CHAPTER **28**

Apert Syndrome: Literature Review and Case Study

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INTRODUCTION

Apert Syndrome is a rare condition involving craniosyntosis and multifaceted abnormalities of the hands and feet. The term acrocephalosyndactyly type 1 has also been used to describe this disorder. It distinguishes itself from other craniosytosis disorders such as Crouzon Syndrome and Pfeiffer Syndrome, by elongation of the skull occurring always with syndactyly of the hands and feet (1, 2). This condition is estimated to occur in 1 of every 64,500 births (3) and is linked to 2 mutations in fibroblast growth factor receptor 2 (3). The occurrence is equally distributed between males and females, and most cases are due to a sporadic mutation, although there does exist a link to advancing paternal age (4).

In addition to syndactyly, the foot of a patient with Apert Syndrome often demonstrates polydactyly, progressive joint fusions, first ray pathology, nail deformities, and soft tissue changes. Cohen and Kreiborg classified the foot type into 3 categories based on the degree of syndactylyzation. In type 1, syndactyly occurs only between toes 2 through 4; in type 2 it occurs between toes 2 through 5; and in type 3 all of the toes are involved (6). Although historically the Apert foot was thought to be bilateral and symmetrical (1, 7), several cases have been published in the literature with asymmetrical deformities (8), which would make this type of classification more complex.

Polydacytly of the feet in Apert syndrome is thought to be widely under-reported due to the fact that little research has been done evaluating the foot radiographically and the clinical picture may be misleading. The first metatarsal is typically bifid proximally but often there is only 1 metatarsal head (9,10). There have however, been reports of completely separate and additional first metatarsals (10).

A constant feature of Apert syndrome is progressive joint fusions throughout the foot. Although the existing literature does not demonstrate a consistent pattern and/or rate of fusion, it is clear that progressive syntosis of the foot is inevitable as the patient matures (7, 11). Anderson et al described the fusions as first involving the tarsal bones, then progressing to the metatarsal shafts during childhood. Finally, fusions between all interphalangeal joints of the toes will eventually occur (12). As the foot becomes progressively fused, it becomes very rigid, therefore reducing the shock-absorbing capacity and ability to adapt to different surfaces. Additionally, the first metatarsal consistently shortens and moves into a varus position (7, 12, 13). The combination of these 2 deformities leads to first ray insufficiency and an increase in sheer forces and transfer of weight to the second and third metatarsals where callous formation and risk of stress fractures occur (11-13).

The nail plate is also affected due to the syndacytly of the toes and deformity of the foot. Mah et al, consistently found a vertically-oriented, onychogryphotic, brittle first toenail, and a plantar curl to the lesser toe nails (7). In toes that are completely syndactylized, a cleft may or may not exist between the nails, and in cases of type 3 Apert feet, the nail can be one single broad nail plate (7, 9, 14). Due to changes in the nail plate, osseous deformities, and irritation of shoe gear, cases of onychocryptosis and even nail sepsis have been reported (7, 14).

The treatment of the foot in in a person with Apert Syndrome varies greatly, and is most consistent with the geographic location that the patient is being treated. While surgery is common in surgical specialty centers, most patients will receive only palliative care for painful callouses. It appears the only benefit in operating on the foot of a person with Apert syndrome is to assist with proper shoe fitting and give psychological effect. Most patients are able to ambulate with custom molded orthopedic shoes and experience a similar quality of life (12, 15).

Until recently, the main research and focus of treatment has been primarily devoted to the management of craniosyntosis and hand abnormalities because these play a critical role in the development and functionality of the maturing child. With improvement in overall care, the life expectancy and quality of life of a patient with Apert Syndrome has dramatically improved, which has led to a need for a standard of care in the lower extremity. This is particularly important because the condition is known to progress with age and can lead to substantial pain and debilitating complications.

CASE REPORT

We examined a 59-year-old man with Apert Syndrome who had been followed by this podiatry clinic for several years. The patient was obese, had congestive heart failure, and was prediabetic but was still able to ambulate without an assistive device.

On clinical examination, there was syndactylization of all of the toes of the feet bilaterally (Figures 1 and 2). The nail plate was coalesced between toes but there was a cleft in the nail plate. The most medial nail was thickened bilaterally, and there was a plantar curl to the nails of the lesser toes. The patient had hyperkeratotic lesions on the distal aspect of the hallux bilaterally, on the plantar aspect of the foot in the area of the third metatarsal head bilaterally, and on the lateral midfoot area of the right foot (Figures 3 and 4). A soft nodule approximately 2 centimeters in diameter was located on the plantar aspect of the right foot just proximal to the second through fourth metatarsal heads (Figure 4). This nodule, present since birth has remained unchanged. The patient denied any pain or discomfort from the nodule. Telangiectasia were present on the medial aspect of the ankle bilaterally. The range of motion to all joints of the foot and ankle were significantly limited, and muscle strength was intact for all major muscle groups of the lower extremity.

