International Journal of Contemporary Pediatrics
Pillai S et al. Int J Contemp Pediatr. 2025 Sep;12(9):1585-1587
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20252617

Case Report

Iso-Kikuchi syndrome: a rare non-hereditary congenital onycho-dysplasia in a new born

Sharadraj Pillai, Anuragsingh Chandel*, Shubham Kamble, Varsha Chauhan

Department of Pediatrics, MGIMS, Sewagram, Maharashtra, India

Received: 22 July 2025
Accepted: 12 August 2025

***Correspondence:**
Dr. Anuragsingh Chandel,
E-mail: anuragchandel@mgims.ac.in

ABSTRACT

Iso Kikuchi syndrome or onychodysplasia is a rare congenital condition characterized by total anonychia or dysplasia of the nails of the index fingers, with underlying bony abnormalities. It is usually benign and rarely associated with other conditions. Despite various hypotheses regarding the pathophysiology, the exact etiology remains unknown. We report a case of a full-term male neonate who presented at birth with bilateral anonychia of the index fingers and dysplastic radial half of the nails on the middle fingers. X rays of the hands showed Y-shaped bifid distal phalanges of the index fingers. Systemic and radiological evaluations were otherwise normal. This case highlights the importance of early recognition of this condition to avoid unnecessary diagnostic work up.

Keywords: Iso Kikuchi syndrome, Anonychia, Bifid phalanx, Congenital anomaly, Onychodysplasia

INTRODUCTION

Congenital onychodysplasia (CO) is a rare, isolated, benign disorder characterized by complete anonychia or dysplastic changes of the nails. First described by Iso in 1969 and later by Kikuchi in 1974, it is a clinical syndrome involving dysplasia or complete absence of fingernails with underlying bony abnormalities.[1,3] It is mostly sporadic, and the complete pathophysiology remains poorly understood. Clinical criteria for diagnosis include unilateral or bilateral hypoplasia of the index fingernails and/or other fingers including toenails, radiographic abnormalities of the distal phalanx of the affected fingers and congenital occurrence, which may be sporadic or hereditary.[7,8] Although isolated CO is typically associated with normal neurodevelopment, clinical follow up is necessary to exclude other multisystem pathologies in some cases.[2]

CASE REPORT

Authors report the case of a full-term male newborn, second of twins, delivered by cesarean section to a non-consanguineous couple with an uneventful antenatal and perinatal course. There was no history of teratogenic drug exposure during pregnancy. On examination, the child had mild tachypnea with grunting. Additionally, there was complete anonychia of both index fingers and dysplasia of the radial half of the nails on both middle fingers.

Systemic examination was unremarkable. Lateral X rays of both hands showed Y-shaped bifurcation of the distal phalanges of the index fingers. Other laboratory investigations were within normal limits. Abdominal ultrasonography was normal. A diagnosis of congenital onychodysplasia was made. The infant was exclusively breastfed and discharged on day 8 of life. Follow ups during vaccination visits revealed normal growth and age-appropriate developmental milestones (Figure 1 and 2).

DISCUSSION

Iso Kikuchi syndrome is a rare, benign, non-syndromic condition characterized by abnormalities of the nails of the index fingers. The clinical presentation is variable,

ranging from complete absence of the nail (anonychia) to non-specific nail dysplasia. The exact etiology is unknown, but a commonly proposed hypothesis is a focal developmental disturbance, possibly due to transient ischemia affecting the radial side of the distal phalanx during intrauterine life. This may interfere with the development of the nail matrix and, in some cases, the underlying bone.[7] Most cases are sporadic, although occasional reports suggest autosomal dominant inheritance with variable expression.[8] It is extremely rare, with an estimated incidence of approximately 4.2 per 100,000 live births, affecting both sexes equally with no ethnic predilection.[5,9]

