Chapter 44

Buccomaxillofacial clinical approach to the patient with pycnodysostosis: case report and literature review

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ABSTRACT
Pycnodysostosis is a rare sclerosing skeletal dysplasia, hereditary and of autosomal recessive character, which is characterized by the presence of dense bones and thick cortical area with high fragility in affected individuals. To report a clinical case of pycnodysostosis, associating its clinical and radiographic features and discussing its causes, physiological repercussions and considerations related to orthodontic management and oral and maxillofacial surgical approaches. An 8-year-old female patient, phaeoderma, with pycnodysostosis, short stature, broad hands, dystrophic nails, and shortened phalanges, hypoplastic middle facial third and mandibular micrognathia. Intraoral examination revealed the presence of mixed dentition, crowding in the mandible, prolonged retention of deciduous teeth, multiple dental gyrations, extensive dental caries, absence of dental unit 11, ogival palate, anterior and posterior crossbite on the left side. Knowledge of the clinical manifestations in the oral and maxillofacial region related to pycnodysostosis is essential to identify and prepare an appropriate treatment plan for the development of these changes, which with time become more evident and difficult to manage.

Keywords: Pycnodysostosis, Dentistry, Cathepsin K, Maxillofacial abnormalities

1 INTRODUCTION

Pycnodysostosis, also known as Toulouse-Lautrec syndrome, is a rare sclerosing skeletal dysplasia that is hereditary in order and autosomal recessive in character (Elyajouri et al, 2018; Mandal et al, 2016; Turan et al, 2014).

The diagnosis of this condition is made from the patient's history, clinical and radiographic findings (Khirani et al., 2020). Clinically, the main features related to this syndrome that suggest the diagnosis are:

increased bone fragility, short stature, rickets, and facial dimorphism, that is often, microcephaly, protrusion of frontal and occipital protrusions, micrognathia, facial hypoplasia, and nasal prominence (Elyajouri et al, 2018; Gray et al., 2019).

In addition, intraoral changes can also be found, such as delayed transition from deciduous to permanent dentition, crowding and dental impaction, enamel hypoplasia, anterior crossbite, as well as unsatisfactory oral hygiene and high prevalence of caries and periodontal problems (Moreira et al., 2019).

On radiographic examinations some findings are quite common such as diffuse or generalized osteosclerosis, delayed closures of fontanelles and cranial sutures, obtuse mandibular angle, absence of pneumatization of the facial sinuses, and increased bone radiopacity (Alves & Cantín, 2014).

Given its clinical features, osteoporosis is the main differential diagnosis of this condition, which can sometimes be confused and lead to inappropriate treatment (Abourazzak et al., 2013). Furthermore, the main differential diagnoses include other bone dysplasias, such as cleidocranial dysostosis and osteogenesis imperfecta (Valdes-Flores et al., 2014).

Therefore, the knowledge of this syndrome is essential, especially by dental surgeons in order to carefully plan clinical and surgical interventions to reduce the risk of osteomyelitis or osteonecrosis, given the defective bone remodeling (Silva et al., 2016). Thus, this paper aims to report a clinical case of pycnodysostosis and review the literature, associating its clinical and radiographic characteristics, in addition to discussing and understanding its causes and physiological repercussions, approaches and orthodontic and buccomaxillofacial orthopedic management of this syndrome.

2 CLINICAL CASE

An 8-year-old female patient, faioderma, with Toulouse-Lautrec syndrome, or pycnodysostosis, came to the Oral and Maxillofacial Surgery and Traumatology Department (MBCT) of the Hospital do Oeste, in Barreiras, with her mother complaining about her badly positioned teeth.

On physical examination, it was possible to observe that the child presented with short stature not compatible with the age range, broad hands, dystrophic nails and shortened phalanges, accentuated projection of the frontal region, hypoplastic middle third and mandibular micrognathia. Intraoral examination revealed the presence of mixed dentition, crowding in the mandible, prolonged retention of deciduous teeth, multiple dental gyrations, extensive dental caries, absence of dental unit 11, ogival palate, anterior and posterior crossbite on the left side (figure 3).

Figure 1. extraoral frontal and lateral images highlighting the frontal fossa and micrognathia.

