Congenital Anomalies of the Hand—Principles of Management

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KEYWORDS

• Congenital hand • Syndactyly • Polydactyly • Thumb hypoplasia • Opponensplasty • Pollicization

KEY POINTS

- Although congenital hand anomalies are relatively rare, pediatric orthopedic surgeons and hand surgeons will frequently see them during the course of clinical practice.
- The clinician should be aware of the associated malformations and conditions that may, in some cases, be fatal if not recognized and treated appropriately.
- The goals of surgery are to improve hand function and cosmesis while limiting complications that could impair function long term.
- The surgeon must balance functional, cosmetic, and cultural goals and align those goals with the proper surgical techniques in order to maximize patient and parent satisfaction following surgical intervention.
- This article discusses syndactyly, preaxial polydactyly and postaxial polydactyly, and the hypoplastic thumb.

INTRODUCTION

Congenital anomalies of the upper extremity, although less common than congenital heart disease, are noted in approximately 2 per 1000 live births.[1,2] This incidence varies by country due to higher incidence of certain malformations in patients of certain ethnic backgrounds, such as polydactyly in those of African descent or amniotic bands in Japanese. Although many of these malformations lead to minor functional deficits, they can pose a concern for the parents and lead to psychological distress in children.[3] In addition, the 1-year mortality of patients with hand malformations is 14% to 16% due to associated malformations, often involving the heart, kidneys, or tracheoesophageal complex.[4] Boys are affected more commonly than girls by a 3:2 ratio, and mothers older than 40 years of age are twice as likely to have children with congenital hand differences as those only 10 years younger.[5,6]

Malformations of the hand and forearm were classified by Swanson[7] in 1964 and adopted by the International Federation of Societies for Surgery of the Hand (**Table 1**). Although this classification has its use, it is generally hard to use in clinical instances, because patients may be classified into several categories at once, and it does not guide treatment or prognosis. Oberg and colleagues[8] proposed a modified classification based on a more recent understanding of the embryology of congenital hand malformations. Using this classification, malformations are divided into malformations, deformations, and dysplasias, and then further subdivided (**Table 2**).

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Table 1
Swanson classification system for congenital hand differences

Type I	Failure of formation • Transverse deficiency • Longitudinal deficiency ○ Preaxial: Hypoplasia of thumb and/or radius ○ Central: Typical and atypical cleft hand ○ Postaxial: Hypoplasia of ulna and/or hypothenar hand
Type II	Failure of differentiation • Soft tissue (syndactyly, Poland syndrome, camptodactyly) • Skeletal (synostosis, carpal coalition, complex syndactyly)
Type III	Duplication (polydactyly, mirror hand)
Type IV	Overgrowth (macrodactyly)
Type V	Undergrowth (radial hypoplasia, symbrachydactyly, brachydactyly)
Type VI	Congenital constriction ring syndrome (amniotic band syndrome)
Type VII	Generalized skeletal abnormalities

From Swanson AB. A classification for congenital malformations of the hand. N J Bull Acad Med 1964;10:166.

Embryology

Fetal limb development is initiated with the appearance of a limb bud, consisting of undifferentiated mesenchyme, at the lateral body wall 26 to 28 days after fertilization. Subsequently, the limb develops rapidly in a proximal-to-distal direction over the next 4 weeks. Complex interactions between signaling centers orchestrate this embryonic differentiation.[9–12] The first axis to form in the limb bud is the preaxial-postaxial (radial-ulnar) axis, which is defined by the zone of polarization activity. The next axis to form is the dorsal-volar axis defined by the apical ectodermal ridge (AER). Finally, the AER defines the limb proximal-distal axis and controls interdigital cellular apoptosis. Signaling pathways critical to limb formation include sonic hedgehog (Shh), wingless-type, and fibroblast growth factors. Ectopic Shh expression is a known source of polydactyly because of its role in digit number and identity.[9–17]

INDICATIONS/CONTRAINDICATIONS

For all congenital hand malformations, the primary surgical indication is to improve hand function and cosmesis. Contraindications include patients with surgical reconstruction that interferes with function, such as for a centralization procedure in a patient with poor elbow function or with a primary

Table 2
Oberg classification system for congenital hand differences

I: Malformations	A. Abnormal axis formation/differentiation: entire upper limb i. Proximal-distal axis ii. Radial-ulnar (anterior-posterior) axis iii. Dorsal-ventral axis iv. Unspecified axis B. Abnormal axis formation/differentiation: Hand plate i. Proximal-distal axis ii. Radial-ulnar (anterior-posterior) axis iii. Dorsal-ventral axis
II: Deformations	Constriction ring syndromes, trigger digits
III: Dysplasia	A. Hypertrophy i. Whole limb ii. Partial limb B. Tumorous conditions i. Vascular ii. Neurologic iii. Connective tissue iv. Skeletal
IV: Syndromes	A. Specified B. Others

From Oberg KC, Feenstra JM, Manske PR, et al. Developmental biology and classification of congenital anomalies of the hand and upper extremity. J Hand Surg Am 2010;35(12):2073; with permission.