Pyle Disease

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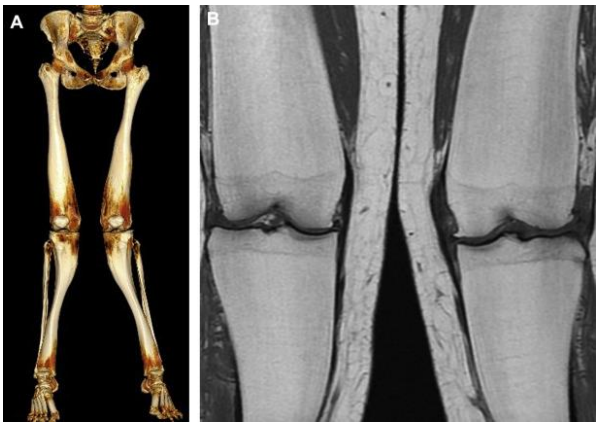
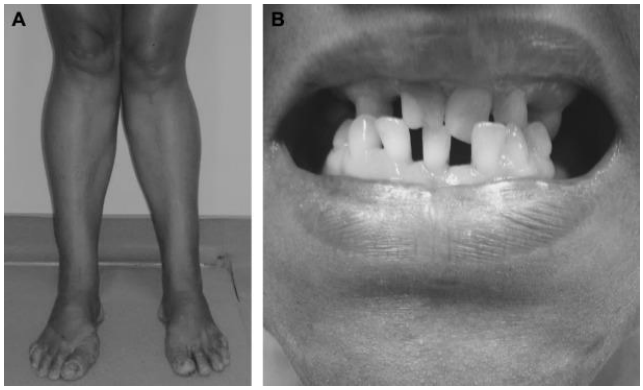
Pyle disease is also known as a "metaphyseal dysplasia" due to a rare, inherited bone disorder that causes abnormalities in the metaphyseal area of the long bones of the upper and lower extremities, especially, the arms and legs. The near-ends of the bones, called metaphysis, become abnormally wide, giving the bones a boat oar or a paddle shape. This is due to an enlargement of the spongy inner layer of bone, while the dense outer layer thins, making the bones fragile and prone to fractures. In the legs, these abnormalities can lead to knock knees, or genu valgus.



Paddle shapped appearance of the metaphysis of the long bones

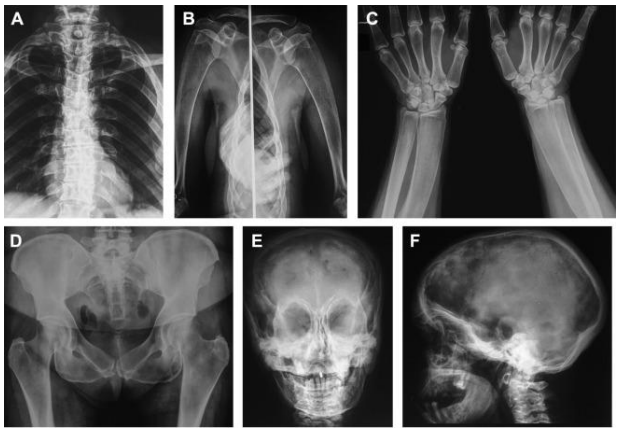
Most cases of Pyle disease are asymptomatic and do not require any treatment. Occasionally, a "genu valgus deformity" can be so pronounced that it requires an orthopedic intervention, like a corrective osteotomy. Other bones and joints deformities can be seen contributing to a lack of elbow extension, or to abnormalities of the thoracic cavities. There may be also widened metaphyses of the long bones with cortical thinning. A sclerosis of the cranial bones as well as dental malformations or delay in eruption of the teeth may be seen. Occasionally, some dental abnormalities may also require repair or corrections. Arthralgia with hypoplastic frontal sinus and absent paranasal sinuses can be seen.



Genu Valgus deformity and delay in erruption of teeth

Pyle disease is an autosomal recessive disorder caused by a bi-allelic loss of function mutations on the “SPRP4”gene located on chromosome 7p14. The gene codes for secreted frizzled-related protein 4 which plays a role in bone remodeling. Biological parents can pass down the genetic mutations which cause or increase the chances of their child to develop the disease which is inherited in an auto-recessive pattern.

The disease maybe also be called “metaphyseal chondrodysplasia” or “metaphyseal dysostosis “which is a rare autosomal chondro-dysplasia characterized by the flaring of the metaphyseal area of the long bones with a relative constriction and sclerosis of the diaphysis. When the disease is also present with a mild cranial sclerosis, it is termed “Craniometaphyseal dysplasia”. This is one of the causes of what is called an “Erlenmeyer flask deformity”. The metaphysis become relatively lucent and patients may remain asymptomatic unless they have also a genu valgus deformity which in itself may also require surgical correction if patient is symptomatic.

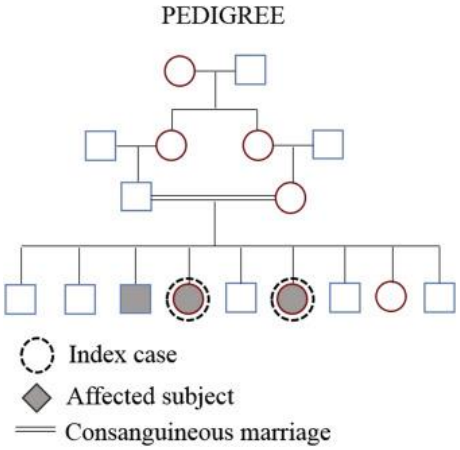


Proximal femur deformity similar to “Legg-Calve-Perthes disease”, with Sclerosis at the base of the skull.

Rarely, the deformity may interest the proximal femur giving the appearance of an osteonecrosis of the proximal femur often seen in Legg-Calve-Perthes disease. By it autosomal recessive genetic pattern of inheritance, Pyle's disease is a rare skeletal disorder with a benign course. Fewer than 30 cases are described in the world literature to date. It has been seen in children issued of consanguineous unions, in which Erlenmeyer flask deformity was discovered at the femoro-tibial metaphyses with typical flaring of the long bones. This rare entity requires appropriate genetic counseling when discovered with concomitant genu valgus deformity.

With an autosomal recessive pattern of inheritance, it is expected to observe a benign course of the disease. We know also little about possible mutations because of the paucity of the cases. The clinical findings and the radiological studies revealing the bony deformities allow us to discover the striking skeletal deformities discussed above but important to asset a diagnosis on these rare cases.

Appropriate genetic counseling has also its place in the evaluation of such rare disease.



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