Polydactyly of the Hand

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Abstract

Polydactyly is considered either the most or second most (after syndactyly) common congenital hand abnormality. Polydactyly is not simply a duplication; the anatomy is abnormal with hypoplastic structures, abnormally contoured joints, and anomalous tendon and ligament insertions. There are many ways to classify polydactyly, and surgical options range from simple excision to complicated bone, ligament, and tendon realignments. The prevalence of polydactyly makes it important for orthopedic surgeons to understand the basic tenets of the abnormality.

Polydactyly is the presence of extra digits. Its incidence is likely underestimated because many practitioners treat simple "nubbins" without referring them to orthopedic specialists.[1-3] Polydactyly can be detected by ultrasound as early as 14 weeks' gestational age, with partial autoamputation seen in most isolated polydactylies.[4] The thumb, responsible for 40% of hand function, must be able to oppose the other digits with a stable pinch.[5] Polydactyly encumbers this motion when the duplicated digits deviate from normal alignment. Ezaki[6] noted that the anatomy is better described as "split" than "duplicated." There are many dichotomous ways to classify polydactyly: preaxial (radial) versus postaxial (ulnar), thumb versus triphalangeal, simple versus complex (**Figure 1**). Mixed polydactyly is defined as the presence of preaxial and postaxial polydactyly.[7] Surgical management seeks to allow normal hand function and to restore cosmesis.

Epidemiology

Sun and colleagues[8] reported the overall polydactyly incidence as 2 per 1000 live births in China from 1998 to 2009, with a slight male predominance; polydactyly was also 3 times more common than syndactyly in this population. Ivy,[9] in a 5-year audit of Pennsylvania Department of Health records, found polydactyly to be the fourth most common congenital anomaly after clubfoot, cleft lip/palate, and spina bifida. Thumb duplication occurs in 0.08 to 1.4 per 1000 live births and is more common in American Indians and Asians than in other races.[5,10] It occurs in a male-to-female ratio of 2.5 to 1 and is most often unilateral.[5] Postaxial polydactyly is predominant in black infants; it is most often inherited in an autosomal dominant fashion, if isolated, or in an autosomal recessive pattern, if syndromic.[1] A prospective San Diego study of 11,161 newborns found postaxial type B polydactyly in 1 per 531 live births (1 per 143 black infants, 1 per 1339 white infants); 76% of cases were bilateral, and 86% had a positive family history.[3] In patients of non-African descent, it is associated with anomalies in other organs. Central duplication is rare and often autosomal dominant.[5,10]

Genetics and Development

As early as 1896, the heritability of polydactyly was noted.[11] As of 2010, polydactyly has been associated with 310 diseases.[12] Ninety-nine genes, most involved in regulation of anterior-posterior formation of the limb bud, have been implicated.[12,13]

The upper limb begins to form at day 26 in utero.[14] Apoptosis in the interdigital necrotic zones results in the formation of individual digits. It is presumed that, in polydactyly, the involved tissue is hypoplastic because of an abnormal interaction between mesoderm and ectoderm.[5] Presence of an apical ectodermal ridge determines the formation of a limb bud, and on it the zone of polarizing activity (ZPA) dictates preaxial and postaxial alignment.[14,15] The ZPA is located on the posterior zone of the developing limb bud. The levels of GLI3, a zinc finger-containing DNA-binding protein, are highest in the anterior area, and HAND2, a basic helix-loop-helix DNA-binding protein, is found in the ZPA. This polarity promotes

Figure 1. Varieties of polydactyly presentations: (A) thumb triphalangism with delta phalanx, (B) synpolydactyly (extra digit within syndactyly), (C) pedunculated polydactyly. Photographs courtesy of Dr. Oakes.

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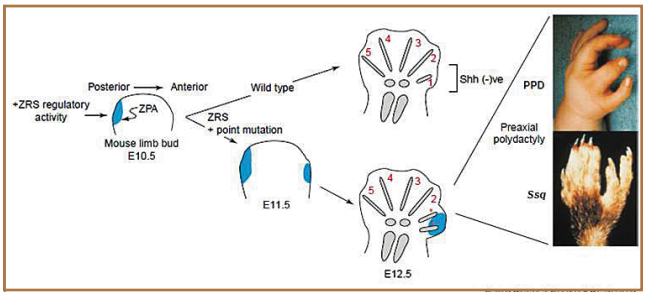

Figure 2. Development of preaxial polydactyly (PPD). Reprinted from *Current Opinion in Genetics & Development*, vol. 15, Lettice LA and Hill RE, "Preaxial polydactyly: a model for defective long-range regulation in congenital abnormalities," pages 294-300, Copyright 2005, with permission from Elsevier.

