

# Dysphagia in Individuals with Huntington's Disease: A Narrative Review

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## ABSTRACT

Huntington's disease (HD) is a neurodegenerative autosomal dominant condition characterized by motor, behavioral, and cognitive symptoms. Aspiration pneumonia stands out as a leading cause of death in HD, primarily attributed to dysphagia, which gets more noticeable as the disease progresses. Dysphagia symptoms in individuals with HD are compounded by noticeable movement problems, including Chorean or rigid-bradykinetic patterns. These symptoms manifest in every phase of swallowing and fluctuate with the progression of HD. Lingual chorea, delayed swallowing initiation, and impaired swallowing-respiratory coordination are key indicators of dysphagia in individuals with HD. The negative impact on eating behaviors is further exacerbated by concurrent cognitive and sensory deficits. Consequently, dysphagia leads to social isolation, restrictions on activities and involvement, and a diminished quality of life for individuals with HD. To minimize these adverse effects, a referral to a speech-language therapist (SLT) for swallowing assessment should be initiated immediately upon the diagnosis of HD by a neurologist. Starting from the earliest stages of the disease, both clinical and instrumental swallowing assessments should be employed to minimize the detrimental consequences of dysphagia. Depending on the assessment results, compensatory and/or rehabilitative (restitutive) strategies can be recommended for treatment. Furthermore, the SLT actively collaborates with other team members, including individuals with HD, caregivers, neurologists, otolaryngologists, gastroenterologists, and others, contributing collectively to the decision-making process regarding both oral and non-oral feeding considerations. Despite the negative impact of dysphagia on individuals with HD and its significant role in individuals' deterioration, the evidence for specific dysphagia interventions remains limited. Clinicians, therefore, rely on well-established general swallowing therapy practices. There is a pressing need for evidence-based research on dysphagia in HD. In this study, the literature on dysphagia in individuals with HD will be examined, with a focus on its pathophysiology and the role of SLT in diagnostic and intervention techniques.

**Keywords:** Huntington's disease, swallowing, dysphagia, chorea

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## INTRODUCTION

Huntington's disease (HD) is a neurodegenerative and autosomal dominant disease that often manifests as progressive symptoms beginning in adulthood [1,2]. This condition typically emerges

between the ages of 30 and 50, impacting 4-10 individuals per 100,000 in the population [3-8]. HD is linked to the huntingtin (HTT) gene on chromosome 4p16.3, arising from an elevated number of cytosine-adenine-guanine (CAG) trinucleotide repeats

within this gene [3,9,10]. In individuals without HD, the number of CAG repeats typically does not exceed 34. However, for those with HD, this number can surpass 40 [9]. The increased count of CAG repeats triggers the production of the huntingtin protein, leading to subsequent neuronal loss [9]. Notably, neuronal loss is more common in the basal ganglia, specifically in the caudate nucleus and putamen, although it is also observable in the cerebral cortex [2]. HD causes motor, cognitive, and psychiatric disorders [11]. The most characteristic feature of the disease is chorea, a motor disorder involving dance-like, involuntary, fast, and non-stereotypic hyperkinetic movements [12]. Other motor disorders include muscle stiffness (rigidity), dystonia (involuntary and prolonged muscle contractions), and slowness of movement (bradykinesia) [9,13,14]. Cognitive and psychiatric disorders associated with the disease include dementia, depression, personality changes, and attention deficit [12]. As the disease advances, a genetic test is employed to confirm the diagnosis when clinical symptoms, encompassing a mix of motor, cognitive, and behavioral disorders, raise suspicion [8,13]. While chorea is a common initial experience, dystonia and rigidity also manifest as the disease progresses [13,14]. These motor disruptions in motor function can result in issues like swallowing difficulties (dysphagia) and hyperkinetic dysarthria [15,16]. Alongside motor challenges which contributes to dysphagia, cognitive problems can influence swallowing as well [15]. Dysphagia, in turn, may lead to malnutrition, dehydration, and aspiration pneumonia [17]. Notably, aspiration pneumonia stands as a primary cause of death in HD [18]. Furthermore, dysphagia can contribute to social isolation, limitations in activity and participation, and a decline in overall quality of life

[16,17]. Hence, dysphagia not only adversely affects individuals with HD but also may amplify the burden on caregivers. Therefore, early diagnosis and treatment of dysphagia are of critical importance.

Dysphagia should be managed by a multidisciplinary team for individuals with HD. Members of the team may include, but are not limited to, speech-language therapists (SLTs), neurologists, otolaryngologists, gastroenterologists, nutritionists, physiotherapists, occupational therapists, and nurses. Referral of individuals to SLTs in the early stages of the disease is particularly crucial in terms of providing information and counseling to individuals, according to European HD guidelines [19]. SLTs assist in clinical decision-making for the immediate assessment and intervention for individuals with HD and dysphagia [19]. Individuals with HD and their caregivers are referred to SLTs and then they go through stages such as assessing swallowing safety of individual, identifying current risks for dysphagia, minimizing the individual's and caregiver's concerns, discussing intervention options during the course of the disease, and effectively communicating with needed health professionals [19].

Little is known about HD-related dysphagia despite the physical, psychological, social, and even deadly consequences. This study aims to provide a wide framework for SLTs and learners through reviewing the literature on the etiology, prevalence, symptoms, assessment, and treatments of dysphagia associated with HD. The goal is also to guide clinical practice and improve awareness of the role of SLTs in HD-related dysphagia among other health professionals on the team.

