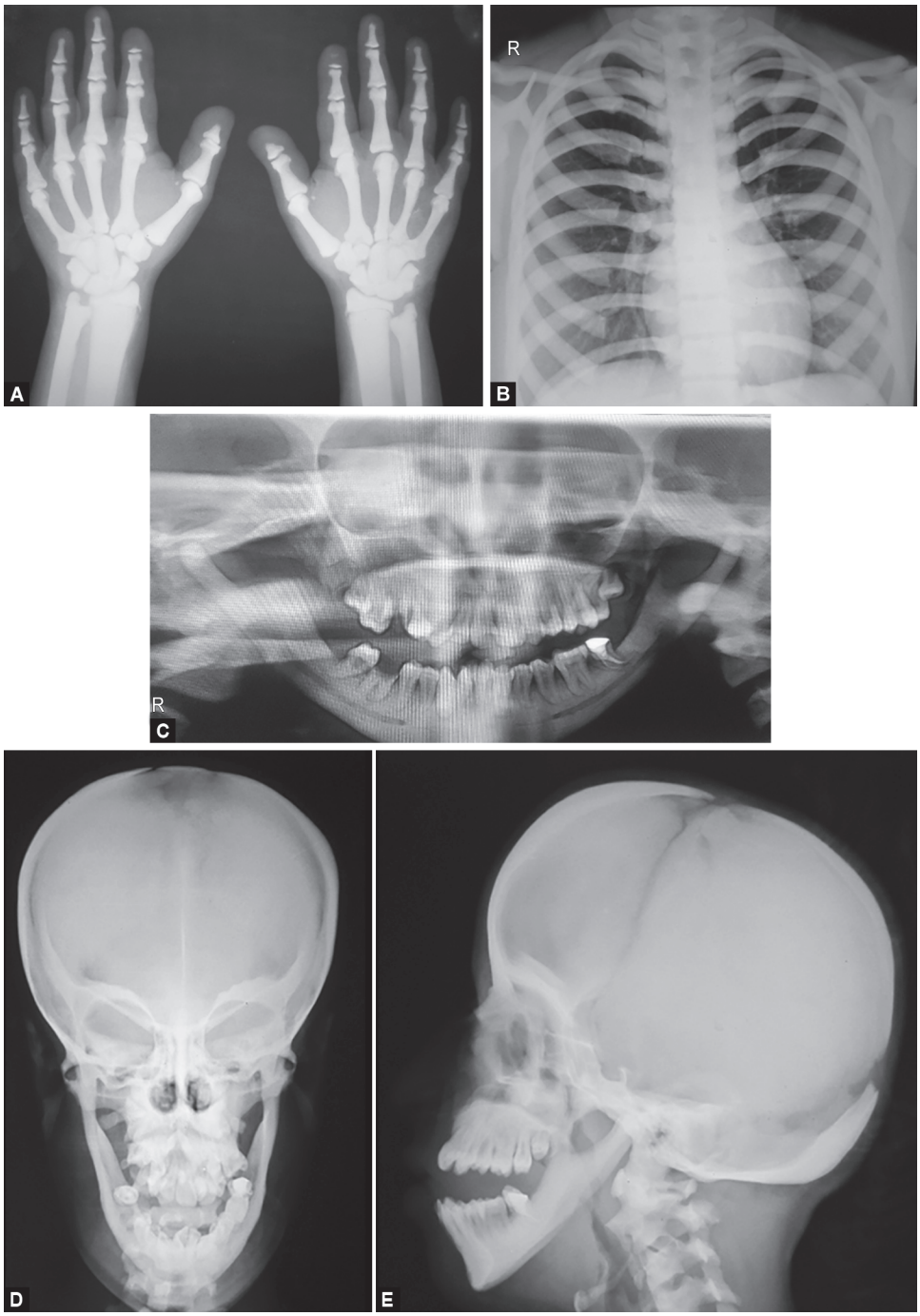
Pyknodysostosis is a lysosomal storage disease of the bone that occurs due to a genetic defect on chromosome 1q21, which codes the enzyme cathepsin K (CTSK).[8] This protease is responsible for degrading type I collagen that constitutes 95% of the organic bone matrix. This genetic disorder is usually diagnosed at an early age. The affected bones are abnormally dense and brittle as a result of insufficient resorption.[6,9,10] The differential diagnosis of pyknodysostosis includes osteopetrosis, cleidocranial dysplasia, and idiopathic acroosteolysis.

