

Pyknodysostosis – A Case report

Nandhini Subramaniam, Arvind Muthukrishnan

Department of Oral Medicine and Radiology, Saveetha Dental college, Saveetha University, Chennai, Tamil Nadu, India

Correspondence: Dr. Arvind Muthukrishnan, Department of Oral Medicine and Radiology, Saveetha Dental college, Saveetha University, 162, Poonamallee High road, Chennai – 600 077, Tamil Nadu, India. E-mail: arvindmuthukrishnan@yahoo.com

ABSTRACT

Pyknodysostosis is a rare autosomal recessive disorder. This disorder has defective gene that codes for enzyme Cathepsin K which inhibits the normal osteoclastic function, making the bones abnormally dense and brittle. Extraoral manifestation includes increased bone density, skull deformities with delayed suture closure, proptosis, and frontal bossing. Intraoral features include anterior open bite, posterior crossbite, crowding of teeth, poor oral hygiene, and periodontal disease. Pyknodysostosis is a rare disorder of bone showing various craniofacial features. These patients usually have normal lifespan. Thorough history and clinical examination aids in diagnosis. There is increased risk of mandibular fractures and osteomyelitis following extraction. Managing with minimally invasive procedures and proper follow-up is required to prevent its complications due to invasive dental treatment.

Keywords: Cathepsin K, craniofacial abnormalities, pyknodysostosis

Introduction

Maroteaux and Lamy in 1962 defined pyknodysostosis (pyknodense, dys-defective, exostosis-bone) as a rare autosomal recessive disorder of osteoclast dysfunction causing osteosclerosis.^[1] This disorder has equal sex distribution with the incidence of 1.7 per 1 million and parental consanguinity in 30% cases.^[2] The first case description was given in 1923 by Montanari; however, Maroteaux and Lamy defined the characteristics of this disorder. This disorder is also known as “Toulouse Lautrec syndrome” named after French artist who was found to be affected with pyknodysostosis.^[3] The genetic defect is located in the chromosome 1q21.^[4] Pyknodysostosis patients present with short stature, increased bone density, skull bone deformities with delayed suture closure, acro-osteolysis of distal phalanges, unossified fontanelles, hypoplastic nails, proptosis, beaked nose, frontal and occipital bossing, hypoplastic maxilla, obtuse mandibular gonial angle, crowding of teeth, high arched palate, anterior open bite and posterior cross bite, retained deciduous, and delayed eruption of permanent teeth.^[2,5] This disorder has defective gene that codes for Cathepsin K that inhibits normal osteoclast function. Defective

osteoclast can cause impaired bone resorption and remodeling leading to dense and brittle bones. There is a greater tendency for mandibular fracture during extraction and post-extraction osteomyelitis.^[6]

Case Description

A 20-year-old female reported to the Department of Oral Medicine and Radiology with a chief complaint of pain in her right posterior tooth region for the past 1 week. On general examination, she had a short stature, frontal and parietal bossing [Figure 1], depressed nasal bridge, hypoplastic midface, proptosis, short fingers, and dystrophic nails in the lower limb [Figure 2].

Intraoral examination revealed high arched palate, crowding of teeth [Figure 3], anterior open bite [Figure 4], posterior cross bite, retained 53, 63, 73, 83, 84, 85, 74, 75, 65. Missing 13, 23, 33, 43, 34, 35, 44, 45, partially erupted 47 with pericoronal flap covering the distal portion and periodontal pocket of 6 mm distal to 47 with exudation of pus suggestive of pericoronal abscess in relation to partially erupted 47. She was given 5 days course of antibiotics (C. amoxicillin + clavulanic acid [b.i.d], T. metronidazole 400 mg [b.i.d], anti-inflammatory drugs T. Aceclofenac + T. paracetamol [t.i.d]) and astringent gum paint (tannic acid + potassium iodide + glycerine + menthol) for topical application.

Panoramic radiograph revealed crowding of teeth, retained 53, 63, 65, 73, 74, 75, 83, 84, 85 impacted 18, 13, 23, 25, 28, 35, 34, 43, 44, 45 [Figure 5] PA and lateral skull X rays showed significant hyperostosis, open fontanelle, open suture, and non-pneumatized paranasal sinuses [Figure 6].

Access this article online

Website: www.japer.in

E-ISSN: 2249-3379

How to cite this article: Subramaniam S, Muthukrishnan A. Pyknodysostosis – A Case report. J Adv Pharm Edu Res 2017;7(2):168-170.

