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Case reports

A longitudinal study of the progression of Huntington's disease and oropharyngeal dysphagia: A four-case follow-up

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ABSTRACT

Huntington's disease (HD) is a neurodegenerative disorder, with dysphagia being a common symptom of the disease. Few studies established a relationship between neuromotor impairment and dysphagia. There is also a lack of described therapeutic approaches for dysphagia in HD. This study aimed to better understand the progression of neurological clinical aspects, instrumental swallowing and dysphagia management in four patients presented with HD in an outpatient follow-up setting. The longitudinal follow-up period was 36 to 43 months through neuroclinical assessments (Unified Huntington's Disease Rating Scale) and fiberoptic endoscopic swallowing evaluations. Case 1 – a moderate decline of independence with safe swallowing. Case 2 – a moderate motor impairment, safe swallowing and moderate dysphagia during follow-up. Case 3 – a longer disease duration and increased motor impairment associated with moderate/severe dysphagia. Case 4 – a longer disease duration, need of assistance to perform daily activities, severe dysphagia and palliative care, considering the family decision of exclusive oral feeding. This longitudinal study revealed that the progression of neuromotor damage was not directly related to dysphagia development. This case series demonstrates the importance of follow-up settings with instrumental swallowing evaluations and careful consideration of early palliative care for HD patients.

Keywords: Deglutition Disorders; Huntington Disease; Rehabilitation; Neurodegenerative Diseases

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INTRODUCTION

Huntington's disease (HD) is a rare, inherited, neurodegenerative genetic disorder that appears between the ages of 35–55 years[1,2]. HD is caused by the expansion of a CAG trinucleotide repeated in the *IT15* gene on chromosome 4[3,4]. Although disease progression is variable, its outcome is fatal, and the average survival is 15–20 years after diagnosis[3]. Clinical symptoms and signs include progressive and nonlinear motor, cognitive, and behavioral impairments that result in complete dependence. There is no treatment capable of reversing the disease, thus, symptom management is currently the only available treatment option[5,6].

Oropharyngeal dysphagia is a symptom of HD and is characterized by changes in the preparatory, oral, and pharyngeal phases of swallowing that lead to malnutrition, dehydration, and bronchoaspiration[7,8]. Aspiration pneumonia is the leading cause of death in later stages[6,9]. Although dysphagia is a common symptom in HD, its progression remains incompletely understood, since few longitudinal studies have been conducted with this objective. Investigations that focus on oropharyngeal dysphagia progression are essential for improving the management of complications associated with morbidity and mortality in HD. The present study aimed to describe the characteristics of swallowing and the neurological and clinical progression in four patients with HD, followed in an outpatient clinic.

CASE REPORT

The study was approved by the Research Ethics Committee of the State University of Campinas - UNICAMP, SP, Brazil, which deals with ethics and guidelines for research involving human beings, with the assent of 95284718.3.0000.5404. All participants and/or guardians signed the informed consent form. This is a longitudinal study of four patients diagnosed with HD, aged between 54 at 68 years, who went through follow-up at the neurology and dysphagia outpatient clinic of a tertiary care hospital.

Data collection took place at four-time points, with an interval of 6 months to 1 year between them. Participants underwent a fiberoptic endoscopic evaluation of swallowing (FEES) and neurological evaluation (data of motor, behavioral performance and independence using the Unified Huntington's Disease Rating Scale – UHDRS). All procedures, in each of the periods, were performed in the same week or with a maximum interval of 1 week. Participants were followed up for 36 to 43 months (mean, 39.6 months). Other clinical data included genetic data, family history, sex, age, time of symptom onset, time of diagnosis, dysphagia symptoms, and performance of speech therapy.

The FEES was submitted by an otorhinolaryngologist accompanied by a speech therapist. The test protocol consisted of blue-stained food in the following consistencies: thin liquid (International Dysphagia Diet Standardization Initiative [IDDSI][10] level 0), nectar (IDDSI level 2), honey (IDDSI level 3) and pudding (IDDSI level 4). These consistencies were obtained by adding two, three, and four teaspoons, respectively, of thickener [Thicken-easy®] to 100 mL of water. Two different quantities (3 and 7 mL) of each consistency were tested. Solid consistency (IDDSI 7) was also evaluated, using cornstarch cookies. As the participants presented difficulties in swallowing, airway protection maneuvers and/or changes in head posture were performed to promote safe oral feeding. The FEES results were judged by two speech therapists and an otorhinolaryngologist.

