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First Arch Syndrome: A Case Report on Crouzon Syndrome

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ABSTRACT

Wide spectrum of anomalies in the first branchial arch gives rise to a variety of congenital syndromes mainly affecting the lower jaw, ear or mouth during early embryonic development. Crouzon syndrome, one of many craniosynostosis syndrome, is an autosomal dominant disorder caused by mutation in Fibroblast Growth Factor Receptor (FGFR)-2 and -3 gene. Here, we report a case of 7 years old male patient who presented to us with features of brachycephaly, exophthalmos, flat broad nasal bridge and low set ears.

Keywords: First arch syndrome, Craniofacial dysostosis, exophthalmos, copper beaten appearance

INTRODUCTION

Development of the craniofacial structures is unique and complex process which proceeds due to a rapid and orderly composition of mesodermal and cranial neural crest cells via a complex signaling network.[1]Syndromes of the first and second branchial arches manifest as combined tissue hypoplasia and aplasia of the derivatives of the first and second arches which include: face, external ear, middle ear and maxillary and mandibular arches during early embryonic development. They are the second most common craniofacial malformation after cleft lip and palate. Derivatives of first arch include mainly the lower jaw – mandible, two bones in the middle ear malleus and incus, muscles for chewing that is muscles of mastication and mandibular nerve.[1,2] On the basis of clinical features, anatomy and embryology, it is clear that all the anomalies arising from abnormal development of the first arch includes:(a) Treacher Collins syndrome or mandibulofacial dysostosis; (b) Pierre Robin syndrome (c) mandibular dysostosis; (d) deformities of the external and middle ear; (e) congenital deaf-mutism; (f) cleft lip and cleft palate; (g) hypertelorism; (h) a recently described syndrome exhibiting congenital deafness and hypertelorism.[3]

Crouzon syndrome, also known as craniofacial dysostosis, is an autosomal dominant disorder that affects the first branchial arch, which serves as the precursor for both Maxilla and Mandible. First described in 1912 by French neurologist Octave Crouzon, a triad of skull deformities, facial anomalies, and proptosis, Crouzon syndrome is caused by a mutation in the fibroblast growth factor receptor (FGFR)-2 and -3 gene on chromosome 10. The incidence is 1:60,000 live births with worldwide prevalence rate of approximately 1 per 25,000 live births with no known race or sex predilection.[4,5]It represents the most common syndromic craniosynostosis and is caused by premature obliteration and ossification of two or more sutures, most often coronal and sagittal resulting in brachycephaly, midface hypoplasia and wider anterior skull base.[6]

Other anomalies include shallow orbit, maxillary hypoplasia, and occasional upper airway obstruction.[7] Besides these features, it is characterized by hypertelorism, exophthalmos, strabismus, beaked nose, short upper lip and relative mandibular prognathism with no digital abnormalities.[8,9,10] Other intraoral manifestations include reverse overjet, V-shaped maxillary dental arch, narrow, high, or cleft palate and bifid uvula with occasional oligodontia, macrodontia, peg-shaped, and widely spaced teeth.[11]

CASE REPORT

A 7 year old male, accompanied by his father reported to the Department of Oral Medicine And Radiology, K.D. Dental College And Hospital, Mathura with a complaint of carious right Mandibular posterior teeth associated with dull aching pain which usually aggravated on mastication and relieved on its own. Patient had a history of cleft lip and palate wrt Right side for which he had undergone surgery for cleft lip at the age of 1 year. Because of low Hemoglobin level, surgery for cleft palate was delayed. He had problem of speech because of which he couldn't even utter words and eat and drink properly. On 2019, at the age of 5 years, he underwent surgery for cleft palate. After that he started speaking and eating food normally. However, his speech was still not clear.

Genealogical examination revealed that the patient was the single child born to a non consanguineous healthy parents. Patient weighed 13 kg and his height was 3 feet 5 inches with normal extremities and normal IQ level. Patient was ectomorphic and poorly nourished. On extraoral examination, patient presented with brachycephalic head, convex facial profile, flat forehead with depressed supraorbital ridges. Besides these features, eexophthalmos, broad and flat nasal bridge and low set ears was also evident. Middle third of face showed decreased prominence due to hypoplasia of malar region.

Intraoral finding revealed narrow and V shaped palate with surgical scar in the mid palatine area. Mandibular arch was observed to be narrow. Patient was in early mixed dentition with anterior crowding. Dentinal caries was noticed wrt 75, grossly decayed wrt 85 and root stumps were observed wrt 74 and 84. Disto - labial rotation irt 11, Mesio - labial rotation irt 21 was also observed. Provisional diagnosis of caries wrt 85, 75 along with Crouzon syndrome was made. Apert syndrome, Carpenter syndrome, Pfeiffer syndrome, Jackson Weiss syndrome were considered for differential diagnosis.

