

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

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## Abstract

Pyknodysostosis (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 multiples 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 span<sup>1</sup>. 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 clavicles<sup>2</sup>. Oral manifestations include anterior crossbite, increase incidence of dental caries, hypoplastic maxilla<sup>2</sup>. Radiographic findings include bone density increase, persistent fontanelles and failure of closure of the cranial sutures<sup>1,2</sup>. This syndrome has been seen in many races and nationalities, including black, Arab and caucasians<sup>3</sup>. 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 3<sup>rd</sup> percentile) and a Occipitofrontal head

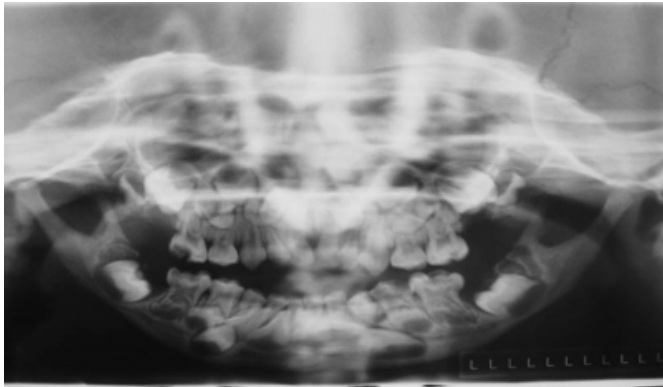
circumference (OFC) of 48.5 cm (3<sup>rd</sup> percentile). He was also noted to present prominent calvaria, open fontanelles, small faces with micrognathia and brachydactyly of fingers with shortening of the distal phalanges. Clinical oral exam showed multiple acute dental cavities, associated with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

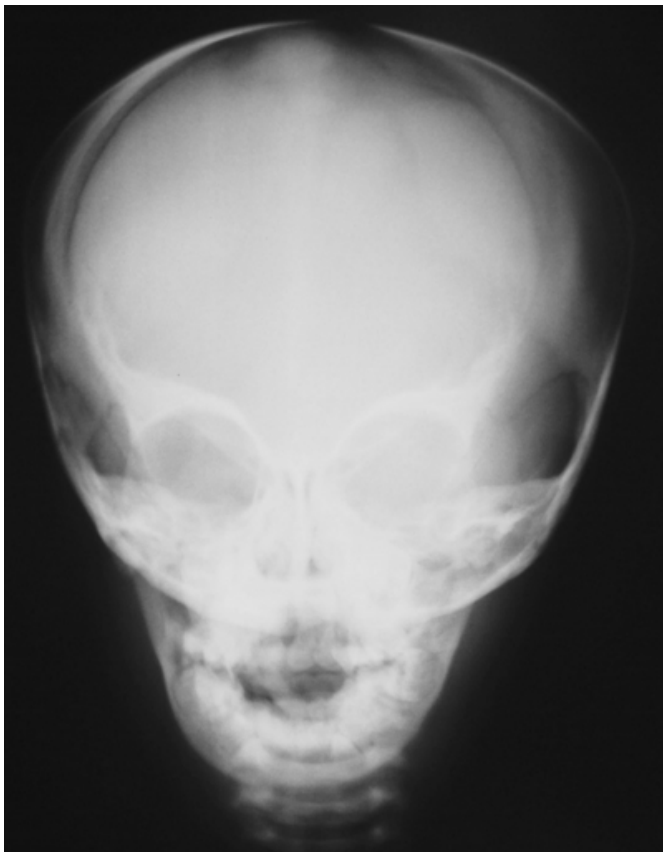
**Figure 1**

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 (Â“harlequin appearanceÂ” or Â“raccoon maskÂ” 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 disease<sub>2</sub>.

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 mandible<sub>2</sub>.

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<sub>1,2</sub>.

Due to the increased morbidity in patients with PKDN, this patients should be enrolled in a prevention oral diseases program from their early years<sub>4</sub>, 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.

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