Clinical Quiz

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Congenital pseudoarthrosis

Clinical Presentation

An 18-month-old male baby was brought to our Outpatient Department with complaints of congenital bowing of his right leg. The patient's mother also gave history of trivial trauma to his right leg at 6 months of age, following which the patient was unable to bear weight on the right extremity. The baby was a first child of the family, born at-term by spontaneous vaginal delivery. Antenatal history was typical. The parents denied any family history of skeletal abnormalities and consanguinity. Examination revealed anterior bowing and shortening of the right leg (**Figure 1**). There was free and painless abnormal mobility present at distal third of the leg. Any attempt to bear weight on right extremity increases the deformity. There were also patches of pigmentation over the trunk (**Figure 2**). There were no features suggestive of dysmorphism. There was no other associated skeletal deformity, and the rest of the systems were within normal limits. Laboratory investigations including complete blood count, erythrocyte sedimentation rate, serum calcium, serum phosphate, and serum alkaline phosphatase were within normal limits.

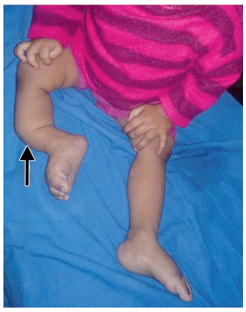

Figure 1 - Clinical photograph of patient showing shortened and deformed right leg (arrow).

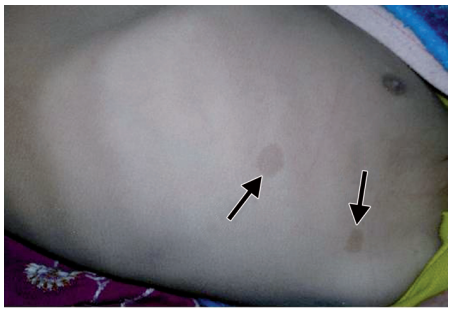

Figure 2 - Clinical photograph of patient showing patches of hyperpigmentation over the trunk - café-au-lait spots (arrow).

Questions

1. What are the patches of pigmentation over the trunk called?
2. What is the diagnosis?
3. What is the management?

Clinical Quiz

Answers

1. Patches of pigmentation over trunk are called as café-au-lait spots. There is a strong association between congenital pseudoarthrosis of tibia (CPT) and neurofibromatosis type I (NF-I). The CPT develops in approximately 6% of patients with NF-I. On the other hand, 55% of patients with CPT were found to have NF-I.[1]

2. The clinical diagnosis of patient is congenital pseudoarthrosis of tibia (CPT). Congenital bowing of leg, inability to bear weight following trivial trauma, increase in deformity on attempt of weight bearing, presence of free painless abnormal mobility at distal third of leg with patches of pigmentation over trunk - café-au-lait spots (**Figure 2**) favors diagnosis of congenital pseudoarthrosis of tibia.

3. Congenital pseudoarthrosis of the tibia poses one of the most challenging management problems in pediatric orthopedics. The goal of treatment is to achieve union and maintenance of union till skeletal maturity. Bracing the extremity before occurrence of pathological fracture is recommended as an early measure for treating CPT. Brace is to be worn at all times, especially during weight bearing and its use is continued all the way through skeletal maturity.[2] Following occurrence of pathological fracture, surgery is the mainstay of treatment. The aim of surgery is to excise the diseased segment of bone and bridging the defect with stable fixation. Intramedullary fixation with nails, bridging the defect with free vascularized fibular graft, and Ilizarov external fixation are amongst the most commonly used means of achieving union in CPT.[2-4]

Discussion

Congenital pseudoarthrosis of the tibia refers to non-union of a tibial fracture that develops spontaneously or after insignificant trauma in a dysplastic bone segment of the tibial diaphysis. In most of the cases the disease develops during the first two years of life. Nevertheless, there are reports of cases in which disease developed before birth and reports of late-onset pseudoarthrosis. The etiology of CPT remains unclear. However, there is a strong association between CPT and NF-I.[1] Several theories have been proposed to explain the etiology of disease (intrauterine trauma, birth fracture, generalized metabolic disease, and vascular malformation), but none of the theory is able to explain the cause. Based on the infrequent reports in the literature of familial and bilateral involvement, some authors have anticipated that CPT is hereditary.[1,2] Congenital pseudoarthrosis of the tibia poses one of the most challenging management problems in pediatric orthopedics. The goal of treatment is to achieve union and maintenance of union till skeletal maturity. Histopathological analysis at the diseased site reveals hyperplasia of fibroblasts with the formation of dense fibrous tissue. The hyperplasic fibrous tissue is located in the periosteum and between the broken bone ends, surrounding the tibia leading to compression, osteolysis, and persistence of pseudoarthrosis.[1,2] Consequently, management of pseudoarthrosis and the associated deformity are on a gamut.

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