Hypoplasia, Brachydactyly, and Other Failures of Formation

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Scott A. Riley and Ronald Burgess

Contents

Abstract

Congenital anomalies affect 1–2 % of newborns, and approximately 10 % of these children have upper extremity abnormalities. Congenital anomalies of the limb are second only to congenital heart disease in the incidence of birth malformations (Bamshad et al. Pediatr Res 45:291–299, 1999). The clinical manifestations of these anomalies in children are extremely variable, and as such, classifying specific patterns of deformities remains an ongoing challenge. However, approximately 10–15 % of these congenital upper extremity anomalies can be grouped into a broad category that represents underdevelopment and/or failure of formation within portions of the upper limb. The deformities represented in this category can range anywhere from a smaller than normal digit (hypoplasia) to a complete absence of the extremity (amelia).

To the medical provider who is unaccustomed to evaluating and caring for these children, describing these anomalies can be difficult, and formulating potential treatment plans is often a difficult task. Therefore, the goals of this chapter are to (1) present accepted terminology used to define specific conditions, (2) discuss the common clinical features, and (3) provide treatment recommendations for some particular diagnostic subtypes seen in this category of deformities.

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Introduction

For the past 50 years, limb malformations in the upper extremity have been classified according to the predominant anomaly identified. Most congenital upper extremity anomalies can fit into one of seven general categories as defined by the International Federation of Hand Societies (Table 1; Swanson 1976). Some upper extremity anomalies are easier to classify than others. For example, a transverse deficiency in the upper extremity is classified as a Type I – failure of formation. The clinical presentation of this condition is usually straightforward, so categorizing the anomaly is not difficult. However, if the limb also has residual nubbin-like structures, like those seen in symbrachydactyly, the classification is more challenging (Cheng et al. 1987). Then consider an anomaly like cleft hand, classically thought to represent a longitudinal-central failure of formation (Type I); but it can also present with anatomic findings that show characteristics of hypoplasia (Type IV – undergrowth) as well as syndactyly (Type II – failure of separation). All in all, there are many instances where descriptive classification schemes do not work very well.

While it is interesting to discuss how different upper extremity anomalies can fit in to certain classification systems, it should be stated that as the understanding of developmental biology improves, these systems will continue to evolve over time (Manske and Oberg 2009). A more recent scheme for classifying congenital hand anomalies, referred to as the Oberg, Manske, and Tonkin (OMT) system (Oberg et al. 2010; Tonkin et al. 2013), has proposed three distinct categories that distinguish malformations from those that represent deformities or dysplasia (Table 2). Ultimately, the hope is that by continuing to explore these developmental differences, the physician community may be able to offer better treatment protocols for a child with a specific deformity. Rather than delving into these categorical differences, the goal of this chapter is to develop an appreciation of the more common anomalies associated with both undergrowth and failures of formation in the upper extremity. To accomplish this, the focus will be on the clinical presentation, associated findings, and treatment considerations for these conditions.

An additional point to consider is that cleft hands, transverse deficiencies, and phocomelia are among the most obvious and disfiguring of all congenital anomalies. With the advent of advanced prenatal imaging, the accuracy in diagnosing congenital upper extremity anomalies in utero is improving. As such, parents may consult the orthopedic surgeon about these disorders in the prenatal period. For a source of guidance about these matters, Bae, et al. provide an excellent discussion about the current status of prenatal screening as well as ethical and treatment considerations involved with the implementation of this technology (Bae et al. 2009).

Hypoplasia

Hypoplasia is defined as a small or underdeveloped body part. In the upper extremity, any anatomic segment such as a digit, a hand, or even the entire arm may be considered hypoplastic when compared to its normal counterpart in the contralateral extremity. Despite its small size, a hypoplastic body part may well be very functional, and when possible, every effort should be attempted to preserve or augment these affected, yet functional units.

The exact incidence of hypoplasia in the upper extremity is difficult to determine because in many congenital anomalies that are encountered, a hypoplastic anatomic part can also be identified as a part of the main condition. However, if one considers isolated hypoplasia of a single upper extremity segment with no other associated musculoskeletal deficits, the condition represents about 8 % of all congenital upper extremity anomalies (Giele et al. 2001). Yet it is suspected that this number may be artificially small due to underreporting of hypoplasia when the upper extremity remains very functional.

The inheritance patterns of hypoplastic anomalies are thought to be sporadic in nature unless the condition is associated with a specific syndrome. For example, many children with Feingold

Table 1 Embryological classification of congenital anomalies

Classification	Subheading	Subgroup	Category
I. Failure of formation			
	A. Transverse arrest		
		1. Shoulder	
		2. Arm	
		3. Elbow	
		4. Forearm	
		5. Wrist	
		6. Carpal	
		7. Metacarpal	
		8. Phalanx	
	B. Longitudinal arrest		
		1. Radial deficiency	
		2. Ulnar deficiency	
		3. Central deficiency	
		4. Intersegmental	Phocomelia
II. Failure of differentiation			
	A. Soft tissue		
		1. Disseminated	(a) Arthrogryposis
		2. Shoulder	
		3. Elbow and forearm	
		4. Wrist and hand	(a) Cutaneous syndactyly
			(b) Camptodactyly
			(c) Thumb-in-palm
			(d) Deviated/deformed digits
	B. Skeletal		
		1. Shoulder	
		2. Elbow	Synostosis
		3. Forearm	(a) Proximal
			(b) Distal
		4. Wrist and hand	(a) Osseous syndactyly
			(b) Carpal bone synostosis
			(c) Symphalangia
			(d) Clinodactyly
	C. Tumorous conditions		
		1. Hemangiotic	
		2. Lymphatic	
		3. Neurogenic	
		4. Connective tissue	
		5. Skeletal	
III. Duplication			
	A. Whole limb		
	B. Humeral		
	C. Radial		
	D. Ulnar		
		1. Mirror hand	

(*continued*)

Table 1 (continued)

Classification	Subheading	Subgroup	Category
	E. Digit		
		1. Polydactyly	(a) Radial (preaxial)
			(b) Central
			(c) Ulnar (postaxial)
IV. Overgrowth	A. Whole limb		
	B. Partial limb		
	C. Digit		
		1. Macrodactyly	
V. Undergrowth			
	A. Whole limb		
	B. Whole hand		
	C. Metacarpal		
	D. Digit		
		1. Brachysyndactyly	
		2. Brachydactyly	
VI. Constriction band syndrome			
VII. Generalized skeletal abnormalities			

syndrome (inherited as autosomal dominant) are found to have small second and fifth fingers as part of the disorder. Thumb hypoplasia, which is discussed in detail in another chapter, can be seen in association with Fanconi anemia, an autosomal recessive disorder. As such, if hypoplasia is seen in conjunction with anomalies, a formal genetic consultation is probably indicated, and additional diagnostic testing may be needed to rule out other organ system involvement.

The clinical presentation of a child with hypoplasia can be quite variable. Obvious size differences are usually easy to detect, but the best way to identify subtler forms is to directly measure the region of concern and compare it to the contralateral upper extremity. In these mild forms of hypoplasia, radiographs may aid in confirming the diagnosis. In addition to noting potential size differences, comparison radiographs can also show other anomalies such as missing carpal bones, carpal coalitions, and malformed phalanges. Additional testing modalities such as MRI and CT scanning are usually not necessary for diagnostic purposes.

Any treatment considerations for the patient with hypoplasia should focus on maximizing function of the involved extremity. Often, this will require following the patient for an extended period of time, before recommending any surgical interventions, in order to understand just how the child uses the extremity most efficiently. In addition to getting subjective input about hand usage from the family, occupational therapists can provide objective functional data by administering age-appropriate testing modalities and thus offer valuable insights during this process.

