Generalized Skeletal Abnormalities

Andrew J. Watt, MD[a], Kevin C. Chung, MD[b,*]

KEYWORDS

- Congenital trigger thumb
- Clasped thumb • Achondroplasia
- Madelung deformity • Congenital hand

The upper extremity is complex and its function relies on the precise arrangement of bone, cartilage, muscle, tendon, nerve and vascular structures acting in concord. This structure is, in turn, reliant upon the highly orchestrated temporal and spatial development, coordinated by molecular signaling and complex cellular interactions. This coordinated process allows undifferentiated mesenchyme and ectoderm to develop into the complex human limb. Given this complexity, it is not surprising that limb and hand abnormalities are among the most common congenital anomalies.

Congenital deformities affect 1% to 2% of all newborns and 10% of these deformities involve the upper extremity.[1,2] Congenital limb anomalies are classified according to the embryonic failure that underlies their clinical presentation. The most widely accepted classification, proposed by Frantz and O'Rahilly[3,4] and modified by Swanson, divides these anomalies into one of seven categories of embryologic failure: failure of formation of parts, failure of differentiation, duplication, overgrowth, undergrowth, congenital constriction band syndrome, and generalized skeletal abnormalities. The category of generalized skeletal anomalies is often applied for disorders that cannot be discretely categorized into one of the other six classes of embryologic failure. This article describes Swanson's broad category of generalized skeletal abnormalities, focusing on anomalies that are commonly seen in practice, including congenital trigger finger, congenital clasped thumb, and Madelung's deformity, as well as skeletal hand deformities that are characteristic of a generalized bone and connective tissue disorders, including achondroplasia and Marfan syndrome.

CONGENITAL FLEXION DEFORMITIES OF THE THUMB

The thumb plays a unique role in hand function. The degrees of freedom in flexion, extension, circumduction, and opposition of the thumb allow the hand to perform eloquent tasks while its strength imparts powerful grip. Thumb function develops gradually over the first year of life. Throughout the first four months of life, the thumb is characteristically held in a flexed, adducted position within the palm of the hand and is only intermittently extended. Thumb extension progresses around four months and ultimately culminates in the highly coordinated fine pinch grasp around one year of life.[5] In a small subset of children, this coordinated progression of thumb function is impaired and congenital flexion deformities of the thumb are present. Congenital flexion deformities result from musculotendinous imbalance, joint abnormalities, and intrinsic tendon and tendon sheath anomalies. The most common causes of persistent congenital thumb flexion are congenital trigger thumb and congenital clasped thumb.

Supported in part by a Midcareer Investigator Award in Patient-Oriented Research (K24AR053120) from the National Institute of Arthritis and Musculoskeletal and Skin Diseases (Kevin C. Chung).

[a] Division of Plastic and Reconstructive Surgery, Department of Surgery, Stanford University Hospitals & Clinics, 770 Welch Road, Suite 400, Palo Alto, CA 94304, USA
[b] Section of Plastic Surgery, Department of Surgery, University of Michigan Medical School, 2130 Taubman Center, 1500 E. Medical Center Drive, Ann Arbor, MI 48109-0340, USA
* Corresponding author.
E-mail address: kecchung@umich.edu (K.C. Chung).

Hand Clin 25 (2009) 265–276
doi:10.1016/j.hcl.2008.12.008
0749-0712/08/$ – see front matter

CONGENITAL TRIGGER THUMB

Congenital trigger thumb is characterized by persistent flexion of the interphalangeal (IP) joint. The thumb is easily flexed; however, extension at the IP joint is impaired. True triggering is rare and motion at the metacarpophalangeal joint is unencumbered. The initial descriptions of triggering are attributed to Notta,[6] a French physician who described nodules occurring within the flexor tendons resulting in pathologic triggering of the affected digit. Notta's original reports were based on his experience with adult trigger finger; however, the term Notta's node has persisted only in descriptions of congenital trigger finger.

