Dysphagia
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REVIEW ARTICLE

Dysphagia in Huntington's Disease: A Review

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Abstract Huntington's disease (HD) is a progressive neurodegenerative autosomal dominant disease characterized by disturbed movements and behavior and cognitive decline. The motor disturbances are both choreiform and hypokinetic. As a result of the combination of these signs, it is known that many patients with HD suffer from dysphagia. Little is known about the frequency and the characteristics of dysphagia in HD. Well-balanced strategies for treatment and prevention of dysphagia in HD are lacking. Therefore, we have performed a detailed survey of the literature. We found that the patient groups studied were heterogeneous and the methods used were highly variable, and no balanced advice for prevention and treatment was systematically proven.

Keywords Huntington's disease · Dysphagia · Deglutition · Deglutition disorders

Huntington's disease (HD) is a progressive neurodegenerative autosomal dominant disease characterized by disturbed movements and behavior and cognitive decline. The motor disturbances are both choreiform and hypokinetic. Choreiform movements are irregular and involuntary in HD and involve not only limbs but respiratory and bucolingual muscles, comparable to dancing (*chorea* = dancing in Greek). HD is caused by a CAG repeat expansion of the HTT gene on the short arm of chromosome 4. The mutant protein huntingtin causes neurodegeneration in the brain, particularly in the caudate nucleus and putamen. Onset of HD on average occurs in the third or fourth decade of life and lasts about 15–20 years [1–8]. Death often results from aspiration pneumonia caused in turn by a progressive dysphagia [9–12]. Previous studies into dysphagia in HD investigated dysphagic features in the different phases of ingestion [13–17]. It is not known in what stage of HD the dysphagia becomes clinically apparent. The frequency of this incapacitating sign is not known. No well-proven strategies to prevent dysphagia are available. Therefore, we reviewed the literature on this clinically important sign in HD, looking for different methods and interventions to find a better strategy for treatment and prevention.

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Methods

Literature searches were performed using OVID Medline (1985–2009), OVID Embase (1985–2009), Omega (1985–2009), PubMed (1985–2009), and EBSCO Cinahl (1985–2009) to identify evidence about the theme of dysphagia in HD. Search terms included aerophagia, aspiration, asphyxiation, chest infection, choking, cineradiography, coughing, deglutition disorders, drinking, dysarthria, dysphagia, eating, drinking, fluoroscopy, food, ingestion, laryngeal chorea, mastication, penetration, phases of ingestion, pneumonia, swallowing, tongue, voice, and voice disorders in HD. The relevant studies [13–17] about dysphagia in HD were evaluated and summarized (Appendix). Studies were relevant if they were directly linked to swallowing, phases of ingestion, and dysphagia in HD; otherwise, they were excluded, such as studies about nutrition and dysarthria. To create a clear picture about the

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Table 1 Phases of ingestion [18]

Preparatory oral	Oral	Pharyngeal	Esophageal
Transport to the mouth	Bolus positioning	Velum elevation and retraction	Peristaltic
Lip closure	Undulation tongue	Velopharyngeal closure	Esophageal transit time (8–20 s)
Mastication	Triggering of pharyngeal swallow	Hyoid and laryngeal elevation	
Tongue rotation	Coating of pharyngeal wall	Total laryngeal closure	
Tongue thrust	Oral transit time (<1–1.5 s)	Cricopharyngeal opening	
Tongue-to-palate contact		Reduce tongue base	
Bolus formation		Contraction pharyngeal constrictors	
		Pharyngeal transit time (<1.0 s)	

overall conclusions of dysphagia in HD according to the literature, the dysphagic features were divided into the four phases of ingestion [18], i.e., preparatory oral, oral, pharyngeal, and esophageal (Table 1).

Results

Five studies [13–17] directly linked to swallowing and dysphagia in HD were found (Appendix). Three studies were about dysphagia in HD [13–15], one case study was about laryngeal chorea in relation to swallowing [16], and one case study was about oropharyngeal dysphagia in HD, measuring the oral transit time, pharyngeal transit time, and esophageal response time [17]. The studies used several objective methods and techniques. Leopold and Kagel [13] used a detailed diagnostic examination. An unweighted dysphagia scale of 0–5 was formulated based on abnormalities of ingestion that were observed during the clinical and radiologic examinations. In this article, a mixed group of HD patients were studied. Hunt and Walker [14] described nursing interventions for dysphagia in HD. The nursing interventions were mealtime tips, suggestions for an environment in which to eat, appropriate aids for stage of disease, food choices and supplements, and methods to control dysphagia. In this study, three undefined patients with HD were illustrated. In another study, Kagel and Leopold [15] used the Crozer-Chester Medical Centre Dysphagia Centre evaluation. In addition, a videofluoroscopic swallowing study was used. Mochizuki et al. [16] used barium cineradiologic examination for laryngeal chorea in HD. The dysphagia was not described in detail: the only characteristic that was given referred to a cough at the beginning and during the swallow. The specific misdeglutition per phase of ingestion according to the cineradiologic examination was not described. Hamakawa et al. [17] employed two procedures to assess dysphagia: one was a general view of self-feeding and the other used videofluorography.

