World Journal of Advanced Research and Reviews
eISSN: 2581-9615 CODEN (USA): WJARAI
Cross Ref DOI: 10.30574/wjarr
Journal homepage: https://wjarr.com/

(CASE REPORT)

Bilateral Congenital agenesis of the Proximal Pole of the Scaphoid: A Rare Variant of Carpal Anatomy

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World Journal of Advanced Research and Reviews, 2025, 28(03), 068-073

Publication history: Received 22 October 2025; revised on 29 November 2025; accepted on 01 December 2025

Article DOI: https://doi.org/10.30574/wjarr.2025.28.3.4022

Abstract

Introduction: Congenital anomalies of the carpus are rare, and scaphoid agenesis is an exceptionally uncommon condition. Most reported cases involve unilateral absence and are frequently associated with other malformations such as thumb hypoplasia or radial hemimelia. No previous reports have described isolated bilateral agenesis of the proximal pole of the scaphoid.

Materials and Methods: We present the case of a 32-year-old male evaluated for intermittent right-wrist pain. Clinical examination was complemented with bilateral wrist radiographs, computed tomography, and magnetic resonance imaging to characterize carpal anatomy and exclude associated abnormalities.

Results: Imaging revealed bilateral congenital agenesis of the proximal pole of the scaphoid with preservation of the distal portion. No additional skeletal, systemic, or soft-tissue anomalies were identified. The patient had no significant medical or family history of congenital disorders. Physical examination demonstrated normal wrist alignment, full range of motion, and no functional limitations. Because symptoms were mild and the wrist remained stable, conservative management with periodic follow-up was recommended.

Discussion: This appears to be the first documented case of isolated bilateral congenital agenesis of the proximal pole of the scaphoid. The patient's benign clinical course supports a non-interventional approach. Recognition of this rare variant is crucial to avoid misdiagnosis, particularly confusion with post-traumatic conditions.

Conclusion: Isolated bilateral congenital agenesis of the proximal pole of the scaphoid represents a previously undescribed anatomical variant. Awareness of this condition may prevent unnecessary interventions and contributes to understanding congenital carpal anomalies.

Keywords: Scaphoid Agenesis; Congenital Anomalies; Bone Malformation; Hand Anomalies

1. Introduction

Congenital anomalies of the upper limb represent approximately 1–2% of all birth defects, and about 10% of these involve the wrist and hand[1, 2]. These alterations encompass a broad spectrum of structural defects, ranging from digital duplications to bony fusions or complete absence of anatomical segments. Within this spectrum, scaphoid agenesis is

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considered an exceptionally rare entity, most commonly described as part of broader syndromes involving radial ray dysplasia or complex genetic alterations[3].

During embryogenesis, the development of the skeletal structures of the hand begins between the fourth and eighth weeks of gestation. During this critical period, mesenchymal cells condense and differentiate to form the cartilaginous templates of the carpal bones[4, 5]. Any disruption of this process—whether due to genetic mutations, teratogenic exposure, infections, hypoxia, or environmental factors—may result in significant structural abnormalities of the limbs.

Scaphoid agenesis has been described either in isolation or as part of syndromes such as Holt–Oram, which combines upper limb malformations with congenital heart disease[6], or the VACTERL association, characterized by vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb anomalies[7]. In the vast majority of cases, its presentation is unilateral and associated with radial deficiency, thumb hypoplasia, or carpal dysplasia.

There are several classifications for congenital anomalies of the upper limb, the most widely used being that of the International Federation of Societies for Surgery of the Hand, based on Swanson's system. It is divided into seven groups: (I) failure of formation; (II) failure of differentiation; (III) duplication; (IV) overgrowth; (V) undergrowth; (VI) congenital constriction band syndrome; and (VII) generalized skeletal anomalies. Scaphoid agenesis or hypoplasia is typically associated with thumb deficiencies and classified within Group I (failure of formation with longitudinal radial deficiency). However, no specific classification for congenital anomalies of the scaphoid itself has yet been described in the literature.

