Congenital Hand Committee report 2007

Longitudinal radial and ulnar deficiencies

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Longitudinal deficiencies are associated with hypoplasia or aplasia of either radius or ulna and/or that of the digits on radial or ulnar side of the hand. Each may be associated with elbow and arm deficiencies. Longitudinal ray deficiency is considered to be caused by a deficit of mesenchymal cells in the early developmental stage of limb bud growth.

References:

Radial longitudinal deficiency

The congenital deformities in which radial digits are hypoplastic or absent and/or the radius is hypoplastic or absent are called radial ray deficiencies. Elbow and arm deficiencies are sometimes associated with these conditions. Radial deficiencies should be expressed addressing the presence or absence and degree of radial dysplasia in the forearm and hand as well as the presence or absence of elbow, arm and shoulder dysplasia, which may occur in various combinations. Radial deficiencies usually appear as an isolated limb deformity, but also with abnormalities of other body systems and may appear as a part of congenital syndrome.
Radial digit hypoplasia/aplasia

Blauth classified hypoplastic thumb into five grades as follows:

Grade 1 - Minimal hypoplasia of the thumb ray, this being short with mild hypoplasia of the thenar muscles.

Grade 2 - The first metacarpal is hypoplastic and lies in an adducted position. There is instability of varying degrees at the metacarpophalangeal joint, particularly in association with loss of ulnar collateral ligament integrity. The first web is narrowed and shallow. There is thenar muscle hypoplasia.

Grade 3 – There is hypoplasia or absence of the proximal part of the first metacarpal. The former has been entitled “3A” and the later “3B” by Manske. MP joint instability may be global, the thenar musculature exhibits a greater degree of hypoplasia and there are extrinsic tendon anomalies.

Grade 4 - The first metacarpal is absent. The thumb containing phalanges is connected to the hand by a skin bridge – the floating thumb or pouce flottant.

Grade 5 - Absence of the thumb.

It should be noted that there are a number of variations in presentation of thumb hypoplasia which make precise classification difficult. For instance, anomalies of extrinsic tendons (Grade 3) may occur in association with mild hypoplasia of the base of the first metacarpal (Grade 2) with a satisfactory, though not a normal, thumb CMC joint. Interphalangeal joint motion is often diminished in Grade 2 thumb hypoplasia in association with flexor pollicis longus anomalies. A pollex abductus (see below) may be present. Some have suggested that Grade 2 be sub-divided according to the degree of instability of the metacarpophalangeal joint.

References:

There are other types of radial digit hypoplasia, including the five fingered hand, absence
of more than two radial digits, and an hypoplastic thumb associated with syndactyly of the thumb and index finger. The five fingered hand may also be termed a non-opposable triphalangeal thumb. There is a spectrum of radiological appearances with graduation from a five-fingered hand in which the radial ray metacarpal resembles that of a finger, to a triphalangeal thumb in which the radial ray metacarpal resembles that of a thumb but has three phalanges.

A number of joint and extrinsic tendon anomalies may be associated with thumb hypoplasia. The radial digits are more likely to exhibit a degree of stiffness, particularly a flexion contraction of the proximal interphalangeal joint of the index finger. The tendency to flexion deformities is more evident in the radial digits than in the ulnar digits. There may be diminished flexion of interphalangeal joints in conjunction with flexor pollicis longus anomalies. A pollex abductus describes an extrinsic connection between flexor pollicis longus and the extensor tendon on the radial aspect of the thumb at the metacarpophalangeal joint. Contracture of the flexor pollicis longus abducts the thumb at the metacarpophalangeal joint.

In thumb hypoplasia the radial artery may be absent and the radial carpal bones are hypoplastic.

References:

Radial deficiency in the forearm

Hypoplasia or absence of the radius has been divided into four grades by Bayne. James, McCarroll and Manske have added a further category in which the radius is fully formed but there is radial deviation at the wrist.

Grade 0 – Radial deviation of the wrist without hypoplasia of the radius, possibly due to radial carpal bone hypoplasia.
Grade 1 – Hypoplasia of the radius with a distal physeal growth deficiency.
Grade 2 – More severe hypoplasia of the radius with distal and proximal physeal growth
deficiencies.
Grade 3 – Partial absence of the radius – most often with some proximal radius present and the existence of a fibrous anlage resulting in curvature of the ulna.
Grade 4 – Absence of the radius.

References:

Associated elbow deficiency

Deficiency of the elbow may appear as part of a radial deficiency. Radio-ulnar synostosis may be associated with a radial ray deficiency, often in Holt-Oram syndrome.

a. Contracture of the elbow joint  
b. Humero-radial synostosis  
c. Radial head dislocation  
d. Radio-ulnar synostosis

Deficiency of the proximal part of the arm

This includes:

a. Hypoplasia and aplasia of the humeral head; and  
b. Hypoplasia of the glenoid.

References:

Common syndromes in which radial deficiency is associated

It is necessary to consider the possibility of a syndrome when other system anomalies are
evident.

VACTERL association
Fanconi anemia
Holt-Oram syndrome
Ventriculoradial dysplasia
Craniosynostosis-radial aplasia syndrome (Baller-Gerold syndrome)
Nager syndrome hemifacial microsomia
Goldenhar syndrome (oculo-auriculo-vertebral syndrome)
Huberg-Haywood syndrome (oro-cranio-digital syndrome)
Rothmund-Thomson syndrome
Duane radial dysplasia syndrome
Levey-Hollister (LARD) syndrome
Seckel syndrome
Trisomy 18(Edward syndrome)
Trisomy 21 (Down syndrome)
Trisomy 13(Patau syndrome)
Thrombocytopenia-absent-radius syndrome
Aase-Smith syndrome
Instituto Venzolano de Investigaciones Científicas (IVIC) syndrome
Okihiro syndrome (Duane radial ray syndrome)

Ulnar longitudinal deficiency

In ulnar deficiency, dysplasia of the hand, that of the forearm, elbow and arm appear in various combinations and deficiency in this category should be expressed with reference to each anatomical region.

Ulnar deficiency in the hand

A great variety of hand anomalies may be associated with ulnar deficiencies. Absence of multiple digits is common. The thumb is always present but sometimes it may be hypoplastic. Abnormality of the first web space with loss of width and depth and a tendency to a planar (pronated) thumb position is common. Other anomalies, such as syndactyly and polydactyly, may be associated.
a. Hypoplasia of the little finger  
b. Absence of the 5th digital ray  
c. Absence of two ulnar digital rays  
d. Absence of three ulnar digital rays  
e. Absence of four ulnar digital rays  
f. Abnormalities of the radial digit  
   1) hypoplasia of the digit  
   2) syndactyly  
   3) polydactyly

Ulnar deficiency in the forearm

a. Ulnar deviation of the wrist without hypoplasia of the ulna  
b. Hypoplasia of the ulna  
c. Partial absence of the ulna with radial bowing  
d. Total absence of the ulna

Reference:

Elbow deficiency in ulnar deficiency

a. Contracture of the elbow joint  
b. Humeroradial synostosis  
c. Radial head dislocation

Ulnar deficiency in association with proximal limb deficiency

a. severe radiohumeral synostosis with distal humeral duplication  
b. severe radiohumeral synostosis with a large medial exostosis that resembles large medial condyle

Reference:
Ulnar deficiency as part of a congenital syndrome

Schinzel syndrome (ulnar-mammary syndrome)
Postaxial acrofacial dysostosis
Femur-fibula-ulna syndrome
Cornelia de Lange syndrome