

Review Article

Clinical and radiographic maxillofacial features of pycnodysostosis

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Abstract: The aim of this study was to review of the literature to determine the radiographic and clinical maxillofacial features of pycnodysostosis emphasizing the main aspects of interest to the dentist in order to make them fit for the proper treatment of this population. It is important to make the diagnosis as early as possible in order to plan the treatment more suitable to provide a better life's quality to the patients. The most frequent clinical maxillofacial features were: grooved palate, midfacial hypoplasia, mandibular hypoplasia and enamel hypoplasia. The most common radiographic maxillofacial features were: obtuse mandibular angle, frontal/parietal/occipital bossing, open fontanels and sutures, multiple impacted teeth. The earlier diagnostic of pycnodysostosis has a fundamental role in general health of the patients. We consider that is very important that the dentist know recognize the radiographic and clinical maxillofacial features of pycnodysostosis, which allows correct treatment planning avoiding risks and ensuring better life's quality to the patients.

Keywords: Pycnodysostosis, maxillofacial abnormalities, radiography

Introduction

Pycnodysostosis is a rare autosomal recessive disorder. The main features of patients with pycnodysostosis are short stature, dysplasia of the clavicles, acro-osteolysis of distal phalanges, deformity of the skull, midfacial hypoplasia, absence or hypopneumatization of the paranasal sinuses, narrow and/or grooved palate, generalizated osteosclerosis and fragility of bone [1, 2].

Patients with pycnodysostosis present several morphological changes of maxillofacial structures, requiring specialized dental care. In the maxilla and mandible, for example, the abnormal growth of the dental arch results in dental crowding [2, 3]. This condition hinders good oral hygiene leading to a high rate of caries and periodontal disease in these patients [3, 4]. In some cases surgical intervention is required to correct malocclusion and esthetics in patients with dentofacial deformities caused by micrognathia and hypoplasia of maxilla [5-7]. In these cases must take preventive care in order

to avoid complications such as osteomyelitis [8].

The goal of this study was to review the literature to determine the radiographic and clinical maxillofacial features of pycnodysostosis emphasizing the main aspects of interest to the dentist in order to make them fit for the proper treatment of this population.

Materials and methods

Were analyzed studies published in the medical literature, after review in Pub-Med/MEDLINE, SCOPUS, ISI Web of Knowledge, SciELO and LILACS. Articles written in Portuguese, English and Spanish were searched using the keywords: pycnodysostosis, maxillofacial abnormalities, oral findings, radiography.

Original articles, systematic reviews and case report were included, longitudinal prospective studies and retrospective studies, which were published between 1970 and 2013. We excluded articles without abstract available.

Maxillofacial features of pycnodysostosis

Table 1. Clinical maxillofacial features of patients with pycnodysostosis

	Grooved palate	Midfacial hypoplasia	Mandibular hypoplasia	Dental crowding	Enamel hypoplasia	Narrow palate	Cross/open bite	Class III Skeletal	Dental abnormalities
Agarwal <i>et al.</i> [16]	++	-	+	-	-	+	-/-	-	-
Alves <i>et al.</i> [13]	+	+	+	-	-	+	-/+	-	-
Bathi & Masur [8]	++	+	-	-	+	++	-/-	-	-
Delgado <i>et al.</i> [17]	+	+	-	+	-	+	-	-	-
Fleming <i>et al.</i> [18]	+	+	+	+	-	-	-/-	-	+
Fonteles <i>et al.</i> [7]	++++	++++	+++	++++	-	-	++++/+++	++++	++++
Francisco & Nicholoff [25]	-	+	-	-	-	+	-/-	-	-
Hunt <i>et al.</i> [15]	+++	++	++	-	-	-	++/-	+	+
Ilankovan & Moos [5]	+	+	+	-	+	-	-/-	+	-
Kamak <i>et al.</i> [19]	+	+	-	-	-	+	+/+	+	+
Kawahara <i>et al.</i> [20]	+	+	+	+	-	-	-/-	-	-
Kshirsagar <i>et al.</i> [21]	+	+	-	+	-	+	-	-	-
Muto <i>et al.</i> [2]	+	+	+	-	+	-	+/-	-	-
Nørholt <i>et al.</i> [6]	-	+	-	-	-	-	+/+	+	-
O'Connell <i>et al.</i> [3]	++	+	+	++	-	-	-/-	-	-
Ramaiah <i>et al.</i> [22]	-	+	-	+	+	-	-/-	-	-
Soares <i>et al.</i> [23]	+	+	+	+	-	+	-/+	-	-
Soliman <i>et al.</i> [24]	-	-	-	-	+++++++	-	-/-	-	-
Number of patients*	22	20	13	12	11	9	9/7	8	7
Percentage (%)	66.66	60.60	39.39	36.36	33.33	27.27	27.27/21.21	24.24	21.21

*n=33; (+) presence (each + represents a patient); (-) absence or no related.

