

Case Report

Isolated polydactyly of hands and feet in neonates: a case report

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ABSTRACT

Polydactyly is a common congenital limb anomaly characterized by the presence of extra digits on the hands, feet, or both. When it occurs without any associated syndromic or systemic abnormalities, it is referred to as isolated polydactyly. In neonates, this condition presents with a wide spectrum of manifestations, ranging from small soft-tissue projections to fully developed digits containing bones and joints, and it may occur unilaterally or bilaterally. Classification is typically based on the anatomical location of the extra digit—preaxial (thumb or big toe side), postaxial (little finger or little toe side), or central—with postaxial polydactyly being the most frequently observed type, particularly among certain ethnic populations. Isolated polydactyly commonly follows an autosomal dominant pattern of inheritance with variable expression, although sporadic cases are also frequently reported. Accurate and early diagnosis is critical to differentiate isolated polydactyly from syndromic forms, as this distinction plays a key role in determining clinical management strategies and providing appropriate genetic counselling to families. In cases involving non-functional or minimally developed extra digits, intervention may not be necessary; however, surgical removal is often considered during infancy or early childhood for functional or cosmetic reasons, especially when the anomaly may interfere with hand or foot function or affect the child's psychosocial development. This case emphasizes the clinical importance of recognizing isolated polydactyly in the neonatal period and underscores the need for thorough clinical evaluation, precise classification, and a coordinated, multidisciplinary management approach involving pediatricians, geneticists, and surgeons. Early intervention and counselling are essential for addressing parental concerns, guiding developmental support, and ultimately enhancing the quality of life and long-term functional outcomes for affected children.

Keywords: Polydactyly, Congenital limb anomalies, Neonatal limb anomaly, Postaxial

INTRODUCTION

Polydactyly is a frequently encountered congenital anomaly defined by the presence of extra digits on the hands, feet, or both, and when it occurs without accompanying syndromic or systemic abnormalities, it is termed isolated polydactyly. It represents one of the most common limb malformations observed in neonates, with clinical presentations that vary significantly depending on genetic, ethnic, and familial influences.¹ The condition may involve fully formed or rudimentary digits and can appear unilaterally or bilaterally. Early recognition and accurate classification are crucial for guiding management

decisions, which may include surgical correction in cases where function or appearance is compromised. While many instances of isolated polydactyly follow an autosomal dominant inheritance pattern, sporadic cases are also common.²

The objective of this work is to present a neonatal case of isolated postaxial polydactyly, highlighting the importance of early diagnosis, proper classification, and multidisciplinary involvement in clinical decision-making. This report aims to contribute to the understanding of isolated polydactyly and support best practices in neonatal care and parental counselling.

CASE REPORT

A full-term male neonate, born at 39 weeks of gestation via spontaneous vaginal delivery with a birth weight of 3.1 kg, with appearance, pulse, grimace, activity, and respiration (APGAR) score of 8 and 9 at 1 and 5 min respectively, was noted at birth to have bilateral postaxial polydactyly of both hands and feet (Figures 1 and 2). All extra digits were soft-tissue in nature with fully formed nail beds and were soft tissue in consistency. There were no other visible anomalies and neurological, cardiovascular, and systemic examinations were normal. Feeding and sucking was normal.



Figure 1: Bilateral postaxial polydactyly of both hands.



Figure 2: Bilateral postaxial polydactyly of both feet.

Antenatal history

23 years old primigravida, booked case, immunized, Hb-12.8 g/dl, blood group-O positive, no h/o PIH, gestational diabetes mellitus (GDM), hypothyroid, no h/o drug intake, radiation exposure, infections, urinary tract infection. Prenatal imaging at 20 weeks had identified a possible limb abnormality but lacked follow-up.

Family history was positive for similar complaints of polydactyly of hands and feet for mother (Figures 3 and 4),

however, there were no other congenital anomalies in three-generation pedigree and there was no consanguinity.

On physical examination, vitals were stable, appropriate for the gestational age, no dysmorphic facies, and no syndactyly or limb asymmetry. Cardiovascular, respiratory, abdominal and neurological examinations were unremarkable; no remarkable skin findings were noted.



Figure 3: Polydactyly of hands of mother.



Figure 4: Polydactyly of feet of mother.