In order to access the findings of the literature to be included in the review, PubMed, Scopus, and Google Scholar were consulted. Conference papers, seminar proceedings, and symposium papers were excluded from the review. Moreover, books were consulted to gather additional information. The article title, abstract, and keyword fields were examined using the following search '*dysphagia in Huntington's disease, dysphagia assessment in Huntington's disease, dysphagia therapy/intervention in Huntington's disease*'. The search encompassed recent publications, specifically those released post-2000, with the exception of four publications including dysphagia-related scales and focusing on HD and serving as primary sources for information on the disease.

### Main Points;

- Dysphagia is a common problem in Huntington's disease, affecting about 90% of individuals with the condition.
- As the disease progresses, symptoms worsen, leading to issues like malnutrition, dehydration, and pneumonia.
- Assessment methods include comprehensive swallowing evaluation like the clinical evaluation and instrumental assessment.
- A team of healthcare professionals, including speech-language therapists, works together to ease dysphagia symptoms using a mix of behavioral and medical approaches, with early involvement of caregivers in the process.

### **Prevalence, Pathophysiology and Symptoms-Findings of Huntington's Disease Related Dysphagia**

Dysphagia, a common disorder in neurodegenerative diseases like Parkinson's, Amyotrophic lateral sclerosis and Alzheimer's, is also prevalent in HD [20-23]. Dysphagia is reported in around 90% of IwHD [21]. It is observed in 35% of people in the early stage, 94% of people in the middle stage, and 100% of people in the late stage [24]. Neuropathological changes in HD involve neurodegeneration in regions such as the striatum, pallidum, cerebral neocortex and allocortex, brainstem, thalamus, and cerebellum [25]. A study highlighted the compromise of swallowing safety due to atrophy in a network comprising parietototalamocerebellar areas [26]. This network is associated with sensory processing, sensorimotor transformation, and cognitive control. Neurodegeneration in these areas leads to the emergence of sensory, motor, cognitive, and psychiatric disorders [26].

It is well known that dysphagia can manifest in the oral, pharyngeal, and esophageal phases in HD [27]. Neuromuscular dyscoordination in the oral and pharyngeal phases is thought to originate from basal ganglia or cerebellar dysfunction [24,25]. The impact of mutant huntingtin on oro-pharyngeal muscles is also presumed [24]. Silent aspiration, as observed in individuals with HD, may arise from sensory disturbances in the epiglottis and posterior wall of the hypopharynx, similar to other neurodegenerative disorders (e.g., Parkinson's disease) [24,28]. Motor and sensory problems affecting swallowing can worsen with motor problems in the upper extremities in individuals with HD, leading to severe restrictions in self-feeding [29]. Dysphagia symptoms in individuals with HD can be hyperkinetic or akinetic-rigid motor symptoms, varying according to the stage of HD [15,30]. Involuntary movements observed in HD can complicate the eating and drinking process by making postural control challenging, increasing the risk of aspiration [15,31]. Rapid lingual chorea and lingual dysfunction associated with swallowing coordination issues result in problems with bolus preparation and delivery [15,32,33]. Weak lingual control of the bolus can lead to premature spilling into the laryngeal vestibule [29]. Coughing due to laryngeal chorea is an early clinical symptom of HD [27]. Loss of voluntary feeding control is associated with late-stage symptoms such as aspiration pneumonia, choking, unintentional weight loss, and cachexia [30,27,34].

In the oral phase of swallowing, in addition to lingual

dysfunction, impaired and involuntary lip and jaw movements can be observed [35]. In the pharyngeal phase of swallowing in individuals with HD, coordination disorders, airway protection issues, prolonged laryngeal elevation, decreased pharyngeal clearance, pharyngeal residue, and penetration/aspiration problems may arise [15,29,30]. Postural instability in HD contributes to problems in both the oral and pharyngeal phases [29]. In the esophageal phase, there may be gastroesophageal inflammation such as gastritis or esophagitis and esophageal dysmotility [15,36].

Psychiatric and cognitive problems can also impact nutrition in addition to motor and sensory issues. Cognitive and psychiatric changes pose the greatest burden on families, are associated with functional decline, and can predict hospitalization [37,38]. Individuals with HD may not be aware of social eating behaviors, may fragment meals during anger bursts, and may struggle to maintain the social aspects of eating [27]. Cognitive inhibition deficiencies affecting eating speed lead to insatiable appetite, impulsively fast eating (tachyphagia), and increased hunger in individuals with HD, resulting in ineffective transportation of food into the oral cavity and inadequate lip closure [33].

Common oral health issues observed among individuals with HD include gum inflammation, tooth decay, poor oral hygiene, and bruxism in a recent review [39]. Additionally, many medications used for psychiatric symptoms induce dry mouth [40]. Early referral to a dentist is essential for maintaining oral health, as dental procedures become more challenging as the disease progresses [39].

In summary, involuntary movements can impact all phases of swallowing in individuals with HD. Symptoms such as throwing food from containers, pushing food out of the mouth with the tongue, breathing during laryngeal swallowing, and regurgitation can occur [27]. Although dysphagia symptoms and findings emerge in all three phases, they are typically observed in the oral and pharyngeal phases [32,36]. Figure 1 emphasizes dysphagia symptoms and findings in individuals with HD based on swallowing stages [15,27,32].