Most patients with mild hypoplasia are very high functioning and require no surgical treatment for the condition. These children may need to alter the manner in which a task needs to be performed; yet they adapt very well in most task-oriented situations. There are, however, a few instances where surgery can offer benefit to these patients. One example would be if a child has a small, floppy digit that gets "in the way" of gripping or pinching tasks. In this case, hand use is being compromised, and amputation of that digit should be considered. In contrast, a small, stable finger with good tendon function might be made more useful by bone lengthening (Arata et al. 2011), and reconstruction is the best surgical option for this particular digit. It is equally important that the

Table 2 Oberg, Manske, and Tonkin (OMT) classification

1. Malformations
A. Failure of axis formation/differentiation – entire upper limb
1. Proximal-distal outgrowth
Brachymelia with brachydactyly
Symbrachydactyly
Transverse deficiency
Intersegmental deficiency
2. Radial-ulnar (anteroposterior) axis
Radial longitudinal deficiency
Ulnar longitudinal deficiency
Ulnar dimelia
Radioulnar synostosis
Humeroradial synostosis
3. Dorsal-ventral axis
Nail-patella syndrome
B. Failure of axis formation/differentiation – hand plate
1. Radial-ulnar (anteroposterior) axis
Radial polydactyly
Triphalangeal thumb
Ulnar polydactyly
2. Dorsal-ventral axis
Dorsal dimelia (palmar nail)
Hypoplastic/aplastic nail
C. Failure of axis formation/differentiation – unspecified axis
1. Soft tissue
Syndactyly
Camptodactyly
2. Skeletal deficiency
Brachydactyly
Clinodactyly
Kirner's deformity
Metacarpal and carpal synostoses
3. Complex
Cleft hand
Synpolydactyly
Apert hand
2. Deformations
A. Constriction ring sequence
B. Arthrogryposis
C. Trigger digits
D. Not otherwise specified
3. Dysplasias
A. Hypertrophy
1. Macrodactyly
2. Upper limb
3. Upper limb and macrodactyly
B. Tumorous conditions

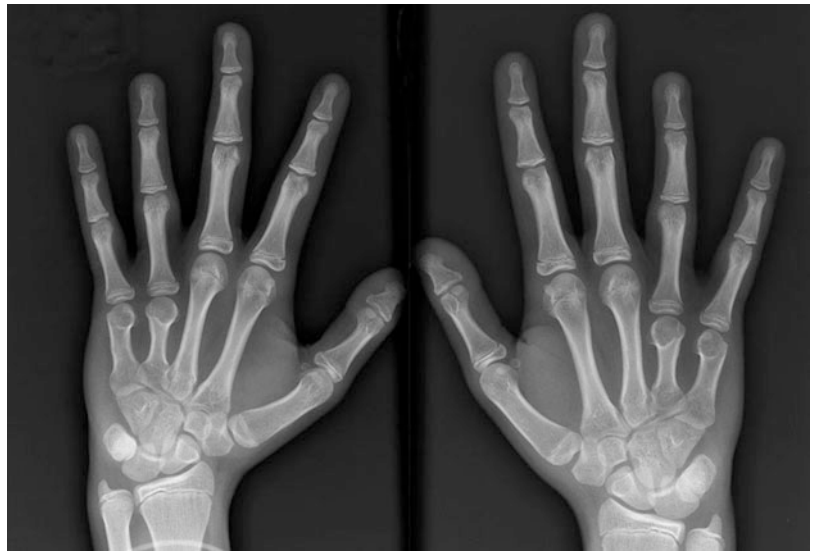

Fig. 1 Fourteen-year-old female with bilateral ring and small short metacarpals otherwise known as "knuckle-knuckle-bump-bump" (Courtesy of Shriners Hospitals for Children, Philadelphia)

patient's family has a very clear understanding of the expected goals of any type of surgery. Maximizing the child's use of the extremity should be the primary goal, and focusing on this goal may help dismiss any unrealistic expectations regarding treatment outcomes.

Treatment Strategy

Because the majority of children with hypoplasia of a portion of the upper extremity are very high functioning, they require no surgical care. Nonetheless, there are two instances where surgical treatments are helpful. The first indication is to remove a "floppy" digit that is interfering with the functional abilities of the hand. In such cases, excising the affected digit and reconstructing the local web space are recommended as surgical care. A short course of occupational therapy following the procedure may be helpful as well. The second circumstance, which is discussed in another chapter, involves reconstructing and improving the function of a hypoplastic thumb.

Brachydactyly and Symbrachydactyly

After considering hypoplasia of the upper extremity in general, a discussion of the more common clinical types is appropriate. Flatt reported that out of 2,758 collected cases of congenitally anomalous hands, brachydactyly (5.2 %) and thumb hypoplasia (3.5 %) were the most frequently seen undergrowth conditions (Flatt 1994a). Because thumb hypoplasia is discussed as a separate topic elsewhere, this section will focus on brachydactyly and symbrachydactyly.

The term brachydactyly as translated from Greek means "short finger." While the whole digit may appear to be small, on radiographic examination, an individual bone segment is often smaller than normal. When the phalanges are involved, the middle phalanx is the most commonly affected bone. In this instance, the condition may be referred to as brachymesophalangia (short middle phalanx). Short metacarpals can be seen in association with some syndromes (e.g., Turner's), so the term brachymetacarpia may be encountered in the literature (Fig. 1). It should be stated that these terms are purely descriptive and do not imply that one type consistently has a more favorable prognosis than others.

Most forms of brachydactyly are inherited in an autosomal dominant manner, usually with variable severity. However, there are some cases of brachydactyly are felt to be sporadic in occurrence because of no well-documented family inheritance pattern. On the molecular level, alterations in BMP cartilage-derived morphogenic protein

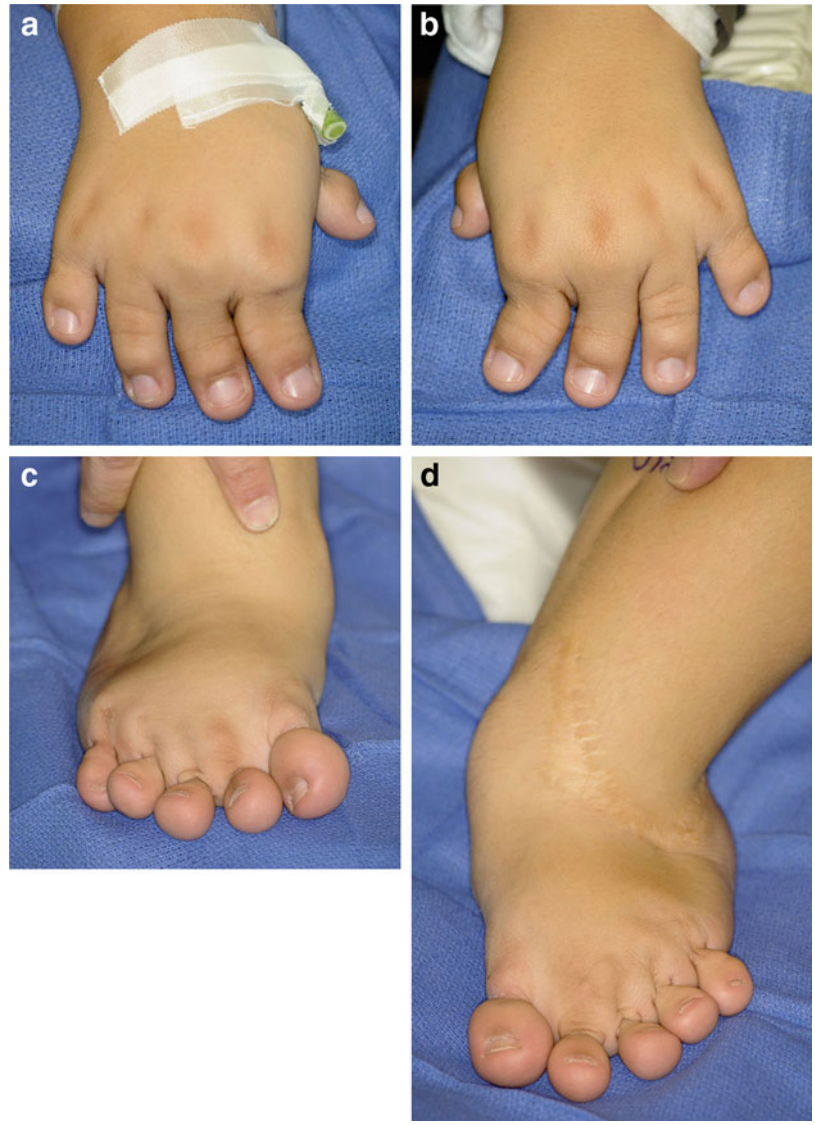

Fig. 2 Fourteen-year-old female with Grebes' chondrodysplasia and severe brachydactyly associated with a cartilage-derived morphogenetic protein deficiency (Courtesy of Shriners Hospital for Children, Philadelphia). (**a**) Right hand, (**b**) left hand, (**c**) right foot, (**d**) left foot

have been shown to be associated with brachydactyly in humans and mouse models (Waters and Bae 2012a; Fig. 2). A classification system for brachydactyly, first proposed by Bell in 1951 (Fitch 1979), focuses on the anatomic location of the hypoplastic segment to categorize the anomaly (Table 3). This system has been most useful in the identification of certain inheritance patterns by being able to follow specific types along family pedigrees. For example, the most common types, A3 and D, have been shown to demonstrate autosomal dominant inheritance with reduced penetrance. Some populations also have a relatively high frequency of certain types of brachydactyly (Temtamy and Aglan 2008). As far as the clinical presentation is concerned, the degree to which any digit is involved may range from a "small, but normal" appearing finger to a tiny residual "nubbin," which may be nothing more than a small pouch of skin attached to the hand.

