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Challenges in the Surgical Orthopedic Treatment of Long Bone Fracture in a Rare Case of Pycnodysostosis

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Abstract **Case Report**

Pycnodysostosis is a rare autosomal recessive disorder caused by an inactivating mutation in cathepsin K (CTSK) and characterized by dysmorphic facial features, a short stature, acro-osteolysis, osteosclerosis with increased bone fragility, and delayed closure of cranial sutures. Although the satisfying healing potential, many issues exist in managing fractures in pycnodysostosis patients. In this paper, we report the challenges faced in managing a fracture of the femoral shaft in a 14-year-old boy coming from a low-income family. The patient was administered to the Paediatric Surgical Emergency Department of the Children's Hospital in Rabat for the management of a closed fracture of the left femur. Despite being of small stature with dysmorphic facial features, a prominent forehead, and brachydactyly, he never had a medical consultation before. The patient then underwent a surgical reduction. We attempted titanium elastic nail fixation, but metaphyseal drilling was not feasible due to the high bone density, and the narrow medullary canal eliminated this option. Therefore we opted for the palate fixation. Sclerotic bone made drilling extremely difficult. The postoperative fracture was successfully healed.

Keywords: Pycnodysostosis, osteosclerosis, short stature, locking compression plate.

INTRODUCTION

Pycnodysostosis, also known as Toulouse-Lautrec syndrome, is an uncommon hereditary autosomal recessive lysosomal disease causing skeletal dysplasia [1, 2]. The prevalence is 1/1.700.000 [3, 4]. Pycnodysostosis is caused by homozygosity or compound heterozygosity for biallelic pathogenic mutations in the cathepsin K gene (CTSK), a lysosomal metalloproteinase highly expressed in osteoclasts and has a role in bone matrix degradation. The skeleton is characterized by osteosclerosis associated with increased bone fragility [5]. Given the rarity of the disease, guidelines on fracture pycnodysostosis are unavailable. As far as we know, we are reporting an uncommon case from the Moroccan population.

CASE PRESENTATION

We report the case of a 14-year-old boy, from a low-income family, who suffered from a low-energy thigh injury. The patient presented with severe pain and could not bear weight on the affected limb. The physical examination reveals short stature with dysmorphic facial (Figure 1: A) features including a prominent forehead, double-rowed teeth, a grooved palate (Figure 1: B), and brachydactyly (Figure 2). Additionally, there is no history of consanguinity, and the patient has never experienced any fractures or had any previous medical examination. The X-ray showed a transversal midshaft femur fracture, with a high-density bone, and a narrow medullary canal (Figure 3, 4). Under general anesthesia, we attempted titanium elastic nail fixation, but metaphyseal drilling was not feasible due to osteosclerotic bone. Then, we opted for open reduction and internal fixation with a 4.5 locking compression plate (LCP) (Figure 5), and non-locking screws drilling the hole was the most difficult part of the procedure, due to the osteosclerotic nature of the bone. To prevent thermal necrosis, continuous cold saline irrigation was done. We also took necessary precautions to avoid iatrogenic fractures. Retrospectively, we made a detailed evaluation of the child, with radiological evaluation showing an obtuse angled mandible and short distal phalanx in a few digits (Figure 6), which established the diagnosis of pycnodystosis. The postoperative period was uneventful, and the patient was discharged after five days.

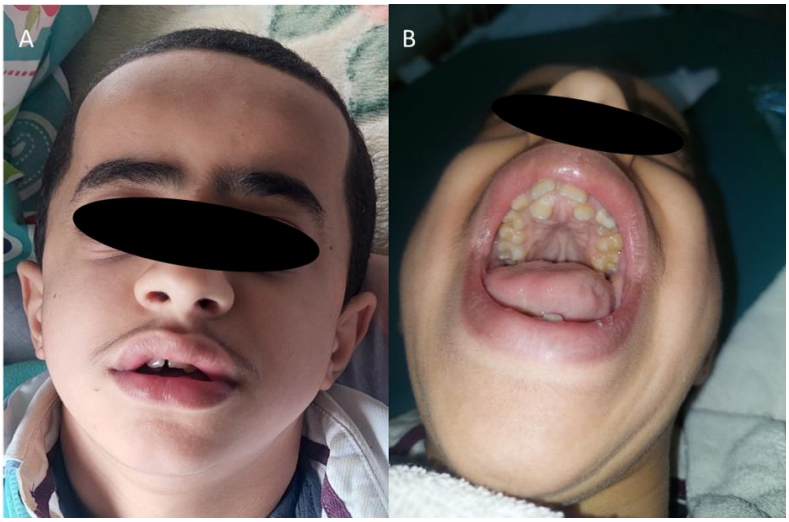

Figure 1: Image Showing: (A) Facial dysmorphism (B) Grooved palate and irregular dentition

