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Contents

Chapter 1

General introduction

Huntington's disease

Huntington's disease (HD) is a progressive neurodegenerative disease, with an autosomal dominant mode of inheritance. The chromosomal abnormality is a CAG repeat expansion on chromosome 4. The mutant protein, huntingtin, causes neurodegeneration in the brain, particularly in the caudate nucleus and putamen. Characteristic features of HD are movement abnormalities (chorea, hypokinesia), cognitive decline and psychiatric disturbance. Once manifest, the patient's life expectancy is about 15-20 years [1]. After disease onset, three clinical stages can be described [2]. Stage I: Patients develop initial symptoms, but are still independent. Stage II: As the disease progresses, patients become more dependent, and the symptoms are more generalised. Stage III is the end-of-life stage: prior to death, the patients are completely dependent for all daily life activities [2]. Patients with HD also suffer from dysphagia, a condition which can have serious consequences, such as weight loss, dehydration and pneumonia leading to death. Studies on causes of death in HD found a variation in the percentage of patients who died of pneumonia, ranging from 33%- 86% [3-8].

Swallowing

The act of swallowing is a complex motor program involving a sequence of activation and inhibition of muscles in the mouth, pharynx, larynx and esophagus [9]. Swallowing is divided into four phases [10]. The first phase is the *preparatory oral phase*, which involves the transport of the bolus in the mouth, and, if necessary, mastication of the bolus and mixing it with saliva. This phase is almost entirely voluntary, and can, therefore, be interrupted when needed. The second phase, the *oral phase*, begins when the proximal part of the bolus is propelled to the oropharynx, until the bolus head is reaching the point where the lower edge of the mandible crosses the tongue base. This leads to the start of the third phase, the *pharyngeal phase*, which ends when the bolus leaves the cricopharyngeal area and the tail of the bolus is in the esophagus. During the last phase of the swallow, the *esophageal phase*, the bolus reaches the stomach by peristaltic movements of the esophagus. The last two phases are completely involuntary [10]. It is known that patients with HD have disturbances in all phases of ingestion.

Dysphagia in HD

Although dysphagia is a very frequent occurrence in HD, only very few systematic studies are available (11-15). One larger study demonstrated that HD patients suffer from dysphagia in all phases of ingestion [11]. During the preparatory oral phase, it was found that patients had postural instability, hyperextension of head and trunk, tachyphagia, inadequate mastication, delayed lingual transfer and lingual chorea. During the oral phase, swallow incoordination, repetitive swallows, swallow latency, intraoral bolus retention, and segmented lingual transfer were seen. In the pharyngeal phase, coughing, choking, aspiration, eructations, aerophagia, audible swallows, prolonged laryngeal elevation, phonation during the swallow, pharyngeal stasis, inability to stop respiration, wet vocal quality, and laryngeal chorea were reported. During the last phase of the swallow, the esophageal phase, it was found that patients vomit, have early satiety, abnormal esophageal motility, diaphragmatic chorea and reflux.

Although many swallowing disturbances have been reported, no follow up-study has since been carried out; it is not known, therefore, when dysphagia commences, or how it progresses during the different stages of the disease. Furthermore, dysphagia-specific features, like spilling, residue and transit times, are not known. As far as we are aware, no study into dysphagia treatment and interventions has been performed. A well-structured study into dysphagia in HD, in a larger group of patients, was, therefore, urgently needed.

Aims of this thesis

The first step was to study the available literature on dysphagia in HD *(chapter 2).* We also investigated whether the most frequently encountered primary cause of death in HD, pneumonia, was due to aspiration *(chapter 3)*. As the existing assessment scales were inadequate, we developed a new scale *(chapter 4)*. This tool was used to evaluate swallowing problems in patients from all three disease stages. In *chapter 5,* we investigated the specific dysphagia problems in HD, and described the severity of dysphagia during the course of the disease. In order to assess this properly, we made 45 videofluoroscopies and measured swallowing features, such as penetration and aspiration, spilling, residue and transit times. The next chapter *(chapter 6)*, describes a guideline on dysphagia developed by a working group of the European Huntington Disease Network (EHDN). Finally, we explored if the commonly used ‘chin tuck’ method that is used in patients with dysphagia, and is also often recommended in HD patients, is a useful intervention for HD patients *(chapter 7)*. The final chapter *(chapter 8)* consists of a summary and concluding remarks.

This thesis aims to contribute to the research into and the knowledge about dysphagia in HD, with the intention of stimulating further research, and, most importantly, helping HD patients to handle their dysphagia and receive the most adequate intervention to finally prevent aspiration.

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Chapter 2

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 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 highly variable, and no balanced advice for prevention and treatment was systematically proven.

Key words: Huntington's disease, dysphagia, deglutition, deglutition disorders

Introduction

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.

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 themes 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 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).

Table 1 Phases of ingestion [18]

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

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 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 oesophageal 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 radiological 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 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 phase 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 the residue after the swallow. In the pharyngeal phase the main problems are coughing, choking, and aspiration. (Table 2)

Table 2 Overall conclusions of the previous studies on dysphagia in HD

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

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 directly 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.

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 the 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 subsequently death [9-12], it is very important to assess and measure the dysphagic features as soon as possible. By investigating dysphagia at early stages of the disease and following patients as the disease progresses, a more accurate categorisation of dysphagia can be established that may lead to its proper management. To date no such studies exist in part because dysphagia is not recognised 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.

