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Case Report

Paediatrics Section

Contralateral Preaxial Polydactyly and Triphalangeal Thumb with a Familial Pattern: A Case Report

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ABSTRACT

Congenital anomalies of the hand arise due to disturbances in limb development during early embryogenesis and demonstrate considerable clinical heterogeneity. Among preaxial hand anomalies, polydactyly and triphalangeal thumb are commonly encountered as isolated entities; however, the occurrence of these anomalies in contralateral hands of the same individual is rare and sparsely described. This report describes a one-year-old male child presenting with preaxial polydactyly affecting one hand and a triphalangeal thumb involving the opposite hand. The child was born at term to non-consanguineous parents following an uneventful antenatal and perinatal period. A positive family history of similar congenital hand anomalies was present. Clinical examination revealed a well-formed supernumerary digit adjacent to the thumb on one hand, while the contralateral hand showed an elongated thumb with three distinct phalanges. The remaining digits and limbs were normally developed, and no dysmorphic features were identified. Radiographic evaluation confirmed the clinical findings, demonstrating an additional preaxial digit on one side and a triphalangeal configuration of the thumb on the other. Comprehensive systemic examination, developmental assessment, and screening investigations did not reveal any associated congenital anomalies, supporting the diagnosis of an isolated familial limb defect. This asymmetric presentation highlights the wide phenotypic spectrum of preaxial hand malformations and underscores the importance of careful evaluation to exclude syndromic associations and radial ray defects. Early recognition is essential for appropriate genetic counselling, prognostication, and planning of surgical management. Documentation of rare familial combinations of congenital hand anomalies contributes to a better understanding of limb developmental disorders and their inheritance patterns.

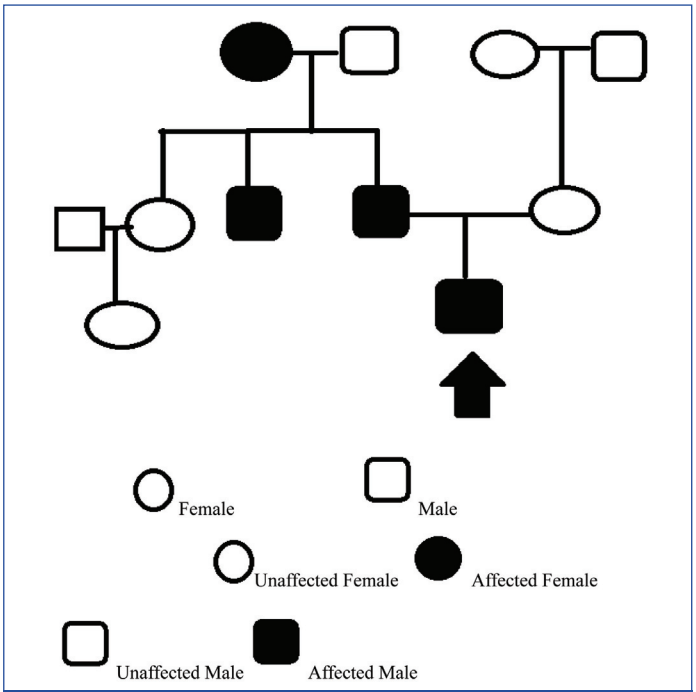
Keywords: Autosomal dominant inheritance, Congenital hand anomaly, Developmental anomaly, Elongated thumb, Radial ray defects

CASE REPORT

A one-year-old male child has been brought to the paediatric outpatient department with complaints of low-grade fever, nasal discharge, and cough for three days duration. Clinical assessment was suggestive of a viral upper respiratory tract infection. The child's vital parameters at presentation were: temperature 98.4°F, pulse rate 82/minute, respiratory rate 26/minute, and oxygen saturation on room air 98%. The child was managed with an antipyretic (paracetamol 15 mg/kg/dose) and an antihistamine (cetirizine 2.5 mg/day) for three days. There was no significant past medical history. During routine general examination, congenital abnormalities of the hands were noted incidentally.

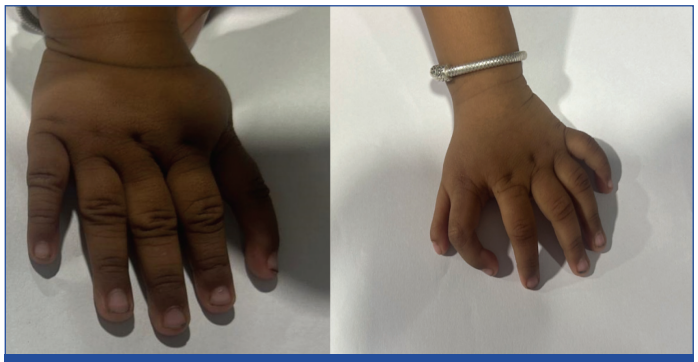
On detailed history, the child was born at term to non-consanguineous parents. Antenatal history was uneventful with no history of maternal illness, drug intake, or radiation exposure. There was no history of previous abortions or stillbirths in the past. A detailed three-generation pedigree analysis [Table/Fig-1] revealed similar congenital hand anomalies affecting the father, paternal grandmother, and paternal uncle. The clinical photographs of the affected individuals are shown in [Table/Fig-2]. All affected members in the paternal lineage were males. Female members in the family were unaffected. The mother and maternal relatives were unaffected, suggesting a familial pattern of inheritance.

