

A 9-year-old Saudi Boy with Cleidocranial Dysplasia: A Case Report

Rabab Ali Jassim AL Buainain¹, Yasin Alavi Aruveetil², Faisal Alsineedi², Abdullah Faraj Alshammari³,
Hussain Gadelkarim Ahmed^{4,*}

¹Saudi Pediatric Dentistry Program, Dammam Medical Complex, Dammam, Kingdom of Saudi Arabia (KSA)

²Department of dentistry, King Fahd Military Medical Complex, Dhahran, KSA

³Department of Dentistry, University of Hail, KSA

⁴College of Medicine, University of Hail, KSA

*Corresponding author: hussaingad1972@yahoo.com

Abstract Cleidocranial dysplasia (CCD) is an uncommon disorder associated with a genetic disorder mainly causing dysplasia of bones and teeth with autosomal dominant inheritance pattern, which has an extremely varied presentation. The dental indicators are principally delayed exfoliation of primary teeth and delayed eruption of permanent teeth, and numerous impacted supernumeraries. This article represent a 9 years old Saudi boy case of CCD and illustrates the clinical and radiological features of this patients. In this case the early diagnosis of the condition was not associated with proper orientation of the dental treatment to offer better quality of life to patient.

Keywords: *Cleidocranial dysplasia, dental treatment, Saudi*

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1. Introduction

Cleidocranial dysplasia (CCD) is an uncommon but well-known genetic skeletal condition, characterized by dental anomalies and bone abnormalities [1]. The disease is triggered by mutation in the gene *RUNX2* (*CBFA1*), located on the short arm of chromosome 6 [2,3]. The causing gene, *RUNX2* codes for a core-binding transcription factor protein (*CBFA1*), which is intricate in the differentiation of osteoblasts and bone construction [4-6]. *RUNX2* has an essential role in the epithelial-mesenchymal communications that control progressive tooth morphogenesis and histodifferentiation of the epithelial enamel organ.

Individuals affected with CCD have a distinctive facial look with a bulky forehead, hypertelorism, and midfacial hypoplasia [4]. Overall health is commonly good and the intellect is unaffected. The adversative overall health effects of CCD are generally not very severe or debilitating and there is no accompanying deficiency in cognitive or intellectual functioning in such patients [7].

CDD is a skeletal dysplasia described by delayed closure of the cranial sutures, hypoplastic or aplastic clavicles, and multiple dental abnormalities. Manifestations may differ among persons in the same family. The most noticeable clinical results are abnormally large, wide-open fontanels at birth that may stay open throughout life; mid-face retrusion; abnormal dentition, comprising delayed eruption of secondary dentition, failure to shed the primary teeth, supernumerary teeth with dental crowding, and malocclusion; clavicular hypoplasia bring about thin,

leaning shoulders that can be opposed at the midline; and hand defects such as brachydactyly, tapering fingers, and short, broad thumbs. Persons with CCD are shorter than their natural sibs and are more to be expected to have other skeletal/orthopedic problems such as *pes planus*, *genu valgum*, and scoliosis. Other medical problems comprise repeated sinus infections and other upper-airway problems, repeated ear infections, high incidence of cesarean section, and mild degree of motor delay in children under age of five years [8].

Diagnosis of CCD is rely on clinical and radiographic results that contain imaging of the cranium, thorax, pelvis, and hands. *RUNX2* (*CBFA1*) is the only gene in which mutation is identified to be associated with CCD. Molecular genetic testing of *RUNX2* detects pathogenic variants in 60%-70% of patients with a clinical diagnosis of CCD.

This article reports the case of a 9 year old child diagnosed with CCD after birth. The aim is to provide the health team with update measures toward dental management of patient with CCD diagnosis and dental treatment.

2. Case Presentation

2.1. Chief Complaint and Medical History

A 9 years old Saudi male attends Pediatric Dental Clinics in Dammam Medical Complex with his mother. The main concerns were about delay in the eruption of permanent teeth and multiple carious teeth. The patient

diagnosed with Clidocranial dysplasia shortly after his birth.

2.2. Clinical Features

The extra-oral examination showed frontal bossing, hypertelorism, convex profile, competent lip, his shoulder could be brought closer together and short stature. His medical history revealed delayed closure of the anterior fontanelle, absence of calvarian bone and partial occipital, as shown in [Figure 1](#) & [Figure 2](#).

Intra-oral findings include normal gingival texture with melanin pigmentation, shallow alveolar bone and decreased vertical growth due to poor alveolar bone development, poor oral hygiene, multiple caries, hypoplastic teeth, retained primary teeth, delayed eruption of permanent teeth, mesial step molar relationship and CL I canine relationship, over jet and overbite are not applicable due to upper anterior remaining roots, as shown in [Figure 3](#).