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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 manometric (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 need to be evaluated 2.Mealtime tips 3.Factors to consider in controlling dysphagia: -Medication -Environment -Equipement -Posture -Food choice -Breath control with swallowing -First aid

Design	Setting	Participant	Intervention	Outcome measures	Results
[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-values > 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	Setting Clinical setting	Participant HD (N=1)	Intervention Barium cineradiologic examination		Descending phase-type misdeglutition Involuntary movements of the larynx Tongue protrusion Cough at the beginning of phonation
[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

Chapter 3

Aspiration pneumonia and death in Huntington's disease

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Abstract

Huntington’s disease (HD) is a progressive neurodegenerative autosomal dominant disease characterized by choreatic and hypokinetic movements, disturbed behaviour, and cognitive decline. Pneumonia is the most common cause of death, followed by cardiovasculair diseases. It has been suggested that choking is the causative underlying factor for pneumonia in HD. As a detailed specification of the type of pneumonia has never been performed, we analyzed the records of our Brain Bank containing 224 cases to determine the exact cause of death and type of pneumonia. The conclusion is that the majority (86.8%) of our HD patients where the cause of death could be identified died from aspiration pneumonia.

Introduction

Huntington's disease (HD) is a progressive neurodegenerative autosomal dominant disease characterized by disturbed movements, changes in behaviour, and cognitive decline. Although the motor disturbances are both choreatic and hypokinetic, chorea is the most characterizing. Choreatic movements are irregular, unwanted involving not only the limbs and the trunk but also the respiratory and buccolingual muscles. 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. The mean age at onset is in the third and fourth decade of life and the disease duration about 15-20 years. [1] Death usually results from respiratory complications. Studies to the cause of death in HD found that the primary cause (variation of 33%-85.7%.) of death is pneumonia. [2] [3] [4] [5] [6] [7] None of the reported studies mentioned any details about the type of pneumonia. Pneumonia has several causative factors of which aspiration seems an important one. Pneumonia can be classified into different types, such as community-acquired pneumonia (CAP), hospital-acquired pneumonia (HAP), chemical pneumonia, and aspiration pneumonia. Although the exact percentage is unknown, most HD patients have dysphagia, especially in the advanced stage of the disease, therefore, aspiration is the most likely cause of the fatal pneumonia. The aim of the present study is to find out how often aspiration pneumonia is the primary type of fatal pneumonia in HD.

Materials and Methods

The records of all deceased HD patients from the Leiden University Medical Center (LUMC) brainbank in the Netherlands were collected. The diagnosis of HD was confirmed by family history and post-mortem pathological investigation, and since 1993 in most cases by DNA analysis. From all files the following information was collected: DNA confirmation, gender, year of birth, year of death, age at onset, location of death, naturally death, total body autopsy, described primary cause of death, other described underlying causes of death, repeated pneumonias, special remarks. The special remarks contained all available information about the type of pneumonia and the macroscopic description of the content of the lungs at autopsy. The clinical data from the patients were analyzed, when available, especially for dysphagia.

Statistical analysis

Descriptive statistics was used to obtain results, and Chi-square distribution was used to compare different variables. (SPSS 18.0)

Results

Two hundred twenty four charts of deceased HD patients were reviewed, of which 216 (111/104 male/female, one unknown) had records available, 147 listed a cause of death (Table 1), which was pneumonia in 81 (55%) cases. 38 of those 81 cases had an autopsy that included a description of the lungs. Of those 38 cases, 12 had gastric juices or food in the lungs, and 4 had giant cells, reaction which we defined as "death due to aspiration pneumonia", 17/38 had hyperemia, which we defined as "possible aspiration pneumonia", and only 5 showed bacteria or viruses, which was defined as "primary infectious pneumonia". For the cause of death aspiration pneumonia, no significant difference was found for gender and aspiration (χ^2 =.995). From all 216 records, in 69 cases no primary cause of death was given. In 15/69 records the clinical information revealed information about dysphagia, and in 8/69 records information about repeated pneumonias in the end stage was present. In 4/69 records, a description of the microscopy revealed of giant cells and hyperemia in the lungs.

Table 1 Primary cause of death in 147 patients with Huntington disease

Cause of death	N=147	100%
Known Cause:		
-pneumonia	81	55.1
-suffocation	6	4.1
-pulmonary embolism	6	4.1
-cachexia	11	7.5
-cardiac diseases	16	10.9
-other neurological diseases	3	2.0
-shock/sepsis	7	4.8
-suicide	2	1.4
-euthanasia	5	3.4
-other causes	10	6.8

Discussion

With the information available, we found it plausible that significant more patients died from aspiration pneumonia, instead of a primary infectious pneumonia. It is most likely that dysphagia is the causative factor for this aspiration pneumonia. The relation between dysphagia leading to aspiration pneumonia is confirmed in patients with Parkinson’s disease (PD) and in elderly people. [8] [9] The leading cause of death in PD is aspiration pneumonia, for as much as 70%. And these patients also suffer from dysphagia. [8] [10] [11] In elderly people it is found that dysphagia is a common problem, and that dysphagia is the major pathophysiologic mechanism leading to aspiration pneumonia. [9] [12] Another interesting point is that it is likely that patients who died from an infectious pneumonia, still died because of aspiration. Because in most cases the cultures consisted of the staphylococcus aureus, klebsiella and candida albicans. When patients aspirate their saliva, patients can develop pneumonia, especially patients in the last stage of HD, who are mostly cachectic, in bad condition and with a poor resistance. Previous studies to the role of chronic conditions, health behaviors, and nutritional status have shown that these factors increases the risk of pneumonia. [13] [14] [15] On investigating the files of the deceased HD patients, a large proportion of the files, 32% did not contain the primary cause of death. Therefore, it seems that the files of the deceased HD patients are relatively inaccurate in recording causes of death. Other studies have also shown an inaccuracy rate. Haines and Conneally [4] had an overall rate of 66%. Alderson [16] found an overall accuracy rate of 61%. Thus, our overall rate of 68.1% is not unusual. Because of this overall rate, it seems likely that more patients died of aspiration pneumonia. Some of these patients were described with suffering from dysphagia, and repeated pneumonias in their last period of life. As said these issues have influence on developing aspiration pneumonia. To conclude, our data suggest that aspiration pneumonia is the most prominent primary cause of death in HD, which is in accordance with the literature. As the source data showed to be rather incomplete, lacking information about the clinical status of the patients regarding dysphagia, we started to develop a dysphagia assessment scale to get better insight in the prevalence of dysphagia and its consequences for the development of pneumonia.