Figure 2. Hand and nail abnormalities characteristic of pycnodysostosis.

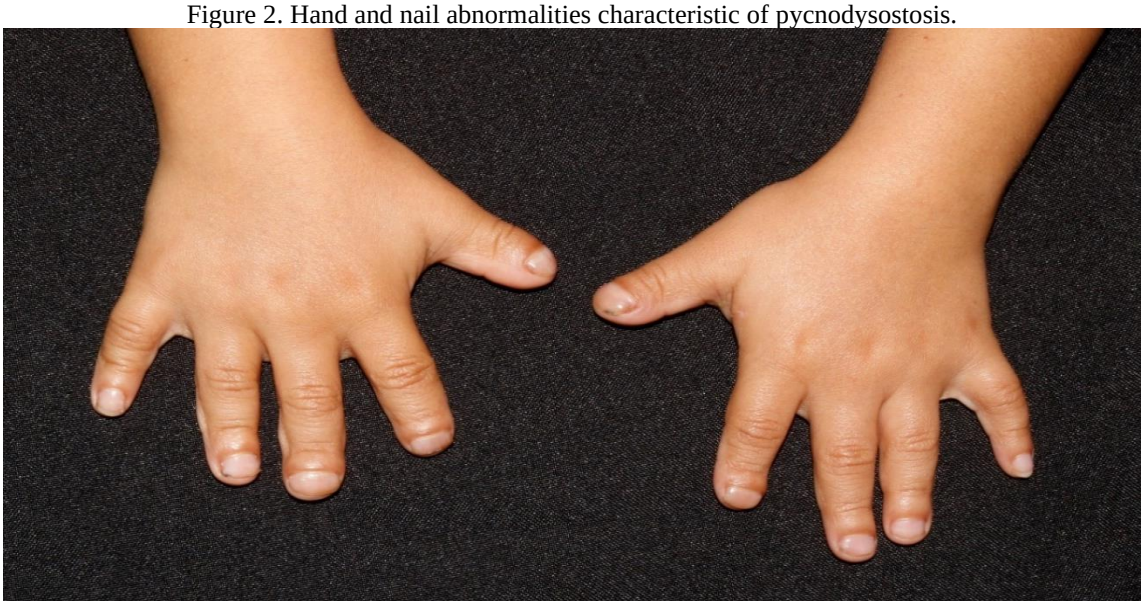
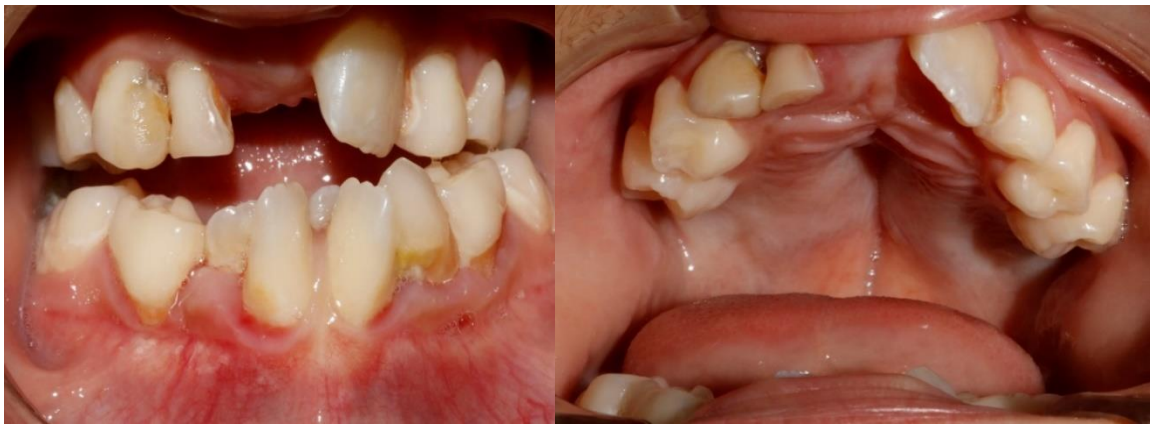


