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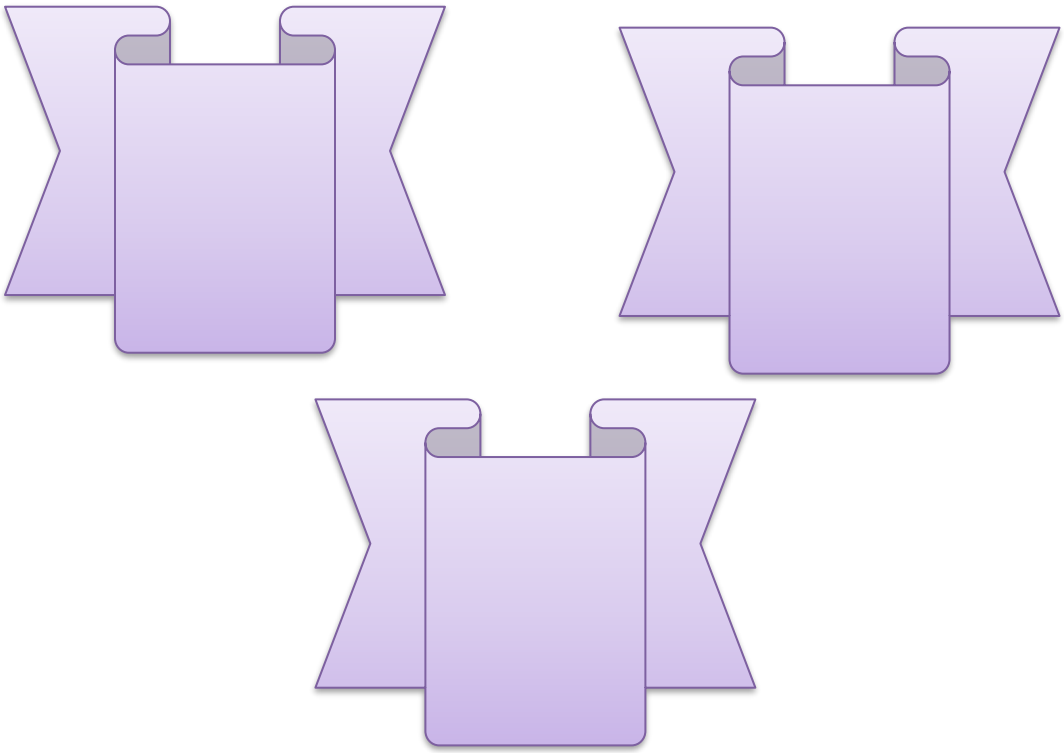

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## Case Report

# Crouzon Syndrome with Hydrocephalus

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## ABSTRACT

**Background:** Hydrocephalous in the presence of primary craniosynostosis is a unique condition with respect to pathogenesis, clinical significance, and morphological appearance. It is rarely observed in nonsyndromic craniosynostosis, and in these cases usually attributable to coincidental disorders. Conversely, it is a common feature of syndromic craniosynostosis. The management of CS requires a multidisciplinary approach, and early diagnosis is vital in order to avoid complications such as blindness and raised intracranial pressure.

**Case presentation:** A 7-years-old boy presented to the Neurosurgery OPD complaining of facial disfigurement, headache, vomiting and one episode of fit. X-Ray Skull films showed typical features of impressions of sulci and gyri demonstrating raised ICP due to fused sutures. Computed tomography scan showed hydrocephalus and prominent markings on the skull's inner table with shallow orbits. Based on all these findings the patient was diagnosed to have the Crouzon syndrome.

**Conclusion:** In true hydrocephalus ventriculo-peritoneal shunting is currently the single promising mode of treatment in case of craniosynostosis case.

**Keywords:** Crouzon syndrome; Hydrocephalous; Craniosynostosis

## INTRODUCTION

Crouzon's syndrome [CS] previously referred to as craniofacial dysostosis, is a rare autosomal dominant disorder that was first described by a French neurosurgeon, Octave Crouzon, in 1912 [1]. It is manifested by premature fusion of coronal and sagittal sutures, which presents at birth and leads to an abnormal shape of the skull [2]. Mutation in the fibroblast growth factor receptor 2 [FGFR2] gene, located on chromosome 10q25-q26 [2] is the main cause of this syndrome. While CS patients with associated acanthosis nigricans can have FGFR3 gene mutation. This syndrome has a prevalence rate of approximately 16.5 cases per million live births and constitutes 4.8% of all cases of this syndrome [3, 4].

## CASE PRESENTATION

A 7-years-old boy [figure 1], presented to the Neurosurgery OPD complaining of facial disfigurement, headache, vomiting and one episode of fit. Parents said that these complaints had been there for the past 3 to 4 years but they never went to visit a healthcare practitioner. The headache was sudden in onset, throbbing in character, sometimes radiating to the neck, aggravated on posture change. In the start it was mild in intensity and had now risen to a moderate severity and is often associated with vomiting. The vomiting was an infrequent symptom and often occurred in association with headache. Past medical and surgical history were unremarkable. The family was a low-class family. Father was a farmer and mother, a housemaid. He was their only son. There was no family history of any congenital abnormality, Diabetes Mellitus, Hypertension, Asthma, etc. The father added "No one from our family ever visited a doctor. We never felt the need to…"

Baby was born via simple vaginal delivery at term. APGAR score was normal and the mother mentioned the abnormal facial appearance of the baby at birth. He was the first baby of the family, and no consanguinity was reported.

