

Case Report

Two cases with pycnodysostosis in a family: a case report

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ABSTRACT

Pycnodysostosis (Greek, pycnos - density, dys - defect, ostosis - bone) is a rare inherited disorder of the bone, first described by Maroteaux and Lamy. Pycnodysostosis is an autosomal recessive disorder, with incidence estimated to be 1.7 per 1 million births. Clinical presentation of this disorder include short stature, dolichocephalic skull, frontal bossing, obtuse mandibular angle, dysplastic clavicles, and short hands and feet, diffuse osteosclerosis, acro-osteolysis along with the finger and nail abnormalities. The main oral aspects are midfacial hypoplasia, a grooved palate, and dental abnormalities include double row of teeth, delayed eruption of permanent dentition, multiple caries. Pathological fractures of the bones occur due to sclerosis. Radiologically, skull bones appear thickened with open fontanels which look like 'lakes of bones', hypoplasia of facial bones, generalized osteosclerosis, open fontanels and cranial sutures, non pneumatization of paranasal sinuses, and fractures commonly in lower limbs.

Keywords: Hypoplasia, Osteosclerosis, Paranasal sinuses, Pycnodysostosis, Short stature

INTRODUCTION

Pycnodysostosis (Greek, pycnos - density, dys - defect, ostosis - bone) is a rare inherited disorder of the bone, first described by Maroteaux and Lamy.¹

Pycnodysostosis is an autosomal recessive disorder, with incidence estimated to be 1.7 per 1 million births; occurs due to lysosomal storage disease of the bone caused by a mutation in the gene that codes the enzyme cathepsin K.²

Phenotypic features of this disorder include short stature, dolichocephalic skull, frontal bossing, obtuse mandibular angle, dysplastic clavicles, and short hands and feet, diffuse osteosclerosis, acro-osteolysis along with the finger and nail abnormalities.³

The main oral aspects are midfacial hypoplasia, a grooved palate, and dental abnormalities include double row of teeth, delayed eruption of permanent dentition,

multiple caries. Pathological fractures of the bones occur due to sclerosis.^{4,5}

Radiologically, skull bones appear thickened with open fontanels which look like 'lakes of bones', hypoplasia of facial bones, generalized osteosclerosis, open fontanels and cranial sutures, non pneumatization of paranasal sinuses, and fractures commonly in lower limbs.⁶

CASE REPORT

Here, we report two siblings (Figure 1) with typical features of pycnodysostosis, both clinical and radiographical.

Case 1

Child 1- A 9 year old male came with complaints of not gaining adequate height. He was born to a second degree consanguineous marriage. Examination revealed height

107cm (<3rd percentile for his age) with US: LS ratio of 1:1.1, weight 16kg (<3rd centile), and head circumference 50.5cm (<3rd percentile). He has coarse facial features with frontal bossing with open fontanels measuring 6x5.5cm; restricted opening of mouth, beaked nose, crowded teeth, with chest deformity. The hands and feet had short terminal phalanges, broad based and dysplastic nails with koilonychia.



Figure 1: Two siblings 7 and 9 years, with frontal bossing, and short stature.

Case 2

Child 2- Younger sibling of child 1, 7 year boy, was also brought for not gaining adequate height. His height was 102 cm (<3rd percentile) with US: LS ratio of 0.86cm, weight 14kgs (<3rd percentile), head circumference 47cm (<3rd percentile) with open anterior fontanels measuring 5.5x5.5cms. He also had coarse facial features, overcrowding of teeth and chest deformity, short and broad phalanges with dysplastic nails.



Figure 2: Case 1 X-Ray skull lateral view showing wide opened anterior fontanel, non pneumotized sinuses, and obtuse angle of the mandible.

The radiological survey of limbs and skull findings (Figure 2 and 4) widely separated cranial sutures with widely open anterior and posterior fontanel; non-pneumatized paranasal sinuses and obtuse angle of the mandible. Skeletal x rays of long bones showing diffuse skeletal hyperostosis with sparing of medullary cavity (Figure 3). The bone age was equal to his chronological age for both the siblings. Both the brothers had moderate bilateral conductive hearing loss. Eye examination findings were normal.

Both the siblings had similar characteristic clinical and radiological findings suggestive of Pycnodysostosis.



Figure 3: Case 1 (A) X-Ray wrist and hand showing thickened cortex with narrow medullary space and osteosclerosis of the phalanges. (B) X-Ray of lower limbs showing healed and calcified fractures with cortical thickening



Figure 4: Child 2 lateral view of X-Ray skull, showing wide opened anterior fontanel, hypoplasia of mid face, obtuse mandibular angle and with non pneumotized paranasal sinuses.

DISCUSSION

Pycnodysostosis is an autosomal recessive disorder of the bone caused by an inactivating mutation in cathepsin K (CTSK), resulting from osteoclast dysfunction. The first case of pycnodysostosis was described in 1923 by Montanari; however, in 1962, typical features of pycnodysostosis was described by Maroteaux and Lamy (Greek: pycnos = dense; dys = defective; osteon = bone). The most common phenotype of pycnodysostosis is short stature, which was reported in 95.9% of the 97 reported cases. The next most common phenotype is an increase in bone density, which was reported in 88.7% of the 97 patients, with an estimated prevalence of 1 to 1.7 per million.^{7,8}

The normal osteoclasts bone cells function is to reabsorb into the bone and build new bone.² Impaired function of this critical enzyme (CTSK) for bone remodeling and resorption by osteoclasts leads to the fragile and sclerosing nature of the bone in affected patients.⁹ It is characterized by diffuse condensation of the skeleton with thickening of the cortex and narrowing of the medullary cavity.¹⁰

Cranial and maxillofacial features include frontoparietal bossing, thick calvaria, open fontanelles and sutures, hypoplastic paranasal sinuses, Wormian bones in the lambdoidal region, relative proptosis, beaked nose, and an obtuse mandibular gonial angle, often with relative prognathism.¹¹ Dental abnormalities such as hypoplasia of the enamel, obliterated pulp chambers, and hypercementosis are some of the most striking features of this anomaly.¹² The most evident clinical and radiographic feature of PYCD, however, is the presence of frontal, parietal and occipital bossing. Beaked nose, groove in midpalate, midfacial hypoplasia, mandibular hypoplasia and overcrowded teeth and obtuse mandibular gonial angle are common findings.¹³ The differential diagnosis is established with osteopetrosis, cleidocranial dysplasia and idiopathic acro-osteolysis, osteogenesis imperfect and mandibuloacral dysplasia.^{14,15}

There is no specific treatment for this anomaly and the current treatment is only supportive.

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