IFSSH Scientific Committee on Congenital Hand Conditions

Chair: Wee Lam (UK, Singapore)

Committee: Charles Goldfarb (USA)

Wiebke Huelsemann (Germany)

David McCombe (Australia)

Lindley Wall (USA)

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ABSTRACT

The Oberg, Manske and Tonkin (OMT) Classification of Congenital Hand and Upper Limb Anomalies was adopted as the official IFSSH classification in 2014 with recommendations for 3-yearly reviews and updates. This report examined the evidence and feedback from the scientific community to see whether changes to the 2020 OMT should be made. The Committee concluded that no current changes are required, but highlighted a number of areas where further research and discussions are needed. These areas include the conditions of symbrachydactyly vs transverse arrest, the ongoing challenge with classifying syndactyly, and the nomenclature of arthrogrypotic conditions. The OMT Classification continues to enjoy high inter- and intra-rater reliability, thus establishing its central place as a robust classification system in various registries around the world.

INTRODUCTION

The sheer heterogeneity of congenital hand differences (CHD) makes it challenging to classify every condition, but various attempts have been made since the 1960s to design a universal classification that will facilitate global communication. In 1976 the International Federation of Society for Surgery of the Hand (IFSSH) adopted the Swanson classification, which remained the preferred system for the next 50 years (Swanson, 1976).

In 2014 a Scientific Committee on Congenital Hand Conditions commissioned by the IFSSH recommended the replacement of the Swanson classification with a new classification system. Developed by a group of surgeons and scientists in the United States and Australia led by Kerby Oberg, Paul Manske and Michael Tonkin (Tonkin et al., 2013), this system subsequently became known as the OMT classification and was adopted as the preferred IFSSH system (Ezaki et al., 2014). The endorsement of the OMT Classification system represented a move away from the eclectic mixture of dysmorphology and aetiology of the Swanson classification to one that was fundamentally based on knowledge about known errors in the major developmental axes or molecular/genetic pathways.

As with every new system, there were varying opinions about the usability of the OMT system as a universal classification. The 2014 IFSSH Committee recommended regular reviews of its use with 3-yearly updates as our knowledge of CHD advanced (Ezaki et al., 2014). Following these recommendations, the OMT Classification underwent various changes in 2015 (Tonkin and Oberg, 2015), 2017 (Tonkin, 2017) and 2020 (Goldfarb et al., 2020). Over the years, various authors reported its improved inter- and intra-rater reliability as compared to the Swanson system (Bae et al., 2018; Ekblom et al., 2010; Goldfarb et al., 2015). Others disagreed and considered it too non-specific and unlikely to aid collaboration with other disciplines, especially those concerned with prevalence and international data collection (Lowry et al., 2017). More recently, there have been reports about difficulties in classifying a small number of conditions as well as disagreements over the use of terminologies or groupings when using the latest 2020 OMT Classification version (Sait et al., 2022; Sletten et al., 2022; Wall et al., 2022).

A further IFSSH Scientific Committee on Congenital Hand Conditions was commissioned in 2022, tasked to review the 2020 OMT Classification for updates and to explore its adoption as an international classification system.

AREAS OF CONSIDERATIONS FOR UPDATES TO THE 2020 OMT

The OMT Classification is designed to accommodate changes based on improved understanding of limb development. The 2020 updated version and its various rationales for change can be found in the article by Goldfarb et al (2020) and the report on the IFSSH website https://www.ifssh.info/scientific_committee_reports.php. One example of change in the 2020 update is the cleft hand, which has been moved from an 'unspecified axis' to its rightful place under the 'proximal-distal' axis, following defining work by various groups (Duijf et al., 2003; Guero and Holder-Espinasse, 2019; Kantaputra and Carlson, 2019).

