ISSN: 2581-5989
PubMed - National Library of Medicine - ID: 101738774

International Journal of Dental Science and Innovative Research (IJDSIR)

IJDSIR : Dental Publication Service

Available Online at: www.ijdsir.com

Volume – 7, Issue – 2, April – 2024, Page No. : 01 - 03

Crouzon syndrome with sickle cell trait: Report of a rare combination

[1]Dr. Rooposhi Saha, Professor, Department of Pedodontics and Preventive Dentistry MARDC, Pune, India.

[2]Dr. Manjiri Joshi, Professor, Department of Oral Medicine and Radiology, MPDC, Vadodara.

[3]Dr. Vanishree B. K., Associate Professor, Department of Pedodontics and Preventive Dentistry, MARDC, Pune, India.

[4]Dr. Shweta Kajjari, Reader, Department of Pedodontics and Preventive Dentistry, KLE VKIDS, Karnataka, India

Corresponding Author: Dr. Rooposhi Saha, Professor, Department of Pedodontics and Preventive Dentistry MARDC, Pune, India.

Citation of this Article: Dr. Rooposhi Saha, Dr. Manjiri Joshi, Dr. Vanishree B. K., Dr. Shweta Kajjari, "Crouzon syndrome with sickle cell trait: Report of a rare combination", IJDSIR- April – 2024, Volume –7, Issue - 2, P. No. 01 – 03.

Type of Publication: Case Report

Conflicts of Interest: Nil

Abstract

Crouzon syndrome is a rare genetic disorder. Features of this syndrome include craniosynostosis, maxillary hypoplasia, hypertelorism, hearing loss, deviation of nasal septum, spine deformities among others. Here we report a case with most of the features of Crouzon syndrome and an additional never before reported sickle cell trait that runs in the family. Along with a multidisciplinary approach for treatment, an early and accurate diagnosis is helpful for such patients.

Keywords: Craniosynostosis, Sickle Cell, Hearing Loss.

Introduction

Craniosynostoses are a rare group of disorders caused primarily due to premature closure of most often coronal and sagittal sutures. In 1912, Crouzon first described this syndrome as craniofacial synostosis and suggested the term "dysostose craniofaciale hereditare". It is also referred to as craniofacial dysostosis, hereditary craniofacial dysostosis, dysostosis craniofacialis, syndromic craniosynostosis and premature craniosynostosis. [1]

Its incidence is estimated at 1 in 25,000 live births and it constitutes 4.8% of all craniosysostosis. In 25% cases it may occur sporadically because of fresh mutation. [2]

This syndrome is caused by malformations of the mesenchyme and ectoderm resulting due to mutation in fibroblast growth factor receptor 2 (FGFR2) and FGFR3 genes on chromosome 10 in both sporadic and inherited cases. [3]

Features of this syndrome include craniosynostosis, maxillary hypoplasia, shallow orbits, ocular proptosis and hypertelorism. There is considerable variation in the extent of these features, but ocular proptosis is always present even in the absence of craniosynostosis which is a prerequisite for the diagnosis of Crouzon syndrome. Other features associated with this condition are hearing

loss, deviation of nasal septum, calcification of stylohyoid ligament, cervical spine anomalies and stenosis of jugular foramen.

Franceschetti coined the term "pseudo Crouzon syndrome" to designate those cases which simulate the Crouzon syndrome, and in which patients do not have relative mandibular prognathism, parrot nose and there is no familial occurrence. [1]

Case Report

A 7-year-old male patient reported to the outpatient department with a chief complaint of decayed teeth. The boy was operated for cleft palate 2 years back and for right eye 1 year back. His medical history disclosed that he also has a sickel cell trait. Family history revealed the similar problem in his older male sibling who could not survive more than two months. Patient has an elder sister who is completely normal. Patient's mother has a sickel cell trait. Patient has defective speech although academic performance in school is good. In general examination a pegion chest appearance was noted. (Figure 1)

Extraoral examination revealed, there was presence of straight profile, maxillary hypoplasia, malar deficiency, shallow orbits, ocular proptosis and hypertelorism. (Figure 2a & 2b)

Intraoral examination revealed dental arches were "U" shaped. Maxilla showed a high vault palate with lateral palatal swellings.

On intraoral examination patient had all the decayed teeth in maxillary arch except for the primary second molars. In the mandibular arch all the teeth except for one or two teeth had caries. (Figure 3a & 3b) Panoramic radiograph revealed presence of permanent teeth buds in delayed eruption. Lateral skull view showed thickening of inner and outer table of cranium and copper metal beaten appearance in occipital region is seen.

