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Pycnodysostosis: A Rare Case of Pathological Fractures

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Abstract: ***Pycnodysostosis is a rare autosomal recessive disorder marked by short stature, distinctive facial characteristics, increased bone fragility, and acro - osteolysis. This case study describes a 50 - year - old female presenting with multiple pathological fractures and characteristic facial features, confirmed by genetic testing identifying a mutation in the CTSK gene. Radiological investigations revealed fractures in the tibia, fibula, and ribs. The patient was managed with bisphosphonates and supportive measures. This case highlights the importance of considering pycnodysostosis in patients with recurrent fractures and distinct craniofacial abnormalities to ensure timely diagnosis and appropriate management. Significance of the article: This article is significant because it documents a rare presentation of pycnodysostosis providing valuable insights for clinicians in diagnosing and managing similar cases. It emphasizes the need for awareness of atypical presentations to ensure timely and appropriate treatment. Purpose of article: The purpose of this article is to report a case of pycnodysostosis presenting as multiple fractures and to highlight the importance of considering this rare condition in the differential diagnosis of patients with similar symptoms.***

Keywords: pycnodysostosis, pathological fractures, genetic disorder, bisphosphonates, acro - osteolysis

1. Introduction

Pycnodysostosis is a rare genetic disorder caused by mutations in the cathepsin K gene leading to impaired bone remodelling and resorption. (1) Cathepsin K is important for normal bone cells called osteoclasts to reabsorb and build new bones (2). Thus this leads to inadequate resorption of bone matrix. This results in skeletal abnormalities, including short stature, abnormally shaped skull and fragile bones prone to fractures. (2)

Individuals with pycnodysostosis often present with distinct physical features such as prominent forehead, underdeveloped mid face and dental anomolies. They may also experience functional impairments including restricted joint mobility, skeletal deformities, and abnormal curvature of spine. (1)

Due to rarity of pycnodysostosis there is limited understanding of its pathophysiology and optimal management strategies. Further research is needed to develop effective interventions and improve support for individuals living with this condition.

2. Case Report

A 50 year old female patient was referred with H/O multiple fractures since childhood. During clinical examination it was noted that patient had a short stature with height weight. Patient had difficulty in walking since childhood. Patient is a recently operated case of ORIF of right tibia.

On general examination, she was conscious and oriented to time, place, and person, with all higher mental functions intact. She had normal IQ.

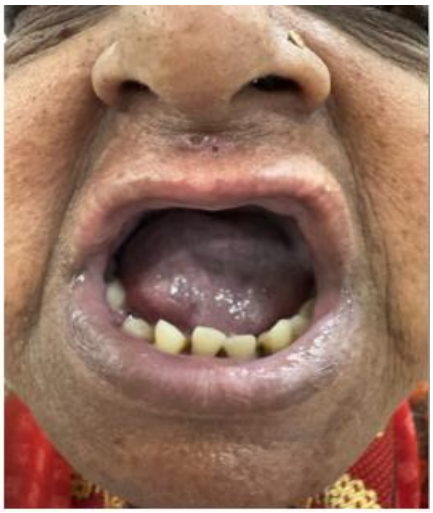
Pulse was 74 bpm, blood pressure was 120/80 mmHg, and SpO2 was 99. All other systemic examinations were within normal limits.

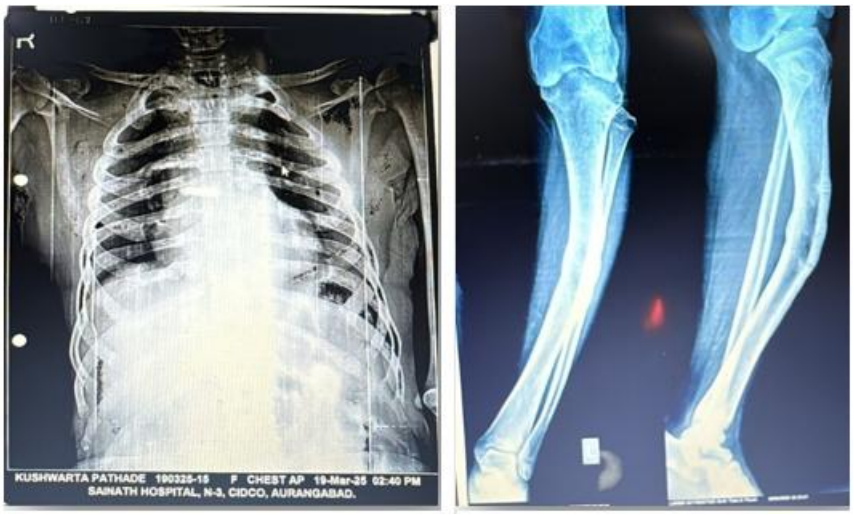
Patient had clinical features of bilateral exophthalmos, facial asymmetry, frontal bossing and micrognathia. Intraoral examination revealed mandible prognathism with absent incisors, presence of canines and dental crowding.

Physical examination of hands showed shortened fingers with spoon shaped nails, giving drumstick appearance. Patient also had hearing loss since the age of 20 years.

