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Ulnar longitudinal deficiency; type D of cole and manske's classification and type v of goldfarb's: a case report

Abstract

Ulnar longitudinal deficiency (ULD) is an infrequent congenital anomaly found in approximately 1 in 100,000 live births. In an incidental discovery, a 57-year-old healthy man, referred to our Orthopedic Center due to a right medial malleolus fracture, was diagnosed with ULD. A comprehensive physical examination revealed notable abnormalities in the patient's right upper extremity, marked by significant shortening, pronation, and internal rotation. Radiological examinations exposed a complex skeletal deformity, including radiohumeral synostosis, the complete absence of the ulna and carpal bones, along a shortened and bowed radius. Remarkably, only the thumb and the first metacarpal bone were present in the hand, accompanied by a distinctive ulnar deviation of the wrist by Cole and Manske's classification (type D) and Goldfarb's classification (type V), the patient's condition was identified through a thorough clinical assessment. This detailed characterization advances our understanding of the specific anatomical features and classifications associated with ULD, offering valuable insights for clinical management and potential interventions in similar cases.

Keywords: ulnar longitudinal deficiency, radial longitudinal deficiency, congenital anomalies

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Introduction

Three distinct types of congenital longitudinal deficits impact the upper extremities: radial, ulnar, and intersegmental. The most prevalent is radial longitudinal deficit (RLD), affecting approximately 1 in 5000 to 1 in 55,000 live infants.[1,2] In contrast, ulnar longitudinal deficit (ULD) is less common, with an estimated occurrence of 1 in 100,000 live newborns.[3,4] ULD encompasses a wide range of conditions, from the absence of ulnar rays and ulnar carpal bones to the complete absence of the ulna. Associated variations may include radial head dislocation, radiohumeral synostosis, and shoulder abnormalities. In this specific case, we present a unique instance in a 57-year-old male patient with right ULD. Notably, this presentation is characterized by radiohumeral synostosis and the absence of four metacarpal bones. Additionally, the carpal bones and ulna are absent in this particular case. The comprehensive nature of this anomaly highlights the intricacy and variability within the spectrum of ULD conditions. This case report aims to provide valuable insights into the understanding and management of such unconventional presentations, contributing to the expansion of our knowledge regarding congenital upper extremity deficiencies presentations, expanding our knowledge of congenital upper extremity deficiencies.

Case presentation

A 57-year-old healthy male with a right medial malleolus fracture was referred to the Orthopedic Center of Boo-Ali Sina Hospital in Sari, Mazandaran, Northern Iran. A congenital anomaly in the patient's right upper extremity was detected during a physical examination. In comparison to the unaffected limb, the affected limb exhibited significant shortening, pronation, and internal rotation. Notably, the shoulder joint remained unaffected. Subsequent radiographic examinations revealed the presence of radiohumeral synostosis, complete absence of the carpal and ulna bones, and a bowed and shortened radius. Furthermore, the hand exhibited only the thumb and the first metacarpal bone, with the wrist showing a distinctive curvature in the direction of the ulna. The elbow's range of motion was limited due to congenital webbing (pterygium), and the fingers displayed restricted mobility. (Figure 1 & 2) Comprehensive assessments of the musculoskeletal system revealed no additional issues, and there were no indications of other organs being affected.

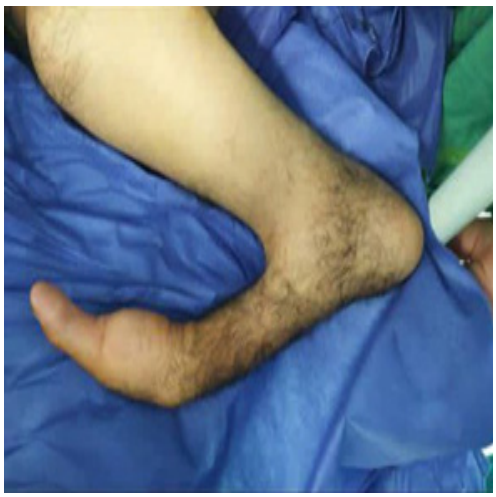

Figure 1 Clinical photograph of a male patient displaying ulnar longitudinal deficiencies on the right side.

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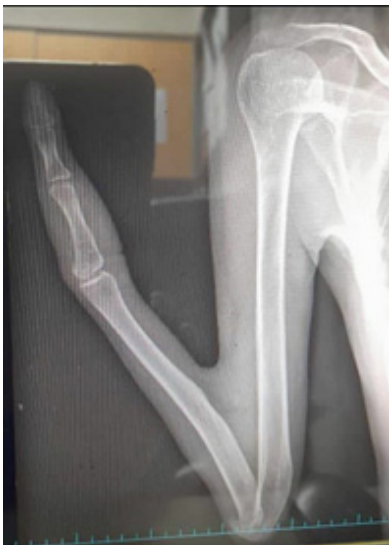

Figure 2 A radiograph of the right upper limb illustrating the absence of the ulna and monodactyly.