When evaluating a patient with small digits, the term symbrachydactyly may be encountered in the congenital hand anomalies literature.

Table 3 Brachydactyly types (After Temtamy and Aglan 2008)

Type	Inheritance	Proposed gene defect	Clinical features
A1	Autosomal dominant	Indian hedgehog	Short middle phalanges (usually digits I–IV)
	Rare		
A2	Autosomal dominant	BMP receptor on q4	Short middle phalanx (digit II)
	Very rare		
A3	Autosomal dominant	Unknown	Short middle phalanx (digit V)
	Common (up to 21 % in some populations)		
A4	Few pedigrees identified	Mutation: HOXD13	Short middle phalanges (digits II and V)
	Rare (autosomal dominant)		
B	Few pedigrees identified	Mutation: ROR2	Absence/hypoplasia of terminal digits (II–V), with nail absence
	Rare (autosomal dominant)		Distal phalanx of thumb may be duplicated
C	Few pedigrees identified	Mutation: CDMP1	Brachymesophalangy of digits II, III, and V
	Rare (? autosomal dominant)		Ring finger (digit IV) is usually normal in size
D	Autosomal dominant (up to 4 % in some populations)	Unknown (possible mutation: HOXD13)	Short distal phalanx of digit I (thumb)
E	Autosomal dominant	Unknown (possible mutation: HOXD13)	Variable shortening of metacarpals (often digit IV)

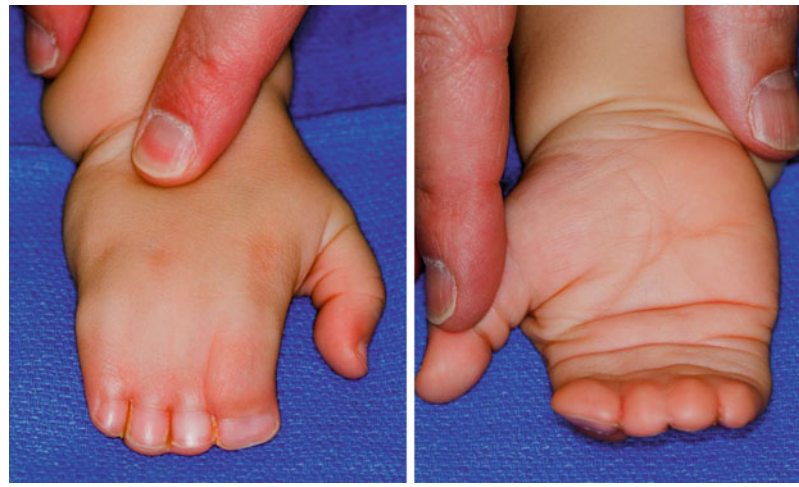

Fig. 3 Short finger symbrachydactyly is characterized by the triad of syndactyly, brachydactyly, and symphalangism (Courtesy of Shriners Hospitals for Children, Philadelphia)

As defined, symbrachydactyly is the condition representing hypoplastic digits (brachydactyly), webbing between fingers (syndactyly), and a general hypoplasia of the hand. To avoid confusion, brachydactyly refers specifically to the digit, whereas symbrachydactyly describes a spectrum of clinical findings seen in the hand.

A descriptive classification system has identified four separate types of symbrachydactyly: peromelic, oligodactylic, short finger, and monodactylic (Nguyen and Jones 2009) based upon the most commonly seen anatomic features (Figs. 3 and 4). Symbrachydactyly is often seen unilaterally, and the occurrence is felt to be sporadic in nature. It is secondary to a failure of formation, but the exact manner by which this occurs continues to be investigated. Symbrachydactyly can be seen in association

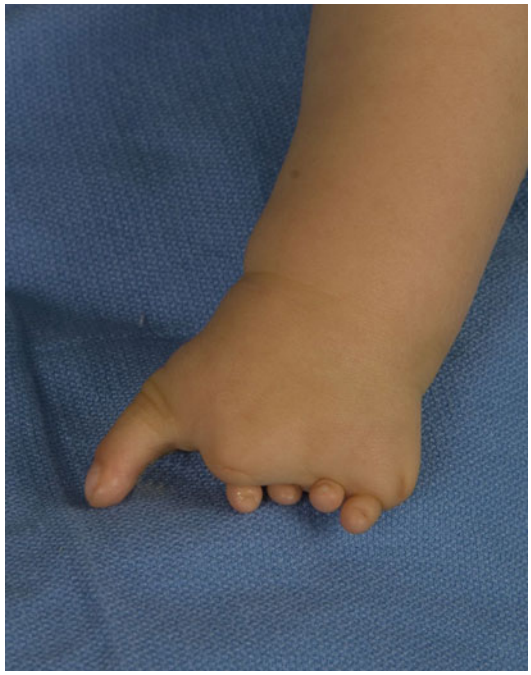

Fig. 4 Monodactylic symbrachydactyly with perseveration of the thumb (Courtesy of Shriners Hospitals for Children, Philadelphia)

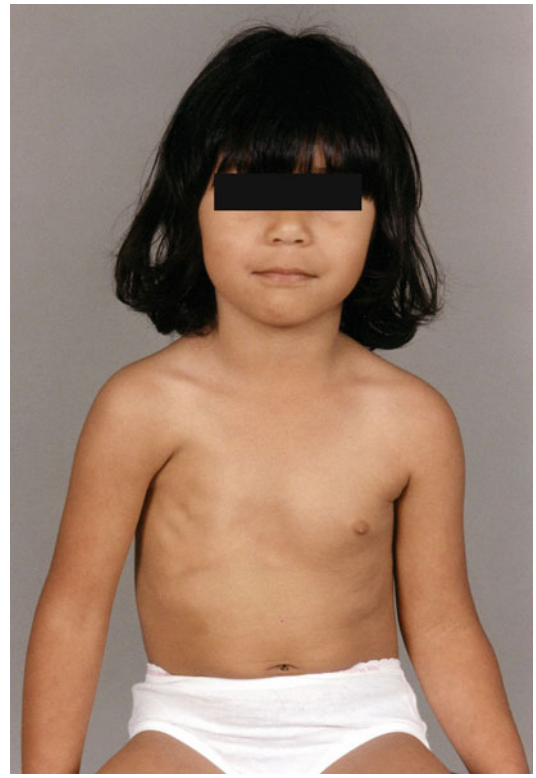

Fig. 5 A young girl with a Poland's syndrome denoted by the absence of the pectoralis muscles and the breast nipple (Courtesy of Shriners Hospitals for Children, Philadelphia)

with other conditions, and the classic example is Poland's syndrome (absence of the sternal head of the pectoralis major muscle with various degrees of ipsilateral hand hypoplasia; Fig. 5). This condition has recently been reported in association with transverse deficiency at the level of the forearm (Kallemeier et al. 2007). The term "atypical cleft hand" is also seen describing cases of symbrachydactyly.

Although symbrachydactyly and constriction band syndrome (CBS, discussed in another chapter) can appear similar in clinical appearance, the conditions have different diagnostic and treatment implications. There are two distinct differences. First, in patients with CBS, actual bands are usually identified elsewhere in the body. However, in symbrachydactyly, only one hand is commonly affected, and no other bands are seen. Second, with symbrachydactyly, the anatomic structures proximal to the small digits are also hypoplastic or abnormal. In CBS, the musculoskeletal structures proximal to a constriction band are normal in size (Fig. 6).

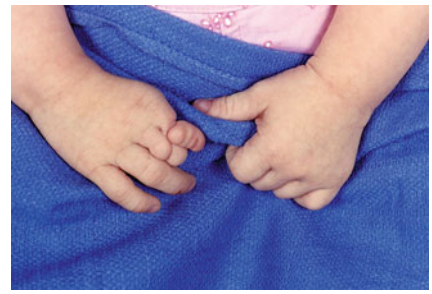

Fig. 6 Constriction band syndrome around the index and long fingers with normal musculoskeletal structures proximal to the banding (Courtesy of Shriners Hospitals for Children, Philadelphia)

While classification schemes regarding brachydactyly and symbrachydactyly are helpful in describing the child's clinical presentation, these systems are unable to predict the child's ability to use the hand. Any surgical recommendations should focus on improving a child's

functional outcome as "function trumps form." Input from the family about hand use, evaluations from therapists, and direct observation of the patient are critical in the decision-making process. Many patients with brachydactyly and symbrachydactyly are highly functional, adapt well to their hand anomaly, and do not warrant surgical intervention. They may occasionally need an assistive device to help with a specific task, however; surgery would not appreciably improve their function. There are cases that warrant surgery to augment hand use.

