A rare Occurrence of Anonychia in a Neonate : A case Report

Deepak Mishra[1], Pankaj Ray[2], Chandra Bhal Jha[3], Rakesh Mishra[4]

[1] Pediatrician, Kanti Childrens' Hospital, Maharajgunj, Kathmandu, Nepal
[2] Chief Consulatant Pediatrician, Kanti Childrens' Hospital, Maharajgunj, Kathmandu, Nepal
[3] Senior Consultant Dermatologist, Koshi Hospital, Biratnagar, Nepal
[4] Orthopaedic Surgeon, National Trauma Center, Kathmandu, Nepal

Corresponding Author

Deepak Mishra,
Pediatrician, Kanti Children's Hospital,
Maharajgunj, Kathmandu, Nepal
Email- 319deepak@gmail.com

Keywords

Anonychia, autosomal recessive, non-syndromic

Online Access

DOI: 10.3126/mjsbh.v22i1.44357

Date of submission - 2022 Apr 08
Date of acceptance - 2023 Sep 16

Abstract

Congenital anonychia in isolated form is a rare anomaly in which there is an absence of nails of fingers and toes since birth. However, other ectodermal and mesodermal defects in the body are absent. Here, we report a case of 30-hour old female with e rare congenital disorder anonychia in fingernails of her left hand and all toenails. All other systemic examinations were normal. The baby was born of a consanguineous marriage, and the father also had absent nails in multiple fingers.

INTRODUCTION

Congenital simple anonychia is a rare anomaly in which there is an absence of nails of some or all fingers and toes since birth without any association with other syndrome.[1] Anonychia may be either acquired anonychia or congenital syndromic anonychia, and both are more common than congenital asyndromic type.[2] Simple congenital anonychia denotes congenital absence of the nails where there is no other congenital defect. This is an extremely rare type of anonychia. Autosomal recessive inheritance is the primary mode of inheritance in simple anonychia.[2] In hyponychia, only a part of the nail is missing, and other tissues at the tips of the fingers and toes, such as nail bed are normal. Other than anonychia congenita, the overall health of the patient remains normal. Its prevalence is unknown. The etiology behind this rarest condition is mutations in the RSPO4 gene on chromosome 20 in exon 3.[3] Fragility on the long arm of the chromosome 10 is also associated with the anonychia.[4] This gene gives instructions for the synthesis of a protein called R-spondin-4. R-spondin-4 has an important action in the Wnt signaling pathway, which is responsible for the growth and development of nails.[4]

CASE REPORT

A female baby at 30 hours of life presented to our emergency room with the chief complaint of delayed passage of meconium and urine. She had normal tone, activity, and reflex with stable vitals on examination. Physical examination revealed an absence of finger nails of left hand (Figure 1) and all toe nails (Figure 2). Nail folds, nail beds and distal phalanges were found to be normally developed. The tips of the involved fingers were soft and compressible.

Fig 1: Absence of fingernails of left-hand

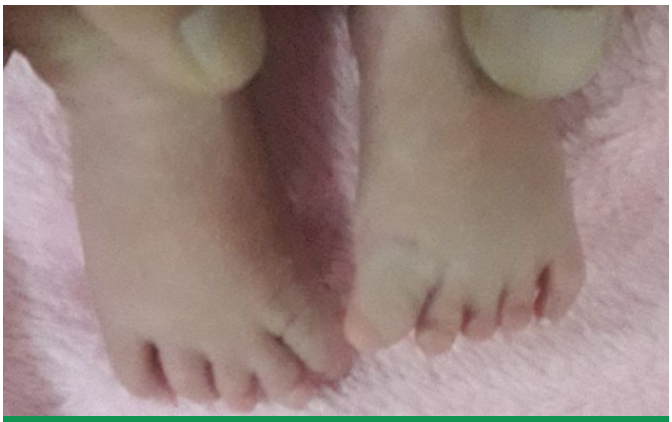

Fig 2: Absence of toenails

Hairs, tongue, oral mucosa, and head circumference all were normal. Ichthyosis, dyshidrosis, skin pigmentation or palmoplantar keratoderma were absent. X-ray of chest, hands and feet were also normal. (Figure 3 & 4)

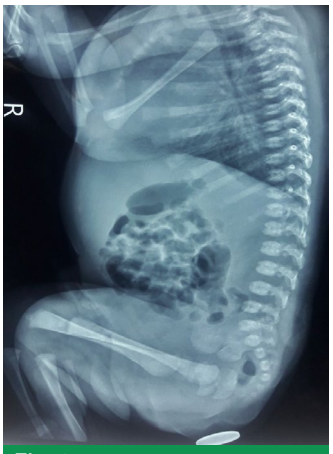

Fig 3

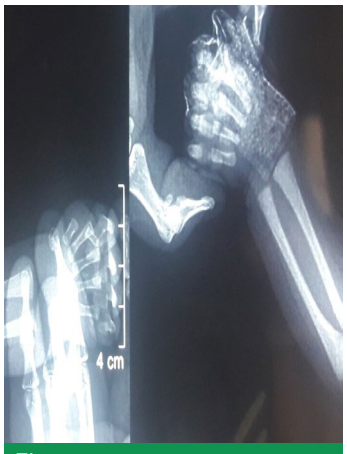

Fig 4

According to her parents, she was born at the hospital via normal vaginal delivery at full term. There was no history of taking any medicine by the mother during pregnancy. As per the mother's history, the baby was born of a consanguineous marriage, and the father also has absent nails in multiple fingers. Further investigations related to genetics could not be done due to financial constraints.

