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CROUZON SYNDROME: PRESENTATION AND DIAGNOSIS OF A RARE CASE IN AN OPHTHALMOLOGY CLINIC

Ophthalmology

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ABSTRACT

Crouzon syndrome accounts for about five percentage of all craniosynostosis cases, commonly presented with distinctive malformations of the craniofacial region manifested by premature fusion of bony sutures. It is a rare genetic disorder attributed to mutation of FGFR2, a gene responsible for fibroblast growth factor receptor-2. This paper reports the clinical presentation and diagnosis of a rare case of crouzon syndrome in a young male patient presented to an ophthalmologic outpatient department with a chief complaint of diminution of vision in both the eyes with the gradual enlargement of eyeballs since one year of age. Timely diagnosis and education provided to patient and family about the management of this syndrome enabled the patient to attain intellectual growth, physical competence and to maximize the opportunities to lead a normal life.

KEYWORDS

crouzon syndrome, craniosynostosis, optic atrophy, proptosis

INTRODUCTION:

The craniosynostoses are a heterogeneous group of syndromes characterized by a premature fusion of cranial sutures, craniosynostoses are a heterogeneous group of syndromes known to be a rare genetic disorder, mainly presented by distinctive malformations of the cranial and facial region may be associated with other anomalies. Fusion of cranial sutures in craniosynostoses limits the conventional growth of brain, leading to its offset growth towards the open sutures. The resultant asymmetrical shape of skull and deformities makes craniosynostoses a host of numbers of syndromes; including but not limited to Carpenter syndrome, Apert syndrome, Jackson-Weiss syndrome and Crouzon syndrome. A young male patient seen for initial consultation in the outpatient Ophthalmology department came with a chief complaint of attenuation of vision along with gradual enlargement of eyeballs noted from the age of one year. Our paper describe the history of present illness, diagnostic measures used to diagnose the Crouzon syndrome and management of this rare case.

Case report:

A male patient aged 8-years, accompanied with his father, came for an initial evaluation at the department of Ophthalmology clinic with complaint of diminution of vision in both eyes and gradual enlargement of both eyeballs since infancy. Severity of symptoms reported to be gradually increased over a period of time as the child grew. The patient's past medical and surgical history was reviewed, according to which, he was born full term with a vaginal delivery without any complication. There was no other significant medical history or contributory family history.

On physical examination, the child had a short stature with a height 110 cm. Shape of the skull was asymmetrical with the flattened forehead and occiput and prominent dome shaped vault. On ocular examination indicated the best corrected visual acuity in the right eye was PL+, PR+4 and in the left eye it was 3/60. There was mild proptosis present in the right and left eye, determined by exophthalmometer, measured 23 mm and 22 mm respectively. Right eye had 45° divergent squint with poor fixation (Figure 1). Slit lamp examination showed normal eyelid, conjunctiva, cornea, anterior chamber, iris and lens of both the eyes. The right eye pupil was sluggishly reactive to light, whereas left eye pupil was normally reactive to light. Interpupillary distance measurement in the patient was 63 mm, which is considered abnormal. Based on ishihara chart, colour vision was normal in left eye (only Seeing eye). Intraocular pressure in the right eye was 18 mmHg and in the left eye was 20 mmHg measured by Goldmann's applanation tonometer within normal range. The fundus examination indicated total optic atrophy in the right eye and partial optic atrophy in the left eye (Figure 2).

Oral examination of the patient suggested high arched palate, hypoplastic maxilla and malocclusion of the teeth. The child did not have any hand or feet abnormalities and other anomalies. Radiograph of the skull demonstrated sclerosis and overlapping edges of the coronal and lamdoidal sutures suggestive of synostosis with prominent cranial markings due to compression of developing brain on the fused sutures appearing as multiple radiolucencies on the inner surface of the cranial vault, which is called beaten metal appearance (Figure 3). Additionally, radiograph of paranasal sinus revealed hypoplastic maxillary bones. Radiographs of spine and chest were normal.