Probably the most common surgical technique to augment function involves altering the skin about the affected digit(s) through syndactyly release and/or web space reconstruction. Because the care of syndactylized fingers is presented in another chapter, a brief summary of web space reconstruction will be discussed here. The main goals of this procedure are to increase digital separation (span) and improve the apparent length of the fingers by providing depth to the web space. The Z-plasty and its varied modifications are the most common methods used to improve the web space (Shaw et al. 1973). Strict attention to detail is needed when designing the angles and arm's length of flap transpositions in order to maximize skin mobilization. Also, great care must be used when handling the transposed flaps so that tension on the skin is minimized. In most cases, well-designed skin flaps do not require additional soft tissue coverage. However, if skin is needed, a full-thickness skin graft can be used to supplement the reconstruction. As mentioned in the treatment of syndactyly, skin grafts should be avoided in the web space commissure due to their propensity to contract.

The actual length of a finger can be improved in two ways. For minor changes in digit length, an osteotomy and interposition bone grafting can be used. While this technique allows for immediate gains in length, the concern is that inserting too large a graft in a small digit may compromise the surrounding skin and soft tissue structures leading to necrosis of the fingertip (Flatt 1994b). An alternative is distraction osteogenesis,which involves gradual lengthening of the bone and the soft tissues. There have been many methods described to achieve this task (Arata et al. 2011), although the basic technique requires an osteotomy and a device to gradually distract (lengthen) the osteotomy site. The most common types of distractors are uni-plane external fixators, attached to the bone by small wires. The slow distraction rate, about 0.5 mm to1mm per day, allows for both fracture callus formation and the accommodation of the soft tissues necessary for bone lengthening. Gains in digital length of up to 3.5 cm have been reported (Seitz and Froimson 1995). After the bone is lengthened to the desired amount, the distraction ceases and the external fixator is left in place until the regenerate callus matures to resemble normal bone (i.e., a cortical shell and medullary cavity). Subsequently, the device is removed and appropriate therapy is begun. Early removal can result in callus deformation or overt fracture through the regenerative bone.

Nonvascularized toe phalangeal transfer is a method used to achieve immediate length in a hypoplastic digit. The technique typically involves harvesting the proximal phalanx from a toe, then transplanting it within the skin of a hypoplastic digit. This procedure is controversial with regard to indications and outcome. The best results are when the procedure is done before 18 months of age, as the rates of physeal arrest and phalangeal resorption increase beyond this age (Nguyen and Jones 2009). Preliminary nonvascularized toe phalangeal transfer followed by distraction lengthening of the phalanx has been performed (Netscher and Lewis 2008). A recent article has shown that there is potential donor site morbidity with the nonvascularized toe phalangeal transfer, thus narrowing the indications for he procedure (Garagnani et al. 2012).

The most immediate way to provide both length and function to the brachydactylous hand is through microvascular toe-to-hand transfer. Although this is a technically demanding procedure, it probably provides a child the best opportunity to develop functional pinch and grasp (Nguyen and Jones 2009). Most toe-to-hand transfers are done at the level of the metacarpophalangeal joint, and the second toe is the most common donor "digit." There is no generally

accepted age for surgical intervention, but due to the size of the structures involved, the procedure is generally performed after 18 months of age. Experienced surgeons are reporting a successful toe-to-hand transfer rate of 95 % or more (Jones et al. 2007), and the long-term outcomes indicate continued growth of the transferred toe as well as improved function in the hand (Nguyen and Jones 2009).

Treatment Strategy

For the brachydactylous digit, the mainstay of surgical treatment for a functioning finger is web space deepening or reconstruction. This procedure is recommended on one side of a finger at a time to lessen the chance of venous or arterial vascular compromise to the digit. The other side of the digit web is usually reconstructed 3 months later. Increasing the length of a phalanx or metacarpal either by direct grafting or by osteotomy distraction can improve both function and appearance of the finger. However, the gains in bony length may result in tendon mechanism dysfunction. As the bone is lengthened, the adjacent tendons become relatively shorter and can promote joint contractures.

In the case of symbrachydactyly, the primary goal is for the hand to be able to hold an object between two stable "posts." For the child that has good thumb function and a reasonably sized ulnar digit, performing a web space reconstruction to optimize the span between the fingers is the only treatment that is required. If the border digits are small, it is important to assess the thumb for a functional carpometacarpal joint that allows a "mobile post." If the thumb acts as a mobile post, then augmenting the size of the digits can improve the hand's overall function. The accompanying digit(s) can be augmented by a nonvascularized toe phalangeal transfer, on-top-plasty of adjacent digit, or distraction lengthening of the metacarpal. For these treatment methods, key points to remember are:

1. The viability and growth potential of a nonvascularized toe phalangeal transfer is maximized when done in a patient less than 18 months of age, although the indications have narrowed.
2. Distraction lengthening, because it is done gradually and over a time period of up to several weeks, requires close monitoring of the patient.
3. On-top-plasty is a relatively difficult procedure requiring competency in pediatric hand surgery (Fig. 7).

Cleft Hand

Cleft hand	
Nonoperative management	
Indications	**Contraindications**
Few indications before surgery	None
Determining prehensile pattern of the child	
Cleft hand	
Physical/occupational therapy recommendations	
Assessment of prehensile function	
Education of parents	
Participate in the decision-making process	

Cleft hand is one of the more striking congenital anomalies encountered by the hand surgeon. The condition represents a failure of formation characterized by a longitudinal deficiency of the central rays of the hand. There are two distinct types of "cleft" hands – typical and atypical cleft hands. Although these forms can appear similar, there are important distinctive differences. Atypical cleft hand is really a form of symbrachydactyly. Atypical cleft hand is usually unilateral, presents with a U-shaped central defect, and occurs spontaneously (Miura and Suzuki 1984; Fig. 8). The classic cleft hand has a V-shaped defect in the central portion with variable proximal extension (Fig. 9). Extreme cases may extend into the carpal bones. The adjacent border digits can have syndactyly (Kozin 2003). The typical patient has bilateral involvement, although the extent is variable. In addition, clefts of the feet as well as cleft lip and palate are

