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Revisiting Radiographic Characteristic of the Hands in Apert Syndrome: A Literature Review with Cases

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ABSTRACT

Introduction: Syndactyly of the hand in Apert syndrome involves all components of the hands. In 1991, Joseph Upton introduced a comprehensive classification of the Apert hands based on its severity. Plain x-ray is the most commonly used to evaluate bony abnormality.

Case Presentation: We present two cases with classic characteristics of Apert syndrome with demonstrated different types of Apert hands; type II and type III. Radiographs exhibited bony deformities with typical misalignment and multiple osseous formations in several parts of phalanges.

Discussion: Apert hands present complex syndactylies involving the index, middle, and ring fingers in different stages regarding Upton's classification system. Almost all Apert hands also lack the active motion of the interphalangeal joints. The ossification allowed the synostosis and symphalangism to become evident on radiographs. The abnormality of the epiphysial growth causes short and deviated fingers.

Conclusion: Apert hands demonstrate complex deformity involving bony structures that developed prenatally and postnatally. Typical deformities including complex syndactyly, clinodactyly, and symphalangism.

KEYWORDS: Apert syndrome, hand deformity, radiographic appearance, syndactyly, clinodactyly, symphalangism

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INTRODUCTION

Apert syndrome, a rare and complex congenital disorder, stands as a paradigm of the intricate relationship between genetics, embryology, and clinical presentation. The syndrome, often referred to as acrocephalosyndactyly type 1, is characterized by a constellation of craniofacial and extremity anomalies, prominently featuring craniosynostosis and syndactyly. First described by Eugène Apert in 1906, the syndrome's hallmark phenotypic features are well-documented, yet the radiographic intricacies of its hand manifestations remain an area of compelling research.

The hands, as a prime example of the intricate orchestration of genetic information, cellular processes, and external influences, provide a unique window into the underlying pathophysiology of Apert syndrome.[1] Radiography, as a critical diagnostic tool, has undergone its own evolution over the years. From early film-based techniques to modern digital imaging modalities, radiographs have enabled us to peer beneath the surface and gain insights into the internal structures of the human body. In the context of Apert syndrome, plain radiographs are the most commonly utilized imaging tool to evaluate abnormal anatomy of the Apert hands.[2] Radiographs offer a non-invasive means to capture the nuances of skeletal development, fusion anomalies, and growth patterns that characterize the hands of affected individuals. Our case report delves into the radiographic nuances of the hands in Apert syndrome, aiming to unearth the hidden complexities that underscore the visually apparent syndactyly and bone deformities. By elucidating the radiographic characteristics, we strive to not only contribute to the understanding of this rare syndrome but also pave the way for improved clinical management and enhanced quality of life for affected individuals.

While considerable literature has been devoted to the clinical and genetic aspects of Apert syndrome, the radiographic characteristics of its hand manifestations remain relatively less explored. Hereby, we present two cases of Apert syndrome by highlighting the radiographic feature of the hands, with objectives to (1) systematically document and describe the radiographic abnormalities in the hands of individuals with Apert syndrome, including syndactyly, bone deformities, and growth plate anomalies, (2) correlate the observed radiographic features with the underlying genetic mutations and molecular pathways implicated in Apert syndrome, thereby unraveling the complex interplay between genetic determinants and skeletal development, (3) highlight the clinical significance of early radiographic assessment in guiding treatment strategies, surgical planning, and postoperative monitoring for individuals with Apert syndrome, (4) enrich the existing body of knowledge on Apert syndrome by providing a comprehensive radiographic perspective, fostering a holistic understanding of the syndrome's multi-systemic manifestations.

In the subsequent sections of this case report, we will delve into the methodology employed for radiographic analysis, present our findings, and discuss their implications in the broader context of clinical management and research. Through this endeavor, we aspire to contribute to the multidisciplinary efforts aimed at unraveling the complexities of Apert syndrome and advancing the care and well-being of those affected by it.

CASE ILLUSTRATION

We presented our first case, a 2 years old girl with a typical characteristic of Apert Syndrome **(Figure 1)**. She is the first child of a non-consanguineous marriage. The mother had a normal delivery with no history of trauma, infection, smoking, and drug use during pregnancy. There was no history of abnormal physical finding or syndromic disorder in both parents and the third decade of life.

