

Helping Hands: The Management of Congenital Hand and Upper Extremity Anomalies

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Congenital hand and upper extremity malformations vary in anatomic location and severity. Many anomalies are slight and present no functional or aesthetic issues, while others are severe enough to cause disability. In addition, some anomalies may serve as indicators of potentially life-threatening systemic disorders. Prompt referral to an upper extremity specialist is recommended so that parents can receive appropriate counseling and reassurance, along with comprehensive explanation of the condition and treatment plan. However, since a pediatrician is likely to be the first physician to speak with the family, it would be ideal for pediatricians to be able to discuss the condition and treatment options, and to recognize the anomalies that require screening for related comorbidities. Also, pediatricians are uniquely positioned to help parents cope with the difficult emotional responses that are prompted by the birth of a child with a physical abnormality.

Educational Objectives:

At the conclusion of this activity participants will be able to:

- Describe the more common hand and upper extremity anomalies.
- Describe treatment approaches for hand and upper extremity anomalies.
- Recognize conditions associated with common hand and upper extremity anomalies.

Congenital upper extremity anomalies occur in 0.2% of live births.^{1,2} Since several minor deformities are underreported because they present insignificant functional or aesthetic problems, the true incidence is certainly much higher. Approximately 10% of diagnosed congenital hand anomalies are serious enough to warrant surgical intervention.³

The majority of upper extremity malformations have no identifiable cause, but genetic and environmental factors are implicated in some cases. It is estimated that 30% of these anomalies result from gene mutations or chromosomal abnormalities, and that environmental and teratogenic agents account for 10%. The remaining 60% are idiopathic.^{3,4}

Treatment approaches

The goal for care of congenital hand and upper extremity anomalies is to optimize the following: ability to orient the hand in space, sensate skin coverage, grasping power, and precise handling of objects.⁵ Ultimately, the objective is to help the child become a fully independent adult. With this in mind, the temptation to create a cosmetically normal hand should be rejected if the child will be left with less functional capacity.

Management of upper extremity anomalies should begin with counseling soon after birth. Initial consultation includes a clear explanation of the child's anomaly and statement of short-term and long-term treatment plans. Depending on the child's anomaly and its severity, the treatment plan may include occupational therapy, stretching, splinting, casting, prostheses, and/or reconstruction surgery. Many minor anomalies, however, may not need any form of treatment.

If reconstructive surgery is needed, timing is critical. Ideally, surgical reconstruction should be completed by school age, so that the child may adapt to and fully use the reconstructed limb before confronting the functional and social challenges of school.⁶ Suggested surgical timing guidelines are listed in Table 1 (page 15). These are general guidelines, and surgical timing may vary according to the emotional preparedness of the family and the status of other medical problems that take precedence. General medical evaluation by the pediatrician is recommended prior to surgery.

Furthermore, embryologic formation of the upper extremities occurs simultaneously with other major organ systems, and thus concomitant

cardiovascular, craniofacial, musculoskeletal, genitourinary, and neurological anomalies may be found. Certain upper extremity anomalies are known to be associated with specific systemic disorders, some of which are not clinically evident until the child is older. As such, the more readily identifiable upper extremity anomaly may serve as a clinical marker for a clinically silent condition. The pediatrician's awareness of these associations may facilitate earlier intervention than might occur otherwise.

Common hand anomalies

Polydactyly

Polydactyly is the most frequently reported congenital hand anomaly. Any digit can be completely or partially duplicated. In many cases, hand function is not diminished.

Radial polydactyly (preaxial): The thumb may be duplicated anywhere from the metacarpal to the thumb tip. Both the radial and the ulnar duplicates always display some degree of hypoplasia. The duplicated thumbs may be abnormally angulated, and the joints are usually stiff. Because of the importance of the thumb in prehensile activities, thumb polydactyly almost always interferes with normal hand function.⁷

The goals of reconstructive surgery are to remove the most hypoplastic thumb of the duplicated pair and to reconstruct the retained thumb so that it has maximal stability, proper alignment, optimized motion, and normal sensation. Most cases of radial polydactyly are not associated with systemic problems.

Central polydactyly: Central polydactyly involves the index, long, and/or ring fingers, and may be coexistent with syndactyly. Central polydactyly is often transmitted as an autosomal dominant trait and may be associated with toe polydactyly. Treatment is almost always surgical, consisting of resection of the least developed digit, and, if necessary, repair of the remaining digits.

