A new documentation system for congenital absent digits

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Abstract

Background Congenital absent digits continue to be described by many confusing terms and are currently classified in categories I, V, and VI of the International Federation of Societies for Surgery of the Hand classification and seven subclassification systems. Very few classification systems provide any logical basis for surgical reconstruction. The purpose of this study was to develop a simple alphanumeric documentation system to reproducibly describe the morphological or radiographic appearance of congenital absent digits and facilitate communication of these children's hand anomalies from one hand surgeon to another.

Methods Dorsal and palmar photographs and PA radiographs of 235 hands in 204 children born with congenital absent digits over a 15-year period were analyzed to determine which digital rays were missing and their level of absence. Each hand can be described by three letters, R (radial), C (central), and U (ulnar), as well as numbers 1–5. The first letter and number designate which rays are missing and the second and third letters and numbers designate which rays remain present.

Results There are 15 morphological phenotypes of congenital absent digits. The three most common phenotypes are

U4R1 (a thumb but absence of all four fingers), R1U4 (absent thumb), and R5 (aplastic hand).

Conclusions This new documentation system allows hand surgeons to describe the simple morphological or radiographic appearance of congenital absent digits; incorporates all the previous subclassification systems that have attempted to describe congenital absent digits in radial, central, and ulnar deficiencies, symbrachydactyly, and congenital constriction ring syndrome; and has subsequently allowed the development of an algorithm which predicts whether conventional or microsurgical reconstruction is indicated for each specific phenotype.

Keywords Classification · Congenital hand anomalies · Radial deficiency · Transverse deficiency · Thumb deficiency · Constriction band syndrome · Symbrachydactyly

Introduction

Congenital absent digits may be described by many confusing terms including transverse absence, transverse arrest, transverse deficiency, radial longitudinal deficiency, radial club hand, cleft hand, split hand, central deficiency, central hand deficit, brachydactyly, symbrachydactyly, ectrodactyly, adactyly, ulnar deficiency, ulnar longitudinal deficiency, ulnar club hand, congenital constriction ring syndrome, and amniotic band syndrome. The classification accepted by most hand surgeons was originally proposed by Swanson et al. [28], who grouped congenital anomalies of the upper extremity into seven categories according to “the anatomical parts of the limb affected by certain embryological failures during development”—failure of formation, failure of differentiation, duplication, overgrowth, undergrowth, congenital constriction band syndrome, and generalized skeletal abnormalities. Flatt [11] described this classification as “properly conceived and usable.” The

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Swanson classification, subsequently adopted by the International Federation of Societies for Surgery of the Hand (IFSSH), classifies congenital absent digits into either category I—transverse or longitudinal failure of formation, category V—undergrowth, or category VI—congenital constriction ring syndrome [28]. Congenital absent digits may be due to a transverse deficiency (category I); a radial, central, or ulnar longitudinal deficiency (category I); symbrachydactyly (category V); or congenital constriction ring syndrome (category VI) and have been further described by seven subclassification systems—Blauth types IV and V thumb hypoplasia within the radial longitudinal deficiencies [3]; types I, II, and III of Nutt and Flatt's [21] classification of cleft hands; Ogino's [22, 23] classification of cleft hands; type V of Manske and Halikis' classification of central deficiency [17]; types B, C, D, and E of Ogino and Kato's classification of hand anomalies in ulnar longitudinal deficiencies [24]; grades 2, 3, and 4 of the Blauth and Gekeler classification of symbrachydactyly [4]; and type 4 of the Patterson classification of congenital constriction ring syndrome [27].

Unfortunately, very few of these classification systems and not even the Swanson classification are practical enough to allow surgeons to communicate a simple picture of the congenital hand anomaly to other surgeons and, furthermore, do not allow hand surgeons to compare the results of their reconstructions. A retrospective analysis of a large series of children with congenital absent digits, referred for both conventional and microsurgical reconstruction from 1995 to 2002, was subsequently continued as a prospective study from 2003, in order to develop a simple documentation system for children with congenital absent digits.

Materials and Methods

Children born with total or partial absence of one or more digits distal to the radiocarpal joint, referred to a single hand surgeon over a 15-year period (1995–2010) for consideration of conventional or microsurgical reconstruction, were included in this study if they had PA radiographs and dorsal and palmar photographs of their hands. The radiographs and photographs were analyzed to determine which digital rays were missing and their level of absence. Each hand can be described by three letters, R (radial), C (central), and U (ulnar), as well as numbers 1–5. The first letter and number designate which rays are missing and the second and third letters and numbers designate which rays are present.