Her hands demonstrated symmetrical fusions of the first to fifth digits in both hands with conjoined nails (synonychia) appearance. The first digit was free with sufficient webspace. The palms formed a concave shape similar to a spoon-like appearance **(Figure 2)**. Plain x-ray revealed complete bony fusion at the distal of the 2nd to the 4th digits with a suggestive osseous union between 3-4 proximal interphalangeal joint of both hands. Metacarpal synostosis of the 3-4 digits was observed in both hands, and at 2-3 digits on the left hand. Radial subluxation of the distal of the 1 digit was seen in both hands too **(Figure 3)**.

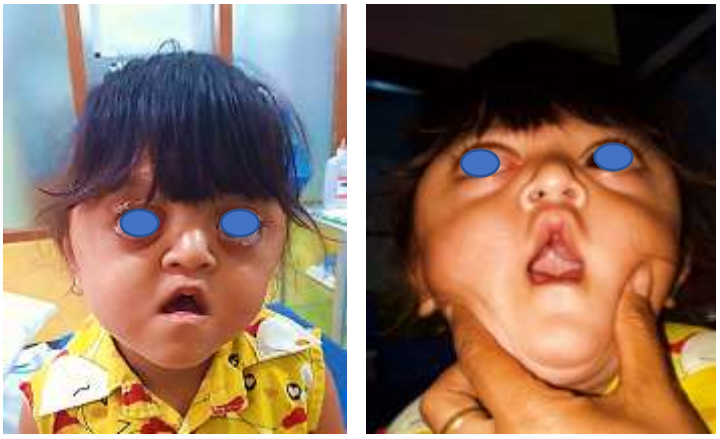

Figure 1. Typical facial disfigurement of Apert Syndrome (left); high-arched palate with pseudopalatoschizis (right)

Figure 2. Spoon-like shape (type II) syndactyly of both hands. Note the concave palms shown by arrows.

Figure 3 X-ray exhibited symmetrical complex syndactyly with symphalangism at proximal and distal interphalangeal joint, and clinodactvlv of thumb

Our second case, a 4 year old girl presented with characteristics of Apert syndrome **(Figure 4)**. She is the first child in the family. There was no history of consanguinity and was delivered normally. There was no history of trauma, infection, drug use, or exposure at the time of pregnancy. There was no history of physical abnormal finding or syndromic disorder in the family. Her parents are well-educated and come from a good economic status. There was no other apparent congenital malformations detected.

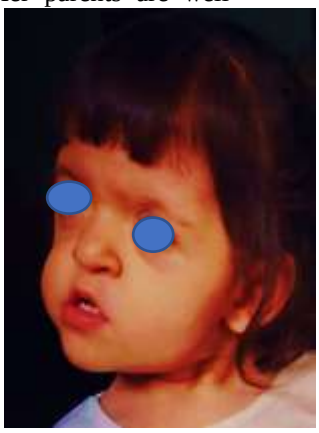

Figure 4 Facial disfigurement with prominent midface hyploplasia with class III malocclusion.

She had severe bilateral syndactyly of both hands and feet. Her hands demonstrated complete fusion and overlapping of all digits (pansyndactyly) which made appearance of hoof-like or rosebud-like hands. Conjoined nails of the 2-3-4-5 digits were seen in both hands. **(Figure 5)**. X-ray of hands revealed only four digits in both hands. Both hands exhibited bony fusion only at the distal portion of all digits. Severe clinodactyly with incomplete and short metacarpal bones were seen **(Figure 6)**.