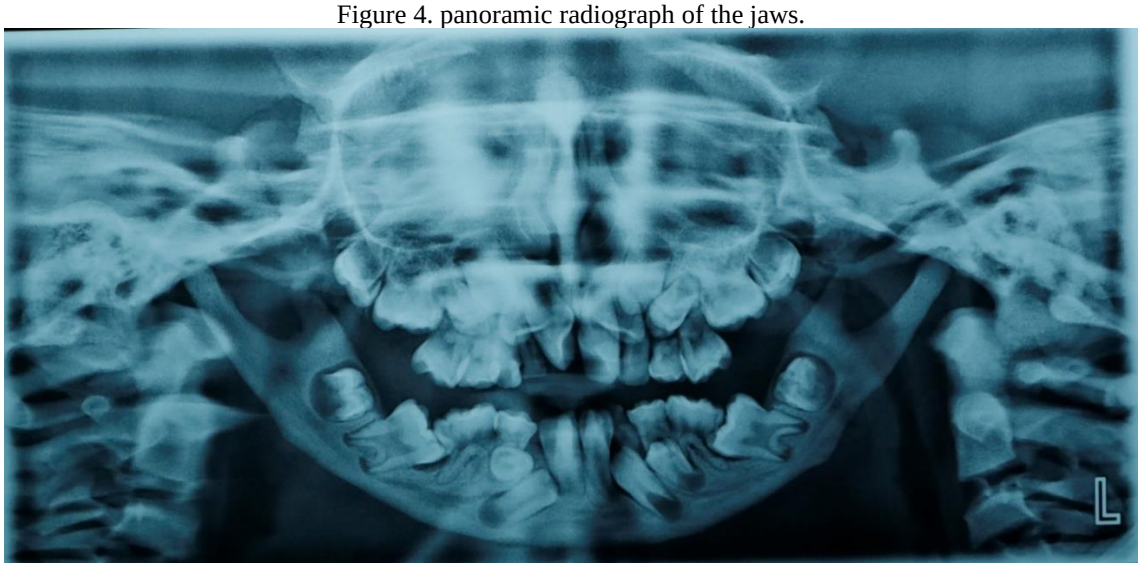
Figure 3. intraoral images showing unsatisfactory oral hygiene, dental gyrusions and crowding, crossbite and ogival palate



The imaging exam, panoramic radiography of the jaws, showed multiple retained and impacted dental units in the mandible and maxilla. In addition, the mandible was atrophic and had an obtuse mandibular angle and poorly defined bilateral sinus contour as shown in figure 4.

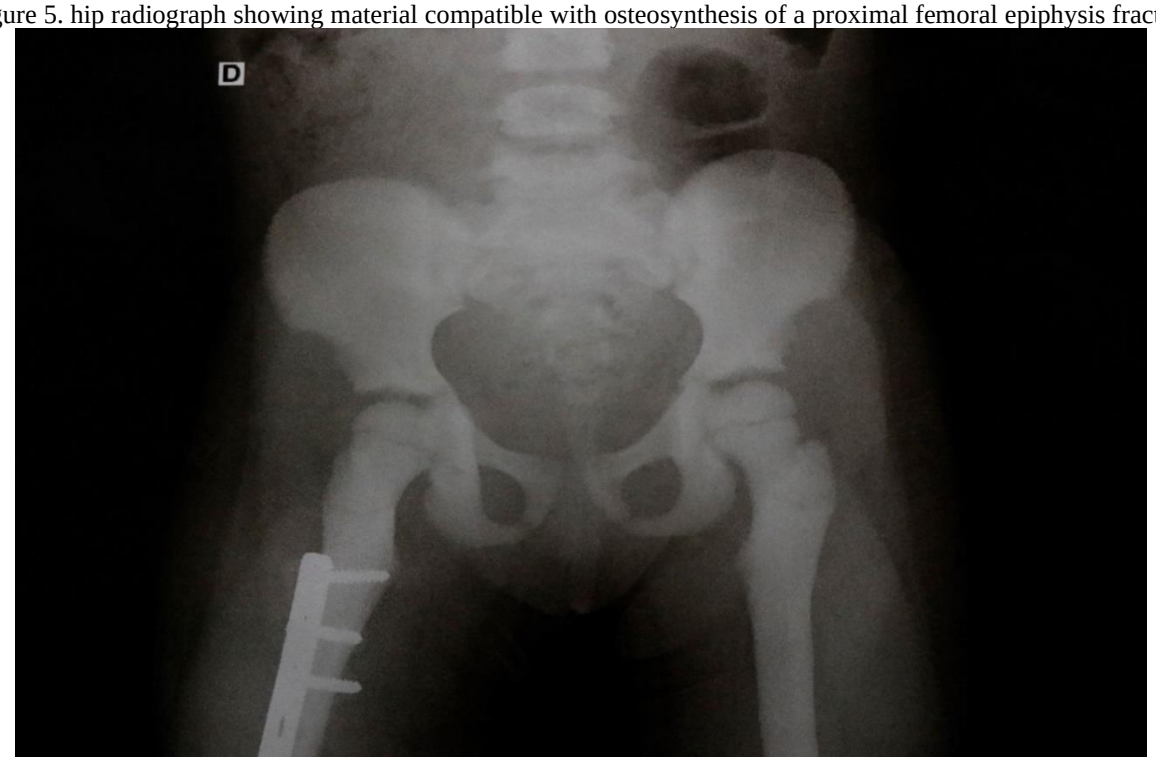
The patient has a history of consanguinity between her parents, that is, her parents are first cousins.

