Surgical treatment results of hand deformities in patients with Apert syndrome

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ABSTRACT

Objectives: Hand deformities in Apert syndrome are complex pathologies and usually consist of complex syndactyly with distal bony fusion in the 2nd, 3rd, and 4th fingers, simple syndactyly in the 5th finger, and short thumb, radial clinodactyly and synphalangism. Other accompanying deformities and the presence of craniosynostosis make the treatment plan even more complex. Because it is such a rare disorder, no standard treatment protocol has yet been developed.

Methods: In this study, our aim was to evaluate the early results of our standard treatment protocol for Apert’s hands. We evaluated 7 patients with Apert’s hands who were treated bilaterally between the years of 2008 and 2013. We performed a two-stage surgical protocol, the first of which was the bilateral opening of the border fingers and the second stage was the opening of the middle fingers and web space deepening, osteotomy and collateral release of the thumb. Patients were evaluated according to grasping and pinching ability, graft-flap necrosis and cosmetic satisfaction.

Results: The mean age at the first operation was 2.7 years and the mean number of operations was 3 per patient. No patient developed graft-flap necrosis and no patients required amputations. All patients were able to perform grasping and pinching functions and families were satisfied with the cosmetic results.

Conclusion: Using a two-stage surgical protocol, achieving satisfactory results with a minimal number of operations is possible in patients with Apert Syndrome.

Key words: Apert syndrome, hand deformity, syndactyly

Introduction

Hand deformities in Apert syndrome are among the most complex pathologies of the upper extremity congenital pathologies. Complex syndactyly involving distal phalangeal levels affecting the 2nd, 3rd and 4th fingers is present. In addition to the distal zone, there is proximal fusion in the 4th and 5th metacarpals. These deformities result in diffuse joint stiffness after some time [1]. The first digit is commonly included in syndactyly and there is an additional clinodactyly at the metacarpophalangeal (MCP) joint level.

Initially, most cases of Apert syndrome are generally referred to major craniofacial clinics. These patients are only referred back to hand surgery departments after their craniofacial treatments are completed. Therefore, most patients are treated after 2 years of age for hand deformities. Although no standard approach presently exists for the treatment of Apert’s hands, most studies have focused on decreasing the number of surgeries with the best acceptable cosmetic and functional results [2-4].

In this study, our aim was to evaluate the early results of a two-stage procedure for treating Apert’s hands.
Patients and Methods
In this study, we evaluated 7 Apert syndrome patients (4 males, 3 females) with bilateral hand deformities between the years of 2008 and 2013. The mean age at the first operation was 2.7 and the mean follow-up time was 2.7 years (min, 1.2; max, 5.4). The mean age at final follow-up was 4.2 years.

Five of the patients treated were classified as having Upton Type 3 deformities, while 2 patients were classified as having Upton Type 2 deformities. According to the Upton classification system, Type 1 (spade hand) patients have a shallow first web space and simple syndactyly in their fourth and fifth digits. Type 2 (spoon hand) patients have simple syndactyly in their first and second digits and prominent concavity in the palm. Type 3 (rosebud hand) patients have complex syndactyly with distal synostosis in their first and second digits.

In this study group all patients were treated with a standard two-stage procedure; a bilateral correction of the second and fourth web spaces, followed by a third web space correction and deepening of the first web space, osteotomy and correction. Patients were evaluated based on a number of factors, including grasping and pinching ability, graft-flap necrosis and cosmetic satisfaction.

Surgical Technique
Under general anesthesia, patients were placed in the supine position. Both upper extremities and donor areas in the groin region were prepared. Dorsal rectangular flaps for 2 and 4 web spaces were created (Figure 1). A zig-zag incision was created on the palmar surface (Figure 2). After dissecting the neurovascular bundle, the web base level was determined. All soft tissue and bony connections were separated up to this level. Syndactyly zones were separated at the level of distal phalanx. Rectangular and zig-zag flaps were paired and partial skin closure was performed. For bare areas on the skin, full-thickness skin grafts were harvested. After donor area closure the bare areas were closed by skin grafts using absorbable sutures (Pegelak Rapid, Doğsan, Turkey) (Figures 3 and 4).

After 6 months to 1 year of initial surgery all patients were prepared for a second surgery. In the sec-
ond surgery, first web space deepening and osteotomy, together with third web space separation, was planned. All surgeries were intended to achieve four- or five-fingered hands. Radial, ulnar or both-sided incisions were used for proximal phalanx osteotomy and reconstruction. An open or closed wedge osteotomy was performed using a blade. After completing the osteotomy, a 0.8 mm k-wire was used to fix the osteotomy site. After subsequent narrowing and widening of the ulnar and radial collateral ligaments, k-wires were left in place subcutaneously (Figures 5 and 6).