Between 2020 and 2023, the current Committee considered various feedback from the literature, discussions during international meetings, comments from various groups which had raised concerns as well as drawing from their own experience in clinical practice and national databases. We examined updates from our scientific colleagues to consider any modification of the 2020 OMT Classification (Appendix 1) based on new knowledge in developmental biology and genetics. Every update to the OMT Classification must be accompanied by

scientific evidence and international consensus. The OMT Classification is embedded in various national registries and any change would result in disruptions to these databases as these require both software updates and linkage from current classification to the previous one. This article outlines a number of areas where changes to the OMT Classification should be considered in the future following more robust research evidence and discussion.

1. Simplifying the current alphanumerical system

The OMT Classification uses a combination of Roman (e.g., I, II, III) and Arabic (e.g., A, B, C; a, b, c; 1, 2, 3) alphanumericals to classify conditions. The Roman system was adopted from the Swanson system (Groups I-VIII). For example, radial longitudinal deficiency of the entire upper limb is classed as 'I-A-2-I'. This combination of alphanumericals may present certain challenges when coding for registry databases and also when searching for a diagnosis. For example, when searching for "IB4I" it may automatically also bring up "IB4II" and "IB4III" In the future, a simpler option will be to convert the coding to a single alphanumerical system, probably the Arabic system. This may make it easier to add new categories/make changes at the end of a classification, i.e., "4" instead of 'IV". Under the new system, radial longitudinal deficiency of the entire upper limb would thus be classed as '1-A-2-1'. Further discussion is needed among the major registries with the involvement of data analysts, as this can represent a significant change to existing databases.

2. Symbrachydactyly vs transverse deficiency of the entire upper limb

Various reports examining the inter-rater reliability of the OMT Classification have shown a high rate of disagreement between the phenotypes of symbrachydactyly and transverse deficiency (Bae et al, 2018; Ekblom et al, 2014; Sletten et al, 2022). The term 'symbrachydactyly' was first suggested by Pol in 1921 (Holmes and Nasri, 2022) to describe a deformity of the hands and feet with shortened fingers or toes, hypoplasia of the middle or distal phalanges and often accompanying syndactyly.

Buck-Gramcko (1999) developed the teratologic line of symbrachydactyly with two lines: the typical regression in symbrachydactyly and transverse arrest. In the original Swanson classification (Swanson, 1976), symbrachydactyly and transverse deficiency were placed under two separate categories: undergrowth and failure of formation, respectively. In the 2020 OMT update (Goldfarb et al, 2020), the term '*with ectodermal element'* was added to

symbrachydactyly (I-A-1-IIa, b and I-B-1-II) to differentiate these from transverse deficiency *('without ectodermal elements'*; I-A-1-IIIa, b and I-B-1-III).

Despite this, various authors considered the two conditions as part of a continuum. Nubbins are usually associated with symbrachydactyly; however, in a recent study by Hu et al., (2023), 52% of extremities categorized as transverse deficiency in the Congenital Upper Limb Differences Registry (CoULD) registry had nubbins. Another interesting finding from the study included a lower incidence of nubbins in more proximal limb differences, e.g., amelia and humeral level transverse deficiencies as compared to distal deficiencies. There was also a 20-times higher chance of surgeons diagnosing symbrachydactyly rather than transverse deficiency if a CHD is distal as compared to proximal. The study indicates that the level of deficiency is more determinant to a given diagnosis of transverse deficiency vs symbrachydactyly than the absence or presence of nubbins. A similar observation was reported by Sletten et al. (2022) where they found that many patients with transverse deficiency proximal to the wrist were classified as such despite having ectodermal elements. A change in terminology to the symbrachydactyly and transverse deficiency categories was discussed at the recent World Congenital Hand Symposium in Minnesota and is currently being considered for the grouping of all proximal deficiencies, including symbrachydactyly, as 'Transverse deficiency - entire upper limb division' but to specify whether these were 'with or without ectodermal elements.' For the *Hand plate division,* the separate classifications of transverse deficiency vs symbrachydactyly should remain as this division remains meaningful in the context of microsurgical toe-transfer reconstruction (Sletten et al, 2022).