This type is characterized by complex pansyndactyly (a tight osseous or cartilaginous union among all 5 fingers), synonychia (broad conjoined nails), rotation, and overlapping of fingers, as well as deficiency of the palmar field. Individual longitudinal ridges demarcating partial separation of the underlying distal phalangeal segments may be present, and the nail of the fifth finger may be marked by a longitudinal ridge or may be separated. In many occasions, the thumb is deficient. If the fifth finger exist, it is mostly spared from the

osseous union but joined by complete, simple syndactyly. Synostosis of the fourth and fifth metacarpal is common, as often as a complex syndactyly with distal synostosis between the thumb and the index finger. Such a complex anatomical structure creates an appearance of a

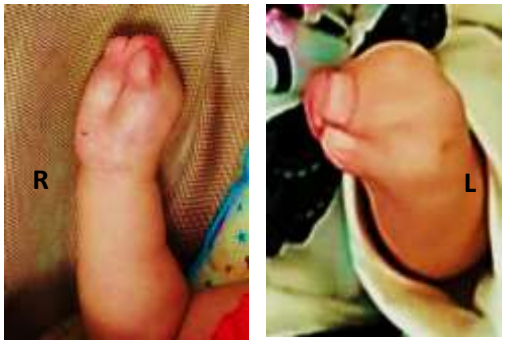

Figure 5 Rosebud-like shape (type III) syndactyly of both hands

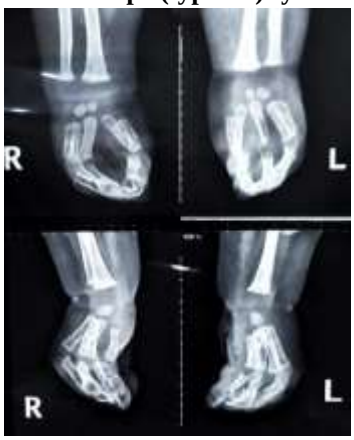

Figure 6. X-ray of hands

DISCUSSION

Children with Apert syndrome usually suffer from the limitation of hand function due to severe syndactyly, which generally affects the upper limb more severely than the lower limb.[3] Syndactyly of the hand in Apert syndrome is a challenging problem since it involves all components of the hands including skin, bones, nerves, tendons, and vessels. These malformations developed prenatally and progressed further after birth, creating more severe deformities.[4]

The metacarpal bones of the Apert hands are particularly shorter than ones of a normal hand and all five bones are more similar in their lengths. Another clinical finding is the fusion of the bases of the fourth and fifth metacarpal bones, more commonly evident after 5 years of age, but sometimes it occurs during the first year of life.[5]

Almost all Apert hands suffer from symphalangism, which literally means: "joined bones" and refers to an ankylosis (bony union) of the interphalangeal joints or the fusion of adjacent phalanges. It is a prominent radiographic feature observed in the hands of individuals with Apert syndrome. This phenomenon results from disrupted endochondral ossification and complete cartilaginous continuity across the digital joints[5] as a result of aberrant Fibroblast Growth Factor Receptor 2 (FGFR2) signaling pathways. The fusion of digital rays contributes to the characteristic syndactyly, while the additional fusion of individual phalanges accentuates the distinctive appearance of the hand. More bone fusion becomes evident with growth. The ossification occurring with growth allowed the symphalangism and synostosis to become evident on radiographs. Clinicians usually observe symphalangism in the middle finger and developed to the adjacent fingers. However, there is still uncertainty if this happens due to spontaneous fusion or is present at birth.[6]

The thumb rays of the Apert hand are particularly short and radially deviated. [6,7] Clinodactyly, which literally means: "deviated finger", is a lateral deviation of a finger frequently caused by an abnormal middle finger (triangular or trapezoidal shape) and presents a C-shape physeal plate or longitudinally bracketed diaphysis.[7] Epiphyseal growth, on the other hand, yields to pressure, and this behavior is the principal mechanism in the plastic response of bone. Pressures, as applied to the epiphysis, may come parallel to the direction of epiphyseal growth or those at right angles to this direction. Bone can only grow to deformed shapes if pressure is applied more or less continuously during the period of growth.[8]

The Upton classification system, initially devised for simplifying the assessment of syndactyly in Apert syndrome, serves as a valuable tool for surgical planning.[1] This classification categorizes the extent of syndactyly into three types, based on the number of fused digits and the presence

of metacarpal synostosis. Radiographic evaluation aids in precisely determining the type of syndactyly, enabling surgeons to tailor their approach accordingly.

