CASE REPORT

Pycnodysostosis: Report of A Rare Case

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ABSTRACT

A 20-year old male patient reported in the outpatient department of Shaheed Suhrawardy Medical College Hospital, Dhaka, Bangladesh, with the complaints of body aches along with pain in the upper and lower limbs for several years. He also gave history of previous hospitalization for the same reason. He also had complaints of failure to thrive in his adolescence. However, he was treated as a case of polyarthritis in his previous admission in the district hospital. On clinical examination, he was found to have short stature, low-weight, peculiar face, prominent forehead, receding jaw and partial dysplasia of the terminal phalanges. He was sent for radiological investigations in the Department of Radiology & Imaging of the same hospital. Plain radiograph of his skull showed brachycephaly with wide sutures and persistent fontanelles with sclerotic changes in the base of skull and orbital rims. The angle of the mandible was obtuse, and the maxilla was found hypoplastic. Lumbar region showed 'spool-shaped' vertebral bodies with quite prominent anterior defects; bones had sclerotic changes. Both hands had acro-osteolysis of distal phalanges of thumb, index, middle and ring fingers (both right and left sides) along with irregular distal fragments; bones were sclerotic, too. Those features were consistent with pycnodysostosis. The patient was kept admitted into the hospital and treated accordingly. After obtaining a written informed consent, he was presented as a special case in clinical seminar.

Keywords: *Pycnodysostosis, osteosclerosis, bone fragility, radiological investigation*

Mugda Med Coll J. 2022; 5(2): 110-113

INTRODUCTION

Pycnodysostosis (also spelled as 'pyknodysostosis'), or osteopetrosis acro-osteolytica (often termed as Toulouse-Lautrec syndrome), is a rare autosomal recessive bone dysplasia, characterized by osteosclerosis and short stature.[1,2] The patients are usually short (below 150 cm) and the skeleton is susceptible to fracture.[2] The disease is found in all races, incidence is rare though. The incidence of pycnodysostosis has been estimated to be 1.0 to 1.7 per million live births, with an equal sex distribution.[3] 30% of the cases are offsprings of consanguineous unions.[4] The principal characteristics of this syndrome are short stature, cranial dysplasia, obtuse angle of mandible, clavicular dysplasia, total or partial dysplasia of the terminal phalanges and generally increased bone density.[5,6] The exfoliation of deciduous teeth is usually altered, as well as the eruption of the permanent dentition.[6] Here we have presented a case of pycnodysostosis (in a young male patient reported with complaints of body aches and pain in the limbs for several years) for academic and clinical interest.

CASE SUMMARY

A 20-year old male patient reported in the outpatient department of Shaheed Suhrawardy

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Medical College Hospital, Dhaka, Bangladesh, with the complaints of bodyache along with pain in the upper and lower limbs for several years. He also gave history of previous hospitalization for the same reason. He was born to unrelated parents. He gave a history of failure to thrive in his adolescence. However, he denied any history of intellectual disability and currently is attending a vocational college. He remains a mouth and nasal breather. However, he was treated as a case of polyarthritis in his previous admission in the district hospital. He was on NSAIDs drugs. On clinical examination, he was found to have short stature, low-weight, peculiar face, prominent forehead, receding jaw and partial dysplasia of the terminal phalanges. He was sent for radiological investigations in the Department of Radiology & Imaging of the same hospital. X-ray of his skull showed brachycephaly with wide open cranial sutures and persistent fontanelles with sclerotic changes in the base of skull and orbital rims. The angle of the mandible was obtuse, and the maxilla was found hypoplastic (Fig. 1), while lumbar region showed 'spool-shaped' vertebral bodies with quite prominent anterior defects; bones had sclerotic changes (Fig. 2). X-ray of both hands revealed acro-osteolysis of distal phalanges of thumb, index, middle and ring fingers (both right and left sides) along with irregular distal fragments; bones were sclerotic, too (Fig. 3). Those features were consistent with pycnodysostosis. The patient was then admitted under the Department of Orthopaedic Surgery and treated accordingly.

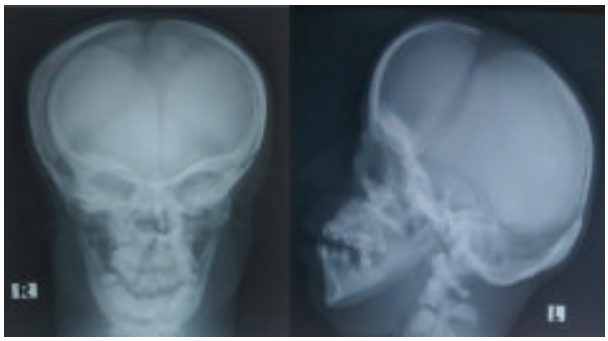

Fig. 1: *X-ray of skull (anteroposterior and lateral views) showing brachycephaly with wide open cranial sutures and persistence of open fontanelles, sclerosis of the base of skull and the orbital rims. The angle of the mandible is obtuse, and the maxilla is hypoplastic.*

