Apert syndrome a rare craniofacial disorder: A case report

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ABSTRACT

This article presents a case of rare syndrome in an 8-year old child who presented to the clinic with complain of toothache. This is type 1 acrocephalosyndactyly syndrome leading to severe syndactyly of the hands and feet, craniosynostosis and dysmorphic facial features. It is caused due to the mutation in the fibroblast growth factor receptor gene. Apert syndrome’s classical features are syndactyly of hands and feet, frontal bossing of head, saddle nose appearance, hypoplastic maxilla, proptosis, vision loss, short stature and mental retardation. Dental anomalies present in the patients are crowding of teeth, a v-shaped maxillary arch and high arched palate with bilateral palatal swelling resulting in a pseudo cleft configuration in the midline of palate.

Key-words: Acrocephalosyndactylia, craniosynostosis, Ectopic tooth eruption, maxillary hypoplasia.

INTRODUCTION

Apert syndrome is a rare congenital type 1 acrocephalosyndactyly syndrome, representing approximately 5% of all craniosynostosis syndromes with prevalence of 1 in 65000 to 160000 live births⁴,⁵, affecting males and females in equal severity⁶,⁷. It was first described by Whearon in 1894 and reviewed extensively by French physician Apert in 1906⁸.

It is caused by allelic mutation in the fibroblast growth factor receptor (FGFR-2) gene at locus 10q26⁹,¹⁰. This disorder is characterised by severe craniosynostosis, craniofacial abnormalities, symmetric syndactyly of the hands and feet and dysmorphic facial features⁶.

CASE HISTORY

An 8-years old boy was presented in my centre with chief complain of toothache, who was suffering from Apert syndrome. During history it was found that his parents were clinically normal. There was no complication during pregnancy and no other family members showed a similar condition. At birth, he had craniosynostosis, brachycephaly and syndactyly of hands and feet. He was photophobic, so tarsorrhaphy was done in both his eyes by ophthalmologist (Fig. 1).
Extraoral examination revealed a brachycephaly skull with midface hypoplasia, a flat forehead, depression of temporal bones, proptosis, hypertelorism and short nose with bulbous tip and trapezoidal shape of the mouth (Fig 1-A and B). Syndactyly of second, third, fourth and fifth digits were present in both hands (Fig 3-A). Both feet showed a fusion of all toes (Fig 3-B). Frontal bossing of forehead was present with acne like eruption on chest (Fig. 2).
Figure 3: Syndactyly: hands (A) and feet (B).

Intraoral evaluation revealed poor oral hygiene with varying degree of periodontal involvement, an arched swelling giving appearance of pseudo cleft configuration, maxillary arch V-shaped and carious teeth (Fig 4-A and 4-B). After primary examination and management, he was referred to specialized centre for further care.
Figure 4: Intraoral view: (A) Occlusal view and (B) Mandibular view.

DISCUSSION

Dr. Eugene Charles Apert, in 1906, described the triad craniosynostosis (premature fusion of the calvarial sutures especially coronal suture), severe syndactyly of the hands and feet, and dysmorphic facial features, characterising the syndrome. It is easily detected in the neonatal period due to craniosynostosis and associated finding of syndactyly in the hands and feet. He described the nine cases of syndactyly associated with acrocephaly.

It is inherited as an autosomal dominant fashion and being associated with advanced paternal age. The risk of a second child being affected is 1%\(^8\). The most cases of apert syndrome result from two specific mutation of a gene located on chromosome 10q23, encoding FGFR2. The two mutations involve C-to G transversion, leading to a change of codon TCG to TGG, producing serine to tryptophan substitution at amino acid 252. The second mutation is a C937G transversion, changing codon CCT to CGT, resulting in proline to arginine substitution at amino acid 253\(^9\). The 252W is most common mutation occurring within 67% of patient and has been associated with more severe craniofacial anomalies whereas the P353R mutation may be associated with more severe syndactyly\(^9\).

