Case Report

Skeletal and Dental Features of Cleidocranial Dysplasia

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Abstract

Cleidocranial Dysplasia is an autosomal dominant condition characterised by partial or complete absence of clavicles, multiple impacted supernumerary teeth, delayed closure of cranial sutures and other skeletal abnormalities. It is caused due to a mutation in the RUNX2 gene which is responsible for bone and tooth formation. Cleidocranial Dysplasia presents with characteristic skeletal and dental features which are diagnostic of the condition. As retention of deciduous teeth and delayed eruption of the permanent teeth lead to aesthetic and functional concerns, these patients often report first to the dentist for evaluation. In this case report, we discuss two cases of cleidocranial dysplasia which were diagnosed by their dental findings.

Keywords: Cleidocranial dysplasia, supernumerary teeth, clavicle.
Introduction
Cleidocranial dysplasia (CCD), also known as Marie and Sainton disease, Mutational dysostosis and Cleidocranial dysostosis, is a rare congenital defect primarily affecting bones that undergo intra-membranous ossification; generally the calvarian but also the clavicular bones[1]. CCD is characterized by delayed closure of the cranial sutures, hypoplastic or aplastic clavicles and multiple dental abnormalities. Patients usually present with open skull sutures, hypoplastic or aplastic clavicles, supernumerary teeth, delayed eruption of permanent dentition, wide pubic symphysis, short stature and a variety of other skeletal changes [2]. The major problems faced by these patients is due to the abnormalities associated with their teeth [3]. Thus the dentist is usually responsible for the diagnosis of this condition. We report two cases of cleidocranial dysplasia diagnosed through dental findings.

Figure-1A: Clinical photograph showing frontal bossing with depressed central part of forehead, flattened superior orbital ridges and depressed nasal bridge.
Figure-1B: Intraoral photograph of the maxillary arch showing a solitary erupted tooth.
Figure-1C: Intraoral photograph of the mandibular arch showing erupted teeth in the mandibular anterior and right premolar region.

Case report
Case 1
A 50-year-old male reported to the department of Oral medicine and Radiology with complaint of missing upper and lower teeth. His history stated that there was delay in the eruption of his permanent teeth and the few teeth present in the mouth were lost by spontaneous fragmentation and exfoliation.
On examination, he had proportionately short stature and pigeon shaped chest with increased anteroposterior diameter. The clavicles were not palpable bilaterally. On extraoral examination he showed frontal bossing with depressed central part of forehead, flattened superior orbital ridges
and depressed nasal bridge. (Figure-1A) Intraoral examination revealed low arched palate with enlargement of the maxillary alveolar ridge. (Figure-1B) In the mandible, this enlargement was predominantly in the anterior region. (Figure-1C) A total of three teeth were visible, one in the maxilla and two in the mandible. All other teeth were clinically absent. Panoramic radiography revealed multiple unerupted teeth predominantly in the maxillary left posterior and mandibular anterior region. (Figure-2A) Posteroanterior skull radiograph showed wormian bones in the skull and open anterior fontanelle. (Figure-2B) Chest radiograph showed bilateral absence of clavicles. (Figure-2C) Based on the above features, we arrived at a diagnosis of cleidocranial dysplasia. Surgical removal of all the unerupted teeth followed by alveoloplasty was then done and a removable complete denture was fabricated.

Case 2
An 18 year old female patient reported to the Department of Oral Diagnosis and Radiology with complaint of missing upper right front tooth. She gave a history of multiple over retained deciduous teeth which were extracted by a local practitioner. Following this, there was eruption of the permanent teeth. Since the anterior permanent teeth did not erupt, she consulted our institution. On extraoral examination, she had frontal bossing with depressed centre of forehead, depressed bridge of nose and flattened supraorbital ridges. (Figure-3A) Intraoral examination showed retained 53,63,73,83 and missing 11.(Figure 3B& 3C) Panoramic radiography showed multiple impacted supernumerary teeth.(Figure-4A) Chest radiograph showed absence of clavicles bilaterally(Figure-4B ) leading to the diagnosis of cleidocranial dysplasia.
Discussion
Cleidocranial dysplasia was first described in 1765 by Martin in a patient with congenital absence of clavicles but it was in 1898 that Marie and Sainton described cases of the disease and associated them with patterns of inheritance [4,5]. This condition is usually caused by a mutation of the RUNX2 (Core Binding Factor-α1) gene, located at chromosome 6p21. 6 This gene encodes a protein which is essential for osteoblast and dental cell differentiation as well as for bone and tooth formation [5, 6]. However, 40% of the cases of CCD appear spontaneously with no apparent genetic cause [5,6].

Figure-3A: Clinical photograph showing frontal bossing with depressed central part of forehead, flattened superior orbital ridges and depressed nasal bridge.
Figure-3B: Intraoral photograph of the maxillary arch showing missing 11.
Figure-3C: Intraoral photograph of the mandibular arch showing retained 73, 83.

CCD is a relatively uncommon disorder with a prevalence of 0.5 per 100,000 live births [6]. Males and females are equally affected [7]. The most characteristic and pathognomonic skeletal feature is the complete or partial absence of one or both clavicles [8]. In both our reported cases, the clavicle was absent. The dental features are another significant part of the disease with delayed eruption and impaction of deciduous and permanent teeth. Case 2 showed over retention of the deciduous teeth. Root resorption is delayed[9]. The permanent teeth lose their eruption stimulus and remain embedded [9]. This was a feature of Case 1. It is thought to be due to the absence of cellular cementum and an increase in the amount of acellular cementum of the roots of the affected teeth [3,5,9]. In addition to the above, delayed closure of the fontanelles and sutures with wormian bones are also reported in cases of CCD [2]. Case 1 showed both of these features.

Less common findings of CCD include short stature, a bell-shaped thorax, hypoplasia of the pelvis, enlargement of the frontal and occipital bones, and phalangeal abnormalities. Shortened or absent nasal bones, paranasal sinus abnormalities, thickening of some segments of the calvaria, small maxillae, and delayed union of the mandibular symphysis [2].

The radiographic evaluation of patients is the most important and reliable means to
confirm the diagnosis, since cleidocranial dysplasia has characteristic radiological features such as broad sutures, large fontanels persisting into adulthood, numerous wormian bones and multiple unerupted supernumerary teeth [1]. In both our cases, cleidocranial dysplasia was diagnosed based of the radiographic findings.

The suggested treatment for CCD includes a combined orthodontic and surgical approach with timely extraction of deciduous teeth, extraction of supernumerary teeth, exposure of selected unerupted permanent teeth and orthodontic forced eruption and fabrication of dentures [1,9].

Figure-4A: Panoramic radiograph showing multiple impacted supernumerary teeth. Figure-4B: Chest radiograph showing bilateral absence of clavicles.

In both cases, surgical removal of the impacted teeth was done before fabrication of prosthesis. Although psychosocial disorders associated with abnormal facial and body features may occur, the subjects with cleidocranial dysplasia have normal intelligence, with an overall good prognosis and normal life expectancy[8].

Cases with cleidocranial dysplasia can be diagnosed easily provided the clinician is alert to the possibility of its existence and the aesthetic and functional problems that these patients face can then be appropriately managed.

Reference