

Crouzon's Syndrome: A Case Report

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Abstract Crouzon's syndrome is a rare autosomal dominant disorder characterized by craniofacial malformations. It's the most common syndrome among the craniosynostosis group accounting for about 4.8 % of all of them. Crouzon syndrome is caused by mutation in the fibroblast growth factor receptor-2 (FGFR-2) gene resulting in premature closure of suture lines. Our article describes a case report of a 3 years old girl who displayed characteristic dysmorphic skull and facial features of Crouzon's syndrome.

Keywords: Crouzon's syndrome, craniosynostosis, maxillary hypoplasia

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1. Introduction

Crouzon's syndrome (CS), otherwise known as craniofacial dysostosis, was first described by the French neurologist Octave Crouzon in 1912 as one of the different varieties of craniosynostosis [1]. It's a rare genetic disorder with prevalence of 15-16 cases in one million newborns. A positive family history is reported to occur in 44-67% of cases. Majority of the cases of CS are autosomal dominant with variable expressivity, but sporadic cases with negative family history has been reported [2]. 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. Crouzon's syndrome is characterized by a triad of premature craniosynostosis, exophthalmos and midface hypoplasia.

Here with the consent of the parents, clinical and radiographic features of CS in a child 3 years old is going to be described in this article

2. Case Report

A 3 years-old female child, brought by her parents with concerns of protruding large eyes for more than one year. Her eyes protrusion has been gradually increasing. Parents didn't mention any other symptoms. No Past medical history or surgical history.

The perinatal history found to be insignificant. Family history revealed Autosomal Dominant inheritance as the father suffers from the same condition (Figure 1). Her developmental mile stones were found to be normal.

Anthropometry was suggestive of short stature and Grade 3 malnutrition.

On extraocular examination, the patient had trigonocephalic skull with frontal bossing. Nasal bridge was depressed, low set ears present without any degree of hearing loss. Ocular manifestations such as shallow orbits, hypertelorism, bilateral ocular proptosis was seen. A mild degree of mid-facial hypoplasia was also present. Her hands and feet appear to be normal. On fundus examination, chronic papilledema features were present. Other systems examination was normal

On radiological examination, skull X- ray revealed an abnormal skull shape with hypoplasia of the maxilla and zygoma. Prominent cranial markings of the inner surface of cranial vault shows the typical copper beaten appearance, seen as multiple radiolucencies appearing as depressions (Figure 1A and Figure 1B). CT scan revealed closure of skull sutures, maxillary hypoplasia and diffuse indentations of inner table of skull (Figure 2A and Figure 2B).

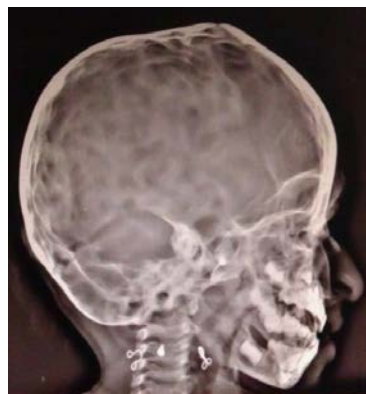


Figure 1A. X-ray skull lateral view of the patient showing typical copper beaten appearance

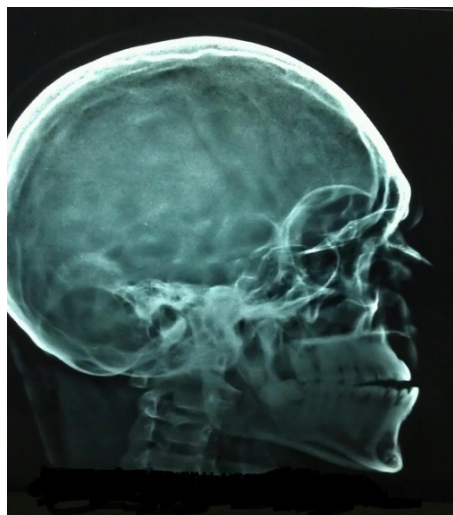


Figure 1B. X-ray of the father retrieved from a previous admission showing the typical copper beaten appearance

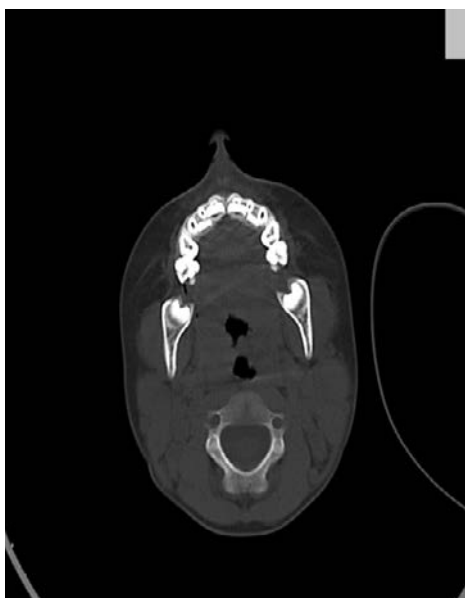


Figure 2A. Computed tomography (CT) image of the skull showing maxillary hypoplasia