Source of Support: Nil, **Conflict of Interest:** None declared.



Figure 1: Clinical photograph showing frontal bossing



Figure 4: Intraoral photograph showing anterior open bite



Figure 2: Clinical photograph showing dystrophic nails in the lower limbs



Figure 5: Orthopantomograph showing multiple impacted teeth and obtuse mandibular gonial angle



Figure 3: Intra oral phototograph showing crowding of teeth in the lower arch



Figure 6: PA and lateral cephalograph showing significant hyperostosis open fontanelle, open suture, and non-pneumatized paranasal sinuses

Hematologic findings revealed normal full blood count values, serum calcium, and alkaline phosphatase levels. Thyroid function test was within normal range.

Correlating the typical clinical and radiographic findings, a diagnosis of pyknodysostosis was made.

Discussion

Pyknodysostosis is an autosomal recessive disorder of bone with genetic defect located on chromosome 1q21 that codes for enzyme Cathepsin K which is responsible for normal osteoclast functioning. As a result of mutation, the defective osteoclasts cause impaired bone resorption and remodeling. Therefore, the affected individuals have dense and brittle bones increasing the risk of fracture.^[3,6]

Pyknodysostosis has few dental implications and dentist should be aware of it. There is increased the tendency for mandibular fracture during extraction and post-extraction osteomyelitis. Taking this into consideration our patient was given a 5 day course of antibiotics, anti-inflammatory drugs and was instructed to maintain oral hygiene. Furthermore, orthodontic treatment is contraindicated in these patients because of low remodeling capacity of bone.^[2]

This case report describes general features of pyknodysostosis along with classical oral features and few dental implications. Other bone diseases that should be considered as a differential diagnosis are cleidocranial dysplasia and osteopetrosis.^[6-8] Unlike pyknodysostosis, cleidocranial dysostosis will show aplasia of one or both clavicle. Osteopetrosis will have a dense bone obliterating the bone marrow leading to evident anemia and hepatosplenomegaly. However, no such features are seen in our case.

The diagnosis of pyknodysostosis is primarily based on clinical features and radiographs. However, CTSK (cathepsin k) gene mutation analysis is the confirmatory test.^[3]

There is no specific treatment. Bone fractures are the primary threat to the affected patients. Hence, care has to be taken to prevent the tendency for fracture to occur. Conservative dental management along with maintenance of oral hygiene and regular dental visits may help the affected individuals to prevent complications.

Conclusion

Pyknodysostosis is a rare disorder of bone showing various craniofacial features. These patients usually have normal lifespan. Thorough history and clinical examination aids in diagnosis. Dental treatment should be planned in such a way to avoid postprocedural complications, and it is important to educate the patient regarding oral hygiene.

Clinical significance - pyknodysostosis has increased the risk of mandibular fractures and risk of osteomyelitis following extraction and therefore requires minimally invasive procedures, and proper follow-up is required to prevent its complications due to invasive dental treatment.

References

1. Sedano HD, Gorlin RJ, Anderson VE. Pyknodysostosis. Clinical and genetic considerations. *Am J Dis Child* 1968;116:70-7.
2. Kamak H, Kamak G, Yavuz I. Clinical, radiographic, diagnostic and cephalometric features of pyknodysostosis in comparison with Turkish cephalometric norms: A case report. *Eur J Dent* 2012;6:454-9.
3. Tummala P, Rao KB, Lakshmi BS. Pyknodysostosis: A case report of radiological analysis. *J Dent Med Sci* 2013;12:6-8.
4. Fonteles CS, Chaves CM Jr, Da Silveira A, Soares EC, Couto JL, de Azevedo Mde F. Cephalometric characteristics and dentofacial abnormalities of pyknodysostosis: Report of four cases from Brazil. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 2007;104:e83-90.
5. Alves N, Cantín M. Clinical and radiographic maxillofacial features of pyknodysostosis. *Int J Clin Exp Med* 2014;7:492-6.
6. Nirupama C, Sarasakavitha D, Palanivelu S, Guhan B. Pyknodysostosis: A case report of rare entity. *J Indian Acad Oral Med Radiol* 2013;25:18.
7. Sanjay SC, Murthy K, Shukla AK, Krishnappa N. Case report-pyknodysostosis. *J Clin Diagn Res* 2015;9:TD09-10.
8. Pakshir H, Najafi HZ. Pyknodysostosis: A case report. *J Dent Tehran Univ Med Sci* 2008;5:88-93.

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.