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Research Article

The Cost of Curiosity: A tale of too much testing in a case of Isolated Macro-syndactyly

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Abstract

Isolated macro-syndactyly is a rare congenital anomaly characterized by enlarged, fused digits, which may exist independently or as part of genetic syndromes. We report the case of a 4-year-old female presenting with congenital enlargement and fusion of the right third and fourth digits. The child was developmentally normal, with no systemic abnormalities or functional impairment, and the parental concern was primarily cosmetic. Despite low syndromic suspicion, the family pursued extensive genetic testing, including chromosomal microarray and sequencing for overgrowth-related genes, which were negative. Radiographs confirmed soft tissue and bony fusion without additional skeletal anomalies. Conservative management with counseling was advised, reserving surgical options for future cosmetic or functional needs. This case highlights the importance of thorough clinical evaluation in distinguishing isolated cases from syndromic associations. It also underscores the potential pitfalls of unnecessary exhaustive testing, which may increase cost and anxiety without influencing management in such localized anomalies.

(A) (B)

Figure 1: A) Enlarged and fused, broad, bulky fingers. B) X-Ray showing soft tissue fusion.

Case description:

A 4-year-old female child presented to our pediatric outpatient department for a routine check-up. She was developmentally normal with no significant medical history or family history of congenital anomalies. The parents had no concerns except for a cosmetic deformity of the right hand noted since birth.

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On examination, isolated macro-syndactyly was seen involving the right third and fourth digits. The affected digits were enlarged and fused, resulting in broad, bulky fingers (Figure 1A). There was no limitation of movement, no tenderness, and no skin changes. The other hand and limbs were normal, and no dysmorphic features or systemic abnormalities were identified.

A detailed history confirmed no functional disability, and the primary concern was cosmetic. Given the isolated anomaly, the clinical suspicion for syndromic association was low. Nonetheless, the family had pursued extensive genetic testing externally, including chromosomal microarray and sequencing for overgrowth syndrome-related genes (e.g., PIK3CA and AKT1). The results were negative for pathogenic mutations.

Radiographs of the affected hand confirmed soft tissue and bony fusion of the involved digits (Figure 1B), without other skeletal abnormalities. Systemic evaluation including abdominal ultrasound and cardiac screening was normal.

Considering the lack of functional impairment and absence of syndromic features, a decision for conservative management was made. The family was counselled on the benign nature of the condition and on possible future surgical options for cosmetic or functional improvement if desired.

Discussion

Macro-syndactyly consists of enlarged, fused digits and is a rare congenital hand anomaly (1). It may present in isolation or as part of genetic syndromes such as Proteus syndrome or PIK3CA-related overgrowth spectrum (2). Our patient's isolated macro-syndactyly with no systemic involvement and normal developmental milestones suggests a localized congenital anomaly. Careful clinical examination and history remain crucial in differentiating isolated cases from syndromic presentations. While genetic testing being easily available in modern era, and is valuable in suspected syndromic or progressive cases, however in isolated macro-syndactyly with minimal symptoms, exhaustive genetic workup may not impact management and can add cost and anxiety (3). The external genetic workup in this case was extensive but did not reveal any abnormalities, affirming the diagnosis of isolated macro-syndactyly. Radiological imaging with plain X-rays remains essential to assess bony involvement and assist surgical planning. Surgical intervention is generally reserved for significant functional impairment, pain, or patient preference for improved cosmesis. Risks of surgery include scarring, contractures, and recurrence, so timing and extent of surgery must be individualized (4).

This case emphasizes avoiding "exaggerated workup" and highlights the importance of clinical judgment to guide appropriate and cost-effective evaluation. Close clinical monitoring and family education are key elements of management.

Conclusion

Isolated macro-syndactyly is a rare congenital anomaly often presenting with cosmetic concerns and minimal functional disability. A focused clinical evaluation supplemented by basic imaging is sufficient for diagnosis in most cases. Extensive genetic testing can be deferred or selectively employed based on clinical suspicion. Management should be individualized, prioritizing function, cosmetic concerns, and patient-family preferences. This case illustrates that unnecessary extensive workups can be avoided with careful clinical assessment, reducing burden and anxiety while ensuring optimal care.

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