The overall conclusion one can draw from these studies is that dysphagic features occur especially in the preparatory oral, oral, and pharyngeal phases of ingestion. The main problems in the preparatory oral phase are the postural instability, rapidly and impulsively consuming food, and poor lingual control. In the oral phase, the main problems are uncoordinated swallow, repetitive swallow, and residue after the swallow. In the pharyngeal phase the main problems are coughing, choking, and aspiration (Table 2).

Discussion

The techniques used to examine dysphagia in HD are different and only some can be beneficial for patients; therefore, their outcomes can be somewhat questionable. For example, along with the videofluoroscopic swallowing study, Kagel and Leopold [15] used compensatory techniques like reducing chorea by manually positioned extremities, a wedge supporting the midthoracic to lumbar spine, and the clinician giving cues to help the patients maintain an erect trunk and neutral neck postures. Furthermore, before the patients ingested a test substance, the patients were fed a lemon ice bolus. This lemon ice bolus was intended to stimulate oropharyngeal sensory systems and slow oropharyngeal movements. Logemann et al. [19] measured the effects of a sour bolus on oropharyngeal swallowing in patients with neurogenic dysphagia. Results showed that there was significant improvement in oral onset of the swallow and a significant reduction in pharyngeal swallow delay and in frequency of aspiration with the sour bolus [19]. Pelletier et al. [20] examined the effect of citric acid (2.7%) on swallowing. Citric acid (2.7%) significantly reduced aspiration and penetration compared with water. Furthermore, a significant increase in spontaneous dry swallows was observed after the taste stimuli. Another compensatory technique used was placing the soft and solid foods directly on the lateral molars. This can be beneficial to the patient because he/she does not have to transport the soft and solid foods from the lips to the lateral molars. In spite of all these compensatory techniques, patients still showed a lot of swallowing problems.

Table 2 Overall conclusions of previous studies on dysphagia in HD

Preparatory oral	Oral	Pharyngeal	Esophageal
Postural instability [15, 17]	Impaired voluntary swallowing [13]	Coughing [13–17]	Vomiting [13, 15]
Abrupt postural changes caused flux to the pharynx [15]	Swallow incoordination [15]	Choking [14, 15]	Early satiety [15]
Hyperextension of head and trunk [13, 15]	Short oral transit time (0.23 s) [17]	Aspiration [13–15]	Abnormal esophageal motility [13, 15]
Difficulty controlling rate and amount of food intake [13]	Repetitive swallows [15]	Eructations [15]	Diaphragmatic chorea [15]
Rapidly and impulsively consuming food [13, 17]	Swallow latency [15]	Aerophagia [13, 15]	Reflux [15]
Tachyphagia [15]	Residue after swallowing solid foods [13, 17]	Audible swallows [15]	
Inadequate mastication [13–15, 17]	Intraoral bolus retention [15]	Prolonged laryngeal elevation [15]	
Poor lingual control [14, 17]	Segmented lingual transfer [15]	Difficulty descent larynx [16, 17]	
Tongue protrusion [16]		Phonation during swallow [15]	
Premature liquid transfer [15, 17]		Pharyngeal stasis [13, 15, 17]	
Delayed lingual transfer [15]		Impaired cricopharyngeal function [13]	
Lingual chorea [15]		Inability to stop respiration [15]	
		Wet vocal quality [15]	
		Laryngeal chorea [15, 16]	
		Epiglottis tilt not inferoposterior [17]	

