Case Report

Crouzon's syndrome in son and father

D K Sindal[1], V H Karambelkar[2], Smita Javadekar[3], B S Joshi[4], Vikrant Narwade[5*]

[1]Professor and HOD, [2]Professor, [3,4]Associate Professor, [5]Residents, Department of Ophthalmology, KIMSDU, Malkapur, Karad, Maharashtra, INDIA.
Email: drvikrant.narwade@gmail.com

Abstract

Crouzon's syndrome is an autosomal dominant disorder with complete penetrance and variable expressivity. Crouzon's syndrome is caused by mutation in fibroblast growth factor receptor 2 gene. The disease is characterised by premature synostosis of coronal and sagittal sutures which begins in first year of life. Case report of son and father is presented with characteristic features of Crouzon's syndrome.
Keywords: Crouzon syndrome, craniosynostosis, exopthalmos.

[*]**Address for Correspondence:**
Dr. Vikrant Narwade, Residents, Department of Ophthalmology, KIMSDU, Malkapur, Karad, Maharashtra, INDIA.
Email: drvikrant.narwade@gmail.com
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INTRODUCTION

Crouzon syndrome accounts for approximately 4.8% of all cases of craniosynostosis, with a prevalance approximately 1 per 25,000 live birth worldwide[1]. It may be transmitted as an autosomal dominant genetic condition, but 25% of cases represent a fresh mutation. The molecular analysis of craniosynostosis syndrome identifies mutation in fibroblast growth factor receptor 2 (FGFR 2) gene, which is mapped on chromosome locus 10q25-q26[2]. Molecular analysis of FGFR gene provides useful information and helps with conforming diagnosis and performing prenatal diagnostics. Crouzon syndrome has no sex or race predilection. The appearance of crouzon syndrome can vary in severity from a mild presentation with subtle midface features to severe forms with multiple fused cranial sutures and marked midface and ocular problems. Varieties of the ophthalmic findings includes: proptosis-due to the shallow orbits, ocular hypertelorism- the consequences of the widening of the sphenoid bone and root of the nose , strabismus-exotropia, V syndrome, hypertrophy, which together with maxillary hypoplasia and protrusion of lower jaw. There have also been rare occurrences of aniridia, anisocoria, blue sclera, cataract, ectopialentis, glaucoma, megalocornea, nystagmus and optic nerve hypoplasia[3] Ocular complications of Crouzon Syndrome include: chronic papillary edema, as a result of increased intracranial pressure with consequential optic atrophy; keratopathy, as a consequences of qualitative changes of tear film due to exposure to the climatic factors.

CASE REPORT

A seven year male child along with his father from Babarmati was sent to the ophthalmology OPD for evaluation of proptosis. The information was provided by his father: the boy had a big eyes since birth (figure 1), three years ago he had head trauma after which one surgery was done and after surgery swelling over forehead starts appearing which is cystic, non-pulsatating, nonreducible (figure 2). Father is also having same big eyes since his birth (figure 4 and 5). The ophthalmological examination was carried out in accordance with standard procedures O/E: Vision (right eye): 6/18, (left eye): 6/18, anterior segment (right eye): mild conjunctival congestion present, anterior chamber normal depth and clear, pupil: normal size and reacting to light. patient had bilateral proptosis with exotropia. Onexophthalmometry (Right eye): 25 mm,(Left eye):27 mm, Non reducible, non pulsatating, No bruie heard. Fundus examination revealed pale disc with mylinated nerve fibre and foveal reflex dull in both eyes. Forehead: Large cystic swelling present over forehead which is non-pulsating, non-reducible.

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Figure 1:

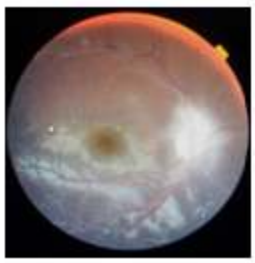
Figure 2:

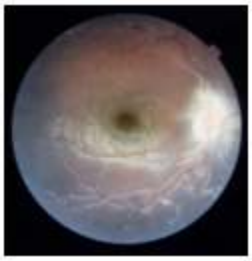
Figure 3:

Figure 4:

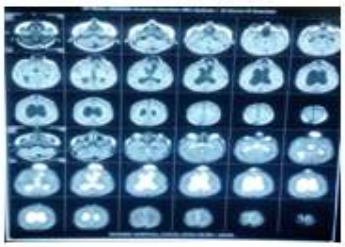
Figure 5:

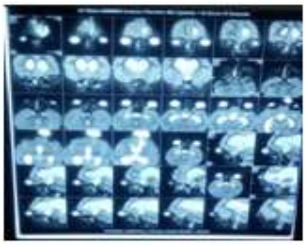
FUNDUS IMAGES





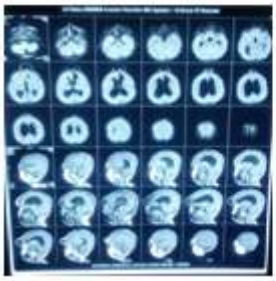
MRI ORBIT

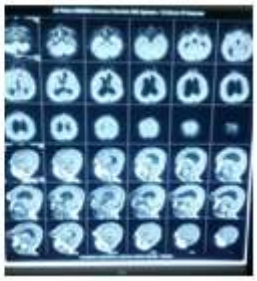




*Proptosis with widely spaces orbits (indicating hypertelorism).
*Prominant subarachnoid spaces noted along bilateral optic nerves with tortuous course bilaterally.....could be secondary to raised intracranial pressure.

MRI BRAIN





*Gross dilatation of bilateral vetricles, 3rd ventrical suggestive of hydrocephalus. 4th ventrical appears normal.

*Herniation of brain parenchyma along with CSF is seen through the defect in the frontal bone most likely suggestive of encephalocele. Assymetrical dilatation of the frontal horn of left lateral ventricale which is seen herniating anteriorly through the defect in the cranial vault.
*Brachicephaly with assymetric and irrgularthining of cranial vault.
*Thining of corpus callosum.

After the full examination, the child was diagnosed with crouzon syndrome. The child was prescribed artificial tears eye drops and gel for the future topical use . The father was explained the nature of the child's disease and the consequential poorsightedness. The child was sent and recommended that he should undergo the surgical treatment. We advised him to regular follow up to neurosurgeon, ophthalmologist, oral and maxillofacial surgeon for proper management and to minimize the complications.

DISCUSSION

Congenital malformation of orbit are usually a part of complex syndrome which affects the bones of the skull and face. Premature craniosynostosis, midface hypoplasia and exophthalmos accounts for the traid of crouzon syndrome. Premature closure of cranial sutures, most commonly coronal and sagittal ones results in abnormal skull growth, affects the growth and development of orbit and maxillary complex. Other clinical features includes hyperteleorism, exophthalmos, strabismus, beaked nose, short upper lip hypoplsia, relative mandibular prognathism. Some other feature commenly seen in these patients are visual disturbances related to imbalance of extraocular muscles, congenital damages due to exopthalmos, refractive error (usually hypermetropic astigmatism), optic nerve complication, hearing loss, ear canal stenosis or atresia. The intelligence of crouzon syndrome patients is usually in normal range. In order to diagnose crouzon syndrome, thorough ophthalmological examination, radiological as well as genetic analysis is needed. The most common differential diagnosis of crouzon syndrome is Apert syndrome characterised by isolated coronal synostosis and syndactaly of all limbs[4].

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