### **Assessment of Huntington's Disease Related Dysphagia**

It is now established that dysphagia is prevalent in HD. Various symptoms associated with dysphagia can manifest during different stages of the swallowing process. For instance, aspiration, even in the early stages of HD, has been reported to

be prevalent, with a notable incidence of silent aspiration [26]. Although individuals with HD may not self-report dysphagia symptoms during assessment, it is crucial for neurologists to address these symptoms explicitly and refer to SLT for a comprehensive swallowing evaluation, such as a clinical evaluation and instrumental assessment. The comprehensive swallowing evaluation, aligned with the International Classification of Functioning, Disability, and Health (ICF)

framework, should commence the evaluation from the early stages of the disease [19,31]. The ICF framework emphasizes that dysphagia assessment should encompass (1) the evaluation of body structures and functions that may influence swallowing, eating, or drinking behaviors, (2) swallowing activities and participation, (3) eating or drinking activities and participation, and (4) personal and environmental factors that may affect swallowing [9,41].

<p><b>Oral preparation phase</b></p> <ul style="list-style-type: none"> <li>• Involuntary movements of the head and trunk</li> <li>• Overfilling the mouth</li> <li>• Reduced lip closure</li> <li>• Inefficient mastication</li> <li>• Involuntary movement of the tongue</li> <li>• Tachyphagia</li> </ul>
<p><b>Oral phase</b></p> <ul style="list-style-type: none"> <li>• Lingual chorea</li> <li>• Lack of coordination in swallowing</li> <li>• Fragmented and slow lingual conduction</li> <li>• Bolus retention</li> <li>• Repeated swallowing</li> <li>• Deficiency of coordination among oral and pharyngeal phases</li> <li>• Impaired swallow initiation</li> <li>• Oral residue</li> </ul>
<p><b>Pharyngeal phase</b></p> <ul style="list-style-type: none"> <li>• Laryngeal and respiratory incoordination</li> <li>• Involuntary laryngeal movements</li> <li>• Prolonged and/or decreased laryngeal elevation</li> <li>• Coughing after and/or before swallows</li> <li>• Residue in vallecula</li> <li>• Residue in the piriform sinus</li> <li>• Penetration</li> <li>• Aspiration</li> </ul>
<p><b>Esophageal phase</b></p> <ul style="list-style-type: none"> <li>• Decreased and/or slow opening of the upper and lower esophageal sphincter</li> <li>• Esophageal dysmotility</li> <li>• Slow bolus movement along the esophagus</li> <li>• Reflux</li> <li>• Burping</li> <li>• Reverse peristalsis</li> <li>• Regurgitation</li> <li>• Vomiting</li> </ul>

**Figure 1.** Dysphagia symptoms and findings in individuals with Huntington's disease

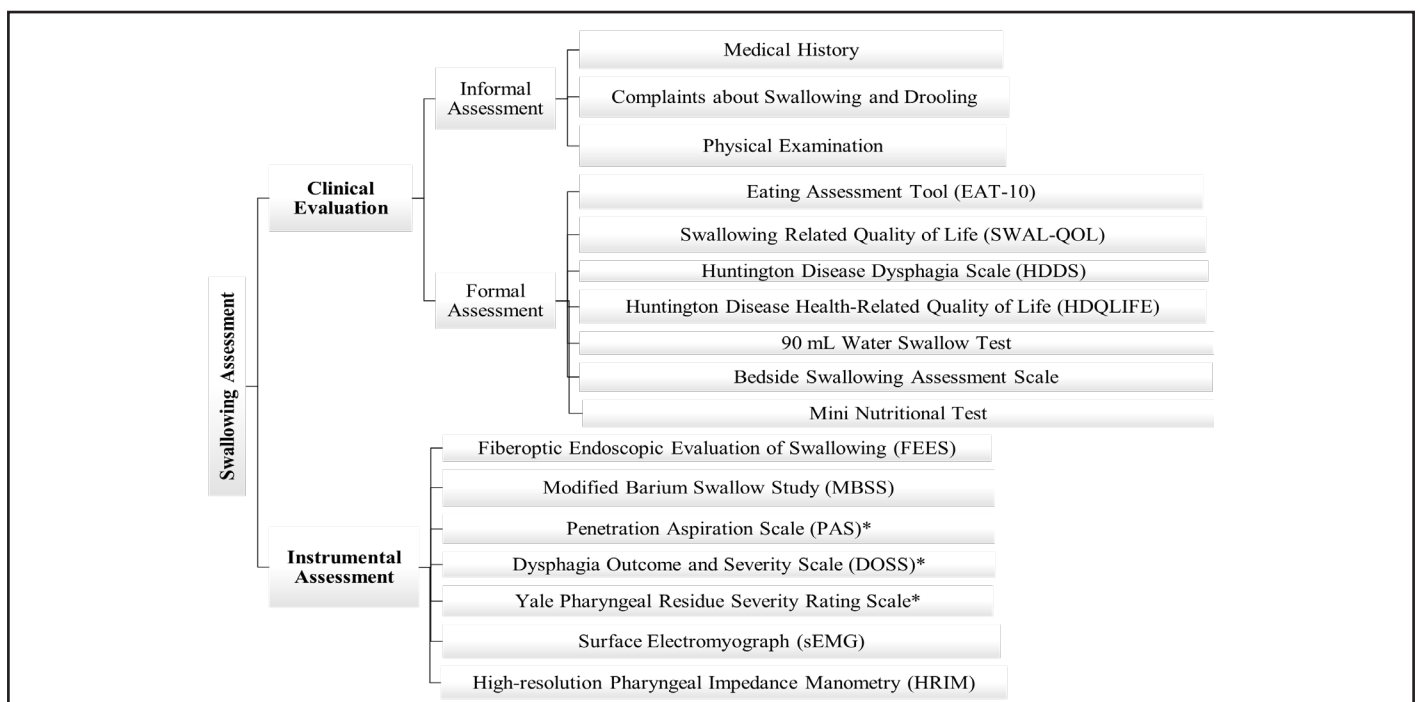
Clinical evaluation of swallowing includes medical history, complaints about swallowing and drooling, physical examination (observation, oral motor assessment, respiration, phonation and resonance assessment, feeding assessment, test swallows) [42]. Dysphagia assessment includes a clinical evaluation (including formal and informal assessments). Information collected during the informal (pre-assessment/preliminary) assessment should adhere to the standards recommended by assessment guidelines

