

Case Report

Pycnodysostosis: A bone dysplasia with unusual oral manifestation

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Abstract

Pycnodysostosis, a sclerosing bone dysplasia, is a rare autosomal recessive disorder with an estimated prevalence rate of one in one million. Patients with pycnodysostosis usually have normal intelligence, sexual development and life span. This condition is characterized by increased bone density and fragility along with oral manifestations like malposition teeth, hypoplastic maxilla, receded chin and delayed eruption of permanent teeth with discharging sinuses in the jaws because of poor blood supply. This is one such rare case report of a 47-year-old patient presenting with a complaint of fractured jaw and reviewing the clinical and radiographic characteristics of pycnodysostosis.

Key words: Bone dysplasia, hypoplastic maxilla, osteosclerosis, pycnodysostosis

INTRODUCTION

Pycnodysostosis was first reported in 1923 by Montanari, and he called it as atypical achondroplasia. Later, Maroteaux and Lamy described it in 1962.^[1] The famous French painter Toulouse Lautrec suffered from this disease.^[2] The main characteristics are short stature, cranial dysplasia, increased bone density and fragility. Other clinical features include open cranial sutures, hypoplastic paranasal sinuses, dysplastic lateral clavicle, shortened terminal phalanges, proptosis, blue sclera and frontal or occipital bossing.^[3]

Oral manifestations include obtuse gonial angle, grooved palate, anterior cross-bite, malpositioned teeth associated with increased incidence of dental caries and periodontitis, hypoplastic maxilla, receded chin, delayed eruption of permanent teeth, delayed exfoliation of deciduous teeth and hypoplasia of root-obiterated pulp spaces.^[4]

CASE REPORT

A 47-year-old man reported to the department with a complaint of deformed lower jaw for the past

10 years. History revealed that the patient had undergone extraction of his teeth that was uneventful, following which there was fracture of jaw at the extraction site. Subsequently, there was frequent exfoliation of teeth with fracture at different sites in the lower jaw. The patient was asymptomatic and therefore negligent about it. He reported to the department for management of his deficient jaw. His medical history revealed that he had multiple fractures of the upper and lower limbs and a history of diabetes mellitus and hypertension. Family history revealed that there was consanguinity in his parents. General examination revealed that the patient's height was 127 cm and weight was around 49 kg, with proportionate dwarfism [Figure 1]. The hand and feet had short digits with overlying cutaneous wrinkles that tapered off with large overriding nails [Figure 2].

On extraoral examination, there was facial dysmorphism with prominent forehead (frontal bossing), proptosis, beaked nose, deep nasolabial skin folds, micrognathia and obtuse mandibular angle on the right side [Figure 3]. Mouth opening was restricted, and was around 25 mm.

Intraoral examination revealed multiple clinically missing teeth, chronic periodontitis, narrow and grooved palate, no features of enamel hypoplasia, malposed teeth [Figure 4] and evidence of sequestrum in relation to the tooth 46 [Figure 5]. Based on the history and clinical presentation, a provisional diagnosis

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of a bone dysplasia, probably pycnodysostosis, was made and differential diagnosis of cleidocranial dysplasia and osteopetrosis was included.

Laboratory findings were within normal limits, including hemoglobin conc., differential count calcium, phosphate, alkaline and acid phosphatase

level. Computed tomography of the bone window of the skull showed open sutures and fontanelles with nonaerated paranasal sinuses, flattening of the mandibular angle on the right side with evidence of fracture and loss of bone architecture on the left side involving the ramus of the body of the mandible and hypoplastic maxilla [Figures 6–8].



Figure 1: A 47-year-old man, 127 cm in height



Figure 2: Shortened digits and cutaneous wrinkles



Figure 3: Mild proptosis and facial dysmorphism



Figure 4: Grooved palate and clinically missing maxillary anteriors



Figure 5: Sequestrum in relation to tooth 46



Figure 6: 3D-reconstructed computed tomography showing open sutures and fontanelles

Orthopantomograph revealed generalized bone loss, multiple missing teeth and obtuse gonial angle with loss of bone structure on the left side of the mandible involving the body and ramus [Figure 9]. Lateral skull revealed open fontanelles with nonaerated paranasal sinus [Figure 10].

The patient was surgically managed for osteomyelitis by removal of the sequestrum and curettage, and further mandibular reconstruction was performed.

DISCUSSION

Pycnodysostosis is an autosomal recessive disorder with a slight male predilection (1.6:1).^[5] The patient presents with characteristic facies, dwarfism, beaked nose, prominent head and generalized increase in the density of bones not sufficient to obliterate medullary canals or cranial orifices.^[1] Frequent fractures due to trauma can aid in diagnosing this condition. In our case, the patient was negligent about the condition and reported to us with a fractured jaw. History also revealed that the patient had previous repeated incidents of fractured jaws.

