

An unusual case of radial polydactyly, (tetraplication of the thumb, duplication of the radial carpal bones and bifurcation of the radius)

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Abstract

Radial polydactyly, the most common digital duplication in Asian and white populations, has a wide range of manifestations. Its classification is useful for planning and assessing surgical treatment. Our patient had four thumbs, duplicated radial carpal bones, and a bifurcated radius. This presentation is not covered by any of the current classifications. To the best of our knowledge, this is the first case of such characteristics reported to date. Consequently, we propose some modifications in the nomenclature and classification of radial polydactyly.

Keywords: Radial polydactyly, Triphalangeal thumb, Tetraplication of the thumb.

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Introduction

Polydactyly is one of the most common congenital deformities of the hand. Radial polydactyly associated with triphalangeal thumb accounts for 2% of congenital anomalies of the upper limbs (1). Radial polydactyly, the most common digital duplication in Asian and white people, has a wide range of manifestations (2-4). Many classifications have been proposed for describing this type of deformity. Our patient had polydactyly on the radial side of the hand and the four ulnar digits with normal appearance. The features of this patient's polydactyly did not fit any of the well-known classifications. The differential diagnosis in such cases includes the 6-fingered hand (nonopposable thumb), multiple duplication of the index finger with absent thumb, mirror hand, and the duplicate or multiple hand deformity (5). The original classification of radial polydactyly was published by Wassel in 1969(6). Various changes have been made to this origi-

nal scheme, but the main structure remains unchanged and it continues to be the most common classification system for this type of deformity. Analysis of our patient's deformity highlights the difficulty of fitting such rare cases into any classification system.

Wassel's classification is based on the level of duplication, from distal to proximal; uneven numbers are bifid duplication and even numbers are complete duplications (type I refers to bifid distal phalanx and type VI to duplicated metacarpal). Type VII refers to cases of duplication involving triphalangism. The Wassel classification is sufficient for simple radial polydactyly but a broader classification is needed to accurately classify triphalangism, triplication, and tetraplication.

Wood and Miura proposed additional subtypes to cover triphalangism and triplication (7, 8). In Wood's classification, type IV is subdivided into two subtypes: type IVA, in which both duplications have a

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Fig. 1. A- Opposition of 3 radial thumbs, B- three radial thumbs in view, C- V shaped deformity of 4 thumbs, D- congenital sinus on radial side of wrist (base of thumb).

triphalaengeal component at the level of the metacarpophalangeal (MP) joint; type IVB, in which the triphalaengeal component is only on the radial side. Miura added type C to cover ulnar triphalaengeism(8). Wassel's type VII was divided to 4 subtypes by Wood (7). Types VII A, B, and C refer to ulnar, bilateral, and radial triphalaengeism, respectively. Type D refers to a central triphalaengeal deformity with a nontriphalaengeal hypoplastic digit on each side (known as triplication). We present an unusual case of radial polydactyly not covered by any of the aforementioned types.

Case report

A 16-year-old boy was seen for assessment of a congenital anomaly of the left hand. His family history was negative for congenital anomalies. Physical examination of the affected limb revealed a hand with 8 digits. The 4 ulnar digits appeared normal and there was 1 separated, deformed thumb, and 3 deformed and syndactylized thumbs on the radial side (Fig. 1, A, B and C). A sinus tract on the radial side of the wrist had been present from birth (Fig.

1-D), and the left wrist was wider than the right. Ulnar deviation at the wrist was limited, range of shoulder motion was normal, and there was a 30 degree lack of extension at the elbow. The patient had no other deformities.

Radiographic examination showed 2 main triphalaengeal thumbs duplicated at the MP joint. Attached to the radial main thumb were 2 hypoplastic thumbs, one starting from the metacarpal and the other from the distal interphalaengeal joint (Fig. 2). There was one metacarpal, duplicated trapezium, scaphoid, and lunate bones, and a bifurcated radius. A degree of fusion between the two radial epiphyses produced relative shortening of the bone compared to the ulnar length at the wrist joint (Fig. 2). Posterolateral dislocation of the radial head at the elbow was also observed (Fig. 3).

After preoperative planning, we excised the three radial thumbs and corrected the deformed ulnar thumb by closing wedge osteotomy, shortening, and IP joint arthrodesis. Extensor and flexor tendons were realigned and abductorplasty was performed (Fig. 4).



Fig. 2. A, Two main triphalangeal thumbs duplicated from the metacarpophalangeal joint. B, Attached to the radial main thumb are two hypoplastic branches, one from the metacarpal and the other from the distal interphalangeal joint. C, Bifurcation of radius bone. D, Duplicated trapezium, scaphoid and lunate bones. E, Ulnar lengthening relative to duplicated radius bone.

