Scripta Scientifica Medicinae Dentalis, 2023;9(1):55-61
DOI: 10.14748/ssmd.v9i1.9035

ISSN 2367-7236 (Print)
ISSN 2367-7244 (Online)

CASE REPORTS

# DENTAL TREATMENT OF PATIENTS WITH CLEIDOCRANIAL DYSPLASIA: TWO CASE REPORTS

**Elitsa Dzhongova, Ivan Gonkov**

*Department of Oral and Maxillofacial Surgery, Faculty of Dental Medicine, Medical University of Varna*

## ABSTRACT

**INTRODUCTION: Cleidocranial dysplasia (CCD) is an autosomal dominant disorder, also referred to as cleidocranial dysostosis. The diagnosis is made on the basis of clinical and radiological findings and is confirmed through a genetic analysis. Cleidocranial dysplasia manifestations in the skeletal system include aplasia or hypoplasia of the clavicles, leading to shoulder joint hypermobility, a cone-shaped chest with short ribs, a wide skull with prominent frontal bone, etc. The dental status of CCD patients is commonly marked by a persistent primary dentition, delayed eruption of permanent teeth, supernumerary teeth, an incorrect proportion between the two jaws, impaired masticatory function and aesthetics.**

**AIM: The aim of the present study is to show the clinical and radiologic findings of two CCD patients and describe the multidisciplinary approach necessary for their treatment. Early diagnosis and awareness of the condition help to determine the optimal treatment and duration as well as to improve patients' quality of life.**

**CASE REPORT: The subject of this clinical study are two closely related patients who sought professional dental help due to persistent primary teeth, delayed eruption of permanent dentition beyond the mean eruption age and resultant occlusal and aesthetics problems.**

**RESULTS: Correction of the dental status in CCD patients is a long process. Timely combination of orthodontic and surgical treatments is crucial for optimal and long-lasting results.**

**CONCLUSION: Dental treatment of CCD patients is a complex endeavor. The type of approach in the orthodontic, surgical and prosthetic treatment is largely determined by the patients' age, their general health as well as their intraoral status (present deciduous teeth, existing permanent teeth and occlusal contacts).**

**Keywords:** *supernumerary teeth, hypoplasia, delayed eruption*

## INTRODUCTION

Cleidocranial dysplasia (CCD) is a rare generalized disorder affecting the skeletal and teeth bones. It was first described by Paul Sainton and Pierre Marie as early as 1898. The diagnosis is made on the basis of clinical and radiological findings and is confirmed through a genetic analysis. Cleidocranial dysplasia manifestations in the skeletal system include aplasia or hypoplasia of the clavicles, leading to shoulder joint hypermobility, and a cone-shaped chest with short ribs (1,2). Another characteristic feature is the delayed closure of sutures and fontanels with the presence of additional small bones (wormian bones),

**Address for correspondence:**
*Elitsa Dzhongova*
*Faculty of Medicine*
*Medical University of Varna*
*84 Tzar Osvoboditel Blvd*
*9002 Varna*
*e-mail: elica.djongova@mu-varna.bg*

**Received**: April 27, 2023
**Accepted**: June 13, 2023

frontal bossing, hypertelorism, and an enlarged nose base with a depressed bridge (3). Cleidocranial dysplasia patients usually have good general health and no mental abnormalities (4).

Cleidocranial dysplasia patients' dental status typically presents persistent deciduous teeth, delayed eruption of the permanent dentition and many impacted supernumerary teeth. Other changes in the masticatory apparatus may include incomplete development of the maxilla, forward and upward projection of the mandible as well as malocclusion. Despite the abnormalities, patients, especially those at a younger age, do not complain of pain, swelling or difficulties in the masticatory functions. Therefore, in most cases, patients seek professional dental help much later in life when the impairments in the masticatory apparatus are the result of delayed exfoliation of deciduous teeth and the eruption of permanent ones.

## AIM

The aim of the present study is to show the clinical and radiologic findings of two CCD patients and describe the multidisciplinary approach necessary for their treatment. Early diagnosis and awareness of the condition help to determine the optimal treatment and duration as well as to improve the patients' quality of life.

