Journal of Endocrinology, Metabolism and Diabetes of South Africa 2025;30(3):134-135
https://doi.org/10.1080/16089677.2025.3777

JEMDSA
ISSN 1608-9677 EISSN 2220-1009

CLINICAL SIGNS

Madelung deformity

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Introduction

Madelung deformity represents an uncommon skeletal anomaly that primarily affects the distal portions of the radius and ulna bones. Originally documented by Otto Wilhelm Madelung in 1878, this condition was traditionally viewed as an isolated orthopaedic disorder. However, modern clinical understanding has evolved to recognise its frequent occurrence in conjunction with various genetic disorders, particularly Turner syndrome and Léri-Weill dyschondrosteosis (LWD). The presence of this deformity has emerged as a significant clinical indicator that may warrant comprehensive genetic testing and endocrinological evaluation.[1]

Pathophysiology

The short stature homeobox-containing (SHOX) gene is located in the pseudo autosomal region of the distal Xp and Yp. Mutations or deletions of SHOX are associated with syndromes of poor growth and skeletal dysplasia, including Léri-Weill dyschondrosteosis (LWD), Turner syndrome (TS), and Langer mesomelic dysplasia (LMD). Homozygous gene defects cause LWD, whereas TS and LMD are caused by haploinsufficiency.[2]

Clinical manifestations

Patients present with varying degrees of pain, reduced grip strength, and the classic "bayonet" deformity. Madelung's deformity presents with characteristic radiographic findings, including radial shortening with accompanying curvature, widening of the distal radioulnar joint, and triangular configuration of the carpal bones. The predominant clinical manifestation is a proximal-volar subluxation of the wrist, creating a distinctive step-off deformity where the hand and carpus appear to be displaced from the forearm in a stepwise configuration.[3] Patients also present with volar and ulnar subluxation of the hand and decreased range of motion in the wrist, commonly with supination difficulty. The presumed cause is an arrested development of the palmar-ulnar side of the distal radius.[4]

Conclusion

Madelung deformity is more than just a skeletal abnormality; it serves as an important clinical sign indicating underlying genetic issues, especially SHOX gene haploinsufficiency. In the context of TS, recognising this deformity highlights the multisystem nature of the condition and helps guide proper management strategies. For clinicians, understanding the importance of this skeletal sign improves patient care and genetic counselling. Early recognition and appropriate multidisciplinary management can optimise functional outcomes and quality of life for affected patients, making Madelung deformity an important clinical sign worthy of systematic evaluation in patients with TS and related genetic conditions.

Bilateral Madelung deformities in a 24-year-old patient with Turner's Syndrome.

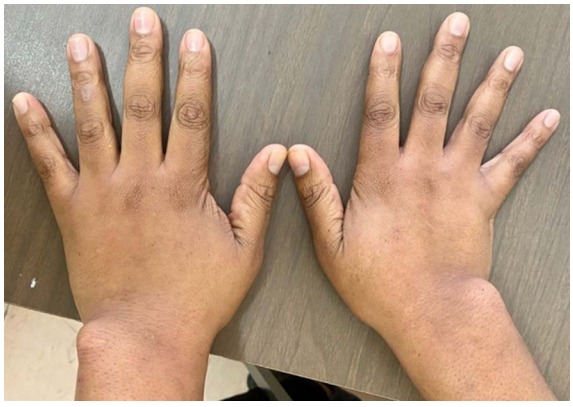

Image A (left): shows distal ulnar dorsal prominence in the prone position.

Image B (right): shows a lateral view of the ulnar anomalies and palmar wrist displacement.

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