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CASE REPORT

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The silent anatomy behind the noise: Structural airway narrowing in pycnodysostosis

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ABSTRACT

Background: Pycnodysostosis, also known as Toulouse-Lautrec syndrome, is a rare autosomal recessive osteosclerotic skeletal dysplasia caused by CTSK gene mutations encoding cathepsin K, leading to defective osteoclastic bone resorption. It is characterized by short stature, craniofacial dysmorphism, acro-osteolysis, and bone fragility.

Case Presentation: We report the case of a 14-year-old girl born to consanguineous parents who presented with persistent snoring despite prior adeno-tonsillectomy, progressive conductive hearing loss, recurrent fractures, and characteristic craniofacial abnormalities. Craniofacial CT demonstrated diffuse osteosclerosis, persistent cranial sutures, mandibular hypoplasia, mastoid sclerosis, and complete agenesis of the frontal, maxillary, and sphenoidal sinuses—an exceptionally rare manifestation. Laboratory findings excluded osteopetrosis.

Conclusion: This case expands the phenotypic spectrum of pycnodysostosis by documenting complete paranasal sinus agenesis and emphasizes the essential role of CT imaging in diagnosing structural causes of persistent upper airway dysfunction and conductive hearing loss in affected patients.

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Introduction

Pycnodysostosis is a rare autosomal recessive osteosclerotic dysplasia first described by Maroteaux and Lamy in 1962 [1].

It results from loss-of-function mutations in the *CTSK* gene on chromosome 1q21, leading to cathepsin K deficiency and subsequent impairment of osteoclastic bone resorption [2]. The disease is characterized by dense yet brittle bones, proportionate short stature, persistent cranial sutures, mandibular hypoplasia, acro-osteolysis of the distal phalanges, and dental anomalies [3]. Despite these characteristic features, the diagnosis is often delayed or initially misattributed to more common skeletal dysplasias.

The disease's estimated prevalence is about 1 in 1 million, but it is more common in regions with high consanguinity rates [4].

Craniofacial involvement has important implications for the upper airway. Hypoplasia of the maxilla and mandible, combined with a relatively long soft palate and glossoptosis, may reduce the pharyngeal airway space, predisposing affected individuals to chronic mouth breathing, snoring, and sleep-disordered breathing [5].

However, these symptoms are frequently attributed to adeno-tonsillar hypertrophy in childhood, and patients may undergo adenoidectomy or tonsillectomy without significant improvement. Persistent snoring after upper airway surgery should therefore prompt evaluation for an underlying craniofacial dysplasia.

Fractures following minor trauma are frequent, though hematopoiesis remains normal, distinguishing the disorder from osteopetrosis [6].

Historically, the syndrome is also known as the "Toulouse-Lautrec syndrome," since the French painter Henri de Toulouse-Lautrec is believed to

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have suffered from pycnodysostosis, given his short stature and recurrent fractures [7]. Radiology plays a pivotal role in identifying the disease and differentiating it from other osteosclerotic conditions such as osteopetrosis and craniometaphyseal dysplasia [8].

Here, we report the case of a 14-year-old girl who presented with persistent snoring despite prior adenoidectomy and tonsillectomy. Radiological evaluation performed for suspected nasopharyngeal obstruction revealed characteristic craniofacial and skeletal features of pycnodysostosis, leading to the correct diagnosis. This case highlights the crucial role of imaging in identifying syndromic causes of upper airway dysfunction and emphasizes the need to recognize snoring as a potential clinical clue to underlying skeletal dysplasia.

Case Presentation

A 14-year-old girl was referred to the radiology department for evaluation of persistent snoring and suspected nasopharyngeal narrowing. She was born to first-degree consanguineous parents (cousins) and has one healthy older sibling. Since early childhood, she had been followed for short but proportionate stature and craniofacial dysmorphism, initially misdiagnosed as achondroplasia at the age of 10. According to her mother, she had experienced multiple fractures following minimal trauma (Fig. 4), suggestive of significant bone fragility. Psychomotor and cognitive development were normal. At the age of 8, she had undergone adeno-tonsillectomy for presumed adenoidal hypertrophy; however, snoring persisted postoperatively.

