Update of dental and maxillofacial alterations in patients with pycnodysostosis

Atualização das alterações dentárias e dos ossos gnáticos em pacientes com picnodisostose

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ABSTRACT

Pycnodysostosis is a rare, autosomal recessive genetic condition, which causes a decrease in bone remodeling, resulting in different clinical and radiographic manifestations. This case series aims to describe two clinical cases diagnosed at the Department of Oral and Maxillofacial Surgery and Traumatology of a University on the Northeast of Brazil. There are two complex cases involving osteomyelitis and dental and bone alterations of the jaws. It is concluded that the knowledge of oral and maxillofacial characteristics of this syndrome are required to plan appropriate treatment for patient in order to avoid complications of dental treatments due to inadequate bone remodeling.

Key words: pycnodysostosis; dentistry; bone disease.

RESUMO

A picnodisostose é uma condição genética rara, autossômica recessiva, que ocasiona diminuição da remodelação óssea, resultando em várias manifestações clínicas e radiográficas. Este estudo pretende descrever dois casos clínicos diagnosticados no Serviço de Cirurgia e Traumatologia Bucomaxilofacial de uma universidade no nordeste do Brasil. São dois casos complexos que envolvem osteomielite e alterações dentárias e ósseas dos maxilares. É necessário o conhecimento das características orais e maxilofaciais dessa síndrome para planejar o tratamento adequado para o paciente, a fim de se evitar complicações de tratamentos dentários em virtude do remodelamento ósseo inadequado.

Unitermos: picnodisostose; odontologia; doenças ósseas.

RESUMEN

La picnodisostosis es una enfermedad genética rara autossómica, recesiva, con disminución de la remodelación ósea, que ocasiona varias manifestaciones clínicas y radiográficas. Este estudio describe dos casos clínicos diagnosticados en el servicio de cirugía y traumatología bucomaxilofacial de una universidad en el nordeste de Brasil. Los casos son complejos y envuelven osteomielitis, alteraciones dentales y óseas de las mandíbulas. Es necesario conocer las características orales y maxilofaciales de este síndrome para planear el tratamiento adecuado al paciente, con la intención de evitar complicaciones de tratamientos dentales debido a la remodelación ósea inadecuada.

Palabras clave: picnodisostosis; odontología; enfermedades óseas.

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INTRODUCTION

Pycnodysostosis was described in 1962 by Maroteaux & Lamy. Also called Toulouse-Lautrec syndrome, it is a rare, autosomal recessive inherited disorder caused by mutation in the gene encoding the cathepsin K enzyme. It can be related to inbreeding between the parents in 30% of cases, with no gender predilection; causes lower bone remodeling, which results in several clinical and radiographic manifestations.

Clinical manifestations of this disease include short stature, generalized sclerotic bone, stunted extremities, and adult height ranging from 134 to 152 cm. The most frequently oral manifestations observed are maxillary atresia, increased angle of the mandible, enamel hypoplasia, increased risk of pathological fractures during dental surgery and tooth extraction due to bone fragility. However, despite the fragility, bone healing is normal. The onset of osteomyelitis related to a tooth with large carious lesion is frequent. These findings reinforce the importance of diagnosing the abnormality for appropriate dental planning of patients, taking necessary precautions.

Radiographic findings include osteosclerosis, wide open cranial sutures and fontanelles, absence of pneumatization of the facial sinuses, and increased radiopacity of all bones (especially the vertebral column), long bones and base of the skull. The disease is often diagnosed at an early stage in patients with short stature and open anterior fontanelle. When identification is late, it is due to bone fracture conditions.

One of the major aspects that require the oral surgeon attention is the need to carefully plan surgical interventions due to defective bone turnover to minimize the risk of osteomyelitis or osteonecrosis.

Our study proposes to report two distinct cases of pycnodysostosis syndrome, associating clinical and radiographic characteristics and elucidating clinical management to avoid complications resulting from poor bone remodeling. In addition, a Medline mapping survey of existing cases was conducted from 1962 to 2018.

CASE REPORT

Case 1

A male patient with pycnodysostosis syndrome sought the Oral and Maxillofacial Surgery and Traumatology service of the Universidade Federal do Rio Grande do Norte (UFRN) with short stature, large hands with short spoon-shaped phalanges, dystrophic nails and history of repeated fractures in the left tibia, associated with low-impact trauma (Figure 1A-B). Intraoral examination revealed ogival palatal, maxillary atresia, rotated teeth and open mandibular angle (Figure 1C-E).

FIGURE 1 – Estroanal and intraoral clinical features of patient case 1
A) patient with short stature; B) shortened phalanges; C) class III malocclusion with overjet and bilateral posterior open bite; D) presence of non-erupted tooth, agenesis of the second lower premolar and obtuse angle of the mandible.

Case 2

A female patient with pycnodysostosis syndrome attended at the Oral and Maxillofacial Surgery and Traumatology service of the UFRN presenting short stature, large hands with short spoon-shaped phalanges (Figure 2A-B), rotated teeth, dental enamel hypoplasia (Figure 2C) and bilateral osteomyelitis of the body of the mandible (Figure 3). She reported painful and pulsating symptoms. The partially dentate patient presented process of local infectious with active drainage and bilateral submandibular fistula.

