Congenital pseudoarthrosis of the radius in Neurofibromatosis Type 1: An entity not to be missed!

Jayakrishnan K. Narayana Kurup, Hitesh H. Shah *

Department of Orthopedics, Kasturba Medical College, Manipal Academy of Higher Education, Manipal, Karnataka, 576104, India

ARTICLE INFO

Keywords:

Congenital pseudoarthrosis
Radius
Neurofibromatosis
Café au lait spots

ABSTRACT

Congenital pseudoarthrosis of the radius is exceedingly rare. We report an isolated pseudoarthrosis of the radius in a five-year-old girl child with Neurofibromatosis type-I (NF-I). She underwent excision of the pseudoarthrosis tissue, ulna osteotomy, iliac crest cortico-cancellous bone grafting, and K wire fixation of both bones. A sound union of the radius and the ulna was achieved after 4 months. At two-year follow-up, mild shortening of the forearm persisted with maintenance of sound union of both bones. Accurate diagnosis of the condition is central in avoiding complications and the need for complex surgical procedures.

1. Introduction

50–80% of patients presenting with congenital pseudoarthrosis of long bones have been associated with an underlying diagnosis of Neurofibromatosis type 1. The prevalence of pseudoarthrosis among NF1 Patients is about 5%.¹ The majority of the cases involve tibia. The incidence of forearm pseudoarthrosis is 2 cases per million population.² Involvement of the ulna is more common than that of the radius. Isolated pseudoarthrosis of the radius is extremely rare. They usually present with progressive deformities of the arm from birth or following a trauma. Bone dysplasia from NF1 also can lead to impaired healing. Delay in the diagnosis of the condition in a growing child can result in a progressive deformity of the forearm, shortening, reduced grip and pinch strength, and loss of range of movement of the elbow and wrist. Herein we report a case of pseudoarthrosis of the left radius in a 5-year-old girl child who presented with deformity of the forearm following a fracture of radius 4 years back. She was diagnosed with NF1 on further examination.

2. Case report

A five-year-old girl presented to us with nonunion of the left radius after she had sustained a fracture following a fall at the age of one year. She had a progressive deformity of forearm and difficulty in using the limb for daily activities (Fig. 1). She was previously operated twice elsewhere for the radius fracture. The previous surgeries were open reduction and k-wire fixation followed by repeat open reduction and

bone grafting after a few months. On further detailed examination, the child was found to have multiple café-au-lait spots and was diagnosed to have Neurofibromatosis Type-I (Fig. 2). She had a shortened forearm with a radial deviation of the left wrist. Wrist range of movements, pronation, and supination of the forearm were severely restricted. Erythrocyte Sedimentation rate was 13 mm/hr, and C-reactive protein was 1 mg/L. Forearm radiographs revealed nonunion with pseudoarthrosis formation of the shaft of the left radius with an intact ulna (Fig. 3). There was dorsal subluxation of the distal ulna at the wrist. She underwent excision of the Pseudoarthrosis tissue, ulna osteotomy, iliac crest bone grafting (5 cm) of both radius and ulna, and intramedullary K wire fixation (Fig. 4). She went on to achieve a complete union of the radius and the ulna in 4 months (Fig. 5). The intramedullary k-wires in the radius and ulna were retained. Physiotherapy was started, once the union was achieved. At two-year follow-up, there was excellent remodeling of both radius and ulna. There was approximately 3 cm of shortening of the forearm compared to the opposite side. The wrist and elbow range of movements improved, and she was able to use the upper limb for all her routine activities.

3. Discussion

Pseudoarthrosis of the long bone is extremely rare in children. It is associated with a pathological lesion. Pseudoarthrosis is often associated with NF1, and hence children presenting with non-union or pseudoarthrosis should be thoroughly examined for features of NF1. The typical features include Café-au-lait spots, Axillary freckling, Lisch

* Corresponding author.

E-mail address: hiteshshah12@gmail.com (H.H. Shah).

<https://doi.org/10.1016/j.jor.2020.09.013>

Received 19 July 2020; Accepted 20 September 2020

Available online 23 September 2020

0972-978X/© 2020 Professor P K Surendran Memorial Education Foundation. Published by Elsevier B.V. All rights reserved.

Fig. 1. 1A and 1B shows the shortening and severe radial deviation deformity of the left forearm.

Fig. 2. Showing multiple large Café au lait spots over the anterior aspect of the trunk.

nodules, and neurofibromas. In a systematic review conducted by Siebelt et al., 74% of cases of congenital pseudoarthrosis of forearm had underlying NF1.³ Missing NF1 can prolong the time for adequate treatment of the fracture or nonunion and can lead to an established pseudoarthrosis with complications like deformity, shortening, stiffness, and loss of range of motion of the adjacent joints. The child in this report had a delay in the detection of congenital pseudoarthrosis. The initial

injury was treated as an uncomplicated radial diaphyseal fracture and later by re reduction, bone grafting, and fixation. The diseased hamartomatous tissue was not excised, which resulted in failure to unite and establishment of the pseudoarthrosis.

NF1 with congenital pseudoarthrosis mainly affects the tibia. In the forearm, it involves the ulna more than the radius. According to Siebelt et al. only 22 cases of isolated radius congenital pseudoarthrosis have been reported to date.³ Various treatment recommendations are available in current literature for the management of forearm pseudoarthrosis. The treatment modalities include immobilization in a cast, cortico-cancellous bone grafting with or without internal fixation, free vascularized fibula grafting (FBFG), or converting to a one bone forearm.^{4–10} Cast immobilization alone has poorer outcomes in achieving union.³ Bone grafting with or without internal fixation has produced variable results in terms of union.⁹ Multiple attempts of bone grafting may be required for the union. FBFG has produced good results. However, the procedure is technically demanding. Non-unions have been reported even with FBFG.³

The primary aim of established pseudoarthrosis of long bones is to achieve union. Siebelt et al. in their study, has mentioned that osteosynthesis and autologous cortico-cancellous bone grafting should be disregarded as a treatment option due to poor results.³ However, this is not the dictum in the treatment of congenital pseudoarthrosis.