A normal hand would be designated as R0. An absent thumb would be designated R1U4, equivalent to Blauth stage V thumb hypoplasia (Fig. 1) or type 4 congenital

constriction ring syndrome (Fig. 2). A hand with a thumb but absent fingers would be designated U4R1 (Fig. 3). Cleft hand would be designated as C1R2U2. Complete absence of all five digits would be designated as R5 (Fig. 4). The spectrum of radial deficiencies includes R1U4, R2U3, R3U2, and R4U1 (Fig. 5). The spectrum of transverse deficiencies and ulnar deficiencies includes U1R4, U2R3, U3R2, and U4R1 (Fig. 6). Central deficiencies include C1R2U2, C1R1U3, C1R3U1, C2R1U2, C2R2U1, and C3R1U1 (Fig. 7). There are 15 different morphological phenotypes of congenital absent digits.

This documentation system can be further refined, if deemed necessary by the individual surgeon, by describing the level of absence of the rays as:

- w—from the radiocarpal joint to the carpometacarpal joints
- m—distal to the carpometacarpal joints to just distal to the metacarpophalangeal joints
- p—distal to the metacarpophalangeal joints out to the proximal third of the middle phalanx of the fingers or the tip of the thumb
- d—distal to the proximal third of the middle phalanx of the fingers out to the tip of the fingers (Fig. 8 and Table 1)

This documentation system was developed after retrospective analysis of children referred from 1995 to 2002 and has since been used prospectively on all children referred from 2003 to 2010.

Results

Two hundred thirty-five hands in 204 children met the inclusion criteria for this study. Diagnoses based on the IFSSH classification system included 54 radial longitudinal deficiencies, 51 symbrachydactylies, 48 cleft hands, 37 congenital constriction ring syndromes, 36 transverse failures of formation, and 9 ulnar longitudinal deficiencies. Confusion between cleft hand and symbrachydactyly may have led to increased diagnoses of cleft hand compared with symbrachydactyly.

Absent thumb, the R1U4 phenotype occurred in 44 hands. The more severe radial deficiencies R2U3, R3U2, and R4U1 occurred in 4, 17, and 8 hands, respectively. The most common phenotype of congenital absent digits seen in 57 hands was a thumb but absence of all four fingers, classified as U4R1, corresponding to a severe transverse deficiency or grade 3 symbrachydactyly [4]. Transverse or longitudinal deficiencies of the ulnar rays occurred much less frequently—the U1R4 phenotype was only seen in 1 hand and the U2R3 and U3R2 phenotypes in 4 and 11 hands, respectively.

Fig. 1 **a** Photograph, **b** radiograph, and **c** diagrammatic documentation of a R1U4(w) hand phenotype

There were 60 central deficiencies of which six different phenotypes are possible. True cleft hand designated as a C1R2U2 phenotype was seen in 19 hands (Fig. 9). Cleft hands with a single missing index finger (C1R1U3) or a single missing ring finger (C1R3U1) were never seen. Absence of the index, middle, and ring fingers, classified as the C3R1U1 phenotype (Fig. 10) and corresponding to grade 2 atypical cleft hand symbrachydactyly [4], was seen in 24 hands, whereas absence of the index and middle rays, the C2R1U2 phenotype, and absence of the middle and ring finger rays, the C2R2U1 phenotype, occurred less frequently in 11 and 6 hands, respectively. Finally, complete absence of all five digits designated as R5 occurred in 29 hands.

Five morphological phenotypes accounted for 173 of the 235 hands (73.6 %). The two most common phenotypes were 57 U4R1 hands and 44 R1U4 hands. The aplastic hand R5 (29) and the central deficiencies C3R1U1 (24) and C1R2U2 (19) were the next most common phenotypes (Table 1).

This simple documentation system has allowed the subsequent development of an algorithm to predict which conventional or microsurgical technique is indicated for each specific morphological phenotype of congenital absent digits. Of the 15 phenotypes, only seven (R1U4, R2U3, R3U2, R4U1, R5, U4R1, and occasionally C3R1U1) are suitable

indications for either conventional reconstruction by pollicization or distraction lengthening, or microsurgical reconstruction by toe-to-hand transfers.

