

# ORAL FEATURES AND ORTHODONTIC TREATMENTS IN SUBJECTS WITH CLEIDOCRANIAL DISPLASIA: a review

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# ORAL FEATURES AND ORTHODONTIC TREATMENTS IN SUBJECTS WITH CLEIDOCRANIAL DYSPLASIA: a review

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## Abstract

The Cleidocranial Dysplasia (CCD), is a rare autosomal dominant disease due to a genetic mutation; it primarily affect skull, clavicles and dentition. Dentists are often the first to encounter patients with CCD, for this reason it is important they have a deep knowledge about the clinical features of this disorder. A review of the international literature have been made to report the cranio-facial and dental features of the CCD and to investigate the orthodontic therapeutic approaches for the functional and aesthetical rehabilitation of this disorder.

## Introduction

Cleidocranial Dysplasia (CCD) is a rare inherited disorder that occurs approximately in one in a million individuals. This pathology is caused by mutations in the RUNX2 gene on chromosome 6p21 that is responsible for osteoblast differentiation and skeletal morphogenesis; it may be autosomal dominant or caused by a spontaneous genetic mutation, with complete penetrance and variable expressivity (1). It is characterised by defective ossification, delayed bone and tooth development, stomatognathic and craniofacial abnormalities (2). The purpose of this review is to collect and analyze data in the literature on orofacial typical manifestations of the syndrome and to present the therapeutic approaches.

## Methods

A systematic review of literature has been carried out on PubMed (Medline) database up to december 2018. Used keywords were: cleidocranial dysplasia, CCD, cleidocranial dysostosis. Only articles regarding epidemiology, role of genetics, anatomical features, diagnosis and treatment were considered for this review, while all the other articles regarding specific aspects of the pathology or a specific treatment of one aspect have been excluded.

## Review

Clinical Features:

CCD prominently affects bones of membranous origin. Affected individuals typically present underdeveloped clavicles ("cleido-" in the condition name refers to these bones). As a result, their shoulders are narrow and sloping, can be brought unusually close together in front of the body, and in some cases can be made to meet in the middle of the body (3). A narrow thorax allows the proximity of the shoulders in front of the chest (4). Defects of the cervical and lumbar vertebrae are included in the clinical findings. Absence of the pubic symphysis and hypoplasia of the pelvis is common in female individuals. Frequently, genua valga and pes planus are found in children younger than 5 years of age (5). Moderately short stature was observed. The arms and legs are abnormally short; furthermore, the defects are revealed in hands, feet, and nails (6). Hearing problems are often described, they are a result of abnormalities in the development of the facial skeleton, a high, arched palate, reduced paranasal sinuses and mastoid cells (or a lack thereof), dysfunction of the auditory tube and frequent inflammation of the middle ear (7). The mental development of these patients is usually normal.

Skull and facial manifestations include delayed ossification of the cranial sutures and fontanelles (that may remain open through out life), brachycephalic head with an increased transverse diameter of cranium, pronounced frontal and parietal bone, occipital bossing, formation of Wormian bone, ocular hypertelorism, and broad-based nose (3). Patients tend to have a skeletal Class III malocclusion due to mandibular hyperplasia along with hypoplasia of mid-face. Vertical facial growth is decreased due to hypoplasia of alveolar bone (8).

Oral features include deficient mid-face, underdeveloped paranasal sinuses, high arched palate, delayed or failed exfoliation of deciduous dentition and a delayed eruption of the permanent teeth; consequently, adults with CCD frequently have a mixed dentition. In addition, patients with this condition often exhibit a high number

of included supernumerary teeth that frequently induce follicular cyst formation. Up to 94% of people with CCD have dental findings including supernumerary teeth (70%) and eruption failure of permanent teeth. The permanent first and second molars are rarely affected, but spontaneous eruption is usually delayed. Teeth abnormalities include enamel and cementum hypoplasia, root dilaceration, and microdontia. Submucous cleft palate as well as complete cleft of hard and soft palates have also been reported (9).

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Treatment:

The clinical manifestations of CCD are often innocuous, but hyperdontia and the other developmental abnormalities of the teeth are a major feature and require a multidisciplinary cooperation between orthodontists, oral and maxillofacial surgeons. The planning of dental treatment in CCD varies from individual to individual and depends on the type and severity of anomalies, the age at diagnosis, and social and economic circumstances. The main objectives remain the restoration of craniofacial and dental function together with esthetics (10). Although there are numerous options, there is a general consensus that the best results are obtained if the condition is diagnosed and treated at an early age.

Therapy may include surgical exposure of impacted teeth in combination with removal of supernumerary teeth. Already during the surgical procedure orthodontic bands and brackets may be attached. The next step in treatment should consist of orthodontic alignment of the teeth and correction of the jaw relations. In most cases, midfacial hypoplasia and corresponding abnormal jaw relations are also present. This deformity is best treated with midfacial osteotomy. Following this second surgical intervention, orthodontic treatment is completed in order to establish an optimal result regarding function and esthetics (11,12).Â

Â The most popular orthodontic-surgical regimes are the Toronto-Melbourne, Belfast-Hamburg, and Jerusalem approaches (13,14).Â

The Toronto-Melbourne approach is based on time. The best period for treatment begins at 5 to 6 years of age. The timing of serial extraction of deciduous teeth depends on the extent of the root lengths developed in permanent teeth. Supernumerary teeth are also extracted with the alveolar bone covering the impacted teeth. The rationale of this approach is to facilitate the spontaneous eruption of impacted permanent teeth (15).Â

The Jerusalem approach needs 2 surgical interventions. In the first, the anterior primary teeth and all supernumerary teeth are extracted, followed by the exposure of permanent incisors at 10 to 12 years of age. In the second phase, the posterior primary teeth are extracted, and the impacted permanent canines and premolars are exposed after age of 13. The surgery removes the barrier on the eruption path and promotes the normal eruption pattern of impacted teeth. However, two-thirds of the roots in permanent teeth have already developed in this approach, further orthodontic traction is usually needed.

The Belfast-Hamburg approach consists in a single surgery under general anesthesia to remove all retained elements and supernumerary teeth, and to expose the impacted permanent teeth. After healing orthodontic traction is performed (16, 17). The advantage of this procedure is that the patient is exposed to only a single surgical operation under general anesthesia.

In the Bronx approach, the first phase is to remove primary teeth as well as supernumerary teeth and expose the impacted teeth. The use of removable partial overdenture is for esthetic and functional purposes. Orthodontic treatment starts after the spontaneous eruption of permanent teeth for sufficient posterior support, then a Le Forte I osteotomy is performed. Finally implants are placed to restore dentition defect (18).Â

Each approach has different indication and outcomes, but something that all procedures have in common is that all are undertaken over a long period; for this reason the patient compliance is essential for any of these modalities. The skeletal anomalies and complex multiple dentition of CCD add much difficulty and uncertainty to the orthodontic treatment. What is the best treatment method of CCD is still under evaluation (19,20).

## Conclusions

Surgical exposure combined with orthodontic traction is an effective treatment for patient with CCD. Many valid orthodontic-surgical approaches are reported in literature; patient's age, demand, economic circumstances, and status of primary and permanent dentition should be considered when making treatment plan. Treatment of patients with CCD requires an early diagnosis and an interdisciplinary approach and collaboration among specialists, the patient and the patient's family.Â

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