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AUTOSOMAL RECESSIVE TOTAL CONGENITAL ANONYCHIA, IN A SAUDI FAMILY

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Abstract
The autosomal recessive total congenital anonychia is a rare genetic disorder. In this manuscript we are reporting the occurrence of this trait in a Saudi Arabian family.

Key words: dermatology; genetic; nails

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Introduction

Total absence of nails since birth, Otherwise called Total Congenital Anonychia; (TCA) is a rare trait.
This rare genetic disorder has been reported in individuals from different countries around the world. Previous report [1], had indicated that it could be found in countries such as, Russia, Great Britain, America, Holland, Iran. Recently, it has been reported in Brazil [2] and Turkey [3].
In this manuscript, we use a Saudi family as a case study.

Case Report

The proband was a 22-year-old boy who had come to hospital for the treatment of acne vulgaris. Complete absence of all nails, had been noticed since birth. His parents are first cousin. One of his sisters have also suffered the same disorder (TCA). Although, she is married to a person from different tribe she has 2 children with normal nails. No other members of the family was affected with nail disorders and no history of the similar condition before in the family.
Family pedigree is shown in Figure 1.The patient had no other obvious case of disorder. However, the parents mentioned that, their affected children always request someone to help scratch their skin whenever they had itchy skin. The parents also noted that the „skin'', in place of the nails (nail bed), get thickened with time.

Physical Examination showed complete absence of all the fingernails, and all toenails. Other examinations, in particular such as hair, teeth, and skeletal system, revealed no abnormality. Skin examination, was also normal and did not show any abnormal pigmentation. Nevertheless, the family declined medical photography and the radiological examination of the hands and feet.
Similarly, his affected sister could not be examined as she lives in another city.

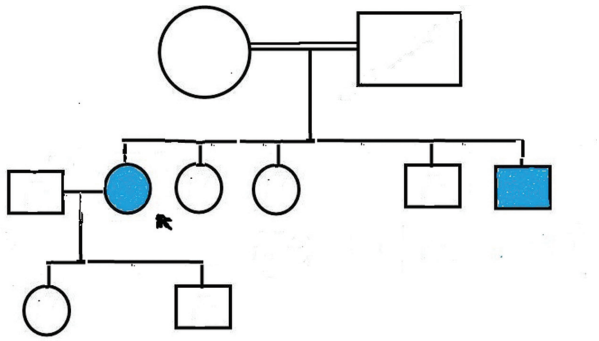

Figure 1. Family pedigree.

Discussion

Genetic disorders are not uncommon in Saudi Arabia. This is because of several factors, and in particular, consanguineous marriages which are very common in Saudi Arabia especially in villages and small towns. The government had launched a pre-marital medical counseling program in order to reduce the burden of genetic disorders especially the genetic blood diseases like sickle cell anemia.

There are several common and rare genetic dermatological disorders reported in families from Saudi Arabia. These include; Kindler syndrome [4], Multiple hereditary trichoepitheliomas [5], Lamellar ichthyosis [6], Hereditary hypotrichosis simplex [7].

Anonychia congenita totalis, had also been reported from Saudi Arabia but in a single patient [8].

Therefore, this is the first report of Total Congenital Anonychia (TCA), in a Saudi family.

Congenital absence of a nail (anonychia) is a rare genetic defect. The first two cases of anonychia were described in 1842 [8].

Anonychia of a single or a few nails can be found in many hereditary disorders and syndromes. For examples in Cooks syndrome which is a combination of anonychia and absence or hypoplasia of distal phalanges [9], also, in Zimmermann-Laband syndrome, which has many features including gingival fibromatosis and hypoplastic or absent nails [10].

However, TCA have been reported with a limited syndromes. A previous review, have listed the following reported associations with TCA:

1. Aplasia or hypoplasia of upper lateral incisor, spaced teeth, loss of some molars.
2. Microcephaly, clinodactyly, single transverse palmar crease, widely spaced teeth.
3. Deafness and onycho-osteodystrophy syndrome, DOOR syndrome (deafness, onycho-osteodystrophy, mental retardation).
4. Glossopalatine ankylosis syndrome (abnormal mouth, tongue being attached to temporomandibular joint).
5. Bizarre flexural pigmentation and hair abnormalies.
6.Dyscephalic mandibulo-oculofacial syndrome and craniofrontal nasal dysplasia.

Nevertheless, TCA can be isolated defect in this family.

TCA is inherited mostly as an autosomal recessive manner like the family in this report. The trait does not affect the life of the affected patients. Yet, patient might be sensitive to touching acidic materials using the tips of their fingers or may feel uncomfortable by scratching the skin with no nails.

It has been found that autosomal recessive form of TCA is caused by loss-of-function mutations in the gene encoding R-spondin 4 (RSPO4), present on chromosome 20p13, which functions in the WNT signaling pathway. This gene plays a crucial role in nail morphogenesis, andacts as a landmark for early nail unit formation [12].

We believe that autosomal recessive TCA is a distinct disorder. However, the affected patients need to be examined thoroughly to exclude systemic or ectodermal defect particularly the teeth.

There is no specific therapeutic procedure for this disorder and the treatment remains masterly inactivity or artificial nails.

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