

A successful dental management in a child with Cornelia de Lange Syndrome: rare case of Cornelia de Lange Syndrome in child

O sucesso do manejo odontológico em criança com Síndrome Cornelia de Lange: um caso raro da Síndrome Cornelia de Lange em criança

Clarissa Christina Avelar Fernandez¹, Queila Braga Oliveira², Helena Freire Romanos², Marta Teresinha Fornasari³, Gloria Fernanda Barbosa de Araújo Castro^{4*}

¹PhD student, Department of Pediatric Dentistry and Orthodontics, School of Dentistry, Universidade Federal do Rio de Janeiro; ²MSD, Department of Pediatric Dentistry and Orthodontics, School of Dentistry, Universidade Federal do Rio de Janeiro; ³DDS, Department of Pediatric Dentistry and Orthodontics, School of Dentistry, Universidade Federal do Rio de Janeiro. ⁴Associate Professor, Department of Pediatric Dentistry and Orthodontics, School of Dentistry, Universidade Federal do Rio de Janeiro.

Abstract

Background: the Cornelia de Lange Syndrome (CdLS) is a rare and complex syndrome characterized, basically, by psychomotor retardation associated with a number of congenital malformations. **Aims:** this paper reports the case of an 11-year-old female child diagnosed with CdLS and her successful dental management. **Case report:** the patient had severe mental retardation, definite negative behavior and the clinical findings included oral and physical changes. The patient's oral hygiene was deficient with the presence of calculus and gingivitis, besides several active caries lesions in permanent and deciduous dental elements. The treatment consisted in guidance for caregivers about oral hygiene and diet, and the dental procedures were performed under general anesthesia. Currently, the patient is accompanied by monthly follow-ups. **Conclusions:** the lack of knowledge about oral hygiene and cariogenic diets was identified as a one of the reasons for the oral diseases present. Due to the need to care for the other more serious and complex health problems, the oral diseases had evolved faster than usual and thus were difficult to treat and maintain thereafter. Under such conditions, the dentist plays a key role within a multidisciplinary team. From the guidance and knowledge provided in the dental clinic, there was a significant improvement in the life quality of the child and her family.

Keywords: Cornelia de Lange Syndrome. Pediatric Dentistry. Mental Disorders.

Resumo

A Síndrome Cornelia de Lange (SCdL) é rara e complexa, e caracteriza-se, basicamente, por retardo psicomotor associado com várias malformações congênitas. **Objetivos:** este artigo reporta o caso de uma menina de 11 anos de idade diagnosticada com Síndrome Cornelia de Lange e o sucesso do seu manejo odontológico. **Relato de Caso:** a paciente possui retardo mental severo, comportamento definitivamente negativo e achados clínicos como alterações físicas e orais. A higiene oral da paciente é deficiente com presença de cálculo e sangramento espontâneo, além da presença de severas lesões de cárie nos dentes permanentes e decíduos. O tratamento consistiu em orientação da dieta e de higiene oral para os responsáveis, e os procedimentos odontológicos foram realizados sob anestesia geral. Atualmente, a paciente é acompanhada por consultas de revisão periódicas a cada mês. **Conclusões:** a falta de conhecimento sobre a importância da higiene oral e hábitos alimentares foi identificada como uma das razões para a condição oral da paciente. Devido à necessidade de cuidar do estado de saúde geral da paciente, as doenças orais evoluíram mais rápido do que o habitual, tornando o seu tratamento um desafio ainda maior. Sob tais condições, o dentista desempenha um papel fundamental dentro de uma equipe multidisciplinar. A partir da orientação e conhecimento fornecido na clínica odontológica, houve uma melhoria significativa da qualidade de vida da criança e de sua família.

Palavras-chave: Síndrome de Lange. Odontopediatria. Transtornos mentais.

INTRODUCTION

The Cornelia de Lange Syndrome (CdLS) presents psychomotor retardation and congenital malformations, including skeletal, gastrointestinal, cardiac and craniofacial deformities¹. Diagnosis is made clinically

based on characteristic facial features². The prevalence is estimated at 01 per 10,000 births³.

CdLS is rare, complex and causes delays in the maturation of most systems of human organs.² Knowledge of these issues is essential for professionals to provide appropriate care, including oral health, and a better quality of life. This paper reports the case of an 11-year-old female child diagnosed with CdLS and her successful dental management.

Correspondente/ Corresponding: *Gloria Fernanda Barbosa de Araújo Castro – End: Caixa Postal: 68066 – Cidade Universitária – CCS CEP: 21941-971 – Rio de Janeiro – E-mail: gfbacastro@yahoo.com.br – Tel: Fax: +55 (21) 39382101

CASE REPORT

An 11-year-old female child, diagnosed with CdLS, was referred to the Special Needs Clinic, Universidade Federal do Rio de Janeiro, with caries lesions and gingival bleeding as the main complaint.

CdLS was clinically confirmed at birth and anamnesis revealed a history of anemia, throat infections and slow healing of cuts and injuries. No allergic reactions to medicines were reported. Currently, the patient is monitored at the Pediatric Hospital of the university and is not under treatment for any other disease. No other genetic disorders were reported in the family.