It is noteworthy that, to date, there are no reports in the medical literature of bilateral congenital agenesis of the proximal pole of the scaphoid in the absence of associated anomalies. Thus, the present case represents a novel contribution to the anatomical and clinical understanding of congenital carpal variants. We describe a young adult with no traumatic history or systemic malformations whose joint function remains normal despite this bilateral structural anomaly. This finding underscores the importance of considering congenital anatomical variants as possible differential diagnoses when encountering atypical radiographic findings, helping avoid diagnostic errors—such as mistaking them for pseudarthrosis or avascular necrosis and supporting more conservative, function-oriented decision-making.

2. Materials and Methods

This is a descriptive case report. The patient was evaluated in the outpatient orthopedic clinic for mild, intermittent pain in the right wrist. There was no history of recent trauma or family history of malformations. The assessment included: Complete physical examination, Plain radiographs of both wrists, Computed tomography (CT), Magnetic resonance imaging (MRI).

Infectious processes, significant perinatal events, consanguinity, growth abnormalities, or prior surgical history were ruled out. All imaging studies were interpreted by musculoskeletal radiologists, and the findings were reviewed by a multidisciplinary team.

3. Results

We present the case of a 32-year-old male who consulted due to mild right wrist pain of two years' duration. He had previously sustained an indirect wrist hyperextension injury, which resulted in mild swelling and dorsal wrist pain that improved with analgesics and home-based physical measures. Since then, he experienced intermittent pain and mild functional limitation in the dorsal wrist region. There was no history of joint or soft-tissue infection, childhood illnesses, or congenital defects. His parents were not consanguineous, and he was born at term without pregnancy or perinatal complications. No neonatal intensive care was required.

Physical examination revealed a healthy-appearing patient with normal nutritional status and unremarkable facial features. The right wrist displayed no scars, deformities, pain, or edema. The thenar and hypothenar eminences of both hands were normal. Range of motion of the wrists and fingers was preserved, with normal muscular strength and no upper extremity length discrepancies or distal neurovascular deficits (Figures 1–3).