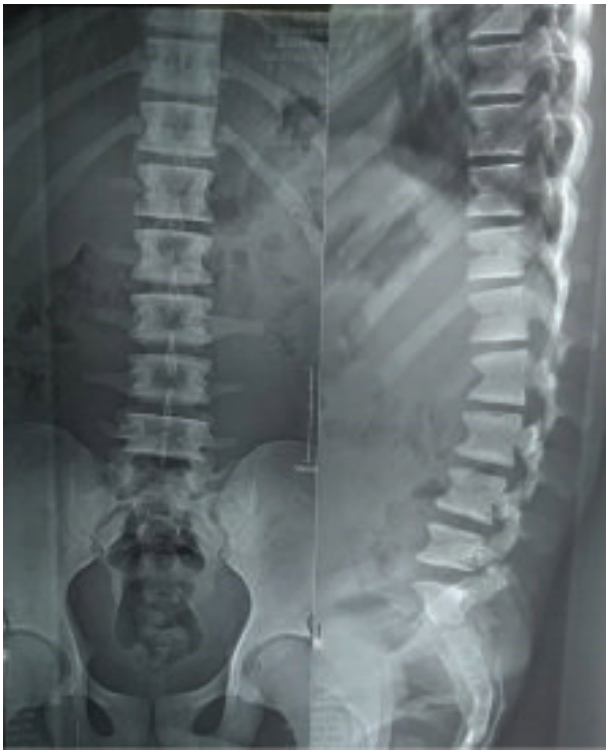

Fig. 2: *X-ray of lumbar region (anteroposterior and lateral views) showing spool shaped vertebral bodies with quite prominent anterior defects; bones have sclerotic changes.*

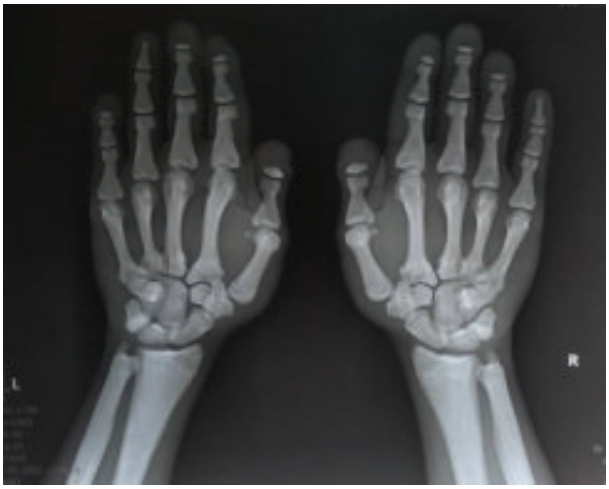

Fig. 3: *X-ray of both hands (anteroposterior view) showing acro-osteolysis of distal phalanges of thumb, index, middle and ring fingers (both right and left sides) along with irregular distal fragments; bones are sclerotic, too.*

DISCUSSION

Our patient had typical imaging findings which include "spool shaped" vertebrae, acro-osteolysis of distal phalanges of hands, obtuse mandibular angles, wide open cranial sutures and persistent fontanelles.

However, pycnodysostosis can be confused with other similar diseases, such as osteopetrosis and cleidocranial dysostosis, because they present some clinical and radiographic similar signs.[7,8] Differentiation from osteopetrosis solely on the basis of plain radiograph is not often possible.[7] Hence, it is important that the physician knows how to make the differential diagnosis in order to indicate the best treatment for each patient. Several studies supported that the most common finding in patients with pycnodysostosis is obtuse mandibular angle.[2-6,8-11] The presence of diffuse sclerosis, cortical thickening of bones, and acro-osteolysis on plain radiographs in the setting of other common clinical features is often sufficient to make the diagnosis.[2,3,8-10] If the disease is not diagnosed in infancy, fractures resulting from trauma due to fragility of bones usually lead to the diagnosis of this disease.[11] However, in our case it was an incidental diagnosis.

Several evidence suggest surgical management of these patients with bone grafting, fixation screws and bone plates.[12] Dental, maxillofacial and orthopaedic surgeons have managed such patients based on suitability of surgery and stability of reconstruction procedures.[12] Besides, looking at relevant biomarkers and evaluation of hormonal status have roles in such cases.[3] Thus, a team of specialists is often involved in treatment that include a paediatrician, an internist, an orthopaedic or dental surgeon, and perhaps an endocrinologist.

CONCLUSION

Patients with pyknodysostosis come to medical attention for a variety of reasons but often go undiagnosed even when presenting with classic features due to the rarity of the condition and the overlap with other skeletal dysplasias. Diagnosis of this condition is strongly implied by clinical and radiographic findings, but genetic testing can confirm the diagnosis. However, treatment should consist of a multidisciplinary approach to address concerning symptoms to improve the patient's quality of life.

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