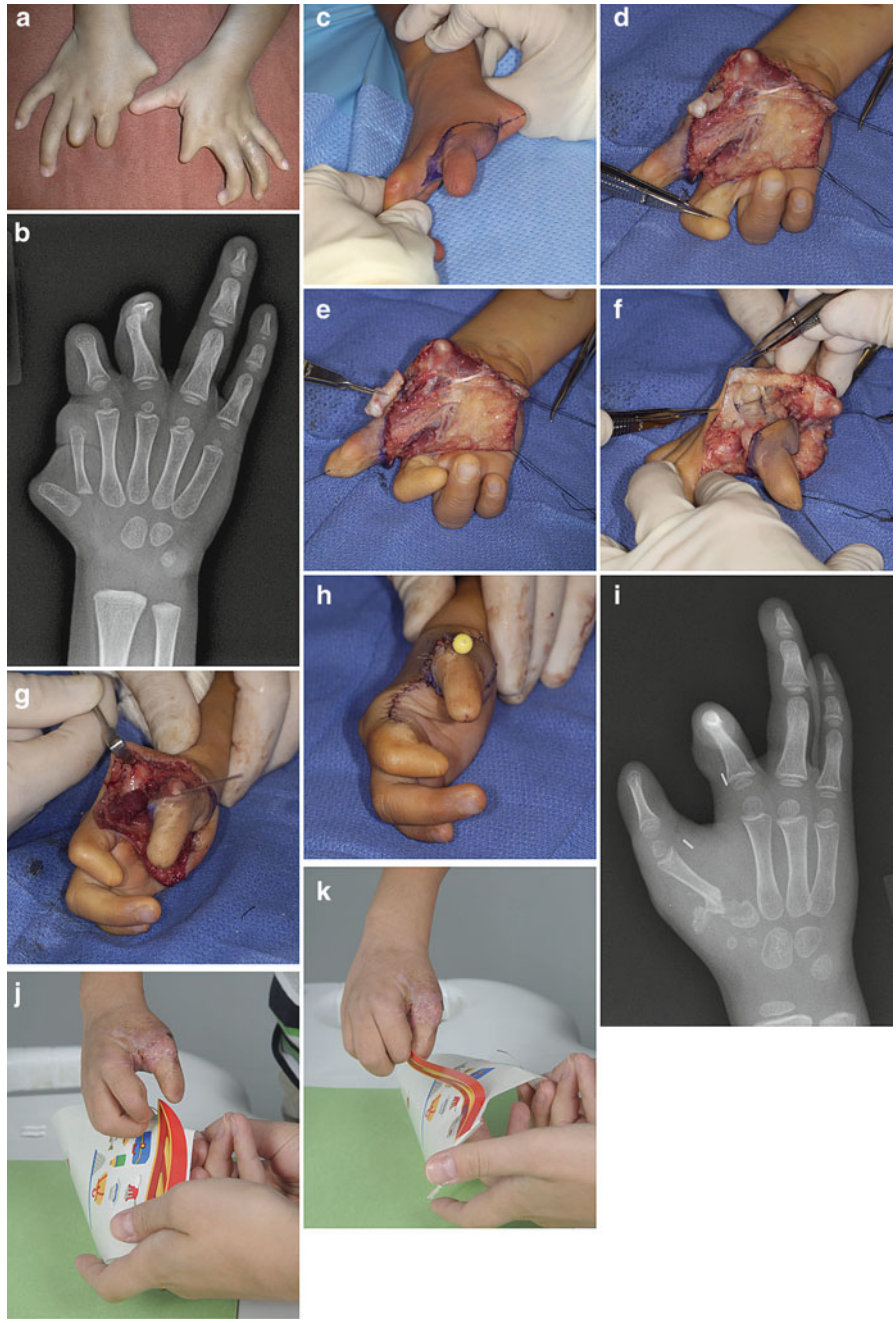

Fig. 7 Three-year-old male with brachydactyly of both hands (Courtesy of Shriners Hospitals for Children, Philadelphia). (**a**) Right hand is missing the terminal portions of the thumb, index, and long finger. On-top-plasty with placement of the index finger on top of the thumb was recommended. (**b**) X-rays coincide with clinical picture. Thumb with adequate carpometacarpal joint and extra metacarpal between thumb and index. (**c**) Skin design for on-top-plasty. (**d**) Exposure of extra metacarpal between thumb and index. (**e**) Removal of extra metacarpal. (**f**) Dorsal dissection with preservation of veins. (**g**) Transposition of index on top of thumb metacarpal. (**h**) Skin closure with wide thumb-index web space and elongated thumb. (**i**) X-ray following healing of index metacarpal to thumb metacarpal. (**j**) Clinical outcome with ability to open for pinch. (**k**) Grasping of sticker

associated with the hand anomaly. The incidence of cleft hands is reported as 1:10,000–1:90,000 live births (Barsky 1964; Ogino et al. 1986). The inheritance pattern is often autosomal dominant with variable penetrance, although autosomal recessive and X-linked forms have been documented. The genetic factors for certain forms of the condition have been identified in patient with split-hand-foot malformation (Dlx homeobox abnormalities) and ectrodactyly-ectodermal dysplasias (transcription factor gene p63 affecting Dlx homeobox regulation) (Waters and Bae 2012b). Failure in the maintenance of the central portion of the apical ectodermal ridge is the proposed etiology (Al-Qattan and Kozin 2013). In an animal model, chemically induced abnormalities in digital ray formation have indicated lead to central polydactyly, syndactyly, and central deficiency (Naruse et al. 2007).

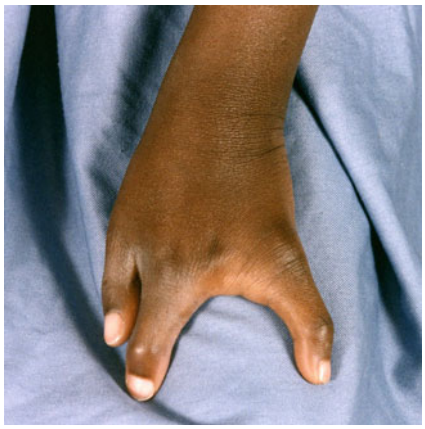

Fig. 8 Atypical cleft hand (a.k.a. symbrachydactyly) with unilateral U-shaped central defect (Courtesy of Shriners Hospitals for Children, Philadelphia)

Classifying the various forms of typical cleft hand continues to be a challenging task due to the wide variability in clinical findings. The three most often-referenced classification schemes describe the deformity according to either the number of digits missing (Nutt and Flatt 1981), the number of rays present together with associated bony syndactyly and polydactyly (Ogino 1990), or the functional integrity of the thumb-index finger web space (Manske and Halikis 1995). While each of these classification systems describe the anomaly differently, they are individually important in that they identify specific elements seen in cleft hand that need to be addressed when considering treatment alternatives.

Because the cleft hand deformity can be striking in appearance, its mere presence can be a source of stress for the family. During the initial evaluation, the examiner can comfort the situation by resisting the temptation to immediately focus on the hand condition. Also, being sensitive to the family's fears and concerns can be beneficial toward establishing a good physician-patient relationship that will last many years. The importance of allowing time for questions cannot be

Fig. 9 Typical bilateral cleft hand with V-shaped defect in the central portion (Courtesy of Shriners Hospitals for Children, Philadelphia)

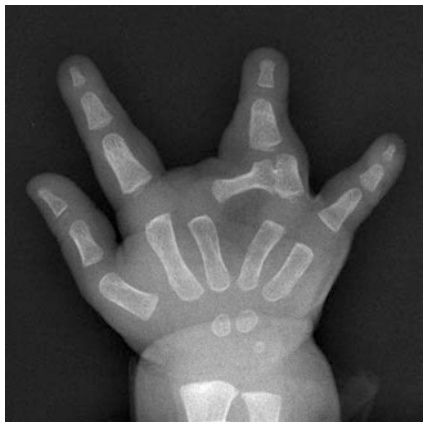

Fig. 10 X-ray of a 3-year-old with cleft hand. Anteroposterior view shows a proximal phalanx within the cleft oriented in a transverse direction (Courtesy of Shriners Hospitals for Children, Philadelphia)

overemphasized. When it can be arranged, most families are referred for an evaluation by a genetics specialist. When evaluating the affected hand, a thorough assessment of every anatomic structure is critical. Take note of what is present and what is missing, of which structures look "normal" and those that appear abnormal. The location and size of the cleft is important as well as any compromise of the thumb-index web space. Most clefts are found in the center of the hand, and various forms of additional phalangeal or metacarpal irregularities (such as hypoplasia, duplications, and unusual shapes) can be present (Falliner 2004) (Fig. 10). Obtaining radiographs of the affected extremities is helpful for assessing bony anatomy, but the findings can be misleading in the very young patient due to incomplete or delayed ossification of abnormal bony elements. Thus, serial radiographs as the child grows provide better information in regard to potential surgical treatment plans.

Before discussing specific operative procedures, it is important to state that a number of these children do not require surgery to improve their function. In spite of the obvious cleft in the hand, they are able to pinch and grasp both small and large objects with remarkable dexterity. Even hands with a single mobile digit can function at a high level (Fig. 11). Therefore, any proposed surgery must be designed to enhance function. Having the patient's hand assessed by a qualified therapist can be very helpful in formulating treatment plans. Lastly, there should be no pressure to "operate early" on cleft hands. As the child grows, observing their abilities over time allows for more confidence when recommending a surgical procedure.

While there is no single "right way" to surgically treat cleft hand, however, there are some general established treatment principles. These principles focus on functional improvement and include altering the configuration of the cleft, maximizing the position of the thumb, and improving the functionality of stiff or malaligned digits (Waters and Bae 2012b). In addition, improving the aesthetic appearance of the hand, particularly those with severe deformities, is a reasonable request of many families. Fortunately, decreasing the depth of the cleft will enhance both the appearance and function. However, the family should be cautioned that surgery done solely for aesthetic purposes may be disappointing as no operation will produce "normal" looking hands.

Before discussing specific surgical procedures, an additional clinical factor needs to be considered. Closure of the cleft can be proposed for any patient with cleft hand. In most hands, reconstruction of the cleft is a worthwhile endeavor because holding small objects within the palm is easier and the aesthetic appearance improved. However, in a few children (e.g., children with only two or three digits), the cleft enhances function for grasping large objects. Cleft closure is contraindicated as function trumps form.

The technique of cleft closure varies with extent and particular anatomy. Minor clefts may be treated by Z-plasty incisions combined with a hexagonal flap (Barsky flap) from a digit adjacent to the cleft for reconstruction of the web commissure. A deeper (or wider) cleft may require reconstruction of intermetacarpal ligament between the heads of the metacarpals. Local tissue or tendon graft can be used to connect the metacarpal heads to lessen the chances of the digits splaying with hand growth (Ogino 1990). The metacarpal physis must be avoided during intermetacarpal ligament reconstruction. If anomalous bony structures are encountered in the cleft, reconstruction

Fig. 11 Six-year-old boy with bilateral cleft hands that have only a single mobile digit that functions at a high level (Courtesy of Shriners Hospitals for Children, Philadelphia). (**a**) Clinical picture of both hands. (**b**) Holding a hockey stick. (**c**) Wakeboarding

becomes more challenging. These abnormal phalanges or metacarpals are often small and poorly positioned, so excision is usually the best treatment. There are occasions, however, when these bones provide structural support to a joint and partial preservation is necessary during cleft reconstruction. When the middle metacarpal is completely absent, closure of the cleft space can be impeded by the carpometacarpal articulations of the adjacent fingers. For these hands, a V-shaped osteotomy in the center of the carpals or transferring the second metacarpal base to a more central position will allow for improved closure and digit alignment (Ogino 1990).

