I J Pre Clin Dent Res 2014;1(3):77-80
July-September

International Journal of Preventive & Clinical Dental Research

Crouzon Syndrome: An Unusual Genetic Trait

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Abstract

Crouzon syndrome is rare developmental deformity, which is described in the clinical entity of craniosynostosis. These are heterogeneous group of conditions, characterized by premature fusion of cranial sutures. The concern relating these cases is primarily esthetics with problems ranging from mild to severe variability. It is important to treat these patient as early as possible for Functional, esthetic and psychological reasons. A series of cases has been reported below which were diagnosed as Crouzon's syndrome based on clinical and radiologic findings.

Key Words

Craniofacial; craniosynostosis; crouzon syndrome

INTRODUCTION

Craniofacial anomalies represent a distinct group of deformities involving the craniofacial skeleton. Although unusual, it usually effects not only the normal functioning of the effected individual but also their social well being.[1] Premature fusion of one or more sutures due to lack of growth at the fused suture and overgrowth at the non fused suture is described as craniosynostosis.[2] Craniosynostosis frequently includes Apert, Crouzon, Pfeiffer, Carpenter, and Saethre-Chotzen. Recognizing and treating the problem is important as it can lead to numerous complications.[3] Crouzon's syndrome represent a rare genetic trait first described in 1912 by a French neurosurgeon Octave. The condition also termed as Type I Crouzon's Disease /Craniofacial dysarthrosis /craniofacial dysostosis.[4] It commonly effects the craniofacial region and is usually diagnosed during birth or early childhood.[5] The incidence is 1:25000 live birth with both gender equally effected and with a positive family history. The mutation in fibroblast growth factor 2 gene (FGFR2), is responsible for the deformities observed. Diagnosis is made based on clinical history and radiographic examination.[6]

CASE REPORTS

CASE 1

A 4 year old female patient reported to our Dental Hospital accompanied by her mother, with a chief complaint of chipping in the upper front teeth since 3 months. Her medical history revealed that she had undergone Tarsorraphy 3 years ago following avulsion of her left eye and reported history of wheezing since 2 years and is on homeopathic medicines. History of snoring present since early childhood. There was no significant family history; no adverse or deleterious habits. On general physical examination no syndactyly of fingers or toes were noted. On extra oral examination, patient had a concave profile with brachycephaly associated with gross asymmetry, nasal bridge was depressed, aduncous nose giving a parrot beak appearance, hypertelorism with exopthalmosis of eyes were seen (Fig 1 & Fig. 2). History from her parents revealed that these features were present since birth. On intraoral examination brownish discoloration was seen along the mesial surface of 51, 61. Mesial terminal plane with anterior cross bite was seen. There was no abnormality detected in the soft tissues. On palpation inspectory findings were confirmed (Fig. 3).

CASE 2 & 3:

Two Siblings from the same family reported to dental hospital after a month for routine dental checkup. On extra oral examination both had mild brachycephaly, maxillary retrusion, malar deficiency,

