SCIENTIFIC ARTICLE

Combined Congenital Radial and Ulnar Longitudinal Deficiencies: Report of 2 Cases

Kerby C. Oberg, MD, PhD, Tyler E. Harris, MD, Montri D. Wongworawat, MD, Virchel E. Wood, MD

Variation in longitudinal deficiencies is likely related to the timing and duration of an insult during early limb development. In experimental models, teratogenic insults induce ulnar deficiencies earlier in gestation than radial deficiencies. In this report, we describe the rare combination of right radial and left ulnar deficiencies in 2 cases. Interestingly, 1 case had a history of 2 separate and apparently distinct episodes of bleeding during early gestation, whereas the other demonstrated associated hematoma formation early in development. These cases also demonstrate the susceptibility for ulnar defects on the left and radial defects on the right. The authors discuss the relationship of prenatal insults on limb development and the mechanisms underlying longitudinal deficiencies. *(J Hand Surg 2009;34A:1298–1302. © 2009 Published by Elsevier Inc. on behalf of the American Society for Surgery of the Hand.)*

Key words Congenital, longitudinal, deficiency, radius, ulna.

Longitudinal deficiencies are abnormalities affecting the forearm, hand, or both, that can exhibit primarily radial- or ulnar-sided deficiencies. Radial-sided defects, or radial longitudinal deficiencies (RLD), are uncommon, occurring in 1 in 9,000 to 1 in 55,000[1,2]; ulnar-sided defects or ulnar longitudinal deficiencies (ULD) are rare, with an estimated incidence of about 1 in 100,000, or 10 times less frequently than RLD.[2] There is a relatively high incidence of bilaterality (50% for RDL and 25% for UDL), suggesting that developmental insults frequently affect both forelimbs. However, when unilateral, RLDs show a predilection for the right limb, whereas ULD occurs more frequently on the left. Furthermore, combined RLD and ULD in the same patient are exceedingly rare and none of the previous reports of this malformation have attempted to link these combined defects with an underlying etiology.[3–5]

In this report, we present 2 cases with combined right-sided RLD and left-sided ULD and discuss potential etiologic mechanisms.

From the Departments of Pathology and Human Anatomy, and Orthopaedic Surgery, Loma Linda University; and the David Grant Medical Center, Travis Air Force Base, CA.

Received for publication February 20, 2009; accepted in revised form April 20, 2009.

No benefits in any form have been received or will be received related directly or indirectly to the subject of this article.

The views expressed in this material are those of the authors, and do not reflect the official position of the U.S. Government, the Department of Defense, or the Air Force.

Corresponding author: Virchel E. Wood, MD, Department of Orthopaedic Surgery, Loma Linda University Medical Center, 11234 Anderson Street, Room A537, Loma Linda, CA 92354; e-mail: lkent@som.llu.edu.

0363-5023/09/34A07-0019$36.00/0
doi:10.1016/j.jhsa.2009.04.042

CASE 1

A 3.1-kg boy of 39 weeks' gestation was born to a G4P3A0 24-year-old woman. Between the 5th and 6th weeks of gestation (3–4 weeks' developmental age), the mother reported an episode of pelvic cramping with vaginal bleeding. This resolved spontaneously without medical attention. At 8 weeks' gestation (6 weeks' developmental age), the mother presented to the emergency room reporting a second episode of pelvic cramping and bright red vaginal bleeding. Pelvic ultrasound demonstrated a viable 8-week intrauterine pregnancy with a small subchorionic hemorrhage, and the patient was sent home and prescribed bed rest. The next afternoon, the patient reported to the emergency room after passing a large amount of blood in the morning. She also reported persistent suprapubic cramping and continued bleeding. Repeat ultrasound confirmed enlargement of the subchorionic hemorrhage, but the fetus remained viable. The bleeding subsequently subsided.

Evaluations later during pregnancy showed normal growth and no abnormalities were identified. The mother was rubella immune. Screens for hepatitis B,

