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

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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/a plastic 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)
      6. Carpenter (MIM #201000)
      7. Catel-Manzke (MIM #616145)
      8. Cornelia de Lange (5 types)
         Type 1) (MIM #122470)
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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))

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) Frontometafysal 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 abnormalities (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. VACTERL 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”.