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Case Report

Dentistry Section

Clinical, Radiographic and Histopathological Evaluation of Cleidocranial Dysplasia in a Child: A Case Report

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ABSTRACT

Cleidocranial Dysplasia (CCD) is a rare autosomal dominant skeletal disorder characterised by clavicular hypoplasia, delayed closure of cranial sutures and multiple supernumerary unerupted teeth. A 12-year-old girl presented with a chief complaint of multiple mobile lower anterior teeth and delayed eruption of permanent teeth. General examination revealed short stature, partial shoulder approximation and genu valgum. Extraoral findings included frontal bossing and depressed nasal bridge. Intraoral examination showed multiple retained deciduous teeth with only one erupted permanent tooth. Radiographic evaluation {Orthopantomogram (OPG), lateral cephalogram, chest and skull radiographs} demonstrated approximately 49 impacted and supernumerary teeth, clavicular hypoplasia, nasal bone hypoplasia and delayed fontanelle closure. Based on characteristic clinico-radiographic features, a diagnosis of CCD was established. The patient was referred for paediatric evaluation and genetic testing; however, molecular confirmation could not be obtained. Oral prophylaxis and extraction of mobile deciduous teeth were performed and comparative histopathological analysis was undertaken. Early recognition of such characteristic dental findings facilitates timely diagnosis and interdisciplinary management.

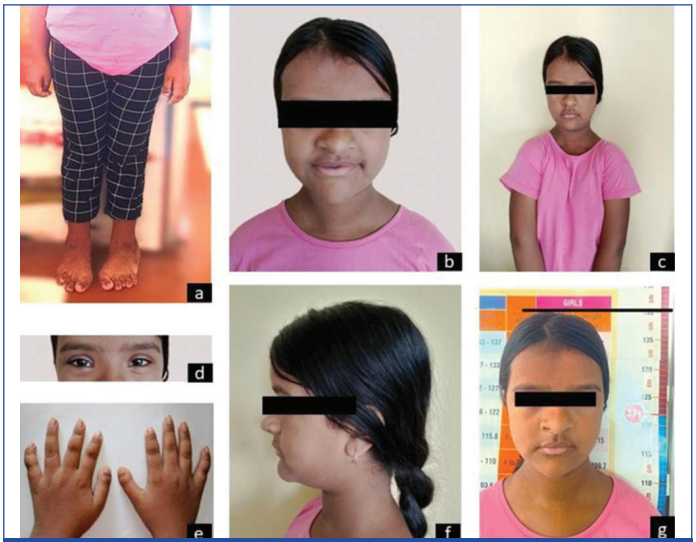
Keywords: Clavicle, Delayed tooth eruption, Impacted tooth, Supernumerary teeth

CASE REPORT

A 12-year-old girl reported to the Department of Paediatric and Preventive Dentistry, with a chief complaint of multiple loose teeth in lower front teeth region in the last 4-5 months. Her parents noticed that several deciduous teeth had become mobile but failed to exfoliate naturally over the past 4-5 months. They expressed concern regarding delayed eruption of the permanent successors. The patient reported no history of pain, spontaneous bleeding, or trauma associated with the mobile teeth. Patient also experienced difficulty in chewing food because of those mobile teeth. The patient's medical history was non contributory. There was no history of systemic diseases, hospitalisations, or surgical interventions, no history of medication use. No known allergies to drugs, food, or environmental allergens were reported. The parents provided a history of the child snoring during sleep. Aside from that, no deleterious oral habits were reported. The child was born to a non consanguineous couple. Both parents were clinically evaluated and no skeletal or dental abnormalities suggestive of any disorder were observed. There was no reported familial history of skeletal dysplasia, genetic syndromes, or developmental disorders among parents/siblings. The patient was a student living with her parents.

General examination revealed genu valgum [Table/Fig-1a], broad forehead with frontal bossing and malformed nares [Table/Fig-1b], drooping shoulders with excessive mobility allowing approximation towards the midline [Table/Fig-1c], hypertelorism [Table/Fig-1d], broad thumbs with conical phalanges and malformed metacarpal and phalangeal joints [Table/Fig-1e], a straight lateral profile with depressed metopic suture and nasal bridge, slightly retrognathic mandible with prominent chin [Table/Fig-1f] and short stature (height 140 cm, weight 32 kg) [Table/Fig-1g].

