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(CASE REPORT)

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# Pycnodysostosis: A rare case report

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## Abstract

Pycnodysostosis is a rare genetic disorder characterized by dense but brittle bones, short stature, and dental anomalies. This review summarizes the clinical presentation, genetic basis, diagnostic methods, and management strategies for pycnodysostosis. Early recognition and multidisciplinary care are crucial for optimizing outcomes and improving the quality of life for affected individuals. Further research into the pathophysiology of pycnodysostosis may lead to novel therapeutic approaches. We report a case of pycnodysostosis in a 23-year-old male sustaining a low energy right femoral shaft fracture.

**Keywords:** Bone Density; Short Stature; Genetic Disorder; Dental Abnormalities.

## 1. Introduction

Pycnodysostosis is an exceedingly rare genetic disorder characterized by dense but fragile bones, short stature, and dental abnormalities. It poses significant challenges in diagnosis and management due to its diverse clinical manifestations and skeletal abnormalities. Treatment is primarily supportive, focusing on managing fractures, correcting bone deformities, and addressing dental issues. Although there is no cure for pycnodysostosis, early diagnosis and appropriate management can improve quality of life for affected individuals. Further research into the genetic mechanisms of pycnodysostosis may lead to the development of targeted therapies in the future.

This review aims to provide a comprehensive overview of pycnodysostosis, highlighting its clinical features, genetic basis, diagnostic approaches, and treatment options.

## 2. Case presentation

We report a case of a 23-year-old male presented at our Emergency Department with right femoral fracture which occurred after a minor fall with the complaints of hip pain and the inability to walk. Physical examination found an intact skin without neurovascular injury, radiographs revealed a right femoral shaft spiral fracture. [Figure 1]

Also, his antecedents included a consanguineous first-degree relative and repeated spontaneous fractures, predominantly in the two tibias, since the age of 3 years. Examination revealed a dysmorphic syndrome with frontal hump, micrognathia, finger malformations, dental malposition with multiple caries, curved nails, asymmetrical thorax, scoliotic attitude of the dorsal spine. Skeletal X-rays showed densification of the skull base bones [Figure 2], dental malposition, diaphyseal and metaphyseal densification of the long bones, predominantly in the lower limbs with the presence of malunited right tibial fracture, and small phalanges of the hands [Figure 3]. Bone densitometry was normal.

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Given the clinical signs and radiological manifestations, the diagnosis was pycnodysostosis. Genetic counseling was offered to the family, as well as dental and orthopedic management.

The patient underwent delicate internal osteosynthesis. Therefore, drilling was highly difficult and time-consuming, also one of the drill tips was bent. However, osteosynthesis was successfully performed using femoral plate without any other complications.

A short- and long-term follow-up of two years showed a complete femoral fracture consolidation. [Figure 4].

