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Congenital Radioulnar Synostosis with Metacarpal Fusion - A Unique Presentation

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Abstract

Congenital skeletal dysplasias and dysostosis are very rare conditions that lead to bone and joint abnormalities, including growth deficiencies, fractures, and vertebral defects that can be debilitating to affected patients. Although many disorders are well recognized, there exist patterns and combinations of skeletal defects which have not yet been described. We present the case of a 17-year-old male with a unique combination of congenital radioulnar synostosis, fourth and fifth metacarpal fusion, carpal bone fusion, and thumb aplasia not previously described in the literature.

Keywords: *Radioulnar synostosis, metacarpal fusion, thumb aplasia, skeletal dysplasia, congenital*

Introduction

Proper bone development is complex and requires the correct activation of multiple genes and growth factors. The osseous tissues are formed by two different processes during embryogenesis: endochondral and intramembranous ossification. Endochondral ossification involves chondrocyte formation of cartilage models that will later form ossified bone, while intramembranous ossification involves direct bone formation by osteoblasts.[1] Disruption of the necessary genes or molecules can result in skeletal dysplasia or dysostosis.

Congenital skeletal dysplasias include diverse conditions that affect bone or joint growth and encompass over 400 disorders.[2] These dysplasias typically present during the prenatal period and result from genetic defects, but can evolve throughout life and involve previously unaffected bones and joints. In contrast, skeletal dysostosis does not progress and remains static as the patient ages. Regardless of the underlying etiology, there is a large spectrum of bony abnormalities that can develop throughout the skeleton from these conditions, including growth deficiency, bowing or shortening of the bones, fractures, vertebral defects, and rib abnormalities.[3] Many syndromes have been reported, but there still exist patterns of skeletal abnormalities yet to be described. We report a unique case of an upper extremity congenital skeletal abnormality with a constellation of radioulnar synostosis, fourth and fifth metacarpal fusion, carpal bone fusion, and thumb aplasia not previously described together in the literature.

Case Presentation

The patient is a 17-year-old male with a history of bilateral upper extremity congenital abnormalities who presented complaining of lifelong bilateral hand pain and stiffness, most severe on the left side. His medical history is significant for in utero recreational drug exposure, and possible unspecified developmental delay. He reports no familial history of congenital abnormalities (skeletal or otherwise) in his parents or eight siblings.

On physical examination, his bilateral humeri appear equal in length. The right forearm is short in comparison to the left. Passive range of motion of the right elbow is 30 to 140 degrees and the left elbow is 15 to 140 degrees. The right forearm is unable to supinate or pronate, but the left forearm shows supination to 80 degrees and pronation to 30 degrees. The patient has flexion to 80 degrees and extension to 90 degrees at the right wrist. His left wrist flexes to 60

degrees and extends to 70 degrees. There are palpable radial and ulnar artery pulses bilaterally. He has an absent thumb on the right hand, with four remaining digits. His right fourth metacarpophalangeal joint is receded with shortening of the fourth and fifth metacarpals. On the left hand, his thumb is dysplastic and the fifth proximal interphalangeal joint displays camptodactyly.

On radiographic examination the left upper extremity appears normal except for hypoplasia of the left scaphoid, trapezium, and first ray with chronic flexion deformity of the left fifth digit at the proximal interphalangeal joint. The right forearm displays a 3cm synostosis proximally between the radius and ulna. The right radial head is posteriorly dislocated with respect to the capitellum, likely representing congenital displacement **(Figure 1).** The right wrist and hand display multiple abnormalities. There is hypoplasia of the distal right radius and ulna with absence of the ulnar styloid. The head of the right fifth metacarpal is hypoplastic, with the fifth digit displaying ulnar angulation at the metacarpophalangeal joint. The right first digit, first metacarpal, and trapezium are absent. The right scaphoid is either absent or hypoplastic with fusion to the capitate bone. Multiple bones in the right hand appear fused; these include the lunate and triquetrum, fourth and fifth metacarpals, and possibly the scaphoid and capitate **(Figure 2)**.

