

Short Fourth Metacarpal Reaffirms Turner's Syndrome: A Case Report

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ABSTRACT

Turner's syndrome (TS) is a hereditary condition that primarily impacts female infants, characterized by ovarian dysgenesis, short stature, webbed neck, and a variety of phenotypic abnormalities. Among the clinical features, the metacarpal sign has emerged as a notable diagnostic marker in TS. This sign refers to the shortened fourth metacarpal bone, which can be identified through radiographic imaging. The presence of this anatomical variation, particularly when combined with other physical manifestations of TS, aids clinicians in making an accurate diagnosis. This article highlights the significance of the metacarpal sign as a diagnostic tool in the clinical assessment of TS, emphasizing the importance of thorough physical examination and radiological evaluation in patients suspected of having this condition.

Keywords: Amenorrhea, Case report, Short fourth metacarpals, Turner syndrome, X chromosome.

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INTRODUCTION

Turner syndrome (TS) occurs when an X chromosome is absent, either totally or partially, with an estimated incidence of one in 2,000–2,500 live births of females.¹ While it presents with a constellation of clinical features, the diagnosis often relies on cytogenetic analysis to confirm the structural abnormalities or absence of the X chromosome. Among the hallmark physical characteristics observed in patients with TS is short stature, with adult heights typically falling below the third percentile for age and gender. This short stature stems from multiple factors, including growth hormone deficiency, ovarian failure, and skeletal abnormalities. Among these skeletal anomalies, attention has recently been drawn to the presence of a distinct radiographic finding, "the metacarpal sign" also known as "positive Archibald's sign." This sign involves a line drawn tangent to the heads of the 4th and 5th metacarpals (bones in the hand) that intersects the 3rd metacarpal, indicating the shortening of the 4th metacarpal.² A short fourth metacarpal is also associated with pseudohypoparathyroidism (PH), pseudopseudohypoparathyroidism (PPH), hereditary multiple exostoses, homocystinuria, basal cell nevus syndrome and, rarely, due to infection, an idiopathic cause.³ Here we present a case of a young girl who presented with primary amenorrhea and was found to have a short 4th metacarpal. On further evaluation and investigation, she was diagnosed with TS.

CASE REPORT

A 22-year-old female presented with primary amenorrhea in our outpatient department. On physical examination, we noted a shortened fourth metacarpal in both hands, resulting in dimpling over the knuckles when the hands are clenched into a fist, hypertelorism, widely spaced nipples, an underdeveloped mammary gland (Tanner stage II), downy pubic hair (Tanner stage II), absent axillary hair, and short stature (136 cm height), with normal heart sounds and mental status (Fig. 1). Both hearing and speech were normal. Her kidney function tests, liver function tests, and thyroid profile were normal. Parathyroid hormone (PTH), phosphate, and ionized calcium levels were within normal ranges. Serum LH

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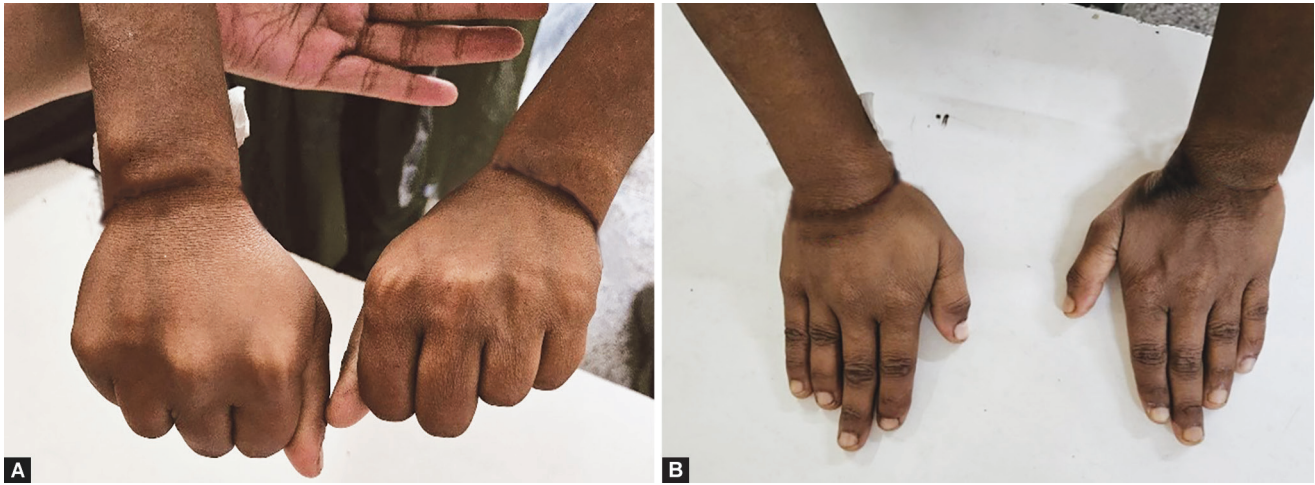
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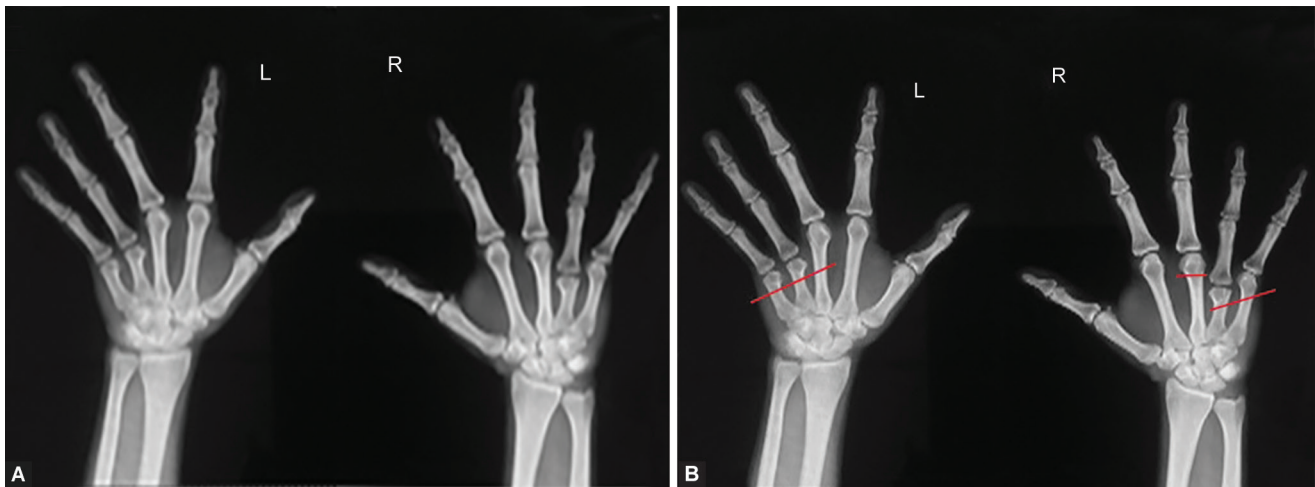
was elevated at 160 (5–25) IU/L and follicle-stimulating hormone (FSH) was 28.6 (4.7–21.5) IU/L. As viewed on the X-ray of both hands, the short fourth metacarpal and distortion of the tangential line joining the 3rd and 5th metacarpals (X-ray right hand) (positive Archibald's sign/metacarpal sign) (Fig. 2). On ultrasonography, the uterus and ovaries were absent. The diagnosis was further confirmed by chromosomal analysis, which showed the presence of a 45, XO karyotype.

