Case 15613

Eurorad

A typical case of pyknodysostosis

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Section: Musculoskeletal system
Area of Interest: Musculoskeletal bone Musculoskeletal soft tissue Head and neck Musculoskeletal spine
Procedure: Diagnostic procedure
Special Focus: Congenital Case Type: Clinical Cases
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Patient: 17 years, female

Clinical History:

A 17-year-old girl was referred for taking hand X-ray to check the epiphysis as she had short stature with short finger and toes (Fig. 1a, 1b) and was suspected for low level growth hormone. She also had abnormal teeth, mild proptosis, beaked nose, prominent cranium with frontoparietal bossing.
Imaging Findings:

Hand X-ray showed hyperostosis of bones with partial agenesis of first, second and third distal phalanges (Fig. 2). In order to investigate more and rule out any other bony anomalies, skull X-ray and lumbosacral spine X-ray of the patient were taken after obtaining consent from the patient and her elder sister. Skull X-ray in lateral projection demonstrated enlarged head, thick calvarium, frontal bossing, open coronal and lambdoid suture with wormian bones in the lambdoid suture, shallow mandibular angle with mild-mandibular prognathism and hypoplasia of paranasal sinuses (Fig. 3). In the lumbosacral spine X-ray osteosclerosis of vertebral bodies and visualised parts of pelvis and ribs were observed. Lose of lumber spine normal lordosis with mild kyphotic changes of the upper lumbar spine were seen. Underdevelopment of the upper endplates of L2, L3 and L4 with possible spondylolysis of L5/S1 were also noted. (Fig. 4).
Discussion:

Pyknodysostosis is an autosomal recessive disease of extensive increased bone density. This is a lysosomal storage disorder which is caused by the mutation of a gene responsible for cathepsin K resulting in cathepsin deficiency relevant chromosome 1q21. The cathepsin K is a protease enzyme necessary for normal osteoclast function and degradation of type 1 collagen which produces 95% of the bone organic matrix. With the lack of this enzyme decreased bone resorption occurs which results in increased bone density [1].
The disease is usually diagnosed by typical clinical and radiographic features; however, an exact confirmation is obtained from the analysis of cathepsin kinase gene mutation [2]. The patients have short stature with a height of 150 cm or less, short limbs, wrinkled skin, kyphosis, scoliosis, history of repeated chest infections, mild enlarged head with a beaked nose, slight proptosis and sleep apnea. In the skeletal X-ray general increased bone density is seen. Hand X-ray demonstrates osteosclerosis with acro-osteolysis of the terminal phalanges. In the skull X-ray thick calvarium, frontoparietal bossing, open fontanelles and sutures with wormian bones in the lambdoid suture, hypoplasia of paranasal sinuses, obtuse mandible angle, mandibular prognathism and persistent primary teeth can be appreciated. Sclerosis of vertebral bodies, clavicle hypoplasia with erosion of distal parts can also be detected [3]. In this case, many typical radiographic features of Pyknodysostosis are present while contrary to the literature instead of increased lumbar lordosis which is typical for the disease, loss of normal lordosis with mild kyphosis of the upper lumbar spine is seen. The treatment of Pyknodysostosis is usually supportive including prevention of bone fractures and dental hygiene. The regular dental check-up is helpful to avoid dental problems. Special cares should

be taken during a tooth extraction procedure to reduce the rate of infection and decrease mandibular fracture. Some patients may get long bone fractures as well as mandibular fractures commonly caused by post-osteomyelitis, trauma and exodontia. The intellectual and sexual development of patients is usually normal. This anomaly always has a good prognosis and the patients have good life expectancy [4]. In conclusion, the early diagnosis of patients with Pyknodysostosis with their typical clinical and radiographic features would help to avoid unnecessary treatments. Alternatively, the physicians should advise patients to take specific precaution to prevent bone fracture during exercise and recommend particular dental hygiene. Also, parents need specific guidance regarding handling their affected children.

