CASE REPORT

HAMPERING THE HAMPERED – A SYNDROMIC APATHY

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ABSTRACT
Crouzon's syndrome is an autosomal dominant condition characterized by craniosynostoses with associated dentofacial anomalies, although it is an inherited condition many cases are sporadic and present as de novo mutations arising from unaffected parents. This paper reports a case of crouzon's syndrome in a 22 year old male with particular reference to characteristic clinical features.

Key words: Crouzon syndrome (CS); Craniosynostoses

Introduction
Crouzon's syndrome was first described in 1912 by French neurologist Octave Crouzon as a hereditary syndrome characterized by triad of premature fusion of the cranial sutures, exophthalmos and midface hypoplasia.1-2 It is a rare genetic disorder with an incidence rate of 1 in 25,000 people out of the general population with a frequency rate of 4.5% cases of all craniosynostoses3 and is more common syndrome of a group of more than 100 types of craniosynostoses. Craniosynostoses are a heterogenous group of syndromes characterized by premature sutural fusion which occurs individually4 or relating to other anamolies.5 Crouzon syndrome may be distinguished from craniosynostoses for its association with facial malformations. Majority of the cases of crouzans syndrome are autosomal dominant with large scale of penetrance with variable expressivity.6 It has no sex or race predilection.1 Mutation in the Fibroblast growth factor receptor 2 (FGFR-2) gene is responsible for both sporadic and inherited cases of Crouzon syndrome.7 This paper reports a case of crouzans syndrome in a 22 year old male with particular reference to characteristic clinical features.

Case Report
A 22 year old male reported to the Department of Oral Medicine, Oral Diagnosis and Radiology, Vishnu dental college (Bhimavaram, Andhra pradesh, India) with a chief complaint of missing teeth in upper and lower arches since childhood. Patient had no history of trauma. There were no anomalies reported among any of the sibilings or near relatives. No intellectual or developmental impairment was apparent.

On extraoral examination the patient presented with short stature, brachycephaly, hypertelorism, hypoplasia of the maxilla associated with pseudopagognathism, concave facial profiled front bossing with beaked nose (Figure 1-3). Syndactyly was absent. No obvious dermatological findings such as acanthosis nigricans was observed. Intraoral examination revealed high narrow arched palate, underdeveloped maxilla, prognathic mandible, enlarged tongue. Missing teeth were evident. Occassionally oligodontia, macrodontia, partial anodontia and incomplete eruption of permanent teeth were evident. It is a rare genetic disorder with an incidence rate of 1 in 25,000 people out of the general population with a frequency rate of 4.5% cases of all craniosynostoses3 and is more common syndrome of a group of more than 100 types of craniosynostoses. Craniosynostoses are a heterogenous group of syndromes characterized by premature sutural fusion which occurs individually4 or relating to other anamolies.5 Crouzon syndrome may be distinguished from craniosynostoses for its association with facial malformations. Majority of the cases of crouzans syndrome are autosomal dominant with large scale of penetrance with variable expressivity.6 It has no sex or race predilection.1 Mutation in the Fibroblast growth factor receptor 2 (FGFR-2) gene is responsible for both sporadic and inherited cases of Crouzon syndrome.7 This paper reports a case of crouzans syndrome in a 22 year old male with particular reference to characteristic clinical features.

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In the present case Crouzon syndrome associated with Acanthosis nigricans, mutation mapped to chromosome locus 10q-25-q26. Thirdly, Ocular proptosis which is caused by very shallow orbits, and other features like strabismus and hypertelorism.7 The most common dermatological manifestation seen in crouzon syndrome is acanthosis nigricans which is detectable after infancy in approximately 5% of the patients. It presents as velvety, light brown to black markings usually on the neck arms, or in the groin.