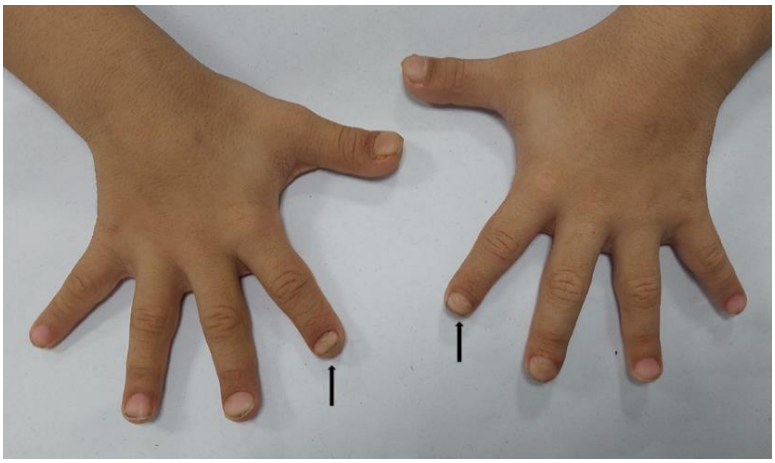

Figure 2: Image showing small fingers and grooved nails

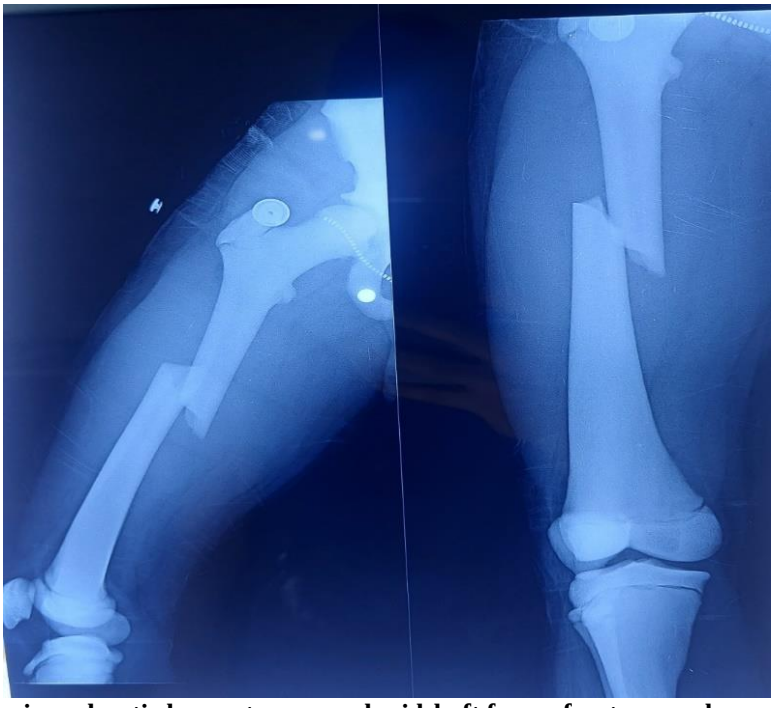

Figure 3: X-ray showing sclerotic bones, transversal midshaft femur fracture, and narrow medullary canal

