

Crouzon Syndrome – A Rare Case Report

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ABSTRACT

Background: A 13 year old boy presented with gradual poor grade in his school performance. Parents of the boy were worried because of his deteriorating academic performance and physical appearance. Patient was subjected to thorough clinical examination and Radiological examination and was found to be having features and findings consistent with Crouzon Syndrome. The patient was properly counselled and was lost to follow up after 6 months due to his social and economical constraints.

Keywords: Radiology, Diagnostics, Orthopaedics.

INTRODUCTION

Crouzon syndrome is rare case and diagnosis is equally challenging and rarity of the case merits its presentation. It's very rare disease and very few cases have been published worldwide.

CASE REPORT

A 13 year old boy presented with gradual poor grade in his school performance. He had no other siblings. The physical characteristic included flat occiput with high prominent forehead with subtle frontal bossing. Patient had shallow orbits with mid facial (maxillary) hypoplasia, Exophthalmos & Occular hypertelorism were quite obvious. The nose had beaked appearance (parrot beaked) with DNS (Deviated Nasal Septum). On examination Mandibular prognathism with malocclusion (overcrowding) of upper teeth was



seen. The ears were low set. The Radiological picture was quite characteristic which showed Craniosynostosis (both coronal and sagittal sutures were involved)

resulting in Brachycephaly with high prominent forehead. Sphenoid bone appeared flattened. Midface (Maxillary) hypoplasia was seen. Exophthalmos (Proptosis) secondary to shallow orbits and occular hypertelorism was seen. Mild DNS (Deviated Nasal Septum) to right with compressed nasal passage was seen. Mandible prognathism with narrow high arched palate and V shaped maxillary dental arch was seen. Overcrowding of upper teeth with malocclusion and single peg shaped tooth seen. However there was no evidence of any Hydrocephalus. Ear canals were bilaterally normal. No evidence of any vertebrae fusion or subluxation of radial heads seen (reported in some 18% of cases)^{5,2}.

INVESTIGATIONS

Investigations included plain skiagrams including skeletal survey and CT scan studies.

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Fig 1: Showing Compressed Nasal Passage.



Fig 4: Skeletal Survey.



Fig 2: Craniosynostosis.



Fig 5: Patient's Picture showing beaked nose and low set ears.



Fig 3: Malocclusion / Overcrowding of Upper teeth.

DIFFERENTIAL DIAGNOSIS

Crouzon syndrome has gamut of findings and differential diagnosis included Carpenter syndrome and Apert syndrome.

TREATMENT

Cranio facial surgeries and dental surgeries are done to prevent mastication problems and preservation of brain function if due to sutural overriding. However this patient had no such

problems and didn't undergo any surgical treatment.

DISCUSSION

Incidence of Crouzon syndrome is currently estimated to occur in one out of every 25,000 people out of the general population^{5,6}. It is caused due to mutations in Fibroblast growth factor receptor -2 (FGFR2) gene, mapped to chromosome locus 10q25-10q26 but exhibits locus heterogeneity with causal mutations in FGFR2 and FGFR3². This is seen in greater frequency in families with a history of the disorder, but that doesn't mean that everyone in the family is affected. It has no race predilection and no sex predilection. Diagnosis of Crouzon syndrome usually can occur at birth by assessing the signs and symptoms of the baby. Further analysis including radiographs, magnetic resonance imaging (MRI) scans, genetic testing, X rays and CT scans can be used to confirm the diagnosis of Crouzon syndrome.

As a very complicating result of the changes to the developing embryo, the symptoms are very pronounced features, especially in the facial areas. Low set ears is a typical characteristic as ears on the fetus are much lower than those on an adult^{6,2}. During normal development, the ears "travel" upward on the head, however in Crouzon patient this pattern of development is disrupted.

The most notable characteristic of Crouzon syndrome is cranial synostosis but it usually presents as brachycephaly, which results in the appearance of a short and broad head⁶. Fusion of different sutures (coronal and sagittal sutures are most commonly involved) leads to different patterns of growth of skull. Examples include Trigonocephaly (fusion of the Metopic suture), Brachycephaly (fusion of coronal suture), Dolicocephaly (fusion of the sagittal suture), Oxycephaly (fusion of coronal and lambdoidal sutures), Plagiocephaly (unilateral premature closure of lambdoid and coronal sutures), Kleeblattschaedel (premature closure of all sutures)^{2,4,5}. Flat occiput is seen. Cloverleaf skull is rare (only 7%). Hydrocephalus may be seen in few cases (Progressive in 30%)² Approximately 73% of patients have chronic tonsillar herniation and syringomyelia may be present.

Midface (maxillary) hypoplasia may be present. Exophthalmos (bulging eyes due to shallow eye sockets after early fusion of surrounding bone) may result in frequent exposure conjunctivitis or keratitis. Ocular hypertelorism (greater than normal distance between the eyes) and Divergent strabismus can also be seen. Psittichorhina (beak-like nose) with compressed nasal passage along with choanal atresia/stenosis and Deviated nasal septum is commonly seen. Hypoplastic maxilla (insufficient growth of the midface) results in relative mandibular prognathism (chin appears to protrude despite normal growth of mandible) and gives the effect of the patient having a concave face³ Overcrowding of upper teeth, malocclusions and V-shaped maxillary dental arch is seen³. There may be Narrow, high or cleft palate and bifid Uvula. Occasional oligodontia, macrodontia, peg-shaped and widely spaced teeth can be seen^{2,3}. The ear canals may be narrow or absent. Cervical fusion (18%) involving C2-C3, C3-C4 and C5-C6 is also found. Block fusions involving multiple vertebrae is also found. Subluxation of radial heads with Ankylosis of the elbows is seen rarely^{2,5,6}.

For reasons that are not entirely clear, most of the Crouzon patients also have noticeable shorter humerus and femur bones in proportion to the rest of their bodies than members of the general population.

Surgery is typically used to prevent the closure of the skull from damaging the brain's development. Without surgery, blindness and mental retardation are the typical outcomes. To treat the midface deficiency, the lower orbit and midfacial bones can be moved forward^{1,3}.

Dental Consideration:

From dental point of view, this disorder is important to understand since many of the physical abnormalities are present in the head, and particularly the oral cavity. Common features are a narrow/ high - arched palate, posterior bilateral crossbite, hypodontia (missing some teeth), and increased spacing between teeth³ due to maxillary hypoplasia. Crouzon patient generally have a considerable permanent underbite and subsequently cannot chew using their incisors^{1,3}.

LEARNING POINTS

1 There is a Greater frequency seen in families with a history of the disorder but that doesn't mean that everyone in the family is affected.

2 Diagnosis of Crouzon syndrome usually can occur at birth by assessing the signs and symptoms of the baby

3 Surgery is used wherever necessary to prevent the closure of the skull from damaging the brain's development. Neurosurgery & Dental surgery help in correcting dentofacial deformities the deformities. A multidisciplinary approach is required.

4. This article enforces the necessity for a dentist to recognise and diagnose genetic diseases.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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