Upton's type I (*spade hand*) Apert hands show the least severe hand configuration, consists of a short, broad, free but radially deviated thumb, a syndactyly of index, long, and ring (2-4) fingers which forms a mid-hand mass through a flat side-to-side union, and shallow web spaces separate the mid-hand mass with the thumb and the fifth finger. This mid-hand 3-fingers union is either bony or cartilaginous and primarily exists at the metaphyseal level of the distal phalanx and the distal interphalangeal joint (the cartilaginous union commonly occurs between the index and middle fingers). The fifth finger is partially or sometimes almost completely joined to the central mid-hand mass by soft tissue. Syndactyly in the fifth finger web space, if any, is always simple. The transverse metacarpal arch is normal. The mid-hand mass creates an appearance of a spade-like hand. [1-3] (see **Figure 8**)

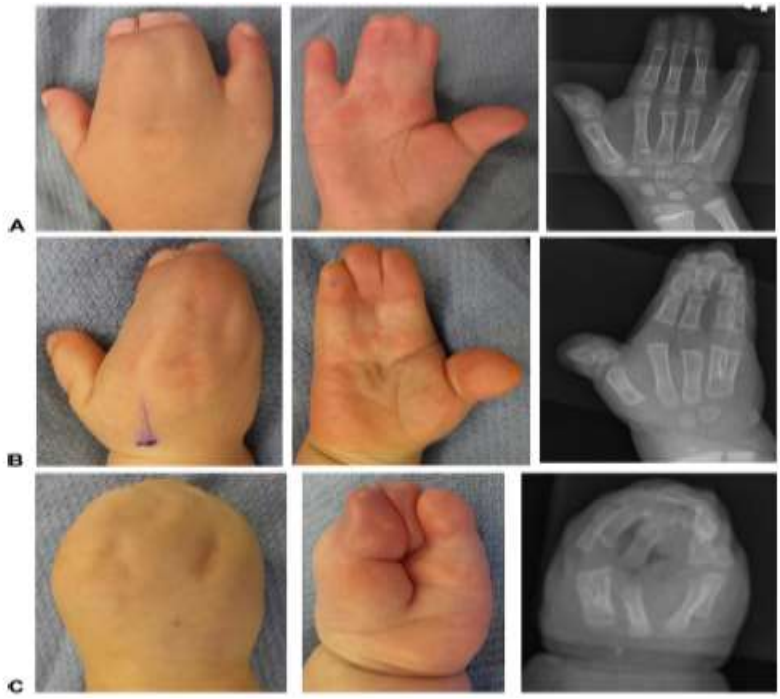

Figure 7 Upton's classfification of syndactyly in Apert hands. 7A, type I (spade hand). 7B, type II (spoon hand). 7C, type III (rosebud hand)

Figure 8. Spade-like hand

Upton's type II (*spoon hand*) Apert hands involves a more severe configuration with the thumb demonstrating a simple complete or incomplete/partial syndactyly of the first webspace without an osseus union between the thumb and the index finger, a fusion involves index, long, and ring (2-4) fingers, and a usually complete, simple syndactyly in the 4th interdigital space. Most Upton type II Apert hands do however contain an adequate first web space. The hand has a large, concave palm as a result of complex finger fusion at the distal phalangeal level, hence creating an appearance of a spoon-like hand. [1-3,] (see **Figure 9**)

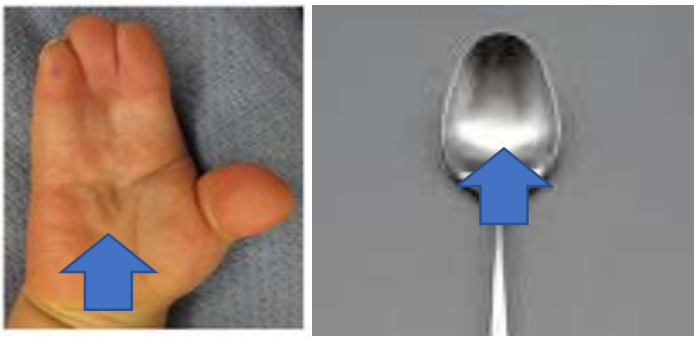

Figure 9. Spoon-like hand. Notice the concave palm.

