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CASE REPORT

Unilateral congenital absence of the carpal scaphoid associated with dysplasia of the capitate. Presentation of a case[☆]

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KEYWORDS

Aplasia carpal;
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Abstract We report on a case of aplasia or unilateral congenital absence of the carpal scaphoid associated with dysplasia of the capitate. Congenital absence of the carpal scaphoid is a rare but well-documented condition. As far as we know, the present case is the seventh one reported in the medical literature. Imaging studies (X-ray and MRI) confirmed the absence of the carpal scaphoid associated with a dysplasia of the capitate and malformation of the radial styloid process. Congenital absence of the scaphoid when other congenital abnormalities (such as hypoplasia or aplasia of either forearm bones or thenar eminence, malformations of the skeletal elements of the thumb, absence of sesamoid bones or abnormal development of the forearm bones) do not exist is probably the main feature of the present case report.

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PALABRAS CLAVE

Aplasia carpiana;
Escafoides;
Congénita

Ausencia congénita unilateral del escafoides carpiano y displasia del hueso grande. A propósito de un caso

Resumen Presentamos un caso de aplasia o ausencia congénita unilateral del escafoides carpiano asociado a displasia del hueso grande. La ausencia congénita del escafoides carpiano es una entidad rara e infrecuente, pero bien documentada. Es el séptimo caso presentado en la literatura. A diferencia de la mayoría de los casos publicados de aplasia/hipoplasia de escafoides carpiano, en nuestro caso no se ha detectado asociación de otras anomalías congénitas tipo hipoplasia o aplasia de musculatura tenar y antebrazo, malformaciones del esqueleto del pulgar, ausencia de huesos sesamoideos o alteraciones del desarrollo de los huesos del antebrazo. El estudio radiológico revela la ausencia congénita del escafoides asociado a una displasia del hueso grande e hipoplasia de la estiloides radial. Se completa el estudio con resonancia magnética nuclear.

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Introduction

Congenital aplasia of the carpal scaphoid is extraordinary rare, but it is much more frequently found in an isolated form.¹⁻⁶ Although this disorder is normally described in association with radial hemimelia or thumb aplasia/hypoaplasia,⁷ it can also coexist with other congenital syndromes, such as the one known by its acronym VACTERL (vertebral defects, anal atresia, cardiac anomalies, tracheoesophageal fistula, oesophageal atresia [“e” from the U.S.A. spelling, esophageal] and renal and limb anomalies)⁸, Holt-Oram syndrome (cardiac defects and upper limb anomalies)⁹ or TAR syndrome (thrombocytopenia-radial absence).¹⁰

The patient described in this article did not present associated anomalies or other developmental defects of the skeleton in the same limb. The patient was the 7th case of isolated unilateral aplasia congenital of the carpal scaphoid bone and the 1st case associated with capitate bone fusion.

Clinical case

The patient was a 40-year-old man, a Traumatologist working in the field of traumatic hand surgery for a mutual traffic accident insurance company. For the past 20 years, he had been having minor pains in the right wrist (his dominant hand), although with “overload synovitis” related to his work. These had limited his activities partially, needing only occasional symptomatic drug treatment. Fifteen years ago,

acroparesthesia appeared in the area of the median nerve, originally occurring only following exertion and normally at night. The condition became gradually worse and continual, to the point that he presented thenar amyotrophy with a positive Tinel’s sign in the wrist.

Upon physical examination, the clinical aspect of right hand (Fig. 1a and b) presented minimal differences with the contralateral one, such as a slight swelling located in the anatomical snuffbox that erased the tendon structures limiting it, as well as a minor thenar flattening secondary to the nerve entrapment. In the last few years, evolution tended toward normality. Joint mobility was 58° for palmar flexion and 40° for dorsal flexion, with the values for the left being 60° and 70°, respectively. For the right wrist, the radial deviation was 10° and the cubital deviation, 30°; for the left wrist, the deviations were 20° and 35°, respectively. Right thumb opposition was up to the interdigital commissure between the 4th and 5th fingers, while it was the base of the little finger on the palmar side for the left thumb. There was no reduction of the 1st commissure, with equal radial and palmar abduction bilaterally. The grip strength of the right hand measured with a Jamar dynamometer 30 kg, compared with 55 kg of the left.

