Case Report

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Management of an Ulnar Nonunion in a Pediatric Case with Rothmund-Thompson Syndrome

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Abstract

Rothmund-Thomson syndrome (RTS) is a rare autosomal recessive genodermatosis characterized by poikiloderma, short stature, juvenile cataracts, sparse hair, skeletal abnormalities, and a predisposition to osteogenic sarcomas and skin cancers. While numerous skeletal abnormalities have been described in patients with Rothmund-Thomson syndrome, nonunion cases are rarely reported in the literature. In this study, we present the case of a patient with RTS who sustained a combined ulna and radius fracture in the proximal forearm after a simple fall. The patient was followed for four months with titanium elastic nails, but despite union of the radius, nonunion of the ulna was observed. A second trauma later resulted in re-displacement of the old fracture line of the ulna. The patient subsequently underwent stable fixation with plate and screw fixation, received additional medical treatment, and union was achieved. With our report, we hope to increase practitioner awareness that delayed bone healing may be a possibility in RTS patients and to encourage consideration of routine imaging and calcium and vitamin D supplementation.

Keywords: Forearm fracture, nonunion, pediatric injury, rothmund-thompson syndrome

Introduction

Rothmund-Thomson syndrome (RTS) is a rare autosomal recessive disorder with heterogeneous clinical features such as characteristic rash (poikiloderma), short stature, sparse hair, juvenile cataracts, gastrointestinal disorders, and skeletal abnormalities including radial ray defects, with approximately 500 cases reported in the literature to date (1, 2).

RECQL4, which encodes an ATP-dependent DNA helicase, is the only known gene associated with RTS. Pathogenic variants in the gene are found in approximately two-thirds of affected individuals, referred to as Type 2 RTS. Causal genes for Type 1 RTS, which lacks pathogenic variants in RECQL4, have not yet been identified (3, 4).

Skeletal anomalies such as radial hypoplasia/aplasia, congenital dislocations, synostosis, brachymesopharyngia, and patellar defects have been reported in the majority of patients with RTS.(5) Widespread osteopenia, pathological fractures, nonunion, and delayed bone union are also reported in the literatüre (6-8). In this case report, a case of proximal third ulna and radius fracture who underwent revision surgery due to nonunion will be presented.

Case Report

A 12-year-old male patient presented to our clinic with deformity and pain in his left forearm following a fall from a standing position. Examination revealed widespread skin lesions. His height and weight were two standard deviations below his peers (height: 130 cm, weight: 29 kg). Imaging revealed bone and joint deformities in both upper extremities, along with fractures of the ulna and radius in the proximal third of the left forearm. Preoperative evaluation revealed that the patient had been diagnosed with RTS and under follow-up for approximately one year, and was taking isotretinoin for skin lesions. The patient underwent surgery. An intramedullary titanium elastic nail (ITEN) was then applied to the patient's radius and ulna fractures (Figure 1). At the 3-month follow-up, the radius fracture was observed to have healed, but the ulna fracture had delayed healing. At 4 months, the patient returned to our clinic with pain and a new deformity following another fall from standing height on his arm. After examination, tenderness was observed in the forearm, and radiographs revealed complete union of the radius fracture, but nonunion and displacement

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Figure 1. Postoperative radiographs after treatment of both radius and ulna fractures with intramedullary titanium elastic nails (a-b). Radiographs of patient's forearm 4 months after surgery. Nonunion and displacement of ulnar fracture is seen (c-d).

of the ulna fracture (Figure-1). The patient underwent reoperation. Intraoperative evaluation revealed that the radius had complete union, but the ulna had not. Therefore, the decision was made to perform primary osteosynthesis on the ulna, ITENs were removed from both bones, and the ulna fracture was fixed with a locking compression plate (Figure-2). During postoperative follow-up, the patient was started on calcium and vitamin D as medical treatment. One-year follow-up examination revealed complete fracture union (Figure 3). The patient had no pain and functional limitations. His elbow and forearm range of motion were equal to the contralateral side (Figure-3). Written informed consent was obtained from patient's parents fort his study.

