

# 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.<sup>1,2</sup> 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.<sup>3</sup> 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.<sup>4</sup>

Those most at risk for Crouzon syndrome are children of parents with either the manifest 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

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).

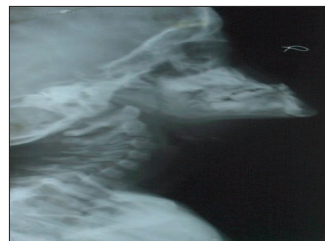


Figure 1: Showing cervical scoliosis.



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 pre-surgical treatment.

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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 paradoxical retrognathia.

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.<sup>2</sup>

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.<sup>5</sup> 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.<sup>3,6</sup> 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.<sup>6</sup> 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.<sup>7</sup>

Five autosomal dominant craniosynostosis syndromes (Apert, Crouzon, Pfeiffer, Jackso-Weiss and Crouzon syndrome with acanthosis nigricans) result from mutations in FGFR genes.<sup>8</sup> The TWIST gene, which is also known to cause Saether-Chatzen 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.<sup>3-5</sup>

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

## REFERENCES

1. Panchal J, Uttchin V. Management of craniosynostosis. *Plast Reconstr Surg* 2003; **111**:2032-48.
2. Rimoin DL, Connor JM, Peritz RE. Principles and practice of medical genetics. 3rd ed. New York: *Churcbill Livingstone*, 1997.
3. Cohen MM. Craniosynostosis and syndromes with

- craniosynostosis: incidence, genetics, penetrance, variability and new syndrome updating. *Birth Defects Orig Artic Ser* 1979; **15**: 13-63.
4. Shotelersuk V, Mahatumarat C, Ittiwut C, Rojvachiranonda N, Srivuthana S, Wacharasindhu S, *et al.* FGFR-2 mutations among Thai children with Crouzon and Apert syndromes. *J Craniofac Surg* 2003; **14**:105-7.
  5. Posnick JC. The craniofacial dysostosis syndromes: current reconstructive strategies. *Clin Plast Surg* 1994; **4**:585-98.
  6. Kreiborg S, Aduss H. Pre- and postsurgical facial growth in patients with Crouzon's and Apert's syndromes. *Cleft Palate J* 1986; (Suppl1):78-90.
  7. Rokicki W, Rokicka A. Coexistence of Crouzon syndrome with ventricular septal defect. *Wilad Lek* 2003; **56**:298-9.
  8. Preising MN, Schindler S, Friedrich M, Wagener H, Golan I, Lorenz B. On the effect of mutations of the fibroblast growth factor receptors as exemplified by three cases of craniosynostosis. *Klin Monatsbl Augenbeilkd* 2003; **220**:669-81.

