

Crouzon Syndrome with Ocular Abnormalities: A Case Report

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ABSTRACT

Crouzon syndrome is a rare genetic disorder characterized by premature closure of cranial sutures, exophthalmos, beak-like nose and mid facial hypoplasia. It was initially described as hereditary syndrome of craniofacial synostosis. There is premature fusion of one or more cranial sutures in craniosynostosis leading to skull deformities. Craniofacial abnormalities are usually present at birth and may progress with time. The most common presentation is brachycephaly but the timing and order of suture fusion contributes to

the shape of head. The orbits are shallow resulting in ocular proptosis with or without divergent strabismus. Hypoplasia of maxilla with curved parrot like nose and orbital hypertelorism are typical facial features. The diagnosis and management of craniofacial abnormalities have always been a challenge therefore understanding of these abnormalities is necessary to monitor subsequent growth and to ensure that the patient receives the best available care. Herein, we report this rare entity of Crouzon syndrome showing characteristic features with ocular abnormalities.

Keywords: Craniosynostosis, Exophthalmos, Facial anomalies

CASE REPORT

A five month old male child presented to departmental OPD with complaint of outward protrusion of both eyeball present since birth. Detailed family and medical history was taken which revealed mother had normal hospital based vaginal delivery. In family, sibling and near relatives are normal. Examination and photos are taken after taking prior consent from parents.

On examination, it was noted that head was elliptical in shape with beak-like nose, broad nasal bridge, high arched palate and low set of ears.

On local examination, patient had bilateral proptosis, hypertelorism, divergent squint both eyes. Anterior

segment including pupil was normal. Fundus showed clear media with hyperemic disc, absence of physiological cup, engorged and tortuous vessels with normal macular reflex [Table/Fig-1-3].

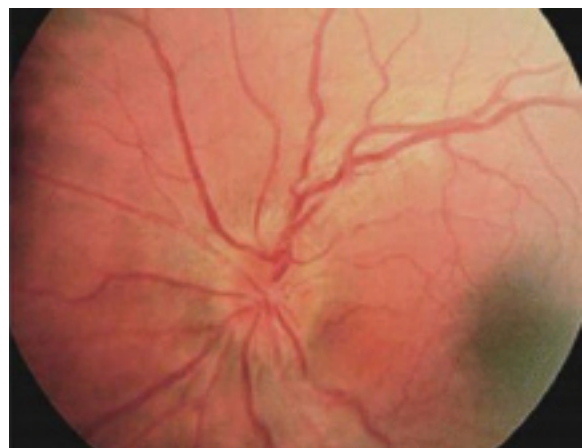
There was no distal abnormality and systemic examination was also normal as reported by Paediatrician.

He was diagnosed as a case of Crouzon syndrome with these ocular abnormalities on clinical basis. Patient was started on lubricating eye drops and ointment to prevent exposure keratopathy and advised close follow-up. Child was referred to Paediatrician and Neurosurgeon for further management.



[Table/Fig-1]: Showing B/L proptosis, hypertelorism, broad nasal bridge and beak nose.

[Table/Fig-2]: Showing elliptical shaped head, beak nose and low set ears.



[Table/Fig-3]: Showing fundus picture with hyperemic disc, absence of physiological cup, engorged and tortuous vessels.

DISCUSSION

Octave Crouzon in 1912 first described Crouzon's syndrome as one of the varieties of craniofacial dysostosis caused by premature closure of two or more sutures, most often coronal and sagittal. It is a rare genetic disorder with autosomal dominant inheritance with the prevalence of 16.5 per million newborns and it constitutes 4.8% of all craniosynostosis [1]. Crouzon syndrome is characterized by deformities of skull, facial hypoplasia and ocular proptosis [2]. More than 90% of cases occur due to mutations in fibroblast growth factor gene (FGFR2) which maps to chromosome 10p 25-q26 [3].

The most common ocular abnormalities are shallow orbits, ocular proptosis, orbital hypertelorism, strabismus, papilloedema, optic atrophy, exposure keratitis and visual loss. Other ocular abnormalities like nystagmus, iris coloboma, aniridia, anisocoria, microcornea, megalocornea, cataract, ectopia lentis, blue sclera, glaucoma and luxation of the eye have been seen in rare cases [4]. Oral abnormalities include short upper lip, hypoplastic maxilla, relative mandibular prognathism, cleft palate and bifid uvula. Among cardiovascular abnormalities, patent ductus arteriosus and aortic coarctation are associated with Crouzon syndrome. Hydrocephalus, seizures and mental retardation may occasionally present in this syndrome [5]. The main dermatological manifestation is acanthosis nigricans and is seen in 5% of cases with Crouzon syndrome [6].

The differential diagnosis of Crouzon syndrome includes Apert syndrome, Carpenters syndrome, Saethre-Chotzen syndrome, Pfeiffer syndrome [7]. There is lack of hand or foot abnormalities in Crouzon's syndrome which distinguish it from other craniosynostosis syndromes.

Various test like MRI, genetic testing, X-rays and CT-scans can be used to confirm the diagnosis. Molecular testing is more accurate and reliable than ultrasonography in diagnosing Crouzon syndrome [8]. CT-scan brain shows signs of raised intracranial pressure, fusion of coronal and sagittal sutures and 3D images will reveal copper beaten appearance.

Management of such patients requires multidisciplinary approach and early diagnosis is important. Increased intracranial pressure leading to optic atrophy may

occur, which produce blindness if the condition is not treated. The primary measure of treatment is to minimize intracranial pressure and release of prematurely fused sutures during the first year of life by a neurosurgeon for proper brain growth and expansion [9]. These primary measures and newer techniques like cosmetic reconstruction of facial bones allow the patient to have normal life span.

Our patient showed characteristic features of Crouzon syndrome and was reported much earlier though had earlier signs of raised intracranial pressure without any sequelae of optic atrophy hence was referred to neurosurgeon for timely management and to prevent any further sequelae of dysostosis.

CONCLUSION

Early detection of Crouzon syndrome and prevention of ocular complications is required to reduce the associated visual loss as optic atrophy remains an important cause of visual impairment in these patients. Timely decompression may prevent optic neuropathy and any visual loss.

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