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CROUZON SYNDROME- A RARE CASE REPORT

Medical Science

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ABSTRACT

We report the case of a major neonatal form of Crouzon syndrome in a newborn of 21 days with no history of consanguinity.

Clinical manifestations were marked by abnormal development of head and face associated with dyspnoea.MDCT brain showed craniosynostosis with widely spaced orbits and retrognathia. 2D-ECHO showed presence of congenital heart disease (PDA).

The major forms of Crouzon syndrome is a source of complications including exposure keratopathy and optic atrophy requiring ophthalmological caring in awaiting for a craniofacial surgery.

Introduction:

Crouzon 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 factor receptor- 2 [FGFR2] gene,which is mapped on chromosome locus 10q25.5-q26,with variable expressivity. 50% of the incidence of Crouzon syndrome are not inherited and are the result of new mutations. [2,3] It accounts for about 4.8% of all cases of craniosynostosis and occurs in approximately 1.6/100,000 live births worldwide. [4,5] They are a rare group of congenital condition in which abnormally shaped skull results from premature closure of the skull sutures.

CASE REPORT

A 21-day old female newborn presented to the OPD with prominent eyeballs since birth. Mother also complained of dyspnoea and poor feeding in the child.

She was a full term baby delivered by normal vaginal route in the hospital and did not cry immediately after birth. There was no history of consanguinity in parents. No other family member or siblings had similar findings.

On examination, the child had abnormal appearance of head and face with short and broad head (brachycephaly), flat nasal bridge, curved parrot like nose,maxillary hypoplasia, high arched palate and low set ears. On ocular examination,bilateral proptosis and hypertelorism was seen. Cornea had no signs of exposure keratitis. Fundus examination was within normal limit. There was no syndactyly which is a differentiating feature between Crouzon and Apert syndrome [fig.1]. Intraoral manifestations included retrognathia and high arched palate. On systemic examination there was increase in respiratory rate and heart rhythm was irregular. Bilateral crepitations were present.

USG revealed large eyeballs [fig.2]. MDCT brain showed craniosynostosis with shallow widely spaced orbits and retrognathia[fig.3].2D-ECHO suggested the presence of congenital heart anomaly(PDA) [fig.4]

Figure 1: Child with crouzon syndrome showing proptosis, mid facial hypoplasia and absence of syndactyly.

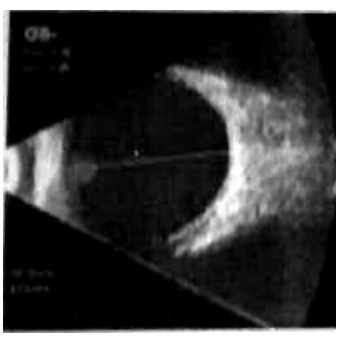

Figure 2:USG showing large eyeballs.

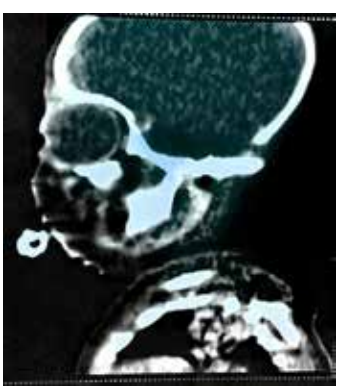

Figure 3:MDCT showing swallow widely spaced orbits with retrognathia.

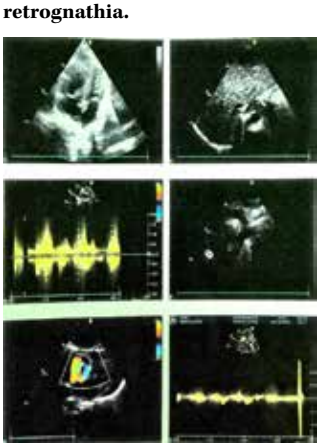

Figure 4:2D-ECHO showing congenital heart disease (PDA).

The karyotype could not be assessed after refusal by parents.
The final diagnosis of craniosynostosis, most likely Crouzon syndrome was made on the basis of clinical and radiological findings.

DISCUSSION

Octave Crouzon (1912) was first to describe Crouzon syndrome. [6] It is caused by mutation in fibroblast growth factor receptor-2 gene.

Premature craniosynostosis, midface hypoplasia and exophthalmos account for the triad of Crouzon syndrome. Premature closure of cranial sutures results in abnormal skull growth which affects the development of orbits and maxillary complex and may cause occasional airway obstruction. Other clinical features include hypertelorism, exophthalmos, strabismus, short upper lip, beaked nose, maxillary hypoplasia and relative mandibular retrognathism. Acanthosis nigricans is the main dermatological manifestation. Optic atrophy due to increased intracranial pressure is seen in 30-80%of patients. It can lead to blindness if not treated. Heart and visceral complications are also described in this syndrome.

The differential diagnosis of Crouzon syndrome includes Simple craniosynostosis, Apert syndrome, Carpenter syndrome, Pfeiffer syndrome and Saethre-chotzen syndrome. [7]

The appearance of an infant with Crouzon syndrome can vary in severity from mild presentation with subtle mid face deficiency to severe forms with multiple cranial sutures fused and marked midface and eye problems. Upper respiratory tract obstructions can lead to acute respiratory distress. In most congenital forms as in our case, maxillary hypoplasia is responsible for respiratory difficulties. Absence of inbreeding parents suggests a sporadic case.

Early diagnosis and management of Crouzon 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 and brain growth. Skull reshaping may need to be repeated as the child grows to give the best possible results. [8]If necessary mid facial advancement and jaw surgery can be done to reduce the exophthalmos and to correct occlusion to an appropriate functional position.Prognosis depends on malformation severity. [9] Prophylactic dental care is essential. Supportive treatment includes special education for children with mental retardation.Preservation of cornea is essential while awaiting craniofacial surgery and involves construction of external tarsorrhaphy.

To conclude an early detection of Crouzon syndrome contributes to the reduction of ophthalmic complications. A proper understanding of these abnormalities can ensure that the patient receives the best available care.

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