Correction of Congenital Upper Extremity Contracture with Flexor Origin Slide

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Abstract

The clinical features seen in patients with chromosome 16p13.11 duplications are poorly described and reports of successful treatment modalities for their musculoskeletal issues are limited. Existing literature has highlighted a clinical picture consistent with a general hypermobility syndrome. We report a 9-year-old male with a 16p13.11 microduplication who presented with worsening contractures of his hands which were present since birth but had gradually worsened and caused dysfunction. He was surgically treated with a flexor origin slide procedure and z-lengthening of the hypothenar skin on his right hand, successfully restoring full motion and function of his fingers.

Introduction

Chromosome 16p13.11 microduplication is a rare disorder with uncertain clinical significance. It has been linked to patients with cognitive impairment, developmental delay, autism, and schizophrenia[3,4,8] . Affected populations have been described as being phenotypically normal to having a diverse set of physical characteristics. In a series of 10 patients with 16p13.11 microduplications, Nagamani et al identified different phenotypic manifestations[4]. They described dysmorphic facial features such as epicanthic folds, low set ears, posterior rotated ears, and nasal abnormalities. Musculoskeletal manifestations included craniosynostosis, hypermobility, polydactyly, syndactyly, arachnodactyly, and pes planus.

We present a case of a 9 year-old male who was referred to our orthopaedic surgery upper extremity clinic for contractures of bilateral hands, right worse than left, with associated pain and difficulties with daily activities. Rather than hypermobility, which is more commonly reported in patients with 16p13.11 microduplications, our patient had stiffness of his fingers. He failed nonsurgical measures including a year of occupational therapy with stretching and nighttime splinting. We performed a flexor origin slide procedure to improve his contractures. At the 1 year follow-up, the patient had full extension of his fingers, resolution of contractures, and improved function.

Case Report

Our patient is a 9-year-old right hand dominant male who presented to our clinic with his mother, a phenotypically normal woman, with worsening flexion contractures of both hands over the past two years. His right hand was more affected than the left. He was found to have mildly abnormal facial findings, including a low, narrowed anterior hairline, synophyrs, flattened cheekbones, and midface hypoplasia. The unusual constellation of findings prompted a referral to a geneticist, who ordered a chromosomal microarray. This exhibited an approximately 819 kb gain of 16p13.11. He lacked other findings consistent with this microduplication, such as intellectual delay, autism, or schizophrenia.

Musculoskeletal examination of his right upper extremity revealed ulnar deviation of the index, long, ring and small fingers at the metacarpophalangeal joints (MCPJs), as well as contractures of the MCPJs, and distal interphalangeal joints (DIPJs) of digits 2-5. The ring and small finger active MCPJ motion was limited to an arc of 70-90° (normal 0-100°), and the DIPJs to 45-90° (normal 0-85°). With wrist extension, the patient was unable to extend his fingers actively or passively. However, he was able to extend them fully with the wrist in neutral, signifying an extrinsic etiology of his contractures (Figure 1). Initial treatment of his contractures consisted of a stretching and splinting therapy program supervised by an occupational therapist. Unfortunately, he had minimal symptomatic improvement, so he and his family elected to proceed with surgical correction.

He underwent a flexor origin slide of the right upper extremity and a z-plasty (local soft tissue rearrangement) of the hypothenar aspect of the palm. The purpose of the flexor origin slide was to move the origin of the wrist and finger flexors distally without affecting resting muscle length. An incision was made along the ulnar aspect of his forearm, starting posterior to the medial epicondyle and extending distally down the forearm to the ulnar wrist flexion crease. The entire flexor-pronator mass was elevated from the humerus, ulna, and interosseous

