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A CLASSICAL CASE OF CROUZON SYNDROME

Pediatrics

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ABSTRACT

The Crouzon syndrome or craniofacial dysostosis is a rare disease that affects the craniofacial skeleton development. Although it is uncommon, it has a transmission risk of 50% when one of the parents is a carrier. One third of cases arise sporadically through fresh mutations.

KEYWORDS

Crouzon Syndrome, Craniosynostosis, Craniofacial dysostosis, Turricephaly, Fibroblast growth factor gene (FGFR) .

INTRODUCTION

As Described by a French neurosurgeon Octave Crouzon in 1912, Crouzon syndrome is a rare genetic disorder with 1 in 25,000 births worldwide and makes up to 4.8% incidence of all the cases of craniosynostosis. The triad composed by cranium deformities, facial anomalies and exophthalmia, described by Crouzon in 1912, forms today the Crouzon's syndrome. 1,2,3,4.

Crouzon's syndrome is caused by mutation in the fibroblast growth factor receptor -2 (FGFR-2) gene 4. Normally, the sutures in the human skull fuse after the complete growth of the brain, but if any of these sutures close early then it may interfere with the growth of the brain. The disease is characterized by premature synostosis of coronal and sagittal sutures which begins in the first year of life. Since the suture becomes cast, the growth perpendicular to it becomes restrict and compensatory growth occurs in the remaining open sutures to allow the continuity of the brain development causing abnormal osseous growth and production of facial deformities.

CASE DETAILS

A 13 months old female child was brought to Alluri Sita Ramaraju Academy of Medical sciences (ASRAM hospital) with the complaints of inability to flex both elbow joints. Developmentally the child is normal and attaining all milestones as per age.

On appearance the patient have a macrocephaly with tower shaped head (turricencephaly), bulging of anterior fontanelle, exophthalmos, low set of ears, parrot beaked nose and short extremities.

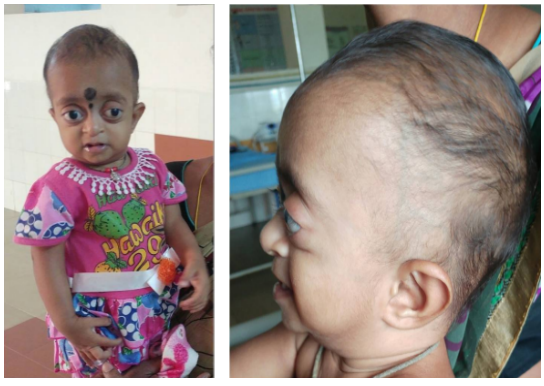

Fig 1. A and B : Frontal and lateral view face showing the frontal bossing, midface hypoplasia, hypertelorism, exophthalmos, depressed nasal bridge giving parrot beak appearance.

RADIOLOGICAL EXAMINATION

Radiological investigations suggested craniofacial dystocia with prominent cranial vault when compared to facial bones. Abnormal convolutional markings on cranial vault seen giving copper beaten appearance. Maxillary hypoplasia, Enlarged Sella, hypertelorism, persistence of anterior fontanelle and metopic suture.

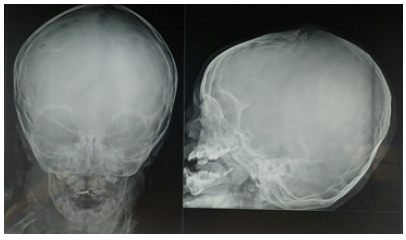

Fig 2 . AP and lateral view of the skull—demonstrating maxillary retrusion, relative mandibular prognathism and Prominent convoluted markings of the skull.

DISCUSSION

Craniosynostosis is the term used to describe premature fusion of one or more cranial sutures. Crouzon syndrome is an autosomal-dominant disorder with premature synostosis of frontal and parietal sutures 2. 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 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 5. FGFR -2 genes are known to affect suture development. FGFRs are transmembrane receptor tyrosine kinase proteins which are composed of an extracellular domain with three immunoglobulin-like domains (IgI, IgII, and IgIII), a transmembrane domain, and a split tyrosine kinase domain.

Crouzon syndrome is characterized by craniosynostosis, hydrocephalus, ocular proptosis, shallow orbits, and maxillary hypoplasia. The patients are often associated with impaired vision and dental malocclusions. The patient might have upper airway obstructions causing respiratory distress 6 and short stature due to growth hormone deficiency 6.

Differential diagnosis of Crouzon's syndrome includes Apert's syndrome, Carpenter syndrome, Pfeiffer syndrome, Seatre-Chotzen syndrome, and Jackson Weiss syndrome. Patients with associated acanthoses nigricans can have FGFR3 mutation.5

CONCLUSION

Management of Crouzon 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.7,8. Mid-facial advancement and jaw surgery can be done to provide adequate orbital volume and reduce the exophthalmoses to correct the occlusion and attain an appropriate functional position and allow a more harmonious appearance.

Craniofacial syndromes, if diagnosed at any early stage, can be managed by orthopedic growth modification, thus avoiding the need for major facial surgeries and additionally benefiting the psychological development of the patient. However, the prognosis depends on malformation severity.

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