CASE REPORT

Diagnosis and Evaluation of Crouzen Syndrome: A Rare Case Report

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Abstract

Background: Crouzen syndrome is a rare genetic disorder characterized by distinctive malformations of the skull and facial region. Premature cranial suture is the most common skull abnormality. Considering the general paucity of cases in the Indian literature, we present a case report of a 18-year-old male having all the features of classical crouzen syndrome.The differential diagnosis of the condition and treatment options are discussed. Although the syndrome has typical clinical features, the relative rarity of the condition still poses a diagnostic dilemma.

Keywords: Craniosynostosis, crouzens syndrome, craniofacial dysostosis, hypertelorism

Introduction

Cranial skeletogenesis is unique. The cranial skeleton is composed of an assortment of neural crest and mesoderm- derived cartilages and bones. The craniosynostosis are a heterogeneous group of syndromes characterized by a premature sutural fusion that occurs individually or relating to other anomalies.[1-3] Crouzon syndrome, is an autosomal dominant disorder with complete penetrance and variable expressivity. Described by a French neurosurgeon in 1912, it is a rare genetic disorder characterized by premature closure of cranial sutures, midfacial hypoplasia, and orbital defects. The triad is composed of cranial defects, facial anomalies and exophthalmus.[4] The Disease of Crouzon may be distinguished from the simple craniosynostosis for its association with facial

malformations. The condition is thought to arise due to a mutation in the Fibroblast Growth Factor Receptor-2 (FGFR-2) gene. The diagnosis is based on clinical findings and radiological examination. The disease is characterized by premature synostosis of coronal and sagittal sutures. Once the sutures become closed, growth potential to those sutures is restricted. However, multiple sutural synostosis frequently extend to premature fusion of skull base causing midfacial hypoplasia, shallow orbit, maxillary hypoplasia, and occasional upper airway obstruction.

Intraoral manifestations include mandibular prognathism, overcrowding of teeth, and V-shaped maxillary dental arch. Narrow, high, or cleft palate and bifid uvula can also be seen. Occasional oligodontia, macrodontia, peg-shaped, and widely spaced teeth have been reported.[5]

Case report

An 18 years old boy reported with the complaint of aesthetic, difficulty in chewing.

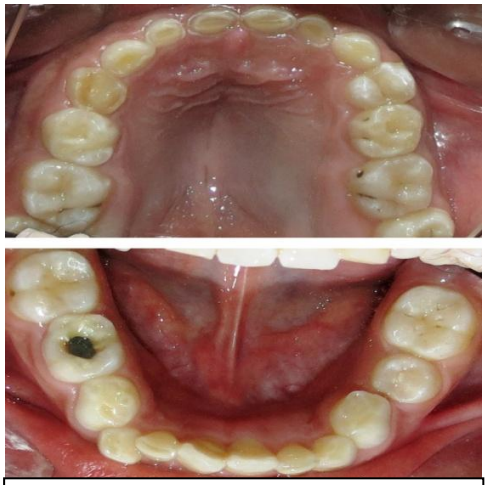

Figure1:Over-retained deciduous teeth with missing permanent teeth.

On examination brachycephaly, maxillary retrusion, malar deficiency, hypertelorism, ocular proptosis and a beaked nose were noted in patient. Medical history was insignificant and the patient appeared to have normal mental level. Intra-oral examination revealed presence of all the primary teeth except for 4 permanent molars. All the primary teeth were attrided and class III incisor relationship. The maxillary and mandibular arches were U-shaped [figure-1]. Masticatory function

was normal with no evidence of temporomandibular dysfunction.

