SCIENTIFIC ARTICLE

Epidemiology of Congenital Upper Limb Anomalies in a Midwest United States Population: An Assessment Using the Oberg, Manske, and Tonkin Classification

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Purpose To examine the relative presentation frequency of children with upper limb congenital anomalies at 3 Midwestern referral centers using the Oberg, Manske, and Tonkin (OMT) classification and to assess the utility of this new classification system.

Methods 641 individuals with 653 congenital upper extremity anomalies were identified at 3 hospitals in 2 large metropolitan areas during a 1-year interval. Patients were identified prospectively and the specific upper extremity anomaly and any associated syndromes were confirmed using medical records and radiographs. We applied the OMT classification that categorizes anomalies using a dysmorphology outline as malformations, dysplasias, deformations, and syndromes, and assessed its utility and ease of use.

Results There were 480 extremities (74%) with a limb malformation including 184 involving the entire limb. Arthrogryposis was the most common of these (53 extremities). Anomalies affecting only the hand plate accounted for 62% (296) of the malformations. Of these, radial polydactyly (15%) was the most common specific anomaly, followed by symbrachydactyly (13%) and cleft hand (11%). Dysplasias were noted in 86 extremities; 55 of these were multiple hereditary exostoses. There were 87 extremities with deformations and 58 of these were trigger digits. A total of 109 children had a syndrome or association. Constriction ring sequence was most common. The OMT was straightforward to use and most anomalies could be easily assigned. There were a few conditions, such as Madelung deformity and symbrachydactyly, that would benefit from clarification on how to best classify them.

Conclusions Malformations were the most common congenital anomalies in the 653 upper extremities evaluated over a 1-year period at 3 institutions. We were able to classify all individuals using the OMT classification system. *(J Hand Surg Am. 2015;40(1):127–132.*

Type of study/level of evidence Diagnostic III.

Key words Congenital upper limb anomalies, hand anomaly, malformation, prevalence, OMT classification.

Additional material is available online.

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An understanding of the epidemiology of congenital anomalies is necessary for public health, for comparative studies, to monitor changes over time, and to facilitate development of treatment guidelines.

Ideally, the incidence of congenital limb deformities would be established based on all births in the United States. Incidence studies or the examination of the number of new cases per 10,000 live births per year are difficult to perform in the United States, however, because of its large population, the many possible delivery sites, and the lack of a centralized birth registry.

The incidence of congenital upper limb anomalies has been presented in other countries. Recently, studies have reported the incidences of these anomalies in Western Australia[1] and Finland[2] using the classification previously endorsed by the International Federation of Societies for Surgery of the Hand (IFSSH), the modified Swanson system.[3,4] More recently, Ekblom et al[5] reclassified 577 anomalies in Sweden using the Oberg-Manske-Tonkin (OMT) system.[6] Each of these 3 studies used a population-based registry.

In the United States, the epidemiological data that are collected by the U.S. Centers for Disease Control (CDC) include data on birth defects based on 14 population-based state programs. Fourteen conditions are tracked. The only limb deformity birth defect tracked by the CDC is one global condition category of "upper and lower limb reduction deficits", although some states attempt to track birth anomalies with more specificity.[7] From this, the estimated national incidence of reduction deformities of upper limbs is 3.64 per 10,000 live births.[8]

The epidemiology of congenital upper limb anomalies also should include more specificity on the type of limb anomaly present. This can be difficult due to the multiple specialists involved in diagnosis and care and the use of multiple different classifications,[9–13] varying by specialty (eg, pediatricians, geneticists, hand specialists). Furthermore, the predominant system used by hand specialists, the Swanson classification system, has many difficulties, and one noted center recommended a change to a descriptive method of recording anomalies.[14] Recently, in response to the challenges with the Swanson system, the IFSSH adopted the OMT classification for congenital hand anomalies.[15] The utility of OMT classification for congenital hand anomalies has only recently been examined.[5,6]

The purpose of this study was to better understand the relative frequency of upper limb congenital anomalies by assessing patients presenting to 3 large referral centers in 2 Midwestern cities the United States. We classified the anomalies using the OMT system and assessed the ease of use and utility of this new system.