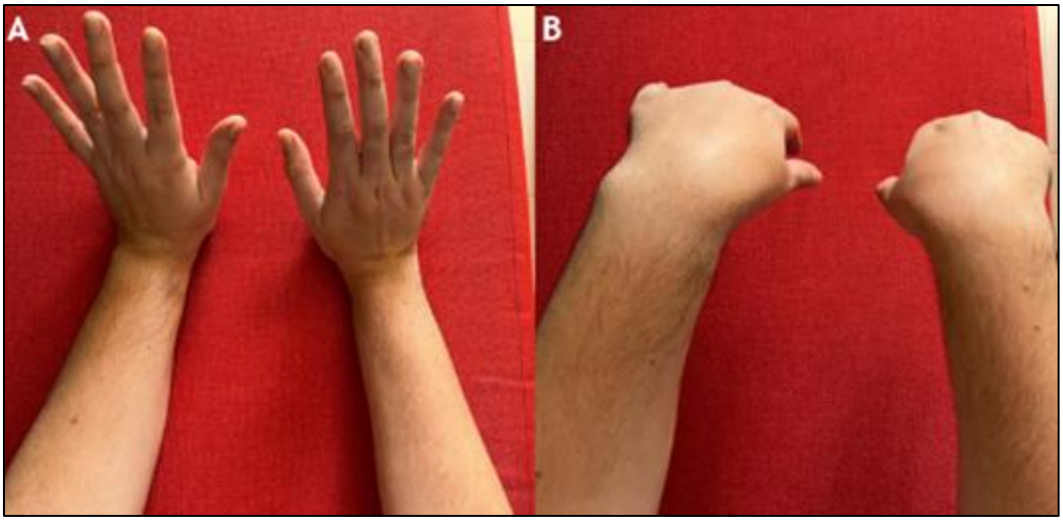

Figure 1 A. Patient's wrists in extension. B. Patient's wrists in flexion

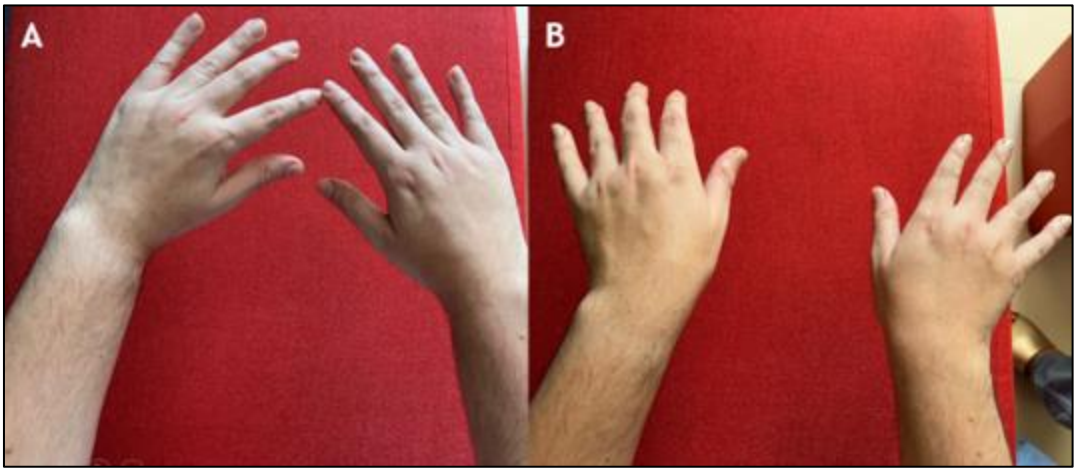

Figure 2 A. Wrists in radial deviation. B. Wrists in ulnar deviation

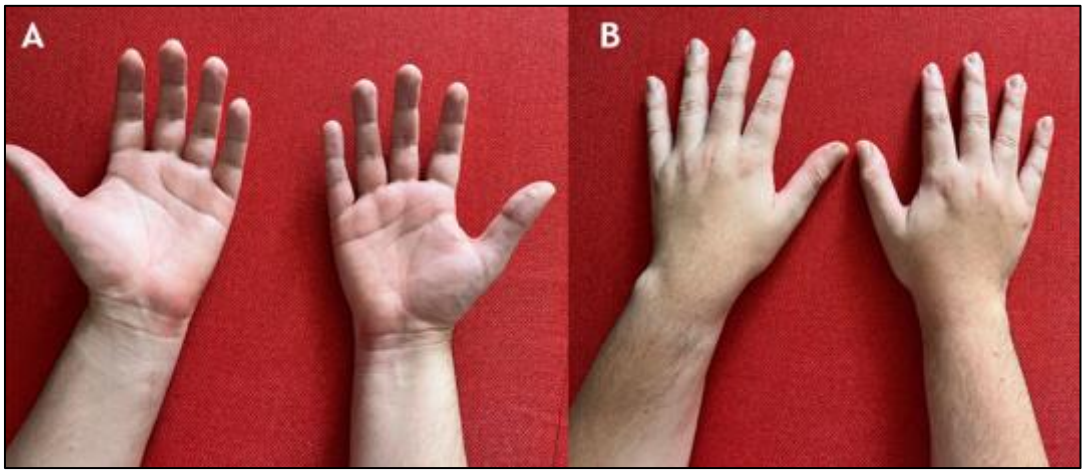

Figure 3 A. Hands in supination. B. Hands in pronation

Radiographs of the right wrist demonstrated agenesis of the proximal pole of the scaphoid with marginal osteophytes along the distal radial articular surface and scaphoid articular surface. Early arthritic changes were also noted at the scaphoid–trapezium joint. Given these findings, a CT scan and MRI of the right wrist were obtained, confirming the

radiographic abnormalities and ruling out additional alterations. Subsequently, radiographs of the left wrist were ordered, revealing agenesis of the proximal pole of the scaphoid as well (Figures 4–6).