Radiographic examination revealed significant osseous fusions throughout the foot and bilateral polydactyly (Figures 5 and 6). A duplication of the first metatarsal and adjacent phalanges was present. In the forefoot, osseous fusion and bony bridging was present in all the interphalangeal joints and between the proximal phalanx of the first toe and metatarsal head of the second toe. The metatarsal shafts were fused between the 2 first metatarsals distally, and between the third and fourth metatarsals proximally. The bases of all the metatarsals were fused with the exception of the more medial first metatarsal. The first metatarsal cuneiform joint was not fused and laterally deviated. The first metatarsal is in a slightly varus position. In the midfoot and rearfoot nearly every joint is fused.

The patient had been treated with custom molded extra depth, extra width diabetic shoes for the foot deformities. Urea 40% cream and regular podiatry visits with sharp debridement of the callouses and nails had been performed. The patient was able to ambulate with a nonantalgic gait and denied any pain in his feet and lower extremity. Although the patient is mildly active, he is able to perform all of his activities of daily life without limitation.

DISCUSSION

This case demonstrates that with advancements in treatment, patients with Apert Syndrome now have a much longer life expectancy, which leads to a need for a focus on the feet. Although the craniosytosis aspect of the disease can be life threatening, once the patient reaches skeletal maturity there is little that will change in that respect. Conversely, throughout development and well past skeletal maturity, the deformities of the foot are progressive and require regular care.

This case would be classified, as a type 3 foot as there is syndactyly of all the toes (6). Additionally, it is one of only a few reported cases of additional first metatarsal bones (9, 10). Otherwise, the foot type appears to be consistent with

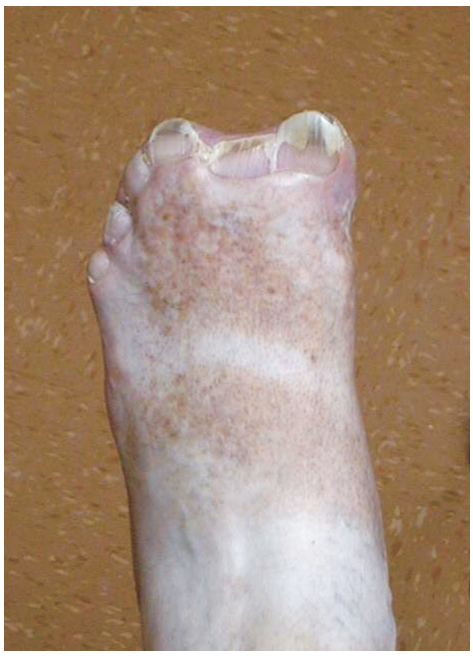

Figure 1. Clinical appearance of a patient with Apert's syndrome.

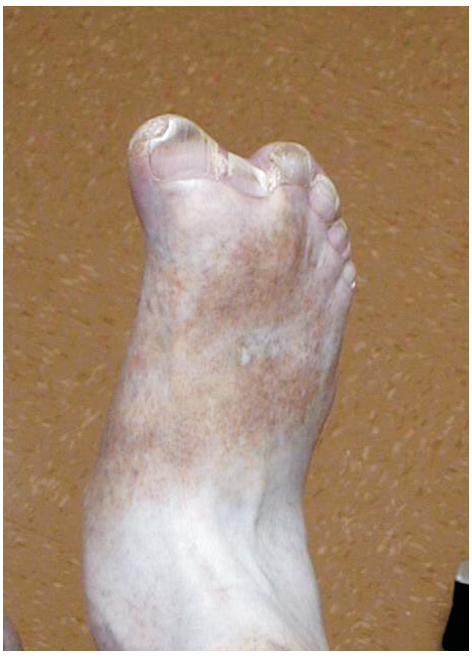

Figure 2. Note the coalesced nail plate.

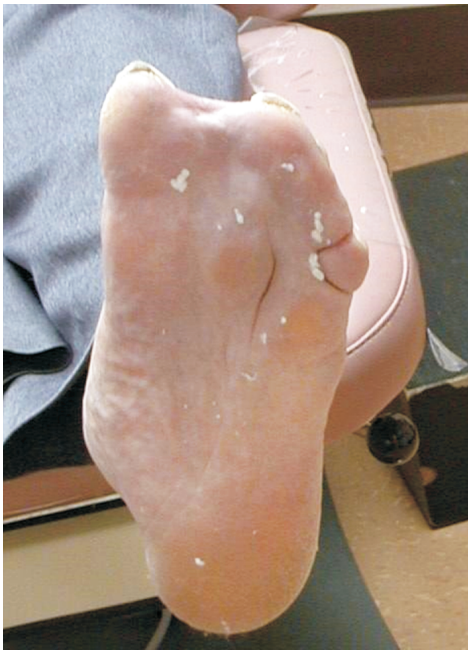

Figure 3. Plantar view.