A postoperative soft cast was applied for 3 weeks. After 3 weeks, all casts were removed and the patient was provided with skin care instructions. K-wires were removed postoperatively within 4 to 6 weeks.

Results
With additional interventions the mean number of operations per patient was 3 (min, 2; max, 4). While two of the planned, staged surgeries were major, one involved minor surgery, such as scar revision, implant removal, contracture release or a Z-plasty.

No amputations were performed in the study group and no patient suffered from flap or graft necrosis. All patients were able to grasp after the first web space was deepened. No complications were encountered at the osteotomy zone.

All families were cosmetically satisfied with the final appearance of the hands (Figure 7).

Discussion
Apert syndrome was first described by Eugene Apert [5]. While cranial and hand deformities are dominant components of the syndrome, other facial deformities can accompany the condition. Apert syndrome occurs at a rate of 1 in 65,000 live births [6-8]. The syndrome is reported to effect both genders equally and is related to a high paternal age [8,9]. While the syndrome is reported to be inherited as an autosomal
dominant trait, most cases are related to two spontaneous mutations in the FGFR-2 gene in the 10th chromosome.

Treatment is difficult and requires a multi-disciplinary approach due to the complex craniofacial and hand deformities associated with the syndrome. Common hand deformities are operated upon at approximately two years of age because patients are primarily treated for craniofacial deformities shortly after birth.

The main treatment strategy in Apert syndrome is to obtain an optimal result with a minimum number of operations. Because of the surgeries required to repair the maxillofacial deformities and the risks resulting from airway and pulmonary problems due to the syndrome itself, a functionally and cosmetically acceptable hand should be achieved with a minimum number of operations. These risks are more prominent in Upton stage 3 patients.

All of the patients in this study group had two-stage surgery. Although during the first operation a first and fourth web space release is commonly recommended, leaving the 2., 3. and 4th fingers attached requires radial and ulnar web space release of the 3rd finger during the second operation. This may impose vascular risks for the 3rd finger [3]. For this reason, special attention was given in our practice to release the 3rd finger radial web space during the initial procedure. This practice is very important to obtain a 5-fingered hand.

With the presence of complex synonychia and synostosis (Upton Type 3), release of some digits may be problematic because of the common vascular supply, which can create circulatory risks. For such patients a four-fingered hand with an acceptable cosmetic and good functional outcome can be the ultimate aim of treatment [4]. A four-fingered hand can successfully achieve grasping and pinching. For hands with relatively simple deformities (Upton Types 1 and 2), this surgical strategy can achieve a 5-fingered functional hand.

In this study, the main goal was to achieve a functional hand with deep web spaces having a long and radially deviated thumb. There are studies suggesting external fixator correction or lengthening for thumb deformities [10]. However, for clinodactily treatment, open wedge osteotomies were preferred to gain a length of thumb in this study. Additionally, radial collateral release and ulnar collateral plication is very important to achieve a pinching hand that can oppose the second finger. Despite all interventions regarding the thumb and 1st web space, restricted thumb movements are inevitable.

In this study, group priority was given to craniofacial deformities. Although some authors recommend operating on hand deformities simultaneously with cranial surgery at approximately 2 months of age, we strictly avoid this approach because of possible upper respiratory and pulmonary malformations.

Studies showed many advantages of operating on patients at early ages [11]. According to Upton, late surgery can impose growth disorders and an impaired body image [10]. Holten et al. reported that an earlier recovery of hand movements can protect hand functions [1]. From our perspective, we also think that these basic principles are important for improved results; however, none of the patients in this study were operated on before cranial treatments. Families also gave priority to craniofacial deformity treatment. For these reasons, patients were operated on between 2 and 3 years of age. Despite this relatively late correc-
tion, we observed enough range of motion for grasping and pinching. Even though increased contracture risk is present with advanced age, we believe that this does not affect reasonable functional result expectations.

In this study, all patients were operated on bilaterally. Although a bilateral approach is important to reduce the number of operations, this approach increases the duration of the operation. For this reason finger separation, graft extraction and graft application should be performed simultaneously by multiple surgeons consecutively.

As a result, there is no standard treatment protocol for Apert syndrome, especially in Upton stage 3 patients in which hand surgery can be performed after craniofacial procedures. With a two-stage surgical protocol, achieving a functionally and cosmetically acceptable hand with a minimum number of operations is possible.

**Conflict of interest statement**

The authors have no conflicts of interest to declare.

**References**


