International Journal of Farmacia

Journal Home page: www.ijfjournal.com

ISSN: 2455-8108

IJF |Volume 8 | Issue 3 | July - Sept - 2022
www.ijfjournal.com

CASE STUDY *Disease*

A REVIEW ON PYLE DISEASE

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ABSTRACT

Metaphyseal dysplasia, or Pyle disease is a disorder of the bones. It is a rare disease in which the outer part of the shafts of long bones is thinner than normal and there is an increased chance of fractures. Its hallmark feature is an abnormality of the long bones in the arms and legs in which the ends (metaphyses) of the bones are abnormally broad; the shape of the bones resembles a boat oar or paddle. The broad metaphyses are due to enlargement of the spongy inner layer of bone (trabecular bone). Although trabecular bone is expanded, the dense outermost layer of bone (cortical bone) is thinner than normal. As a result, the bones are fragile and fracture easily. The bone abnormalities in the legs commonly cause knock knees (genu valgum) in affected individuals. Other bone abnormalities can also occur in Pyle disease. Affected individuals may have widened collar bones (clavicles), ribs, or bones in the fingers and hands. Dental problems are common in Pyle disease, including delayed appearance (eruption) of permanent teeth and misalignment of the top and bottom teeth (malocclusion).

Keywords: Pyle disease, Genetics, Diagnosis management, Advanced treatment methods

INTRODUCTION

Pyle's disease is a metaphyseal dysplasia of benign course, inherited with an autosomal recessive pattern. Pyle disease is an innocuous autosomal recessive disorder in which mild clinical manifestations contrast with the radiological appearances of gross metaphyseal under modelling. It was described in 1931 by Edwin Pyle. an orthopaedic surgeon from Connecticut (U.S.A.), on a 5 year old boy who presented with severe genu valgum in both knees and cranial deformities. [1][2]

There is striking clinic radiological dissociation, with mild clinical manifestations. Genu valgum is the most common feature on physical examination and is frequently the only consistent finding. Others findings that may be observed on physical examination include bilateral and symmetrical enlargement of the knee, proximal two-thirds of the humerus, distal two-thirds of the radius and ulna, as well as the proximal phalanges and distal metacarpal bones. Others clinical manifestations include caries, poor denal implantation, malocclusion, manibular prognathism, limited extension of the elbow, muscle weakness, arthralgia. Some cases may presented deformities such as scoliosis.[3]

Synonyms

Metaphyseal, Pyle's syndrome, Pyle-cohn syndrome, Pyle diease, Bakwin-krida syndrome [1]

Signs and symptoms

Despite its rarity clinical interest in PD has recently risen on discovering its relationship with mutations in a soluble inhibitor of the Wnt5,6,7 pathway. Analysis of the phenotype of these patients and the events observed in mouse models which reproduce the dysfunction of the gene sFRP reveal a differential control in the homeostasis of cortical and trabecular bone. We now present a case with distinctive images of the cortical and trabecular bone and the relevant interest of the clinical symptoms of these patients is commented upon. [4][5]

It is an autosomal recessive disorder in which mild clinical manifestations contrast with radiological appearances of gross metaphyseal under modeling. Most patients present with mild genu valgum. The elbows are unable to extend fully. There may be widening of the lower femora and clavicles. Bones can sometimes be fragile, but fracturing is usually not common. Patients may present with dental caries, mandibular prognathism, spinal alignment, and disproportionate limb lengthening. Mental development, physical development, and height are usually normal. Most patients with Pyle's diseases will have symptoms that vary from person to person. Symptoms are.5%–29% of people have Delated eruption of teeth, Other people may have Abnormality of thorax, Absent paranasal sinuses, Arthralgia, Caries teeth, Genu valgum, Hypoplastic frontal sinuses, Limited elbow extension, Mandibular prognathia, Metaphysealdyplasia, Muscleweakness[6].

