Research Paper Medical Science

Crouzon's Syndrome: A Case Report

Dr. Gopikishan Sharma	PG Resident, department of Paediatrics , GMC Kota
Dr. Dinesh kumar Meena	PG Resident, department of Paediatrics , GMC Kota
Dr. R. K. Gulati	Professor and Head of Department , GMC Kota

ABSTRACT

Crouzon's syndrome is an autosomal dominant disorder with complete penetrance and variable expressivity. Crouzon's syndrome is caused by mutation in the fibroblast growth factor receptor 2 (FGFR2) gene.. The disease is characterized by premature synostosis of coronal and sagittal sutures which begins in the first year of life. Case report of a male newborn is presented with characteristic features of Crouzon's syndrome

KEYWORDS Crouzon's syndrome, fibroblast growth factor, premature synostosis

INTRODUCTION;-

Cranial skeletogenesis is unique. The cranial skeleton is composed of an assortment of neural crest and mesoderm- derived cartilages and bones that have been highly modified during evolution. Cranial malformations, although uncommon, compromise not only function but also the mental well-being of the person. Recent advances in human genetics have increased our understanding of the ways particular gene perturbations produce cranial skeletal malformations.[1] However, an abnormal head shape resulting from cranial malformations in infants and children continues to be a diagnostic and therapeutic challenge. Crouzon's syndrome is an autosomal dominant disorder with complete penetrance and variable expressivity or can appear as a mutation.[2] Described by a French neurosurgeon Octave Crouzon in 1912,[3] it is a rare genetic disorder. It may be transmitted as an autosomal dominant genetic condition. Crouzon syndrome is caused by mutation in the fibroblast growth factor receptor 2 (FGFR2) genes.[4] The disease is characterized by premature synostosis of coronal and sagittal sutures which begins in the first year of life. Once the sutures become closed, growth potential to those sutures is restricted. However, multiple sutural synostoses frequently extend to premature fusion of skull base causing midfacial hypoplasia, shallow orbit, maxillary hypoplasia, and occasional upper airway obstruction.[5] Intraoral manifestations include mandibular prognathism, overcrowding of upper teeth, and V-shaped maxillary dental arch.[3] Narrow, high, or cleft palate and bifid uvula can also be seen. Occasional oligodontia, macrodontia, peg-shaped, and widely spaced teeth have been reported.[3,5]Crouzon's syndrome occurs in approximately 1 in 25,000 births worldwide.[6] Crouzon syndrome makes up approximately 4.8% of all cases of craniosynostoses.[7] No known race or sex predilection exists.[5] The differential diagnosis of Crouzon's syndrome includes simple craniosynostosis as well as Apert syndrome, Carpenters syndrome, Saethre-Chotzen syndrome, Pfeiffer syndrome.[8] While cases have been documented, seldom have reported with mental retardation and also very few have been found on the oral rehabilitation inclusive of preventive procedures in these children.

CASE REPORT;-

A male neonate [Fig; 1]birth weight 2.75 kg admitted on day of life first with respiratory problem.On general physical examination patient had abnormal head shape {plagiocephaly},bilateral exomthalmos ,high arched palate ,maxillary hypoplasia with craniosynosyosis. Head circumference was 30 cm.Patient had no significant family history.Both parents and 3 other siblings were normal .CT Bone window[Fig:2] was suggestive of saggital and coronal suture craniosynostosis. All above mentioned features confirmed this Crouzon syndrome



Fig: 1



Fig:2

Discussion:
Craniofacial abnormalities are often present at birth and may progress with time.

Crouzon's syndrome is an autosomal-dominant disorder with complete penetrance and variable expressivity, but about one third of the cases do arise spontaneously. The male to female preponderance is 3:1.[2,10] With the advent of molecular technology, the gene for Crouzon's syndrome could be localized to the fibroblast growth factor receptor II gene (FGFR 2) at the chromosomal locus 10q 25.3-q26, and more than 30 different mutations within the gene have been documented in separate families.

Differential diagnosis of Crouzon's syndrome considers Apert syndrome and other problems including Carpenter syndrome, Pfeiffer syndrome, Seatre-Chotzen syndrome, and Jackson Weiss syndrome. Patients with associated acanthoses migricans can have FGFR3 mutation. [9]

The appearance of an infant with Crouzon's syndrome can vary in severity from a mild presentation with subtle midface deficiency to severe forms with multiple cranial sutures fused and marked midface and eye problems. Upper airway obstruction can lead to acute respiratory distress and the presence of mental retardation is rare in these children.[3,10] Increased intracranial pressure leading to optic atrophy may occur, which can produce blindness if the condition is not treated.

Management of Crouzon's disease is multidisciplinary and early diagnosis is important. 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.[6,10] If necessary, mid-facial advancement and jaw surgery can be done 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 depends on malformation severity.[6,011]

We have diagnosed the case early with the syndrome.We advised him to regular follow up and timely visit to neurosuergon, dentist, opthalmologist and oral and maxillofacial suergon for proper management and minimuize complication.

Contributors: All authors were involved in data collection, analysis and drafting of the paper.

Competing interests: None stated.

REFERENCES

1. Helms JA, Schneider RA. Cranial skeletal biology. Nature. 2003;423:326–31[pubmed] | 2. Fogh-Andersen P. Craniofacial dysostosis (Crouzon's disease) as a dominant hereditary affection. Nord Med. 1943;18:993–6. | 3. Crouzon LE. Dysostose cranio-faciale héréditaire. Bulletin de la Société des Médecins des Hôpitaux de Paris. 1912;33:545–55. | 4. Fries PD, Katowitz JA. Congenital craniofacial anomalies of ophthalmic importance. Surv Ophthalmol. 1990;35:87–119[pubmed] | 5. Hlongwa P. Early orthodontic management of Crouzon Syndrome: A case report. J Maxillofac Oral Surg. 2009;8:74–6. [PMC free article] [PubMed] | 6. Cohen MM., Jr Craniosynostosis update 1987. Am J Med Genet Suppl. 1988;4:99–148. [PubMed] | 7. Gray TL, Casey T, Selva D, Anderson PJ, David DJ. Ophthalmic sequelae of Crouzon syndrome. Ophthalmology. 2005;112:1129–34. [PubMed] | 8. Regezi JA, Sciubba JJ. 4th ed. Philadelphia: W.B. Saunders co; 1999. Oral pathology–Clinical pathologic correlations; pp. 477–8. | 9. Gorlin RJ, Cohen MM, Levin LS. 3rd ed. Oxford: Oxford University Press; 1990. Syndromes of the head and neck; pp. 516–26. | 10. Jarund M, Lauritzen C. Craniofacial dysostosis: Airway obstruction and craniofacial surgery. Scand J Plast Reconstr Surg Hand Surg. 1996;30:275–9.[pubmed]