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Clinicoradiological Correlation of Crouzon Syndrome – A Case Report

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Authors' contributions

This work was carried out in collaboration between all authors. Author PVA designed the study, performed the statistical analysis, wrote the protocol and wrote the first draft of the manuscript. Authors FM and AP managed the analyses of the study. Author DKSL managed the literature searches. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

Crouzon's syndrome is a rare autosomally dominant genetic disorder with complete penetrance and variable expressivity. In 1912 a French neurosurgeon first described this disorder. There is a mutation in the fibroblast growth factor receptor 2 (FGFR2) gene which causes this syndrome. Crouzon's syndrome is characterised by premature closure of sutures in the skull predominantly involving coronal and sagittal suture resulting in interference in the growth of the brain.

Keywords: Crouzon's syndrome; fibroblast growth factor; craniofacial synostosis; copper beat appearance.

1. INTRODUCTION

The term "craniofacial anomaly" implies all congenital deformities of the cranium and face. More specifically, they interfere with the physical and mental well-being of the patient. Most craniofacial dysmorphisms occur due to inherited mutations and aberrant environmental

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modulation of multiple genes. Crouzon's syndrome is a rare autosomally dominant genetic disorder with complete penetrance and variable expressivity. In 1912 a French neurosurgeon first described this disorder [1]. There is a mutation in the fibroblast growth factor receptor 2 (FGFR2) gene which causes this syndrome. Crouzon's syndrome is characterised by premature closure of sutures in the skull predominantly involving coronal and sagittal suture resulting in interference in the growth of the brain [2-3]. There is a restriction on the growth potential of the sutures once the sutures are closed. Intraoral manifestations are mandibular prognathism, reverse overjet, V-shaped narrow high maxillary arch, cleft palate and bifid uvula. Occasional oligodontia, macrodontia, peg-shaped and widely spaced teeth have been reported. The incidence of Crouzon syndrome is approximately one in 25,000 births worldwide. It constitutes 4.8% of all cases of craniosynostoses. Crouzon syndrome does not have any gender predilection.

2. CASE REPORT

A patient 26 years' old female came with the complaints of deposits in all the teeth for the past ten years. Presenting illness revealed that patient was apparently normal ten years back after which the patient noticed whitish deposits in all the teeth which gradually increased to attain the present state in the past 10 years. No history of bleeding gums and bad breath. On general examination, the patient was of short stature. No relevant familial history. On extraoral examination, frontal bossing present. Eyes do not point in the same direction, increased intra ocular distance, abnormal protrusion of the eyeball out of the orbit was present. Incompetent lips present. Maxillary hypoplasia and relative mandibular prognathism present. Hands and feet appeared to be normal. On hard tissue examination, the presence of generalised supragingival calculus and subgingival calculus was present. Presence of anterior open bite with posterior crossbite. Angle's class III malocclusion were present.

The patient was subjected to radiographic investigations. On radiographic investigation, OPG revealed anterior open bite. Lateral cephalograph revealed hypoplastic maxilla with shallow orbits, small paranasal sinuses. A standard impression of the gyri on the inner table of the skull, predominantly in the anterior part of the calvarium resulting in hammered silver/beaten metal/ copper beaten appearance. PA cephalograph revealed increased thickness in the outer and inner table of the skull, copper beat skull appearance, anterior open bite.

Correlating the clinical features, positive family history and radiological features and it is finally diagnosed as crouzon syndrome.

Fig. 1. Photograph of the patient showing low set ears, parrot beaked nose, maxillary hypoplasia, relative mandibular prognathism, lips incompetent

Fig. 2. Photograph of the patient showing trigonocephaly, maxillary hypoplasia, lip incompetent present

Fig. 3. Hands appeared to be normal

3. DISCUSSION

Crouzon syndrome was first described by a French neurologist Octave Crouzon, in 1912. Crouzon syndrome is an autosomal dominant genetic disorder with complete penetrance and variable expressivity, which is characterised by abnormal fusion between bones in the skull and face, which results in an abnormally shaped head and face [4]. At birth no phenotypic features of Crouzon syndrome are appreciable. The clinical features are gradually noticed during the first few years of life. With the advent of molecular technology, the gene for the Crouzon's syndrome could be localised to the Fibroblast Growth Factor Receptor II gene (FGFR2) at the chromosomal locus 10q 25.3-q26, and more than 30 different mutations within the gene have been documented in separate families [5-7]. Premature fusion of the cranial sutures result in craniosynostosis, and this initiates changes in the brain and adjoining structures, such as an increase in intracranial pressure, reduced orbital volume, exophthalmos (proptosis), severe maxillary hypoplasia and occlusal derangement. Complications of Crouzons syndrome may include conjunctivitis or keratitis, luxation of the eye globes, exotropia, poor vision due to optic atrophy and corneal injury, blindness. Frequent headaches, seizures, mental deficiency, increasing hydrocephaly, conductive hearing deficit, upper airway obstruction develop secondary to septal deviation, midnasal abnormalities, choncal abnormalities and nasopharyngeal narrowing. Ultrasonic prenatal diagnosis of exophthalmos has been reported, which might give a clue regarding the forthcoming developing problems [8]. Thorough clinical and radiological analyses are required for early recognition and diagnosis of Crouzon syndrome. Posteroanterior radiographs reveal obliterated sutures, hypoplastic maxilla with shallow orbits, shortened cranial fossa, enlarged hypophyseal cavity, and small paranasal sinuses. Prominent cranial markings on the inner surface of cranial vault may be seen as multiple radiolucencies appearing as depressions resulting in hammered silver/beaten metal/ copper beaten appearance [8]. A copper beaten skull was seen in the radiograph of our patient indicating internal remodelling of the calvaria due to an increase in intracranial pressure, as a result of premature cranial suture fusions. Differential diagnosis with features of craniosynostosis such as Pfeiffer's syndrome, Apert syndrome, Saethre–Chotzen syndrome, Carpenter syndrome and Jackson–Weiss syndrome. All these involve craniofacial abnormalities, as well as other abnormalities including the hands or feet. Patients with Crouzon syndrome usually have an average lifespan [9,10].

