Case Report

Relato de Caso

Qualitative and quantitative analysis of oropharyngeal swallowing in Down syndrome

Análise qualitativa e quantitativa da deglutição orofaríngea na Síndrome de Down

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Keywords

Swallowing
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ABSTRACT

Purpose: To describe the qualitative and quantitative temporal analysis of oropharyngeal swallowing in children diagnosed with Down syndrome (DS) through a case series study of six individuals aged 4 to 17 months (mean age = 11.16 months; median = 12 months). **Methods:** Qualitative and quantitative temporal analysis of swallowing using videofluoroscopy and specific software. The following parameters were assessed: presence or absence of oral incoordination, labial sphincter sealing incompetence, oral residue, posterior oral spillage, laryngotracheal penetration and aspiration, pharyngeal and total oral transit time (TOTT). **Results:** Qualitative analysis identified individuals with disorders in at least four of the swallowing parameters investigated. Only one individual presented total oral transit time (TOTT) different from the others. No difference was observed between the cases regarding pharyngeal transit time. **Conclusion:** Qualitative swallowing disorders are observed in children with DS, with difference in TOTT only in the case report of the youngest infant.

Descritores

Deglutição
Transtorno de Deglutição
Síndrome de Down
Fluoroscopia
Análise Quantitativa
Software

RESUMO

Objetivo: Este estudo tem por objetivo descrever a análise qualitativa e quantitativa temporal da deglutição orofaríngea em crianças com diagnóstico de Síndrome de Down (SD). **Método:** Estudo de série de seis casos, com idade variando de quatro a 17 meses (Média de 11,16 meses e mediana de 12 meses). Realizada análise qualitativa e quantitativa temporal da deglutição orofaríngea por meio de videofluoroscopia de deglutição e *software* específico. Foram analisados os parâmetros qualitativos de coordenação oral, resíduos orais, escape oral posterior, penetração, aspiração laringotraqueal e realizada análise do tempo de trânsito oral total (TTOT) e faríngeo. **Resultados:** Verificou-se alteração em pelo menos quatro dos parâmetros qualitativos investigados. Somente um dos indivíduos apresentou diferença no TTOT quando comparado com os demais e não houve diferença no tempo de trânsito faríngeo entre os casos. **Conclusão:** Houve alterações qualitativas na deglutição em crianças com SD e diferença no TTOT somente no caso de menor faixa etária.

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INTRODUCTION

Down syndrome (DS), also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. It is the main cause of mild to moderate intellectual disability, occurring in 1:600-800 live births each year[1]. The DS phenotype is well defined, known, and widely described in the specific scientific literature, presenting physical characteristics of easy recognition, clinical identification, and confirmation of genetic diagnosis.

Regarding oral motor performance, children with DS present reduced oral cavity, hypotonicity of oral and facial muscles, dental malocclusion, and macroglossia[1,2]. In addition, these children may present alterations in the central nervous system development and esophageal motility, leading to impairments in the stomatognathic functions such as oropharyngeal swallowing[2].

The videofluoroscopic swallowing exam (VFSE) is considered the gold standard method for assessment of deglutition, including in pediatrics, allowing visualization of all phases and enabling qualitative and quantitative analyses of distance and time. Although several previous studies have used VFSE for the qualitative analysis of oropharyngeal swallowing in DS, they investigated variables different from those proposed in this study. These studies fragmented the investigation of dysphagia: some with greater emphasis on the oral phase[3], others on the pharyngeal phase, highlighting the presence or absence of tracheal aspiration[2].

Quantitative temporal analysis of oropharyngeal swallowing has been used less frequently in pediatric cases than in the adult population[4], with few studies investigating healthy children[5] and individuals with pathological conditions such as cerebral palsy. No studies on temporal analysis of dysphagia in this population measured by software have been found in the literature as proposed herein.

Therefore, this study aimed to describe the qualitative and quantitative temporal analysis of oropharyngeal swallowing in children diagnosed with DS.

PRESENTATION OF THE CLINICAL CASES

The present study was approved by the Ethics Committee on Human Research of the Universidade Estadual Paulista - UNESP - Campus de Marília under protocol no. 1755/2009. The parents and/or legal guardians of the participants signed an Informed Consent Form (ICF) prior to study commencement in which they agreed with the proceedings and dissemination of the research and its results.

Six videofluoroscopic swallowing exams (VFSE) of children diagnosed with Down syndrome (DS) performed in specialized clinics between 2015 and 2016 were included in the study. To characterize and describe the cases of DS confirmed by laboratory examination (karyotype), the following information was collected during clinical investigation: age, gender, presence of eating and/or swallowing complaints, previous speech-language therapy, history of pulmonary complications, presence of gastroesophageal reflux (GER), history of cardiac conditions, and need for cardiac surgical intervention (Chart 1).