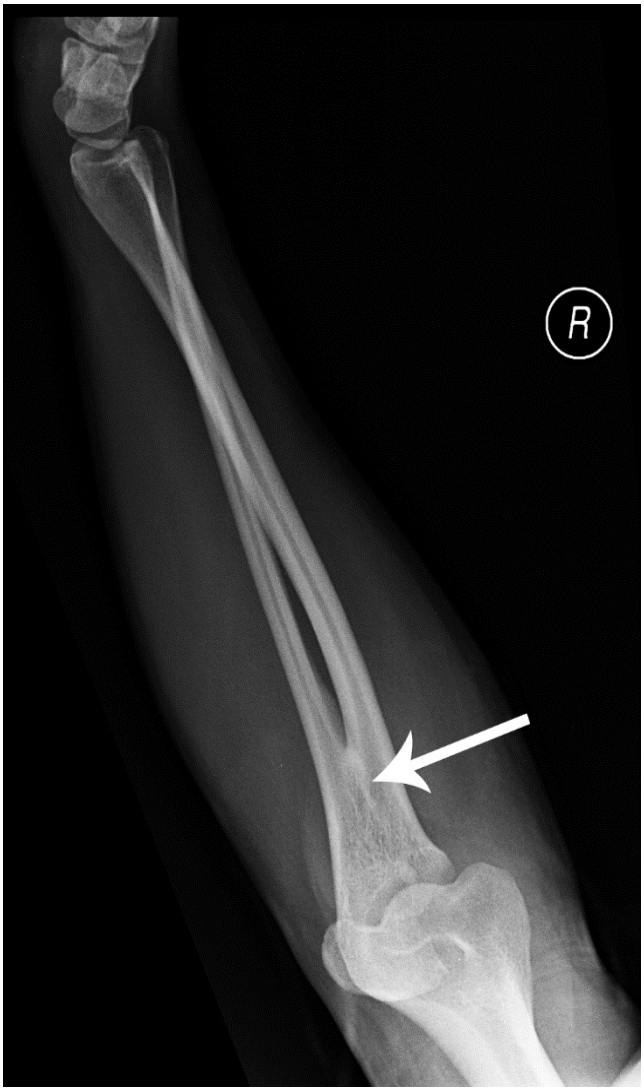

Figure1. *17-year-old male who presented with bilateral hand pain. Lateral radiograph of the right forearm and elbow. There is a 3cm synostosis between the proximal portions of the radius and ulna (arrow). The radial head, coronoid process, and olecranon process are intact. The radial head projects 7.5mm proximal to the capitellum.*

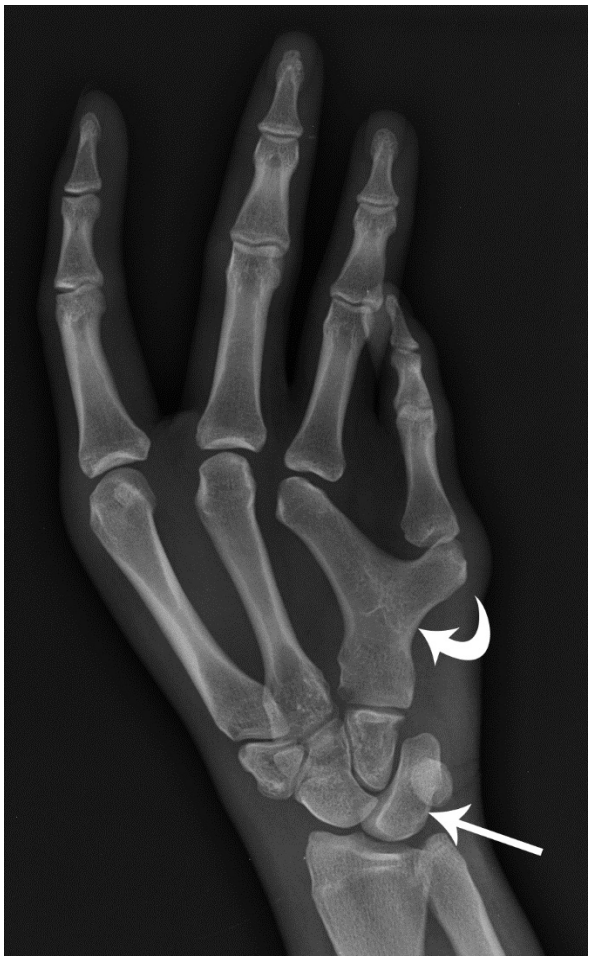

Figure2. *17-year-old male who presented with bilateral hand pain. PA plain radiograph of the right wrist and hand. The distal aspects of the radius and ulna are hypoplastic with absence of the ulnar styloid process. The first metacarpal and entire thumb are absent. The fourth and fifth metacarpals are fused (curved arrow) along with the lunate and triquetrum (straight arrow).*

Discussion

Congenital radioulnar synostosis (CRS) is a rare congenital disorder with only about 350 cases reported in the literature.[4] About 60% of cases are bilateral and typically caused by a failure in separation of the fused precursors to the radius and ulna during the seventh week of gestation.[4] CRS has been described in the setting of other congenital deformities, including syndactyly, polydactyly, clubfoot, and thumb hypoplasia.[5] Furthermore, CRS is often associated with posterior dislocation of the radial head, as in this patient.[5] Patients with CRS may report a family history of the disorder as it can be associated with Carpenter syndrome and Apert syndrome. Patients with Carpenter syndrome and Apert syndrome, though, typically have craniosynostosis and facial abnormalities which were not present in our case.

