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**Case Report**

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# Pycnodysostosis with recurrent long bone fractures

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## ABSTRACT

Pycnodysostosis is a rare autosomal recessive sclerosing bone disorder characterized by generalized diffuse osteosclerosis. Patients usually have a large head with separated sutures, open fontanels, aplasia of frontal sinuses, obtuse mandibular gonial angle and acroosteolysis of distal phalanges and multiple long bone fractures. We report this case of a 30-year-old female with repeated multiple long bone fractures and other clinico-radiological pathognomonic features of pycnodysostosis.

**Keywords:** Pycnodysostosis, Pathological fractures, Proximal femoral nail

## INTRODUCTION

Pycnodysostosis is a rare autosomal recessive disorder due to mutation of cathepsin K gene causing osteosclerosis because of osteoclastic dysfunction.[1] This disorder is characterized by increased bone density, short and stubby fingers, dysplastic nails, fragile long bones that may fracture easily, craniofacial abnormalities and increased lumbar lordosis.[2] Radiographic features includes generalised osteosclerosis, dense vertebral bodies with sparing of transverse processes, failure of segmentation of lower lumbar or lumbosacral vertebra, widening of distal femur.[3] These patients usually present with recurrent fractures even after trivial trauma.

## CASE REPORT

A 30 year old female presented to us with subtrochanteric fracture of left femur following history of fall. She had past history of repeated fractures at multiple anatomical locations. She fractured right tibia at 7 years of age following healed on conservative management. She had similar history of fracture shaft of right femur when she was 24 years old (6 years ago) for which she was operated with open reduction and internal fixation with Kuntscher nail at another institute. She fractured her right tibia again at 28 years of age following trivial trauma. This was managed conservatively.

On physical examination there is a facial asymmetry and beaked nose, short little finger bilaterally, clawed toes of feet and hallux valgus left great toe, procurvatum deformity right leg, increased lumbar lordosis and high-arched palate (Figure 1-4).

Roentgen graphic examination showed blocked vertebrae of L5-S1, increased lumbar lordosis, diffuse osteosclerosis of vertebral body with characteristic sparing of transverse process, deviated nasal septum and non-pneumatised bilateral frontal sinuses, cortical sclerosis and narrowing of medullary canal of phalanges, metacarpal and carpal bones (Figure 5-7). United fracture of right femur was seen with diffuse cortical sclerosis and K nail *in situ* (Figure 8). Right tibia showed malunited fracture in diaphysis (Figure 9 (a and b)). Left femur showed subtrochanteric fracture with diffuse sclerosis with narrow medullary canal, coxa-vara and excessive

bowing of distal shaft in both sagittal and coronal planes (Figure 10 (a)).

Blood parameters including thyroid profile, parathyroid hormone and vitamin D were normal. Histopathology specimen collected from medullary canal intraoperatively reveals proliferative fibro collagenous tissue with degenerated osteoid material**.**

Our pre-operative plan was to open up the fracture site to enable us to obtain tissue for histopathological examination, reaming of the medullary canal and reduce the fracture for fixation with long proximal femoral nail (LPFN). Reamed nailing was planned to facilitate nail insertion considering gross cortical thickening and narrowing of the medullary canal. Authors also thought that the process of reaming may help in opening up the vascular channels in the pathological bone and thereby aid in bony union. LPFN was pre-operatively chosen as fixation device so as to take care of any probable future pathological fracture in the shaft as well as the neck of the affected femur.