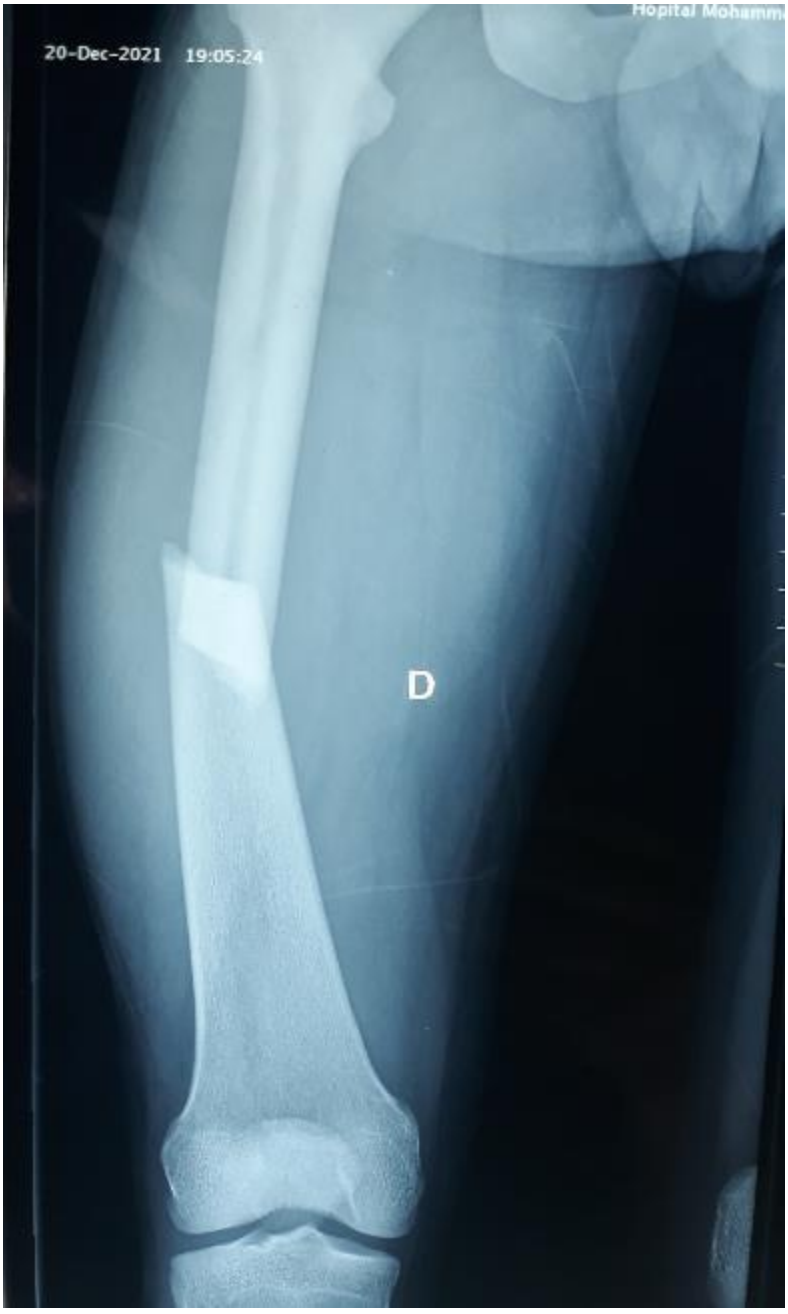


**Figure 1** X-rays showing right femoral spiral shaft fracture

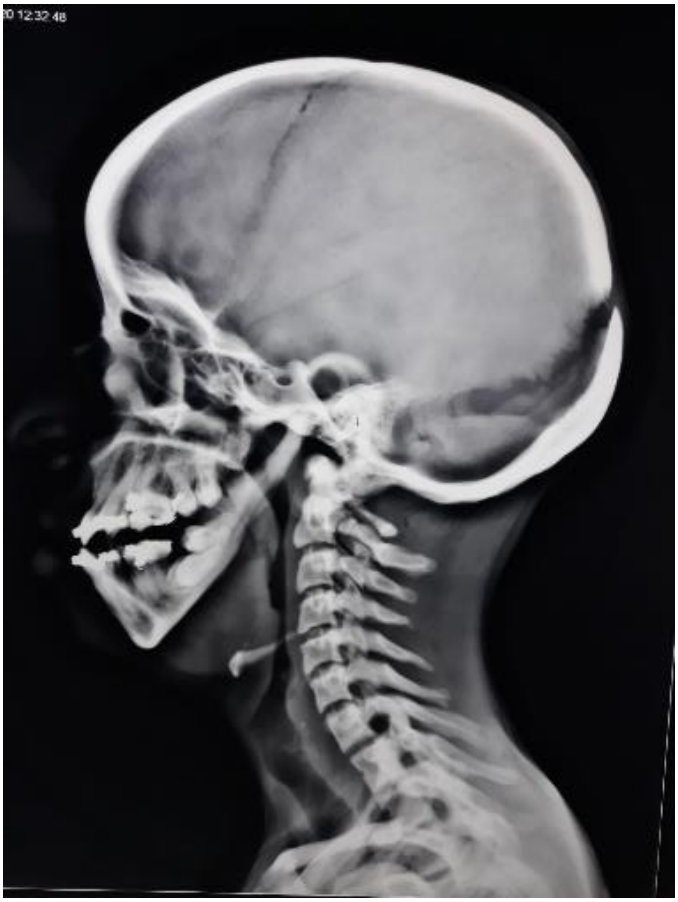

**Figure 2** Radiographs showing densification of the skull base bones

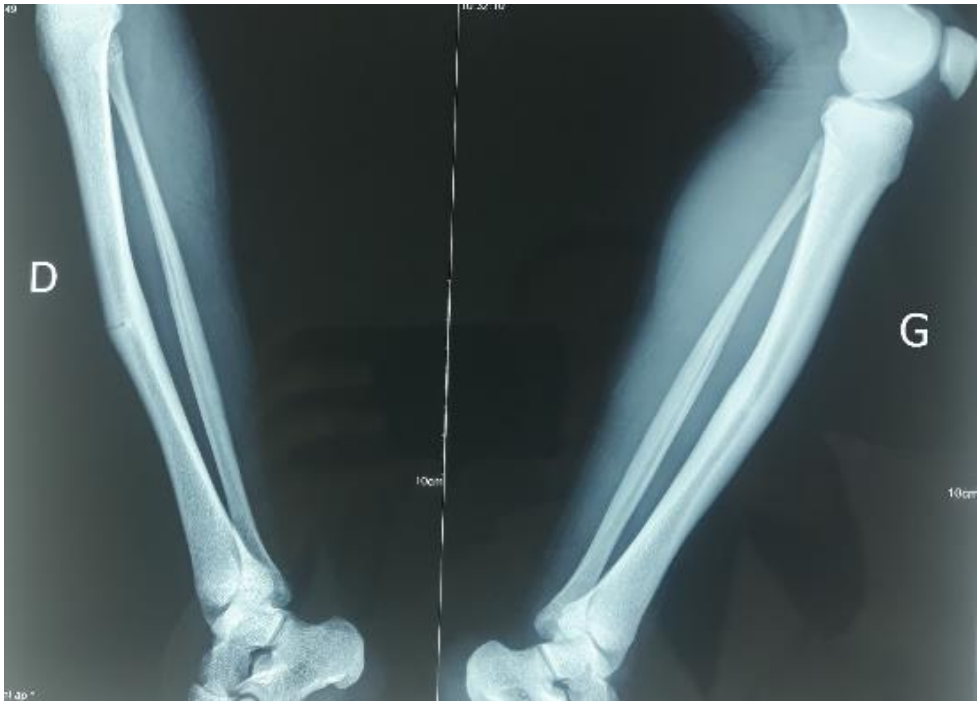


**Figure 3** X-rays showing densification of tibial bones with the presence of malunited right tibial shaft fracture

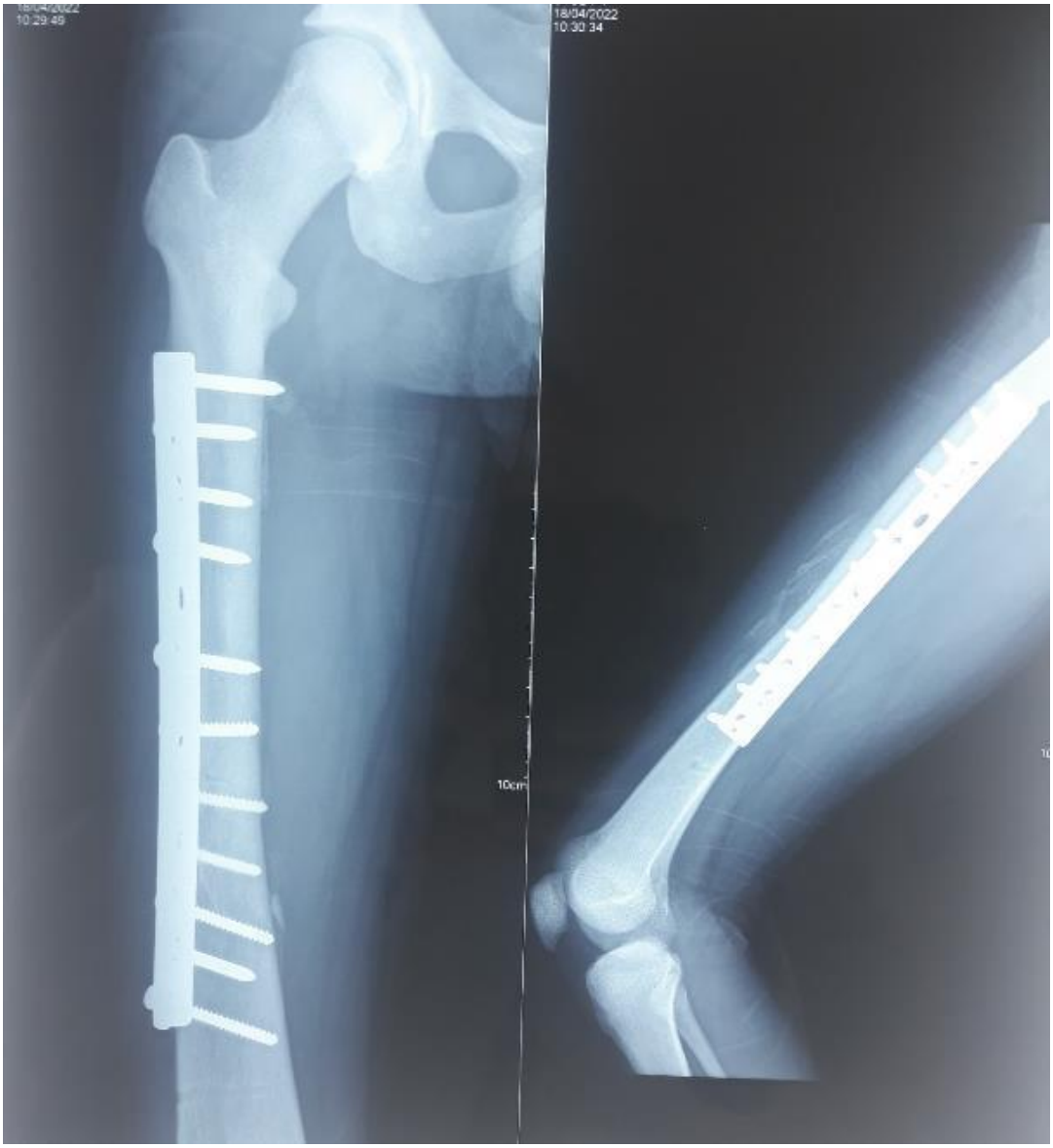


**Figure 4** Post-operative X-rays showing femoral fracture osteosynthesis**.**

## 3. Discussion

Pycnodysostosis is a rare genetic disorder that was first described in 1962 by Maroteaux and Lamy, and since then, our understanding of its pathophysiology has improved significantly. Pycnodysostosis is caused by mutations in the gene encoding cathepsin K, a lysosomal protease enzyme crucial for bone remodeling. The characteristic features of pycnodysostosis include dense but fragile bones, short stature, and dental anomalies. Diagnosis is based on clinical evaluation, radiological findings, and genetic testing. It is also characterized by clinically minor trauma-related fractures [1,2]