As the severity of the cleft increases, the status of the thumb-index web space usually becomes more narrowed. The thumb-index web space must be addressed. If this web interval is merely contracted, employing single or multiple Z-plasties widens the web space (Riley and Burgess 2009). However, when the thumb and index finger are almost syndactylized (Manske and Halikis Types IIB & III), reconstructing the web space requires the use of additional skin. In these cases, additional soft tissue coverage for the web is achieved by using a dorsal transposition skin flaps (Flatt 1994c).

Eloquent and technically demanding procedures have been described for treating both the deep cleft and the thumb web contracture simultaneously (Snow and Littler 1967; Rider et al. 2000; Miura and Komada 1979; Upton and Taghinia 2010). The basic concept is to widen the thumb-index web space, close the cleft, and resurface the thumb-index commissure with skin from the cleft. In order to optimize hand function,

additional abnormalities like interphalangeal joint flexion contractures, angulation deformities, or anomalous bony structures can also be addressed at the same time. Subsequent hand therapy and splints designed to maintain the thumb-index web space can be beneficial. Potential complications include skin flap necrosis and pin tract infection.

Technique: Snow-Littler Reconstruction of Cleft Hand

Cleft hand Preoperative planning
OR table: regular
Position/positioning aids: supine
Fluoroscopy location: ipsilateral
Equipment: standard, wire driver, K-wires
Tourniquet: sterile

The patient is placed supine on the operating room table (Fig. 12a, b). The procedure is usually performed under general anesthesia. A single dose of intravenous preoperative antibiotics is administered. The limb is prepped and draped in sterile fashion. Chlorhexidine gluconate and alcohol prep (ChloraPrep; CareFusion, Leawood, Kansas, USA) is preferred, which may be more effective in eliminating bacteria and avoids iodine that can migrate beneath the tourniquet and cause burns (Saltzman et al. 2009). A sterile pediatric tourniquet (Delfi Medical Innovations, Vancouver, Canada) is placed on the upper arm that exsanguinates during application.

The hand is carefully examined in anticipation of web space widening and cleft closure (Fig. 12c, d) Initial attention is toward the design of the palmar-based skin flap from the cleft (Fig. 12e, f). Lines are drawn along the dorsum of the ring and index fingers coalescing in a V-shaped apex and the bottom of the cleft. The incisions course around the sides of the ring and small metacarpophalangeal joint toward the palm. The proximal extent is to the level of the apex of the dorsal V. A small Barsky flap is designed from the adjacent ring finger for commissure reconstruction after clef closure. An additional incision is necessary to separate or widen the thumb-index web space. In cases that require only widening, a straight incision is sufficient. In cases with syndactyly, then a zigzag incision will need to be incorporated into the design to accommodate syndactyly separation.

The web space flap is elevated first from the dorsal side (Fig. 12g). The skin is sharply incised and dissection carried directly to the paratenon. The dorsal veins are ligated and preserved within the flap. Once the dorsal aspect is raised, dissection proceeds from the dorsal side to the palmar side (Fig. 12h). On occasion, an artery entering the flap van be preserved, which changes the flap from random to axial. This axial flap is much more robust compared to the random flap. The neurovascular bundles to the index and ring finger are identified and protected (Fig. 12i).

Once the flap is raised, attention is directed toward separation of the thumb and index and widening of the thumb-index web space. The skin is incised and the fibrous interconnections cut (Fig. 12j). The web space is widened with slow and deliberate dissection. The princeps pollicis artery and its braches must be identified. The intervening muscles, such as the first dorsal interosseous and adductor pollicis, may require release until adequate widening has been obtained (Fig. 12k, l).

Attention is then directed to the index finger. In cleft hands with a long finger metacarpal, the index finger is transpose to the long position. In cleft hands without a long finger metacarpal, a closing wedge osteotomy at the base of the index is performed to close the cleft and align the finger and widen the thumb-index web space (Fig. 12m, n). Fixation is usually accomplished with Kirschner wires (Fig. 12o). Addressing the bony alignment negates the necessity of intermetacarpal ligament reconstruction. The surgeon must ensure that bony alteration does not result in digital scissoring.

A small Barsky flap from the long finger is raised and inset to reconstruct the commissure (Fig. 12p). The cleft skin is transposed into the widened thumb-index web space (Fig. 12q). The skin is trimmed and closed with 5–0 plain suture (Fig. 12r, s). Following closure, the tourniquet is

