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Congenital Pseudarthrosis of the Tibia a Rare Pathology Presenting As Posterior Bowing: A Case Report

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Abstract
Congenital pseudarthrosis of the tibia (CPT) is a rare pathology with reported incidence between 1:140,000 and 1:250,000. The natural history of the disease is extremely unfavourable and once a fracture occurs, there is a little or no tendency for the lesion to heal spontaneously. Histologically tibia shows area of segmental dysplasia resulting in anterolateral bowing of the bone Treatment is mainly surgical. The key to get primary union is to excise hamartomatous tissue and pathological periosteum. Each child with CPT must be followed up till skeletal maturity to identify and rectify residual problems after primary healing. We are illustrating a case of severe neonatal form with posterior bowing at birth.
Keywords- *Congenital pseudarthrosis, Fibrous dysplasia, Hamartoma.*

Introduction

Congenital pseudarthrosis of the tibia (CPT) is a rare pathology, which is usually associated with neurofibromatosis type I (NF1). The reported incidence of Congenital pseudarthrosis of the tibia (CPT) varies between 1:140,000 and 1:250,000 and bilateral forms are extremely rare[1]. The disease usually becomes evident within a child's first year of life but can have a later onset where the anterior bowing starts between the ages of 4 and 12 years [2]. Severe neonatal forms the signs are present at birth. Histologically tibia shows area of segmental dysplasia resulting in anterolateral bowing of the bone. There is a marked cuff of fibrous tissue in the area of pseudarthrosis, also called the fibrous hamartoma[3], in continuity with abnormal periosteal thickening. Most of the cells of this hamartoma have been identified as fibroblasts [1]. The osseous dysplasia leads to a tibial nonunion and, because of tibial bowing and reduced growth in the distal tibial epiphysis, shortening of the limb usually occurs[2]. The natural history of the disease is extremely unfavourable and once a fracture occurs, there is a little or no tendency for the lesion to heal spontaneously. Treatment is mainly surgical. The key to get primary union is to excise hamartomatous tissue and pathological periosteum.

Case Report

Patient encountered during the routine rounds of postnatal ward. Neonate was born by normal vaginal delivery, all the antenatal records along with ultrasonography were unremarkable. Posterior

bowing of the left leg was noticed (Fig. 1). X-ray was done which revealed posterior bowing of both tibia as well as fibula along with an increase in cortical density and narrow sclerotic medulla (Fig. 2). Although anterior bowing is present in majority of cases but the diagnosis of congenital pseudarthrosis type III was made as per Crawford classification and referral to higher institution was made for early surgical intervention before the child starts the weight bearing.

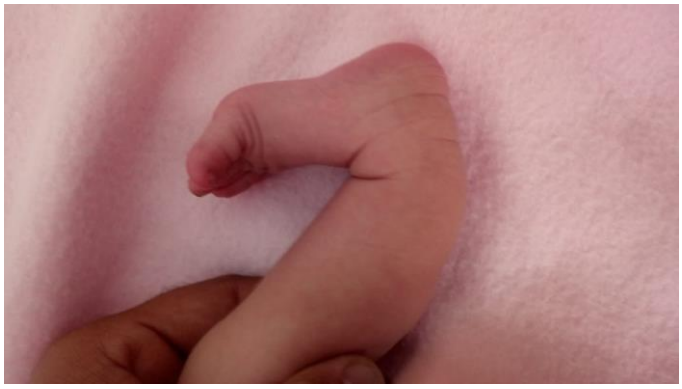

Fig.1: Posterior bowing of leg in a neonate.

