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Cleidocranial Dysostosis - A Case Report

Case Report

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Abstract

Cleidocranial dysplasia (CCD) is a rare autosomal dominant disorder that exhibits several skeletaldefects and dental abnormalities.The characteristic features seen in cleidocranial dysplasia are aplastic or hypoplastic clavicles,delayedclosureor open fontanelles, open skull sutures and dental abnormalities, which includes over retention of primary dentition and delayed eruption of permanent dentition, with presence of multiple impacted supernumerary teeth. In this article we report 2 cases of cleidocranial dysplasia reported in our dental outpatient department with an aim to highlight the clinical features,dental abnormalities and radiological features.

Keywords: Cleidocranial Dysostosis; Cleidocranial Dysplasia; Impacted Permanent Teeth; Autosomal Dominant.

Introduction

Cleidocranial dysplasia is a rare congenital defect affecting the bones which undergo intramembranous ossification primarily. The bones usually involved are skull,clavicles and jaws [1, 2].Cleidocranial dysplasia is rare in occurrence with an incidence of 1:10,00,000 individual and has an autosomal dominant inheritance pattern [3-5]. It was first described by Pierre Marie and Paul Sainton in the year 1898,subsequently more than 1000 cases have been documented in the medical literature [6, 7]. Cleidocranial dysplasia is also known as mutational dysostosis,cleidocranial dysostosis and Marie and Sainton disease [8].

Individuals affected with CCD exhibit multiple skeletal defects,with striking characteristic features of partial or complete absence of clavicles, delayed closure of the fontanelles,presence of open skull sutures,wide pubic symphysis and multiple wormian bones [9]. Typically, the clavicles are underdeveloped in varying degrees resulting in excessive mobility allowing them to approximate the shoulders anteriorly [10]. Delayed closure of fontanelles and presence of metopic sutures result in frontal bossing.

Patients with CCD usually exhibit short stature, hypertelorism, depressed nasal bridge, maxillary hypoplasia and multiple dental abnormalities.The thoracic cage is small and bell shaped. The clinical and dental features of CCD are unique and may lead to initial diagnosis in most cases [11-13].

The classic dental features seen in CCD includes retention of the primary dentition, delayed eruption with consequent impaction of the permanent teeth and presence of multiple impacted supernumerary teeth, crown and root abnormalities [14-20].

Successful dental treatment of CCD requires a compliant patient and an interdisciplinary team approach involving orthodontics,prosthodontic and maxillofacial surgeon.Since early diagnosis of CCD is essential for initiating the appropriate treatment approach,the clinicians should be aware of the characteristic features.Hence the aim of the article is to describe the clinical features,dental abnormalities and radiological features of two cases of cleidocranial dysplasia reported in our dental outpatient department.

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Case Description

Case 1

A 25-year-old male reported to the department of Oral Medicine and Radiology with a complaint of multiple missing teeth in the upper front teeth region for the past 8 years and wanted replacement. His past dental history revealed that he had undergone restoration in a private dental clinic 5 years before and no dental extractions done previously. His past medical history and family history were non-contributory.

On general examination the patient was apparently well with moderately built and short in stature.On physical examination the patient was able to approximate the shoulders anteriorly demonstrating the hypermobility of the joint (Figure Ia). On extraoral examination the patient had frontal bossing,hypertelorism,depressed nasal bridge and hypoplastic maxilla leading to relative prognathic mandible (Figure Ib & Ic). Intraoral examination revealed narrow high arched palate,multiple over-retained deciduous teeth with multiple unerupted permanent teeth(Figure IIa &IIb). Based on the clinical and intra-oral features a provisional diagnosis of cleidocranial dysplasia was given.

The patient was advised to undergo panoramic imaging,posterio-anterior skull view and posterio-anterior chest view for further evaluation. Panoramic imaging (Figure IIIa) revealed the presence of a total 56 teeth including multiple impacted permanent and supernumerary teeth distributed along the entire region of maxillary and mandibular alveolar bone.Posterio-anterior skull view (Figure IIIb) revealed brachycephaly withpresence of wormian bones,open fontanelles,thickened calvarium and poorly developed sinuses.The posterio-anterior view of the chest (Figure IIIc) revealed bell shaped rib cage and complete absence of the clavicle on both the sides.