CASE REPORT

A 53-year-old woman was referred to the radiology department complaining of genu valgum and joint pain. Her family had noted widening of the knees at the age of 8 years, which progressed over time. There was no functional disability. The patient sought medical care several times, receiving the diagnosis of osteoarthritis with no further investigation. Over the last year, she had developed a more pronounced arthralgia that was not relieved by common analgesics. There was no visual, auditory, cognitive, or psychomotor disturbance. The most evident finding on physical examination was the marked genu valgum (Fig.1) [7].

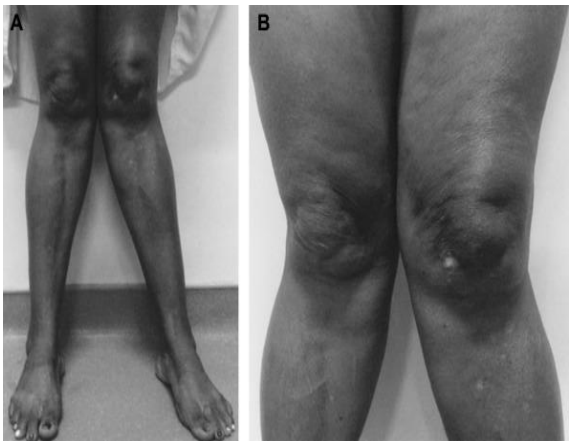


Fig.1 (A) Genu valgum deformity. Fig.1 (B) Zoomed-in view. Greater detail of the bilateral widening of distal thighs and knees, accompanied by genu valgum

Fig 1: Photographs of the lower limbs of patient.

Mild facial dysmorphism, prognathism, and prominent ears and frontal bossing were also noted. The patient showed partial edentulism with caries and poor hygienic condition of the remaining teeth. There were no signs of anemia, jaundice, organomegaly, fractures, or motor disturbances.

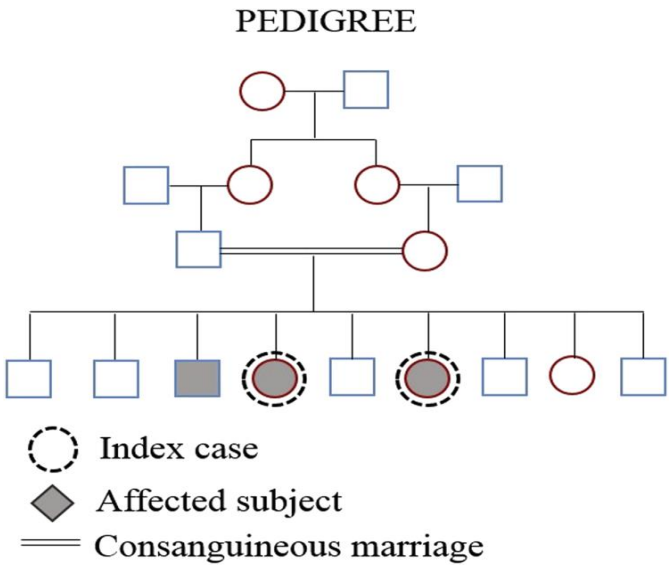
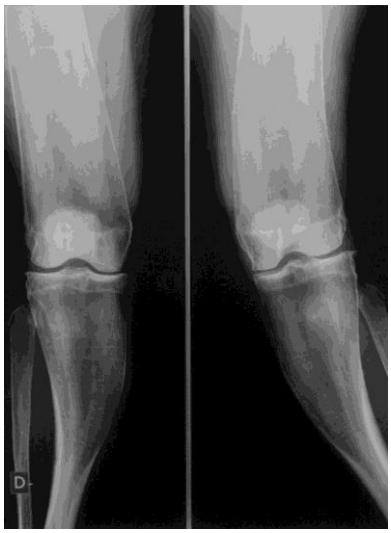