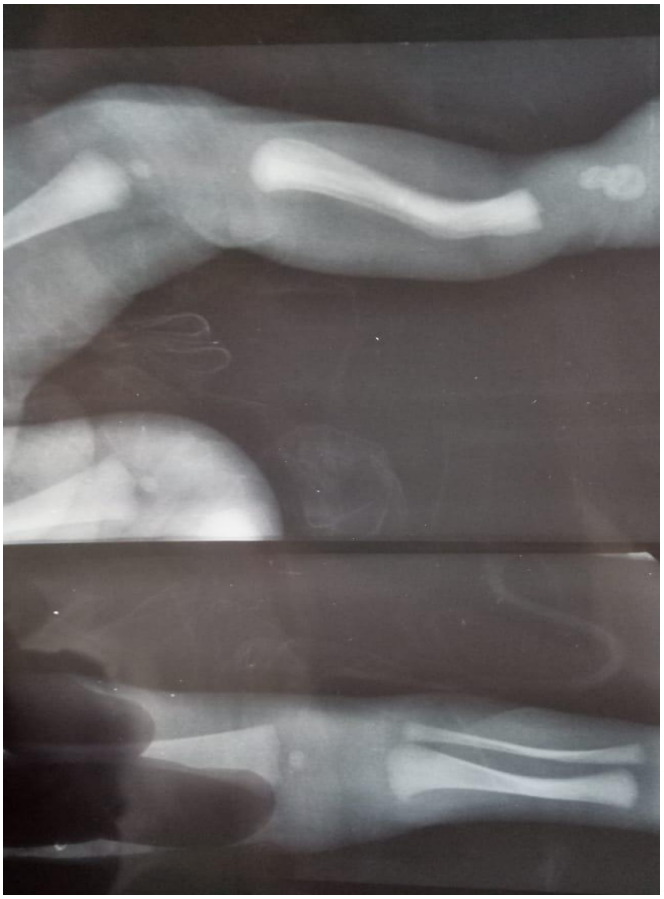

Fig.2 X-ray findings showing posterior bowing of both tibia as well as fibula along with an increase in cortical density and narrow sclerotic medulla.

Discussion

Congenital pseudarthrosis of tibia is associated with osseous dysplasia which is mostly unilateral, located at the junction of the middle and distal thirds of the tibial segment. There is no predilection for sex or side. The fibula also affected in more than half the cases. The disease has association with neurofibromatosis type I, fibrous dysplasia and congenital ring syndrome. While the incidence of bowing and CPT is less than 4% in NF1[4], half of the patients (40–80%) presenting with CPT are NF1 carriers[4],[5]. Clinically bowing may be appreciated at birth or in the first weeks in life which is usually convex anterolaterally or as a discontinuity between the two bones of the tibial segment[6]. Severe neonatal forms or primary pseudarthrosis, in which signs are present at birth, can be distinguished from secondary pseudarthrosis, which is revealed by a pathological fracture when the child begins walking[7]. Our case illustrated a contrasting feature of posterior bowing of both tibia and fibula in a severe neonatal form.

There are several classification systems, of them Crawford classification[5], which is the most frequently used today (Table 1) as it has the advantage of being descriptive and identifying the different stages as CPT progresses.

Table 1: Crawford classification

type I: anterior bowing with an increase in cortical density and a narrow medulla;
type II: anterior bowing with narrow, sclerotic medulla
type III: anterior bowing associated with a cyst or signs of a prefracture
type IV: anterior bowing and a clear fracture with pseudarthrosis often associating the tibia and fibula;

MRI is the investigation of choice and it describes the morphology of the pseudarthrosis and the adjacent soft tissue more precisely than a standard radiographs. The area of the pseudarthrosis is hyper intense on fat-suppressed and T2-weighted images and slightly hypo intense on T1-weighted images with contrast enhancement after administration of gadolinium. The periosteum in the area of the pseudarthrosis appears as a thickened soft-tissue layer. The natural history of the disease is extremely unfavourable and once a fracture occurs, there is a

little or no tendency for the lesion to heal spontaneously. Treatment is mainly surgical and it aims to obtain a long term bone union, to prevent limb length discrepancies, to avoid mechanical axis deviation, soft tissue lesions, nearby joint stiffness, and pathological fracture. The key to get primary union is to excise hamartomatous tissue and pathological periosteum. The prognosis of CPT has changed considerably in the last few decades after the advent of vascularised fibular transfers and the Ilizarov technique. Age at surgery, status of fibula, associated shortening, and deformities of leg and ankle play important role in primary union and residual challenges after primary healing. Despite these advances, multiple surgical interventions are often necessary to obtain union of the pseudarthrosis, and the risk of amputation is never entirely eliminated. Each child with CPT needed to be followed up until the skeletal maturity to identify and rectify residual problems after primary healing.

Conclusions

Although the congenital pseudarthrosis of the tibia (CPT) is a rare pathology and in severe neonatal forms the signs may be present since birth but a meticulous antenatal ultrasound in second trimester may detected the milder deformities. Early detection of the entity can facilitate early surgical intervention for the better outcome.

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