Based on the clinical,extraoral,intraoral and radiographic features a final diagnosis of cleidocranial dysplasia was given.The patient is currently being treated by a team comprising of oral and maxillofacial surgeon,orthodontist and prosthodontist, planned for orthognathic surgery to correct the malocclusion and surgical removal of impacted teeth to be followed by replacement of missing teeth.

Case 2

A 48-year-old female reported to the department of Oral Medicine and Radiology with a complaint of multiple missing teeth in the upper and lower jaws and wanted replacement. Her past dental history revealed she had undergone extractions of retained deciduous teeth 25 years before.Her past medical history and family history were non-contributory.

On general examination the patient was well-oriented, moderately built and short in stature.On physical examination when she was asked to bring her shoulders forward, she was able to approximate both the shoulders anteriorly demonstrating the hypermobility of the joint (Figure IVa). On extraoral examination she had frontal bossing,hypertelorism with depressed nasal bridge (Figure IVb). Intraoral examination revealed multiple missing teeth,multiple decayed over-retained deciduous teeth with multiple unerupted permanent teeth (Figure Va&Vb). Based on the clinical and intra-oral features a provisional diagnosis of cleidocranial dysplasia was given.

Radiographic investigations including panoramic imaging,posterio-anterior skull view and posterio-anterior chest view was advised for further evaluation. Panoramic image (Figure VIa) revealed the presence of a total 30 teeth including multiple missing, decayed over-retained deciduous teeth,multiple impacted permanent and supernumerary teeth distributed along the entire region of maxillary and mandibular alveolar bone.Posterio-anterior skull view (Figure VIb) revealed brachycephaly,presence of wormian bones,open fontanelles,thickened calvarium and poorly developed sinuses.The posterio-anterior view of the chest (Figure VIc) revealed bell shaped rib cage and complete absence of the clavicle on both the sides.

Based on the clinical,extraoral,intraoral and radiographic features a final diagnosis of cleidocranial dysplasia was given.The patient is currently being treated by a team comprising of oral and maxillofacial surgeon and prosthodontist, planned for surgical removal of impacted teeth and replacement of missing teeth.

Discussion

Cleidocranial dysostosis(CCD) is a rare genetic syndrome with autosomal dominant inheritance.CCD is usually caused by haploinsufficiency in the RUNX2 (runt related transcription factor 2) gene located on the short arm of chromosome 6 which has an important role in osteoblasts differentiation and maturation of chondrocytes [21-23]. This syndrome can be identified during prenatal ultrasonography but sometimes it is detected only after birth due to cranial deficiencies or presence of any not related medical pathologies or cranial deficiencies [24].

For a definitive diagnosis of CCD the pathognomonic triad of multiple impacted supernumerary teeth,partial or complete ab-

Figure 1. Ia showing approximation of shoulders, IIb & IIc showing extra-oral features of frontal bossing, hypertelorism, depressed nasal bridge.

Figure 2. IIa showing maxillary arch showing missing permanent teeth in the anterior region and palate appearing narrow and constricted and figure IIb showing mandibular arch.

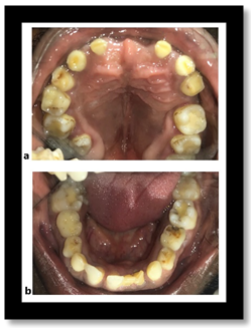


Figure 3. IIIa shows panoramic image showing a total of 63 teeth with multiple retained deciduous teeth, multiple impacted and supernumerary teeth in the maxillary sinus region, cuspid, bicuspid and tricuspid region. Figure IIIb showing PA view of the skull showing brachycephaly, open fontanelles, multiple wormian bones and poorly developed sinuses. Figure IIIc PA view of the chest showing absence of clavicles bilaterally (white circles).