The Penetration-Aspiration Scale (PAS)[11] was used for the classification of penetration and/or aspiration, considering a range of levels from 1 to 8. The functionality of swallowing was classified using the Functional Oral Intake Scale (FOIS)[12] (1 to 7 - total oral intake) and the classification of the dysphagia severity was performed using the Dysphagia Outcome and Severity Scale (DOSS)[13] (1 to 7 – normal diet, no strategies).

Demographic and clinical information of four patients with HD followed up longitudinally will be presented.

RESULTS

The four cases will be presented below, along with information regarding the demographic data of each patient, as well as the histories of neurological assessments and swallowing.

Clinical information of each case is organized in a table format to aid in visualizing the clinical findings, including sex, age, duration of illness, age of onset of initial symptoms, number of CAG repetitions, family history, details of the initial swallowing assessment, UHDRS scale data at the initial assessment, and longitudinal follow-up duration (Table 1).

Table 1. Characterization of Huntington's disease patients with clinical and demographic data

Participants	Age	Time of the disease	Sex	Age at first symptom	CAG	HD family history	Swallowing*	UPHRS *	Longitudinal follow-up
Case 1	58	6	M	52	42	Grandfather, two aunts	Feeding orally and without consistency restriction, complaining of choking on liquids	Motor: 40 points; Independence: 90; Behavior: 4	3 years and 4 months
Case 2	64	7	M	57	39	Four sisters, father and grandmother	Feeding with increased speed and coughing with solids	Motor: 27 points; Independence: 100; Behavior 0 points	3 years and 6 months
Case 3	68	14	F	54	38	A sister, father and grandmother	Feeding with increased speed and coughing while eating	Motor: 20 points; Independence: 90; Behavior: 4	3 years and 3 months
Case 4	66	25	F	41	42	Two brothers, father, two uncles, grandmother and others	Difficulty feeding in all consistencies. The patient and her family decided for exclusive oral feeding, despite the indication of alternative feeding.	Motor: 60 points; Independence: 20; Behavior 60.	3 years and 7 months

Captions: Male = M; Female = F; Huntington's disease = HD; cytosine-adenine-guanine = CAG repeat expansion; Unified Huntington's Disease Rating Scale = UPHRS;
* Data regarding the First assessment

Case 1

In Table 1, the clinical information of Case 1 is presented.

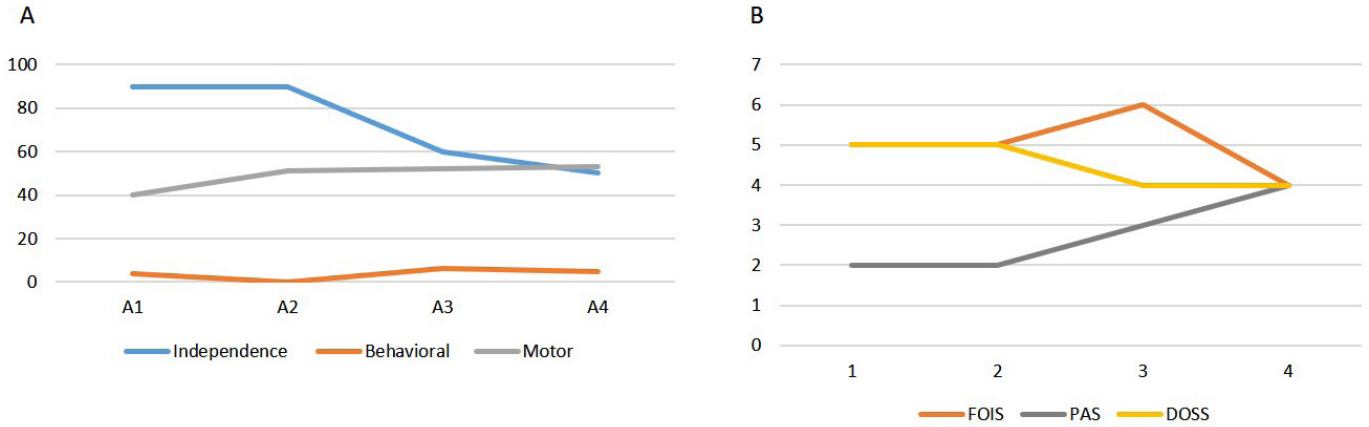
Figure 1 represents the progression of HD, related to the UHDRS scale, in four moments of the longitudinal evaluation, with a slight decline in motor function and further evidence of independence.