METHODS

We prospectively assessed patients with congenital upper extremity anomalies for the calendar year 2011. One of 3 pediatric upper extremity surgeons examined each patient at one of 3 pediatric hospitals in 2 metropolitan cities Gillette Childrens Hospital in Minneapolis, Minnesota, and St Louis Shriners Hospital and St Louis Childrens Hospital in St Louis, Missouri. Institutional review board approval was obtained. The catchment areas varied; the St Louis Shriners Hospital for Children attracts patients from 9 Midwestern states and up to 600 miles away, the St Louis Childrens Hospital attracts patients from 6 states and up to 250 miles away, and the Gillette Childrens Hospital attracts patients from 6 states and up to 250 miles away. None of the studied patients were seen at more than one of these institutions.

The diagnosis for each patient was obtained and documented at the time of the initial visit and confirmed subsequently with medical record review that included clinical notes and radiographic images. We included patients with congenital conditions and excluded those with injuries, birth brachial plexus palsies, and lower extremity–only diagnoses. Additionally, we excluded patients with osteogenesis imperfecta, single chromosome disorders, mucopolysaccharidosis, idiopathic multicentric osteolysis, and children with syndromes evaluated without specific upper extremity anomalies. We also documented confirmed syndromes and associations.

Each patient was classified according to the modified OMT classification, which uses a dysmorphology framework.[15] The OMT consists of 4 general categories: malformations, deformations, dysplasias, and syndromes (Table 1). *Malformations* are defined as the abnormal formation of a tissue or part, *deformations* as an alteration to a normally formed tissue or part, and *dysplasias* as a change to the number, size, or shape of the cells of a tissue or part. At each institution, 2 upper extremity surgeons with congenital/pediatric expertise applied the classification to each of the patients independently. When a discrepancy was encountered, a discussion led to a resolution. We recorded the problematic diagnoses and syndromes. Patient sex and laterality were recorded. If the upper extremity anomaly was the same for both upper extremities, the patient was considered as a single case. If

TABLE 1. Categories

Malformations		
	Abnormal Axis Formation/Differentiation—Entire Upper Limb	N
I.A.1	Proximal distal axis	31
I.A.2	Radioulnar axis	83
I.A.3	Dorsoventral axis	5
I.A.4	Unspecified axis (including arthrogryposis)	65
	Abnormal Axis Formation/Differentiation—Hand Plate	
I.B.1	Proximal distal axis	76
I.B.2	Radioulnar axis	101
I.B.4	Unspecified	119
Deformations		
II.A	Constriction ring sequence	29
II.B	Trigger digits	58
Dysplasias		
IIIa	Hypertrophy	7
IIIb	Tumorous conditions	79
Total		**653**

TABLE 2. Selected Diagnoses, > 10 Patients Affected

	Anomaly	Extremities Affected
I.A.1.iii	Symbrachydactyly (forearm)	21
I.A.2.i	Radial longitudinal deficiency	43
I.A.2.ii	Ulnar longitudinal deficiency	17
I.A.2.iv	Radioulnar synostosis	13
I.A.4.ii	Arthrogryposis	53
I.B.1.i	Brachydactyly	13
I.B.1.ii	Symbrachydactyly	38
I.B.1.iii	Transverse deficiency	22
I.B.2.i	Radial longitudinal deficiency, hand plate	21
I.B.2.iii	Radial polydactyly	44
I.B.2.vi	Ulnar polydactyly	25
I.B.4.i.a	Cutaneous syndactyly	20
I.B.4.i.b	Camptodactyly	21
I.B.4.ii.a	Clinodactyly	14
I.B.4.iii.c	Cleft hand	32
II.A	Constriction band sequence	29
II.B	Trigger digit	58
III.B.4.1	Multiple herditary exostosis	57

the anomalies were different, then the extremities were classified separately.

RESULTS

Our cohort included 653 upper extremity anomalies affecting 641 patients. One center had 345 different anomalies in 336 patients, and the other 2 centers (located in same city) had a total of 308 different anomalies in 305 patients. There were 330 boys (51%). The left upper extremity was affected in 178 patients, the right was affected in 153, and both limbs were affected in 304. The laterality could not be confirmed in 18. We considered patients with arthrogryposis, multiple hereditary exostoses, and all syndromes to be affected bilaterally. The 304 patients affected bilaterally had the same anomaly on both sides, whereas there were 12 additional children affected differently for each upper limb. We classified these children by each side affected.

There were 480 malformations (74% of total anomalies), 87 deformations (13%), and 86 dysplasias (13%). The category subdivisions (Table 1), the most common diagnoses (Table 2), and the entire list of diagnoses (Appendix A, available on the *Journal's* Web site at www.jhandsurg.org) are provided. The most common diagnoses were trigger digit (58), multiple hereditary exostoses (55), arthrogryposis, whole limb (53), radial polydactyly (44), and radial longitudinal deficiency, whole limb (43).