The term 'ectodermal elements' remains unclear but is synonymous with 'nubbins' to most surgeons, consisting of pedunculated soft tissue attachments, usually with bone, cartilage, and skin +/- nails but without bony articulations. These represent hypoplastic digits that have partially formed after an insult to the apical ectodermal ridge (AER) and underlying mesoderm. From studies of Poland syndrome, in which there is a high incidence of symbrachydactyly, the popular theory remains that a symbrachydactyly results from a partial or complete blockage of blood flow in the subclavian or vertebral arteries or their branches (Bavinck and Weaver, 1986), whereas a complete insult to the AER results in a transverse deficiency with no nubbins (Farr et al., 2018; Hu et al, 2023). Others have suggested an association of symbrachydactyly with the pathogenesis of brachydactyly, and therefore the development of symbrachydactyly may be closely related to mutations in the bone morphogenetic protein (BMP) pathway (Holmes

and Nasri, 2022). In the future, vascular development at the embryonic level or the BMP pathway may be points for research to further define the relationship between transverse deficiency and symbrachydactyly, and thus guide possible OMT Classification changes.

3. Arthrogryposis nomenclature and classification

Currently, the classification of arthrogrypotic conditions is under 'Dysplasia' and 'Congenital contracture' (III –C–I–a, b and c). Under the subheading of 'Arthrogryposis Multiplex Congenita', the conditions are divided into a) Amyoplasia, b) Distal arthrogryposis and c) Other.

The term 'arthrogryposis' covers a very heterogenous group of over 400 known conditions. Classification of all these conditions remain challenging. Lowry et al. (2017) commented that arthrogryposis is a general term that could be a malformation, deformation or dysplasia and proposed a classification system that took into account the different phenotypes as well as aetiology, including possible syndromic associations. Hall and colleagues (2019) suggest approaching it in four ways: clinically, genetically, aetiologically and functionally. They suggested a multi-layered approach to classification as one system is unlikely to address all the needs of various specialties. The OMT Classification is designed to improve communication between surgeons and other disciplines, but it is primarily surgeons who will use the classification. Consequently, most would be familiar with the different presentations of amyoplasia (more than one major joint involved usually including the elbow and wrist) or distal arthrogryposis (can involve more than one joint but typically affecting the hand, including camptodactyly and thumb-in-palm deformity) (Alzahrani and Farr, 2022). Most would see these as distinct entities and hence the conditions are classified as such. At present, we do not recommend any further reorganization or movement of this group of conditions within the OMT Classification. The only changes worth considering are related to terminology; the terms Arthrogryposis Multiplex Congenita and amyoplasia are used interchangeably and therefore in the future, a more generic main heading can be used, i.e., 'Arthrogryposis' rather than 'Arthrogryposis Multiplex Congenita', as the latter is considered by several surgeons to be the same as amyoplasia.

4. The ongoing challenge with classifying syndactyly

Various authors have expressed views on the aetiology of syndactyly as resulting from an error in the proximal-distal axis, (e.g., Al Qattan, 2023). The process of patterning the autopod (hand plate) is the most complicated portion of limb development. Subsequently, classifying four digits, a thumb, and variations in the interdigital web spaces according to the development axis is far more complicated than more proximal anomalies.

Digital patterning starts with anlagen formation influenced by the sonic hedgehog (Shh)-Gli3 counter gradients and the Hoxd9-13 gradients (Pérez-Gómez et al., 2018). As the digits extend and the corresponding interdigital spaces progress, retinoic acid, bone morphogenetic protein (BMPs) and Notch signalling target interdigital regression, while concurrently Wnt and fibroblast growth factor (FGH) signalling pathways counter interdigital cell death. This delicate balance was shown in the elegant experiments by Bandyopadhyay et al., (2006) where BMP2-deificent mice exhibited a soft tissue type syndactyly.

Cutaneous syndactyly has multiple patterns that do not appear to follow a specific axis; rather the patterns appear to demonstrate gene-specific embryological "watershed regions" of contribution that are most deficient with mutation and highlight syndactyly between the digits of these watershed regions. The molecular array of these genes and how they contribute to typical digit and interdigital space formation has not been well characterized. Nevertheless, some are AER-related such as WNT, Notch or FGF while others appear to be unlinked to an axis, at least at present (Cassim et al., 2022).

