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Apert Syndrome- A Case Report With Review

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Abstract
Apert syndrome is the rare acrocephalosyndactyly syndrome type 1, characterized by craniosynostosis, severe syndactyly of hands and feet, and dysmorphic facial features. It demonstrates autosomal dominant inheritance assigned to mutations in the fibroblast growth factor receptor gene. The rarity of the syndrome and similarity of features with other craniosynostosis syndromes makes it a diagnostic dilemma. Genetic counselling and early intervention form an essential part of treatment. Because of the paucity of reported cases in Indian literature and typical features in oral cavity, a dentist should be competent to diagnose and form a part of the multidisciplinary management team. The present case report is about an Apert's syndrome patient highlighting the craniofacial characteristics and oral health care measures for these patients.
Keywords: *Apert's syndrome, autosomal dominant, fibroblast growth factor, craniosynostosis.*

Introduction

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. In 1851, Virchow noted that there is cessation of growth in a direction perpendicular to that of the affected suture while growth proceeds in a parallel direction. There are also distinct craniofacial synostosis syndromes that share common features such as suture synostosis, midface hypoplasia, and facial and limb abnormalities. Apert's syndrome is one such syndrome which is characterized by craniosynostosis, midface hypoplasia, and symmetric syndactyly of both hands and feet.

Case Report

A 21 year-old male patient came to the department of oral medicine and radiology with the chief complaint of difficulty in mouth opening from three months. Patient gives history of decreased mouth opening which was gradual in onset and associated with burning sensation during intake of spicy foods. Personal history revealed chronic gutkha chewing habit since 1 year with a frequency of 10 times per day. He cessated the habit 3 months back. On Physical examination he was thin built, short stature, moderately nourished with normal gait and erect posture. No facial asymmetry with facial form, Leptoprosopic and convex profile. Slight increase in intercanthal distance is seen (Figures 1&2). Syndactyly observed in both hands (Figure 3).

Syndactyly seen in right foot (Figure 4). No relevant medical and family history. Intra oral examination revealed short maxilla, high arched palate (Figure 5) crowding of teeth, blanching of mucosa was present. On palpation vertical fibrous bands were present on right and left buccal mucosa (Figure 7). Generalised bleeding on probing and gingival recession was present Grade I mobility in relation to 12, 21, 31, 32, 34, 41, 42, 44, 45 was seen. Missing teeth in relation to 11, 25, 37, 47. Based on the history and clinical features it was provisionally diagnosed as Apert's syndrome and oral submucous fibrosis. The hand wrist radiographs demonstrate an extra phalange synostosing with index finger and phalanges of middle finger with ring finger of right hand; synostosis involving phalanges of index and middle fingers of left hand (Fig 6).The radiographs of lower extremities demonstrate synostosis of big toe and second toe and also synostosis of second and third metatarsals of right foot (Fig 7).

Treatment plan was Patient was advised to quit gutkha chewing habit. Intralesional injections including hyaluronidase, dexamethasone and placentrix to be given. As for apert syndrome there is no cure. Conventional treatment includes early release of the coronal suture and fronto-orbital advancement and reshaping. As the patient exhibited no severe facial deformities no such treatments are indicated or needed. Advised Oral prophylaxis, scaling procedure. Advised prosthesis, removable or fixed denture for the missing teeth.