The radiological study of the right wrist (Fig. 1c and d) revealed the complete absence of the scaphoid bones, together with a capitate dysplasia, as well as hypoplasia of the radial styloid and trapezium. Likewise, compensatory changes were seen in the carpal bone, with proximal migration and radial movement of the 2nd carpal bone. The lunate bone articulated proximally with the radius in correct

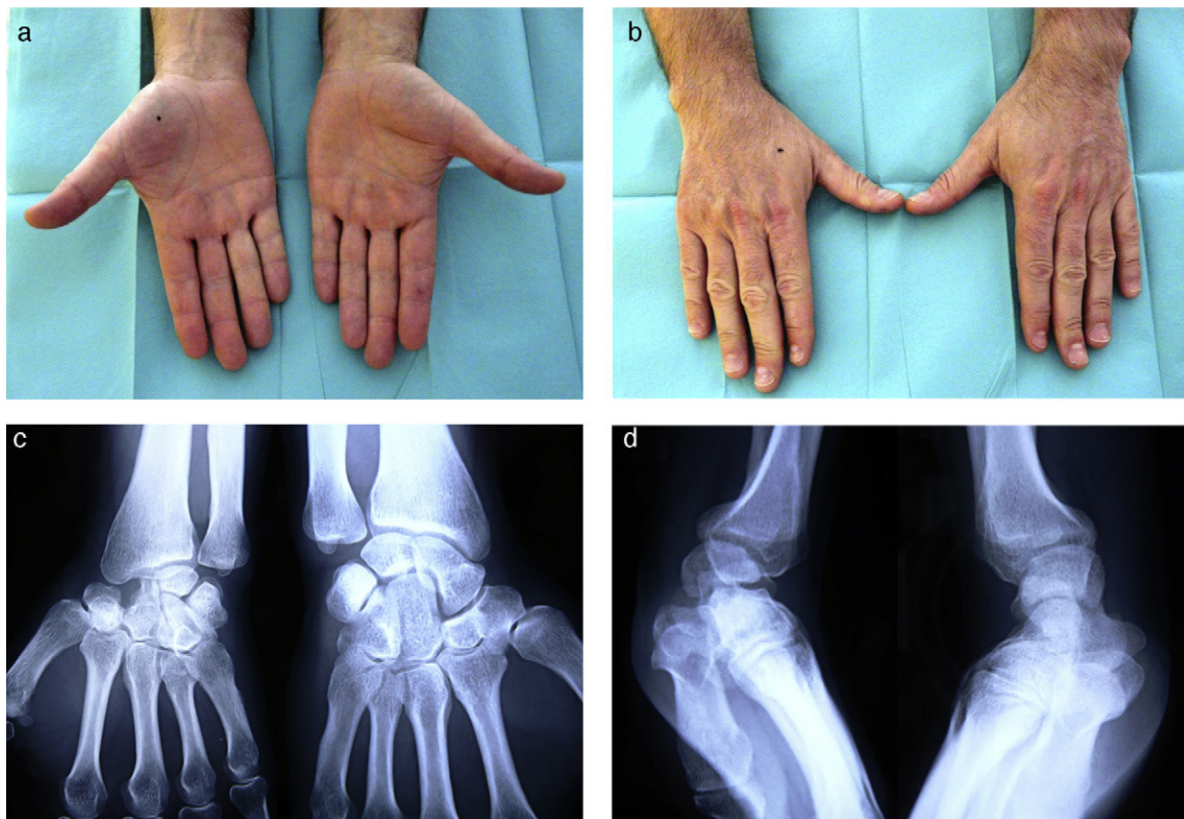


Figure 1 Clinical aspect of the hand (a and b). Radiological study of the right wrist (c and d).