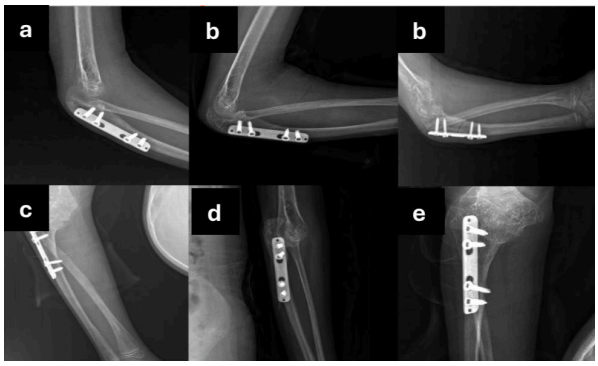

Figure 2. Postoperative radiographs after revision surgery with plate fixation (a-c). Comlete union is seen at postoperative 12th month (c-e).

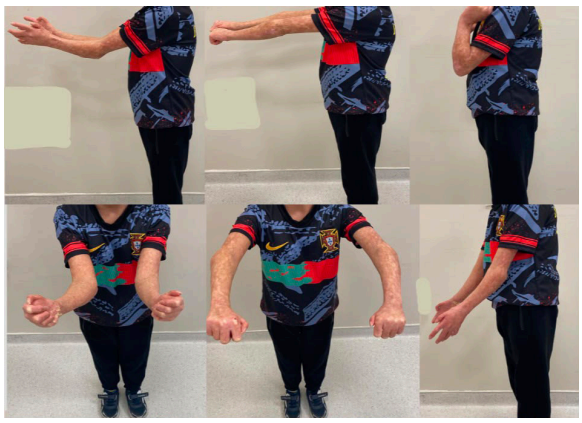

Figure 3. In the physical examination at the first postoperative year, it is observed that the joint range of motion is equal on both sides.

Discussion

In this case report, the management of radius and ulna fractures in a patient with RTS is discussed. Fracture union in the ulna could be achieved with stable fixation in the second surgery. RTS patients with skeletal abnormalities are at increased risk for pathological fragility fractures and delayed fracture healing due to the associated decreased bone mineral density (5). When the numerous skeletal abnormalities associated with RTS was taken into consideration, stable fixation may be more likely to achieve fracture union.

The literature reports an incidence of skeletal abnormalities as high as 75% in patients with RTS (9). There are two cases of fracture nonunion reported in RTS patients. In one case, a 5th metatarsal fracture was evaluated for nonunion following conservative treatment, and the patient was subsequently lost to follow-up before union could be monitored following intramedullary screw surgery (6). In another case, a patient with RTS who was being followed up for osteosarcoma had multiple spontaneous fractures and a nonunion of the tibia. However, in this case, the patient did not receive any treatment for the tibia fracture (7). Although our patient had a RECQL4 mutation in addition to clinical findings, he was not diagnosed with osteosarcoma. However, due to his young age, follow-up for osteosarcoma was necessary. The risk of osteosarcoma in cases associated with the RECQL4 gene has been reported to be as high as 30% (10).

Management of RTS primarily consists of preventive care. Annual ophthalmic examinations to identify cataracts and follow-up radiographs to identify underlying skeletal dysplasias are recommended. Calcium and vitamin D supplements may also be prescribed to RTS patients with bone demineralization or a history of fractures. Patients are advised to avoid excessive sunlight, and close monitoring for changes in skin lesions is recommended due to the increased incidence of skin cancer. Patients with RTS encompass a diverse clinical spectrum. A multidisciplinary clinical team, including a dermatologist, ophthalmologist, orthopedic surgeon, and oncologist, is necessary to optimally manage their follow-up (3).

Although impaired bone biology associated with RTS likely played a major role in the development of nonunion, a potential mechanical factor should also be considered.