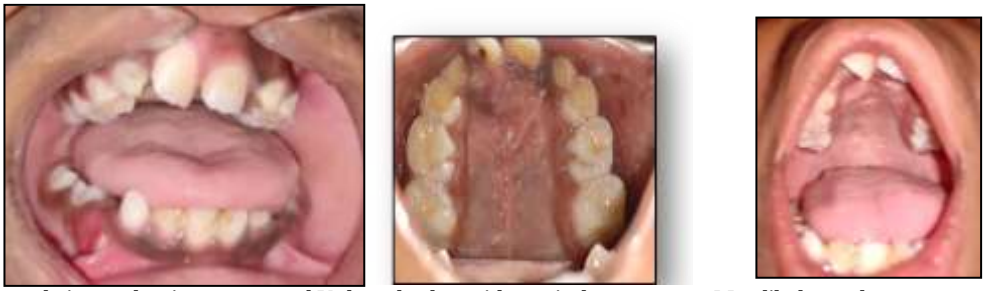
The patient was subjected to radiographic investigations. Panoramic radiograph showed caries wrt 75, 85, root stumps wrt 74,84. All permanent tooth buds were in erupting stage. Lateral cephalogram showed shallow orbits, sutural diastasis of bilateral coronal sutures, depressed nasal bridge, copper beaten skull, asymmetrical calvarial thickening and deviated nasal septum and obliterated sutures. PNS view showed small paranasal sinuses, prominent cranial markings of the inner surface of cranial vault seen as multiple radiolucencies appearing as depressions and deviated nasal septum. On the basis of history and records, clinical features and radiographic examination, final diagnosis of Crouzon Syndrome was established.

Patient was advised for oral prophylaxis at regular intervals and pulpectomy irt 85. Extraction was advised for 74 and 84 along with restoration wrt 75. Patient was also advised for speech therapy. To rule out any other deformities, the patient was advised for a complete long bone radiographic survey and PA view of the chest along with a paediatric general examination. Radiographic examination of metacarpal bones and fingers were unremarkable. On anteroposterior spine radiograph, no remarkable abnormality was detected. No abnormality was reported in long bones of bilateral lower limbs. On, paediatric evaluation, vitals were within normal parameters. Bilateral air entry was present

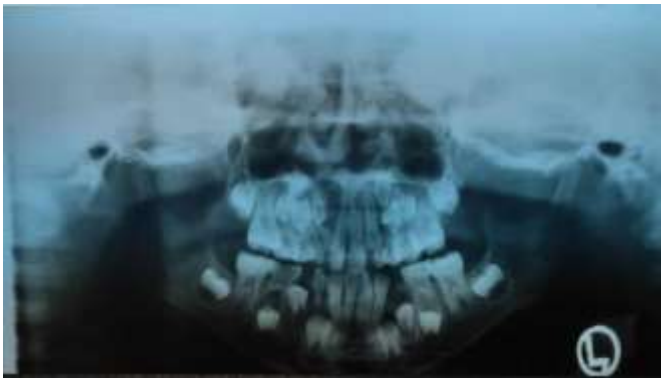
on chest examination. On CVS examination, S1 and S2 sounds were heard with no heart murmur. On palpation of the liver, it was soft approximately 2 cm and no abnormalities were felt in the spleen.

Frontal and lateral profile of the patient showing brachecephalic head, exopthalmos, low set ears, broad and flat nasal bridge, hypoplasia of malar region

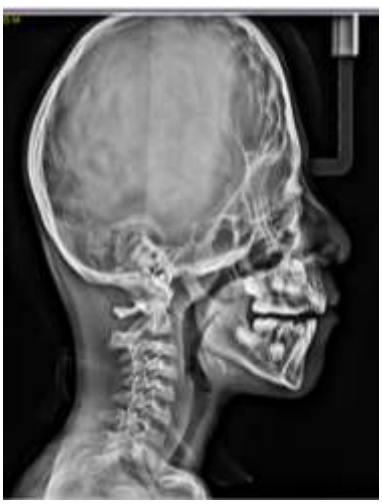
Extraoral pictures showing normal extremities

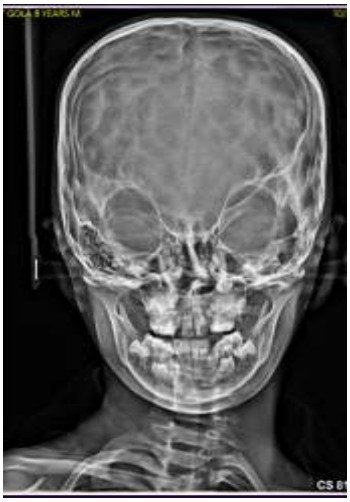


Intraoral picture showing narrow and V shaped palate with surgical scar, narrow Mandibular arch, root stump wrt 74,84, caries wrt 75,grossly decayed wrt 85