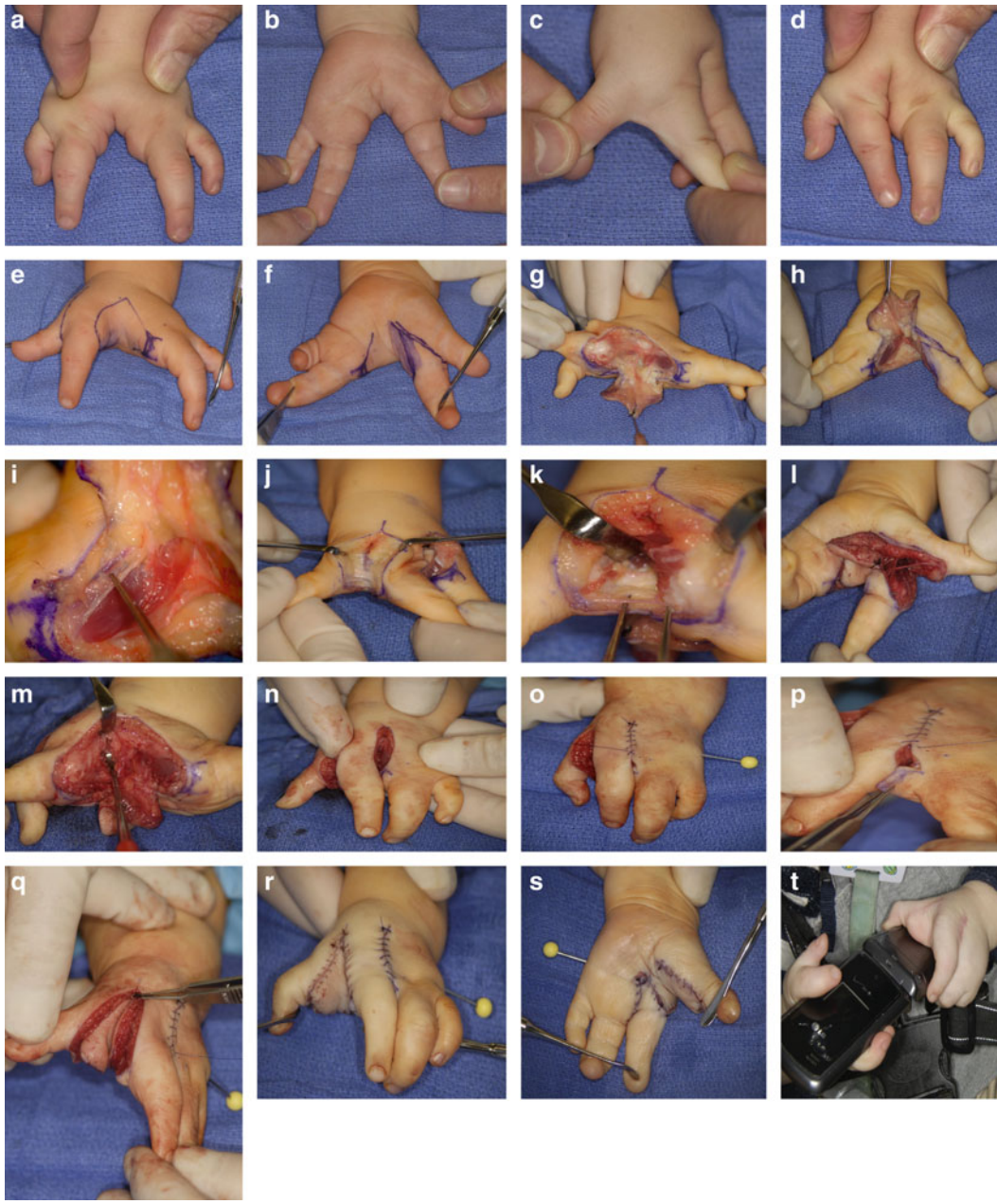

Fig. 12 Sixteen-month-old child with bilateral cleft hand (Courtesy of Shriners Hospitals for Children, Philadelphia). (**a**) Left hand dorsal view. (**b**) Left hand palmar view. (**c**) Narrow thumb-index web space. (**d**) Intended cleft closure. (**e**) Dorsal skin design. (**f**) Volar skin design. (**g**) Flap elevation from dorsal to palmar with preservation of an axial artery to the flap. (**h**) Continued dissection along the palmar aspect of the hand. (**i**) Protection of the adjacent neurovascular bundles. (**j**) Incision for widening of thumb-index web space. (**k**) Protection of princeps pollicis artery and division of intervening muscle. (**l**) Volar view following adequate release of the thumb-index web space. (**m**) Closing wedge osteotomy at the base of the index to close the cleft and align the finger. (**n**) Improved alignment of index finger and cleft closure following osteotomy. (**o**) Kirschner wire fixation of osteotomy. (**p**) Barsky flap elevated from long finger for commissure reconstruction. (**q**) Cleft skin rotated into widened thumb-index web space. (**r**) Dorsal closure. (**s**) Palmar closure. (**t**) Incorporation of hand into activities of daily living, such as grasping a large object

deflated to ensure capillary refill to all digits. Adequate fluffy dressings are necessary to equalize the anterior-posterior and medial-lateral dimensions of the hand. This dressing allows uniform compression without constriction. A long-arm soft cast (3M™ Scotchcast™ Soft Cast Casting Tape, St. Paul, Minnesota, USA) is applied with the elbow flexed to greater than 100° to decrease the chance of accidental removal. This fiberglass casting tape does not harden completely, but remains slightly flexible when cured. More importantly, soft cast can be unwrapped in the clinic avoiding the petrifying cast saw. The child is admitted overnight with the arm elevated to promote venous drainage. The entire hand can be covered the next day if vascularity has been maintained throughout.

Cleft hand Surgical steps
Limb is exsanguinated and the tourniquet is inflated
Cleft flap design along with skin incision for thumb-index web space release
Elevate dorsal cleft flap at the level of paratenon with venous preservation to the flap
Elevate volar portion of flap with protection of index and longer finger neurovascular bundles
Look for potential arterial axial supply to flap
Incise skin between thumb and index finger
Gradual release and widening of thumb-index web space. Protect princeps pollicis artery
Index finger transposition or wedge osteotomy dependent upon osseous anatomy
Kirschner wire fixation
Inset cleft flap into thumb-index web space
Cleft closure with Barsky flap for commissure
Skin is closed with absorbable suture, and the limb is immobilized in a long cast for 4 weeks

Cleft hand Postoperative protocol
Limb is immobilized in a long-arm cast for 4 weeks
Kirschner wire is removed and a short arm splint is fabricated
Active range of motion and scar care is initiated
Gradual incorporation of hand into activities of daily living (Fig. 12t)
Passive range of motion is begun to prevent digital stiffness once bony union is confirmed

In summary, the child with cleft hand anomaly has a challenging condition. Some cleft hands represent "functional triumphs and aesthetic disasters" (Upton and Taghinia 2010, p. 480). Treating cleft hand is not for the inexperienced surgeon or for the faint of heart. A detailed evaluation of the child, his or her hand, and their functional abilities is critical prior to recommending any surgery. As function trumps form, not recommending surgery may be difficult for the family to accept. When surgery is advised, one must make certain that the family understands the details of the procedure and the anticipated benefits. Unrealistic aesthetic and functional expectations must be avoided. The family must understand that as the child's hand grows, revision operations to address joint contractures, web space contractures, and angular deformities of the digits may be necessary. Hence, frequent and regular follow-up evaluations are until the child's growth is complete.

Transverse Deficiencies

Another failure is congenital transverse deficiency. This anomaly can occur at any level from the phalanges to the humerus and is named according to the most distal remaining bone segment. In the typical patient, the condition is unilateral and is not seen with other congenital abnormalities. This deformity occurs sporadically, and familial involvement has not been demonstrated. The condition should not be confused with constriction band syndrome (discussed elsewhere in this textbook), whereby the entrapment of developing tissue by an amniotic band may lead to a transverse-type amputation of the extremity. The clinical appearance of these two conditions is at times quite similar, but if the diagnosis is in question, the patient with amniotic band syndrome often shows signs of additional bands or creases in the affected limb or other extremities. Another difference is that transverse deficiencies generally occur more proximal compared to amputations secondary with constriction band syndrome (Ogino and Saitou 1987). In a review of published series, the incidence of the

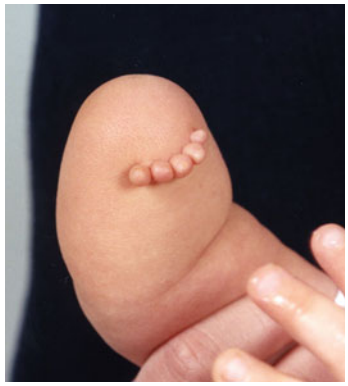

Fig. 13 A transverse deficiency of the proximal one-third of the forearm with terminal nubbins (Courtesy of Shriners Hospital for Children, Philadelphia Unit)

transverse deficiencies has been estimated to be about 6 % of all congenital upper extremity anomalies (Jain and Lakhtakia 2002).

Amputation at the level of the forearm is the most commonly encountered transverse deficiency of the upper extremity. The proximal one-third of the forearm is most frequent (Kozin 2003; Fig. 13). The deficiency can also be found at the hand and wrist regions, but transverse loss at the humeral level is rare (Knight et al. 2012). There are variations in the underlying bony structures and the type of tissues seen at the distal portion of the limb. When the amputation occurs in the forearm, there have been additional bony anomalies identified such as radioulnar synostosis and radial head dislocation (Jain and Lakhtakia 2002). With reference to overlying skin, some patients have a bulbous-like skin and soft tissue coverage. Additionally, structures like finger nubbins (that may have nails), skin invaginations, and even varying degrees of bone hypoplasia are commonly seen. Because the rudimentary nubbin-like structures are similar to those seen in symbrachydactyly, recent work has investigated the relationship between the two conditions. The authors propose that symbrachydactyly and transverse deficiency represent a continuum with different severities of a similar mesodermal process (Kallemeier et al. 2007). The etiology of the transverse deficiency is thought to be due to disruptive events affecting the apical ectodermal ridge after the limb bud forms (Al-Qattan and Kozin 2013).

In terms of functional adaptation, the child that presents with an isolated upper extremity transverse deficiency is often able to adjust to their difference. In fact, a recent study evaluated children with unilateral congenital below-elbow deficiency found that compared to the general population, they "do not perceive their health-related quality of life to be diminished" (James et al. 2006). Regardless of this fact, there will inevitably be questions raised about the applicability of a prosthetic device for the child. The discussion should include that consistent wearing of any upper extremity prosthesis is a challenging and unrealistic goal. Of note, James et al. (2006) looked at various outcome measures in patients with congenital below-elbow amputations. These patients took a battery of assessment examinations, and the results indicated that non-wearers of a prosthesis performed as well or better than wearers on both musculoskeletal health questionnaires and functional tests. In their patient population, actual prosthetic wear was found to be about 65 %. The authors concluded that while there may be social situations where a prosthesis can provide some benefit, the devices themselves did not significantly improve the child's function. It is also important for the family to realize that because the device covers the residual limb, sensation from the distal aspect of the extremity is negated. This loss of "sensory feedback" appears to be a substantial reason for a child's reluctance to wear any type of device. The classic recommendation for prosthetic prescription is to fit the patient with a passive terminal prosthesis around 6 months of age to assist with bimanual tasks and independent sitting. As the child matures and functional needs change, a more active (body-powered or myoelectric) terminal device prosthesis is to be used. This standard recommendation is no longer applicable based upon the data available. One study examined the types of prostheses used by below-elbow congenital amputees who were determined to be consistent wearers. The authors found that these successful users generally choose among multiple devices based upon functional need and as such should be provided with multiple prosthetic device options

Fig. 14 Eight-year-old competitive swimmer with right below the elbow transverse deficiency (Courtesy of Shriners Hospitals for Children, Philadelphia). (**a**) Clinical picture. (**b**) Swimming prosthesis to facilitate competition

(Crandall and Tomhave 2002). Ultimately, it is often difficult to predict whether or not a child will become a consistent prosthetic user. An adaptive prosthesis is useful when a child wants to succeed in a particular task or activity (Fig. 14). Fitting the child with a device to accomplish a specific goal is rewarding.