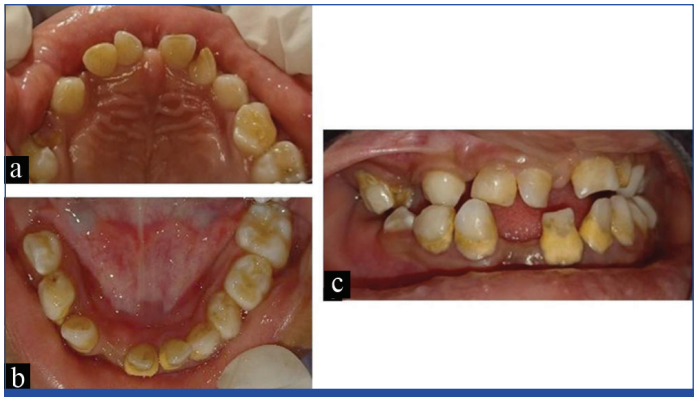
Intraoral findings included a high-arched palate, multiple retained primary teeth, delayed eruption of permanent teeth and poor oral hygiene with grade III calculus. The only erupted permanent teeth in oral cavity was left mandibular first molar [Table/Fig-2a-c].



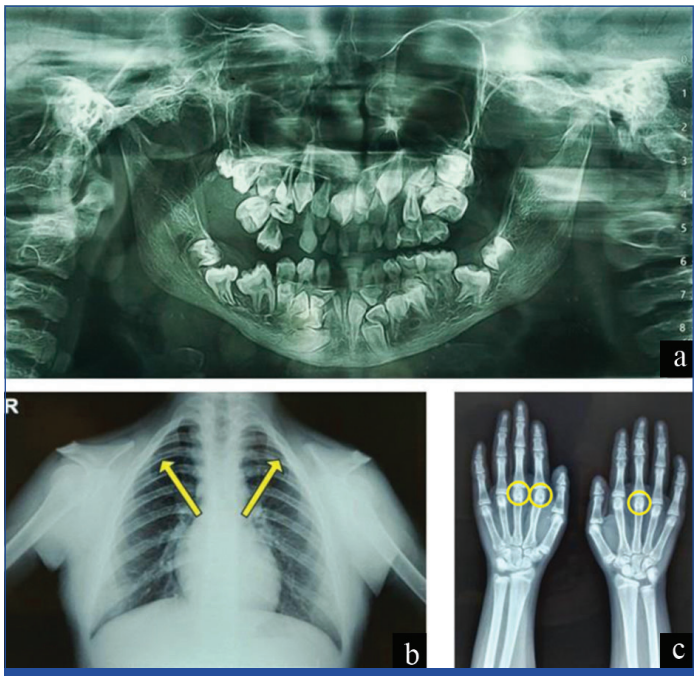
[Table/Fig-1]: Extraoral photographs showing characteristic craniofacial and skeletal features observed in the patient: a) Knocked knees (genu valgum); b) Broad forehead with frontal bossing, broad malformed nares, pentagonal facial form; c) Approximation of shoulders; d) Hypertelorism; e) Malformed metacarpal and phalangeal joints and broad thumbs with conical phalanges; f) Lateral profile showing straight profile, prominent nose and chin, depressed metopic suture and nasal bridge; g) Short stature (height 140 cm).

Radiographic findings: Panoramic radiograph showed approximately 49 teeth in total, mostly unerupted. OPG revealed multiple impacted permanent and supernumerary teeth and several malformed permanent teeth, along with deviated nasal septum [Table/Fig-3a]. Chest Posteroanterior (PA) view showed bilateral clavicular hypoplasia [Table/Fig-3b]. Hand and wrist radiograph revealed pseudo-epiphyses of metacarpals [Table/Fig-3c].