Clinical examination revealed proportionate short stature, frontal bossing, a prominent nasal bridge, and micrognathia, resulting in a characteristic craniofacial appearance (Fig. 1). There was dental malalignment with crowding (Fig. 5). The hands were short and broad, showing acro-osteolysis of the distal phalanges and spoon-shaped nails (Fig. 2). A dorsal scoliosis with thoracic asymmetry was also noted (Fig. 3). No hepatosplenomegaly or neurological abnormalities were present. The patient also reported progressive hearing impairment. CT imaging demonstrated bilateral sclerotic, non-pneumatized mastoid air cells, leading to poor ventilation of the middle ear and reduced ossicular chain mobility, consistent with conductive hearing loss of structural origin.

A craniofacial CT scan performed to evaluate persistent snoring demonstrated diffuse craniofacial osteosclerosis (Fig. 6), persistent cranial sutures, particularly in the lambdoid region (Fig. 7), and mandibular hypoplasia with an obtuse mandibular angle (Fig. 8). Notably, there was complete agenesis of the maxillary, sphenoidal, and frontal sinuses, replaced by dense compact bone, along with poor pneumatization of the mastoid air cells (Fig. 9). Multiple impacted teeth with associated dentigerous cysts were also present.

Skeletal imaging showed generalized osteosclerosis, without cortical thickening or medullary



Figure 1. Clinical appearance: proportionate short stature and craniofacial dysmorphism.



Figure 2. Acro-osteolysis: resorption of the distal phalanges with spoon-shaped nails.

narrowing. Laboratory investigations revealed vitamin D deficiency, while serum calcium, phosphate, and alkaline phosphatase levels were within normal ranges. Complete blood count showed no anemia or cytopenia, and there was no evidence of bone marrow failure, effectively excluding osteopetrosis.

Based on the combination of proportionate short stature, recurrent fractures, characteristic craniofacial dysmorphism, acro-osteolysis observed clinically, and further supported by the patient's history and morphotype, the diagnosis of pycnodysostosis (Bouderet's disease, or Toulouse-Lautrec syndrome) was strongly suspected before imaging. The confirmation was then established by the Radiology Department of 20 August Hospital, as the craniofacial CT findings, particularly the complete sinus agenesis, mandibular hypoplasia, and persistent cranial sutures, were the decisive features that definitively unveiled the syndrome.

Following this radiologic identification, the patient and her family were referred for genetic counseling and multidisciplinary management, including orthopedic, dental, and otolaryngology management.

Results and Discussion

Pycnodysostosis is a rare autosomal recessive osteosclerotic dysplasia caused by loss-of-function mutations in the *CTSK* gene, which encodes cathepsin K,

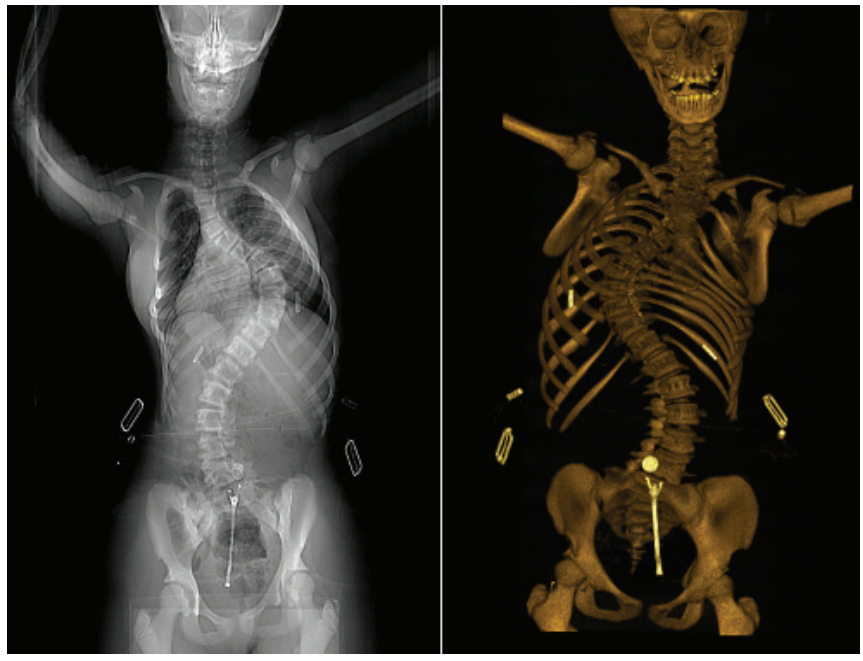


Figure 3. Dorsal scoliosis: full-spine radiographic and 3D CT reconstructions demonstrating a dorsal scoliosis with associated thoracic cage asymmetry.

