**Pyles disease - a case report**

Kapil Ghorpade¹, Sunny Pazhayattil²

**Abstract**

Pyle’s disease is also known as metaphyseal dysplasia, Pyle-Cohn syndrome, Bakwin-Krida syndrome.² It is a rare genetic skeletal disorder which is conventionally classified with craniotubular dysplasias. The case report a 6 yr boy with Pyle’s disease is presented. He had mild facial dysmorphism, dental malformation & mild genu valgum. Skeletal radiology revealed the characteristic Erlenmeyer flask sign³ at distal femoral and proximal tibial metaphysis. The case is reported because of the rarity of the condition.

**Key words:** Bakwin-Krida syndrome, Pyle’s disease, Metaphyseal dysplasia, Erlenmeyer flask sign

**Introduction**

Pyle’s disease is a rare autosomal recessive skeletal disorder with mild clinical manifestations in contrast with the radiological appearances of gross metaphyseal defect. The disorder was first reported by Pyle in 1931 as “a case of unusual bone development”.¹ The same case and his affected sister were restudied by Bakwin and Krida who designated the disorder ‘familial metaphyseal dysplasia’.² Its confusion with Craniometaphyseal dysplasia was clarified by Gorlin et al.³ The term ‘metaphyseal dysplasia’ was used in the 1983 Paris Nomenclature and in deference to popular convention, the eponym Pyle was added in brackets.

There has been considerable confusion with Craniometaphyseal dysplasia which is far more common. This paper presents a case report and review of literature about Pyle’s disease.

**Case report**

The boy aged 6 yrs, came to us with complaints of deformity of bilateral knee joints and restricted extension of bilateral elbow joints. There was no history of trauma or fever. Milestones achieved normally according to age. On examination the child was of average build, mental status was normal for his age. Cranial nerve examination was normal. He was having dental malocclusion. Local examination of knee joints revealed mild bilateral genu valgum deformity. There was terminal restriction of extension of elbow joints bilaterally. Spine, hip, shoulder and wrist examination were normal. Radiography of both knee joints revealed uniform broadening of bilateral metaphysis of proximal distal femur & proximal tibia showing Erlenmeyer flask sign.(Figure 2) There was broadening of metaphysis of other long bones.(Figures 2,3,5) Genu valgum in this case is due to sinuous bowing of Tibia.(Figure 4)

There was minimal sclerosis of the base of skull. Spine radiograph were normal. Haemogram, biochemical, serological investigations were within normal limits.

**Discussion**

There are considerable numbers of disorders
both genetic & acquired, which are associated with metaphyseal
flaring. Pyle's disease is a rare disorder with striking radiographic
manifestations in contrast with the relatively normal clinical
features. It has an autosomal recessive inheritance. Clinically
genu valgum develops early in life, dental malocclusion is
common. Mild limitation of extension of elbows is present.
Muscle weakness & joint pain are common. There may be mild
scoliosis. There may be somewhat greater tendency to fracture
the bones.

Figure. 2. X-ray showing broadening of the metaphysis of Femur.

Figure. 3. X-ray showing broadening of the metaphysis of Tibia.

Figure. 4. X-ray showing broadening of the metaphysis of Radius
and Ulna.

Figure. 5. X-ray showing broadening of the metaphysis of Humeras.

Radiographically the distal femur & proximal tibia shows
Erlenmeyer flask appearance. The humerus exhibits
undermodeling of proximal two thirds, while radius & ulna are
undermodeled at distal one third. The tubular bones of hands
show distal flaring of metacarpals & proximal flaring of
phalanges. In spine mild platyspondyly, dorsal scoliosis may
present. Marked thickening of the ribs, clavicles and ischial bone
is evident. The skull is only mildly involved. There is distinct
supraorbital bulge, occasionally mild hyperostosis of the vault and often mild prognathism.

It should be differentiated from Craniometaphyseal dysplasia, which is characterised by occult hypertelorism and broadening of the base of nose, increased bone sclerosis and often narrowing of neural foramina leading to mixed type deafness. In autosomal recessive form, facial paralysis and defective vision are common sequelae. Radiographically flaring of metaphysis is milder than Pyle’s disease. There is diaphyseal sclerosis. In skull frontal, paranasal, occipital sclerosis is common feature.5

Persons with Pyle’s disease are often asymptomatic but genu valgum & sequelae of fractures may require surgical correction.5 We are reporting one more case of Pyle’s disease with bilateral genu valgum.

References