As far as other treatment options are concerned, there are some specific instances when surgical intervention may be indicated. For example, there are cases where the soft tissue coverage at the terminal end of the amputation site shows substantial invaginations (or creases) that can be concerning for potential skin maceration issues. If this becomes a consistent hygiene problem, excision of the involved skin "crease" and rotation of a local full-thickness skin flap to cover the defect is the most effective treatment. Another possible surgical intervention that parents may consider involves excising the skin nubbins or "tiny digits" that can occur at the distal portion of the congenital amputation site. This is a reasonable treatment alternative, although some children use their nubbins to assist in the holding of objects and provide additional sensory feedback to the extremity. Lastly, there are more extensive procedures that have been proposed to treat terminal deficiencies. When the deficiency occurs at the hand level, microvascular toe-to-hand transfer has been reported to help supplement the extremity's function (Kozin 2003). Distraction osteogenesis is another option for children with short residual limbs. Lengthening a short amputation segment allows better prosthesis fitting (Seitz 1989; Jasiewicz et al. 2006; Alekberov et al. 2000). There are a limited number of cases reported, but most patients benefited from this treatment.

Treatment Strategy

The majority of patients with unilateral transverse deficiency require no surgical treatment. Even children that have "tiny digits" at the terminal end of the limb seem to appreciate the added sensory feedback that these appendages provide and opt to not have them removed. The exception to nonsurgical care is in the case of a child with a short forearm segment in a below-elbow terminal deficiency. Lengthening of this short forearm yields an improved ability to hold objects in the crook of the elbow and allows for a greater variety of prosthetic device alternatives.

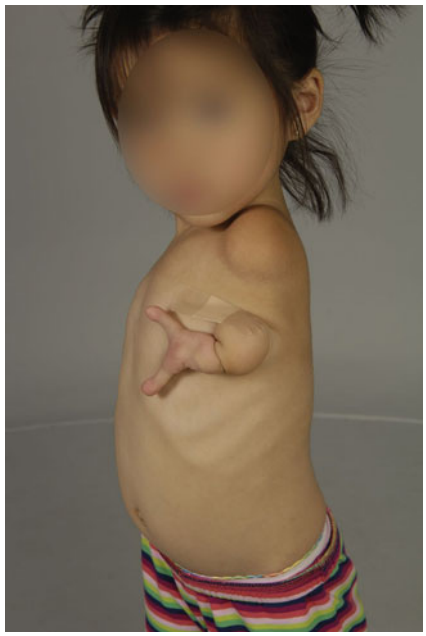

Fig. 15 Five-year-old child with phocomelia variant and the hand attached directly to the trunk (Courtesy of Shriners Hospitals for Children, Philadelphia)

Phocomelia

Phocomelia is a developmental anomaly characterized by the presence of a normal hand and the absence or hypoplasia of the proximal portion of a limb. A patient with this anomaly has the physical appearance of the hand being attached directly to the trunk (Fig. 15). This failure of formation deficiency is classified as an intercalary defect, which implies the loss of an intervening segment in the extremity. The condition is a rare deformity, comprising less than 1 % of all upper extremity congenital anomalies (Flatt 1994a). In about half of cases that have been identified, the children also have organ system defects (Bermejo-Sanchez et al 2011a). There is no specific inheritance pattern for phocomelia as most cases are due to either a spontaneous mutation or occur as part of a congenital syndrome. The specific factors that cause phocomelia continue to be investigated. Current theories have focused on deficient cell division in the limb bud (Al-Qattan and Kozin 2013), as well as inhibition of angiogenesis and/or progenitor cell survival in the developing limb (Galloway et al. 2009). Unfortunately, the anomaly is probably best known for its association with maternal use of the drug thalidomide for emesis during early pregnancy in the late 1950s. It is estimated that over 10,000 infants may have been born with severe birth defects related to thalidomide. Although thalidomide was banned in 1962, the drug has been used recently as a treatment for leprosy and cancer. Regrettably, there have also been new cases of thalidomide-related phocomelia reported in leprosy-endemic regions (Bermejo-Sanchez et al 2011a; Vargesson 2009). Strict birth control is necessary for women taking thalidomide. Other medications have been implicated as a possible cause of phocomelia. For example, a recent report described an infant with a limb reduction disorder similar to phocomelia that was born to a mother taking carbamazepine for control of seizures (Dursun et al. 2012).

On the clinical level, phocomelia was originally described to have three basic morphological typologies (Frantz and O'Rahilly 1961). Type I (sometimes referred to as true phocomelia) is the form wherein the hand is attached directly to the trunk and no other intervening osseous structures are found. The other two types have additional hypoplastic bone elements that can be identified between the hand and thorax. A child with type II phocomelia has the hand and forearm connected to the trunk; while in type III, the hand is attached to a humerus. However, our current understanding of the embryological development of the limb along three longitudinal axes of formation (see chapter ▶ "Embryology") will likely change the way scientists come to classify phocomelia. In a recent paper, the authors reviewed 60 cases that were diagnosed as upper extremity phocomelia. After a critical analysis of the clinical and radiographic findings, they determined that phocomelia may actually "represent a spectrum of severe longitudinal dysplasia" because none of their cases had a true intercalary deficiency (Goldfarb et al. 2005). Another study questioned the existence of "true" phocomelia because of the fact that when intercalary defects are seen, the hand and glenoid were abnormal as well (Tytherleigh-Strong and Hooper 2003). Ultimately, it is felt that as the medical community develops a greater

understanding of both the developmental biology and the specific genetics of this particular limb deficiency, the ability to classify what is called "phocomelia" will improve as well.

All discussions about potential treatments for this condition should focus on making the child as independent with activities and self-care as possible. Most commonly, these children will benefit from using prostheses, however; the specific types prescribed will depend both on his or her functional requirements and the ability of the other extremities to accomplish those needs. Ample shoulder control and good stability of the trunk are advantages to children with phocomelia. Fitting of the device can be challenging because of the size and orientation of the residual limb. Surgical care is rarely needed in this anomaly (Kozin 2003). There may be certain indications for surgery to promote prosthetic device wear, such as excision of painful bony prominences about the residual limb.

Amelia

Amelia is defined as the congenital anomaly characterized by the complete absence of one or more limbs. Since this diagnosis is very rare, little information has been reported about this condition. A recent review study from 20 congenital anomaly surveillance programs found that among both live and stillbirths, the incidence of amelia is 1.41 per 100,000. The upper limbs are affected slightly more frequently than the lower extremities, and single extremity involvement occurs about 65 % of the time. Interestingly, this same study determined that the frequency of amelia was higher among mothers less than 20 years of age, and 69 % of the reported cases had multiple congenital anomalies including anencephaly, cardiac septal defects, and other musculoskeletal deformities (Bermejo-Sanchez et al 2011b).

The exact mechanism causing amelia is unknown. Because the condition represents a failure of formation, the prevailing theories focus on either an error in the molecular biology processes of the developing limb (such as loss of specific fibroblastic growth factors (Al-Qattan and Kozin 2013)) or an interruption in the vascular supply to the limb during the early embryological stages. The actual causes are likely multifactorial; however, there have been cases of amelia seen in monozygotic twins that have vascular anastomoses between their placentas. The subsequent limb anomalies were thought to be due to alterations in the local arterial supply (Bermejo-Sanchez et al 2011b).

The treatment principles for the child with amelia are similar to those mentioned for phocomelia, except that there are really no surgical indications. As was seen in patients with transverse deficiency and phocomelia, the children with unilateral upper extremity involvement adapt remarkably. The option of prosthetic device use is routinely considered, although there are unique challenges with regard to device construction and usage in these patients. Difficulties include the lack of scapulohumeral motion to power the prosthesis, limited bony and soft tissue anchor points to keep the device in place, and limited sensory feedback to the patient. In addition, for children with bilateral amelia, the prosthesis can be "heavy" and the harnessing apparatus even more complicated. In cases of unilateral amelia, the child may decide not to wear a well-designed prostheses because they can function well without the device.

Treatment Strategy

Since amelia is such a rare condition, most physicians will never encounter a case in their career. Even though operative care is not required for these children, the surgeon and his or her team play an important role in coordinating additional evaluation, providing suggestions to enhance function, and supporting their psychosocial needs.

Summary

Congenital upper extremity anomalies that are characterized by undergrowth and/or failure of formation represent a broad category of deformities. The majority of these malformations occur in a sporadic fashion without a family history.

This group of anomalies has a wide range of clinical manifestations, and each affected child presents with unique capabilities and challenges. As such, treatment strategies must be individualized and proposed only after a careful assessment of the child's anatomy, functional abilities, and long-term needs.

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