Differential Diagnosis List: Pyknodysostosis, Osteopetrosis, Osteogenesis imperfecta, Cleidocranial dysplasia, Idiopathic acro-osteolysis

Final Diagnosis: Pyknodysostosis

References:

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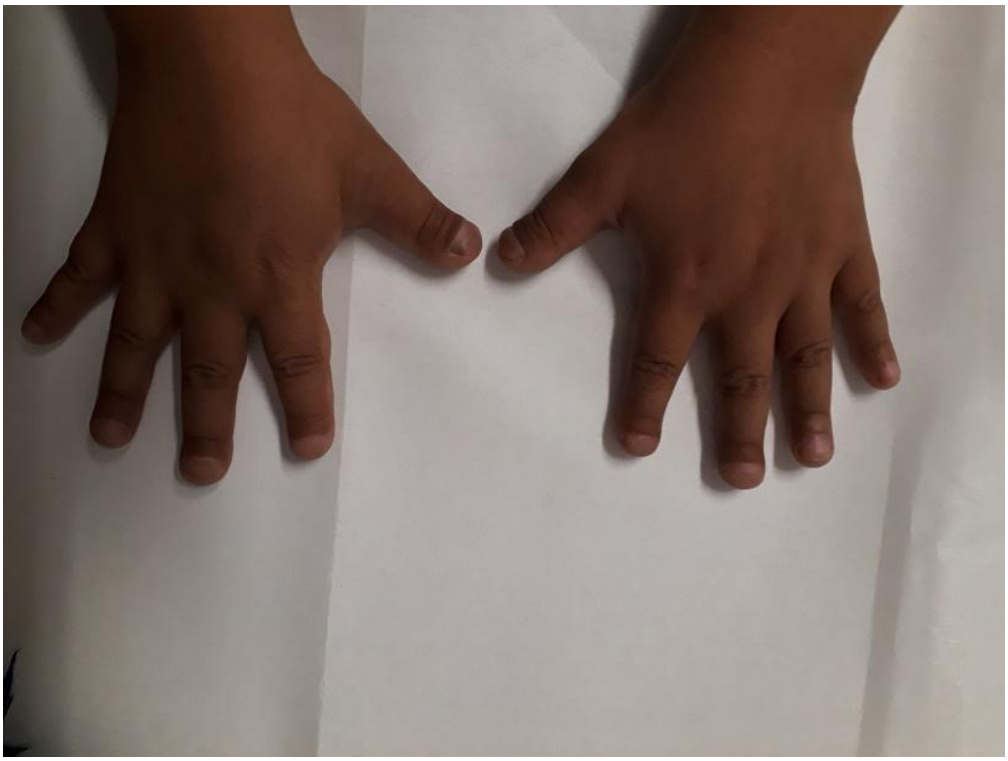
Hunt NP, Cunningham SJ, Adnan N, Harris M. (1998) The dental, craniofacial, and biochemical features of pyknodysostosis: a report of three new cases. Journal of oral and maxillofacial surgery, 56(4), pp.497-504. (PMID: 9541353)

Ramaiah, K.K.K., George, G.B., Padiyath, S., Sethuraman, R. and Cherian, B. (2011) Pyknodysostosis: report of a rare case with review of literature. Imaging science in dentistry, 41(4), pp.177-181. (PMID: 22232728)