Figure 4. panoramic radiograph of the jaws.



The mother also reported that the patient has bone fragility due to the syndrome, with a history of a left femur fracture in 2018 with subsequent removal of bone synthesis material, and a right femur fracture in 2020, as can be seen in Figure 5.

Figure 5. hip radiograph showing material compatible with osteosynthesis of a proximal femoral epiphysis fracture



Furthermore, the patient's laboratory tests show altered calcium levels: 12.8 mg/dl (reference 8.5 to 10.5) and alkaline phosphatase: 194 IU/L (reference 20 to 150), indicating a degree of osteoporosis. At the moment, the patient is in follow-up with the CTBMF team at Hospital do Oeste, in Barreiras.

3 DISCUSSION

Pycnodysostosis is an autosomal recessive genetic condition caused by mutations in the gene responsible for encoding the enzyme Cathepsin K, which is expressed in osteoclasts and plays a key role in bone resorption and remodeling processes (Elyajouri et al, 2018; Turan et al., 2014). With this, individuals affected by this genetic alteration do not perform resorption of the organic bone matrix, and are characterized by having dense bones with thick cortical area. However, despite their high density, the bones of these patients are fragile and more susceptible to fractures (Deake et al., 2017; Ketterer et al., 2017). In the case at hand, this finding described in the literature was also observed in this patient, given the recurrence of fractures in long bones in two distinct periods, suggesting her systemic bone fragility.

According to Silva et al. (2016), this syndrome is usually associated with consanguinity between parents or a positive family history, and does not have a predilection for gender. Similarly, the patient in the present report was the result of a consanguineous relationship between her parents, since they are first cousins.

According to Rodrigues et al. (2017), pycnodysostosis is a very rare condition that has an estimated prevalence of 1 in every 1.7 million births. According to Moreira Junior et al. (2019), cases of this syndrome are most commonly found in the Asian continent, South America, and Europe, respectively. Also according to these authors, Brazil is one of the countries with the highest prevalence, accompanied by India, and in the Brazilian case, the northeastern region of the country concentrates the largest amount of cases. As described by these epidemiological findings, our patient characterizes this high regional prevalence, since she is a resident of the Brazilian northeastern region.

The clinical findings related to this syndrome are vast; short stature is a characteristic that is intrinsically related to pycnodysostosis, being reported in almost all cases previously published in the literature. Although there are also proven cases of this condition in which the individuals had normal height[9,14] . In the case of this patient, she also had a short stature, not compatible with her age group, corroborating the data presented in the literature.

Other clinical and radiological characteristics quite common in individuals with pycnodysostosis are also presented by the patient of this study, such as digital acroosteolysis, generalized bone sclerosis, delayed closure of cranial sutures and fontanelles, craniofacial deformities and midface bone hypoplasia. Besides the oral manifestations: maxillary atresia, grooved palate, obtuse mandibular angle, enamel hypoplasia, hypercementosis, dental malposition and crowding, prolonged tooth retention, increased bone density, and lack of demarcation between trabecular and cortical bone (Moreira et al., 2019).

