University Journal of Surgery and Surgical Specialties

ISSN 2455-2860 Volume 3 Issue 3 2017

# Pyle's Metaphyseal Dysplasia-A case report

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**Abstract :** Of all the skeletal dysplasias known, Pyles dysplasia is an extremely uncommon and rare variety. It is primarily a metaphyseal dysplasia. We report here a case of a 60 year old woman who presented with pathological fracture of her right tibia and right humerus. The tibial fracture was managed with interlocking nailing and the humerus was treated conservatively.

**Keyword :**Pyle, Metaphyseal dysplasia, Interlocking Nailing.

## PYLE'S METAPHYSEAL DYSPLASIA A CASE REPORT:

**INTRODUCTION**: Pyle first described this skeletal anomaly in 1931.Since then, less than 35 genuine cases have been reported worldwide. Pyle-type skeletal dysplasia is an extremely uncommon and rare skeletal disorder primarily affecting the metaphysis where the remodeling is affected. It is a genetic disorder inherited in an autosomal-recessive pattern, where both parents are affected, but there is a 25% chance that the affected alleles will pass to a developing embryo and produce the disorder. The actual gene or genetic defect is unknown, and cannot therefore be tested. The disorder is characterized by certain striking radiological features with relatively unremarkable phenotypic features. Genu valgum is the only consistent clinical finding [Fig-1].

**CASE REPORT:**

In October 2009, a 60 year old woman presented with complaints of pain and swelling in the proximal right leg and right arm following a trivial fall. On examination of her right arm and leg, swelling, tenderness along with crepitus was noted. There was no neurovascular involvement at either site. Routine antero-posterior and lateral radiographs of the affected tibia and humerus showed widening of the metaphysis with fracture [fig-6, 7]. Other features observed in the patient were scoliosis [Fig-2], dental caries, genu valgum and prognathism. She had normal stature and normal IQ. Other long bone radiographs showed splaying of the

metaphysis with constriction of diaphysis [Fig-5]. The constellation of all the above clinical and radiological features was suggestive of Pyle's Metaphyseal dysplasia [Fig-4, 5]. Closed interlocking nailing by a midline patellar tendon splitting approach was done for the proximal tibial fracture [fig-10]. Patient was immobilized in a long leg plaster slab for 4 weeks following which knee and ankle mobilization were begun. At 6 weeks, partial weight bearing was allowed with gradual full weight bearing at 8 weeks. The proximal humeral fracture was managed conservatively by a plaster slab for 3 weeks with subsequent conversion into functional cast bracing. The striking feature in this case was that both the fractures showed abundant callus formation by 4$^{th}$ week with complete radiological union by 5-7 weeks which seemed to be relatively early [Fig-8, 9].

**DISCUSSION:**

Pyle-type metaphyseal dysplasia is an extremely uncommon and rare genetic skeletal disorder inherited in an autosomal - recessive pattern. It is characterized by a genetic defect in metaphyseal remodeling that leads to grossly widened metaphyses of long bones with marked cortical thinning and osteoporosis (Erlenmeyer-flask deformity) especially in the distal end of femur and the proximal tibia. Proximal two-thirds of humerus and distal two thirds of radius and ulna may show similar changes. Less commonly these changes can be noted in other distal long bones, distal metacarpals and proximal phalanges[10]. Clinical signs and symptoms are usually vague like muscle weakness, joint pain, genu valgum, scoliosis, and limited extension of elbow but the common presentation is a pathological fracture after a minor injury[7].Caries and misplaced teeth, and patchy sclerosis of the cranial vault [Fig-3][5, 6], with associated moderate supraorbital prominence and prognathism are other features that have been reported in Pyle-type metaphyseal dysplasia.

Gorlin et al noted that Pyle-type metaphyseal dysplasia is clinically, radiographically and genetically distinct from craniometaphyseal dysplasia, a relatively uncommon condition with which it has been often confused. Pyle-type metaphyseal dysplasia must be distinguished and separated from craniometaphyseal dysplasia, for prognosis and treatment, since deafness, facial paralysis and occasional impairment of vision may result from cranial nerve compression in the latter [1,2], whereas, they are unusual in the former.Other differential diagnoses of Erlenmeyer flask deformity are Gaucher's disease, osteoporosis, thalassemia and cartilage dysplasias such as Ollier's disease and diaphyseal actasis ,Niemann Pick disease[7].Absence of cranial involvement excludes first two diagnoses in our patient. Moreover, our patient did not have anemia, jaundice, hepatosplenomegaly, increased bone density or any history of blood transfusion ruling out these conditions. Heselen et al noted that spinal malalignment or scoliosis [fig-2] is infrequent but are well-documented complications. Spinal involvement varies from moderate platyspondyly to biconcave lens appearance of the vertebral bodies[5]. The stature is usually normal with normal intelligence. General health is not affected except for a tendency for pathological fracture following a trivial trauma [4]. Shibuya et al described metaphyseal dysplasia in two families with the involvement of both flat and long bones. They also performed a quantitative measurement of the bone mineral content and reported a marked reduction of the value (one quarter of the

normal)[9]. Lindberg has raised concerns about the ability to obtain and maintain fixation and the uncertain nature of healing in patients with this disease based on the abnormalities of the bone. The results of two surgical procedures (corrective osteotomies for genu valgum) in one teenage boy shows that the post-operative healing is similar to that of patients without the disease. This is the only report in English literature that documents the post operative course after surgical procedures in these patients.[8] The observation in our case was that in Pyle's disease, all the bones being uniformly osteopenic, pose greater technical difficulties to the treating surgeon in reduction and adequate fixation of fractures in such osteoporotic bones. Hence the surgeon should have a proper preoperative planning in implant selection, fixation and anticipate intraoperative difficulties in treating such fractures. The post operative bone healing was found to be rather quicker than that in the ordinary patient.