Discussion

The ideal classification system for congenital anomalies of the hand “should provide a simple description of the appearance of the hand to enhance communication between hand surgeons, and at the same time incorporate the embryological mechanism, as well as dictate the surgical reconstruction and predict the prognosis” [18]. Unlike most other classifications, our documentation system provides a simple description of either the morphological or radiographic appearance or functional status of a child’s hand to facilitate communication between physicians, but it does not attempt to imply any underlying embryological causation, although it does incorporate all the previous subclassification systems. Our study cohort of 235 hands is even larger than Swanson’s original study and all other classification systems [7, 14, 25], except for one study of 321 hands by De Smet [9]. The most recent prospective study of 585 upper limb anomalies in Sweden [10] only included 130 hands with absent digits.

Fig. 2 **a** Photograph, **b** radiograph, and **c** diagrammatic documentation of a R1U4(m) hand phenotype

Fig. 3 **a** Photograph, **b** radiograph, and **c** diagrammatic documentation of a U4R1(w) hand phenotype

Many hand surgeons experience difficulty classifying congenital hand differences according to the Swanson system. A large study of 650 children including 321 hands within categories I, V, and IV reported that 15 % of anomalies could not be classified by the current IFSSH classification [9]. The Japanese Society for Surgery of the Hand (JSSH) has even suggested adding a category of “unclassifiable cases.” Tonkin [31] has argued that a classification system based on causation is inappropriate and proposed an alternative system based purely on the location and morphological features of the anomaly, which is precisely what our documentation system provides.

Within the IFSSH classification [28], there are several individual subclassification systems that describe congenital absent digits. Category I failure of formation may be subdivided into longitudinal and transverse deficiencies. Longitudinal deficiencies are further subdivided into radial, central, and ulnar deficiencies. Bayne and Klug’s classification of radial deficiencies [2] is based on the radiographic appearance or absence of the radius, but James et al. [12] expanded this classification to include type N—normal radius, normal carpus but hypoplastic thumb, and type O—normal radius, radial-sided carpal abnormality, and hypoplastic thumb. Should hypoplastic thumb be classified as a

radial longitudinal deficiency (category I) or as undergrowth (category V)? The Blauth classification [3] certainly helps guide surgical reconstruction of hypoplastic thumbs. Blauth types IV and V floating and absent thumbs correspond to our R1U4 phenotype.

Many physicians have difficulty distinguishing between transverse deficiency, symbrachydactyly, and constriction ring syndrome. The first controversial issue with the IFSSH classification relates to the terminology of cleft hand and symbrachydactyly. Multiple classification systems have been proposed for cleft hand which has usually been considered as a central longitudinal deficiency within category I. Barsky [1] originally differentiated typical and atypical cleft hands. Maisels [15] proposed a “centripetal suppression theory” in which cleft hand results from an insult to the developing hand plate, initially with the absence of the middle finger, followed by progressive suppression of the radial rays, eventually leaving a small finger, corresponding exactly to our C1R2U2, C2R1U2, C3R1U1 and eventually R4U1 phenotypes. Watari and Tsuge [32] postulated that cleft hand was due to polydactyly of the middle finger. The radial component of the middle finger combines with the index finger and the ulnar component of the middle finger combines with the ring finger resulting in an absent middle

Fig. 4 **a** Photograph, **b** radiograph, and **c** diagrammatic documentation of a R5(w) hand phenotype

Fig. 5 Diagrammatic representation of **a** the morphological spectrum of radial deficiencies and **b** the radiographic spectrum of radial deficiencies

finger. Nutt and Platt [21] proposed the term “congenital central hand deficit” with four groups based on the number of absent metacarpal rays and phalanges and the extent of their involvement. Group I had one absent ray, group II had two absent rays, and group III had three absent rays. Involvement seemed to spread predictably from the middle finger phalanges to the middle finger metacarpal, then the index finger phalanges to the index finger metacarpal, and finally the ring finger phalanges to the ring finger metacarpal, which corresponds exactly with our proposed phenotypes C1R2U2 progressing to C2R1U2 and finally to C3R1U1. Tada et al. [30] argued that cleft hand should remain as a central longitudinal deficiency within category I failure of formation, since the single digit in a cleft hand always represents the small finger and single digits are never found on the radial side in cleft hand—the most extreme form of a cleft hand always corresponds to a R4U1 phenotype. Ogino [22] demonstrated experimentally that typical cleft hand, central polydactyly, and syndactyly are induced by the same teratogenic mechanism and proposed a classification of typical cleft hand based on the number of missing finger rays [23]—missing one finger usually the middle finger, missing two fingers either the index and middle fingers or the middle and ring fingers, and missing three fingers. Consequently, the JSSH has argued that typical cleft hand is not a central longitudinal deficiency at all and has