For the correction of masticatory dysfunctions associated with CCD, several treatment approaches have been described in dental literature, involving surgical procedures along with orthodontic treatment, implant treatment or the placement of removable/fixed dental prostheses. Modern procedures include extraction of impacted supernumerary teeth, exposure of retained permanent teeth and their subsequent alignment by an orthodontist. In the past, restoration of the masticatory apparatus was carried out through surgery followed by orthodontic treatment with fixed/removable prostheses (5,6). The particular treatment approach is mainly determined by the patient's age as well as their general and local health condition.

## CASE REPORTS

The present case report describes two male patients with a close family relationship (brothers) who sought dental assistance at the University Medical and Dental Center, at the Faculty of Dental Medicine in 2017. Both patients had complaints as a result of persistent primary teeth, delayed eruption of the permanent teeth, and bite dysfunctions. After the parents signed an informed consent form, extra- and in-

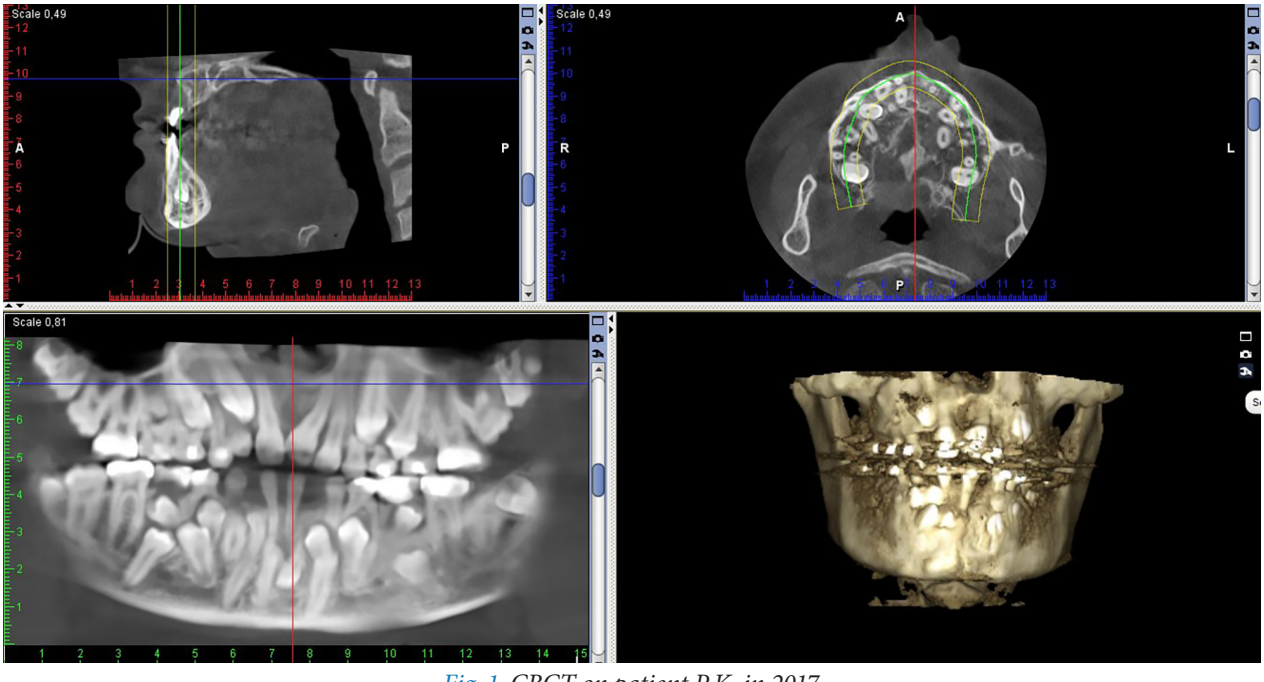


*Fig. 1. CBCT on patient P.K. in 2017.*

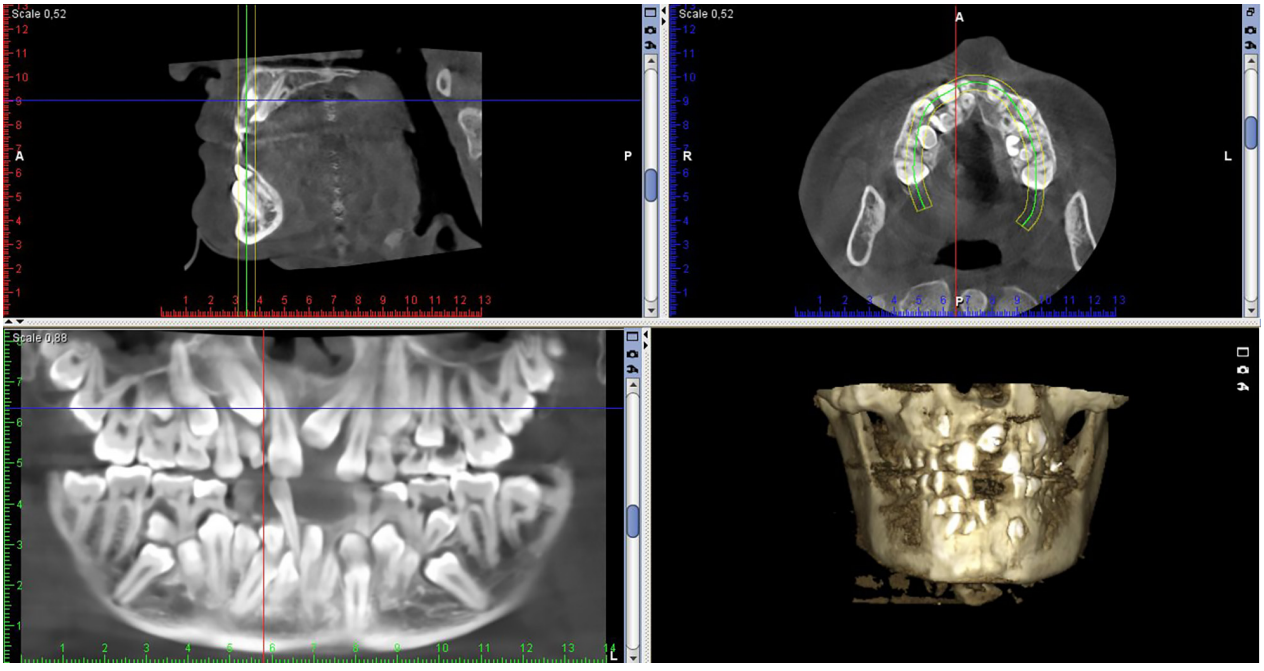