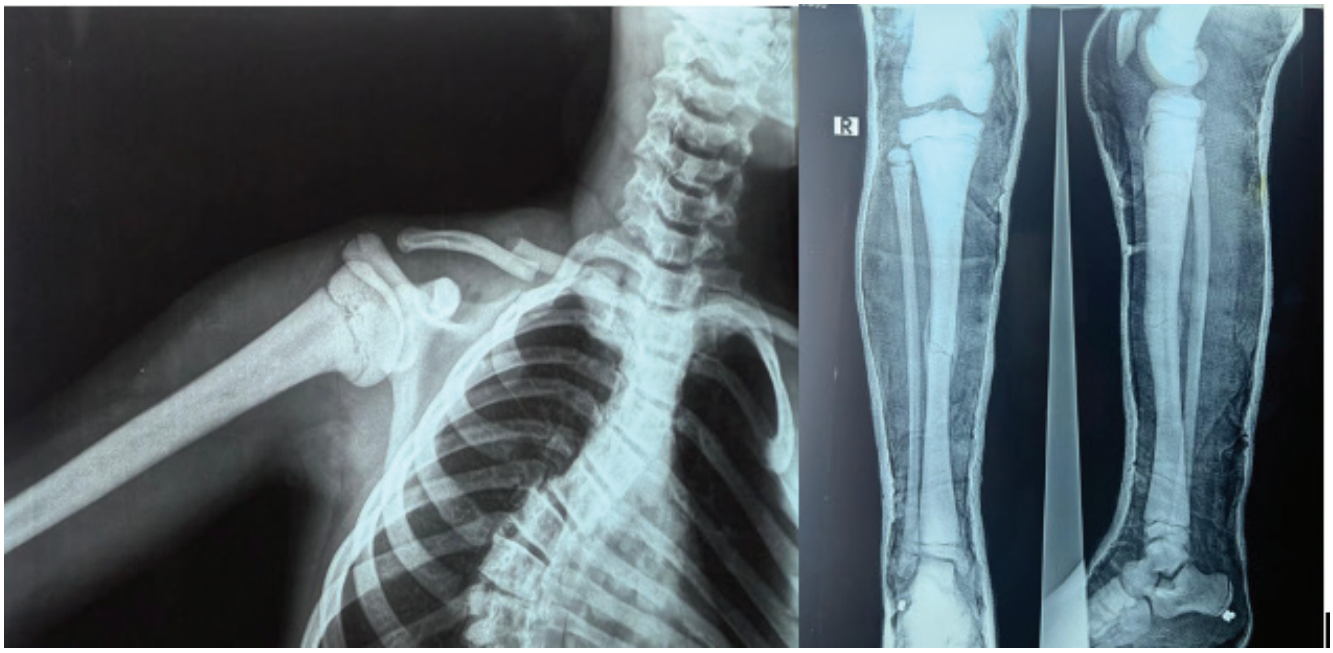


Figure 4. Recurrent fractures due to bone fragility.



Figure 5. Dental malalignment and tooth crowding.

a cysteine protease essential for osteoclastic degradation of type I collagen [2,9]. Impaired osteoclastic bone resorption leads to the accumulation of dense but brittle bone, explaining the high frequency of fractures despite normal calcium and phosphate metabolism.

The clinical phenotype is characteristic: proportionate short stature, frontal and occipital bossing, persistent open sutures, mandibular hypoplasia, acro-osteolysis, and dental crowding [3]. Spinal deformities such as scoliosis may occur secondarily to compromised bone strength [10]. Hematopoiesis typically remains preserved, distinguishing pycnodysostosis from osteopetrosis, in which bone marrow failure and cranial-nerve compression are common [11].

Radiologically, pycnodysostosis demonstrates generalized osteosclerosis, open or partially fused sutures, an obtuse mandibular angle, and hypoplasia of the maxilla and mandible [10]. In our patient, the lambdoid sutures remained widely open, reflecting impaired skull vault remodeling due to osteoclastic dysfunction, a typical but often overlooked radiological clue to pycnodysostosis.