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On retrospective evaluation of the initial radiographs, the diameter of the elastic nail used during the first surgery appeared relatively small in relation to the medullary canal, which may have resulted in insufficient mechanical stability. In pediatric forearm fractures treated with elastic intramedullary nailing, adequate canal fill and stable three-point fixation are critical for successful fracture healing (11, 12). A nail with insufficient diameter may lead to micromotion at the fracture site, delayed union, or nonunion, particularly in patients with compromised bone quality such as those with RTS. Therefore, the failure observed in this case was likely multifactorial, involving both impaired bone biology and suboptimal mechanical stability. Stable fixation achieved during revision surgery appears to have played a key role in obtaining successful union.

Conclusion

In the initial treatment of a seemingly simple fracture in a 12-year-old patient followed up with RTS, union was not achieved with routine techniques, and the need for additional surgery arose. Fracture union was achieved with stable fixation performed in the second surgery. With this report, we hope to raise practitioner awareness of the high likelihood of delayed bone healing or nonunion in patients with RTS and encourage consideration of routine imaging and calcium and vitamin D supplementation. Additionally, given their poor bone healing capacity, we suggest that patients with RTS may benefit from early and more aggressive surgical interventions for fractures.

References

1. Vennos EM, James WD. Rothmund-Thomson syndrome. Dermatologic clinics. 1995;13(1):143-50.
2. Lindor NM, Devries EM, Michels VV, Schad CR, Jalal SM, Donovan KM, et al. Rothmund- Thomson syndrome in siblings: evidence for acquired in vivo mosaicism. Clinical genetics. 1996;49(3):124-9.
3. Larizza L, Roversi G, Volpi L. Rothmund-Thomson syndrome. Orphanet J Rare Dis. 2010;5:2.
4. Kitao S, Shimamoto A, Goto M, Miller RW, Smithson WA, Lindor NM, et al. Mutations in RECQL4 cause a subset of cases of Rothmund-Thomson syndrome. Nature genetics. 1999;22(1):82-4.
5. Mehollin-Ray AR, Kozinetz CA, Schlesinger AE, Guillerman RP, Wang LL. Radiographic Abnormalities in Rothmund-Thomson Syndrome and Genotype–Phenotype Correlation with RECQL4 Mutation Status. American Journal of Roentgenology. 2008;191(2):W62-W6.
6. Barisonek KL, Protzman NM, Wobst GM, Brigido SA. Delayed union of a Jones fracture in a patient with Rothmund-Thomson syndrome: a case report and review of the literature. The Journal of Foot and Ankle Surgery. 2016;55(2):291-3.
7. Carlson AM, Thomas KB, Kirmani S, Lindor NM. Chronic tibial nonunion in a Rothmund–Thomson syndrome patient. American Journal of Medical Genetics Part A. 2012;158(9):2250-3.
8. Beckmann N. Multiple Low Energy Long Bone Fractures in the Setting of Rothmund- Thomson Syndrome. Case Reports in Medicine. 2015;2015(1):495164.
9. Wang LL, Levy ML, Lewis RA, Chintagumpala MM, Lev D, Rogers M, et al. Clinical manifestations in a cohort of 41 Rothmund- Thomson syndrome patients. American journal of medical genetics. 2001;102(1):11-7.
10. Martins DJ, Di Lazzaro Filho R, Bertola DR, Hoch NC. Rothmund-Thomson syndrome, a disorder far from solved. Front Aging. 2023;4:1296409.
11. Tella AO, Aldhilan MM. The outcome of titanium elastic nail fixation of pediatric long bone fractures - a retrospective analysis. Orthop Rev (Pavia). 2024;16:118449.
12. Jain S, Mohanachandran J, Mohan R. Outcomes and complications of Titanium elastic nailing for forearm bones fracture in children: our experience in a district general hospital in the United Kingdom. Acta Orthop Belg. 2023;89(3):539-46.