*Fig. 2. CBCT on patient P.K. in 2022.*

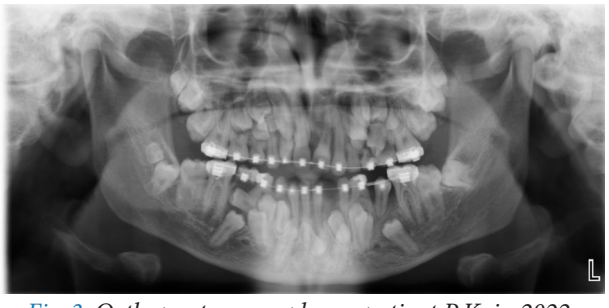

*Fig. 3. Orthopantomography on patient P.K. in 2022.*

traoral examinations were performed. The extraoral examination of the head and neck revealed a prominent frontal bone, enlarged nasal base with a slightly depressed bridge. The intraoral examination of both patients revealed the presence of many deciduous teeth still in the mouth, their replacement being long overdue, a narrow high-arched palate, anterior open bite, incorrect occlusal relationships, and mandibular prognathism. The orthopantomogram (OPG) showed many impacted permanent teeth in either jaws, and their eruption time did not correspond to the patients' chronological age. Following an orthodontic examination, it was agreed to conduct a combined treatment, including the placement of brackets on both jaws, extraction of persistent deciduous and supernumerary teeth under local anesthesia, gradual exposure, positioning and orthodontic traction of impacted permanent teeth. Cone-beam computed tomography (CBCT) scans of both patients proved crucial in deciding on the sequence of the extractions and the degree of impaction.

The orthodontic movement and alignment of the dental rows are still on-going for both patients.