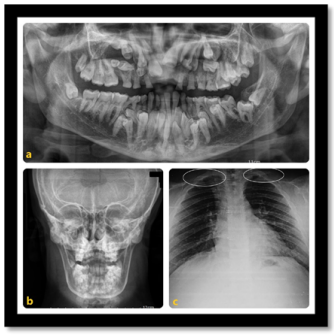
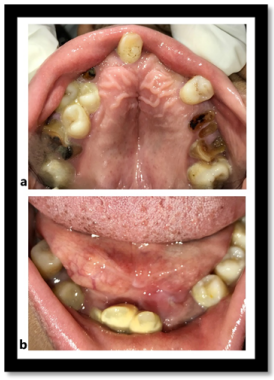


Figure 4. IVa showing approximation of shoulders, IVb showing extra-oral features of frontal bossing, hypertelorism, depressed nasal bridge.

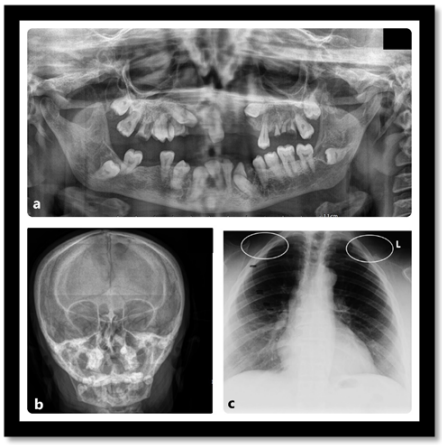
Figure 5. Va showing maxillary arch showing multiple missing and decayed teeth. Palate appearing narrow and constricted figure Vb showing mandibular arch.



sence of clavicles and presence of open fontanelles & cranial sutures must be present [25]. The genetic disturbances in the osteoblast differentiation results in abnormal dentition with over-retained primary dentition,multiple impacted supernumerary and permanent teeth.

The clavicle is the first bone to undergo ossification and exhibits many deformities ranging from various degrees of hypoplasia to complete absence of clavicles. When the clavicles are completely absent which occurs in 10 percent of cases with cleidocranial dysplasia it results in hypermobile and drooping shoulders.

Figure 6. VIa showing panoramic image showing a total of 30 teeth with multiple missing, over-retained decayed teeth, impacted permanent and supernumerary teeth. Figure VIIb showing PA view of the skull showing brachycephaly, open fontanelles, multiple wormian bones and poorly developed sinuses. Figure VIIc showingPA view of the chest showing absence of clavicles bilaterally (white colored cirlces).



Pycnodysostosis also known as Marteau lamy syndrome exhibits clinical features similar to cleidocranial dysplasia, but it can be differentiated by presence of dwarfism. The affected patients have dense and fragile bones.Mandibulo-acral dysplasia (MAD) is a rare genetic disorder, characterized by short stature, delayed closure of cranial sutures, mandibular hypoplasia and dysplastic clavicles.The scalp hair becomes sparse by the third decade and some patients may develop alopecia.Micrognathia results from osteolysis of the body and ramus of the mandible.Crowding of teeth can be seen and early loss of tooth due to hypoplastic roots may occur.

Dental management of patients with CCD can be challenging. The fundamental treatment goal is to establish an aesthetic appearance and functional occlusion.The treatment plan depends on the chronological and dental age of the patient.The timing of the diagnosis is not only important in deciding an appropriate treatment plan but also in obtaining a successful result.The principles of treatment are focused on surgical intervention,orthodontic correction of malocclusion and prosthodontic rehabilitation [26, 27]. Dental management requires an interdisciplinary approach involving orthodontics, maxillofacial surgeon and prosthodontists.Every child born to an individual with CCD has a chance of inheriting the mutation.Hence it would be appropriate to provide genetic counselling to young patients who are affected [28-30].

Conclusion

The present case report highlights the need for awareness among dentists about CCD syndrome. When CCD is diagnosed in the early stages of life, a permanent dentition with proper functional occlusion, as well as an aesthetically satisfying facial appearance,motivation and psychological supportfor the patients and their family members, may be achieved by interdisciplinary team approach.

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