Discussion

The limb bud development occurs roughly between the 4th and 8th week after conception, involving three distinct axes guided by specialized cells, embryonic stem cells, and signaling molecules. The apical ectodermal ridge (AER) plays a crucial role in establishing the proximal-distal axis and regulating normal limb development, facilitated by numerous fibroblast growth factors (FGFs). In the zone of polarizing activity (ZPA), responsible for radial-ulnar differentiation, FGFs regulate the expression of Sonic Hedgehog (SHH) molecules. the interaction between ZPA and AER is intricate, with ZPA secreting SHH molecules controlling AER activity, and AER responding with a positive feedback loop via FGFs. According to current theories, Ulnar Longitudinal Deficiency (ULD) may occur when ZPA is malformed or the SHH signaling pathway is dysfunctional.[3,5]

The nature of ULD is still debated, generally considered a sporadic and non-inherited condition characterized by unilateral involvement of the upper limb. While radial side involvements are well-documented, ULD typically presents with ulnar side abnormalities. Systemic issues in ULD primarily affect the musculoskeletal system, sparing other systems compared to Radial Longitudinal Deficiency (RLD) in recent study, Sara Velasquez Restrepo's case report, detailing ulnar longitudinal deficiency (ULD) in an 11-month-old female infant, provides a valuable comparative perspective alongside our case involving a 57-year-old healthy male. Both instances underscore the diverse spectrum of ULD presentations across age and gender, sharing common features such as ulnar deviation and limb shortening. However, specific skeletal abnormalities vary, with our case revealing radiohumeral synostosis, the complete absence of the ulna and carpal bones, and a shortened and bowed radius. This detailed comparison enhances our medical understanding of ULD's intricate manifestations, reinforcing the necessity for tailored diagnostic and therapeutic approaches.

Recognizing the complexity of ULD, these insights guide clinicians in developing individualized strategies to address the unique challenges posed by this congenital skeletal anomaly. The collaborative analysis by Restrepo and our case contributes significantly to refining treatment strategies for individuals affected by this rare condition, promoting advancements in the medical management of ULD.[6] Musculoskeletal disorders like scoliosis, phocomelia, fibular insufficiency, and proximal femoral focal deficit are commonly associated with ULD.[7] Various methods have been proposed to categorize ULD, often emphasizing the increasing deficiency of the elbow and ulna. Ogino and Kato's categorization focused on the degree of ulnar defect[8] while Cole and Manske classified patients based on thumb involvement and the first web space.[9] Swanson's method and Goldfarb et al.'s classification introduced additional criteria, including significant carpal/digital deficits, straight radius, radiohumeral synostosis, and complete absence of the ulna.[10] Based on clinical results, the patient is likely classified as type V in Goldfarb's categorization and type D in Cole and Manske's classification.

Conclusion

In conclusion, this case report highlights a distinctive instance of ulnar longitudinal deficiency (ULD) in a 57-year-old male with a right medial malleolus fracture. The intricate skeletal abnormalities, encompassing radiohumeral synostosis and the absence of the ulna and carpal bones, correspond to type D in Cole and Manske's classification and type V in Goldfarb's classification. The insights gained from this case contribute valuable information to the diverse spectrum of ULD conditions, thereby enhancing our comprehension of clinical management and potential interventions. The documentation of such uncommon presentations emphasizes the significance of ongoing research to refine diagnostic and therapeutic approaches for individuals affected by ULD.

Statement of ethics

Verbal informed consent was obtained from the patient to publish this case report and any accompanying images. The patient signed the free and informed consent form.

Ethical approval and consent to participate

Available

Acknowledgments

None.

Conflicts of interest

The authors declare that there is no conflicts of interest.

References

1. Colen DL, Lin IC, Levin LS, et al. Radial longitudinal deficiency: recent developments, controversies, and an evidence-based guide to treatment. *J Hand Surg Am*. 2017;42(7):546–563.
2. Wall LB, Ezaki M, Oishi SN. Management of congenital radial longitudinal deficiency: controversies and current concepts. *Plast Reconstr Surg*. 2013;132(1):122–128.
3. Oberg KC, Harris TE, Wongworawat MD, et al. Combined congenital radial and ulnar longitudinal deficiencies: report of 2 cases. *J Hand Surg Am.* 2009;34(7):1298–1302.
4. Srinivasan P. Ulnar club hand: A rare case report. *J Orthop Assoc South India State*. 2021;18(1):18–20.
5. Afshar A. Combined congenital radial and ulnar longitudinal deficiencies: a case report. *J Hand Microsurg*. 2015;7(1):191–193.
6. Velasquez RS, Oboli VN, Kumar D, et al. Ulnar longitudinal deficiency: a case report. *Cureus*. 2023;15(6):e40111.

7. Bednar MS, James MA, Light TR. Congenital longitudinal deficiency. *J Hand Surg Am*. 2009;34(9):1739–1747.
8. Jerome TJ, Prabu R, Terrence TK. A new variant of type i congenital ulna deficiency with the normal thumb, webspace, hand, and elbow. *Cureus*. 2020;12(12):e12261.
9. Cole RJ, Manske PR. Classification of ulnar deficiency according to the thumb and first web. *J Hand Surg Am*. 1997;22(3):479–488.
10. Goldfarb CA, Manske PR, Busa R, et al. Upper-extremity phocomelia reexamined: a longitudinal dysplasia. *J Bone Joint Surg Am*. 2005;87(12):2639–2648.

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