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Crouzons syndrome: A case report

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Abstract

Human skull is made up of many bone joints connected by sutures. The sutures fuse in later life after the complete growth of the brain. If any of these sutures closes early, it may interfere with the normal growth of the brain. The developing brain may exert pressure on the skull and may grow in the direction of the other open sutures. Premature sutural fusion may occur alone or together with other anomalies, making up various syndromes. Crouzons syndrome is an example of such a syndrome that is associated with premature synostosis of the sutures of the skull. Presented in this article is a case of Crouzons syndrome seen in a boy aged 9 years.

Key words: Craniofacial syndromes, Crouzon, premature synostosis

Introduction

The skull is composed of many bones that are separated by sutures. These sutures allow the skull to expand and develop in synchrony with the growth of the brain. If one or more of the sutures close early, especially before the complete growth of the brain, the developing brain may exert pressure on the skull and may grow in the direction of the other open sutures. This can result in an abnormally shaped head and in severe cases can also cause increased pressure on the growing brain.

Premature synostosis (fusion of the suture) commonly involves the sagittal and coronal suture. Lambdoidal sutures are occasionally involved. The order and rate of suture fusion determines the degree of deformity and disability.[1,2] Various sutures may be prematurely synostosed, and multiple sutural involvement is found eventually in most cases.[3]

Premature sutural fusion may occur alone or together with other anomalies, making up various syndromes.[4]

In this article we present Crouzons syndrome, which is one of the syndromes associated with synostosis of cranial sutures and is seen 1 in 60,000 persons. The differential diagnosis of Crouzons syndrome includes simple craniosynostosis as well as the Apert, Pfeiffer and Saethre-Chotzen syndromes. Unlike the other forms, Crouzons syndrome presents with no digital abnormalities.[5]

Case Report

A 9-year-old male child with normal intelligence studying in standard five visited the Department of Pedodontics and Preventive Dentistry for routine dental checkup through the school dental health program. His appearance was different from other children of his age, with protruding eyes and enlarged calvarium. History from the parents revealed that these features started developing since he was a small child, and the severity has gradually increased. No positive familial history was obtained.

On careful examination, following features were identified [Figure 1]:

1. Exophthalmos
2. Strabismus
3. Hypertelorism
4. Irregularly shaped vault with left side of the head enlarged, involving the sagittal and coronal sutures
5. Relatively large mandible
6. Retruded maxilla, resulting in midface retrusion
7. Parrot beak nose
8. Asymmetrical enlargement of the skull
9. Everted lower lip
10. Anterior cross bite with posterior open bite
11. Deep and narrow palate
12. No digital abnormalities were seen
13. Reduced vision
14. No dental aplasia was present

X-ray report

Radiographs of the skull [Figures 2A and 2B] revealed obliteration of sagittal and coronal suture lines with obvious bony continuity. A hammered-silver ('beaten metal/ copper beaten') appearance was seen in the regions of the skull due to compression of the developing brain on the fused bone.

Features of the CT scan were as follows [Figure 3]

1. Craniosynostosis with premature closure of sutures
2. Shallow orbits with proptosis
3. Hypoplastic maxilla and zygoma
4. Nonpneumatized right middle ear cavity with acellular mastoids
5. Moderate degree of hydrocephalus with diffuse