Figure 1 B shows the data regarding the patient's performance in the FEES with the worsening of PAS, DOSS, and FOIS scores with exclusive oral feeding at the follow-up. During the FEES maneuvers and interventions, aspects of swallowing were tested, which can be seen in Chart 1. These strategies were aimed at caregivers who performed the training at home. The patient was monitored weekly by the speech therapist.

Chart 1. Therapeutic strategies used at the four time points in all patients (N=4)

	1	2	3	4
Case 1	- Postural adequacy - Maneuvers: HD, MD, DE	- Postural adequacy - Maneuver: MD - Volume and speed control	- Maneuver: HD - Volume and speed control	- Maneuvers: HD, MD - Volume and speed control
Case 2	- Maneuvers: HD, MD - Volume control - Tongue counter resistance exercise and strengthening of the orbicularis oris muscle	- Maneuvers: MD, DE - Volume and speed control	- Maneuvers: HD, MD - Volume control - Ejection exercises of the bolus and lip seal	- Maneuver: DM - Volume and speed control. - Laryngeal lifting exercise and increased ejection force with counter resistance
Case 3	- Volume and speed control, - Posture adequacy	- Speed and volume control - Liquid and nectar in the straw, - Supraglottic maneuver, Mendelsohn	- Maneuver: HD - Volume and speed control	- Maneuver: MD - Volume and speed control
Case 4	-Volume and speed control, drinking liquid using a straw	- Volume and speed control	- Maneuver: MD - Volume and speed control	- Maneuver: MD - Volume and speed control, macerated medication and mashed food

Captions: HD = Head Down; MD = Multiple Deglutition; DE = Deglutition Effort



Captions: UPHRS = Unified Huntington's Disease Rating Scale; FOIS = Functional Oral Intake Scale; DOSS = Dysphagia Outcome and Severity Scale; PAS = Penetration-Aspiration Scale

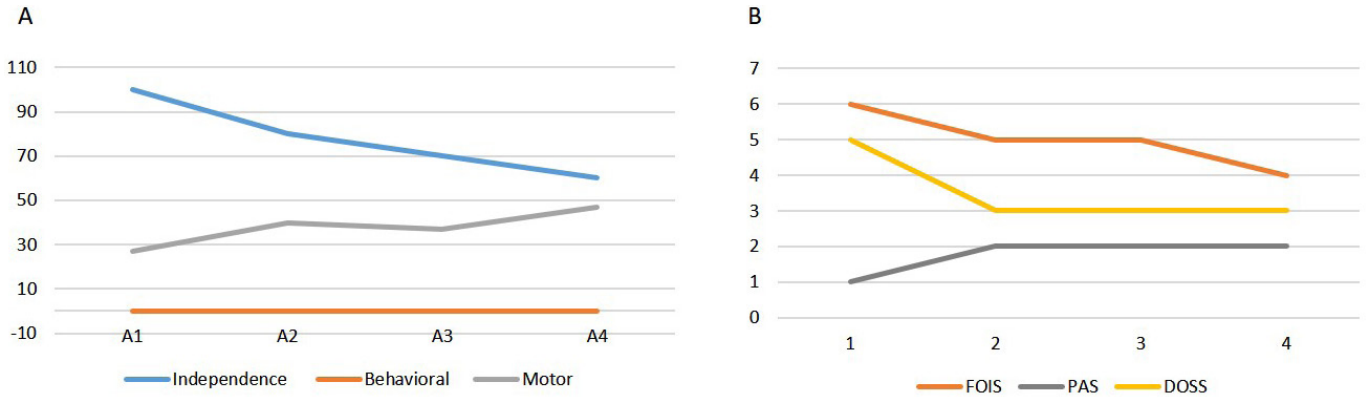
Figure 1. Case 1 - **A.** Neurological scales (UHDRS): Motor score, Independence and Behavioral in the four evaluation moments. **B.** Swallowing scales: FOIS, PAS and DOSS in the four evaluation moments.

