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Case Report — Pyknodysostosis

ABSTRACT

Pyknodysostosis is an extremely rare lysosomal storage disease of the bone. Osteosclerosis of the skeleton is due to a decrease of the bone turnover. A plethora of clinical and radiological findings are observed. Patients are short statured with acroosteolysis and dense bones and develop multiple spontaneous bone fractures. A female patient presented with frontal bossing, small and thick fingers and toes. The classical clinical and radiological findings confirmed it as pyknodysostosis.

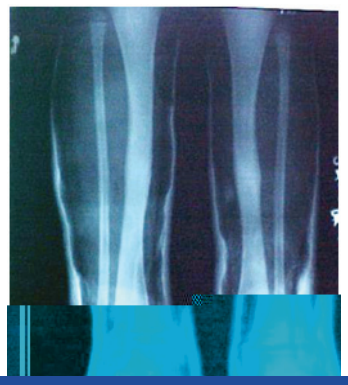
with chief complaints of a short stature and abnormality of hands and feet as noticed by her parents. The girl was normal till three years of her age afterwards abnormal nails with swollen and stained fingers and toes were noted by parents. The girl belonged to a low socioeconomic background. There is history of second degree consanguinity of parents. Other siblings of the girl were normal.

Weight was 50 percentile, height was 70 percentile. This case belongs to grade III malnutrition. She did not have any hearing or visual impairments. Head circumference was 49 cm. Anterior fontanel was wide-open 8X2.5 cms, posterior was open 2X3cms. Sutures were wide open with frontal and occipital bossing. Eyes show bilateral proptosis.

She had short neck, thin mandible, underdeveloped maxillae, malocclusion teeth and deviated nasal septum along with macroglossia. Examination of bony thoracic cage revealed pigeon chest deformity. Her limbs examination showed broad and short upper and lower limbs. Fingers and toes were broad due to swelling of terminal phalanges of all the twenty digits bearing hypoplastic nails. Sandal Gap deformity of first & second toes was seen bilaterally on both feet [Table/Fig-1]. She did not have any neurological deficit.

[Table/Fig-1]: Anterio Posterior radiograph of the feet showing sandal gap deformity and acroosteolysis

With these clinical presentation radiographs of skull, thorax, pelvis, hands, legs and feet were taken. All bones showed generalized increased density. Cortices were thick and dense with preservation of the medulla. Both legs X-rays showed fractures of mid shafts of both tibiae, there is no displacement and is known as "Banana Fractures" because if you tear the fruit with peel intact it will not show on the covering [Table/Fig-2]. Lateral radiograph of the skull



[Table/Fig-2]: Anterio Posterior radiograph of legs showing Banana Fractures of mid shaft of both tibiae

revealed dense calvarium, wide opened sutures, open fontanels and a number of wormian bones. Para nasal sinuses revealed lack of aeration and hypoplastic maxillae. Poorly developed mastoid was also noticed with prominent sella turcica [Table/Fig-3]. Radiographs of hands and feet delineated classical "Acro-osteolysis" – due to this phalanges look short and aplastic [Table/Fig-4]. No specific treatment was given to the patient.

[Table/Fig-3]: Lateral radiograph skull showing open fontanelle, acute angled mandible and wormian bones

[Table/Fig-4]: Anterio Posterior radiograph of the hands showing acroosteolysis

DISCUSSION

Pyknodysostosis is observed more in males, male: female ratio is 2:1 [1]. There is no racial predilection noted. This disease is seen in all age groups but observed mainly in children and young adults, cases as young as 8 month old are seen [2]. It has an incidence of 1.7 per million births [3]. The presentation of this disease is very distinctive and it shows similarity in appearance of all affected persons regardless of age, sex and race. Dwarfism is characteristic and people affected rarely attain 150 inches height [4].

This disease was first recognized about two centuries ago. Nineteenth century's famous french author Henri-de-Toulouse-lautrel was diagnosed to be suffering from pyknodysostosis. Pyknodysostosis a greek term, was described originally in 1923, but it was in year 1962 recognized as an entity by Maroteaux & Lamy. "Pykno" means dense bones and "Dysostosis" means abnormal bone formation [2,4,5]. It is an autosomal recessive disease. Cytoplasmic abnormalities with short arm deletion of a G-22 chromosome have been reported in association with this disease. A defect in enzyme "Cathepsin K" is observed [1,2].

It present with repeated bone fractures and typical finger abnormalities [4]. Anaemia is not a feature of this disease. Some patients also present with dysplastic clavicles having acromial end hypoplasia and shallow acetabulum. Spinal radiological abnormalities like lack of segmentation, spondylolysis, spondylolysthesis and spool-shaped vertebrae are also described. Madelung's deformity of wrists is also described. Our case has "Sandal Gap" deformity of 1 & 2 toes [6]. Current case did not show hearing loss or orbits vitreous hyperplasia as seen in only few cases [6,7].

Pyknodysostosis should be differentiated from other similar disease. Osteopetrosis is characterized by lack of periodicity of production of dense bone, anaemia, normal mandible, no evidence of acro-osteolysis. Cleido-cranial dysostosis will not have acro-osteolysis. In Hajdu-Cheney Syndrome sucked barley sugar appearance is observed due to extension of acro-osteolysis upto metatarsals. In vinyl chloride exposure acro-osteolysis will have a history of exposure will be significant. Pathologically there is disorganization of bone structure at the level of lamellar bone formation and of the osteum. There are few osteoclasts and an excessive number of osteoblasts on the trabecular surface. It is also suggested that cells do not respond normally to the demands of the stress on the skeleton [5].

CONCLUSION

Pyknodysostosis is a rare bone disorder presenting with a number of clinical and radiological features. Whatever is the case the normal life expectancy of the affected patients is good. We hope that biotechnology in future can bring a cure to this disease.

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REFERENCES

[1] Odhiambo A, Moshy J, Otieno W, et al. Pyknodysostosis; Early and Delayed clinical Manifestation. *Professional Med J*. 2011;18(2):331-35.
[2] Kamak H, Kamak G, Yavuz I. Clinical, radiographic, diagnostic and cephalometric features of Pyknodysostosis in comparison with Turkish cephalometric norms: A case report. *Eur J Dent*. 2012;6:454-56.
[3] Ramaiah K, George G, Padiyath S, Sethuraman R, Cherian B. Pyknodysostosis : report of a rare case with review of literature. *Imaging Science in Dentistry.* 2011;41(4):177.
[4] Ahari Z. Pyknodysostosis. *Journal of bone and joint surgery*.1969;51B(2):307.
[5] Sutton D. A Textbook of radiology and imaging. Edinburgh: Churchill Livingstone; 1998.
[6] Mujawar Q, Naganoor R, Patil H, Thobbi A, Ukkali S, Malagi N. Pyknodysostosis with unusual findings: a case report. *Cases J*. 2009;2(1):6544.
[7] Ashraf M, Farooq A, Dar TI, Wani KA. Pyknodysostosis with Hearing Impairment. *J Medicine*. 2010;11(2):202-04.

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