Clinical Medicine Insights: Arthritis and Musculoskeletal Disorders

CASE REPORT

Bilateral Congenital Absence of Flexor Pollicis Brevis and Abductor Pollicis Brevis Muscles with Bilateral Thenar Atrophy: A Case Report

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Abstract: In this paper, we report a case of a 23-year-old male patient with bilateral absence of the flexor pollicis brevis and abductor pollicis brevis muscles with an intact functioning opponens pollicis and flexor pollicis longus muscles with bilateral thenar atrophy due to its rarity. All physical, neurological, ultrasonographic, direct radiographic, electromyographic and MRI studies were used to confirm and document this congenital anomaly.

Keywords: hand, flexor pollicis brevis, abductor pollicis brevis, thenar atrophy

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Introduction

The thenar eminence constitutes the intrinsic muscles of the hand that are responsible for complex movements of the thumb. Thenar atrophy is the term that denotes the wasting of these muscles and is associated with various clinical situations.[1] Although it is almost always mentioned along with carpal tunnel syndrome, sometimes it may be associated with muscle anomalies that are often seen in connection with other syndromes or congenital anomalies.[1] It can be an isolated defect, as in Cavanagh's syndrome,[2] can be present with cardiac (Holt-Oram syndrome) or eye (Okihiro's syndrome) disorders,[3,4] or can be associated with a hand anomaly, as in Haas's malformation.[5] Vascular abnormality may be associated with thenar hypoplasia, which has been demonstrated in Okihiro's syndrome.[6] Congenital absence of the thenar muscles with or without absence of the flexor pollicis longus and unilateral absence of abductor pollicis brevis and flexor pollicis brevis has been reported previously.[7] No treatment is indicated for this selective deficit in view of the satisfactory hand function. A rare case is presented here of selective bilateral absence of the flexor pollicis brevis and abductor pollicis brevis with an intact functioning opponens pollicis and flexor pollicis longus, which has not been previously reported.

Case Report

A 23 year-old male patient presented at our orthopaedic clinic with a complaint of mild deficiencies in some hand functions; he especially had mild dysfunction in touching the thumbs to the other fingers or performing tasks that require fine skills. The patient was aware of the deformity ever since he was a child but was not concerned about this. There was no history of any trauma in childhood and adulthood and no family history of congenital anomalies. All physical, neurological, ultrasonographic, electromyographic and MRI studies were done. His routine X-rays of the hand did not show any abnormalities. No organ anomaly was present on systemic examination. In the neurological examination, there was mild disability in bilateral thumb abduction and opposition. In the EMG study, the median nerve was found to be intact. Moreover, there was bilateral thenar atrophy (Figs. 1 and 2). No vascular anomaly was determined from the Doppler study. MRI, axial and coronal cross-sectional images showed the absence of bilateral flexor pollicis brevis and abductor pollicis brevis muscles (Fig. 3 and Fig. 4). All other thenar muscles and the flexor pollicis longus muscle were intact. In our case, the patient refused any surgical treatment for the the absence of muscles as there was only mild hand dysfunction and the deformity is of little functional significance to him.

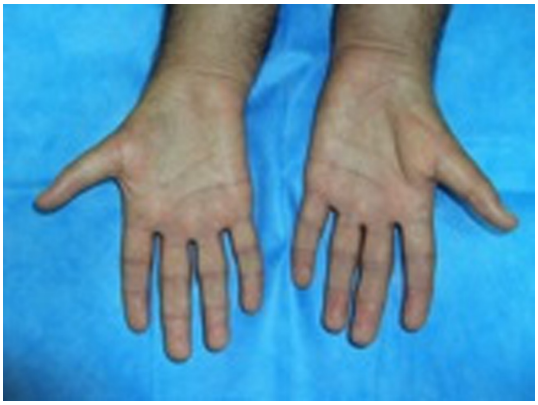

Figure 1. Patient with bilateral thenar atrophy.

Discussion

Together with the abductor pollicis muscle, the opponens pollicis and flexor pollicis muscles that form the thenar eminence, play an important role in hand functions, especially for fine motor functions of the thumb.[8] The flexor pollicis brevis is a small, narrow muscle consisting of two portions, the outer and inner. The outer portion is superficial and originates from the flexor retinaculum of the wrist. The inner portion is deep and originates from the ulnar side of the metacarpal bone. These two portions join to form a tendon that inserts in the proximal phalanx of the thumb. This muscle is innervated by motor branches

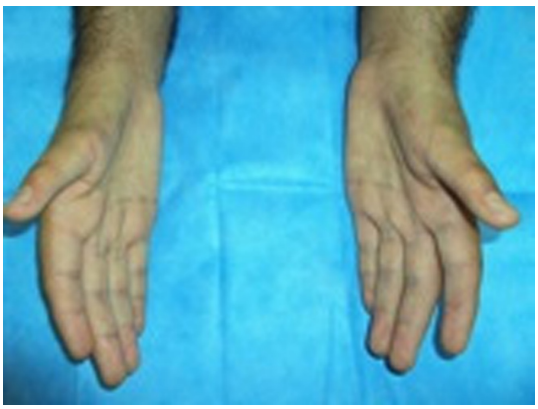

Figure 2. Patient with bilateral thenar atrophy.