The management of pycnodysostosis presents several challenges due to the complexity of the disorder. Internal fixation can be technical challenging due to increased bone density in such diseases, furthermore a multidisciplinary approach is essential involving orthopedic surgeons, dentists, geneticists, and other healthcare professionals to address the various aspects of the condition. Early diagnosis is crucial as it allows for timely interventions to prevent complications and improve the quality of life for affected individuals [3].

Orthopedic management focuses on the prevention and treatment of fractures, which are common in pycnodysostosis. Prompt recognition and stabilization of fractures are essential to minimize pain, deformity, and functional impairment [4]. Surgical interventions such as bone grafting or osteotomy may be necessary in severe cases to correct bone

deformities and improve mobility [5]. Dental care is another important aspect of managing pycnodysostosis. Individuals with pycnodysostosis often experience dental abnormalities, including overcrowding of teeth and delayed eruption. Regular dental visits are essential to monitor these issues and prevent complications such as dental caries and periodontal disease [6].

Genetic counseling is recommended for individuals with pycnodysostosis and their families to understand the genetic basis of the disorder and the risk of passing it on to future generations [7]. Advances in genetic testing have improved the accuracy of diagnosing pycnodysostosis and have facilitated prenatal diagnosis in some cases [8].

Despite these challenges, the prognosis for individuals with pycnodysostosis is generally favorable with appropriate management. With ongoing research into the underlying genetic mechanisms of pycnodysostosis, there is hope for the development of more targeted therapies that can further improve outcomes and quality of life for affected individuals [9,10].

## 4. Conclusion

Pycnodysostosis is a rare genetic disorder that requires a multidisciplinary approach for effective management. Early diagnosis, orthopedic interventions, dental care, and genetic counseling are key components of the management strategy. Continued research is essential to advance our understanding of pycnodysostosis and improve outcomes for individuals affected by this condition.

## Compliance with ethical standards

### *Disclosure of conflict of interest*

No conflict of interest to be disclosed.

### *Sources of funding*

This study was not supported by any institution and company.

### *Statement of informed consent*

Written informed consent was obtained from the patient for publication of this case report.

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