[19]. A comprehensive preliminary assessment should include case history, medical history, dysphagia symptoms, nutrition/swallowing history, and patient-specific information at the beginning of the clinical evaluation [42]. Medical reports, discussions with healthcare professionals, and verbal information from the individual or caregiver can be utilized to obtain a thorough medical history [42]. The formal assessment aims to identify the presence of dysphagia symptoms, such as mealtime

performance and accompanying environmental factors, during the clinical evaluation [9]. Patient-reported scales that provide preliminary information for advanced assessments can be employed as screening tools to assess the risk of dysphagia [31]. Widely used scales include the Eating Assessment Tool (EAT-10) and Swallowing Related Quality of Life (SWAL-QOL) [43-45]. Self-assessment tools specifically designed for individuals with HD, such as the Huntington Disease Dysphagia Scale (HDDS) and Huntington Disease Health-Related Quality of Life (HDQLIFE), can also be utilized [16,46]. A 90 mL water swallow test and bedside swallowing assessment, including clinician observation, can provide preliminary information for more advanced assessments [31,47]. Additionally, observing feeding in the home environment can be beneficial for assessing the impact of environmental factors [19]. When clinical evaluation symptoms in the digestive/upper respiratory tract, an appropriate medical referral should be made. In the final stage of clinical assessment, should use the gathered information to decide on further assessment procedures and determine the safety of food trials [19].

Fiberoptic Endoscopic Evaluation of Swallowing (FEES) and Videofluoroscopic Swallowing Study (VFSS)/Modified Barium Swallow Study (MBSS) are the most used instrumental methods for diagnosing dysphagia in individuals with HD [26,27,29].

However, these diagnostic approaches may be contraindicated if the individual cannot collaborate or participate due to involuntary movements or cognitive issues [19]. FEES may be more challenging to use in individuals with HD because it does not visualize the oral phase, and choreic movements may complicate evaluation [19]. However, various scales and procedures can be employed and integrated for specific purposes. In some studies where FEES is used for dysphagia in individuals with HD, it has been combined with the Penetration Aspiration Scale (PAS), Dysphagia Outcome and Severity Scale (DOSS), or Yale Pharyngeal Residue Severity Rating Scale [24,26,31,48-50]. Oral intake can be recorded based on the Functional Oral Intake Scale (FOIS) [51]. In a study using VFSS for dysphagia in individuals with HD, the Penetration Aspiration Scale and Bethlehem Assessment Scale (BAS) were used [29]. Additionally, high-resolution pharyngeal impedance manometry (HRIM) and surface electromyograph (sEMG) can be used [32,52]. The advantage of pharyngeal HRIM is that it allows the objective evaluation of swallowing biomechanics, contributing to understanding the pathogenesis of dysphagia and developing a treatment plan. Surface electromyography (sEMG) can be used to measure the activity of the submental muscles during swallowing and as a biofeedback tool. Methods and scales that can be used in swallowing assessments in individuals with HD are provided in Figure 2.



**Figure 2.** Swallowing assessment in individuals with Huntington's disease.

\*Scales that are used during the instrumental assessment.

Collaborative analysis of the information obtained from swallowing assessments with medical professionals can enhance the effectiveness of diagnosis, appropriate food selection, postural modification, and maneuver or exercise options. Gaining insights into the impact of dysphagia on eating and drinking activities and participation, as well as the burden it places on caregivers, will provide a foundation for a holistic, sensitive, and collaborative intervention. Certainly, the assessment process should involve a multidisciplinary team comprising a neurologist, SLT, and other relevant professionals. Each opinion and perspective are crucial for planning the therapy phase.

### **Intervention of Huntington's Disease Related Dysphagia**

Behavioral, medical interventions and a combination of both can be employed in the management of dysphagia among individuals with HD. SLTs play a pivotal role, particularly in addressing oral and pharyngeal dysphagia. Their responsibilities encompass tailoring treatment plans for swallowing disorders based on individual performance, delivering treatment, and establishing criteria for concluding interventions. Furthermore, their coordination with other healthcare professionals facilitates the selection of the most effective intervention method for the individual [42].

