

Apert Syndrome: A Case Report and Treatment Protocol

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ABSTRACT

Apert's syndrome (AS), a type 1 acrocephalosyndactyly syndrome, is a rare congenital disorder characterized by craniosynostosis, midface hypoplasia and syndactyly of hands and feet. The rarity of the syndrome and similarity of features with other craniosynostosis syndromes make it a diagnostic dilemma. Because of the multiple alterations in patients with AS, a multidisciplinary approach consisting of dentists, neonatologists, neurosurgeons, plastic surgeons, ophthalmologists, otolaryngologists and geneticists is essential for a successful planning and treatment. A case of Apert's syndrome is presented in a 5 year old girl with an emphasis on multidisciplinary approach to treatment.

KEYWORDS: Apert's syndrome, craniosynostosis, syndactyly

INTRODUCTION

Apert's syndrome (AS) is a rare congenital type I acrocephalosyndactyly disorder characterized by craniosynostosis, syndactyly of the hands and feet combined with dysmorphic facial features.¹ AS has a prevalence rate of 15.5 per million live births and represents 4.5% of all cases of craniosynostosis.² AS was first described in 1894 by Wheaton but the term "Apert syndrome" was given by Dr. Eugene Apert in 1906 for the presentation of a series of nine cases.^{3,4}

AS is an autosomal dominant disorder caused due to mutation of fibroblast growth factor receptor-2 (FGFR-2) mapping to chromosomal bands 10q (10q25-26). Suture progenitor cells with mutated fibroblast growth factor receptors (FGFR2) cannot transduce signals from extracellular fibroblast growth factors (FGFs) and therefore do not produce the necessary fibrous material essential for a normal calvarial suture. In Apert cases, the spheno-occipital and spheno-ethmoidal synchondroses and the fronto-ethmoidal suture fuse early, resulting in a severely shortened posterior and anterior cranial base with a resultant hypoplastic midface.⁵

Apert patients have a characteristic pseudo prognathic appearance due to a reduction in the size of the maxilla, particularly in the antero-posterior direction. This small maxilla results in tooth crowding and an anterior open-bite. The mandible is within normal size and shape. Characteristic intraoral findings include impacted teeth, delayed eruption, ectopic eruption, congenitally missing teeth, supernumerary teeth, and thickened gingiva.^{6,7} Cleft of the soft palate, bifid uvula, byzantine-arch palate, choanal stenosis, and anomalies of the tracheal cartilage are some of the other frequent findings.¹ Mouth

breathing, observed in most cases of Apert's syndrome, is related to alteration in facial growth.⁶ Another key feature of AS include syndactyly or webbing of the fingers and toes resulting in a spoon-like deformity of the hands and feet.¹

CASE REPORT

A 5-year-old girl presented to the Department of Pedodontics and Preventive dentistry complaining of malaligned teeth, difficulty in chewing food, bleeding from gums and facial and limb deformity.

She was the second child of her parents who were nonconsanguineously married. Her mother had a normal delivery without any history of trauma, infection and drug use during the term. No other family members had similar complaints or any other congenital abnormality. Her intelligence quotient was in the range of 75 with slightly incoherent speech.

Examination revealed flattened occiput with frontal prominence, an abnormal contour of the head (brachycephaly), shallow and downward slanting orbits with bilateral proptosis and hypertelorism. She had depressed nasal bridge and a thick nose with a bulbous tip (parrot beak shaped) and cross bow-shaped lips (Figure 1). Limb deformities in the form of syndactyly of the hands and feet are noted (Figure 2a, b, and c).

Intraoral examination revealed normal mouth opening, anterior open bite and a high arched palate associated with lateral swellings of the palatine processes, one on either side of the middle miming a pseudo-cleft in the midline (Figure 3a). The oral hygiene was poor and generalized chronic gingivitis, and gingival enlargement with spontaneous bleeding was present (Figure 3b).

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Figure 1. Extraoral view of the patient showing parrot beak nose, hypertelorism, downward slanting outer canthus of eyes, depressed nasal bridge with cross bow shaped lips.



Figure 2. (a,b and c) Photographs showing syndactyly in hands and syndactyly of feet.



Figure 3a. Intraoral front view showing gingival swelling with spontaneous bleeding. Figure 3b. Intraoral maxillary view showing high arched palatal vault with deep pseudo cleft in midline.

Dental caries on anterior and posterior teeth was present. On investigation, panoramic view radiograph showed high arched palate with anterior open bite (Figure 4).



Figure 4. Panoramic radiograph showing anterior open bite.

Initial dental treatment consisted of oral hygiene instructions, demonstration of tooth brushing technique with the aid of electrically powered toothbrushes along with adjunctive use of chlorhexidine mouth rinse 0.12% two times per day for one month. Patient was recalled monthly for professional oral prophylaxis and preventive treatments in the form of topical fluoride therapy. Restorations were performed on carious teeth. The patient was advised to visit a plastic surgeon for the treatment of her extremities.

DISCUSSION

The two most common of the syndromic craniosynostosis, collectively known as acrocephalo-syndactyly, are Crouzon's and Apert's, which constitute 70% of such cases.⁸ The spectrum of abnormalities in Apert syndrome consists of craniosynostosis, midface hypoplasia, and symmetric syndactyly of the hands and feet. Also, cleft or pseudocleft palate is a frequent finding in AS; whereas these traits are extremely rare in Crouzon syndrome. These features favor the diagnosis of Apert syndrome over Crouzon syndrome in the current case.⁹

Treatment of Apert syndrome involves a multidisciplinary approach and ideally should begin at birth with proper diagnosis. For the correction of craniosynostosis, craniectomy is performed during 6 months of age. Cosmetic correction for syndactyly of digits is done in first year of life and completed by 3 to 4 years of age whereas for midface deficiency and pseudo cleft between 4 to 6 years age. Orthodontic and orthognathic surgery is performed after permanent teeth eruption and completion of growth.

By using selective inhibitors of the FGFR-kinase domain, nonsurgical management of Apert syndrome may be a possibility in the future.¹⁰ Genetic counseling is an important factor as recurrence risk for an affected individual to have an affected offspring is 50%.

The rarity of the syndrome along with the characteristic craniofacial and dental anomalies as well as the genetic transmission makes it necessary to carry out genotyping and genetic advising of each diagnosed case. With the discovery of various prenatal diagnostic modalities, it has been possible to detect AS early and provide a timely multidisciplinary care, thus offering a better quality of life to affected individuals.

CONCLUSION

This paper highlights the importance of dentist as well as the specialist in the early detection of this syndrome and timely provision of multidisciplinary treatment with the purpose of reducing the duration and complexity of further treatment.

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