Apert Syndrome: A Case Report

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Abstract

Apert syndrome is a rare type I acrocephalosyndactyly syndrome characterized by dysmorphic facial features, craniosynostosis, and severe syndactyly of the hands and feet. It represents an autosomal dominant inheritance and occurs due to the gene mutations in the receptors of the fibroblast growth factor. The oral cavity features includes tooth crowding, reduction in the size of the maxilla, impacted teeth, anterior open-bite, ectopic eruption, delayed eruption, thick gingiva and supernumerary teeth. The mandible simulates a pseudoprognathism but usually is within normal shape and size. In the present case report, a 54 year's old female patient reported with the features of Apert's syndrome such as dysmorphic facial features, occular anomalies, syndactyly and oral features. The case was referred to a specialized centre of clinical care for further treatment.

Keyword: Acrocephalosyndactylia, Apert syndrome, Craniosynostosis

Introduction:

A French physician Apert, in 1906, first described a rare type I acrocephalosyndactyly syndrome known as Apert syndrome that was characterized by severe syndactyly of the hands and feet, craniosynostosis, and dysmorphic facial features.[1,2,3] It is an autosomal dominant inheritance assigned to mutations in the fibroblast growth factor receptors (FGFR-2) gene at locus 10q26.[2,4]

Fibroblast growth factor receptors have a high affinity for fibroblast growth factors that, play a role in signalling pathways with multiple biologic effects including cranial growth and development when bound to their specific receptors.[3,5]

Most of the cases shows disorder results from a mutation in the father and its prevalence at birth is 1:65,0003[2,6] and it equally affects the males and females.[3,5]

Apert syndrome show distinctive clinical features in which the coronal suture fuses prematurely (at less than 3 months), leading to a cone-shaped head known as acrocephalic with a high prominent forehead and shortened antero-posterior diameter. The mid face is hypoplastic. Hypertelorism, down slanting palpebral fissures and proptosis are the occular anomalies. The nose is wide and short due to which the nasal bridge is depressed.[1,2,5,7] Anomalies of the elbows and shoulders, skeleton, viscera, and impaired mental function due to central nervous system anomalies have been reported by previous studies.[5,6]

Apert patient's oral cavity is also characteristic. The clinical findings of patients include a reduction in the size of the maxilla, in the antero- posterior direction particularly, which results in tooth crowding and an anterior open-bite of the maxilla. Size and shape of the mandible is within normal, and it simulates a pseudoprognathism. Impacted teeth, ectopic eruption, delayed eruption, supernumerary teeth, and thick gingiva are also common dental anomalies.[3,5,7,8]

Upper and lower respiratory tract abnormalities include bifid uvula, cleft soft palate, choanal stenosis,

Byzantine-arch palate, and anomalies of the tracheal cartilage.[1] Due to the alteration in facial growth, mouth breathing is observed in most of the cases of Apert's syndrome[3]. Spoon-like deformity of the hands and feet due to the partial to complete fusion of the digits results in Syndactyly[1,2].

A case of Apert syndrome in an adult female patient is presented in this article.

Case Description:

Female patient of 54 years old, came to the camp for regular dental check-up. The patient reported Apert syndrome and was the only case of her family.

The clinical triad of craniosynostosis, midface hypoplasia, and syndactyly of the hands and feet which is the characteristic of Apert Syndrome was present in the patient.

The patient had abnormal shaped skull which was characterised by a high, full forehead and a flat occiput which may be due to the premature closure of one or more of the joints (fissur es) between the bones of the skull and early fusion of the coronal sutures representing Craniosynostosis.

The midface of the patient was incompletely developed. She had flat face, orbits were shallow, small nose, maxillary hypoplasia, and a narrow palate with a bifid uvula **(Figure No. 1)** along with cleft soft palate **(Figure No. 2)**. The patient had varying forms of osseous and cutaneous syndactyly leading to fusion of fingers **(Figure No. 3)**. Other clinical features which were present in the patient includes occular anomalies (proptosis, hypertelorism, slanting palpebral fissures and right eye divergent strabismus) **(Figure No. 4)**, depression of the nasal bridge with short and wide nose **(Figure No. 5)**. Reduction in the size of the maxilla, anterior open-bite of the maxilla, supernumerary tooth, tooth crowding with ectopic tooth eruption were features present in oral cavity **(Figure No. 6).**

To receive a comprehensive healthcare treatment, the patient was referred to a specialized centre of clinical care for special needs patients.