## DISCUSSION

Cleidocranial dysplasia is a rare congenital skeletal disease characterized by triggering aplasia or hypoplasia of the clavicles, delayed exfoliation of primary teeth and delayed eruption of the permanent dentition. The disease poses many challenges as to its diagnosis and relevant treatment approaches. Its diagnosis can be difficult due to the variability of the clinical and radiological findings associated with the syndrome. The most common CCD manifestations may include relatively short stature, narrow chest, shoulder joint hypermobility as a result of underdeveloped clavicles, dysplasia of the distal phalanges, and/or pelvic bones deformities. The clinical forms of CCD are highly variable and inconsistent and not all symptoms are manifested (7). With regard to teeth/jaw abnormalities, retained deciduous teeth, delayed eruption of permanent teeth, presence of supernumerary teeth, and occlusal disturbances vary among patients (8).

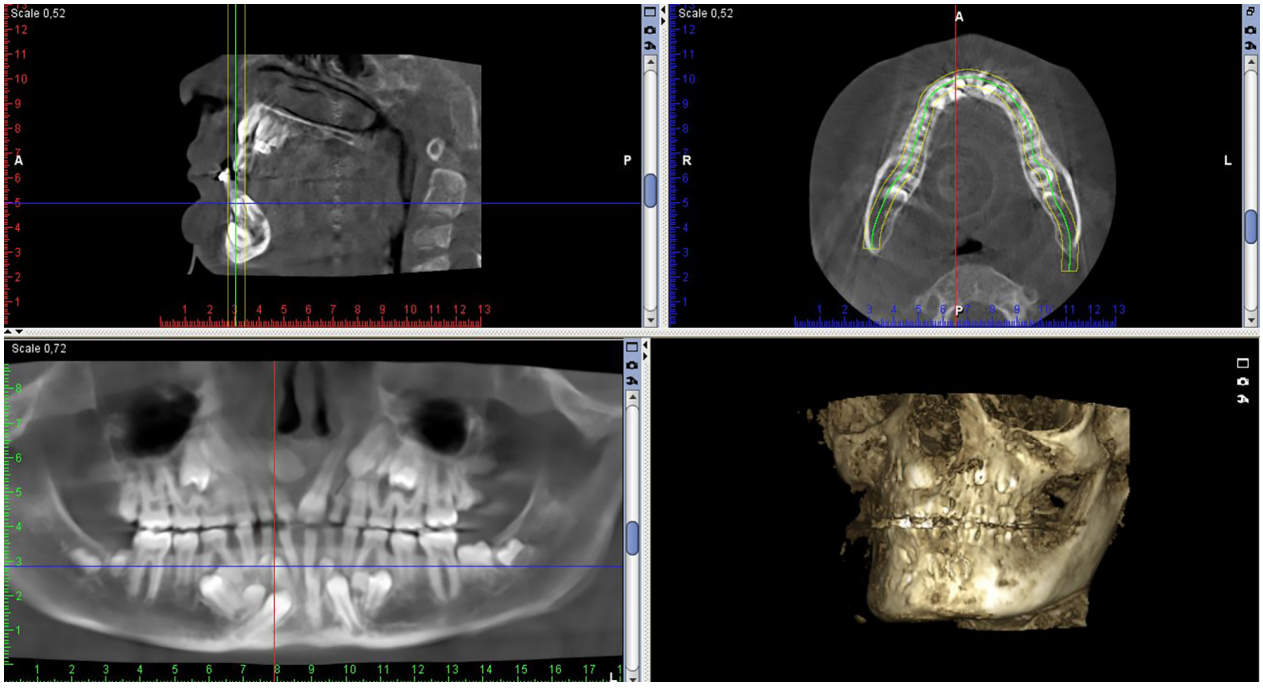


*Fig. 4. CBCT on patient A.K. in 2017.*

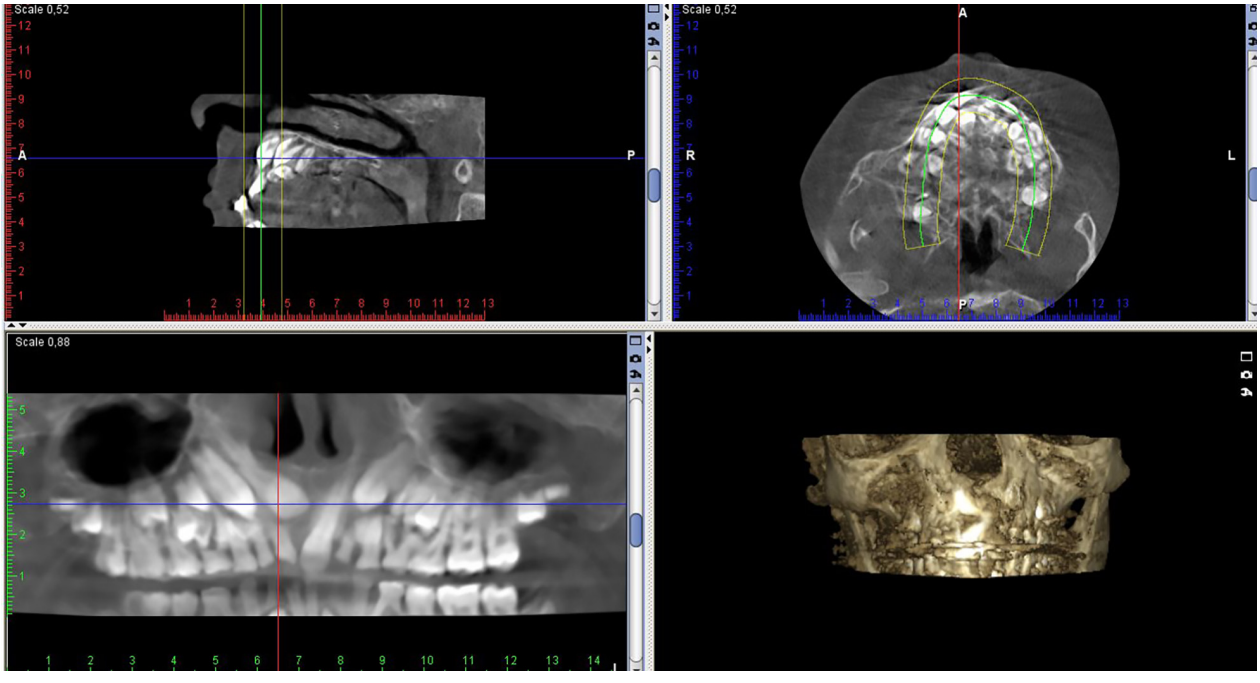


*Fig. 5. CBCT on patient A.K. in 2022.*

It has been hypothesized that supernumerary teeth in CCD are the result of unresorbed fragments of dental lamina during the odontogenesis (9). However, their accumulation in the jawbones leads to delayed eruption of the permanent teeth or alters their eruption path in the dental rows.

Cleidocranial dysplasia is a genetic disorder caused by mutation in the RUNX2 gene on chromosome 6p21. This gene guides osteoblastic differentia-