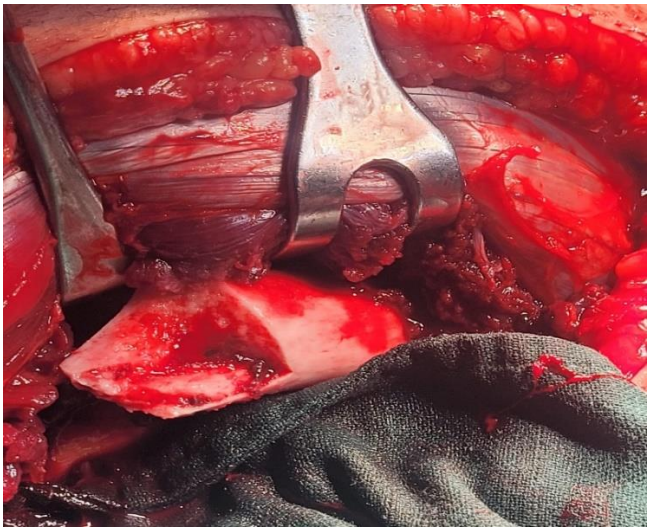

Figure 4: Perioperative images showing a narrow medullary canal

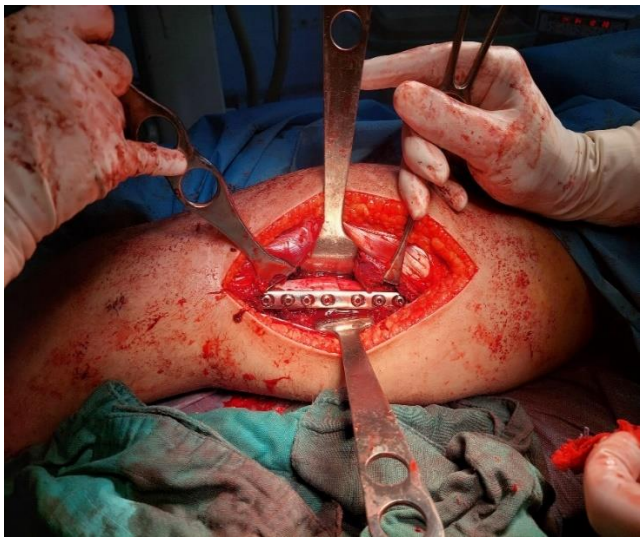

Figure 5: Perioperative images showing internal fixation with a 4.5 locking compression plate (LCP)

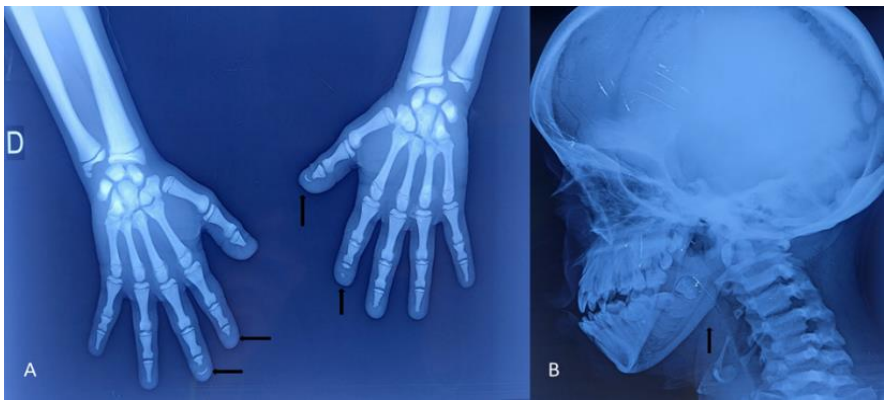

Figure 6: (A) Bilateral hand X-ray showing short distal phalanx (B) Black arrow showing the obtuse angle of the mandible

DISCUSSION

Pycnodysostosis is an uncommon genetic autosomal recessive lysosomal storage disorder that causes skeletal dysplasia, caused by a deficiency in the cathepsin K (CTSK) gene [6]. Cathepsin K is a key enzyme generated locally by osteoclastsis, which is responsible for the breakdown of collagen type I, and makes up 95% of the organic bone matrix [7]. This osteoclast deficiency causes inadequate bone turnover, resulting in weak and brittle bones. This condition is characterized by short stature, osteosclerosis,

micrognathia, open fontanelles, uneven teeth with hypodontia, grooved palate, obtuse angle of the jaw, acroosteolysis of the terminal phalanges resulting in short fingertips, and grooved nails [8]. Frequent fractures, sclerotic and malformed bones, as well as limited medullary canals are key considerations for orthopedics [9, 10].

For patients with pycnodysostosis, Grewal *et al.*, [9] and Romans *et al.*, [10] suggest that the ideal fracture treatment strategy should involve open reduction combined with rigid internal fixation. There was no intramedullary fixing because of the intramedullary canal's extremely small diameter. However, this is also not an easy process to follow. Due to the challenging sclerotic bone drilling, a suitable quantity of sharp and powerful drill bits is required. This process is time-consuming and might lead to thermal necrosis, causing additional complications. In addition, bone deformation can make it more difficult to place a plate over it. By bringing these concerns to our attention preoperatively, we can better prepare for these intraoperative issues. Before the surgery in this uncommon situation, we should always reflect on a few factors, including selecting the appropriate implant, keeping a sufficient supply of sharp drill bits—preferably newer ones—and providing ample saline irrigation while drilling. In a related osteopetrosis case report, Kumar *et al.*, emphasized the need for individualized treatment plans incorporating detailed preoperative planning to improve patient outcomes [11].