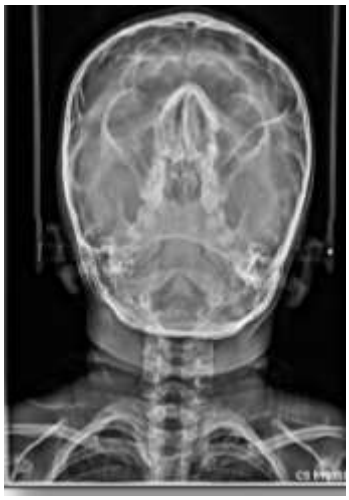


Panoramic radiograph showing mixed dentition stage, carious wrt 75,85, root stump wrt 74,84

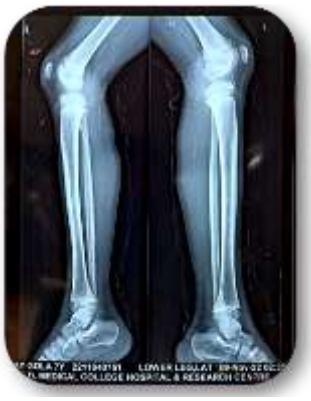


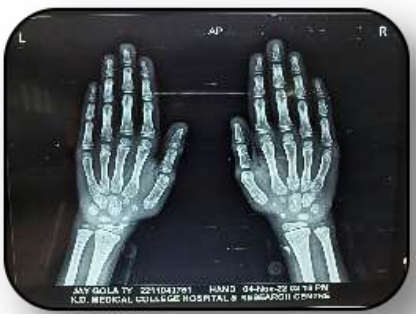


Lateral cephalogram and PA view showing shallow orbit, diastasis of bilateral coronal suture, copper beaten appearance, asymmetrical calvarial the inner surface of cranial vault



PNS view showing obliterated sutures, small paranasal sinuses, prominent cranial markings of thickening





Lower limb radiograph, Hand wrist radiograph, , Chest PA view

DISCUSSION

Crouzon syndrome is an autosomal-dominant disorder with complete penetrance and variable expressivity. Crouzon syndrome: triad of skull deformities, facial anomalies, and exophthalmos, similar characteristic features of varying degrees were evident in our patient. All the differential diagnosis considered in this case involved craniofacial abnormalities in which signs and symptoms can have significant overlap. An important detail to be considered is the normal hands and feet found in a Crouzon patient in contrast to those with Apert syndrome, a similar but more severe craniosynostosis syndrome, where syndactyly of the extremities is present. Pfeiffer syndrome, another craniosynostosis, is also characterized by short, broad big toes and thumbs.[12,13] In Crouzon syndrome brachycephaly (widened and shortened) is the most common presentation due to bi-coronal suture fusion. A copper beaten appearance of the skull is also a common finding in this syndrome due to multiple radiolucencies of the skull bones which was evident in our patient.

If the history, physical, and imaging are still inconclusive, molecular testing can be pursued. For those patients with positive family history of Crouzon or one of the other craniosynostosis syndromes, prenatal genetic testing and 2-dimensional and 3-dimensional ultrasounds can be done to confirm the diagnosis before the birth. Amniocentesis and/or chorionic villus sampling can be performed in high-risk pregnancies.[14]

Surgical management remains the mainstay of treatment for Crouzon syndrome depending upon the variable expressivity of the diseases and the age of the patient which

includes cranial vault remodeling and amendment of facial anomalies. Remodeling of the skull vault during infancy, surgery for facial and orbital correction at an age of 5–7 years, and advancement surgery for maxilla/ mandible during teenage life comes under staged surgical approach. Correction of midface anomalies include Le Fort III osteotomy, Monobloc procedure, bipartition osteotomy and combination surgeries. A new technique, craniofacial disjunction, followed by gradual bone distraction (Ilizarov procedure) can be carried out to correct exophthalmos and improve the functional and esthetic aspects of the middle third of the face without requirement for bone graft in patients aged 6-11 years.[5,6]

CONCLUSION

Since Crouzon syndrome has variable manifestations, combination of detailed family history, findings of specific physical examination, use of various imaging modalities and genetic testing can be helpful for early detection. Proper treatment plan by multidisciplinary approach utilizing modern methods will achieve good outcome and improve quality of patient's life.[5,6]

Declaration by Authors

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