To complicate matters, the genetics of osseous versus cutaneous syndactyly differ. Osseous syndactyly can start at the level of metacarpal development and disruption of the pattern can yield metacarpal fusion (more associated with the radioulnar axis). A well-known feature of HoxD13 mutations is osseous syndactyly, usually of digits 3-4 with polydactyly (synpolydactyly). In Apert syndrome, it seems the ongoing activation/mutation of fibroblast growth factor receptor (FGFR2) (Andersen et al. 1998) is associated with terminal phalangeal fusion, i.e., osseous acrosyndactyly of the terminal phalanges, but which also decreases the hand plate size during metacarpal anlagen formation causing frequent fusion of metacarpal 4 and 5.

In summary, knowledge of the morphogenetic relationships underlying syndactyly is accumulating, but a clear relationship has not yet emerged. The most consistent contributing axis are factors from the AER overlying the interdigital space. But the proximal-distal axis

does not appear to correlate with the variable patterns of syndactylies that occur along the radioulnar axis. For this reason, it is recommended that syndactyly remains under the 'Unspecified axis' until further details emerge.

Symbrachydactyly vs complex syndactyly vs syndromic syndactyly vs synpolydactyly

Both Wall et al. (2022) and Sletten et al. (2022) found a lack of consensus with the classification of a specific non-syndromic complex syndactyly phenotype: symbrachydactyly with a polydactylous element. Opinions differ as to whether these should be placed under complex syndactyly or synpolydactyly or symbrachydactyly. Under the 2020 OMT Classification, symbrachydactyly (hand plate) is classified under the proximal-distal axis: IB-1-II, whereas syndactyly with its variants is classified under the Unspecified axis, including Osseous subheading: complex syndactyly (I-B-4-II-a). The complex syndactyly subheading (I-B-4-III) is further subdivided into a) Syndromic syndactyly (e.g., Apert hand), b) synpolydactyly or c) Not otherwise specified.

As mentioned, the classification of syndactyly is far from straightforward. The findings of symbrachydactyly with a polydactylous element represents a phenotype that is not commonly seen and which at present cannot be placed in a distinct category. Symbrachydactyly does not usually present with more digits and synpolydactyly, the result of HoxD13 mutations, does not usually present with missing phalanges. These 'brachy-synpolydactyly' cases may be worthy of a distinct category under the 'Unspecified axis and Complex subheading'. At present, they should be classified under the 'Not otherwise specified' category until more information is obtained.

REVISTING THE PURPOSE OF THE OMT CLASSIFICATION

The OMT Classification was designed to be a universal system that addresses some of the shortfalls of the Swanson system and increase inter-rater reliability in the classification of CHDs. An international survey was conducted among congenital hand surgeons to assess the current status of OMT usage in clinical and research settings (Goldfarb et al., 2023). From the survey, 61% of international experienced congenital hand surgeons use the OMT Classification regularly in their practice. The OMT Classification appears to be favoured by those who

regularly use registries, whereas those who do not find the classification of CHDs according to developmental axes largely to be an exercise without relevance to patient care.

It is a long-held assumption that an effective classification should be both easy to apply and be an aid in treatment decision (Tonkin et al., 2013). However, the OMT Classification, like the Swanson classification before it, has always served a different purpose. Rather than provide a direct guide for the management of different conditions, these broad classifications give a more general overview of the different types of CHDs and the categories (seven for the Swanson and four for the OMT) cater to the vast heterogeneity of anomalies and facilitate comparisons between registries. The OMT system should not be compared to other condition-specific classification systems designed more specifically to guide treatment, e.g., the Blauth classification for thumb hypoplasia. When used in registries, the OMT system helps categorize patients with a high inter-rater agreement to allow investigation of specific cohorts of patients. Moving forward, this purpose of the OMT Classification will perhaps need to be explained to future congenital hand surgeons to increase its usability and adoption.