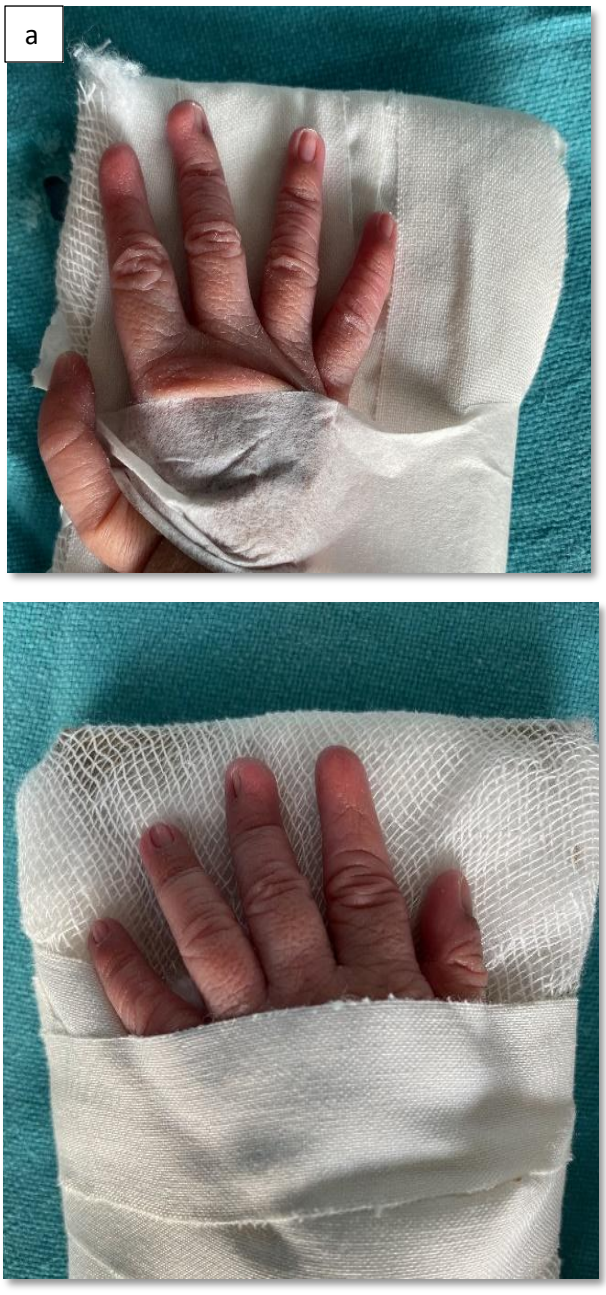

Figure 1: (a, b) Total anonychia of index fingers and dysplasia of radial half of middle finger nails in both hands.

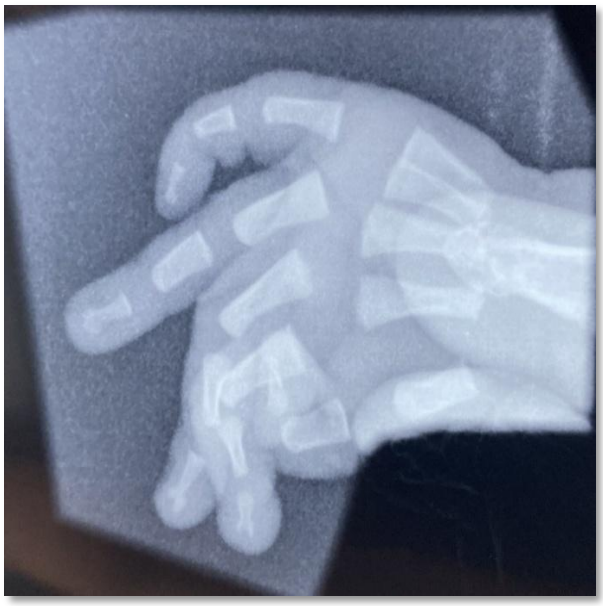

Figure 2: Plain radiograph of right-hand showing Y-shaped bifurcation of distal phalanges of index and middle fingers.

