

CROUZAN SYNDROME – A CASE REPORT

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ABSTRACT

Genetic disorders account for a significant amount of morbidity and mortality in children and are of primary interest to the dentist. Crouzan syndrome is one of a rare group of syndromes characterized by craniosynostosis or premature closing of the cranial sutures. The major features are Brachycephaly, ocular proptosis, under developed maxilla, midface hypoplasia, rare cleft lip, palate. Early Craniectomy is often needed to alleviate the raised intracranial pressure. This paper discusses a case report of five year old girl with the features of crouzan syndrome and a multidisciplinary approach to be followed in managing the situation.

KEY WORDS: Craniofacial syndromes, Crouzon, Premature Synostosis

INTRODUCTION

In 1912, a French neurologist, Octave Crouzon (1874-1938)¹ first described a hereditary syndrome of craniofacial dysostosis in a mother and son, which included a triad of skull deformities, facial anomalies and exophthalmos.^{2,3}

The skull is composed of many bones that are separated by sutures. Premature synostosis (fusion of the suture) commonly involves the sagittal and coronal suture. Lambdoidal sutures are occasionally involved. The order and rate of suture fusion determines the degree of deformity and disability.^{4,5} Various sutures may be prematurely synostosed, and multiple sutural involvement is found eventually in most cases.⁶ Premature sutural fusion may occur alone or together with other anomalies, making up various syndromes.⁷

Crouzon syndrome is an autosomal dominant disorder with complete penetrance and variable expressivity. It is characterized by premature closure of calvarial and cranial base sutures as well as those of the orbit and maxillary complex (craniosynostosis). Other clinical features include hypertelorism, exophthalmos, strabismus, beaked nose, short upper lip, hypoplastic maxilla, and relative mandibular prognathism. Unlike some other forms of autosomal dominant craniosynostosis, no digital abnormalities are present.⁸

In this article we present Crouzon's syndrome, which is one of the syndromes associated with

synostosis of cranial sutures. The differential diagnosis of Crouzon's syndrome includes simple craniosynostosis as well as the Apert, Pfeiffer and Saethre-Chotzen syndromes.

Case report:

A five year old female (Fig.1) accompanied with her mother was referred to the Department of pedodontics complaining of presence of tooth in the nose (Fig. 2) and wanted removal of the same. History revealed that mother had a full term, normal uneventful pregnancy, medical and dental history was not contributory. No positive familial history was obtained. However patient's mother gave a history of surgical repair for cleft lip, when the patient was

six months old. Her appearance was different from other children of her age (Fig.3), with protruding eyes and enlarged calvarium. History from the parents revealed that these features started developing since she was a small child.

On careful examination, following features were identified:

1. Exophthalmos
2. Hypertelorism
3. Retruded maxilla, resulting in midface retrusion
4. Asymmetrical enlargement of the skull

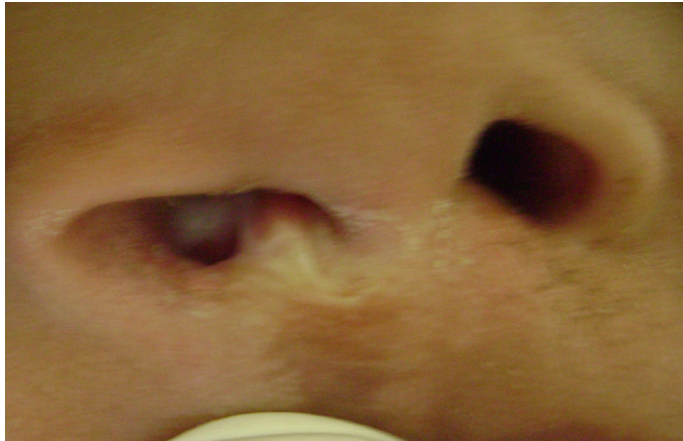


Fig. 1. Tooth in the nasal cavity



Fig. 2. Five year old female



Fig. 3: Extra oral appearance of the patient



Fig. 4. clinical features of the hands



Fig. 5: Clinical features of the feet



Fig. 6: Hand wrist radiograph



Fig. 8: Submentovertex view



Fig. 7: Lateral cephalogram



Fig. 9: Extracted tooth from the nasal cavity

5. Cleft involving soft palate, uvula
6. Ankyloglossia
7. Surgically treated cleft lip
8. Bending of Metacarpels, Flat foot, Widened space between hallux and the rest of the toes. (Fig.4 and Fig. 5)

Investigations:

Hand – Wrist radiograph:(Fig.6) findings included

- Presence of Dactyly in relation to thumb
- Bending of metacarpels in few fingers

Lateral cephalogram: (Fig. 7) revealed

- Severe frontal bossing
- Boat shaped skull
- Thickening of calvarium
- Maxillary hypoplasia
- Decreased facial height

Submentovertex view: (Fig. 8) showed

- Unfused sutures in the frontal bone.
- Broadening of skull.

A syndrome due to the complexity of symptoms as mentioned above always demands a multidisciplinary approach for successful outcome. The aim of treatment in this case was removal of tooth from the nasal cavity.

Following thorough blood and radiographic investigations, tooth from the nasal cavity was removed under local anesthesia (Fig. 9) and post extraction instructions were given.

Discussion:

Crouzon syndrome is a genetic disorder, commonly inherited as an autosomal dominant trait, with complete penetrance and variable expressivity, but about one-third of the cases do arise spontaneously. The male-to-female preponderance is 3:1. With the advent of molecular technology, the gene for the Crouzon syndrome could be localized to the Fibroblast Growth Factor Receptor II gene (FGFR2) at the chromosomal locus 10q 25.3-q26, and more than 30 different mutations within the gene have been documented

in separate families. Management of such a problem requires multidisciplinary approach. Treatment includes measures to minimize intracranial pressure and secondary calvarial deformities. Orthodontic treatment with subsequent orthognathic surgical intervention has to be followed in managing the dentofacial deformity.

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