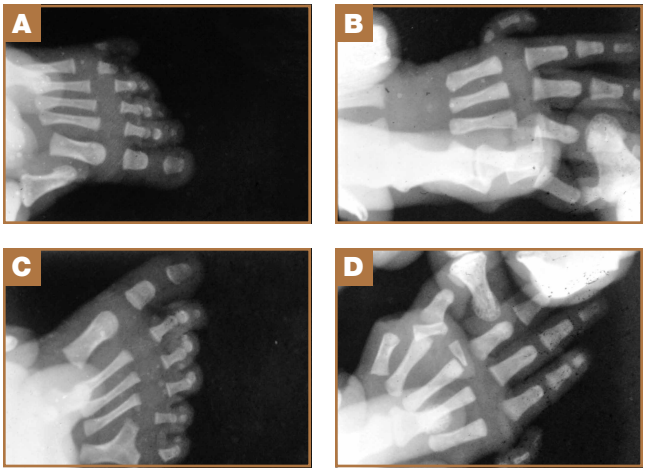

Figure 3. Radiographs of polydactylous hands and feet. If polydactyly is detected, full physical examination is required. Photograph from files of Daniel Riordan, MD, courtesy of Dr. DC Faust.

sonic hedgehog (Shh) gene expression in the posterior region, which in turn prevents GLI3 cleavage into its repressed form. GLI3R (repressed) and GLI3A (active) concentrations are highest, therefore, in the anterior and posterior portions of the bud, respectively. The GLI3A:GLI3R ratio is responsible for the identity and number of digits in the hand (ie, the thumb develops in regions of high GLI3R). GLI and Shh mutations lead to polydactylous hands with absent thumbs (**Figure 2**).[16]

Ciliopathies have also been shown to cause postaxial polydactyly, possibly because of the role that nonmotile cilia play in hedgehog signaling.[17] Mutations in Shh genomic regulators cause preaxial polydactyly.[18] HoxD activates Shh in the ZPA; HoxD13 mutations are associated with synpolydactyly.[16,19] In each of these mutations, Shh production is altered, and some form of polydactyly results.

Associations

Many syndromes have been associated with polydactyly. Not all polydactyly is associated with other disorders, but the more complex the polydactyly, the more likely that other anomalies are present. Every patient who presents with polydactyly should have a full history taken and a physical examination performed (**Figure 3**). Any patient with syndromic findings or atypical presentations (eg, triphalangism, postaxial polydactyly in a patient of non-African descent, central and index polydactyly) should be referred to a geneticist.

Classifications

The Wassel[20] classification describes the anatomical presentation of thumb duplication on the basis of 70 cases in Iowa (**Figures 4, 5**; **Table 1**). Because some duplications fall outside the Wassel classification, many researchers have proposed modifications (**Figure 6**).[21-25]

The Temtamy and McKusick[10] classification, which is the product of geneticists, classifies duplications by grouping genetically related presentations (**Table 2**). It provides the most commonly used postaxial classification, with type A being a fully developed digit and type B a rudimentary and pedunculated digit, informally referred to as a *nubbin*. Type B is more common than type A. Given inheritance patterns, it is assumed that type A is likely multifactorial and type B autosomal dominant.[10] Thumb polydactyly inheritance is still unclear. The other types of preaxial polydactyly and high degrees of polydactyly are rare but seem to be passed on in an autosomal dominant fashion on pedigree analysis.[10]

