Familial Crouzon syndrome

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Abstract

Crouzon syndrome is an autosomal dominant condition of the craniosynostotic syndromes without syndactyly and with various dentofacial anomalies. Craniosynostosis, maxillary hypoplasia, shallow orbits, ocular proptosis and hypertelorism are the characteristic features of Crouzon syndrome. This report describes the variable clinical features in affected individuals over two generations of a family with dentofacial deformities and review of literature.

Keywords: Craniosynostosis, Crouzon syndrome, dentofacial anomalies

Introduction

In 1912, Crouzon first described this syndrome as one of the varieties of craniofacial dysostosis caused by premature obliteration and ossification of two or more sutures, most often coronal and sagittal[1] and suggested the term “dysostose craniofaciale hereditare”. It is also referred to as craniofacial dysostosis, hereditary craniofacial dysostosis, dysostosis craniofacialis[2], syndromic craniosynostosis and premature craniosynostosis.[3]

Its incidence is estimated at 1 in 25,000 births.[4,5] Of these cases, 67% are familial,[6] whereas 33–56% may arise as a consequence of spontaneous mutations.[5] In a study conducted by Kriberg, new mutations due to increased parental age played a significant role in the etiology of this syndrome.[6]

The condition is thought to arise due to mutation in fibroblast growth factor receptor 2 (FGFR2) and FGFR3 genes on chromosome 10[1,5,6] in both sporadic and inherited cases.[1,4–8] The severity of expression of the disease increases in successive siblings, with the youngest child being the most severely affected.[7]

Features of this syndrome include craniosynostosis, maxillary hypoplasia, shallow orbits, ocular proptosis and hypertelorism. There is considerable variation in the extent of these features, but ocular proptosis is always present even in the absence of craniosynostosis which is a prerequisite for the diagnosis of Crouzon syndrome.[4,5] Other features associated with this condition are hearing loss,[2,4,6] deviation of nasal septum,[6] calcification of stylohyoid ligament,[6] cervical spine anomalies[2,4,6] and stenosis of jugular foramen.[4,6]

Franceschetti coined the term “pseudo Crouzon syndrome” to designate those cases which simulate the Crouzon syndrome, and in which patients do not have relative mandibular prognathism, parrot nose and there is no familial occurrence.[2]

Case Report

A 13-year-old male patient reported to the outpatient department with a chief complaint of irregular arrangement of his teeth. The boy had large eyes at the time of birth and they became more prominent as he grew [Figure 1a]. His parents had noticed the discrepancy in the size of the jaws at the age of 6 months and the lower jaw was forwardly placed when compared to the upper jaw [Figure 1b]. His medical history was not contributory. Family history revealed 2nd degree consanguity, with the same problem in his mother.

On extraoral examination, there was presence of trigonocephaly, concave profile, maxillary retrognathism, malar deficiency [Figure 1b], shallow orbits, ocular proptosis and hypertelorism. Intraoral examination revealed Angle’s class III molar relation with a negative overjet in the anterior tooth region [Figure 1c]. The dental arches were “U” shaped with bilateral posterior crossbite. Maxilla showed a high vault palate with lateral palatal swellings [Figure 1d, Chart 1].
Radiographic examination revealed the presence of impacted teeth 13, 17, 23, 27 in panoramic radiograph [Figure 1e]. The lateral skull radiograph showed beaten metal appearance in the periphery [Figure 1f].
Considering the family history, the patient’s mother was also reviewed. On general examination, her gait and posture were normal and she was moderately nourished. On extraoral examination, she showed brachycephaly [Figure 2a], concave profile [Figure 2b], maxillary retrognathism [Figure 2c], decreased malar prominence, shallow orbits, ocular proptosis and hypertelorism. Intraoral examination revealed anterior open bite [Figure 2c], Angle’s class III molar relation, crowding of the maxillary anterior teeth and lateral palatal swellings [Figure 2d]. Orthopantomograph

**Chart 1**: Pedigree chart. Note: solid blocks are affected with Crouzon syndrome

**Figure 2a**: Frontal view of case 2 showing concave profile, ocular proptosis and hypertelorism

**Figure 2b**: Lateral profile of case 2 showing maxillary deficiency with relative mandibular prognathism

**Figure 2c**: Intraoral photograph of case 2 showing a negative overjet in the anterior region

**Figure 2d**: Photograph of case 2 showing high arched palate with lateral palatal swellings

**Figure 2e**: Orthopantomograph of case 2 showing prominent gonial angles
revealed prominent gonial angles [Figure 2e]. Lateral skull showed characteristic copper beaten appearance [Figure 2f].

Discussion

The correct identification of a craniosynostosis syndrome necessitates an extensive physical and radiographic examination to detect all the associated anomalies. Craniosynostosis commonly begins during the first year of life and is usually completed by the age of 2–3 years. Abnormalities of calvarial shape in Crouzon syndrome are dependent on the sutures involved. Premature fusion of synchondroses of cranial base, subsequent lack of bone growth perpendicular to the synchondroses and cranial base produces characteristic cranial shapes like brachycephaly, trigonocephaly, and scaphocephaly. The most severely affected patients can demonstrate a “clover leaf” skull (Kleeblatt schadel deformity). In the cases reported above, the child had a trigonocephalic head and mother showed a brachycephalic head.

Exophthalmosis is stated to be a prerequisite for Crouzon syndrome and is said to be caused by a lack of forward sutural growth in the temporal and cranial base region. This produces a relative prominence of eyeball, which sometimes results in blindness due to increased intracranial pressure. The cases reported above showed prominent exophthalmos with normal vision.

Hypertelorism which was seen in our cases is a prominent finding in the affected individuals and is thought to arise due to decrease in growth of the sphenozygomatic and sphenotemporal sutures.

In Crouzon syndrome, maxilla is hypoplastic, which causes relative mandibular prognathism. Underdevelopment of maxilla is most severe in premaxillary area, causing crowding in the maxillary anterior teeth region. Both of our cases showed these features.

Patients with Crouzon syndrome show a high vault palate sometimes associated with lateral swellings. Both our cases showed these findings.

Cranial markings, a normal finding in growing individuals, are seen more prominently in patients with Crouzon syndrome because of increased intracranial pressure from the growing brain. These markings may be seen as multiple radiolucencies giving a beaten metal appearance in the periphery, which was classically seen in our case.

By the early recognition of the syndrome, the sutures can be guided before their fusion or artificial sutures can be placed to allow for the growth of brain. Patients reporting after cessation of growth can be subjected to Le Fort III osteotomy to correct maxillary retrusion and associated hypoplasia of infraorbital and malar regions. Forward movement of infraorbital margin reduces the degree of proptosis. Spectaclesplasty followed by a Le Fort I maxillary osteotomy at skeletal maturity also yields better esthetic results. In our cases, the child was advised to go for surgical rapid maxillary expansion followed by Le Fort I osteotomy and his mother was advised to go for posterior segmental osteotomy followed by Le Fort I osteotomy to correct the dentofacial anomalies.

To conclude, Crouzon syndrome should be managed as early as possible as it results in poor cosmetic appearance and results in other complications like mental retardation, airway obstruction and decreased visual acuity as the age advances.

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