Upton type III (rosebud hand) represents the severest and the rarest configuration. This type is characterized by complex pansyndactyly (a tight osseous or cartilaginous union among all 5 fingers), synonychia (broad conjoined nails), rotation, and overlapping of fingers, as well as deficiency of the palmar field. Individual longitudinal ridges demarcating partial separation of the underlying distal phalangeal segments may be present, and the nail of the fifth finger may be marked by a longitudinal ridge or may be separated. In many occasions, the thumb is deficient. If the fifth finger exist, it is mostly spared from the osseous union but joined by complete, simple syndactyly. Synostosis of the fourth and fifth metacarpal is common, as often as a complex syndactyly with distal synostosis between the thumb and the index finger. Such a complex anatomical structure creates an appearance of a rosebud-like hand. [1-3,] (see **Figure 10**)

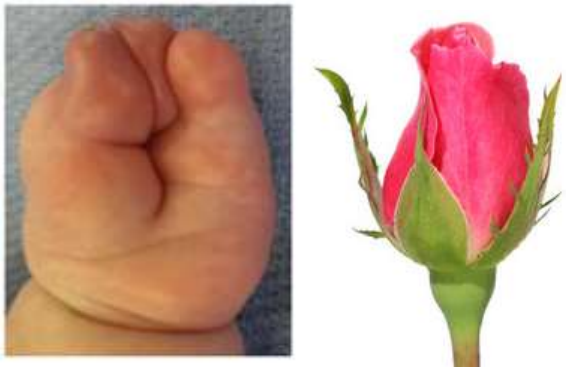

Figure 10. Rosebud-like hand

Our first Apert syndrome patient showed the hand appearance of Upton type 2. We can observe clearly a spoon-like appearance of a concave palm, a sufficient first webspace, and a fusion involves index, long, and ring (2^{nd} to 4^{th}) fingers, and a complete, simple syndactyly in the 4^{th} interdigital space. (see **Figure 2)**. Radiological examination revealed complete bony fusion at the distal of the index, long and ring (2^{nd} to 4^{th}) digits with a suggestive osseous union between the 3^{rd} and the 4^{th} proximal interphalangeal joint of both hands. Metacarpal synostosis of the the 3^{rd} and the 4^{th} digits was observed in both hands, and at the 2^{nd} and the 3^{rd} digits on the left hand. Radial subluxation of the distal of the thumb was detected in both hands as well. (see **Figure 3**).

Our second patient showed the Upton type 3 appearance. The radiographic assessment supporting the Upton classification guides the selection of appropriate surgical techniques. In cases of complete syndactyly, where multiple digits are fused, staged procedures might be required to achieve optimal outcomes. Partial syndactyly cases may necessitate web space deepening or reconstruction of individual digits. Radiographs offer a roadmap for the intricate surgical maneuvers, enhancing the precision and success of procedures. However, while the Upton classification system provides a valuable framework for surgical planning, its application might be limited in cases with atypical presentations or complex hand anomalies. In such scenarios, a nuanced approach that considers both the radiographic and clinical aspects is essential.

Most children with Apert will require surgical reconstruction and physical-occupational therapy to maximize the use of their hands secondary to hand surgeries. The timing of surgical corrections varies with the complexity of the deformity and the web space involved.[2,9] In some cases, delayed release of complex syndactyly of the third web in Apert syndrome patients causes compression on epiphyses, with early epiphyseal closure leading to symphalangism and reduced capitate ossification. The failure of normal distal migration of third metacarpal appeared to occur until the third web is release. But still cannot be concluded if an earlier release of the third web is recommended and further research is still needed. The rationale for commencing surgery earlier included deformity reduction and the type of hand present – e.g. a type III hand underwent surgery at 5 months.[2,6,10] Although there is lacking formal protocol, experts generally agreed that restoration of maximum functional potential and

liberation of all the fingers should be completed before school age (12-24 months of age).[9,10]

What is beyond surgical consideration?

The clinical implications of symphalangism extend beyond its aesthetic aspects. The restriction of joint mobility due to fused phalanges impacts hand function and dexterity, posing challenges in daily activities. This highlights the necessity of a multidisciplinary approach, wherein plastic surgeons collaborate with hand therapists to optimize postoperative outcomes and restore functionality.