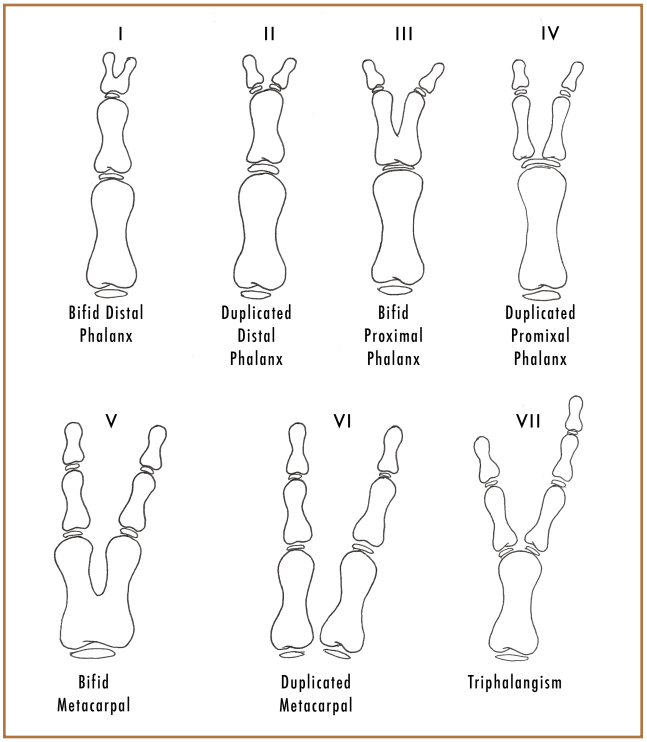

Figure 4. Wassel classification. Drawing courtesy of Ray Kimbrough.

The Stelling and Turek classification presents the duplication from a tissue perspective: Type I duplication is a rudimentary mass devoid of other tissue elements; type II is a subtotal

Figure 5. Preaxial polydactyly clinical examples. Photographs (A) courtesy of Dr. Oakes, (B–D) from files of Daniel Riordan, MD, courtesy of Dr. DC Faust, (E) courtesy of Dr. DC Faust.

Table 1. **Wassel Classification**

Type	Description	% of Polydactyly[a]
I	Bifid distal phalanx	2%-6%
II	Duplicate distal phalanx	15%-17%
III	Bifid proximal phalanx	6%-9%
IV	Duplicate proximal phalanx	43%-46%
V	Bifid metacarpal	10%-12%
VI	Duplicate metacarpal	3%-4%
VII	Triphalangism	6%-20%

[a]Data were derived from Cohen.[5]

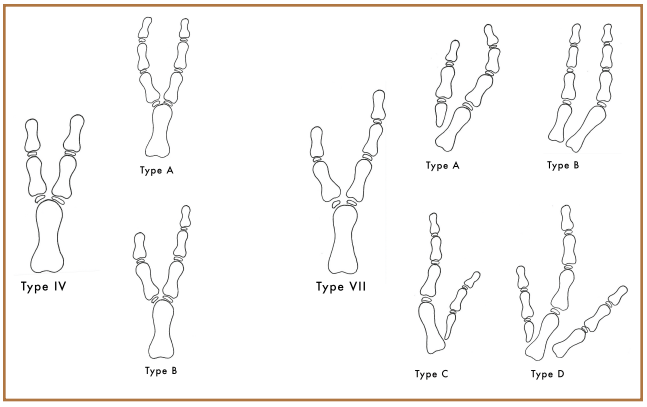

Figure 6. Wood modification of Wassel classification. Drawing courtesy of Ray Kimbrough.

duplication with some normal structures; and type III is a duplication of the entire "osteoarticular column," including the metacarpal.[1] It is interesting to note that histology of type I duplications shows neuroma-like tissue.[26-28] Again, *normal* is a relative term because, in polydactyly, the duplications are hypoplastic and deviated, with anomalous anatomy.

The Rayan classification describes ulnar polydactyly and was derived from a case study series of 148 patients in Oklahoma (**Table 3**).[29]

There are also some complex polydactylies that are not easily classified: ulnar dimelia, cleft hand, pentadactyly, and hyperphalangism. Ulnar dimelia, also known as "mirror hand," is typically 7 digits with no thumb, but other variations are seen. The radius is often absent, and the elbow is abnormal. There is some debate about whether it is a fusion of 2 hands. Pentadactyly, or the 5-fingered hand, appears as 5 triphalangeal digits with no thumb (**Figure 7**).

Isolated thumb triphalangism might appear similar to pentadactyly. Miura[30,31] pointed out that the radial digit in the pentadactylous hand may be opposable (thumb-like) or nonopposable; in his studies, the patients with the opposable thumb had a metacarpal with a proximal epiphysis (**Figure 8**). Consequently, the triphalangeal thumb metacarpal with a distal epiphysis is true pentadactyly, whereas that with a proximal epiphysis is hyperphalangism (**Figure 9**). Treatment of these complex polydactylies involves the same underlying principles as for preaxial and postaxial polydactyly, albeit with additional proximal upper extremity considerations.

