



Case Report

Crouzons syndrome- A rare genetic disorder

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ABSTRACT

Background: Crouzon syndrome is a rare genetic autosomal dominant disorder characterized by distinctive malformations of the skull and cranio-facial region.

Case Description: The parents of a 7 years old boy reported with a complaint of pain in the upper front and back teeth region since 3-4 months. He also presented with typical features of craniofacial deformity, maxillary hypoplasia, exophthalmos, hypertelorism, ptosis are present among other manifestations. The overall presentation gave an indication towards a clinical diagnosis of Crouzon's syndrome. This case report describes the different clinical features and management under general anesthesia of a pediatric patient who reported with characteristic findings of this syndrome.

Conclusion: This case report emphasizes the important role of a pediatric dentist in diagnosing such syndrome and to provide corrective measure along with co-ordination of a multi-disciplinary team. The team may comprise a geneticist, neurosurgeon, psychologist, speech and language therapist, nose surgeons and respiratory care specialist.

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1. Introduction

Crouzon syndrome is rare genetic autosomal disorder, also known as branchial arch syndrome, which can be evident at time of birth or during pregnancy. This was first described in 1912 by a French physician 'Octave Crouzon'. He described four essential characteristics that include exorbitism, retromaxillism, infra-maxillism and paradoxical retrognathia.¹ There is premature closure of cranial sutures, mid-facial hypoplasia and orbital deformities. The syndrome with reported incidence of 1:25,000 live births is the most common of over 70 conditions in which pre-mature fusion of the cranial sutures may be a feature.² However

it affects both genders equally. It may be inherited as a genetic trait or it may arise as a new condition in the family. In the familial type, the condition is inherited as an autosomal dominant condition.³

The autosomal disorder is thought to arise due to mutation in the genes that codify type-2 fibroblast growth factor receptor (FGFR2) both in sporadic and the inherited cases.⁴ However, 50% of the Crouzon syndrome new cases are not inherited but result from new spontaneous mutations during phase of development.

The diagnosis and management is entirely depends on clinical findings and examination. Thus this case report encompasses management of a 7 years old pediatric patient who was managed under general anesthesia with the consent of pediatrician and physician.

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2. Case Report

A 7 year-old boy reported to our department with his parents complaining of pain in upper front and back tooth region since 3-4 months. Parents also complaints of difficulty on eating and mastication. There was no history of similar disease in any of the siblings and parents of affected child.. There were no systemic manifestations such as fever or weight loss. The patient was mentally challenged and his normal developmental milestones were delayed. He started walking at age of 4 years and started speaking at age of 2 years (slurry speech). Patient was not on any long term medications.

On clinical examination, the patient presented with the typical manifestations of craniofacial dysostosis. Brachycephalic head form, prominent forehead, concave profile, decreased molar prominence, ocular proptosis, hypoplastic midface, flat and broad nasal bridge, exophthalmos hypertelorism, maxillary retrognathism and a short upper lip (Figure 1). No anomalies of the upper or lower extremities were noted.

Intra-oral examination reveals grossly carious teeth wrt 54, 64, 74, 75, 83, 84, 85 (FDI notation system), root stumps wrt 51, 52, 61, 62, 81, smooth surface caries wrt 71, 73, 82 and Occlusal caries wrt 65 (Figure 2). The diagnosis of Chronic irreversible pulpitis wrt 51, 52, 61, 62, 64, 73, 74, 75, 84, 85 and reversible pulpitis wrt 65, 72, 73, 82 was initiated. Treatment plan included single sitting pulpectomy wrt 52, 54, 62, 64, 74, 75, 83, 84, 85, Glass Ionomer Cement/Composite restoration wrt 65, 72, 73, 82 and extraction wrt 51, 61 under general anesthesia considering mental status of patient. Due mentally challenged status of patient intra-oral periapical radiographs and ortho pantomogram were not possible to obtain. Investigation like Complete blood picture and nasal patency test were performed and physician consent was obtained. Post-operative care was done following procedure and medications were prescribed and patient was made under follow up. After 7 days follow up patient parents were satisfied with treatment.

