

CASE REPORT

K.W. Fleming
G. Barest
O. Sakai

Dental and Facial Bone Abnormalities in Pyknodysostosis: CT Findings

SUMMARY: Pyknodysostosis is an autosomal-recessive disorder of osteoclast dysfunction causing osteosclerosis, with associated maxillofacial anomalies. Multidetector CT with multiplanar and 3D reconstruction illustrated the pathologic findings in this case. Abnormalities included multiple retained deciduous teeth, unerupted teeth with associated follicles, an irregularly expanded alveolus and body of the mandible, and an obtuse mandibular angle. Volume-rendered imaging better delineated the irregular dentition, with crowding and retention of deciduous teeth.

P yknodysostosis is a rare autosomal-recessive disorder of osteoclast dysfunction causing osteosclerosis. The name derives from the Greek “pyknos,” meaning dense. The disorder is also known as Toulouse-Lautrec syndrome, named for the famous French artist who was thought to be afflicted with pyknodysostosis.¹

Case Report

A 20-year-old man with pyknodysostosis reported for a CT of the face, requested for preoperative planning. The patient had presented to the oral-maxillofacial surgery clinic with the complaint, “I need some teeth.” Panorax radiograph revealed multiple disorganized crowded deciduous and permanent teeth within the expanded maxillary and mandibular alveolar processes (Fig 1A). Many of the teeth were unerupted or only partially erupted. Precise evaluation of anatomic relationships among teeth was difficult secondary to overlap. This problem was worst in the areas of tooth crowding. Lucencies around the unerupted teeth, possibly representing normal follicles, were present; however, ill-defined borders may have indicated a superimposed inflammatory/infectious process.

Noncontrast CT of the maxillofacial bones was performed on a 4-detector-row CT scanner (Phillips MX-8000, Phillips Medical Systems, Andover, Mass). Axial images of the face were obtained by using 1.3-mm collimation, and multiplanar and 3D reconstructions were generated on a workstation (Voxar, Framingham, Mass). The scout topogram revealed Wormian bones within the lambdoid suture, a finding often associated with pyknodysostosis (Fig 1B). Axial (Fig 1C, -D), coronal (Fig 1E), and sagittal (Fig 1F) reformatted images demonstrated multiple dental abnormalities. There was persistence of the deciduous teeth within the maxilla and mandible, causing marked crowding. The teeth were misaligned and disorganized, and multiple teeth were unerupted or only minimally erupted. Unerupted teeth were surrounded by well-defined lucent areas, consistent with normal follicles. Poorly marginated lucencies in this location suggested inflammation and/or infection. Along with the dental abnormalities, marked hypoplasia of the maxillary, frontal, and sphenoid sinuses was noted. Unerupted maxillary teeth were seen in the floor of the hypoplastic maxillary sinus, with a few teeth protruding into the antrum (Fig 1E, -F). The hard palate was relatively deep and grooved. The gonial angle of the mandible was obtuse. Note was also made of mild proptosis. Sagittal reformatted images revealed a relatively beaked nose. The surface-rendered 3D image (Fig 1G) gave an overview of these deformities. Irregular dentition was best delineated with volume-

rendered display (Fig 1H). Interactive review at the workstation with rotation in any axis and simultaneous orthogonal/oblique multiplanar reconstructions allowed a thorough understanding of all the abnormalities.

Discussion

Pyknodysostosis is an autosomal-recessive disorder of osteoclast dysfunction causing osteosclerosis. Some features of pyknodysostosis overlap the more common osteopetrosis and cleidocranial dysostosis.² It is believed that the first case description was in 1923 by Montanari; however, it was not until 1962 that Maroteaux and Lamy defined the characteristic features of pyknodysostosis.³ General features include short stature (<150 cm), generalized diffuse osteosclerosis with a tendency for fracture after minimal trauma, hypoplastic clavicles,^{3,4} as well as acro-osteolysis with sclerosis of the terminal phalanges—a feature that is considered essentially pathognomonic.^{1,5} Cranial and maxillofacial features include frontoparietal bossing, thick calvaria, open fontanelles and sutures, hypoplastic paranasal sinuses, Wormian bones in the lambdoid region, relative proptosis, beaked nose, and an obtuse mandibular gonial angle, often with relative prognathism.^{3,4,6} These findings were seen in our patient, with hypoplastic sinuses being a prominent feature.