Results

A total of 54 articles were found, being 18 in Pub-Med/MEDLINE, 18 in SCOPUS, 4 in ISI Web of Knowledge, 4 in SciELO and 10 in LILACS. Were selected a total of 18 articles and based on these selected articles were included 35 patients, who had analyzed their clinical and radiographic features reported by the authors. The **Table 1** shows the clinical maxillofacial features and the **Table 2** shows the radiographic maxillofacial features of patients with pycnodysostosis, with their respective percentages and according to the authors. **Figure 1** shows a panoramic radiograph of a patient with some radiographic maxillofacial features in the pycnodysostosis: thin mandible, obtuse mandibular angle, malposed teeth, elongation of the condyle and coronoid process.

Discussion

Pycnodysostosis is a rare autosomal recessive disorder primarily characterized by osteosclerosis, bone fragility, short stature, acro-osteolysis, and delayed closure of the cranial sutures [4].

The pycnodysostosis can be confused with other diseases, such as osteopetrosis (OP) and

cleidocranial dysostosis (CD), because they present some clinical and radiographic similar signs, so it is important that the dentist knows how to make the differential diagnosis in order to indicate the best treatment for each patient. The delayed closure of sutures; frontal, occipital and/or parietal bossing; hypopneumatization of the paranasal sinuses; narrow and deep palate are features that can be observed in pycnodysostosis and in CD [8, 9]. Patients with CD present supernumerary teeth [9], however, in pycnodysostosis the appearance of supernumerary teeth is not very frequent, being reported by only 5.71% of the authors (**Table 2**). Increase of bone density and bone fragility are present in both pycnodysostosis and OP [8, 10]. The differential diagnosis between pycnodysostosis and OP is performed through the following analysis: the size of the skull, since in OP the patients have no frontal/parietal bossing; the size of the maxilla and mandible, the maxillary sinus, the sutures and the angle of the mandible, which do not show changes in OP [8].

When not diagnosed in infancy, fractures resulting from trauma lead to the diagnosis of Pycnodysostosis. Mandibular fractures have been described in adults following extractions, other important potential risk in these cases is

Maxillofacial features of pycnodysostosis

Table 2. Radiographic maxillofacial features of patients with pycnodysostosis

	Obtuse mandibular angle	Frontal/parietal/occipital bossing	Open fontanelle and sutures	Multiple impacted teeth	Absence or hypopneumatization of the paranasal sinuses	Wormian bones	elongation of the coronoid process/mandibular condyle	Supernumerary teeth
Agarwal <i>et al.</i> [16]	+	-	++	-	+	++	-/-	-
Alves <i>et al.</i> [13]	+	+	+	-	+	-	+/+	-
Alves-Pereira <i>et al.</i> [4]	+++	+++	-	-	++	++	++/++	-
Bathi & Masur [8]	++	+	++	-	++	-	-/-	-
Delgado <i>et al.</i> [17]	+	+	+	-	+	-	-	-
Fleming <i>et al.</i> [18]	+	-	-	+	+	+	-/-	-
Fonteles <i>et al.</i> [7]	+++	++++	-	++	-	-	-/-	-
Francisco & Nicholoff [25]	+	+	+	+	-	+	-/-	+
Hunt <i>et al.</i> [15]	+++	++	+++	++	+++	-	-/-	-
Ilankovan & Moos [5]	+	+	+	-	+	-	-/-	-
Kamak <i>et al.</i> [19]	+	+	+	-	-	-	-/-	-
Kawahara <i>et al.</i> [20]	+	+	+	+	-	-	-/-	-
Kshirsagar <i>et al.</i> [21]	+	+	+	+	+	-	-	-
Muto <i>et al.</i> [2]	+	+	+	-	+	-	+/-	-
Nørholt <i>et al.</i> [6]	+	-	-	-	-	-	-/-	-
O'Connell <i>et al.</i> [3]	++	++	++	++	-	-	+/+	-
Ramaiah <i>et al.</i> [22]	+	+	+	-	+	+	-/-	+
Soares <i>et al.</i> [23]	+	+	+	-	-	-	-	-
Soliman <i>et al.</i> [24]	+++++++	+++++++	+++++++	+++++++	-	-	-/-	-
Number of patients*	33	28	27	17	15	7	5/4	2
Percentage (%)	94.28	80.00	77.14	48.57	42.85	20.00	14.28/11.42	5.71

*n=35; (+) presence (each + represents a patient); (-) absence or no related.



