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Orofarineal dysfunction in children with the Cornélia De Lange Syndrome: series of cases

Disfagia orofaríngea em crianças com síndrome Cornélia De Lange: série de casos

Disfagia orofaríngea en niños con síndrome de Cornelia De Lange: serie de casos

Vanessa Souza Gigoski de Miranda[*]
Andressa de Moura Barbosa[*]
Lisiane de Rosa Barbosa[*]
Gilberto Bueno Fischer[*]
Rafaela Soares Rech[*]

Abstract

Introduction: Cornélia De Lange Syndrome (CdLS) is characterized by being polymalformative that involves facial anomalies, growth and psychomotor development retardation, behavioral changes and associated malformations. It is known that children affected by this syndrome have swallowing disorders, but there are few studies presented in the literature due to the rarity of the disease, with a case report being found and mostly with description of the findings. **Objective:** To identify swallowing disorders in children with Cornelia de Lange Syndrome, through videofluoroscopy. **Methodology:** Case series, retrospective. This is a convenience sample with children, diagnosed with Cornelia de Lange Syndrome, who had swallowing videofluoroscopy. Medical records of patients who were not complete were excluded. The sample characterization data were obtained from physical records and the study outcome data through clinical reports of patients' swallowing videofluoroscopies. **Results:** Of the 6 individuals, 5 were male, in which 3 (50%) had laryngotracheal aspiration, silently. The median age was 5.50 months. According to the findings in the swallowing videofluoroscopies, swallowing difficulties were identified, such as

* Universidade Federal de Ciências da Saúde de Porto Alegre, RS, Brazil.

Authors' contributions:
VSGM: Study design; Methodology; Article outline; Critical review.
AMB: Study design; Data collection; Article outline.
LRB: Study design, Methodology, Critical review; Orientation.
GBF: Methodology, Critical Review; Guidance.
RSR: Study design, Methodology, Article outline; Critical review; Guidance.

Correspondence e-mail: Vanessa Souza Gigoski de Miranda - vanessa_gigoski@hotmail.com
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premature posterior escape of food, inefficient ejection and difficulties in the formation of the bolus, such as delay in triggering the pharyngeal reaction, reflux to the nasopharynx, stasis in the valleys and peripheral sinuses and tracheal aspiration. **Conclusion:** All children with Cornelia de Lange Syndrome in this study had dysphagia to some degree, and half of them had silent laryngotracheal aspiration.

Keywords: De Lange Syndrome; Deglutition Disorders; Speech, Language and Hearing Sciences.

Resumo

Introdução: A síndrome Cornélia De Lange (CdLS) é caracterizada por ser polimalformativa que envolve anomalias faciais, atraso de crescimento e desenvolvimento psicomotor, alterações comportamentais e malformações associadas. Sabe-se que as crianças acometidas por essa síndrome apresentam alterações de deglutição, mas são poucos os estudos apresentados na literatura devido à raridade da doença, sendo encontrado relato de um caso, e na maioria das vezes, com descrição dos achados. **Objetivo:** Identificar as alterações de deglutição em crianças com a Síndrome Cornélia de Lange, por meio da videofluoroscopia. **Metodologia:** Série de Casos, retrospectiva. Trata-se de uma amostra de conveniência com crianças, diagnosticadas com Síndrome Cornélia de Lange, que apresentassem videofluoroscopia da deglutição. Foram excluídos prontuários de pacientes que não estivessem completos. Os dados de caracterização da amostra foram obtidos através de prontuários físicos e os dados de desfecho do estudo através de laudos clínicos de videofluoroscopias da deglutição dos pacientes. **Resultados:** Dos 6 indivíduos, 5 do sexo masculino, em que 3 (50%) apresentaram aspiração laringotraqueal, de forma silente. A mediana de idade foi de 5,50 meses. Conforme os achados nas videofluoroscopias da deglutição, identificou-se dificuldades de deglutição como escape posterior prematuro de alimento, ejeção ineficiente e dificuldades de formação do bolo alimentar, como atraso no acionamento da reação faríngea, refluxo para nasofaringe, estase em valéculas e seios periformes e aspiração traqueal. **Conclusão:** Todas as crianças com Síndrome Cornélia de Lange deste estudo apresentaram disfagia em algum grau, e metade delas apresentou aspiração laringotraqueal de forma silente.

Palavras-chave: Síndrome de Lange; Transtornos de Deglutição; Fonoaudiologia.