Case 2

In Table 1, the clinical information of Case 2 is presented.

Figure 2 A presents the progression of HD, related to the UHDRS scale, in four moments of the longitudinal evaluation, with a decline in independence and motor function.

Figure 2 B shows the data regarding the patient's performance in the FEES with maintenance of penetration (considered normal), with a decline in DOSS and exclusive oral feeding (FOIS 4). During the FEES maneuvers and interventions, aspects of swallowing were tested, which can be seen in Chart 1. These strategies were aimed at caregivers who performed the training at home and the patient was monitored weekly by the speech therapist.



Captions: UPHRS = Unified Huntington's Disease Rating Scale; FOIS = Functional Oral Intake Scale; DOSS = Dysphagia Outcome and Severity Scale; PAS = Penetration-Aspiration Scale

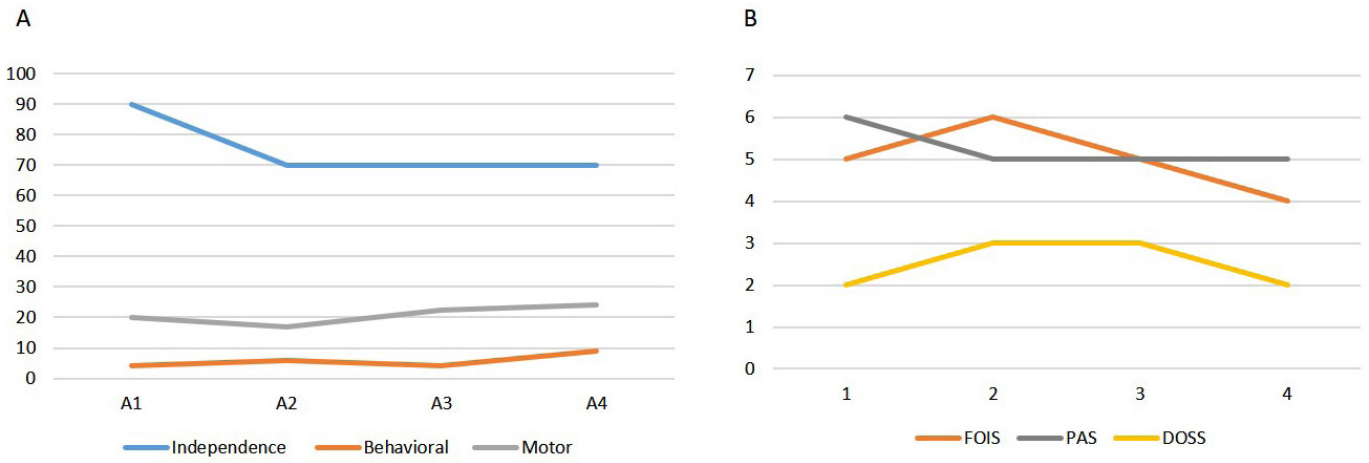
Figure 2. Case 2 - **A**. Neurological scales (UHDRS): Motor score, Independence and Behavioral in the four evaluation moments. **B.** Swallowing scales: FOIS, PAS and e DOSS in the four evaluation moments of the four moments of the follow up.

Case 3

In Table 1, the clinical information of Case 3 is presented.

Figure 3 A presents the progression of HD, related to the UHDRS scale, in four moments of the longitudinal evaluation, with a slight decline in independence and maintenance of motor function.

Figure 2 B shows the data regarding the patient's performance in FEES with the presence of aspiration. However, consistency management yielded controlled DOSS and FOIS scores. During the FEES maneuvers and interventions, aspects of swallowing were tested, which can be seen in Chart 1. These strategies were aimed at caregivers who performed the training at home and the patient was monitored weekly by the speech therapist.