Figure 1

Figure 2

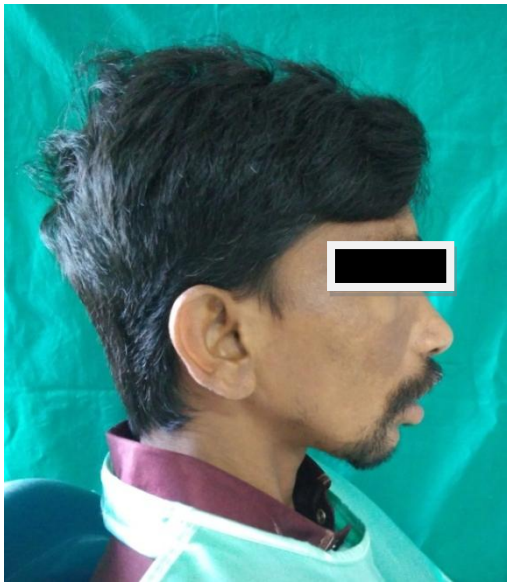


Figure 3

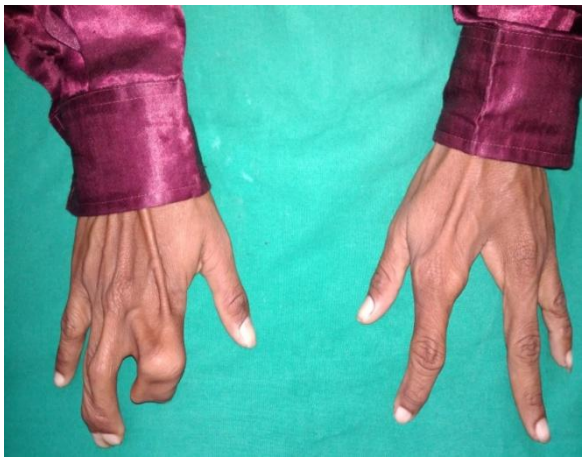


Figure 4

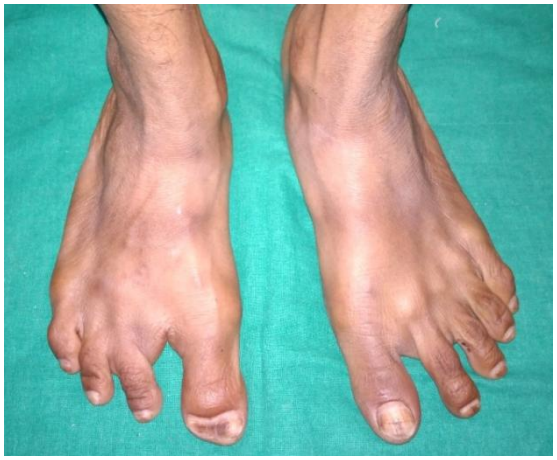


Figure 5

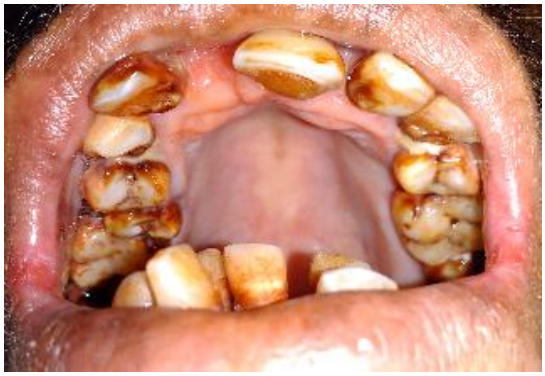
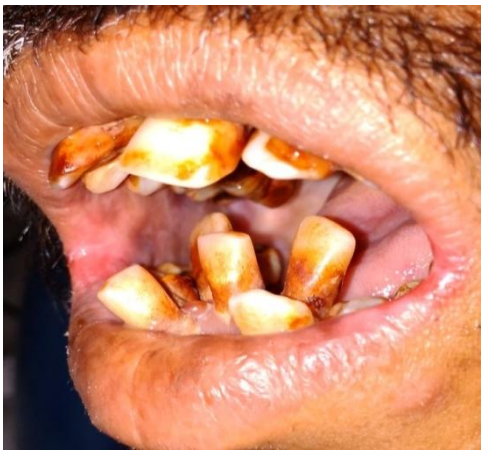


