Case Report

The congenital absence of the radius, scaphoid, trapezium, thumb and hypoplasia of the lunate

Ayfer Mavi, PhD, Bilal Cagman, MD, Ayhan Ozkur, MD, Metin Bayram, MD, Erdem Gumusburun, PhD.

ABSTRACT

The congenital absence of the right radius, scaphoid, trapezium, thumb and hypoplasia of the lunate are described in a 9-year-old boy. He had full extension of his elbow, while flexion motion was limited to 90 degrees. His hand was radially deviated. The thumb was absent. The index and middle fingers were united and immobile. The ring finger had movement only at the metacarpophalangeal joint, while the little finger had full motion. Complete blood count fell within normal ranges. He had no other malformations or mental retardation. No hereditary family history was found and there was no consanguineous marriage. This case is different from cases mentioned in the literature due to lunate hypoplasia and the anomalies of the index, middle and ring fingers.

Keywords: Radial longitudinal deficiency, congenital deformity, hypoplasia of lunate.

Saudi Med J 2002; Vol. 23 (8): 1005-1007

Congenital radius deficiency is a deformity of the upper extremity characterized by hypoplasia or absence of the radius. In the literature, many different names were given for example, club hand or radial hemimelia.[1,2] The incidence is one in 30,000 to one in 100,000. It is bilateral in approximately half the cases; and involvement is unilateral, the right side is affected twice as frequently as the left. Males are more often affected than females.[2,3] The exact cause of the longitudinal defect in the radial elements of the upper limb bud is unknown. Gegenbauer[4] suggested that the upper limb consisted of a main stem and 4 accessory rays. The humerus, the ulnar, the 2 carpal bones, the 5th metacarpal, and the 3 phalanges of the 5th finger made up the main stem. The radius, the scaphoid and trapezium, the first metacarpal, and the 2 phalanges of the thumb constitute the first accessory ray; the 2nd, 3rd, and 4th accessory rays consisted of the index finger, the middle finger, and the ring finger, with their respective metacarpals and carpal bones. It was proposed that congenital absence of the radius was due to the suppression of development of the first accessory ray. The upper limb develops in an orderly proximal to distal sequence. An intact, healthy apical ectoderm is a prerequisite for differentiation of the underlying mesenchymal tissues. Saunders[5] removed part of the apical ectodermal ridge in the developing wings of chick embryos and produced anomalies similar to congenital absence of the radius. It appears that the most probable cause of congenital absence of the radius is damage to the apical ectoderm or the deeper

From the Department of Anatomy, (Mavi, Gumusburun), Department of Radiology, (Ozkur, Bayram), Faculty of Medicine, Gaziantep University and the Department of Orthopedic & Traumatology, (Cagman), Government Hospital, Gaziantep, *Turkey*.

Received 27th August 2001. Accepted for publication in final form 26th January 2002.

Address correspondence and reprint request to: Dr. Ayfer Mavi, Department of Anatomy, Faculty of Medicine, Gaziantep University, 27310 Gaziantep, *Turkey*. Fax. +90 (342) 3601617. E-mail: mavi@gantep.edu.tr

mesenchymal tissues on the anterior aspect of the developing upper limb bud.[2] Environmental factors known to cause radial deficiencies are chemicals (such as thalidomide), irradiation, and viral infections.[2] In the embryo, bones appear during the 5th week as mesenchymal condensations in the limb buds. During the 6th week, the mesenchymal models of the bones in the limbs undergo chondrification to form hyaline cartilage models; therefore, the teratologic factor must affect the limb bud during the first 5 weeks of fetal life.[3]

Case Report. The boy was admitted to the Department of Orthopedic and Traumatology, Gaziantep Government Hospital, Turkey, as his right hand and forearm were shorter than the left. On examination he had shorter right forearm and bowed radially with a prominent knob at its lower end but his arm was not shorter than the other side. He had full extension of his elbow, while flexion motion was limited to 90°. His hand which radially deviated. The thumb was absent. The index and 3rd finger was united and immobile. The 4th (ring) finger had movement only at the metacarpophalangeal joint, while the little finger had full motion **(Figure 1).** Ossification of the carpal (wrist) bones began during the first year after birth. In a 9-year-old healthy boy, he had complete ossified carpal bones except

