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A Case Report on Ellis–van Creveld Syndrome: Clinical, Embryological, Anesthetic, and Surgical Implications

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Abstract

Postaxial polydactyly (PAP) in the form of rudimentary soft tissue masses is quite common. Management involves ligation or surgical excision. Rarely do literature discussions cover complex variants in which the extra finger is fully developed. Ellis–van Creveld syndrome (EVC) or chondroectodermal dysplasia is a rare disorder characterized by PAP. When chondral dysplasia encompasses PAP, dwarfism, and genu valgum, ectodermal dysplasia involves nails and teeth. We describe two EVC cases. When one had cardiac malformations, the other had dental anomalies. One of them underwent genu valgum correction and removal of the additional finger. The hand surgery proved complex due to fusion of its metacarpal with the fifth metacarpal. Pediatric Outcomes Data Collection Instrument (PODCI) scores at 1.5 and 2.5 years after hand and knee surgeries were excellent. A multidisciplinary approach is essential to achieve comprehensive care. Additional embryological research is necessary to elucidate the clinical manifestations described in this report.

Keywords

- postaxial polydactyly
- ulnar polydactyly
- Ellis–van Creveld syndrome

Introduction

Polydactyly is one of the most common congenital hand differences. Postaxial polydactyly (PAP) is the predominant form.[1] When much discussion happens on the management of the more common soft tissue nubbins, complex types associated with syndromes like Ellis–van Creveld syndrome (EVC) that hinder hand function are seldom discussed. Only less than 300 EVC cases are documented to date.[1] This study describes two EVC cases in detail (►**Fig. 1**).

Case Report

Case 1

A 11-year-old boy, born to parents of consanguineous marriage, had PAP of both hands, genu valgum, and congenital heart disease. The right extra digit interfered with his writing. Clinical manifestations in the hands are shown in ►**Fig. 2**. Medial angulation at all the metatarsophalangeal joints, metacarpal synostoses, symphalangism, and great toe duplication were present.

His height was 142 cm, below the 50th percentile on height chart for age and gender.[2] Although he had undergone cardiac surgery, preoperative evaluation revealed residual cardiac lesions. He was advised tricuspid valve replacement before limb surgery. He has had three cardiac operations. His parents were now hesitant for limb surgery due to his cardiac issues.

Case 2

A 7-year-old boy, born to parents of nonconsanguineous marriage, had clinical and radiological features similar to

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Fig. 1 Children with features of Ellis–van Creveld syndrome: bilateral postaxial polydactyly, genu valgum, and reduced growth. (**A**) Case 1 and (**B**) Case 2.

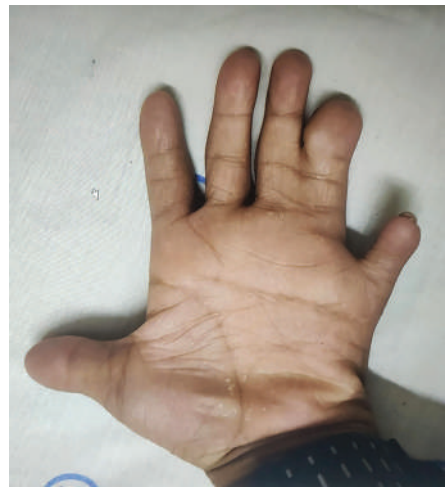

Fig. 3 Left hand of case 2 showing the hypoplastic sixth digit with simple incomplete syndactyly between the ring and little fingers. The middle and ring finger tips are at the same level.

those of case 1. His primary concern was the challenge of mixing rice and eating. He had difficulty in putting his hand in the pants pocket. His left hand had additional features like syndactyly between the ring and little fingers, and duplicated distal phalanx of the little finger (►**Fig. 3**). Genu valgum was corrected by bilateral proximal tibial hemiepiphysiodesis. His feet were malformed with six toes each. Dysplastic nails and dental anomalies are shown in ►**Fig. 4**. He was 111 cm tall, which corresponds to the third percentile for age and gender.[2] Extremities were disproportionately short with an upper-to-lower segment ratio of 1.22 (normal: 1.01).[3]

The right extra digit was removed under general anesthesia, regional block, and tourniquet. An incision made around the base of the digit was extended distally as Z-plasty along the junction of glabrous and nonglabrous skin. Blunt dissection exposed the sixth metacarpal. In the subperiosteal plane, hypoplastic ligamentous and muscular attachments to the extra metacarpal were divided and suture-tagged for reattachment. The extra digit was removed by dividing the metacarpal synostosis junction. The elevated periosteal sleeve was used to cover the raw bony amputation stump with nonabsorbable sutures. The detached structures were sutured to the existing hypothenar musculature. Skin was closed and Z-plasty flaps were transposed (►**Fig. 5**). The wounds healed well. The Pediatric Outcomes Data Collection Instrument (PODCI) was administered to the parent at the final follow-up, 1.5 years and 2.5 years after hand and knee surgeries, respectively. PODCI scores were 96 for upper extremity, 100 each for transfer/mobility, sports/physical functioning,