Examination: On inspection there was a healthy baby lying comfortably on the examination table with a blue colored beautiful dress and glasses. He had coarse facial features with frontal bossing, craniosynostosis of the coronal sutures, overriding skull bones, shallow orbits, proptosis, maxillary hypoplasia, hypertelorism, wide nasal bridge, low set ears and mandibular prognathism. X-Ray Skull films in lateral and PA view [figure 2 &3] showed typical features of impressions of sulci and gyri demonstrating raised ICP due to fused sutures. Computed tomography scan showed hydrocephalus and prominent markings on the skull's inner table with shallow orbits. Based on all these findings the patient was diagnosed to have the Crouzon syndrome. The presenting symptoms pointed towards raised ICP due to Hydrocephalus so Ventriculoperitoneal shunt was planned. Patient was put on O.R list and medium pressure Medtronics VP Shunt was passed. Patient was discharged at 2nd post-op day. At follow-up on 14th post-op day the baby's symptoms resolved. Patient was vomiting and fit free since the operative intervention. The shunt was working properly. At 6 months follow-up [figure 4] visit baby's shunt was in good working condition with good push and recoil. The parents were very much satisfied with the surgery.

**Figure [1]:** Facial features of Crouzon syndrome

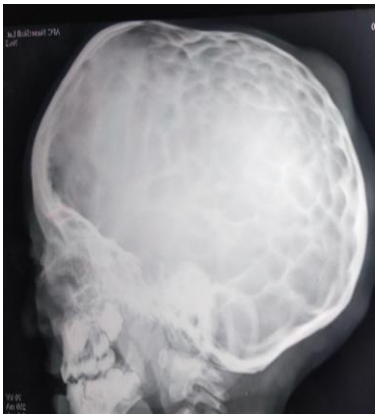

**Figure [2]:** Lateral view X-ray of skull

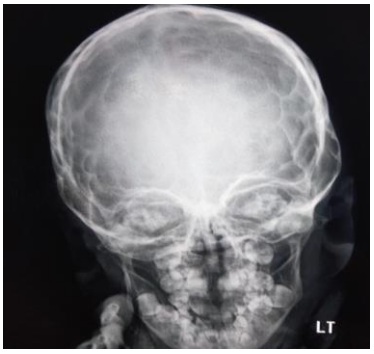


**Figure [3]:** Antero-Posterior view x-ray of skull

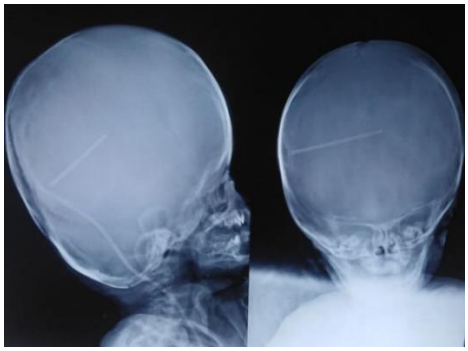

**Figure [4]:** X-ray skull of patient after 6 months showing VP shunt in-situ with resolution of typical features of impressions of sulci and gyri demonstrating raised ICP

## DISCUSSION

The growth of the brain tissue perpendicular to the sutures is limited by craniosynostosis, and hence compensatory growth occurs in the direction of open sutures. This results in characteristic deformities as the shape of the cranial base and vault are affected by asymmetrical growth. The features of this syndrome include craniosynostosis, facial abnormalities, brachycephaly, exophthalmos and other eye abnormalities. [5]. These manifestations could either progress or even regress over time. Additionally, these syndromic patients can also present with progressive hydrocephalus, hearing loss, fusion of C2 and C3 along with intraoral complications such as mandibular, teeth, palate, and uvular abnormalities [4]. However, infants with CS can phenotypically vary from a range of mild to severe forms as this syndrome has complete penetrance but variable expressivity [5, 6]. Mental retardation is usually not seen [5].

The differential diagnosis of Crouzon's syndrome includes Muenke cranial craniosynostosis, Apert syndrome, Pfeiffer syndrome, Carpenter syndrome, Jackson-Weiss syndrome, and Saethre-Chotzen syndrome [7, 8]. Apert syndrome presents with all manifestations as seen in CS, except for syndactyly of hands and feet, as for Pfeiffer syndrome, it will have additional manifestations which include broad thumbs and broad big toes along with variable syndactyly [4, 10].

Early prenatal non-invasive testing [NIPT] and fetal ultrasonography may play a role in determining the risk that the fetus will be born with genetic abnormalities, thereby helping couples who would like to continue pregnancy with the choice of termination or optimal postnatal management [5]. The treatment of CS is usually done by combined effort from different disciplines and early diagnosis is vital in order to avoid complications such as blindness and raised intracranial pressure. In the early life, adequate cranial volume can be redeemed for uncompromised brain growth by surgically releasing the synostotic skull sutures, with further skull reshaping procedures done throughout the child's growth period so optimal results are achieved. CS has good cosmetic results of plastic surgery have proven to be remarkably very satisfiable [5, 6]. Research and development of gene therapy in the field of craniosynostosis is considered to be desirable as it can help discover non-surgical managements and further expand new advancements [5-9].

**Conclusion:** In true hydrocephalus associated with Crouzon syndrome, ventriculo-peritoneal shunting is currently the single promising mode of treatment. But multidisciplinary approach is needed to solve other associated deformity associated with Crouzon's syndrome.

**Conflict of Interest and Financial Disclosure:** None.

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