2.3. Radiographic Features

A panoramic radiograph revealed unerupted permanent teeth and supernumerary teeth in maxilla and mandible jaws. Dental age is 5 years old much delayed to the chronological age (9.4 years old) as eruption but as root formation dental age is 7 years old, which is also delayed than chronological age. Bitewings radiographs and

periapical radiographs were decided to check proximal caries and any pathology related to roots and/or bifurcation area since the teeth are extensively decayed, as shown in [Figure 4](#).



Figure 1. A 9 years old boy with Clidocranial dysplasia



Figure 2. Showing extra oral examination: head (Frontal bossing), Face (Symmetrical), Eye(Hypertelorism).



Figure 3. Showing oral examination: Primary dentition, Plaque deposits, Multiple caries, Hypoplastic teeth, Supernummary teeth

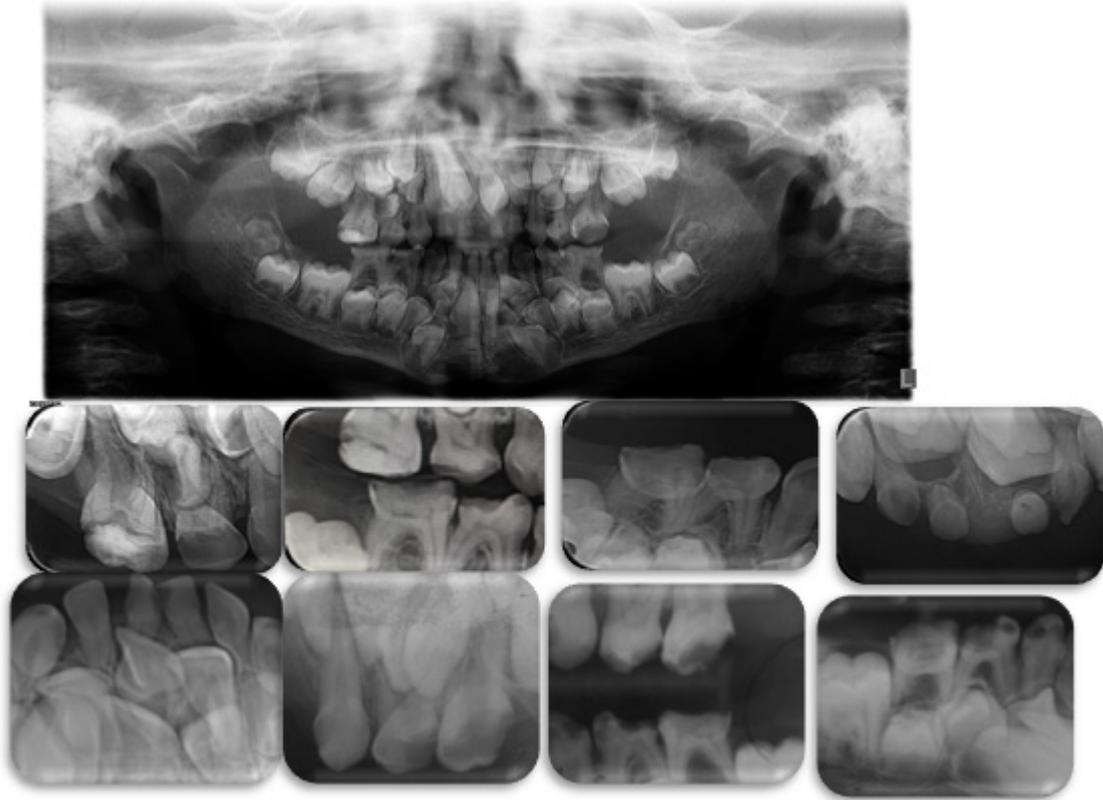


Figure 4. Panoramic Radiograph Evaluation

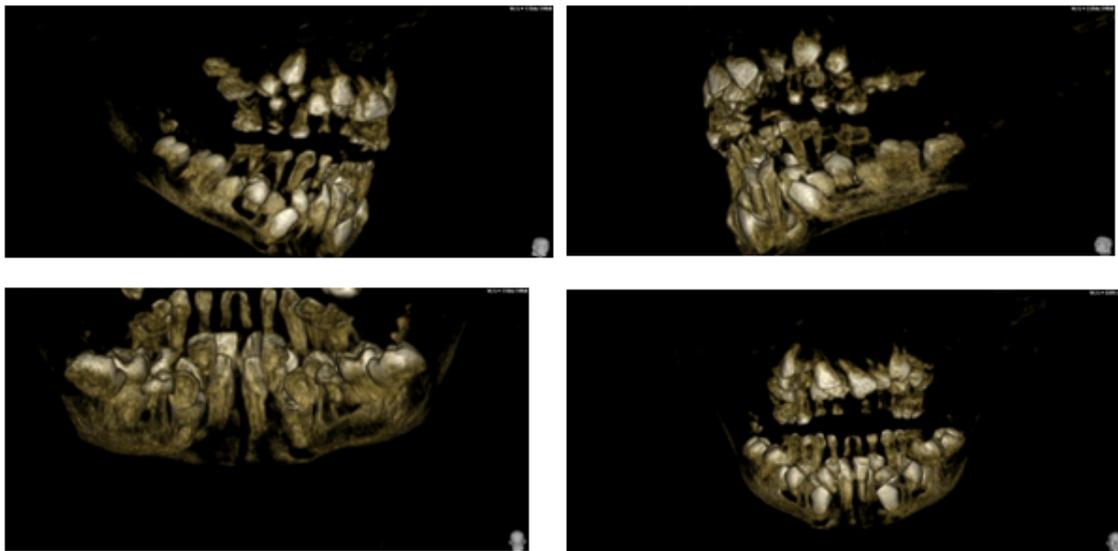


Figure 5. Panoramic Radiograph: Supernummary dental charting

Cone beam CT scan were decided to correctly account and localize supernumerary teeth, it revealed in: The upper right posterior teeth; one permanent 1st, 2nd molar and three premolars. The upper right anterior teeth; one permanent canine, lateral incisor and two permanent central incisors. The upper left posterior teeth; one permanent 1st, 2nd molar and three premolars. The upper left anterior teeth; one permanent canine, lateral incisor and two permanent central incisors. The lower right anterior teeth; two permanent canines, two permanent lateral incisors one of them is lacerated and two permanent central incisors. The lower right posterior teeth; one permanent 1st, 2nd molar and two 2nd premolars and

two 1st premolars. The lower left anterior teeth; two permanent canines, two permanent lateral incisors one of them is lacerated and two permanent central incisors. The lower left posterior teeth; one permanent 1st, 2nd molar and two 2nd premolars and two 1st premolars.

3. Discussion

The CCD is an uncommon disorder, with distinctive clinical features. Oral indexes display a hypoplastic maxilla with high-arched palate. Crowding of teeth is formed by custody of deciduous teeth, delayed eruption of

permanent teeth, and the presence of a large number of unerupted supernumerary teeth [9]. Therefore, It is essential to institute early diagnosis in such patients in order to offer an improved quality of life, and if required, suitable treatment.

However, in the present study, we reported dental inquires referred to a case of a 9 years old child diagnosed with CCD in eastern Saudi Arabia. During our comprehensive search in literature we didn't come across such case report from Saudi Arabia.

