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CASE REPORT

Congenital Pseudarthrosis of Index Metacarpal Bone Treated with Distraction Osteogenesis Followed by Autologous Grafting

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Introduction

The term "congenital pseudoarthrosis" refers to the semblance of a false joint formation which is present from birth. The soft tissue at the pseudoarthrotic site is composed of a variable admixture of fibrous tissue, fibrocartilage, and hyaline cartilage with evidence of enchondral ossification. Spaces and clefts are lined by a synovial-like tissue [1].

Congenital pseudoarthrosis is mostly seen in the lower extremity, particularly involving the tibia, fibula and femur [2]. In upper extremities, it may also involve clavicle and one or both bones of the forearm [3, 4]. However, metacarpal bone involvement is exceedingly rare with only three previous case reports in current literature [5–7]. But, to the best of our knowledge, there have been two cases of bilateral congenital pseudarthrosis of index metacarpals reported in literature up to date [5, 6]. Congenital pseudoarthrosis is usually associated with genetic syndromes and presented as a component of several other abnormalities [2]. In 1950, Aegerter suggested a possible relationship between neurofibromatosis, congenital pseudarthrosis, and fibrous dysplasia [8].

In 1949, Moore stated that "the treatment of congenital pseudarthrosis will probably never be entirely effective until the etiology has been determined. Osteosynthesis by bone-grafting still offers the principal means of attacking the problem" [9]. The treatment of congenital pseudoarthrosis is challanging because achieving complete union of pseudoarthrosis site, providing an acceptable alignment and functional extremity is difficult and refracture rate is high. This is probably due to low osteogenic capacity of the affected whole segment. The principle of the treatment is complete resection of the pseudoarthrosis, grafting the bone defect and ensure bony union either with internal or external fixation. Currently Ilizarov external fixation has gained wide acceptance as a gold standard treatment for the management of congenital tibial pseudoarthrosis [2].

Herein, we intend to present a rare case of bilateral congenital pseudoarthrosis involving bilateral index metacarpal bones. This is not a unique case and previously two other cases has been published. But what makes our case different than previous cases is the treatment of choice. We have treated this patient with excision of the pseudoarthrosis, gradual distraction to overcome the short metacarpal length without neurovascular compromise, followed by tricortical grafting and internal fixation.

Case Report

A 21-year-old male patient was referred to the orthopaedic outpatient clinic with complaints of shortness, pain during activity and motion limitation of both index fingers since he was a baby after a normal pregnancy. There was no history suggestive of any teratogenic exposure in the first trimester nor any adverse events in the antenatal, perinatal, neonatal periods or any consanguinity in the parents. No kind of medical treatment had been previously applied. No additional comorbidities or blood analysis abnormalities were apparent. There was no family history of such a deformity or of neurofibromatosis.

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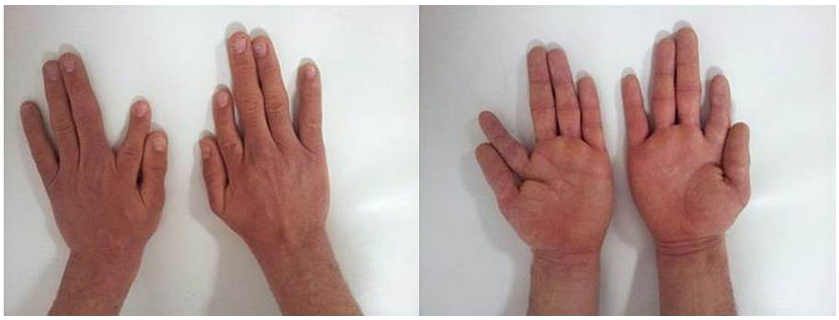

Fig. 1 Initial clinical examination

Physical examination revealed bilateral second metacarpal head retroplacement, causing short index fingers and radial deviation in the left index finger, with slight flexion limitation and pain during motion in the second metacarpophalangeal joints (Fig. 1). The wrists and the other fingers were all normal. No brownish spots were apparent on his whole body which may resemble "café au lait" spots. On palpation the second metacarpal bones were rigid, and there was painful bossing on the dorsal aspect of the hand. There was a slight painless, passive motion at the bosses between the proximal and distal segments. Neurovascular examination was normal. Radiological findings revealed discontinuity of the second metacarpal bones with corticated margins that showed pseudarthrosis of bilateral index metacarpal bones with broadening, shortening, and disruption in the metacarpophalangeal joints (Fig. 2). The remaining metacarpals and the rest of the hand bones appeared to be radiologically normal.