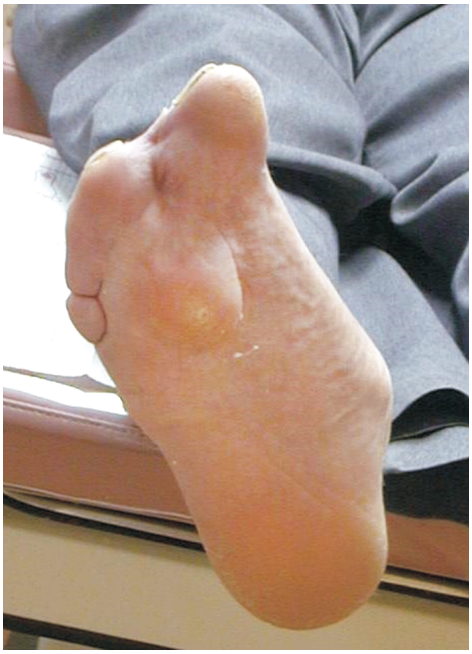

Figure 4. Note the asymptomatic soft nodule.

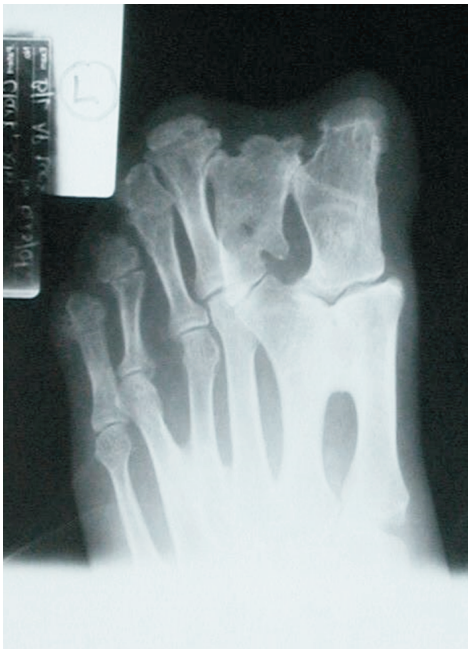

Figure 5. Radiographic appearance.

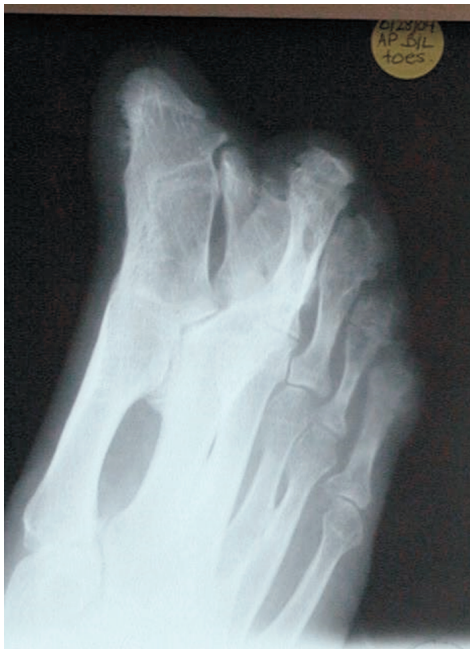

Figure 6. Note the significant osseous fusions.

the typical Apert feet in that there is a varus alignment of the hallux, progressive joint fusions throughout the foot, and nail and soft tissue changes (7, 12-14). Transfer callouses due to altered weightbearing from a shortened first ray and an increase in shear forces due to a rigid foot type were demonstrated.

In this case, radiologic examination was crucial in determining the extent of deformity of the foot. Because the progressive nature of the foot deformity is well documented (7-11), we can assume that the high degree of syntosis throughout the foot was due to the advancing age of this patient. Also, the additional first metatarsal could only be determined radiographically.

The treatment of the feet in this patient appears to be successful in that he was able to perform all activities of daily life and was pain free. This result was achieved through regular debridement of callouses, the use of Urea cream, custom molded diabetic shoes, and regular nail care. This prevented the callous formation from becoming too thick and painful, reduced the friction forces on the foot, and prevented onychocryptosis or the potential for nail sepsis. Although surgical options are often used in patients with Apert syndrome, we do not deem it necessary in this case as a conservative approach has worked well. A potential benefit to surgery in this case would be limited to better shoe fitting and the psychological benefit of improving the cosmetic appearance of the foot (15), however the patient would also be exposed to the risk of surgical complications.

Patients with Apert syndrome experience many unique deformities in their feet that are progressive as the patient ages. Podiatric care of these deformities is critical to proper management of the patient's health especially as care of other aspects of the disease have lead to an increase in life expectancy. A conservative approach has worked well in this case and should be attempted in the podiatric management of most patients with Apert Syndrome.

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