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Chapter 4

The Huntington’s Disease Dysphagia Scale

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Abstract

Background
Little is known about the swallowing disturbances of patients with Huntington's Disease; therefore, we developed the Huntington's Disease Dysphagia Scale.

Methods
The scale was developed in four stages: 1) item generation, 2) comprehension testing, 3) evaluation of reliability, 4) item reduction and validity testing. The questionnaire was presented twice to 50 Huntington's disease patients and their caregivers. The Kruskal-Wallis test was used to evaluate whether the severity of swallowing difficulties increased with advancing disease. Pearson's correlation coefficient was used to examine the construct validity with the Swallowing Disturbance Questionnaire.

Results
The final version contained 11 items with five response options and exhibit a Cronbach's alpha coefficient of 0.728. The severity of swallowing difficulties was significantly higher in more advanced Huntington's disease. The correlation with the Swallowing Disturbance Questionnaire was 0.734.

Conclusion
We developed a valid and reliable 11-item scale to measure the severity of dysphagia in Huntington's disease.

Introduction

Huntington’s disease (HD) is a progressive neurodegenerative disease characterized by disturbed movements, behaviour, and cognition.[1] Patients with HD suffer from dysphagia, and patients have swallowing difficulties in all phases of ingestion.[2-7] Carefully designed longitudinal studies on dysphagia in HD are required to clarify when dysphagia emanates, what the dysphagia symptoms in the different stages of ingestion are, and whether the severity of dysphagia increases with advancing HD. This may lead to increased awareness and a treatment that is more tailored to the patient’s individual needs. A first requirement to address these questions is the availability of a good measurement instrument tailored to the specific swallowing impairments of this population, but because such an instrument does not exist, we had a goal of developing a patient-reported outcome measure, the Huntington’s Disease Dysphagia Scale (HDDS).

Patients and methods

Expert team

The task of the HD expert team was to generate items that evaluated swallowing difficulties in general and to add items that were specific to HD. The items were complemented with items drawn from the literature. The experts were then asked to rate the relevance of each item (0=not important, 5=very important). Items with a score of 3 or greater were included in the initial scale. The professions of the experts were neurologists (n=2), speech-and-language therapists (n=4), an ear-nose-throat specialist (n=1), radiologist (n=1), nursing home physicians (n=2), and a methodologist (n=1). Almost all experts were experienced with HD, except one speech-and-language therapist, the ear-nose-throat specialist, and the radiologist, but they were experienced in dysphagia.

Participants

Patients with a CAG repeat size of 36 or greater and without other diseases that could affect swallowing were included. Patients from all three clinical stages[8] - in our study defined as stage I: patients living at home (n=23); stage II: patients living at home who attended day care (n=18); and stage III: patients living in a nursing home (n=14)- were enrolled. Stage I patients were consecutively selected from the outpatient neurology department of the Leiden University Medical Centre (LUMC), whereas patients in stage II and III were consecutively selected from a Huntington Care center, a center specializing in care and treatment of HD patients. The center provides specialized units for HD patients who are in the mid or end-stage, and also offers the possibility for early HD patients to receive day care.

Proxy information on the patient's swallowing problems was obtained from the partner or caregiver, or from a close friend.

The study was approved by the medical ethics committee of the LUMC. All patients gave informed consent.

Scale development

The scale was developed in Dutch in four stages: (1) item generation, based on a literature search and input by experts; (2) item comprehension testing in patients; (3) evaluating reliability in patients and relatives; (4) item reduction and construction of the final scale, followed by examining the scale for the presence of floor and ceiling effects (i.e., 15% or greater of patients scoring the minimum or maximum possible total score), overall reproducibility, as well as construct validity by comparison with the Swallowing Disturbance Questionnaire (SDQ).[9]

Item generation

A large pool of questions regarding dysphagia in relation to HD at the level of disability was collected. Items were drawn from other existing dysphagia scales,[9-21] the literature on dysphagia in HD,[2-7] and from our clinical experience with HD.

Item comprehension testing

Intelligibility of questions was investigated in 10 HD patients sampled from the various clinical stages using the "Thinking aloud" procedure. Patients were also encouraged to add items. During this phase, items were rephrased continuously until all items were easily understood and interpreted correctly. The response options addressed the frequency of the problem: (0) almost never, (1) seldom, (2) sometimes, (3) frequently, (4) almost always.

Evaluating reliability in HD patients

The initial questionnaire was sent to all of the included 55 patients with HD. To measure intra-rater reliability, the same questionnaire was sent again to these patients 2 weeks after the first questionnaire had been received. Test-retest reliability of the individual items and total score were analysed with the Intraclass Correlation Coefficient (ICC; one-way random effects model). Items with ICC's less than 0.30 were removed. Relatives of the included patients also received the questionnaire twice and rated their impression of the patients’ swallowing difficulties. The aim of this proxy information was to find out whether close relatives were able to identify the patient’s swallowing difficulties. When patients did not return the first questionnaire, but their relatives did, both patients and relatives received the second questionnaire after 2 weeks. The ICC was used to calculate the agreement between both outcomes. A total score was calculated if three or fewer values were missing, in which the mean of the other values was used to replace the missing values; no total score was computed if four or more values were missing.

Item reduction, scale construction, and construct validity

After removal of items with low intra-rater reliability, additional item reduction was based on the strength of the inter-item correlation, where items with high (>0.80; indicating redundancy) or low (<0.20; indicating incoherence) item-total correlations were removed. Additionally, items whose presence resulted in a decrease of Cronbach’s alpha were also removed.

To examine the construct validity, the final questionnaire was correlated with the SDQ[9] (Pearson’s correlation coefficient), which was completed simultaneously with the HDDS by all patients and relatives. The SDQ was chosen because this scale emerged as a valid tool to detect early dysphagia in patients with Parkinson’s disease.