In osteopetrosis, the bone marrow may be absent; therefore, hematopoietic alterations may appear frequently. Signs of compression of the cranial nerves exist, such as facial paralysis, deafness, or pain. Cleidocranial dysplasia may seem like pyknodysostosis in the presentation of clavicular agenesis or aplasia, as well as alterations of the skeletal bone membranes; however, bone density is not increased. In idiopathic acroosteolysis, the appearance of the patients is typical, with hypotelorism, exophthalmos, and an upturned nose. The angle of the mandible is acute, and increased bone density is not present.[11]

However, diagnosis of pyknodysostosis is primarily based on the clinical features and radiographs, a CTSK gene mutation analysis is often confirmatory.[12]

This anomaly consists of 12 different mutations.[13] When not diagnosed in infancy, fractures resulting from trauma usually lead to the diagnosis of this disease.[14,15]

The oral and maxillofacial manifestations of this disease are very clear. The exfoliation of deciduous teeth is usually altered, as well as the eruption of the permanent dentition.[16] Mandibular fractures have been described in adults following extractions.[17]



Figs 3A to E: Extraoral and intraoral radiograph: (A) Hand wrist radiograph; (B) Chest radiograph; (C) Orthopantomogram; (D) Frontal cephalogram; (E) Lateral cephalogram

There may be dental abnormalities, with hypoplasia of the enamel, obliterated pulp chambers, and hypercementosis. Protrusion of the incisors with anterior open bite may be found, and dental crowding associated with extensive caries and periodontitis is frequent. These conditions cause the premature loss of dentition that may already be complete by the fourth decade of life[16] greater bone density increases, the probability of developing post-extraction osteomyelitis.[15]

Schilling in a study determined a volumetric bone density of 686 mg/cm in patients with pyknodysostosis vs 290 mg/cm in the control group. Norholt affirmed that due to the maxillary hypoplasia, these patients often present a class III dentition. These authors defend the orthognathic correction by osteogenic distraction. Occasionally exophthalmos and blue sclera coexist.[18] Soliman et al. suggest the increased bone volume of the sella turcica compresses the pituitary gland, causes its hypoplasia and deficient production of the growth hormone.

Exceptionally, hepatosplenomegaly and hematologic alterations have been observed.[15] Another important alteration that usually affects these patients are respiratory problems. These

conditions are due, above all, to a very long soft palate that may even come into contact with the base of the tongue.[11]

Radiological findings may show some degree of widening of the distal femur. The skull shows open anterior fontanelle and sutures with small facial bones, non-pneumatized paranasal sinuses, and flattened mandibular angle. Terminal phalanges in the hand are partially or totally aplastic with loss of ungual tufts. The acromial ends of the clavicles may be aplastic. Other abnormalities include failure of complete segmentation of the atlas, axis, and the lower lumbar spine, coxa valga, and abnormal radioulnar articulation. Histologically, the appearance is similar to that of osteopetrosis, but the medullary canals are present, and microscopic evidence of attenuated haversian canal system is seen. Life expectancy for a pyknodysostosis patient is normal.

There is no specific treatment as of date for this disorder, and treatment is supportive. Since bone fractures are a primary threat to those affected by pyknodysostosis, it is important that care is taken to prevent or minimize tendencies for a fracture to occur. Such precautions include careful handling of an affected child, along with caution to avoid any inadvertent injuries while performing day-to-day activities. Dental hygiene and regular dental checkups are especially helpful for affected individuals due to various dental anomalies.[2]

Conclusion

Pyknodysostosis is a rare condition that is diagnosed basically on its clinical and radiographic features. It is important to recognize these features so that the correct diagnosis can be made. This allows treatment and prevention of future complications and ensures a better quality of life to the patient.

Clinical Significance

It is important to recognize these features so that the correct diagnosis can be made. This allows treatment and prevention of future complications and ensures a better quality of life to the patient.

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