Sir,

Congenital Trigger Thumb (CTT; OMIM-190410) is characterized by fixed flexion at the Interphalangeal Joint (IPJ). It is a rare malformation and unknown etiology.\(^1\) Thumb Aplasia (TA; OMIM-188100) is another rare limb anomaly occasionally associated with radial deficiency syndromes.\(^2\) These anomalies occur independently and may cause mild to severe arrest in the hand’s function and the quality of life of the individual. Here, we report an unusual observation of the concomitant presence of CTT and TA in a subject who had a positive family history of polydactyly (Figure 1A).

A female individual of 35 years came from the suburbs of Islamabad and was initially ascertained from Pakistan Institute of Medical Sciences, Islamabad. She was observed to have a complete absence of the thumb in the right hand (Figure 1 and 2). The palm was thin and reduced and the index finger depicted medial inclination. Additionally, there was camptodactyly of third and fourth digits. Radiographs revealed a complete absence of first digital ray including phalangeal elements, metacarpal and certain carpals, while the remaining carpals were crowded (Figure 2A and B). The radius and ulna demonstrated shorting and dysplastic distal heads. They, however, did not show signs of curvature.

In the left hand, there was trigger thumb at the IPJ and it was extendable only up to 90 degrees (Figure 1 and 2). There was soft tissue contracture along the volar aspect of the thumb. The condition was non-progressive and was not associated with swelling or pain. Radiographic examination depicted a hypoplastic first digital ray, displaced pisiform and triquetral, and absent trapezoid and scaphoid (Figure 2). There were signs of symphalangism at the distal IPJ of the index finger.

Accordingly, the phenotype in the right hand was concordant with hypoplastic thumb type-5, as per revised classification of James et al.\(^3\) The contracture phenotype of left thumb was consistent with trigger thumb stage-IV.\(^3\) The individual had normal IQ and no other symptoms. Her parents were first cousins (inbreeding-coefficient F=0.0625). Family history was negative for thumb aplasia/hypoplasia or trigger thumb. However, one of her brothers had unilateral polydactyly (pre-axial type-I) in his left hand (Figure 1A).

The association of CTT with other digit anomalies has not been well-documented. On the other hand, complete absence of thumb has been reported to be associated with limb anomalies like syndactyly, symbrachydactyly, pre-axial polydactyly, mirror hand, and constriction band syndrome.\(^2,4\) Malik and Jabeen reported a male individual with unilateral TA associated with zygodactyly, lean physique and low weight,\(^4\) with a family history of polydactyly. The present subject also had a first-degree relative with polydactyly, though other symptoms were not concordant. There are a number of well-characterized syndromic conditions with thumb hypoplasia/aplasia and polydactyly, i.e., hypoplasia of tibia with polydactyly (OMIM-188770), Townes-Brocks syndrome (OMIM-107480), and Fuhrmann syndrome (OMIM-228930); however, these syndromes have other symptoms which were not witnessed in our subject.
Recently, Riaz et al. reported a collection of 6 cases with TA and none of those had trigger thumb as an associated anomaly. Nonetheless, the molecular bases of isolated forms of CTT and TA remain to be discovered.

REFERENCES

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