DISCUSSION

Overall, the metacarpal sign is positive in 33.8% of people with TS.⁴ A short fourth metacarpal may be a normal finding in 9.6% of individuals older than 6 years.⁵ Seventy-five percent of people with PH and ninety percent of patients with PPH may have it.⁶ Additionally, it has been documented in cases of homocystinuria and primary hypoparathyroidism.^{7–9} Post-infective, post-traumatic, and idiopathic are some additional frequent causes. Rare causes include juvenile idiopathic arthritis, inherited multiple exostosis syndrome, Langer-Giedion syndrome, and



Figs 1A and B: (A) Clinical photograph of both hands displaying dimpling of the fourth knuckle due to the short fourth metacarpal (Archibald's sign); (B) Clinical photograph depicting bilateral shortening of the fourth finger due to a shortened fourth metacarpal



Figs 2A and B: (A) X-ray of both hands (anteroposterior view) depicting a short fourth metacarpal; (B) Metacarpal sign/positive Archibald's sign and distortion of the tangential line joining the third and fifth metacarpals [R]

basal cell nevus syndrome. In our case, we have eliminated other causes through a comprehensive history, physical examination, and investigations. The metacarpal sign in TS refers to a characteristic finding related to the hands and fingers in individuals with this genetic condition. In TS, the metacarpals (bones in the palm) tend to be shorter than normal, contributing to a specific hand morphology. This sign is part of the broader physical features associated with TS, which include a webbed neck, hearing loss, short stature, infertility, low-set ears, and heart and kidney defects. The metacarpal sign is often identified through a physical examination, where the healthcare provider assesses the ratio between the metacarpal bones and the overall length of the hand. While the metacarpal sign is not the sole diagnostic criterion for TS, it adds to the clinical picture and aids in the comprehensive evaluation of individuals. Early detection of TS is crucial for timely intervention and management, including hormonal therapy to

address growth and development issues. As with many aspects of TS, a multidisciplinary approach involving medical, psychological, and social support is essential for optimal care and the well-being of individuals with this condition.

This report is limited by its single-case design, which restricts the generalizability of the findings. Furthermore, the metacarpal sign can be seen in other clinical conditions and is not exclusive to Turner syndrome. To further prove the diagnostic importance of short fourth metacarpal as a clinical marker of Turner syndrome, more research with bigger patient cohorts is needed.

CONCLUSION

Early recognition of TS is crucial for timely intervention and management to address associated health concerns and optimize patient outcomes.

DECLARATIONS

Patient Consent for Publication

Written informed consent was obtained from the patient for publication of clinical details, radiological findings, and images.

Ethics Waiver/Approval

Ethical approval was waived as this is a single case report in accordance with institutional guidelines and does not require formal ethics committee approval.

AI Use Declaration Statement

No AI tools were used in the preparation of this manuscript.

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AUTHORS' CONTRIBUTIONS (CREDIT FORMAT)

- Patient management: Prabhat Agrawal, Nirmal Kumari, Ranjeet Singh, Priyanka Joshi
- Data collection: Prabhat Agrawal, Nirmal Kumari, Ranjeet Singh, Priyanka Joshi, Ruchika Garg
- Writing – Original draft preparation: Prabhat Agrawal, Nirmal Kumari
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