

Clinical and Radiological Evaluation of Cleidocranial Dysplasia in Two Trinidadian Siblings

AK Bissoon, K Moze

ABSTRACT

Cleidocranial dysplasia is a rare developmental disorder of the skeleton and teeth that may be inherited as an autosomal dominant trait or occur spontaneously. This is a report of two Trinidadian, East Indian brothers aged 13 and 15 years referred from a private dental practice with the chief complaint of retained deciduous teeth. Subsequent clinical and radiographic investigations led to the diagnosis of cleidocranial dysplasia. The clinical and radiographic findings are discussed.

Keywords: Cleidocranial dysplasia, radiography, supernumerary

Evaluación Clínica y Radiológica de la Displasia Cleidocraneal en dos Hermanos Trinitarios

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RESUMEN

Displasia cleidocraneal es un raro trastorno del desarrollo del esqueleto y los dientes, que puede ser heredado como un rasgo autosómico dominante o ocurrir espontáneamente. Éste es un reporte acerca de dos hermanos indotrininitenses de 13 y 15 años, remitidos por una clínica dental privada por causa de dos dientes deciduos retenidos. Las investigaciones clínicas y radiográficas posteriores llevaron al diagnóstico de una displasia cleidocraneal. Se discuten los resultados clínicos y radiográficos.

Palabras claves: Displasia cleidocraneal, radiografía, supernumerario

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INTRODUCTION

Cleidocranial dysplasia (CCD) or cleidocranial dysostosis is an autosomal-dominant malformation syndrome, but can occur spontaneously in about 40% of cases (1). Its incidence is very rare, estimated at one per million (2, 3). It affects both genders equally (3). The disease primarily affects the skull, clavicles and dentition, although a variety of anomalies may be found in other bones. Patients tend to have a short stature, a large head with frontal and parietal bossing and ocular hypertelorism. The sutures and fontanelles of the skull show delayed closure or may remain open and wormian bones may be observed on skull radiography. The midfacial skeleton may be hypoplastic, resulting in relative mandibular prognathism. The paranasal sinuses may also be underdeveloped and the bridge of the nose broad and depressed.

There may be complete absence of clavicles to a partial absence or marked thinning of one or both clavicles. This usually leads to hypermobility of the shoulders so much so that some patients can approximate their shoulders.

Dental abnormalities include multiple supernumerary teeth, prolonged retention of deciduous teeth and delay or failure of eruption of permanent teeth. Thus, dental panoramic radiography is a variable adjunct in confirming the diagnosis of cleidocranial dysplasia (4). Although cleidocranial dysplasia is rare, it is integral for dental practitioners to be aware of its features because the dental abnormalities produce the most morbidity associated with the disease and are often the reason for diagnosis in some individuals. This report highlights and discusses the clinical and radiographic features of the disease as seen in two siblings, only diagnosed at age 13 and 15 years of age after a visit to a general dental practitioner.

From: School of Dentistry, The University of the West Indies, St Augustine, Trinidad and Tobago.

Correspondence: Dr AK Bissoon, #6 John Peter Road East, Chaguanas, Trinidad and Tobago, West Indies. E-mail: ktlou556@hotmail.com

CASE REPORT

A 13-year old boy with no significant medical history presented to the Paediatric Dental Clinic at the University of the West Indies Dental School referred by a general dental practitioner. His chief complaint was multiple retained deciduous teeth. His mother indicated in the history that his brother, aged 15 years, also has the same problem and that she still had deciduous teeth. She also indicated that her older brother and her mother suffered with the same problem.

On clinical examination, it was noted that the patient had a short stature, frontal bossing, a broad nose bridge, an increased lower facial height and clavicles that were reduced in size. The patient also exhibited hypermobility of his shoulders (Figs. 1–3). Intra-oral examination revealed that



Fig. 1: Frontal view of 13-year old.



Fig. 2: Right side view of 13-year old.