All study participants underwent VFSE with liquid consistency offered in usual nursing bottle at the habitual continuous flow of each child (which does not allow volume control), at side view in a sitting position, with the heads positioned in the direction of the body axis, without flexion or rotation, adapting to their posture when necessary.

The tests were analyzed qualitatively and quantitatively by two experienced speech-language pathologists with expertise in the area. After measurement of the swallowing times by each rater, statistical analysis was conducted to verify the agreement between them using the Student's *t*-test at 5% significance level ($p<0.05$). No statistically significant difference was observed between the values reported by the raters ($p=0.009$). The inter-rater agreement mean was calculated for oral and pharyngeal transit times. The following parameters were considered in the VFSE qualitative analysis: presence or absence of oral incoordination, labial sphincter sealing incompetence, oral residue, posterior oral spillage, and laryngotracheal penetration and aspiration[6]. Quantitative temporal analysis of swallowing was conducted using specific software[7] to measure, in milliseconds (ms), the total oral transit time (TOTT) and pharyngeal transit time (PTT).

Chart 1. Clinical description of the study cases

Case	Age	Gender	Clinical history					
			Swallowing complaints	Pulmonary complications	Gastroesophageal reflux	Cardiac conditions	Cardiac surgical intervention	Previous Speech-language therapy
1	1 to 5 months	Male	✓			✓		✓
2	1 to 2 months	Female		✓	✓	✓		✓
3	7 months	Male	✓	✓	✓			✓
4	1 to 3 months	Female		✓	✓	✓		✓
5	10 months	Female	✓	✓	✓	✓	✓	✓
6	4 months	Male	✓	✓	✓	✓		✓

Captions: ✓= present

Chart 2. Qualitative and quantitative temporal parameters of oropharyngeal swallowing analyzed by videofluoroscopy

Parameters	Description
Labial sphincter sealing incompetence	Incomplete occlusion between the lips and the nursing bottle nipple with or without presence of anterior oral spillage.
Oral incoordination	Incoordination of the oral structures along the anterior-to-posterior transit of the liquid bolus.
Oral residue	Contrasted material remains in the oral cavity after swallowing.
Posterior oral spillage	Presence of premature liquid escape to the hypopharynx, surpassing the region in which pharyngeal response should occur.
Laryngotracheal penetration	Presence of contrasted material in the laryngeal structures above the vocal folds[6].
Laryngotracheal aspiration	Presence of contrasted material in the laryngeal structures below the vocal folds[6].
Total oral transit time (TOTT)	Total oral transit time (TOTT) was described by Gatto et al.[8] based on the definition of oral transit time (OTT) proposed by Logemann et al.[9]. TOTT was defined as an interval in milliseconds (ms) between the first frame showing the food inside the oral cavity and the first frame showing the head of the bolus in the end of hard palate and begin of soft palate (posterior nasal spine) and the point where the lower margin of the mandible ramus.
Pharyngeal transit time	Pharyngeal transit time (PTT) was defined based on Kendall et al.[10]. PTT begins in the first frame, and it is defined by the interval between the moment the head of the bolus passes the anterior nasal spine and the moment the tail of the bolus passes through the pharyngoesophageal sphincter, with time up to 1150 ms considered as normal[5].

Table 1. Frequency of findings in the qualitative swallowing parameters

Case	Oral Phase				Pharyngeal Phase			
	Oral incoordination	Labial sphincter sealing incompetence	Oral residue	Freq.	Posterior oral spillage	Laryngotracheal penetration	Laryngotracheal aspiration	Freq.
1	+	-	+	66.66%	+	+	-	66.66%
2	+	+	+	100%	-	-	-	0
3	+	+	+	100%	+	-	-	33.33%
4	+	+	+	100%	-	-	-	0
5	+	+	+	100%	+	+	-	66.66%
6	+	+	+	100%	+	+	+	100%
Freq.	100%	83.33%	100%		66.66%	50%	16.66%	

Captions: + = present; – = absent; Freq = frequency

Table 2. Frequency of findings in the qualitative parameters and temporal analysis of each swallowing phase

Case	Frequency of qualitative parameters - Oral phase	Frequency of qualitative parameters - Pharyngeal phase	Total oral transit time in milliseconds	Pharyngeal transit time in milliseconds
1	+ +	+ +	680	600
2	+ + +		800	920
3	+ + +	+	550	750
4	+ + +		890	590
5	+ + +	+ +	800	720
6	+ + +	+ + +	2240*	440

Captions: Frequency of qualitative alterations: Oral phase: +++ = 100%; ++ = 66.66%; Pharyngeal phase: +++ = 75%; ++ = 50%; + = 25%. * TOTT with longest sample time

For both the qualitative and quantitative parameters, the criteria were considered as described in Chart 2.