Intra-oral examination reveal several features, with the most being, hypoplastic teeth, retained primary teeth, delayed eruption of permanent teeth, mesial step molar relationship and CL I canine relationship, which were previously reported in several studies [10-12]. Moreover, multiple caries was observed in this case which might be attributed to the poor oral hygiene and diet habit of this patient.

In the present case, Cone-beam computed tomography (CBCT) [13] were decided to correctly account and localize supernumerary teeth, it revealed distinct features in this patient. CBCT done to check any developmental anomalies, orthodontic assessment, Supernumerary teeth exact numbers and positions, any pathological abnormalities. Use of CBCT in orthodontics is increasing; however, some patients started treatment with conventional images. The images can be useful for the assessment of mesiodistal root angulations if the volume is properly manipulated to create a pan-like image [14].

Treatment plan for all findings in radiographs were determined by removing all carious lesions, fractured restoration and application of the suitable restoration for each affected tooth. Supernumerary teeth in all quadrants, needed to choose the well-formed teeth and remove the malformed ones and extra teeth to allow eruption of permanent teeth (Surgical exposure with orthodontic traction). Posterior primary teeth needed to be restored & kept as possible to be used as anchorage for orthodontic appliance for traction of anterior permanent teeth in the 1st phase. Such approaches were previously described [15]. However, for eruption of impacted teeth, particularly in young patients, surgical introduction is favored as it can be concluded in one or two sessions. Orthodontic occlusal measure of the tooth will restore the height of the alveolar ridge to that compatible with normal dental and skeletal growth. Normal periodontal attachment and gingival margins can be achieved, thus eliminating the need for additional periodontal therapy [16,17].

Using the right behaviour management techniques to motivate and encourage the patient to change from his bad lifestyles like irregular tooth brushing, carving a lot of snacks etc.

Improvement of oral hygiene to avoid any further caries in the future. Keep the patient under routine dental follow up every three months to avoid any further caries and keep good oral hygiene status with topical fluoride application every 6 months.

4. Conclusion

Dental treatment plan for CDD were determined by removing all carious lesions, fractured restoration and application of the suitable restoration for each affected tooth. Improvement of oral hygiene is essential for future better quality of life.

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