Table 2. **Temtamy and McKusick Classification[a]**

Polydactyly	Type	Description
Postaxial	Type A	Fully developed extra digit
	Type B	Rudimentary extra digit, or pedunculated postminimi
Preaxial	Type 1	Thumb polydactyly
	Type 2	Polydactyly of triphalangeal thumb
	Type 3	Polydactyly of index finger
	Type 4	Polysyndactyly
High degree	Tibial defect with preaxial polydactyly	
	Agenesis of tibia and mirror foot	
	Ulnar and fibular dimelia	
	Other types of duplication of limbs	

[a]Derived from Temtamy and McKusick.[10]

Table 3. **Rayan Classification of Ulnar Polydactyly**

Rayan Classification	Description
I	Cutaneous "nubbin"
II	Pedunculated digit (most common)
III	Articulating digit with fifth metacarpal
IV	Fully developed digit with sixth metacarpal
V	Polysyndactyly (least common)

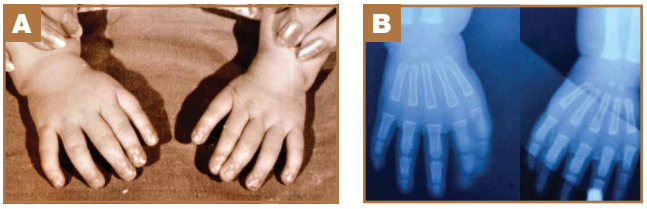

Figure 7. Pentadactyly clinical photographs and radiographs from files of Daniel Riordan, MD, courtesy of Dr. DC Faust.

When to Operate (Timing)

Ezaki[6] recommended surgical intervention at age 6 to 9 months, before fine motor skills have developed with the abnormal anatomy. Cortical learning occurs as the child begins prehensile activities before 6 months, but the risks of anesthesia outweigh functional benefits until the child is older. Waiting until 1 year of age is not uncommon, though surgery at an earlier age may be beneficial if the polydactyly affects hand function.[32] It is not uncommon to wait with the more balanced thumb polydactylies to assess thumb function. Hypoplasia might also delay surgical intervention until there is enough tissue inventory for reconstruction. Wassel[20] noted that surgical intervention ideally occurs before the supernumerary elements displace the normal elements, as tends to happen with growth. Suture ligation is an option in the neonatal unit for some pedunculated digits.[33] Studies have shown satisfactory results in adults treated for polydactyly, if the patient presents later than expected.[34]

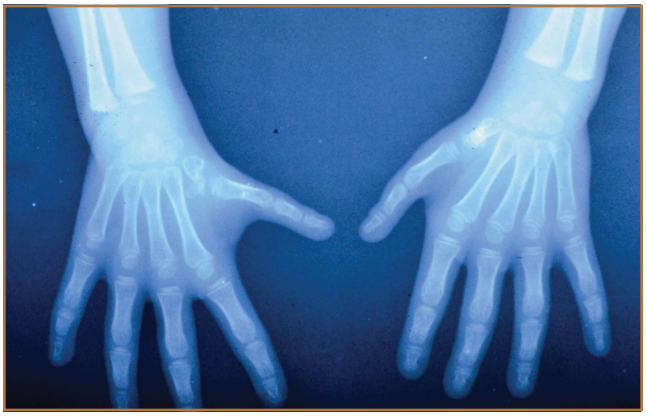

Figure 8. Thumb triphalangism, contrasting with pentadactyly (Figure 7). According to Miura,[30] this is not pentadactyly because metacarpal epiphysis is proximal. Radiograph from files of Daniel Riordan, MD.

Surgical Considerations

Knavel recommended simple excision, stating that "ablation requires no ingenuity and creates no problems."[5] This belief, though true for some duplications, will not lead to the best outcome for more complex polydactylies. The goal of surgery is a stable and well-aligned thumb for pinch and prehensile activity, as well as a cosmetically pleasing hand. Incisions should not be made linearly along the axis of the digit, as the scar will cause deviation with growth.[24]

