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A Rare case of CROUZON SYNDROME in a 4-Month-Old baby: Case Report

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Abstract: *Crouzon syndrome is an autosomal dominant disorder characterized by craniosynostosis, exophthalmos and hypoplastic maxilla with relative mandibular prognathism. Crouzon syndrome is caused by mutation in the fibroblast growth factor receptor-2 (FGFR-2) gene. The present article describes a case report of a 4-Month-old male baby with characteristic skeletal, facial and ocular features of Crouzon syndrome.*

Key words: *Craniofacial dysostosis, craniosynostosis, rouzon syndrome, exophthalmos*

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I. Introduction

Crouzon syndrome is a rare autosomal dominant genetic disease with an incidence of 1 in 25,000 live births. This syndrome is named after Octave Crouzon, a French physician. It is also known as craniofacial dysostosis and acrocephalosyndactyly Type II.

It is characterized by a triad of skull deformities (due to premature closure of cranial sutures: craniosynostosis), midface hypoplasia with mandibular prognathism and ocular abnormalities usually manifesting as exophthalmos. It is an autosomal dominant disorder with complete penetration but variable expressivity. It arises due to mutation in FGFR2 gene. The growth of the skull and brain are impaired in a direction perpendicular to the fused sutures, giving rise to the craniofacial abnormalities.

II. Case Report

A 4-month-old male child was brought with the complaints of abnormal facies, abnormal skull shape and protruding eyeballs since birth, with left eye protrusion being rapidly progressive in the last 10 days. Antenatal history and birth history were non-contributary. This baby is the first child born to non-consanguineous parents. There is no hereditary background and parents appeared normal.

On general examination, brachycephaly, tower skull with steep forehead(figure 1), hypertelorism, marked proptosis(figure 2), depressed nasal bridge with parrot beak-like nose, maxillary hypoplasia, mandibular prognathism, high arched palate and cleft palate(figure 3) were observed.

On ophthalmic examination, eyes were grossly proptosed – Right eye 25mm and Left eye 28mm with Luedde's exophthalmometer(figure 4).

Figure 1:

Figure 2:

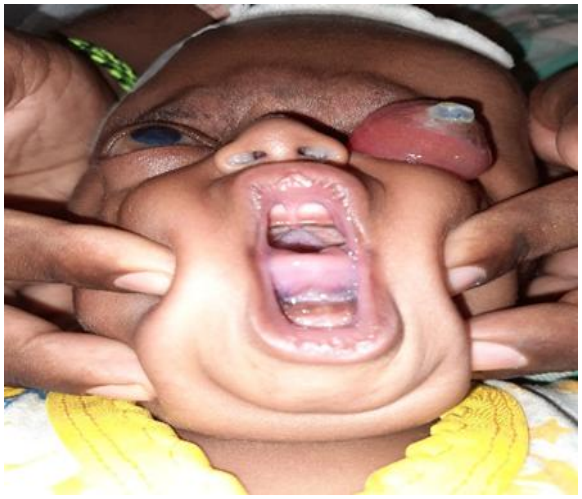

Figure 3:

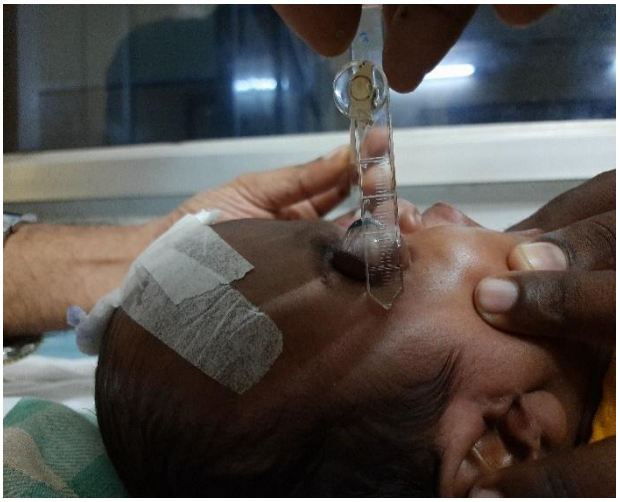

Figure 4:

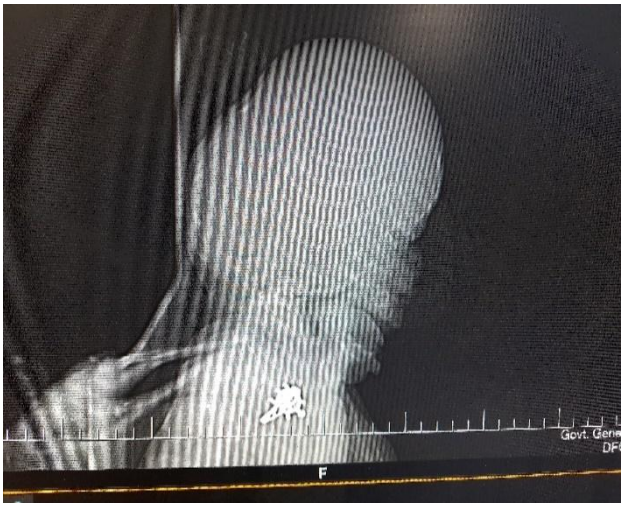

Figure 5:

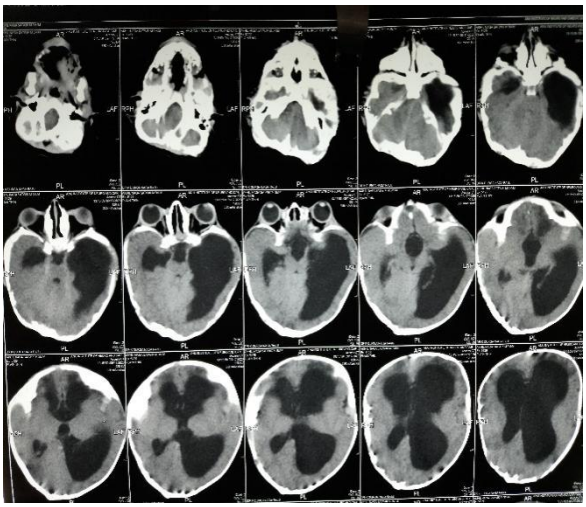

Figure 6:

Ocular motility appears to be grossly restricted in both eyes.
The palpebral aperture of the right eye was wide and the bulbar conjunctiva in the lower part was dry and mildly congested. The cornea showed early signs of exposure keratopathy. The pupil reacted sluggishly to light. On fundoscopy, optic disc pallor is seen.
The palpebral aperture of the left eye was wider with dryness and marked severe congestion of entire bulbar conjunctiva. The cornea showed signs of exposure keratitis, with an oval corneal ulcer with irregular margins involving the entire cornea. Pupil and fundus details couldn't be seen.

Investigations: Except for the hemoglobin being 10.9 gm% and WBC count 18,700 cells/cumm, all other routine blood investigations were non-contributory.
Roentgenographs revealed asymmetrical calvarial thickening and oxycephaly(figure 5)
Cranial CT showed non communicating hydrocephalus with colpocephaly, and with convolutional markings in the inner table of skull(figure 6)
MRI scan showed bilateral proptosis and posterior half of left lateral ventricle grossly dilated due to left foramen of Monro obstruction.
The screening of chest, spine and limbs showed no abnormalities.

Diagnosis: Based on the above mentioned clinical and radiographic features, the diagnosis of craniofacial dysostosis syndrome, most likely Crouzon syndrome was made.

Treatment: Parents were adequately counselled about the nature of the disease and complications. Conservative treatment with antibiotic eyedrops, atropinization and Lubricant eye gel were prescribed. Even Eyelid taping was not possible because of extreme proptosis.

The child was referred to neurosurgeon for hydrocephalus, for which the baby underwent Ventriculo-Peritoneal shunting procedure, and regular follow-ups were advised regarding management of any sequalae of raised ICT.

The child was also referred to oculoplastic surgeon to consider any temporary measures for protection of cornea and proptosis correction. Guarded visual prognosis was explained to the parents regarding visual outcome in left eye and were advised regular follow-up visits.

The child was further referred to oral and maxillofacial surgeon for correction of cleft palate and other sequalae.

III. Discussion

Crouzon syndrome, a rare genetic disorder, accounts for 4.8% of all craniosynostosis. It was first described by a French neurosurgeon Louis Edouard Octave Crouzon in 1912, as a hereditary craniosynostosis syndrome, with a triad of skull deformities, facial anomalies, and exophthalmos. Additionally, cleft lip, cleft palate, and bifid uvula are known associations.

It is an autosomal dominant disorder with complete penetration but variable expressivity. It arises due to mutation in FGFR2, which is mapped to chromosome locus 10q25-10q26, causing skull bones to fuse prematurely. Locus heterogeneity with mutation in FGFR3 genes results in Crouzon syndrome with acanthosis nigricans.

The diagnostic radiographic features of Crouzon syndrome are premature craniosynostosis, with characteristic presence of craniofacial anomalies, with absence of digital and limb anomalies. Premature craniosynostosis usually involves either coronal or lambdoid and occasionally the sagittal sutures. The abnormal process may also extend to involve sutures at the skull base. The latter explains mid face hypoplasia and upper airway obstruction. Occasional spine deformities, such as craniovertebral junction abnormalities, butterfly vertebrae, and fused cervical vertebrae have been reported. Therefore, radiographs of limbs and spine are important, in addition to skull radiographs, for definitive diagnosis.

Anteroposterior and lateral skull radiographs and CT not only reveal abnormal skull shape but also show sclerosis and fusion of sutures. Other radiographic features of craniofacial bones are hammered silver beaten/copper beaten skull vault, enlarged hypophyseal cavity, shallow orbits, small paranasal sinuses, and hypoplastic maxilla.

The differential diagnosis of Crouzon syndrome includes Crouzon syndrome with acanthosis nigricans, Pfeiffer's syndrome, Apert syndrome, Saethre–Chotzen syndrome, Carpenter syndrome, Treacher Collins syndrome and Jackson–Weiss syndrome. These syndromes show presence of limb and digital abnormalities, unlike Crouzon syndrome.

The treatment of Crouzon syndrome patients begins during the child's first year of life, with cranial decompression, for prevention of raised intracranial pressure, to prevent mental retardation and impaired vision. Multi - disciplinary and multistage surgeries are recommended for management of Crouzon Syndrome.

IV. Conclusion

Management of craniofacial deformities often requires multidisciplinary team involvement. Clinicians must be able to recognize the characteristic features of the various craniofacial syndromes so that an early diagnosis and referral for specialized care can be done. Craniofacial syndromes, if diagnosed at any early stage, can be managed properly, thereby protecting the eyeball and preserving the visual function and also the need for major facial surgeries can be avoided, additionally benefiting the psychological development of the child and reducing the financial and mental burden on the parents.

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