Resumen

Introducción: El síndrome de Cornélia De Lange (CdLS) se caracteriza por ser polimalformativo que involucra anomalías faciales, retraso del crecimiento y desarrollo psicomotor, cambios de comportamiento y malformaciones asociadas. Se sabe que los niños afectados por este síndrome presentan trastornos de la deglución, pero existen pocos estudios presentados en la literatura debido a la rareza de la enfermedad, encontrándose un reporte de caso y la mayoría de las veces con descripción de los hallazgos. **Objetivo:** identificar los trastornos de la deglución en niños con síndrome de Cornelia de Lange, mediante videofluoroscopia. **Metodología:** Serie de casos, retrospectiva. Se trata de una muestra de conveniencia con niños, diagnosticados de Síndrome de Cornelia de Lange, que habían ingerido videofluoroscopia. Se excluyeron los registros médicos de los pacientes que no estaban completos. Los datos de caracterización de la muestra se obtuvieron de los registros médicos físicos y los datos de los resultados del estudio a través de informes clínicos de videofluoroscopias de deglución de los pacientes. **Resultados:** De los 6 individuos, 5 eran varones, de los cuales 3 (50%) tenían aspiración laringotraqueal, en silencio. La mediana de edad fue de 5,50 meses. De acuerdo con los hallazgos en las videofluoroscopias de deglución, se identificaron dificultades de deglución, como escape posterior prematuro de alimentos, eyección ineficiente y dificultades en la formación del bolo, como retraso en el desencadenamiento de la reacción faríngea, reflujo a la nasofaringe, estasis en los valles y senos periféricos y aspiración traqueal. **Conclusión:** Todos los niños con síndrome de Cornelia de Lange en este estudio tenían disfagia en algún grado y la mitad de ellos tenían aspiración laringotraqueal en silencio.

Palabras clave: Síndrome de Lange; Trastornos de la deglución; Terapia del habla.

Introduction

Cornélia De Lange syndrome (CdLS) is characterized by being polymalformative involving facial anomalies, growth retardation and psychomotor development, behavioural changes and associated malformations (cardiac, gastrointestinal and musculoskeletal)[1] . Patients with CdLS may have cleft palate, cognitive impairment, visual and auditory changes[2,3]. It refers to a rare syndrome, in which, epidemiologically, the incidence is 1: 10,000 to 1: 30,000 live births[4,5]. In addition, they usually have low life expectancy due to cardiac abnormalities, apneas, food aspiration[4].

In a study developed by Orenstein (2009) [6], changes were identified in the oral and pharyngeal phase of swallowing, and it was suggested that these could be associated with changes in the integrity of the central nervous system and in the structures involved in the swallowing process . Also in this study, changes in tone and orofacial sensitivity and in the mobility of structures were observed. All these changes in tone and sensitivity of oropharyngeal structures may be causes of difficulties in the swallowing process[7]. The limited laryngeal excursion observed in these individuals may be associated with the neuromuscular alteration present in patients with CdLS[7].

Another important factor identified in the syndrome is gastroesophageal reflux (GERD). The reflux of food into the nasopharynx indicates an inefficiency of the velopharyngeal seal and causes pressure leakage during the pharyngeal phase of swallowing, which can lead to laryngotracheal aspiration[8]. It is known that pneumonia and GERD were determined to be the most common cause of death (31%) in 295 people with CdLS over a 41-year period[9].

Considering the functional impairments of swallowing and consequences in respiratory conditions, it is necessary that there be more studies on oropharyngeal dysphagia of patients with CDLS to get to know the scientific and clinical community that work with these patients. The present study aims to identify swallowing changes in children with Cornelia de Lange Syndrome, through swallowing videofluoroscopy.

Methodology

Retrospective case series, carried out in a philanthropic institution that provides care to children and adolescents with multiple disabilities, in partnership with Universidade Federal de Ciências Médicas de Porto Alegre after approval by the university's Research Ethics Committee under number 3,288,377 and approval by the institution of origin of the students. It is a convenience sample made up of 6 children, diagnosed with Cornelia de Lange Syndrome, who had swallowing videofluoroscopy and had been screened by the multidisciplinary rehabilitation centre in question. Medical records of patients who were not complete were excluded. The sample characterization data were obtained from physical medical records and the study outcome data through clinical reports of the patients' swallowing videofluoroscopies. The examinations were carried out in different clinics and hospitals, according to the coverage of the health plan of each patient. Each of the services presents the report in a different way, describing the changes in the oral and pharyngeal phase of swallowing and the presence of laryngotracheal penetration and / or aspiration. In addition, the completion of each exam may or may not include the degree of severity of dysphagia - when present - according to the standardization of the service.

