Journal of Hand Surgery Global Online 5 (2023) 852–855

Contents lists available at ScienceDirect

Journal of Hand Surgery Global Online

journal homepage: www.JHSGO.org

Case Report

A Rare Presentation of Postaxial Polydactyly in a 2-Year-Old Female with Ellis-van Creveld Syndrome

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ARTICLE INFO

Article history:
Received for publication August 17, 2023
Accepted in revised form August 22, 2023
Available online September 20, 2023

Key words:
Ellis-van Creveld syndrome
Postaxial
Polydactyly
Ulnar

Postaxial or ulnar polydactyly is the most common form of polydactyly that may present with the duplication of soft-tissue structures only or with additional bony involvement. Surgical excision is the only viable treatment option for postaxial polydactyly with bony involvement, and psychological or cosmetic reasons are the main rationale for treatment. Ellis-van Creveld syndrome (EVC) is a rare congenital disorder characterized by chondral and ectodermal dysplasia, particularly postaxial polydactyly. The exact prevalence of EVC is unknown, and fewer than 300 cases have been reported. We present a case of a 2-year-old Hispanic female with EVC who presented with bilateral postaxial polydactyly and complete duplication of the metacarpal and phalanges. We describe the presentation and treatment of this patient, who ultimately underwent staged resection of the duplicated digits with reconstruction of the abductor muscle.

Polydactyly is a congenital hand or foot deformity involving the development of additional digits.[1,2] Polydactyly can vary in appearance of the additional digit from simple cutaneous appendages to complex bony involvement and can be classified as preaxial, central, or postaxial. Postaxial or ulnar polydactyly is the most common, affecting 4 in 10,000 live births per year, and has been reported to be genetic, sporadic, or syndromic.[2] Treatment for postaxial polydactyly with soft-tissue duplication has historically been suture ligation postnatally or surgical excision.[1,3] Surgical excision is the only viable treatment option for polydactyly with bony involvement.[1,4] Since ulnar polydactyly is rarely associated with functional disabilities, psychological and cosmetic reasons are the main rationale for surgical treatment.[1]

Ellis-van Creveld syndrome (EVC) is a rare congenital disorder characterized by chondral and ectodermal dysplasia, particularly postaxial polydactyly.[5,6] Children with EVC exhibit a complex variety of comorbidities including cardiac defects, growth retardation, narrow thorax, and dysplasia of the teeth and fingernails. Approximately half of the patients with EVC do not live beyond childhood primarily due to cardiac abnormalities. The exact prevalence of EVC is unknown, and fewer than 300 cases have been reported.[6,7] We introduce a case of a 2-year-old Hispanic female with EVC who presented with bilateral postaxial polydactyly and complete duplication of the metacarpal and phalanges. We describe the presentation and treatment of this patient, who ultimately underwent staged resection of the duplicated digits with reconstruction of the abductor muscle. Written informed consent was obtained from the patient's legal guardian regarding the publishing of this report and associated figure images.

Case Report

A 2-year-old Hispanic female with EVC was brought to our clinic by her mother with concerns about duplicated digits of the patient's bilateral hands (Fig. 1). The patient's comorbidities included atrioventricular septal defect, hypoplastic aortic arch,

Declaration of interests: No benefits in any form have been received or will be received related directly to this article.

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https://doi.org/10.1016/j.jhsg.2023.08.007

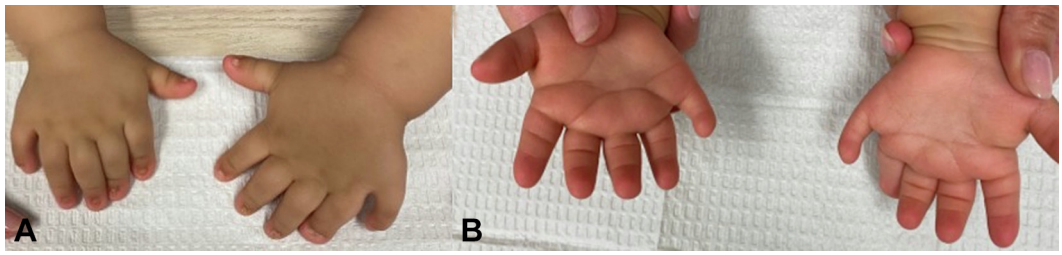

Figure 1. Preoperative clinical images of the **A** dorsal surface and **B** volar surface of bilateral hands demonstrating postaxial polydactyly bilaterally.