There were 109 children diagnosed with a syndrome, 17% of the population. We excluded syndromic children without a defined upper extremity anomaly. There were 82 patients labeled as IV.A, with a specifically defined syndrome (Table 3). The most common syndromes were constriction ring sequence and vertebral anomaly, cardiac, tracheaesophogeal fistula, and renal, and limb (VACTERL). There were 27 patients labeled as IV.B (ie, syndrome not otherwise recorded) (Table 3).

We were able to classify all anomalies with the OMT classification system. In general, we found the classification system intuitive and easy to apply with little potential overlap in the categories of limb anomalies. Nevertheless, there were several specific limb anomalies that we found difficult to classify. The correct classification of transverse deficiency and symbrachydactyly for both the whole limb and for just the hand plate is unclear. We differentiated the 2 by using prevailing expert consensus and classifying

TABLE 3. Syndromes and Associations

Syndrome	Number of Patients
IV.A	**Total 82**
Aperts	1
Constriction Ring Sequence	29
Cornelia de Lange	4
Ectrodactyly-Ectodermal Dysplasia-Clefting	3
Fanconi Pancytopenia	4
Goldenhar	1
Holt-Oram	4
Larsen	1
Leri Weill Dyschondrosteosis	1
Nail Patella Syndrome	3
Mobius	1
Oculodentodigital Dysplasia	1
Orofacialdigital	1
Poland Syndrome	6
Rubinstein-Taybi	1
Thrombocytopenia Absent Radius	8
Trisomy 21	1
VACTERL Association	12
IV.B-Others	**Total 27**
Pierre Robin	3
Beals Syndrome	2
Others	22
Total	**109**

an anomaly as symbrachydactyly only if nubbins with fingernails were present. The appropriate classification of Madelung deformity was also unclear as it could be considered an epiphyseal abnormality (III.B.4.iv) or a longitudinal deficiency (I.A.2), as it can involve the entire limb. After a discussion with Michael Tonkin (personal communication, July 2014), we categorized Madelung deformity as a longitudinal deficiency (see Appendix A, I.A.2.vii—a category we added for this purpose). Finally, the appropriate classification of patients with multiple digits with camptodactyly was a dilemma. No matter the number of digits affected, we categorized such patients as camptodactyly (I.B.4.i.b) unless there was a confirmed distal arthrogryposis diagnosis (I.B.4.i.d). Although not specifically listed, we classified clavicular pseudoarthroses as I.A.4.i.c.

In addition, the syndromes were challenging in several ways. First, the diagnoses of constriction ring sequence (II.A), nail patella syndrome (I.A.3.i.b), and Poland syndrome (I.A.1.ii.a) were specific diagnosis options but were also listed as named syndromes (IV.A.7, IV.A.25, and IV.A.32, respectively). The appropriate placement of these 3 diagnoses in particular needs clarification. We included these diagnoses under the syndromes category *and* as specific anomalies. Arthrogryposis, in contrast, is listed only as a specific anomaly. Second, the mucopolysaccharidoses are not included but may bear specific inclusion, perhaps under III.B.3. We excluded such patients from inclusion in this investigation. Third, although IV.B is an appropriate catch-all for "other" syndromes and cannot be all-inclusive, syndromes of particular relevance to the hand, such as Pierre Robin and Beals syndromes, may bear inclusion under IV.A.

DISCUSSION

Understanding the epidemiology of a medical condition or syndrome is paramount to determining its impact on society. A paucity of information exists on the incidence of congenital upper extremity anomalies in the United States. The classic and heavily cited reference is Flatt's *The Care of Congenital Hand Anomalies*,[16] which describes the prevalence of patients seen in the state of Iowa over his career. He reported the relative frequency of anomalies for the 2,758 children seen over 34 years: syndactyly (18%), polydactyly-all types (15%), and camptodactyly (7%) were most frequent. The majority (75%) of anomalies affected the hand. Radial clubhand (currently referred to as radial longitudinal deficiency) was the most common anomaly involving the entire extremity at 5%. Decades later, Flatt's book remains one of the few detailed assessments of the relative frequency of upper extremity anomalies in the United States.

