

The Classification of Congenital Hand Abnormalities

Konjenital El Anomalilerinin Sınıflandırılması

Tuba Tülay Koca¹, Aydın Arslan²

¹State Hospital, Physical Medicine and Rehabilitation, Malatya, Turkey

² State Hospital, Orthopedics and Traumatology, Malatya, Turkey

Dear Editor,

Congenital hand anomalies may present in a varying range from slightly recognisable issues to complete absence of the upper extremity. The majority of upper limb deformities cause some minor neurological deficits. When these deformities prompt functional or cosmetic losses, they should be considered for surgical intervention (1).

Although there are many classifications for congenital hand anomalies, Swanson's classification based on embryological development and clinical findings is the most accepted one (2). In this study, we aim to present the case of a 25-year-old male patient with congenital hand deformity, who we failed to fit in a single Swanson category due to underlying multiple embryogenic development issues, in the light of the notable clinical findings. As the 25-year-old male patient presented in our clinic, we immediately detected deformities in the hands in the first examination. We learnt that he had undergone surgery for polydactyly (six fingers) as a small boy and shared similar findings with his brother. The patient was normal size. Cognitive assessments were within normal limits. All the fingers had clinodactyly; the patient also had camptodactyly in the second left, third and fifth right fingers, expanded base in both forth fingers, and hypoplasia in both thumbs. The fourth and fifth fingers of the left hand showed aplasia while all the nails of all fingers had dysplasia. The dermatology clinic evaluated the patient as "nail patella syndrome" (Figure 1).

Radiological examination revealed shortening (brachydactyly) in all the metacarpi of fingers on both hands, hyper-segmentation (poly-phalangy) in the second, third, and fourth left fingers, metacarpal duplication-related polydactyly in the fourth right finger, metacarpus and duplication-induced polydactyly in the proximal phalanx of the fourth left finger, and aplasia in the distal phalanx of the third left finger (Figure 2).

Limb development takes place in the 3rd-8th gestational weeks. Development of congenital anomalies are usually related to genetic, environmental, and unknown causes (4).



Figure 1. Anteroposterior conventional radiography showing shortening (brachydactyly) in all the metacarpi of fingers on both hands, hyper-segmentation (poly-phalangy) in the second, third, and fourth left fingers, metacarpal duplication-related polydactyly in the fourth right finger, metacarpus and duplication-induced polydactyly in the proximal phalanx of the fourth left finger, and aplasia in the distal phalanx of the third left finger.



Figure 2. Photograph showing clinodactyly in all the fingers of both hands, camptodactyly in the second left and third and fifth right fingers, hyper-segmentation in the ring fingers on both hands, hypoplasia in both thumbs, aplasia in the fourth and fifth left finger nails, and displasia in all finger nails.

Although there are at least 112 syndromes defining hand anomalies, this amount only covers 5% of congenital hand anomalies. Mccusick-Kaufman syndrome, Bardet-Biedl syndrome, Meckel-Gruber syndrome, Apert syndrome, Pfeiffer syndrome, Turner syndrome, Rubenstein-Taybi syndrome, Albright hereditary osteodystrophy, Holt-oram syndrome, and Cornelia de Lange syndrome are some of these syndromes (3).

In Swanson classification (2), the first group contains developmental arrest-related anomalies. Development can be longitudinal or transverse. Pause in transverse growth causes amputated limbs in several degrees. Whereas, pause in longitudinal development can be pre-axial, post-axial, central, and intercalated. While pause in pre-axial development can bring about hypoplasia (radial clubbing) in the thumb or radius, post-axial pause can result in ulnar hypoplasia (ulnar clubbing). Correspondingly, pause in central development may cause cleft hand while intercalated pause can result phocomelia (3, 5).

In the second group, as a result of inefficient differentiation and synostosis of the structures, several deformities surface such as dislocated radial head, syndactyly, symphalangism, congenital trigger finger, clinodactyly, camptodactyly, carpal coalitions, and tumour formations (3, 5, 6). Our patient had clinodactyly in all the fingers of both hands and camptodactyly in the second left and third and fifth right fingers, which suggested that the patient could be categorised in the second group.

