Case Report

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Cleidocranial Dysplasia: A Rare Case Report

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ABSTRACT

Background: Cleidocranial dysplasia is a rare familial disorder characterized by the presence of numerous supernumerary and unerupted teeth, partial or complete absence of clavicles and characteristic craniofacial deformities. Here, the article reviews the clinical and radiographic findings of cleidocranial dysplasia in a 16-year-old adolescent boy who presented with delayed teeth eruption. This patient was first noticed by the dentist because of the aesthetic problems i.e. delayed eruption of teeth. Here, we report a typical case of CCD in a 16-year-old male who had classical diagnostic feature of this syndrome.

Keywords: Cleidocranial dysplasia, supernumerary and unerupted teeth.

INTRODUCTION

Cleidocranial dysplasia (Ccd) is a rare congenital disorder and was referred to as cliedocranial dysostosis previously. The syndrome was first described by Marie and Sainton in 1898 as a rare development condition in which the leading features were aplasia of clavicles, exaggerated development of the transverse diameter of cranium, delayed closure of fontanelles and disorders of jaws and dentition. It affects men and women equally. The prevalence is less than 1 per million. The defect often appears in several successive generations. It is characterized by abnormalities of skull, dentition, jaws and clavicles. It is seen that fontanels are open despite the age of the patient far beyond the normal closure time. The profile of the patient is concave due to underdeveloped maxilla.The bridge of the nose is broad and depressed. The mandible appears to be prognathic with a depressed forehead. The most striking oral abnormalities are the failure of eruption of the permanent teeth with multiple unerupted supernumerary teeth.[1-8] Ccd shows an autosomal dominant inheritance. It is associated with a spontaneous mutation in the gene coding for osteoblast transcription factor Runx 2, which is essential for osteoblasts and dental cell differentiation as well as for bone and tooth formation. Runx 2 also known as OSH2, Cbfa1, PEBP2Aa, AML-3 belongs to runt domain gene family. [9]

CASE REPORT

A 16-year-old male patient was reported to the Department of Oral Medicine and Radiology, Narsinhbhai patel Dental college and hospital, Sankalchand Patel University, Visnagar with the

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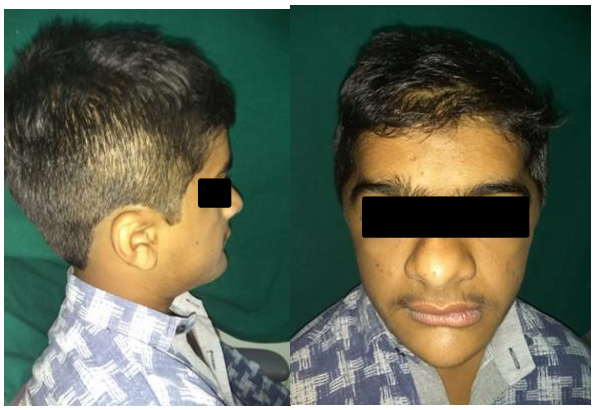

Fig 1: Clinical photograph showing hypertelorism, frontal bossing, deviated nasal septum, progrnatic mandible and midfacial hypoplasia.

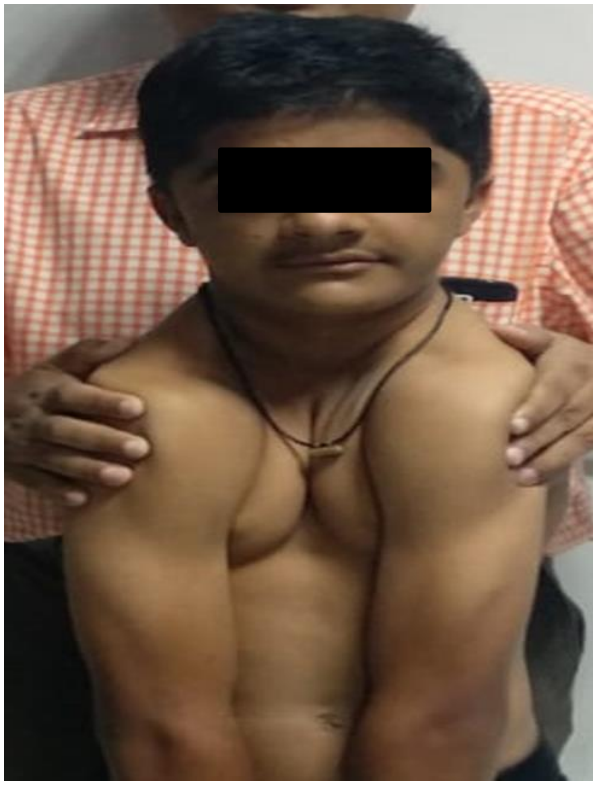

Fig 2: patient with hypermobility of the shoulder girdles and frontal bossing.

chief complain of missing teeth in upper and lower jaws. He gave history that he did not have any teeth in these regions of the jaws since childhood after his milk teeth shed away. He gave the history of falling of these teeth few years back. Upon general examination, he was of average height and normal gait.

