ARTIGO ORIGINAL / ORIGINAL ARTICLE

CLINICAL EVALUATION OF OROPHARYNGEAL DYSPHAGIA IN MACHADO-JOSEPH DISEASE

Sabrina Mello Alves CORRÊA1, Valter Nilton FELIX1, Jonas Lírio GURGEL2, Rubens A. A. SALLUM1 and Ivan CECCONELLO1

ABSTRACT – Context - In Machado-Joseph disease, poor posture, dystonia and peripheral neuropathy are extremely predisposing to oropharyngeal dysphagia, which is more commonly associated with muscular dystrophy. Objective - To evaluate the clinical characteristics of oropharyngeal dysphagia in Machado-Joseph disease patients. Method - Forty individuals participated in this study, including 20 with no clinical complaints and 20 dysphagic patients with Machado-Joseph disease of clinical type 1, who were all similar in terms of gender distribution, average age, and cognitive function. The medical history of each patient was reviewed and each subject underwent a clinical evaluation of deglutition. At the end, the profile of dysphagia in patients with Machado-Joseph disease was classified according to the Severity Scale of Dysphagia, as described by O’Neil and collaborators. Results - Comparison between dysphagic patients and controls did not reveal many significant differences with respect to the clinical evaluation of the oral phase of deglutition, since afflicted patients only demonstrated deficits related to the protrusion, retraction and tonus of the tongue. However, several significant differences were observed with respect to the pharyngeal phase. Dysphagic patients presented pharyngeal stasis during deglutition of liquids and solids, accompanied by coughing and/or choking as well as penetration and/or aspiration; these signs were absent in the controls. Conclusions - Oropharyngeal dysphagia is part of the Machado-Joseph disease since the first neurological manifestations. There is greater involvement of the pharyngeal phase, in relation to oral phase of the deglutition. The dysphagia of these patients is classified between mild and moderate.


INTRODUCTION

Machado-Joseph disease (MJD) is a very common cerebellar ataxia and was initially described by Nakano et al.13) in a descended from Guilherme Machado, that emigrated from the Azores to Massachusetts, in the United States of North America (USA), afterwards moving on to other countries16, 26.

Nowadays many families without Portuguese descent are recorded as suffering from the disease. Japan has become one of the main center of the disease, which is also found in Brazil, India, China, Australia, Spain and France12.

MJD patients often display progressive cerebellar ataxia, external ophthalmoplegia (for eyelid retraction), pyramidal syndrome of spasticity (exaggerated reflexes), and extra pyramidal signals (postures and movements with an abnormal twist), dysarthria, distal muscle atrophy and small fasciculations of the face and tongue13.

The first signs of the disease are shown in the 25 to 55 year age group, with an average of 40 years. In 1993, Takiyama et al.20) located the MJD gene in the chromosome 14q (the MJD chromosome) and, in 1994, Kawagushi et al.9) identified the defective gene as an expansion of the sequence of the trinucleotide CAG repeat.

With application of molecular genetic technology, it is possible to establish the diagnosis of MJD with great sensitivity and precision11. Genomic DNA can be isolated from peripheral lymphocytes by conventional methods and the fragment containing the CAG repeat sequence of the MJD gene can be treated by polymerase chain reaction, using reaction agents DMJ52 and DMJ25. The reactive products in the polymerase chain are defined using gelatinous polyacrylamide plates 6%, in parallel with markers of known molecular weight. The sum of the differences of the various pairings is equal to T, the allele size, and the equation $N = \left[\frac{(T-121)}{3}\right]$ shows the number of repeated CAG.

Chronic back pain, persistent and annoying in its nature is a frequent MJD patients complaint, possibly due to central changes, particularly in the dopaminergic circuits tract and striatum of the diencephalon, the
processing of sensory impulses leading to the poor posture, dystonia and peripheral neuropathy\(^{(23)}\).

In classic literature dealing with this clinical disease, patients with MJD are considered normal in terms of their mental state\(^{(24)}\). However, Ishikawa et al.\(^{(40)}\) reported dementia and delirium in four patients with MJD who presented excitement, uncooperative attitude, crying, disorientation, a slow thought process, hallucinations, in which the sequence of CAGs repeated in the MJD chromosome were much longer than the average found in patients with the disease. The electroencephalography of these patients showed reduced brain activity, and computed tomography as well as magnetic resonance imaging of the skull ruled out cortical atrophy.

Poor posture, dystonia and peripheral neuropathy are extremely predisposing to oropharyngeal dysphagia, which is more commonly associated with muscular dystrophy. Thus, with the exception of those rare cases of cognitive impairment, patients with MJD, fully aware of what is happening to them, with evidence of dysphagia, have significant additional commitment to their quality of life.