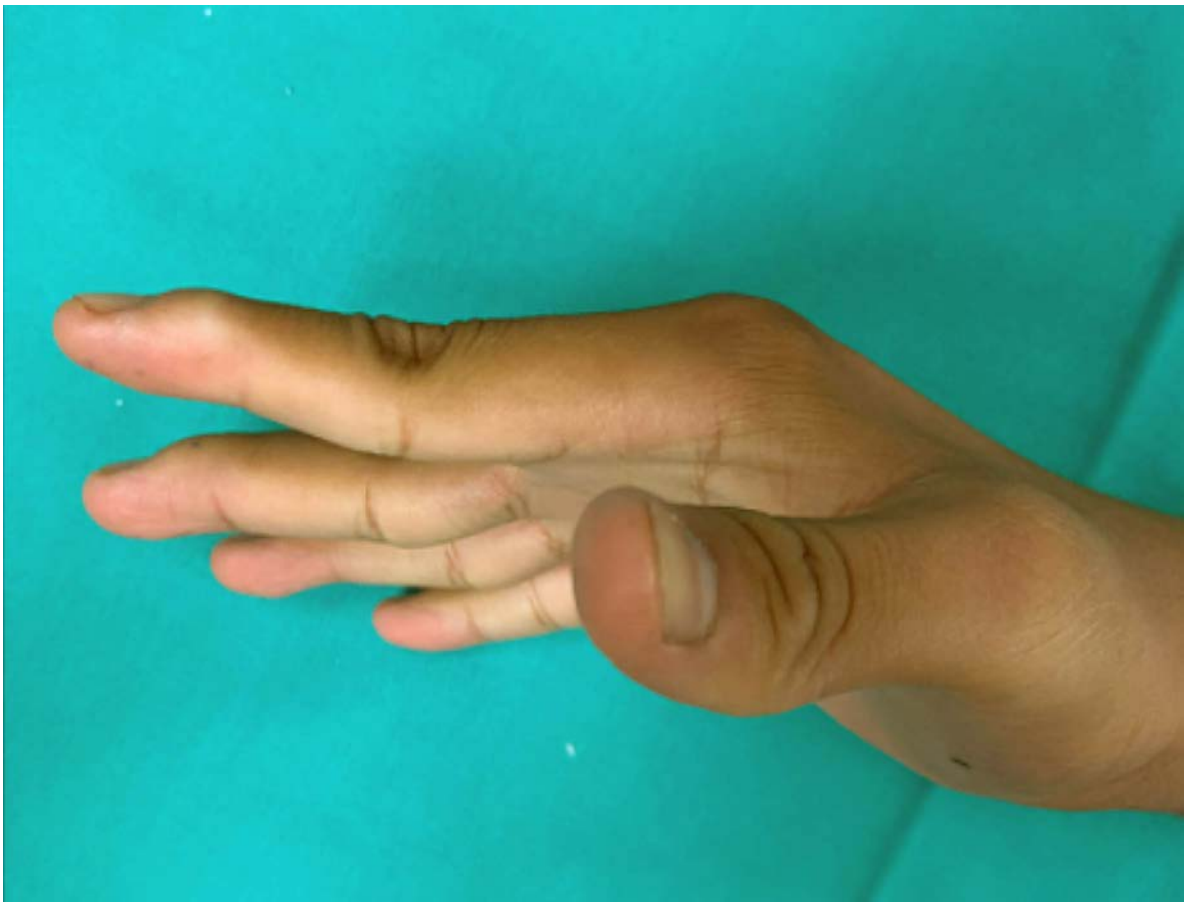

Figure 1. Clinical photograph of hand deformity prior to surgery.

membrane in a subperiosteal fashion, taking care to protect the neurovascular structures. After the release was performed, full passive extension of the wrist and fingers was observed in the operating room (Figure 2).

Postoperatively, the patient was casted with the wrist and fingers in full extension for 6 weeks. He was then referred to an occupational therapist for stretching, ROM, and night splinting. At his 1 year follow-up visit, he had full active and passive ROM of all digits and resolution of his pain and discomfort (Figure 3). He was able to perform all of his desired activities including playing basketball.