Psychometric performance SDQ

Test-retest reliability of the SDQ was calculated with an ICC (one-way random effects model). Cronbach’s alpha of the SDQ was also calculated.

Dysphagia progression during the disease

To evaluate whether patients in more advanced stages of HD perceived more swallowing abnormalities than those in the earlier stages, a Kruskal-Wallis test was used.

Results

Item generation

In total, 14 items were considered relevant by the experts. These items related to the four different phases of ingestion: preparatory oral phase (3 items), oral phase (2 items), pharyngeal phase (7 items), and esophageal phase (2 items). The questions were all found intelligible by the patients.

Patients

Fifty (out of 55) patients (91%; 22 men; mean ± SD age: 52 ± 11 years; mean ± SD disease duration: 10 ± 5 years) and 50 relatives (29 partners, 15 carers, 6 close friends) returned the questionnaires (Table 1). The five nonresponding patients were in stage II (n=1), and stage III (n=4). Two (out of 55) patients partially completed the questionnaire (≥4 missings). Forty-eight (out of 55) patients (87%; 21 men; mean ± SD age: 54 ± 11 years; mean ± SD disease duration: 10 ± 5 years) and 45 relatives (26 partners, 15 carers, 4 close friends) completed the second set.

Table 1 Baseline sociodemographic, clinical and functional characteristics of the Huntington's disease patients

Patients	
N (Males/Females) (n=50)	50 (22/28)
Age, y	52.2 (11.3)
Age at onset, y	43.1 (9.8)
Disease duration, y	9.6 (5.2)
CAG repeat length (n=46)	43.9 (3.1)
N Stages of HD (n=50)	
Clinical stage I	23
Clinical stage II	17
Clinical stage III	10
N Proxies (n=50):	
Partners	29
Carergivers	15
Close friends	6

Unless otherwise stated, values are means (SD).
Abbreviations: SD, standard deviation; HD, Huntington's disease; CAG, repeat length triplets number (≥36, pathological range)

Item reduction, scale construction, and construct validity

The ICC of one question ("wet voice after swallowing") was very low (ICC=0.189) and therefore removed. Cronbach's alpha for the 13 remaining questions was 0.670, which increased to 0.728 after removal of the items "residue in the mouth, cheek or teeth" and "producing sounds during eating or drinking" (Table 2). Finally, 11 items were included in the HDDS (Table 2). These 11 items related to the various swallowing phases as follows: preparatory oral phase (3 items), oral phase (1 item), pharyngeal phase (5 items), and esophageal phase (2 items), indicating that all phases of ingestion were still covered.

Of all patients, 9.1% had a total score of 11, whereas none of the patients had the maximum possible score, indicating absence of floor and ceiling effects. The test-retest reliability of the final HDDS as measured with the ICC was 0.754. The correlation with the SDQ was 0.734. The test-retest reliability (ICC) of the SDQ was 0.597.

Table 2 Final Huntington's Disease Dysphagia Scale

	Item	Cronbach's alpha if item deleted
1.	Do you drool during the day?	0.697
2.	Do you feel you have too much food in your mouth, so that you cannot swallow anymore?	0.705
3.	Does food come out of your mouth?	0.709
4.	Does food or beverage come out of your nose?	0.710
5.	Are you always successful in swallowing food or beverage?	0.714
6,.	Do you choke food or beverage?	0.693
7.	Do you cough while swallowing food or beverage?	0.701
8.	Do you sneeze while swallowing food or beverage?	0.718
9.	Do you sometimes unexpectedly breathe while you swallow food or beverage? In other words: Suddenly breathe when you swallow?	0.715
10.	Does food or beverage sometimes come back in your mouth, while you had actually swallowed it?	0.702
11.	Do you sometimes feel if there is a lump in your throat?	0.724

Cronbach's alpha of the final scale = 0.728 (n=48). 'Cronbach's alpha if item deleted' indicates Cronbach's alpha value of the total scale if the particular items were removed. This shows that further removal of any item would result in a decrease of Cronbach's alpha.
Response options;
Question 1-4 and 6-11: 1 No, almost never; 2 Yes, seldom; 3 Yes, sometimes; 4 Yes, frequently; 5 Yes, almost always
Question 5: 1 Yes, almost always; 2 Yes, frequently; 3 Yes, sometimes; 4 Yes, seldom; 5 No, almost never

Dysphagia severity across the disease stages

The Kruskal-Wallis test, used to evaluate whether the severity of the swallowing difficulties measured with the HDDS increased with increasing disease severity according to the three clinical stages, was significant (*P* = 0.037). Kruskal-Wallis was also significant for the proxy scores (*P* = .0005).

Discussion

We developed an 11-item scale to measure dysphagia in HD. The instrument can be used to monitor swallowing difficulties from early manifest to end-stage disease. The HDDS demonstrates good construct validity, as shown by the high correlation with the SDQ.

Nevertheless, for evaluating dysphagia in HD, the HDDS may be preferred, not only because it more fully covers the specific domain (i.e., better content validity), but also because of its superior reproducibility in this population. Agreement between the patients' and partners' HDDS scores was low, which indicates that relatives have a quite different perception of the patient's swallowing difficulties than the patients themselves. Because reproducibility and construct validity of the HDDS was assessed in patients and exhibited high values-indicating that one can be confident of the patients' ability to complete this scale- completion by proxies is not recommended. However, when HD patients in the very last stage are confronted with cognitive decline, it is recommended that carers fill out the HDDS. At present the HDDS scale is developed and validated in Dutch, but the scale has been translated into English.

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Chapter 5

Swallowing impairment in Huntington's disease: videofluoroscopic findings

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Submitted

Abstract

Objective

Dysphagia is a frequent finding in Huntington's disease (HD) and aspiration pneumonia is known to be a frequent cause of death. However, it is unknown what the specific dysphagia features in HD are, and if dysphagia is more severe in more advanced disease. The goal of our study was to identify the specific dysphagia features in HD, and to investigate the severity of dysphagia in the different stages of HD.

