***Case report***

**A RARE CASE OF CROUZON SYNDROME**

**ABSTRACT**

Crouzon’s syndrome is a rare autosomal dominant genetic disorder characterized by craniosynostosis and distinct craniofacial abnormalities. It results from mutations in the FGFR2 gene, leading to premature fusion of skull bones and subsequent facial dysmorphology. This case presents a 9-year-old girl with characteristic phenotypic features of Crouzon’s syndrome, including a towering skull, hypertelorism, midface hypoplasia, bilateral proptosis, and retrognathia. The patient also exhibited ENT complications, including recurrent sore throat, nasal discharge, tonsillar hypertrophy, and mixed hearing loss. Radiographic findings confirmed cranial abnormalities consistent with the syndrome, while biochemical tests revealed altered metabolic markers. Given the absence of limb anomalies and positive family history, the diagnosis was established based on clinical and radiological findings. The treatment approach included antibiotic therapy, symptomatic relief, and a planned adenotonsillectomy surgery to manage airway obstruction and improve the patient's quality of life. This case underscores the importance of early diagnosis, multidisciplinary intervention, and long-term follow-up to optimize outcomes in individuals with Crouzon’s syndrome.

**Keywords:** Crouzon’s Syndrome, Craniosynostosis, FGFR2 Mutation, Craniofacial Abnormalities

**INTRODUCTION**

Crouzon’s syndrome is a rare genetic disorder that affects the development of the skull and facial bones. It is a rare autosomal dominant condition with multiple mutations of the fibroblast growth factor receptor (FGFR2) gene, which accounts for 4.8% of all cases of craniosynostosis. Crouzon syndrome is primarily caused by mutations in the FGFR2 gene. This gene provides instructions for making a protein called fibroblast growth factor receptor, which is involved in the development and maintenance of bone and tissue. Mutations in the FGFR2 gene lead to the production of an overactive protein, causing the premature fusion of skull bones (craniosynostosis).

**CASE PRESENTATION**

A 9-year-old girl presented to the pediatric clinic with a chief complaint of recurrent sore throat, fever, cold, intermittent headache since 1month, nasal cavity bleed since 2days, nasal discharge, she also had ear discharge 6months back, partial hearing loss. A detailed family and medical history were collected because the child's appearance and head size were abnormal. The child was born prematurely at 28weeks due to early labor pains, The mother had a Lower Segment Caesarean Section (LSCS) delivery and was the second child from a non-consanguineous marriage. The first female child passed away after 1day of birth. The child was taking no medications and the parents denied any history of allergies. The child was developing normally for her age and all milestones were achieved at the appropriate age. A general head-to-toe examination of the child revealed a towering skull with dolichocephaly, long face, high sloping forehead, up slanting palpebral fissure, hypertelorism, midface hypoplasia, bilateral proptosis, retrognathia. Ear examination revealed that left ear has moderately severe mixed hearing loss and there was a wax in both the ears, patient had no finger abnormalities. Intraoral examination revealed a high arched palate, grade 3 tonsillar hypertrophy, nose examination revealed Bilateral inferior turbinate hypertrophy. History showed that these features began to develop from the birth of the baby and gradually increased in severity over time. There was no significant positive family history. Given the above findings, radiographs of the skull were taken, showing copper beaten skull, Grade 2 adenoid hypertrophy, CT brain were also taken which was found to be normal. Other systemic examination

was found to be normal. Routine Biochemical tests revealed that there was an decrease in urea levels[4mg/dl], creatinine levels [0.54mg/dl] and uric acid levels [2.82mg/dl] and there was an increase in alkaline phosphate levels[289mg/dl] and globulin levels[3.84mg/dl]and hematological findings were within normal limits.

Based on the above clinical and radiological findings and in the absence of hand and feet anomalies, a diagnosis of Crouzon’s Syndrome was made.

Treatment plan- Patient was treated with tablet AMOXYCLAV 375MD/PO/BD, TAB PAN 20MG/PO/OD, TAB PCM 250MG/PO/BD, TAB LEVOCETRIZINE 5MD/PO/HS, NASOCLEAR DROPS 2DROPS TID, SYP AMBROXYL 5ML BD, DEWAX EAR DROPS 3DROPS QID and planned for ADENOTONSILLECTOMY Surgery.

