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Case Report

Multidisciplinary management of cleidocranial dysplasia

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ABSTRACT

Background: Cleidocranial dysplasia (CCD), also known as *cleidocranial dysostosis* or *osteodental dysplasia,* is an autosomal dominant disorder caused by a microdeletion defect in chromosome 6p21. The causative factor of this disorder is a mutation in the gene encoding in the transcription factor core binding factor α 1 (*CBFA1*), and its incidence is 1 in 100,000 population. The aims of this clinical paper are to illustrate the ways of diagnosing and treating CCD as stated in the literature, and to report the multidisciplinary management of a patient with this condition.
Methods: Clinical and radiographic examinations are primordial for diagnosis because the most frequent features of CCD are a pathognomonic deformity of the skull (open skull sutures and delayed closing of cranial fontanels, presence of multiple Wormian bones), midfacial hypoplasia, a wide forehead, pronounced frontal bossing and hypertelorism, supernumerary and impacted teeth, hypoplastic or absent clavicles, and short stature. The diagnosis of CCD is obvious when there is a family history.
Results: Many solutions for the treatment of dental disorders in CCD have been suggested, and the intervention of several dental specialists, namely, the oral surgeon, orthodontist, periodontist, and maxillofacial surgeon, was required to achieve optimal results.
Conclusion: Orthodontic treatment brought all of the teeth on the arches, and the patient was able to sustain orthognathic surgery to correct the skeletal open bite and Class III relationship.

1. Introduction

According to Meckel [1], "rudimentary clavicle" and accompanying "skeletal abnormalities" were quoted and recognized as early as 1760, but the first clinical case addressing clavicular defects was reported by Martin in 1765 [2]. Another case depicting an affected skull and both clavicles was published by Scheuthauer in 1871 [3], and in 1897, Marie and Sainton [4] coined the descriptive title "cleido-cranial dysostosis" (CCD), while in 1926, Hesse [5] was the first author to describe, in detail, the dental and jaw defects associated with CCD. In fact, many authors assert that this disease—or similar ones—were described in earlier literature [6–8]. Indeed, CCD is a well-defined and rare congenital disorder of the bones, distinguished by abnormalities of the clavicles, skull, jaws and teeth, and occasionally, by a limitation of growth of the long bones. Oral expression of this disorder is characterized by failure of eruption of the secondary dentition and multiple (unerupted) supernumerary teeth [9]. CCD diagnosis relies on radiographic and clinical features and findings (imaging of thorax, skull, pelvis, and hands are mandatory for review and final diagnosis). Its transmission is an autosomal-dominant mode involving a mutation in the gene that encodes the transcription factor Runx2/core binding factor α 1 (Cbfa1) [10–12] (located on chromosome 6p21), essential for the proliferation of osteoblasts and dental cells for bone and tooth formation, respectively [13]. *RUNX2* (*CBFA1*) has proved to be the only gene associated with CCD, and *RUNX2* molecular genetic testing is able to detect mutations in 60% to 70% of individuals diagnosed with a CCD. The disease gene was mapped to chromosome 6p21, within a region containing *CBFA1,* the gene that encodes a member of the runt family of transcription factors, and many studies have shown that haploinsufficiency of Cbfa1 causes the CCD phenotype. Cbfa1 controls the differentiation of precursor cells into

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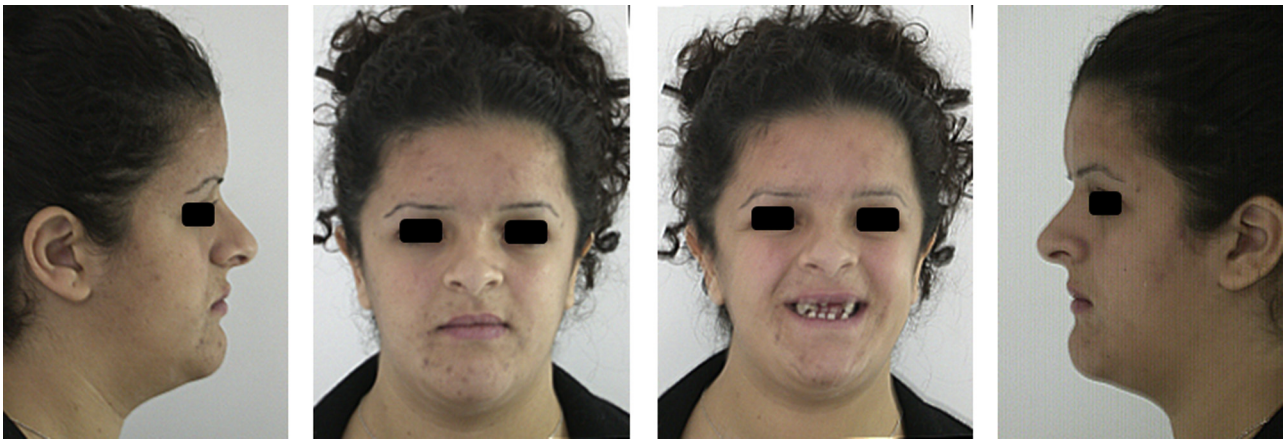