Table 1 shows that all the cases presented one or more qualitative alterations in the oral phase of deglutition and that Cases 2 and 4 presented alterations only in the oral phase, whereas the other cases presented impairment in both phases; however, laryngotracheal aspiration was only detected in Case 6.

In the analysis of the TOTT, only Case 6 presented a different value compared with those of the other cases. As for the PTT, all the cases presented values within the normality standards for the infantile population (Table 2).

DISCUSSION

The characteristic face, oral and cervical cavity phenotype, associated with the presence of hypotonicity in DS, has already been referred to as one of the aspects responsible for the presence of oropharyngeal swallowing losses in this population[2].

In this study, changes were observed in the oral and pharyngeal phases of swallowing in most of the qualitative parameters analyzed. These results corroborate those in the literature, which preconizes that both the anatomical changes and the neuromotor control of swallowing in this population may negatively affect deglutition[3,11].

A relevant aspect in the discussion of qualitative changes in the oral phase of swallowing in DS is associated with the importance of the brain areas in the modulation of this swallowing phase. The specialized literature suggests that this modulation occurs with the activation of various cortical and subcortical areas of the central nervous system. Thus, it is worth emphasizing that not only the presence of alterations in the myofunctional orofacial aspects of DS, but also other changes in morphophysiological bases of the central nervous system (CNS) could contribute to impair oral phase modulation[12,13].

The presence of laryngotracheal penetration and aspiration occurred in only one of the cases, and although this is a case series study, it is possible to observe that pulmonary complications are more frequently reported than laryngotracheal aspiration in the investigated sample. Pulmonary complications were observed in five cases, as described in the casuistics profile, and the results indicated presence of laryngotracheal aspiration in only one of the cases. These findings suggest that the pulmonary complications present in this population are not only the result of presence of laryngotracheal aspiration, and that issues such as GER and cardiac conditions deserve emphasis in the investigation of pulmonary complications[13]. Aspiration of reflux content is common in the pediatric population diagnosed with GER[14], generating complaints about gagging during feeding, which may be mistaken for oropharyngeal symptoms. However, it is relevant to conduct an objective investigation on swallowing in this population, considering that other studies with larger samples have already demonstrated higher frequency of laryngotracheal aspiration[2,3].

With respect to the quantitative temporal analysis of swallowing, only the case report of the youngest infant showed total oral transit time (TOTT) different from the other cases, and no changes were observed as for pharyngeal transit time (PTT). Considering that significant difference between the youngest individual of the sample and the others was only observed in the TOTT, it is necessary to investigate deglutition performance regarding maturation of the CNS in this population because of the marked difference between this age group and the others. Therefore, we suggest that the influence of age on the biomechanics of swallowing be addressed in further studies on DS, because it is known that the processes of cerebral maturation and orofacial development cause changes in the oral structures, coordination of swallowing mechanisms, and oral modulation which can directly impact this function in the general population, including the DS population[14].

Another aspect that should be considered when measuring the time of the swallowing phases, especially in Pediatrics, is the utensil used in feeding. Different utensils, such as cups, nursing bottles and spoons, affect the modulation of the oral phase of swallowing differently, and can interfere in effective performance in the different variables of this function, as in the transit times[5,15]. In the present study, the measurements were taken using a nursing bottle, and show that the presence of differentiated TOTT, even increased in relation to the normality standard for OTT cited in the literature for children[5], is not associated with the utensil used, but probably with the age group, because it occurred only in the case report of the youngest individual of the sample.

FINAL CONSIDERATIONS

The authors are aware that the sample size imposes limitations to this study, but research on swallowing with this population is still difficult to conduct, considering that screening for eating and/or swallowing disorders at birth and during childhood is not frequent in this population. Furthermore, many family members have no complaints about gagging, and thus do not seek investigation early. When chewing complaints appear with advancing age, changes in swallowing may have already been compensated and are concentrated in the oral phase of deglutition and in the oral preparation phase. In view of what has been exposed, we suggest that investigation of swallowing abilities be performed early in this population and may contribute to identify cases of greater risk. Future studies with larger samples of children with DS at different age ranges are needed to facilitate understanding the findings on this important genetic disease.

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Author contributions

AVMNS was the main author of the article, responsible for the study design, literature search, collection and analysis of data, and writing, processing and submission of the manuscript; PCC, CMG, and RGS were the study advisers, contributed in the writing, correction, and approval of the final version of the manuscript.