

Congenital Upper Extremity Differences: Understanding the Etiology and Treatment

When a child is born with an upper extremity or hand difference, the primary-care physician is often the first to discuss the condition with the family. The child's parents are often looking for some type of reassurance in the midst of their emotional distress. A basic understanding of the etiology of, and treatment options for, upper extremity congenital differences can help the primary-care physician offer preliminary information, reassurance and an appropriate referral for the baby.

Embryonic Origin

The majority of upper extremity malformations occur during the embryonic period of gestation — weeks three through eight. By the third week of gestation, the limbs begin to form as buds. Differentiation begins at the shoulder and proceeds down the limb to the hand in linear progression. By the eighth week, each of the distinct components of the arms and hands exist, from the shoulders, elbows and wrists down to the tiny fingernails. It is during this crucial period of formation that most congenital differences occur.



Photo top left: This child was born with an absent thumb. Photo top right: This child had "index pollicization" surgery to rotate the index finger into the thumb position. The child demonstrates the new thumb's function by pinching a coin.

In the subsequent weeks of pregnancy — weeks nine through term, or the fetal period — the upper extremities grow and mature, and malformations are much less likely. If they do occur, they are often the result of either overgrowth of the limb or amniotic bands wrapping around the limb, thereby constricting growth (constriction band syndrome).

Clinical Evaluation

Table 1 lists some common limb differences involving malformations of the upper extremities. Although many of these, such as polydactyly and congenital amputation, are readily apparent to the naked eye, others

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Table 1: Limb Differences

Type	Description
Failures of formation <ul style="list-style-type: none"> • Transverse deficiencies • Longitudinal deficiencies • Central deficiencies 	Congenital amputations (e.g., below-elbow) Deficiency of the radius or ulna, absent digits Cleft hand
Failures of differentiation <ul style="list-style-type: none"> • Proximal radioulnar synostosis • Metacarpal synostosis • Digital syndactyly 	Fusion of the ulna and radius Fusion of the hand bones Webbed fingers
Duplication <ul style="list-style-type: none"> • Preaxial polydactyly • Postaxial polydactyly 	An extra thumb An extra little finger
Overgrowth <ul style="list-style-type: none"> • Macroductyly 	Enlarged finger(s) or limb
Undergrowth <ul style="list-style-type: none"> • Thumb hypoplasia 	Incomplete development of the thumb
Congenital constriction band(s)	Constriction of the limb by amniotic bands

might not be discovered until there is a history of dysfunction. Most congenital differences are not painful, but they may cause the child to use the limb “differently.”

Physical examination can help determine the location and possible cause of the dysfunction. The examiner will want to evaluate both active and passive motion in each part of the upper extremity, including the shoulder, elbow and wrist and the finger and thumb joints. This will also help the child (if old enough) to pinpoint the area of loss of motion. Congenital deformities or subsequent limitations of motion indicate the need for X-rays. Radiographs should include views of both ends of the involved bones, including joints.

Treatment for hand and upper extremity differences is often surgical and is highly individualized to the patient’s needs. Surgical reconstruction is usually carried out between 6 and 12 months of age. Attainment of maximum function, through all stages of growth and development, is the ultimate goal.

Sprengel's Undescended Scapula

Sprengel’s is a good example of a disruption in normal fetal development. The scapula forms and differentiates during the embryonic period and then fails to descend into the normal position along the vertebral column. The result can be a sort of webbing of the neck, with fullness at the base of the neck. A thorough evaluation of the child should include an examination of the surrounding structures, including the clavicles, ribs and shoulder musculature. Ninety-eight percent of children with Sprengel’s also have other anomalies.

In minor cases, cosmetic differences may be relatively unnoticeable, with only minor functional impairment. In more serious cases, cosmetic and functional impairment may be significant. Surgery can include removal of any bony prominence to improve cosmetic appearance, as well as derotation and relocation of the scapula to improve function. Whenever possible, surgery should take place after 3 years of age, when the child has better tolerance for an extensive procedure.

Pseudarthrosis of the Clavicle

With pseudarthrosis of the clavicle, a child is typically born with a prominent bump at the midpoint of the clavicle, usually on the right side of the body. There is little if any associated pain. An X-ray will show a lack of bone continuity in the center third of the bone, but without the telltale callus formation noted with a birth fracture. With an older child, there may be a drooping of the shoulder on the affected side. Spontaneous healing does not occur. Although surgery effectively unites the bone, however, some studies have shown that children can go untreated and remain asymptomatic with no functional defects. Surgery is best reserved for children with a bump that is cosmetically unacceptable or for rare cases in which pain or dysfunction is present.

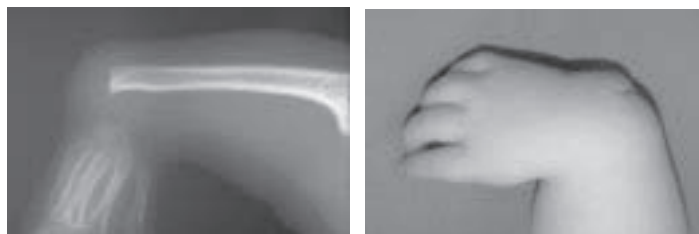
Failures of Formation

Failures of formation fall into two categories — transverse deficiencies and longitudinal deficiencies. Transverse deficiencies include all congenital amputations, with the name of the amputation describing the point at which the remaining limb ends. The most common manifestation is a unilateral below-elbow amputation.