Under spinal anaesthesia the patient was put in a fracture table. Fracture was opened through lateral approach and medullary canal was found to be obliterated. First, drill bits of size 3.2 and 4 mm were used to open up the obliterated medullary canal. To widen the medullary canal further, reaming was done with increasing reamer up to size 11 mm. Reamed out contents of medullary canal were collected and sent for histopathological examination. Due to excessive bowing of the femur in sagittal and coronal planes the reamer was found to be hitting the anterior-lateral cortex. Though we tried to insert a LPFN it was not feasible as there was a tendency of the distal nail tip to abut the anterolateral cortex due to excessive bowing at distal femur. Hence intraoperatively we changed our plan and fixed it with a short proximal femoral nail (10×25), (Figure 10 (b)). The hard cortical bone made drilling difficult and resulted in breakage of the drill bit and splitting of far cortex at the site of proximal locking bolt at the distal end. Post operatively on day two patient was advised to do active knee bending and static quadriceps exercises. Weight bearing was delayed as there was splitting of far cortex at the distal end.


**Figure 1 (a, b): Beaked nose and prominent maxillary prominence left side and facial asymmetry.**

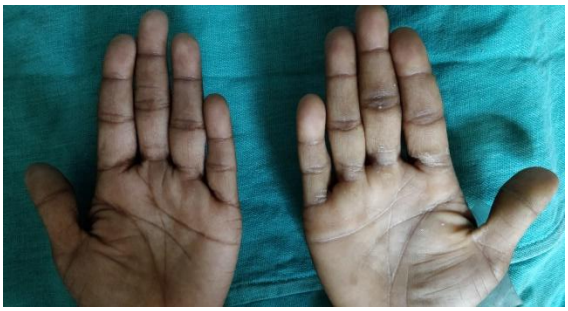

**Figure 2: Short little finger bilaterally.**

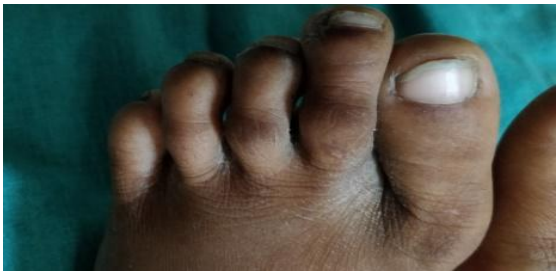

**Figure 3: Clawed toes of feet and Hallux valgus left.**

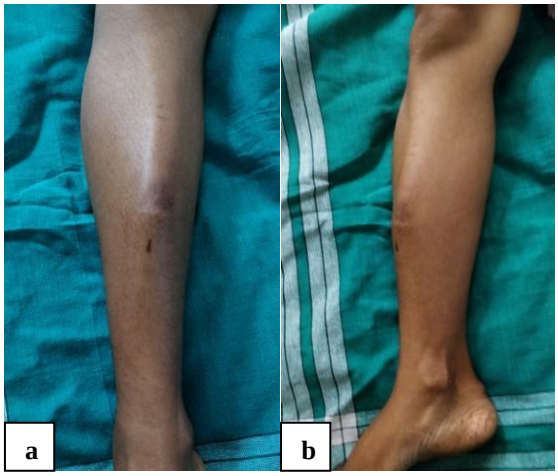


**Figure 4 (a and b): Procurvatum deformity right leg.**

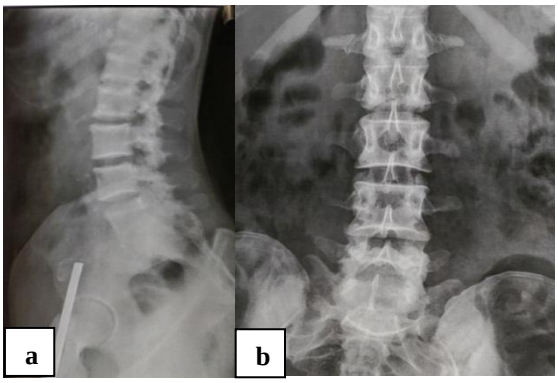


**Figure 5: Vertebral features: (a) blocked L5-S1 vertebra and exaggerated lumbar lumbar lordosis and (b) diffuse osteosclerosis of vertebral body with sparing of transverse process.**

