

A rare association of Pyle disease with ventricular septal defect

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ABSTRACT

Pyle's disease is a rare autosomal recessive disorder manifested by metaphyseal dysplasia. It is characterized by defect in metaphyseal remodeling that leads to grossly widened metaphyses of long bones with marked cortical thinning and osteoporosis, especially in the distal end of femur and proximal tibia. The other features include genu valgum, broadening of the long bones extending into the diaphyses, widening of the ribs and clavicles, platyspondyly, and cortical thinning. We report this rare disorder in a 5-year-old boy associated with ventricular septal defect which has never been reported in the literature. It may be an incidental finding or some association with this rare disorder.

Key words: *Dysplasia, Metaphyseal, Pyle, Ventricular septal defect*

Pyle disease is a rare autosomal recessive disorder manifested by metaphyseal dysplasia and few phenotypic features. 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]. It is characterized by 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 the femur and proximal tibia. The other features are facial dysmorphism, genu valgum, widening of the ribs, and mild skull involvement with hyperostosis of the vault. We report a rare association of Pyle's disease with ventricular septal defect (VSD) in a 5-year-old male child.

CASE REPORT

A 5-year-old boy presented to OPD with history of bony swelling over right wrist, left ankle, right knee with restriction of movements at ankle and knee joint for last 1-year. The swelling was insidious in onset, progressive in nature. There was no history of trauma, fever, and pain. The child was delivered by normal vaginal route and cried immediately after birth. The antenatal and postnatal period was uneventful. The initial milestones were normal. There was no family history of similar illness. The child has a good scholastic performance.

The examination revealed a weight of 18 kg (25th percentile), height of 115 cm (25th percentile), and head circumference of 50 cm. The upper segment to lower segment ratio was normal for age. The vital parameters revealed pulse of 92/min, respiratory rate of 28/min, temperature of 98°F, SpO₂ of 96% in all 4 limbs, and blood pressure of 92/64 mmHg with normal jugular venous pressure. Local examination of right wrist revealed 3 cm×2 cm bony swelling, and similar swelling was seen at left ankle and right knee with restriction of movements at right knee and left ankle joint. The cardiovascular system revealed normal S1 with pansystolic murmur (grade III/VI). The other system was essentially normal.

The investigations revealed normal complete blood count and biochemistry. The chest X-ray was normal. The X-ray of right wrist, left ankle, and right knee revealed splaying of metaphysis with thinning of cortex at the end of long bones and relative constriction of central portion of shafts (Figs. 1-3). The lower end of femur shows Erlenmeyer flask deformity on the right side (Fig. 4). The X-ray of skull and thoracolumbar region was essentially normal (Fig. 5). The echocardiography showed perimembranous VSD of 4 mm with normal pulmonary vasculature.

Based on the clinical features and radiological evidence diagnosis of Pyle's disease with VSD was made. This case report highlights the rare association of Pyle's disease with VSD which has never been reported in the literature.

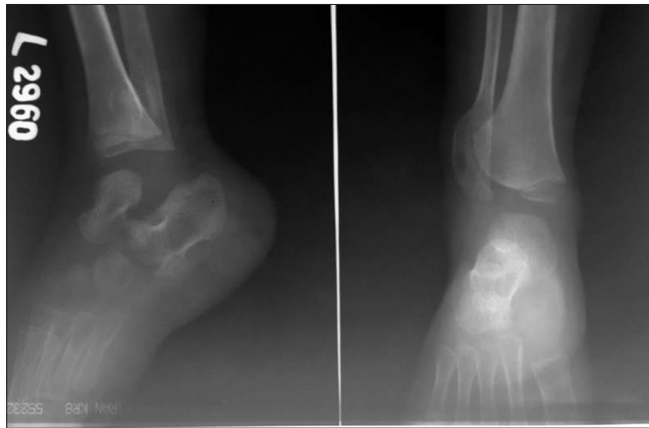


Figure 1: Lower end of fibula showing splaying of metaphysis with thinning of cortex



Figure 2: Lower end of ulna showing splaying of metaphysis with thinning of cortex

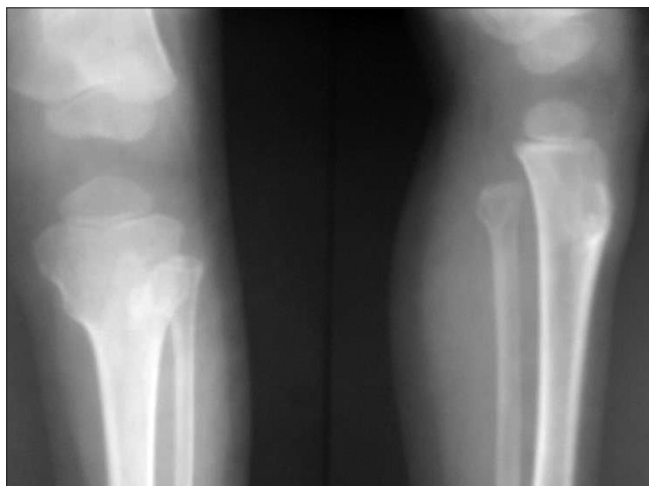


Figure 3: Upper end of tibia showing splaying of metaphysis with thinning of cortex

DISCUSSION

Pyle metaphyseal dysplasia has typical X-ray findings with relatively less phenotypic features. It is manifested by defect



Figure 4: Lower end of femur showing Erlenmeyer flask deformity

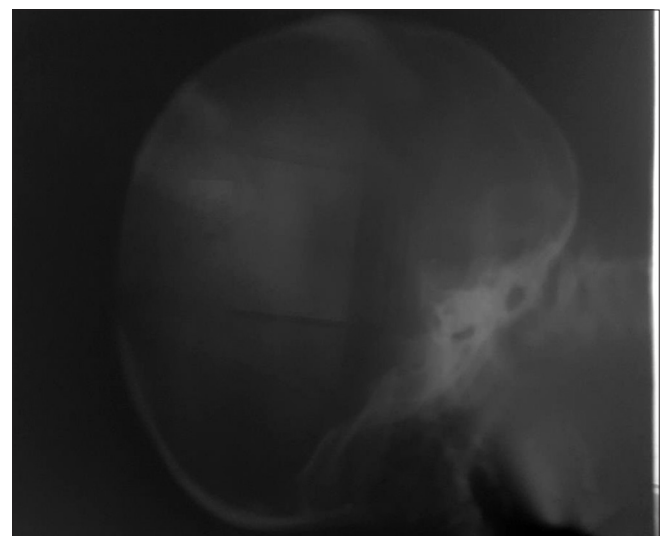


Figure 5: Normal X-ray of skull

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 proximal tibia. Spinal involvement varies from moderate platyspondyly to biconcave lens appearance of the vertebral bodies [3,4]. Jaw involvement has also been documented. Skeletal survey of obligate heterozygotes may show minor skeletal changes. Along with these roentgenographic changes, there may be few clinical signs and symptoms such as muscle weakness, joint pain, genu valgum, scoliosis, and limited extension of elbow. The genu valgum deformity can be corrected by surgery [5,6].

It should be differentiated from craniometaphyseal dysplasia, which is characterized by occult hypertelorism and broadening of the base of nose, increased bone sclerosis, and often narrowing of neural foramina leading to mixed type deafness. The other differential diagnosis includes Gaucher disease, osteopetrosis, thalassemia, and Niemann-Pick

disease. Since our patient did not have anemia, jaundice, hepatosplenomegaly, increased bone density, or any history of blood transfusion, so the last four possibilities were ruled out. The presence of ventricular septal defect makes this case report unique, which further increases the association spectrum of the disease.

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