

Clinical Implications In Pyle`s Disease, (Familial Metaphyseal Dysplasia), Report Of Five Cases with Review Of The Literature.

Sameer A. Kitab* FICMS Orth.

Summary :

Background: Pyle`s disease is a distinct skeletal dysplasia with defective metaphyseal modeling and Erlenmeyer-flask deformity of tubular bones.

Objectives: Clinical implications in five reported cases with mechanical bone properties different from normal bones were presented. One patient with Genu-valgum deformity had corrective osteotomy done with internal fixation showing osteotomy healing and sustained deformity correction with more than twelve years of follow up. Two patients presented with fractures and angular malalignment with abundant callus .The other two had insufficiency fractures due to repetitive low energy activities.

Results: Patients with Pyle`s disease have less than normal mechanical bone properties, insufficiency fracture or gross fractures may develop after low energy or repetitive trauma. Internal fixation can be used and healing may progress with abundant callus.

Conclusions: Pyle`s disease stands as a distinct type of metaphyseal dysplasia with characteristic clinical-radiological criteria.Genu valgum deformity may warrant osteotomy correction, Internal Fixation can be used and osteotomy may be rapid to heal with the widened bone surfaces. Protection against potential angular malalignment is important until there is sound healing of the fractured bone. Because of increased fragility of bone, a word of caution may be appropriate in the young patient indulged with sports activity.

Key words: Pyle`s , Metaphyseal, Osteotomy

Introduction:

Pyle`s in 1931 described a 5-year old boy with “unusual bone development” and large bone metaphyseal flaring (7).Since then many cases were reported with certain descriptive or surgical implications.(2,3,4,8,9,10,12) The pathophysiology of Pyle`s disease or familial metaphyseal dysplasia is incompletely understood (1,6,9,10,11,14,17),radiologically it has peculiar (Erlenmeyer flask) metaphyseal flaring of tubular bones with relatively unremarkable phenotypic features (5,8,12,13,15,16).It may arise as an autosomal recessive heritable form(5,9). The biochemical markers of skeletal homeostasis are typically normal. Clinically Pyle`s disease runs a relatively benign course ,however ,cortical thinness and increased fragility of bone as well as progressive genu-vagum deformity and limb length discrepancy may have some surgical implications . Healing potential and fixation problems are important concerns when surgical correction is indicated (3,4). The clinical implications of five patients with Pyle`s disease with main tubular bone affection and no radiological evidence of skull or spine features, all with normal body proportions and unremarkable facial features were reported with review of the literature.

Case No1:

A 16 year old female presented with progressive genu- valgum and Knock-knees with 32 degree angle on the right and 16 degrees on the left , she has a sister with the same disorder .The patient height was 167 cm with normal body proportions. A right sided supracondylar femoral medially based closed wedge osteotomy was done. Internal fixation using a contoured round hole plate and screws was done. Per operatively the bone was soft to the cut with a thin cortex and rectangular features. Healing progressed with little visible callus, and the patient was full weight bearing 3 months postoperatively, with full knee ROM. Her recent follow up twelve years postoperatively showed no deformity progression (Figure 1).The patient is married now with 3 normal daughters.

*Dept. of Orthopedic, Ad-Diwaniyah Medical College, Al-Qadisiyah University

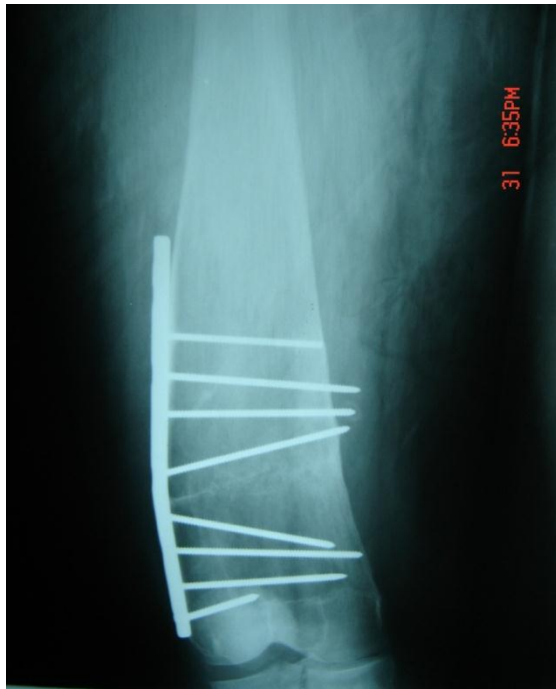


Figure (1) depicting osteotomy healing in case No. one.

