

Congenital Differences of the Upper Extremity: Classification and Treatment Principles

Moon Sang Chung, MD

Department of Orthopedic Surgery, Seoul National University College of Medicine, Seoul, Korea

For hand surgeons, the treatment of children with congenital differences of the upper extremity is challenging because of the diverse spectrum of conditions encountered, but the task is also rewarding because it provides surgeons with the opportunity to impact a child's growth and development. An ideal classification of congenital differences of the upper extremity would reflect the full spectrum of morphologic abnormalities and encompass etiology, a guide to treatment, and provide prognoses. In this report, I review current classification systems and discuss their contradictions and limitations. In addition, I present a modified classification system and provide treatment principles. As our understanding of the etiology of congenital differences of the upper extremity increases and as experience of treating difficult cases accumulates, even an ideal classification system and optimal treatment strategies will undoubtedly continue to evolve.

Keywords: *Upper extremity, Congenital differences, Classification, Treatment principles*

One to two percent of newborns are born with congenital defects, and 10% of them have congenital differences of the upper extremity.^{1,2)} As congenital differences of the upper extremity are a significant challenge, the reconstructive surgeon has a unique opportunity to positively affect the child's growth and development.³⁾

The primary purpose of a classification system is to increase communication about the specific features of a condition between physicians, and provide the basis for discussion and comparison of information regarding epidemiology and treatment results. Therefore, an ideal classification should reflect the full spectrum of morphologic abnormalities and should be simple and logical for physicians to remember and to use, and would also incorporate etiology, guide treatment, and provide prognosis,^{4,5)} which is still unavailable in congenital differences of the upper extremity.

In this article, I review the current classification systems for congenital differences of the upper extremity, discuss the limitations of obtaining the goals mentioned, and suggest a classification system that was modified after the Swanson/International Federation of Societies for Surgery of the Hand (IFSSH) classification.^{6,7)} I also present principles of treatment of congenital difference of the upper extremity according to the suggested system.

CURRENT CLASSIFICATION SYSTEMS

Various classification systems are divided according to their basis of foundation; descriptive, anatomic or topographical, embryologic, teratologic sequencing, genetic, and different combinations of these.

Descriptive classification is based on findings of deformity, such as radial clubhand, which describes a radially deviated hand looking like a golf club, or camptodactyly, meaning a flexed finger. Although this system is intuitive and commonly used as a diagnosis in clinical practices, it depends on the confusing Greek and Latin terminology and has little scientific value.

A more developed system is the anatomic or topo-

Received July 26, 2010; Accepted October 5, 2010

Correspondence to: Moon Sang Chung, MD

S Seoul Orthopedic Specialty Hospital, Metan-dong, Suwon 443-370, Korea

Tel: +82-31-225-7000, Fax: +82-31-213-2600

E-mail: moonsang@snu.ac.kr

Copyright © 2011 by The Korean Orthopaedic Association

This is an Open Access article distributed under the terms of the Creative Commons Attribution Non-Commercial License (<http://creativecommons.org/licenses/by-nc/3.0>) which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited.

Clinics in Orthopedic Surgery • pISSN 2005-291X eISSN 2005-4408

graphical classification based on anatomic findings or the extent of involvement. Syndactyly is a kind of descriptive classification for a finger deformity, but complete, incomplete, simple or complex syndactyly is an anatomic classification and indicates the extent of involvement. This can guide treatment recommendations and sometimes prognosis. Similarly, most sub-classifications for various diagnostic conditions are based on anatomy, such as the Wassel classification for thumb polydactyly which is based on the level of thumb bifurcation.⁸⁾ Frantz and O’Rahilly^{9,10)} further developed the anatomic or topographic classification and expanded the concept of intercalary deficiencies. They divided limb deficiencies into terminal and intercalary, and each into transverse and paraxial.

