

QUANTITATIVE ANALYSIS OF OROPHARYNGEAL SWALLOWING IN NEURONAL CEROID LIPOFUSCINOSIS WITH GASTROSTOMY: CASE REPORT

Análise quantitativa da deglutição orofaríngea em indivíduo gastrostomizado com lipofuscinose ceróide neuronal: relato de caso

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ABSTRACT

The presence of oropharyngeal dysphagia in the pediatric population with genetic diseases it is still poorly studied. The aim of this study was to analyze the oral total transit time and pharyngeal transit time, in an individual with neuronal ceroid lipofuscinosis (NCL) with severe oropharyngeal dysphagia. Individual with NCL, 3 years old, 2 years with gastrostomy and no oral feeding, weighting loss, but without pulmonary complications. Oropharyngeal swallowing was studied by videofluoroscopy and it was realized a quantitative analysis using software. Changes were observed throughout the whole biomechanics of swallowing. The quantitative analysis of total oral transit time was found 45.37 seconds (default normality in children is 4 seconds) and for pharyngeal transit time was 4.53 seconds. It was found that beside the changes in the biomechanics of oropharyngeal swallowing in the case studied, an increase in total oral transit time and pharyngeal transit time was also observed, which can significantly compromise the nutritional status and pulmonary these individuals.

KEYWORDS: Neuronal Ceroid-Lipofuscinoses; Deglutition Disorders; Quantitative Analysis

■ INTRODUCTION

The neuronal ceroid lipofuscinoses (NCL) are a group of neurodegenerative diseases, clinically

and genetically heterogeneous, characterized by accumulation of autofluorescent material within the cells, resulting in distinct ultrastructural¹ patterns.

Clinically these diseases are characterized by progressive visual loss and epilepsy. Their clinical ratings are established based on age at onset of the appearance of ultrastructural findings, and the late infantile NCL, also known as Jansky-Bielschowsky disease, has its onset between the second and fourth year of life^{1, 2}.

The studies that have been conducted with this genetic condition are concentrated in the diagnostic process and the genetic aspects of this condition, and few have studied the characterization of diverse symptomatology which may be part of the clinical NCL³. Some clinical studies have indicated the presence of oropharyngeal swallowing difficulties in this population, however, few have investigated this function through gold standard examinations such as deglutition videofluoroscopy^{1, 2, 4}.

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Quantitative analytic studies of total oral and pharyngeal transit time in children are scarce, and such measurements are still in the standards-setting⁵ phase. Moreover, the implication that the change in these parameters may result in altered pulmonary or nutritional status deserves investigation.

Therefore, the aim of this study was to analyze the total oral transit time (TOTT) and pharyngeal transit time (PTT) in an individual with a clinical genetic diagnosis of Neuronal Ceroid Lipofuscinosis (NCL) with severe oropharyngeal dysphagia.

■ CASE PRESENTATION

JET, 3 years and 9 months, female, was referred to the Laboratory of Dysphagia in a reference center specialized in this investigation, in the state of São Paulo – Brazil in April 2010. The patient had clinical genetic diagnosis of late infantile neuronal ceroid lipofuscinosis performed at a referral center for genetic diagnosis. The family reported complaints of difficulty swallowing and feeding from 2 years and 3 months. At the time of this evaluation, the patient had been fed exclusively by alternative means of feeding for two years.

Prior to the investigation of oropharyngeal swallowing, full phonoaudiological clinical and neurological complementary investigations were performed, and in these it was found that the patient had severe impairment of comprehension and expression of language, intellectual disability and severe visual impairment.

For analysis of the TOTT and PTT a videofluoroscopic evaluation of swallowing, applying low-risk protocol, which recommends the use of thin paste consistency in volume of 5 ml for 3 consecutive offers and the individual in his best state of alertness⁵. The consistency was prepared with the use of instant food thickener of one of the brands available on the market, consisting of starch containing 375 kcal per 100g, and 125mg of sodium for each 100g of carbohydrates. This thickener was added to the patient's usual food before undergoing gastrostomy and was added to the consistency of barium (BaSO₄) sulphate in a ratio of 50% barium to 50% food without altering the previously standardized consistency.

After the videofluoroscopic evaluation of swallowing, a computerized analysis of the total oral and pharyngeal transit time was performed with the aid of specific software for this analysis, which provided the recording of the time in milliseconds⁶.

The total oral transit time (TOTT) was defined as the interval, in milliseconds, between the first frame showing the food inside the oral cavity to the first frame showing the proximal part (head) of the bolus in the hypopharynx or when the head of the bolus

makes angle with the mandible. For the pharyngeal transit time (PTT), what was considered was the presence of food in the posterior region of the nasal spine, located at the end of the hard palate, beginning of the soft palate, until complete passage through the superior esophageal sphincter⁴.

