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Karnataka Paediatric Journal

Letter to the Editor

Isolated macrodactyly in an infant

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Received: 24 August 2025
Accepted: 28 September 2025
EPub Ahead of Print: 19 November 2025
Published: 30 December 2025

DOI
10.25259/KPJ_60_2025

Quick Response Code:

Dear Editor,

Macrodactyly is an uncommon congenital condition characterised by abnormal enlargement of one or more digits. It may present as an isolated anomaly or in association with syndromes that involve other congenital malformations. The estimated incidence ranges from 1 in 50,000 to 1 in 100,000 live births, with variability noted across ethnic and regional populations.[1-3] Beyond the visible deformity, cultural stigma and social perceptions often contribute to significant psychological challenges, some of which may persist into adulthood. Due to its rarity and wide spectrum of clinical presentations, universally accepted treatment guidelines are lacking, making surgical management particularly complex.[4] Current therapeutic options include soft-tissue debulking, epiphysiodesis (growth plate arrest) or, in selected cases, amputation, aimed at achieving acceptable cosmetic and functional outcomes. However, the need for newer and more refined approaches remains.

A 1-year-old girl was brought to the outpatient department by her parents with complaints of an enlarged second digit of the right foot present since birth. According to the father, the digit appeared larger and longer than the others at birth and continued to grow disproportionately thereafter. The parents denied any history of congenital anomalies, limb malformations, overgrowth syndromes, or similar conditions in the family. On general physical examination, the child was healthy, well-nourished and developmentally appropriate for age. A head-to-toe examination revealed frontal bossing and prominent Mongolian spots on the back. No other dysmorphic features were present. Systemic examination was unremarkable, and no additional defects were identified. Digits of both upper limbs and the left foot were normal. Local examination of the right foot revealed diffuse hypertrophy of the second digit with a slight curvature [Figure 1]. The distal phalanx could be hyperextended abnormally without pain. No syndactyly was observed. The remaining toes of the right foot were normal in size, shape, and function. Radiological findings confirmed disproportionate phalangeal and metatarsal enlargement, demonstrating enlarged bony structures with overgrowth, which will guide surgical planning, such as considering ray resection or amputation [Figure 2].

There were no features of associated syndromes such as neurofibromatosis, Sotos syndrome or other overgrowth disorders. Based on these findings, a diagnosis of isolated macrodactyly was made.

In addition to clinical management, it is important to recognise the psychosocial aspects associated with isolated macrodactyly. Parents often experience significant anxiety regarding the child's appearance, possible functional limitations and social acceptance. Concerns about stigma and peer interactions may emerge as the child grows, underscoring the need for early counselling and long-term psychological support. Multidisciplinary care, including access to counselling

Figure 1: Clinical image of right foot revealing diffuse hypertrophy of the second digit with a slight curvature.

Figure 2: Radiograph of second toe showing enlarged bony structures, with disproportionate overgrowth of the phalanges and metatarsals.

services and parent support groups, can help families cope better with these challenges and improve overall quality of life.

Following a multidisciplinary consultation with the surgical teams, it was decided to defer surgery until after puberty due to the child's current ability to tolerate footwear, the potential for further skeletal growth, the risk of multiple repeat procedures if operated on too early and the psychosocial concerns associated with repeated surgical interventions in infancy.

The primary objective was to create a painless, functional and cosmetically acceptable digit that can be accommodated by footwear. Alternatively, surgery may be deferred until skeletal maturity.[2,3] The choice of treatment is guided by both functional and psychosocial considerations, often influenced by parental perceptions of the condition's impact on the child. The child has been scheduled for regular follow-up every 6 months, which will include assessment of foot growth, functional evaluation with particular attention to gait and footwear needs and periodic radiological monitoring. Counselling of the parents forms an integral part of each visit to address concerns related to progression and long-term outcomes. The expected clinical course is a gradual enlargement of the affected digit, which typically continues until skeletal maturity.

Early diagnosis and individualised treatment planning are crucial for addressing both functional limitations and cosmetic concerns, thereby improving long-term outcomes.

Ethical approval: Institutional Review Board approval is not required.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship: Nil.

Conflicts of interest: There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation: The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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How to cite this article: Jayachandra B, Arun Babu T. Isolated macrodactyly in an infant. Karnataka Paediatr J. 2025;40:258-9. doi: 10.25259/KPJ_60_2025