Captions: UPHRS = Unified Huntington's Disease Rating Scale; FOIS = Functional Oral Intake Scale; DOSS = Dysphagia Outcome and Severity Scale; PAS = Penetration-Aspiration Scale

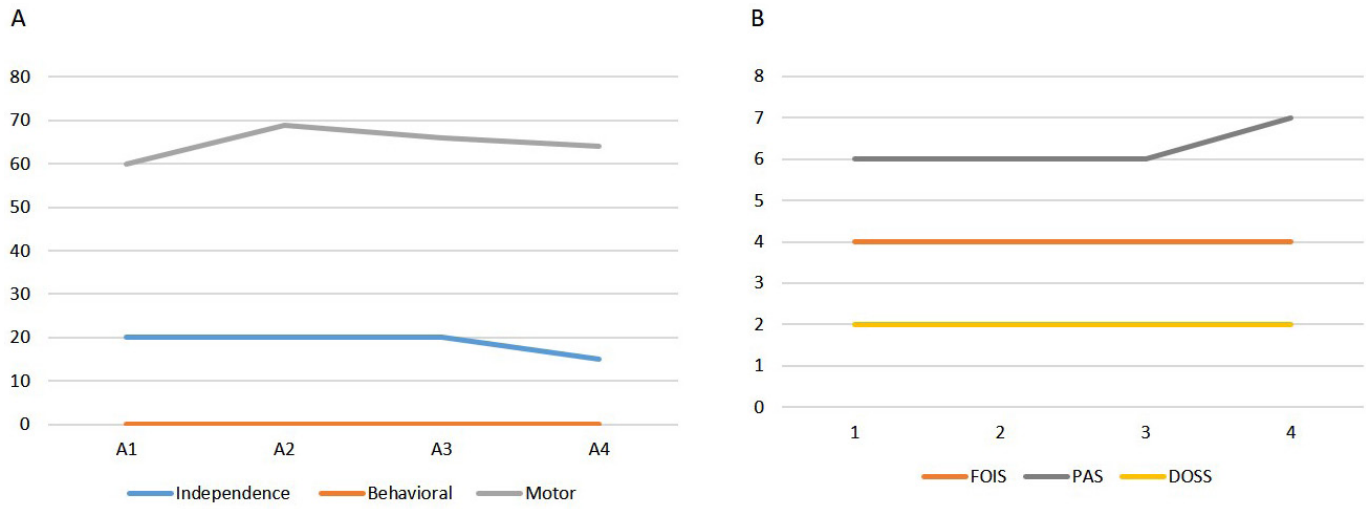
Figure 3. Case 3 - **A**. Neurological scales (UHDRS): Motor score, Independence and Behavioral in the four evaluation moments. **B.** Swallowing scales: FOIS, PAS and DOSS in the four evaluation moments of the four moments of the follow-up.

Case 4

In Table 1, the clinical information of Case 4 is presented.

Figure 4 A illustrates the progression of HD related to the UHDRS scale at four points in the longitudinal evaluation, showing maintenance of motor function, behavior, and independence.

Figure 4 B shows the data regarding the patient's performance in the FEES with the maintenance of PAS, DOSS, and FOIS scores, along with time of the disease and severity of the disease. During the FEES maneuvers and interventions, aspects of swallowing were tested, which can be seen in Chart 1. These strategies were aimed at caregivers who performed the training at home and the patient was monitored weekly by the speech therapist.



Captions: UPHRS = Unified Huntington's Disease Rating Scale; FOIS = Functional Oral Intake Scale; DOSS = Dysphagia Outcome and Severity Scale; PAS = Penetration-Aspiration Scale

Figure 4. Case 4 - **A**. Neurological scales (UHDRS): Motor score, Independence and Behavioral in the four evaluation moments. **B.** Swallowing scales: FOIS, PAS and DOSS in the four evaluation moments of the four moments of the follow-up.

