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Anonychia congenita totalis with epilepsy and mental retardation

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CASE REPORT

ABSTRACT

Anonychia congenita is a rare disorder characterized by absence of several or all fingernails and/or toenails since birth. A 35-year-old man with mental retardation presented with a history of absence of all nails since birth. There was also a history of epileptic fits since early childhood.

Key Words

Anonychia, anonychia congenita, RSPO4 gene

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INTRODUCTION

Absence of nails from birth (congenital anonychia) is a rare congenital anomaly. It may occur as an isolated symptom or be a part of other developmental defects of digits or any structures,[1] which are sometimes hereditary.[2] Anonychia may not be complete and often there are rudimentary nails on some fingers and toes (hyponychia). Anonychia occur sporadically, it may have dominant or recessive inheritance pattern.[3] Very few cases have been reported till now. We herein, report a rare case of anonychia congenita totalis with epilepsy and mental retardation.

CASE REPORT

A 35-year-old product of non-consanguineous marriage presented with a history of complete absence of nails of all fingers and toes since birth. (Fig 1 & 2) There was no other associated bony abnormality of hands or feet. He was otherwise physically normal, except that he had mental retardation.

On further questioning from family members, he was found to be on treatment for epilepsy since early childhood. There was no history of any drug intake by his mother during pregnancy. His parents and other siblings were normal.

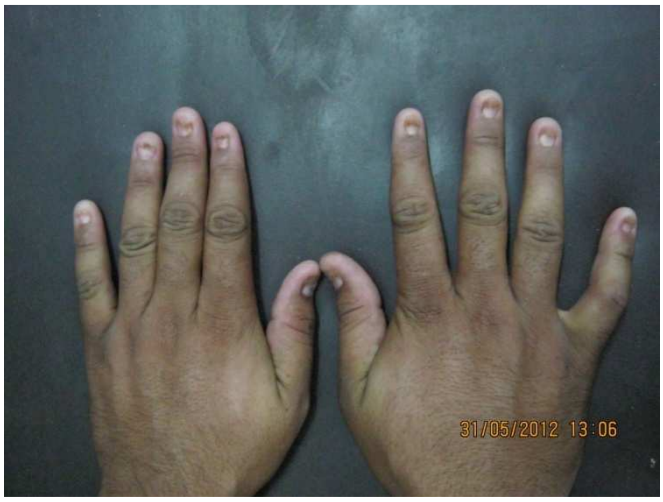

Figure 1: Hands showing absence of all nails

The skin around the absent nails was normal. Hair and teeth of the subject were also normal. Systemic examination was normal. Routine laboratory investigations including complete blood count, liver function tests, urine examination were normal. Radiological examination of hands and feet were unremarkable. Based upon the history and clinical features, a diagnosis of anonychia congenita totalis with epilepsy and mental retardation was made.

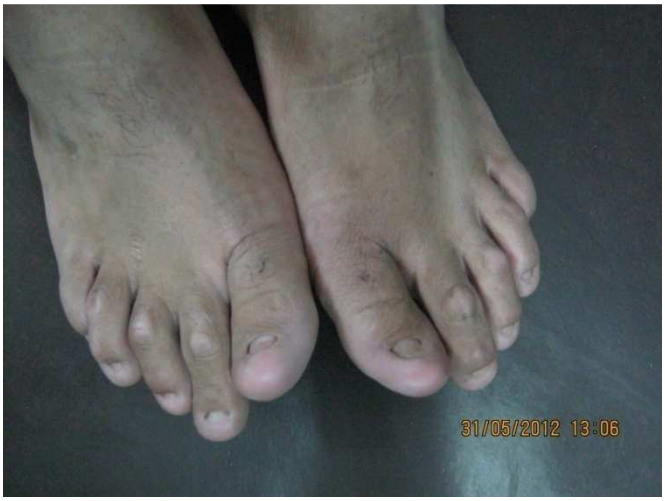

Figure 2: Feet showing absence of all nails

DISCUSSION

Anonychia congenita is an extremely rare disorder characterized by complete absence of fingernails and toenails.[1] It may occur singly or as a part of some syndromes like nail patella syndrome, hypohidrotic ectodermal dysplasia, tricho-odonto-dental syndrome, DOOR syndrome (deafness, onychodystrophy, osteodystrophy and mental retardation), AEC syndrome (ankyloblephron, ectodermal defects, cleft lip/palate), and various craniofacial malformation syndromes.[4] Simple anonychia means congenital absence of nails without any other coexisting major congenital anomaly.[5] Non-syndromal anonychia can present as partial or total anonychia. The partial form involves any thumb with autosomal dominant inheritance while the involvement of second third and fourth digits represents an autosomal recessive trait.[6]

Development of nails begins at 8-10 weeks of expected gestational age and is completed by fifth month.[4] Drugs consumed by the mother during first or second trimester are extremely significant for nail formation. Phenytoin or warfarin may cause hypoplasia of nails, while alcohol may lead to anonychia.[7] Although, mother of our patients had not taken any medicines.

Absence of nails probably represents a mesenchymal dysplasia occurring during the morphogenesis of the digits. The gene RSPO4 has been identified responsible for anonychia, which is a member of R-spondin family of secreted proteins. Due to frameshift and non-conservative missense mutation in the exon 2 of R-spondin 4 gene present on chromosome 20p13 which helps in activating the Wnt/beta-catenin signalling pathway, that affects the highly conserved first furin-like cysteine-rich domain which plays a crucial role in nail morphogenesis, resulting in absence of nails.[8] Interestingly there is no family history of anonychia in our case and this condition seems to be caused by sporadic mutation.

Several syndromic variants of this disorder have been reported in the literature.[3,5,6] Our case was unique as congenital anonychia was associated with epilepsy and mental retardation. Whether it an association or mere coincidence is a matter of debate?

CONCLUSION

As this condition does not interfere with daily activity of the affected individuals, he/she should be assured about the condition. The patients of congenital anonychia should be examined thoroughly because of possible association, some of which are mentioned above. This case is presented in view of its rarity of this condition.

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