

## Crouzon Syndrome: A Case Report

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### ABSTRACT

Crouzon syndrome is the most common syndrome in the craniosynostosis group. Crouzon syndrome accounts for about 4.8% of all cases. It usually has autosomal dominant inheritance with full penetrance and variable expressiveness from subtle to severe forms and is caused by maxillary hypoplasia with craniosynostosis, proptosis, and relative mandibular protrusion. be characterized. Mutations in the fibroblast growth factor receptor 2 gene have been implicated in the development of this rare genetic disorder. Our work reports the diagnosis of this rare syndrome in young patients based on clinical and radiological features. Prompt and timely treatment of the syndrome has allowed this patient to lead a normal life despite the syndrome.

**KEYWORDS:** Crouzon's Syndrome, Craniosynostosis, Craniofacial dysostosis

### INTRODUCTION

Premature union of one or more sutures causes a craniofacial malformation known as craniosynostosis. This asymmetric growth alters the shape of the skull base and calvaria, resulting in characteristic deformities. There are over 100 syndromes associated with craniosynostosis. Among them are Crouzon Syndrome, Apert Syndrome, Pfeiffer Syndrome, Carpenter Syndrome, Sessle-Chotzen Syndrome, and Jackson-Weiss Syndrome.

#### Case description:

An 1 year-old boy presented to the pediatric clinic for a general health evaluation. A detailed family and medical history was collected because the child's appearance and head size were abnormal. The mother had a normal vaginal delivery and was the fourth child from a non-consanguineous marriage. No abnormalities have been reported in any of the siblings or close relatives. The child was taking no medications and the parents denied any history of allergies. The child was developing normally for his age and all milestones were achieved at the appropriate age.

**How to cite this paper:** Dr. Kala Barathi. S | Mr. Azrudheen. B "Crouzon Syndrome: A Case Report" Published in International

Journal of Trend in Scientific Research and Development (ijtsrd), ISSN: 2456-6470,

Volume-7 | Issue-1, February 2023, pp.1240-1241,

URL: [www.ijtsrd.com/papers/ijtsrd53851.pdf](http://www.ijtsrd.com/papers/ijtsrd53851.pdf)



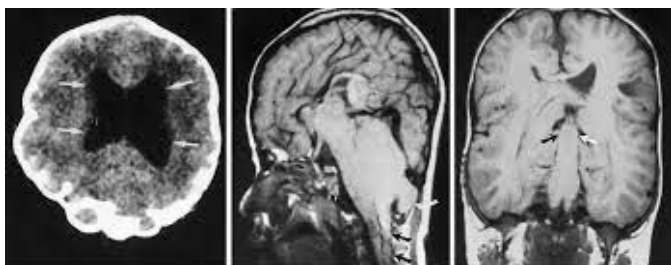
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A general head-to-toe examination of the child revealed an oval head with a convex facial profile (acrocephaly). Ocular findings include prominent eyeballs (proptosis), bilateral proptosis, and hypertelorism. The patient had no finger abnormalities or hearing loss.

Intraoral examination revealed a high arched palate, hypoplastic maxilla leading to pseudoprognathism of the mandible. History showed that these features began to develop from the birth of the baby and gradually increased in severity over time. There was no significant positive family history. Given the above findings, radiographs of the skull were taken, showing fusion of the coronal and sagittal sutures, shallow orbits, and an enlarged intercanthal space.

A brain MRI was also performed, which showed cortical thinning with fusion of the coronal and sagittal sutures, and a pronounced sulcus interconjunctival space with widening of the lateral ventricles. There was also evidence of thinning of the corpus callosum with upper swelling. Gross radiographic features confirmed the diagnosis of Crouzon syndrome.



**Figure 1: crouzon syndrome Brain MRI**

### Discussion:

Crouzon syndrome is an inherited disorder, also known as brachial arch syndrome. This syndrome affects the embryonic first humeral arch, the progenitor of the maxilla and mandible. In its early stages, the syndrome was called "craniofacial dysostosis". This disorder is characterized by three features: craniosynostosis, midfacial hypoplasia, and proptosis. Premature closure of cranial sutures causes abnormal growth of the skull, affecting growth and development of the orbital and maxillary complexes.

The cause is due to mutations in the fibroblast growth factor receptor 2 (FGFR2) gene, which maps to chromosomal locus 10q25-10q26, although FGFR-2 (Crouzon syndrome) and FGFR3 (Crouzon syndrome with hyperepidermal hyperplasia) Locus heterogeneity due to causative mutations is shown. nigricans) in various patients. The most common ocular abnormalities are shallow orbit, proptosis, orbital schisis, strabismus, papilledema, optic nerve atrophy, exposure keratitis, and decreased vision. Rarely, nystagmus, iris coloboma, aniridia, anisocoria, microcornea, macrocornea, cataract, ectopia lentis, blue sclera, glaucoma, and dislocation have also occurred.

Treatment is multidisciplinary and multistage surgery is recommended. Early craniectomy with frontal advancement is most commonly indicated to prevent or treat increased intracranial pressure. If necessary, midface replacement and maxillofacial surgery can be performed to provide adequate orbital volume, reduce proptosis, and correct the occlusion to proper

functional position. Prognosis is limited to malformations depends on the severity.

### Conclusion:

Early detection, patient education and timely intervention are critical steps in the management of this syndrome. Early initiation of a multidisciplinary approach improves survival in children with Crouzon syndrome and also helps subjects overcome social stigma, as subjects with Crouzon syndrome have a near-normal lifespan.

### Conflict of Interest

None

### Funding

None

### Consent for publication

Informed consent was obtained from the parents of the patients to publish this case in medical journal.

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