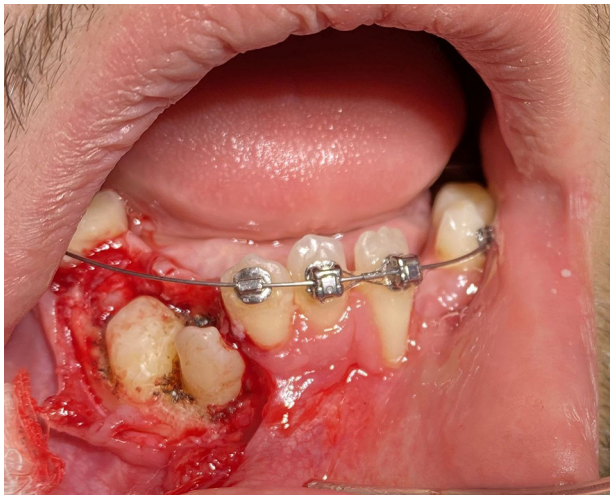

*Fig. 6. Exposure of impacted teeth 43 and 44—patient P.K. in 2022.*

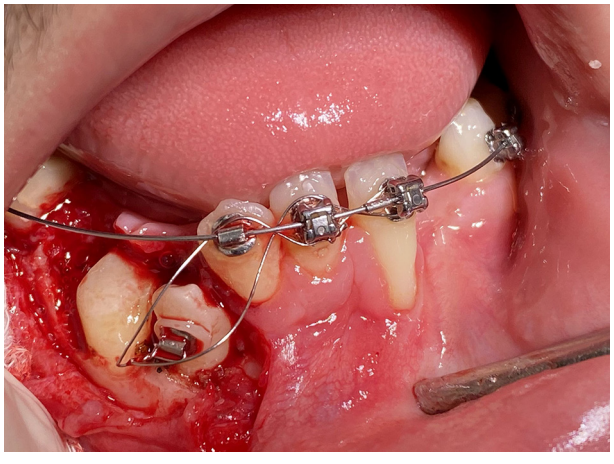

*Fig. 7. Placement of brackets and orthodontic traction—patient P.K. in 2022.*

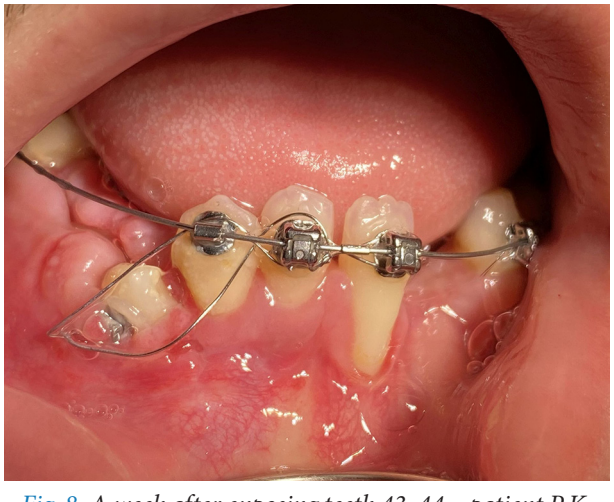

*Fig. 8. A week after exposing teeth 43, 44—patient P.K.*

tion and chondrocytes maturation, triggering bone formation processes. It also determines the induction of CBFA1 protein in new bone formation (10). The RUNX2 gene plays a crucial role in signaling the epithelial-mesenchymal interactions during tooth bud morphogenesis and histodifferentiation of the epithelial enamel organ (11). The prevalence of this mutation is 1:100,000 births with no predilection for gender (12,13).

Diagnosing CCD by clinical and radiological findings alone is difficult since other diseases present matching symptoms. Differential diagnosis should be made with Crane-Heise syndrome, mandibuloacral dysplasia, pycnodysostosis, Yunis-Varon syndrome, etc.

Reports from histopathological analyses differ among researchers. One histological examination found out distorted dentin tubules, prominent interglobular dentin and acellular cementum, with absence of cellular cementum in the apical region (14). Yet, another study reported no changes in the structure and arrangement of dentinal tubules (15).

For the treatment of teeth/jaw deformities in CCD, no universal approach and specific treatment duration exist. The approach may involve orthodontic, surgical, or prosthetic treatments. It is yet debatable as to which approach yields best results. In the past, restoration of the masticatory apparatus was mainly achieved through prosthetic treatment (removable and/or fixed dental prostheses) with relatively rapid restoration of the aesthetics and functional disorders (6). However, it should be noted that prosthetic rehabilitation is not always an option as this syndrome is characterized by completely retained deciduous and permanent teeth and incomplete eruption and short clinical crowns do not provide stable support for either removable or fixed prosthetic devices. Furthermore, restoration of the masticatory apparatus via removable prostheses is not very appealing to younger patients. However, this treatment approach is widely implemented and its proponents maintain that the application of orthodontic and surgical treatments is time-demanding, requiring many expensive and complex surgeries (8,16). Another study in dental literature describes a non-invasive approach in the correction of the dental status: a composite build-up is carried out to restore

the shape and size of teeth and improve occlusion. However, conservative treatment is temporary and additional surgical treatment is further required (17).

In addition to the exposure and extraction of supernumerary teeth in the orthodontic phase, CCD surgical treatment may also include restoration of the masticatory apparatus with dental implants (18, 19). Genetically, it affects not only the dentition but also the jaw bones. The biological properties of bone tissue and particularly its ability for osseointegration in CCD cases are still being investigated (20).

A number of strategies have been described as to the appropriate commencement time of dental treatment based on the patient's age (21, 22, 23, 24). According to Jensen and Kreiborg, the optimal time period to start CCD dental treatment is 5–7 years, as it should be possible to diagnose supernumerary incisors at that age (6). Beyond that age, the gradual formation of permanent tooth roots and the delayed resorption of the primary teeth would cause delayed eruption or impaction of the permanent teeth and therefore complicate the treatment process (25). Early diagnosis enables immediate treatment of any complications associated with this syndrome. In most cases, however, patients seek professional help much later in life, which necessitates an individual approach for each clinical case.