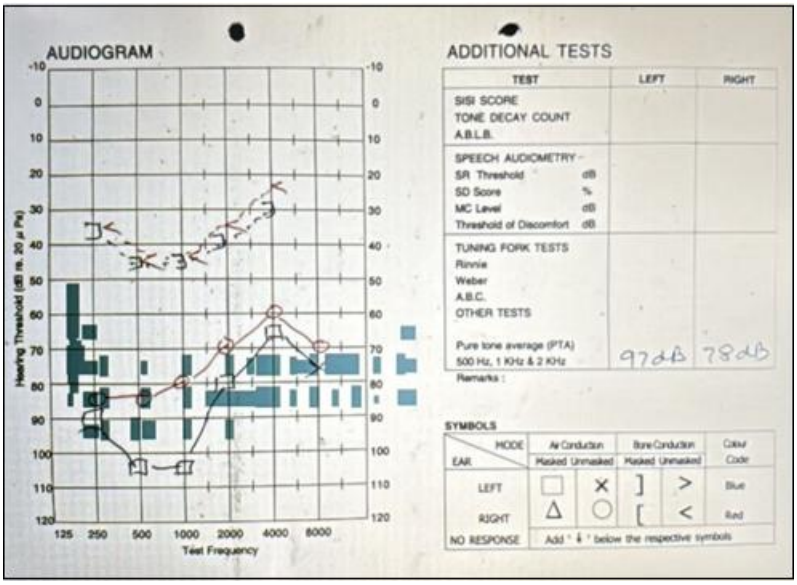
Patient had history of multiple pathological fractures secondary to mild trauma since childhood & she was managed for same with operative procedures.

Clinical Photographs:





Chest X - ray Both Lower limb Xray



Pure tone audiometry suggestive of bilateral sensorineural hearing loss

When asked about family history there were no relatives with similar clinical features.

Calcium and phosphorous levels were evaluated and lab values showed no alterations. Same results were found regarding osteocalcin and ALP.

Chest X ray revealed multiple level fractures in multiple ribs. Right leg X ray revealed proximal 1/3rd tibia and fibula fracture.

ECG was suggestive of normal sinus rhythm.

Pure Tone Audiometry (PTA) was suggestive of bilateral sensorineural hearing loss.

Treatment:
Patient was started on tablet Alendronate 70 mg once weekly. Calcium and vit D supplementation were given. Growth hormone therapy remains the mainstay of the treatment in growing age group. Treatment is mostly supportive. It is important that care is taken to prevent or minimise tendencies for future fractures to occur.

3. Conclusion

In summary, pycnodysostosis remains a rare yet important differential diagnosis in patients with multiple pathological fractures and craniofacial anomalies. Early recognition and a multidisciplinary approach involving orthopaedic, genetic, and supportive care teams can improve patient outcomes and quality of life. (4)

References

[1] Shannon LeBlanc [1], Ravi Savarirayan [1]
[2] Margaret P Adam, Jerry Feldman, Ghayda M Mirzaa, Roberta A Pagon, Stephanie E Wallace, Anne Amemiya
[3] Arman A, Bereket A, Coker A, Özlem P, Kiper S, Güran T, Özkan B, Atay Z, Akçay T, Haliloglu B, Boduroglu K, Alanay Y, Turan S. Cathepsin K analysis in a pychnodysostosis cohort: demographic, genotypic and phenotypic features. Orphanet J Rare Dis.2014; 9: 60.
[4] Bertola D, Amaral C, Kim C, Albano L, Aguena M, Passos - Bueno M. Craniosynostosis in

pycnodysostosis: Broadening the spectrum of the cranial flat bone abnormalities. Am J Med Genet Part A.2010; 152A: 2599–603.

[5] Bizaoui V, Michot C, Baujat G, Amouroux C, Baron S, Capri Y, Cohen - Solal M, Collet C, Dieux A, Genevieve D, Isidor B, Monnot S, Rossi M, Rothenbuhler A, Schaefer E, Cormier - Daire V. Pycnodysostosis: Natural history and management guidelines from 27 French cases and a literature review. Clin Genet.2019; 96: 309–16.

[6] Campeau PM, Lu JT, Sule G, Jiang MM, Bae Y, Madan S, Högler W, Shaw NJ, Mumm S, Gibbs RA, Whyte MP, Lee BH. Whole - exome sequencing identifies mutations in the nucleoside transporter gene SLC29A3 in dysosteosclerosis, a form of osteopetrosis. Hum Mol Genet.2012; 21: 4904–9.

[7] Caracas HP, Figueiredo PS, Mestrinho HD, Acevedo AC, Leite AF. Pycnodysostosis with craniosynostosis: case report of the craniofacial and oral features. Clin Dysmorphol.2012; 21: 19–21.

[8] Grewal S, Kilic O, Savci - Heijink D, Kloen P. Disturbed remodeling and delayed fracture healing in pediatric pycnodysostosis patients. J Orthop.2019; 16: 373–7.