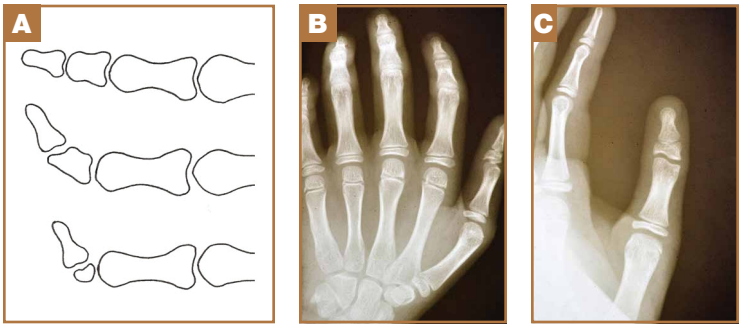

Figure 9. (A) Delta phalanx types in hyperphalangism, which most often occurs in the thumb.[23] Drawing courtesy of Ray Kimbrough. (B, C) Radiographs show delta phalanx in skeletally immature patient. From files of Daniel Riordan, MD, courtesy of Dr. DC Faust. See Figure 1A for a clinical photograph of delta phalangism of the thumb.

Wassel type I polydactyly might appear incidentally as a broad thumb, in which case it requires no intervention (**Figure 10**). However, in Wassel types I and II polydactyly with deformity, the Bilhaut-Cloquet procedure is useful for both bifid and duplicated phalanges (**Figure 11**).[5,6,30,32,35] Collateral ligaments may need to be released in type II because of difficulty in opposing the tissue. Cosmetic results with Bilhaut-Cloquet are unpredictable. The original technique required symmetrically sized digits; results today have been improved with microtechniques and preservation of an entire nail.[36] Another option is ablation of the more hypoplastic osseous element and soft-tissue augmentation of the residual digit. The theme of ablation and augmentation is seen throughout the literature for the surgical treatment of polydactyly (**Figure 12**).[1]

For type III polydactyly, the bifid proximal phalanx is narrowed by resection and realigned with osteotomy of the remaining diaphysis. Type IV polydactyly, the most common thumb duplication, often requires advancement of the abductor pollicis brevis to the base of the proximal phalanx to aid in metacarpophalangeal (MCP) stabilization, abduction, and opposition. The metacarpal head, if broad and with 2 facets, can be shaped to form a single articulating surface. The metacarpal, occasionally with the proximal phalanx, often requires realignment by closing wedge osteotomy. Last, tendons on the

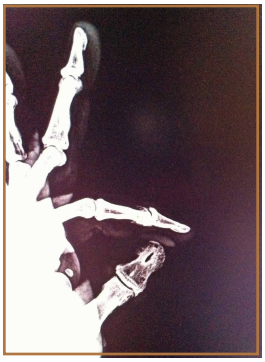

Figure 10. Incidental finding of Wassel type 1 thumb polydactyly. Radiograph courtesy of Dr. DC Faust.

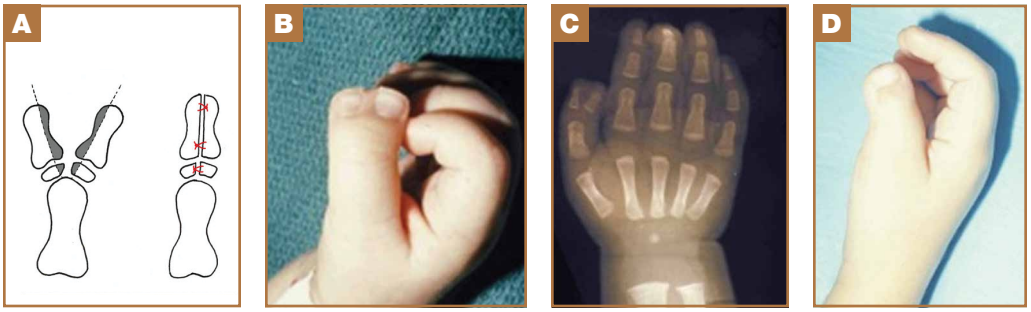

Figure 11. (A) Bilhaut-Cloquet procedure. Drawing courtesy of Ray Kimbrough. (B) Preoperative clinical photograph of Wassel type 2 polydactyly. (C) Radiograph of same hand. (D) Broad thumb after Bilhaut-Cloquet surgical treatment. Photograph courtesy of Dr. DC Faust.

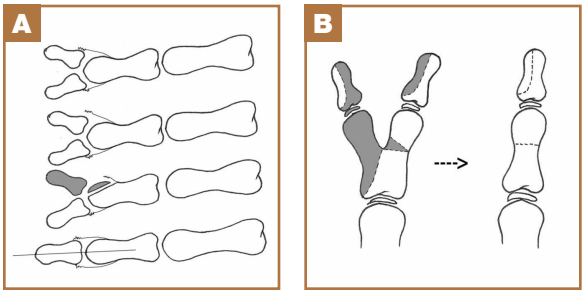

Figure 12. Ablation and augmentation procedures. Drawings courtesy of Ray Kimbrough. Adapted from *Journal of Hand Surgery*, vol. 14, Manske PR, "Treatment of duplicate thumb using a ligamentous/periosteal flap," pages 728-733, Copyright 1989, with permission from Elsevier.