■ RESULTS

Once the quantitative analyses of total oral and pharyngeal transit times had been carried out we noted the values of 45.37 seconds for the TOTT and 4.53 seconds for the PTT. Associated with the quantitative measurement of oropharyngeal transit times, analysis was performed of the videofluoroscopic findings from this swallowing. We verified the presence of alterations in the uptake of food, change in the labial sphincter sealing, oral incoordination with increased oral transit time, abnormal pharyngeal swallowing response, causing posterior oral leak, as noted in Figure 1. Moreover, we found the presence of decreased laryngeal elevation and laryngeal penetration of the pasty food offered with no laryngotracheal aspiration (Figure 2).

■ DISCUSSION

Oropharyngeal swallowing depends upon a complex neuromotor mechanism, in which anatomical, physiological, and cognitive aspects all participate. Any change in one of these components may lead to impairment of oropharyngeal swallowing synchronic process, which may cause pulmonary complications, and problems with hydration, nutritional status and with social aspects of the individual⁷.

It is known that the Neuronal Ceroid Lipofuscinosis is a disease of degenerative character, which compromises neurological motor and cognitive functions, and may also change the oropharyngeal swallowing, however reports of this function in this population are scarce.^{1, 2, 3, 4}

In the case studied we find, both in the qualitative and the quantitative analyses, changes in the biomechanics of oropharyngeal swallowing. In the analysis of PTT and TOTT we find that both values are found higher than expected. According to the literature the TOTT is approximately 2 to 4 seconds, on average^{4, 8} for the pasty consistency. In the case studied this time was 45.53 seconds, ie well above the standard suggested by the literature, despite there being few studies of normality⁵.

The increase in TOTT can be attributed to many factors in the analysis of the biomechanics of swallowing. For some authors, the presence of cognitive abnormalities associated with lack of oral



Figure 1 – Videofluoroscopic image showing the presence of posterior oral leaking.



Figure 2 – Videofluoroscopic image showing the presence of penetration.

coordination, can exacerbate the performance of the oral phase of swallowing^{9,10}, such changes are common in this genetic disease¹¹.

Moreover, the use of alternative feed path for prolonged periods without partial oral feeding, such as in the case studied, tends to minimize oral swallowing training. The greater the usage time

of the alternative pathway and exclusive means, the greater the impact on performance of the oropharyngeal swallowing biomechanics during the execution of this function¹².

As for the PTT, we found in the literature that in most studies, few have been standardized in the infant population⁵, and the normal range was 1-2

seconds⁵. In the case of this individual with genetic diagnosis of NCL, PTT was 4.53 seconds. Although this swallowing time is above the normal range, this is less than the altered TOTT. This PTT may suggest that this change may even be suffering the impact of the severe impairment of the oral phase of swallowing this case being still efficient enough to protect the lower airway, since laryngeal penetration found in the consistency of food has not been tested frequently. Quantitative and qualitative analysis of the pharyngeal phase of swallowing suggests that the serious increase in TOTT may have caused delay in triggering the pharyngeal response, which consequently contributed to the increase in PTT this individual.

■ CONCLUSION

Accordingly, significant increase in oropharyngeal transit times were observed in this subject, and oropharyngeal dysphagia, a part of the phenotype of this syndrome, should be investigated and monitored during the course of the disease. Thus, we suggest that individuals with the genetic diagnosis of Neuronal Ceroid Lipofuscinosis be referred for the investigation of the oropharyngeal swallowing performance as soon as possible. The use of gastrostomy is necessary in the presence of severe impairment of TOTT, due to the impact of this change on the nutritional status, even in the absence of severe a case of laryngotracheal penetration or aspiration.

RESUMO

A presença de disfagia orofaríngea infantil na população com afecções genéticas ainda é pouco estudada. O objetivo deste estudo foi analisar o tempo de trânsito oral total (TTOT) e o tempo de trânsito faríngeo (TTF) em um indivíduo com diagnóstico genético clínico de Lipofuscinose Ceróide Neuronal (LCN) com disfagia orofaríngea grave. Indivíduo com LCN, 3 anos de idade, gastrostomizado há dois anos e sem via oral parcial, histórico de déficit de ganho de peso anterior a via alternativa de alimentação, porém sem complicações pulmonares. A deglutição orofaríngea foi estudada por meio de videofluoroscopia de deglutição e análise quantitativa da deglutição com uso de software específico para tal avaliação. Na análise quantitativa do TTOT e TTF constatou-se, respectivamente, 45,37 segundos (padrão de normalidade em criança é de 4 segundos) e de 4,53 segundos para o TTF. Constatou-se significativo aumento nos tempos de trânsito orofaríngeo neste indivíduo, sendo que a disfagia orofaríngea, parte do quadro desta criança com diagnóstico de LCN, deve ser investigada e acompanhada durante a evolução da doença. Uma avaliação da deglutição orofaríngea e acompanhamento nos indivíduos com esta condição genética deve ser realizada, considerando que essa alteração pode fazer parte do fenótipo desta condição e também pelo impacto que esse aumento nos tempos da deglutição pode ocasionar na condição nutricional e pulmonar desta população.

DESCRITORES: Lipofuscinoses Ceróides Neurais; Transtornos de Deglutição; Análise Quantitativa

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