Food, which not only supports the maintenance of life, is listed as one of the greatest pleasures which can be experienced by man, and this makes it possible to understand the problem that dysphagia causes for patients and their families\(^{(40)}\).

However, this problem is not commonly found in current literature, with works that only scratch the surface of deglutition of patients\(^{(17, 19)}\), or only describe cases\(^{(14, 22)}\), with conflicting data, motivating the creation of this, a prospective randomized survey, which aims to clinically categorize the dysphagia of patients with MJD, seeking to improve the interdisciplinary therapeutic approach.

**METHODS**

Twenty patients with MJD, previously diagnosed by the Neurology Department and confirmed by molecular testing, with moderate ataxia gait, external ophthalmoplegia, postural changes, presenting sporadic fasciculations of face and tongue, and intermittent low back pain, fit the clinical type I of the disease\(^{(21)}\), all of normal weight, but with clinical complaints of dysphagia since the onset of neurological manifestation, and 20 controls, individuals who did not report any complaints regarding the health and particularly of deglutition, were included.

The survey was conducted in the University of São Paulo Medical School, Department of Gastroenterology, São Paulo, SP, Brazil, and in the Laboratory of Research and Evaluation in Physical Activity, Catholic University of Rio Grande do Sul, Porto Alegre, RS, Brazil.

The patients had no other diseases, nor received any medication routinely, with the exception of analgesics.

The study was approved by the Scientific-Ethical Committees of both institutions and all participants signed informed consent forms.

The patients and the controls, of both sexes over 18 years of age with preserved cognitive responses, having been approved in the mini-exam of mental capacity\(^{(13)}\), had attended at least until the fourth grade of elementary school and had not previously undergone any deglutition rehabilitation programme.

Twenty patients with MJD, complaining of high dysphagia, and 20 controls equated in terms of gender distribution and average age (Table 1). The MJD patients were submitted to anamnesis and all the participants in the survey, both patients and controls, underwent clinical evaluation of their deglutition.

**TABLE 1. Material**

<table>
<thead>
<tr>
<th></th>
<th>Age (years)</th>
</tr>
</thead>
<tbody>
<tr>
<td>MJD Patients</td>
<td>44.75 ± 10.37</td>
</tr>
<tr>
<td>Controls</td>
<td>38.55 ± 11.36</td>
</tr>
</tbody>
</table>

**Anamnesis**

The anamnesis consisted of questioning concerning the time since the MJD diagnosis and the appearance of dysphagia, the presence or absence of coughing or choking during or after meals, the feeling of food stuck in the throat.

Average meal times were also investigated as so as if there had been weight loss or bronchial aspiration after the onset of dysphagia, which food textures proved easier or more difficult to swallow, if the eating itself was pleasurable and if patients fed by themselves, totally independently or with the help of others.

**Clinical evaluation of deglutition**

A clinical evaluation was carried out on all subjects, using latex gloves, flashlight, plastic spatula, Littman stethoscope\(^9\) and portions of 10 mL of foods: liquid (water/ juice), paste (yogurt) and solid (biscuit).

By means of visual observation, the ability of lip sealing and bilateral lateralization, protrusion and retraction of the tongue were all observed. The tongue tonus was assessed by a motor test, which asked the patient to externalize the tongue as much as possible and resist the force from the spatula. Using spatula, the sensitivity of the forehead, cheeks, chin, and of tip, body, bottom and side portions of the tongue (which was left in the intra-oral resting position) and the gag, cough, palatal and bite reflexes were all tested.

Cervical auscultation was adopted for the clinical evaluation of the pharyngeal phase of deglutition. The stethoscope was positioned on the left side of the neck, the region corresponding to the laryngotracheal junction, anterior to the carotid artery, in order to hear the effectiveness or not of the mechanism for airway protection and if the transit of the bolus was normal or not.

Pharyngeal stasis was identified when there was increased duration of deglutition apnea and multiple gulps at cervical auscultation.

Besides this, the presence or absence of cough and/or choking during the intake of each element swallowed was observed. The issue of high-pitched sounds was asked to evaluate if there was preserved ability of laryngeal elevation.

At the end, the profile of dysphagia in patients with Machado-Joseph disease was classified according to the Severity Scale of Dysphagia, as described by O’Neil et al.\(^{(15)}\).
**Statistical analysis**

Data was analyzed using the c² test, adopting a significance level of 5%.

**RESULTS**

In the anamnesis, the MJD patients showed high frequency of coughing or choking during the meal and feeling of food stuck in the throat and it could be observed that the food with the paste texture was easier to swallow and is the preference of all dysphagic patients (Table 2).