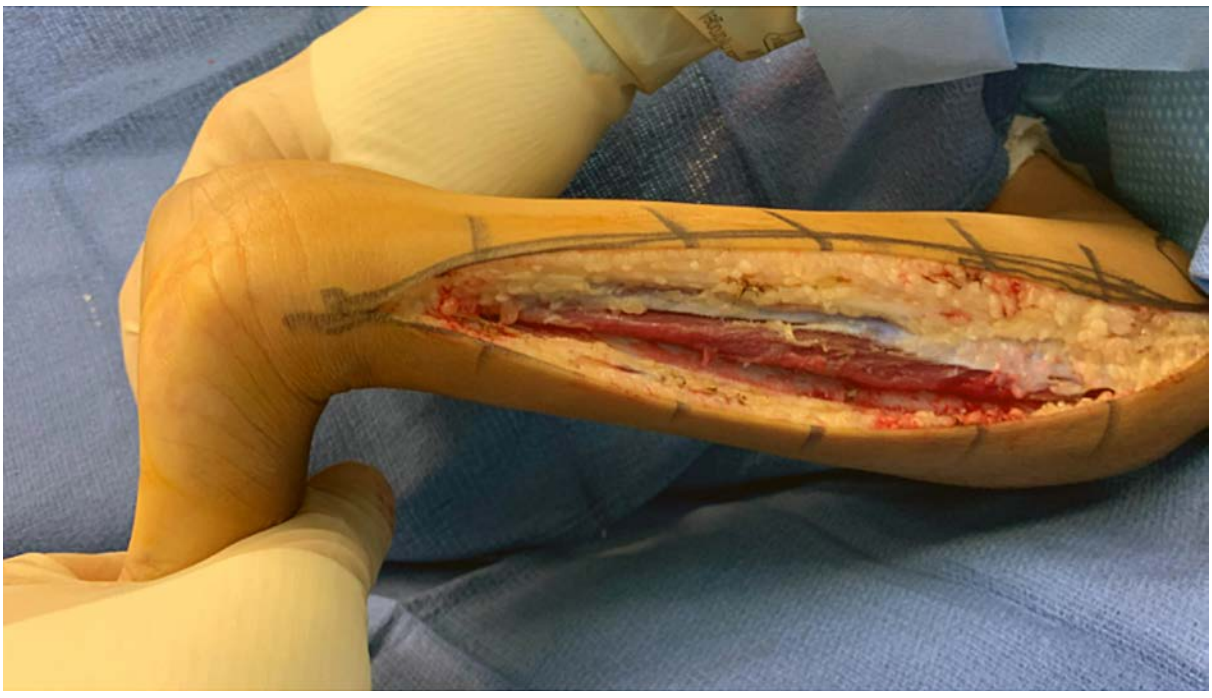

Figure 2. Clinical photograph intraoperatively demonstrating full passive extension of wrist and fingers after flexor origin slide procedure.