The third group consists of duplications (polydactyly, mirror hand). Polydactyly is embryologically divided into three subgroups in itself: radial (pre-axial), central, and ulnar (post-axial) (7). Central polydactyly includes duplications of index, long, and ring fingers. Our patient had duplicated metacarpal finger on the right hand (the fourth finger) as well as duplicated metacarpal finger and polydactyly characterised by proximal phalanx on the left hand (the fourth finger). Wassel classification is based on thumb duplications only. Our patient had thus had central polydactyly and hyper-segmentation in his ring fingers on both hands. With these characteristics, the patient also corresponds to the third group. Ulnar polydactyly includes the little finger (7-9).

The fourth group includes excessive growth or gigantism; it refers to macrodactyly and hemihypertrophy which is characterised by the expansion of all parts of the whole finger. Also, it is characterised by lipofibromatosis hamartomas, which are seen as excessive fat accumulation in all the tissues (1, 3, 8, 9).

The fifth group designates hypoplasia of the thumb, radial hypoplasia, and brachydactyly, all of which are caused by poor growth and hypoplasia. This group has been divided into five subgroups by Blauth (1, 2, 3, 3A, 3B, 4, 5) (8-10). Our patient had carpometacarpal instability in both thumbs which is associated with hypoplasia of the thumb characterised by metacarpal hypoplasia (type 3B). Besides, the patient also presented distal phalanx in the third left finger and aplasia in both of the little toes. In addition, all the fingers on both hands and the fourth and fifth toes on both feet showed brachydactyly due to metacarpal and metatarsal shortening. The sixth group is made up of the sporadic congenital inherited adherent band syndrome. Several intrinsic (amniotic band syndrome) and extrinsic factors are held responsible for its formation. It can present amputations at any given level. It can be associated with clubfoot, cleft lip, and cleft palate. The seventh group covers common anomalies and syndromes. Characterised by multiple joint contractors, "arthrogriposis multiplex congenital" is placed in this groups (1, 3, 5, 8, 9, 10).

We failed to come up with a corresponding syndrome since our patient did not show any symptoms apart from congenital hand deformities on the hands. However, we hope that our study can still contribute to the literature since the patient was an exceptional case with clinical and radiological findings corresponding to Swanson classification types 1, 3, and 5 all at the same time.

REFERENCES

1. Bowe J, Conway H. Congenital deformities of the hands. *Plast Reconstr Surg* 1956;18(4):286-90.
2. Swanson AB, Swanson GD, Tada K. A classification for congenital limb malformation. *J Hand Surg Am* 1983;8(5-2):693-702.
3. Flatt AE In: The Care of Congenital Hand Anomalies. 2nd ed. St Louis: Quality Medical Publishing 1994.
4. Rogala EJ, Wynne-Davies R, Littlejohn A, Gormley J. Congenital limb anomalies: frequency and aetiological factors. Data from Edinburg Register of the Newborn (1964-68). *J Med Genet* 1974;11(3):221-33.
5. Sammer DM, Chung KC. Congenital hand differences: embryology and classification. *Hand Clin* 2009;25(2):151-6.
6. Manske PR, Oberg KC. Classification and developmental biology of congenital anomalies of the hand and upper extremity. *J Bone Joint Surg Am* 2009;91(4):3-18.
7. Wassel HD. The result of surgery for polydactyly of the thumb. A review. *Clin Orthop Relat Res* 1969;64:175-93.
8. Blauth W. Congenital Deformities of the Hand. New York: Springer-Verlag 1976.
9. Kozin SH. Upper-extremity congenital anomalies. *J Bone Joint Surg Am* 2003;85:1564-76.
10. Eaton CJ, Lister GD. Toe transfer for congenital hand defects. *Microsurgery* 1991;12(3):186-95.

Received/Başvuru: 30.01.2015, Accepted/Kabul: 19.03.2015

Correspondence/İletişim

Tuba Tülay KOCA
Devlet Hastanesi, Fiziksel Tıp ve Rehabilitasyon, MALATYA,
TÜRKİYE E-mail: tuba_baglan@yahoo.com

For citing/Atıf için

Koca TT, Arslan A. The classification of congenital hand abnormalities. *J Turgut Ozal Med Cent* 2015;22:282-3 DOI: 10.7247/jtomc.2015.2813