<table>
<thead>
<tr>
<th>TABLE 2. Anamnesis of the MJD patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Average disease and dysphagia time</td>
</tr>
<tr>
<td>Coughing or choking during meals</td>
</tr>
<tr>
<td>Coughing or choking after meals</td>
</tr>
<tr>
<td>Feeling of food stuck in the throat</td>
</tr>
<tr>
<td>Average meal time</td>
</tr>
<tr>
<td>Weight loss</td>
</tr>
<tr>
<td>Bronchial aspiration</td>
</tr>
<tr>
<td>Paste food texture easier to swallow</td>
</tr>
<tr>
<td>Liquid food texture more difficult to swallow</td>
</tr>
<tr>
<td>Solid food texture more difficult to swallow</td>
</tr>
<tr>
<td>Pleasure of eating</td>
</tr>
<tr>
<td>Independence in feeding habits</td>
</tr>
</tbody>
</table>

Weight loss was no more than 3 kilograms, most of the sufferers were able to maintain independence in feeding habits and indeed got pleasure from it, but all of them stressed the additional effort and excessive time taken to ingest meals since the beginning of the neurological condition.

With reference to the clinical evaluation of the oral phase of deglutition, comparison with the controls showed significant differences only in regards to less protrusion and retraction ability and higher tonus of the tongue of the patients (Table 3).

**DISCUSSION**

Patients with oropharyngeal dysphagia commonly present muscle dysfunction in the lips, tongue, palate, and/or pharynx, as well as incoordination of deglutition movements. The controls did not show any changes whatsoever to the clinical evaluation of deglutition, in both the oral phase and that of the pharyngeal, but as was expected, according to the classic features of MJD, the dysphagic patients demonstrated deficiencies in the oral phase of deglutition, related to the protrusion, retraction and tonus of the tongue, which, however, had adequate sensitivity and lateral mobility, with the lips closing and reflexes maintained throughout the various tests performed.

As for the analysis of the pharyngeal phase, cervical auscultation is a tool that enables the hearing of the distinctive sounds of deglutition, with the use of a stethoscope. The sounds resulting from the movement of food towards the airway are different from those of a normal deglutition.

Cervical auscultation is inexpensive, noninvasive and easy to use. Cervical sounds of adult and infant swallowing have been qualitatively described. It is a method of listening to the sounds of swallowing during the pharyngeal phase to detect patients with dysphagia. Typically, a stethoscope is placed at the lateral aspects above the cricoids cartilage in front of the sternocleidomastoid muscle and the large vessels.

When clinicians listen to the swallowing sounds with a stethoscope, qualitative judgments are made about what is heard. Based on the sharpness of the sound, the characteristic swishing double-click as the bolus passes through the pharynx and into the esophagus is judged for normalcy. However, all recordings from dysphagic patients show multiple gulps.

However, with regards to the pharyngeal phase, there were several significant differences noted, with dysphagic patients presenting pharyngeal stasis at the deglutition of liquids and solids, often accompanied by coughing and/or choking and penetration or aspiration (Table 4).

In the same table it is observed that all subjects showed, at the issue of high-pitched sounds, preserved ability of laryngeal elevation, and the deglutition of paste not resulting in pharyngeal stasis, coughing, choking, signals of penetration or aspiration of food.

Considering the data coming from the history of patients and the variables of clinical evaluation where statistically significant differences in the ratio patients/controls could be seen, the profile of the dysphagia of patients with MJD was compatible with the level 4 (mild/moderate dysphagia) on the O’Neil et al. Severity Scale of Dysphagia.
Activamente continuaram esforços terão sido efetuados neste campo, focando na prestação objetiva, mensurável e reproduzível de técnicas para aumentar a aceitação e utilidade da ausculta cervical na pesquisa e aplicações clínicas(2).

Pode ser notado que os pacientes disfáicos apresentaram distúrbios mais marcados, com base em estatísticas, em relação aos controles, no deglutição de líquidos e sólidos, com estase faríngea, com tosse e/ou engasgo, acompanhada de penetração e/ou aspiração, típica de disfagia moderada(5), que são ausentes nos controles, embora a laringe esteja elevada efetivamente, refletindo um des coordenação de movimentos das estruturas envolvidas no processo.

Foi clara a final de avaliação clínica, por que as pastas, como mencionado no anamnese, foram preferidas pelos pacientes, uma vez que não promoveu estase faríngea, a presença de tosse e/ou engasgo ou sinais de penetração ou aspiração se exibida.

O Severity Scale of Dysphagia descrito por O’Neil et al.(15), que classifica disfagia do 7 (deglutição normal) a 1 (disfagia grave), considera que a restrição de dois ou mais consistências de comida, e penetração e/ou aspiração são presentes em disfáicos e ausentes em controles, embora a laringe esteja elevada efetivamente, refletindo um des coordenação de movimentos das estruturas envolvidas no processo.


DESCRITORES - Doença de Machado-Joseph. Transtornos de deglutição.
REFERENCES