Abstract:
Pycnodysostosis is a rare sclerosing skeletal dysplasia, first described in 1962 by Maroteaux and Lamy. It is a genetic disorder, usually diagnosed due to pathological bone fractures. Short stature, facial morphological characteristics, certain radiological features aid in the diagnosis. The head is usually large, the nose beaked, the mandibular angle obtuse, and both maxilla and mandible hypoplastic. Dental abnormalities and impaction are observed, as well as alterations in eruption and frequent dental crowding. The differential diagnosis includes osteopetrosis, cleidocranial dysplasia, Engelmanns disease and idiopathic acro-osteolysis. We report a case of pycnodysostosis who presented as a pathological fracture femur and review its clinical and radiographic features with the literature.

Keyword: Pycnodysostosis, pathological, acro-osteolysis

Introduction:
Pycnodysostosis is a rare sclerosing skeletal dysplasia with prevalence of 1 per 1 million. It is a genetic disorder with autosomal recessive trait. The primary features of this syndrome are generalized skeletal sclerosis, short stature, open fontanelles, obtuse angled mandibles, dysplastic clavicles, dysplastic terminal phalanges and generally increased bone density, chronic oro-maxillary infections. The most important presenting features are chronic periodontitis and recurrent pathological fracture of long bones due to the increased bone fragility. It closely resembles osteopetrosis and cleidocranial dysostosis but can be distinguished by its morphological and radiological features. We report one such case of pycnodysostosis, who presented with pathological fracture of right femur and review its clinical and radiographic features with the literature.
Case Report
A 28 years male presented with a periprosthetic fracture of the right Shaft of femur following a trivial fall. He had past history of fracture of the same bone for which he was done compression plating elsewhere 1 year back. He was short statured with facial dysmorphism with parrot beaked nose, obtuse angled mandibles (Fig 1), crowded teeth(Fig 2), dysplastic skull with open anterior fontanelles(Fig 3)

He had short stubby hands & feet(fig 4-5) and hypoplastic terminal phalanges of digits(fig 6). He had radiologically denser bones with Erlenmeyer flask deformity of distal femur(fig 7). He had multiple episodes of long bone fractures for which he had undergone native treatment for variable period of time resulting in multiple deformities (Fig 8,9). Radiograph of the lumbar spine showed spondylolysis of L5 vertebra and spool shaped vertebrae (Fig 9)

fig 1 dysmorphic face with parrot beaked nose and obtuse angled mandibles
fig 2 crowded teeth fig 3 dysplastic skull with open anterior fontanelles
fig 4 short stubby hands

fig 5 hypoplastic terminal phalanges of digits
fig 5 short stubby foot with toes

fig 7 distal femur showing Erlenmeyer Flask deformity

fig 8 X-ray of both legs showing multiple healed deformities of both tibia due to native treatment
We managed the patient by doing an implant exit of the previously done DCP. Then we did a Precontoured Proximal Femur Locking Compression Plate Osteosynthesis. Union was achieved in 18 weeks with complete disappearance of fracture line in about 6 months. After 16 months follow up, patient is walking with full weight bearing with good hip range of movements (fig 11-16).
immediate postop radiograph after implant exit, ORIF with proximal femur LCP

fig 16 Good Range of movements at 12 months follow up

Hematological parameters were normal with no evidence of hepatosplenomegaly by clinical and ultrasonographical studies. Our patient had history of treatment of frequent dental infections.

Discussion

-Pycnodysostosis was first described by Maroteaux and Lamy, who also coined the term. Henri Toulouse-Lautrec, a famous French painter was thought to be afflicted by this disorder, hence this disorder is also referred by his name (1,2). Recent Studies have identified that this dysplasia is related to genetic mutations in the lysosomal cysteine protease enzyme cathepsin K which is synthesized by osteoclasts, a key molecule for bone resorption. Hence a decreased bone turn-over occurs in this condition (5,6,7). The locus for the dysplasia has been mapped to chromosome 1q21(17). This dysplasia is characterized by short-limbed short stature.