To remove the painful pseudoarthrosis and to lengthen the index metacarpal bones to improve appearance and motion, surgery was recommended. With the knowledge about acute lengthening and grafting, it was decided to make gradual distraction and iliac tricortical grafting. Informed consent was obtained from the patient and a two-step-operation was planned beginning with the right hand. In the first operation, the non-union area of the right index metacarpal bone was explored through a dorsal approach and the well vascularized fibrocartilogenous tissue was excised and sent for histopathological examination. A monolateral external distractor with two Schanz screws in each segment was applied through the dorsolateral aspect to protect the extensor tendon (Fig. 3). On postoperative Day 1 distraction was started at 0.5 mm per day by making a quarter turn of the distractor twice daily. The pathological result was joint formation of cartilage covered with synovium (Fig. 4).

When the soft tissue limitation for distraction (18 mm of distraction) (Fig. 5) was reached, the process was ceased and the second step was undertaken. In the second operation, the distractor was removed and the gap between the fragments was filled with contralateral tricortical iliac crest graft. After providing a proper alignment of the metacarpal bone, internal stabilization with plate and screws was performed. The hand was immobilized in a short arm resting splint cast for 3 weeks, after which the patient received 3 weeks of physiotherapy.

After 6 months he had full range of motion and good callus formation (Fig. 6). After 1 year the graft union was solid and the hand was able to stand full weight-bearing (Fig. 7). The patient was pain-free, and happy with the surgical approach which had been taken for his hand. It is planned to apply the same procedure to the left hand.

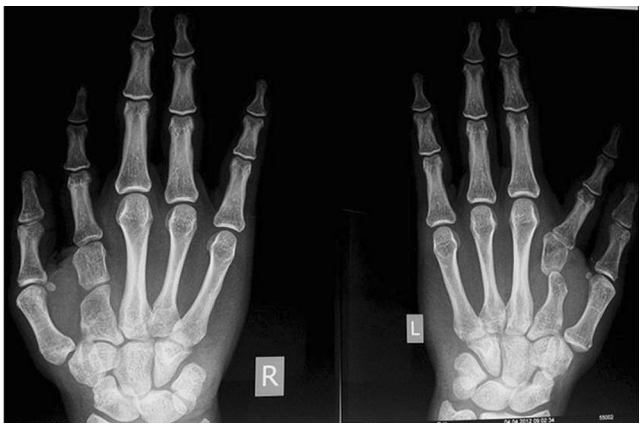

Fig. 2 Bilateral congenital pseudoarthrosis of the second metacarpal bones with broadening, shortening, and disruption evident in the metacarpophalangeal joints

Discussion

A shortness in a metacarpal bone can be related to many diseases. When a line is drawn along the heads of the fourth and fifth metacarpals, if that line does not intersect with the head of the third metacarpal then this is called a "positive metacarpal sign or Archibald's sign" [10] which shows a short fourth metacarpal, possibly related to disorders such as achondroplasia, Turner syndrome, various types of dwarfism, gigantism, hyperparathyroidism, pseudohypoparathyroidism, or pseudo–pseudo-hyperparathyroidism [11].

When the London Dysmorphology Database was searched for syndromes associated with congenital pseudoarthrosis, only five syndromes were found: neurofibromatosis Type 1, Noonan features–neurofibromatosis, oral–facial–digital syndrome Types one and two, and osteofibrous dysplasia [7].