Figure 6. Craniofacial CT demonstrating diffuse generalized osteosclerosis affecting the skull base and facial bones, characteristic of pycnodysostosis.

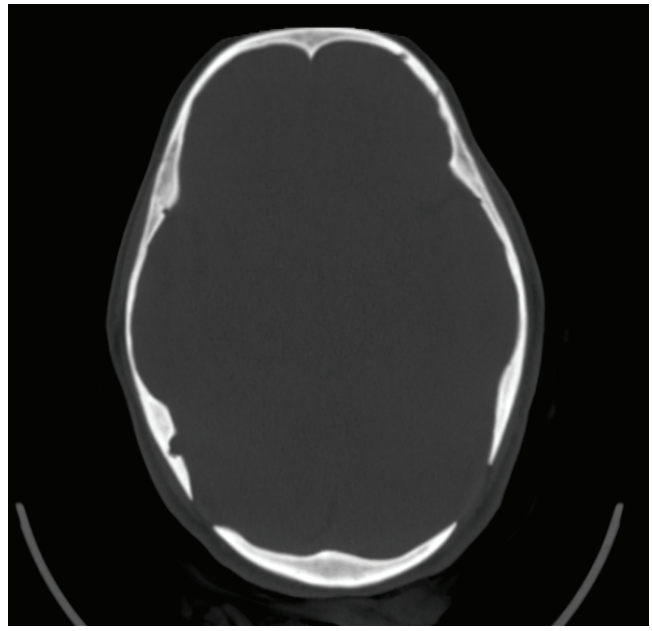


Figure 7. Craniofacial CT demonstrating persistent open lambdoid sutures due to defective skull vault remodeling, a characteristic radiologic feature of pycnodysostosis.



Figure 8. Sagittal and 3D CT reconstructions demonstrating mandibular hypoplasia with characteristic obtuse mandibular angle, contributing to craniofacial disproportion and upper airway narrowing.

In addition, complete agenesis of all paranasal sinuses, including the maxillary, sphenoidal, and frontal sinuses, was observed. Although sinus hypopneumatization has been previously reported, total sinus agenesis remains exceptional [8], and reflects the underlying osteoclastic dysfunction that prevents normal pneumatization of craniofacial bone.

The paradoxical coexistence of hyperostotic craniofacial bones with persistent open sutures reflects region-specific consequences of impaired osteoclastic remodeling. Excessive bone deposition within paranasal sinus regions likely prevents normal pneumatization during craniofacial development, whereas defective skull vault remodeling contributes to delayed suture closure. This

distinctive pattern helps differentiate pycnodysostosis from other osteosclerotic disorders.

Importantly, our case also highlights upper airway functional consequences. Despite prior adeno-tonsillectomy, the patient continued to exhibit significant snoring, which was likely multifactorial, with craniofacial structural narrowing related to mandibular hypoplasia, glossoptosis, and reduced nasopharyngeal space serving as major contributing factors. This observation aligns with cephalometric studies showing reduced upper airway dimensions in pycnodysostosis, predisposing to snoring and sleep-disordered breathing [12].

Additionally, the patient reported progressive conductive hearing loss, explained by the bilateral sclerosis and lack of pneumatization of the mastoid air cells, leading to middle ear under-ventilation and reduced ossicular chain mobility. This mechanism emphasizes that hearing impairment in pycnodysostosis may be structural.

This case has several limitations. Genetic confirmation through CTSK mutation analysis was not available at the time of reporting due to resource limitations. Functional assessments, including polysomnography, formal audiometry, and 3D pharyngeal airway volumetric analysis, were also unavailable. Therefore, while imaging strongly supports structural contributors to the patient's symptoms, functional severity could not be objectively quantified. The key clinical, radiologic, and functional features of pycnodysostosis are summarized in Table 1. The main differential diagnoses and their distinguishing features are summarized in Table 2.



Figure 9. Axial craniofacial CT image showing complete absence of the maxillary, sphenoidal, and frontal sinuses, replaced by dense compact bone, associated with bilateral mastoid sclerosis and marked hypopneumatization. These findings support impaired craniofacial pneumatization related to osteoclastic dysfunction.

