65. **PYCNODYSOSTOSIS- A RARE DIAGNOSIS NOT TO MISS**

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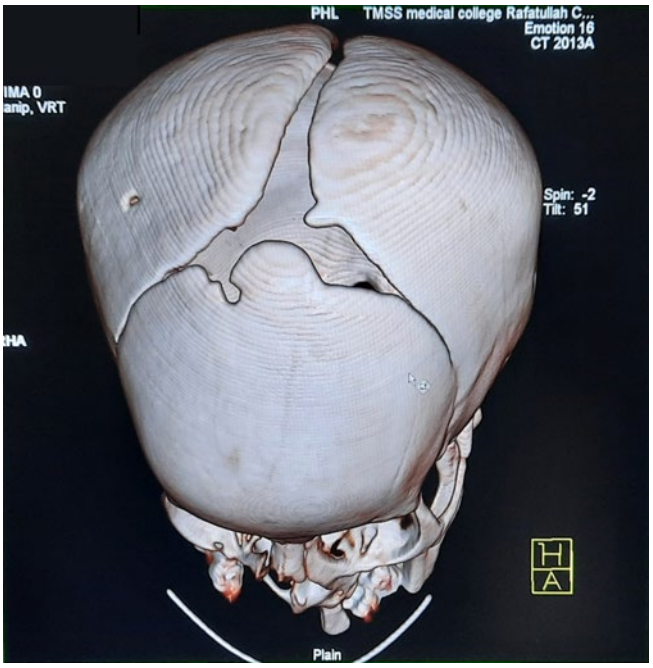
INTRODUCTION: Pycnodysostosis derived from the Greek words pycnos-density, dys-defect, ostosis-bone is a rare inherited disorder of the bone with an incidence of 1.7 per million births. It is one of the lysosomal storage disorders with a deficiency of enzyme cathepsin K. Along with a history of repeated fractures of the bones, patients with Pycnodysostosis have a short stature, dolichocephalic skull, obtuse mandibular angle, short terminal phalanx. They also have dental abnormalities such as the delayed eruption of permanent teeth, and double rows of teeth. Mild psychomotor problems were noted in a few individuals. Pycnodysostosis is diagnosed by clinical and radiological features as there is no diagnostic criterion. Skulls show open fontanels, generalized osteosclerosis, loss of the normal angle of the jaw, and hypoplasia of facial bones. Social and occupational support needed to prevent recurrent fractures. Orthopaedic and orthodontic cares are needed. **CASE:** A non-diabetic normotensive male of 63 years presented to the medicine outpatients department (OPD) with complaints of burning type, non-radiating moderate intensity pain in the epigastric region for three months following taking of NSAIDs. He gave a history of fractures of different bones following minimal trauma since his childhood and used to take NSAIDs every now and then. He did take orthopaedic consultation for those fractures but the underlying pathophysiology was not explored. Malunion of long bones resulted in a deformed limb. The rest of his past medical history and family history was unremarkable. On clinical examination, we found the patient had short stature with proportionately short limbs. He had dysmorphic features with a large head, small facial structures, frontal and bilateral parietal bossing. Examination of the oral cavity revealed a narrow high arched palate and hypoplastic, overcrowded teeth. His digits were short and stubby. The lower limbs of the patient have angular deformities that resulted from malunion of bones from previous fracture. Haematological and biochemical investigations were normal. Chest X-ray showed multiple ribs fracture on the right side. The patient's clavicle was normal in X-ray. Both tibia and fibula showed a malunited fracture in diaphysis. Computed tomography (CT) of the skull, as well as 3D reconstruction of CT, found open fontanelles and widening of sutures (Figure). Frontal and parietal bossing was noted. There was hypoplasia of the air sinuses. CT scans showed dysplasia of the bones of the face and hands. Hypoplasia was noted in the terminal phalanx of fingers. An endoscopy of the upper gastrointestinal tract found multiple erosion around the antrum of the stomach. Considering the clinical and radiological feature diagnosis of Pycnodysostosis with NSAIDs-induced antral erosion was made. Patient and his caregivers were briefed about the diagnosis. Consultation from a multidisciplinary team was done concerning his treatment. **CONCLUSION**: It is important to diagnose Pycnodysostosis early as it allows proper planning to prevent future fractures. While assessing a patient with short stature with a history of repeated bone fracture Pycnodysostosis should be considered along with other differentials.

***Figure*.** 3D Reconstruction of Skull (Anterior Fontanelle and Sutures are open and wide).



Key words: Pycnodysostosis; Fracture; Autosomal Recessive.