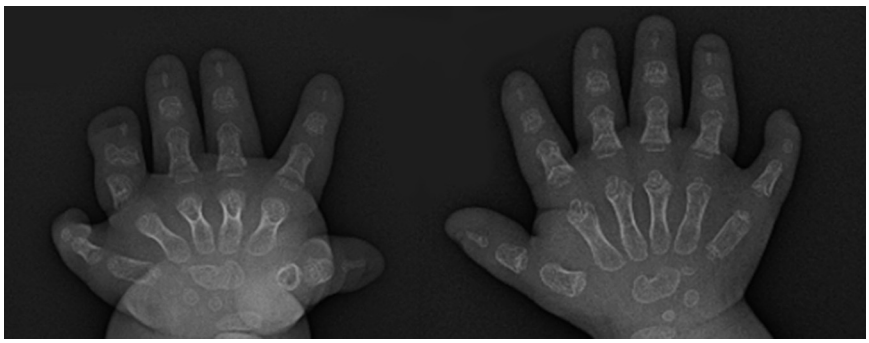

Figure 2. Preoperative posteroanterior radiographs bilateral hands demonstrating complex polydactyly with a complete duplication of the 6th metacarpal and the proximal, middle, and distal phalanges. There is an incidental finding of capitohamate coalition.

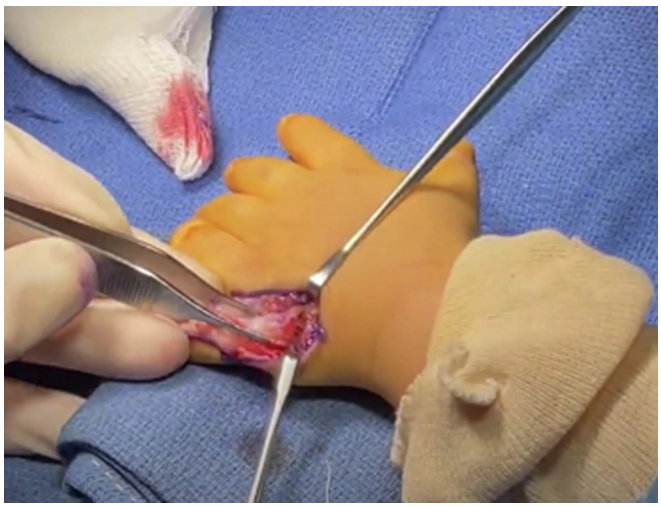

Figure 3. Intraoperative image of the left hand demonstrating resection of the 6th metacarpal at the level of the mid-shaft.

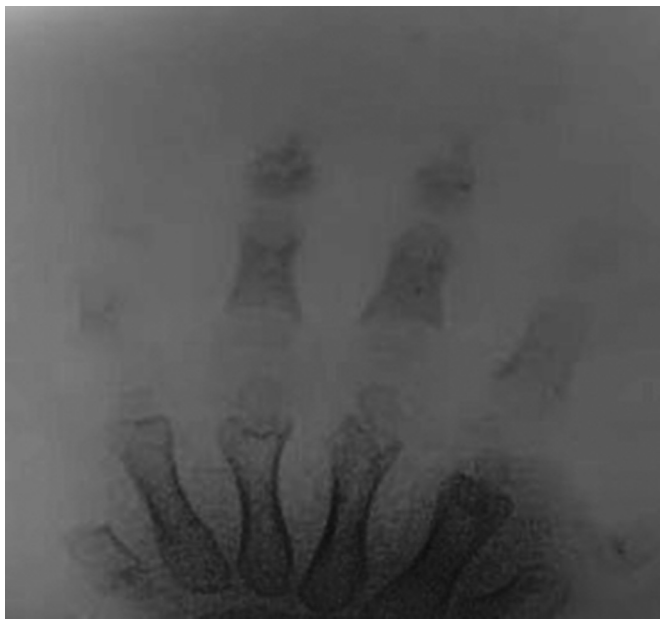

Figure 4. Intraoperative fluoroscopic image of left hand demonstrating successful resection of the 6th metacarpal at the level of the mid-shaft.