Examination of the upper limbs revealed the presence of preaxial polydactyly involving the left hand, characterised by a well-developed supernumerary digit located adjacent to the thumb. Evaluation of the right hand showed an elongated thumb with abnormal configuration, raising suspicion of a triphalangeal thumb [Table/Fig-3a,b]. The remaining digits of both hands appeared normally formed. Functional assessment of the child showed adequate grasp and palmar grip. There were no functional limitations noted in the child. No abnormalities were detected in the lower limbs. The child's growth parameters were within normal limits for age, and developmental milestones were appropriate. No dysmorphic facial features or other congenital abnormalities were observed.



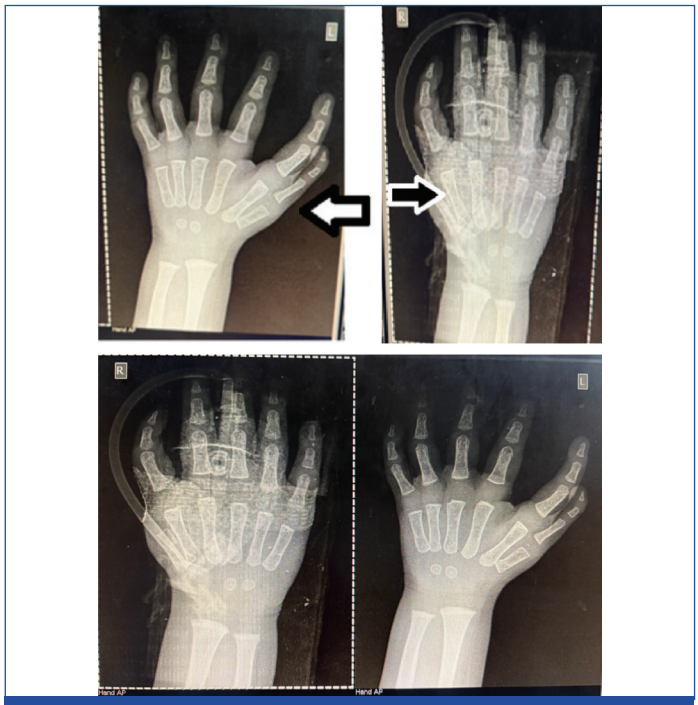
[Table/Fig-1]: This pedigree chart shows three-generation paternal transmission of preaxial hand anomaly. Square- male; Circle- female; Filled symbols- affected individuals; Unfilled symbols- unaffected individuals. The arrow mark indicates the affected child.

[Table/Fig-2]: Clinical photographs of affected family members demonstrating similar preaxial hand anomalies across generations.



[Table/Fig-3]: 3a, b. Clinical photographs showing preaxial polydactyly of the left hand and elongated thumb of the right hand, suggestive of triphalangeal thumb.

Plain radiographs of both hands were obtained to assess skeletal anatomy. The left hand radiograph confirmed preaxial polydactyly with duplication of the radial digit. The right-hand radiograph demonstrated a thumb composed of three phalanges, consistent with the diagnosis of a triphalangeal thumb. The carpal bones and remaining metacarpals were appropriate for the child's age [Table/Fig-4].



[Table/Fig-4]: Radiographs of both hands showing preaxial polydactyly on the left hand and a triphalangeal thumb on the right hand.

Ultrasonography of the abdomen was performed to screen for associated visceral anomalies and revealed no abnormalities. Cardiovascular and neurological examinations were unremarkable.

On examining the affected adults, they had a mildly weaker grip but were able to perform routine activities without major difficulties. When the affected family members were questioned about the deformity, they expressed concern mainly regarding the cosmetic appearance rather than functional limitation. Some adult relatives reported social attention and mild bullying during childhood. No genetic analysis was performed due to financial reasons; however, genetic counselling was offered to the family.

Based on the clinical findings, radiological features, and positive family history, a diagnosis of isolated familial preaxial hand anomaly was established.