Figure 1. The panoramic radiograph showing thin mandible, obtuse mandibular angle, malposed teeth, elongation of the condyle and coronoid process.

the occurrence of osteomyelitis [2, 11]. The osteomyelitis can be caused by oral conditions of the patient [8], after the teeth extraction, due to poor bone vascularity of these patients [4], or may be result of iatrogenic fracture [10]. Ilankovan and Moos [5] indicate that the surgical procedures in pycnodysostosis patients have enhanced possibility of infection, impaired wound healing, osteomyelitis and pathological fracture. The treatment of osteomyelitis is very difficult in these patients, especially when it involves the mandible [3].

We agree with Sedano *et al.* [12] and Muto *et al.* [2] when they say that the most common finding in patients with pycnodysostosis was obtuse mandibular angle. Elmore [1] reviewed 33 cases described in the literature and in 27 cases it was reported the loss of the mandibular angle, Alves *et al.* [13] reported that this finding was present in 87.09% of cases. To Elmore [1] the most common feature was separated cranial sutures, which was present in all cases. Maxillary hypoplasia, grooved palate, maxillary sinus hypoplasia were common findings to Muto *et al.* [2], Alves *et al.* [13] and also in our study. Open fontanelle was also a significant finding in our study, being in agreement with the findings of Elmore [1]. We found 80% of cases with frontal/parietal/occipital bossing reported, however this finding was considered inexpressive to Elmore [1]. For this author the grooved palate was also found in a small number of patients, which is in disagreement with our findings. Wormian bone was found in a few patients in the study of Elmore [1], as well as in our study. Crossbite was a little expressive feature found by Muto *et al.* [2]. Furthermore, Class III skeletal, dental abnormalities, elonga-

tion of the coronoid process and the mandibular condyle were reported by few authors.

The main clinical maxillofacial features in the pycnodysostosis include a grooved palate (66.66%), midfacial hypoplasia (60.60%), mandibular hypoplasia (39.39%), enamel hypoplasia (33.33%), dental crowding (36.36%), narrow palate (27.27%), cross bite (27.27%), class III skeletal (24.24%), open bite (21.21%) and dental abnormalities (21.21%) (**Table 1**). The main radiographic maxillofacial features in the pycnodysostosis include obtuse mandibular angle (94.28%), large head with frontal, parietal and occipital bossing (80%), open soft cranial sutures and fontanelles (77.14%), multiple impacted teeth (48.57%), absence or hypopneumatization of the paranasal sinuses (42.85%), wormian bones (20%), elongation of the coronoid process (14.28%) and the condyle (11.42%), supernumerary teeth (5.71%) (**Table 2**).

Others oral findings may consist of premature or delayed eruption teeth, persistence of deciduous teeth, malposition of teeth, hypodontia and hypercementosis [4, 10, 14]. It is also quite frequent presence of caries and periodontal disease in patients with pycnodysostosis, probably due to dental crowding [7, 8, 14, 15].

Alves *et al.* [13] affirm that the clinical and radiological features are the bases diagnosis of this disease. We agree with these authors when affirm that it is important to make the diagnosis as early as possible in order to plan the treatment more suitable to provide a better life's quality to the patients. Furthermore, the earlier diagnostic of pycnodysostosis has a fundamental role in general health of the patients.

Conclusion

The higher rates of maxillofacial features in patients with pycnodysostosis reported by authors were: obtuse mandibular angle, large head with frontal, parietal and occipital bossing, open soft cranial sutures and fontanelles, grooved palate and midfacial hypoplasia. We consider that is very important that the dentist know recognize the radiographic and clinical maxillofacial features of pycnodysostosis, which allows correct treatment planning avoiding risks and ensuring better life's quality to the patients.

Disclosure of conflict of interest

None.

