# **Case Report**

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# Crouzon syndrome: a case report

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#### **ABSTRACT**

Crouzon syndrome is an example of syndromes caused by premature obliteration and ossification of two or more sutures, most commonly coronal and sagittal. Described by a French neurosurgeon in 1912, it is a rare genetic disorder. Crouzon syndrome is caused by mutation in the Fibroblast Growth Factor Receptor 2 (FGFR2) gene. This disorder is characterized by distinctive malformations of skull and face (craniofacial region). Premature cranial suture closure is the most common skull abnormality. The case of an 8-month-old boy with Crouzon syndrome which is one of syndromes associated with synostosis, is presented. He presented in OPD with a cranial deformity, maxillary hypoplasia and exophthalmos. The clinical, characteristic radiological features and investigations carried out, along with treatment of this patient are discussed as part of multidisciplinary management.

**Keywords:** Crouzon syndrome, FGFR2

# INTRODUCTION

Crouzon syndrome is a rare genetic disorder characterized by premature closure of cranial sutures, exophthalmos and mid facial hypoplasia. Crouzon syndrome occurs in approximately 1 in 25000 births worldwide. It makes up approximately 4.8% of all cases of craniosynostosis, making it the most common syndrome within the craniosynostosis group. It may be transmitted as an autosomal dominant genetic condition or appear as a mutation.1 No known race or sex predilection exists. The majority of patients with Crouzon syndrome have mutations in the extracellular immunoglobulin III domain of the Fibroblast Growth Receptors 2 (FGFR2) gene. The differential diagnosis of Crouzon syndrome includes Apert syndrome, Pfeiffer syndrome, Jackson-Weiss syndrome, Carpenter syndrome. Crouzon syndrome is distinguishable from other craniosynostosis syndromes by lack of hand and/or foot abnormalities. Multiple staged surgeries are the general treatment plan for patients with Crouzon syndrome. Hereby a case report of 8-month-old boy with Crouzon syndrome is presented.

# **CASE REPORT**

8-month-old boy presented to Paediatric OPD for general health checkup. Since child's appearance and head size was not normal, family and medical history was taken in detail. The mother had normal vaginal delivery, and this was her fourth child from non-consanguineous marriage. There were no anomalies in any of the siblings or near relatives reported. The child was not on any medications and parents denied any history of allergy. The child was developmentally normal according to his age and all the milestones were achieved at appropriate age.

General head to toe examination of the child revealed elliptical shaped head (acrocephaly) with convex facial profile. Ocular findings include prominent eyeballs (exophthalmos), bilateral proptosis and hypertelorism. The patient did not have any digital abnormalities or hearing deficit.

Intraoral examination revealed high arched palate, hypoplastic maxilla leading to pseudoprognathism of mandible. History revealed that these features started developing since baby was born and the severity gradually increased over a period of time. There was no significant positive family history. Taking into consideration the above-mentioned findings, X ray skull was done which showed fusion of the coronal and the sagittal sutures, shallow orbits and widened intercanthal distance.



Figure 1: Images of 8-month-old child with Crouzon syndrome.

MRI Brain was also done which revealed thinning of cortex with fusion of coronal and sagittal sutures, prominent sulco-gyral spaces with dilatation of lateral ventricles. There was also evidence of thinning of corpus callosum with superior bulge. The overall radiological features confirmed the diagnosis of Crouzon syndrome though the genetic study was not done.

### **DISCUSSION**

Woctave Crouzon (1874-1938), a French neurologist, in 1912, described the hereditary syndrome of craniofacial synostosis in a mother and son. He described the triad as skull deformities, facial anomalies and exophthalmos now known as Crouzon syndrome. It is an autosomal dominant disorder with complete penetrance and variable expressivity.

The cause is attributable to mutations in the fibroblast growth factor receptor-2 (FGFR2) gene which is mapped to chromosome locus 10q25-10q26, but exhibits locus heterogeneity with causal mutations in FGFR-2 (Crouzon syndrome) and FGFR3 (Crouzon syndrome with Acanthosis nigricans) in different affected individuals.

The most common ocular abnormalities are shallow orbits, ocular proptosis, orbital hypertelorism, strabismus, papilledema, optic atrophy, exposure keratitis and visual loss. There also have been rare occurrences of nystagmus, iris coloboma, aniridia, anisocoria, microcornea, megalocornea, cataract, ectopia lentis, blue sclera, glaucoma and luxation of the eye.<sup>3</sup>

In the mouth, short upper lip, hypoplastic maxilla, relative mandibular prognathism, malocclusions and v shaped maxillary dental arch have been reported.

Headaches and seizures are attributable to elevated intracranial pressure. Conductive hearing loss is common owing to ear canal stenosis or atresia. Acanthosis nigricans is the main dermatological manifestation in 5% cases of Crouzon syndrome.<sup>4</sup>

Radiographs, MRI scans, genetic testing and X rays can be used to confirm the diagnosis.

The treatment is multidisciplinary and multiple staged surgeries are recommended. Early craniectomy with frontal bone advancement is most often indicated to prevent or treat increased intracranial pressure. If necessary, midfacial advancement and jaw surgery can be done to provide adequate orbital volume, reduce the exophthalmos and correct the occlusion to appropriate functional position.<sup>5</sup> Prognosis depends on severity of malformations.

#### **CONCLUSION**

Crouzon syndrome should be managed as early as possible as it results in poor cosmetic appearance and results in other complications like mental retardation, airway obstruction and decreased visual acuity as the age advances. With proper treatment, these patients can be productive and active members of the main stream society. Genetic counselling plays important role in these casest.

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