supraventricular tachycardia, chronic respiratory failure, and skeletal abnormalities including bilateral symmetric sixth digit duplication consistent with bilateral postaxial polydactyly. Radiographs at the time of her initial visit demonstrated complex polydactyly with a complete duplication of the sixth metacarpal and the proximal, middle, and distal phalanges (Fig. 2). Examination demonstrated full digital range of motion, strength, and sensation at the duplicated digits bilaterally. However, the patient's mother was concerned about potential injury as the duplicated digits often caught onto objects in the house and potential psychological effects as the patient grew. After the discussion of the risks and benefits of operative versus nonoperative treatment, the family elected to proceed with surgery. Excision was planned in a staged fashion with the right hand addressed first, followed by the left hand 5 months later to minimize complications associated with prolonged anesthesia.

The arm was exsanguinated, and the tourniquet was inflated to 150 mm Hg. An incision was made on the dorsal side extending from the area of the base of the duplicated metacarpal up to the web space. Tenotomy of the duplicated extensor tendon was performed in zone 6 at the mid-metacarpal shaft. Attention was turned to the volar aspect of the hand where the wedge resection was

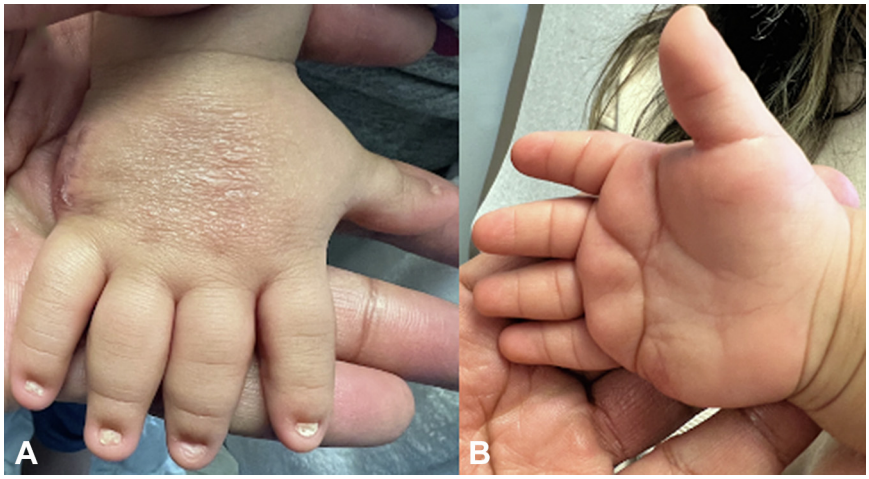

Figure 5. Postoperative clinical photos of the **A** dorsal surface and **B** volar surface of the right hand showing a well-healed incision and acceptable digit alignment without complications.

carried out with an incision down to the metacarpal. Tenotomy of the superficial and deep flexor tendons was then performed. The abductor musculature to the sixth metacarpal was dissected free as well as the digital neurovascular bundles and were transected. The collapsible portion of the metacarpal was excised with an oscillating saw, and the distal portion of the excised digit was removed (Fig. 3). The abductor muscle was sutured to the periosteum of the fifth metacarpal. Intraoperative radiographs demonstrated successful amputation of the duplicated ulnar digit without retained bony fragments (Fig. 4). The incision was closed with absorbable sutures, and the patient was placed in a well-padded ulnar gutter splint to be worn until her initial postoperative clinic visit.

The patient was seen weekly after surgery for the first 2 weeks and then again at 4 weeks. The splint was discontinued 1 week after surgery, and a soft dressing was applied allowing a gentle range of motion as tolerated. At 4 weeks, the patient had full sensation and range of motion and demonstrated excellent dexterity appropriate to her age. The incision healed appropriately without complications (Fig. 5). The left hand was addressed 5 months later after ample recovery of the patient's right hand. The postoperative course was similar to her right hand without complications.