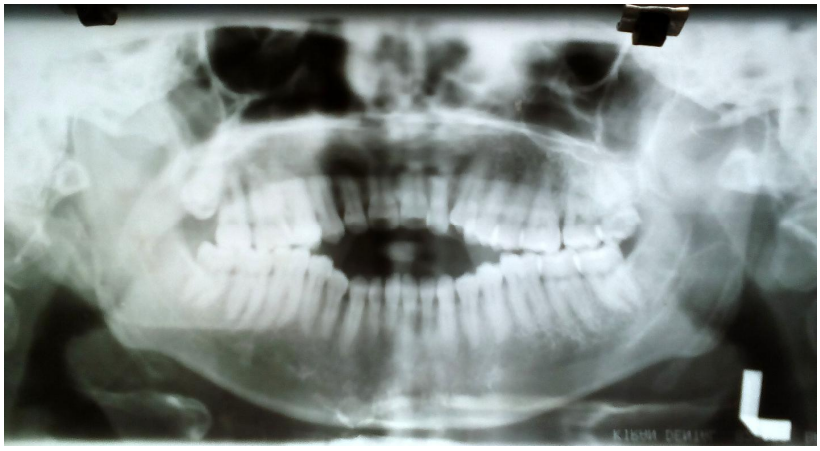

Fig. 4. OPG of the patient showing anterior open bite

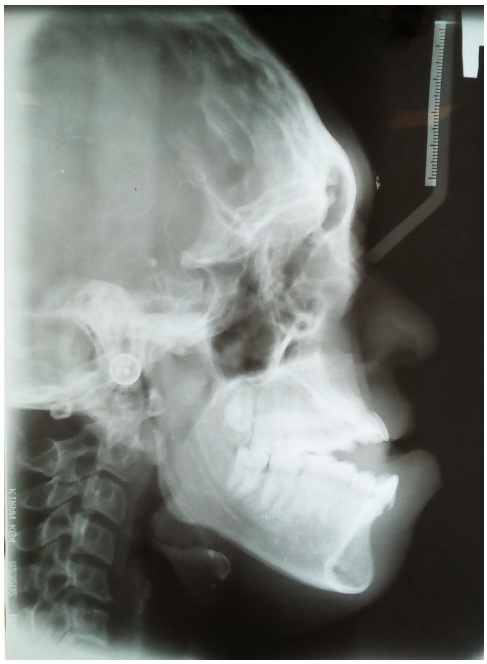

Fig. 5. Lateral cephalograph radiograph showing copper beaten appearance, maxillary hypoplasia, relative mandibular prognathism, anterior openbite

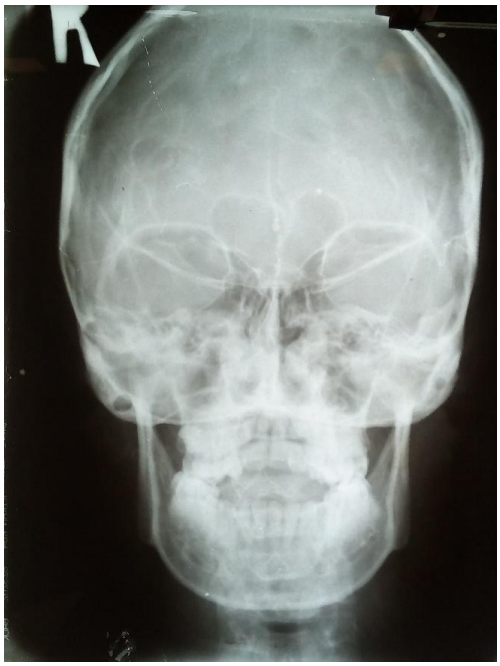

Fig. 6. PA cephalograph radiograph showing copper beaten appearance, small paranasal sinuses, small orbits, anterior openbite

Management of Crouzon syndrome is multidisciplinary and early diagnosis paves the way for the betterment of the patient. 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. Orthodontic evaluation should begin at an early age in these children. Early intervention would help in correction of the developing anterior crossbite, the development of skeletal class III deformity, and associated functional abnormalities. The orthopaedic force of the rapid maxillary expansion (RME)-facemask therapy stimulates cellular activity in circummaxillary sutures and maxillary tubercula, and this facilitates maxillary forward displacement [10]. Surgical intervention is usually required for midfacial advancement to improve the sagittal discrepancies and to relieve the intraocular pressure, thus improving the aesthetics and function of the patient.

4. CONCLUSION

The dentist must have a thorough knowledge about the craniofacial synostosis. Genetic counselling can be carried out. Early diagnosis and planning for the treatment is crucial in the management aspect of crouzon syndrome as it involves a multidisciplinary approach.

CONSENT

As per international standard or university standard, patient's written consent has been collected and preserved by the authors.

ETHICAL APPROVAL

As per international standard or university standard written ethical permission has been collected and preserved by the authors.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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