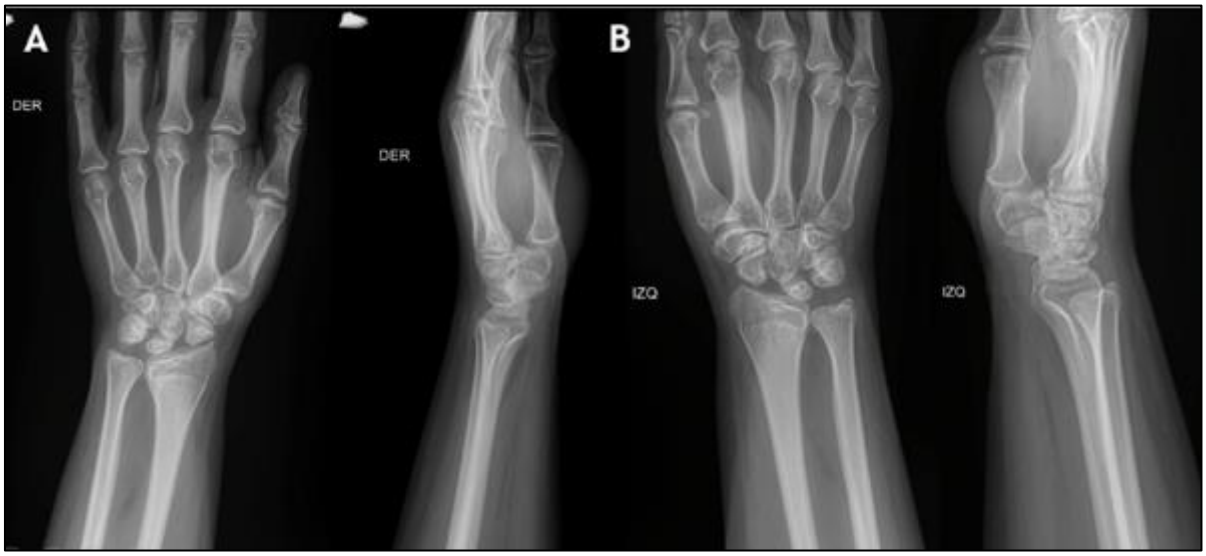

Figure 4 A. AP and lateral radiographs of the right wrist showing agenesis of the proximal pole of the scaphoid. B. AP and lateral radiographs of the left wrist also showing agenesis of the proximal pole of the scaphoid

