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The role of the pediatric dentist in managing the oral health needs of patients with Cleidocranial Dysplasia

The role of pediatric dentists in managing the oral needs of patients with Cleidocranial Dysplasia

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SUMMARY

Introduction: Cleidocranial dysplasia (CCD) is a rare congenital skeletal condition, inherited in an autosomal dominant pattern, with wide clinical variability. The prevalence is estimated at 1:1,000,000, with no gender predilection and no impact on cognitive abilities. Objective: This study aims to analyze the role of the pediatric dentist in the management of CCD in children, a genetic condition that affects craniofacial formation and causes functional and aesthetic alterations. Materials and Methods: This is a narrative review based on articles available in the PubMed database. Studies on CCD in pediatric patients were included, focusing on etiopathogenesis and clinical reports relevant to dental management. There was no time limitation. Literature Review: The diagnosis of CCD occurs through the analysis of clinical and radiographic signs, generally visible since childhood.

Prolonged retention of deciduous teeth, delayed eruption of permanent teeth, and supernumerary teeth are common. The pediatric dentist is often the first to identify these signs, facilitating early diagnosis and effective therapeutic strategies. Final Considerations: The pediatric dentist plays a central role in the diagnosis and monitoring of CCD (Central Density of the Teeth), working in a multidisciplinary way to promote oral health and quality of life. Although there have been advances, there are still gaps in the literature, reinforcing the need for more research and specific clinical protocols for the care of these patients.

Keywords: Cleidocranial Dysplasia, Pediatric Dentistry, Dental Anomalies, Dental Management, Child Oral Health.

ABSTRACT

Introduction: Cleidocranial dysplasia (CCD) is a rare congenital skeletal condition with autosomal dominant inheritance and wide clinical variability. The prevalence is estimated at 1:1,000,000, with no gender predilection and no impact on cognitive abilities. Objective: This study aims to analyze the role of pediatric dentists in the management of CCD in children, a genetic condition that affects craniodentofacial formation and causes functional and aesthetic changes. Materials and Methods: This is a narrative review based on articles available in the PubMed database. Studies on CCD in pediatric patients were included, focusing on etiopa-thogenesis and clinical reports relevant to dental management. There was no temporal delimitation. Literature Review: The diagnosis of CCD is made through the analysis of clinical and radiographic signs, usually visible since childhood. Prolonged retention of deciduous teeth, delayed eruption of permanent teeth, and supernumerary teeth are common. Pediatric dentists are often the first to identify such signs, favoring early diagnosis and effective therapeutic strategies. Final Considerations: Pediatric dentists play a central role in the diagnosis and monitoring of CHD, working in a multidisciplinary manner to promote oral health and quality of life. Although there have been advances, there are still gaps in the literature, reinforcing the need for more research and specific clinical protocols for the care of these patients.

Keywords: Cleidocranial Dysplasia, Pediatric Dentistry, Dental Anomalies, Dental Management, Children's Oral Health.

1. INTRODUCTION

Cleidocranial dysplasia (CCD), first reported in 1766 by Monrad[1], is a rare congenital skeletal disease, inherited in an autosomal dominant pattern[2], with wide variability of clinical expression[1]. Its prevalence rate is approximately 1:1,000,000[3], it does not interfere with

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cognitive abilities of the individual and shows no preference for genderÿ

The diagnosis of DCC is made through the detection of clinical and radiographic signs. characteristic of the condition, noticeable since childhood, with emphasis on prolonged retention. of deciduous teeth, late eruption of permanent teeth, and the presence of supernumerary teeth2 . Given these factors, the pediatric dentist tends to be the first professional in the area. from the health department to having contact with the patient, favoring early diagnosis and allowing a most effective therapeutic intervention.

It should be added that the main cause of DCC is a genetic mutation in the RUNX2 gene, which, Consequently, it undergoes alteration or loss of its functions related to differentiation. osteoblasts, dental lamina regression, and the contribution to osteoclastogenesis in the dental follicle and periodontal ligament2 .