Soliman et al. (7) reported that short stature is caused by the increased bone volume of the sella turcica that, on compressing the pituitary gland, causes its hypoplasia and a deficient production of the growth hormone. There is hypoplasia or absence of the lateral portion of the clavicles, and hypoplasia of the terminal phalanges of the digits (termed acro-osteolysis), leading to short, stubby hands with bulbous distal phalanges. The skull has widened sutures and persistent open fontanelles with frontal and occipital bossing. The nose is protuberant, parrot-like. The mandible is small, and the angle of the mandible is described as obtuse, leading to a very small chin. The teeth are delayed in appearance, crowded and disordered when present leading to chronic alveolitis and related complications. (18,19) Helfrich et al (8) asserts that in diseases where the formation and function of the osteoclasts is reduced, as is the case with Pycnodysostosis, dental eruption is affected. This situation is confirmed in the both clinical cases. There may be dental abnormalities, with hypoplasia of the enamel, obliterated pulp chambers and hypercementosis. Radiographs show generalized osteosclerosis. The medullary canal though present is small and irregular. Spinal radiographs may show spool shaped vertebra. Spondylolisthesis and spondylolysis are common(4). Hand radiographs show the hypoplasia or resorption of the distal phalanges as seen in our case reports. Coxa vara, coxa valgum, genu valgum, kyphosis, and scoliosis are also reported. The distal femur in a patient with Pycnodysostosis usually has an Erlenmeyer’s flask deformity(22) The sclerotic bone is fragile as the increased bone density was mainly in the trabecular bone and not in the cortical bone. Hence the cortex will be of normal thickness, whereas the medullary cavity will be limited as a result of the increase in trabecular bone.
Hence these bones though sclerotic are notorious for fractures. Pathological fractures are more common in lower extremities especially in the disphysisal region. By age 22 years, one patient had sustained more than 100 fractures (21). The fractures are usually transverse and diaphyseal, and heal with scanty callus. Both bone formation and resorption are simultaneously diminished at the fracture site. Meredith et al. (10) suggested that cells at the fracture sites fail to respond normally to the demands of stresses on the skeleton. However, their microscopic observation showed all the elements of fracture healing process. Delayed union and non union of long bone fractures could be problems in adulthood requiring surgical intervention (14). Non-union has been reported in the ulna, clavicle, and tibia. A variety of fracture fixation methods like IM nailing, plating, Ilizarov external fixator (IEF) with or without bone grafting have been described in the literature for Pycnodysostosis (14). But nothing proves to be superior than the other. IM nailing has been effective in preventing refracture due to permanent fixation and in obtaining physiological alignment. However, longer persistence of fracture line, failure of callus to bridge a large bone gap resulting even in non union have been reported. In such cases, auto iliac bone graft will be effective in achieving bone union. Also, IM nailing of fractures in Pycnodysostosis poses great difficulties in reaming of sclerotic bone requiring a high degree of surgical skill. Some reports have shown successful outcomes using open reduction and internal fixation with compression plating but refractures and periprosthetic fractures are common (11, 12, 13). A rigid Ilizarov external fixator (IEF) could be a good alternative for IM nailing and plating whereas pin site infections, longer fixation periods and high re-fracture rate were serious disadvantages of IEF for treatment of fractures in Pycnodysostosis. (14) Life expectancy is normal. Adult height reaches 130 to 150 cm. Growth hormone has been used in physiologic replacement doses to accelerate growth in these short patients. Laboratory parameters are normal as haematopoiesis is not usually affected.

**Differential diagnosis:**
The differential diagnosis includes osteopetrosis. Unlike osteopetrosis, Pycnodysostosis does not lead to aplastic anaemia because the medullary canal is partially preserved. In osteopetrosis signs of compression of the cranial nerves exist such as facial paralysis, deafness or pain occur. Cleidocranial Pycnodysostosis may be considered because of the hypoplasia of the clavicles; however, osteosclerosis is not seen in cleidocranial Pycnodysostosis. Engelmann’s disease can also be considered, but clinically those patients are tall and eventually develop muscle weakness. Other rare disorder that may be confused with Pycnodysostosis is idiopathic nonfamilial acroosteolysis (15, 20) where the appearance of the patients is typical, with hypotelorism, exophthalmos and an upturned nose. The angle of mandible is acute and increased bone density is not present. (16, 20)

**Conclusion:**
Pycnodysostosis is a rare genetic disorder with characteristic radiological and morphological features. It can be considered as a differential diagnosis of closely related disorders like osteopetrosis, cleidocranial dysplasia, idiopathic acroosteolysis & Engelmann’s disease. Surgical treatment is indicated in long bone fractures in adults due to higher incidence of delayed and non unions and associated secondary deformities. Compression Plate osteosynthesis can be a good alternative.
fixation to IM nailing where it is technically demanding.

References:


