Chapter 12
Polydactyly and Syndactyly of the Hand

Erik C. B. King and Muhammad Y. Mutawakkil

Brief Overview of Condition

Hand anomalies are the second most common musculoskeletal congenital conditions encountered in the newborn, and polydactyly and syndactyly are the most common hand anomalies. Polydactyly is defined as a duplication of fingers in the hand, and syndactyly is defined as a fusion of the soft tissue and/or skeletal elements of adjacent digits. Polysyndactyly refers to syndactyly and polydactyly in the same hand. Both polydactyly and syndactyly can occur in isolation or as part of a syndrome. Because both congenital differences can potentially cause significant limitations in normal hand function, it is important to establish the diagnosis early and develop a treatment plan. Corrective surgery is usually recommended and should be timed so as to minimize functional limitations. Discussions about timing usually focus on ensuring that the child achieves developmental milestones, while also minimizing the risks of surgery and anesthesia.

E. C. B. King (✉)
Department of Orthopaedic Surgery, Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, USA
e-mail: eking@luriechildrens.org

M. Y. Mutawakkil
Department of Orthopaedic Surgery, Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, USA

Department of Orthopaedic Surgery, Northwestern University Feinberg School of Medicine, Chicago, IL, USA
e-mail: muhammad.mutawakkil@northwestern.edu

J. F. Sarwark, R. L. Carl (eds.), *Orthopaedics for the Newborn and Young Child*,
https://doi.org/10.1007/978-3-031-11136-5_12

Background Including Epidemiology and Pathophysiology

The incidence of polydactyly varies by ethnic groups. It is estimated to be 1 in 300 in African-descent populations and 1 in 3000 in Caucasian populations [1, 2]. In children of African descent, polydactyly is more common on the ulnar aspect of the hand, whereas in Caucasian children, polydactyly is more common on the radial aspect of the hand. Indeed, the presence of ulnar polydactyly in a Caucasian child should raise suspicion that the child has an underlying syndrome. Polydactyly can be an isolated congenial anomaly or a component of a syndrome. Through advances in the genetics and molecular biology, several genes that play a role in the development of polydactyly have been identified. The responsible genes include GLI3 gene, the ZNF141 gene, the MIPOL1 gene, and the PITX1 gene [3]. Syndromes associated with polydactyly include Apert syndrome, Carpenter syndrome, Pfeiffer syndrome, Smith Lemli Opitz syndrome, Pallister Hall syndrome, Poland syndrome, Bardet Biedl syndrome, Ellis–van Creveld syndrome, Laurence-Moon-Biedl syndrome, and trisomy 13 [1, 2, 4].

The incidence of syndactyly is reported to be 1 in 2000 live births [4]. Like polydactyly, the incidence of syndactyly varies by ethnic group. It is ten times more common in Caucasian individuals than in African-descent individuals [1]. Positive family history is reported in 10–40% of cases, and approximately 50% of syndactyly cases are bilateral. Although syndactyly exhibits an autosomal dominant pattern of inheritance, variable expressivity and incomplete penetrance is seen, resulting in variable phenotype within a family and a 2:1 preponderance in males. Syndactyly can be an isolated congenital anomaly or a component of a syndrome.

During the early stages of fetal development, hand and feet are initially webbed together extending to the fingertips resembling a mitten. The expression of apical ectodermal ridge (AER) maintenance factor causes the webbing to persist early in gestation. However, during normal embryologic development, the production of AER ceases in a programmed coordinated fashion. Cessation of AER production results in apoptosis (also known as programmed cell death) of the cells in the web. In turn, the tissue in the web recedes beginning distally and progressing proximally, and the web space is formed. Embryologic events that lead to the failure of normal apoptosis cause syndactyly.

Clinical Presentation: History and Physical

Polydactyly

Polydactyly is classified based on the radial to ulnar location of the duplicated digit. Radial polydactyly, also known as preaxial polydactyly, occurs along the radial side of the hand. Ulnar polydactyly, also known as postaxial polydactyly, involves the

ulnar side of the hand. Central polydactyly is less common and involves duplication of one of the three central digits. Central polydactyly is often coexistent with syndactyly.

Radial Polydactyly (Preaxial Polydactyly)

On clinical examination, radial polydactyly resembles a duplication of the thumb (Fig. 12.1). However, parents should be counseled that this is in fact not a duplication of a normal thumb, but rather abnormal development of two thumbs. Neither of the thumbs is normal, and even following reconstructive surgery, the residual thumb is always more narrow and stiffer than a normal thumb. Some hand specialists believe that "split thumb" is a more appropriate term for this anomaly.

Radial polydactyly may involve any level of the thumb and is commonly described by the Wassel classification [5] (Fig. 12.2). This classification scheme is based on the anatomic level of duplication and the number of supernumerary bones. Type IV is the most common type.

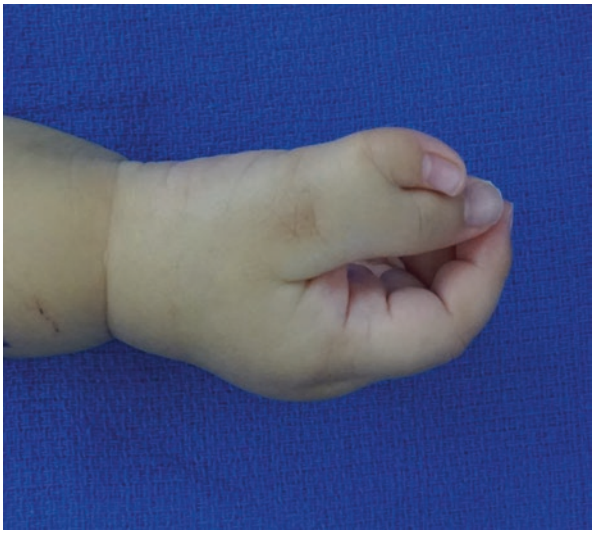

Fig. 12.1 Radial polydactyly, Wassel type IV

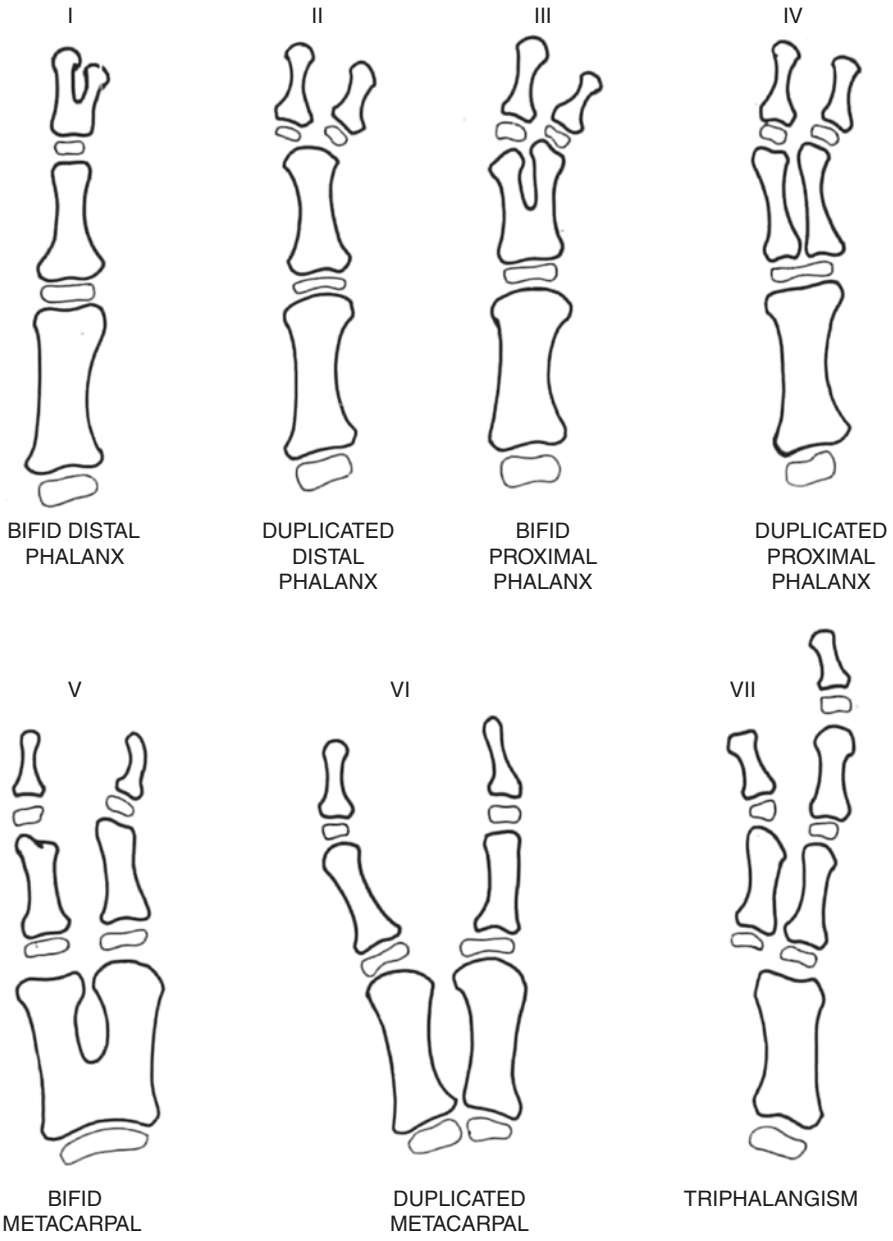

Fig. 12.2 Seven types of thumb polydactyly distinguished [5]. Reprinted with permission from Wassel HD. The results of surgery for polydactyly of the thumb: A review. Clinical Orthopaedics and Related Research, Vol. 64, pgs. 175–193, © 1969, with permission from Wolters Kluwer Health, Inc.

Central Polydactyly

Central polydactyly is an uncommon form of polydactyly that affects the index, ring, or long fingers (Fig. 12.3). Central polydactyly makes up approximately 5–15% of all polydactylies [6, 7]. Central polydactyly is classified into three types. Type I polydactylies are ones in which the supernumerary finger is not attached to the adjacent finger by osseous or ligamentous attachments. Type II polydactylies have normal-appearing osseous and soft tissue structures within the supernumerary digit and share a joint, a bifid metacarpal, or phalanx with the adjacent finger. Type II is further subdivided by the absence (type IIa) or presence (type IIb) of associated syndactyly. Type III polydactylies consist of completely duplicated rays, including fully formed metacarpals.

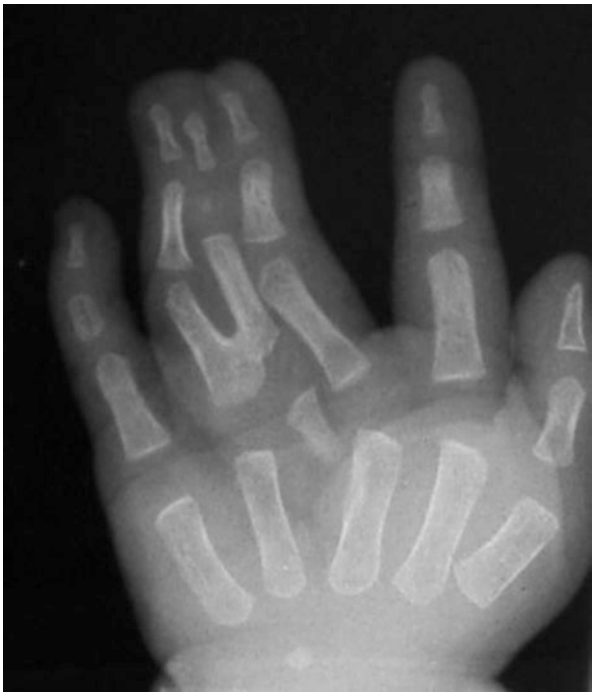

Fig. 12.3 Central polydactyly between middle and long fingers. There is also syndactyly involving all three fingers

Ulnar Polydactyly (Postaxial Polydactyly)

Ulnar polydactyly is the most common form of polydactyly. This anomaly presents as a supernumerary digit on the ulnar border of the affected hand. Ulnar polydactylies are classified as Type A, which is a fully developed finger, or Type B, which comprises a rudimentary nubbin or pedunculated finger attached to the lateral aspect of the finger via a small soft tissue stalk (Fig. 12.4).

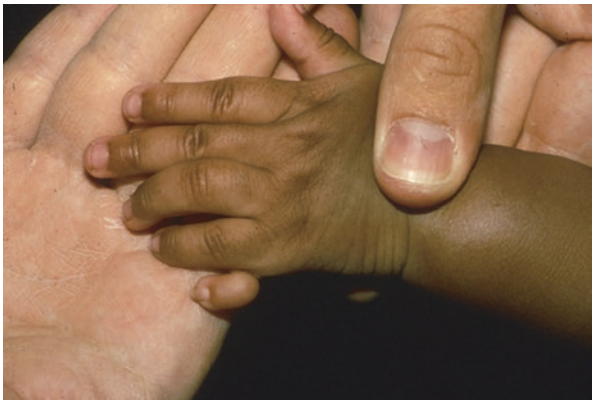

Fig. 12.4 Ulnar polydactyly, type B. The rudimentary polydactyly digit (nubbin) is connected by a narrow soft tissue stalk with no articulation to the hand

Mirror Hand

Mirror hand is an extremely rare form of polydactyly in which there is symmetric duplication of the hand around the mid-axis of the hand. The central finger is often bordered by three symmetric fingers on each side (Fig. 12.5). Despite the seven digits, there is no functional thumb. Mirror hand is also associated with duplication of the ulna bone in the forearm without a true radius.

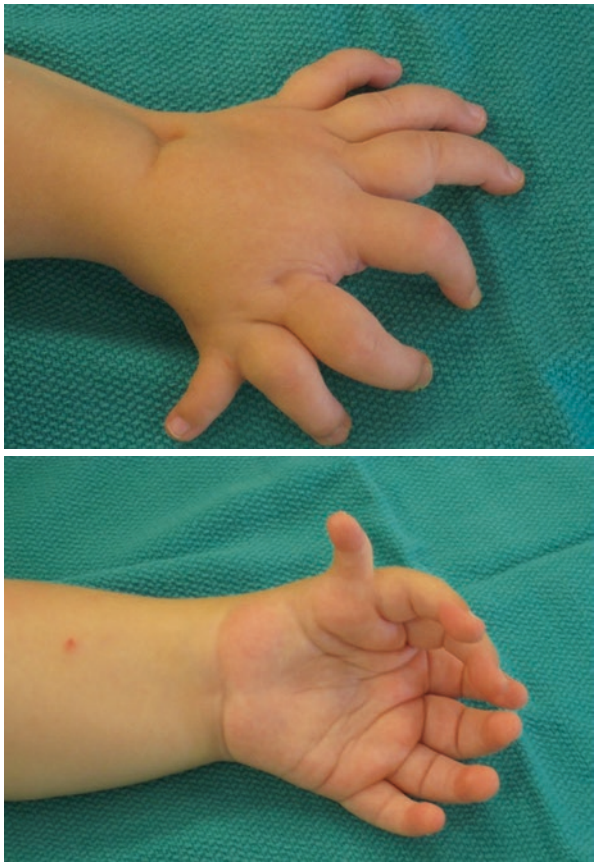

Fig. 12.5 Mirror hand (ulnar dimelia). Note that the hand lacks a functional thumb

Clinical Presentation: History and Physical

Syndactyly

Syndactyly is defined as simple if there is fusion of soft tissue components only, and complex if there is fusion of the bone in addition to the soft tissues. Additionally, syndactyly is considered complete if the finger fusion extends from the hand all the way to the fingertip, and incomplete if the fusion ends proximal to the fingertip. The most common location for syndactyly is between the middle (third) and ring (fourth) fingers (Fig. 12.6). A syndactyly that has additional skeletal anomalies, e.g., abnormally oriented bones, is further classified as complicated. Acrosyndactyly is a type of syndactyly in which there is fusion at the distal ends of the finger but with fenestration proximally. Acrosyndactyly is most commonly seen in amniotic band syndrome and Apert syndrome (acrocephalosyndactyly).

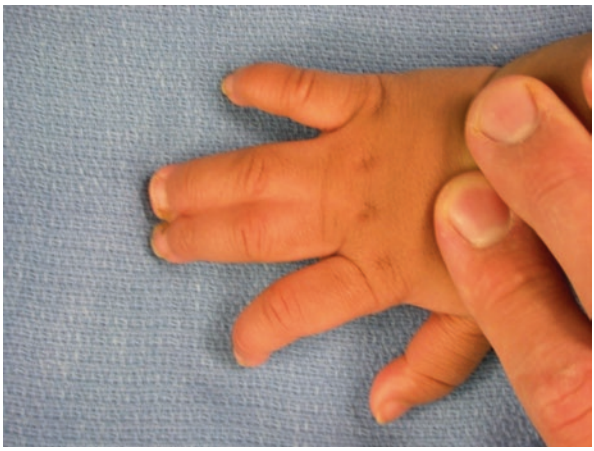

Fig. 12.6 Simple complete polydactyly between the middle and ring fingers

Evaluation

Both polydactyly and syndactyly of the hand are typically identified at birth or shortly thereafter. When these anomalies are present, special attention should be paid to look for other congenital abnormalities that would suggest the presence of a syndrome. There is no immediate urgency during the postnatal period to obtain radiographs of the affected hand, since surgery is generally deferred until after 6 months of age. During subsequent evaluation by the hand surgeon, radiographs will be obtained in order to delineate the skeletal anatomy. This information will aid in surgical planning and the determination of prognosis.

Management

In general, most congenital polydactyly and syndactyly problems can be managed surgically to improve hand function and appearance. Surgical procedures can vary from simple to complex reconstructions, depending on the complexity of the abnormal anatomy. Although timing for surgical intervention is a matter of surgeon preference, most surgeons recommend performing surgery for these conditions when the child is 6 to 18 months of age. Timing considerations include anesthesia safety, size of the hand, and developmental milestones. The risks of anesthesia-related complications decrease greatly after the child is 6 months old. For the more complex cases, multiple surgical procedures spread over many months or years may be recommended.

Due to its importance in normal hand function, radial (thumb) polydactyly reconstruction is recommended in virtually every case. The surgical goal is to create the best functioning thumb possible by utilizing components of both thumbs. Often this means using components of the most hypoplastic thumb to augment those of the more normal retained thumb. Surgical techniques include ligament reconstruction, transfer of the intrinsic hand muscles to the preserved digit, and reinforcement of the extrinsic flexor and extensor tendons.

In the treatment of ulnar polydactyly, ligation, or "tying off," in the newborn nursery of some type B ulnar polydactylies is feasible if there is a small, underdeveloped digit connected by a narrow soft tissue stalk. Ligation causes necrosis and eventual auto-amputation of the nubbin. Parents should be advised that it may take 2–4 weeks for the necrotic digit to fall off. However, there are many reports of an unpleasing scar, a small mound of tissue, or painful neuromas following simple ligation. In order to avoid these complications, some physicians no longer routinely recommend ligation of type B polydactylies. Certainly, type B anomalies that have a broad connection are best treated with formal surgical excision in the operating room. Similarly, the more extensive skeletal connection of type A ulnar polydactyly does not lend itself to tying off in the newborn period. Type A ulnar polydactylies are best treated surgically later in infancy.