When addressing the changes in dental development in patients with DCC, the challenges associated functional and aesthetic aspects, clinical management strategies, and therapeutic alternatives. available for this condition, this review aims to contribute to the expansion of scientific knowledge about the specificities of dental care in children with DCC, promoting a more humanized, efficient, and integrated approach.

2. THEORETICAL FRAMEWORK

2.1 Cleidocranial Dysplasia: Concept and Characteristics Definition and etiology

Cleidocranial dysplasia (ICD-10 Q74.0), first reported in 1766 by Monrad1 , It is a rare congenital skeletal disease, inherited in an autosomal dominant pattern. , with ample variability in expression1 , its prevalence rate is approximately 1:1,000,003 , without gender preference4 .

The genetic disorder was mapped to chromosome 6p21 in 1995, when it was discovered. a mutation in the RUNX2 gene, which results in an alteration in its transcription factor, the protein CBFA1, active in the differentiation of osteoblast cells for the regression of the dental lamina, in osteoclastogenesis in the dental follicle, periodontal ligament, and bone remodeling of the craniofacial complex2 .

Furthermore, it is important to emphasize that DCC does not affect the conscious intelligence of individual3 .

General radiographic and clinical manifestations

The diagnosis of cleidocranial dysplasia (CCD) is made based on the detection of signs. clinical and radiographic features characteristic of the condition, among which the following stand out in the clinical aspects General characteristics: narrow chest, valgus knees and valgus thighs³, short stature, prominent forehead, hypoplasia

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mid-facial, hypertelorism, retrognathic maxilla, prognathic mandibleÿ, brachydactyly and scoliosisÿ; in
Oral clinical aspects: prolonged retention of deciduous teeth, delayed tooth eruption.
permanent teeth, presence of supernumerary teeth, impacted teeth, cysts, odontomas[2], palate
ogival (narrow and deep), maxillary atresia, and absence of cleft palateÿ; and in the radiographic findings:
Hypoplastic or aplastic clavicles, delayed closure of the anterior fontanelle, presence of bones.
Wormian bones exhibit late ossification of the pubic and carpal bones, as well as the absence of fusion of the rami.
ischiopubic musclesÿ, ÿ.

Genetic mapping also emerges as a reliable option to support diagnosis.
DCC1 .

Physiopathological, genetic, and phenotypic aspects

The primary cause of cleidocranial dysplasia (CCD) is a genetic mutation in the RUNX2 gene.
which, consequently, suffers alteration or loss of its functions. This affects 60% to 70% of patients with DCC, while
the remaining percentage (30% to 40%) is attributed to genetic inheritance .
Delving deeper into the genetic aspect, it was found that the mutation can affect any of the eight
exons of the RUNX2 gene, since each exon is located in one of the gene's three regions: QA,
RHD and PST, responsible for specific amino acid sequences, each affected part will consequently have a different
phenotypic expression within what is expected for dysplasia6 .

Given this, the understanding of the genotypic spectrum of DCC has expanded.
considerably, leading to the conclusion that there is great phenotypic variability among patients with CCD, including
in the intrafamilial context.6 .

2.2 Dental Manifestations of Cleidocranial Dysplasia: Alterations in dental development

The replacement of deciduous teeth with permanent teeth is common among mammals, being
called diphyodonty, which is strictly controlled by genetic factors. Because of this,
Humans have incisors, canines, and premolars in their deciduous dentition, with the addition of molars in their
permanent dentition .

The formation of tooth germs, starting from the primary dental lamina, begins during the
Intrauterine life, around the 6th week. As a result, it takes 3.5 years for the formation.
Complete shedding of the deciduous dentition. As for the permanent tooth germs, they begin to form.
starting from the successor dental lamina, between the 13th and 17th week of intrauterine life. Thus, the formation
of the permanent dentition takes about 14 years2 .