Intraoral clinical presentation included altered pattern of exfoliation of deciduous teeth and eruption of the permanent dentition. The disease is diagnosed at an early age, wherein the main reasons for consultation are generally short stature and open anterior fontanelles.^[6] In later stages, consultation is usually for fracture resulting from slight or moderate trauma, given the severe bone fragility. Symptoms include 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.^[7,8] In our case, multiple clinically missing teeth, chronic periodontitis, narrow and grooved palate, features of enamel hypoplasia and malposed teeth were observed. These conditions cause the premature loss of dentition that may already be complete by the fourth decade of life, similar to our patients.



Figure 7: 3D-reconstructed computed tomography showing fractured body of the mandible and obtuse gonial angle



Figure 8: 3D-reconstructed computed tomography showing fractured body of the mandible

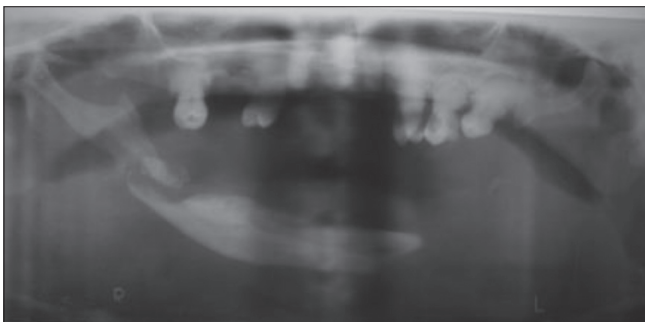


Figure 9: OPG showing generalized bone loss, multiple missing teeth, and obtuse gonial angle

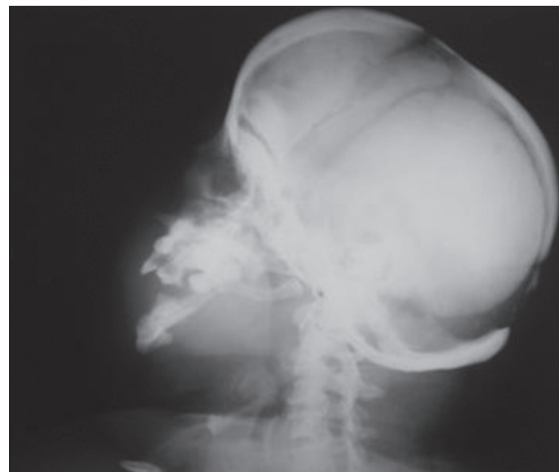


Figure 10: Lateral skull showing open fontanelles with nonaerated paranasal sinus and fractured body of the mandible

Greater bone density increases the probability of developing postextraction osteomyelitis.^[1] Our patient showed evidence of sequestrum in relation to the tooth 46 as a result of osteomyelitis. Orofacial infections are commonly encountered by the dentist and there are wide ranges of modalities that can be implemented in managing them.

A constant characteristic is the short stature of the patients. This is caused by the increased bone volume of the sella turcica that, on compressing the pituitary gland, causes its hypoplasia and a deficient production of the growth hormone. Exceptionally, hepatosplenomegaly and hematologic alterations have been observed.^[7] Our patient's height was 127 cm and weight was around 49 kg, with proportionate dwarfism. The hand and feet had short digits with overlying cutaneous wrinkles that tapered off with large overriding nails. One of the important alterations that usually affect these patients is respiratory problems.^[9]

Diagnosis of pycnodysostosis is based on the clinical presentation, and medical treatment for the condition is symptomatic. The differential diagnosis of pycnodysostosis includes osteopetrosis, acroosteolysis, mandibular acral dysplasia and cleidocranial dysplasia. Unlike osteoporosis, hepatosplenomegaly and anemia are rare in pycnodysostosis due to the presence of active medullary hematopoiesis. Difference between pycnodysostosis and cleidocranial dysplasia is that dense and brittle bones are found in pycnodysostosis but not in cleidocranial dysplasia.^[10] New treatment modalities like gene therapy and bone marrow transplant can be expected to be the mainstay in the future, now that the abnormal expression of cathepsin K and the gene defect has been located.^[11]

CONCLUSION

Now-a-days, symptomatic treatment is provided for patients with pycnodysostosis, with the main intention of prevention of fractures. Early diagnosis is mandatory to avoid complications as it is difficult to manage. As

pycnodysostosis is associated with inappropriate bone remodeling, it can pose a challenge for a dental health care professional to provide treatment as there can be serious complications, such as osteomyelitis arising as a result of dental infections. Tooth extractions in these patients demand certain special care, such as carrying out the surgery atraumatically with proper asepsis. Oral hygiene practices and frequent visits to the dentist can prevent serious complications.

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