



Proceeding

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Pycnodysostosis: A Rare Cause of Short Stature, A Case Report

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Introduction

Pycnodysostosis is a rare genetic lysosomal storage disease characterized by osteosclerosis of the skeleton, wide anterior fontanelle, short stature and bone fragility. We aim to highlight the clinical and radiological features of pycnodysostosis.

Case Presentation

A 3 year old girl presented with short stature. She was the product of a consanguineous marriage. Clinical examination revealed severe short stature and dysmorphic features, with short large hands and feet with dystrophic nails. The skull was large with frontal and occipital prominence and wide open anterior fontanelle. The mandible was hypoplastic. The teeth were decayed. She had permanently obstructed nostrils.

Radiological examination showed a generalized increase in bone density without fault modelling of metaphyseal bone. The phalanges were short with acro-osteolysis of the distal phalanx of the second finger. There was increased density of the skull base,

wide coronal sutures with hypoplastic facial bones. The lower jaw was poorly developed and the mandibular angle was obtuse. Radiograph of the dorso-lumbar spine showed a spondylolisthesis.

Discussion

Pycnodysostosis is a rare autosomal recessive osteosclerosing bone disorder, first described by Maroteaux and Lamy in 1962. Pycnodysostosis is characterized by proportionate short stature, wide open sutures and fontanelles, short stubby fingers, hypoplastic distal phalanges, retrograde mandible, bone fragility with multiple fractures and osteosclerosis.

Conclusion

We have presented a case of Pycnodysostosis which presented with short stature. Early diagnosis is essential for appropriate management of bone deformity and its complications. Appropriate counselling of the patient and family is important to prevent osteosclerosis related complications.



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