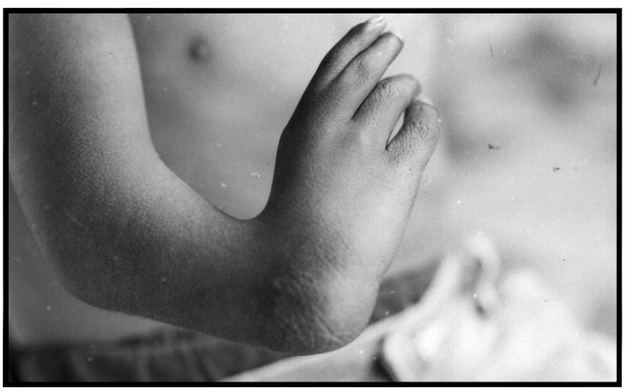

Figure 1 - The morphologic appearance of his right upper extremity. His elbow had full extension while flexion motion was limited to 90 degrees. His hand was radially deviated. the thumb was absent. The index and 3rd finger was united and immobile. The 4th finger had movement only at the metacarpophalangeal joint, while the little finger had full motion.

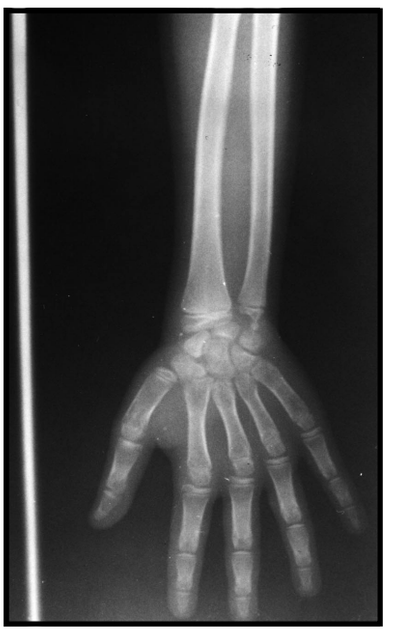

Figure 2 - Ossification of the carpal (wrist) bones begins during the first year after birth. The radiograph appearance of carpal bones in a 9-year-old healthy boy, he had complete ossified carpal bones except pisiform bone.

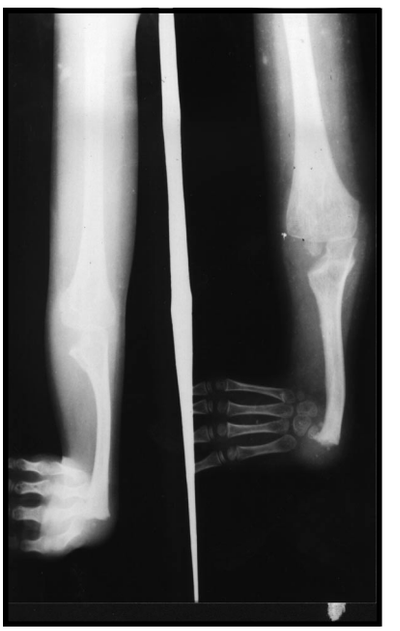

Figure 3 - The radiograph appearance of his right forearm and hand. His radius, scaphold and trapezium were missing and his lunate was hypoplastic. The distal humeral epiphysis was early fused. Humero-ulnar joint was normal. The distal ulnar epiphysis and processus styloideus were displastic. The radiographic appearance of the metacarpophalangeal and interphalangeal joints were normal.

pisiform bone,[6] (**Figure 2**) while on the right forearm and hand radiography of our patient, radius, scaphoid, and trapezium were missing and lunate was hypoplastic. The distal humeral epiphysis was early fused. Humero-ulnar joint was normal. The distal ulnar epiphysis and processus styloideus were displastic. The radiographic appearance of the metacarpophalangeal and interphalangeal joints were normal (**Figure 3**). His complete blood count fell within normal ranges. He had no another malformations or mental retardation. No hereditary family history was found and there was no consanguineous marriage.

