Unilateral congenital lunate absence: A case report

Alden H. Newcomb, Jessica Frankenhoff

Department of Orthopaedic Surgery, Virginia Commonwealth University, Richmond, United States

Received: November 9, 2020  Accepted: December 20, 2020  Online Published: July 29, 2021
DOI: 10.5430/css.v7n1p1  URL: https://doi.org/10.5430/css.v7n1p1

ABSTRACT

Isolated congenital aplasia of carpal bones in the absence of an associated congenital syndrome is exceedingly rare. Since the first documented case report of congenital carpal bone absence in 1911, only 12 additional case reports have been published. Here we present a case report of an otherwise healthy 43-year-old male with unilateral congenital absence of the lunate, which has only been previously reported once in the literature.

Key Words: Lunate, Congenital absence, Carpal bones, Aplasia

1. INTRODUCTION

Isolated congenital aplasia of carpal bones in the absence of an associated congenital syndrome is exceedingly rare. Since the first documented case report of isolated congenital carpal bone absence in 1911,[1] only 12 additional case reports have been published. Here we present a case report of an otherwise healthy 43-year-old male with unilateral congenital absence of the lunate, which has only been previously reported once in the literature.

2. CASE PRESENTATION

A 45-year-old right-hand dominant male laborer presented to the hand surgery clinic complaining of chronic left wrist pain. The onset of the pain was insidious and had been progressing over the past few years. He denies any previous injury, infection, or surgery to the extremity. He has no past medical history and has never taken prescription medications. His prenatal and childhood and adult medical history are unremarkable.

His physical exam was generally unremarkable. He appeared healthy in no acute distress. His head was normocephalic and without abnormal facies. Cardiovascular and pulmonary exams were grossly normal. Examination of the left upper extremity showed a scar overlying the thumb metacarpal from a childhood laceration, but otherwise no evidence of trauma. There were no surgical scars over the dorsal or volar wrist. There was brisk capillary refill in all digits and palpable radial pulse. He had full range of motion with active flexion and extension at all metacarpophalangeal and interphalangeal joints. Range of motion of the left wrist showed 45° of flexion and 20° of extension compared with the right at 60° and 60 degrees, respectively. The DRUJ was stable in pronation and supination compared to the right without clicking, laxity, or tenderness to palpation. Finger cascade was normal. There was mildly tender palpable synovitis over the dorsal central aspect of the wrist. There was no thenar, interosseous or forearm atrophy.

Plain radiographs of the left wrist were obtained, followed by high-resolution computerized tomography scan. Figures 1 and 2 show the PA and oblique radiographs of the left wrist, with a representative coronal CT slice of the left wrist shown in Figure 3. Imaging of the left wrist demonstrates absence
of the lunate and a dysplastic appearing scaphoid. There was no suggestion of any remnant lunate to suspect advanced Keinbock’s disease. The proximal pole of the scaphoid had eroded into the distal radius and the proximal surface of the capitate appears flattened. There were widespread degenerative changes throughout the entirety of the wrist including subchondral cysts and arthritic changes at the midcarpal, trapeziometacarpal, and radiocarpal joints. Imaging of the contralateral wrist demonstrated a full complement of anatomically correct carpal bones with no abnormality.

Figure 1. PA radiograph of the left wrist

He was treated conservatively with oral nonsteroidal anti-inflammatory medication, a splint and a corticosteroid injection into the radiocarpal joint. At 4-month follow-up he continued to have pain in the left wrist. Surgical management including wrist fusion was discussed at this time. He again opted for conservative management with a second corticosteroid injection. He did not return to clinic thereafter.

3. DISCUSSION

Congenital carpal absence is often seen as a part of the constellation of the presentation of an underlying syndromic congenital disorder such as VACTERL (vertebral defects, anal atresia, cardiac abnormalities, trachea-esophageal fistula, renal and radial abnormalities and lung abnormalities). Other associated syndromes include thrombocytopenia-absent radius, Holt-Ohram, Fanconi’s anemia, and thumb hypoplasia/aplasia.[2] Carpal absence is also seen in congenital extremity malformations such as intercalary radial hemimelia, which consists of not absence of the scaphoid, trapezium, radial styloid, thumb metacarpal and thenar musculature.[3]
Congenital aplasia of carpal bones in the absence of additional syndromic or congenital abnormalities is exceedingly rare. To the best of the knowledge of the authors there are only 13 total case reports of congenital carpal absence, ranging from isolated absence of a single carpal bone to complete absence of the entire carpus, as summarized in Table 1.

### Table 1. Summary of published case reports of isolated congenital carpal absence

| Absent Carpal Bones | Reference |
|---------------------|-----------|
| Scaphoid, bilateral | Patankar [4] |
| Scaphoid, unilateral | Schick [5] |
| Scaphoid, unilateral | Srivastava & Kochhar [6] |
| Scaphoid, unilateral | Kuz & Smith [7] |
| Scaphoid, unilateral | Papanikolaou & Haddadin [8] |
| Scaphoid, unilateral | Panciera & Le Viet [9] |
| Lunate, bilateral | Kobayashi et al. [10] |
| Lunate, unilateral | Roche [11] |
| Scaphoid + lunate, bilateral | Hoffman [1] |
| Scaphoid + capitate, unilateral | Eraltug [12] |
| Scaphoid + lunate + capitate, unilateral | Postacchini & Ippolito [13] |
| Pan-carpal absence (left), trapezium (right) | Gurav & Patil [14] |
| Scaphoid + lunate (unilateral) | De Smet [15] |

The scaphoid is involved in 11 of these 13 reports, with isolated unilateral scaphoid aplasia/agenesis being the most commonly reported deficiency (5 of 13) and a single report of bilateral isolated scaphoid absence. Of the remaining 7 case reports, 5 involve absence of the scaphoid in conjunction with other carpal bones. The lunate is the second most commonly involved carpal bone, reported as absent in 7/13 case reports. Unlike the scaphoid, only one other report of a unilaterally absent lunate exists in the literature.

