Case Report

Autosomal Dominant Inheritance Pattern for Trigger Thumb

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Notta, in 1850, provided the first clinical description of trigger thumb.[1,2] He reported the presence of a nodule on a flexor tendon that was interfering with its gliding motion. Since this first description, the etiology of trigger fingers has been debated. Although a genetic basis has been suspected, the evidence for such in reported cases in the English literature is sparse. We present a pedigree analysis with multiple cases of trigger thumb that supports a genetic basis for this condition.

Case Report

The proband (III-6), a 6-year-old girl, was referred to the plastic and hand surgery service for evaluation of a flexion deformity of her left thumb. The grandparents recalled noticing the deformity when she was 6 months old. She had no other congenital deformities and was in good health. Physical examination showed the interphalangeal joint of the left thumb in a flexed position and a nodule at the A-1 pulley. Another nodule was palpated over the right thumb flexor sheath. No restriction of motion was noted for the right thumb. During the operation, trigger thumb release by division of the A-1 pulley was performed. Ten days postoperatively, the patient had full range of motion of her left thumb.

The family history was significant for a number of other affected family members (Fig. 1). The patient's 2-year-old sister also presented with bilateral nodules and a right trigger thumb. After release of the right trigger thumb, she presented 6 months later with triggering of the left thumb, which was also released surgically. The proband's father had a congenital trigger thumb, surgically released at age 2 years, and his brother had bilateral trigger thumbs, surgically corrected at age 3 years. The proband's cousin also had bilateral trigger thumbs that were repaired when he was 3 years old. The patient's second cousin also had a trigger thumb by report. No other congenital deformities were identified in this pedigree.

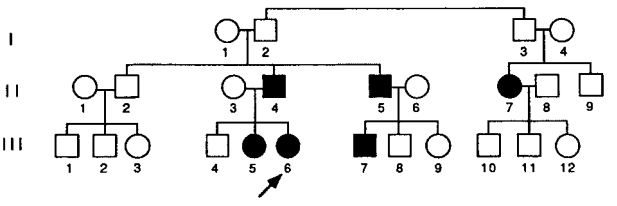

FIG. 1. The proband (III-6) is a 6-year-old girl who presented with a left trigger thumb. The patient's sister (III-5) was eventually treated for bilateral trigger thumbs. The proband's father (II-4) had been treated for a unilateral trigger thumb at age 2. Her uncle (II-5) had bilateral trigger thumbs released at 3 years of age. A more distant relative (II-7) was also reported to have had a trigger thumb released at an early age.

Discussion

The incidence of trigger finger has been quoted as 2 percent of all congenital upper extremity anomalies or one in 2000 office visits.[3,4] In children and adults, the thumb is the most common digit involved.[1,5,6]

The etiology of trigger finger is not clear. Rodgers and Waters examined 1046 newborns prospectively to determine the incidence of trigger digits at birth but found no trigger digits in any of the newborns.[2] They concluded that trigger digits in children are acquired. Among 4719 newborns prospectively examined by Slakey and Hennrikus, trigger thumb was not found at birth.[7] Others share the view that trigger thumb is an acquired deformity.[6–8]

However, Ger et al. suggest trigger thumbs are not reported at birth because of the normal flexion posture of the thumb, which persists until the age of 3 months.[4] Weber reported a 24-month-old boy with unilateral trigger thumb, and his father with untreated bilateral trigger thumbs, with spontaneous improvement.[9] Bollinger and Fahey also alluded to a congenital predisposition when trigger thumbs were seen in fraternal twins.[10]

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The genetic predisposition was also supported by Neu and Murray's report of identical twins with involvement of the same digit.[11]

After reviewing the literature, our report is the largest series of family members affected by congenital trigger thumbs. Although not diagnosed at birth, most family members recall the affected individuals' thumbs being fixed in a flexed position at an early age. The direct transmission of trigger thumb from parents to offspring in our pedigree suggests an autosomal dominant inheritance. However, reduced penetrance is suggested because, by history, neither I-2 nor I-3 had trigger thumb.

A spontaneous recovery of 30 to 50 percent has been reported in the literature.[12,13] Splint therapy also has been advocated.[14] However, the currently accepted treatment for trigger thumb is its surgical release by division of the A-1 pulley.[4,6,7,12,15] No consensus has been reached on the age at which surgical release of trigger thumb is recommended. Some reports showed residual joint contracture in patients who had the operation after 3 years of age.[12,15] In our series, the family members were diagnosed with trigger thumbs after age 3 and underwent surgical treatment. No residual contracture was noted in the series.

Trigger thumb is an unusual problem in children. We report a pedigree that supports an autosomal dominant mode of inheritance with reduced penetrance. No other associated anomalies were found. Surgical treatment of trigger thumb by division of the A-1 pulley was successful in all patients with no residual contracture.

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Discussion

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Discussion by Christopher Cunniff, M.D.

Over the past decade, there has been a staggering increase in the number of medical disorders that have been found to have a genetic basis. With the recent completion of a refined draft of the human genome, these numbers are sure to increase.[1,2] In the accompanying article by Shim et al., trigger thumb is now added to this group of disorders, at least for some families. Documentation of such a family may provide information about etiology, recurrence risk, and natural history of this rare condition.

In the investigation of the genetic basis for any disorder, it is first necessary to have a good phenotypic description. Once the phenotype is established, further genetic investigation can be undertaken to localize the causative gene and, eventually, to clone this gene and recognize its place within other developmental pathways. Although recognition of trigger thumb as a genetic trait is only a first step, it is an important one.

The recurrence risk for genetic disorders is important to communicate to affected individuals or to at-risk individuals within families. Information from this family indicates that the possibility of autosomal dominant inheritance should be considered and discussed with patients who have trigger thumb or with families with a history of trigger thumb. Furthermore, knowing that a family member is at risk may provide for earlier detection and a better outcome. As the authors indicate, although not seen in their series, some studies have suggested that residual contractures are commonly seen when trigger thumb is released after age 3 years. Early recognition and surgical correction may therefore produce a better outcome in affected individuals.

Finally, it should be recognized that discovery of the genetic basis for one disorder may shed light on other related conditions. For example, both achondroplasia, a common dwarfing condition, and isolated craniosynostosis are associated with mutations of one of the fibroblast growth factor receptors.[3,4] Recognizing these relationships has broader implications for both diagnosis and recurrence risk. Although the family presented by Shim et al. does not yet bring us to an understanding of how trigger thumb might relate to other surgical disorders, it provides the basis for further investigation of etiologic relationships between this condition and other joint and connective tissue abnormalities.

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