The process of tooth eruption, extensively studied by Marks (1996), is divided into five
stages:

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Pre-eruptive: stage that extends from the beginning of tooth germ formation until the Crown formation. When the root begins to form, the dental follicle recruits activity. Osteoclastic resorption of the overlying bone and deciduous root allows the tooth to move apically .

Intraosseous: the duration of this stage depends on the type of tooth in question, as its Length directly affects the path and speed of the eruption. This stage corresponds to The distance traveled by the tooth from its original formation site to the top of the alveolar process. The main determining factor in this process is genetics. In the context of DCC, this is understood to mean that... The reason for the delay in the eruption of permanent teeth, since this disease directly impacts the Bone remodeling due to reduced osteoclastic activity. In addition, local factors, such as supernumerary teeth, odontomas, and cysts, can also influence the delay of this phase .

Penetration into the mucosa: the relevant factor at this stage is the formation of the junctional epithelium, which is continuously renewed on the tooth surface during eruption.8

Pre-occlusal: at this stage, the root grows and bone formation occurs at the base of the crypt and/or in the intraradicular septa8 .

Post-occlusal: the tooth erupts completely, promoting bone formation. circumferential, the consolidation of the lamina dura, the maturation of the periodontal ligament fibers, and the establishment of the root8

Abnormalities in dental morphology

Dental anomalies are frequent in patients with DCC and, in addition to clinical examination, it is possible to map them using intraoral radiographs, panoramic radiographs, and cephalometric films.9 The most commonly found dental irregularities include multiple teeth. supernumerary5 , malformed roots with absence of cellular cementum, maxillary bone excessively dense and defective resorption of bone and roots of deciduous teeth2 .

Given this, Jensen and Kreiborg (2018) collected longitudinal data from a database of studies with clinical reports of patients with CCD, analyzing works published in 1980. , 19909 and the most recent one from 20182The researchers found that, due to a mutation in the gene In RUNX2, the remnants of the dental lamina do not regress within the expected time after the formation of the crown, which can be reactivated and give rise to supernumerary teeth.

On the other hand, Stonehouse-Smith (2024) suggests that high levels of signaling pathway Wnt in the mesenchymal component of the developing dentition may be directly related to the occurrence of supernumerary teeth in individuals with DCC11 .

Furthermore, the researchers deduce that, due to the presence of supernumerary teeth On the path of permanent teeth, these need to move to another position, leading them to...

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to occupy ectopic sites in the mouths of patients with DCC2 .

Functional and aesthetic implications

From this perspective, it becomes evident that unwanted functional and aesthetic changes These affect patients with DCC . Therefore, the therapeutic approach should focus on both masticatory function and the patient's personal aesthetic satisfaction.12

Treatment approaches range from less invasive ones, such as restorative interventions, in-office whitening13 and orthodontic traction14, to more aggressive ones, such as extraction of unerupted teeth, surgical exposure of these teeth, tooth removal supernumerary14 and insertion of implant-supported fixed prostheses12 .

2.3 EPIDEMIOLOGICAL SCENARIO OF CLEIDOCRANIAL DYSPLASIA IN SOUTH AMERICA

Documented cases in South American countries

A group of researchers from the Faculty of Medicine in Cartagena, Colombia15, gathered Data from 72 cases of patients with cleidocranial dysplasia in South American countries. The distribution was as follows: Brazil, with 54 cases; Colombia, with 9 cases; Chile, with 5 cases; and Argentina and Venezuela, with 2 cases each.

The distribution by sex and age group of the patients included 36 women and 36 men, with ages ranging from babies between 0 and 5 months old to seniors over 60 years old15 .

Prevalence of craniofacial and oral clinical manifestations

It was found, through comparison of the data, that clavicular dysplasia is the the most prevalent feature in patients with CCD, corresponding to 98.6% of cases, followed by the presence of open fontanelles in 92.6%15 .

