ABSTRACT

Pycnodysostosis is a rare, genetic bone disorder characterized by generalized bone sclerosis, short stature and increase in mandibular angle. Here, we report a case of pycnodysostosis presented with classical intraoral features.

Keywords: Pycnodysostosis, Bone sclerosis, Cathepsin K.


CASE REPORT

A 11-year-old boy reported to outpatient department of Karpaga Vinayaga Institute of Dental Sciences with chief complaint of multiple decayed teeth. History revealed that the patient had undergone extraction of a decayed tooth 2 months back. On general examination, the patient appeared to be short stature, large head with frontal bossing (Figs 1 and 2). Patient gave history of repeated fracture in right leg (4 times) from the age of 3. The last fracture was 8 months back in the right femur. Aplastic terminal phalanges were also observed in the upper and lower limbs (Figs 3 and 4). On intraoral examination, high arched palate, crowding of teeth, palatally erupted 12, 25, buccally erupted 24, multiple teeth. Other intraoral features include high arched palate, enamel hypoplasia, obliterated pulp chambers.

INTRODUCTION

The term pycnodysostosis meaning ‘dense defective bone’, was first described in 1962 by Maroteaux and Lamy. The syndrome is inherited as autosomal recessive trait occurring due to mutated gene located on chromosome 1q21. Normally, the gene is responsible for the function of an enzyme lysosomal cystine protease, cathepsin K, which in turn helps in the degradation of collagen fiber type 1, a major component of bone matrix. The mutation of the gene lowers the bone remodeling capacity resulting in sclerosis of bone.

Pycnodysostosis occurs in both sexes in varying age groups. The principle features of the syndrome like short stature, open fontanels, frontal and parietal bossing helps in the early diagnosis of the syndrome. Fracture is the common presenting symptom in the adults. Large head with frontal and parietal bone bossing, open sutures and fontanels, straightened mandible (obtuse angle of the mandible), dysplastic clavicles, total or partial aplasia of terminal phalanges are the other classical features of the syndrome to be mentioned. The bone becomes sclerotic with high fracture susceptibility.

Radiographically, skull bone appears thickened with open fontanels which look like ‘lakes of bones’. Hypoplasias of facial bones, non-pneumatization of paranasal sinuses, partial or complete aplasia of the distal phalanges are also seen. Fracture occurs most commonly in lower limb, and the fracture line is characteristically transverse and tends to occur in the middle of the affected bone.

Double row of teeth or crowding may be the prominent intraoral feature of the disease. Crowding occurs due to failure in the exfoliation of deciduous dentition with premature or delayed eruption of permanent dentition. Poor oral hygiene, multiple caries are inevitable due to crowding.
decayed teeth involving 65, 85, 16, 24 with fractured restoration in 75 were observed (Fig. 5). Orthopantomogram showed straightening of the mandibular angle with obliterated pulp chambers in anterior teeth (Fig. 6). Posteroanterior view of lower limbs showed sclerotic tibia and fibula (Fig. 7). With this classical presentation, we made a diagnosis of pycnodysostosis. Necessary restoration and extraction was carried with caution in department of pedodontics and patient was asked to report for regular periodic visit.

**DISCUSSION**

Pycnodysostosis is a benign, relatively uncommon autosomal recessive disorder characterized by osteoclastic dysfunction and hence it is a disease of low bone remodeling. Classical features of the syndrome like short stature, repeated fracture helps in the diagnosis.4

Crowding, improper oral hygiene can be dealt with planned extraction of retained deciduous teeth. Extraction of tooth for these patients should be carried out with caution to avoid fracture of jaw bone or postextraction osteomyelitis. Orthodontic treatment for crowding in these patients is controversial, because low remodeling capacity of the bone puts these patients at a high risk for orthodontic treatment failure.2-4

Pycnodysostosis should be distinguished from other two similar bone disorder, such as osteopetrosis and cleidocranial dysplasia. Presence of dense bone in osteopetrosis is similar to pycnodysostosis but unlike pycnodysostosis distinction between cortex and marrow is difficult in osteopetrosis. Due to the obliteration of marrow in osteopetrosis, anemia and hepatosplenomegaly would dominate the clinical picture.1,3,6,7 In cleidocranial dysplasia, partial or total aplasia of the one or both the clavicle is present with no predisposition to fractures.3,6,7

In our case, history of repeated fracture, short stature, frontal bossing, aplastic terminal phalanges, sclerotic bone, absence of anemia and hepatosplenomegaly along with intraoral features, like retained deciduous teeth, crowding with multiple decayed teeth, radiographic features of straightened mandibular angle, sclerotic long bones drove us toward the diagnosis of pycnodysostosis. We carried out restoration of carious teeth and planned atraumatic extraction for unpromising and retained teeth. Importance of proper oral hygiene was educated to them. Patient was kept under follow-up.

**CONCLUSION**

History, complete generalized examination, recording of extra and intraoral features may help in the diagnosis of underlying core diseases, according to which the treatment plan or procedures may be tailored. Dental treatments for those with pycnodysostosis should be framed in such a way that post procedural complication should be avoided or minimized. Educating the patients will help them to maintain good oral hygiene.
Pycnodysostosis: A Case Report of Rare Entity

Fig. 6: Orthopantomogram

Fig. 7: Radiograph showing sclerosis of bone

REFERENCES


ABOUT THE AUTHORS

C Nirupama
Professor, Department of Orthodontics, Karpaga Vinayaga Institute of Dental Sciences, Kanchipuram, Tamil Nadu, India

D Sarasakavitha
Reader, Department of Pedodontics, Karpaga Vinayaga Institute of Dental Sciences, Kanchipuram, Tamil Nadu, India

Sivakumar Palanivelu (Corresponding Author)
Reader, Department of Oral Pathology, Karpaga Vinayaga Institute of Dental Sciences, Kanchipuram, Tamil Nadu, India, Phone: 04427598484, e-mail: sivakumarpalanivelu@yahoo.com

Bala Guhan
Reader, Department of Oral and Maxillofacial Surgery, Karpaga Vinayaga Institute of Dental Sciences, Kanchipuram, Tamil Nadu, India