Discussion

Embryologically, limb buds are an outgrowth of mesoderm into the overlaying ectoderm. This bulging ectoderm, known as the apical ectodermal ridge (AER), appears 26 days after fertilization (9). The terminal portion of the mesoderm is called the zone of polarizing activity (ZPA). Sonic hedgehog (SHH), which is expressed in the posterior portion of the ZPA, is the main regulator of the anteroposterior axis (the formation of ulnar and radial structures) (10). Zone of polarizing activity regulatory sequence (ZRS) is a limb specific SHH en-

hancer on chromosome 7q36. ZRS regulates SHH expression and point mutation in ZRS results in exaggerated SHH activity (11). Ectopic anterior expression of SHH results in various forms of preaxial polydactyly and triphalangeal thumb. Thumb triplication and triphalangeal thumbs are known to be more prevalent in populations with genetic isolates and mutations located at chromosome 7q36 (10). The basic defect that produces radial polydactyly, triphalangeal thumb and 6-fingered hand is the appearance of ectopic SHH in the anterior mesoderm (12). Al-Qattan reviewed the

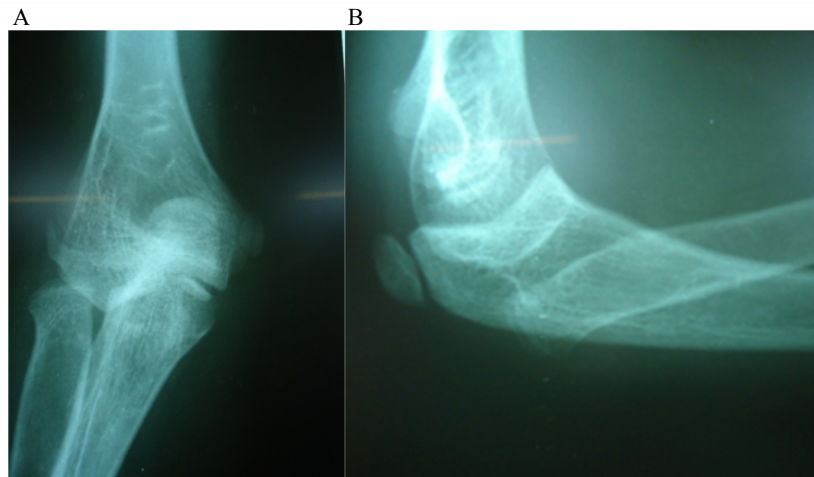


Fig. 3. A and B, Anteroposterior and lateral view of left elbow.



Fig. 4. The physical appearance of hand after operation.

pathogenesis of these malformations viewed as a spectrum of severity of embryonic events (11). Biphalaengeal duplicated thumb is usually unilateral, sporadic and resulted from developmental error can cause minor anterior SHH expression. Thumb triplication, radial polydactyly with triphalangism, and isolated triphalangism are the result of moderate anterior expression of SHH and are related to point mutation of ZRS on the 7q36 chromosome. In animal models, elevated levels of SHH in the radial aspect of the limb bud cause mirror hand or ulnar dimelia (12). In humans, there is a relationship between radial polydactyly and ZRS mutations.

In the Southwestern region of the Netherlands, an area known for the high rate of ZRS point mutation, triplication and radial polydactyly with triphalangism accounts for about one third of cases of radial polydactyly (13), while in other populations they account for around 7%. In a study of 5 generations of a family, Radhkrishna and coworkers strongly suggest that radial pol-

ydactyly, triphalangeal thumb and duplication of the big toe can be manifestations of the same autosomal dominant gene (14). It is likely that other factors modify the expression of this gene. In our case the patient had no deformity of the foot and no family history for any type of deformity. It is likely that a gene mutation (ZRS on 7q36 chromosome) is the cause of our patient's polydactyly.

Wassel's simple classification of radial polydactyly has been generally adopted. In 1978, Wood showed that many other forms of thumb polydactyly exist, including a double triphalangeal thumb sharing common metacarpal and different patterns of thumb polydactyly with triphalangeal components.

Wood type VIID designates thumb triplication with a triphalangeal component. Mennen described another variant of thumb triplication; all thumbs were fully developed but triphalangeal and shared an enlarged first metacarpal (15). An essential feature of the triple thumb deformity is that

the extra digits be opposable or partially opposable at birth. When the duplicated radial digits are not opposable and lie in the same plane as the other digits (the fingers), the deformity is referred to as the 6-fingered hand (16). Finally Hentz suggested that some cases of non-opposable triphalangeal radial polydactyly be classified as absent thumb with multiple duplication of the index, especially if the radial metacarpal shows a distal epiphyseal plate (the normal thumb metacarpal has a proximal epiphyseal plate) (17). This concept has not gained popularity, however, because epiphyseal anomalies of thumb metacarpals are not uncommon, including the presence of double epiphysis.

An essential feature of both classic and non-classic mirror hand is the presence of symmetry (although not necessarily perfect) around the midline. The hand deformity in our case lacked this essential feature.

On the basis of Wassel's classification and adapting Upton's nomenclature, Zuidam et al proposed a universal classification that included combined radial polydactyly, triphalangism, deformities, and symphalangism. They used "T" for triplication, "Tph" for triphalangism or a ray with three phalanges, "H" for a hypoplastic or floating ray, "D" for deviation of duplicates from the longitudinal axis, "S" for symphalangism, a condition with fusion of the joint. The location of the affected ray is designated as follows, "u" for an ulnar ray, "m" for a middle ray, and "r" for a radial ray (18).

The Zuidam's classification and nomenclature covers a broad range of manifestations of radial polydactyly, (including triplication, triphalangism, hypoplastic deformities, and symphalangism). Duplication up to trapezium (type VIII) is covered by this classification. In our case there was duplication of all radial-sided carpal bones, including the scaphoid and lunate bones as well as bifurcation of the radius bone. We propose the addition of type IX to designate duplication of all radial-sided carpal bones (trapezium, scaphoid and lunate), and type

X for bifurcation of the radius bone. Because in the case of tetraplicated thumb, radial, middle and ulnar side are not applicable, we propose the use of the numbers 1,2,3 and 4 from radial to ulnar thumbs. We further propose the use of "Te" to designate tetraplication.

Our case would thus be classified as type X Te, IV_u, II_m Tph_{2/4}, H_{1/3}.

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