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Crouzon syndrome – Role of a dentist

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Abstract

Crouzon syndrome is a genetic disorder characterized by premature closure of one or more cranial sutures resulting in a variable number of craniofacial abnormalities. It is frequently diagnosed on the basis of clinical features. However, radiographic examination plays adjuvant role and support the clinical diagnosis. The management includes a team approach in which each organ system is addressed independently. This article describes a case of Crouzon syndrome in a 21-year-old girl. Dental considerations are discussed in detail.

Keywords

craniosynostosis; crouzon syndrome; hypoplasia; proptosis

Introduction

Crouzon syndrome (CS) is a rare genetic disorder characterized by premature closure of one or more cranial sutures and produces the characteristic craniofacial and other associated abnormalities [1, 2]. It is one of the craniosynostosis syndrome that is caused by a mutation in the fibroblast growth factor receptor 2 gene (FGFR2) [1]. Although CS is inherited as an autosomal dominant trait, many cases are sporadic and present as de novo mutations arising from unaffected parents. It has a worldwide prevalence rate of approximately 1 per 25,000 live births [1, 2]. This syndrome comprised of a triad of skull deformities, facial anomalies, and exophthalmos. Nevertheless, manifestations of CS may vary in severity from a mild presentation with just midface hypoplasia to severe forms like early fusion of multiple cranial sutures and thus related several complications [3]. Treatment protocol vary depending on the age, signs, symptoms and the severity of the condition in a particular patient.

The management of CS requires a multidisciplinary approach for successful outcome. Crouzon syndrome almost always requires surgery to enable the skull to expand properly and to align the jaws, along with numerous other surgeries to repair face, orbital and ear defects In the first year of life, it is preferred to release the synostotic sutures of the skull to allow adequate cranial volume thus allowing for brain growth and expansion. Skull reshaping may need to be repeated as the child grows to give the best possible results. If necessary, midfacial advancement and jaw surgery can be done to provide adequate orbital volume and reduce the exophthalmus to correct the occlusion to an appropriate functional position and to provide for a more normal appearance. Prognosis depends on malformation severity [1,2,8].

As far as management of dental manifestations in growing CS patients is concerned, an attempt to

correct the maxillary hypoplasia and anterior crossbite by means of rapid maxillary expansion and protraction of the maxilla using facemask or distraction osteogenesis can be done.

Case Report

A 21 year old girl reported to the department of Oral Medicine and Radiology with the chief complaint of pain in upper left posterior region of jaw since 4-5 days. As the girl's facial appearance was not normal, medical and family history was taken in detail. Review of medical history was insignificant, specifically, her mother reported normal labor and delivery. There were no similar or any other anomalies reported in any of the family members or near relatives. No intellectual or developmental impairment was apparent. On extraoral examination, she had frontal bossing, depressed and wide nasal bridge, proptosis of the eyes, downslanting of palpebralfissures, hypertelorism, hypoplasia of the maxilla, concavefacial profile, incompetent lips, and short upperlip [Figures 1&2]. Noobvious dermatological or any other findings were observed.

Intraoral examination revealed narrow high arched palate and median diastema [Figure 3]. The chronology of eruption and eruption status of teeth was normal. There was caries with 26 for which she had reported to us. Also, there was extensive caries with 46. The radiographic evaluation of the patient was carried out. The orthopantomogram revealed presence of all permanent teeth with no obvious abnormality (except carious 26 and 46). Lateral skull view demonstrated a concave facial profile and copper beaten appearance on skull. [Figure 4]. Clinico - radiographic findings were diagnostic of CS. For a carious tooth intraoral periapical radiograph was also taken which revealed deep occlusal caries with periapical pathology which was treated by an endodontic treatment.

As an important part of the management, the counseling of the patient and her parents was done regarding the nature of the disease from which she is suffering. Patient was advised orthodontic treatment that involved correction of proclinated teeth and median diastema. Even though the patient didn't have signs and symptoms related to vision and cranial problems, expert opinion of the ophthalmologist and neurosurgeon was sought to evaluate for the same.

