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Ocular involvement of Crouzon Syndrome: A Case Report

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Abstract: Crouzon syndrome is a rare genetic disorder characterized by distinctive malformations of the skull and facial region. Though Crouzon syndrome is inherited as autosomal dominant disorder, many of them are sporadic. We report a case with ocular involvement in A-9-year old female.
Keywords: Crouzon syndrome, ocular involvement

INTRODUCTION

Octave Crouzon, a Frecnchof neurologist was the first who described a hereditary syndrome of craniofacial craniofacial dysostosis in a mother and son, which includes a triad: skull deformities, facial anomalies and exophthalmos [1]. 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 [2].The prevalence of this disease is 333–476 per million births. Bilateral severe proptosis and corneal exposure are due to arrested growth of the maxilla and zygoma resulting in shallow orbits [3] Crouzon syndrome is caused by mutation in the fibroblast growth factor receptor 2 (FGFR2) gene at the chromosomal locus 10q 25-q26 [4**].** It is characterized by craniosynostosis, shallow orbits, ocular proptosis, midface hypoplasia, and a curved, beaklike nose. It accounts for approximately 4.8% of all cases of craniosynostosis making it the most common syndrome within the craniosynostosis group [5]. In most individuals with Crouzon syndrome, there is unusual shallowness of the orbits that accommodate the eyeballs. As a result, the eyeballs appear to protrude; so affected individuals are unusually susceptible to developing exposure keratitis as well as exposure conjunctivitis [6]. Rarely the globe may be luxated anterior to the lids. The differential diagnosis of Crouzon syndrome includes simple craniosynostosis as well as the Apert, Pfeiffer and Saethre-Chotzen syndromes. Unlike the other forms, Crouzon syndrome presents with no digital abnormalities [7].

CASE REPORT

A-9-year old female presented with history of bilateral prominent eyes since birth. She was mentally retarded. The prenatal, the delivery and the postnatal history revealed no abnormality. The family history was normal. The clinical findings were: hypertelorism, bilateral proptosis with exotropic right eye. The visual acuity was 6/18 in the right eye and 6/9 in the left eye. The cornea had exposure keratitis. The fundus was normal.

Fig-1: Anterior view of a 9-year-olf child with Crouzon syndrome

DISCUSSION

Craniosynostosis is a feature seen in around 100 syndromes wherein premature fusion of one or more cranial sutures occur, which reduces intracranial

space and leads to skull deformities and disturbances in normal brain development [8]. Crouzon syndrome patients have 3 distinctive features: craniosynostosis, underdeveloped midface, proptosis [9].

There may be associated chronic papilloedema causing optic atrophy. Other associations are aniridia, blue sclera, cataract, coloboma, ectopia lentis and optic nerve hypoplasia.Visual impairment in at least one eye may occur in 35 % and 9% in both eyes [10]. The proptosis found in this patient is present in most of the patients worldwide [11]. There also have been rare occurrences of nystagmus, anisocoria, microcornia, megalocornea, glaucoma and luxation of the eye [12]. There is no race or sex predilection. The appearance of an infant with Crouzon syndrome can vary in severity from mild presentation with subtle midface characteristics to severe forms with multiple cranial sutures fused and marked midface and eye problems [13].

CONCLUSION

Crouzon syndrome is a rare congenital abnormality with very frequent ocular features. The proptosis that is found in most of the patients presenting this issue exposes them to ocular complications due to the exposure of the cornea. A thorough ophthalmological follow up of these patients is mandatory in order to protect them from blindness.

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