In the context of managing dysphagia in individuals with HD the primary objective is to alleviate symptoms rather than tackling the underlying cause [27]. As the disease advances, considerations for cognitive levels and behaviors become increasingly vital. Consequently, the treatment plan should be adjusted according to the individual's cognitive level. Behavioral treatment for dysphagia falls into two categories: compensatory or rehabilitative (restitutive) intervention [42]. Compensatory intervention involves a range of strategies such as postural adjustments, proper equipment utilization, oral hygiene practices, considerations for meal quantity and frequency, bolus size and placement, maneuvers, feeding rate, nutritional modifications, and caregiver support. It is crucial to customize and apply these strategies with a specific focus on individuals with HD [19,27]. Creating a conducive feeding environment also contributes to improving swallowing function and safety [31]. Exercises and maneuvers aimed at enhancing swallowing function are integral components of rehabilitative treatment [42]. Exercises that enhance airway closure could be helpful [52-54]. A randomized controlled pilot study, for instance, examined the impact of a 4-month respiratory muscle

strength training on 18 participants with HD, revealing benefits for pulmonary function in individuals with HD but no effect on swallowing function [53].

Speech-language therapists can offer recommendations and implement compensatory and rehabilitative interventions for both individuals and caregivers early on [19]. These recommendations may encompass raising awareness about swallowing, reducing concerns related to swallowing and eating, providing guidance on postural adjustments, alerting about hazardous foods and textures, minimizing distractions during mealtime, and establishing a comfortable environment [19]. Recommendations pertaining to diet modifications are incorporated in addition to early-stage interventions. Importantly, safe swallowing advice and information should be communicated effectively, considering both oral and written formats for individuals and caregivers of individuals with HD. Furthermore, SLTs are crucial in providing suggestions to modify the viscosity, textures, and temperatures of food and liquids for enhanced swallowing safety [19].

Active collaboration of SLTs in the multidisciplinary decision-making process regarding non-oral nutrition (e.g., nasogastric tube [NG], percutaneous endoscopic gastrostomy [PEG]) is important. However, this decision-making process should be initiated with due consideration of the cognitive abilities of the individual, involving individuals with HD and their caregivers. Until advanced stages, oral feeding is generally feasible in HD. Even in advanced stages, efforts can be made to continue diet modifications, compensatory, and rehabilitative swallowing maneuvers as much as possible. Additionally, maintaining oral hygiene and ensuring that caregivers possess knowledge about safe nutrition for individuals are emphasized [19,39].

Medical interventions encompass pharmacological treatments and non-oral nutrition alternatives. Pharmacological treatments (e.g., antipsychotics, valbenazine, and tetrabenazine) can be advantageous in managing motor and behavioral symptoms in HD. However, their usage should be approached cautiously in individuals with mid to late-stage dysphagia [31]. Especially for individuals facing challenges with swallowing pills and capsules, alternative administration methods may be explored. Data from controlled laboratory studies suggest that the crushed content of valbenazine capsules can be incorporated into soft foods or liquids, including acidic ones, or administered through a G-tube. This holds promise for patients struggling with capsule

ingestion, and further evaluation in a clinical setting is warranted [55]. Generally, in scenarios where oral intake becomes problematic, and airway protection is uncertain, nutritional

options like NG or PEG are discussed with the collaboration of other team members. Dysphagia intervention recommendations for individuals with HD are given in Figure 3 [19,27,39,52-54].

<b>Compensatory Strategies &amp; Techniques</b>
<ul style="list-style-type: none"> <li>• <b>To reduce or prevent involuntary movements:</b> <ul style="list-style-type: none"> <li>• <i>Keep the feet grounded</i></li> <li>• <i>Support the hands, arms, and lower back</i></li> <li>• <i>Chin tuck</i></li> <li>• <i>Use stabilized and small-sized food containers</i></li> <li>• <i>Lidded cups</i></li> <li>• <i>Adjustable-length and shaped straws</i></li> <li>• <i>Presentation of small amounts and bite-sized food portions</i></li> </ul> </li> <li>• <b>To prevent body weight loss and dehydration</b> <ul style="list-style-type: none"> <li>• <i>Eating small and frequent meals</i></li> <li>• <i>Reducing bolus size</i></li> </ul> </li> <li>• <b>In the presence of dysphagia in the oral and pharyngeal phases</b> <ul style="list-style-type: none"> <li>• <i>Diet modification</i></li> <li>• <i>Reducing bolus size</i></li> <li>• <i>Serving cold-sour bolus before meals</i></li> <li>• <i>Verbal cues</i></li> </ul> </li> </ul>
<b>Rehabilitative Strategies &amp; Techniques</b>
<ul style="list-style-type: none"> <li>• <b>To maintain oral motor function:</b> <ul style="list-style-type: none"> <li>• <i>Oral motor exercises</i></li> </ul> </li> <li>• <b>To reduce swallowing symptoms in the pharyngeal phase:</b> <ul style="list-style-type: none"> <li>• <i>Appropriate swallowing exercises/maneuvers</i></li> <li>• <i>Respiratory exercises</i></li> </ul> </li> </ul>
<b>Medical Strategies &amp; Techniques</b>
<ul style="list-style-type: none"> <li>• <b>For inadequate esophageal opening:</b> <ul style="list-style-type: none"> <li>• <i>Botulinum toxin injections</i></li> </ul> </li> <li>• <b>Nasogastric (NG) tube</b></li> <li>• <b>Percutaneous endoscopic gastrostomy (PEG)</b></li> <li>• <b>Oral hygiene</b></li> </ul>