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References

- [1] Elmore SM. Pycnodysostosis: A review. *J Bone Joint Surg Am* 1967; 49: 153-62.
- [2] Muto T, Michiya H, Taira H, Murase H, Kanazawa M. Pycnodysostosis. Report of a case and review of the Japanese literature, with emphasis on oral and maxillofacial findings. *Oral Surg Oral Med Oral Pathol* 1991; 72: 449-55.
- [3] O'Connell AC, Brennan MT, Francomano CA. Pycnodysostosis: orofacial manifestations in two pediatric patients. *Pediatr Dent* 1998; 20: 204-7.
- [4] Alves-Pereira D, Berini-Aytés L, Gay-Escoda C. Pycnodysostosis. A report of 3 clinical cases. *Med Oral Patol Oral Cir Bucal* 2008; 13: E633-5.
- [5] Ilankovan V, Moos KF. Pycnodysostosis: case report with surgical correction of the facial deformity. *Br J Oral Maxillofac Surg* 1990; 28: 39-42.
- [6] Nørholt SE, Bjerregaard J, Mosekilde L. Maxillary distraction osteogenesis in a patient with pycnodysostosis: a case report. *J Oral Maxillofac Surg* 2004; 62: 1037-40.
- [7] Fonteles CS, Chaves CM Jr, Da Silveira A, Soares EC, Couto JL, de Azevedo MF. Cephalometric characteristics and dentofacial abnormalities of pycnodysostosis: report of four cases from Brazil. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 2007; 104: e83-90.
- [8] Bathi RJ, Masur VN. Pycnodysostosis - a report of two cases with a brief review of the literature. *Int J Oral Maxillofac Surg* 2000; 29: 439-42.
- [9] Alves N, Oliveira R. Cleidocranial dysplasia - A case report. *Int J Morphol* 2008; 26: 1058-68.
- [10] Zachariades N, Koundouris I. Maxillofacial symptoms in two patients with pycnodysostosis. *J Oral Maxillofac Surg* 1984; 42: 819-23.
- [11] Iwu CO. Bilateral osteomyelitis of the mandible in pycnodysostosis. A case report. *Int J Oral Maxillofac Surg* 1991; 20: 71-2.

Maxillofacial features of pycnodysostosis

- [12] Sedano HG, Gorlin RJ, Anderson VE. Pycnodysostosis: clinical and genetic considerations. *Am J Dis Child* 1968; 116: 70-7.
- [13] Alves N, Oliveira RJ, Deana NF, Sampaio JCA. Morphological features of pycnodysostosis with emphasis on clinical and radiographic maxillofacial findings. *Int J Morphol* 2013; 31: 921-4.
- [14] Yamada N, Inomata H, Morita K. Two cases of pycnodysostosis with special emphasis on maxillo-facial findings. *Dentomaxillofac Radiol* 1973; 2: 12.
- [15] Hunt NP, Cunningham SJ, Adnan N, Harris M. The dental, craniofacial, and biochemical features of pycnodysostosis: a report of three new cases. *J Oral Maxillofac Surg* 1998; 56: 497-504.
- [16] Agarwal I, Kirubakaran C, Sridhar G. Pycnodysostosis: a report of two siblings with unusual manifestations. *Ann Trop Paediatr* 1999; 19: 301-5.
- [17] Delgado W, Beltrán J, Arrascue M, Oré I. Pycnodysostosis: Un síndrome óseo de interés en estomatología. *Rev Estomatol Herediana* 1999; 9: 5-14.
- [18] Fleming KW, Barest G, Sakai O. Dental and facial bone abnormalities in pycnodysostosis: CT findings. *AJNR* 2007; 28: 132-4.
- [19] Kamak H, Kamak G, Yavuz I. Clinical, radiographic, diagnostic and cephalometric features of pycnodysostosis in comparison with Turkish cephalometric norms: a case report. *Eur J Dent* 2012; 6: 454-9.
- [20] Kawahara K, Nishikiori M, Imai K, Kishi K, Fujiki Y. Radiographic observations of pycnodysostosis. Report of a case. *Oral Surg Oral Med Oral Pathol* 1977; 44: 476-82.
- [21] Kshirsagar VY, Ahmed M, Nagarsenkar S, Sahoo K, Shah KB. Ichthyosis vulgaris and pycnodysostosis: An unusual occurrence. *Acta Med Acad* 2012; 41: 214-8.
- [22] Ramaiah KK, George GB, Padiyath S, Sethuraman R, Cherian B. Pycnodysostosis: report of a rare case with review of literature. *Imaging Sci Dent* 2011; 41: 177-81.
- [23] Soares LF, Souza IPR, Cardoso AS, Pomarico L. Pycnodysostosis: Oral findings and differential diagnosis. *J Indian Soc Pedod Prev Dent* 2008; 26: S23-5.
- [24] Soliman AT, Ramadan MA, Sherif A, Aziz Bedair ES, Rizk MM. Pycnodysostosis: clinical, radiologic, and endocrine evaluation and linear growth after growth hormone therapy. *Metabolism* 2011; 50: 905-11.
- [25] Francisco JV, Nicholoff TJ. Pycnodysostosis: An unusual presentation in a denture wearer. A case report. *Oral Surg Oral Med Oral Pathol* 1991; 72: 693-5.