IMPROVING THE UNIFORMITY OF CLASSIFICATION/CODING WHEN USING THE OMT SYSTEM

At present, the central place of the OMT system as a universal classification tool within major registries appears to be cemented; it has the highest inter-rater reliability of any classification at present. On the whole, regular users of the OMT system should find it straightforward to classify the majority of conditions. Criticism that its use requires a detailed knowledge of embryology is unfounded, as the user simply needs to search for a condition that has already been classified, e.g., radial polydactyly belongs under 'I-B-2-III, i.e., Malformation of the hand plate in the radial-ulnar axis'. Furthermore , the 'OMT App' (https://www.ifssh.info/OMT-Classification-App.php) which can be freely downloaded, simplifies this process by using the 'search' function to match the clinical diagnosis or phenotype to its place within the OMT ontology. Regular use of the OMT system allows the user to gain a deeper understanding of embryology at the same time.

There remain difficulties with classifying a small number of conditions. Sletten et al. (2022) found an almost perfect inter- and intra-reliability for conditions with easily distinguishable characteristics which they termed Group 1 but only a moderate reliability for Group 2 (all other conditions). They chose not to use a consensus group or give instruction to the raters beforehand in order to test the reliability of the OMT system in settings as close to everyday practice as possible. Wall et al. (2022) conducted a Delphi-like consensus exercise to discuss cases that even experienced surgeons find difficult to classify. Most of these diagnoses were resolved following further discussions.

Other reported difficulties, unrelated to diagnostic difficulties but rather coding difficulties, include confusions over how to classify a hand with two CHDs, e.g., cleft hand with associated syndactyly, or a child with bilateral CHDs. To improve the uniformity of classification across registries, this committee has produced a video (led by Charles Goldfarb) to provide step-by-step recommended instructions on the use of the OMT Classification. The video is available here: https://ifssh.info/OMT-Classification-App.php

The following points are highlighted in the video:

1. Overview of the OMT system and how to classify a condition based on the phenotype.
2. When classifying an upper limb with two CHDs, use the main phenotype, e.g., cleft hand as the primary diagnosis and other anomalies to be considered as secondary changes, e.g., syndactyly or camptodactyly.
3. When classifying a child with bilateral CHDs, list both*.
4. When classifying a child with a syndrome, list both the phenotype and also the syndrome*.

*These may be more relevant for registries where listing all phenotypes make it easier to search for a condition.

Lessons continue to be learnt about how to improve the uniformity of classification when using the OMT system. One suggestion for increasing inter-rater reliability is to merge certain

conditions and remove the division of Malformation IA and IB. Using the example of radial longitudinal dysplasia (RLD), Sletten et al. (2022) argued that the separation of this condition into IA (entire upper limb) and IB (hand plate only) is somewhat artificial as all patients with RLD have some degree of thumb hypoplasia, and most patients with thumb hypoplasia have at least some degree of carpal abnormalities (considered a more proximal abnormality). They expressed concerns that splitting conditions such as RLD can lead to more coding variations among surgeons. While simplification and merging of groups together may potentially increase consensus, there is a need for a balanced approach; the OMT system is designed according to pathoembryology in which one of the major considerations is the timing of insult. Merging conditions that have been formed during early and late patterning would reduce the fundamental strength of the OMT system to combine knowledge with diagnosis. It runs the risk of taking a step backwards and resembling the confusing and oversimplified categories of the Swanson system.

THE IMPORTANCE OF REGISTRIES

Patient registries have the potential to overcome research limitations inherent to rare conditions like CHDs. These use multi-centre observational study methods for the collection of uniform data and to evaluate the presentation of conditions, specified outcomes and associations for a population defined by a hand phenotype or syndrome. The data generated can be helpful especially where retrospective studies are lacking, or when randomized controlled trials are difficult or ethically impossible to conduct in children.

