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CROUZON SYNDROME: A CASE REPORT

Ophthalmology

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KEYWORDS

INTRODUCTION

Crouzon syndrome is primarily an autosomal dominant disorder that belongs to a group of conditions known as craniosynostosis syndromes. Although craniosynostosis occurs in approximately 1 in 2,500 births, Crouzon syndrome is relatively rare with an estimated incidence of 1 in 60,000 newborns.[1] It is characterised by premature closure of one or more cranial suture (craniosynostosis) mainly coronal sutures, which leads to abnormal development of head (brachycephaly, scaphocephaly,or a cloverleaf skull in severe cases) and prominent facial features like midfacial hypoplasia, proptosis, strabismus, beaked nose along with middle ear abnormalities, hearing loss and recurrent otitis media.

First described by French neurologist Louis Édouard Octave Crouzon[2] in 1912, this syndrome is caused by missense mutations in the fibroblast growth factor receptor 2 (FGFR2) gene (chromosome locus 10q25e -10q26)[3-4]. The lack of suture expansion causes secondary deformities, including increased intracranial pressure (ICP), midfacial hypoplasia, and airway obstruction.[5] Additionally, the shallow orbits can lead to severe ocular complications, including inadequate closure of lids, secondary exposure keratopathy,and optic atrophy. The involvement of the maxilla often results in malocclusion, cleft lip and/or palate and dental abnormalities, further impacting speech and feeding. Eventhough limb deformities are not typically a hallmark of crouzon syndrome, some individuals with the condition can have subtle or less common limb involvement.[6] Patients may also experience intellectual disabilities and require emotional support due to the social stigma associated with their appearance. Other craniosynostosis syndromes include Apert syndrome, Saethre-Chotzen, Pfeiffer syndrome , and most common type Muenke syndrome.[6,7]

Case Report

A 13-year-old boy presented to the pediatric department with a one-week history of fever and right hip pain, leading to an inability to move his right lower limb. Evaluation confirmed a diagnosis of septic arthritis of the right hip, with cultures testing positive for MRSA. Patient was referred to ophthalmology for consultation in view of proptosis and dystopia.

Born as the fifth of seven siblings in a low-income family, he had no immediate relatives with a similar history or any diagnosed syndromic disease. He was born full- term via LSCS in a hospital and was diagnosed with anal agenesis at birth. He underwent surgery at one month of age. Normal developmental milestones achieved according to age.

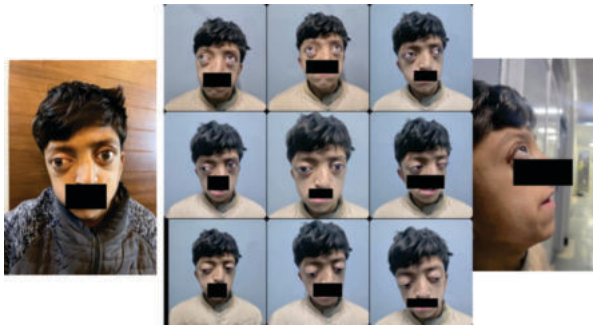

Fig 1 : Anterior view of patient with movement in all gazes and lateral view.

On examination, patient vision was 6/6(P) in both eyes. Colour vision was normal. Patient had proptosis of both eyes with globe dystopia in right eye. Right eye was displaced inferiorly and laterally. Pupilary distance from midline was increased in right eye but, Intercanthal distance was normal. (unilateral hypertelorism?). Exotropia of approximately 16 degrees was present in the right eye with abaxial proptosis. Hertel's examination showed proptosis: right eye – 24mm, left eye – 23mm (base 118). There was inadequate lid closure present in right eye with right globe subluxation. Extra occular movements were normal. Right pupil was mid dilated and was sluggishly reacting to light. Left pupilary reflex was normal. Anterior segment examination was normal. Corneal topography done showed no abnormality. CCT (R-514µm, L-502µm). ACD (R-3.96mm, L-3.86mm). No lens abnormality noted. Pallor of the neuroretinal rim was observed in the right fundus, while the rest of the fundus findings were normal. No optic nerve edema seen. Axial length (R-23.84mm, L-23.60mm). AS-OCT and OCT-RNFL test were normal. IOP was normal in both eye.

NCCT of patient was done. Report showed bilateral shallow and wide bony orbit with bilateral proptosis of eyeball . All sutures are fused with frontal bossing and inner cortex intentation. Right pliagocephaly with prominent ventricles noted. Styloid ligament calcification noted.

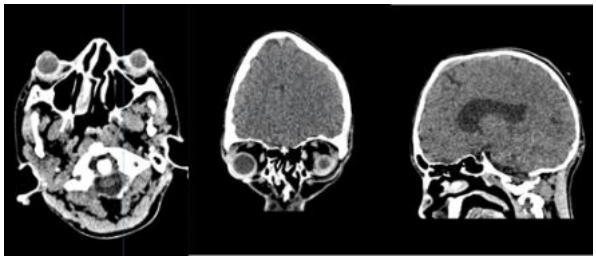

Fig 2: radiological images of NCCT head of patient in transverse, coronal and sagittal axis.

Patient also had high arched palate. Maxillary arch was constricted and underdeveloped. Alignment of maxillary teeth showed crowding of teeth. The lateral profile view shows maxillary retrognathism. Maxillary central incisors were retroclined. Both upper and and lower limb extremities were normal. No deformities of the fingers were noted. The ENT examination showed no signs of hearing loss. However, external deviation of nose towards left and deviation of the nasal septum to the left was noted, though no airway obstruction was present. Bilateral grade 1 hypertrophy of tonsils was noted. The ophthalmological, radiological, and dental findings described above are consistent with patients with Crouzon syndrome.

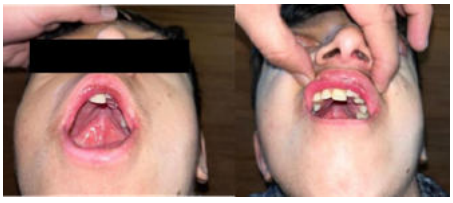

Fig 3: shows high arched palate and crowding of teeth.

DISCUSSION

Crouzon syndrome is a complex craniofacial disorder with significant implications for affected individuals and their families. Early intervention is the key to restore quality of life in patients with crouzon syndrome.[8] Adequate family counselling about the condition of child is vital for better treatment compliances and social rehabilitation.[9] A multi desciplinary approach is to be taken to manage complication arising from crouzon syndrome. Surgical approaches like midfacial distraction osteogenesis at early stages of life offers successful treatment option especially in patients with airway obstruction.[6] Occular complication like exposure keratopathy can be managed with frequent use of lubricating drop, lid taping at night. For cosmetic improvement, strabismus can be treated with surgical approach.[10]

Social stigma due to facial appearance is an important factor affecting the psychological well being of patient. Especially during the adolescent age, discrimination, social exclusion, negative assumptions about their abilities or character can negatively affect the social interactions and quality of life. Support groups, therapy and self esteem building activities can help patient maintain good mental health.[11,12]

CONCLUSION

Crouzon syndrome is a genetic disorder that predominantly impacts craniofacial development. While it poses notable medical challenges, timely diagnosis and effective interventions can significantly enhance outcomes for those affected. Ongoing advancements in genetic research and surgical procedures continue to improve the quality of life for individuals with Crouzon syndrome.

Consent: Informed consent was obtained from patient prior to data collection and publication of case report.

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