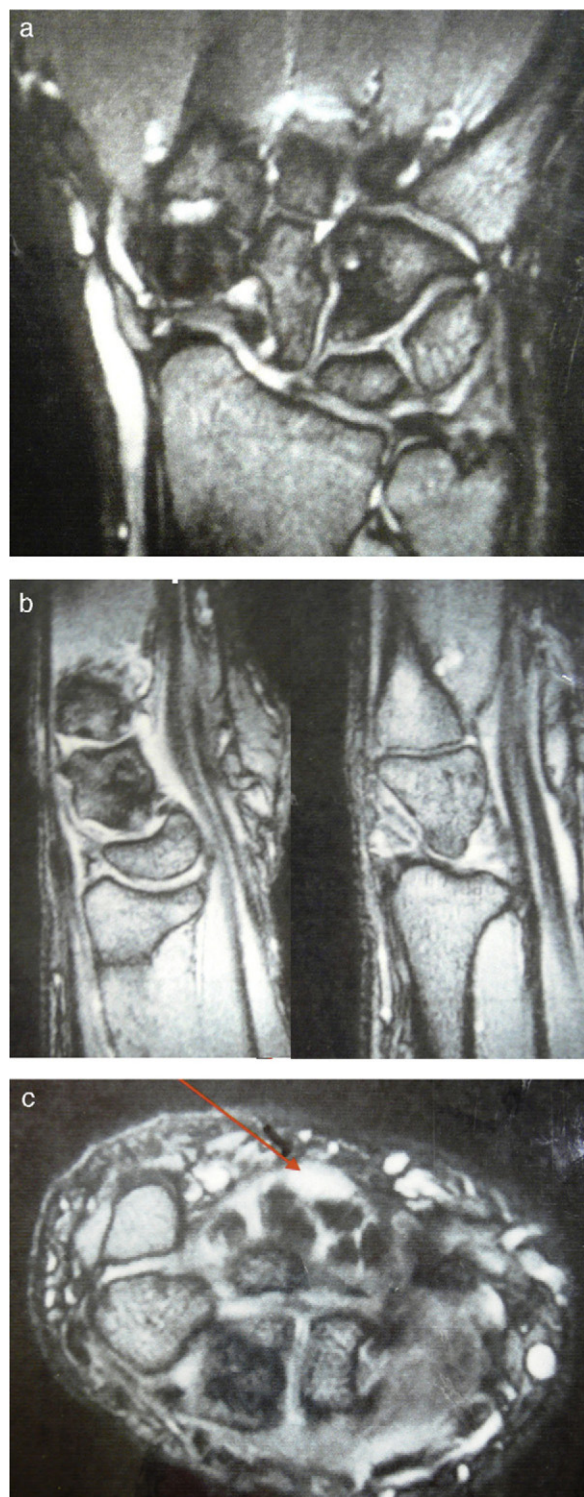


Figure 2 Nuclear magnetic resonance study.

anatomical form, with dorsal inclination (dorsal intercalated segment instability [DISI]), and articulated distally with the hamate. The capitate articulated with the radius as the lunate bone was cubital to it. The carpal height index was 0.43 in the right wrist and 0.60 in the left; that is, it was 28% less on the side without scaphoid bones. The radiocarpal

and intercarpal joint spaces were free from degenerative changes. The contralateral wrist was completely normal.

The nuclear magnetic resonance (NMR) study (Fig. 2a–c) displayed the existence of a small rudiment of the carpal scaphoid in the form of dense fibrous tissue, a slight osteochondral lesion in the capitate that articulated with the radius, subchondral sclerosis in the radius and trapezium (probably secondary to impingement), an inclination in lunate dorsiflexion (DISI pattern) and slight oedema in the median nerve. The electromyography detected severe neuropathy from median nerve entrapment in the carpal tunnel. Given the function liberation that the patient presented, he was operated on and the median nerve was liberated by endoscopic release. After the surgery, the neuropathic pain and episodes of acroparesthesia disappeared.