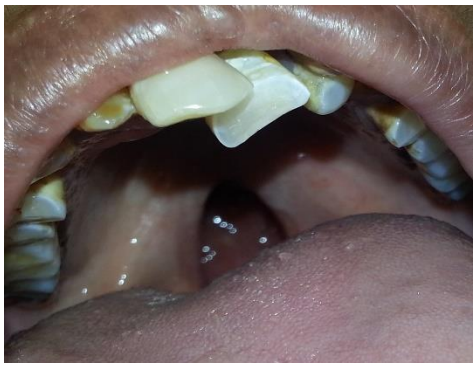

Figure No. 1: Bifid Uvula

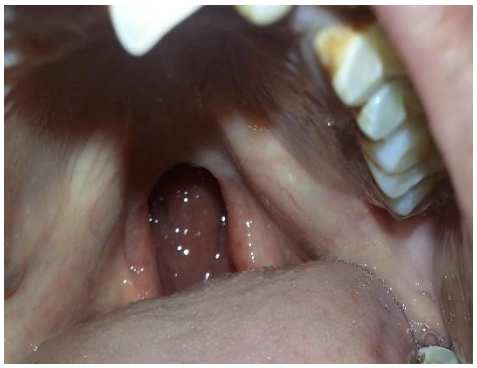

Figure No. 2: Cleft Soft Palate

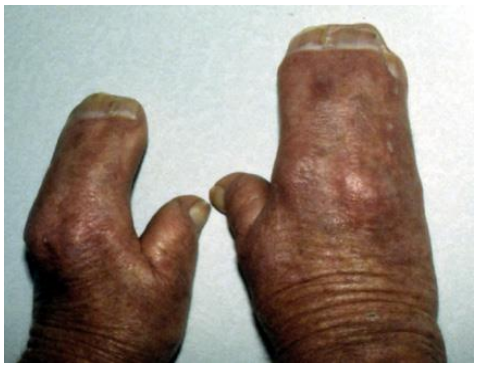

Figure No. 3: Syndactyly of the Hands

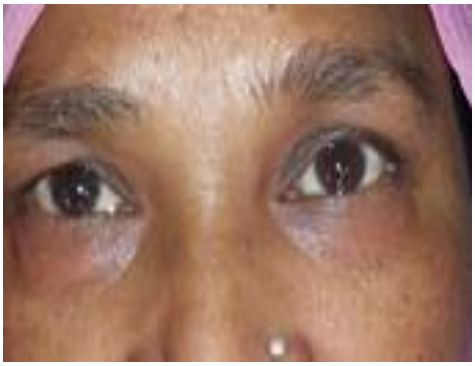

Figure No. 4: Proptosis, Slanting Palpebral Fissures and Right Eye Divergent Strabismus

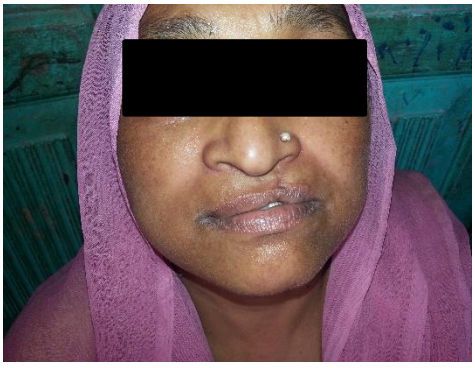

Figure No. 5: Incomplete Cleft Lip with Wide, Short Nose and Depressed Nasal Bridge

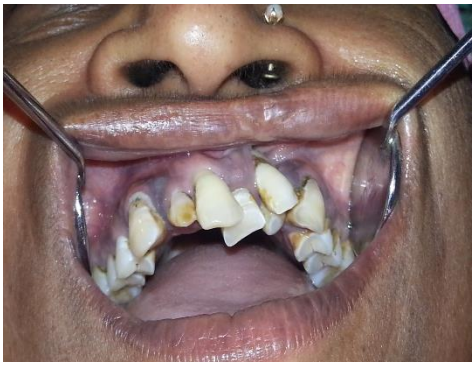

Figure No. 6: Supernumerary Tooth, and Crowding with Ectopic Tooth Eruption

Discussion:

Apert, in 1906, described the triad syndactyly of the hands and feet, dysmorphic facial features, and craniosynostosis characterizing the syndrome.[1,2,3] With the mutations in the fibroblast growth factor receptors (FGFR-2) gene at locus 10q26[2,4], a rare autosomal dominant heritage was linked to the syndrome. Apert syndrome have well established clinical and oral features and are in agreement with the case described in the present report. Clinically the syndrome is characterized by premature fusion of the coronal suture and hypoplastic midface.[1,2,5,7]

Short nose with depression of the nasal bridge and occular anomalies, could also be observed. Tooth crowding and an anterior open-bite of the maxilla due to the reduction in the size of the maxilla are the characteristics oral cavity findings.[3,5,7,8] as seen in the patient. Joint work of an orthodontist and a periodontist are required for the effective clinical management to improve oral health.

Anomalies of the elbows and shoulders, viscera, skeleton and central nervous system[5,6] or

abnormalities of the upper and lower respiratory tracts[1,3] have been reported in some affected individuals. However, the case reported here did not present any related complaint of these anomalies during clinical examination.

Conclusion:

Apert syndrome is a rare autosomal heritage with many affected parts of the body. For the effective planning and treatment of such patients, a multidisciplinary approach provided by dentists, plastic surgeons, neurosurgeons, ophthalmologists and geneticists should be included in the integral healthcare delivery system.

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