Descriptive analyses were performed using absolute and relative frequencies for categorical and median variables and interquartile range for the numerical variable (age). Fisher's exact test was used to investigate the association between landmarks in the swallowing process and laryngotracheal aspiration considering a significance level of 95%. To analyze the binomial distribution, the probability of each milestone in the swallowing process taking place among the sample studied was analyzed. The statistical software used was SPSS version 21.0.

Results

Six children with CdLS were identified, who had complete medical records and a videofluoroscopy examination of swallowing. Table 1 shows the description of the cases, with age, reason for referral for the exam, consistency and tools used during the exam, and the completion of the swal-

lowing videofluoroscopy. Of the 6 individuals, 3 (50%) had laryngotracheal aspiration. The median age was 5.50 months and the interquartile range was (3.13 - 27.75). Of these, 5 were male. The most frequent reasons for the exam were choking, coughing and difficult oral acceptance of food.

Table 1. Description of the cases Cornelia de Lange Syndrome referred for swallowing videofluoroscopy. Porto Alegre, 2019. (n = 6)

Subject	Age (months)	Reason for the exam	Consistency	Tool used	Conclusion of the exam
Case 1	7	Choking	Thin liquid	Flow M orthodontic nipple bottle	Oropharyngeal Dysphagia
Case 2	5	Use of nasoenteric probe	Liquid nectar and pudding	Cup and spoon	Moderate to Intense Oropharyngeal Dysphagia
Case 3	90	Side dish	Thick pasty, fine pasty, thin liquid and thickened liquid	Cup and spoon	Mild to Moderate Oropharyngeal Dysphagia
Case 4	0,53	Speech therapy for consistency progression	Thin liquid	Duckling and syringe	Mild Oropharyngeal Dysphagia
Case 5	6	Difficulty in oral acceptance	Liquid and pasty	Baby bottle and spoon	Oropharyngeal Dysphagia
Case 6	4	Has cough during oral	Pasty	Spoon	Oropharyngeal Dysphagia

According to the findings in the swallowing videofluoroscopies, difficulties in the oral swallowing phase were identified, such as premature posterior escape of food, inefficient ejection and difficulties in forming the bolus, which were presented in Figure 1. In Figure 2, the changes identified in pharyngeal phase, such as delay in triggering the pharyngeal reaction, reflux to the nasopharynx, stasis in valecules and peripheral sinuses and tracheal aspiration. As identified, half of the cases presented, laryngotracheal aspiration of food was found. In all exams, this aspiration was observed silently.

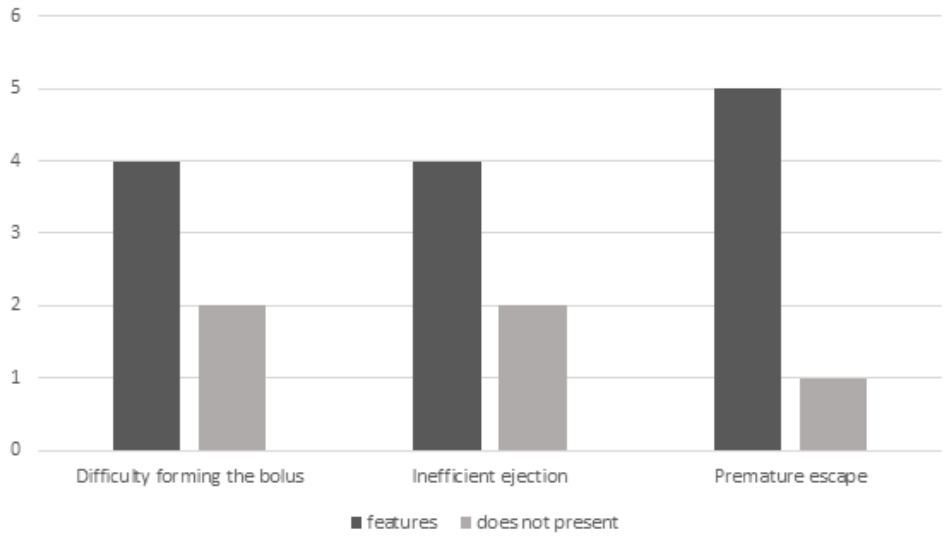

Figure 1. Oral swallowing phase of patients with Cornelia de Lange Syndrome. Porto Alegre, 2019.

