

Pycnodysostosis: An Early Case Report With Emphasis On The Radiographic Findings

A Ferreira Costa, S Pereira de Castro Lopes, S Maria de Almeida, C Steiner

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Abstract

Pycnodysostosis (PKND) is a rare sclerosing bone disorder, characterized with generalized diffuse osteosclerosis. The head is usually large with fronto-parietal bossing and open fontanelles, hypoplastic malar and maxillary bones.

We report a case of PKND in a 4-year-old male child, first referred for evaluation with a possible diagnosis of osteopetrosis, because of his multiple members and clavicles fractures. We describe the clinical findings, dentomaxillofacial radiologic exams and differential diagnosis from osteopetrosis.

INTRODUCTION

Pycnodysostosis (PKND) is an esquelletal autosomal recessive disorder. Patients with PKND usually have normal intelligence, sexual development and life span¹. Other clinical features are skull deformities, an obtuse gonial angle, hypoplastic paranasal sinus, shortened terminal phalanges and a history of multiple fractures of the members and clavicles². Oral manifestations include anterior crossbite, increase incidence of dental caries, hypoplastic maxilla². Radiographic findings include bone density increase, persistent fontanelles and failure of closure of the cranial sutures^{1,2}. This syndrome has been seen in many races and nationalities, including black, Arab and caucasians³. The medical literature reports some cases in pediatric patients. We report here a case in a 4-year-old male child, the youngest reported in the literature to our knowledge.

CASE REPORT

The patient is a four years-old male of mixed Portuguese, Afro-Brazilian and Amerindian ancestry. He is the second child of a consanguineous couple (first-cousins) who had a previous miscarriage. He also has a healthy maternal half-sister. Family history is otherwise unremarkable. Pregnancy was complicated by amniorrhexis in the second trimester. Delivery was at term in good conditions, birth weigh 3,700 g, height 50 cm and Apgar scores 9/10.

At the clinical evaluation his weight was 12 kg and stature 90 cm (both below the 3rd percentile) and a Occipitofrontal head

circumference (OFC) of 48.5 cm (3rd percentile). He was also noted to present prominent calvaria, open fontanelles, small faces with micrognathia and brachydactyly of fingers with shortening of the distal phalanges. Clinical oral exam showed multiple acute dental cavities, associated with malpositioned teeth and anterior crossbite.

Neuropsychomotor development was normal. At age four, both fontanelles remain open. The patient also presented with multiple fractures of the members and clavicles being referred for evaluation due to a possible diagnosis of osteopetrosis.

The radiological evaluation consisted in a panoramic (fig 1), postero-anterior skull x-ray (fig 2) and hand radiograph (fig 3). These exams revealed generalized sclerosis of the skeleton, including skull, being more pronounced in the periorbital region ("harlequin appearance" or "raccoon mask" sign), wormian bones, open fontanelles and cranial sutures; absence of facial sinuses, and obtuse mandible angle and clubbing of the terminal phalanges.

Figure 1

Figure 1: Panoramic radiography revealing obtuse mandible angle, acute caries and malpositioned teeth.

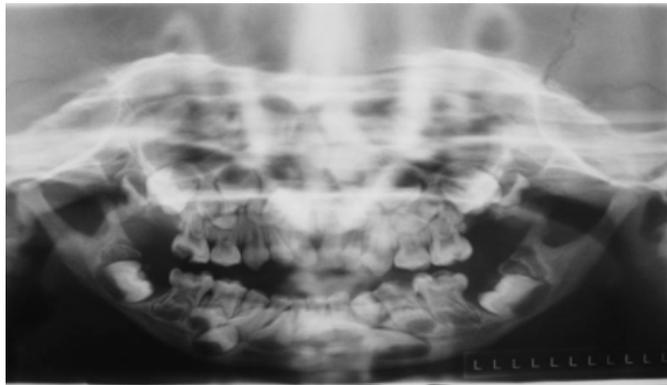


Figure 2

Figure 2: Postero-anterior skull radiography revealing generalized sclerosis of the skeleton, more pronounced in the periorbital region (the "harlequin appearance" or "raccoon mask" sign), open fontanelles and cranial sutures, absence of facial sinuses.



Figure 3

Figure 3: Hand radiography showing clubbing of the terminal phalanges.



The patient described here had the clinical and radiological features of pycnodysostosis. Features with differentiate osteopetrosis (Albers-Schönberg disease) from PKDN are normal stature, cranial nerve compression, myelophthitic anemia. Skull dysplasia and hipoplasia of the distal phalanges are not commonly reported with osteopetrosis.

The patient was referred to the pediatric dental office for clinical and orthodontic treatment.

DISCUSSION

A normal life span can be expected in individuals with PKDN, but the manifestations in the oral region can affect the morbidity and mortality from this disease₂.

Complications expected during the treatment of oral diseases such as dental cavities and teeth extractions are jaw fractures and osteomyelitis. The treatment of osteomyelitis is very difficult in PKDN patients, especially in the mandible₂.

Deficient growth of the maxilla and mandible makes normal tooth alignment impossible, with dental crowding and difficult oral hygiene.

Early intervention to relieve dental crowding may be indicate to the pediatric patient to allow better alignment of primary and erupting permanent teeth. An ideal alignment will lead to easier dental hygiene and avoid osteomyelitis due to dental extraction later in life. No recommendations are available in the literature regarding the efficacy and safety of orthodontics treatment in children with PKDN_{1,2}.

Due to the increased morbidity in patients with PKDN, this patients should be enrolled in a prevention oral diseases program from their early years₄, consisting of oral hygiene instruction and frequent visits to the dentist. The earlier diagnostic of this pathology has a fundamental role in general health of the patients and the dentists should be able recognize its signs in radiographs.

CORRESPONDENCE TO

Andre Luiz Ferreira Costa PhD student Laboratory of Neuroimaging, UNICAMP Campinas, Brazil 13.081-970
Work phone: 55 19 3788 92 17 E-mail:
alcosta@fcm.unicamp.br

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Author Information

André Luiz Ferreira Costa

Ph.D. Student, Laboratory of Neuroimaging, UNICAMP

Sergio Lucio Pereira de Castro Lopes

Ph.D. Student, Department of Dental and Maxillofacial Radiology, UNICAMP

Solange Maria de Almeida, D.D.S., Ph.D.

Department of Dental and Maxillofacial Radiology, UNICAMP

Carlos Eduardo Steiner, M.D., Ph.D.

Department of Medical Genetics, UNICAMP