Tirelli et al and Valerio et al reported cases limited to nail involvement without skeletal changes.[1,2] In contrast, our case presented with bilateral anonychia of the index fingers and dysplasia of the radial halves of the middle fingernails. Y-shaped bifurcation of the distal phalanx on X-ray, a hallmark radiological finding, was present in our case as well.[7] In a case reported by Padmavathy et al there was significant hypoplasia of the distal phalanx, whereas our patient showed milder skeletal findings and no systemic anomalies.[3] The child showed normal development during follow up, further affirming the benign nature of the condition.

Although congenital onychodysplasia of the index finger (COIF) usually involves only the index fingers, reports exist of involvement of other digits. Our patient had additional middle finger nail dysplasia. Krishnegowda et al described a similar case with associated brachydactyly and azure lunula, which were not seen in our patient.[4] Jindal et al also reported bilateral index finger anonychia with bony hypoplasia, closely resembling our findings.[9] Imaging is crucial for diagnosis, especially in cases with subtle or atypical nail changes. The bifid or Y-shaped distal phalanx on X ray is a key diagnostic feature, differentiating this entity from other causes of congenital nail abnormalities, such as ectodermal dysplasia or amniotic band sequence.[7,10]

CONCLUSION

This case highlights the rare entity of congenital onychodysplasia. It is a benign condition with excellent prognosis. Early recognition is important to avoid extensive investigations and unnecessary interventions.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Tirelli LL, Luna PC, Cano R, Giraldo JP, Larralde M. Iso-Kikuchi Syndrome: Report of 3 Pediatric Cases. Actas Dermosifiliogr (Engl Ed). 2018;109(5):33–6.
2. Valerio E, Favot F, Mattei I, Cutrone M. Congenital isolated Iso-Kikuchi syndrome in a newborn. Clin Case Rep. 2015;3(10):866–9.
3. Padmavathy L, Rao L, Ethirajan N, Kanthimathi R, Adaikappan M. Iso-Kikuchi syndrome with absence of ring fingers and metacarpal bone abnormality. Indian J Dermatol Venereol Leprol. 2008;74(5):513–5.
4. Krishnegowda SY, Kumar NS, Vasudevan P. Iso-Kikuchi syndrome in a newborn associated with azure lunula and brachydactyly. Indian J Paediatr Dermatol. 2015;16(3):168–9.
5. Di Chiacchio N, Jasso-Olivares JC, Di Chiacchio NG, Jacinto JA, Restrepo MVS. Syndrome in question. An Bras Dermatol. 2015;90(3):423–5.
6. Banerjee A, McLarney RM, Duffy RF, Manders SM. Congenital onychodysplasia of the ring finger presenting as a bifid nail: report of a 6-year-old girl. Pediatr Dermatol. 2021;38(6):1336–7.
7. Milani-Nejad N, Mosser-Goldfarb J. Congenital onychodysplasia of index fingers: Iso-Kikuchi syndrome. J Pediatr. 2020;218:254.
8. L'Orphelin JM, Picard C, Andre J, Richert B, Dompmartin A. Syndrome d'Iso-Kikuchi: onychodystrophie congénitale des index ou syndrome COIF. Ann Dermatol Venereol. 2018;145(4):304–6.
9. Jindal R, Gandhi N, Yadav A. Iso-Kikuchi syndrome in a neonate with bilateral index finger anonychia and distal phalanx hypoplasia. Clin Pediatr (Phila). 2021;60(5):228–30.
10. Xiao Z, Zhang Y, Liu Y. A 10-year-old girl with a melon seed–like nail on her index finger. Pediatr Dermatol. 2024;41(1):33–5.

Cite this article as: Pillai S, Chandel A, Kamble S, Chauhan V. Iso-Kikuchi syndrome: a rare non-hereditary congenital onycho-dysplasia in a new born. Int J Contemp Pediatr 2025;12:1585-7.