IMAGES IN MEDICINE

Pseudoarthrosis in neurofibromatosis type-1

A 27 year old man presented with multiple papules, nodules, and hyperpigmented spots on the body, which he had had since early childhood. In addition, he had a deformity of the right forearm. Examination revealed multiple large sized café-au-lait macules, axillary freckling, and soft cutaneous neurofibromas (mollusca fibrosa). The right forearm was grossly shortened with a length of only 12 cm (fig 1). It presented a globular appearance and on palpation the forearm bones had a break in their continuity. There was hypermobility at

Figure 1 Shortening of the right forearm with globular deformity, multiple cutaneous neurofibromas, and café-au-lait macules over the shoulder.

Figure 2 Anteroposterior radiograph of the right forearm showing pseudoarthrosis, dislocation of humeroulnar joint, and osteoporosis.

the wrist and elbow joints in addition to abnormal movements through the forearm. The left forearm was 28 cm in length and bones were normal. Radiological examination revealed pseudoarthrosis in the mid-part of right forearm involving both the radius and ulna, dislocation of humeroulnar joint, radioulnar diastasis, and a decrease in the bone density (fig 2). On ophthalmological examination, there were multiple bilateral Lisch nodules. A diagnosis of neurofibromatosis type-1 and pseudoarthrosis as a complication was thus established.

Pseudoarthrosis, congenital or acquired, is usually associated with neurofibromatosis type-1 or fibrous dysplasia. Rarely, no cause may be found in the congenital type when it is regarded as idiopathic. Pseudoarthrosis of long bones occurs in about 13% of patients with neurofibromatosis type-1. However, more than 50% of the patients with pseudoarthrosis have been observed to have some or other manifestation of neurofibromatosis type-1. Pseudoarthrosis of tibia is not uncommon in neurofibromatosis type-1 but involvement of forearm bones is extremely rare and only 60 cases of pseudoarthrosis of forearm bones had been reported up to 1999. In forearm bones, pseudoarthrosis involves the ulna alone most commonly, followed by the radius alone, and rarely both bones may be affected. Pathogenesis of pseudoarthrosis, irrespective of its aetiology is not clearly understood. It has been postulated that neurofibromatous tissue or some abnormality in the soft tissue surrounding the site of pseudoarthrosis plays some part in its development. However, histology of tissues taken from affected site has failed to confirm this. It is one of the most difficult conditions to treat as multiple surgical approaches have been applied repeatedly without much success. Recently use of vascularised bone grafts have shown some ray of hope.

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