Case Report

Crouzon Syndrome With Ophthalmological Complications

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Crouzon Syndrome is characterized by premature craniosynostosis. It has an autosomal dominant inheritance but represents fresh mutation also. Other craniofacial abnormalities include ocular proptosis caused by shallow orbits with or without divergent strabismus. 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.

Split craniotomy was performed by a neurosurgeon to relieve raised intracranial pressure and to enhance brain growth. Crouzon Syndrome was originally described in 1912 by Crouzon in a mother and her daughter. It is an autosomal dominant inherited disorder but represents fresh mutation also. Crouzon syndrome is characterized by premature craniosynostosis which is quite variable but the coronal suture is nearly always bilaterally involved. Craniofacial abnormalities include brachycephaly, shallow orbits and maxillary hypoplasia. Other facial features include prominent nose, frontal bossing, ocular proptosis with or without divergent strabismus and hypertelorism, although age related modifications have been reported. Here we report a case of 6-year-old boy who presented with visual problems and was diagnosed clinically as a case of Crouzon Syndrome with ocular complications.

Case Report

A six year old child presented to Paediatrics Department of Capital Development Authority Hospital with complaints of progressive bulging of both eyes and decreased vision in the left eye since one month (Fig.1) There was no history of fever, headache, vomiting or fits. The vision of the right eye was normal. His birth was uneventful. Mother had no antenatal problem. He attained his milestones at appropriate age. Other siblings were alive and healthy. For the past one month he was having visual problems. On examination child was fully oriented in time, place and person. He was microcephalic with abnormal shape of skull and hypoplastic orbital ridges. There was bilateral proptosis with left divergent strabismus. Horizontal nystagmus was observed in both eyes but was more marked in left eye. He had reduced vision. Funduscopy revealed bilateral optic atrophy. Other facial features included a prominent nose and deep and narrow palate. No digital abnormalities were seen. No dental aplasia was present. Systemic examination revealed no abnormality.

He was diagnosed as a case of Crouzon syndrome with ocular complications on clinical basis. He was investigated for microcephaly and suspected craniosynostosis. Radiographs of skull showed small sized skull with early closure of sutures and fontanelle suggestive of craniosynostosis(Fig 2). A hammered-silver (beaten metal/ copper beaten) appearance was also seen due to raised intracranial pressure and compression of the developing brain on the fused bone. CT scan brain with contrast was done which confirmed the features suggestive of craniosynostosis with premature closure of sutures and pinched ventricular chain. It also showed shallow orbits with proptosis(Fig 3).

The final diagnosis of craniosynostosis most likely Crouzon syndrome with ocular complications was made. Child was referred to Neurosurgeon for split craniotomy. The surgery was uneventful. The child has improved, performing his daily chores appropriately and is school going.

Discussion

Crouzon Syndrome (craniofacial dysostosis) was originally described in 1912 by French Neurologist, Octave Crouzon in a mother and her daughter. It is an autosomal dominant inherited disorder but represents fresh mutation also. The male-to-female preponderance is 3:1. Crouzon syndrome, along with Apert, Carpenter, Chotzen, and Pfeiffer syndromes, is one of the most common genetic disorders associated with a craniofacial syndrome. Mutations in fibroblast growth factor receptor gene (FGFR2) which maps to chromosome 10p 25-q26, is responsible for greater than 90% of cases. Incidence of Crouzon syndrome is 1 in 25000 in the general population.

This syndrome is characterized by premature craniosynostosis which is quite variable but the
Coronal suture is nearly always bilaterally involved. The degree of craniosynostosis as well as the age of onset is variable. The shape of the head depends on the timing and order of suture fusion but brachycephaly is the most common finding. The orbits are shallow and ocular proptosis with or without divergent strabismus are prominent. Hypoplasia of maxilla with or without curved parrot-like nose and orbital hypertelorism are typical facial features. The phenotypic features of Crouzon syndrome may be absent at birth and evolve gradually during the first few years of life.

Other craniofacial abnormalities include exposure conjunctivitis or keratitis, unexplained poor visual acuity, optic atrophy, nystagmus, inverted V shaped palate and conduction hearing loss. Patients of Crouzon syndrome may occasionally present with hydrocephalus, seizures, and mental retardation. Management of such patients requires multidisciplinary approach. Treatment includes measures to minimize intracranial pressure and secondary calvarial deformities. Surgical morcellation 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.

In conclusion, early detection of Crouzon syndrome and its ocular complications is required to reduce the associated ophthalmic problems. Optic atrophy remains an important cause of visual impairment in these patients before decompressive craniotomy.

References
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