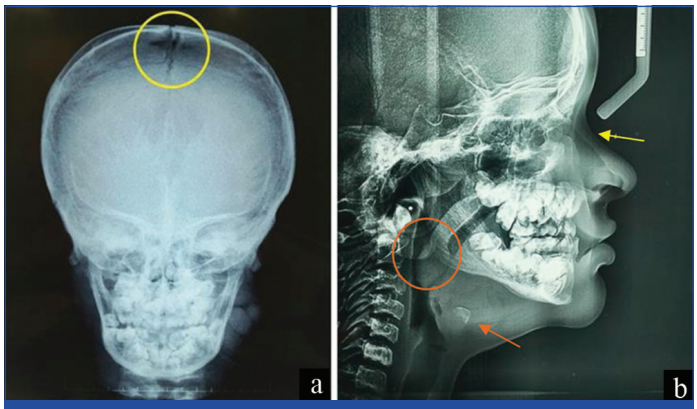
Skull PA view showed delayed anterior fontanelle closure, parietal and occipital bossing, hypoplastic maxilla and reduced frontal sinus pneumatisation [Table/Fig-4a]. Lateral cephalogram revealed nasal bone hypoplasia, broad short Anterior Nasal Spine (ANS), undefined Point A, hypoplastic hyoid, narrow pharyngeal airway and prominent posterior clinoid process [Table/Fig-4b].



[Table/Fig-2]: Intraoral photographs of the patient showing: a) Maxillary arch showing high-arched palate, multiple retained primary teeth, multiple unerupted/impacted permanent teeth; b) Mandibular arch showing multiple retained primary teeth, multiple unerupted/impacted teeth, stain and calculus, only one erupted permanent tooth; and c) Poor oral hygiene with stain and calculus, multiple retained primary teeth, multiple unerupted/impacted teeth and spacing between teeth.



[Table/Fig-3]: Radiographic findings of the patient: a) Orthopantomogram (OPG) showing multiple impacted permanent and supernumerary teeth and several malformed permanent teeth, along with deviated nasal septum; b) Chest PA view showing bilateral clavicular hypoplasia (yellow arrows); c) Hand and wrist radiograph revealing pseudo-epiphyses of metacarpals (yellow circles).



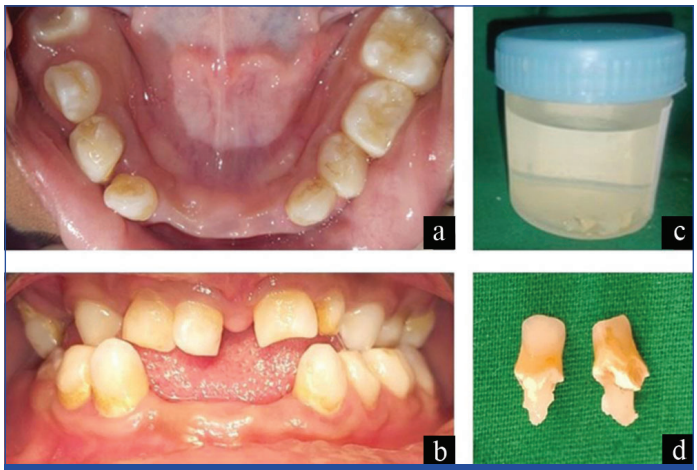
[Table/Fig-4]: Radiographic findings: a) Skull PA view showing delayed anterior fontanelle closure (yellow circle), parietal and occipital bossing; b) Lateral cephalogram showing nasal bone hypoplasia (yellow arrow), broad short Anterior Nasal Spine (ANS), undefined Point A, hypoplastic hyoid (orange arrow), narrow pharyngeal airway (orange circle) and prominent posterior clinoid process.

The differential diagnoses considered included pycnodysostosis, mandibuloacral dysplasia, Yunis–Varon syndrome and Crane-Heise syndrome. Based on the absence of characteristic radiographic and systemic findings associated with these conditions, they were excluded. The presence of bilateral clavicular hypoplasia with shoulder approximation, delayed closure of cranial sutures, multiple impacted and supernumerary teeth, pseudo-epiphyses of metacarpals and characteristic craniofacial features collectively favoured the diagnosis of CCD.

Based on the clinical and radiographic features, a provisional diagnosis of classic CCD was made. Parental consent was obtained for use of the patient's photographs and data.

Patient is referred for the genetic testing for mutation of Runt-related Transcription Factor 2 (RUNX2) gene to confirm the diagnosis of CCD. However, the patient did not return with molecular confirmation, likely due to financial constraints. The definitive diagnosis of CCD was therefore established based on the comprehensive evaluation of the characteristic clinical-radiographic findings.

Treatment: Ultrasonic scaling was done followed by oral hygiene maintenance instructions were given to the patient. Grade III mobile deciduous left lower central and lateral incisors were extracted and sent for histopathological evaluation [Table/Fig-5a-d]. The histopathological features of the extracted teeth were compared with that of a control tooth (extracted from a non CCD patient of similar age). Patient was recalled for periodic follow-up to ensure the eruption of permanent successors.