Discussion. In congenital deficiencies of the radius, it may be absent totally or partially. In complete absence of radius, the bones of the radial ray the scaphoid, trapezium, first metacarpal and phalanges of the thumb are frequently absent.[1,3] O'Rahilly[7] surveyed the literature for radial deficiency and found that the scaphoid was absent in 81% (65 of 80 cases) and the trapezium in 84% (62 of 73 cases). Skerik and Flatt[8] studied the anatomy of 38 cases of deficiency of the radius and found that the scaphoid was absent in 80% (17 of 21 cases) and the trapezium was absent in 66% (14 of 21 cases). Lamb[2] examined 68 patients with radial club hand; scaphoid was absent in 98% and the trapezium in 100%. Silverman[9] expressed in his textbook that the trapezoid, lunate, and pisiform are involved in 10% with hypoplasia, fusion or delayed ossification rather than total absence; but the trapezium is missing in apparently 67% and the scaphoid in 80%. In this case, the radius, scaphoid, trapezium and thumb were absent, the lunate was also hypoplastic. As seen above hypoplasia of lunate is very rare. Tachdjian[3] and Frantz et al[1] reported that first metacarpal and phalanges of the thumb might be frequently absent. Silverman[9] reported that the first metacarpal and its phalanges were missing in more than 80% of cases. But James et al[10] and Lamb[2] observed that all the affected limbs had thumb hypoplasia. In our case, the patient did not have the first metacarpal and thumb. Regarding the other fingers, Tachdjian[3] reported that the radial 2 fingers, especially the index were abnormal with varying degrees of flexion contracture, weakness of active motion, and hypoplasia. The ulnar 2 digits were almost always normal and more functional. Silverman[9] reported that the ulnar 4 metacarpals and phalanges were present and free of defect in the majority of cases. In the present case, the patient had 4 metacarpals. The index and middle fingers were united and immobile. The ring finger had movement only at the metacarpophalangeal joint, while the little finger had full motion. Tachdjian[3] reported that ossification of the distal humeral epiphysis might be delayed. Silverman[9] reported that there may be varying abnormalities of the distal humeral end. In the present case, the distal humeral epiphysis was early fused.

Congenital longitudinal radial deficiencies are frequently associated with other malformations. This is explained by the fact that many organs develop at the same time as the upper limb buds. It behooves the orthopedic surgeon to be aware of these associations and to be sure that the patient has no serious anomalies that would make surgery hazardous and inadvisable. According to Goldberg and Meyn,[11] associated with these malformations are classified into 7 groups: 1. Chromosomal abnormalities, 2. Mental deficiency, 3. Cranio-facial defect, 4. Cardiac anomalies, 5. Blood dyscrasias, 6. Vertebral anomalies and 7. Teratogenic syndromes. In our case, our patient did not have any associated anomaly. We think that this case is different from the cases reported in literature in the presence of lunate hypoplasia and the anomalies of index, middle and ring fingers.

References

1. Frantz CH, O'Rahilly R. Congenital skeletal limb deficiencies. *J Bone Joint Surg Am* 1961; 43A: 1202-1224.
2. Lamb DW. Radial club hand: A continuing study of sixty-eight patients with one hundred and seventeen club hands. *J Bone Surg Am* 1977; 59A: 1-3.
3. Tachdjian MO. Pediatric Orthopedics. 2nd ed. Philadelphia (PA): WB Saunders Company; 1990. p. 188-198.
4. Gegenbauer C. Zur morphologie der gliedmassender wirbelthiere. *Morph Jahrb* 1976; 2: 396.
5. Saunders JW. The proximo-distal sequence of origin of the parts of the chick wing and the role of the ectoderm. *J Exp Zool* 1948; 108: 363-368.
6. Williams PL, Warwick R, Dyson M, Bannister LH. Gray's Anatomy. 37th ed. Edinburgh (Scotland): ELBS Churchill Livingstone; 1989. p. 421.
7. O'Rahilly R. Morphological patterns in limb deficiencies and duplications. *Am J Anat* 1951; 89: 135-193.
8. Skerik SK, Flatt AE. The anatomy of congenital radial displasia: its surgical and functional implications. *Clin Orthop* 1969; 66: 125-143.
9. Silverman FN. CAFFEY'S pediatric X-ray diagnosis. Volume 1. 8th ed. Chicago (IL): Yearbook Medical Publishers Inc; 1985. p. 500-501.
10. James MA, McCarroll HR Jr, Manske PR. The spectrum of radial longitudinal deficiency: A modified classification. *J Hand Surg Am* 1999; 24A: 6: 1145-1155.
11. Goldberg MJ, Meyn M. The radial club hand. *Orthop Clin North Am* 1976; 7: 341.