**DISCUSSION:**

In 1912, French neurologist Octave Crouzon (1874–1938) reported that a mother and son had a genetic condition called craniofacial synostosis. He defined the trifecta as exophthalmos, facial anomalies, and skull deformities—now referred to as Crouzon syndrome. It is a fully penetrant, autosomal dominant condition with varying expressivity [1]. Mutations in the fibroblast growth factor receptor-2 (FGFR2) gene, which is mapped to chromosome locus 10q25–10q26, are the cause. However, there is locus heterogeneity, with different affected individuals having causal mutations in FGFR-2 (Crouzon syndrome) and FGFR3 (Crouzon syndrome with Acanthosis nigricans) [2]. Shallow orbits, ocular proptosis, orbital hypertelorism, strabismus, papilledema, optic atrophy, exposure keratitis, and vision loss are among the most prevalent ocular disorders. Rare cases of nystagmus, iris coloboma, cataract, ectopia Lentis, blue sclera, glaucoma, luxation of the eye, aniridia, anisocoria, microcornea, and megalocornea have also been reported [1].

The sequence, pace, and development of sutural synostosis determine the degree of craniofacial deformity in Crouzon's syndrome. Typically, craniosynostosis starts in the first year of life and is finished by the time a child is three years old [3]. When a suture is fused, its ability to grow upright is limited. Consequently, compensatory development occurs at the remaining open sutures, leading to aberrant bone formation [4].

Premature craniotomies, exophthalmos (optic disc edema and proptosis), and midface hypoplasia are the three hallmarks of Crouzon's syndrome. Brachycephaly, hypertelorism, divergent squint, cloverleaf skull, nasal septal deviation, wide-beaked curved nose that resembles a parrot's nose, and cleft lip are further craniofacial traits observed [5]. Towering skull with dolichocephaly, long face, high sloping forehead, up slanting palpebral fissure, hypertelorism, midface hypoplasia, bilateral proptosis, retrognathia, left ear has moderately severe mixed hearing loss, arched palate, grade 3 tonsillar hypertrophy, nose examination revealed bilateral inferior turbinate hypertrophy, radiographs of the skull showed copper beaten skull these are the findings were observed in our patient.

The narrow orbit causes exophthalmia. Although the height of the palate is normal by measurement, maxillary hypoplasia causes the anterior posterior dimension of the upper arch and the breadth of the dental arch to decrease, producing the impression of a strongly arched palate [6]. Crowding of the teeth, crossbite, anterior open bite, and cleft palate are further intraoral symptoms.

Symptoms of the central nervous system include conductive hearing impairment, headache, dizziness, epilepsy, hydrocephalus, and bilateral jugular foraminal stenosis. These individuals often have normal levels of mental capacity and psychomotor development. However, mental retardation can result from elevated intracranial pressure. Significantly frequent side effects include jugular foramen stenosis with venous blockage, chronic tonsillar herniation, and progressive hydrocephalus [7].

Crouzon's condition can lead to blindness, luxation of the eye globes, exotropia, conjunctivitis or keratitis from proptosis, and impaired vision from corneal damage and optic atrophy [8].

The diagnosis of cranio-synostosis and other related skeletal abnormalities depends heavily on radiographic assessment. The first radiological indicators of cranial suture synostosis are overlapping margins and sclerosis. Radiolucent sutures that often appear on the skull on radiography, the patient had a copper-beaten skull and obliteration of the sagittal suture, which suggested internal remodeling of the calvaria brought on by an increase in intracranial pressure from premature sutural fusion [9].

Saethre-Chotzen syndrome, Apert syndrome, Pfeiffer syndrome, and Carpenter syndrome are all differential diagnoses for Crouzon's syndrome. All of CS's symptoms, as well as hand and foot syndactyly, are present in Apert syndrome. Along with characteristics of CS, Pfeiffer syndrome will also have wide large toes, either with or without soft tissue syndactyly of the hands and feet [10].

Early diagnosis is crucial for the interdisciplinary management of Crouzon's illness. The patient's age and the severity of the illness determine how they are managed. In order to provide sufficient cranial room for brain growth and expansion, it is preferable to surgically remove the prematurely fused sutures of the skull within the first year of life. For optimal effects, skull reshaping may need to be repeated as the kid gets older. Jaw surgery and mid-facial advancement may be required to decrease the exophthalmos and give sufficient orbital volume. A psychiatrist may be required in certain cases when psychological issues brought on by the cosmetic defect are observed [11].

**CONCLUSION**

Crouzon syndrome is an uncommon condition in which the patient presents with distinct facial features. Clinicians must be able to recognize these distinctive symptoms in patients who are ignorant of their condition in order to provide early care and prevent consequences caused by late diagnosis.

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