reclassified typical cleft hand, central polydactyly, and syndactyly in a separate category IV as an “abnormal induction of finger rays.” Finally, Manske and Halikis [17] have proposed classifying central deficiencies based on progressive narrowing of the thumb–index finger web space to provide a logical basis for reconstruction. All these subclassifications of cleft hand correspond exactly with our proposed phenotypes C1R2U2 progressing to C2R1U2 or C2R2U1 and finally to C3R1U1.

It has been proposed that the description “typical cleft hand” be replaced by the term “central longitudinal deficiency” and the term “atypical cleft hand” has been reclassified as grade 2 oligodactylic symbrachydactyly [4, 5, 16]. However, this reclassification does not seem to correlate with the embryology—reduction of rays in cleft hand proceeds radially leaving a single small finger corresponding to a R4U1 phenotype. Conversely, the “reduction theory” of symbrachydactyly [4, 6] postulates that hypoplastic or absent middle phalanges progress proximally, so that the distal phalanges only remain as nubbins with rudimentary nails (grade 1). Further progression results in the absence of the proximal and middle phalanges of the index, middle, and ring fingers—grade 2 oligodactylic symbrachydactyly previously described as “atypical cleft hand,” which corresponds to our C3R1U1 phenotype. Reduction proceeds ulnarly from the central three fingers to absence of all four

Fig. 6 Diagrammatic representation of **a** the morphological spectrum of transverse and ulnar deficiencies and **b** the radiographic spectrum of transverse and ulnar deficiencies

fingers but leaving the thumb, resulting in grade 3 monodactylic symbrachydactyly, and finally involves the thumb resulting in grade 4 peromelic symbrachydactyly, with absence of all five digits leaving only nubbins or nail remnants, which corresponds to our U4R1 and R5 phenotypes. The problem with incorporating atypical cleft hand in symbrachydactyly is that based on the published literature, reduction of rays in symbrachydactyly proceeds ulnarly from the central three digits leaving only a thumb (grade 3 monodactylic symbrachydactyly), corresponding to our U4R1 phenotype, whereas in cleft hand, reduction proceeds radially leaving only a small finger, corresponding to our R4U1 phenotype.

A second controversial issue is whether symbrachydactyly is classified in category I [13, 20, 26] or in category V [9, 16] of the IFSSH classification. The two transverse anomalies—transverse deficiency and symbrachydactyly—are classified in different categories: transverse deficiency in category I failure of formation and symbrachydactyly in category V undergrowth. However, it is becoming accepted that symbrachydactyly is a distal manifestation of transverse deficiency, whereas the acarpia and congenital forearm amputation forms of transverse arrest are proximal

manifestations of transverse deficiency. Ogino et al. [26] argue that grade 1 symbrachydactyly is a mild form of an intercalary transverse deficiency, whereas grades 2–4 represent terminal transverse deficiencies. Kallemeier et al. [13] argued the opposite theory that transverse deficiency of the forearm represents a more proximal manifestation of symbrachydactyly after finding manifestations of symbrachydactyly—nubbins and skin invagination in 93 % of transverse deficiencies of the forearm. Yamauchi and Tanabu's [33] seven-stage classification of symbrachydactyly, with stage 7 being the most severe—a forearm amputation—is compatible with Kallemeier's hypothesis. The JSSH considers symbrachydactyly to be synonymous with transverse failure of formation and therefore believes that symbrachydactyly should be moved to category I of the IFSSH classification. However, this means combining transverse deficiencies in which the terminal elements are missing, with symbrachydactyly, in which the initial deficiency is hypoplasia of the middle phalanges and the terminal elements remain.