Discussion

Polydactyly is one of the most common congenital hand abnormalities with a wide array of radial, central, or ulnar involvement. The African form of postaxial polydactyly is more common with a reported incidence of 1 in 150–300 live births and is more likely to occur bilaterally in a familial inheritance pattern. The Caucasian form is less common with an incidence of 1 in 1,500–3,000 live births, and most of these cases are unilateral and sporadic.[1] Temtamy and McKusick classified postaxial polydactyly as type A consisting of a duplicated digit with well-defined metacarpal articulation and bony connection to the hand. In contrast, type B consists of a poorly formed rudimentary tag on a pedunculated digit without any duplication of osseous structures.[8] Postaxial polydactyly can be further classified by the Stelling and Turek classification system. Type I includes the duplication of soft tissues only which are often removed by suture ligation or surgical excision depending on the surgeon's preference.[3,4] Surgical excision is necessary for treating types II and III, including the duplication of osseous structures with type III exhibiting a complete duplication of the ray and a separate metacarpal.[4] Given the potential for social esteem and societal issues and possible injury by catching on objects and clothes as the patient matures, the family was offered surgical excision which was accepted.

There has been debate in recent literature on whether suture ligation or surgical excision is optimal for Stelling type I duplicated soft tissue prominences. Although suture ligation has been considered more convenient and cost effective, previous literature reports an increased risk of an unaesthetic amputation stump in addition to painful neuroma formation.[1] Watson and Hennrikus performed suture ligation postnatally and reported parent satisfaction in cosmesis in all cases despite residual bumps occurring in 43% of the ligated digits.[3,9] Singer et al[1] recommended surgical excision for all forms of postaxial polydactyly after demonstrating a 10% residual remnant rate with good postoperative outcomes.[3] In contrast to the differing opinions on treatment for type I polydactyly, the only treatment option for types II and III is surgical excision. Depending on the extent of osseous duplication of digit, previously described techniques involve corrective osteotomy, ligament, soft tissue reconstruction, and articular contouring.[1,4] Although previous studies have been heterogeneous in regard to polydactyly type, surgical excision of types II and III have demonstrated satisfactory results.[1] The patient in our case had an uneventful recovery with full sensation, range of motion, strength, and dexterity of both hands. There were no complications.

Currently, no gold standard exists regarding the optimal age of patients undergoing surgical excision and reconstruction. Most authors recommend an age of 12 months because patients at this age have a lower risk of complications with general anesthesia, a deficient functional pinch, and have not developed concerns with their self-image.[4] Patients with polydactyly due to a syndrome should be optimized before their elective hand surgery as was performed with our patient with EVC who had cardiac and respiratory abnormalities. Our decision to stage these procedures within 6 months was to limit the time under general anesthesia.

References

1. Singer G, Thein S, Kraus T, Petnehazy T, Eberl R, Schmidt B. Ulnar polydactyly - an analysis of appearance and postoperative outcome. *J Pediatr Surg*. 2014;49(3):474–476.
2. Wessel LE, Daluiski A, Trehan SK. Polydactyly a review and update of a common congenital hand difference. *Curr Opin Pediatr*. 2020;32(1):120–124.
3. Chopan M, Sayadi L, Chim H, Buchanan PJ. To tie or not to tie: a systematic review of postaxial polydactyly and outcomes of suture ligation versus surgical excision. *Hand (N Y)*. 2020;15(3):303–310. https://doi.org/10.1177/1558944718810885

4. Comer GC, Potter M, Ladd AL. Polydactyly of the hand. *J Am Acad Orthop Surg*. 2018;26(3):75–82.
5. Ellis RW, van Creveld S. A syndrome characterized by ectodermal dysplasia, polydactyly, chondro-dysplasia and congenital morbus cordis: report of three cases. *Arch Dis Child*. 1940;15(82):65–84.
6. Baujat G, Le Merrer M. Ellis-van Creveld syndrome. *Orphanet J Rare Dis*. 2007;2:27.
7. Baghianimoghadam B, Arabzadeh A, Fallah Y. Ellis-van Creveld syndrome in Iran, a case report and review of disease cases in Iran, Middle East. *Acta Med Litu*. 2021;28(2):317–324.
8. Temtamy SA, McKusick VA. The genetics of hand malformations. *Birth Defects Orig Artic Ser*. 1978;14(3):1. i-xviii, 1–619.
9. Watson BT, Hennrikus WL. Postaxial type-B polydactyly. Prevalence and treatment. *J Bone Joint Surg Am*. 1997;79(1):65–68.