Fig. 3: The 13-year old patient exhibiting hypermobility of shoulders.

all the deciduous teeth were still present. There was also an erupted mesiodens (Fig. 4). The maxillary right first permanent molar was erupted and the left first permanent molar was partially erupted. The maxillary anterior teeth had



Fig. 4: Anterior teeth in occlusion of 13-year old. Mesiodens present.

severe tooth wear and the mandibular anterior teeth to a lesser extent (Figs. 5, 6). Caries were present on the upper second deciduous molars and the upper right first permanent molar.



Fig. 5: Mandibular teeth in 13-year old. Tooth wear evident on lower incisors.



Fig. 6: Maxillary teeth in 13-year old. Severe tooth wear evident on maxillary incisors and canines.

Panoramic radiography revealed the presence of multiple supernumerary and unerupted teeth in both the mandible and maxilla (Fig. 7). The supernumeraries were mostly

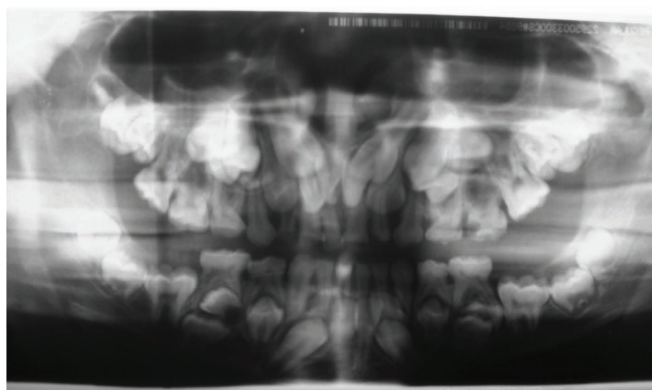


Fig. 7: Panoramic radiograph of 13-year old. Multiple unerupted and supernumerary teeth are evident.

localized to the anterior maxilla and lower premolar regions, however, one was present in the left mandibular third molar region (Fig. 8) and another was developing in the right maxillary third molar region (Fig. 9). A maxillary standard occlusal radiograph was also taken (Fig. 10). A chest X-ray was performed to assess the clavicles and they were found to be markedly thin and partially absent (Fig. 11).

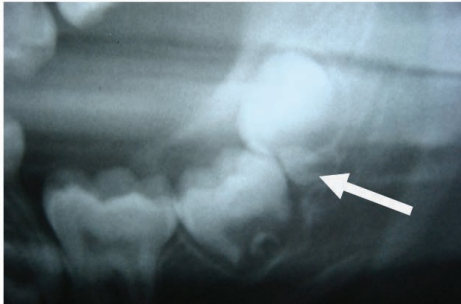


Fig. 8: Part of panoramic radiograph of 13-year old showing developing supernumerary in left mandibular third molar region (arrow).

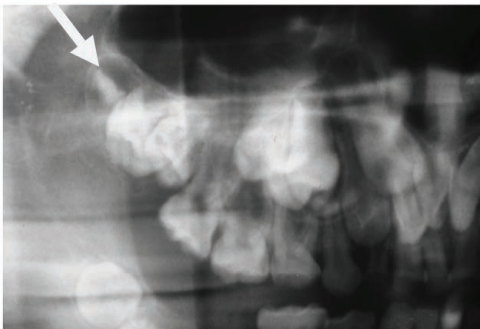


Fig. 9: Part of panoramic radiograph of 13-year old showing developing supernumerary in right maxillary third molar region (arrow).

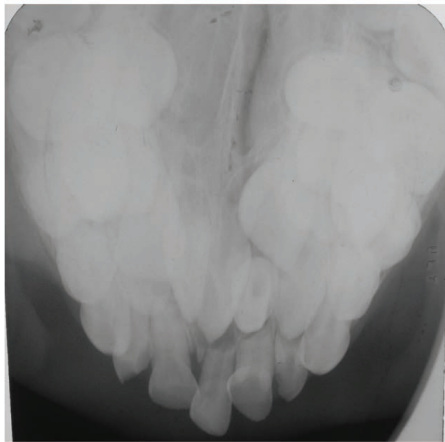


Fig. 10: Standard maxillary occlusal radiograph of 13-year old showing multiple impacted teeth and supernumeraries.