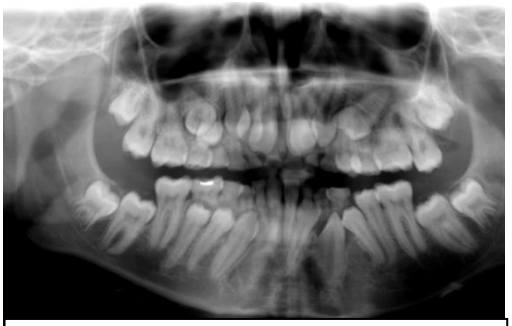

Figure2: OPG shows multiple impacted permanent teeth and retained deciduous

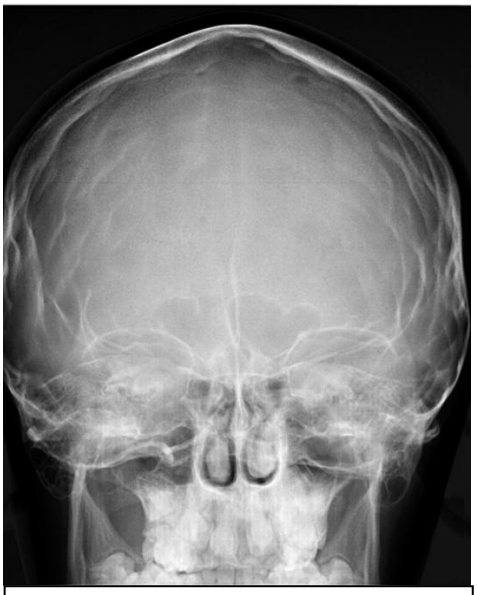

Figure3: PA Skull radiograph showing beaten metal appearance.

Orthopantomograph showed patient had multiple impacted teeth in the maxillary and mandibular arches with over retained deciduous teeth [figure-2].On radiographic examination of PA skull, early closure of the sutures was soon giving a beaten metal appearance on the radiograph [figure-4].

Ophthalmic examination showed:

- Gross proptosis that measured 20mm on exophthalmometry.
- Incomplete lid closure.
- Interpupillary distance was 68mm.
- Color vision testing was normal.
- Pupils were equally round and reactive to light and accommodation without afferent pupillary defect.
- Intra occular pressure was 15 mm HG OU by Goldman appliance.
- Dilated fundus examination was found to be normal.
- Shallow orbits.

On basis of classical clinical and radiographic features, diagnosis of gorlin- goltz syndrome was made. Our patient reported late with the syndrome and was never treated surgically. His oral hygiene was maintained using various oral

prophylaxis techniques. In order to improve aesthetics surgical exposure of impacted teeth with orthodontic correction is advised. We expect him to have a normal and healthy life span after all his treatment is completed.

Discussion

Overview and etiopathogenesis

The Crouzon's Syndrome is the most frequent of the craniofacial diseases and is characterized for being a rare genetic disorder that can be diagnosed upon the birth or during the childhood. The dominant transmission range is of 100% and the large scale penetrance with phenotypic expression is highly variable.[6-10] It is responsible for about 4.8% of all the cases of craniosynostosis, and is the most common syndrome of a group of more than 100 types of craniosynostosis.[9] The mutation in the genes that codify receptor 2 of the (FGFR2) fibroblast growth factor, is responsible for the deformities observed. [8,9] It is also postulated that a cranium basis malformation causes the premature fusion of the cranial sutures evolving with midfacial hypoplasia and cranium shape changing according to the sutures involved. [2,7]The suture fusion order and range determine the degree of deformity and inability.[9] The triad composed by cranium deformities, facial anomalies and exophthalmia, described by Crouzon in 1912, forms today the Crouzon's syndrome.[8-10] In this disease, the premature closure of cranial sutures and midfacial sutures and the cranium basis premature synostosis, give it a brachiocephalic configuration [7,9]. Since the suture becomes cast, the growth perpendicular to it becomes restrict and the cast bones act like a sole osseous structure [9]. Compensatory growth occurs in the remaining open sutures to allow continuity to the brain development causing abnormal osseous growth and production of facial deformities [9]. Such syndrome is progressive, from the beginning in the first year of life appearing frequently only at two

years of age [8,9]. There are also congenial premature forms in which the synostosis begins inside the uterus and is manifested at the birth with facial deformities like upper maxillary hyperplasia, responsible for respiratory difficulties and exophthalmia [8,9.]