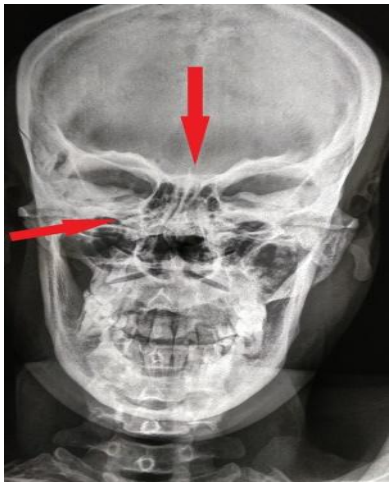

**Figure 6: Deviated nasal septum, non pneumatised frontal sinuses.**

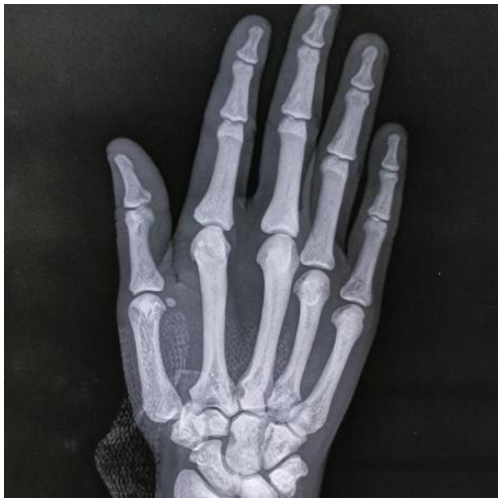

**Figure 7: Diffuse sclerosis and narrowed medullary canal of phalanges, metacarpal, carpal bones.**

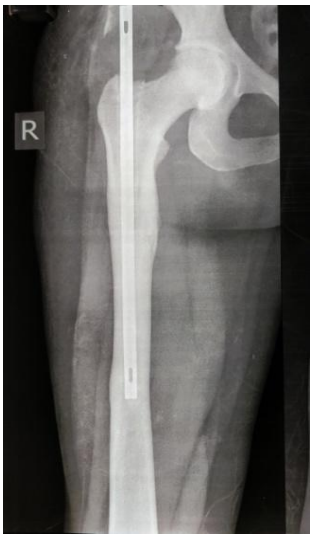


**Figure 8: United fracture of right femur was seen with diffuse cortical sclerosis and k nail *in situ*.**

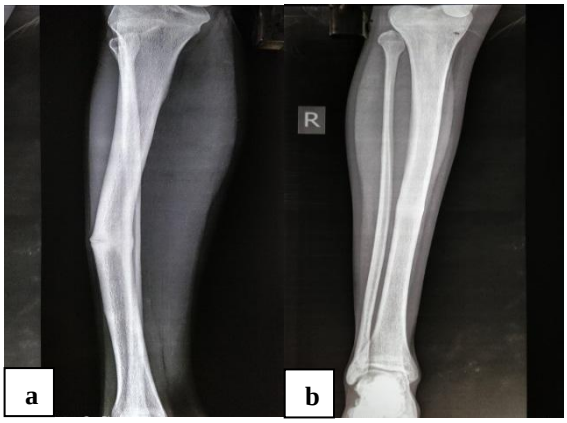


**Figure 9 (a and b): Right tibia showed malunited fracture in diaphysis.**

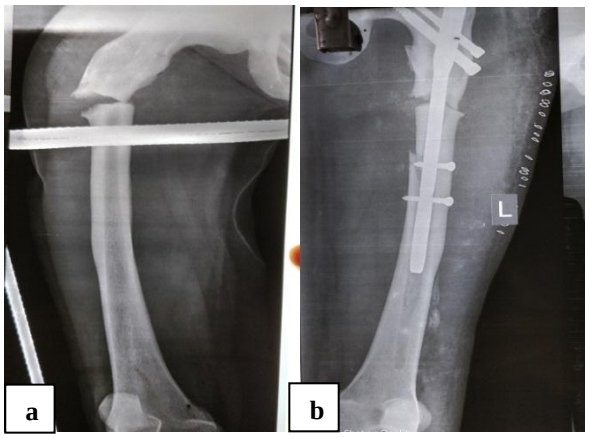


**Figure 10: (a) Left femur showed subtrochanteric fracture with diffuse sclerosis with narrow medullary canal, coxa-vara and excessive bowing of distal shaft in both sagittal and coronal planes and (b) subtrochanter fracture left femur fixed with short PFN and excessive bowing in distal shaft of femur in coronal plane.**