At present there are a number of major registries for CHDs such as the CoULD based in the United States, the Australian Hand Difference Register (ADHR), the Congenital Upper Limb Anomalies North (CULA North) project in Scandinavia and Germany, and the British Society for Surgery of the Hand (BSSH) registry. Both the CULA North and the BSSH registries are based on the International Consortium of Health Outcomes Measurement (ICHOM). Some of these registries have compiled data that have resulted in a number of important publications; for example, the CoULD comprehensively collects functional outcomes and health-related quality of life data using the Paediatric Outcomes Data Collection Instrument (PODCI) and the Patient Reported Outcomes Measurement Information System (PROMIS) which has allowed valuable insights into children's overall well-being despite their CHDs (Bae et al., 2018b) or

insights into rare clinical associations involving radial longitudinal deficiency (Forman et al., 2020).

The value of registry studies is undisputed, and every centre that treats these children should aspire to set up a registry to collect data. The amount of data and format of collection depends on the available resources but at the very minimum, these should include epidemiological data, hand conditions classified according to the OMT system, and the use of at least one outcome measure, e.g., the PROMIS.

CONCLUSION

In this report, the Committee has initially aimed to provide an update on the OMT Classification but as the project develops, it becomes apparent that any future discussion of the OMT system should be placed in the context of CHD registries. At present, the OMT Classification remains the best universal classification system with an excellent inter-rater reliability, allowing effective communication across registries. Resources such as the OMT App, or 'How to use the OMT' video should further facilitate the classification process and reduce discrepancies.

Any update must be substantiated by major progress in knowledge and an international consensus. This update has therefore recommended *areas of consideration for change* rather make definitive changes, as the evidence for these is lacking. The OMT Classification remains unchanged from the 2020 version (Appendix 1). These areas are recommended future points of collaborative research between surgeons and scientists with updates, if appropriate, at every major conference such as the World Congenital Hand Symposia, IFSSH congresses or the Limb Development Conferences (Lam, 2019). The committee therefore recommends that the 3-yearly updates should be revised to at least 5-yearly to coincide with these conferences and to await the results of ongoing research in limb development.

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Appendix 1:

The Oberg, Manske and Tonkin (OMT) Classification of Congenital Hand and Upper Limb Anomalies, last updated October 2020.

OMT CLASSIFICATION OF CONGENITAL HAND AND UPPER LIMB ANOMALIES
Last update 5th October 2020

I. MALFORMATIONS
A. ENTIRE UPPER LIMB - ABNORMAL AXIS FORMATION (EARLY LIMB PATTERNING)
1. Proximal-distal axis
i. Brachymelia
ii. Symbrachydactyly Spectrum (with ectodermal elements)
a) Poland syndrome
b) Whole limb excluding Poland syndrome (various levels – humeral to phalangeal)
iii. Transverse deficiency (without ectodermal elements)
a) Amelia
b) Segmental (various levels – humeral to phalangeal)
iv. Intersegmental deficiency (Phocomelia)
a) Proximal (humeral – rhizomelic)
b) Distal (forearm – mesomelic)
c) Proximal + Distal (hand to thorax)
v. Whole limb duplication/triplication

2. Radial-ulnar (anterior-posterior) axis
i. Radial longitudinal deficiency
ii. Ulnar longitudinal deficiency
iii. Ulnar dimelia
iv. Radiohumeral synostosis
v. Radioulnar synostosis
vi. Congenital dislocation of the radial head
vii. Forearm hemi-physeal dysplasia, radial (Madelung
Deformity), or ulnar

3. Dorsal-ventral axis
i. Ventral dimelia
ii. Dorsal dimelia

4. Unspecified axis
i. Shoulder
a) Undescended (Sprengel)
b) Abnormal shoulder muscles
ii. Upper to Lower limb transformation

B. HAND PLATE - ABNORMAL AXIS DIFFERENTIATION (LATE LIMB PATTERNING/DIFFERENTIATION)

1. Proximal-distal axis
i. Brachydactyly
ii. Symbrachydactyly (with ectodermal elements)
iii. Transverse deficiency (without ectodermal elements)
iv. Cleft hand (Split Hand Foot Malformation)