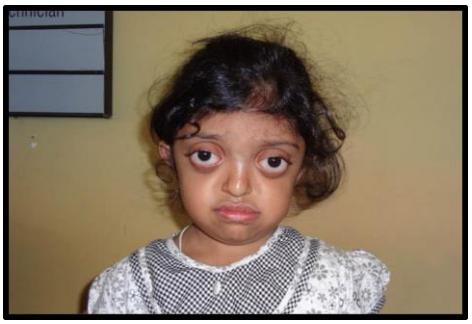

Fig. 1: Extra-oral frontal view

Fig. 2: Extra-oral lateral view

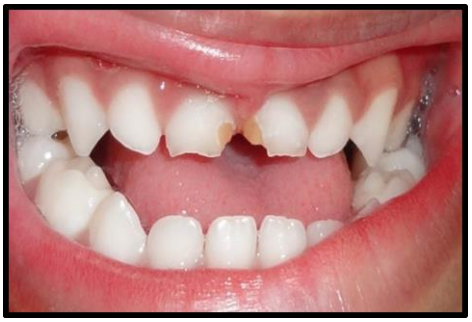

Fig. 3: Intra-oral view

Fig. 4: Extra-oral frontal view

Fig. 5: Extra-oral lateral view

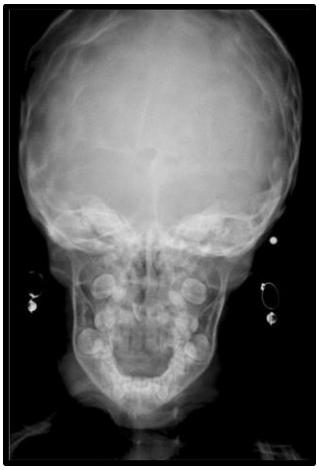

Fig. 6: Posterio-anterior view

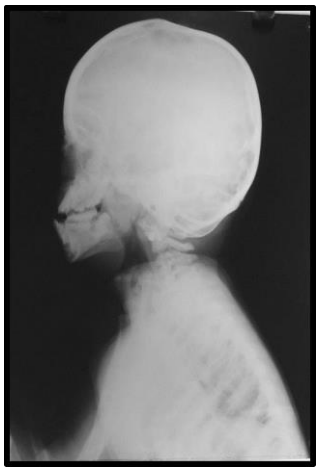

Fig. 7: Lateral skull view

hypertelorism, mild ocular proptosis and a beaked nose (Fig. 4 & Fig. 5). Intra-oral examination revealed a Class III incisal relationship with an anterior open bite from teeth 13 to 23.U-shaped arch with mild crowding of the lower arch was noted. Masticatory function was normal with no evidence of temporomandibular dysfunction. No other anomalies such as polydactyl or syndactyl were noted. Provisional diagnosis for all these three cases was given as Crouzon syndrome. Comprehensive history of the family was taken to discover if any other members were affected. While in case 1 this was the first incidence in the family, cases 2 & 3 reported with a positive family history where Mother and her father were also known to have similar facial appearance. Differential diagnosis of Apert syndrome, Pfeiffer, Carpenter and Sayre-Chotzen syndrome which has similar clinical presentation were considered. Mutations in FGFR2 are observed in these conditions which lead to the development of synostosis of the cranial sutures and Symmetric syndactylus of the hands and feet generally involving the second, third and fourth

Table I: Abnormalities associated with Crouzon syndrome

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

digits. These features were not elicited in our case. The Apert syndrome which has similar clinical features was ruled out because it was also associated with malformation of hands and feet, with symmetric syndactylus generally including the second, third and fourth digits. Radiographs of the skull in all the 3 cases revealed obliteration of sagittal and coronal suture. In posteroanterior view due to compression of the developing brain on the fused bone a hammered-silver ('beaten metal/ copper beaten') appearance was seen. On lateral Cephelogram hypophyseal cavity enlargement, small paranasal sinuses and the maxillary hypoplasia with shallow orbits, and relative mandibular prognathism were noted (Fig. 6 & Fig. 7). Treatment plan was made which included restoration of 51 and 61 in case 1 and a multidisciplinary approach to relieve the intracranial pressure and restore facial symmetry. A ventriculoperitoneal shunting (V-P shunt), by a neurosurgeon along with Cranial vault contouring and monobloc advancement of midface with LeFort I advancement followed by Orthodontic correction of the occlusion was planned in all the 3 cases.

DISCUSSION

A normal birth is always regarded as a reasonable, normal, occurrence. But when a deviation occurs for any reason; the malformation is received with a sense of fear and horror, but at the same time as an evincing example of the power of God. Crouzon's syndrome can be described as triad of cranium deformities, facial anomalies and exophthalmia. French neurosurgeon Crouzon in 1912 first described this condition as an autosomal hereditary disease with four characteristics features: exorbitism, retromaxillism, inframaxillism and paradoxic retrogonia.[7,8] The clinical manifestation of a case of Crouzon's syndrome can vary in severity ranging from mild midface deficiency to severe forms presenting as fusion of multiple cranial sutures to obvious midface and eye problems. Acute respiratory distress can lead to upper airway obstruction. Mental retardation is rarely seen.[7,8] Blindness can occur if increased intracranial pressure leading to optic atrophy is not treated.[4] The associated features of Crouzon syndrome are given in Table I.[9-10] This paper reports on the variable craniofacial findings in Crouzon syndrome in correlation with radiological findings which help in arriving at quicker diagnosis which is also quite economical. 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, orthodontics, genetics and audiology can address the patient's multiple needs.

CONCLUSION:

The degree of abnormalities may vary from case to case as well as between members affected in the same family. The suture fusion order and range determine the degree of deformity and inability. The early diagnosis of Crouzon's syndrome is critical to

avoid cranial hypertension, visual disturbances, blindness, etc. Therefore, it is important to closely monitor patients diagnosed with Crouzon's syndrome and an early multidisciplinary approach with specific treatment for each condition to prevent late diagnosis effects.

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