Fig 2: Pedigree

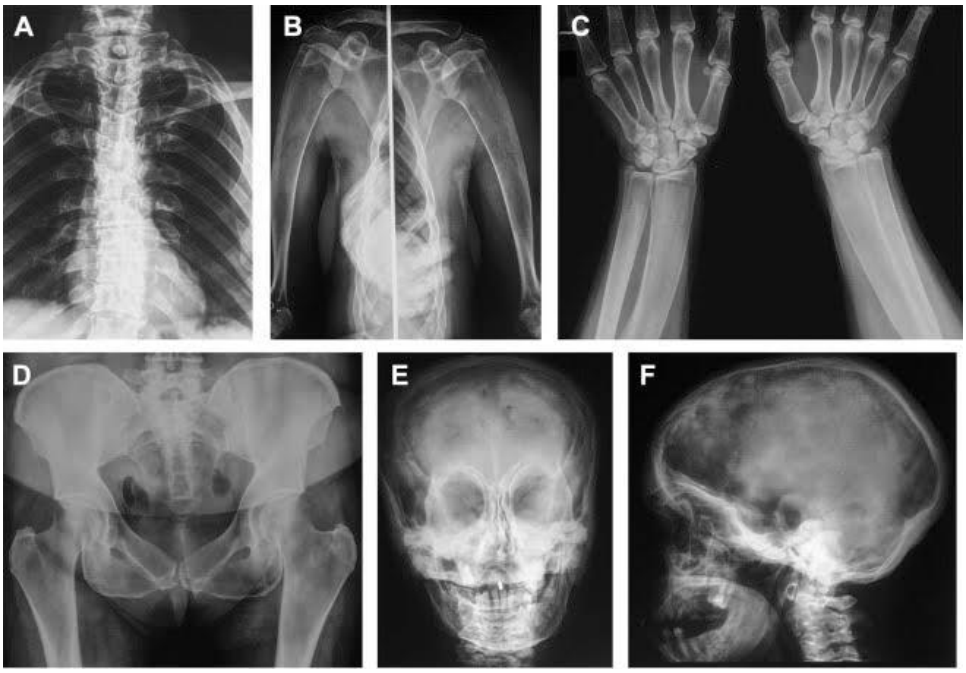
Parental consanguinity is demonstrated, and of the 9 offspring's, 3 are affected. Family history reveled parental consanguinity and similar phenotypic characteristics in 2 of her 8 siblings. The parents had no musculoskeletal deformities. Pedigree. Parental consanguinity is demonstrated, and of the 9 off springs 3 are affected (fig.2)



Conventional knee radiographs in anteroposterior view showing Erlenmeyer flask deformity, characterized by femorotibial metaphyseal flaring, cortical bone thinning, and genu valgum.

Fig 3: Knee radiographs of patient 1.