The anatomic classification system was further developed into embryologic classification by Swanson et al.¹¹⁾ in 1964, which was based on the concept that anomalies should be grouped according to the part that were affected during development. In the Swanson classification, each limb malformation is classified according to the most predominant anomaly and is placed into one of seven categories (Table 1). This system was accepted by the American Society for Surgery of the Hand (ASSH), the IFSSH, and the International Society for Prosthetics and Orthotics (ISPO), and is now termed as the IFSSH classification.⁶⁾

Recently, the Japanese Society for Surgery of the Hand (JSSH) suggested a modification of the IFSSH system, adding two groups; “Abnormal induction of rays” and “Unclassifiable cases.”¹²⁾ The category of “Abnormal induction of rays” includes syndactyly, the central polydactyly-cleft hand-osseous syndactyly complex, and triphalangeal thumb. This concept was based on recent embryologic studies supporting a common etiology for central polydactyly, syndactyly, and typical cleft hand.¹³⁾ Although this makes sense from an embryologic standpoint, the new category separates central polydactyly from radial and ulnar polydactyly that are classified as “Duplication,” resulting in

Table 1. The Classification System Developed by the International Federation of Societies for Surgery of the Hand^{6,7)}

I. Failure of formation
II. Failure of differentiation
III. Duplication
IV. Overgrowth
V. Undergrowth
VI. Congenital constriction band syndrome
VII. Generalized skeletal abnormalities

the separation of morphologically similar congenital differences and creating a major contradiction to accept intuitively.^{5,14)} Tonkin¹⁴⁾ points out that the difference between abnormal formation and abnormal induction appears to be one of semantics.

Congenital differences can be classified according to their severity of expression, which is called teratologic sequence classification. Although this classification is not comprehensive of all anomalies, this system is often used for classification for specific conditions, as the extent of pathology can determine function and guide treatment. For example, teratologic sequencing of thumb hypoplasia as used in Blauth and Manske’s 5 category system classifies thumb hypoplasia according to the severity of hypoplasia and is helpful for directing the treatment (Table 2).^{15,16)}

Finally, there can be a classification system based on particular genetic or molecular abnormalities. For instance, defects in the HOXD13 have been implicated in several common congenital hand differences such as syndactyly and polydactyly.¹⁷⁾ However, this type of classification may be cumbersome as the genetics are extremely complex and involve multiple steps and interactions between many genes and proteins, in addition to the interaction with environmental factors.⁵⁾ Progress in understanding of these complex genetic and molecular interactions may contribute to a better classification scheme and guide potential genetic treatment options.

LIMITATIONS OF THE IFSSH CLASSIFICATION

At present, the most widely accepted classification is the IFSSH classification. However, it has been criticized for its inherent limitations because it attempts to incorporate etiology into morphologically-based classification,^{4,14)} and is difficult to classify complex cases, especially with the

Table 2. Classification of the Hypoplastic Thumb^{15,16)}

Type 1	Minimally shortened and narrowed structures
Type 2	Mild underdevelopment of all structures; short bones; small diameters; mild thenar muscles hypoplasia; unstable thumb MCP joint; narrow first web space
Type 3A	Stable CMC joint; significant decrease in the thumb size; severe intrinsic and extrinsic muscle hypoplasia; unstable MCP joint; narrow first web space
Type 3B	Type 3A with an unstable CMC joint
Type 4	Pouce flottant; rudimentary thumb
Type 5	Complete aplasia of the thumb

MCP: metacarpophalangeal, CMC: carpometacarpal.

Table 3. The Author's Classification of Congenital Differences of the Upper Extremity

Control failure	Formation failure	Under-formation	Longitudinal: radial/central/ulnar deficiency, madelung deformity
			Transverse: congenital amputation, constriction band syndrome
			Mixed: symbrachydactyly
		Over-formation	Longitudinal: hyperphalangism
			Transverse: polydactyly, mirror hand
			Mixed: polydactyly with hyperphalangism, polydactyly with triphalangeal thumb
	Separation failure	Under-separation	Longitudinal: symphalangism
			Transverse: syndactyly, synostosis (radioulnar/metacarpal)
			Mixed: carpal coalition, camptodactyly, delta bone, arthrogyposis
		Over-separation	Longitudinal: congenital shoulder dislocation, congenital laxity
			Transverse: congenital radial head dislocation
	Growth failure	Under-growth	Longitudinal: brachydactyly
			Transverse: microdactyly
			Mixed: small hand
		Over-growth	Longitudinal: macrodactyly (long)
			Transverse: macrodactyly (wide)
			Mixed: macrodactyly (typical), hemihypertrophy
Structural failure	Generalized		Osteogenesis imperfecta, achondroplasia
	Localized		Melorheostosis, congenital tumorous conditions

complex spectrum of cleft hand and symbrachydactyly.¹⁸⁾

The IFSSH classification system separates “Failure of formation” from “Failure of differentiation” on the basis of timing of the causative insult, but creates separate groups for “Duplication,” “Overgrowth,” and “Undergrowth,” although duplication is a kind of failure of formation and “Overgrowth” and “Undergrowth” are examples of differentiation or development failure (failure of differentiation).¹⁴⁾