Case No.2:

A 45 year old female patient presented with progressive left lower thigh and knee pain. The patient noticed increasing swelling in the lower thigh of 2 months duration , she reported her symptoms as due to an (activity in a wedding event). The patient has an affected sister with the same disorder. Her X-rays showed typical Erlenmeyer- Flask deformity of all tubular bones with no facial features and normal spine radiological features. There was a fracture line in the Rt. Femoral supracondylar region . She had 20 degree supracondylar femoral flexion deformity (Figure 2).There was abundant callus formation surrounding the fracture (Figure 3).The patient refused a corrective osteotomy and was treated with cast immobilization for 30 days with resolution of pain.



Figure 2 depicting fracture mal alignment with abundant callus formation in case No. two.

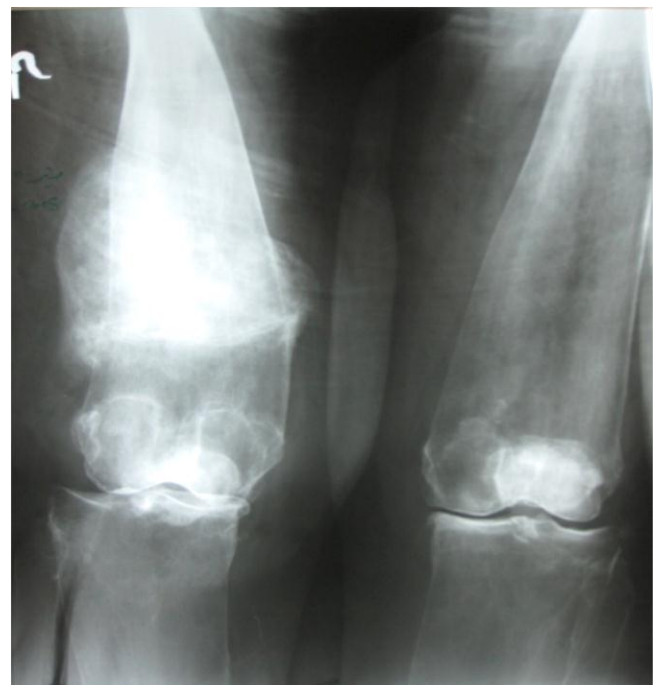


Figure 3 depicting a supracondylar femoral fracture treated conservatively with abundant callus formation in case No. two.

Case No3:

A 23 year old male patient presented with lower leg local pain and tenderness after a sport activity .He was referred because of an abnormal (bone shape) of his tibiae bone. X-ray showed a classical Erlenmeyer flask shape deformity with no visible fracture line. The patient was treated as a case of insufficiency fracture with resolution of pain on cast immobilization for one month.



Figure 4 depicting the radiologic AP and Lateral views of the leg in case No. 3.

Case No. 4:

An 11 year old male patient presented at the age of two years because of delayed walking with bilateral swollen elbows. Six months later he presented with Rt. sided Monteggia fracture with posterior angulations' of the fractured ulna and radial head dislocation. The patient had an abnormal swelling of bone ends with the typical Erlenmeyer deformity and marked osteopenia .Shortly after, the patient presented with left thigh pain and proximal thigh swelling. X-ray showed a left femur subtrochanteric fracture with varus angulations' and shortening (figure 5) .There was abundant callus surrounding the fracture site. At the most recent follow up, the patient has both elbows with short flared and angulated ulna and persisted bilateral radial head dislocations.



Figure 5 depicting the subtrochanteric femoral fracture with abundant callus in case No. 4.

Case No. 5:

A 32 year old male patient presented with frequent bilateral lower legs pain controlled with analgesics .His X-rays showed typical Erlenmeyer –Flask deformity of all tubular bones with normal facial features and no remarkable vertebral radiological features (Figures 6 and 7)



Figure 6 depicting the radiologic AP view of both legs in patient No. five.

