

Pyknodysostosis (Rare Skeletal Defect): A Case Report.

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ABSTRACT

Pyknodysostosis is a defective bone disease that is responsible in many bone deformities. We report a case with recurrent urti, growth retardation, facial dysmorphism, delayed eruption of teeth & inability to gain weight properly since childhood and successful treatment of the patient by supportive measures.

Keywords: cathepsin K, prognathism, wormian bones, potters thumb, sandal gap.

INTRODUCTION

Pyknodysostosis (dense defective bone) is a rare clinical entity was first described by Maroteaux and Lamy in 1962.^[1] The disease has also been named Toulouse-Lautrec syndrome, after the French artist Henri de Toulouse-Lautrec, who was believed to suffer from the same disease. The frequency of pyknodysostosis is expected to be 1.7 per million live births.^[2,3] Pyknodysostosis can be classified in the large group of genetic diseases that are individually uncommon, but collectively important because of the sum of their numbers, and their heavy impact upon affected individuals. It is an autosomal recessive osteochondrodysplasia following a typical Mendelian pattern of inheritance. In 1996 the gene responsible for the disease was labelled over chromosome 1 q 21 and the enzyme related was cathepsin K and the disease was classified under the category of lysosomal storage disorder.^[2] It's a lysosomal cysteine protease, cathepsin K (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.^[4-6]

General features of pyknodysostosis include short stature under 150 cm, generalized diffuse osteosclerosis with a tendency to fracture after minimal trauma, hypoplastic clavicles, as well as acro-osteolysis with sclerosis of the terminal phalanges which is an essential pathognomonic feature.

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Other features include wrinkled skin, finger and nail abnormalities, kyphosis and scoliosis, history of repeated chest infections, and sleep apnoea. The intellectual and sexual development is usually normal in the patients.^[4] 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, hypoplastic midface, and obtuse mandibular gonial angle, often with relative prognathism.^[5]

CASE REPORT

A 20 yrs old male student by occupation resident of Kasganj (UP) presented in the OPD medicine of Jawahar Lal Nehru Medical College & Hospital, Aligarh, with the chief complaints of recurrent urti, growth retardation, facial dysmorphism delayed eruption of teeth & inability to gain weight properly since childhood, but the acute condition that made patient to attend the hospital was coughing with expectoration since last 3 weeks associated with high grade fever of around 103°F (as told and measured by pts mother). Associated with generalised weakness and bony pains. There is also a history of multiple trauma to the patient, but none was so significant leading to hospitalisation of the patient, but followed by a deformity in an injured joint, however no associated history of delayed wound healing. There wasn't any associated history of anaemia, rashes, pain abdomen, swelling over the body, swelling over the body, drug intake, obesity, mental retardation, disproportionate growth any similar complaints in any other family member.

On examination patient is a young aged male of lean built and poor nutritional status. His vitals were stable except for the temp. of 103°F. Anthropometry reveals height of 149 cms BMI of 16 kg/sqm and upper segment to lower segment ratio of 1 without any evidence of a short neck.

General examination reveals a deformity in skull in the form of trigonocephaly with jaw prognathism with poorly developed middle part of the face with beaked nose. There is also the presence of the left sided DNS with nasal spur.

Examination of both hands, suggestive of ulnar bowing of forearm with broadening of wrist with deformity of fingers in form of atrophy of distal phalanx of second finger bilateral. With a deformity of thumb in the form of increased

transverse diameter (known as potter thumb or killer thumb or club thumb) lower limb suggestive of sandal gap and presence of club thumb in both feet with atrophy of the 4th distal phalanx.

Chest reveals the deformity in the form of pectus excavatum without any associated spinal deformity without any swelling rosary, sulcus, erythema . Systemic examination reveals normal status of CNS & CVS & GIT.

Respiratory system evaluation s/o chest deformity with right sided lower zone stony dullness and decreased air entry and adventitious sounds in the form of crepts. Investigations were done.

Baseline investigation didn't reveal any significant abnormality and the calcium and phosphate level came out to be normal. With normal Hb level and slightly raised total counts with predominantly lymphocytes.

Radiological evaluation was done. Biochemical evaluation in the form of VIT D and iPTH were done & pleural fluid evaluation was also done to rule out infection.

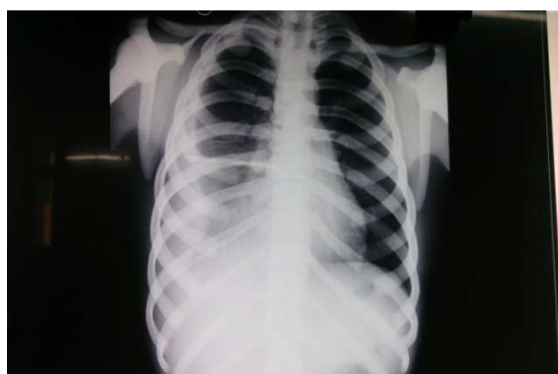


Figure 1: CXR PA.

CXR PA revealed increased bone density with atrophy of medial end of clavicle. Also Rt sided pleural effusion and consolidation with normal cardiac shadow.



Figure 2: X-ray Skull lateral.

X-ray skull lateral view suggestive of increased angle of mandible with malocclusion in the lower jaw with normal sella turcica and opened anterior

& posterior fontanelle .With increased bone density.



Figure 3: X-ray both hands.

X-ray both hands S/o increased bone density and atrophy of distal Px of 2nd finger (acro-osteolysis).



Figure 4: X-ray both knee.

X-ray both knee AP shows widening of lower end of both femurs leading to flask like deformity i.e Erlenmeyer Deformity.^[10]

25 OH VIT & iPTH levels came out to be normal.

USG ABD:-MILD hepatomegaly & few subcentimeteric lymph nodes.

Pleural fluid evaluation revealed tubercular pleural effusion.

Clinical Impression:-Pyknodysostosis With Rt Lower Lobe Consolidation With Synpneumonic Effusion (Tubercular).

DISCUSSION

Patient was put on ATT under DOTS cat 1. There is no specific treatment as of date for this disorder and treatment is supportive. Since bone fractures are a primary threat, it is important that care to be taken to prevent or minimize tendencies for a fracture to occur.^[7,8] Tooth extraction demands special care, such as carrying out the surgery as atraumatically as possible and with proper asepsis, due to the risk of fracture, especially in the mandible^[9] However, bone healing is normal despite the fragility of bones. As a real complication, mandibular fracture and subsequent osteomyelitis should be considered when seemingly minor surgery such as tooth extraction

or dental implants are indicated and osteomyelitis could become refractory due to osteosclerosis. Child patients should receive special dental care, particularly preventive treatment because of the group of mouth alterations described, in addition to periodic follow-up of their growth and craniofacial development^[8-10] The prognosis of the disease is good and no more serious systemic alteration has been noted. Also, life expectancy for a pyknodysostosis patient is normal.^[7]

CONCLUSION

In patients with Pyknodysostosis we advocate adequate supportive treatment which is helpful in providing a better prognosis for the patient.

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