INTRODUCTION

Temtamy and McKusick defined terminal transverse limb defects as absence or hypoplasia of distal structures of limbs with more or less normal proximal structures. It is a rare, congenital deficiency which manifests itself as an abrupt truncation through the transverse axis of limb and produces an amputation like stump. The truncation through the hand may involve only certain phalanges, only the digits or the full hand. The malformation may be unilateral or bilateral and may affect upper limbs and/or lower limbs. Most of the cases reported are sporadic and isolated, however, the condition may appear as a part of syndrome. The genetic bases of isolated terminal limb deficiencies are not well understood.

Here, we present a sporadic male subject with an isolated, congenital, unilateral terminal reduction deformity of the left hand.

CASE REPORT

The index subject, a 47 years old male, belonged to a rural area of the Southern-Punjab. He is self-employed as a kitchen chef and has adequately adapted to perform his routine activities. The subject is the third in a sibship of 6 individuals (3 brothers, 3 sisters) and has 5 normal children (3 boys, 2 girls). Parental consanguinity was denied.

The left arm of the subject was short and lacked a proper formed hand, the forearm abruptly ending in a stump (Figures 1A and 1B). The hand was amputated through the palm. Five bead-like finger remnants were attached through cutaneous bridges to the distal rim of reduced palm. They appeared as non-functional, under-developed nubbins and harbored nails at their dorsum (Figures 1A and 1B). The amputated autopod had compromised extension and flexion movements.

In the X-ray, left radius and ulna were observed to be short (Figures 1C and 1D, Table I). Their distal heads gave evidence of hypoplasia. Particularly, the radius was thin and its distal extremity revealed retarded growth and decalcification. A bony synostosis of dysplastic epiphyses covered the distal heads of radius and ulna (Figures 1E and 1F). The malformation may be unilateral or bilateral and may affect upper limbs and/or lower limbs. Most of the cases reported are sporadic and isolated, however, the condition may appear as a part of syndrome. The genetic bases of isolated terminal limb deficiencies are not well understood.

Here, we present a sporadic male subject with an isolated, congenital, unilateral terminal reduction deformity of the left hand.
ulna. Carpals/metacarpals, and the proximal/middle phalanges were completely missing. Each nubbin had a single hypoplastic bony element (Figures 1C and 1D, Table I). In the upper-arm, shoulder, humerus, and elbow joints were unremarkable. Additionally, the right arm, lower limbs and feet appeared unaffected. The subject had no other clinical symptoms of orofacial, neurological, skeletal or internal organs.

**DISCUSSION**

The clinical presentation in this subject is similar to the subjects reported by Neumann et al., Limont and Salisbury, and Graham et al. In all those cases the transverse anomaly was affecting the autopod only while the proximal limb segment demonstrated minimal symptoms. Likewise, the cases reported by Phakde et al., Horn and Kolb also depicted trans-carpal amputations, however, all these cases were observed to be bilateral (Table I). Additionally, few of them involved transverse anomalies of lower limbs. Furthermore, certain other subjects reported in the medical literature had atypical transverse anomalies and their phenotypic presentations are not consistent with the clinical and roentgenographic observations in the present case. A summary of selected reports on congenital amputation is given in Table I.

The etiology and pathogenesis of isolated limb amputations is not clear. For the asymmetrical and unilateral cases, the extragenic factors and teratogens could not be ruled out. On the other hand, symmetrical terminal transverse deficiencies involving all 4 limbs or bilateral upper or lower limbs may suggest genetic predisposition. Additionally, transverse limb deficiencies have also been reported as a part of several well-characterized syndromic malformations like Adams-Oliver syndrome, Poland syndrome, and achiropody, etc., which reiterates the genetic etiology of transverse malformations.

**REFERENCES**