In addition, the “cleft hand” is difficult to explain by the IFSSH system. Cleft hand can be divided into typical and atypical. A typical cleft hand (central deficiency) is not associated with forearm anomalies unlike radial or ulnar deficiencies, and is usually bilateral, familiar, and associated with polydactyly, syndactyly and clefting of the feet, while atypical cleft hand or symbrachydactyly is usually unilateral, not hereditary, and not associated with foot anomalies, suggesting different etiologies for similar conditions. In the original Swanson/IFSSH classification, typical cleft hand was classified under “Failure of formation” and symbrachydactyly was classified as “Undergrowth,”

suggesting that Swanson initially considered the etiologies of the two conditions as different. However, in the current modification of the IFSSH classification, symbrachydactyly is brought back to “Failure of formation” category,^{19,20)} highlighting the fact that the IFSSH classification is truly morphological and not etiologically based.⁵⁾ Concerning the typical cleft hand, which is usually associated with central polydactyly and syndactyly, the JSSH modification tried to overcome the problem by introducing another category of “Abnormal induction of rays,” but also has its own contradictions as we noted before.

THE AUTHOR'S CLASSIFICATION SYSTEM

I suggest a new classification for congenital differences of the upper extremity, which is modified after the current IFSSH classification system (Table 3). This classification is also an embryologic classification, and is based on the concept that an organ develops and matures through three distinct stages, which are formation, separation, and growth, and each stage can have two types of control

failure, which are early control failure (resulting in under-formation, under-separation, or under-growth) and late control failure (resulting in over-formation, over-separation, or over-growth). These 6 control failures have three morphological types, which are longitudinal, transverse, and mixed. By having these morphological types for each control failure category, my classification system makes it easy to intuitively classify complex deformities.

Concerning the “Generalized skeletal abnormalities” in the IFSSH system, my system classifies the conditions as “Structural failure” as opposed to “Control failure,” because those anomalies have abnormal tissue from structural or cellular failures while conditions of “Control failure” have normal tissue quality. Therefore, congenital differences of the upper extremity can be first divided into “Control failure” (with normal tissue) and “Structural failure” (with abnormal tissue), and then the “Control failure” can be further classified into “Formation failure,” “Separation failure,” and “Growth failure,” and the “Structural failure” into “Generalized structural failure” and “Localized structural failure.”

The most significant difference of my classification from the IFSSH system is that it divides the concept of “Differentiation” of the IFSSH system into “Separation” and “Growth.” This system has three large categories under “Control failure” and can avoid placing separate categories of “Overgrowth” and “Undergrowth” (embryologically failure of differentiation) at the same level of hierarchy. Furthermore, by placing “over-formation” and “under-formation” under the large category of “Formation failure,” this system removes the redundant category of “Duplication,” which is also embryologically a kind of formation failure. In addition, this classification system removes constriction band syndrome from the 7 categories of the IFSSH system and places it under the category of “Formation failure” (“transverse type under-formation”), because the main pathology of distal tissue agenesis or coalescence is a vascular compromise at some point of intra-uterine organ formation.

The advantage of my classification is that physicians can intuitively classify most of the congenital anomalies because the 3 stages (formation, separation, growth), the 3 types (longitudinal, transverse, and mixed) and the 2 prefixes (under-, over-) are anatomically or morphologically straightforward, although the system is based on embryology. For example, the complex symbrachydactyly, which has mixed longitudinal and transverse deficiency by the anatomical classification, and can be simply classified into “Mixed type under-formation” by my classification system, successfully including both anatomical and embryological

concepts. In my experience, most of the congenital differences could be categorized reliably with some reasonable explanations.

In addition, my classification system can categorize some disease entities, which have not been included in any classification schemes previously, such as congenital laxity of joint, which is not included in the IFSSH system, can be classified as “Transverse type over-separation.” Also, delta bone is not classified into any category in the IFSSH system, but it can be classified as “Longitudinal type under-separation,” because the anomalous orientation of the growth plate causing coronal deviation is thought to be caused by incomplete separation of the epiphysis from the primary ossification center.