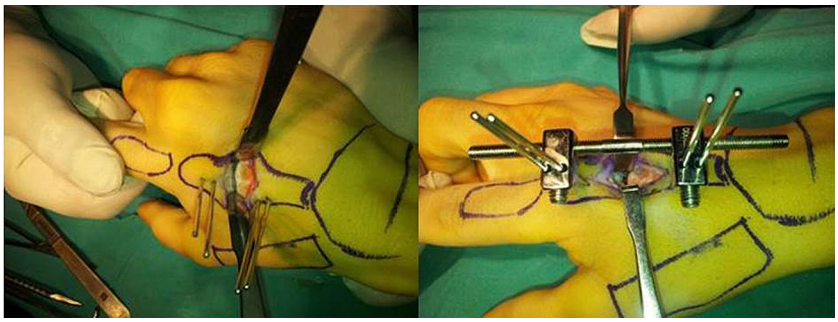

Fig. 3 Exploring pseudoarthrosis and applying the distractor through the dorsolateral aspect

However, the pseudoarthrosis in these conditions mostly involves the lower limb bones. Therefore, this report can be considered to make a new contribution to the congenital pseudoarthrosis issue.

Shortness in the other metacarpals can also be seen in various disorders, including Basal cell nevus syndrome, Beckwith–Wiedemann syndrome, Biedmon syndrome, Larsen syndrome, multiple exostoses syndrome, epiphyseal dysplasia, tricho-rhino-phalangeal syndrome, cri-du-chat syndrome, Russell–Silver dwarfism, Patterson–Lowry rhizomelic dysplasia or or other congenital problems like pseudarthrosis [12].

Devriendt et al. in 2000, and Koenig et al. in 2005 reported cases of bilateral short second metacarpals with an additional phalanx as a part of other symptoms in a new syndrome [13, 14]. Demirci et al. reported that the epiphyseal hypertrophy of the first metacarpal can simulate pseudarthrosis in Fanconi's anaemia [15].

Incecik et al. described a 16-year-old boy with Neurofibromatosis Type 1(NF1) who developed pseudoarthrosis of the 4th and 5th metacarpals and metacarpophalengeal joints in the left hand. He had specific cutaneous lesions and Lisch nodules in the iris. To their knowledge, this was the first report of pseudoarthrosis of the fingers and metacarpals in the hand in a pediatric patient with NF1. No treatment was discussed in this article [7].

Deviation of the index fingers can also be seen in Desbuquois syndrome and Catel–Manzke syndrome. In Desbuquois syndrome, the deviation of the index fingers is caused by an additional phalanx between the metacarpal and the proximal phalanx of the index finger [16], whereas in Catel–Manzke syndrome, the deviation of the index fingers is caused by duplication of the proximal phalanx [17].

In a very recent study, Ranganath and Dalal reported a case of bilateral congenital pseudoarthrosis of the second metacarpals [5]. However, in that case the patient also had camptodactyly, cleft palate, short stature, advanced bone age, genu valgum, and borderline intellectual dysfunction that led them to think it was a variant of Devriendt syndrome.

The most similar case in literature to the case reported here, was the first reported case of bilateral congenital pseudoarthrosis of the index finger metacarpal bones, reported by Taheri et al. in 2009 [8]. The left hand of an 8-year-old girl with bilateral congenital pseudarthrosis of the second metacarpal bones was treated with corticocancellous iliac graft and K wire fixation. The K wires were removed after 6 weeks in a short palmar splint, and good results were reported after 6 months. The case presented here differs from that one as we applied gradual distraction with an external fixator and made the fixation with mini plate and screws then applied a splint for 3 weeks.

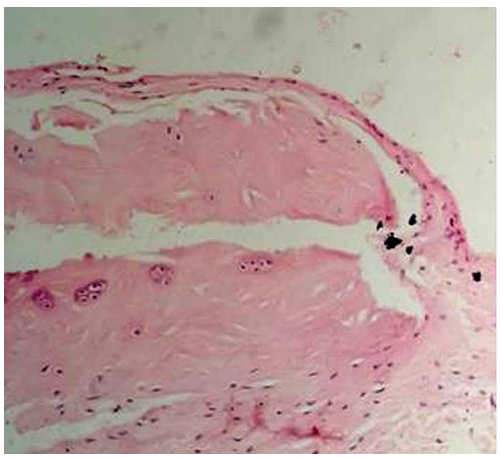

Fig. 4 The pathological view of pseudarthrotic joint: formation of joint with cartilage covered with synovium (H&E ×200)

