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CROUZON SYNDROME WITH OPHTHALMOLOGICAL COMPLICATIONS

Ophthalmology

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ABSTRACT

Crouzon Syndrome is characterized by premature craniosynostosis. It has an autosomal dominant inheritance but represents fresh mutation also.There may be increased intracranial pressure for which surgical morcellation procedures are indicated. A case of craniosynostosis is reported which is diagnosed as Crouzon Syndrome with ocular complications on clinical grounds. Postnatal growth of the skull is constrained along the closed suture leading to deformations of skull shape, distortion of facial features, orbital dystopia, and increases in intracranial pressure. Head shape is typically brachycephalic with associated midface retrusion. The orbits are shallow and the globe is protruded, predisposing the cornea to exposure and intermittent protrusion of the globe equator anterior to the eyelid margins in infancy. Increased intracranial pressure can result in optic neuropathy with occult visual loss for which these children require serial monitoring. Distortion of head shape, abnormal facial appearance, and concerns about increased intracranial pressure restricting brain growth prompt surgical intervention. Expansion and reshaping of the skull, frontal bone, and superior orbits is performed between 9 and 15 months of life when the infant capacity for bone remodeling of the surgical cranial defects is balanced with the stability of the bone. Midface advancement is usually performed at 7 to 10 years of age after completion of upper midface growth.

KEYWORDS

INTRODUCTION

Crouzon syndrome is a rare genetic disorder with autosomal dominant inheritance with the prevalence of 1 in 25,000 live births, and it constitutes 4.8% of all craniosynostosis. In 25% of cases, it may also occur sporadically because of a fresh mutation. The underlying pathological process is premature synostosis of the coronal, sagittal and occasionally lambdoid sutures beginning in the first year of life and completed by 2-3 years of life. This fusion does not allow the bones to grow normally, affecting the shape of the head, appearance of the face and the relationship of the teeth. The diagnosis is based on clinical findings and radiological examination. This syndrome was described by Crouzon in 1912 who described a patient with a characteristic group of deformities which were then observed in other individuals. Crouzon syndrome is caused by malformations of the mesenchyme and ectoderm. Mutation of the gene for fibroblast growth factor receptor 2 (FGFR2) is responsible for Crouzon syndrome and this gene has been mapped to the long arm of chromosome 10 and mutations in exon B of FGFR2 gene have been described [1] Craniofacial abnormalities include brachycephaly, shallow orbits and maxillary hypoplasia. Other facial features include prominent nose, frontal bossing, and ocular proptosis with or without divergent strabismus and hypertelorism, although age related modifications have been reported [6].

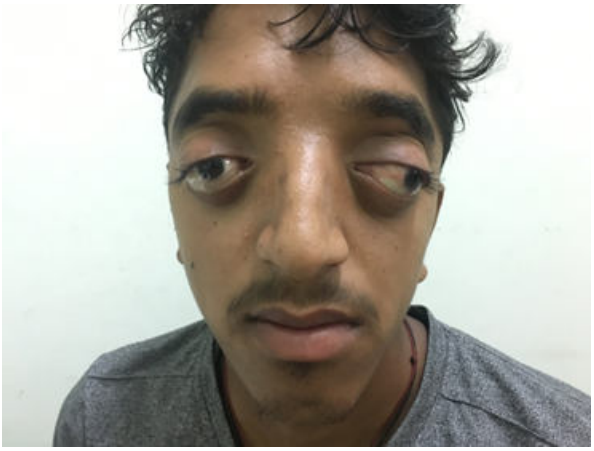

Figure.1: Crouzon syndrome: Characteristic facial appearance. Note microcephaly, bilateral proptosis with left divergent strabismus, prominent nose.

Case Report

A 25 year old male patient presented to eye department of Civil Hospital Palampur for visual disability certificate. He was a known case of Crouzon syndrome with negative family history. On local examination, the patient had a brachycephalic head formation, maxillary hypoplasia, a wide parrot beaked nose with deviated nasal septum. On dental examination, he had abnormal dental occlusionand maxillomandibular relation. On ophthalmic examination, the patient had bilateral proptosis, microcornea, slanted palpebral fissure, shallow orbits, alternate divergent squint >40°, horizontal jerk nystagmus and high myopia. He had hypertelorism. On Slit lamp examination of the patient, there was evidence of healed exposure keratopathy in the left eye with bilateral Bitot spots and posterior embryotoxon bilaterally. Fundus examination revealed myopic degeneration in both the eyes. On gonioscopy angles were closed in both eyes. There were peripheral anterior synaechae and pigment present in both angles. Cardiovascular system, central nervous system, respiratory and abdominal examination was normal. Routine hematological and biochemical tests were within normal limits.

DISCUSSION:

The main clinical feature of Crouzon syndrome is a skull deformity which may be brachycephaly, oxycephaly or trigonocephaly. Hydrocephalus and mental retardation may be present becauseof premature fusion of cranial sutures.[2] There is maxillary hypoplasia with relative mandibular prognathism and dental mal occlusion. There can be deviation of the nasal septum,narrowed or obliterated anterior nares, wide parrot beaked nose and rhinolalia. Midfacial anomalies like cleft lip and cleft palate might be present. Hearing loss may be there which is usually conductive. Orbital defects include bilateral proptosis because of shallowing of orbits secondary to arrested growth of the maxilla and zygoma, and anterior positioning of the greater wing of the sphenoid. There might be spontaneous luxation of the globes also. There is strabismus (esotropia or exotropia) and hypertelorism. There might be vision threatening complications like exposure keratopathy and optic atrophy due to compression of the opticforamen. Acanthosis nigricans can also be associated with Crouzon syndrome. In radiological examination, antero-posterior, lateral and cephalometric views of the skull are taken. It shows premature suture closure and provides information about maxilla-mandibular relation. `Paw marking' of the skull is seen due to raised intracranialpressure.[3] CT scan confirms the standard radio graphical findings and provides information on ventricular size. Suture closure can be graphically displayed by 3-dimensional CT scans.Cephalometric studies measure the dimensions of somefunctional spaces like orbits, rhinopharynx and nasal cavities .[4]

This syndrome is characterized by premature craniosynostosis which is quite variable but the procedures are indicated to reduce raised intracranial pressure and to allow normal brain development. Newer techniques allow for cosmetic reconstruction of facial bones.

Prognosis depends on severity of malformation. Patients usually have normal lifespan. [6]

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