

Anonychia congenita

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A 15-year-old female presented with absence of toenails since birth [Figure 1]. On examination, there was absence of nine toenails; the right big toenail was normal. Fingernails were also normal. No abnormality of the bone and teeth was present. Radiography of both feet showed no abnormality. The patient had normal intelligence. There was no family history. Based on the clinical features and history, a diagnosis of congenital nonsyndromic simple partial anonychia was made.

Anonychia (absence of nails) is a very rare congenital or acquired anomaly. It may occur as a single feature or as part of a syndrome.



Figure 1: Absence of all toenails except the right big toe nail

Nonsyndromic anonychia has been reported in either partial or total forms. Simple anonychia means congenital absence of the nails without any other coexisting major congenital anomaly, and is extremely rare.^[1] It is caused due to frameshift and nonconservative missense mutation in the exon 2 of R-spondin 4 gene present on chromosome 20p13, which affects the highly conserved first furin-like cysteine-rich domain that plays a crucial role in nail morphogenesis, resulting in absence of nails.^[2]

Treatment remains masterly inactivity or artificial nails.

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