Two previously published studies have reported the incidence of congenital anomalies, both using the modified Swanson classification to characterize their populations. In 2001, Giele et al reported the prevalence of congenital upper limb anomalies in Western Australia using a population-based registry (as reported by any provider who had contact with the child before age 6 y).[1] Over an 11-year period, 509 children were included—this denotes a prevalence of 1 in 506 live births. The most common anomalies were failure of differentiation in 35% of the population and duplications in 33%. A Finnish study[2] also using a population-based registry over 12 years found the incidence of congenital anomalies to be 5.25 in 10,000 live births based on 419 cases. Radial longitudinal deficiency was the most common upper limb deformity in their population (138 cases, 33%), followed by undergrowth conditions (91 cases, 22%).

Both studies help our understanding of congenital anomalies, but both were limited by relatively small numbers of cases over a long time interval with a variety of providers contributing data.

More recently, Ekblom et al assessed the utility of the OMT classification system by re-assessing a previously reported population of congenital upper extremity anomalies identified between 1997 and 2007. The 562 Swedish children identified by that population-based registry and classified previously using the modified Swanson system were re-examined.[5] Malformations were documented in 76% of their cohort, deformations in 22%, and dysplasias in 2%. These results were similar to the current study where malformations constituted 74% and deformations and dysplasia were less common. In contrast to the Swedish population, dysplasias were more common in our population, affecting 13% of patients. This difference may be explained by the fact that Ekblom et al excluded tumorous conditions and only included individuals with hypertrophy within the dysplasia category. Despite its inclusion in the OMT, arthrogryposis was also excluded in Ekblom et al's report as the authors were concerned about diagnostic accuracy. We identified a syndrome in 17% of our cohort compared with 2% of the Swedish population. This difference is likely multifactorial, but our inclusion of older children increased the likelihood of syndrome diagnosis.

Ekblom et al found the OMT highly usable and applicable to their patient population. The authors noted several issues with the system including a lack of a specific subgroup for complex syndactyly and cervical spine and/or shoulder anomalies (other than Sprengel deformity). They also reported difficulties classifying the windblown hand. They requested clarification within the classification system for placement of generalized limb hypoplasia and suggested that brachydactyly and transverse deficiency without proximal involvement be delegated to the section of conditions affecting the hand plate only. At least in part in response to their investigation, the OMT was modified and subsequently accepted by the IFSSH.[15] We believe the modification of the OMT allowed our classification of such anomalies without difficulty.

Applying the most recent, modified OMT classification (as adopted by the IFSSH) to our patient cohort, we were able to classify all patients. The OMT classification required considerable work and is based on developmental biology and known causative mechanisms. We believe the OMT approach to categorization improves our ability to organize individuals with like anomalies, rather than classifying purely on phenotype. Some of the challenges that we experienced during classification are truly limitations in our understanding of limb development rather than limitations of the classification system. With additional research, the correct categorization of these anomalies will become straightforward, and the OMT classification seems appropriately designed to incorporate our increasing knowledge over time. One specific challenge was the appropriate listing of certain diagnoses such as constriction ring sequence. We included these patients as a deformation (II.A) *and* as a syndrome (IV.A.7). Perhaps more specific anomaly classification options (ie, acrosyndactyly, constriction band) would decrease the sense that these anomalies are counted repetitively. Finally, in a population study such as this one, the appropriate handling of involvement of both upper extremities was not clear. We chose to count bilateral symmetrical limb involvement (ie, same category) as one case; but if the limbs were diagnosed differently, the child was considered as 2 cases.

The current study has several limitations. First, this report does not provide incidence or prevalence data for these anomalies. Incidence and prevalence require population data, information that was not available to provide a denominator for the 641 patients studied. Population data are often limited to broad categories without the specificity provided herein based on the OMT classification.

Instead of incidence data, this report provides epidemiology data for 3 large congenital hand referral centers. The patients included are affected by referral patterns. For example, post axial polydactyly and cutaneous syndactyly are certainly 2 of the most common congenital hand anomalies but were less common than expected in our population as many were likely cared for in the newborn nursery or by a community provider. There were a large number of patients with arthrogryposis, an uncommon diagnosis. Such families often seek and travel for care at specialized centers such as the 3 centers included in this study. Despite this limitation, we believe that the large number of patients included in this investigation provide new data on the presentation patterns of patients with congenital upper extremity anomalies for pediatric hand surgeons in the United States.

We found the OMT classification system easy to use and we believe it to be an improvement compared with the Swanson system. As the Congenital Committee of the IFSSH will review the OMT classification system periodically, we hope that the data and experience provided herein will help to resolve some of its difficulties and allow appropriate expansion.