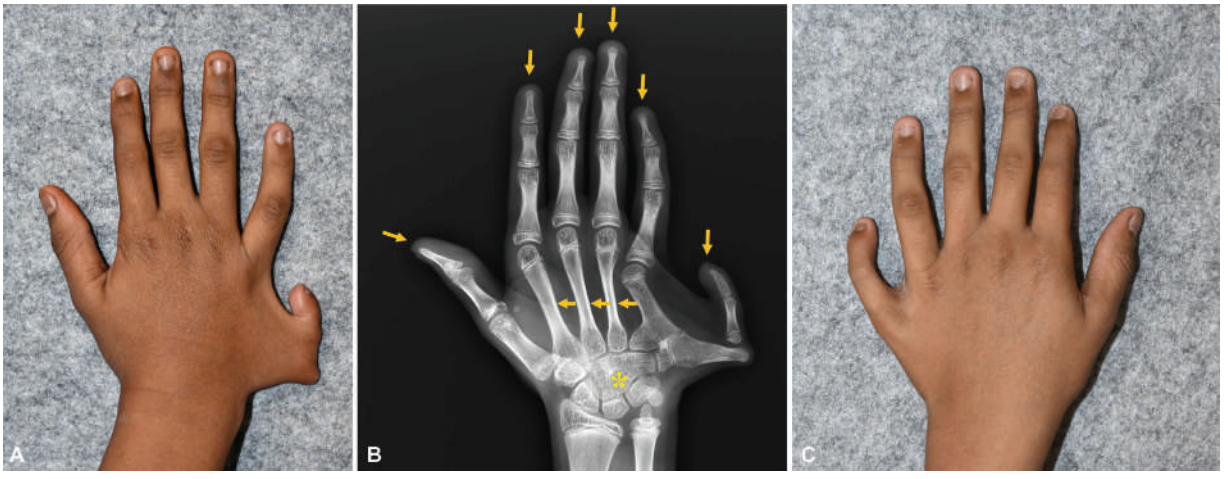

Fig. 2 Bilateral postaxial hexadactyly of the hands of case 1. (**A**) The immobile extra digit in the right hand arising from the ulnar border at right angles. (**B**) Radiograph showing metacarpal synostosis at the bases of the fifth and sixth metacarpals extending up to the mid-shaft of the fifth. The *yellow arrows* point to conical distal phalanges in all the digits and metacarpals with features of osteolysis. The *yellow asterisk* is on the capitate–hamate fusion. Dislocation of the metacarpophalangeal joint and symphalangism between the proximal and middle phalanges of the extra finger. (**C**) Left hand showing a hypoplastic short extra digit.