A combined surgical and orthodontic approach stands out as the most effective in overcoming CCD teeth/jaw abnormalities. The four most successful and popular surgical and orthodontic treatments are the Toronto-Melbourne, Belfast-Hamburg, Jerusalem, and Bronx approaches. Although each of them shows different indications and results, they all obey the same sequence of surgical and orthodontic stages, with little difference in the commencement time (17,22).

In the case reported in the present study, a gradual removal of deciduous teeth was undertaken and the permanent teeth were exposed by flap retraction. A bracket was then bonded to the permanent teeth to achieve prompt traction and arrangement in the dental row. The extraction of supernumerary teeth was combined with the exposure of the impacted ones based on a 3D imaging, which assessed the bone amount and density, the direction and axes of teeth as well as the root development stage. The orthodontic treatment corrected the height and axis of teeth, their arrangement and occlusal relationships. The patients' speech and masticatory functions as well as the aesthetics improved. The combined surgical and orthodontic treatment of CCD teeth/jaw deformities leads to good results while conserving the natural teeth. The main disadvantage proves to be the duration of treatment, which demands great effort and patience on the part of the patient and is quite challenging for the dental specialist.

## RESULTS

Correction of the dental status in CCD patients is a long process. The best and longest-lasting results are obtained through timely combination of orthodontic and surgical treatments. The two treatment approaches enable teeth alignment, occlusal corrections and restoration of the impaired masticatory function. The combined treatment also allows for optimal symmetry and aesthetics of hard and soft tissues.

## CONCLUSION

Dental treatment of CCD patients is a complex endeavor. The choice of an optimal treatment approach, involving orthodontic, surgical, and prosthetic treatments, is largely determined by the patients' age, their general health as well as their intraoral status (present deciduous teeth, existing permanent teeth and occlusal contacts).

## REFERENCES


1. Carvalho CCS, Heimlich FV, Freire NA, Ramos MEB, Rosemiro MM, Israel M. Cleidocranial dysplasia: 2 case reports. Oral Surg oral Med oral Pathol Oral Radiol. 2020;129:e-41. doi: 10.1016/j.oooo.2019.06.131.
2. Jensen B. Somatic development in cleidocranial dysostosis. Am J Med Genet. 1990;35(1):69-74. doi: 10.1002/ajmg.1320350113.
3. Butterworth C. Cleidocranial dysplasia: modern concepts of treatment and report of an orthodontic resistant case requiring a restorative solution. Dent Update. 1999;26(10):458-62. doi: 10.12968/denu.1999.26.10.458.
4. Cooper SC, Flaitz CM, Johnston DA, Lee B, Hecht JT. A natural history of cleidocranial dysplasia. Am J Med Genet. 2001;104(1):1-6. doi: 10.1002/ajmg.10024.

**5.** D'Alessandro G, Tagariello T, Piana G. Craniofacial changes and treatment of the stomatognathic system in subjects with cleidocranial dysplasia. Eur J Paediatr Dent. 2010;11(1):39-43.

**6.** Jensen B.L., Kreiborg S. Dental treatment strategies in cleidocranial dysplasia. Br Dent J. 1992;172(6):243-7. doi: 10.1038/sj.bdj.4807836.

**7.** Haese K, Le Toux G. Surgical strategy for patient with late-diagnosed form of cleidocranial dysplasia: three cases. J Oral Med Oral Surg, 2020;26:4. doi: 10.1051/mbcb/2019033.

**8.** Mabrouk Y, Ammar S, Labidi A, Mansour L, Ghoul S. Dental prosthetic treatment in cleidocranial dysplasia: case report and literature review. Case Rep Dent. 2020;2020:8910798. doi: 10.1155/2020/8910798.

**9.** Jensen BL, Kreiborg S. Development of the dentition in cleidocranial dysplasia. J Oral Pathol Med. 1990;19(2):89-93. doi: 10.1111/j.1600-0714.1990.tb00803.x.

**10.** Leite PB, Carvalho CCS, Heimlich FV, Freire NA, Ramos MEB, Israel MS. The role of dentistry in the diagnosis of cleidocranial dysplasia: reports of two cases. J Oral Diag. 2020;05:e20200019. doi: 10.5935/2525-5711.20200019.