The patient has severe mental retardation, she lacks coordination although hearing is normal and she manages to mumble sounds and a few words, but with impaired pronunciation. She has deficiencies in learning, communication and behavior. Physically she has malformed upper limbs, synophrys, snub nose, low ears, thin lips with clear down slope of the labial commissures "carp mouth", microstomia, widespread hypertrichosis and small feet with an interdigital membrane (Figure 1).

Figure 1 – Clinical features



Fonte: Autoria própria

The patient first visited a dentist as a baby but due to extremely negative reactions did not return until now. The oral clinical examination showed active caries lesions in sixteen teeth, deciduous and permanent; elements 71 and 81 presented prolonged retention with moderate mobility; deficient oral hygiene showing calculus, gingivitis and spontaneous bleeding (Figure 2). The lack of collaboration and need for physical restraint have prevented

normal procedures for oral hygiene and toothbrushing. The child presented negative behavior, compulsive crying, screaming, kicking, throughout the consultation, requiring physical restraint. So that, radiographic examinations and photographs could not be carried out due to the averse/unfavorable behavior of the patient.

Figure 2 – Dental findings: Deficient hygiene – Active caries lesions and gingivitis with spontaneous bleeding



Fonte: Autoria própria

The patient's nutritional habits include a baby bottle of milk containing sugar and cereal between meals and before bed.

The caregivers received guidance on oral hygiene and diet. The dental treatment was performed under general anesthesia according the following plan: scraping both arches, dental prophylaxis, total removal of caries and restoration with glass ionomer cement. The caries of the element 54 evolved into a large coronary destruction and pulpal involvement, and it was chosen to dental extraction of this element. 71 and 81, elements that presented greater mobility (with risk of swallowing) than the observed during the previous clinical examination, also were extracted. Finally, the topical application of neutral fluoride gel was performed. During the dental treatment, there were no complications in anesthesia and/or in process of intubation, as well as, it was confirmed that there were no complications during the post-operative period.

The patient is in monthly follow-up for supervised toothbrushing, reinforcement of oral hygiene and nutritional instructions, dental prophylaxis, and topical fluoride applications. A significant improvement in oral hygiene has been noted in the follow-ups, and physical restraint for toothbrushing has become easier.

DISCUSSION

The hallmark of CdLS is the characteristic features of the patient's face (synophrys, "carp mouth", thin upper lip, long philtrum, microstomia).^{1,2} Furthermore, severe mental retardation, hirsutism, and small feet, among

other congenital malformations are also present as observed here.^{1,2} Severe malformation of the upper limbs is considered a clinical indicator of a worse prognosis and can lead to severe cognitive disorders,⁴ as observed in our patient.

Children with CdLS have difficulties in behavior, socialization and communication.^{2,3} Hyperactivity may decrease concentration, but does not affect hearing². Aggressive behavior is probably due to frustration of poor verbal communication.⁴ In our case, this behavior influenced our choice for treatment under general anesthesia.

Children with special health care needs usually have poor oral hygiene, increased risk of dental problems and unmet medical and dental care.⁵ Parents or caregivers usually seek treatment for caries, only after the development of caries disease rather than preventive care as more care is needed for the other complex health problems, resulting in severer dental needs.⁵

The government does not usually provide specialized dental care for this population, which makes access to dental treatment difficult.⁵ Thus, these children are referred to specialized services where the caregivers are encouraged to seek oral health care. In our case, the caregivers were not well informed and had made no effort to obtain the necessary care for the child and the periodontal disease was at an advanced stage. The caregivers' lack of knowledge about oral hygiene and cariogenic diets was identified as a partial cause of the patient's condition.

Nutritive advice must be given with oral hygiene instructions. Here, the dental treatment and maintenance was affected by the bottle feeding between meals and before bedtime. Such habits are difficult to stop but an effort to stop is necessary.

CdLS causes significant limitations for such patients. Thus, a multidisciplinary approach, where the dentist plays a key role, is necessary. The guidance and knowledge given

at the dental clinic provided a significant improvement in quality of life of the child and her family. Making the caregivers aware of the needs was the first step for improving the child's oral health.

CONCLUSIONS

The CdLS is responsible for a series of limitations in the lives of affected children. In this context, a multidisciplinary approach, where dentists play a key role, is extremely necessary. From the guidance and knowledge provided in the dental clinic, there was a significant improvement in quality of life of the child and her family. The caregiver's realizing was the first step for the changes and the benefits in child's oral health.

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REFERENCES

1. TOKER, A.S. et al. Dental findings in cornelia de lange syndrome. *Yonsei Med. J.*, Seoul, v. 50, n.2, p.289-292, 2009.
2. SCARPELLI A.C. et al. Cornelia de lange syndrome: a case report of a Brazilian boy. *Cleft Palate Craniofac. J.*, Leviston, v. 48, n. 4, p. 490-493, 2011.
3. BHUIYAN Z.A.; ZILFALIL B.A.; HENNEKAM R.C.M. A Malay boy with the Cornelia de Lange syndrome: clinical and molecular findings. *Singap. Med. J.*, Singapore, v. 47, n. 8, p.724-727, 2006.
4. BARBONI C. et al. A New Report of Cornelia de Lange Syndrome Associated With Split Hand and Feet. *Am. J. Med. Genet.*, New York, v.A, p.1-3, 2012.
5. SALLES P.S. et al. Dental Needs and Management of Children With Special Health Care Needs According to Type of Disability. *J. Dent. Child.*, Chicago, v.79, n.3, p.46-50, 2012.