Central polydactyly is treated by surgical excision of the most hypoplastic digit. When associated with syndactyly, separation of the digit is usually performed concurrently. This can be surgically challenging. Underlying bone and joint abnormalities may cause decreased joint mobility and abnormal alignment after otherwise successful surgery. If the central supernumerary digit is fully developed and has normal function, non-surgical observation can be considered.

In cases of mild incomplete simple syndactyly, surgery may not be necessary. For those syndactylies that do require surgery, better outcomes have been reported when syndactyly surgery is performed after 18 months of age [8]. The larger hand size compared to the younger child makes the technical aspects of the surgery easier. When surgery is performed in patients younger than 18 months of age, there is a greater risk for the development of post-surgical web creep as the child grows. Web creep is the undesired process in which the commissure between the fingers is drawn over time in a distal direction by scar contraction.

In cases in which syndactylies span multiple contiguous digits, surgical reconstruction should be performed in multiple stages (Fig. 12.7). The digital blood vessels course along the ulnar and radial sides of each finger. A basic principle of hand surgery is the avoidance of operating on both sides of a finger at a single surgical encounter. Instead, separating surgeries by at least 3 months allow collateral

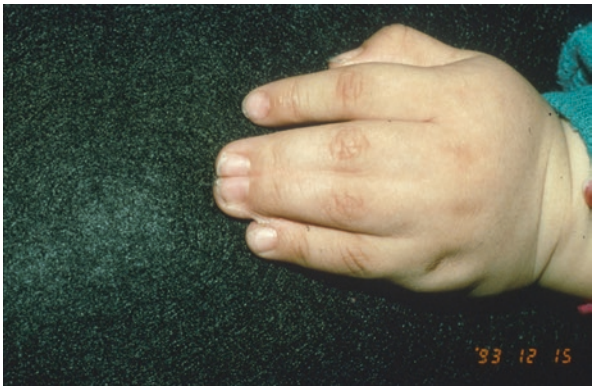

Fig. 12.7 Syndactyly between the middle and ring fingers (complete complex) and between the ring and small fingers (complete simple). Surgical correction was performed in two stages in order to minimize the risk of vascular compromise to the ring finger

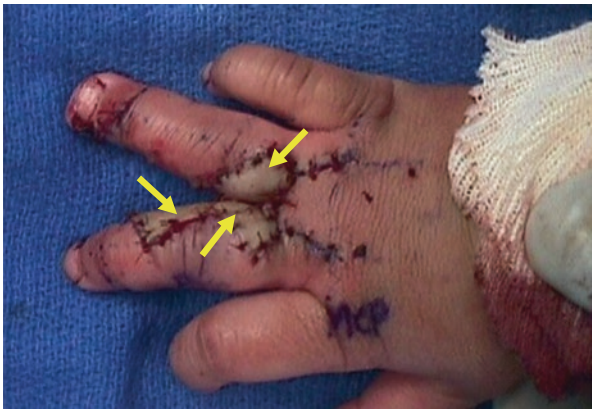

Fig. 12.8 Patient from Fig. 12.6. Surgical separation was performed at 16 months of age. Because of the greater surface area after finger separation, full thickness skin graft was placed during closure (yellow arrows)

circulation and revascularization to occur, and thus minimizes the risk of vascular compromise and iatrogenic necrosis. The goal is to complete all stages of surgery by school age.

When syndactyly surgery is performed, full thickness skin grafting is usually required (Fig. 12.8). This is because the surface area of the skin around conjoined fingers is less than the surface area of skin needed to cover the fingers once they are separated. Skin grafting fills the deficit.

