Diagnosis and Evaluation of Crouzon Syndrome

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ABSTRACT

Crouzon syndrome is an autosomal dominant condition characterized by craniosynostosis with associated dentofacial anomalies. This report describes the different clinical features in two affected individuals of different families with particular reference to characteristic findings of this syndrome.

Key words: Crouzon syndrome. Autosomal dominant. Craniosynostosis. Dentofacial anomalies.

INTRODUCTION

Crouzon syndrome is an autosomal dominant disorder characterized with pre-mature closure of cranial sutures, midfacial hypoplasia and orbital deformities.^{1,2} Crouzon syndrome with a reported incidence of 1:25000 live births is the most common of over 70 conditions in which pre-mature fusion of the cranial sutures may be a feature.³ A positive family history is reported to occur between 44-67% of cases.

The diagnosis is based on clinical findings and radiological examination. Both genders are equally affected. The condition is thought to arise due to a mutation in the Fibroblast Growth Factor Receptor-2 (FGFR-2) gene in both the sporadic and the inherited cases.⁴

Those most at risk for Crouzon syndrome are children of parents with either the menifestus disorder parents or carrier of the gene and fathers at an older age at the time of conception.

CASE REPORT

Case 1: An 11 years old boy reported with the complaint of esthetic, difficulty in chewing and snoring. On clinical examination, he was found to have brachycephaly, maxillary retrusion, malar deficiency, hypertelorism, ocular proptosis and a beaked nose. Intra-oral examination revealed a class III incisor relationship. The maxillary and mandibular arches were U-shaped with bilateral posterior crossbite. Masticatory function was normal with no evidence of temporomandibular dysfunction. He had history of sleep apnoea with upper airway problems. On radiological examination, patient

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had cervical scoliosis, (Figure 1). No other anomalies were observed. There was positive family history and the father also had frontal bossing without maxillary hyperplasia. One of his cousin was also suffering from skull deformity. In the absence of hand and feet lesions a provisional diagnosis of Crouzon syndrome was made.

Case 2: A boy, 14 years of age, reported with the complaint of esthetic, cleft in the palate and difficulty in speaking and eating. On clinical examination, patient had brachycephalic head, maxillary retrusion, malar deficiency, hypertelorism, divergent strabismus, ocular proptosis (Figure 2) and moderate mental retardation. Intra-oral examination revealed class III incisor relationship. The dental arches were U-shaped with unilateral cleft in the upper arch (Figure 3). Upper left lateral incisor and canine was missing. In the absence of hand and feet lesions, a provisional diagnosis of Crouzon syndrome was made.

The treatment of both the patients started with the correction of posterior crossbite by maxillary expansion and fixed brace (Figure 4).



Figure 1: Showing cervical scoliosis.

Figure 2: Patient with brachycephalic head showing maxillary extension malar deficiency.



Figure 3: Showing U-shaped with unilateral cleft in the upper area.



Figure 4: Fixed braces for presurgical treatment.

Case 1 received treatment with reverse pull headgear for orthopaedic effect. But the results were not successful as only dental movements were achieved. Finally, orthognathic surgery was planned for the patient which involved high Lefort II osteotomy for the correction of maxillary hypoplasia and malar deficiency.

Both the patients underwent orthognathic surgery including midfacial advancement for the correction of facial dysmorphisms. The case 2 was managed surgically in two stages. Firstly, repair of cleft palate was done and after one year, second stage was executed including orthognathic surgery.

DISCUSSION

This syndrome was originally described in 1912 by a French neurosurgeon as an autosomal hereditary disease. He described four essential characteristics including exorbitism, retromaxillism, inframaxillism and paradoxic retrogonia.

The affected individuals in this case report presented with varying degree of craniosynostosis, ocular proptosis, hypertelorism and class III malocclusion strongly suggestive of Crouzon syndrome.²

The associated features of Crouzon syndrome are given in Table I. Abnormalities of the calvarial shape in Crouzon syndrome are dependent on the sutures involved.⁵ The most common clinical appearance as seen in both the patients, is brachycephaly. Hydrocephaly and mental retardation may develop due to premature closure of cranial sutures.

Exophthalmos, which was evident in both the cases, is regarded as a universal feature of Crouzon syndrome.^{3,6} Hypertelorism was also a universal finding and is thought to arise due to a decrease in the growth of sphenozygomatic and sphenotemporal sutures.

A class III malocclusion reported in 75% of patients with Crouzon syndrome. The etiology is generally due to a retrusive and short maxilla with relative mandibular prognathism.⁶ Co-existence of Crouzon syndrome with congenital heart malformation (ventricular septal defect) is rarely reported and description of such congenital malformations co-existence is not accessible in literature.⁷

Five autosomal dominant craniosynostosis syndromes (Apert, Crouzon, Pfeiffer, Jackso-Weiss and Crouzon syndrome with acanthosis nigricans) result from mutations in FGFR genes.⁸ The TWIST gene, which is also known to cause Saether-Chotzen syndrome, serves as transcription factor and work with FGFR gene family, affect head and limb region.

Patients with Crouzon syndrome are often best cared for by a team of craniofacial experts in which professionals in plastic surgery, ear/nose/throat surgery, dentistry,

Table I: Abnormalities associated with Crouzon syndrome. ³⁻⁵
Cranium
Craniosynostosis
Brachycephaly and acrocephaly
Palpable ridge
Flat occiput
Frontal bossing
Facial Features
Maxillary retrusion
Malar deficiency
Relative mandibular prognathism
Ear
Low set ear
Conductive hearing loss
Bilateral atresia of auditory meatus
Eye
Downslanting palpebral fissure
Exophthalmos
Iris-coloboma
Ptosis
Exposure keratitis
Hypertelorism
Divergent strabismus
Nystagmus
Nose
Beaked nose
Deviated nasal septum
Mouth
Short upper lip
Class III malocclusion with maxillary crowding
High arched and narrow palate
Cleft palate and bifid uvula
Neurological
Headache
Mild to moderate mental retardation
Seizures
Musculoskeletal
Cervical spine abnormalities (scoliosis)
Calcification of stylohyoid ligament
Meniere'disease: (vertigo, dizziness, and/or ringing in the ear).
Respiratory system
Breathing difficulty
Sleep apnoea
Cutaneous
Acanthosis nigricans

orthodontics, genetics and audiology can address the patient's multiple needs.

The reported cases required repair of cleft and orthognathic surgery.

The pre-surgical orthodontics include decompensation of both the maxillary and mandibular arches. The decompensation results in more severe dental as well as skeletal class III pattern but it will help the surgeon to correct the true skeletal disharmony, which is marked by dental and soft tissue compensation.

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