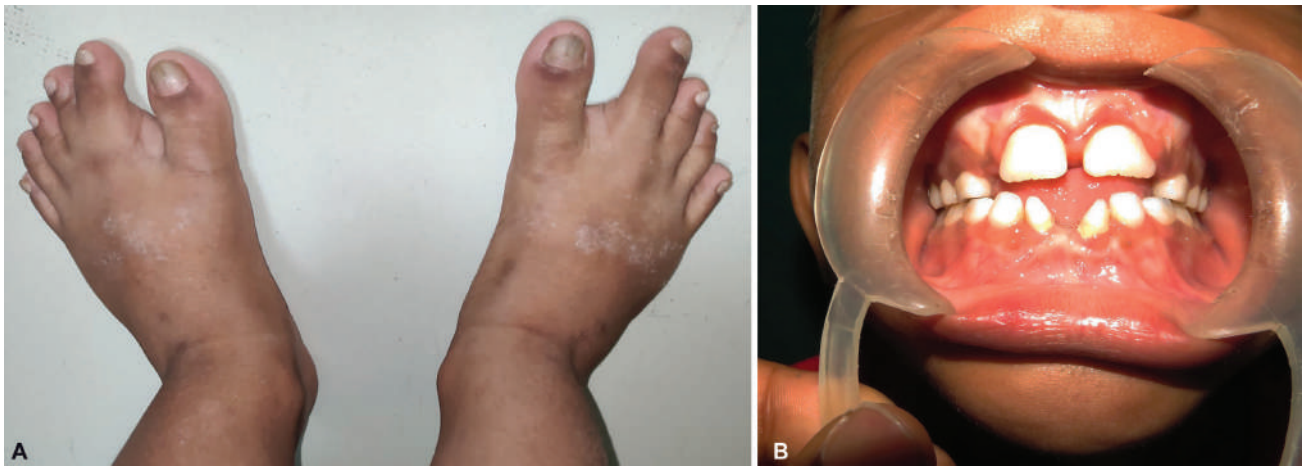

Fig. 4 Ectodermal manifestations of case 2. (**A**) Dysplastic toe nails with longitudinal ridges. (**B**) Congenitally absent upper central incisors erupted as broad central incisors (fused teeth) at around 8 years of age. Absent upper arch lateral incisors. Malformed canines. Lower arch showing peg-shaped deciduous lateral incisors and absent central incisors.

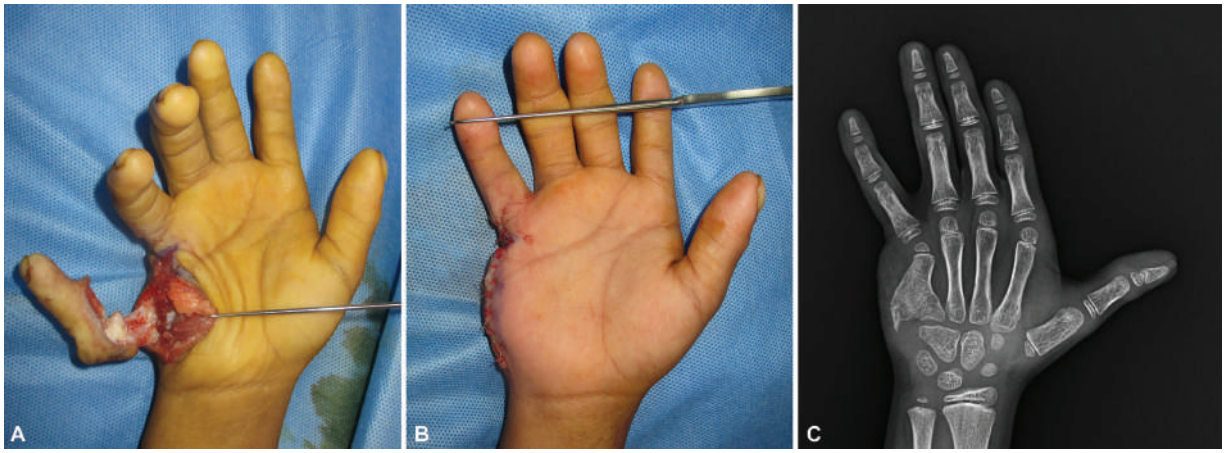

Fig. 5 Intraoperative findings. (**A**) Subperiosteal dissection and exposure of the base of the extra metacarpal. (**B**) Closure of the skin and Z-plasties. (**C**) Postoperative radiograph showing the hand after complete excision of the extra digit with its metacarpal.

pain/comfort, and happiness domains and 99 for global functioning. His mother was pleased that his handwriting improved after surgery. He had no trouble eating or reaching into his pocket. He plays football and participates in tournaments. He has no functional deficit in his left hand and is unlikely to come for surgery.

Discussion

EVC or chondroectodermal dysplasia was first described by Simon van Creveld and Richard Ellis, after they coincidentally met on a train and compiled their cases of PAP, dwarfism, and dental abnormalities.[4] Our cases had disproportionate dwarfism, postaxial hexadactyly of the hands, and genu valgum, distinguishing EVC from its allelic, Weyers acrofacial dysostosis.[5,6] Recent publications on EVC often focus on the molecular biology and report the novel genetic discoveries. A limited amount of literature on clinical hand signs or hand surgery is available. This article sheds light on the assessment and surgical intervention of EVC.

Clinical Features

Short limb disproportionate dwarfism seen in case 2 is the main chondrodysplastic feature.[5] Case 1 had a below-average height. About 58% of cases in another study had normal height.[5] Limb shortening is acromesomelic due to shorter middle and distal limb segments. PAP is typical and having more than six digits is uncommon.[5] Our cases showed a well-developed extra digit with fifth/sixth metacarpal synostosis. As shown in our radiographs, osteolysis can cause cone-shaped tubular bone epiphyses. In 2013, Al-Qattan and Al-Motairi presented their article as the only one to show this.[7] Because distal phalanges are shorter than proximal ones, a tight fist is impossible[5] (►**Fig. 6**). Our cases exhibited textbook findings like symphalangism, small broad middle phalanges, metacarpal synostosis, and capitate–hamate fusion. Nail dysplasia seen in case 2 is a distinctive feature.[5] Genu valgum, a frequent finding, occurs due to a defect in the lateral aspect of the proximal tibial epiphysis and the sloping of the lateral tibial metaphysis.[5] Acetabular

