

**MANAGEMENT OF CLOSED SUBTROCHANTERIC FRACTURE OF LEFT FEMUR IN
A PATIENT WITH PYCNODYSTOSIS: A CASE REPORT****Tan Chen Liang* and Surinder Singh**

International Medical University, Kuala Lumpur, Malaysia.

***Corresponding Author: Tan Chen Liang**

International Medical University, Kuala Lumpur, Malaysia.

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ABSTRACT

Pycnodysostosis is a rare autosomal recessive disorder caused by a mutation in cathepsin K (CTCK) found in osteoclasts. Individuals with pycnodysostosis have characteristic clinical features such as short stature, stubby fingers and narrow palate. Radiologically, the bones appear to have a sclerotic cortex with relative sparing of the medullary canal, making surgical option for internal fixation of fracture rather difficult. This article reports a closed transverse left subtrochanteric fracture in a patient with pycnodysostosis that was managed conservatively.

KEYWORDS: Pycnodysostosis, clinical features, fracture, management.**INTRODUCTION**

Pycnodysostosis is a rare autosomal recessive disorder, which is also known by other names such as Toulouse-Lautrec syndrome and Maroteaux-Lamy syndrome.^[1,2,3] It is a lysosomal storage disease of the bone, primarily caused by inactivating mutation in cathepsin K (CTSK) found in osteoclasts which is a genetic defect located in chromosome 1q21. This causes impaired bone resorption and remodeling leading to generalised osteosclerosis.^[1,2,3,4,5]

It affects both males and females with prevalence rate of 1 in 1.7million and is characterized by the clinical findings of short stature, stubby fingers (osteolysis of distal phalanges), spondylolysis, frontal and parietal bossing, narrow palate, overcrowding of teeth and open fontanels. Radiologically, there are osteosclerotic bones with relative sparing of the medullary canals. Individuals with this disorder have high risk of fractures. The sclerotic cortex of their bone and narrow medullary canal makes surgical management of fractures difficult. However, hematopoietic function is preserved.^[1,2,3,4,5]

CASE REPORT

A 28 years old Malay woman presents with pain and swelling over left hip following a fall in a sitting position. She was previously ambulating with crutches and was unable to do so after the fall.

Past medical history revealed a history of pycnodysostosis which was diagnosed since young. She had a past history of fracture of her left femur in 2013, which resulted in malunion and angulation. Following

that she has been ambulating with crutches. She also has a history of fracture of her left tibia in 2015 which was also treated conservatively.

Upon examination, she is of a short stature, with frontal bossing, prominent eyes, dental crowding as well as a narrow palate. She had stubby fingers and feet with a sandal gap deformity.

X-ray pelvis, left hip and left femur were done. X-ray revealed a closed transverse subtrochanteric fracture of her left femur with an angulation of 60°. Her X-rays revealed bones with thickened cortices and narrow medullary canals.

She was given the option to undergo surgery for her fracture. However, she refused fixation of her fracture and was managed conservatively. She was advised that her fracture could go into nonunion or heal at an acute angle with an inherent risk of refracture on weight bearing.



Fig. 1: Closed transverse subtrochanteric fracture of left femur. Sclerotic cortex with narrow canal is also evident.



Fig. 2: Stubby fingers.

DISCUSSION

Fractures in individuals with pycnodysostosis is rather challenging as opposed to management of fractures in normal individuals.^[1,2,3,4,5] This is because the osteosclerotic cortex and narrow medullary canal makes canal reaming difficult if not impossible due to the hard cortex and unavailability of reamers for small canals.^[1,2,4] However, union after fixation is still possible as there is no compromise of hematopoiesis.^[1,2,3,4,5] Closed reduction and immobilization is also an option. However, immobilization with plaster cast may lead to pressure sores. Malunion is another complication of conservative management of fractures in these individuals, such as that which had occurred in this patient after conservative management of her previous fracture.^[4]

There have been reports on different managements that have been attempted to treat fractures in patients with pycnodysostosis. One patient with an atypical subtrochanteric fracture of her right femur was treated through internal fixation with a plate and screws. The fracture healed in 3 months and patient was able to bear full weight. However, one year later, she developed osteonecrosis of the femoral head which subsequently resulted in her undergoing a right total hip arthroplasty and was able to resume normal activities one year later.^[3]

In another report, a 30 years old lady with a displaced transverse fracture of her right femoral shaft was treated

with plate osteosynthesis due to pycnodysostosis. At 10 weeks, it was identified that there were no signs of union. Additional stabilization was done with plating and with autogenous cancellous bone grafting. However, the patient developed an infection at the surgical site which was treated aggressively and three months postoperatively, signs of union were noticed clinically and radiologically.^[5]

Moving on, a 6 years old boy with diaphyseal femur fracture was initially planned to be treated surgically with an elastic stable intramedullary nailing (ESIN). However, the cortical bone was too hard to penetrate and false routes were easily created owing to the narrow intramedullary cavity. Eventually, the boy was treated through closed reduction and spica plaster cast immobilization. The fracture was later stabilised using a wrist external device. Complete union was achieved clinically in 6 weeks.^[4]

It is clearly evident that surgical management still remains as an option for management of fractures in patients with pycnodysostosis. However, as the case report suggests, there are chances of non union in these patients especially as their bony anatomy has been altered, with a sclerotic cortex and narrow intramedullary canal.^[1,2,3,4,5] At the same time, conservative treatment has also proven to be effective as seen in the case of the 6 years old boy. However it is undeniable that such a treatment has posed a higher risk of complications such as infections and malunion especially in individuals of the older age group.^[4] However, the management of the fracture must also consider the altered anatomy of the patient.

CONCLUSION

Pycnodysostosis is a rare autosomal recessive disorder leading to a distorted bone matrix. It causes cortical sclerosis with relative sparing of the intramedullary canal, leading to difficulties due to the sclerotic cortex and narrow canal. For example, there were no reamers small enough for a narrow canal. Besides, false routes may be created and problems regarding union during casting may also arise. Surgical decisions should be made bearing in view the patient's condition and available expertise. It is best if these cases are managed by senior consultants.

The pictures were obtained with the consent of the patient. The patient was informed that it would be included in a report with assurance that her confidentiality would be preserved.

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