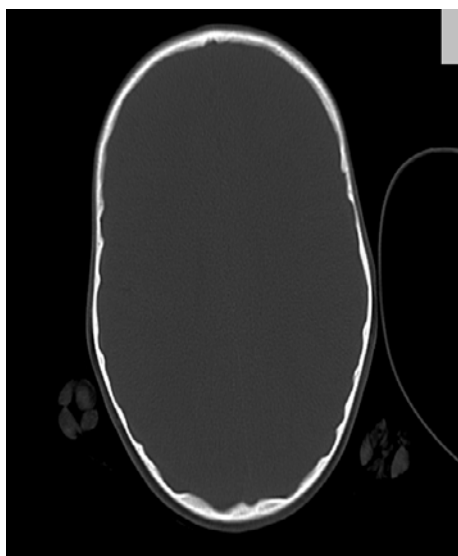


Figure 2B. Computed tomography (CT) image of the skull showing diffused indentation of inner table of the skull

As for management, multistep surgical correction procedures for the craniofacial deformity were advised for the patient. However, despite adequate counseling, the parents refused. Patient was lost to follow-up.

3. Discussion

Crouzon syndrome is a genetic disorder, characterized by premature fusion of sutures in the skull resulting in an abnormally shaped head and face. It is commonly inherited as an autosomal dominant trait, with complete penetrance and variable expressivity, but some cases may present as a *de novo* mutation arising from unaffected parents [3]. Usually, these mutations are associated with increased paternal age and these mutations exhibits a near exclusive paternal origin. The genetic defect that is seen in Crouzon's syndrome appears to originate from the mutation of fibroblast growth factor receptor 2 (FGFR2) located at the chromosomal locus 10q25q26 resulting in early fusion of skull sutures during fetal development [4].

Cranial malformation in Crouzon's syndrome depends on the order and rate or progression of sutural synostosis. Craniosynostosis usually begins during the first year of life and is usually completed by the age of 3 [5]. When a suture gets fused, growth upright of the suture gets restricted. As a result, compensatory growth happens at remaining open sutures causing abnormal bone growth [6].

Crouzon's syndrome is characterized by a triad of premature craniostosis, exophthalmos (optic disc edema and proptosis) and midface hypoplasia. Other craniofacial features seen includes: brachycephaly, hypertelorism, divergent squint, cloverleaf skull, deviation of the nasal septum, wide-beaked curved nose resembling a parrot nose and cleft lip [7]. Our patient presented with cranial features along with some characteristic features of CS which were exophthalmia and maxillary hypoplasia. Exophthalmia arises due to the shallow orbit. Because of maxillary hypoplasia, the anterior posterior dimension of the upper arch along with dental arch width are reduced giving the appearance of a highly arched palate, although palate height is normal by measurement [8]. Other intraoral manifestations include crowding of the teeth, crossbite, anterior open bite and cleft palate.

Central nervous system manifestations include hydrocephalus, headache, epilepsy, dizziness, bilateral jugular foraminal stenosis and conductive hearing impairment. Mental ability and psychomotor development are generally within normal limits in such patients. However, increased intracranial pressure can lead to mental retardation. Progressive hydrocephalus, chronic tonsillar herniation and jugular foramen stenosis with venous obstruction may also occur with significant frequency [9].

Complications of Crouzon's syndrome may include conjunctivitis or keratitis due to proptosis, luxation of the eye globes, exotropia, poor vision due to optic atrophy and corneal injury, blindness [10].

Radiographic evaluation plays a crucial role in diagnosis of cranio - synostosis and other associated skeletal anomalies. The earliest radiographic signs of cranial suture synostosis are sclerosis and overlapping edges. Sutures that normally look radiolucent on the skull

film will not be detectable or will show sclerotic changes [11]. Obliteration of the sagittal suture and a copper beaten skull were seen radiographically in this patient, indicating internal remodeling of the calvaria due to an increase in intracranial pressure owing to premature sutural fusion.

Differential diagnosis of Crouzon's syndrome includes Apert syndrome, Pfeiffer syndrome, Carpenter syndrome and Saethre-Chotzen syndrome. In Apert syndrome, all of the manifestations seen in CS along with syndactyly of hands and feet are present. While in Pfeiffer syndrome, broad big toes with or without soft tissue syndactyly of the hands and feet will be present, in addition to features of CS [12].

Management of Crouzon's disease is multidisciplinary and early diagnosis is important. Management varies according to the age of the patient and severity of the disease. In the first year of life, it is preferred to surgically release the premature fused sutures of the skull to allow adequate cranial volume to be present for brain growth and expansion. Skull reshaping may need to be repeated as the child grows to give the best possible results. If necessary, mid-facial advancement and jaw surgery can be done to provide adequate orbital volume and reduce the exophthalmos. Sometimes psychiatric disorders caused by the cosmetic deformity is seen and a psychiatrist is needed [13].

4. Conclusion

Crouzon syndrome is a rare syndrome in which the patient usually presents with characteristic facial features. Clinicians must be able to recognize these characteristic features in patient who are unaware of their condition, so that early management is given and complications due to late diagnosis are prevented.

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