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APPENDIX A. LIST OF DIAGNOSES

I. Malformations

A. Abnormal axis formation/differentiation—entire upper limb
   1. Proximal-distal axis
      i. Brachymelia with brachydactyly
      ii. Symbrachydactyly
         a) Poland syndrome-6
         b) Whole limb excluding Poland syndrome-1
      iii. Transverse deficiency
         a) Amelia-1
         b) Clavicular/scapular
         c) Humeral (above elbow)-1
         d) Forearm (below elbow)-20
         e) Wrist (carpal absent/at level of proximal carpals/at level of distal carpals) (with forearm/arm involvement)-1
         f) Metacarpal (with forearm/arm involvement)
         g) Phalangeal (proximal/middle/distal) (with forearm/arm involvement)
      iv. Intersegmental deficiency
         h) Proximal (humeral-rhizomelic)
         i) Distal (forearm-mesomelic)
         j) Total (phocomelia)-1
      v. Whole limb duplication/triplication
   2. Radial-ulnar (anterior-posterior) axis
      i. Radial longitudinal deficiency-thumb hypoplasia (with proximal limb involvement)-43
      ii. Ulnar longitudinal deficiency-16
      iii. Ulnar dimelia-1
      iv. Radioulnar synostosis-12
      v. Congenital dislocation of the radial head-7
      vi. Humeroradial synostosis-elbow ankylosis
      vii. Madelung deformity-4
   3. Dorsal-ventral axis
      i. Ventral dimelia
         a) Furhmann/Al-Awadi/Raas-Rothchild syndromes
         b) Nail patella syndrome-3
      ii. Absent/hypoplastic extensor/flexor muscles-2
   4. Unspecified axis
      i. Shoulder
         a) Undescended (Sprengel deformity)-9
         b) Abnormal shoulder muscles
         c) Not otherwise specified-3
      ii. Arthrogryposis-53

B. Abnormal axis formation/differentiation-hand plate
   1. Proximal-distal axis
      i. Brachydactyly (no forearm/arm involvement)-13
      ii. Symbrachydactyly (no forearm/arm involvement)-41
      iii. Transverse deficiency (no forearm/arm involvement)
         a) Wrist (carpals absent/at level of proximal carpals/at level of distal carpals)-6
         b) Metacarpal-9
         c) Phalangeal (proximal/middle/distal)-7
   2. Radioulnar (anterioposterior) axis
      i. Radial deficiency (thumb-no forearm/arm involvement)-21
      ii. Ulnar deficiency (no forearm/arm involvement)-7
      iii. Radial polydactyly-44
      iv. Triphalangeal thumb-4
      v. Ulnar dimelia (mirror hand-no forearm/arm involvement)
      vi. Ulnar polydactyly-25
   3. Dorsoventral axis
      i. Dorsal dimelia (palmar nail)
      ii. Ventral (palmar) dimelia (including hypoplastic/aplastic nail)
   4. Unspecified axis
      i. Soft tissue
         a) Syndactyly-20
         b) Camptodactyly-21
         c) Thumb in palm deformity-9
         d) Distal arthrogryposis-5
      ii. Skeletal deficiency
         a) Clinodactyly-14
         b) Kirners deformity
         c) Synostosis/symphalangism (carpal/metacarpal/phalangeal)-5
      iii. Complex
         a) Complex syndactyly-6
         b) Synpolydactyly-6
         c) Cleft hand-32
         d) Apert hand-1
         e) Not otherwise specified

II. Deformations

A. Constriction ring sequence-29
B. Trigger digits-58
C. Not otherwise specified

III. Dysplasias

A. Hypertrophy
   1. Whole limb

i. Hemihypertrophy-4
ii. Aberrant flexor/extensor/intrinsic muscle-1

2. Partial Limb
i. Macrodactyly-2
ii. Aberrant intrinsic muscles of the hand

B. Tumorous conditions
1. Vascular
i. Hemangioma-3
ii. Malformation-5
iii. Others-1

2. Neurological
i. Neurofibromatosis-2
ii. Others

3. Connective Tissue
i. Juvenile aponeurotic fibroma
ii. Infantile digital fibroma-2
iii. Others

4. Skeletal
i. Osteochondromatosis-57
ii. Enchondromatosis-2
iii. Fibrous dysplasia-2
iv. Epiphyseal abnormalities
v. Others-5

IV. Syndromes

A. Specified
B. Others