Discussion

CS was first described by Octave Crouzon in 1912 which bears his name to the entity. In CS, multiple sutural synostoses frequently extend to premature fusion of skull base causing midface hypoplasia, shallow orbit, and occasional upper airway obstruction [3,4]. Other clinical features include hypertelorism, exophthalmos, strabismus, beaked nose, short upper lip, maxillary hypoplasia, and relative mandibular prognathism with no digital abnormalities [5,6].

CS must be differentiated from simple craniosynostosis and other syndromic craniosynostosis. The syndromes to be considered in differential diagnosis are Pfeiffer's syndrome, Apert syndrome, Saethre-Chotzen syndrome, Carpenter syndrome, and Jackson-Weiss syndrome [2,3] CS can be differentiated from other craniosynostosis syndromes by lack of abnormalities in hand or foot or both. It is stated that unlike most other craniofacial syndromes caused by fibroblast growth factor receptor mutations, the limbs are typically unaffected in CS [1]. In the present case also characteristic absence of limb abnormality is noted.

Diagnosis of Crouzon syndrome can be done at birth by assessing the signs and symptoms of the

baby. However, radiographs, CT scans, magnetic resonance imaging, genetic testing can be used to confirm the diagnosis of Crouzon syndrome [7]. The radiographs will also evaluate the severity of the condition and thus the expected complications. Periodic radiographic examination is required to monitor progress of the disease. On conventional radiographs like skull views, typical radiographic feature is presence of beaten metal appearance which is nothing but the cranial markings of the inner surface of cranial vault [8]. These are also called as digital impressions. Such beaten metal appearance is noted in the present case also. The earliest radiographic signs of suture synostosis are sclerosis and overlapping edges [8].

The management of CS is multidisciplinary and early diagnosis is important.This condition compromises not only the function but also the mental well-being of the person.Treatment of Crouzon syndromeis complex, since there are many aspects of the syndrome to manage.Multiple surgeries are required to enable the skull to expand properly. Crouzon patients generally have a normal lifespan [9]. Prognosis depends on the severity of the condition.

Role of Dentist: Every dentist should have an adequate knowledge about CS because in few patients the dentist might be a first person to diagnose this rare entity. Accordingly he has to do counseling of the patient and his/ her parents and to coordinate the multidisciplinary team for corrective measures. With proper and timely management, these patients can be provided with better quality of life. Genetic counseling is also an important part of management.

In CS, the midline third of the face and the upper jaw are under developed, owing to the disturbance in the growth of the skull sutures. This may affect breathing, swallowing and speech.

As far as the oral cavity is concerned, the manifestations are crowding of teeth, crossbite, anterior open bite, high arched palate, cleft palate, and bifid uvula. Occassionaloligodontia, macrodontia, peg-shaped and widely spaced teeth. In the present case, high arched palate was the only intraoral finding. These patients usually exhibit an Angle Class III malocclusion due to the midface deficiency, while the mandibular growth potential is normal. Treatment might require extraction of some permanent teeth to alleviate the crowding, as well as expansion of the maxilla, either surgically or through the use of orthodontic devices.

Initiation of treatment at younger age, gives an opportunity to a patient to live normal life with negligible complications with reference to cranio-orofacial, region.

Conclusion

Craniofacial abnormalities are usually present at birth and may progress with time. Dentist should have an adequate knowledge about these rare entities including CS, as early diagnosis will aid in planning the corrective measures at younger age, and thus preventing the complications like mental retardation, airway obstruction, blindness and hearing deficit. Though the treatment approach in CS is multidisciplinary, pedodontist plays an integral role in this team.

Figures

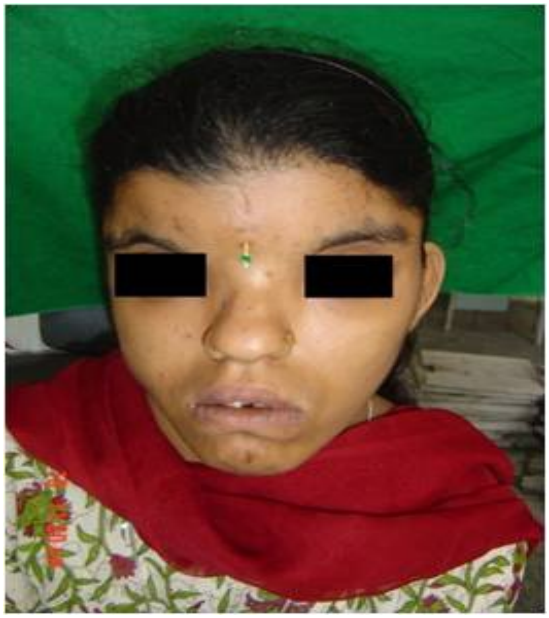

Figure 1: Front view showing down sloping palpabral fissures, frontal bossing, depressed nasal bridge and hypertelorism.