The presence of these signs facilitates the early diagnosis of the disease; when this identification is delayed, it is usually associated with bone fracture conditions (Aynaou et al., 2016). According to the same authors, the diagnosis is based mainly on clinical and radiographic characteristics. Therefore, it is essential to establish the differential diagnosis with other diseases such as cleidocranial dysostosis, osteogenesis imperfecta and osteoporosis.

According to Reimão & Diament (1979), the laboratory findings associated with individuals with pycnodysostosis are usually uniformly normal. However, as mentioned, in the present case the patient in question presented some alterations in the levels of calcium and alkaline phosphatase, alterations that are indicative of a certain degree of osteoporosis.

Some authors state that besides the bone fragility that is usually associated with a history of fractures, as a result of the oral changes caused by this syndrome, osteomyelitis also ends up being a serious oral complication that requires greater attention from the professionals accompanying this patient, mainly due to previous dental extractions or infections (Rodrigues et al., 2017; Moreira et al., 2019).

Osteomyelitis is the main complication of jawbone related to pycnodysostosis, usually arising from previous dental extractions or infections (Mandal et al., 2016). This susceptibility to osteomyelitis occurs because of decreased blood flow in the bone, resulting from decreased osteons and obliteration of Havers' canals, which increases the chances of infection (Rodrigues et al., 2017; Moreira et al., 2019).

There are no specific dental treatment protocols for patients with pycnodysostosis reported in the literature, being recommended the supportive treatment (Kamak et al., 2012; Suassuna et al., 2021). That is, the dental therapeutic approach to this anomaly consists mainly in the adoption of measures to prevent complications such as minimizing trauma, evaluating the need for antibiotic prophylaxis in case of painful dental procedures and avoiding surgical procedures with bone manipulation. Thus, it tends to avoid the emergence of complications such as gnatic bone fracture injuries and osteomyelitis (Rodrigues et al., 2017; Suassuna et al., 2021).

Moreover, as in the present case, crowding and malocclusions, caused mainly by the shape of the palate of these individuals, are often found in patients with this genetic condition (Moreira et al., 2019). These intraoral findings are usually indicative for orthodontic treatment, in an attempt to improve the occlusal conditions of individuals (Fonteles et al., 2007; Rodrigues et al., 2017).

However, due to the absence of cathepsin K and, consequently, low bone remodeling, orthodontic and orthopedic strategies are hampered (Moreira et al., 2019). This is because, the movements adopted in these strategies depend on the normal activity of osteoclasts, bone resorption and remodeling capacity (Fonteles et al., 2007; Suassuna et al., 2021). Therefore, there are no recommendations or protocols available in the literature for performing these procedures aimed at managing the aesthetic and functional demands in patients with pycnodysostosis (Aghili et al., 2017; Mujawar et al., 2009; Rodrigues et al., 2017).

Despite the risks and the absence of protocols, there are few successful reports in the literature of maxillofacial orthodontic and surgical approaches to these patients. For example, orthognathic correction using osteogenic distraction (Nørholt et al., 2004); Le Fort III subcranial advancement with internal osteogenic distraction (Raposo-Amaral et al, 2013); rigid external distraction of the middle third of the face (Varol et al., 2011); orthognathic surgery (Hernández-Alfaro et al., 2011); and orthodontic correction using a removable maxillary spring appliance associated with extractions (Khoja et al., 2015).

Moreover, strict oral hygiene with regular medical and dental monitoring are very useful for the prevention of the main complications associated with the syndrome, given the bone conditions and oral and maxillofacial findings of individuals with this anomaly (Aynaou et al., 2016; Rodrigues et al., 2017).

4 CONCLUSION

Thus, it is clear that the knowledge of oral and maxillofacial characteristics related to pycnodysostosis is essential for the adequate and specific planning and clinical management of patients with this condition, in order to offer a humanized, systematic and individualized approach. In order to enable the prevention and treatment of common complications that affect these patients.

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