Radiographs were taken in order to assess the bone involvement, which confirmed soft tissue duplication without bony involvement in all four limbs.

Genetic testing, including chromosomal microarray, was found to be normal and targeted gene panel for limb development genes, negative for known pathogenic variants (GLI3, SHH, ZRS). Whole exome sequencing was not performed due to isolated presentation and normal development.

Other investigations like echocardiogram were employed to rule out congenital heart disease and abdominal ultrasound for renal anomalies in view of syndromic association, were found to be normal and newborn metabolic screening also were found to be normal.

Surgical excision of extra digits is planned at 12 months of age with realignment and repair of ligaments as crucial for achieving stability of digits with no residual deformity, patient will be followed up for any contracture and deformities for ensuring good functioning, along with quality of life (QoL) and genetic counselling in order to plan for further pregnancies.

DISCUSSION

Our patient presented with isolated bilateral postaxial polydactyly involving both hands and feet, with no associated dysmorphic features or internal anomalies. This presentation is consistent with previously reported non-syndromic familial cases, such as those by Ullah et al, who described similar multi-limb involvement in the absence of systemic features.³ In contrast, syndromic forms of polydactyly, like those associated with Ellis-van Creveld syndrome or Bardet-Biedl syndrome, typically present with additional anomalies including congenital heart disease or renal dysfunction.⁴

The patient's positive family history and ethnic background suggested a possible autosomal dominant inheritance pattern, aligning with the observations made by Fatima et al, who reported a series of siblings with similar inheritance in non-syndromic postaxial polydactyly.⁵ However, unlike cases linked to syndromic causes such as Greig cephalopolysyndactyly syndrome or Trisomy 13, our patient tested negative on chromosomal microarray (CMA), supporting an isolated diagnosis.⁶

A comprehensive genetic evaluation was undertaken. CMA showed no abnormalities, ruling out microdeletions and duplications typically seen in syndromic cases.⁴ Targeted gene testing for common mutations (e.g., *GLI3*, *ZRS*, *SHH* pathway) was also negative, similar to the diagnostic approach followed by Sun et al.⁷ While whole exome sequencing can identify novel or rare gene variants associated with limb anomalies, it may be reserved for complex or syndromic cases.⁸

The timing of surgical intervention plays a critical role in optimizing hand function and cosmetic appearance. In this case, surgery was scheduled around 12 months of age, in alignment with recommendations by Goldfarb et al, who advocate individualized timing based on digit function and complexity.⁹ Early excision is preferred when digits lack bony involvement, especially in soft-tissue nubbins, to minimize complications and improve surgical outcomes.¹⁰

Genetic counselling was also provided to inform the family about recurrence risks and prenatal testing options.¹¹ This is particularly relevant in populations with a higher prevalence of consanguinity or familial transmission.¹² In such contexts, pedigree analysis and risk assessment are critical components of preconception and antenatal care.

Additionally, detailed physical examination and screening for internal organ anomalies (cardiac, renal, neurological) are essential to distinguish isolated polydactyly from syndromic forms.¹³ This comprehensive approach ensures appropriate resource utilization and avoids unnecessary anxiety or interventions in families of otherwise healthy children.

Finally, although isolated polydactyly is often benign, its psychosocial impact should not be underestimated. Parents often express concerns regarding aesthetics, social stigma, or future functionality. Surgical correction generally yields excellent cosmetic and functional results, particularly in cases without osseous involvement.¹⁴

CONCLUSION

This case of isolated bilateral postaxial polydactyly involving all four limbs in a neonate, with a positive family history and absence of syndromic features or genetic abnormalities, highlights the importance of thorough clinical evaluation and tailored genetic testing in the assessment of congenital limb anomalies. By ruling out syndromic associations through detailed physical examination, imaging, and targeted genetic analysis, this case reinforces the distinction between non-syndromic familial polydactyly and more complex genetic syndromes. The planned surgical intervention and genetic counselling underscore the multidisciplinary approach required to ensure optimal functional and aesthetic outcomes, as well as informed reproductive planning. This report contributes to existing literature by documenting a rare presentation of isolated, multi-limb soft-tissue polydactyly with a likely autosomal dominant inheritance, thereby enhancing our understanding of the phenotypic spectrum and management strategies for non-syndromic digital anomalies in neonates.

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