The differential diagnosis of pycnodysostosis includes several skeletal dysplasias with overlapping osteosclerotic or craniofacial features. Osteopetrosis may show diffuse osteosclerosis and cranial nerve involvement, but it is typically associated with medullary canal narrowing, bone marrow failure, anemia, hepatosplenomegaly, and absence of acro-osteolysis. Craniometaphyseal dysplasia is characterized by progressive craniofacial hyperostosis with metaphyseal widening of the long bones, rather than the combination of acro-osteolysis, open cranial sutures, and obtuse mandibular angle. Cleidocranial dysplasia may also present with delayed suture closure, dental abnormalities, and craniofacial dysmorphism, but clavicular hypoplasia or aplasia and supernumerary teeth are more typical, while generalized osteosclerosis and acro-osteolysis are not characteristic. Osteogenesis imperfecta with acro-osteolysis may be considered in patients with recurrent fractures, but it usually presents with osteopenia, blue sclerae, dentinogenesis imperfecta, and ligamentous laxity rather than diffuse osteosclerosis. In the present case, the association of generalized osteosclerosis, preserved hematopoiesis, open cranial sutures, mandibular hypoplasia, acro-osteolysis, recurrent fractures, and complete paranasal sinus agenesis strongly favored pycnodysostosis.

This case is noteworthy for several reasons. First, the patient was born to first-degree consanguineous parents, supporting the autosomal recessive inheritance pattern. Second, she exhibited the classical skeletal manifestations of the disease, including multiple spontaneous fractures and scoliosis. Most importantly, the diagnosis was made incidentally on imaging performed for persistent snoring, and CT revealed complete paranasal sinus agenesis, a radiological finding reported only exceptionally in the literature.

Table 1. Summary of key clinical, radiologic, and functional features of pycnodysostosis.

Feature	Pycnodysostosis
Genetics	CTSK mutation
Inheritance	Autosomal recessive
Key clinical signs	Short stature, fractures, craniofacial dysmorphism
Radiologic hallmarks	Osteosclerosis, open sutures, acro-osteolysis
ENT manifestations	Snoring, hearing loss
Differential diagnosis	Osteopetrosis, CMD, CCD

Table 2. Main differential diagnoses of pycnodysostosis.

Disorder	Similar features	Distinguishing features
Osteopetrosis	Diffuse osteosclerosis	Marrow failure, anemia, hepatosplenomegaly, cranial nerve compression, no acro-osteolysis
Craniometaphyseal dysplasia	Craniofacial hyperostosis	Metaphyseal widening, no typical acro-osteolysis
Cleidocranial dysplasia	Open sutures, dental anomalies	Clavicular hypoplasia/aplasia, supernumerary teeth, no diffuse osteosclerosis
Osteogenesis imperfecta	Recurrent fractures	Osteopenia, blue sclerae, dentinogenesis imperfecta, no osteosclerosis

This underscores that, although the diagnosis of pycnodysostosis is primarily clinical—based on its characteristic morphotype and patient history—the radiologist plays a crucial role in establishing the lesional assessment and confirming the diagnosis. Craniofacial CT provides the decisive structural details that characterize the syndrome and guide multidisciplinary management.

There is no curative treatment for pycnodysostosis; management is supportive and preventive [13]. Patients benefit from: trauma prevention and orthopedic monitoring, dental and maxillofacial follow-up to prevent osteomyelitis, and correction of vitamin D deficiency to optimize bone metabolism.

With appropriate multidisciplinary care, life expectancy and cognitive development remain normal, although fractures may recur throughout life.

Conclusion

Pycnodysostosis is a rare but recognizable skeletal dysplasia whose diagnosis relies heavily on careful clinical and radiologic correlation. In our case, the condition was revealed incidentally during evaluation for persistent snoring, highlighting that upper airway symptoms may reflect underlying craniofacial structural abnormalities. The association of recurrent fractures, distinctive craniofacial morphology, acro-osteolysis, complete paranasal sinus agenesis, and mastoid sclerosis leading to conductive hearing loss is particularly characteristic. CT imaging plays a central role, not only in confirming osteosclerotic changes but also in revealing airway and auditory consequences that may explain persistent ORL symptoms. Early recognition is essential to allow genetic confirmation, family counseling, and preventive orthopedic and maxillofacial management, enabling affected individuals to maintain normal life expectancy and cognitive function despite lifelong bone fragility.

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