Methods

Forty-five HD patients (clinical stage I: n=13, stage II: n=18, stage III: n=14) participated and underwent videofluoroscopy using a variety of liquids and solid food. Oral, pharyngeal, and esophageal characteristics were analyzed per clinical stage.

Results

Dysphagia was found in 78% of the patients in all stages and was more severe for liquids (p=0.003) than solid food (p=0.035). Disturbances were found in especially the oral, and oropharyngeal phase of the swallow. Also disturbances in the duration times of the swallow were found in the oral and pharyngeal phase.

Conclusions

Swallowing abnormalities due to oral phase problems and oropharyngeal dysphagia are a frequent finding in HD patients. Dysphagia features are present in all three clinical stages of the disease. Given the high rate of dysphagia and the early onset of this problem in the course of HD potential swallowing disturbances deserve continuous attention from the earliest stages of the disease.

Introduction

The act of swallowing is a complex coordination of the sequence of activation and inhibition of muscles in the mouth, pharynx, larynx, and esophagus [1]. Swallowing is divided into four phases: the preparatory oral phase, oral phase, pharyngeal phase, and esophageal phase [2]. The preparatory oral phase includes transport from the bolus in the mouth, and mixing it with saliva. During the oral phase the bolus is propelled from the oral cavity to the pharynx. The pharyngeal phase refers to the passage through the pharynx and into the esophagus. During the esophageal phase the bolus is transported to the stomach by the peristaltic movements of the esophagus.

In Huntington's disease (HD) dysphagia is a frequent finding. HD is an autosomal, progressive, neurodegenerative disease, and is characterized by disturbed movements, behaviour, and cognition [3]. HD is caused by a CAG repeat expansion of the *HTT* gene on chromosome 4. The mean age at onset is in the third or fourth decade of life, and the disease duration is about 15 to 20 years [3-10]. Three clinical stages of the disease are described [11]. During stage I patients are independent and develop their first symptoms; in stage II patients start to be dependent and the symptoms are more generalized; during the last stage patients are completely dependent for all daily-life activities. Many patients with HD die of aspiration pneumonia, which has a close relation with dysphagia. In HD aspiration pneumonia is even the most frequently encountered primary cause of death [12]. It is therefore important to study the specific dysphagia features in HD, and their progress during the disease. It has been reported that patients with HD have dysphagia in all phases of ingestion [13-18]. However, it is not known when the dysphagia starts and how it progresses in the different stages of the disease. Further, specific dysphagia features, like aspiration, penetration, spilling, residue have not systematically been studied in HD. Also duration times, like oral transit time (OTT), pharyngeal transit time (PTT), oropharyngeal transit time (OPTT), pharyngeal delay time (PDT), pharyngeal passage time (PPT), and velopharyngeal closure time (VPCT) are unknown. The goal of our study was to identify these specific dysphagia features in HD at all three stages of the disease.

Methods

Subjects

Forty-five HD patients (23 men; mean ± SD age: 54 ± 12 years; mean ± SD disease duration: 10 ± 5 years) participated in the study (Table 1). Patients with a CAG repeat size ≥36 and without other diseases that could affect swallowing were included. Patients from all three clinical stages- in our study defined as stage I: patients living at home (n=13); stage II: patients living at home and having day care (n=18); and stage III: patients living in a nursinghome (n=14) - were enrolled. Patients in stage I were consecutively selected from April 2012 until June 2012 from the outpatient department of neurology of the Leiden University Medical Center (LUMC), while patients in stage II and III were consecutively selected in the same period from a Huntington Care center. The study was approved by the medical ethics committee of the LUMC. All patients gave informed consent.

Table 1 Clinical and demographic information of the Huntington's disease patients

Patients	
N (males/female) (n=45)	45 (23/22)
Age, y	53.7 (11.8)
Age at onset, y	43.8 (10.1)
Disease duration, y	9.8 (5.2)
CAG repeat length (n=37)	44.2 (3.2)
N Stages of HD (n=45)	
Clinical stage I	13
Clinical stage II	18
Clinical stage III	14

Unless otherwise stated, values are means (SD).
Abbreviations: SD, standard deviation; HD, Huntington's Disease; CAG, repeat length triplets number (≥36, pathological range)

Imaging acquisition and analysis

Each patient underwent videofluoroscopy (VFSS). The protocol for the VFSS involved presentation of two swallows each of 3cc and 10cc thin liquid, 5cc thick liquid, and a piece of bariumbread. For safety reasons water soluble liquids were used. Thick liquids were thickened with Nutilis Clear®. VFSS were recorded on a Toshiba Ultimax-i. The video recordings had 15 frames per second. Each video was analyzed in slow motion mode to identify any swallowing disorder. The videos were analyzed separately by two raters, one rater was experienced in HD and dysphagia, and the second rater was experienced in dysphagia. The following features were analyzed: tongue protrusion, hyperextension of the head, adequate mastication; spilling before and during the swallow; penetration and aspiration; residue in the valleculae, and piriform sinus. The following duration times (in sec) were calculated: OTT, PTT, OPTT, PDT, PPT, and VPCT. For penetration and aspiration the Penetration-Aspiration-Scale (PAS) [19] was used; a score between 2 to 5 on this scale was defined as penetration, while a score of 6 to 8 was defined as aspiration [19]. The duration measures were defined as listed in Table 2. These measures all start with the first frame showing the first movement and end with the last frame on which the movement is detected. Investigating if dysphagia increased across the different stages of HD was performed for the following features: tongue protrusion, hyperextension of the head, inadequate mastication, and for PAS-score. Based on all analyzed features patients were also classified as dysphagic or not if they had disturbances in swallowing liquids or solid food. The examination was also recorded on video to assess the oral intake and oral preparatory phase.