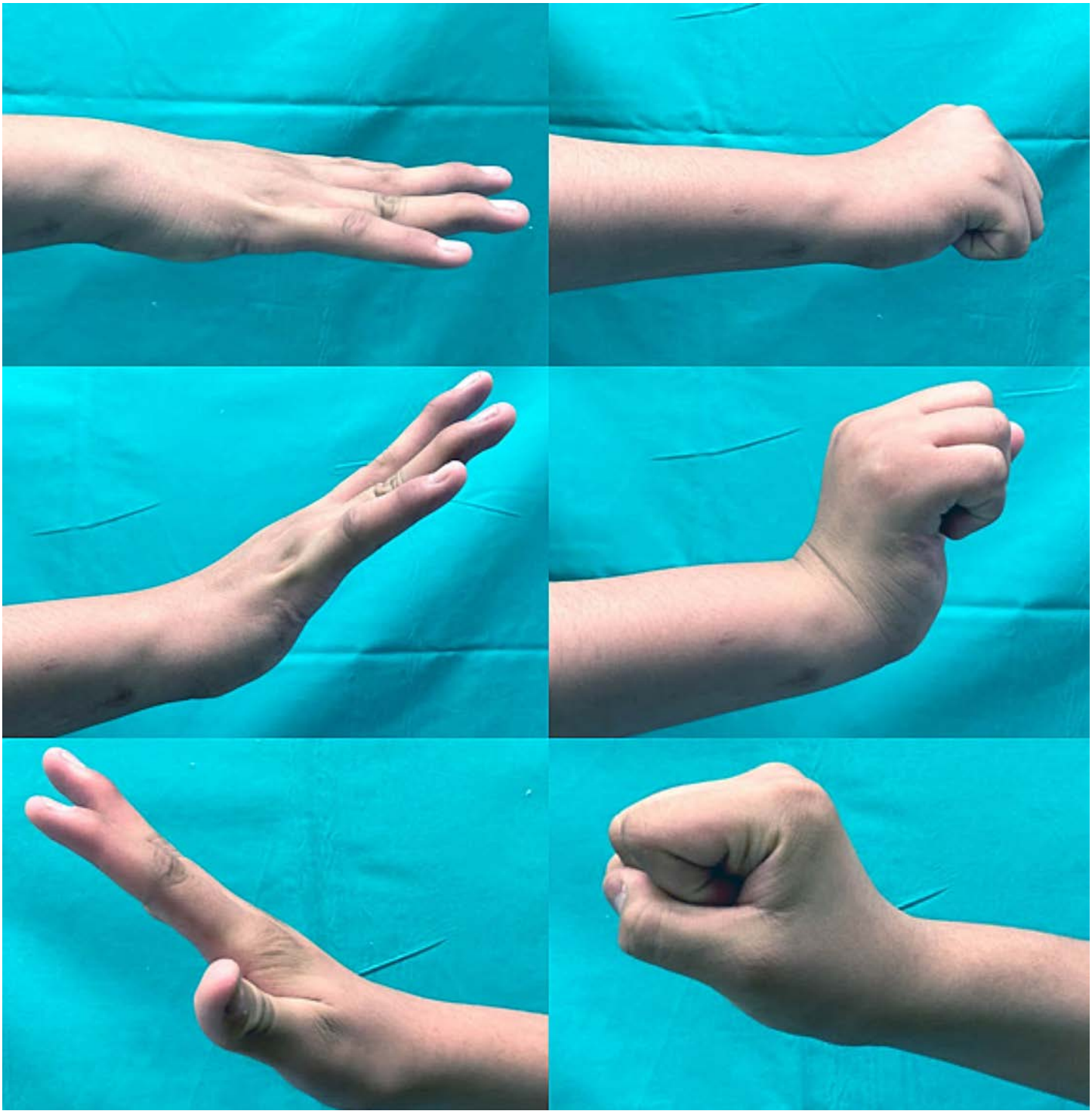

Figure 3. Clinical photos 1 year postoperatively demonstrating full active ROM.

Discussion

We present a patient with a rare genetic disorder, a microduplication of chromosome 16p13.11, with clinical manifestations not previously reported. Specifically, our patient was found to have contractures of his hands and fingers, with the right side being more involved than the left, which led to significant functional impairment. Recent literature have highlighted clinical findings including schizophrenia, intellectual disability, and urinary tract problems in this patient population, but have not reported on specific musculoskeletal manifestations[2,3]. To our knowledge, successful surgical treatment of a patient with musculoskeletal pathology due to a chromosome 16p13.11 microduplication has yet to be published.

Hand and finger contractures in children are often a complication of compartment syndrome, referred to as

Volkmann's contracture. The etiology is a reduction in tissue perfusion caused by increased intracompartmental pressures, leading to muscle ischemia and eventual necrosis. The volar forearm muscles (which include the wrist and finger flexors) contract, leading to dysfunction of the hand[6]. While nonsurgical treatment options such as stretching and bracing play an important role, surgical intervention is often required for meaningful recovery.

First described by Page in 1923, the flexor origin slide is a surgical procedure in which the extrinsic flexor and pronator muscle origins are subperiosteally released from the forearm and allowed to heal in a more distal origin to permit increased finger and wrist extension[5]. It has been shown to be effective in both congenital and ischemic contractures[7]. Geissler et al also reported good results for 3 patients with entrapment and adhesions of the flexor tendons after a forearm fracture, or so-called Pseudo-Volkmann contracture[1].

The relationship between chromosome 16p13.11 microduplication and the development of the contractures remains unclear. Our patient was unique in that the onset of his hand contractures did not correlate with a traumatic injury and were not present at birth. It is difficult to draw conclusions on the etiology because of the rarity of this syndrome and lack of other reports in the literature due to the wide phenotypic variations seen. Further research and case series may illuminate this relationship.

In summary, we present a 9-year-old male with bilateral hand and finger contractures found to have a chromosome 16p13.11 microduplication. He underwent a flexor origin slide with local soft tissue arrangement of his right hand with excellent results. To our knowledge, this is the first report of this specific pathology in this population, and the first documentation of successful treatment.

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