**11.** Roberts T, Stephen L, Beighton P. Cleidocranial dysplasia: a review of the dental, histological, and practical implications with an overview of the South African experience. Oral Surg Oral Med Oral Pathol Oral Radiol. 2013;115(1):46-55. doi: 10.1016/j.oooo.2012.07.435.

**12.** Arun SP, Simon SS, Karthik AK, Chacko RK, Salitha S. A review of clinical and radiological features of cleidocranial dysplasia with a report of two cases and dental treatment protocol. J Pharm Bioall Sci. 2015;7(Supp 2):428-32. doi: 10.4103/0975-7406.163490.

**13.** Gupta S, Gupta S, Singh K, Arora P, Goel S. Cleidocranial dysplasia: spectrum of clinical and radiological findings in seven cases. J Oral Maxillofac Radiol. 2015;3(2)58-62. doi:10.4103/2321-3841.157525.

**14.** Vij R, Batra P, Vij H. Cleidocranial dysplasia: complete clinical, radiological and histological profiles. BMJ Case Rep. 2013;2013:bcr2013009015. doi: 10.1136/bcr-2013-009015.

**15.** Lukimnaa PL, Jensen BL, Thesleff I, Andreasen JO, Kreisborg S. Histological observations of teeth and peridental tissues in cleidocranial dysplasia imply increased activity of odontogenic epithelium and abnormal bone remodeling. J Craniofac Genet Dev Biol. 1995;15(4):212-21.

**16.** Daskalogiannakis J, Piedale L, Lindholm TC, Såndor GKB, Carmichael RP. Cleidocranial dysplasia: Two generations of management. J Can Dent Assoc. 2006; 72(4):337-42.

**17.** Ayub NFAM, Hamzah SH, Hussein AS, Rajali A, Ahmad MS. A case report of cleidocranial dysplasia: A noninvasive approach. Spec Care Dentist. 2021;41(1):111-7. doi: 10.1111/scd.12532.

**18.** Petropoulos VC, Balshi TJ, Blshi SF , Wolfinger GT. Treatment of patient with cleidocranial dysplasia using ossteointegrated implants: a patient report. Int J Oral Maxillofac Implants. 2004;19(2):282-7.

**19.** Schnutenhaus S, Götz W, Luthardt RG. Prosthetic rehabilitation of a patient with cleidocranial dysplasia using dental implants – a clinical report. Int J Implant Dent. 2021;7(1):5. doi: 10.1186/s40729-020-00287-7

**20.** Schnutenhaus S, Luthardt RG, Rudolph H, Götz W. Histological examination and clinical evaluation of the jawbone of on adult patient with cleidocranial dysplasia: a case report. Int J Clin Pathol 2015;8(7):8521-31.

**21.** Angle AD, Rebellato J. Dental team management for a patient with cleidocranial dysostosis. Am J Orthod Dentofacial Orthop. 2005;128(1):110-7. doi: 10.1016/j.ajodo.2004.05.019.

**22.** Becker A, Lustman J, Shteyer A. Cleidocranial dysplasia: Part 1- General principles of the orthodontic and surgical treatment modality. Am J Orthod Dentofacial Orthop. 1997;111(1):28-33. doi: 10.1016/s0889-5406(97)70298-1.

**23.** Becker A, Sheteyer A, Bimstein E, Lustman J. Cleidocranial dysplasia: Part 2 - Treatment protocol for the orthodontic and surgical modality. Am J Orthod Dentofacial Orthop 1997;111(1):173-83. doi: 10.1016/s0889-5406(97)70213-0.

**24.** Berg RW, Kurtz KS, Watanabe I, Lambrakos A. Interim prosthetic phase of multidisciplinary management of cleidocranial dysplasia: „the Bronx Approach". J Proshtodont 2011;20:S20-5. doi: 10.1111/j.1532-849X.2011.00786.x.

**25.** Chang H, Wei J, Wang Y, Jia J, Gao X, Li X, Feng H. Restorative treatment strategies for patient with cleidocranial dysplasia. Acta Odontologica Scandinavica 2015;73(6):447-53. doi: 10.3109/00016357.2014.983541.