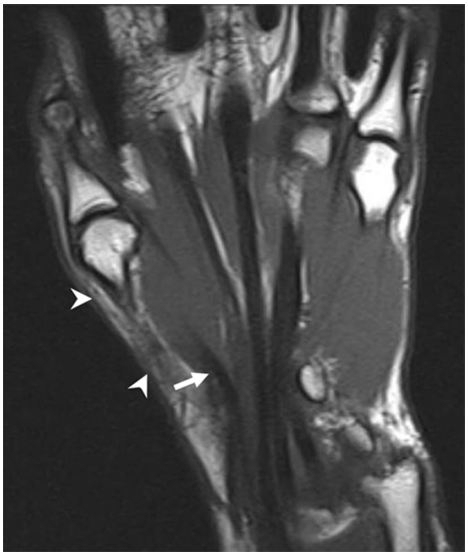

Figure 3. MRI coronal image shows abductor pollicis brevis muscle agenesis.

of the median nerve and is supplied by branches of the radial artery. The flexor pollicis brevis flexes and rotates the thumb. The abductor pollicis brevis is one of the muscles forming the thenar eminence. The abductor pollicis brevis is the largest of the thenar eminence muscle group and is flat, elongated and triangular in shape. It lies just below the skin and slightly overlaps the flexor pollicis brevis and mostly covers the opponens pollicis. The abductor pollicis brevis originates from the tubercle of the trapezium and from the flexor retinaculum. It inserts in the proximal phalanx of the thumb. This muscle is innervated by the median nerve and supplied by palmar branches of the radial artery. It moves the metacarpal bone of the thumb away from the palm. Incidence of bone -muscle anomalies of the hand and forearm is within the range of 1/30.000 and 1/100.000.[9] Whilst male-female ratio

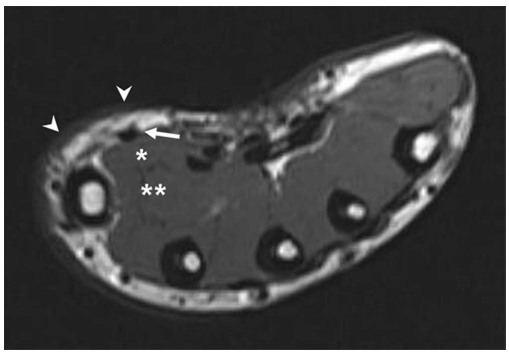

Figure 4. MRI axial image shows flexor pollicis brevis and abductor pollicis brevis agenesis.

is 3:2, thenar muscle anomalies and thumb hypoplasias can be seen in isolation, as in Cavanagh's syndrome and are commonly seen with syndromes such as Holt-Oram, TAR, Fanconi, VACTERL and radial bone anomalies.[2,10–13] Cavanagh's syndrome is a rare anomaly of the upper extremities that presents with unilateral or bilateral hypoplasia of the thenar eminence.[2] Typical clinical, radiographic, and electrophysiologic findings confirm the diagnosis. Differentiation from carpal tunnel syndrome is important to prevent unnecessary intervention. Electrophysiologic and radiographic findings are necessary tools for the physician to establish a correct diagnosis and make an appropriate referral. This syndrome may clinically present with absent radial artery.[2] Holt-Oram syndrome, also called heart-hand syndrome, is an inherited disorder characterized by abnormalities of the upper limbs and heart. Although the clinical manifestations are variable, upper limb abnormalities are always present.[13,14] Thrombocytopenia-absent radius (TAR) syndrome is a rare condition in which thrombocytopenia is associated with bilateral radial aplasia. Thenar atrophy is usually seen in this congenital malformation.[15] Clinodactyly, hypoplastic thenar eminence, 6 fingers, absent first metacarpal, enlarged abnormal fingers, and short fingers may be seen in Fanconi anemia.[16] When deciding treatment choices of these congenital anomalies, Blauth and Scneider- Sickret classification must be kept in mind.[8,9,17] In this classification, anomalies are classified as Type 1 through to Type 5 (Type 1-2-3A, 3B, 4, 5). Whilst Types 1,2, and 3A anomalies need corrective surgery, Types 3B,4, and 5 deformities must be treated by amputation of the thumb and 2nd finger reconstruction. In our case, the constituents of the thenar area (flexor pollicis brevis and abductor pollicis brevis) were absent bilaterally. No other musculoskeletal, neurological and vascular anomalies were present. We found no embryological basis to substantiate the selective absence of these muscles. To the best of our knowledge, isolated bilateral absence of the flexor pollicis brevis and abductor pollicis brevis muscle has not been previously reported in English literature. Isolated unilateral absence of flexor pollicis brevis and abductor pollicis brevis muscles was reported by Iyer et al in 1982.[7] Our patient had mild thumb dysfunction in opposition and pinching, and as he rejected the suggestion of corrective surgery, no treatment was applied to this elective patient.

Author Contributions

Conceived and designed the experiments: KK. Analysed the data: KK, TE, MK, BB. Wrote the first draft of the manuscript: KK, TE, MK. Contributed to the writing of the manuscript: SE, SO. Agree with manuscript results and conclusions: KK, SE, TE, SO, MK, BB, MB. Jointly developed the structure and arguments for the paper: KK, SE, TE, MB. Made critical revisions and approved final version: KK, SO, SE. All authors reviewed and approved of the final manuscript.

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Competing Interests

Author(s) disclose no potential conflicts of interest.

Disclosures and Ethics

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