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Case Report

# Premature Craniosynostosis in a Rare Genetic Disease- A Case Report

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**Abstract**

***Background:*** Crouzon syndrome is a rare genetic disorder inherited in autosomal dominant pattern with complete penetration and variable expressivity.Its most notable characteristic feature is premature synostosis of cranial sutures The case presented is of a 4 yr old boy with box like head with microcephaly, protuberant eyes, hydrocephalus, low visual acquity diagnosed as a case of crouzon syndrome after clinical and radiological assessment.

***Keywords:*** Crouzon syndrome, Autosomal dominant, Premature craniosynostosis

## Introduction

Crouzon syndrome is a rare genetic disorder inherited in autosomal dominant pattern with complete penetration and variable expressivity and a reported incidence of 1:25000 live births (1). Its pathophysiology lays in mutation in the fibroblast growth factor receptor 2 (FGFR2) gene (2). In some cases, an affected gene is inherited from one affected parent. Other cases occur sporadically because of newer mutations in the gene of normal people with negative family history for the disorder (3). This mutation in *FGFR2 gene* leads to premature fusion of cranial sutures and facial bones before the brain of the child has fully developed causing facial asymmetry, abnormal head shape and dental growth. The diagnosis is based on clinical grounds and radiology (4).

The case presented here is of a 4 years old boy-labeled as a case of crouzon syndrome after detailed clinical and radiological assessment.

## Case Report

A 4 years old boy presented to Pediatric Department of Baqai University Hospital with non-specific complains of fever, cough, loss of appetite and pallor since 2 months.

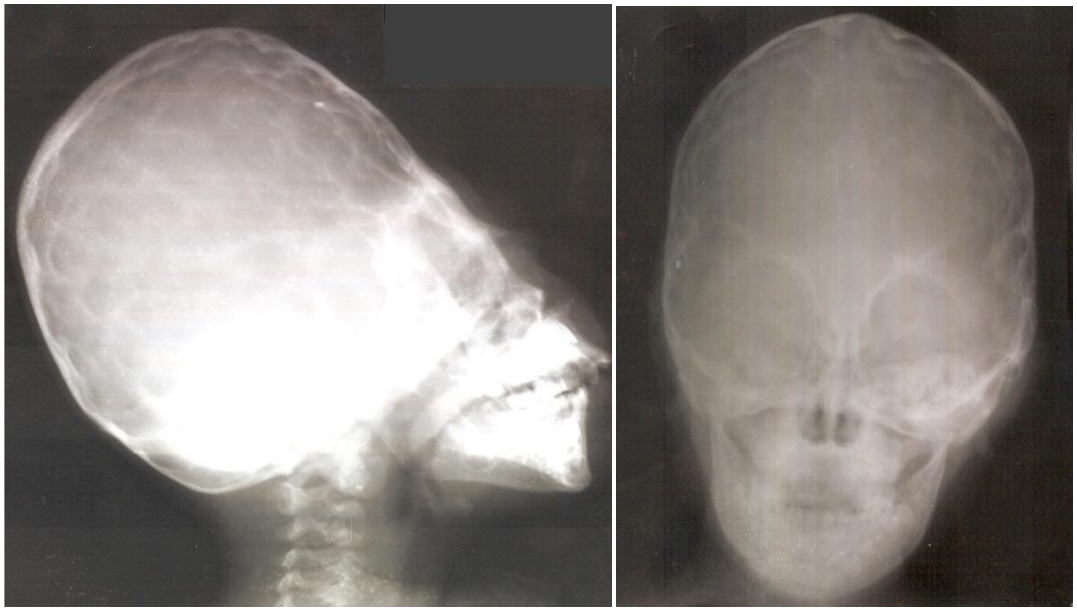
He was blind since birth, had delayed milestones and was not vaccinated due to family issues. His family history was negative for any genetic disease or syndrome.

On GPE examination, he was found anemic and have box like head with microcephaly and protuberant eyes. His anthropometric measurements were also considerably lower than normal indicating severe malnutrition. His CVS examination revealed some systolic murmur. His chest examination revealed HVB and crepitations. Rest of the systems appeared normal on examination. CBC

showed pancytopenia not related to crouzon syndrome.