[Table/Fig-5]: a) Mandibular arch after extraction of mobile deciduous teeth (71 and 72); b) Maxillary and mandibular arch after the extraction; c) Extracted teeth in 10% neutral buffer formalin; d) The extracted teeth 71 and 72.

Histopathology: Ground section of extracted tooth the patient (case, sample 1) was compared with a control deciduous tooth, extracted from a healthy child of similar age (control, sample 2) under 100x magnification.

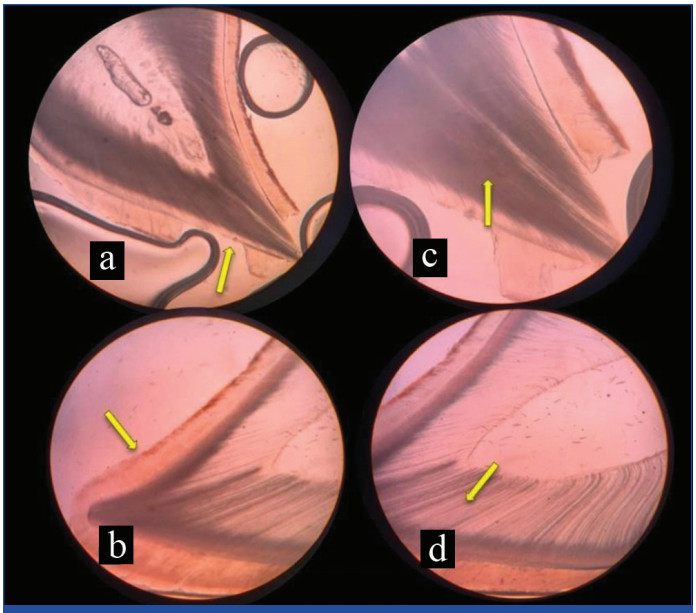
Case: The ground section of tooth (sample 1) revealed the presence of hypoplastic enamel and multiple enamel lamellae along with dead tracts. Two free pulp stones in the coronal portion of the pulp cavity were noted. Dentino-enamel junction was relatively flat with absence of scalloping. Acellular cementum layer is thin and the cellular cementum is absent. The root is resorbed apically [Table/Fig-6,7].

Control: The ground section of (sample 2) tooth revealed the presence of normal enamel with multiple enamel lamellae and tufts. The pulp chamber appears normal. Scalloping was present in the Dentino-Enamel Junction (DEJ). Acellular and cellular cementum layer are present in normal thickness. The root resorption was not found [Table/Fig-6,7].

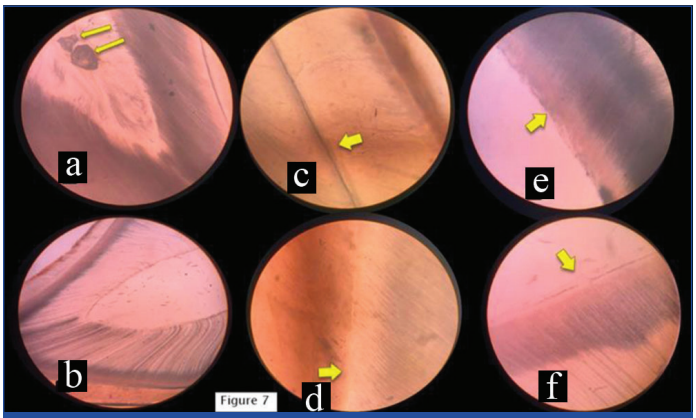
The patient was referred to paediatric medicine department for further systemic evaluation and confirmation of the diagnosis and also for the genetic testing. However, the patient did not report back for the follow-up appointments with the molecular confirmation.

DISCUSSION

The CCD is a rare autosomal-dominant skeletal disorder characterised by clavicular hypoplasia, delayed closure of cranial



[Table/Fig-6]: a) Ground section of sample 1 tooth showing presence of hypoplastic enamel; b) Ground section of sample 2 tooth showing presence of enamel of normal thickness; c) Ground section of sample 1 tooth showing presence of multiple enamel lamellae along with dead tracts in dentin; d) Ground section of sample 2 tooth showing presence of normal enamel with multiple enamel lamellae and tufts (Yellow arrows).



