Crouzon's Syndrome-Clinico-Radiological Illustration of A Case

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Abstract- Crouzon's syndrome [CS], an autosomal dominant disorder with complete penetrance and variable expressivity is caused by mutation in the fibroblast growth factor receptor 2 (FGFR2) gene. Normally, the sutures in the human skull fuse after the complete growth of the brain, but if any of these sutures close early then it may interfere with the growth of the brain. The disease is characterized by premature synostosis of coronal and sagittal sutures which begins in the first year of life. Case report of an 8 year old boy is presented with characteristic features of Crouzon's syndrome. The patient presented with exophthalmos, hypertelorism, beaked nose, deviated nasal septum,high vault palate, crowding in maxillary premolar region, copper-beaten appearance of skull and diffuse indentation of inner table of skull. In addition to the classical features an interesting finding was the presence of prominent commissural lip pits.

Keywords- Crouzon syndrome, craniosynostosis, exophthalmos, commissural lip pits.

Introduction

In 1912, French neurosurgeon Octave Crouzon first described the hereditary syndrome of craniofacial synostosis, which includes a triad of skull deformities, facial anomalies, and exophthalmos and is now known as Crouzon syndrome.[1].It is one of the varieties of craniofacial dysostosis caused by premature obliteration and ossification of two or more sutures, most often coronal and sagittal.[2]

Crouzon's syndrome is also referred to as craniofacial dysostosis, hereditary craniofacial dysostosis, dysostosiscraniofacialis, syndromiccraniosynostosis and premature craniosynostosis. The condition is thought to arise due to mutation in fibroblast growth factor receptor 2 (FGFR2) and FGFR3genes on chromosome 10. Its incidence is estimated at 1 in 25,000 births. Of these cases, 67% are familial, whereas 33–56% may arise as a consequence of spontaneous mutations. Kriberg conducted a study where he described that new mutations due to increased parental age may play a significant role in the etiology of this syndrome.[2,3] Crouzon syndrome makes up approximately 4.8% of all cases of craniosynostoses. No known race or sex predilection exists. However, when the craniosynostosis is of sagittal or metopic types, the predominance increases in boys, while coronal craniosynostosis is more common in girls.The condition is usually detected in the first year of life. However,there are also congenital premature forms in which thesynostosis begins inside the uterus and is evident at birth with facial deformities.[1]

The differential diagnosis of Crouzon's syndrome includes simple craniosynostosis as well as Apert syndrome, Carpenters syndrome, Saethre-Chotzen syndrome, Pfeiffer syndrome.[3]CS can be distinguished from other craniosynostosis syndromes by lack of hand and/or foot abnormalities.1

Case Report

An 8-year-old male patient came to our department with the chief complaint ofdecayed teeth associated with pain in right and left, upper and lower back tooth region since past 6 months.Patient had not undergone any previous dental treatment. Review of medical history was unremarkable. There were no anomalies in any siblings or near relatives. The boy had large eyes at the time of birth and they became more prominent as he grew.

On extraoral examination, there was presence of dolicocephalic head [Fig A],straight facial profile [Fig B], exophthalmos, hypertelorism, beaked nose, wide nasal bridge, deviated nasal septum and malar deficiency. [Fig C]. Commissural lip pits were prominent in our patient. [Fig D]

Intraoral examination revealed high arched palate and crowding in right maxillary premolar region [Fig E]. Dental arches were "U" shaped. There was a negative overjet in the anterior tooth region [Fig F] and Angle's Class III malocclusion bilaterally.

The patient was subjected to radiographic investigations. The orthopantomogram revealed crowding in maxillary premolar region on right and left side and absence of right and left second mandibular premolar tooth buds. [Fig G]

PA skull and lateral skull view demonstrated cranial markings which were seen as multiple radiolucencies giving a beaten metal appearance. [Fig H].

Computed tomography (CT) image of the skull showed diffuse indentation of the inner table of skull [Fig I]. Cone Beam Computed Tomography 3-D view shows the profile of the patient [Fig J].

The clinical, dental, ophthalmologic features, and radiographic findings pointed to the diagnosis of Crouzon syndrome.