Regarding oral clinical manifestations, supernumerary teeth accounted for 88% of cases, followed by delayed eruption of permanent teeth, with 78.8%15 .

In parallel, an analysis of the family history of patients with dysplasia was carried out. cleidocranial dysfunction (CCD), with the following findings: in the first group, consisting of 24 probands, Eight reported no family history of the disease, ten had no concrete information, and six They reported a positive family history for CCD. In the second group, composed of 48 In cases with multiple presentations, six were considered sporadic cases within a family context. while 42 showed a clear pattern of inheritance1ÿ.

Additionally, molecular analyses were performed on only 14 of the 72 cases evaluated. of which 11 presented mutations in the RUNX2 gene. It was observed that the mutations located in Runt domain was associated with an increased incidence of impacted permanent teeth and

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supernumerary individuals, compared to individuals without alterations in this gene domain1ÿ.

Challenges in diagnosis

Of the total cases analyzed, 74.6% were diagnosed before the age of 25 through a Consultation with a dentist due to delayed exfoliation of deciduous teeth, delayed eruption of... permanent teeth and the presence of supernumerary teeth, while 25.4% were diagnosed in adulthood or old age, due to chewing problems, speech impediments, missing teeth, impacted teeth, and unsatisfactory aesthetics15, 12 .

Among the limitations observed, the low frequency of molecular studies stands out, which They could broaden the understanding of genetic patterns among patients with cleidocranial dysplasia. (DCC), contributing to the development of more specific clinical protocols for the condition. Furthermore, most of the cases analyzed lacked detailed descriptions and follow-up. longitudinal, which restricts a more in-depth assessment of the clinical characteristics of dysplasia1ÿ.

2.4 The Pediatric Dentist as a Gateway to Therapeutic Success: Influence of Clinical Management

A pediatric dentist is a trained and highly compassionate professional equipped to deal with the most advanced patients. various childhood conditioning factors. Its role is extremely relevant, given that anxiety Pediatric dental issues are a frequent concern and sometimes even an impediment to clinical treatment, affecting 13.3% to 36.5% of children in various countries16 .

In this way, it becomes clear that the clinical approach employed by dental surgeons Pediatric care is the primary tool for building trust with both the patient and their caregivers. being, consequently, essential for the success of the therapeutic treatment17 .

Importance of early diagnosis

Therefore, pediatric dentistry is, in most cases, the first specialty to address the patient with DCC17. Given the complexity of the treatment, early diagnosis of the disease is This is crucial because it allows for the availability of more therapeutic alternatives.

An example of this is the presence of supernumerary teeth, which often become visible. Radiographically, on average, after 6 years of age. Therefore, if the follow-up is If done correctly, it will be possible to map the location of these teeth and remove them promptly. together with the overlying bone and deciduous teeth, favoring the spontaneous eruption of permanent teeth.

The application of this approach, combined with a shorter orthodontic treatment period,

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It demonstrated satisfactory results, according to the studies by Jensen and Kreiborg2 .

Another early therapeutic option in children is the use of orthopedic and orthodontic oral appliances, since the bony bases are not yet fully formed in this age group14 .

Thus, alterations such as crossbite, dental crowding, and maxillary atresia They can be corrected with less treatment time and often avoid the need for more complex interventions, such as surgery14 .

Multidisciplinary referral and intervention

Given the wide individual phenotypic variability of patients with dysplasia In cleidocranial lesions, the therapeutic approach to oral clinical manifestations must be holistic, requiring communication and cooperation between the professionals involved such as pediatric dentists, orthodontists, Prosthodontists, implantologists, oral and maxillofacial surgeons, and dentists specializing in care for patients with disabilities, so that integrated treatment and clinical success are possible17, 13, 12 .

3. MATERIALS AND METHODS

This work is based on a literature review, as defined by Pautasso (2013). as a highly relevant tool in the scientific community, as it allows the compilation of general and up-to-date information, serving as a consistent database for the in-depth study of a particular topic.