Subsequently, the parent was asked to bring in the older brother and younger sister for examination. Examination of the 15-year old brother revealed similar findings. He exhibited a short stature, frontal bossing, clavicles that were reduced in size and hypermobility of the shoulders (Figs. 12, 13). Intra-orally, all of the deciduous teeth were still present but the right permanent central incisor was erupted buccally to the maxillary deciduous incisors (Fig. 14). All permanent first molars were present except for the mandibular right first molar, which was extracted previously (Fig. 15). The maxil-

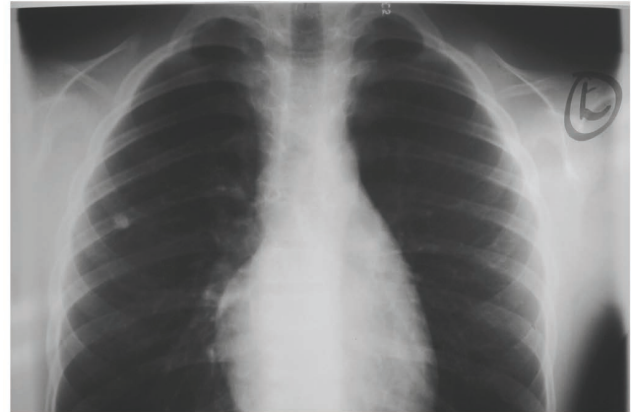


Fig. 11: Chest X-ray of 13-year old showing partially absent and thinned clavicles.



Fig. 12: Frontal view of 15-year old.

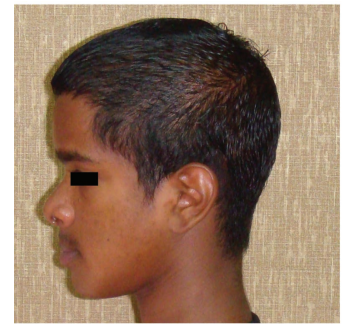


Fig. 13: Left side view of 15-year old.



Fig. 14: Intraoral view of 15-year old. Hypoplastic maxilla is evident.



Fig. 15: Intra-oral view (mandibular teeth) of 15-year old.

lary right deciduous incisor showed grade II mobility. There was a class III incisor relationship and an anterior open bite with the maxillary arch being hypoplastic and retrognathic (Fig. 14). The maxillary deciduous canines and first molars had moderate tooth wear (Fig. 16). Panoramic radiography and standard occlusal radiographs of the maxilla and mandible revealed multiple unerupted teeth and supernumeraries (Figs. 17–19). Supernumeraries were localized to the anterior maxilla and mandibular premolar region. None was observed in the molar regions. Chest X-ray also revealed partially absent clavicles, which were markedly thinned (Fig. 20). However, they were less thin than those of his sibling. Based on the family history and clinical and radiographic



Fig. 16: Intra-oral view (maxillary teeth) of 15-year old.

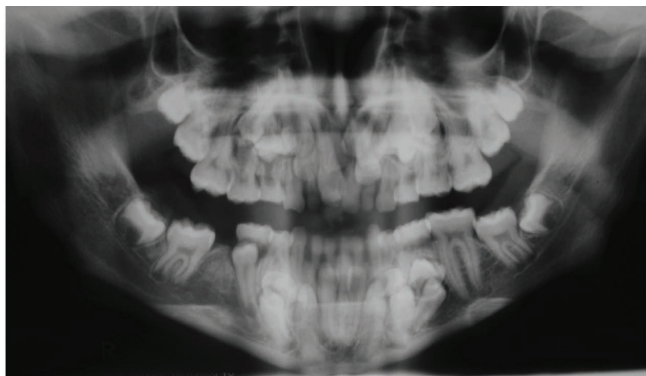


Fig. 17: Panoramic radiograph of 15-year old. Multiple unerupted and supernumerary teeth are evident.