2. Radial-ulnar (anterior-posterior) axis
i. Radial longitudinal deficiency, hypoplastic thumb
ii. Ulnar longitudinal deficiency, hypoplastic ulnar ray
iii. Radial polydactyly
iv. Triphalangeal thumb
a) Five finger hand
v. Ulnar dimelia (mirror hand)
vi. Ulnar polydactyly

3. Dorsal-ventral axis
i. Dorsal dimelia (palmar nail)
ii. Ventral dimelia (hypoplastic/ aplastic nail)

4. Unspecified axis
i. Soft tissue
a) Cutaneous (simple) syndactyly
ii. Skeletal
a) Osseous (complex) syndactyly
b) Clinodactyly
c) Kirner deformity
d) Synostosis/symphalangism
iii. Complex
a) Syndromic syndactyly (e.g., Apert hand)
b) Synpolydactyly
c) Not otherwise specified
II. DEFORMATIONS
A. Constriction ring sequence
B. Not otherwise specified

III. DYSPLASIAS

A. Variant Growth
1. Diffuse (Whole limb)
i. Hemihypertrophy
ii. Aberrant flexor/extensor/intrinsic muscle
2. Isolated
i. Macrodactyly
ii. Aberrant intrinsic muscles of hand

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

2. Neurological
i. Neurofibromatosis
ii. Others

3. Connective tissue
i. Juvenile aponeurotic fibroma
ii. Infantile digital fibroma
iii. Others

4. Skeletal
i. Osteochondromatosis
ii. Enchondromatosis
iii. Fibrous dysplasia
iv. Epiphyseal abnormalities

v. Pseudoarthrosis
vi. Other

C. Congenital Contracture
i. Arthrogryposis Multiplex Congenita
a) Amyoplasia
b) Distal arthrogryposis
c) Other
ii. Isolated
a) Camptodactyly
b) Thumb in palm deformity
c) Other

IV. SYNDROMES*

A. Specified
1. Acrofacial Dysostosis 1 (Nager type) (MIM #154400)
2. Apert (MIM #101200)
3. Al-Awadi/Raas-Rothschild/Schinzel phocomelia (MIM #276820)
4. Baller-Gerold (MIM #218600)
5. Bardet-Biedl (21 types)
Type 1) (MIM #209900)
Type 2) (MIM #615981)
Type 3) (MIM #600151)
Type 4) (MIM #615982)
Type 5) (MIM #615983)
Type 6) (MIM #605231)
Type 7) (MIM #615984)
Type 8) (MIM #615985)
Type 9) (MIM #615986)
Type 10) (MIM #615987)
Type 11) (MIM #615988)
Type 12) (MIM #615989)
Type 13) (MIM #615990)
Type 14) (MIM #615991)
Type 15) (MIM #615992)
Type 16) (MIM #615993)
Type 17) (MIM #615994)
Type 18) (MIM #615995)
Type 19) (MIM #615996)
Type 20) (MIM #617119)
Type 21) (MIM #617406)
6. Carpenter (MIM #201000)
7. Catel-Manzke (MIM #616145)
8. Cornelia de Lange (5 types)
Type 1) (MIM #122470)