Radial ray anomaly is a spectrum of congenital anomalies resulting in absence or hypoplasia of the radius, radial carpal bones, and/or thumb.[6] This anomaly can occur in isolation or in syndromes involving the musculoskeletal, cardiothoracic, gastrointestinal, or genitourinary systems.[6] While hypoplastic radius, fused carpal bones, and aplasia of the thumb can be seen with radial ray anomaly, the systemic findings of the usual associated syndromes are absent in our patient. Fanconi anemia and Holt-Oram syndrome are two well-known disorders usually associated with radial ray anomalies. Patients with Fanconi anemia have bone marrow failure while patients with Holt-Oram syndrome have congenital heart defects (especially left-to-right shunts).

Fusion of the fourth and fifth metacarpals (MF4) may occur as an inherited disorder. Syndactyly V and

syndactyly VIII are two autosomal dominant disorders associated with this abnormality.[7] X-linked recessive MF4 has been shown with fibroblast growth factor 16 (FGF-16) mutations and in Kallman syndrome.[7] The radial abnormalities in our patient and absence of family history of skeletal dysplasia make these disorders unlikely.

The unique finding in our case is the combination of radial abnormalities with MF4 which has not been previously described. Our patient was exposed to recreational drugs in utero, although the exact drug is not known. We speculate that alcohol exposure might have a role as fetal alcohol syndrome (FAS) is the closest drug-related syndrome with findings that resemble those of our patient. While our patient lacks the characteristic facial abnormalities associated with FAS, FAS has been shown to present with CRS, carpal bone fusions, and developmental delay.[8] However, to our knowledge, MF4 is not associated with FAS and the explanation for the combination of radial ray abnormalities and MF4 remains elusive.

Many patients with CRS, similar to this patient, have difficulty with supination and pronation of the forearm. This patient in particular did not feel the need for treatment, but there are surgical options which can increase the range of motion in patients with CRS. Hiroshi et al. found that simple rotational osteotomy in affected patients resulted in an increased range of forearm motion and a marked improvement in performance of activities of daily living.[9] Similarly, proximal radioulnar derotational osteotomy and internal fixation is also effective for increasing the range of motion in patients with CRS.[10]

Conclusions

In summary we describe the findings in a patient with CRS, MF4, and absent thumb, a group of rare skeletal dysplasias not previously described as occurring in combined form in the same patient.

References

[1] Berendsen AD, Olsen BR. Bone development. Bone. 2015 Nov;80:14-18.

[2] Mortier GR, Cohn DH, Cormier-Daire V, Hall C, Krakow D, Mundlos S, et al. Nosology and classification of genetic skeletal disorders: 2019 revision. Am J Med Genet A. 2019 Dec;179(12):2393-2419.

[3] Bacino A. Skeletal Dysplasias: Approach to Evaluation. *UpToDate*, 3 June 2020, www.uptodate.com/contents/skeletal-dysplasias-approach-to-evaluation.

[4] Tsai J. Congenital radioulnar synostosis. Radiol Case Rep. 2017 Apr 13;12(3):552-554.

[5] Elliott AM, Kibria L, Reed MH. The developmental spectrum of proximal radioulnar synostosis. Skeletal Radiol. 2010 Jan;39(1):49-54.

[6] James MA, McCarroll HR Jr, Manske PR. The spectrum of radial longitudinal deficiency: a modified classification. J Hand Surg Am. 1999 Nov;24(6):1145-55.

[7] Malik S. Syndactyly: phenotypes, genetics and current classification. Eur J Hum Genet. 2012 Aug;20(8):817-24.

[8] Jaffer Z, Nelson M, Beighton P. Bone fusion in the foetal alcohol syndrome. J Bone Joint Surg Br. 1981;63B(4):569-71.

[9] Satake H, Kanauchi Y, Kashiwa H, Ishigaki D, Takahara M, Takagi M. Long-term results after simple rotational osteotomy of the radius shaft for congenital radioulnar synostosis. J Shoulder Elbow Surg. 2018 Aug;27(8):1373-1379.

[10] Pei X, Han J. Efficacy and feasibility of proximal radioulnar derotational osteotomy and internal fixation for the treatment of congenital radioulnar synostosis. J Orthop Surg Res. 2019 Mar 20;14(1):81.

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