Table 2 Definitions of the duration measures

Duration times	Applied definitions
Oral transit time (OTT) <1.5sec*	The first frame showing the first movement of the head of the bolus to the oropharynx until the first frame showing the bolus head reaching the point where the lower edge of the mandible crosses the tongue base
Pharyngeal transit time (PTT) <1sec*	The first frame showing the bolus head reaching the point where the lower edge of the mandible crosses the tongue base until the first frame showing the bolus leaves the cricopharyngeal area and the tail of the bolus is in the esophagus
Oropharyngeal transit time (OPTT) <2.5sec	The first frame showing the first movement of the head of the bolus to the oropharynx until the first frame showing the bolus leaves the cricopharyngeal area and the tail of the bolus is in the esophagus
Pharyngeal delay time (PDT) 0-0.2sec*	Time from the bolus head reaching the point where the lower edge of the mandible crosses the tongue base until the first laryngeal elevation in the swallow is seen
Pharyngeal passage time (PPT) <1sec*	The first frame where laryngeal elevation in the swallow is seen until the first frame showing the bolus leaves the cricopharyngeal area and the tail of the bolus is in the esophagus
Velopharyngeal closure time (VPCT) <1sec**	The first frame showing the first contact of the palatum to the pharyngeal wall until the last frame showing contact of the palatum to the pharyngeal wall.

normal value [2]
**Logemann described no specific number for the VPCT, but states that the velopharynx must be closed, just the few moments when the bolus is passing [2]. We therefore took <1sec.

Statistics

A 0ne-sample t test was used to compare the mean outcomes of the duration measures to the maximal normal values described by Logemann [2]. The Kruskal-Wallis test was used to investigate if the dysphagia is more severe in more advanced stages of the disease. For the statistical analyses SPSS 20 was used.

Results

All 45 HD patients underwent VFSS. Thirty-five of all participants (77.8%) were diagnosed as dysphagic, from which 32 (71.1%) had disturbances for solid food, and 30 (66.7%) had disturbances for liquids. The outcomes of the swallowing abnormalities and the duration times are shown in Table 3. The outcomes of the duration times compared to the maximal normal values are shown in Table 4. Significant shorter duration times were found in the OTT for all consistencies. The OPTT was shortened for liquids. Also a decreased VPCT was found for liquids (3cc and 5cc) and solid food. Significant longer duration times were seen in the PTT for solid food, and in the PPT for liquids. The findings of the analysis that examined if dysphagia is more severe in the more advanced stages of the disease are presented in Table 5. Results showed that tongue protrusion and inadequate mastication increased significantly (respectively p=.028, and p=.012). Hyperextension of the head does not increase significantly (p=.081). A score of ≥2 on the PAS showed a significant increase (p=0.018) across HD stages. The analyses further showed that dysphagia in patients was more severe for liquids (p=0.003) and solid food (p=0.035) across the various stages of HD.

Table 3 Videofluoroscopic findings of the preparatory oral phase, oral phase and pharyngeal phase of ingestion

	3cc thin liquid	10cc thin liquid	5cc thick liquid	Solid	Percentage
	%[1]	%[1]	%[1]	%[1]	%[1]
Tongue protrusion					12.2
Hyperextension					31.8
Mastication					
- moderate				29.3	
- inadequate				14.6	
Total				43.9	
Spilling					
- before	32,6	23.8	25.6	20.5	
- during	23.3	45,2	30,8	10,0	
Total	55.9	69.0	56.4	30.5	
PAS					
- penetration	15.9	14.0	4.8	0	
- aspiration	6.8	13.9	7.3		
Total	22.7	27.9	12.1		
Residue					
- valleculae					
mild	20.5	27.9	25.0	7.3	
severe	20.5	25.6	25.0	24.4	
Total	41.0	53.5	50.0	31.7	
- piriform sinus					
mild	27.3	30.2	22.5	2.4	
severe	4.5	20.9	22.5	7.3	
Total	31.8	51.1	45.0	9.7	
	sec	sec	sec	sec	
OTT (<1.5 sec)[2]	.585 (.606)	.367 (.292)	.502 (1.111)	.851 (1.066)	
PTT (<1 sec)[2]	.954 (.403)	.999 (.432)	1.199 (1.145)[1]	1.651 (1.552)[1]	
OPTT (<2.5 sec)[2]	1.558 (.724)	1.334 (.498)	1.699 (.879)	2.187 (1.882)	
PDT (0-0.2 sec)[2]	-.387 (.605)[1]	-.172 (.443)[1]	-.196 (1.372)[1]	.189 (1.179)	
PPT (<1 sec)[2]	1.319 (.567)[1]	1.165 (.475)[1]	1.413 (.544)[1]	1.336 (1.114)[1]	
VPCT (<1 sec)[3]	.705 (.320)	.850 (.659)	.788 (.405)	.781 (.521)	

[1] patients with abnormal values

[2] normal value [2]

[3] Logemann described no specific number for the VPCT, but state that the velopharynx must be closed, just the few moments when the bolus is passing [2]. We therefore took <1sec.

Abbreviations: PAS, Penetration Aspiration Scale; OTT, Oral Transit Time; PTT, Pharyngeal Transit Time; OPTT, Oral Pharyngeal Transit Time; PDT, Pharyngeal Delay Time; PPT, Pharyngeal Passage Time; VPCT, VeloPharyngeal Closure Time

