

CASE REPORT

CROUZON SYNDROME: A CASE REPORT

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ABSTRACT: Crouzon's Syndrome is a rare autosomal dominant disorder. Normally, the sutures in the human skull fuse after the complete growth of the brain. But, if any of these sutures closes early then it may interfere with the growth of the brain. Premature sutural fusion most commonly involves sagittal suture followed by coronal suture. We report a case of 6-year-old male child presented with characteristic features of Crouzon's syndrome. Diagnosis was made on the basis of clinical and radiological findings.

KEYWORDS: Crouzon Syndrome, Exophthalmos, Craniosynostoses, and Fibroblast Growth Factor.

INTRODUCTION: The craniosynostoses are a rare group of congenital condition in which an abnormally shaped skull results from premature closure of the skull sutures. Crouzon's syndrome is a rare autosomal dominant disorder with complete penetrance and variable expressivity or can appear as a mutation.^[1] This syndrome is caused by mutations in the Factor Receptor-2 (FGFR2) gene, which is mapped on the chromosome locus 10q25-10q26, with variable expressivity; 50% of the incidence of Crouzon syndrome are not inherited and are the result of new mutation.^[2,3] It has an incidence of approximately 1 in 25,000 birth worldwide.^[4] It makes up approximately 48% of all cases of craniosynostoses, making it more common syndrome among craniosynostoses. No known sex or race predilection. The syndrome is characterized by premature synostosis of coronal and sagittal sutures, which usually begins in the first year of life. Once the sutures become fused, growth potential to those sutures is restricted. However, multiple sutural synostoses frequently extend to premature fusion of the skull base causing midfacial hypoplasia, shallow orbit, exophthalmos, maxillary hypoplasia, and occasional upper airway obstruction.^[5] Intraoral manifestations include mandibular prognathism, overcrowding of upper teeth, and V-shaped maxillary dental arch.^[6] Narrow, high, or cleft palate and bifid uvula can also be seen. Occasional oligo-dontia, macrodontia, peg-shaped, and widely spaced teeth have been reported.^[5,6] In this article we present a case of crouzon syndrome in a 6-year-old boy.

CASE REPORT: A 6 year-old boy with his parents presented to the outpatient department with diminution of vision in both eye and prominent eyes since birth. He was a full term baby, delivered by normal vaginal route in the hospital and cried immediately after birth. There was no history of consanguinity in parents. No other family member or sibling was affected with the same complaints. He was also fully vaccinated.

On examination, the child was oriented in time, place, and person. He was microcephalic with abnormal shape of skull and hypoplastic orbital ridges. On ocular examination, visual acuity was Counting Finger (CF) 6 Ft right eye and CF 4 Ft in left eye. Bilateral proptosis with a large

CASE REPORT

degree of exotropia in the left eye was seen in primary gaze and hypertelorism (Fig. 1). His cornea had no signs of exposure keratitis. Fundus examination revealed bilateral optic atrophy (Fig. 2).

Intraoral manifestations included mandibular prognathism (Fig. 3) and inverted V-shaped palate with dental abnormalities (Fig. 4). Facial features included a curved, parrot-like nose and mid-facial hypoplasia. There was no syndactyly seen which is a differentiating feature between Crouzon and Apert syndrome. (Fig. 5)

Computed tomography (CT) brain reveals bilateral proptosis with shallow orbits and narrow bilateral bony optic canals, supratentorial ventriculomegaly, bilateral maxillo-sphenoid sinusitis, craniosynostosis (Fig. 6). The final diagnosis of craniosynostoses, most likely Crouzon syndrome with ocular complications was made, on the basis of clinical and radiological findings.

No cardiac abnormality was detected.

DISCUSSION: Octave Crouzon (1912) was first to describe Crouzon syndrome. The Crouzon's Syndrome is the most frequent of the craniofacial diseases and is characterized for being a rare genetic disorder that can be diagnosed upon the birth or during the childhood. The dominant transmission range is of 100% and the large-scale penetrance with phenotypic expression is highly variable.^[7] It is responsible for about 4.8% of all the cases of craniosynostosis and is the most common syndrome of a group of more than 100 types of the craniosynostoses. Crouzon syndrome is caused by a mutation in the fibroblast growth factor receptor-2 (FGFR2) gene.^[2] Premature craniosynostosis, midface hypoplasia and exophthalmos account for the triad of Crouzon syndrome. Premature closure of cranial sutures, most commonly the coronal and sagittal ones, results in abnormal skull growth, affects the growth and development of the orbits and maxillary complex. Other clinical features include hypertelorism, exophthalmos, strabismus, short upper lip, beaked nose, maxillary hypoplasia and relative mandibular prognathism. Acanthosis nigricans is the main dermatologic manifestation of Crouzon syndrome. Optic atrophy is frequently seen and has been reported in 30-80% of patients. Increased intracranial pressure may lead to optic atrophy, which can produce blindness, if the condition is not treated.