Ulnar polydactyly (postaxial): Polydactyly involving the little finger can vary from

rudimentary nubbins (skin "tags") to well-formed, completely duplicated fingers. Ulnar polydactyly is usually transmitted in autosomal dominant pattern, but penetrance is variable. Nubbins are 10 times more common among African American patients compared to other groups. Concomitant anomalies are uncommon in African American children, but white children with ulnar polydactyly are more likely to have an associated anomaly or syndrome.⁸ Several disorders and syndromes are associated with ulnar polydactyly in whites, including polycystic kidney disease, bladder obstruction, cardiac defects, imperforate anus, cataracts, deafness, hydrocephalus, and mental retardation.

The historic treatment of skin nubbins has been suture ligation in the newborn nursery. However, a more cosmetic scar and less bleeding can be achieved when these nubbins are removed with local anesthesia and sharp resection. In the more complete ulnar polydactyly, the most hypoplastic digit is removed, and the remaining digit is reconstructed as necessary.

Syndactyly

Syndactyly is the failure of separation between 2 or more digits. It is classified as complete syndactyly when the connection between fingers extends to the finger tips, and as incomplete when the connection terminates proximal to the finger tip. Syndactyly is further classified as simple if only skin and subcutaneous tissues are involved, and as complex if the bones of adjacent fingers are conjoined as well. The most common location of syndactyly is between the middle and ring finger, followed by ring-small, index-middle, and thumb-index.

Syndactyly is most common in white, male children.⁹ It may occur sporadically, or as an autosomal dominant trait with variable expressivity and incomplete penetrance. Complex syndactyly can be associated with a number of syndromes, and its presence should trigger a search for other syndromic features.

Timing of surgery to correct syndactyly depends on the fingers involved (Table 1). If the involved fingers are of equal length, surgery can be

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Syndactyly: Before and after surgery

deferred until 18 months of age. Skin grafting from a donor site is usually necessary during corrective surgery because the circumference of 2 conjoined fingers is less than the subsequent circumferences of the separated fingers.

Radial longitudinal deficiency (radial clubhand)

This deficiency involves the radial side of the forearm and the thumb side of the hand. The radius is short or absent, and the ulnar is short and bowed. As a result, the forearm is short, and the wrist is radially deviated. This forearm abnormality is variably accompanied by complete or partial absence of the thumb, radial carpus, and thenar muscles.

Radial deficiency is usually associated with systemic disorders, cardiac abnormalities (eg, Holt-Oram syndrome), aplastic anemia (Fanconi's anemia), thrombocytopenia (TAR), and VACTERL pattern of congenital anomalies (vertebral, anal, cardiac, tracheal, esophageal, renal, and limb).¹⁰

Treatment for radial deficiency begins with stretching, serial casting, and splinting. Surgical reconstruction is performed between 6 months and 1 year of age.

Ulnar longitudinal deficiency (ulnar clubhand)

Ulnar deficiency is 4 times less common than radial deficiency.^{11,12} This anomaly involves the ulnar side of the forearm and the medial digits of the hand. It is characterized by partial or complete ulnar absence. Half of the patients with ulnar deficiency have associated defects of the musculoskeletal system, including syndactyly, proximal femoral focal deficiency, and clubfoot.

In contrast to the radial longitudinal deficiency, systemic conditions rarely accompany ulnar deficiency. However, at initial presentation, the physical appearance of an ulnar deficiency may be similar to that of a radial deficiency. It is adequate for a general pediatrician to simply identify the presence of a

longitudinal deficiency and refer the child to an upper extremity specialist who will differentiate between ulnar and radial deficiencies. In making the definitive diagnosis, the upper extremity specialist will perform diagnostic radiographs and other testing.

Mild ulnar deficiencies respond to splinting or casting, and many of these patients can be treated without surgery. Because these patients have a normal thumb and normal radial structures to support the thumb, prehensile function is much better than in children with radial deficiency. Patients with severe deformities or those with associated problems (eg, syndactyly) undergo surgery after 1 year.