Otorhinolaryngologic Manifestations

In the affected individuals there are almost always a high and large forehead, with convexity in the region of the anterior frontanelle, flattening of the occipital region and a certain front occipital protuberance. Hypoplastic maxilla, midfacial and maxillary hypoplasia are responsible for a number of alterations in the face aspect [9]. The upper lip is short and the inferior lip, along with the tongue, are prominent; the maxilla is hypoplastic and there is relative maxillary prognathism and micrognatia [2,9]. The conductive hearing loss is common. The nose shows an aduncous aspect, due to the strong maxillary hypoplasia, recalling a "parrot beak" due to the frontal shortening of the dorsum of nose. [7-9] The obstruction of the upper respiratory passages develops, following the septal diversion, abnormalities to the centre of the nose and epipharynx narrowing.[9] It can lead to acute respiratory anxiety [9], dyspnea of the type polypnea and even sleep apnea, mainly when connected to upper maxillary hypoplasia. [8]

Other clinical manifestations

There are several ocular abnormalities and the most common already reported for such disease are: shallow orbits, bilateral ocular proptosis, hypertelorism, divergent strabismus, optical atrophy, conjunctivitis or exposure keratoconjunctivitis and a non-explained loss of visual accuracy.[7-9] There rarely may occur nystagmus, coloboma of the iris, anisocoria, microcornea or megalocornea, cataract, blue sclera, glaucoma and globe luxation.[9] Despite the exophthalmia is constantly verified in the patients affected by the

Crouzon's disease, the ocular proptosis is not clearly present at birth and develops gradually in the first years of life. Blindness following the optical atrophy by the intracranial hypertension may also occur. Acanthosis nigricans, a disorder that causes brown to black velvet stains, generally on the neck, under the arm or in the groin region is the main Crouzon's syndrome dermatologic manifestation, and it is detectable after the childhood.[9] Generally, the psychomotor development is normal and the mental ability of these patients is usually within the normality. [8,9] However, some reports of mental retardation have been related to the increased intracranial pressure, which develops due to the brain growth restriction by the several synostoses. [9]

Diagnostic tests- imaging studies

Skull radiographs are used to show synostosis, craniofacial deformities, digital markings of skull, widening of hypopyseal fossa, small paranasal sinuses, and maxillary hypoplasia with shallow orbits. The coronal, sagittal, lambdoid, and metopic sutures may be involved. Cervical region radiologic abnormalities include butterfly-shaped vertebra and fusion of the posterior bodies and elements, present in about 18% of the patients. C2-C3 and C5-C6 are equally affected.[4,5] Comparative CT scan 3D reconstruction analysis of the cranium is used to precisely define the pathologic anatomy and to permit specific operative planning. Magnetic resonance imaging (MRI) is used to show occasional corpus collosum agenesis and optic atrophy.

Treatment:[8-10]

Treatment by a multidisciplinary team working together with the family provides the best results with any craniofacial disorder. The goal is to stage reconstruction to coincide with facial growth patterns, visceral function and psychological development. The treatment of patients presenting complex facial deformities is one of

the most challenging multidisciplinary tasks. Due to advances in medical technology and surgical techniques in the last 20 years, correction of severe malformations has become possible and is performed by highly specialized teams, frequently in a single operation. Craniotomy is often performed during the first year of life to treat the craniosynostosis.Frontofacial advancement and midface-advancement can be performed later to correct the proptosis and mid-face hypoplasia. Coordinated orthodontic therapy is often necessary to bring unerupted teeth into place and improve occlusion. Surgery can also be used to separate the fused fingers.

Conclusion

Treatment of craniofacial syndromes consists of a team approach in which each organ system is addressed independently and problems are prioritized on the basis of relative urgency.An understanding of these abnormalities is necessary for the dental team to make the appropriate referrals to insure the patient receives the best available care. Prognosis depends on severity of disease.

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