Table 4 Duration measures in relation to the maximal normal values

Test variable	Max normal value	n	t	95% CI	p
OTT	1.5 sec				
3cc thin liquid		40	-9.548	-1.109_-.721	<0.001
10cc thin liquid		38	-23,917	-1.229_-1.037	<0.001
5cc thick liquid		38	-5.536	-1.363_-.633	<0.001
solid		33	-3.496	-1.027_-.271	.001
PTT	1 sec				
3cc thin liquid		40	-.725	-.175_ .083	.473
10cc thin liquid		41	-.017	-.137_ .135	.987
5cc thick liquid		39	1.085	-.172_ .570	.285
Solid		37	2.552	.134_ 1.169	.015
OPTT	2.5 sec				
3cc thin liquid		39	-8.129	-1.177_-.707	<0.001
10cc thin liquid		38	-14.443	-1.330_-1.002	<0.001
5cc thick liquid		38	-5.614	-1.090_-.512	<0.001
Solid		33	-.955	-,981_ .354	.347
PDT	0.2 sec				
3cc thin liquid		40	-6,135	-.780_-.393	<0.001
10cc thin liquid		41	-5.381	-.512_-.233	<0.001
5cc thick liquid		37	-1.758	-.854_ .061	.087
Solid		37	-.055	-.404_ .382	.956
PPT	1 sec				
3cc thin liquid		40	3.556	.138_ .500	.001
10cc thin liquid		41	2.220	.015_ .315	.032
5cc thick liquid		37	4.621	.232_ .595	.000048
Solid		37	1.883	-.036_ .707	.075
VPCT	1 sec**				
3cc thin liquid		40	-5.830	-.397_-.192	<0.001
10cc thin liquid		39	-1.422	-.364_ .064	.163
5cc thick liquid		38	-3.228	-.345_-.079	.003
Solid		37	-2.560	-.393_-.046	.015

One sample T Test with 95% Confidence Interval of the difference. Transit times as test variables. *, normal values in sec described by Logemann [2]. **Logemann described no specific number for the VPCT, but state that the velopharynx must be closed, just the few moments when the bolus is passing [2]. We therefore took 1sec.

Abbreviations: n, numbers of patients; SD, standard deviation; t, T Test: one sample; CI, Confidence Interval; OTT, Oral Transit Time; PTT, Pharyngeal Transit Time; OPTT, Oral Pharyngeal Transit Time; PDT, Pharyngeal Delay Time; PPT, Pharyngeal Passage Time; VPCT, VeloPharyngeal Closure Time; sec, seconds; p, p-value

Table 5 Dysphagia during the progression of Huntington’s Disease

	n*	Mean rank	Chi-square	p	p**
Tongue protrusion					
- stage I	12	18.50^{1}	7.129	0.028	.401^{1}
- stage II	17	19.71^{2}			.032^{2}
- stage III	12	25.33^{3}			.058^{3}
Mastication					
- stage I	12	19.29^{1}	8.282	0.016	.520^{1}
- stage II	18	17.36^{2}			.029^{2}
- stage III	11	28.82^{3}			.008^{3}
Hyperextension of the head					
- stage I	13	17.19	5.018	0.081	
- stage II	18	24.06			
- stage III	13	25.65			
PAS					
- stage I	13	16.19^{1}	8.041	0.018	.054^{1}
- stage II	18	23.06^{2}			.005^{2}
- stage III	13	28.04^{3}			.221^{3}
Dysphagia for Liquids					
- stage I	13	14.92^{1}	11.841	0.003	.024^{1}
- stage II	18	24.25^{2}			.001^{2}
- stage III	14	28.89^{3}			.144^{3}
Dysphagia for solid					
- stage I	13	17.93^{1}	6.680	0.035	.266^{1}
- stage II	18	22.17^{2}			.020^{2}
- stage III	14	28.79^{3}			.050^{3}

Kruskal-Wallis test for measuring the severity of dysphagia in the three different stages. *some measurements have a number of patients <45. The missing patients could not be judge properly on that issue. **Asymp sig (2-tailed) for post-hoc differences using the Mann-Whitney U test: ^{1} stage I-II; ^{2} stage I-III; ^{3} stage II-III.
Abbreviations: n, numbers of patients; df, degrees of freedom; p, p-value; PAS, Penetration Aspiration Scale

Discussion

A videofluoroscopic study was done to evaluate the specific dysphagia problems in HD patients. Our results show that swallowing abnormalities due to problems in the oral phase as well as oropharyngeal dysphagia are frequent findings in patients with HD. Dysphagia features were present in all three clinical stages of the disease, and were more severe in the more advanced stages. In the preparatory oral phase some patients exhibited tongue protrusion during swallowing, and almost half of patients had difficulty with mastication of solid food. During the oral phase many patients had spilling before and during the swallow. Further, patients had a significantly shorter OTT in comparison to the maximal normal value. This is in line with existing knowledge that HD patients have the habit to rapidly and impulsively consume food and beverage. The shorter duration time may reflect the inability to masticate properly. It is also possible that patients do not have the possibility to masticate properly due to poor lingual control or chorea. Irrespective of the precise mechanism it follows that insufficiently chewed food is swallowed. It is striking that patients in stage I seems to masticate poorer than patients in stage II. We do not have a clear explanation for this finding. Our results further show that more than a quarter of patients have residue of food in the valleculae during the pharyngeal phase. This bears the risk of suffocation when the residue falls into the trachea. An earlier study on aspiration pneumonia and death in HD showed that 4.1% of the patients died due to suffocation [12]. Complaints of patients about a lump in their throat when swallowing should therefore be taken very seriously and timely dysphagia management is warranted in such cases. Another finding in the pharyngeal phase was penetration and aspiration for all fluid consistencies. Also spilling before or during swallowing, and the presence of residue were seen for liquids and solid food. Spilling during swallowing, aspiration and residue were typically more pronounced with larger boluses of liquids (10cc versus 3cc). This may be related to the temporal coordination of swallowing; typical for the swallow of a small amount of beverage is that the swallow starts with the oral phase, followed by triggering the pharyngeal phase. For swallowing a large amount of beverage, in contrast, a simultaneous effectuation of the oral and pharyngeal phase is needed [2]. Based on our results smaller amounts of fluid intake seem therefore recommendable for HD patients. Another important issue is the bolus viscosity. When the viscosity of the bolus increased - in this study 5cc thick liquid was offered - there was less penetration and aspiration compared to small or large amounts of fluid. It is known that as the bolus viscosity increases to the thickness of pudding, the tongue base and the pharyngeal walls require more pressure to trigger a safe swallow [2]. In Parkinson's disease (PD) it was observed that when patients swallowed pudding-thickened liquids, they had residue in the valleculae and piriform sinus, and thus had the risk to aspirate after the swallow [2]. When the viscosity of liquids increased to the thickness of honey, Butler et al. [20] found that less pharyngeal pressure was needed. Honey-thick liquids seem the ideal viscosity to pass the pharynx without much pharyngeal driving force [20]. In our study we found less aspiration while swallowing honey-thick liquid, although piriform sinus and vallecular residue was present in 45-50% of the patients. Since considerably less penetration and aspiration in comparison to thin liquid was found, we think it would be best to recommend honey-thick liquids as soon as HD patients complain of dysphagia. Given the common finding of residue, we also recommend to trigger a dry swallow after each swallow, to empty both valleculae and piriform sinuses and reduce the risk of aspiration after the swallow.