In our experience we have found similar abnormalities in all the swallowing problems summarized in Table 2. However, we think much depends on the stage of HD. Unfortunately, all five studies used a mixed group of HD patients and none systematically examined dysphagia through the successive stages of HD. Therefore, the specific dysphagic features in HD and the prevalence of dysphagia in HD are not known. In our experience, patients in the early stages of HD may develop overt dysphagia. Furthermore, we think we can delay symptomatic dysphagia with early and regular intervention from a speech and language therapist. Compensatory techniques seem to help when patients are treated early, even before the dysphagia starts. It is necessary to give information about dysphagia and its consequences. Dysphagia is potentially a frightening issue for patients, particularly because many remember other family members having swallow difficulties or may have seen someone aspirate. For these reasons, the information must be introduced in a sensitive manner, and discussion about current problems should be encouraged. None of the published studies discussed these important points. Because of the serious consequences of dysphagia in HD, namely, pneumonia and acute respiratory distress and subsequent death [9–12], it is very important to assess and measure the dysphagic features as soon as possible. By investigating dysphagia at the early stages of the disease and following patients as the disease progresses, a more accurate categorization of dysphagia can be established that may lead to its proper management. To date no such studies exist in part because dysphagia is not recognized in the Unified Huntington's Disease Rating Scale (UHDRS), an internationally used research scale that assesses the clinical features and course of HD [21]. Therefore, at first it is necessary to develop a validated dysphagia assessment scale for HD patients. Manor et al. [22] developed a Swallowing Disturbance Questionnaire (SDQ) for Parkinson's disease (PD). This questionnaire asks about experiencing swallowing disturbances and compares its findings to an objective assessment. This SDQ emerged as a validated tool to detect early dysphagia in PD patients [22]. Since some important issues are missing in the SDQ with respect to HD, e.g., the choreiform movements of the tongue, we have initiated such a pilot study for HD patients.

Recommendation

The majority of articles that examined therapeutic outcomes for HD were derived from observational studies with few patients and poor methodology. Lack of randomized control trials is apparent. There is further need for research on treatment outcome in HD so that clinicians may use evidence-based practice to assist clinical decision making.

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Appendix

See Table 3.

Table 3

Design	Setting	Participant	Intervention	Outcome measures	Results
[13]					
	Clinical setting	HD ($N = 12$)	1. Detailed diagnostic examination including: • Neurologic history and examination • Dysphagia examination • Barium cineradiography of ingestion • Pulmonary function testing 2. Esophageal manometry ($N = 8$) 3. Indirect laryngoscopy ($N = 5$) 4. Unweighted dysphagia scale (0-5)		Main problems: 1. Dysarthria and impaired voluntary swallowing and coughing 2. Multiple abnormalities of ingestion 3. Difficulties controlling rate and amount of food 4. Dysphagia always the greatest for liquids 5. Retaining oral material after swallow 6. Aerophagia ($N = 8$) 7. Vomiting ($N = 3$) Barium cineradiologic examination confirmed many of the clinical abnormalities
[14]					
Case studies	Clinical setting	HD ($N = 3$)	Describing case stories Describing nursing interventions		1. Current medication needs to be evaluated 2. Mealtime tips 3. Factors to consider in controlling dysphagia: • Medication • Environment • Equipment • Posture • Food choice • Breath control with swallowing • First aid
[15]					
	Clinical setting	HD group ($N = 35$) Hyperkinetic HD-h ($N = 30$) Rigid bradykinetic, HD-rb ($N = 5$)	Crozer-Chester Medical Center Dysphagia Center evaluation, including: 1. Dysphagia history 2. Clinical assessments of oral motor, sensory, and respiratory system functions 3. Liquid and food ingestion analysis 4. Evaluation of independent feeding 5. Videofluoroscopic swallowing study	Fisher's exact test Bonferroni procedure $p > 0.002$ rejected the null hypothesis	Clinical assessments: HD-h: Dysphagia in 29, 27, 11 patients during swallowing stages I, II, III HD-rb: Dysphagia in 5 patients during stage I; coughing, choking, and swallow latency in stage II; no significant disorder in stage III Videofluoroscopic swallowing study HD-h ($N = 29$) HD-rb ($N = 5$): In most instances, the results correlated with clinical assessments
[16]					
Case study	Clinical setting	HD ($N = 1$)	Barium cineradiologic examination		Descending phase-type misdeglutition Involuntary movements of the larynx Tongue protrusion Cough at the beginning of phonation

Table 3

Design	Setting	Participant	Intervention	Outcome measures	Results
[17]					
Case study	Clinical setting	HD ($N = 1$)	General view of self-feeding Videofluorography	Videofluorography: 1. OTT: oral transit time 2. PTT: pharyngeal transit time 3. PRT: pharyngeal response time	5 ml barium: OTT: 0.77 s, PTT: 0.7 s, PRT: 0.7 s 20 ml barium: OTT: 0.23 s, PTT: 0.71 s, PRT: 0.54 s Pudding swallow: OTT: 0.43 s, PTT: 0.6 s, PRT: 0.8 s Main problems: 1. Inability for smooth transportation of food into oral cavity 2. Disorganization of tongue movement 3. Tendency to eat rapidly

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