**Figure 3.** Recommendations for dysphagia intervention in Huntington's Disease

## CONCLUSION

A referral and a teamwork are crucial for a thorough assessment of swallowing upon the neurologist's confirmation of HD diagnosis, regardless of the presence of symptoms, while clinical evaluations and patient-reported scales are preferred for comprehensive swallowing assessments, they cannot replace instrumental evaluations. Given the neurodegenerative nature of the disease, MBSS should be utilized early in the progression of HD to assess and monitor the individual's advancement. Even in the early stages, proactive swallowing maneuvers and exercises can be beneficial, with modifications adjusted over time based on the individual's condition and preferences. The inclusive involvement of caregivers in the dysphagia management process

at every stage can provide valuable support. Collaborating with experts from various disciplines is essential when making decisions about dysphagia management, especially in situations where oral intake is not possible. Considering the limited nature of existing literature summaries on dysphagia interventions in individuals with HD, further evidence-based research on this subject is necessary.

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## REFERENCES

- [1] Bates GP, Dorsey R, Gusella JF, Hayden MR, Kay C, Leavitt BR, Nance M, Ross CA, Scahill RI, Wetzel R, Wild EJ, Tabrizi SJ (2015) Huntington disease. *Nat Rev Dis Primers*. 23(1):15005. <https://doi.org/10.1038/nrdp.2015.5>
- [2] Freed DB (2020) Motor speech disorders: diagnosis and treatment. Plural Publishing.
- [3] A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes (1993) The Huntington's Disease Collaborative Research Group. *Cell*, 72(6):971–983. [https://doi.org/10.1016/0092-8674\(93\)90585-e](https://doi.org/10.1016/0092-8674(93)90585-e)
- [4] Ross CA, Aylward EH, Wild EJ, Langbehn DR, Long JD, Warner JH, Scahill RI, Leavitt BR, Stout JC, Paulsen JS, Reilmann R, Unschuld PG, Wexler A, Margolis RL, Tabrizi SJ. Huntington disease: natural history, biomarkers and prospects for therapeutics. *Nat Rev Neurol*. 2014 Apr;10(4):204-16. <https://doi.org/10.1038/nrneurol.2014.24>
- [5] Squitieri F, Griguoli A, Capelli G, Porcellini A, D'Alessio B. Epidemiology of Huntington disease: first post-HTT gene analysis of prevalence in Italy. *Clin Genet*. 2016 Mar;89(3):367-70. <https://doi.org/10.1111/cge.12574>
- [6] Evans SJ, Douglas I, Rawlins MD, Wexler NS, Tabrizi SJ, Smeeth L. Prevalence of adult Huntington's disease in the UK based on diagnoses recorded in general practice records. *J Neurol Neurosurg Psychiatry*. 2013 Oct;84(10):1156-60. <https://doi.org/10.1136/jnnp-2012-304636>
- [7] Medina A, Mahjoub Y, Shaver L, Pringsheim T (2022) Prevalence and Incidence of Huntington's Disease: An Updated Systematic Review and Meta-Analysis. *Mov Disord*. 37(12):2327–2335. <https://doi.org/10.1002/mds.29228>
- [8] Roos RA (2010) Huntington's disease: a clinical review. *Orphanet J Rare Dis*. 5:40. <https://doi.org/10.1186/1750-1172-5-40>
- [9] Murry T, Carrau R, Chan K (2020) Clinical management of swallowing disorders, 5th ed. Plural Publishing.
- [10] Nance MA (2017) Genetics of Huntington disease. *Handb Clin Neurol*. 144: 3–14. <https://doi.org/10.1016/B978-0-12-801893-4.00001-8>
- [11] Bachoud-Lévi AC (2020) On behalf the Multicentric Intracerebral Grafting in Huntington's Disease Group. Human Fetal Cell Therapy in Huntington's Disease: A Randomized, Multicenter, Phase II Trial. *Mov Disord*. 35(8):1323-1335. <https://doi.org/10.1002/mds.28201>
- [12] Duffy J (2013) Motor Speech Disorders: Substrates, Differential Diagnosis, and Management. 3 ed. St. Louis: Elsevier Mosby.
- [13] Walker FO (2007) Huntington's disease. *Lancet*. 369(9557):218–228. [https://doi.org/10.1016/S0140-6736\(07\)60111-1](https://doi.org/10.1016/S0140-6736(07)60111-1)
- [14] Mahant N, McCusker EA, Byth K, Graham S (2003) Huntington Study Group. Huntington's disease: clinical correlates of disability and progression. *Neurology*. 61(8):1085-92. <https://doi.org/10.1212/01.wnl.0000086373.32347.16>
- [15] Kagel MC, Leopold NA (1992) Dysphagia in Huntington's disease: a 16-year retrospective. *Dysphagia*. 7(2):106-14. <https://doi.org/10.1007/BF02493441>
- [16] Carlozzi NE, Schilling SG, Lai JS, Perlmutter JS, Nance MA, Waljee JF, Miner JA, Barton SK, Goodnight SM, Dayalu P (2016) HDQLIFE: the development of two new computer adaptive tests for use in Huntington disease, Speech Difficulties, and Swallowing Difficulties. *Qual Life Res*. 25(10):2417-2427. <https://doi.org/10.1007/s11136-016-1273-y>
- [17] Ekberg O, Hamdy S, Woisard V, Wuttge-Hannig A, Ortega P (2002) Social and psychological burden of dysphagia: its impact on diagnosis and treatment. *Dysphagia*. 17(2):139-46. <https://doi.org/10.1007/s00455-001-0113-5>
- [18] Heemskerk AW, Roos RA (2012) Aspiration pneumonia and death in Huntington's disease. *PLoS Curr*. 30;4:RRN1293. <https://doi.org/10.1371/currents.RRN1293>
- [19] Hamilton A, Heemskerk AW, Loucas M, Twiston-Davies R, Matheson KY, Simpson SA, Rae D (2012) Oral feeding