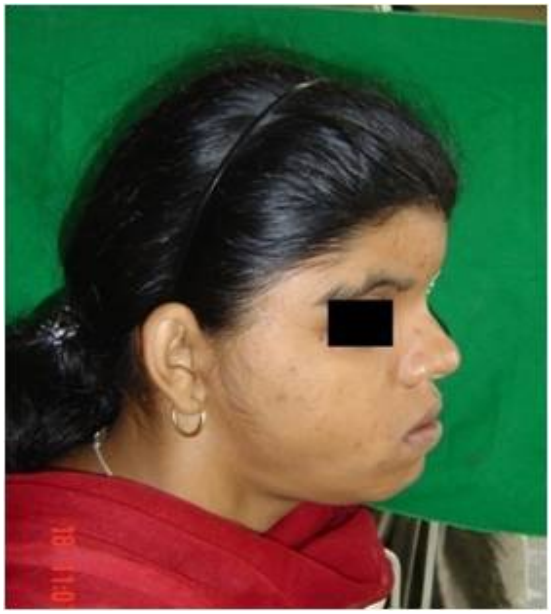

Figure 2: Lateral view showingfrontal bossing, concave facial profile.

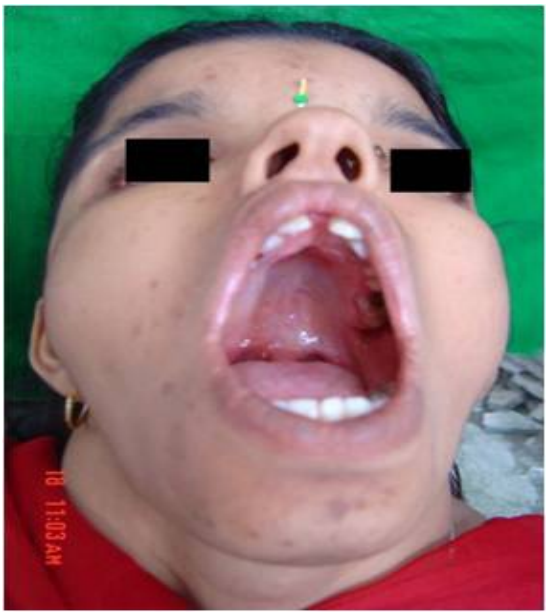

Figure 3: Intraoral photograph showing high arched palate and median diastema

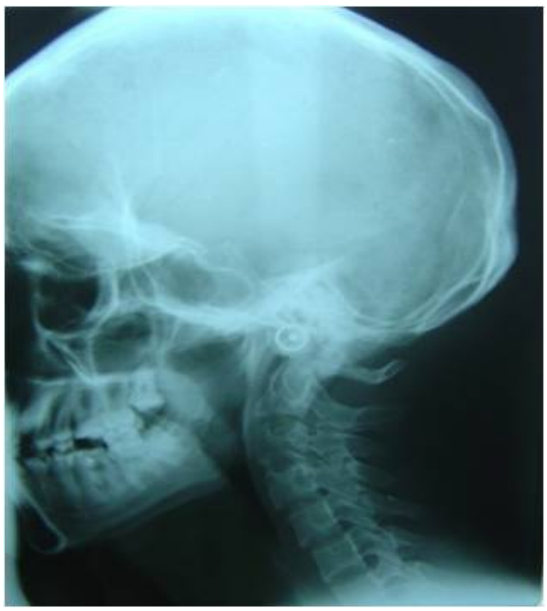

Figure 4: Lateral skull view showing convex profile and typical beaten metal appearance of skull

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