Figure 6



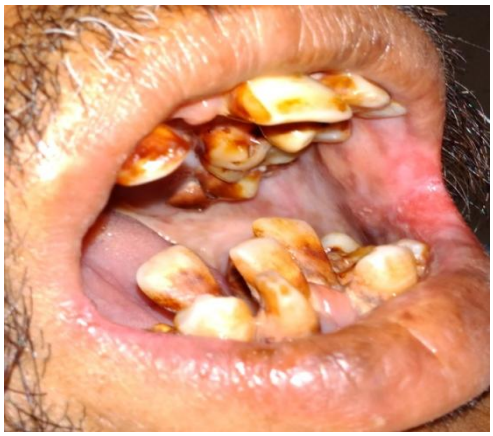


Figure 7

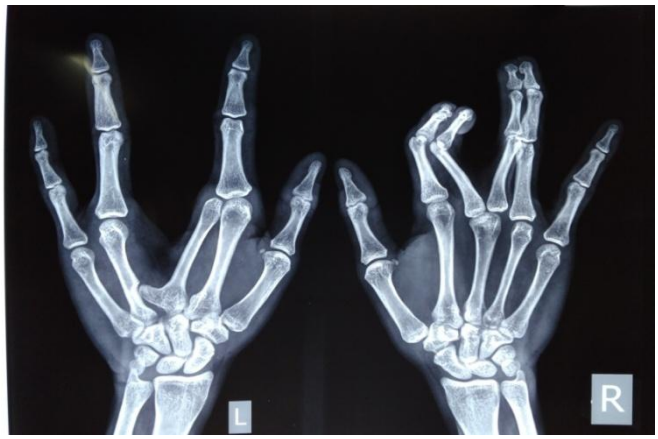
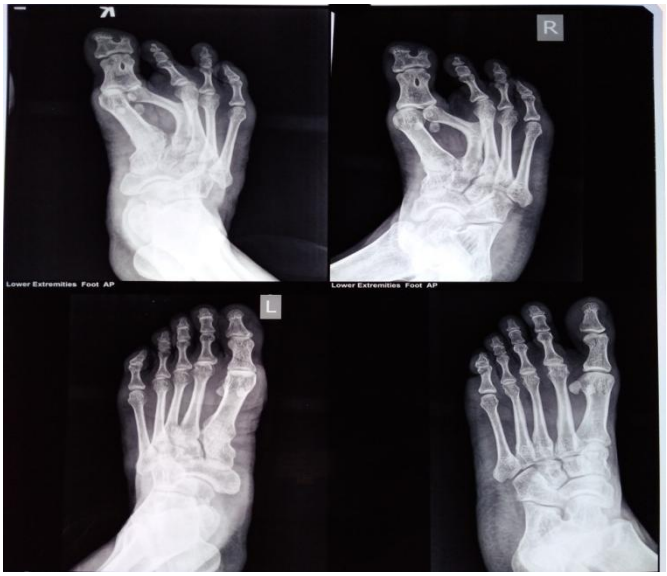


Figure 8



Discussion

Apert's syndrome was described by Wheaton in 1894.[1] In 1906, Apert published a summary on nine cases.[2] Apert's syndrome makes up approximately 4% of all cases of craniosynostosis. The incidence is reported to be 1/160,000 live births.[3]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 gene.[4] Radiology has an important role to play in the evaluation, management, and follow- up of these patients. Plain radiographs are sufficient for diagnosis, but CT has added a new dimension to the evaluation of these disorders.[5] The clinical and oral features of Apert's syndrome are well established and in agreement with the case described in the present report. The syndrome is clinically characterized by premature fusion of the the main reasons for the previous surgical procedures mentioned by the patient. Ocular anomalies and short nose with depression of the nasal bridge could also be observed. Syndactyly, as described by Apert,[6,7] was also present. The oral cavity characteristics included reduction in the size of the maxilla, which may result in tooth crowding and an anterior open bite of the maxilla.[8] Effective clinical management to improve oral health

usually requires the joint work of a periodontist and an orthodontist. For the patient with Apert's syndrome, oral hygiene is as important as it is difficult. Hand deformities make it difficult to brush the teeth. The new generation of electric tooth brushes and fluoride mouth rinses may make the task easier. Professional care, including frequent dental examinations, oral hygiene prophylaxis, fluoride treatments, and dental sealants, is very important.[9] Some affected individuals have anomalies of the viscera, elbows and shoulders, skeleton, and central nervous system, or abnormalities of the upper and lower respiratory tracts.[10]

In the differential diagnosis other genetic disorders which can be seen apart from craniosynostosis include Crouzon, Carpenter (acrocephalosyndactyly type 2), Chotzen, and Pfeiffer syndromes. Specific association of craniosynostosis has been correlated with mutation in the FGFR gene. In the Crouzon syndrome characterized by craniosynostosis, and dysmorphic face, acrocephaly, brachycephaly, exophthalmos, proptosis, hypertelorism, hooked nose, hypoplasic maxilla, ear, and palatine deformities occur. AS especially demonstrates similar characteristics with Crouzon syndrome. In the Crouzon syndrome, contrary to AS, extremities are not involved, and craniofacial deformities lead a milder course. However in AS, multiple sutures fuse prematurely. The face is asymmetric, forehead is more prominent,and exophthalmos

is not so severe. Hand, and foot deformations, and especially extreme cases of syndactyly is its discriminative feature.

Conclusion

Since symptoms of Apert Syndrome demonstrate large variability, the diagnosis, and treatment of the disease require treatment by a multidisciplinary team in collaboration with neurosurgeon, plastic, and reconstructive surgeon, ophthalmologist, psychiatrist, neurologist, perinatologist, and genetician. Diagnosis is based on clinical, radiological, and genetic evaluation. Though its definitive treatment is not available, for anatomical deformities corrective surgery can provide cure. Cranial corrective operations, and fronto-orbital surgery are advised for infants aged 6–9 months, and generally reconstruction of synostosis is considered for patients older than 6 years. Besides these patients can manifest emotional, and behavioural disorders and because of severe craniofacial anomalies, they require psychiatric consultation.[11]

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