General physical examination demonstrated a medium build, short stature, sloping and shrugged shoulders which were easily apposable. Extraoral examination revealed brachycephaly and an ocular hypertelorism. Parietal and occipital bossing was present, giving the skull a large globular shape. Depressed nasal bridge and mid-facial hypoplasia was evident. Mandibular prognathism, prominent chin, straight facial profile with competent lips were noted. (Figure 1 &2)

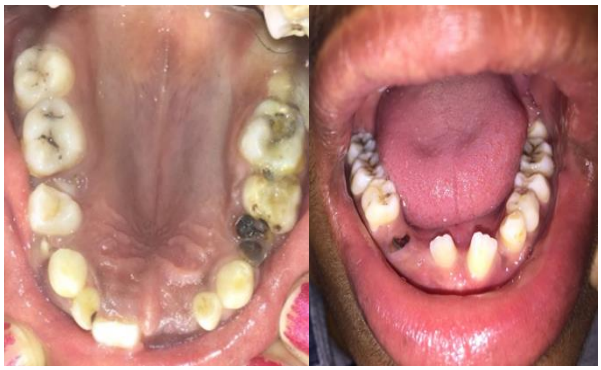

Fig 3: Intraoral showing numerous over-retained deciduous and some missing teeth.

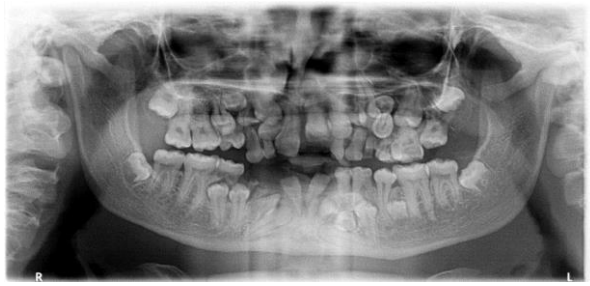

Fig 4: Panoramic view of the jaws showing multiple unerupted supernumerary teeth mimicking premolar. rounding of gonial angle.

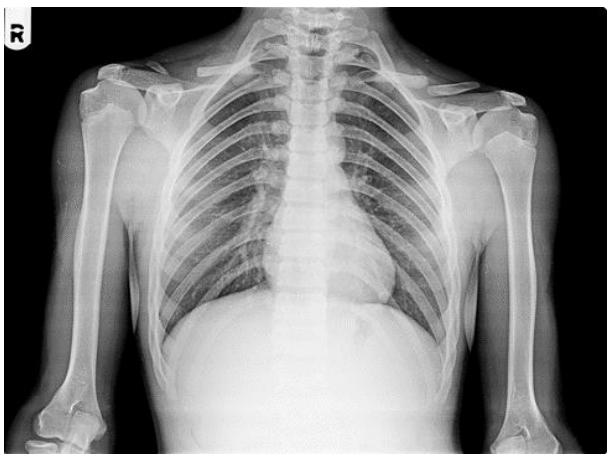

Fig 5: Chest radiograph (PA View) of the patient showing thinning and hypoplasia of the clavicles and bell shaped rib-cage.

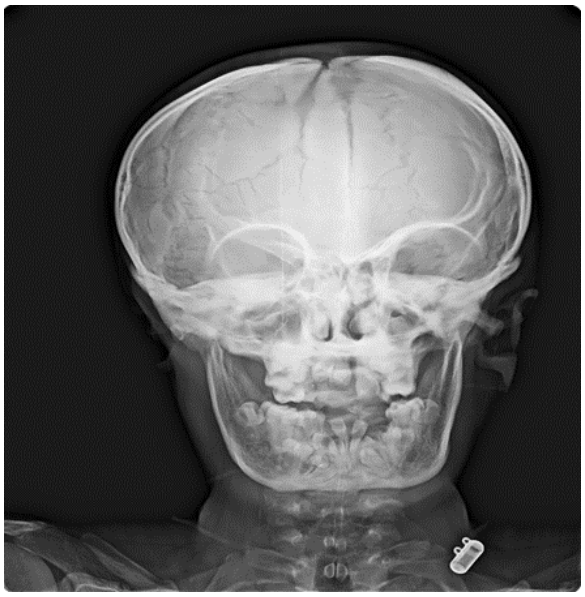

Fig 6: Brachycephaly of the skull with Open sagittal suture.

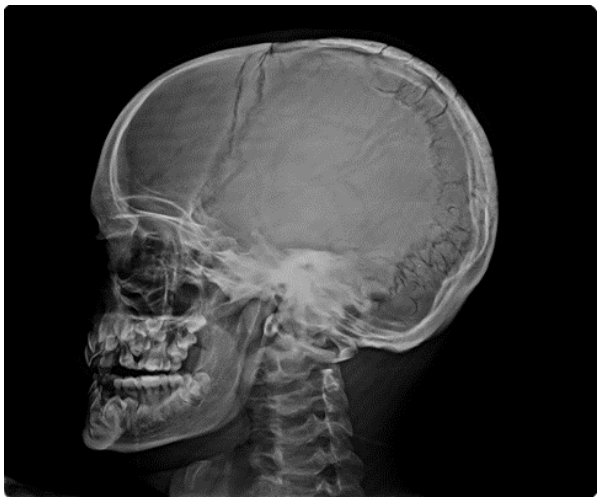

Fig 7: Gonial angle is replaced by a rounded contour of the mandible, Wormion bone evident at the anterior fontanelle region and in the lambdoid suture region.