The patient is currently free from algesic symptoms, which allows him to perform his daily work and sport activities without any limitations or problems.

Discussion

The isolated absence of the carpal scaphoid without other congenital anomalies is very rare; in fact, we found only 6 cases of isolated lesion in the literature,^{1–6} and none of them associated with dysplasia of the capitate. The condition has been described in association with hypoplasia of the radius or the thumb, with the absence of other carpal bones, or as part of congenital syndromes (VACTERL, HOLT-ORAM or TAR) with these constituting the majority of the cases published. In 1953, in an attempt to classify carpal and tarsal anomalies, O’Rahilly¹¹ described 4 categories: (1) severe anomalies associated with abnormalities of the digital or antebrachial segments, or both; (2) fusions; (3) accessory ossicle cases, including the sesamoid; and (4) bipartition cases. Within the 1st group, he described the “radial hemimelia” as the absence of the scaphoid-trapezium-thumb metacarpal. He also described specific conditions in which the proximal and distal zones of the area affected were not themselves necessarily affected, and defined them as intercalary defects. Intercalar radial hemimelia radial is an unusual condition in which the intercalary defect occurs in the radial aspect of the wrist. There is generally a radial styloid deficiency accompanied by an underdevelopment or failure of the scaphoid-trapezium-metacarpal complex. That author reported an incidence of carpal scaphoid and thumb absence of 80%. In 1962, Davison¹² established a classification of 6 different degrees of carpal scaphoid hypoplasia, from a very slight hypoplasia of the 1st radius up to a hand with 4 fingers and radial hypoplasia. He explained that the defect was as a result of an embryological insult that affected the skeleton in a distal proximal sequence. No description was given of the carpal scaphoid associated with a normal thumb. In this author’s classification, the congenital absence of the scaphoid bone was always associated with severe thumb hypoplasia. The International Federation of Societies for Surgery of the Hand (1999)¹³ divided congenital hand deformities into 7 categories or groups. Aplasia or hypoplasia of the scaphoid was always associated with thumb deficiencies, and they included it in the first group (failure in development of parts), subgroup 1b

(radial longitudinal detention deficiency).¹⁴ In 1999, James et al.¹⁵ after reviewing a lengthy series of radial longitudinal deficiency cases, did not find carpal scaphoid aplasia with a normal thumb. It would be normal to expect a carpal collapse and degenerative wrist changes in our case of congenital scaphoid bone absence, but that was not the case. In the literature there are 2 cases of carpal collapse associated with scaphoid anomalies that developed degenerative arthropathy.^{16,17} We do not feel that the median nerve entrapment (carpal tunnel syndrome) originated from the congenital scaphoid absence, with secondary narrowing of the carpal tunnel; if that were the case, this process would have shown up clinically previously.¹⁸ We should conclude by indicating that this isolated congenital absence of the carpal scaphoid bone is characterised by its good tolerance throughout life, with no job or sport limitations. In addition, in spite of the "biomechanical alteration" or harmonic adaptation of the rest of the carpal bone, it withstands the loads transmitted from the hand to the forearm. It is interesting to highlight that the collapse produced has found a well-tolerated balance point, probably because the ligament system guarantees appropriated joint mobility.

Level of evidence

Level of evidence IV.

Ethical disclosures

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this investigation.

Confidentiality of Data. The authors will declare that they have followed the protocols of their work centre on the publication of patient data and that all the patients included in the study have received sufficient information and have given their informed consent in writing to participate in that study.

Right to privacy and informed consent. The authors must have obtained the informed consent of the patients and /or subjects mentioned in the article. The author for correspondence must be in possession of this document.

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