Surgical Treatment of Macrodactyly

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Macrodactyly, enlargement of one or multiple digits, was described in the literature nearly 200 years ago. This is an exceptionally uncommon diagnosis that has led to a paucity of descriptive literature on the treatment options. Because the literature is scarce, and the frequency with which hand surgeons encounter macrodactyly is even scarcer, treatment can be a formidable task often left exclusively to those trained in congenital hand deformity. This article presents our algorithm and surgical techniques for dealing with children with macrodactyly in such a way that should make a complex problem more easily approachable. *(J Hand Surg Am. 2015;40(7):1461−1468. Copyright © 2015 by the American Society for Surgery of the Hand. All rights reserved.)*

Key words Macrodactyly, overgrowth, PI3K-AKT, osteotomy, epiphysiodesis.

Macrodactyly constitutes less than 1% of upper extremity congenital anomalies.[1] Since its description by Von Klein in 1824,[2] there have been only a handful of case series describing the diagnosis and treatment of macrodactyly. As our understanding of the disease process of macrodactyly has evolved, treatment has become more proactive and simultaneously more complex. Humphry's[3] early description of macrodactyly in 1892 displayed basic understanding of the disease, and amputation was the primary method of treatment when the overgrowth grossly interfered with function. Barsky [4]and Tsuge[5] refined treatment options with similar methods to shorten the digit while attempting to maintain aesthetics and function.

The past 50 years have seen a number of modifications and improvements in treatment, although none have truly been able to create a normal digit in terms of appearance and function. Recent studies investigated the disease process and the etiology of a number of overgrowth syndromes, including macrodactyly. Somatic mosaicism of a mutation in a protooncogene in the PI3K-AKT pathway[6] has been implicated. This postzygotic mutation occurs in the early embryo and affects some but not all of the cells in the involved areas. Typically the abnormality is within a nerve territory, as is often exemplified by macrodactyly. Although not a malignancy, the tumor-like condition results from abnormal regulation of growth, and consequently no 2 cases are identical. Although gene-targeted therapy is a goal, this remains in the future, and current options are still limited to surgery. Our preferred treatment incorporates elements from our forebears and includes observation, amputation, and everything in between.

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INDICATIONS/CONTRAINDICATIONS

The primary indication to operate on a digit with macrodactyly is to improve function. Although aesthetics are important, the scarring and stiffness that accompany most surgical techniques should be stressed when counseling the parents or a patient with mild macrodactyly (Fig. 1). A distinction must also be made as early as possible as to whether the patient has static or progressive-type macrodactyly. Although the treatment principles are the same, patients with progressive macrodactyly can expect to undergo surgery earlier, have a higher number of subsequent procedures,[2] and develop earlier interphalangeal joint arthrosis.[7]