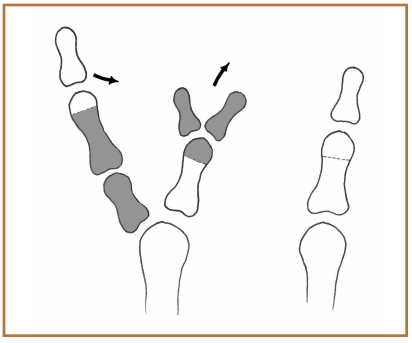

Figure 13. On-top plasty (acral transposition). Drawing courtesy of Ray Kimbrough.

resected bony elements should be rebalanced on the remaining digit, and anomalous slips must be released. For instance, given a radial insertion of the long flexor tendon on the distal phalanx, the tendon should be moved centrally. A strong flexor or extensor tendon on the amputated digit should be transferred to the remaining digit.[24]

Types V and VI are treated similarly to type IV, with the addition of a first web space Z-plasty or web widening if there is thenar eminence contracture. Acral transposition has also been described, with transposition of the tip of the ablated digit in place of the tip of the kept digit; this technique is ideal if one digit has a more normal proximal part while the other has a more normal distal part (**Figure 13**).[35]

Type VII thumb polydactyly, the type most likely inherited and associated with other disorders, should be treated like type VI. The nail should be preserved; amputation of the distal phalanx is not advised. Resection of the delta phalanx or 1 interphalangeal (IP) joint is an option. Articular surfaces will remodel if done before the age of 1 year. If the thenar eminence is hypoplastic, then Huber transfer of the abductor digiti minimi should be considered.[37] Resection of the triphalangeal thumb is also advised, even if the biphalangeal thumb is more hypoplastic, with transfer of the ligaments and tendons, as described earlier.[5,6,24,30,32,35]

Thumb triphalangism, if isolated, and hyperphalangism in the other digits, can be treated with resection of the delta phalanx or one of the IP joints if it is affecting function or cosmesis.[1,6] Wood and Flatt[23] recommended early resection of a thumb delta phalanx because of the likelihood of deviation that impedes thumb function. For children, they recommended delta phalanx resection and Kirschner wire fixation for 6 weeks; for adults, they recommended resection or fusion of the joint, with osteotomy as needed for deviation.[23,24] For thumb triphalangism, multiple surgeries are the norm, as Wood[24] reported in his study of 21 patients who underwent 78 operations in total.

Index polydactyly may present as a simple pedunculated skin tag, which can be simply excised, or as a more complex musculoskeletal duplication. More complex presentations can be treated with procedures similar to those used for the thumb. Typically, the additional digit is radially deviated and angulated, eventually leading to impingement of thumb pinch and the first web space. Ray amputation is also an option if no reconstructive surgery will produce the stable, sensate radial pinch that is essential to hand function.[32]

Ring-finger polydactyly and long-finger polydactyly are often complicated by some element of syndactyly, resulting in a relative paucity of skin (**Figure 14**). There is failure of both formation (hypoplasia) and differentiation (syndactyly). The hypoplasia particularly affects the function of these digits by tethering them; multiple surgeries to restore proper hand function are the norm.[1] Reconstructive surgery for these digits requires preoperative tissue inventory followed by resection and augmentation; as in syndactyly, skin for coverage is at a premium. Creation of a 3-fingered hand is an option.[23]

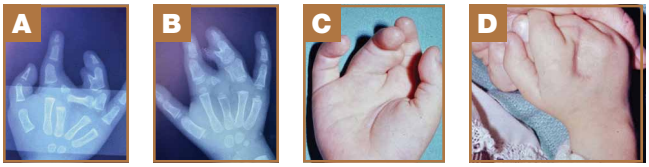

Figure 14. (A) Preoperative and (B) postoperative radiographs of cleft hand with associated polydactyly. (C) Preoperative clinical photograph. (D) Postoperative clinical photographs show improved web-space depth but persistent contracture and deformity. From files of Daniel Riordan, MD, courtesy of Dr. DC Faust.

