Pseudoarthrosis of the hand in neurofibromatosis type 1: a case report

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Neurofibromatosis (NF) is a disorder with a wide spectrum of clinical manifestations. Here, we describe a 16-year-old boy with NF1 who had pseudoarthrosis of the 4th and 5th fingers of the left hand. He had specific cutaneous lesions and Lisch nodules in the iris. Because NF1 affects multiple organ systems, patients are likely to benefit most from a multidisciplinary treatment strategy.

Key words: neurofibromatosis type 1, pseudoarthrosis, hand.

Neurofibromatosis type 1 (NF1) is one of the most common genetic disorders, affecting 1 in 3,000 individuals, and is an autosomal dominant trait linked to chromosome 17. However, about 50% of the cases arise sporadically as new mutations[1].

In order to definitively diagnose NF1, a patient must have two of the following characteristic features: more than six café-au-lait macules greater than 5 mm, neurofibromas, axillary or inguinal freckling, optic nerve glioma, Lisch nodules, a distinctive osseous lesion, and family history of NF1 in a first-degree relative. However, patients with NF1 may present with a wide variety of lesions.

Orthopedic manifestations of NF1 including bony dysplasia, pseudoarthrosis and scoliosis, are found in 10-25% of all individuals with NF[12]. Congenital pseudoarthrosis of the tibia is the most common types of pseudoarthrosis. Pseudoarthrosis in conjunction with NF1 has also been noted to occur with less frequency in other long bones, such as the humerus, radius, ulna, and clavicle[3,4].

Here, we describe a 16-year-old boy with NF1 who developed pseudoarthrosis of the 4th and 5th fingers and metacarpals in the left hand.

Case Report

A 16-year-old boy presented with curvature deformity of the 4th and 5th fingers of the left hand since his first year of life. His parents were non-consanguineous. The developmental milestones were normal.

On his examination, vital signs including blood pressure were normal. More than six café-au-lait spots over 15 mm in diameter could be seen throughout his skin. Axillary and inguinal freckles and subcutaneous neurofibromas were also noted. Ophthalmic evaluation showed bilateral Lisch nodules in the iris. On the neurological examination, there was curvature deformity of the proximal phalanges of the 4th and 5th fingers of the left hand. None had any pain or sensory loss. The remainder of the physical and neurological examination was unremarkable. With these findings, he diagnosed with NF1.

Radiographs of the left hand revealed pseudoarthrosis of the 4th and 5th metacarpocarpal and 5th metacarpophalangeal joints. Metacarpal and proximal phalangeal bone enlargement and intraosseous lytic lesions of the os hamatum, os capitatum and proximal portions of the 3th, 4th and 5th metacarpals (neurofibroma bone involvement) were also noted (Fig. 1).

Discussion

Neurofibromatosis type 1 (NF1) is a genetically transmitted disorder characterized by abnormalities of the skin, nervous tissue and bone. These may include café-au-lait spots, neurofibromas, Lisch nodules, optic gliomas, osseous lesions, macrocephaly, short stature, and mental retardation. Pseudoarthrosis of

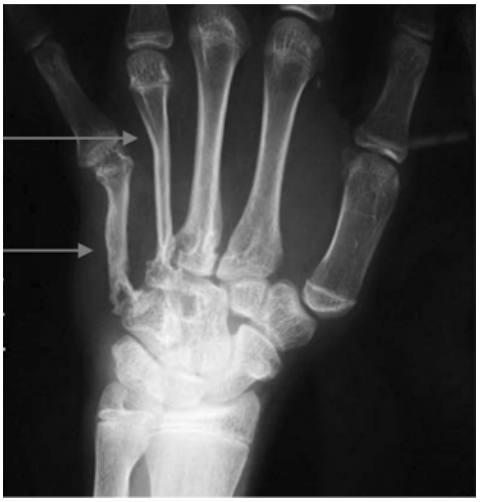

Fig. 1. AP radiographs demonstrating a pseudoarthrosis of the 4th and 5th finger of the left hand in a 16-year-old boy.

the tibia, dystrophic kyphoscoliosis, spinal manifestations, protrusio acetabuli, dislocation of the hip, hypertrophy of a limb, atlantoaxial dislocation, and tendon ruptures may also occur[5,6].

Most children with NF1 have no major orthopedic problems. For those with musculoskeletal involvement, the most important issue is early recognition. In the NF1 population, congenital tibial dysplasia, spinal deformity, and disorders of excessive bone and soft tissue growth are the three types of common musculoskeletal manifestations that require evaluation. Although only 5% of patients with NF1 are diagnosed with congenital tibial dysplasia, 75% of patients with congenital tibial dysplasia have NF12. Diagnosis of this clinical condition is simple, and nearly one-third of patients are reported shortly after birth; the remaining were detected after trauma. The trauma in these cases becomes just an incident that directs the attention of the patient to the underlying problem.

The physiopathology of pseudoarthrosis has still not been defined clearly. However, it probably involves the various different hypotheses, with signal anomalies causing increased osteoclast activity associated with an osteoblastic differentiation anomaly resulting in a bone remodeling defect, which is also favored by the decrease in local vascularization[7,8]. None of them provides an entirely satisfying explanation for the pathogenesis of pseudoarthrosis or its location, probably because of the heterogeneity of the disease and its inconsistent association with NF1.

There are a few previously published reports of NF1 with pseudarthrosis of the humerus, radius, ulna, and clavicle[2,4]. Congenital pseudarthrosis of other bones, such as ulna, radius, and pelvis, is rare. Maffulli et al.[9] reported ulnar pseudarthrosis with NF1 in four children. Kameyama et al.[10] described an 11-year-old child with pseudoarthrosis of the radius associated with NF1. Our patient with NF1 had pseudoarthrosis of the 4th and 5th fingers and metacarpals in the left hand and NF1. To our knowledge, this is the first report of pseudoarthrosis of the fingers and metacarpals in the hand in a pediatric patient with NF1.

Neurofibromatosis type 1 (NF1) affects multiple organ systems, and a multidisciplinary approach to treatment is required. Management of the orthopedic manifestations of NF1 is often difficult. Careful implementation of strategies to manage musculoskeletal disabilities can vastly improve the quality of life in patients with NF1.

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