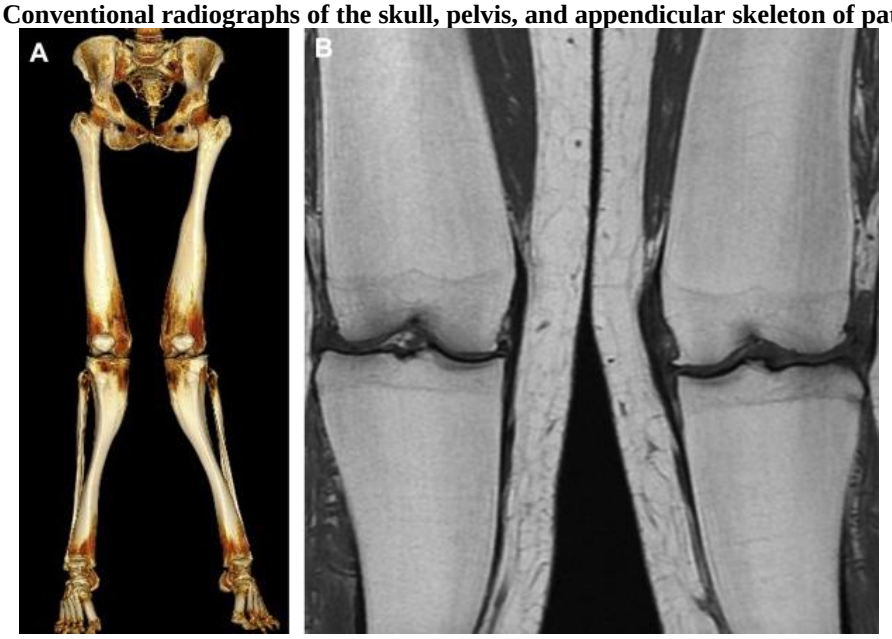
Laboratory tests did not show any significant alterations. Conventional radiography plain films of the patient's knees revealed Erlenmeyer flask deformity, characterized by marked femorotibial metaphyseal flaring, and associated cortical bone thinning.(fig.3) The additional radiographic survey, which included other long bones, skull, and spine, exposed the symmetry and the systemic nature of the skeletal disorder. Most of the long bones showed under tubulation and loss of their usual morphology. The proximal portions of the clavicles and ribs were expanded (fig.4A), as well as the proximal two-thirds of the humerus, which presented an arcuate shape (fig.4B). Other tubular bones shared similar characteristics, such as the distal portions of the radius and ulna, proximal and distal portions of the fibula, head of the metacarpals, and base of the phalanges (fig.4C).The ischiopubic rami were also diffusely enlarged, thereby narrowing the obturator foramina (fig4D). Despite striking changes in tubular bones, skull radiographs showed only mild basal sclerosis, mandibular prominence, and poor pneumatization of paranasal sinuses and mastoid air cells (fig.4E and F).



(A) Proximal metaphyseal widening of the clavicles and ribs. (B) Proximal humeral metaphyseal expansion and bowing. (C) Under modeling of the distal radius and ulna, as well as the proximal phalanges and distal metacarpal bones. (D) Expanded morphology of the ischiopubic rami and narrowed obturator foramina. (E) And (F) Skull radiographs in anteroposterior and lateral

views showing mild basal sclerosis, mandibular prominence, and poor pneumatization of the paranasal sinuses and mastoid air cells. Preoperative three-dimensional tomographic reconstruction of the lower limbs for surgical planning provided detailed analysis of the Erlenmeyer flask deformity and highlighted the S-shaped aspect of the tibia (Fig. 5A)

Fig 4: Conventional radiographs of the skull, pelvis, and appendicular skeleton of patient 1.



(A) Three-dimensional tomographic reconstruction of the lower limbs and pelvis showing detail of the Erlenmeyer flask deformity, as well as the S-shaped aspect of the tibia. Note the narrowing of the obturator...Additonal T1- weighted magnetic resonces imaging (MRI) of knees revealed normal bone marrow signal intensity (fig.5B) [7]

Fig 5: CT and MRI of the lower limbs of patient 1.

Epidemiology

Pyle diease is thought to be a rare disorder, although its prevalence is unknown. More than 25 cases have been described in the medical literature. it has been described in four german families originating from the same town in Bohemia and in a 7-year-old japanese girl[8].

Diagnosis

There are two clinical molecular genetic test that are available to those thought to have pyle's disease. These test are sequence analysis of the entire coding region and deletion/duplication analysis.[9]

Differential diagnosis

Pyle disease may be confused with craniometaphyseal dysplasia. The two however are clinically, radiographically and genetically distinct from one another. [10]

Treatment

People with pyle disease are often asymptomatic. Dental anomalies may require orthodontic intervention. Skeletal anomalies may require orthopedic surgery.[11]

CONCLUSION

Pyle's metaphyseal dysplasia is a rare genetic diease with an estimated prevalence of less than one case per million. Until the 1980s, there were only 20 cases described in the literature, including countries like the USA, France, Germany, South Africa, India and Italy ,with fewer than 30 geniune cases reported to date .the mutation involved is still unknown, with a lack of comprehensive genetic studies on the subject.

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