3. Discussion

Craniofacial abnormalities are mostly present at time of birth and may progress with time. Decreased mental function is present approximately 12% of cases.⁵ Primary features like headache, seizures, visual disturbances, ear canal stenosis or atresia and upper airway obstruction due to nasal septum deviation can be seen. Physical findings like craniosynostosis begins during the first year and usually completes by second and third year.⁶ Coronal and sagittal sutures are most commonly involved leading to high prominent forehead. The most common ocular abnormalities reported are shallow orbits, exposure keratitis, optic atrophy, ocular proptosis and orbital hypertelorism.⁷



Fig. 1: Shows Brachycephalic head form, prominent forehead, ocular proptosis, hypoplastic midface, flat and broad nasal bridge, exophthalmos hypertelorism,



Fig. 2: Shows root stumps wrt 51,52,61,62

Intra-orally delayed eruption, malocclusion, crowding of teeth, v- shaped maxillary dental arch, high arched palate, gingival enlargement, multiple carious teeth (poor oral hygiene) and bifid uvula have been reported.

Differential diagnosis of Crouzon's syndrome considers Apert syndrome and others like Carpenter syndrome, Pfeiffer syndrome, Seatre-Chatzen syndrome, and Jackson Weiss syndrome. Diagnostic tests such as skull radiographs, Comparative CT scan 3D reconstruction analysis, Magnetic

resonance imaging and molecular analysis (for FGFR2 mutation) have been used.⁵

Management of Crouzon's disease includes multidisciplinary approach. In the first year of life, it is preferred to release the synostotic sutures of the skull to allow adequate cranial volume thus allowing for brain growth and expansion. Skull reshaping may need to be repeated as the child grows to give the best possible results.⁸ Mid-facial advancement and jaw surgery can be done in later stage to provide adequate orbital volume and reduce the exophthalmoses to correct the occlusion to an appropriate functional position and to provide for a more normal appearance. Prognosis is said to be fair and entirely depends on malformation severity.

Our patient reported late and was never explained before and treated for syndrome. Oral hygiene was compromised owing to mental retardation and lack of awareness in parents. Under GA complete oral rehabilitation was performed. The child did recover uneventfully and is undergoing regular follow-ups for further preventive oral hygiene measures. For good oral hygiene specially designed handle for his toothbrush can be fabricated so that he will be comfortable using it and regular use of mouthwash Kiddodent (0.05% Sodium fluoride, 0.03% Triclosan and 5% xylitol) can be prescribed.

He was also referred to psychologist, speech and language therapist, nose surgeons and pediatricians. In such patients parent cooperation is very much important as they should not take it as a social burden. However in our case both the parents cooperate very well and understood importance of good oral hygiene.

4. Conclusion

An understanding of such disorder is necessary for the dental team to make the appropriate referrals to insure the patient receives the best available care at appropriate time. The Pediatric Dentist should be an integral part of the multidisciplinary team to provide a normal and healthy life to special child.

5. Source of Funding

None.

6. Conflict of Interest

None.

References

1. Crouzon LE. Dysostose cranio-faciale hereditaire. *Bulletin de la Societe des Medecins des Hopitaux de Paris*. 1912;33:545–55.
2. Cohen MM. Craniosynostosis and syndromes with raiosynostosis: incidence, genetics, penetrance, variability 6. and new syndrome updating. *Birth Defects Orig Artic Ser*. 1979;15(5B):13–63.
3. Kanaparthi R, Kanaparthi A. Craniofacial dysostosis- The dental perspective: A case report . 2012;39:196.
4. Fries PD, Katowitz JA. Congenital craniofacial anomalies of ophthalmic importance. *Surv Ophthalmol*. 1990;35(2):87–119.
5. Bowling EL, Burstein FD. Crouzon syndrome. *Optom - J Am Optom Assoc*. 2006;77(5):217–22.
6. Rice DP. Clinical features of syndromic craniosynostosis. *Front Oral Biol*. 2008;12:91–106.
7. Padmanabhan V, Rai K, Hegde A. Crouzon's syndrome: A review of literature and case report. *Contemp Clin Dent*. 2011;2:211–4.
8. Jarund M, Lauritzen C. Craniofacial dysostosis: Airway obstruction and craniofacial surgery. *Scand J Plast Reconstr Surg Hand Surg*. 1996;30:275–9.

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