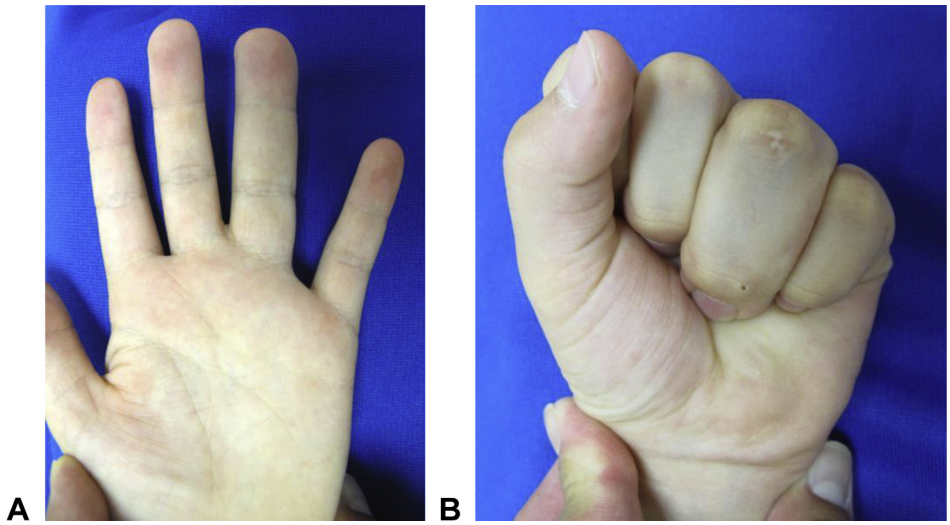

FIGURE 1: Mild macrodactyly of the ring finger with **A** minimal cosmetic and **B** no functional deficits.

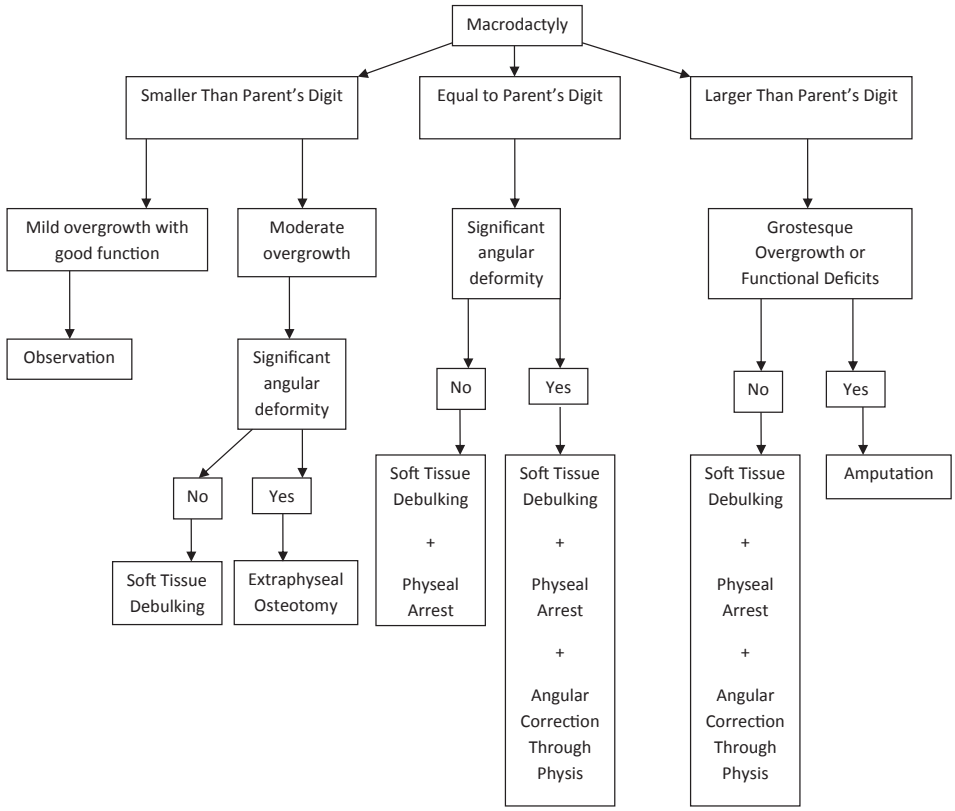

FIGURE 2: Depiction of our treatment algorithm.

A major branch point in deciding how to treat children with macrodactyly is determined by the length of the digit compared with the same digit on the parent of the same sex. If the digit is circumferentially overgrown but shorter than the parent's digit, we offer soft tissue debulking alone. If the digit is approaching the size of the parent's, we recommend debulking along with ephysiodeses to halt further longitudinal growth. We rarely attempt angular correction as an earlier procedure, separate from epiphysiodesis. Rather, we wait until the time of growth arrest, so that osteotomies can be done through the physes and both procedures can be performed simultaneously. For the child who presents with a digit that has already outgrown that of the parent, we avoid shortening procedures such as those described by