On clinical Examination of the oral cavity revealed multiple over-retained deciduous teeth and missing teeth, particularly in anterior maxilla and mandible (Figure 3). For further investigation we decided to take special radiographs included, Orthopantomogram, lateral and postero-anterior view of the skull. We had also advised Chest, spine radiographs.

Orthopantomogram shows Multiple impacted permanent and supernumerary teeth in the incisor and bicuspid regions of the maxilla and mandible and over retained deciduous teeth. Follicular spaces of some impacted teeth were enlarged in the mandibular left parasymphysis region, suggesting cystic transformation. Rounded contour of the angle of the mandible was evident. Thin zygomatic arch was also seen.(figure 4) Chest radiograph shows bilateral hypoplastic clavicles, cone-shaped thorax, low placed scapulae, and spine appeared to be normal. (figure 5)Postero-anterior view (figure 6) shows brachycephaly of the skull with light bulb shaped appearance. Craniofacial disproportion evident, patent sutures with wide separation of the sagittal suture which appears broad with irregular margins, flattening of the skull on the superior surface, inner and outer tables of the skull cannot be well differentiated. Agenesis of frontal sinus and hypoplastic mastoid process seen bilaterally. Nasal septum appears deviated to the right side. Hypertrophy of the left inferior turbinate noted along with deficient zygomatic bone and hypoplastic maxillary sinus. Lateral skull view (figure 7) showed gonial angle is replaced by a rounded contour of the mandible referred to a banana shape. Widening of the coronal suture evident with patent anterior fontanelle. Wormion bone evident at the anterior fontanelle region and in the lambdoid suture region which are formed by secondary ossification centres. Sphenoid sinus shows complete opacification and obliteration of the mastoid air cells was noted. Maxillary retrognathism with shortened nasal bone was evident. Pure tone audiometry test revealed mild conductive hearing loss on right ear and left ear showed moderate conductive hearing loss.

After completing all the necessary investigations, the patient was confirmed as having cleidocranial dysplasia. He is currently being treated by a team of pedodontists, orthodontists, oral surgeons and prosthodontists.

DISCUSSION

CCD involves mutation in the transcription factor, Runx2 (essential for osteoblast and dental cell proliferation as well as for bone and tooth formation)/Cbfa1 (Core Binding Factor A1), located on chromosome 6p21. It primarily affects the bones undergoing intramembraneous ossification, especially skull, clavicles, and flat bones hence termed cleidocranial.

Clavicle is the first bone to ossify (fifth to sixth week of fetal life) and exhibits many deformities ranging from various degrees of hypoplasia and hypocalcification to complete the absence of clavicles. When the clavicles are completely absent, which occurs in 10% of the cases, the neck appears long, and the shoulders are drooping and hypermobile. We reported hypermobile shoulders in our patient.Because of delayed mineralization, there may be abnormal dentition with late eruption and impaction of the deciduous and permanent teeth. The resorption of their root is delayed. Many of the deciduous teeth are retained throughout life and lie among the permanent teeth. The permanent teeth generally lose their eruption stimulus and stay embedded, while the deciduous teeth are retained. Extraction of deciduous teeth does not stimulate the eruption of the permanent teeth. Suggested factors of over retained deciduous teeth are lack of eruption potential and lack of cellular cementum on roots of permanent teeth, delayed mineralization of teeth, physical barrier - abnormal density of bone overlying the succedaneous teeth, and failure of bony crypt to resorb. The presence of multiple supernumerary teeth mainly in the mandibular premolar and anterior maxilla is common which appear similar to premolars. Suggested etiology for supernumerary teeth is incomplete or severely delayed resorption of the dental lamina, which is then reactivated at the time of crown completion in the normal permanent teeth. In present case, we found multiple supernumerary teeth in maxilla and mandible.The current treatment involves combination of orthodontics and maxillofacial surgery with timely extraction of deciduous teeth, surgical removal of supernumerary teeth, exposure of selected unerupted permanent teeth and orthodontic forced eruption, and fabrication of partial denture. Patient is under above mentioned treatment .[10-16]

CONCLUSION

The clinical findings of cleidocranial dysplasia, although present at birth, are often either missed or diagnosed at a much later time. Some cases are diagnosed through incidental findings by oral physicians, treating patients for unrelated conditions. In conclusion, dentists should be aware of these findings in order to diagnose CCD at an early stage so as to provide early treatment for an aesthetically satisfying facial appearance along with psychological benefit and better life style to the patients.

CONFLICTS OF INTEREST

The authors declare they have no potential conflict of interests regarding this article.

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