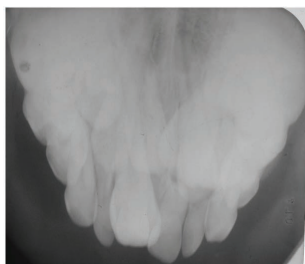


Fig. 18: Standard maxillary occlusal radiograph of 15-year old showing multiple impacted teeth and supernumeraries.

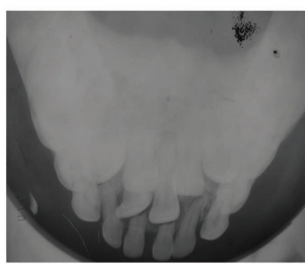


Fig. 19: Standard mandibular occlusal radiograph of 15-year old.



Fig. 20: Chest X-ray of 15-year old showing partially absent and thinned clavicles.

findings, diagnoses of cleidocranial dysostosis were made. The patients were referred for combined orthodontic, paediatric and oral surgery consultations to allow treatment planning and further management.

DISCUSSION

Cleidocranial dysplasia, also known as Scheuthauer-Marie-Sainton syndrome (5), is an autosomal-dominant disease affecting the skeleton and dentition which can be inherited or arise as a result of sporadic mutation. The responsible gene has been identified as *RUNX2* on chromosome 6 (6). Defects chiefly affect membranous bones with the skull and clavicles being the chief site of the disorder.

Characteristically, patients with this disease show prolonged retention of the deciduous dentition and delayed or failure of eruption of the permanent dentition. Multiple supernumerary teeth are usually seen and it has been suggested that there is an involvement of a non-genetic or epigenetic regulation in supernumerary tooth formation when dental characteristics of siblings with the identical gene mutation were examined (6). The number of supernumerary teeth has been correlated significantly with short stature in some studies (7). There is a predisposition to develop numerous supernumerary teeth in the mandibular premolar and maxillary anterior regions (5), but they are rare in the molar regions (8).

Cleidocranial dysostosis may be diagnosed by the family history, excessive mobility of the shoulders, clinical examination of the skull and radiographic findings that are pathognomonic of the disease, like prolonged retention of deciduous teeth and multiple supernumerary teeth. Differential diagnoses include pycnodysostosis or Maroteaux-Lamy syndrome (3). Maroteaux-Lamy syndrome may be differentiated by the presence of dwarfism and patients affected by the syndrome have dense and fragile bones.

Management is largely dependent on the chronological and dental ages of the patient and is mainly supportive with no treatment of the underlying disorder. A multidisciplinary approach should be employed. Treatment includes multiple surgical exposures of unerupted teeth and orthodontic treatment to establish an intact and aligned dental arch. At skeletal maturity, the underlying skeletal deformity correction can be done by maxillary Le Fort I osteotomy. Extraction of all teeth followed by the fabrication of dentures or autotransplantation of selected impacted teeth followed by prosthetic restoration can also be performed. Endosseous implants are given to restore the mandibular and maxillary arch with fixed prosthesis for edentulous patients (9–11).

It is integral to report cases such as those presented in this paper because of the rarity of this disease. The clinical and radiographic findings were characteristic of cleidocranial dysplasia and it is noteworthy to mention that the 13-year old presented with supernumeraries in the molar regions which is rare (5). The presence of severe tooth wear in his case necessitates proper treatment planning and follow-through

with planned procedures. The 15-year old brother may have a better functional prognosis as tooth wear is less and there are apparently fewer supernumeraries than his sibling. However, his maxilla is hypoplastic, resulting in aesthetic effects as well as malocclusion. Thus, careful orthodontic and oral surgery consideration must be given to his case. Further investigations are to be performed on their mother to identify the extent of her condition and possible treatment options if required. Thus far, investigations on their six-year old sister have revealed that she is unaffected.

In conclusion, although cleidocranial dysplasia is a rare disease, it is important for general dental practitioners to recognize its features to enable early treatment planning and intervention if necessary. An interdisciplinary approach to diagnosis and treatment should be emphasized.

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