Update to the 2014 and 2015 Reports of the Scientific Committee on Congenital Conditions

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2014 report: https://ifssh.info/pdf/2014_Congenital_conditions.pdf

2015 report: https://ifssh.info/pdf/2015_OMT_Classifiction_Congenital_Report.pdf

Classification of Congenital Hand and Upper Limb Anomalies

The Swanson Classification was proposed in the 1960s and subsequently adopted by the IFSSH Committee on Congenital Conditions. In 2014 the Committee recommended adoption of a revised classification system for Congenital Anomalies of the Hand and Upper Limb, as proposed by Drs Oberg, Manske and Tonkin, popularly known as the 'OMT' (see previous report).

The Committee recommended review of the 'OMT' system at 3-year intervals according to responses from the hand surgery community. The Committee invited comments and advice based on improvements in knowledge at the time of these reviews when the classification may be modified.

The current update was chaired by Dr. Charles Goldfarb. The 2015 OMT was examined to address comments given by others through different publications and also through the committee's own experience when using the OMT. The following are the main changes:

(1) Simplification of morphology to single conditions rather than combined ones

(2) Addition of useful terms, e.g., ectodermal elements, early or late patterning

(3) Removal of conditions no longer considered as congenital in origin, e.g., trigger digit

(4) Movement of conditions to new classification following improved knowledge in etiology, e.g., cleft hand

(5) Movement of conditions to new classification for clarity and improved clinical use, e.g., grouping arthrogryposis, camptodactyly and thumb in palm in the same grouping under 'Dysplasias'.

(6) Adding the Online Mendelian Inheritance of Men (OMIM) links to the various syndrome

NB: This report contains the 2020 OMT Classification as approved by the IFSSH Executive Committee. The full update and rationale for changes are in Goldfarb et al., JHS (American), 2020 as a Landscaped article. An outline is also given in Lam et al, JHS (European) as a Further Knowledge article, with accompanying letters from different perspectives: European (Wiebke et al, 2020) and Australian experiences (McCombe et al).

OMT CLASSIFICATION OF CONGENITAL HAND AND UPPER LIMB ANOMALIES

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 - clavicular to phalangeal)
III. Transverse deficiency (without ectodermal elements)
   a) Amelia
   b) Segmental (various levels - clavicular to phalangeal)
IV. Intersegmental deficiency (Phocomelia)
   a) Proximal (missing or shortened humerus – rhizomelic)
   b) Distal (missing or shortened 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

1. Arthrogryposis Multiplex Congenita

I. Amyoplasia
II. Distal arthrogryposis
III. Other

2. Isolated

I. Camptodactyly
II. Thumb in palm deformity
III. 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 most relevant; however, many other syndromes have a limb component categorized under "B. Others".