Congenital absent digits may also be due to congenital constriction ring syndrome which is classified in a separate

Fig. 7 Diagrammatic representation of the radiographic spectrum of central deficiencies

Fig. 8 Levels of congenital absence of the fingers and thumb

Table 1 Phenotypes and levels of absence of congenital absent digits in 235 hands

Classification	Level of missing digits				Total
	w	m	p	d	
R1U4	27	13	4	—	44
R2U3	—	2	2	—	4
R3U2	1	12	4	—	17
R4U1	2	5	1	—	8
R5	7	20	2	—	29
U4R1	10	28	18	1	57
U3R2	1	1	9	—	11
U2R3	4	—	—	—	4
U1R4	—	1	—	—	1
C1R2U2	11	7	1	—	19
C2R1U2	3	2	4	2	11
C2R2U1	1	2	2	1	6
C3R1U1	6	11	6	1	24
					235

w—from the radiocarpal joint to the carpometacarpal joints; m—distal to the carpometacarpal joints to just distal to the metacarpophalangeal joints; p—distal to the metacarpophalangeal joints out to the proximal third of the middle phalanx of the fingers or the tip of the thumb; d—distal to the proximal third of the middle phalanx of the fingers out to the tip of the fingers

Fig. 9 **a** Photograph, **b** radiograph, and **c** diagrammatic documentation of a C1R2U2 (m) central deficiency phenotype

category VI of the IFSSH classification system. Patterson [27] divided congenital constriction ring syndrome into four subtypes, with type 4 describing amputation of the digits.

Ulnar longitudinal deficiencies are usually classified on the radiographic appearance of either the ulna or the elbow joint and absent digits are rarely addressed [19, 29]. Miller et al. [19] reported that 56 % of ulnar deficiencies had only three digits, 21 % had only two digits, and 41 % had a hypoplastic or absent thumb. Ogino and Kato [24] classified ulnar deficiencies according to the number of absent digits, proceeding sequentially from ulnar to radial—type B had absence of the small finger; type C had absence of the ring and small fingers; type D had absence of the middle, ring, and small fingers; and type E had absence of the index, middle, ring, and small fingers. Types B–E correspond exactly with our U1R4, U2R3, U3R2, and U4R1 phenotypes. Cole and Manske [8] also reported 90 absent digits in 55 ulnar deficiencies—38 absent small fingers, 30 ring fingers, 10 middle fingers, 2 index fingers, and 10 absent thumbs. All these missing digit configurations correlate exactly with our U1R4, U2R3, U3R2, and U4R1 phenotypes.

Current classification systems and even the IFSSH classification are based on theoretical embryological mechanisms and do not allow a pediatric hand surgeon to communicate a simple reproducible description of a child's hand to other

physicians. Very few classification systems provide any logical basis for surgical reconstruction. The primary purpose of this study was therefore to simplify the documentation of congenital absent digits, currently classified in three different categories of the IFSSH system, as well as by seven different subclassification systems, based on a large cohort of children. The documentation system relies purely on the morphological or radiographic appearance or functional status of a child's hand. The two or three letter–number combination then allows very easy “short-hand” communication of a child's congenital hand anomaly from one surgeon to another. It illustrates perfectly De Smet's concept that “an ideal classification system should allow all physicians and investigators to speak the same language” [9] and is analogous to being able to describe the exact location and geometry of a hand fracture over the telephone to another physician. Our system does not imply any embryological mechanism and therefore renders many of the arguments regarding causation, which incidentally have never been proven in humans, completely superfluous. It also incorporates all the other subclassification systems that have attempted to describe congenital absent digits in radial, central, and ulnar deficiencies, symbrachydactyly, and congenital constriction ring syndrome.

Another major advantage of this documentation system is that it has allowed the development of an algorithm, which predicts which type of surgical reconstruction—whether

Fig. 10 **a** Photograph, **b** radiograph, and **c** diagrammatic documentation of a C3R1U1(m) central deficiency phenotype

conventional or microsurgical, will provide optimal hand function. For example, an absent thumb at the wrist level R1U4(w) is best reconstructed by index finger pollicization, whereas an absent thumb with an intact carpometacarpal joint and remnant of thenar muscles R1U4(m) or R1U4(p) is best reconstructed by microsurgical toe transfer or distraction lengthening. This algorithm will be described later in a separate report.

Conflict of interest The authors declare that they have no conflicts of interest, commercial associations, or intent of financial gain regarding this research.

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