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Familial polysyndactyly type-IV variant, a large Indian family

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ABSTRACT

Haas type syndactyly type-IV is a rare familial syndactyly and it is even rarer to have its variant. We had an unique family having multiple members affected with polysyndactyly of both hands, triphalangeal thumb and variable involvement of feet. About 58 members in five generations are affected with male to female ratio 1.4:1. The vertical transmission from male to male in subsequent generations is suggestive of autosomal dominant inheritance. There are no other morphological abnormalities and the life expectancy was normal.

KEYWORDS familial polysyndactyly, Haas type syndactyly, triphalangeal thumb

Introduction

Haas type syndactyly (type-IV) has a prevalence of 1/300 000 and segregates in an autosomal dominant fashion. It manifests itself as complete cutaneous fusion of all fingers accompanied by the presence of pre- or postaxial polydactyly[1]. The nails may be fused completely or give an impression of separation by a groove only. Flexion of fingers is limited and the union of contiguous fingers gives the hand a cup-shaped appearance. Phalanges may fuse as a conglomerate mass of bones; however, metacarpal synostosis is absent. In the literature, at least two variants of type IV syndactyly have been reported: (a) typical Haas type without involvement of feet; and (b) complete fusion of all fingers with variable fusion of all five digits in feet[2] .52 Haas type syndactyly has been shown to be allelic to triphalangeal thumb polysyndactyly, which is present at a milder end of phenotype. Both entities are caused by mutations in the ZRS locus at chromosome 7q36 (LMBR1), encompassing a long-range regulator of SHH[3, 4].

Case series

A six months male child(fig.1) was admitted to the department of Pediatrics, Government medical college, Kota, India, with complains of fever, cough and loose stools for 3 days. On routine physical examination bilateral pre-axial polydactyly with cutaneous syndactyly of thumb and extra pre-axial digit; 3rd, 4th and 5th finger were present. The index finger was spared bilaterally. The thumb and extra pre-axial digit were triphalangeal. Pre-axial polysyndactyly of right foot was also present. There was no other significant anatomical abnormality detected. On asking family history it was revealed that the mother of the child had pre-axial polysyndactyly of bilateral hands and triphalangeal thumb in similar fashion. Further enquiry into family revealed that 58 members(fig.2) among five generations are affected with variable degree of polysyndactyly of both hands with triphalangeal thumb with or without polysyndactyly of feet. The male to female ratio was 1.4:1. There were no other morphological abnormalities detected. The life expectancy of affected members was normal as the maximum age reported was 75 years in one of the grandparents of the child.

On radiological evaluation of one of the maternal uncle showed following abnormalities (fig. 3):

- Pre-axial polydactyly with extra metacarpal.
- Triphalangeal extra digit and thumb bilaterally.
- Bilateral syndactyly of thumb and extra digit; syndactyly of 3rd, 4th and 5th fingers.
- Bilateral duplication of 2nd and 3rd phalanges of thumb with ulnar deviation of 3rd, 4th and 5th fingers.

Discussion

Syndactyly is a digital malformation in which adjacent fingers and/or toes are webbed because they fail to separate during

limb development. According to the current classification at least nine non-syndromic syndactylies with additional subtypes have been characterized[5]. Most of

these entities segregate in Mendelian dominant fashion. However, two autosomal recessive and an X-linked recessive type have also been described. It is one of the most common hereditary limb malformations depicting a prevalence of 3–10 in 10 000 births, though higher estimates ranging from 10–40/10 000 have been reported[6, 7]. However, certain specific types of syndactyly syndromes are rare in the general population and Haas type IV polysyndactyly is one of them. It is even rarer to have variants of type IV polysyndactyly.

We had such a large family with type IV polysyndactyly with certain variability in the phenotypic expression. First, the syndactyly was incomplete i.e., all the six digits were not completely fused as described in Haas type. Second, the terminal phalanges were also not fused even in fingers described with syndactyly. Third, there was presence of duplication of 2nd and 3rd phalanges of thumb bilaterally. Fourth, there was duplication of hallux in some of the members affected with polysyndactyly.

The reported phenotypic abnormalities in this series are not fitting in to any of the current classifications proposed for syndactyly; however, the close resemblance to Haas type IV will lead to placing these abnormalities as "variant" of type IV syndactyly. As newer cases are discovered day by day there will be a need to review the current classification from time to time.

Conflict of interest:None

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