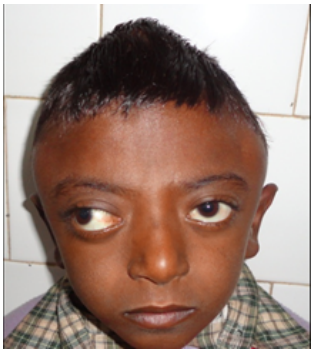

Figure 1: Mild proptosis in both the eyes with right eye divergent squint

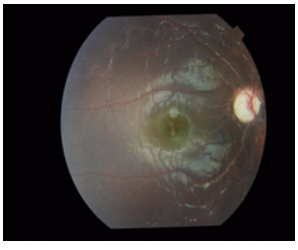

Figure 2: Right optic atrophy in fundus photograph

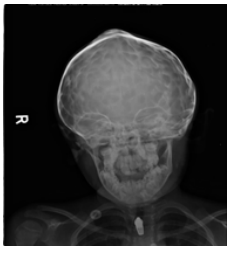

Figure 3: 'Beaten metal appearance' in digital X-ray skull

X-ray spine and chest were normal. X-ray paranasal sinus showed hypoplastic maxillary bones. Computed Tomography (CT) scan of the brain with orbit was also suggestive of coronal and lamdoidal synostosis, and chronically elevated intracranial pressure with shallow orbits and mild proptosis of both globes. On the bases of these characteristics and findings, the patient was diagnosed with Crouzon syndrome.

Treatment options:

The patient and family was explained and provided with the education

about Crouzon syndrome, and treatment options to alleviate the chief complaint of the patient which was reduced vision. [6] The following multistage surgical approach involving multidisciplinary team was explained:

- Cranioplasty that includes skull remodelling for cosmetic benefit.
- Morcellation procedure which consists of drilling numerous burr holes in the skull and connecting them with lines of craniotomy.
- Ilizarov procedure:- A new technique of craniofacial disjunction followed by gradual bone distraction has been reported to correct proptosis and improve the anatomy of mid face.
- Le Fort III osteotomy, monobloc frontofacial advancement or bipartition osteotomy to reduce orbital volume and proptosis, protect against breathing difficulty and provide cosmetic benefit.
- Squint surgery to correct exotropia.
- Orthodontic dental treatment for teeth malocclusion.
- Psychological and social support for coping.

DISCUSSION:

Crouzon syndrome is a rare autosomal dominant disorder with complete penetrance and variable expressivity is named after Octave Crouzon (1874-1938), who was a French neurologist. He first described the hereditary syndrome of craniosynostoses in a case of a mother and a son showing cranial deformities, recession of the maxillae with relative prognathism, marked exophthalmos, divergent squint and optic atrophy with visual impairment [1,2]. Crouzon syndrome is caused by mutation in the fibroblast growth factor receptor-2 (FGFR-2) and/or fibroblast growth factor receptor-3 (FGFR-3) gene which is located on chromosome 10q26 and 4p16.3 respectively.[2,7] These genes regulate the production of fibroblast growth factor receptors. Mutations in these genes cause an increase in the production of FGFR proteins that leads to an abnormal rise in the number of cells forming bones, which results in the premature fusion of cranial bones during the prenatal period. Thus, premature synostosis of the coronal and sagittal and in some cases, lambdoid sutures begins in utero and is manifest at birth. Once a suture becomes fused, growth perpendicular to that suture becomes restricted, leading to compensatory growth in the direction of remaining open sutures to allow continued brain growth, resulting in abnormal bone growth and facial deformities. The order and rate of fusion of the sutures determine the degree of deformity and disability.

Prevalence of the Crouzon syndrome if rare, estimated to 1.6 per 100000 births worldwide and contributing about 5% of all craniosynostoses cases. The patient presented to our outpatient Ophthalmologic clinic had oxycephaly with ocular features including mild proptosis and total optic atrophy in right eye and partial optic atrophy in left eye with right eye divergent squint. Associated features were high arched palate, hypoplastic maxilla, prognathism of mandible and malocclusion of teeth. These significant findings draw the diagnosis of a rare condition, Crouzon syndrome.

Detection of FGFR-2 gene mutation through DNA testing and ultrasonography could help early diagnosis and can provide families with an option of termination or opportunity to make an informed decision about postnatal management if not terminate. Diagnosis made during an early stage prevent poor cosmetic appearance and complications like decreased visual acuity, airway obstruction and mental retardation as the age advances as intervention can be made as early as possible.

The prognosis depends on the severity of malformations and the timing of intervention. In our case, patient was present in late stage with severe visual disability. Innovations in craniofacial surgery explained to the patient can help attain intellectual growth, physical competence and an opportunity to lead normal life.[8,9]

CONCLUSION:

Crouzon syndrome is a rare autosomal dominant disorder characterized by cranial-facial anomalies. The study is to be done to increase awareness and promote progress in the early diagnosis and specific treatment with multidisciplinary approach and thus preventing the serious complications.

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