Type 2) (MIM #300590)
Type 3) (MIM #610759)
Type 4) (MIM #614701)
Type 5) (MIM #300882)
9. Beals (MIM#121050)
10. CLOVE (MIM #612918)
11. Crouzon (MIM #123500)
12. Down (MIM #190685)
13. Ectrodactyly-Ectodermal Dysplasia-Clefting (MIM #129900)
14. Fanconi Pancytopenia (MIM #227650)
15. Freeman Sheldon (#MIM 193700)
16. Fuhrmann (MIM #228930)
17. Goltz (Focal Dermal Hypoplasia - FDH) (MIM #305600)
18. Gorlin (Basal Cell Nevus Syndrome – BCNS) (MIM #109400)
19. Greig Cephalopolysyndactyly (MIM #175700)
20. Hajdu-Cheney (MIM #102500)
21. Hemifacial Microsomia (Goldenhar syndrome) (MIM #164210)
22. Holt-Oram (MIM #142900)
23. Lacrimoauriculodentodigital (Levy-Hollister) (MIM #149730)
24. Larsen (MIM #150250)
25. Laurin-Sandrow (MIM #135750)
26. Leri-Weill Dyschondrosteosis (MIM #127300)
27. Liebenberg Syndrome (MIM #186550)
28. Moebius sequence (MIM #157900)
29. Multiple Synostoses (4 types)
Type 1) (MIM #186500)
Type 2) (MIM #610017)
Type 3) (MIM #612961)
Type 4) (MIM #617898)
30. Nail-Patella (MIM #161200)
31. Noonan (2 types
Type 1) (MIM #163950)
Type 2) (MIM #605275)
32. Oculodentodigital dysplasia AD (MIM #164200); AR (MIM #257850)
33. Orofaciodigital (18 types)
Type 1) (MIM #311200)
Type 2) (MIM #252100)
Type 3) (MIM #258850)
Type 4) (MIM #258860)
Type 5) (MIM #174300)
Type 6) (MIM #277170)
Type 7) (MIM #608518)
Type 8) (MIM #300484)
Type 9) (MIM #258865)
Type 10) (MIM #165590)
Type 11) (MIM #612913)
Type 12) (No MIM yet (Moran-Barroso et al., 1998))
Type 13) (No MIM yet (Degner et al., 1999))
Type 14) (MIM #615948
Type 15) (MIM #617127)
Type 16) (MIM #617563)
Type 17) (MIM #617926)
Type 18) (MIM #617927)
34. Otopalatodigital Spectrum (FILAMIN A – FLNA)
Type 1) Otopalatodigital Type 1 (Gain of function) (MIM #311300)
Type 2) Otopalatodigital Type 2 (Disruption) (MIM #304120)
Type 3) Frontometaphyseal dysplasia (MIM

#305620)
Type 4) Melnick-Needless (MIM #309350)
35. Pallister-Hall (MIM #146510)
36. Pfeiffer (MIM #101600)
37. Pierre Robin (4 subtypes)
Type 1) Pierre Robin (MIM #261800)
Type 2) Pierre Robin with campomelic dysplasia (MIM #602196)
Type 3) Pierre Robin with oligodactyly (MIM #172880)
Type 4) Pierre Robin with facial and digital anomalies (MIM #311895)
38. Poland (MIM #173800)
39. Proteus (MIM #176920)
40. Roberts (MIM #268300)
41. SC Phocomelia (MIM #26900)
42. Rothmund-Thomson (MIM #268400)
43. Rubinstein-Taybi (2 types)
Type 1) (MIM #180849)
Type 2) (MIM #613684)
44. Saethre-Chotzen (MIM #101400)
45. Split-hand-foot malformation (7 types)
Type 1) (MIM #183600)
Type 2) (MIM #313350)
Type 3) (MIM #246560)
Type 4) (MIM #605289)
Type 5) (MIM #606708)
Type 6) (MIM #225300)
Type 7) (MIM #220600)
46. Thrombocytopenia Absent Radius (MIM #274000)
47. Townes-Brock (2 types)
Type 1) (MIM #107480)
Type 2) (MIM #617466)
48. Trichorhinophalangeal (3 types)
Type 1) (MIM #190350)
Type 2) (MIM #150230)
Type 3) (MIM #190351)
49. Ulnar-Mammary (MIM #181450)
50. VACTERLS association (3 types)
Type 1) VACTERL (MIM #192350)
Type 2) VACTERL X-Linked (MIM #314390)
Type 3) VACTERLH (with hydrocephalus) (MIM #276950)

B. Others

*The specified syndromes are those considered mostrelevant; however, many other syndromes have a limb component categorized under “B. Others”.