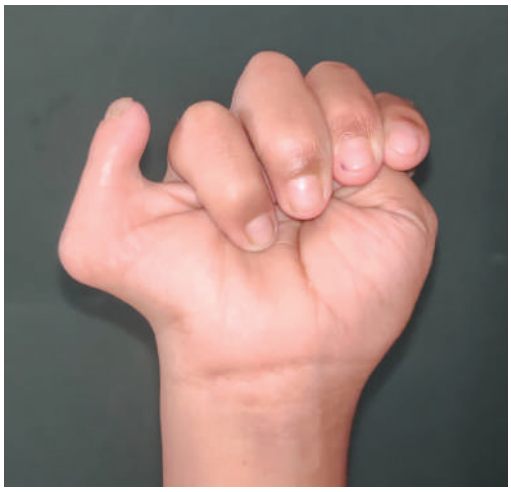

Fig. 6 Inability to make a tight fist due to disproportionate shortening of the phalanges.

osteophytes have been reported before.[5] Case 2 had tibial osteophytes.

Oral manifestations include tethering of the upper lip to gingiva, ankyloglossia, congenitally absent teeth, natal teeth, fused/supernumerary teeth, and eruption irregularities.[5] Hypodontia usually affects incisors as in case 2. Cardiac, renal, and hepatic anomalies may be present. When case 1 lacked dental anomalies, case 2 lacked cardiac malformations, a feature reportedly absent in 40% of EVC cases.[6] Close monitoring is essential as half of EVC children die in childhood from cardiac defects.[1]

Embryological Implications

Sonic hedgehog (*SHH*) secreted by the zone of polarizing activity controls the development of ulnar-sided structures: ulna, ulnar carpal bones, little finger, ring finger, and medial half of the middle finger. *EVC*/*EVC2* gene mutations with its effect on the *SHH* pathway via Gli3 protein dysfunction result in PAP.[5] Genu valgum can be explained by the *EVC* mRNA in tibial epiphyseal chondrocytes.[7] The role of *EVC* gene in preaxial duplication, conical distal phalanges, short/broad middle phalanges, and syndactyly needs further research.

Anesthetic Implications

As in case 1, cardiac involvement causes anesthetic concerns. Preoperative myocardial function evaluation is essential. Infective endocarditis prophylaxis should precede any procedure. Dental anomalies, upper lip mucobuccal folds, cleft lip/palate, thoracic deformities, and hepatic/renal defects can impact anesthesia. These children require multiple surgeries and hence repeated anesthesia. Strategic planning entails scheduling surgeries 6 months apart to minimize the effects of general anesthesia.

Surgical Implications

The extra finger in EVC is well developed (Temtamy type A) with phalangeal and metacarpal elements (Stelling type 3).[8,9] Type A PAP with metacarpal synostosis is labeled as type 3 by Pritsch et al[10] (►**Table 1**). Management of type B PAP, notably clipping or excision, is often debated, although complex varieties of type A are rarely discussed. Metacarpal synostosis and hypothenar attachments to the extra metacarpal complicate it and should be addressed. Z-plasties can restore the ulnar border through mid-lateral skin incisions. Metacarpal osteotomy is needed to maintain the contour. Metacarpal angulation can be corrected with wedge osteotomies if requested by parents.

Conclusion

It is essential to identify syndromes associated with PAP since these children need a multidisciplinary therapy. A fully developed extra digit makes removal complex. Prior to elective procedure, it is necessary to optimize the child. Additional research is necessary to analyze the etiology of the clinical manifestations.

Table 1 Classification systems used in postaxial polydactyly[8–10]

Temtamy and McKusick	Type A: Well-formed and functioning digit on ulnar side of the small finger
	Type B: Small nonfunctioning digit that may be pedunculated or a nubbin
Stelling and Turek	Type 1: Digit with soft tissue only
	Type 2: Digit with phalangeal elements
	Type 3: Digit with phalangeal and metacarpal elements
Pritsch et al (for type A only)	Type 1: A fully developed sixth ray that articulates separately with the carpals (metacarpal type)
	Type 2: The extra digit occurs on the lateral side of the fifth digit with an intercalated distal metacarpal remnant (metacarpophalangeal type)
	Type 3: The supernumerary digit arises from a hypoplastic sixth metacarpal or is fused to the fifth metacarpal (phalangeal type)
	Type 4: The extra digit originates from the fifth metacarpophalangeal joint (intercalated type)
	Type 5: The ulnar digit originates from a bifid fifth proximal phalanx (fully developed type)

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Conflict of Interest
None declared.

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