

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 :

- Ogino T, Kato H. Clinical and experimental studies on ulnar ray deficiency. *Handchir Mikrochir Plast Chir.* 1988 Nov;20(6):330-7.
- Ogino T, Kato H. Histological analysis of myleran induced oligodactyly of longitudinal deficiency in rats. *Handchir Mikrochir Plast Chir.* 1988 Sep;20(5):271-4.
- Kato H, Ogino T, Minami A, Ohshio I. Experimental study of radial ray deficiency. *J Hand Surg [Br].* 1990 Nov;15(4):470-6.
- Otsuji M, Takahara M, Naruse T, et al: Developmental abnormalities in rat embryos leading to tibial ray deficiencies induced by busulfan. *Birth Defects Res A Clin Mol Teratol.* 2005 Jun;73(6):461-7.

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:

- Blauth W. The hypoplastic thumb. *Arch Orthop Unfallchir.* 1967;62(3):225-46.
- Manske PR, McCarroll HR Jr. Reconstruction of the congenitally deficient thumb. *Hand Clin.* 1992 Feb;8(1):177-96.
- Smith P. Congenital. In: Smith P (ed): *Lister's The Hand: Diagnosis and Indications* (4th Ed). London, England: Churchill Livingstone. 2002: 505.

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:

- Blauth W. Die hypoplastische Daumen. Arch Orthop Unfallchir. 1967;62(3):225-46.
- Manske PR, McCarroll HR Jr, James M: Type III-A hypoplastic thumb. J Hand Surg [Am]. 1995; 20(2):246-53
- Tupper JW: Pollex abductus due to congenital malposition of the flexor pollicis longus. J Bone Joint Surg Am. 1969;51(7):1285-90.
- Lister G: Pollex abductus in hypoplasia and duplication of the thumb. J Hand Surg 1991;16(4):626-33.

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 physal growth deficiency.

Grade 2 – More severe hypoplasia of the radius with distal and proximal physal 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:

Bayne LG, Klug MS. Long-term review of the surgical treatment of radial deficiencies. *J Hand Surg [Am]*. 1987;12(2):169-79.

James MA, McCarroll HR Jr, Manske PR. The spectrum of radial longitudinal deficiency : a modified classification. *J Hand Surg [Am]*. 1999;24(6):1145-55.

Mo JH, Manske PR. Surgical treatment of type 0 radial longitudinal deficiency. *J Hand Surg [Am]*. 2004;29(6):1002-9.

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:

Goldfarb CA, Manske PR, Busa R, Mills J, Carter P, Ezaki M: Upper-extremity phocomelia reexamined: A longitudinal dysplasia. *J. Bone Joint Surg [Am]* 87:2639-2648, 2005.

Common syndromes in which radial deficiency is associated

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

evident.

VACTERLS 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 Venezolano de Investigaciones Cientificas (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:

Bayne L. Ulnar Club Hand. In: Green DP: *Operative Hand Surgery* (2nd ed.) New York, Churchill Livingstone. 1998:291-305

Havenhill TG, Manske PR, Patel A, Goldfarb CA. Type 0 ulnar longitudinal ray deficiency. *J Hand Surg [Am]*. 2005;30(6):1288-93

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:

Goldfarb CA, Manske PR, Busa R, Mills J, Carter P and Ezaki M: Upper-Extremity

Phocomelia Reexamined: A Longitudinal Dysplasia. *J. Bone Joint Surg. Am.*87:2639-2648, 2005.

Ulnar deficiency as part of a congenital syndrome

Schinzel syndrome (ulnar-mammary syndrome)

Postaxial acrofacial dysostosis

Femur-fibula-ulna syndrome

Cornelia de Lange syndrome