The differential diagnosis of CS includes simple craniosynostosis as well as Apert syndrome, Carpenter syndrome, Pfeiffer syndrome and Saethre-Chotzen syndrome.^[8]

Early diagnosis and management of Crouzon's syndrome is important, and a multidisciplinary approach is required. 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. Skull reshaping may need to be repeated as the child grows to give the best possible results.^[4,9] Prophylactic dental care is essential, and some teeth may need to be extracted to relieve crowding as the child develops. Functional space maintainer can also be used. Placement of orthodontic braces to expand the palate and align the teeth is often required. Preventive regimen consists of regular follow-up.

Speech therapy can help children learn to communicate. Supportive treatment includes special education for children with mental retardation. People with this syndrome usually have a normal lifespan.

CASE REPORT

As our patient reported late and was diagnose with bilateral optic atrophy, his visual acuity was compromised.

To conclude an early detection of Crouzon syndrome is a precondition for the reduction of amblyopia by means of refractive error correction and an appropriate treatment of strabismus, while optic nerve atrophy remained the main cause of the decrease of a patient's visual acuity.



Fig. 1: Bilateral Proptosis with hypertelorism and divergent squint

Fig. 2: Fundus examination reveals optic atrophy in (BE)

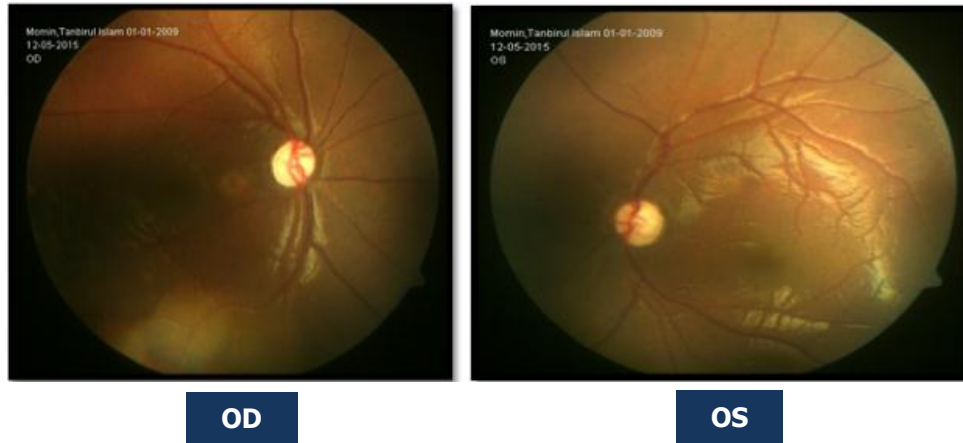


Fig. 3: Mandibular prognathism

CASE REPORT



Fig. 4: High arched palate with dental abnormalities



Fig. 5: Normal fingers with no syndactyly

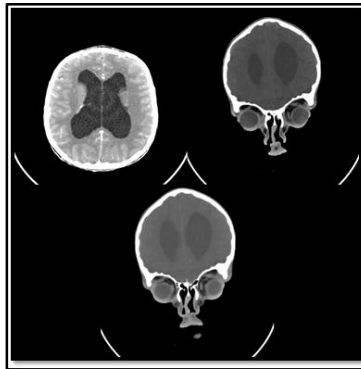


Fig. 6: CT brain shows shallow orbits and supratentorial ventriculomegaly

REFERENCES:

1. Fogh-Andersen P. Craniofacial dysostosis (Crouzon's disease) as a dominant hereditary affection. *Nord Med.* 1943; 18: 993–6.
2. Bowling EL, Burstein FD. Crouzon syndrome. *Optometry* 2006; 77: 217-22.
3. Arnaud-Lopez L, Fragoso R, Mantilla-Capacho J, BarrosNúñezP. Crouzon with acanthosis nigricans. Furtherdelineation of the syndrome. *Clin Genet* 2007; 72: 405-10.
4. Cohen MM Jr. Craniosynostosis update 1987. *Am J Med Genet Suppl* 1988; 4: 99-148.
5. Hlongwa P. Early orthodontic management of Crouzon Syndrome: A case report. *J Maxillofac Oral Surg* 2009; 8: 74-6.
6. Crouzon LE. Dysostose cranio-faciale héréditaire. *Bulletin de la Société des Médecins des Hôpitaux de Paris* 1912; 33: 545-55.
7. Fogh-Andersen P. Craniofacial dysostosis (Crouzon's disease) as a dominant hereditary affection. *Nord Med.* 1943; 18: 993–6.
8. Regezi JA, Sciubba JJ. 4th ed. Philadelphia: W.B. Saunders co; 1999. Oral pathology–Clinical pathologic correlations; pp. 477–8.
9. Jarund M, Lauritzen C. Craniofacial dysostosis: Airway obstruction and craniofacial Surgery. *Scand J Plast Reconstr Surg Hand Surg* 1996; 30: 275-9.

CASE REPORT

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