[Table/Fig-7]: a) Ground Section of sample 1 tooth showing two free pulp stones in coronal portion of pulp cavity; b) Ground Section of sample 2 tooth showing normal pulp chamber; c) Ground Section of sample 1 tooth showing absence of scalloping in DEJ; d) Ground section of sample 2 tooth showing scalloped DEJ; e) Ground section of sample 1 tooth showing thin acellular cementum and no cellular cementum; f) Ground Section of sample 2 tooth showing presence of acellular and cellular cement layers in normal thickness. (Yellow arrows)

sutures and multiple impacted or supernumerary teeth. Classical panoramic radiographic findings of CCD, including numerous impacted permanent teeth and supernumerary teeth, were described by McNamara CM et al., [1]. Cooper SC et al., further documented the natural history of CCD and reported characteristic skeletal findings such as short stature, hypermobility of shoulders due to clavicular hypoplasia and delayed cranial ossification [2]. Dentomaxillofacial variability including midface deficiency and altered craniofacial proportions in CCD patients was later described by Golan I et al., [3].

Histopathological alterations in CCD teeth have also been documented in the literature. Manjunath K et al., reported enamel hypoplasia, altered dentinal tubules, pulp calcifications and deficient cementum in affected teeth [4]. These findings correspond with the microscopic features observed in the present case, where ground section analysis revealed hypoplastic enamel, multiple enamel lamellae, dead tracts in dentin, pulp stones and a thin cementum layer.

At the molecular level, CCD is primarily associated with pathogenic variants in the RUNX2 gene, which plays a key role in osteoblast differentiation and skeletal morphogenesis. Thaweesapphithak S et al., demonstrated a strong phenotypic-genotypic correlation in 453 patients, confirming that RUNX2 haploinsufficiency is directly associated with classical skeletal abnormalities such as clavicular hypoplasia and delayed ossification of cranial sutures [5].

Dental anomalies such as delayed exfoliation of primary teeth, delayed eruption of permanent teeth and multiple supernumerary teeth are among the earliest and most characteristic manifestations of CCD. Using three-dimensional radiographic evaluation, Lu Y et al., reported a significantly increased number of impacted and supernumerary teeth with delayed eruption patterns in CCD patients [6].

Several treatment approaches have been described for the dental management of CCD. Orthodontic and surgical traction of impacted teeth has been reported by Soheilifar S et al., [7], while prosthodontic rehabilitation approaches in CCD patients have been described by Karakaya M et al., [8]. Comprehensive reviews have also emphasised the importance of early diagnosis and multidisciplinary management of CCD [9]. Surgical-orthodontic approaches aimed at facilitating eruption of impacted teeth in growing patients were highlighted by Álvarez I et al., [10]. More recently, Shi Y et al., emphasised that failure of eruption and multiple impactions remain the most consistent dental characteristics requiring early orthodontic intervention [11].

In the present case, clinical examination revealed typical skeletal and craniofacial features such as clavicular hypoplasia, delayed cranial suture closure, genu valgum and multiple impacted and supernumerary teeth. Radiographic evaluation revealed a total of 49 teeth, including numerous unerupted permanent and supernumerary teeth, which is consistent with previously reported findings in CCD patients.

Management of CCD is multidisciplinary and may involve surgical exposure of impacted teeth, orthodontic traction and long-term follow-up depending on the severity of dentofacial anomalies [10,11]. In the present case, only oral prophylaxis and extraction of mobile retained deciduous teeth were performed and the patient was advised periodic follow-up and referred for further multidisciplinary evaluation.

The uniqueness of this case lies in the comparative histopathological evaluation of extracted deciduous teeth from a CCD patient and a non CCD patient of similar age. Such microscopic comparison has rarely been documented in the literature and therefore contributes additional insights into the dental tissue alterations associated with CCD.

CONCLUSION(S)

The present case highlights that the combination of bilateral clavicular hypoplasia with shoulder approximation, delayed cranial suture closure and approximately 49 teeth in total including several impacted and supernumerary teeth on panoramic radiograph was diagnostic of CCD in this 12-year-old patient. Comparative histopathological evaluation further demonstrated enamel hypoplasia, altered dentinal architecture, pulp calcifications and deficient cementum. The patient was referred for medical evaluation and genetic testing; however, molecular confirmation could not be obtained. Early recognition of such characteristic dental findings by paediatric dentists is crucial, as they may represent the first clinical indicator prompting timely diagnosis and interdisciplinary management.

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