Apert’s Syndrome: A Rare Case in India

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ABSTRACT
Craniosynostosis syndrome is characterized by premature craniosynostosis occurring in association with a variety of other abnormalities. The most common craniosynostosis disease occurring without syndactyly is craniofacial dystosis or Crouzen’s syndrome. Most common craniosynostosis disease occurring with syndactyly is Apert’s syndrome. Apert’s syndrome is characterized by craniosynostosis, midfacial malformations, symmetric syndactyly of hands and feet minimally involving digit second, third and fourth. We report a rare case of 12-year-old girl in India with all features of Apert’s syndrome like hypertelorism, exophthalmos, strabismus, steep forehead, depressed nasal bridge, parrot beak nose and retruded middle third of the face.

Keywords: Craniosynostosis, Syndactyly, Aperts, Hypertelorism, Exophthalmos, Micrognathia.

INTRODUCTION
Over the past several decades tremendous advances have been made in the prevention and treatment of developmental anomalies. This metamorphosis in our conceptualization of developmental malformations has led to an improved ability to handle and prevent them. Despite such improvements, developmental malformations remain a significant cause of morbidity worldwide.1 Even when the mode of inheritance is well established, some conditions continue to exhibit a large number of sporadic occurrences, which make their eradication virtually impossible. As such, it is incumbent on us to learn as much as possible about these conditions. In this way, we can become better clinicians and impart better care to those who so desperately need it. Here, we report one such rare anomaly of Apert’s syndrome.

CASE REPORT
A 12-year-old girl reported to the department with a chief complaint of halitosis. Extraoral examination of the patient showed hypertelorism, exophthalmos, strabismus, steep forehead, depressed nasal bridge, parrot beak nose and middle third of the face is retruded resulting in relative mandibular prognathism (Fig. 1). On examination of hands and feet showed syndactyly of second, third and fourth digits of the hands and all the toes of the feet (Figs 2 and 3). Mental status of the patient showed no abnormality. Intraoral examination revealed soft debris, such as plaque and calculus, cleft palate with high palatal vault and constriction of the maxillary arch. Maxilla was V-shaped with severe crowding of the teeth, bulged alveolar ridges and class III malocclusion (Fig. 4). A provisional diagnosis of Apert’s syndrome was established and radiological investigations were performed.

On radiographic examination maxillary occlusal radiograph shows a vertical radiolucent area in between the roots of central incisors suggestive of cleft palate. Orthopantomograph shows normal eruption of all permanent teeth with tooth crypts of third molars in both the arches and erupting canines, second molars in maxilla and mandible (Fig. 5). Hand-wrist radiographs show synostosis of first, second, third and fourth digits of right and left hands (Fig. 6). Skulls PA view shows multiple radiolucencies appearing as depressions of the inner surface of the cranial vault, which results in a ‘beaten metal appearance’.

Fig. 1: Clinical photograph of patient’s frontal view showing midfacial hypoplasia
DISCUSSION

Craniosynostosis is the term that designates premature fusion of one or more sutures. Reduced or asymmetrical skull growth ensues, causing deformity of the skull vault or the base. Virchow in 1851 noted that there is a cessation of growth in a direction perpendicular to that of the affected suture while growth proceeds in a parallel direction. Apert’s syndrome is one such syndrome which is characterized by craniosynostosis, exorbitism, midface hypoplasia and symmetric syndactyly of both hands and feet. It was named after a French physician E. Apert, who first described it in 1906, as a relatively uncommon craniofacial anomaly. It occurs with frequency of 1/160,000 live births and has been rarely reported from India. The condition may be inherited with a frequency of 50% in the offspring of an affected adult or more commonly develops as a result of spontaneous mutation. The molecular basis of this syndrome appears remarkably specific: Two adjacent amino acid substitutions (either S252W or P253R) occurring in the linking region between the second and third immunoglobulin domains of the fibroblast growth factor (FGR) 2 genes.

An association between this disorder and high parental age has been observed. It is thought to occur as a result of androgen end organ hyperresponse affecting the epiphyses and sebaceous glands resulting in early epiphyseal fusion-short stature, fused digits and acrocephaly.

The clinical features are characterized by early fusion of skull bones (synostosis), mainly coronal sometimes lambdoid,
midface regression and webbed digits (syndactyly). Syndactyly always involves fusion of the soft tissues of the first, middle and ring digits. Commonly associated features include cardiac anomalies, visual and hearing defects, cleft palate and varying degree of acne.

The oral findings in Apert patients are also characteristic, which includes micrognathia of maxilla resulting in crowding of teeth. Cleft palate or bifid uvula is found in approximately 75% of those affected patients. Dental anomalies, such as impacted teeth, delayed eruption, ectopic eruption, supernumerary teeth and thick gingiva are also common⁶ which correlated with our case report.

Early surgical intervention to correct the craniosynostosis is crucial in order to maximise the chances for normal development. Correction of hypertelorism can be undertaken by a facial advancement operation.

We treated the patient with full mouth prophylaxis and patient was educated about the maintenance of oral hygiene measures as she had severe malocclusion. Regular dental check up and maintenance was reinforced and patient was referred to department of orthodontics for correction of malocclusion.

In conclusion, until there is a means to correct the molecular defect, we must rely on a strong multidisciplinary approach to patients with Apert syndrome. A team approach is essential to determine the best collaborative corrective plan for the deficiencies of child which involves neurosurgeons, plastic surgeons, otorhinolaryngologists, orthodontists, ophthalmologists, radiologists, geneticists, pediatricians and dermatologists. These children invariably need speech therapy after the surgical correction of abnormalities is completed.

REFERENCES