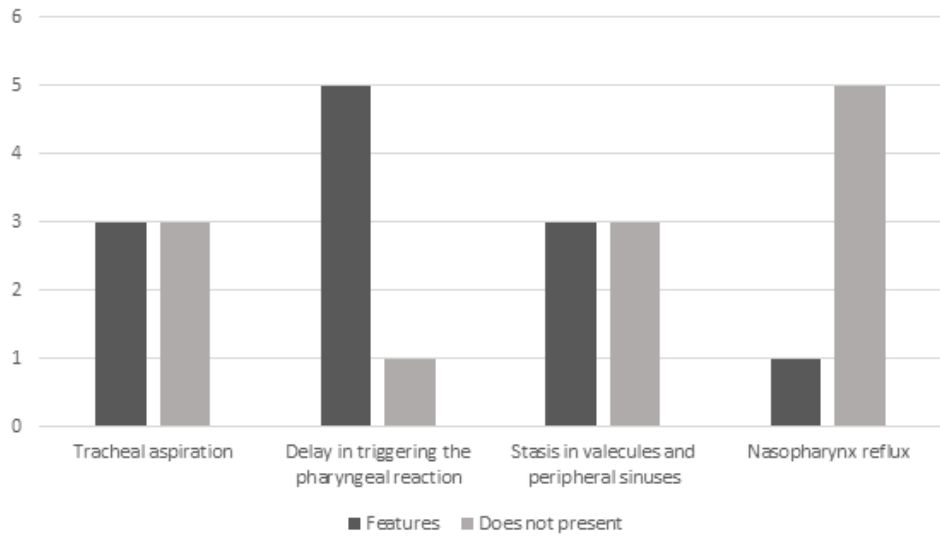

Figure 2. Pharyngeal phase of swallowing in patients with Cornelia de Lange Syndrome. Porto Alegre, 2019. (n = 6)

It is observed that there was a predominance of aspiration in cases of premature posterior escape, difficulties in the formation of the bolus of food, delayed activation of the pharyngeal reaction and stasis in valleys and piriform sinuses. The relationship described is shown in Table 2.

Table 2. Association between swallowing characteristics investigated from videofluoroscopy with laryngotracheal aspiration in children with Cornelia de Lange Syndrome, Porto Alegre, 2019. (n = 6)

Variable	Laryngotracheal Aspiration		p-value
	Yes	No	
Premature Later Escape			0,500
Yes	2 (66,7%)	3 (100,0%)	
No	1 (33,3%)	0 (0,0%)	
Inefficient Ejection			0,200
Yes	3 (100,0%)	1 (33,3%)	
No	0 (0,0%)	2 (66,7%)	
Difficulties in forming the bolus			0,200
Yes	1 (33,3%)	3 (100,0%)	
No	2 (66,7%)	0 (0,0%)	
Delay in triggering the pharyngeal reaction			0,500
Yes	2 (66,7%)	3 (100%)	
No	1 (33,3%)	0 (0,0%)	
Nasopharynx reflux			0,500
Yes	1 (33,3%)	0 (0,0%)	
No	2 (66,7%)	3 (100,0%)	
Stasis in valecules and piriform sinuses			0,500
Yes	1 (33,3%)	2 (66,7%)	
No	2 (66,7%)	1 (33,3%)	

Figure 3 shows that the swallowing milestones that are most likely to occur among study participants are: premature posterior escape (5/6) and delay in triggering the pharyngeal reaction (5/6).