DISCUSSION

Anonychia, a rare anomaly, may be congenital or acquired. The development of a normal nail requires the underlying bone to be normoplastic. An abnormality occurs when the bone is either absent or not normally developed. These abnormalities range from a slight change to the complete absence of fingernails and toenails (complete anonychia). Anonychia may appear as an isolated abnormality or as a feature of genetic syndromes. Complete anonychia generally follows an autosomal recessive pattern and has been mapped to chromosome 20p135. Recently, mutations in the RSPO4 gene, including a novel missense mutation c.178C > T (p. R60W) were identified in three Pakistani families presenting with combinations of anonychia and hyponychia.[5] Intake of drugs like warfarin, phenytoin, and alcohol by the pregnant mother during the first and second trimester leads to nail anomalies.[6] Viral infection during the same period also causes nail anomalies.[2] In this case, mother's pregnancy was normal without any history of medication intake. Important congenital causes include various syndromes like nail-patella syndrome, AEC syndrome (ankyloblepharon, ectodermal defects, cleft lip/palate), TOOD syndrome (tricho-odonto-onychodermal), DOOR syndrome (deafness, onychodystrophy, osteodystrophy, and mental retardation), EEC syndrome (ectodactyly, ectodermal dysplasia, cleft lip/palate), hypo hidrotic ectodermal dysplasia including multiple anomalies and craniofacial malformation syndromes.[2] Anonychia congenita, is an inherited non-syndromic nail disorder that is characterized by isolated abnormalities of the nails. There are ten different types of "Nail disorder, Nonsyndromic Congenital."[7] Anonychia congenita is characterized by the complete absence or severe hypoplasia of all nails of the fingers and toes with no associated physical or mental abnormality. In simple anonychia, there is an absence of nails since birth, while no coexisting anomaly is found. Among nail disorders, isolated simple anonychia is an extremely rare condition. The autosomal recessive pattern is the primary mode of inheritance.[2] Consanguineous marriage should be avoided to reduce the chances of recessive disorders like anonychia.

CONCLUSIONS

Congenital anonychia in isolated form with no other associated anomalies is a rare anomaly in which there is an absence of nails of fingers and toes since birth. It exhibits autosomal recessive inheritance and should be considered when assessing a child born from a consanguineous union.

ACKNOWLEDGEMENT: None

FUNDING SOURCES: None

CONFLICT OF INTEREST: None

REFERENCES

1. Afsar FS, Karakuzu A. Total congenital anonychia. Pediatr Dermatol. 2014;31(6):743-4.
DOI:10.1111/pde.12474
PMID:25424215

2. Yadalla HK, Nitya R, Sujatha C. A rare case of isolated congenital complete simple anonychia. Int J Heal Allied Sci. 2012;1(1):32.
DOI: 10.4103/2278-344X.96419

3. Bruchle NO, Frank J, Frank V, Senderek J, Akar A, Koc E, et al. RSPO4 is the major gene in autosomal-recessive anonychia and mutations cluster in the furin-like cysteine-rich domains of the Wnt signaling ligand R-spondin 4. J Invest Dermatol. 2008;128(4):791-6.
DOI: 10.1038/sj.jid.5701088
PMID:17914448

4. Ozyazgan I, Ozyazgan I, Dundar M. Isolated congenital anonychia cases with coincident chromosomal fragility. Ann Genet. 2004;47(4):381-6.
DOI: 10.1016/j.anngen.2004.03.004
PMID:15581836

5. Khan TN, Klar J, Nawaz S, Jameel M, Tariq M, Malik NA, et al. Novel missense mutation in the RSPO4 gene in congenital hyponychia and evidence for a polymorphic initiation codon (p.M1I). BMC Med Genet [Internet]. 2012;13(1):1.
DOI: 10.1186/1471-2350-13-120
PMID:23234511 PMCID:PMC3532313

6. Lawry M, Daniel CR. Nails in systemic disease. In: Scher RK, Daniel CR, editors. Nails: Diagnosis, Therapy, Surgery. 3 rd ed. Philidelphia: Elsevier Science Limited; 2005; 147-169. DOI:10.4103/0378-6323.86472
PMID:22016270

7. Khan S, Basit S, Habib R, Kamal A, Muhammad N, Ahmad W. Genetics of human isolated hereditary nail disorders. Br J Dermatol. 2015;173(4):922-9.
DOI: 10.1111/bjd.14023
PMID:26149975