Central longitudinal deficiency (cleft hand)

Central deficiency is the result of the unsuccessful formation of the central components of the hand (index, long, and ring fingers). Central deficiency is classified as typical when the long ray is absent, resulting in a V-shaped cleft.^{13,14} It is classified as atypical when the 3 central rays are absent or hypoplastic, resulting in a U-shaped cleft. Typical central longitudinal deficiency may be transmitted as an autosomal dominant trait, in which case it is usually bilateral. However, the majority of typical and atypical cases occur sporadically and are unilateral.

Other anomalies associated with central deficiency include congenital heart disease, cleft foot, cleft lip, cleft palate, imperforate anus, and deafness. Most patients with central deficiency do well functionally and do not require surgery. Indeed, surgery motivated by aesthetic reasons alone may do the child a disservice by improving appearance, but diminishing prehensile function. Reconstructive surgery is recommended for more severe cases.

Trigger thumb

Congenital trigger digit is a flexion deformity of the interphalangeal joint caused by constriction of the A1 pulley, which interferes with the normal gliding of the flexor pollicis longus tendon. As a result, the thumb is held in a flexed position at the interphalangeal joint. When attempts are made to passively straighten the thumb, the metacarpophalangeal joint goes into hyperextension. As pressure is increased, the interphalangeal joint finally yields (sometimes with a palpable clunk at the metacarpophalangeal joint), and the child experiences pain. Although this problem can occur in other fingers, it is much more common in the thumb. Congenital trigger thumbs are not associated with systemic conditions or other anomalies.

Controversy exists regarding treatment for trigger thumbs. In the literature, spontaneous resolution is reported to be as high as 30%.¹⁵ At Children's Memorial Hospital, however, we have rarely seen spontaneous resolution, although our patient population

may be biased toward children who have failed early observation by their pediatricians.

Observation is acceptable when the child presents during the first year of life. Trigger thumbs that are not corrected by age 4, however, may predispose the child to permanent deformity of the thumb.¹⁶ We have found that splinting is not effective for correcting trigger thumbs. Definitive treatment is surgical division of the A1 pulley.

Address emotions when talking to parents

The birth of a child with a major congenital anomaly of the upper extremity is a psychologically traumatic event for parents. Most parents fear that a parental misdeed during pregnancy has caused their child's condition. Often the most important thing that parents can be told is that the child's anomaly is not their fault. Helping parents work through their guilt allows them to begin to accept their child's difference and to develop an optimistic outlook regarding the child's future. Also,

pediatricians can help address the profound concerns that parents may have about their child's physical disability, as well as fears about the social challenges that accompany it. For parents with concerns about the risk of having more children with similar anomalies, a referral to a genetic specialist should be offered.

Conclusion

When first speaking to parents of a child born with a hand anomaly, pediatricians must attempt to alleviate parental guilt and anxieties. Pediatricians can introduce treatment options, including occupational therapy, stretching, splinting, casting, prosthesis, and reconstructive surgery. When indicated, pediatricians should also screen for associated cardiovascular, hematopoietic, gastrointestinal, or genitourinary disorders. Consultation with an upper extremity and hand specialist is recommended for all cases, even when surgery is not needed. As early as possible, short-term and long-term treatment plans should be carefully explained to the family. ■

Table 1	SURGICAL TIMING GUIDELINES
AGE	Congenital hand anomaly
INFANCY	<ul style="list-style-type: none"> • Ulnar polydactyly • Strangulating amniotic bands
6 MONTHS TO 1 YEAR	<ul style="list-style-type: none"> • Radial polydactyly • Central polydactyly • Border syndactyly: thumb-index finger web and ring-small finger web • Complex osseous syndactyly: especially types with transversely positioned bones • Radial longitudinal deficiency
AFTER 1 YEAR	<ul style="list-style-type: none"> • Simple or cutaneous syndactyly: long-ring fingers, long-index fingers • Ulnar longitudinal deficiency: severe cases • Central longitudinal deficiency: severe cases • Trigger thumb
<p><i>Adapted from Herring JA. Tachdjian's Pediatric Orthopaedics, 3rd ed. Philadelphia: WB Saunders Company; 2002:384.</i></p>	

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The congenital hand and upper extremity multi-specialty team collaborates in diagnosis, treatment, and rehabilitation of children with congenital hand and upper extremity problems. The team also counsels parents regarding their child's diagnosis and provides access to support groups and special programs for children. The congenital hand and upper extremity team includes:

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