In this case the patient presented with Brachycephaly, maxillary hypoplasia and pseudo mandibular prognathism. Frontal bossing and depressed nasal bridge with narrowed nares were evident. It is well known fact that hypertelorism is thought to arise due to decrease in the growth of sphenozygomatic and sphenotemporal sutures. No dermatological findings were observed in this case. Crouzon’s syndrome is caused by mutations in fibroblast growth factor receptor-2 gene which is mapped to chromosome locus 10q-25-q26.8 In cases of Crouzon syndrome associated with Acanthosis nigricans, mutation is noted in FGFR3 gene (locus4p16.3).9-10 In the present case the patient presented with Brachycephaly, maxillary hypoplasia and pseudo mandibular prognathism. Frontal bossing and depressed nasal bridge with narrowed nares were evident. It is well known fact that hypertelorism is thought to arise due to decrease in the growth of sphenozygomatic and sphenotemporal sutures. No dermatological findings were observed in this case. Crouzon’s syndrome is caused by mutations in fibroblast growth factor receptor-2 gene which is mapped to chromosome locus 10q-25-q26. In cases of Crouzon syndrome associated with Acanthosis nigricans, mutation is noted in FGFR3 gene (locus4p16.3). In the present case scenario, anterior open bite and high arched palate along with partial anodontia and incomplete eruption of permanent teeth were evident. Occassionally oligodontia, macrodontia, peg-shaped and widely spaced teeth have been reported.

Common complication of Crouzon syndrome is conjunctivitis or keratitis secondary to exophthalmos.7 There were rare occurrences of nystagamus, coloboma, microcornea, macrocornea, cataract, glaucoma, blue sclera and luxation of the eye globes.7 Blindness from optic atrophy secondary to intracrani-
al hypertension can also occur and optic atrophy may be due to narrow optic channel.11 Radiographic investigations which includes orthopantomograph to rule out the impacted teeth present, skull radiographs to show synostosis, widening of hypophyseal fossa, small paranasal sinuses, maxillary hypoplasia with shallow orbits as in this case are commonly used to aid in the diagnosis. Cervical radiographic abnormalities like butterfly shaped vertebrae, fusion of bodies and posterior elements are reportedly to be seen often in crouzon syndrome.3 C2-C3 and C5-C6 are affected equally. Other investigations like CT scan and MRI can be advised. CT scan of brain shows signs of raised intracranial pressure, fusion of coronal and sagittal sutures, and three dimensional images might reveal copper beaten appearance. Magnetic resonance imaging (MRI) is used to show corpus callosum agenesis and optic atrophy.

Crouzon syndrome can be diagnosed prenatally by ultrasonography and molecular genetic testing.12 Unusual head shape and binocular and interlobular diameter can be assessed through ultrasound.13 Molecular gene testing for FGFR-2 gene can be performed by amniocentesis and using amniotic fluid for DNA isolation and to detect mutation.12 The differential diagnosis of Crouzon syndrome includes Craniosynostosis, Apert syndrome, Pfeiffer syndrome, Carpenter syndrome and Saethre Chotzen syndrome. Management of Crouzon syndrome patients varies according to the age of the patient and severity of the disease.

A multidisciplinary approach is required to provide the best result with any craniofacial disorder. The goal of the treatment is stage reconstruction to coincide with the facial growth patterns, visceral function and psychosocial development. The prognosis depends upon on the severity of malformation. As for the dental management of the patient two options are advised for midface hypoplasia.

Use of distraction osteogenesis for reconstruction and LeFort III osteotomy for mid facial advancement and this have to be supplemented by rhinoplasty, genioplasty and bone grafts. The other option may be orthopedic force of Rapid maxillary expansion (RME)-face mask therapy stimulates cellular activity in circummaxillary sutures and maxillary tubercula and this facilitates maxillary forward displacement. A combination of orthopedic and orthodontic treatment was found to be effective for improving the appearance and occlusion of the patients with mild Crouzon syndrome without surgery.

Conclusion
In conclusion, syndromic patients pose a social neglect and consequently hampering quality of life. A multidisciplinary approach is bestowed to arrive at a proper diagnosis and prompt relief of the patient from the associated oral ailments and improving the quality of life of an affected individual.

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References
How cite this article

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Source of Support: Nil
Conflict of Interest: None Declared