Patients may present with frequent fractures, recurrent dental abscesses, or obstructive sleep apnea. Parental consanguinity is recognized as a cause of this autosomal-recessive disorder, the responsible gene being located on chromosome 1q21. This gene encodes cathepsin K, a cysteine proteinase that is expressed in normal osteoclasts and is mutated in patients with pyknodysostosis.^{5,7} Intraoral features include persistence of deciduous teeth, with premature or delayed rupture of permanent teeth, which can cause crowding. Also seen are tooth misalignment, enamel hypoplasia, and a grooved palate.^{3,4} Dental abnormality was the most impressive finding in this patient, with persistence of deciduous teeth and delayed rupture of permanent teeth. As mentioned previously, the follicles of these unerupted teeth may become infected, leading to abscess formation.

There is limited published material describing the CT findings in patients with pyknodysostosis. CT findings including hypoplastic sinuses, poor dentition, and thickening of the calvaria have been described for osteopetrosis—a similar more common entity.⁸ Compared with Panorax radiographs, CT with reconstructions demonstrates greater anatomic detail. Axial imaging defines exact relationships between the unerupted teeth not possible on Panorax alone. Coronal and sagittal reformatting facilitates interpretation of anatomic relationships in the craniocaudal direction. Surface rendering gives an overall perspective of the underlying

Received December 7, 2005; accepted December 10.

From the Department of Radiology, Boston University Medical Center, Boston, Mass.

Please address correspondence to Keith W. Fleming, MD, New York Presbyterian Hospital/Weill Cornell Medical Center, 525 E. 68th St, New York, NY 10021; e-mail: keithwfleming@gmail.com

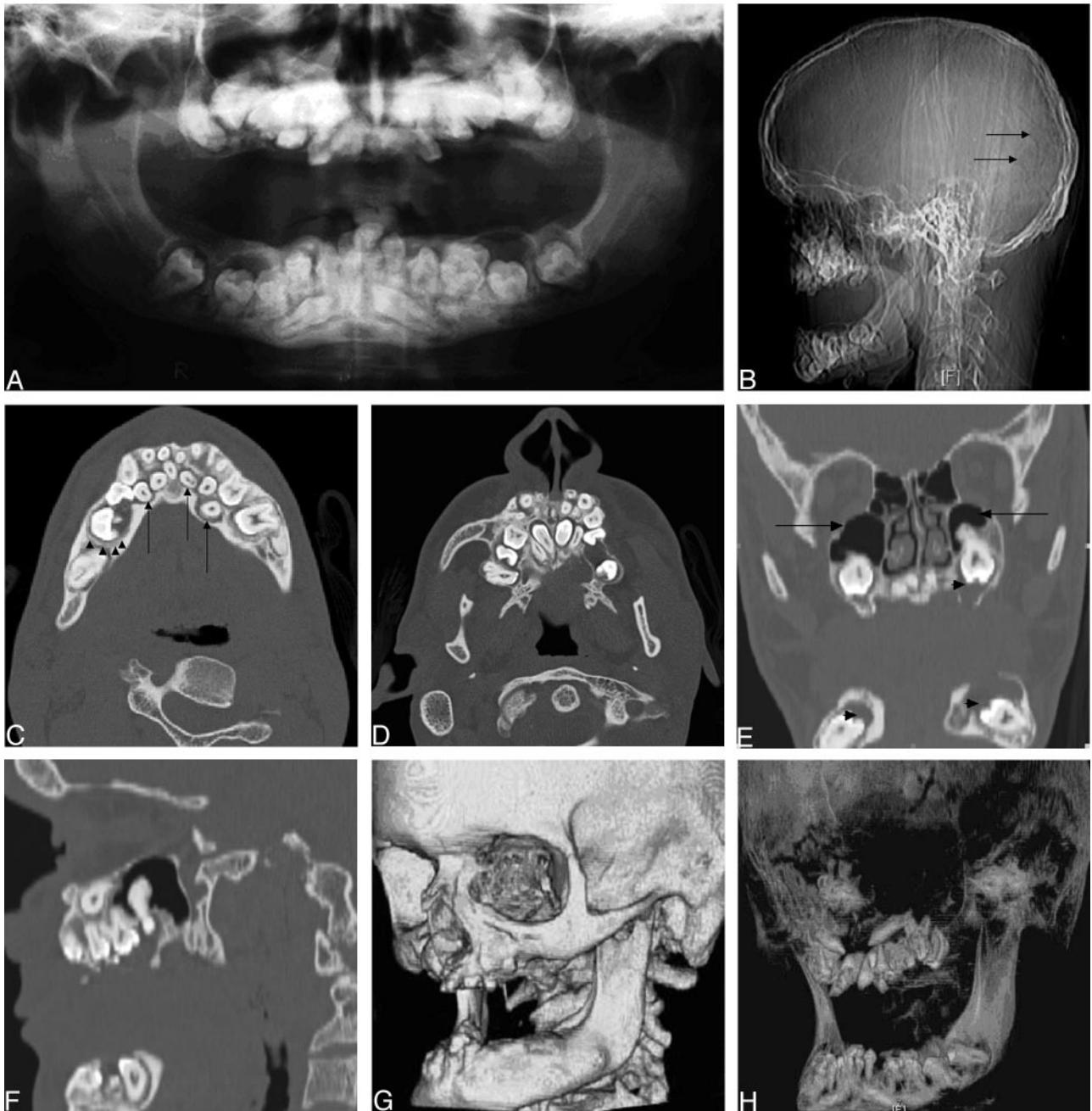


Fig 1. A 20-year-old man with pyknodysostosis.