Clinical Pearls

Polydactyly

- Polydactyly may be isolated disorder or an element of a syndrome.
- The most common forms of polydactyly are radial polydactyly and ulnar polydactyly.
- Radial polydactyly resembles thumb duplication or split thumb.
- Only the most rudimentary polydactyly should be considered for tying off in the newborn period. Complications of tying off including an undesirable scar, a mound of tissue, and/or a painful neuroma.
- Most polydactylies are removed surgical after 6 months of age.

Syndactyly

- Syndactyly may be isolated or an element of a syndrome.
- Syndactyly is described as simple vs. complex, complete vs. incomplete, or complicated.

- The most common location for syndactyly is between the middle and ring fingers.
- Delaying reconstructive surgery to closer to 14 months of age may minimize the complication of web creep.
- If a child has multiple syndactylies, surgical reconstruction will be staged.
- Virtually every syndactyly surgery will require skin grafting to cover skin deficits.

References

1. Ho C. Chapter 15: Disorders of the upper extremity. In: Tachdjian's pediatric orthopaedics: from the Texas Scottish Rite Hospital for Children. 5th ed. Dallas: Elsevier; 2014. p. 388–452.
2. Malik S. Polydactyly: phenotypes, genetics and classification. Clin Genet. 2014;85(3):203–12.
3. Deng H, Tan T, Yuan L. Advances in the molecular genetics of non-syndromic polydactyly. Expert Rev Mol Med. 2015;17:e18.
4. Ahmed H, Akbari H, Emami A, Akbari MR. Genetic overview of syndactyly and polydactyly. Plast Reconstr Surg Glob Open. 2017;5(11):e1549.
5. Wassel HD. The results of surgery for polydactyly of the thumb: a review. Clin Orthop Relat Res. 1969;64:175–93.
6. Wood VE. Treatment of central polydactyly. Clin Orthop Relat Res. 1971;74:196–205.
7. Tada K, Kurisaki E, Yonenobu K, Tsuyuguchi Y, Kawai H. Central polydactyly: a review of 12 cases and their surgical treatment. J Hand Surg Am. 1982;7(5):460–5.
8. Kay SP, McCombe DB, Kozin SH. Chapter 36: Deformities of the hand and fingers. In: Green's operative hand surgery. 7th ed. Philadelphia: Elsevier; 2017. p. 1217–32.

Suggested Readings/Additional Resources

Baek GH, Gong HS, Chung MS, Oh JH, Lee YH, Lee SK. Modified Bilhaut-Cloquet procedure for Wassel type-II and III polydactyly of the thumb. J Bone Joint Surg Am. 2007;89(3):534–41.

Canizares MF, Feldman L, Miller PE, Waters PM, Bae DS. Complications and cost of syndactyly reconstruction in the united states: analysis of the pediatric health information system. Hand (New York, NY). 2017;12(4):327–34.

Comer GC, Potter M, Ladd AL. Polydactyly of the hand. J Am Acad Orthop Surg. 2018;26(3):75–82.

Dao KD, Shin AY, Billings A, et al. Surgical treatment of congenital syndactyly of the hand. J Am Acad Orthop Surg. 2004;12:39–48.

Holmes LB, Nasri H, Hunt AT, Toufaily MH, Westgate MN. Polydactyly, postaxial, type B. Birth Defects Res. 2018;110(2):134–41.

Kozin SH, Zlotolow DA. Common pediatric congenital conditions of the hand. Plast Reconstr Surg. 2015;136(2):241e–57e.

Malik S. Syndactyly: phenotypes, genetics and current classification. Eur J Human Genet. 2012;20(8):817–24.

Perez-Lopez LM, la Iglesia DG, Cabrera-Gonzalez M. Radial polydactyly. What's new? Curr Pediatr Rev. 2018;14(2):91–6.

Temtamy SA, McKusick VA. The genetics of hand malformations. Birth Defects Orig Artic Ser. 1978;14(3):i–xviii, 1–619.

Winter RM, Tickle C. Syndactylies and polydactylies: embryological overview and suggested classification. Eur J Human Genet. 1993;1(1):96–104.