X-ray of skull showed flattened frontal bones with high vertex due to premature fusion of coronal suture. There was copper beater appearance of skull vault bone indicating hydrocephalus as shown in Fig. 1a and b**.** Chest X-ray was normal with normal sized heart and no active lung lesions as shown in Fig. 2.

On ophthalmologic assessment, he had bilateral proptosis, 6$^{th}$ cranial nerve palsy, and low visual acuity of at least 1/60 on both sides. Besides, the median arc was clear, retina was normal and optic nerve was intact on both sides.



**a)** **b)**

**Fig. 1**: (a) X-ray skull lateral view; (b) X-ray skull AP view

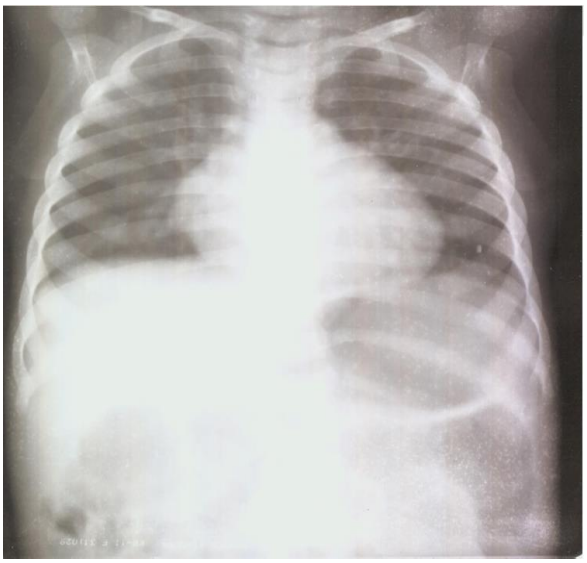


**Fig. 2:** X-ray chest AP view of a 4 year old boy diagnosed with crouzon syndrome

## Discussion

Crouzon syndrome is an autosomal dominant disorder with complete penetration and variable expressivity. However, it occurs sporadically in some cases. Its most notable characteristic feature is premature synostosis of coronal and sagittal sutures, which most often occur prior to birth. However, it can occur lately in infancy or early childhood period.

Closure of sutures prematurely reduces the growth potential at those suture sites on the skull limiting the growth of brain as well leading to hydrocephalus and mental retardation (5). Besides craniosynostosis it usually presents as brachycephaly (short and broad head), exopthalmos(due to shallow eye sockets), hypertelorism and beak-like nose. Hypoplastic maxilla also occurs in some affected

children due to insufficient growth of the midface. Hypoplastic maxilla and normal growth of mandible leads to relative mandibular prognathism (protrusion of chin) and gives the effect of the patient having a concave face. Other features may include short height, spinal malformations, dental problems, and high-arched, narrow or cleft palate.

Association of crouzon syndrome with congenital heart malformations is rarely reported and description of such congenital malformations coexistence is not accessible in literature (6).

Crouzon syndrome is usually diagnosed at birth on clinical grounds by assessing the signs and symptoms of the baby. Further analysis is aided by radiology and genetic testing.

Treatment of crouzon syndrome involves multidisciplinary team working in collaboration. Treatment is planned in view of the severity of skull's deformity and the child's individual needs. In the neonates, surgical release of the synostotic sutures is always the first priority to allow for adequate brain growth and expansion.

Crouzon syndrome patients also present with potential issues with respect to respiration, feeding, neurology such as hydrocephalus, and the potential risk of developmental delay that also needs to be addressed as a part of treatment.

Early surgical intervention and an appropriate treatment plan offers favorable prognosis with better functional and cosmetic outcomes.

## Ethical considerations

Ethical issues (Including plagiarism, informed consent, misconduct, data fabrication and/or falsification, double publication and/or submission, redundancy, etc.) have been completely observed by the authors.

## Acknowledgements

The authors declare that there is no conflict of interests.

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