DISCUSSION

This longitudinal follow-up of four patients with HD showed an absence of a direct relationship between motor, behavioral, and independence aspects with dysphagia progression. By comparing the data from the first to the last evaluation, a worsening of the swallowing pattern, represented by the DOSS, FOIS, and PAS scales, can be verified. The follow-up showed progression of independence (cases 1 and 2) and motor scales (case 3), as well as swallowing safety (case 1), dysphagia severity (case 2) and functional oral intake (cases 1, 2 and 3) Case 4 presented advanced disease and severe dysphagia with stabilization of FOIS and DOSS. This adds deglutition to the discussion that has already been brought forth by other investigators regarding the nonlinear path of motor decline, although disease progression is inevitable. However, the swallowing function may be associated with the decline, as well as other clinical factors, such as age, time of disease, and number of GAG repeats[9,14].

During the FEES maneuvers, some aspects of swallowing were evaluated to observe their effectiveness in improving the efficiency and safety of patient swallowing. Maneuvers used by the four participants were multiple swallows, effortful swallowing, and chin-tuck position during swallowing of liquids. Mendelsohn and supraglottic maneuvers were tested as rehabilitation maneuvers; however, the results were negative due to the difficulty in the patient's understanding and performance due to behavior and cognitive decline. Thus, intervention strategies used with the participants aimed to establish swallowing biomechanics with reduced risk of broncho-aspiration and provide safe feeding to prolong oral feeding time. Longitudinal follow-up of patients also allowed the identification of therapeutic strategies for swallowing. Literature remains scarce regarding rehabilitation of

dysphagia in HD, the management of which usually combines behavioral approaches and compensatory strategies[8,15]. The indication of therapeutic strategies, however, depends on progression of the disease and the person's ability to understand and execute strategies correctly[1,6].

Patients in the initial phase of HD usually present mild cognitive and motor deficits, with less interference in feeding management[6]. In the initial phase of HD, as also verified in patients in this study, patients benefit from therapeutic intervention consisting of exercises that favor swallowing biomechanics[8,9,16,17]. As the disease progresses, strategies such as changes in oral diet and compensatory maneuvers become more appropriate.

In all participants, changes in the oral bolus control and the presence of tachyphagia were observed and compensatory strategies were used to reduce volume and speed during feeding. Only one patient (case 2, with 7 years of disease) was prescribed lip-sealing exercises, bolus ejection, and laryngeal elevation exercises. Although this patient had a disease duration of around six years, the patient also had a smaller number of CAG repeats (39 repetitions). It is known that the number of CAG repeats is strongly associated with a higher rate of global decline, owing to the more pronounced brain damage[1].

In the follow-up of the cases, strategies described by Kindell (2002)[18] were recommended, such as offering small portions of each type of food with intervals between them and offering softer foods with the use of visual reminders to reduce speed[19]. Volume was controlled by offering the patient smaller portions, with adaptations of utensils (smaller spoons) and auditory and visual cues during swallowing management.

Head and neck postural adequacy were performed in all patients, since patients with HD present hyperextension of head and trunk and choreic movements during feeding, which increases the risk of solid and liquid bronchoaspiration[17]. A strategy that can be used during swallowing training is the chin-tuck postural maneuver, which is favored by placing the tray on the person's lap so that the plate is at waist height. Another possibility is the use of a positioning wedge to stabilize the thoracic and lumbar spine of the patient to reduce trunk hyperextension. Three of the patients who were followed up in this study required strategies to control head positioning[9,20].

Considering the general scenario of the disease and its outcome, we emphasize the need for multidisciplinary follow-up in HD to safely maintain an oral diet for as long as possible. The decreased independence and swallowing functionality, identified in all patients in this study over 3 years, show the importance of discussing palliative care[21].

FINAL CONSIDERATIONS

This longitudinal study revealed the progression of neurological and swallowing aspects and absence of a direct relationship between motor, behavioral, and independence aspects with dysphagia progression. The main objective of dysphagia intervention in HD should be maximizing the maintenance of safe oral feeding for as long as possible. Therefore, discussing with the family and multidisciplinary team is essential for improving the quality of life, while respecting patients' wishes, before dementia symptoms emerge.

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Author Contributions

AP: Conceptualization; Formal analysis; Methodology; Writing - Original draft; Writing - Review & editing.

DPL: Data curation; Funding acquisition; Investigation.

MABL: Data curation; Formal analysis; Writing - Review & editing.

PCA, LFM: Conceptualization; Data curation; Formal analysis; Methodology; Writing - review & editing.