CONCLUSION

The diagnostic suspicion of pycnodysostosis arises in the presence of short stature, skeletal dysmorphism, and a history of repeated fractures. The approach for patients with pycnodysostosis must be multidisciplinary. Orthopedic surgeons must take into account, in their pre-operative planning, the multiple technical difficulties they may encounter concerning the particular bone, which has a marble-like consistency and is fragile at the same time, to ensure the presence of suitable instruments in the operating room.

To date, there are no standardized guidelines. Despite the rarity of this condition, further studies should be carried out to define the optimal surgical management not only of fractures but also of the frequent cases of refracture.

Declaration of Patient Consent: The authors certify that they have obtained all appropriate patient consent forms.

Conflicts of Interest: The authors declare no conflicts of interest.

Author Contributions: All authors participated in this work. All have read and approved the final version of the manuscript.

REFERENCES

1. Gelb, B. D., Shi, G. P., Chapman, H. A., & Desnick, R. J. (1996). Pycnodysostosis is a lysosomal disease caused by cathepsin K deficiency (in Eng). *Science (New York, NY), 273*, 1236–1238.
2. Polymeropoulos, M. H., Ortiz De Luna, R. I., Ide, S. E., Torres, R., Rubenstein, J., & Francomano, C. A. (1995). The gene for pycnodysostosis maps to human chromosome 1cen–q21. *Nature genetics*, *10*(2), 238-239. https://doi.org/10.1038/ng0695-238
3. Xue, Y., Cai, T., Shi, S., Wang, W., Zhang, Y., Mao, T., & Duan, X. (2011). Clinical and animal research findings in pycnodysostosis and gene mutations of cathepsin K from 1996 to 2011. *Orphanet journal of rare diseases*, *6*, 1-10.
4. Song, H. Y., Sohn, Y. B., Choi, Y. J., Chung, Y. S., & Jang, J. H. (2017). A case report of pycnodysostosis with atypical femur fracture diagnosed by next-generation sequencing of candidate genes. *Medicine*, *96*(e6367), 12.
5. Arman, A., Bereket, A., Coker, A., Kiper, P. Ö. Ş., Güran, T., Özkan, B., ... & Turan, S. (2014). Cathepsin K analysis in a pycnodysostosis cohort: demographic, genotypic and phenotypic features. *Orphanet journal of rare diseases*, *9*, 1-8.
6. Turan, S. (2014). Current research on pycnodysostosis. *Intractable Rare Dis Res*, *3*, 91e93.
7. Drake, M. T., Clarke, B. L., Oursler, M. J., & Khosla, S. (2017). Cathepsin K inhibitors for osteoporosis: biology, potential clinical utility, and lessons learned (in eng). *Endocr Rev*, *38*, 325–350. https://doi.org/10.1210/er.2015-1114.
8. Rodrigues, C., Gomes, F. A., Arruda, J. A., Silva, L., Álvares, P., da Fonte, P., ... & Silveira, M. (2017). Clinical and radiographic features of pycnodysostosis: a case report. *Journal of clinical and experimental dentistry*, *9*(10), e1276- e1281.
9. Grewal, S., Kilic, Ö., Savci-Heijink, C. D., & Kloen, P. (2019). Disturbed remodeling and delayed fracture healing in pediatric pycnodysostosis patients. *J Orthop*, *16*(5), 373- 377.
10. Romans, M., Sambandam, B., Moses, J., & Prakash, M. (2020). A rare case of pycnodysostosis: Technical difficulties in managing long bone fractures. *J Clin Orthop Trauma*, *11*(2), 332-338. doi: 10.1016/j.jcot.2018.09.012. Epub 2018 Sep 26. PMID: 32099307; PMCID: PMC7026621.
11. Kumar, D., Jain, V. K., Lal, H., Arya, R. K., & Sinha, S. (2012). Metachronous bilateral subtrochanteric fracture of femur in an osteoporotic bone: a case report with a technical note. *JCOT* [serial online]; *3*(2), 103-106.