Anonychia Congenita - Rare Inheritance of a Rare Disorder

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Abstract
Non syndromic Anonychia congenita or congenital absence of finger and toe nails is a rare disorder known to occur due to autosomal recessive inheritance of mutation in the R-spondin-4 gene. We present a case of a 32 year old female born of a non-consanguineous marriage presenting with complete absence of finger and toe nails since birth and similar presentation in family members over four generations, suggesting an autosomal dominant inheritance.

Introduction
Absence of finger and toe nails is termed as anonychia.¹ Anonychia or hyponychia is a rare disorder attributed to homozygous or compound heterozygous mutation in the R-spondin-4 gene on chromosome 20p13.²,³ Anonychia or hyponychia may be partial or total and is usually associated with other skeletal or limb anomalies and various genetic syndromes like Coffin-Siris syndrome and Nail–patella syndrome. Non syndromic congenital anonychia is a rarer entity with only a few case reports in literature.⁴,⁵ The presence of an autosomal dominant form of the disease has not been reported in the best of our knowledge. We present a case of a 32 year old female with complete absence of finger and toe nails over four generations.

Case Report
A 32 year old female, presented with complete absence of fingernails and toenails since birth (Figures 1, 2). She did not have any bony abnormalities, dysmorphic facies or abnormal hair or teeth. Her developmental milestones and intelligence were normal. Her family history was significant for complete anonychia over 4 generations (Figure 3). There was no history of consanguineous marriage among family members.

Discussion
Although syndromes associated with anonychia are usually autosomal dominant (like Coffin-Siris syndrome and nail-patella syndrome), isolated non-syndromic congenital anonychia has an autosomal recessive inheritance pattern. A partial autosomal dominant form of the disease affecting only the thumb is also known.⁶

Most of the case reports of non-syndromic anonychia are seen in consanguineous marriages and are not known to affect more than one generation.

In this case, presence of anonychia was observed over 4 generations. This along with the absence of a consanguineous marriage precludes the presence of autosomal recessive inheritance. Whether, this can be attributed to autosomal dominant mutation of the same or different gene will be a question for future research. A detailed genetic evaluation could not be done in our case due to financial and technical limitations.

References

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