Lastly, my classification system can logically and intuitively guide the treatment principle for each category, such as “Under-formation” needs lengthening (or transplantation), and “Over-formation” needs resection (or amputation); “Under-separation” requires separation (or division), and “Over-separation” requires reduction (or arthroplasty or fusion); “Under-growth” needs augmentation and “Over-growth” needs resection (or debulking).

TREATMENT PRINCIPLES ACCORDING TO SUGGESTED CLASSIFICATION SYSTEM

Treatment of upper extremity congenital differences should be initiated as soon as possible, because most anomalies tend to change with time and the human body is considered to maintain skeletal remodeling power until two to three years of age.

Under-formation

Cases of under-formation require ‘lengthening or transplantation,’ because even the most advanced medical technologies cannot create a simple nail plate. Accordingly, even simple cases of under-formation cannot be cured satisfactorily.

For longitudinal under-formations, such as, radial deficiency (radial clubhand), realignment procedures including osteotomy or tendon transfer are necessary to improve function. Hypoplastic or absent thumbs can be treated by pollicization (a type of transplantation from the index finger), and recently bone lengthening and living tissue transfer have been used to replace an absent bone, joint, or muscle.

In cases of transverse under-formation, such as, congenital amputation, digit creation using an autograft or allograft can be attempted. Having created a small digit, lengthening procedures can improve function and

cosmesis. Autogenous bone grafting has the associated limitations of donor site morbidities and of a lack of available bone. Toe transplantation or proximal fibular transplantation can create a digit with growth potential, but the techniques involved are demanding and have a measurable risk of failure. Allografts introduce problems of rejection, and these patients may require lifelong immunosuppressive therapy.

Over-formation

Over-formation can often be treated satisfactorily by simply removing the redundant tissue ('resection or amputation'). Sometimes additional procedures are necessary, such as, tendon realignment, corrective osteotomy to realign residual digit, resection arthrodesis to reduce an extra-bone and joint in cases of hyperphalangism, or combining two small thumbs to create a balanced, normally sized thumb.

Under-separation

Conditions of under-separation, such as symphalangism (longitudinal), syndactyly (transverse), and carpal coalition (mixed) require 'separation,' but these conditions are usually combined with incomplete or deformed bones, joints, and muscles that need additional procedures.

Coalition can be defined as a condition whereby an under-developed joint becomes a syndesmotic or synchondrosic joint rather than a synovial joint. Because the creation of a normal synovial joint is not possible, separation means making a pseudarthrosis to gain mobility. However, such newly created pseudarthroses have problems of reunion, instability, and inadequate motion, since soft tissues around these joints are also inadequate. Sometimes, osteotomy is necessary to obtain a more functional position.

In cases with under-separations of soft tissues, such as, those encountered in camptodactyly, causative tissues (abnormal skin, pulley, fascia, or tendon) that are not critically required for function must be removed.

Over-separation

Conditions of over-separation, such as congenital dislocation or laxity, are treated by 'reduction or stabilization.' The general treatment principle for congenital dislocation requires joint reduction by a closed or open method and the maintenance of reduction until the remodeled joint becomes stable. When a joint has dysplasia or subluxation, attempts are made to make a more stable joint by intra-articular or per-articular osteotomy, as the joint tends to develop early osteoarthritis or become completely dislocated.

When reduction is impossible or too late, arthroplasty or fusion can improve function.

Under-growth

Under-growth is when a portion of an extremity is under-sized but near normal functionally, and requires 'augmentation.' However, it is not possible to increase the size of the affected hand to the normal level, because we are limited to increasing only its length. When the required length is within 1-2 cm, osteotomy and intraoperative distraction with interpositional strut bone grafting and internal fixation provides a straightforward option, and when the required length exceeds 2 cm, external fixation and gradual lengthening can be performed.

Over-growth

Overgrowth needs 'reduction or debulking.' The affected regions include both normal and abnormal tissues, such as, those affected by neurofibromatosis, hemangioma, lymphangioma, or arteriovenous malformation. Sometimes bones and joints are deformed, and muscles and tendons are abnormal. The treatment goal is to decrease length and volume to obtain levels of function and appearance that are similar to those of normal side. However, decreasing size effectively is difficult and postoperative tissue necrosis is common, and thus, in most cases results are less than satisfactory.