We believe that the key surgical step in achieving union in congenital pseudoarthrosis involves the complete excision of the hamartomatous tissue, including diseased bone and periosteum. The long-standing pseudoarthrosis of the radius would be associated with the deformity of the radius. The intact ulna and the excision of the diseased bone of radius would lead to the gap in between two fragments of the radius. It is necessary to get good contact with the two fragments to achieve the union of the pseudoarthrosis site. The osteogenic potential can be enhanced by a bone graft. The cortical bone resists early resorption and provides stability, while the cancellous bone provides osteoinductive, osteoconductive, and excellent osteogenic potential at pseudoarthrosis site.¹¹ The union can be achieved with adequate size cortico-cancellous bone graft from the iliac crest fixed with intramedullary k-wires or nails. It is necessary to retain intramedullary fixation to maintain the sound union and to prevent the re-fracture. Shortening is common due to the excision of the pseudoarthrosis tissue and acute compression at the pseudoarthrosis site. The shortening of the limb causes less of a problem in the forearm compared to the lower limb. The range of movement of the adjacent joints improves with the deformity correction and the union of the fracture. Prolonged focused physiotherapy is often required to improve the joint function.

This surgical technique is safe, effective and easily reproducible. The vascularized fibula grafting is a complex procedure. It must be reserved

Fig. 3. 3A and 3B depicts the antero-posterior and lateral radiographs of the left forearm showing pseudoarthrosis of left distal radius with dorsal subluxation of the distal ulna.

Fig. 4. 4A and 4B depicts the post operative antero-posterior and lateral radiographs of the left forearm. The entire diseased segment of the radius was resected, ulnar osteotomy was done, cortico-cancellous bone grafting from iliac crest followed by intramedullary fixation of radius and ulna using k-wires. The limb was protected in plaster of Paris cast for six weeks.

for complex and failed cases.

4. Conclusion

Pseudoarthrosis of the isolated radius in a child with NF-1 can be

managed with the excision of the sclerotic diseased bone, excision of the diseased periosteum, osteotomy of the intact ulna, cortico-cancellous grafting, and intramedullary fixation.

Fig. 5. 5A and 5B depicts the antero-posterior and lateral radiographs of the left forearm at 4 month follow-up, showing complete union of the radius and ulna with incorporation of the graft.

Informed consent

Informed consent was obtained.

Disclosure

None of the authors received payments or services, either directly or indirectly (i.e., via his or her institution), from a third party in support of any aspect of this work. No author has had any other relationships, or has engaged in any other activities, that could be perceived to influence or have the potential to influence what is written in this work.

Funding

None declared.

Credit author statement

Jayakrishnan Kelamangalathu Narayana Kurup: Conceptualization, Methodology, Data curation, Writing- Original draft preparation. **Hitesh Shah:** Visualization, Supervision, Validation, Reviewing and Editing.

Declaration of competing interest

None declared.

References

- 1 Stevenson DA, Birch PH, Friedman JM, et al. Descriptive analysis of tibial pseudarthrosis in patients with neurofibromatosis 1. *Am J Med Genet.* 1999;84: 413–419.
- 2 Cheng JC, Hung LK, Bundoc RC. Congenital pseudarthrosis of the ulna. *J Hand Surg.* 1994;19:238–243.
- 3 Siebelt M, de Vos-Jakobs S, Koenrads N, et al. Congenital forearm pseudarthrosis, a systematic review for a treatment algorithm on a rare condition. *J Pediatr Orthop.* 2020;40:e367–e374.
- 4 Witoonchart K, Uerpaiojkit C, Leechavengvongs S, Thuvathakul P. Congenital pseudarthrosis of the forearm treated by free vascularised fibular graft: a report of three cases and a review of the literature. *J Hand Surg Am.* 1999;24:1045–1055.
- 5 Allieu Y, Meyer ZU, Reckendorf G, et al. Congenital pseudarthrosis of both bones: long-term results of two cases managed by free vascularised fibular graft. *J Hand Surg Am.* 1999;24:604–608.
- 6 Gregg PJ, Price BA, Ellis HA, Stevens J. Pseudarthrosis of the radius associated with neurofibromatosis. A case report. *Clin Orthop Relat Res.* 1982;171:175–179.
- 7 Lee KS, Lee SH, Ha KH, Lee SJ. Congenital pseudarthrosis of ulna treated by free vascularised fibular graft – a case report. *Hand Surg.* 2000;5:61–67.
- 8 Cleveland RH, Gilsanz V, Wilkinson RH. Congenital pseudarthrosis of the radius. *Am J Roentgenol.* 1978;130:955–957.
- 9 Beris AE, Lykissas MG, Kostas-Agnantis I, et al. Congenital pseudarthrosis of the radius treated with gradual distraction and free vascularized fibular graft: case report. *J Hand Surg Am.* 2010;35:406–411.
- 10 Mathoulin C, Gilbert A, Azze RG. Congenital pseudarthrosis of the forearm: treatment of six cases with vascularized fibular graft and a review of the literature. *Microsurgery.* 1993;14:252–259.
- 11 Dodabassappa SN, Shah HH, Joseph B. Donor site morbidity following the harvesting of cortical bone graft from the tibia in children. *Journal of children's orthopaedics.* 2010;4:417–421.