Fig. 1. Facial frontal and lateral aspects.

osteoblasts and is thus essential for membranous as well as endochondral bone formation [14]. The incidence of this disorder is 1 in 100,000 worldwide population [14,15]. Bones undergoing intramembranous ossification are primarily affected, especially the skull, clavicles, and flat bones, hence the term *cleidocranial* [14].

The characteristics of CCD are statural and head and neck malformations. Statural malformations involve hypoplasia or aplasia of the clavicles [2], osseous malformations in the cranial base and skull (brachycephalic head) [3], narrow and abnormally shaped pelvic and pubic bones, deformations in the thoracic region, vertebral defects, and bossing of the frontal, parietal, and occipital bones. Head and neck malformations entail delayed closure of cranial fontanels [16,17], maxillary hypoplasia, multiple (jaw) follicular cysts [5], multiple impactions and supernumerary teeth [9], unerupted permanent teeth [18], prolonged retention of deciduous dentition (delayed exfoliation), twisted roots, malformed crowns, malocclusion, and crowding. The purpose of this clinical paper is to report a case of CCD because of its rarity. We address the diagnosis and the different stages of treatment of this case, through a multidisciplinary approach to achieve optimal aesthetic and functional goals.

2. Diagnosis and etiology

A female patient aged 18 years and 1 month presented for orthodontic consultation at the Department of Orthodontics and Dentofacial Orthopaedics, Lebanese University School of Dentistry, Beirut, Lebanon. Her chief concern was missing and malpositioned teeth. The patient was in good health, free of any other disease. She was a thumb sucker until 7 years of age and a nail biter until 15 years of age.

2.1. Clinical examination

The diagnosis of CCD was first based on the clinical examination revealing a triangular, symmetrical face; wide forehead; pronounced frontal bossing; wide nasal bridge; Asiatic slant; and hypertelorism. Her profile was concave, with a bulging forehead, large nose, and flat midface (Fig. 1).

As a general feature, the patient was able to approximate her shoulders until they touched each other, disclosing and confirming the absence of clavicles, which is a characteristic aspect of CCD (Fig. 2).

The intraoral examination displayed a highly arched palate, thick oral fibromucosa and alveolar ridges, the absence of many teeth, and the persistence of deciduous ones [19]. Her occlusion was Class III molar on both sides. An anterior open bite and bilateral posterior crossbite were present, along with gingival recessions on her mandibular central incisors (Fig. 3).

2.2. Radiographic examinations

Lateral and posteroanterior cephalograms displayed open cranial fontanels and skull sutures; midfacial hypoplasia; underdeveloped maxillary sinuses, nasal, and zygomatic bones; midfacial hypoplasia (or pseudoprognathism); and the presence of multiple Wormian bones [20–22] (Fig. 4A).

The chest radiograph revealed the absence of both the right and left clavicles (Fig. 4B), which is characteristic of CCD.

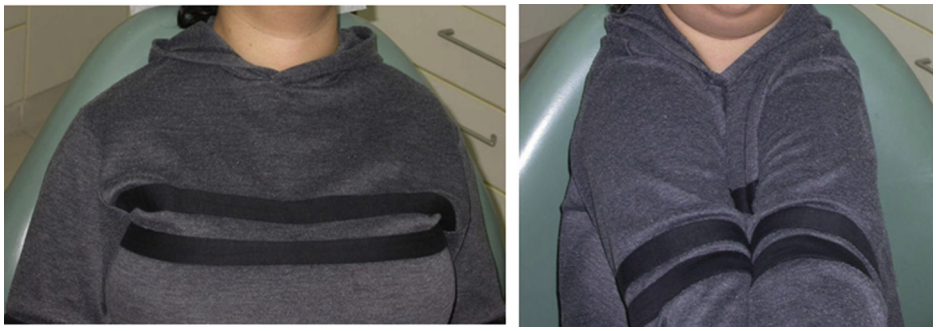

Fig. 2. Shoulder approximation.