The age of onset, etiology, and treatment of congenital trigger thumb remain a matter of supposition and debate. Congenital trigger thumb is estimated to constitute 2.2% of all upper extremity congenital anomalies with an incidence of 1 in 2000 live births.[1,7] The diagnosis is typically made after 6 months of life. Attempts to establish the incidence at birth have provided substantial evidence that congenital trigger thumb is more likely acquired after birth and that the term "congenital trigger thumb" is in itself a misnomer. Rodgers and Waters[8] examined 1046 newborns, whereas Slakey and Hennrikus[9] examined 4719 newborns for the presence of thumb triggering at birth. No trigger thumbs were identified. Kikuchi and Ogino[10] examined 1166 neonates within 14 days of birth and found no congenital trigger thumbs. Of these 1166 patients, two patients went on to develop trigger thumbs within the first year of life. Authors have, however, pointed to cases of fraternal twins,[11] association with trisomy 13 (Pateau Syndrome)[12] and families with generational occurrence suggesting a heritable component or predisposition.[13]

Etiology

From a purely mechanical perspective, congenital trigger thumb results from a discrepancy between the flexor pollicis longus (FPL) tendon and the overlying flexor tendon sheath. Fusiform thickening of the FPL tendon, referred to as Notta's nodule, is pathognomonic of congenital trigger thumb. The nodule is characteristically palpable at the level of the metacarpophalangeal (MCP) joint. The strong flexors of the thumb allow the nodule to be pulled proximally, beneath the retinacular pulley system; however, the weaker extensor apparatus is unable to overcome the stenosis at the level of the first annular (A1) pulley. As a result the thumb takes on a persistent flexed position at the IP joint (**Fig. 1**).

The exact pathologic sequence resulting in congenital trigger thumb is not defined. Theories have focused on the two pathologic components: Notta's node and the A1 pulley. Microscopic examination of Notta's node reveals normal collagenous tendon architecture infiltrated with lymphocytes and monocytes, suggestive of traumatic inflammation.[14] This fusiform enlargement of the tendon resolves following surgical release of the A1 pulley.[15] The A1 pulley itself may be thickened and its anatomy variable. No studies have examined the anatomy of the digital pulley system in infants; however, numerous studies in the adult hand have demonstrated variable anatomy that may predispose the thumb to triggering. Schmidt and Fischer described[16] a Y-shaped fiber complex located at the base of the proximal phalanx in 90% of adult thumbs. They also described thin accessory pulley fibers located between the A1 pulley and this Y-shaped fiber complex, in effect, extending the A1 pulley complex. Bayat and colleagues[17] dissected 14 hands, describing a distinct annular pulley located between the A1 and the oblique pulley, which they designated the variable pulley. Variable anatomy exists within the fibrous pulley system that may contribute to the development of thumb triggering.

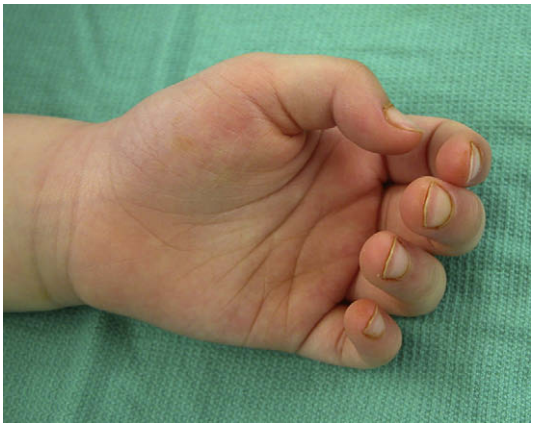

Fig. 1. A typical trigger thumb in a 5-year-old girl. Please note the flexion at the IP joint.

Histologic studies support the theory that tendon inflammation—manifest as enlargement of the FPL tendon—results in a discrepancy in size between the tendon and its associated pulley system. This discrepancy in size then contributes to further trauma and inflammation of the tendon and overlying retinacular pulley. What initiates this cycle of trauma and inflammation is a matter of supposition. Several authors speculate that the neonatal flexed thumb posture may produce chronic irritation of the FPL tendon with impingement occurring at the level of the A1 pulley, and that the stenosis may be exacerbated by the underlying cartilaginous sesamoids.[10,14] Other