A congenital amputation can occur anywhere along the limb from the shoulder to the fingers. With all limb amputations, the treatment of choice is a prosthesis, starting with a paddle prosthesis at 6 months of age, and advancing in sophistication as the child matures and becomes more skilled at manipulating the device.

With longitudinal deficiencies, parts of the limb between the shoulder and fingers are missing, with the name of the deficiency indicating the missing part(s). The spectrum of anomalies ranges and may include the absence of the ulna associated with absent small and/or ring finger; absent central digits with a “cleft hand”; or absence of the radius associated with an absent or deficient thumb. One sign of a deficient radius is a hand that is radially deviated at the wrist. With total absence of the radius, the wrist is unstable; the thumb is also missing in the vast majority of cases. Radial absence is often associated with complex syndromes that may require multidisciplinary management.

Radial longitudinal deficiencies occur bilaterally in 40 to 60 percent of cases. They occur primarily on the right side when unilateral. Surgical intervention involves stabilization of the wrist joint through “centralization” of the wrist on the single forearm bone. Some reconstruction is possible.



X-ray and picture of congenital absence of the radius (longitudinal deficiency of the radius).



X-ray after “centralization” surgery to centralize the wrist on the single forearm bone.

Failures of Differentiation

As the name suggests, failures of differentiation are the incomplete development and separation of specific parts of the upper extremity. This includes fusion, or synostosis, of the radius and ulna, fusion of the metacarpals, and webbing of the fingers. This last condition, known as syndactyly, is the most common upper extremity difference found in the United States. The webbing can be partial or complete. In complex cases, it involves not only soft tissue but also bone. Syndactyly can occur in isolation or as part of more than 20 different syndromes. Treatment is surgical and usually occurs between 6 and 12 months of age, with separation and skin grafting.



Simple complete syndactyly of the ring and long fingers ("webbed fingers").

Duplication

Polydactyly is not necessarily "duplication" as we generally understand it. Rather, it is believed to result from injury to the original embryonic part, causing it to split in two. It can affect the thumb, little finger or central finger. Small digit polydactyly typically manifests as a rudimentary extra finger attached by a skin fold. Tying it off in the nursery (like an umbilical cord) leads to necrosis and a subsequent falling-off. With all other types of polydactyly, the least functional finger is removed surgically, with reconstruction of the remaining digit to maximize its use. "Splint thumb" (thumb duplication) is the most common congenital hand difference in the Southeast Asian population.



Finger duplication



Thumb duplication



Thumb duplication

Small finger polydactyly is the most common congenital hand difference in the African American population.

Overgrowth

The most common presentation of overgrowth is an enlargement of one or more fingers, called macrodactyly. Often there is an enlargement pattern, with corresponding enlargement of the nerve and the surrounding vessels, fat, soft tissues and bone. It can be static or progressive. Joint stiffness and deviation of the affected digit(s) are common. Amputation of the affected finger improves hand function and appearance in severe cases, although debulking of the size may be more effective in less severe cases.

Undergrowth

Also known as hypoplasia, undergrowth describes a part that was completely formed in the embryonic period, but that did not develop normally during the fetal period. It can affect the entire limb or an isolated part. Thumb hypoplasia is most common and can range from a mild diminution in size, with total function, to complete absence of the digit. Surgical consultation to discuss treatment is indicated in all cases.

Congenital Constriction Bands

Children with constriction bands have limbs that have formed and developed normally, but that are then diminished or completely lost as the result of compression by amniotic bands that have wrapped around the limb. With the exception of complete amputations, surgical intervention may be recommended for all types of constriction bands. Otherwise, surgery is recommended by 18 months of age.

Unlocking the Mystery

In recent years, scientists have isolated specific genes (sonic hedgehog and HOX) and proteins produced by the ridge of cells that direct the development of the upper extremities. These discoveries offer hope for the future. Scientists may eventually be able to control limb growth as an understanding emerges of how normal differentiation and development of the upper extremities occur.

A note about congenital syndromes and neuromuscular disorders:

Although the scope of the present article has been the etiology and treatment of specific shoulder and limb malformations, it should be noted that neuromuscular disorders, such as obstetric brachial plexopathy, cerebral palsy and arthrogryposis, can also result in upper extremity dysfunction. The same is true of congenital syndromes such as cleidocraniodyosotosis and Poland's syndrome. Whenever a child presents with an upper extremity difference, an evaluation for associated anomalies is prudent.

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Author's Profile



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Dr. Van Heest specializes in treating pediatric upper extremity conditions, including congenital disorders (syndactyly; digit duplications) and acquired disorders (fractures; growth disorders). She has a special interest in upper extremity problems related to neuromuscular disorders (arthrogryposis; cerebral palsy; spinal cord injury).

Dr. Van Heest received her medical degree from the University of Minnesota, where she later completed her orthopaedic surgery residency. She trained at Harvard University for a fellowship in hand and upper extremity disorders. She has been practicing at Gillette since 1993. Dr. Van Heest is board-certified by the American Board of Orthopaedic Surgeons and the American Society for Surgery of the Hand. To refer a patient, or for more information about the upper extremity services at Gillette, please call: (651) 229-3944 or toll-free (800) 719-4040.