While most discussions has mainly centered on surgical implications, it is imperative to acknowledge that the impact of symphalangism extends beyond the surgery room. The psychosocial aspects of hand anomalies cannot be understated, particularly in the context of syndactyly and its visual impact for patients and parents. Addressing psychological well-being of the patients through counseling and support is a vital component of comprehensive care.

Furthermore, collaboration with geneticists is paramount in providing genetic counseling for affected individuals and their families. As the understanding of the genetic basis of Apert syndrome continues to evolve, future research may explore correlations between specific Fibroblast Growth Factor Receptor 2 (FGFR2) mutations and the radiographic manifestations of symphalangism. This could shed light on genotype-phenotype relationships and potentially refine treatment strategies based on underlying genetic variations. Radiographic analysis of the hands itself, as a tangible representation of the genetic anomalies, can aid in explaining inheritance patterns and prognosis of this syndrome.

CONCLUSION

Apert hands demonstrate complex deformities involving bony structure which developed prenatally and postnatally. Clinicians may find typical hand abnormalities as a complex syndactyly involving the index, middle, and ring fingers, a shortened thumb with radial clinodactyly, symphalangism of central fingers, and simple, complete syndactyly of the ring and little fingers. The radiographic assessment of hand deformities in Apert syndrome and its classification according to Upton offers profound insights into the complexities of this rare disorder. The fusion of phalanges not only contributes to the clinical phenotype but also influences surgical planning and patient outcomes. The integration of radiographic findings with surgical expertise and holistic patient care paves the way for optimal outcomes and improved quality of life for individuals with Apert syndrome. As the field of plastic surgery and genetics progresses, our study serves as a stepping stone towards a deeper understanding of the interplay between genetics, radiographic manifestations, and clinical management.

REFERENCES

I. Upton J. Apert syndrome. Classification and pathologic anatomy of limb anomalies. *Clin Plast Surg*. 1991;18(2):321-355.

II. Pettitt DA, Arshad Z, Mishra A, McArthur P. Apert syndrome: A consensus on the management of Apert hands. *Journal of Cranio-Maxillofacial Surgery*. 2017;45(2):223-231. doi:10.1016/j.jcms.2016.11.018

III. Cohen MM, Kreiborg S. Hands and feet in the Apert syndrome. *Am J Med Genet*. 1995;57(1):82-96. doi:10.1002/ajmg.1320570119

IV. Green SM. Pathological anatomy of the hands in Apert's syndrome. *The Journal of Hand Surgery*. 1982;7(5):450-453. doi:10.1016/S0363-5023(82)80038-5

V. Hoover GH, Flatt AE, Weiss MW. The hand and Apert's syndrome. *J Bone Joint Surg Am*. 1970;52(5):878-895.

VI. Kim JH, Rhee SH, Gong HS, Lee HJ, Kwon ST, Baek GH. Characteristic radiographic features of the central ray in Apert syndrome. *J Hand Surg Eur Vol*. 2013;38(3):257-264. doi:10.1177/1753193412462895

VII. Caouette-Laberge L, Laberge C, Egerszegi EP, Stanciu C. Physiolysis for correction of clinodactyly in children. *The Journal of Hand Surgery*. 2002;27(4):659-665. doi:10.1053/jhsu.2002.33702

VIII. Arkin AM, Katz JF. The Effects of Pressure on Epiphyseal Growth: The Mechanism of Plasticity of Growing Bone. *The Journal of Bone & Joint Surgery*. 1956;38(5):1056-1076. doi:10.2106/00004623-195638050-00009

IX. Guero SJ. Algorithm for Treatment of Apert Hand: *Techniques in Hand and Upper Extremity Surgery*. 2005;9(3):126-133. doi:10.1097/01.bth.0000185378.87246.61

X. Theman TA, Upton J, Taghinia AH, Firriolo JM, Nuzzi LC, Labow BI. Central Coalition Osteotomy of Phalangeal Synostoses in the Management of the Type III Apert Hand. *The Journal of Hand Surgery*. 2018;43(11):1042.e1-1042.e8. doi:10.1016/j.jhsa.2018.03.050