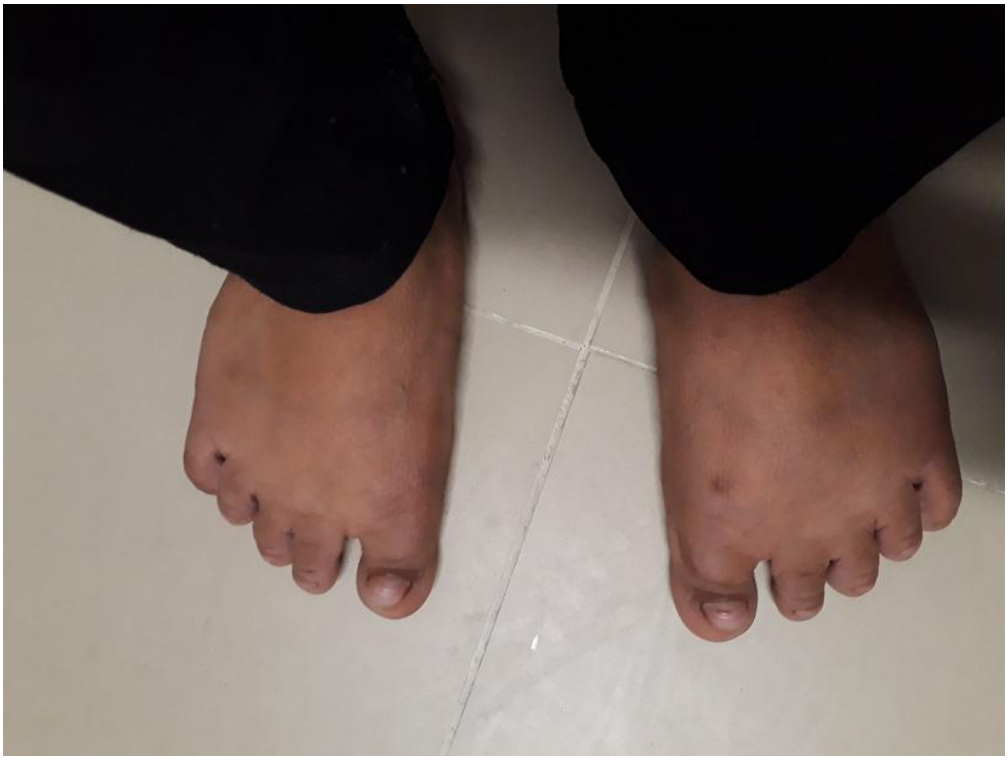
Figure 1

a



Description: Image of hands represents short fingers. **Origin:** Radiology department of French Medical Institute for Mothers and Children.