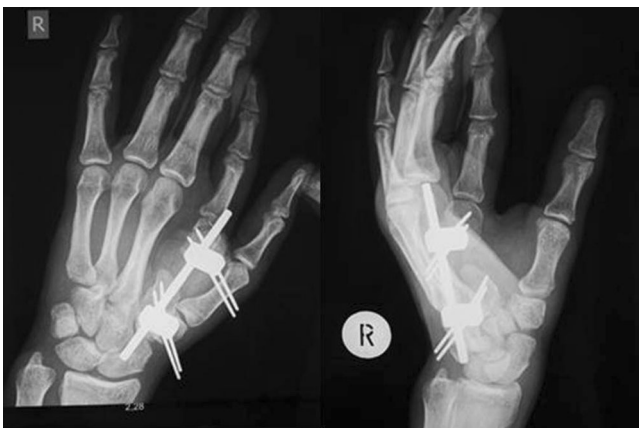

Fig. 5 Distraction with the external fixator

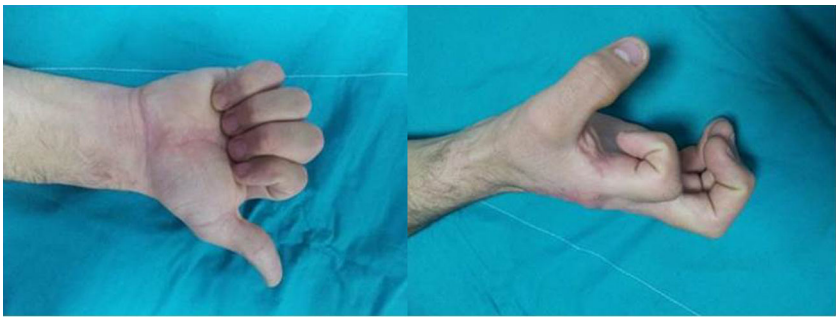

Fig. 6 Functional outcome after 6 months

Distraction osteogenesis has been used by several authors for metacarpal lengthening [18, 19]. In some cases, complications such as stiffness, angulation, subluxation of the metacarpophalangeal joint and delayed union or non-union have been associated with this procedure if certain rules are not followed such as protection of the periosteum, refraining from distraction rates of more than 2×0.25 mm/day and, avoiding lengthening the bone more than 40 % or not more than 20 mm of the preoperative bone length [19]. In the current case, solid union was obtained with full range of motion.

To the best of our knowledge, the case reported here is the the third case in English literature of bilateral congenital pseudoarthrosis of index metacarpals and the first case of congenital metacarpal pseudoarthrosis treated with gradual distraction and tricortical grafting. Whether it is part of a syndrome is as yet unknown and should be investigated. Although the treatment of congenital pseudarthrosis has been reported to be challenging for other bones, avascular bone grafts may be useful for reconstruction in metacarpals. The application of gradual distraction can be considered a more effective technique for postoperative motion and pain relief, as rigid fixation is also necessary for early motion and this results in fewer complications.

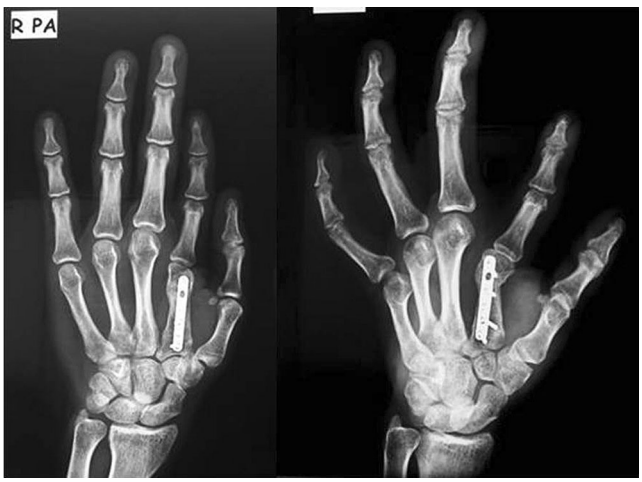

Fig. 7 Solid healing of graft after 1 year

Conflict of Interest None declared.

Source of Funding None declared.

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