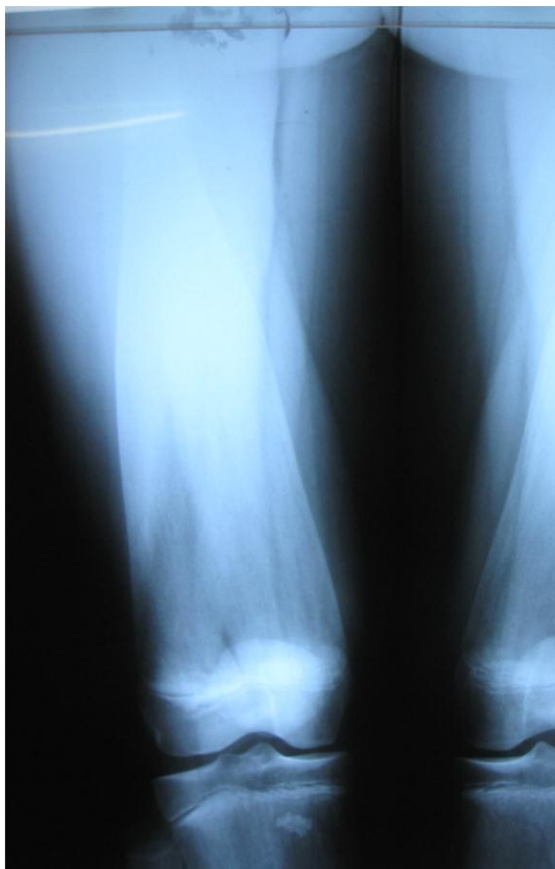


Figure 7 depicting the radiologic AP view of both femora in case No. five.

Discussion:

Pyle's disease is a rare skeletal dysplasia with certain radiological and clinical criteria (2,3,4,5,8,10,13,18). The international nosology and classification of genetic and skeletal disorders ; 2006 revision, considers a widened , flared and/or irregular metaphyses as suggesting metaphyseal dysplasias (17), however, this group encompasses a heterogeneous group with possible cranial involvement and certain facial features or with vertebral involvement (platyspondyly) and short stature.(2,4,5,7,12,15,18). During the past decade, enormous progress has been made concerning the biochemical and genetic basis of skeletal dysplasias. The frequently used radiological identification is being supplemented with biochemical-molecular classification with an over 370 distinct type of skeletal dysplasias . The differential diagnosis of distinct types of Metaphyseal dysplasia subgroup based on clinical-radiological criteria seems blurred and further understanding of genetic-molecular basis may enhance prompt recognition and intervention(1,6,11,14,17). Pyle's disease stands as a distinct type of metaphyseal dysplasia with characteristic clinical-radiological criteria. This is suggested by defective long bone remodeling with the Erlenmeyer flask deformity

,normal body height and proportions and indistinct facial or spine features (4,7,18).The literature is blurred in describing a heterogeneous group of metaphyseal dysplasias with concomitant cranial and spondylotic features. These different types may have different genetic –molecular background and different prognosis (1,4,6,7,11,14,17).

With the above mentioned clinical-radiological criteria , five cases were recognized that met these features, furthermore , these patients presented with distinct clinical presentation regarding the mechanical and fragility characteristics of bones involved .Two patients presented with pain after repetitive ,low energy activity less than that required to fracture a normal bone. They had fracture lines at metaphyseal ends of the femur with abundant stabilizing callus that kept them ambulating , but with pain. The widened bone ends distributes forces over greater surface area and decrease stresses at fracture surfaces. Both of these patients had angular malalignment of the fractures with interestingly little remodeling power over time as presented in patient No. four. Two patients presented with pain in the lower legs after sport activity or walking long distances. The clinical presentation suggests an insufficiency fracture, however with indistinct radiological features.

Another common presentation is the Genu valgum deformity that may warrant osteotomy correction (3).The soft nature of bone with thin cortices that is soft to the cut is characteristic , internal fixation is applicable ,but the poor screws purchase and less than sound fixation may necessitate additional protection. Osteotomy may be rapid to heal with the widened bone surfaces , the patient has painless full weight bearing ambulation three months postoperatively .With the more than 10 years follow up of the 1st case there is no further progression of the valgus deformity. While postosteotomy healing was documented in Pyle's original patient(7) , Eric , presented a case in which bilateral osteotomies were performed through the proximal tibia for progressive genu –valgus deformity .He used external fixation to hold the osteotomy that healed within 12 weeks(3). The soft bone characteristics in Pyle's disease may predispose the patient to insufficiency fractures with repetitive activities. Protection against potential angular malalignment is important until there is sound healing of the fractured bone. Osteotomy for deformity correction may be safely performed and fixed with adequate healing potentials. A word of caution may be appropriate in the young patient indulged with sports activity.

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