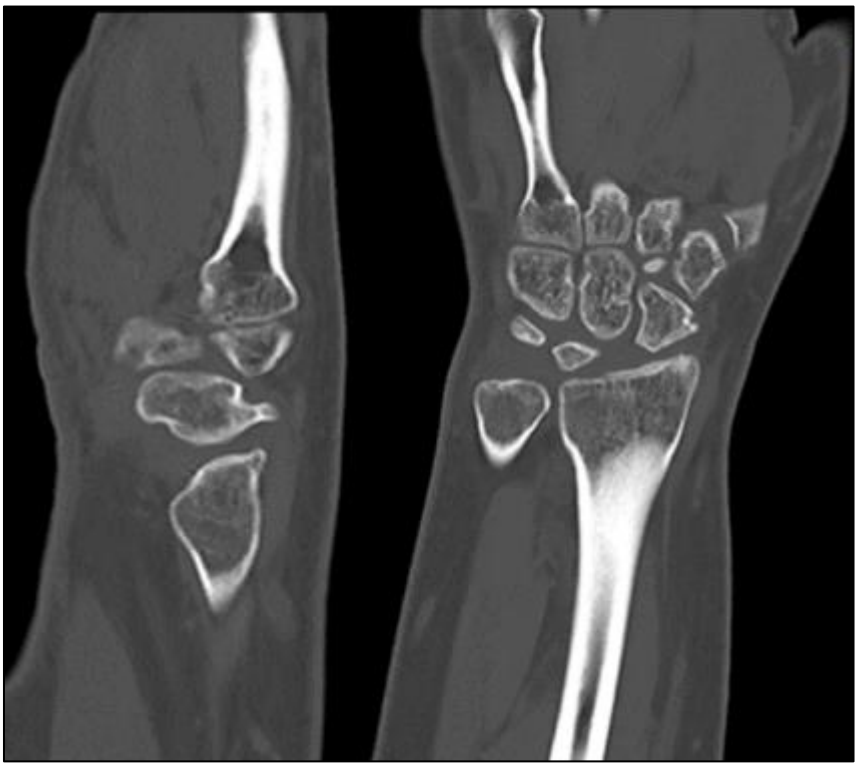

Figure 5 CT scan of the right wrist in coronal and sagittal planes

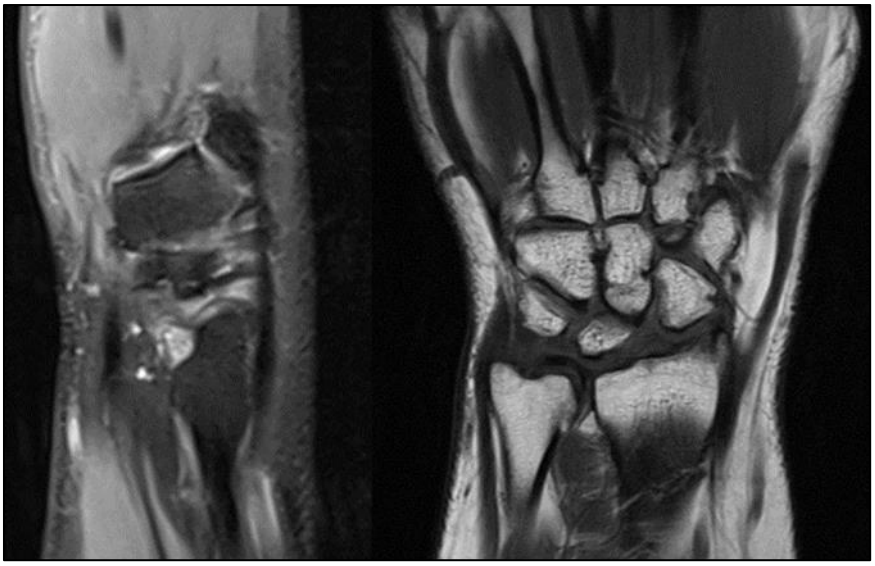

Figure 6 MRI of the right wrist in coronal and sagittal planes

The anatomical and imaging findings were consistent with early radiocarpal and intercarpal arthrosis secondary to biomechanical adaptation related to this anatomical variant. The diagnosis was explained to the patient, and conservative management was recommended. Any surgical intervention could potentially cause pain, reduce mobility, or accelerate arthritic progression by disrupting the established biomechanical adaptation. The patient was managed with analgesics and physical therapy and currently remains asymptomatic with preserved range of motion.

4. Discussion

Agenesis of the scaphoid, particularly of the proximal pole, is an extremely rare congenital anomaly within carpal malformations. In the medical literature, most described cases are unilateral and associated with genetic syndromes or radial ray abnormalities[3]. Therefore, this case constitutes a unique contribution, representing congenital, bilateral proximal scaphoid agenesis in the absence of associated defects and without perinatal, familial, or traumatic factors that could explain the condition.

The current classification of congenital upper limb anomalies, proposed by Swanson and adopted by the International Federation of Societies for Surgery of the Hand, categorizes these malformations as Group I: failure of formation with longitudinal radial deficiency[8]. However, there is no specific category addressing isolated scaphoid agenesis, suggesting potential underdiagnosis or misclassification. Some previous cases may have been misinterpreted as traumatic sequelae or avascular necrosis, especially in asymptomatic patients or in those lacking detailed clinical history.

The clinical relevance of this case lies not only in its rarity but also in the patient's preserved wrist function. Despite bilateral structural alteration, the patient exhibited no significant symptoms or range-of-motion limitations. This demonstrates that absence of the proximal scaphoid pole can be compatible with functional wrist biomechanics. The literature describes how the carpus may adapt to congenital anatomical variations through compensatory remodeling that preserves functionality[1, 7].

These findings highlight important diagnostic and therapeutic implications. When confronted with unusual radiographic findings or wrist pain, it is essential to differentiate between congenital variants and acquired pathology. CT and MRI play a crucial role in characterizing bone morphology and articular surfaces[5].In this case, the integration of imaging with clinical evaluation allowed exclusion of conditions such as scaphoid pseudarthrosis or avascular necrosis, both of which require different surgical approaches.

From a therapeutic standpoint, conservative management is consistent with the principle of avoiding anatomical intervention in the absence of significant functional impairment. Surgical treatment in such patients may produce pain, limit mobility, or accelerate arthrosis by altering the established compensatory biomechanics[2, 4].

5. Conclusion

Bilateral congenital agenesis of the proximal pole of the scaphoid without associated anomalies represents a previously unreported anatomical variant. Its recognition is essential to prevent diagnostic errors or unnecessarily aggressive treatment. This case broadens current knowledge of congenital carpal anomalies and highlights the importance of advanced imaging in the differential diagnosis of wrist pathologies.

Compliance with ethical standards

Disclosure of conflict of interest

The authors declare that there is no conflict of interest regarding the publication of this article.

Statement of informed consent

The patient provided written informed consent authorizing the use of his data and images for the publication of this case.

Funding

This article did not receive funding from any public or private source.

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