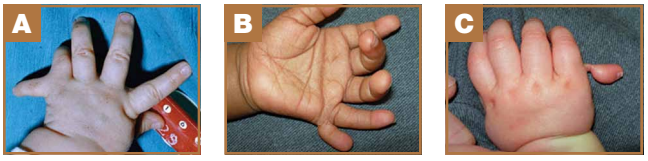

Figure 15. (A,B) Postaxial polydactyly examples. In older patients, broad-stalked duplication should be excised surgically. (C) Typical "nubbin" that can be treated with suture ligation. Photographs courtesy of Dr. DC Faust.

Temtamy and McKusick[10] type A little-finger polydactyly is treated similarly to the thumb, with the caveat that hypothenar and intrinsic muscles that insert on the resected little finger are transferred to the remaining digit. In contrast to thumb polydactyly, the extrinsic musculature tends to be in good position. Suture ligation of type B polydactyly, as described by Flatt, is likely more common than orthopedists appreciate, as pediatricians and neonatal unit practitioners commonly perform this procedure in the nursery.[1-3] It has been described with 2-0 Vicryl[3] (Ethicon, Somerville, New Jersey) and 4-0 silk sutures,[32] with the goal of necrosis and autoamputation. Parents should be told the finger generally falls off about 10 days (range, 4-21 days) after ligation.[3] Multiple authors have cited a report of exsanguination from suture ligation, but we could not locate the primary source. It is advisable to wait until a patient is 6 months of age if planning to resect the nubbin in the operating room, given the anesthesia risk and the lack of functional impairment. Katz and Linder[33] indicated they remove type B polydactyly in the nursery suite used for circumcisions; they use anesthetizing cream on the skin, and sharp excision with a scalpel, followed by direct pressure and Steri-Strip (3M, St. Paul, Minnesota) application. Suture ligation is recommended only if there is a narrow, thin (<2 mm) soft-tissue stalk; any broad or bony stalk necessitates surgical removal to avoid neuroma formation and failure of autonecrosis (**Figure 15**).[27] Other options are a single swipe of a scalpel and elliptical excision; sharp transaction of the digital nerve with subsequent retraction is advised to avoid neuroma formation.[2]

Barton described ulnar dimelia operations as "spare parts surgery."[1] Extra digits are ablated and a thumb created (**Figure 16**). The hand might have a digit in relatively good rotational position for thumbplasty, or the principles of pollicization may need to be used. If the patient is already using the hand,

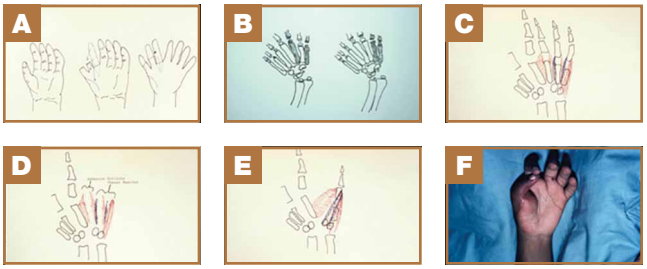

Figure 16. Ulnar dimelia surgical planning and final outcome. From files of Daniel Riordan, MD, courtesy of Dr. DC Faust.

the surgeon should note which finger the patient uses as a thumb.[24] Any accompanying wrist flexion contracture must be corrected with careful attention to musculotendinous balancing. Because the forearm and elbow, and occasionally even the more proximal limb, will be abnormal in this disorder, multiple surgeries are again the norm.[1]

Pentadactyly is treated like thumb hypoplasia, with first web space creation.[1]

Complications

In polydactyly, a reoperation rate of up to 25% has been reported, with most reoperations performed because of residual or subsequent deformity.[5,30,31,38] Risk factors for reoperation are type IV thumb duplication, preoperative "zigzag" deformity, and radially deviated thumb elements at presentation.[5] The delta phalanx may not show on radiographs until the patient is 18 months old, but functional deformity will worsen as long as it is present. Zigzag deformity may be due to the delta phalanx or to musculotendinous imbalance, such as a radially inserted flexor pollicis longus (FPL) or lack of stable MCP abduction. Miura[31] found that careful reconstruction of the joint capsule and thenar muscles from the ablated digit to the remnant digit