## DISCUSSION

Pyknodysostosis is an rare autosomal recessive disorder of bone, causing osteoclast dysfunction resulting in osteosclerosis. Cases have been reported in Negroes, Arabs, Jews and Caucasian population .Patients of age group ranging from 9 months to 50 years have been reported.[4] The estimated prevalence is 1–1.7 per million. The sclerosing activity of pyknodysostosis is due to a genetic defect on chromosome 1q21 lead to mutational changes in a lysosomal cystine protease, CTSK, the expression of which is reduced in the osteoclasts. This protease is responsible for degrading type 1 collagen that constitutes 95% of the organic bone matrix. The affected bones are abnormally dense and brittle as a result of insufficient resorption causing pathological long bone fractures.[4-5]

The clinic-radiological diagnostic features found in our case have been described in the literature by various

authors. Our case had typical history of recurrent fractures of multiple long bones. Cranial and maxillofacial features reported in our case included beaked nose, facial asymmetry with maxillary prominence on left side.[6] Other features present were finger and nail abnormalities in phalanges and intraoral features like high arched palate which have been reported by Hunt et al and Schmitz et al.[7,8] And our case had hypoplastic frontal sinuses as reported by Alves-Pereira et al and Landa et al.[9,10] Our case also had clawed toes bilaterally and hallux valgus in left side. These skeletal abnormalities, however, have not been described by other authors. Hence the association of these features with pyknodysostosis can neither be acknowledged nor refuted.

The radiological findings in our case showed excessive anterior-lateral bowing of the distal femur, failure of complete segmentation of the lumbosacral spine i.e., block vertebra L5-S1, increased Lumbar lordosis and diffuse osteosclerosis of vertebral body with sparing of transverse process. The same features were reported in the study by Maroteaux et al and Fleming et al.[5,11]

Coxa valga was reported by Maroteaux et al but our case had coxa vara in the left femur.[11] Cortical thickening and narrowing of medullary canal in the metacarpal and phalanges were present in this case, however, these finding in these bones have not been reported in the literature.

The differential diagnosis of pyknodysostosis is made with osteopetrosis, cleidocranial dysplasia and idiopathic acro-osteolysis. In osteopetrosis, an autosomal dominant disorder, the bone marrow may be absent but had organomegaly and myelophthisic type anaemia. Signs of compression of the cranial nerves like pain, facial palsy, deafness may exist. Our patient had no anaemia or organomegaly. Cleidocranial dysplasia had presentation of agenesis or clavicular aplasia as well as alterations of the skeletal bone membranes like pyknodysostosis however bone density is not increased. In idiopathic acro-osteolysis, the appearance of the patients is typical, with exophthalmos, hypotelorism and beaked nose. The angle of the mandible is acute but increased bone density is not present.[10]

The diagnosis is primarily based on clinical features and radiographs .The analysis of CTSK gene mutation is the gold standard confirmatory test but it was not done in our case because of non-availability at our institute.[12] The prognosis of the disease is good and no other serious systemic complications have been reported. Life expectancy for the affected patient is normal.[13]

## CONCLUSION

The importance of early recognition of clinical features in the diagnosis allows correct treatment, planning and reduces the chance of complications in future, thus ensuring a better quality of life to the patient. The therapeutic approach should prevent complications and fracture injuries with exercise and activities that are safe and do not require too much impact.

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