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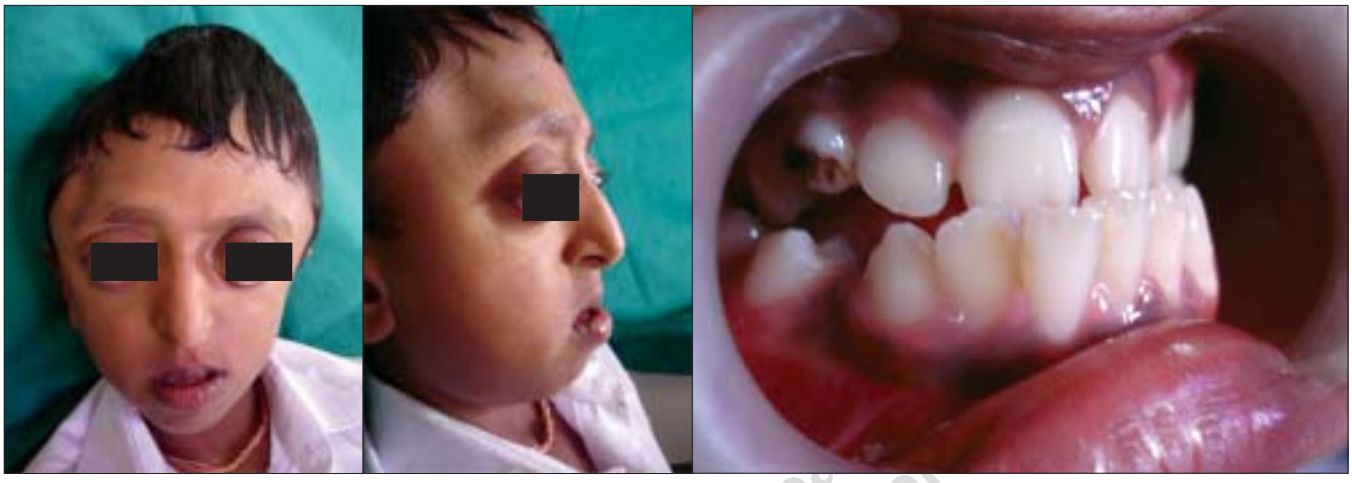

Figure 1: Frontal, lateral and occlusal view showing the typical features of Crouzons syndrome

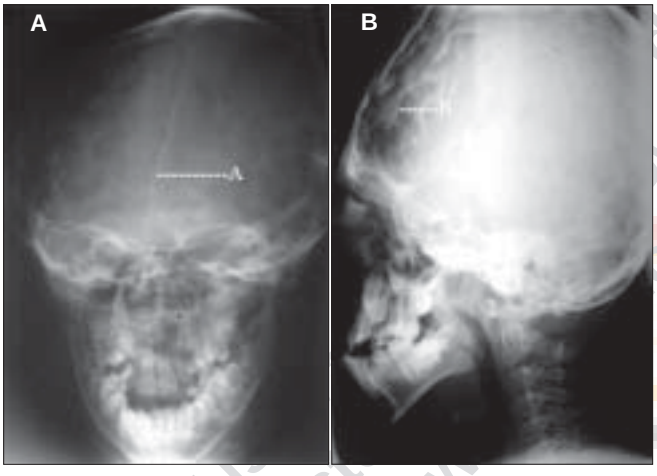

Figure 2: A. Obliteration of sagittal suture lines with obvious bony continuity. B. Hammered-silver ('beaten metal/ copper beaten' appearance) appearance seen in the regions of the skull

indentation of inner table of skull and narrowing of diploic space and prominent convolutional margins [Figure 4].

Treatment plan

A syndrome due to the complexity of symptoms always demands a multidisciplinary approach for successful outcome. The aim of treatment in this case is to relieve the intracranial pressure, which can be done by creating drainage for cerebrospinal fluid, and to retroposition the globe to allow adequate eye closure and prevent exposure keratitis and restore facial symmetry.

The treatment plan for this patient was divided into different stages as follows:

1. Performing a ventriculoperitoneal shunting (V-P shunt), which will be done by a neurosurgeon
2. Cranial vault contouring and monobloc advancement of midface with LeFort I advancement
3. Orthodontic correction of the occlusion