Discussion

Craniofacial abnormalities are often present at birth and may progress with time. Extensive physical and radiographic examination is required to identify a craniosynostosis syndrome. Craniosynostosis commonly begins during the first year of life and is usually completed by the age of 2–3 years.[2] With the advent of molecular technology, the gene for Crouzon's syndrome could be localized to the fibroblast growth factor receptor II gene (FGFR 2) at the chromosomal locus 10q 25.3-q26, and more than 30 different mutations within the gene have been documented in separate families.[4]Abnormalities of calvarial shape in Crouzon syndrome are dependent on the sutures involved. Premature fusion of synchondroses of cranial base,] and

subsequent lack of bone growth perpendicular to the synchondrosesleads to characteristic cranial shapes like brachycephaly, trigonocephaly, and scaphocephaly. The most severely affected patients can demonstrate a "clover leaf" skull (Kleeblattschadel deformity). In the case reported above, the child had adolicocephalic head which could be due to premature fusion of saggital suture.[2]

Exophthalmos 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 case 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. Other clinical features in a patient with this syndrome include strabismus, deviated nasal septum, narrowed or obliterated anterior nares, wide parrot beaked nose, rhinolalia, short upper lip, maxillary hypoplasia and relative mandibular prognathism with no digital abnormalities. There can be occasional upper airway obstruction.Hearing loss may be there which is usually conductive.[1,2,3,4,5] Our patient had beaked nose, wide nasal bridge, deviated nasal septum and malar deficiency.

An interesting finding in our patient was the presence of very prominent commissural lip pits which has never been reported in any case of crouzonsyndrome before to the best of our knowledge.

In Crouzon syndrome, underdevelopment of maxilla is most severe in premaxillary area, causing crowding in the maxillary anterior teeth region.[2] In our patient, crowding could be appreciated in maxillary premolar region.

Patients with Crouzon syndrome show a high vault palate, as was seen in our case. Sometimes it is associated with lateral swellings.[2]

Sometimes cleft palate and bifid uvula can be seen in patients. Occasional oligodontia, macrodontia, peg-shaped, and widely spaced teeth have been reported.[5] In our patient the tooth-buds of mandibular second premolars were absent.

Cranial markings are seen more prominently in patients with Crouzon syndrome because of increased intracranial pressure from the growing brain.[1,2,6,7] These markings may be seen as multiple radiolucencies giving a copper-beaten appearance which was classically seen in our case. Indentations in the inner table of the skull were present.

Enlarged hypophyseal cavity, small paranasal sinuses, abnormal craniocervical junction, butterfly-shaped vertebrae, and fusion of cervical vertebrae (C2-C3 and C5-C6), calcification of stylohyoid ligament and stenosis of jugular foramenhave also been reported in patients suffering from this syndrome.[1,2]

Mental retardation may be present because of premature fusion of cranial sutures[8] but our patient showed normal intelligence.

Prenatal real-time ultrasonographic diagnosis of exophthalmus has been reported in literature. Diagnosis was made at the 35th week of gestation in a fetus of a patient affected with Crouzon syndrome. Recognition of exophthalmus as a part of Crouzon syndrome and the easy visualization of the eye balls and palpebrae in the third trimester made the diagnosis possible.[9]

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. "Spectacles" is a reference to the bilateral circumferential periorbital bony skeleton, and a spectaclesplasty is a differential rotation and advancement of this complex. Ilizarov Procedure i.e. craniofacial disjunction, followed by gradual bone distraction has been reported to produce complete correction of exophthalmos and improvement in the functional and esthetic aspects of the middle third of the face without the need for bone graft in patients aged 6-11 years.[1,2,5,6,10]

Our patient was referred to the department of paedodontics for restoration of the teeth. Orthodontic treatment was advised to prevent the developing malocclusion. He was also advised to consult the oral and maxillofacial surgeon to reduce his skull deformity so as to prevent complications which may arise as a result of increased intracranial pressure. We expect him to have a normal and healthy life span after all his treatment is completed.

Conclusion

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. In our patient there was presence of commissural lip pits which is an interesting intraoral finding as it has never been reported in this syndrome before. More case reports are required to consider commissural lip pits as a consistent finding.

References

References are available on request
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