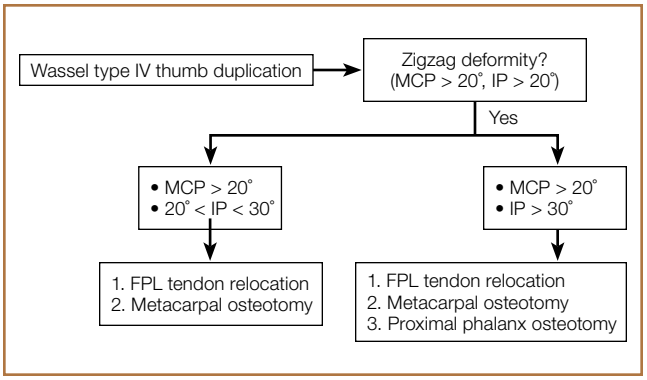

Figure 17. Algorithm of surgical procedures for zigzag deformity. Figure from *Journal of Hand Surgery, European Volume*, vol. 38, Lee CC, Park HY, Yoon JO, Lee KW, "Correction of Wassel type IV thumb duplication with zigzag deformity: results of a new method of flexor pollicis longus tendon relocation," pages 272-280, Copyright Elsevier 2013. Reproduced with permission.

Abbreviations: IP, interphalangeal angulation; FPL, flexor pollicis longus; MCP, metacarpophalangeal angulation.

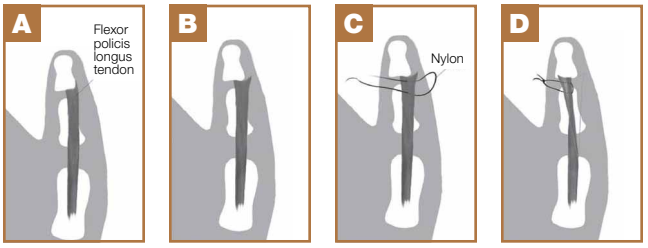

Figure 18. Pullout suture technique for flexor pollicis longus tendon relocation. Figure from *Journal of Hand Surgery, European Volume*, vol. 38, Lee CC, Park HY, Yoon JO, Lee KW, "Correction of Wassel type IV thumb duplication with zigzag deformity: results of a new method of flexor pollicis longus tendon relocation," pages 272-280, Copyright Elsevier 2013. Reproduced with permission.

is the key to a successful initial surgery. Lee and colleagues[39] defined zigzag deformity as more than 20° MCP and IP angulation; for cases present before surgery, they recommended FPL relocation by the pullout technique in addition to osteotomies to prevent further interphalangeal deviation (**Figures 17, 18**).

Abnormal physeal growth, joint instability, and stiffness can all occur. Stiffness is particularly difficult to treat but seldom presents a functional problem. Joint enlargement, which is not uncommon, results from either broad articular surfaces or retained cartilage from the perichondral ring after resection that later ossifies.[5,38] Nubbin-type duplications may not fall off after suture ligation, necessitating further excision, and a cosmetic bump is seen after 40% of suture ligations.[3] Patillo and Rayan[28] and Rayan and Frey[29] warned against suture ligation unless the nubbin has a small stalk because of the possibility of infection and gangrene. The excised nubbin tissue is histologically nervous, and there have been reports of painful neuromas in the remaining scar of a ligated nubbin that respond well to excision.[26,27,40] It is thought that these painful lesions form because the ligature prevents the digital nerves to the vestigial digit from retracting.[27] Nail deformity and IP joint stiffness are seen with the Bilhaut-Cloquet procedure, though often finger function remains satisfactory.

Conclusion

Polydactyly is a common congenital hand abnormality. Its true incidence is unknown because of inconsistent documentation. Surgeons must strive for a functional, cosmetic hand, given a diverse set of possible anomalies. Hypoplasia is the rule; tissue should be ablated and augmented as necessary. Musculotendinous insertions may need to be centralized. Patients' family members should always be counseled that more surgery may be needed in the future, as further deformity can occur with growth. Surgically corrected thumb duplications will be stiffer, shorter, and thinner than their normal counterparts. Nail ridges are common. However, it should be noted that 88% of these patients are satisfied with their results.[41] Some amount of contracture and abnormal function should be expected with index-, long-, and ring-finger duplications. The only remnant of type B postaxial duplications may be a slight discoloration or bump, though stiffness and deformity can happen with a type A deformity. A "duplicated" digit that requires surgical correction will never be completely normal, but acceptable function is routinely achievable.

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This paper will be judged for the Resident Writer's Award.