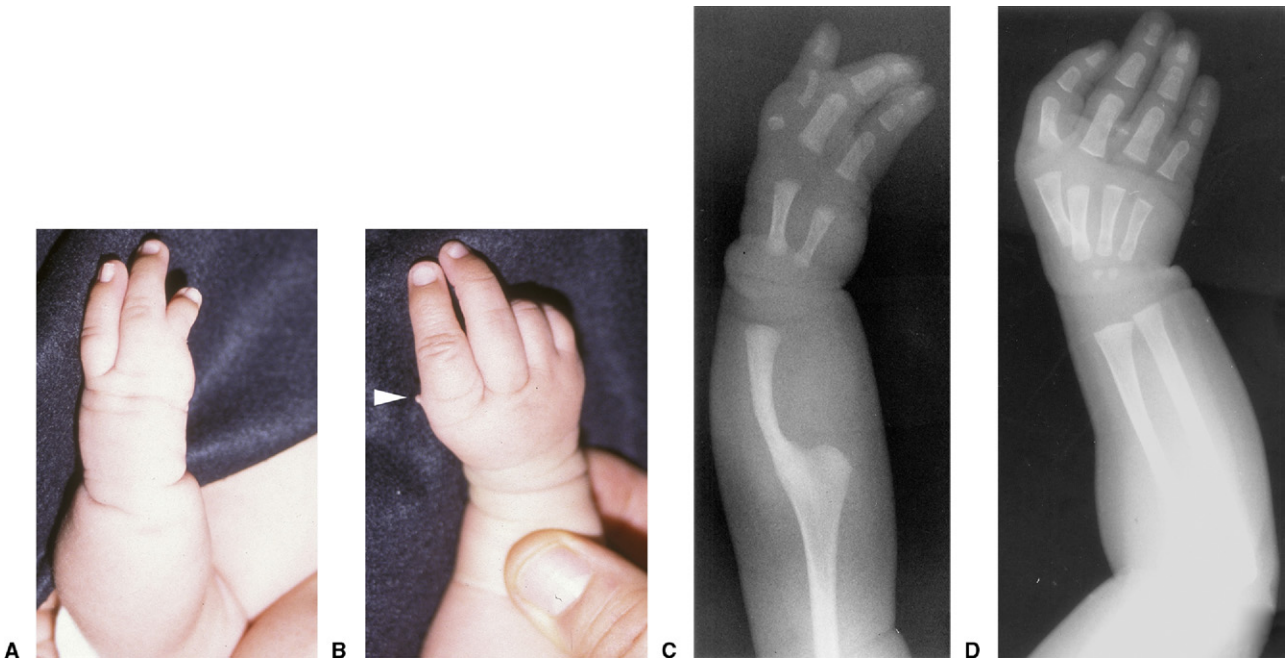

FIGURE 1: Photographs and x-rays from case 1. The gross photographs of **A** the left and **B** right hands illustrate the ulnar and radial defects. A white arrowhead indicates the thumb nubbin. X-ray examination further demonstrated the radial humeral fusion on **C** the left and **D** right-sided radial shortening. Skeletal elements were absent in the thumb nubbin.

rapid plasma reagin, gonococcus, chlamydia, and beta-streptococcus were negative. Alpha-fetoprotein level was normal. The patient underwent a planned caesarean section without complications and delivered a boy with bilateral limb anomalies. Postnatal chromosomal analysis of the infant was reported as normal male karyotype.

At 3 weeks of age, the infant presented for evaluation. Initial examination of the right hand revealed 4 triphalangeal fingers and a nubbin in place of the thumb (Fig. 1). The index finger exhibited early pronation and abduction. Radiographs confirmed the triphalangeal fingers, the lack of osseous structures in the thumb, and shortening of the radius. Both the radius and ulna were present on forearm radiographs. The elbow appeared normal. We classified the RLD as radius deficiency Bayne type I,[6] thumb hypoplasia Blauth type IV.[7]

On the left side, the patient demonstrated absence of the ulnar 2 digits. The thumb was hypoplastic, whereas the index and long fingers were relatively normal (Fig. 1). Radiographic evaluation revealed a hypoplastic thumb and the complete absence of the ulna along with the ring and little fingers. The bowed radius was fused to the humerus. We classified the ULD as forearm/elbow Bayne type IV,[8] hand type C, as described by Ogino and Kato,[9] with absence of ring and little fingers.

CASE 2

A hydropic macerated male fetus with bilateral limb anomalies was delivered to a 21-year-old woman, G1P0, during her 18th week of gestation. The delivery followed premature spontaneous rupture of membranes and vaginal bleeding. The fetus was from a dizygotic twin gestation, with both fetuses suffering intrauterine demise before delivery. The other fetus was macerated, but a normal female fetus. The placenta associated with the hydropic male fetus had a large subchorionic hematoma.

The male fetus had multiple anomalies, including an absent radius on the right with polysymbrachydactyly (5 shortened identical fingers but no thumb) (Fig. 2). Musculoskeletal anomalies are nearly 10 times more common in aborted previable fetuses (53 in 1,000 compared with 5 in 1,000 newborns) with "limb reduction" defects occurring in only 11 in 1,000 in this population and occasionally associated with vasculopathy/coagulopathy.[10] In our case, there was a 2.0 × 2.5-mm blood clot (asterisk in Fig. 2D) that disrupted formation of the central or diaphyseal portion of the second metacarpal. In addition, skeletal analysis by Alcian green cartilage staining revealed proximal absence of the fourth metacarpal. We classified this RLD as radius