Multiple theoretical explanations for the findings in these case reports have been made, but the pathophysiological mechanisms behind carpal bone agenesis are unknown. If carpal absence is a true congenital malformation, it could arise from failure of differentiation of the carpal bone chondrification centers.

The carpal bones are formed during embryogenesis by separation and cavitation of the cartilaginous precursors of the carpals. Embryonic chondrogenic differentiation of carpal bones can be seen at approximately 42-45 days gestational age. The capitate is the first carpal bone to appear with immature pre-cartilage at 47-50 days gestational age. Pre-cartilage of the future scaphoid, lunate, hamate, and triquetrum appear over the next 2-3 days followed by the trapezium and trapezoid. The lunate itself begins to chondrify at Streeter 18th and 19th horizons of embryonic development with the center of ossification appearing at 2 years. Ossification is complete by 15-25 years old. The pisiform is the last carpal bone to appear. Chondrification centers for all carpal bones can be seen by 58 days gestational age. There are no reports of carpal agenesis in infants.

Another proposed mechanism of absence of carpal bones is idiopathic non-hereditary osteolysis of the carpal chondrification center after its development, which could occur in utero or during childhood. In that instance, the inheritance is autosomal dominant with variable expressivity presenting as in our patient with marked deformity in the third decade as the bone completely dissolves. This is in distinction to primary idiopathic osteolysis, which has an onset in early childhood, is associated with foot defects, facial abnormalities, kidney disease and hypertension.

Given the rarity of congenital carpal bone absence, the underlying pathophysiology will likely never be known.

**CONFLICTS OF INTEREST DISCLOSURE**

The authors declare they have no conflicts of interest.
REFERENCES

[1] Hoffmann D. Missbildungen der oberen Extremitat. Arch Klin Chir. 1911; 17: 303-5.

[2] Smith JG, Weiss A-PC, Weiss YS. Congenital Anomalies of the Hand. Clin Pediatr (Phila). 1998 Aug 1; 37(8): 459-67. PMid: 9729700. https://doi.org/10.1177/000992289803700801

[3] O’Rahilly R, Müller F. Developmental stages in human embryos: revised and new measurements. Cells Tissues Organs (Print). 2010; 192(2): 73-84. PMid: 20185898. https://doi.org/10.1159/000289817

[4] Patankar H. Bilateral Congenital Aplasia of the Scaphoid. Journal of Hand Surgery. 1998 Dec 1; 23(6): 817-9. https://doi.org/10.1016/S0266-7681(98)00108-3

[5] Schick N. Congenital Absence of Carpal Scaphoid. Br Med J. 1972 Jul 22; 3(5820): 236. https://doi.org/10.1136/bmj.3.5820.236-b

[6] Srivastava KK, Kochhar VL. Congenital Absence of the Carpal Scaphoid: A Case report. JBJS. 1972 Dec; 54(8): 1782. https://doi.org/10.1016/S0266-7681(72)80108-3

[7] Kuz JE, Smith JM. Congenital absence of the scaphoid without other congenital abnormality: A case report. The Journal of Hand Surgery. 1997 May 1; 22(3): 489-91. https://doi.org/10.1016/S0165-2105(97)80017-2

[8] Papanikolaou P, Haddadin M. Congenital absence of the scaphoid (letter). British Medical Journal. 1972; 2: 292. PMid: 5022023. https://doi.org/10.1136/bmj.2.5808.292-c

[9] Panciera P, Le Viet D. Intercarpal Degenerative Arthritis in Adulthood as a Late Consequence of Unilateral Congenital Aplasia of the Scaphoid: A Case Report. The Journal of Hand Surgery. 2008 Feb 1; 33(2): 213-6. PMid: 18294543. https://doi.org/10.1016/j.jhsa.2007.10.010

[10] Kobayashi H, Kosakai Y, Usui M, et al. Bilateral deficiency of ossification of the lunate bone. A case report. The Journal of Bone & Joint Surgery. 1991 Sep; 73(8): 1255-6. https://doi.org/10.2106/00004623-199173080-00017

[11] Roche AF. Absence of lunate. Roentgenol. 1967; 100: 523-5. PMid: 4961620. https://doi.org/10.2214/ajr.100.3.523

[12] Eraltug U. An unusual variety of carpal agenesis. Int Surg. 1966: 46: 594-5.

[13] Postacchini F, Ippoplito E. Isolated absence of human carpal bones. Teratology. 1975 Jun; 11(3): 267-71. PMid: 1154291. https://doi.org/10.1002/tera.1420110306

[14] Gurav RM, Patil AB. Bilateral hand malformations with absence of carpal bones with fusion of proximal metacarpals. Clin Dysmorphol. 2012 Jan; 21(1): 56-7. PMid: 22064728. https://doi.org/10.1007/s10822-011-0109-2

[15] De Smet L. Unilateral congenital absence of the lunate and scaphoid. Genet Couns. 2005; 16(4): 413-5.

[16] Streeter GL. Developmental horizons in human embryos; a review of the histogenesis of cartilage and bone. Contrib Embryol. 1949 Feb; 33(213-221): 149-68.

[17] Naranjo A, Muniaín MA, Martín I, et al. Primary idiopathic osteolysis: description of a family. Ann Rheum Dis. 1992 Sep; 51(9): 1074-8. PMid: 1417141. https://doi.org/10.1136/ard.51.9.1074