The present research aims to conduct a literature review on the performance This study investigates the role of pediatric dentists in the care of children with cleidocranial dysplasia. exploratory in nature, using secondary data obtained through scientific articles. Published in the PubMed database, with no defined time limit.

The keywords used in the search were: *cleidocranial dysplasia, supernumerary teeth. delayed eruption, RUNX2, skeletal disorders* and *pediatric dentistry.*

The selection of articles for this literature review followed inclusion and exclusion criteria. previously established criteria were included. Studies that addressed cleidocranial dysplasia with Focus on pediatric patients, discussing genetic aspects of the condition, case reports. that contributed to the understanding of the clinical management of children with CCD, as well as articles that presented a universal and up-to-date view of the disease, relevant to the foundation Theoretical aspects of this work were excluded. Articles that exclusively addressed cases in adults were also excluded. studies without full-text access and publications that addressed the topic in a way superficial, without clinical depth.

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4. RESULTS AND DISCUSSION

Knowledge regarding pathologies involving the development of structures
Craniodentofacial disorders, such as cleidocranial dysplasia (CCD), are of extreme relevance to
dentists, especially pediatric dentists, as they are the professionals who most often...
They are dedicated to the study of the root, dental, and bone development and maturation phase.

In the discussion about the etiology of supernumerary teeth in DCC, different hypotheses have been put forward.
have been proposed over time. Kreiborg and Jensen (2018) suggest that remnants of the blade
Dental caries that do not regress properly after crown formation can be reactivated, leading to...
to the formation of these additional teeth. However, more recent studies, such as that of Stonehouse-Smith
(2024) introduce a new perspective by relating this phenomenon to the exacerbated activation of the pathway.
of Wnt signaling in the developing dental mesenchyme. This molecular hypothesis does not
It not only complements the previous morphological understanding, but also points to the complexity.
of the mechanisms involved, reinforcing the need for interdisciplinary approaches in management.
DCC clinician.

In view of this, the authors Kreiborg and Jensen (2018) highlight that, around the age of six
As we age, supernumerary teeth begin to form in the bone. Therefore, if the patient has a
With consistent monitoring, the planning for the removal of these teeth will be more effective, reducing the risk of complications.
the negative repercussions that could occur in the future.

Another author who contributes to this theme is Costa (2017), who emphasizes the importance of
Early diagnosis is crucial, as the timing of diagnosis significantly impacts the course of the disease.
therapeutic approach to be followed.

There are several treatments available, as DCC exhibits wide variability.
phenotypic. Thus, there are authors such as Cunha (2014), who assert restorative treatment as
a less invasive and effective option in certain cases. In contrast, Atil (2017)
He advocates for surgical and prosthetic interventions, as well as the placement of dental implants, as a path forward.
efficient therapeutic, while Rocha (2014) highlights that orthodontic follow-up is
indispensable for clinical success.

Furthermore, the studies analyzed demonstrated a consensus regarding the need for a
An interdisciplinary approach in the treatment of patients with chronic cardiovascular disease.

Given the above, it is clear that, over the years, research on the subject has...
Moving forward, making significant contributions to the scientific community and to dental practice,
expanding knowledge about its clinical manifestations, improving protocols of
diagnosis and promoting the development of more effective therapeutic approaches and
individualized.

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FINAL CONSIDERATIONS

The pediatric dentist plays an essential role in the management of Cleidocranial Dysplasia. in children. Their work involves early diagnosis, ongoing monitoring, and management of Dental anomalies and a multidisciplinary approach to ensure oral health and well-being. The child's overall well-being. Appropriate treatment and careful attention throughout development. Dental care for children with DCC is essential to minimize the impacts of this condition and to improve the patient's quality of life. Despite recent advances, the literature on the subject... It still has considerable gaps, which makes future research essential. focused on the development of specific clinical protocols for the management of patients with CHD, as well as conducting longitudinal studies that allow for a deeper understanding the impact of therapeutic intervention on different age groups.

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