A paediatrician's consultation was also obtained for a complete systemic evaluation and to confirm the diagnosis. A complete blood picture was suggested before commencing the treatment. All the teeth that needed restorations were completed; followed by extractions and space maintainers. An esthetic rehabilitation was done using an acrylic denture.

Discussion

Craniosynostosis commonly begins during the first year of life and is usually completed by the age of 2–3 years. Abnormalities of calvarial shape in Crouzon syndrome are dependent on the sutures involved. Premature fusion of synchondroses of cranial base, subsequent lack of bone growth perpendicular to the synchondroses and cranial base produces characteristic cranial shapes like brachycephaly, trigonocephaly, and scaphocephaly. The most severely affected patients can demonstrate a "clover leaf" skull (Kleeblatt schadel deformity). [1]

Exophthalmosis is stated to be a prerequisite for Crouzon syndrome and is said to be caused by a lack of forward sutural growth in the temporal and cranial base region. This produces a relative prominence of eyeball, which sometimes results in blindness due to increased intracranial pressure. In the present case also the patient was operated for problems in the right eye.

Hypertelorism which was seen in our case is a prominent finding in the affected individuals and is thought to arise due to decrease in growth of the sphenozygomatic and sphenotemporal sutures.

In Crouzon syndrome, maxilla is hypoplastic, which causes relative mandibular prognathism. In our case maxilla was hypoplastic but without significant mandibular prognathism.

Patients with Crouzon syndrome show a high vault palate, which was present in our case although cleft

palate surgery had already been performed on presentation.

An exceptional observation in the present case is a familial sickle cell trait in association with the Crouzon syndrome. The trait seems to have passed on from the generations to the opposite sexes; like the father and the daughter were absolutely normal whereas the mother and the sons were affected. In all the previously reported cases association of Crouzon with sickle cell has not been documented which makes it a rare review.

Management of a patient of Crouzon syndrome has two components:

First is the release of prematurely fused sutures based on evidence of raised intracranial pressure. Surgery is mainly carried out early after 3-6 months. The principle is the release of bony ankylosis by exposure of fused sutures via a coronal flap.

2) Craniofacial reconstructive surgery including advancement of the maxilla and frontonasal complex; and other surgeries depending upon the deformities in the patient like rhinoplasty, oculoplasty and cleft lip and cleft palate repair can be done.

Early and accurate diagnosis of a patient of Crouzon syndrome is essential. Genetic counselling plays an important role. The need, extent and timing of treatment depend upon the severity of the disease and age of the patient. For complete evaluation, optimum treatment planning and comprehensive services, a multidisciplinary approach to the management of a patient of Crouzon syndrome is needed. [2]

References

1. Samantha Y, Vardhan Harsha T, Kiran Ravi A, Sankar Sai J A & Ramakrishna B. Familial Crouzon Syndrome. Contemp Clin Dent. 2010 Oct-Dec; 1(4): 277–280.
2. Kaur Haroop, Waraich Singh Harmeet, Sharma M C. Crouzon Syndrome: A Case Report and Review of Literature. Indian J of Otolaryngology and Head and Neck Surgery. 2006 Dec; 58(4): 381-382
3. Pandey N, Pandey K R, Singh K R, Shah K N. Crouzon Syndrome: A Social Stigma. BMJ Case Reports. 2012; 10: 1-3

Legend Figures

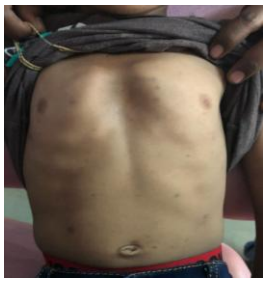

Figure 1: Pigeon chest appearance

Figure 2a Figure 2b

Figure 2a & 2b: Extra oral front and side profile and features

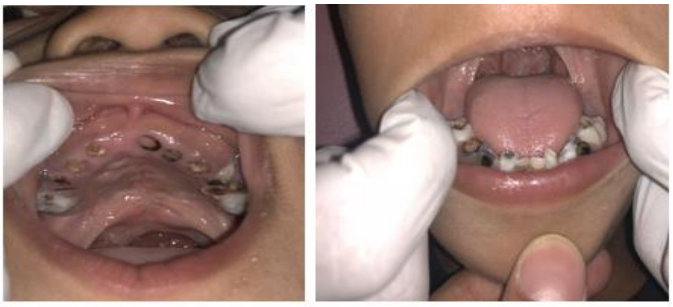

Figure 3a Figure 3b

Figure 3a & 3b: Maxillary and mandibular arches