DESCRIPTION OF CONGENITAL HAND ANOMALIES: A PERSONAL VIEW

M. A. TONKIN

From the Department of Hand Surgery, University of Sydney, Royal North Shore Hospital, Sydney, Australia

A series of four congenital hand cases exhibiting central clefting are presented. The cases are morphologically similar and exhibit characteristics of both symbrachydactyly and central longitudinal deficiency. The cases demonstrate difficulties in classification by either the IFSSH classification system or the JSSH modification of it. An alternative descriptive approach to classification is suggested.

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Congenital anomalies in the hand demand a reproducible and consistent terminology, a universal language which allows discussion of complex clinical entities, indications for treatment and comparisons of results. In 1976, Swanson's classification was adopted as the standard system by which congenital hand anomalies are described by the International Society for Surgery of the Hand (IFSSH) (Swanson, 1976; Swanson et al., 1983) (Table 1). It was derived from ideas existing in the 1970s regarding limb embryology and is largely based on morphological appearance. More recently, Knight and Kay (2000) presented an extended version, attempting to incorporate a list of all congenital anomalies.

The two major groups of this classification do offer an indication of the timing of the causative insult, by separating ''Failure of formation'' from ''Failure of differentiation''. However, the need to create separate groups for ''Duplication'', ''Undergrowth'' and ''Overgrowth'' is illustrative of the limitations inherent in this system, as these are also examples of a failure of formation or differentiation.

Our knowledge has increased significantly since this valuable contribution by Swanson. Limb growth and development has become one of the main models for genetic research. This is to the advantage of all hand surgeons, whether they have an overwhelming passion for congenital hand anomalies, or not. As a consequence, alterations to the Swanson classification are suggested and have been made according to theories, or facts, of causation and aetiology. This author believes that a classification based on morphology (appearance) is unable to incorporate changes based on these parameters.

THE CLEFT HAND

A consideration of the ''cleft hand'' may best illustrate the contradictions that have arisen. It is also helpful to review our understanding of this fascinating condition. Historically, cleft hands were divided into ''typical cleft hands'' and ''atypical cleft hands''. In the Swanson/IFSSH classification, cases previously considered as ''typical cleft hands'' are classified as a ''Failure of formation of parts'' – IFSSH Group 1 – and are a form of longitudinal deficiency, representing failure of development of the central portion of the hand. However, unlike radial or ulnar deficiencies, they are not usually associated with proximal forearm anomalies. Classically, absence of central ray(s) in the hand is usually bilateral, familial and associated with polydactyly, syndactyly and, also, clefting of feet (Buck-Gramcko, 1985; De Smet and Fabry, 1998; Kay, 1999). In the most minor cases, the middle finger is absent, although some children retain all of the digits, exhibiting clefting only. More severe cases involve an increasing number of absent rays. The central cleft is V-shaped and the first web space is deficient. Transverse bones are common and, when multiple digits are absent, the last remaining digit is the little finger (Fig 1).

Those cases previously considered as ''atypical cleft hands'' are classified as symbrachydactyly – central absence type, under ''Hypoplasia'' – IFSSH Group 5 (Manske, 1993). Symbrachydactyly is usually unilateral, with no foot deformities and no family history (Buck-Gramcko, 1985; De Smet and Fabry, 1998; Kay, 1999). It has been described by Blauth and Gekeler (Buck-Gramcko, 1985) as occurring in a teratological sequence. The initial abnormality is shortening of the middle phalanges of the central three digits, progressing to absence and, then, increasingly, severe deficiency of adjoining parts of the hand. When only one digit remains, it is the thumb. Terminal ectodermal elements of ''absent'' digits usually remain as nubbins with nail remnants. This sequence of increasing severity may be sub-classified morphologically into the following types (Fig 2):

1. Short finger type (Brachymesophalangy + syndactyly)
2. Central absence type
3. Monodactylous type
4. Adactylous type
5. Forearm type

Table 1—Swanson classification of congenital limb anomalies (Simplified from Swanson et al., 1983)