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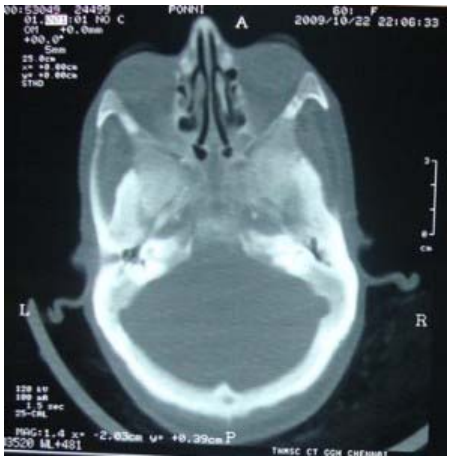
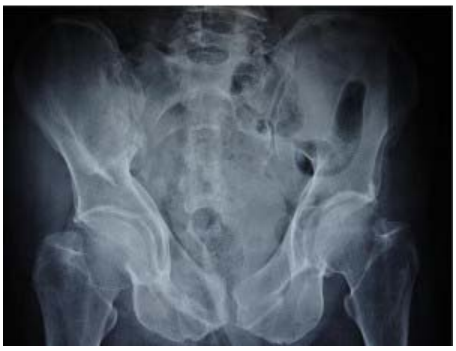


**fig-5**
**fig-6[#humerus]**



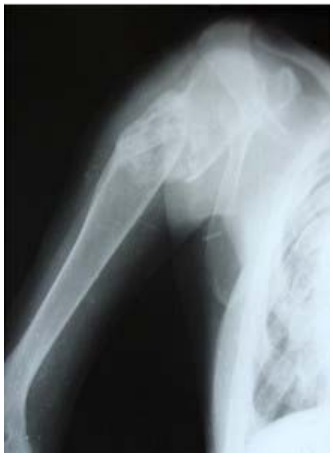
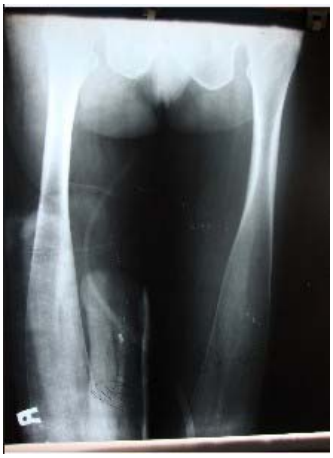
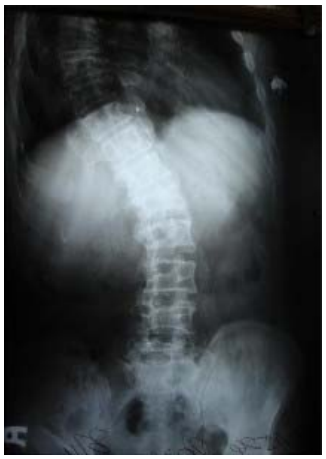


**fig-3**
**fig-4**





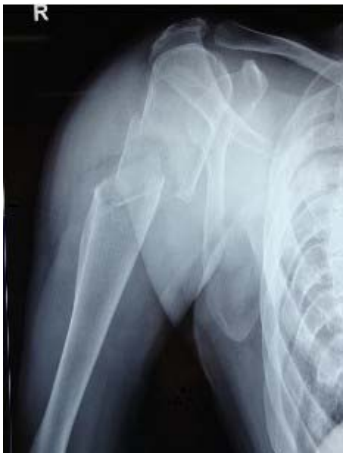


**fig-8[2 months follow up]**
**fig-2**

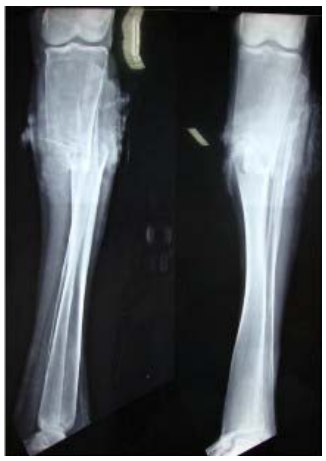


**fig-7 [pre-op]**

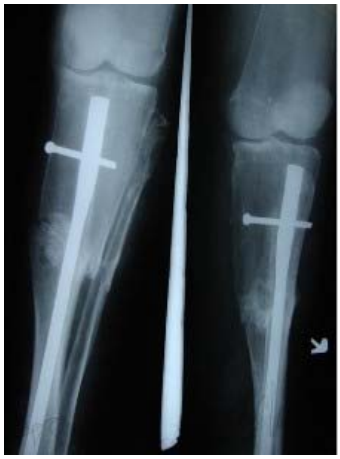


**fig-10 [immediate post op ]**
**fig-1**

**fig-9 [2 month follow up ]**