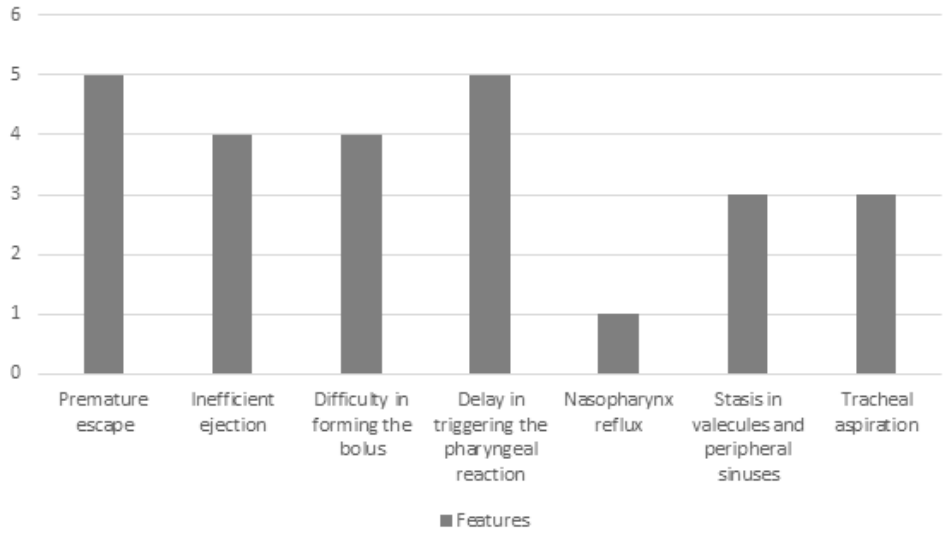

Figure 3. Binomial distribution of deglutition milestones by probability of presence in participants

Discussion

The aim of this research was to identify swallowing disorders in children with Cornelia de Lange Syndrome. These children, as already presented, may present cognitive alterations and delay in neuropsychomotor development[2,3]. It is known that neurological dysfunctions can affect the formation and transport of the bolus to the posterior portion of the oral cavity and the muscle action responsible for transporting the bolus from the oral cavity to the esophagus, leading to changes at the oral and / or pharyngeal level[10].

Regarding the difficulties found in the oral phase of swallowing, such as premature posterior escape of food, inefficient ejection and difficulties in forming the bolus, this same finding was found in a study in patients with cerebral palsy in which it was observed that 100% of children had inadequate cake uptake, absence of lip sealing and inefficient cake preparation in clinical evaluation[11]. It has also been described that children with encephalopathy have a higher frequency of changes in the stomatognathic system and adaptations to the use of related structures[12], corroborating the findings of our study.

Oral ejection in oropharyngeal dysphagia is often deficient due to the inability to pressurize the oral cavity. In this case, the hypopharynx almost always shows marked residue and it is not uncommon for laryngotracheal penetration or aspiration to be associated with these deficient ejections[13].

Regarding the alterations identified in the pharyngeal phase, such as delay in triggering the pharyngeal reaction, reflux to the nasopharynx, stasis in valleys and peripheral sinuses and tracheal aspiration, it can be explained in some studies that the presence of residues in valleys and pyriform recesses for pasty can occur due to the viscosity properties of the food, associated with the decrease in the pressure wave, common in children with neurological disorders[14,15]. In some situations, such as a reduction in pharyngeal contraction and alteration of the closure of the upper esophageal sphincter, they can result in food stasis in the vallecula, pyriform recesses and posterior pharyngeal wall. The greater the delay in swallowing, the greater the risk of silent aspiration[16], a finding found in all children who aspirated in our study.

The reflux of food into the nasopharynx indicates an inefficiency of the velopharyngeal seal and causes pressure leakage during the pharyngeal

phase of swallowing, which can lead to laryngotracheal aspiration[8]. The patient may also have nasal reflux, together with other changes, both in the oral and pharyngeal phases, thus being able to contribute to the aspiration episode[7]. Despite being a frequent finding in the population with CdLS3, only 1 child in our study had such a finding. It is believed that we did not find a greater number of GER changes, due to swallowing videofluoroscopy not being the gold standard for the diagnosis of reflux, but rather, the esophagus and stomach radiography.

It is suggested that the reduced laryngeal elevation associated with the limited opening of the pharyngoesophageal transition also causes the presence of food trapped in the vallecula and in the piriform recesses, in cases of the syndrome. This may be due to muscle hypertonia and a reduction in muscle contraction at the base of the tongue and pharyngeal wall[8].

The swallowing changes found, both in the oral and pharyngeal phases, may be related to changes in the integrity of the central nervous system and in the structures involved in the swallowing process[6]. These clinical manifestations of swallowing disorders are not specific to each etiology, but they constitute a syndrome, which can lead to food refusal, fatigue and coughing during feeding, oral escape, nasal regurgitation, choking, suffocation, asphyxia, cyanosis and quality change vocal, in addition to lung and aspiration problems[17] which can lead to nutritional deficits, dehydration, resulting in weight loss, pneumonia and death[18].

Conclusion

Oropharyngeal dysphagia was identified in all children with Cornelia de Lange Syndrome, with oral and pharyngeal phase impairments. In cases where laryngotracheal aspiration of food was identified, this occurred silently.

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