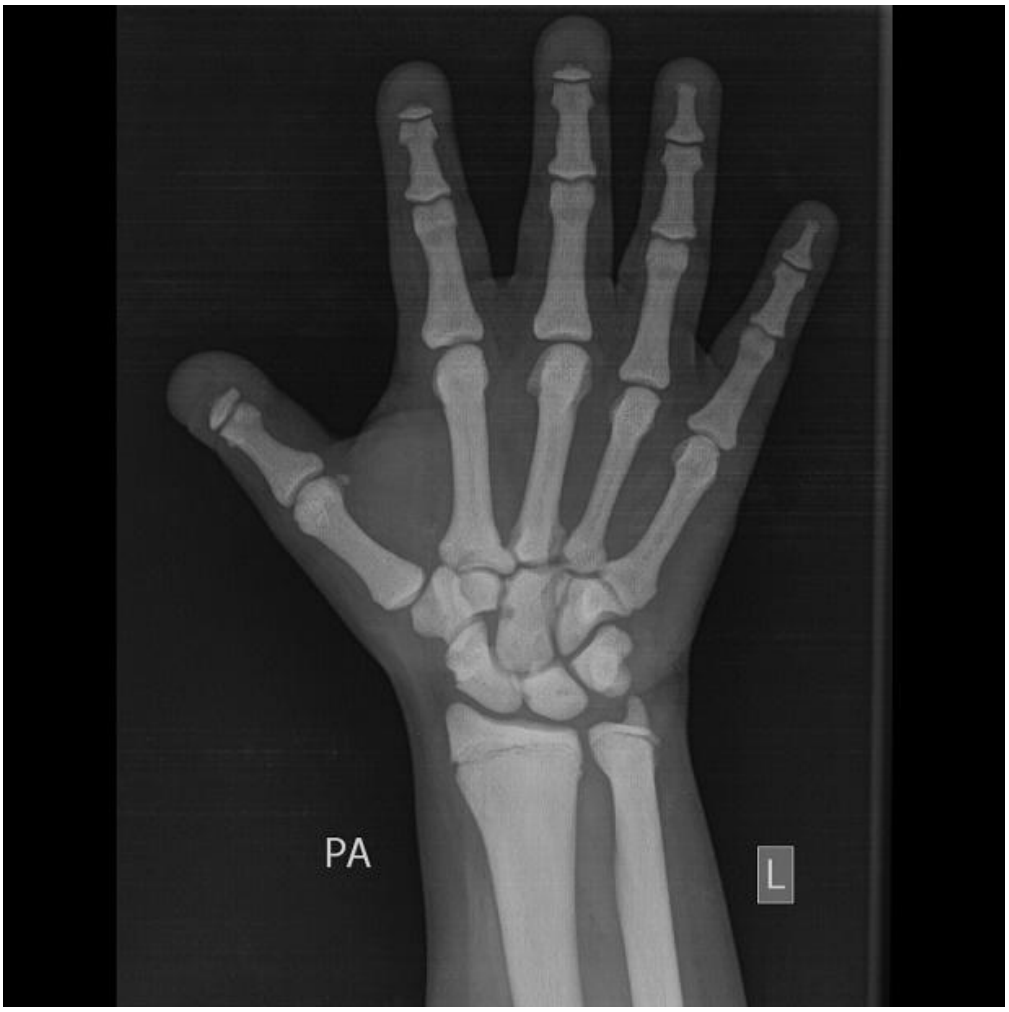
b



Description: Image of feet demonstrate short toes. **Origin:** Radiology department of French Medical Institute for Mothers and Children

Figure 2

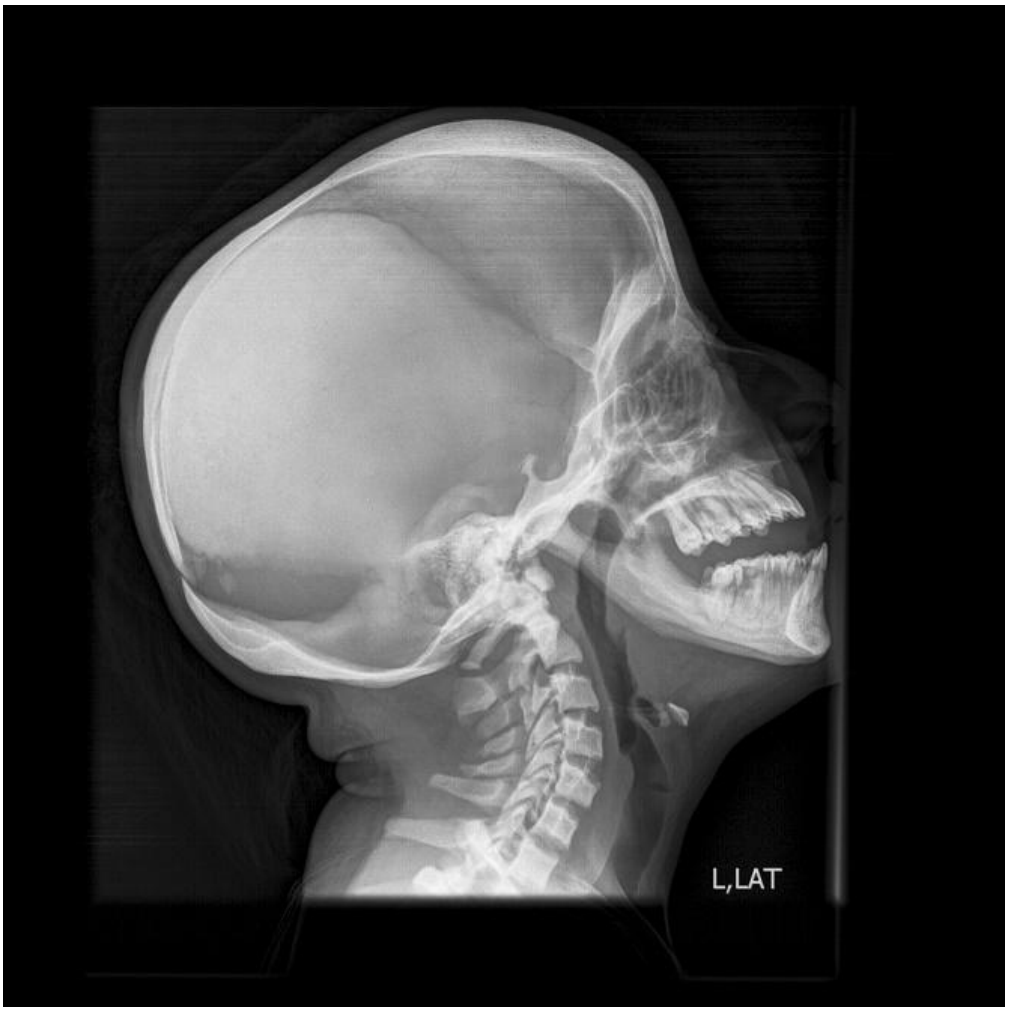
a



Description: Left hand in PA projection shows increased bone density (hyperostosis) with acro-osteolysis of first, second and third phalanges. **Origin:** Radiology department of French Medical Institute for Mothers and Children