- in Huntington's disease: a guideline document for speech and language therapists. *Neurodegener Dis Manag.* 2(1), 45-53.
- [20] Parlak MM, Babademez MA, Alicura Tokgöz S, Bizpınar Ö, Saylam G (2022) Evaluation of Swallowing Function according to the Stage of Alzheimer's Disease. *Folia Phoniatr Logop.* 74(3):186-194. <https://doi.org/10.1159/000519263>
- [21] Kalkers K, Schols JMGA, van Zwet EW, Roos RAC (2022) Dysphagia, Fear of Choking and Preventive Measures in Patients with Huntington's Disease: The Perspectives of Patients and Caregivers in Long-Term Care. *J Nutr Health Aging.* 26(4):332-338. <https://doi.org/10.1007/s12603-022-1743-6>
- [22] Sapmaz Atalar M, Genç G (2022) Parkinson Hastalığında Yutma Bozukluklarına Yaklaşım. *Muğla Sıtkı Koçman Üniversitesi Tıp Dergisi.* 9(2), 189-203.
- [23] Perry BJ, Nelson J, Wong JB, Kent DM (2021) The cumulative incidence of dysphagia and dysphagia-free survival in persons diagnosed with amyotrophic lateral sclerosis. *Muscle Nerve.* 64(1):83-86. <https://doi.org/10.1002/mus.27244>
- [24] Schindler A, Pizzorni N, Sassone J, Nanetti L, Castaldo A, Poletti B, Solca F, Pirola F, Lazzari L, Stramba-Badiale M, Rossi A, Silani V, Mariotti C, Ciammola A (2020) Fiberoptic endoscopic evaluation of swallowing in early-to-advanced stage Huntington's disease. *Sci Rep.* 10(1):15242. <https://doi.org/10.1038/s41598-020-72250-w>
- [25] Rüb U, Seidel K, Heinsen H, Vonsattel JP, den Dunnen WF, Korf HW (2016) Huntington's disease (HD): the neuropathology of a multisystem neurodegenerative disorder of the human brain. *Brain Pathol.* 26(6):726-740. <https://doi.org/10.1111/bpa.12426>
- [26] Schumann-Werner B, Dogan I, Mirzazade S, Mall B, Overbeck R, Honrath P, Schulz JB, Reetz K, Werner CJ (2021) Clinical predictors and neural correlates for compromised swallowing safety in Huntington disease. *Eur J Neurol.* 28(9):2855-2862. <https://doi.org/10.1111/ene.14953>
- [27] Stewart C (2012). Dysphagia Symptoms and Treatment in Huntington's Disease: Review. *Perspectives on Swallowing and Swallowing Disorders (Dysphagia).* 21(4): 126. <https://doi.org/10.1044/sasd21.4.126>
- [28] Rodrigues B, Nóbrega AC, Sampaio M, Argolo N, Melo A (2011) Silent saliva aspiration in Parkinson's disease. *Mov Disord.* 26(1):138-41. <https://doi.org/10.1002/mds.23301>
- [29] Keage M, Baum S, Pointon L, Lau J, Berndt J, Hopkins J, Maule R, Vogel AP (2020) Imaging and Clinical Data on Swallowing Function of Individuals with Huntington's Disease and Dysphagia. *J Huntingtons Dis.* 9(2):163-171. <https://doi.org/10.3233/JHD-190390>
- [30] Heemskerk AW, Roos RA (2011) Dysphagia in Huntington's disease: a review. *Dysphagia.* 26(1):62-6. <https://doi.org/10.1007/s00455-010-9302-4>
- [31] Pizzorni N, Pirola F, Ciammola A, Schindler A (2020) Management of dysphagia in Huntington's disease: a descriptive review. *Neurol Sci.* 41(6):1405-1417. <https://doi.org/10.1007/s10072-020-04265-0>
- [32] Lee TH, Lee JS, Kim WJ (2012) High resolution impedance manometric findings in dysphagia of Huntington's disease. *World J Gastroenterol.* 18(14):1695-9. <https://doi.org/10.3748/wjg.v18.i14.1695>
- [33] Hamakawa S, Koda C, Umeno H, Yoshida Y, Nakashima T, Asaoka K, Shoji H (2004) Oropharyngeal dysphagia in a case of Huntington's disease. *Auris Nasus Larynx.* 31(2):171-6. <https://doi.org/10.1016/j.anl.2003.11.001>
- [34] Trejo A, Tarrats RM, Alonso ME, Boll MC, Ochoa A, Velásquez L (2004) Assessment of the nutrition status of patients with Huntington's disease. *Nutrition.* 20(2):192-6. <https://doi.org/10.1016/j.nut.2003.10.007>
- [35] Manor Y, Oestreicher-Kedem Y, Gad A, Zitser J, Faust-Socher A, Shpunt D, Naor S, Inbar N, Kestenbaum M, Giladi N, Gurevich T (2019) Dysphagia characteristics in Huntington's disease patients: insights from the Fiberoptic Endoscopic Evaluation of Swallowing and the Swallowing Disturbances Questionnaire. *CNS Spectr.* 24(4):413-418. <https://doi.org/10.1017/S1092852918001037>
- [36] Andrich JE, Wobben M, Klotz P, Goetze O, Saft C (2009) Upper gastrointestinal findings in Huntington's disease: patients suffer but do not complain. *J Neural Transm (Vienna).* 116(12):1607-11. <https://doi.org/10.1007/s00702-009-0310-1>