A, Panorax radiograph reveals multiple disorganized crowded deciduous and permanent teeth within the expanded alveolar processes. Many of the teeth are unerupted or only partially erupted. Evaluation of follicles is limited.

B, Scout topogram demonstrates Wormian bones (*arrows*) within the lambdoid suture. Obtuse gonial angle of the mandible and dental abnormalities are also visualized.

C and *D*, Axial CT images at the level of the mandible (*C*) and maxilla (*D*) reveal multiple unerupted crowded deciduous and permanent teeth within the expanded alveolar processes. Lucencies around several of the teeth have well-defined margins, very likely representing follicles (*arrows*). However, bandlike zones of demineralization suggest an active inflammatory/infectious process (*arrowheads*).

E and *F*, Coronal (*E*) and sagittal (*F*) reformatted CT images demonstrate the crowded teeth in the maxilla and mandible with associated lucencies (*arrowheads*) surrounding the unerupted teeth. Note the hypoplastic maxillary sinuses bilaterally (*arrows*).

G, Surface-rendered 3D image shows irregularly expanded alveolar processes and mandibular body and a flattened obtuse mandibular angle. The technique limits evaluation for irregular dentition with crowding and retention of deciduous teeth.

H, Volume-rendered 3D image better delineates irregular dentition with crowding and retention of deciduous teeth.

bony abnormalities, and volume rendering aids in determining the 3D relationships among the abnormal teeth by making the alveolar process less conspicuous. Demonstrating the exact relationships among abnormal teeth can aid the surgeon in planning

extractions and/or reconstructions. Also, improved visualization of cortical bone can aid in distinguishing infected follicles and dental abscesses from the normal well-corticated follicles of unerupted teeth. This helps the surgeon gain a more complete under-

standing of the abnormal anatomy and is useful for planning of tooth extraction and/or implantation of prosthetics. MR imaging findings in patients with pyknodysostosis generally reveal normal cortical thickness in the calvaria; however, there is increased trabecular bone within the medullary cavity, which causes decreased marrow signal intensity.⁹

In summary, axial multidetector CT with multiplanar reformatted 3D surface-shaded and volume-rendered images demonstrated detailed anatomic and pathologic features of the teeth and facial bones in this patient with pyknodysostosis. Excellent delineation of unerupted and partially erupted teeth was obtained.

References

1. Helms CA. **Metabolic bone disease.** In: Brant WE, Helms CA, eds. *Fundamentals of Diagnostic Radiology*. Philadelphia: Lippincott Williams & Wilkins; 1999; 1049–59
2. Beighton P, Horan F, Hamersma H. **A review of osteopetroses.** *Postgrad Med J* 1977;53:507–16
3. Bathi RJ, Masur VN. **Pyknodysostosis: a report of two cases with a brief review of the literature.** *Int J Oral Maxillofac Surg* 2000;29:439–42
4. Hunt NP, Cunningham SJ, Adnan N, et al. **The dental, craniofacial, and biochemical features of pyknodysostosis: a report of three new cases.** *J Oral Maxillofac Surg* 1998;56:497–504
5. Krane SM, Schiller AL. **Hyperostosis, fibrous dysplasia, and other dysplasias of bone and cartilage.** In: Fauci AS, Braunwald E, Isselbacher KJ, et al, eds. *Harrison's Principles of Internal Medicine*. New York: McGraw-Hill; 1998: 2269–75
6. Glass RBJ, Fernbach SK, Norton KI, et al. **The infant skull: a vault of information.** *RadioGraphics* 2004;24:507–22
7. Gelb BD, Shi GP, Chapman HA, et al. **Pyknodysostosis, a lysosomal disease caused by cathepsin K deficiency.** *Science* 1996;273:1236–38
8. Elster AD, Theros EG, Key LL, et al. **Cranial imaging in autosomal recessive osteopetrosis. Part 1. Facial bones and calvarium.** *Radiology* 1992;183: 129–35
9. Karabi S, Reis ND, Linn S, et al. **Pyknodysostosis: imaging and laboratory observations.** *Calcif Tissue Int* 1993;53:170–73