Figure 3

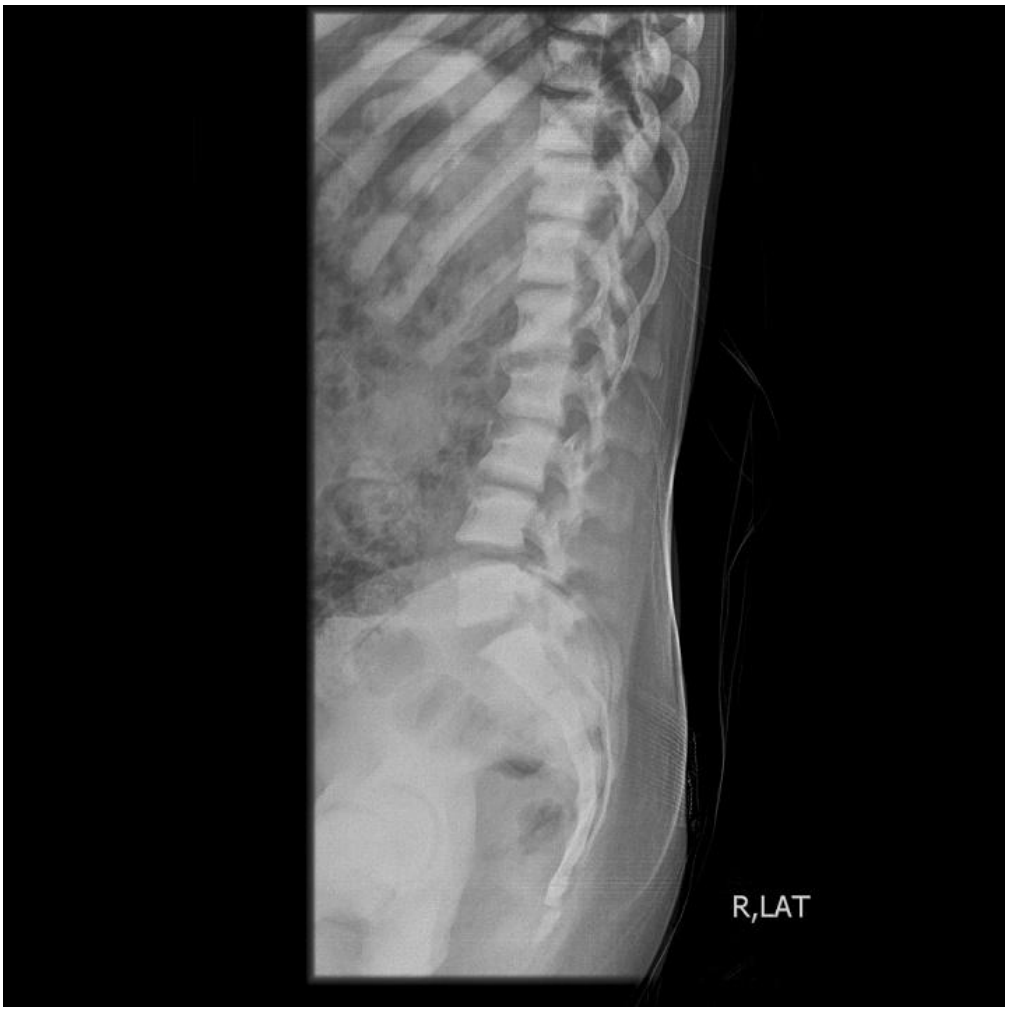
a



Description: Left lateral projection of skull and cervical spine demonstrates front?al bossing, thickened calvarium, open sutures with wormian bones in a lambdoid suture, hypoplastic paranasal sinuses, shallow mandibular angle and mandibular prognathism. **Origin:** Radiology department of French Medical Institute for Mothers and Children

Figure 4

a



Description: Right lateral X-ray of distal thoraco-lumbo-sacral spine shows osteosclerosis of bones, with loss of lordosis and mild kyphosis of upper lumbar spine. Underdevelopment of upper endplates of L2, L3 and L4 are also seen. **Origin:** Radiology department of French Medical Institute for Mothers and Children