DISCUSSION

Congenital anomalies of the hand result from disturbances in the tightly regulated process of limb development during early embryogenesis. Normal digit formation depends on coordinated molecular signalling along the anteroposterior axis of the limb bud, predominantly mediated by the Sonic Hedgehog (SHH) signalling pathway and its distal limb-specific enhancer, the zone of polarising activity regulatory sequence (ZRS) [1]. The ZRS element, located within intron 5 of the LMBR1 gene, plays a critical role in the spatial and temporal regulation of SHH expression. Alterations such as point mutations or duplications involving this region can lead to abnormal anterior limb bud signalling, resulting in preaxial anomalies including polydactyly and triphalangeal thumb [2,3]. Polydactyly is among the most frequently encountered congenital limb malformations and demonstrates marked phenotypic variability, ranging from minor bifurcation of a phalanx to complete duplication of a digit [4]. Triphalangeal thumb is comparatively rare, with an estimated incidence of approximately 1 in 25,000 live births, and may present either as an isolated anomaly or in association with other preaxial defects [5]. Familial aggregation of preaxial anomalies has been well documented, most commonly following an autosomal dominant inheritance pattern with variable penetrance and expressivity [3,6].

Recent pedigree-based and molecular studies have strengthened the genetic basis of combined polydactyly and triphalangeal thumb phenotypes. Duplications involving the ZRS region and copy number variations within the LMBR1 locus have been associated with such presentations [7,8]. Additionally, mutations affecting downstream components of the SHH signalling cascade, including transcription factors such as GLI3, contribute to the wide phenotypic spectrum of limb patterning disorders [9].

In the present case, the occurrence of similar hand anomalies across three generations supports a hereditary aetiology. Similar cases of combined preaxial polydactyly and triphalangeal thumb have been described in familial clusters, most commonly linked to mutations in the ZRS regulatory element. Álvarez LFG described a large autosomal dominant family with variable phenotypic expression ranging from isolated polydactyly to triphalangeal thumb [7]. Li Y et al., reported copy number variations in the LMBR1 locus associated with asymmetric limb anomalies similar to the present case [8]. The absence of associated systemic abnormalities and normal abdominal ultrasonography findings favours an isolated familial limb defect rather than a syndromic disorder.

The differential diagnosis of preaxial hand anomalies with triphalangeal thumb includes both isolated congenital variants and syndromic radial-ray malformations. Conditions that need consideration include Holt-Oram syndrome, Fanconi anaemia, VACTERL association, Townes–Brocks syndrome, and Bardet-Biedl syndrome, all of which may present with thumb anomalies or radial ray defects. These syndromic conditions are typically associated with additional systemic abnormalities such as cardiac defects, renal anomalies, vertebral

anomalies, anal malformations, haematological abnormalities, or dysmorphic facial features [4,9]. In the present case, these conditions were excluded based on normal cardiovascular evaluation, normal abdominal ultrasonography, absence of vertebral or renal anomalies, and normal developmental profile, with no dysmorphic features identified on examination.

Other non-syndromic differentials include isolated triphalangeal thumb, radial polydactyly (Wassel classification variants), and Triphalangeal Thumb–Polysyndactyly Spectrum (TPT-PS) [1,5,10]. These were distinguished on the basis of clinical asymmetry, radiographic confirmation of a well-formed extra preaxial digit in one hand and a three-phalangeal thumb in the contralateral hand, and the absence of syndactyly or additional limb anomalies. The presence of vertical familial transmission across three generations confined to the paternal lineage, with no associated systemic abnormalities, supports the diagnosis of an isolated familial preaxial hand anomaly rather than a syndromic radial ray defect [7,8].

Surgical management of preaxial polydactyly and triphalangeal thumb aims to create a single, stable, and functional thumb with an acceptable cosmetic appearance. The procedure typically involves excision of the less developed duplicated digit with reconstruction of the collateral ligaments and tendons, along with corrective osteotomy when required [10]. Triphalangeal thumb deformity is managed by excision of the accessory phalanx with shortening osteotomy and soft-tissue balancing to restore thumb opposition. The optimal timing of surgery is between six months and 18 months of age [11,12], with recent evidence suggesting improved functional and cosmetic outcomes when performed before three years of age. Early surgical intervention is associated with better joint mobility, improved grip and pinch strength, and better cosmetic outcomes.

Recent studies have further demonstrated genotype-phenotype correlations in LMBR1-associated preaxial anomalies and ZRS regulatory mutations, highlighting variable expressivity and familial clustering in such cases [13-15]. Similar contralateral presentations of preaxial polydactyly and triphalangeal thumb have also been rarely reported in the literature [16,17].

CONCLUSION(S)

Contralateral occurrence of preaxial polydactyly and triphalangeal thumb is an uncommon presentation. Detailed clinical examination combined with careful family history assessment is essential to distinguish isolated familial limb anomalies from syndromic conditions. Early identification allows appropriate counselling, avoids unnecessary investigations, and facilitates timely surgical planning.

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