FIGURE 2 – Estroanal and intraoral clinical features of patient case 2
A) patient with short stature and stunted extremities; B) shortened phalanges; C) biprotrusion, bilateral posterior open bite, enamel hypoplasia, dental agenesis and rotated premolar.
(Figure 3A). She underwent general anesthesia in hospital setting. Submandibular extraoral approach with bilateral retromandibular extension was performed; surgical debridement of areas of bone sequestration was performed until the bleeding bone was found for the placement of reconstruction plates (2.4 mm) (Figure 3B), in addition to coronectomy of the molars in left mandibular body (Figure 3C). Clindamycin was administered one week before the surgery and six weeks after surgery. Two years after surgery the patient is under follow-up showing no signs of recurrence of the mandibular infection (Figure 3D-E).

**DISCUSSION**

Papers on pycnodysostosis published from 1976 to November 2018 in the English language were analyzed, and the cases mapped according to the continent of occurrence of the disease and the geographical region of Brazil. Medline data (PubMed) were searched using the keywords: pycnodysostosis; pycnodysostosis and oral; pycnodysostosis and oral features; pycnodysostosis and dentofacial characteristics. A total of 289 articles were found of which 73 articles excluded. The remaining cases (216) were selected according to the inclusion criteria and are shown in [Figure 4](#).

Oral and maxillofacial findings of patients with pycnodysostosis were not reported in all cases, and only 22 articles detailed addressed the oral characteristics of the syndrome. It is common to observe dental findings in pycnodysostosis, such as hypercementosis – reported in two articles(5, 6); class III malocclusion – reported in three articles(7-9); overjet(7, 10, 11); bilateral open lateral bite(7, 10); anterior open bite(12); posterior crossbite(4, 7, 12); delayed eruption(4, 11-13); dental agenesis(9); enamel hypoplasia(8, 10); osteomyelitis(5, 8, 14-16); osteosarcoma(17); narrow and grooved palate(4, 8-12, 18); poor positioning of teeth or dental crowding(4, 6, 8, 9, 11, 12, 18, 19); ectopic teeth(4, 13). Regarding the radiographic findings, they are: mandibular hypoplasia(3, 4, 8, 10, 12, 18); increased bone density(5, 6); radiodensity around the roots(5, 6, 20); tooth retention(5); no boundary between trabecular and cortical bone(6); maxillary retrusion with hyperdivergent mandibular growth(18); alveolar crest hypoplasia(8); narrow pulp chamber(6); root canal stenosis or atresia(6); root canal calcifications(6); obtuse angle of the mandible(5, 8, 11, 18, 19); pathological fractures(5, 6, 15, 19); poor root development of the lower first permanent molars(6); and laceration of the lateral incisor roots(6).

Osteomyelitis is the most severe oral complication of pycnodysostosis due to previous dental extractions or infections. The authors speculate that inappropriate bone remodeling with osteoclast dysfunction does not prevent bone deposition, however causes fragile bone formation and reduced vascularization, worsening with age. The greater susceptibility to osteomyelitis due to improper bone remodeling may contribute to the high prevalence of oral osteomyelitis in patients with pycnodysostosis.
to the decrease in the number of osteons and the large obliteration of Havers canals impairs blood flow through the vessels, increasing the infection. Our patient underwent previous dental extraction and presented history of pericoronitis on the contralateral side.

Another common finding is enamel hypoplasia, which plays an important role in oral health. Areas of enamel demineralization are likely to cavities, so proper care must be taken to prevent them. Good oral hygiene, clinical evaluation and periodic radiographic examinations are recommended.

Hypercementosis occurs due to the increase of cementocytes and cementoblasts between the cementum layers, which suggests a hypocalcified cementum feature demineralization. This may be caused by improper collagen degradation. Cement consists 90% of I, III and ground substance.

The bite pattern of patients is predominantly class III due to the atypical horizontal insertion of the mandibular condyle into the glenoid fossa; such a pattern tends to project the mandible forward.

Malocclusions are frequent in patients with pycnodysostosis, caused mainly by the palate shape (deep and grooved). Low bone remodeling, caused by the absence of cathepsin K, hampers orthodontic strategies; bone resorption and neoformation are necessary events for successful orthodontic treatment and no protocol is available. Orthodontic treatments consist of maxillary expansion and orthognathic surgery, recommended to correct malocclusion.

In the reported cases, we observed short stature with stunted extremities, shortened phalanges, biprotrusion, bilateral open bite and dental crowding. In addition to these characteristics, case 2 presented amelogenesis imperfecta and osteomyelitis secondary to dental extraction. Several bone diseases should be considered in the differential diagnosis of pycnodysostosis, especially cleidocranial dysostosis, acroosteolysis, osteogenesis imperfecta and osteopetrosis. In cleidocranial dysostosis, the clavicle is often affected; in pycnodysostosis, this bone is not affected. The absence of the mandibular angle is fundamental to distinguish pycnodysostosis from acroosteolysis.

It is concluded that knowledge of the oral and maxillofacial characteristics of pycnodysostosis syndrome is necessary to plan the appropriate treatment for the patient in order to avoid complications resulting from previous tooth extractions, caries, periodontal disease and orthodontic appliance installation, since bone repair of these patients is hampered by inadequate bone remodeling.

REFERENCES


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