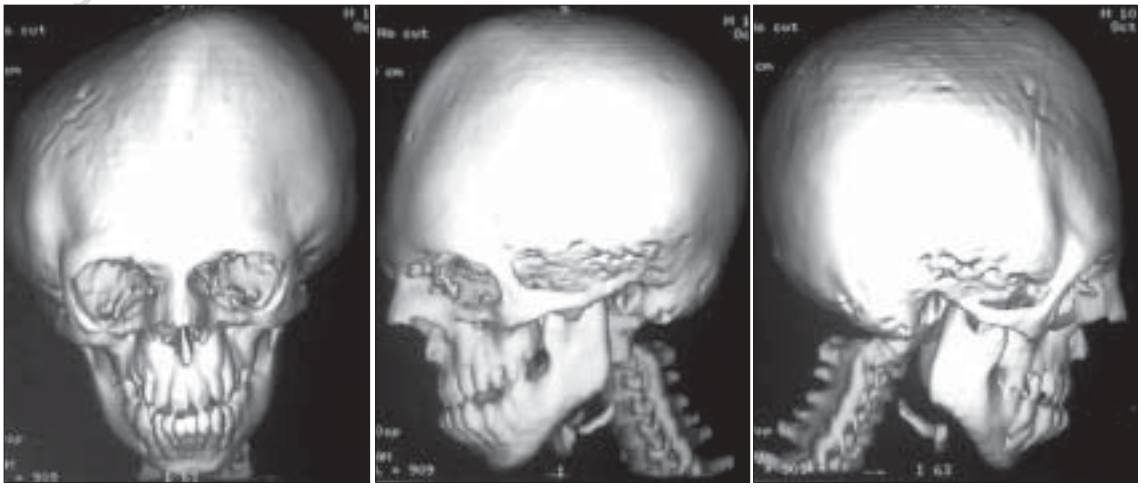

Figure 3: Frontal, left and right lateral CT scan of the skull revealing obliteration of sagittal and coronal suture lines with obvious bony continuity

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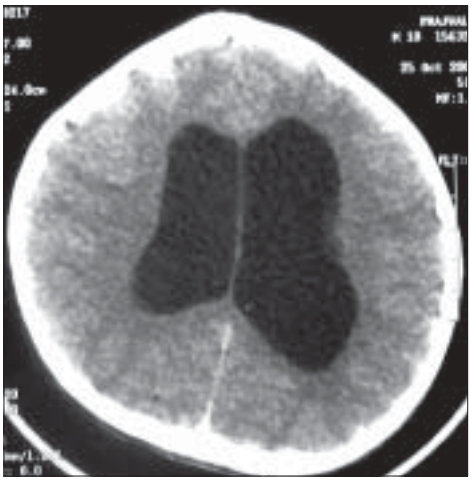

Figure 4: Moderate degree of hydrocephalus with diffuse indentation of inner table of skull as seen in the CT scan

Discussion

First described by Octave Crouzon (1874-1938), who was a French neurologist in 1912, Crouzon syndrome is a genetic disorder, characterized by abnormal fusion between bones in the skull and face, resulting in an abnormally shaped head and face. The phenotypic features of Crouzon syndrome may be absent at birth and evolve gradually during the first few years of life.[6,7]

It is commonly inherited as an autosomal dominant trait, with complete penetrance and variable expressivity, but about one-third of the cases do arise spontaneously. The male-to-female preponderance is 3:1.[2,3]

With the advent of molecular technology, the gene for the Crouzons syndrome could be localized to the Fibroblast Growth Factor Receptor II gene (FGFR2) at the chromosomal locus 10q 25.3-q26, and more than 30 different mutations within the gene have been documented in separate families.[8]

Premature fusion of the cranial sutures results in craniosynostosis, and this initiates changes in the brain and adjoining structures, such as increase in intracranial pressure, reduced orbital volume, exophthalmos (proptosis), severe maxillary hypoplasia and occlusal derangement.[5]

Complications of Crouzons syndrome may include conjunctivitis or keratitis, luxation of the eye globes, exotropia, poor vision due to optic atrophy and corneal injury, blindness. Frequent headaches, seizures, mental deficiency, increasing hydrocephaly, conductive hearing deficit, upper airway obstruction develop secondary to septal deviation, midnasal abnormalities, choncal abnormalities and nasopharyngeal narrowing, Others include nystagmus, iris coloboma, aniridia, anisocoria, corectopia, microcornea, megalocornea, keratoconus, cataract, ectopia lentis, blue sclera and glaucoma.[3]

Ultrasonic prenatal diagnosis of exophthalmos has been reported, which might give a clue regarding the forthcoming developing problems. Early recognition is essential to guide growth and development of the face and cranium.[6]

Management of such a problem requires multidisciplinary approach. Treatment includes measures to minimize intracranial pressure and secondary calvarial deformities. Orthodontic treatment with subsequent orthognathic surgical intervention has to be followed in managing the dentofacial deformity.

Prognosis depends on severity of malformation. Innovations in craniofacial surgery have enabled patients to achieve their full potential by maximizing their opportunities for intellectual growth, physical competence and social acceptance. Patients usually have a normal lifespan.

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