- [37] Ishihara L, Oliveri D, Wild EJ. (2021) Neuropsychiatric comorbidities in Huntington's and Parkinson's Disease: A United States claims database analysis. *Ann Clin Transl Neurol.* 8(1):126-137. <https://doi.org/10.1002/acn3.51252>
- [38] Paulsen JS (2011) Cognitive impairment in Huntington disease: diagnosis and treatment. *Curr Neurol Neurosci Rep.* 11:474–483.
- [39] Munhoz L, Jabbar AQ, Silva Filho WJE, Nagai AY, Arita ES (2023) The oral manifestations of Huntington's disease: A systematic review of prevalence. *Oral Dis.* 29(1):62-74. <https://doi.org/10.1111/odi.14076>
- [40] Rada RE (2008) Comprehensive dental treatment of a patient with Huntington's disease: literature review and case report. *Spec Care Dentist.* 28(4):131-5. <https://doi.org/10.1111/j.1754-4505.2008.00027.x>
- [41] Threats TT (2007) Use of the ICF in dysphagia management. *Semin Speech Lang.* 28(4):323–333.
- [42] Groher ME, Crary MA (2016) *Dysphagia: Clinical Management in Adults and Children.* 2nd ed. USA: Elsevier Health Sciences.
- [43] Belafsky PC, Mouadeb DA, Rees CJ, Pryor JC, Postma GN, Allen J, Leonard RJ (2008) Validity and reliability of the Eating Assessment Tool (EAT-10). *Ann Otol Rhinol Laryngol.* 117(12):919-24. <https://doi.org/10.1177/000348940811701210>
- [44] Cordier R, Joosten A, Clavé P, Schindler A, Bülow M, Demir N, Arslan SS, Speyer R (2017) Evaluating the Psychometric Properties of the Eating Assessment Tool (EAT-10) Using Rasch Analysis. *Dysphagia.* 32(2):250-260. <https://doi.org/10.1007/s00455-016-9754-2>
- [45] McHorney CA, Robbins J, Lomax K, et al. (2002) The SWAL-QOL and SWAL-CARE outcomes tool for oropharyngeal dysphagia in adults: III. Documentation of reliability and validity. *Dysphagia.* 17 (2): 97–114.
- [46] Heemskerk A, Verbist BM (2014) The Huntington's disease dysphagia scale. *Mov Disord* 29:1312–1316.
- [47] de Tommaso M, Nuzzi A, Dellomonaco AR, Scirucchio V, Serpino C, Cormio C, Franco G, Megna M (2015) Dysphagia in Huntington's disease: correlation with clinical features. *Eur Neurol* 74:49–53.
- [48] Rosenbek JC, Robbins JA, Roecker EB, Coyle JL, Wood JL (1996) A penetration-aspiration scale. *Dysphagia.* 1996 Spring;11(2):93-8. <https://doi.org/10.1007/BF00417897>
- [49] O'Neil KH, Purdy M, Falk J, Gallo L. (1999) The Dysphagia Outcome and Severity Scale. *Dysphagia.* 14(3):139-45. <https://doi.org/10.1007/PL00009595>
- [50] Neubauer PD, Rademaker AW, Leder SB (2015) The Yale Pharyngeal Residue Severity Rating Scale: An Anatomically Defined and Image-Based Tool. *Dysphagia.* 30(5):521-8. <https://doi.org/10.1007/s00455-015-9631-4>
- [51] Crary MA, Mann GD, Groher ME (2005) Initial psychometric assessment of a functional oral intake scale for dysphagia in stroke patients. *Arch Phys Med Rehabil.* 86(8):1516-20. <https://doi.org/10.1016/j.apmr.2004.11.049>
- [52] Reyes A, Cruickshank T, Thompson J, Ziman M, Nosaka K (2014) Surface electromyograph activity of submental muscles during swallowing and expiratory muscle training tasks in Huntington's disease patients. *J Electromyogr Kinesiol.* 24(1):153-8. <https://doi.org/10.1016/j.jelekin.2013.09.009>
- [53] Reyes A, Cruickshank T, Nosaka K, Ziman M (2015) Respiratory muscle training on pulmonary and swallowing function in patients with Huntington's disease: a pilot randomised controlled trial. *Clin Rehabil.* 29(10):961-73. <https://doi.org/10.1177/0269215514564087>
- [54] Clark J (2021) A practical approach to movement disorders: diagnosis and management. In: Fernandez, H. H., Walter, B. L., Rush, T., & Ahmed, A. (eds.), Springer Publishing Company.
- [55] Sajatovic M, Patel A, Hebert M, Mar A, Moore R, Bristow A, Farahmand K, Siegert S (2023) Crushing the Contents of Valbenazine Capsules for Potential Addition to Soft Foods or Administration via Gastrostomy Tube. *Clin Ther.* 45(12):1222-1227. <https://doi.org/10.1016/j.clinthera.2023.09.024>

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