Structural Failure

The primary goals of the treatment of generalized structural failure are to extend life expectancy and to prevent progression of the anomaly. In cases with a hormone or enzyme deficiency, hormone or enzyme replacement therapy can achieve the treatment goal. However, in most cases, the causes are unknown and medical treatment is rarely effective. If the anomaly is severe enough to impair walking or daily activities, a child's quality of life can be improved by treatments, such as, bracing, cast immobilization, or surgery, and in cases with a localized structural failure, correction of the localized anomaly alone may achieve its long-term correction. However, the molecular events that underlie such abnormalities have not been elucidated.

CONCLUSIONS

I have reviewed the current classification systems for congenital differences of the upper extremity, discussed the contradictions and limitations of the IFSSH classification, and presented a modified classification system. In addi-

tion, I provided treatment principles according to the suggested classification system. As our understanding of the etiology of congenital differences of the upper extremity increase and as experience of treating difficult cases accumulates, even an ideal classification system and optimal treatment strategies will undoubtedly continue to evolve.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

REFERENCES

- Lamb DW, Wynne-Davies R, Soto L. An estimate of the population frequency of congenital malformations of the upper limb. *J Hand Surg Am.* 1982;7(6):557-62.
- Kozin SH. Upper-extremity congenital anomalies. *J Bone Joint Surg Am.* 2003;85(8):1564-76.
- Bates SJ, Hansen SL, Jones NF. Reconstruction of congenital differences of the hand. *Plast Reconstr Surg.* 2009;124(1 Suppl):128e-143e.
- Manske PR, Oberg KC. Classification and developmental biology of congenital anomalies of the hand and upper extremity. *J Bone Joint Surg Am.* 2009;91 Suppl 4:3-18.
- Sammer DM, Chung KC. Congenital hand differences: embryology and classification. *Hand Clin.* 2009;25(2):151-6.
- Swanson AB. A classification for congenital limb malformations. *J Hand Surg Am.* 1976;1(1):8-22.
- Swanson AB, Swanson GD, Tada K. A classification for congenital limb malformation. *J Hand Surg Am.* 1983;8(5 Pt 2):693-702.
- Wassel HD. The results of surgery for polydactyly of the thumb: a review. *Clin Orthop Relat Res.* 1969;64:175-93.
- Frantz CH, O'Rahilly R. Congenital skeletal limb deficiencies. *J Bone Joint Surg Am.* 1961;43(8):1202-24.
- O'Rahilly R. Morphological patterns in limb deficiencies and duplications. *Am J Anat.* 1951;89(2):135-93.
- Swanson AB, Barsky AJ, Entin MA. Classification of limb malformations on the basis of embryological failures. *Surg Clin North Am.* 1968;48(5):1169-79.
- De Smet L; IFSSH. International Federation for Societies for Surgery of the Hand JSSH. Japanese Society for Surgery of the Hand. Classification for congenital anomalies of the hand: the IFSSH classification and the JSSH modification. *Genet Couns.* 2002;13(3):331-8.
- Ogino T. Teratogenic relationship between polydactyly, syndactyly and cleft hand. *J Hand Surg Br.* 1990;15(2):201-9.
- Tonkin MA. Description of congenital hand anomalies: a personal view. *J Hand Surg Br.* 2006;31(5):489-97.
- Blauth W. The hypoplastic thumb. *Arch Orthop Unfallchir.* 1967;62(3):225-46.
- Manske PR, McCarroll HR Jr, James M. Type III-A hypoplastic thumb. *J Hand Surg Am.* 1995;20(2):246-53.
- Muragaki Y, Mundlos S, Upton J, Olsen BR. Altered growth and branching patterns in synpolydactyly caused by mutations in HOXD13. *Science.* 1996;272(5261):548-51.
- Cheng JC, Chow SK, Leung PC. Classification of 578 cases of congenital upper limb anomalies with the IFSSH system: a 10 years' experience. *J Hand Surg Am.* 1987;12(6):1055-60.
- Miura T, Nakamura R, Horii E. The position of symbrachydactyly in the classification of congenital hand anomalies. *J Hand Surg Br.* 1994;19(3):350-4.
- Manske PR. Symbrachydactyly instead of atypical cleft hand. *Plast Reconstr Surg.* 1993;91(1):196.