These mutations affects the linking the immunoglobulin link domain 2 and 3 of FGFR 2 and results increase affinity and altered specificity of ligand binding\(^10\). This in turn leads to deregulation and ultimately to pre mature osteogenesis and skeletal abnormalities that characterised the syndrome.

Apert syndrome causes fusion of coronal suture prematurely, leading to an acromcephalic head with flattened occiput, shortened anteroposterior diameter, a high prominent forehead, a characteristic form of nose and mouth\(^6\). The midface of this patient is hypoplastic, resulting in respiratory distress especially in the young child. Because of this, most infants become mouth breather giving ‘Open mouth’ appearance. Ocular anomalies are hypertelorism, proptosis, and strabismus with down slanting palpebral fissure are often present and are due to shortening of bony orbit. The patients may suffer from photophobia and visual loss\(^8,11\). Visual loss can result due to chronic exposure of the unprotected eyes, increased intracranial pressure and compression of the optic nerves. Our patient presents almost all of above mentioned features including photophobia.

The characteristic limb defect helps to distinguish Apert syndrome from other craniosynostosis syndrome. Syndactyly of second, third and fourth digit of the hands and feet always observed in these patients. These often are termed as Mitten hands and Sock feet. In our patient, there is syndactyly of second, third, fourth and fifth digits of both hands and all the digits of feet present. The average height of the affected patient is below that of the general population. This patient has eruption mainly on the chest region. These patients are usually mentally retarded.

Our patient had all the typical oral manifestation of an Apert patient like an high arched palate with bilateral swelling of the palatine processes resulting in pseudo cleft configuration, class 3 malocclusion, maxillary arch V-shaped, tongue appeared excessively large and crowding of teeth due to maxillary hypoplasia\(^3,12\). Dental crowding was so severe in these cases that the malpositioned teeth generated a parallel row with the normal erupted teeth. Kreiberg and Cohen, in
their clinical study of Apert syndrome’s oral manifestation, found pseudo cleft of the soft palate or bifid uvula in approximately 75% of the cases. According to them these patients may present several dental abnormalities, including delayed eruption, ectopic eruption and shovel-shaped incisors\textsuperscript{12}. Letra et al detected a high incidence of tooth agenesis in Apert syndrome patients\textsuperscript{3}.

Apert syndrome may be confused with other craniosynostosis syndromes like Crouzon syndrome, Pfeiffer syndrome, Jackson-Weiss syndrome and Beare-Stevenson syndromes. The presence of broad thumbs and cutaneous syndactyly differentiate Pfeiffer syndrome from apert syndrome\textsuperscript{13} while Jackson-Weiss syndrome can be differentiated on the basis of abnormality in the clinical or radiographic appearance of the feet\textsuperscript{14}. The presence of cutaneous disorder, cutis gyrata and acanthosis nigricans are major factors for the correct diagnosis of Beare-Stevenson syndrome\textsuperscript{15}. So, proper evaluation and characterization of the clinical features are important for the correct diagnosis and treatment of the affected patient.

The cosmetic and functional defects of Apert syndrome can be treated by a multi disciplinary approach using multiple surgeries. These patients generally require lifelong management by a multidisciplinary team of health care specialists. Management of children with Apert syndrome includes surgical correction of craniosynostosis, midfacial hypoplasia and syndactyly. Craniotomy often is performed during the first year of life to treat craniosynostosis to get better results while midface advancement can be done at later to correct the proptosis and midfacial hypoplasia. Early cranioplasty corrects both functional and aesthetics consequences of craniosynostosis.

Now a day, prenatal sonography detection of structural abnormalities associated with Apert syndrome is usually present. The specialist should inform the parents that prognosis is not optimal, so that the parents could opt for termination of pregnancy before the stage of fetal viability. As far as dental treatment is of concern, it requires joint work of oral surgeon, periodontist and orthodontist. The aggressive oral prophylaxis plan plays an important role for the management of preventable oral diseases such as dental caries and periodontal diseases.

REFERENCES