- (I) Failure of formation
  - (A) Transverse arrest
    - Shoulder
    - Arm
    - Elbow
    - Forearm
    - Wrist
    - Carpal
    - Metacarpal
    - Phalanx
  - (B) Longitudinal arrest
    - Radial ray
    - Ulnar ray
    - Central ray (cleft)
  - (C) Intersegmental (phocomelia)
- (II) Failure of differentiation
  - (A) Soft tissue involvement
    - Arthrogryposis
    - Shoulder
    - Elbow and forearm
    - Wrist and hand
      - Cutaneous syndactyly
      - Camptodactyly
      - Thumb-in-palm
      - Deviated finger
  - (B) Skeletal involvement
    - Shoulder
    - Elbow synostosis
    - Forearm synostosis
    - Wrist and hand
      - Osseous syndactyly
      - Carpal synostosis
      - Symphalangia
      - Clinodactyly
  - (C) Congenital tumorous conditions
    - Haemangiotic
    - Lymphatic
    - Neurogenic
    - Connective tissue
    - Skeletal
- (III) Duplication
  - Whole limb
  - Humerus
  - Radius
  - Ulna (mirror hand)
  - Digit
    - Radial polydactyly
    - Central polydactyly
    - Ulnar polydactyly
- (IV) Overgrowth
  - Whole limb
  - Partial limb
  - Digit (macrodactyly)
- (V) Undergrowth
  - Whole limb
  - Whole hand
  - Metacarpal
  - Digit
    - Brachysyndactyly
    - Brachydactyly

Table 1. (*continued*)

- (VI) Congenital constriction band syndrome
  - Constriction band
  - Acrosyndactyly
  - Intrauterine amputation
  - Combination of above
- (VII) Generalized skeletal abnormalities
  - Chromosomal abnormalities
  - Other generalized abnormalities

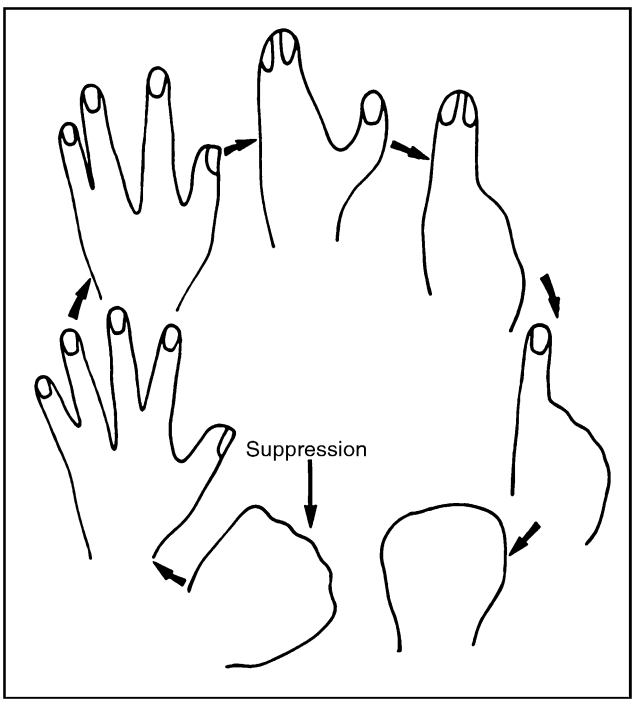

Fig 1 Increasing severity of symbrachydactyly (from Maisels, 1970).

"Atypical cleft hand", therefore, represents one part of the teratological sequence of symbrachydactyly (Buck-Gramcko, 1985; De Smet and Fabry, 1998; Kay, 1999). The appearance is of a broad flat cleft (the central three rays are often absent, although the index finger may be present) between a relatively normal thumb and a little finger, which is typically radially deviated, contains only one interphalangeal joint and is short. The cleft contains finger nubbins that usually possess nail remnants (Fig 2(2)).

If we are to accept this terminology, the descriptions "typical" and "atypical" cleft hands are replaced by "central longitudinal deficiency" and "symbrachydactyly – central absence type" respectively. This reflects the current IFSSH classification (Manske, 1993). However, the Japanese Society for Surgery of the Hand (JSSH)