Pharyngeal duration times were particularly disturbed. An increased PTT was found for thick liquids and solid food. It is likely that thick liquid and solid food need more time to pass the esophagus because of their thick consistency. Yet, this was not associated with an increased frequency of residue. Further, an accelerated laryngeal elevation (PDT) was measured in all fluid swallows and a prolonged PPT in all consistencies. We hypothesize that the latter serves as a compensation strategy for the early initiation of the laryngeal elevation and prevents aspiration of hypopharyngeal residue.

In examining the clinical stages of HD we not only noted a significant increase of tongue protrusion as well as poor mastication, but also an increase of the PAS-score. The increased swallowing dysfunction with a higher incidence of penetration and aspiration in higher stages of the disease is consistent with the reported high number of patients that die of aspiration pneumonia in the end stage of HD. However, swallowing abnormalities in HD can already be found in clinical stage I. It is our experience that patients in stage I sometimes complain of swallowing abnormalities, but that they are seldom referred for a swallowing examination by a speech and language therapist, although there is general consensus that early detection and effective intervention can help to prevent choking [21]. In this respect it is helpful for patients to receive information and advice about the progression of dysphagia during the disease [21]. This information can now be offered more specific based on our findings.

To conclude, disturbances in oral intake and swallowing abnormalities due to oral phase problems and oropharyngeal dysphagia are a frequent finding in HD patients in all three clinical stages of the disease. Based on our findings the following recommendations for the treatment of swallowing disturbances are made: decreased volume and increased viscosity of liquid intake, and triggering a dry swallow after each swallow in all food consistencies.

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Chapter 6

Oral feeding in Huntington´s disease: a guideline document for speech and language therapists

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Practice Points

- Speech and language therapy has an important role in the management of Huntington's disease.
- Management of oral feeding difficulties in Huntington's disease is a challenge but implementation of a systematic program of intervention can improve swallow safety and efficiency and reduce the occurrence of pneumonia.
- Early referral to speech and language therapy is recognized as good practice. It facilitates accumulation of baseline data, which aid clinical decision making on timely assessment and/or intervention. This is crucial in Huntington's disease because of the multiple factors involved, including cognitive and behavioral changes.
- In early-stage disease the individual with Huntington's disease may have no symptoms of dysphagia but speech and language therapists may still have a role in the provision of information and advice.
- In mid-stage disease there is wide variability in the presentation of oral feeding difficulties and regular assessment and review is required to identify changes and their impact.
- There is significant overlap in presenting oral feeding difficulties in the mid and late stages and as the disease progresses the challenge of maintaining adequate nutrition and hydration becomes greater due to the complexities involved.
- The speech and language therapist will contribute to the discussions and multidisciplinary decision making regarding non-oral feeding/supported feeding. Decisions should involve the individual and significant carers, the discussions having been introduced whilst the individual's cognitive abilities allowed full participation.

Summary

Speech and language therapists play an important role in the management of communication difficulties in Huntington’s disease (HD). As the disease progresses the effectiveness of communication becomes increasingly compromised by a combination of changes in motor function, diminishing cognitive linguistic abilities and neuropsychiatric changes, such as depression and apathy. The complexities and challenges presented by communication breakdown in HD require comprehensive assessment and interventions that are responsive to the changing motor, cognitive and emotional needs of the individual. The European Huntington’s Disease Network Standards of Care Speech and Language Therapy Working Group has brought together expert speech and language therapists from across Europe to produce guidelines to improve the management of communication disorders for individuals with HD. The guidelines were developed with the aim of promoting timely and appropriate assessment and focused management throughout all stages of the disease. Literature was thoroughly searched and evaluated in an attempt to ensure that the guidelines are based on available evidence. However as there is a paucity of good-quality, high-level evidence the guidance is based predominantly on expert opinion and consensus. The provision of care varies widely between countries in Europe and the implementation of these guidelines aims to help improve the quality of care delivered to individuals with HD.

Huntington’s disease (HD) is a mainly adult-onset, familial disorder that can affect both men and women [1]. Symptom onset usually occurs by the age of 40 years, but this can be very variable. Age of onset is related to the size of the mutation, which is an unstable expansion of a CAG sequence in the gene [2]. The classical triad of clinical signs and symptoms include a movement disorder, cognitive impairment and behavioral changes. These features cause complex management problems. Described by George Huntington in 1873, this disease continues to devastate families and perplex the clinicians who care for them.

Changes in eating and drinking in HD are impacted by physical, cognitive and affective factors. Management of oral feeding needs to take account of these, as well as the more traditional approach, which may have a narrower focus on oral and pharyngeal dysfunction. The guidelines presented here adopt this more holistic approach.

Dysphagia is a common problem in HD [3,4]. As the disease progresses the swallowing function can become greatly impaired, raising the risks of aspiration and associated bronchopulmonary infections, airway obstruction, dehydration and malnutrition. Dysphagia and aspiration are cited as common triggers for complications leading to death in the later stages of HD

[5]. Management of swallowing difficulties in HD is a challenge but implementation of a systematic program of diagnosis and management of dysphagia can improve swallow safety and efficiency and reduce the occurrence of pneumonia. Some studies support the implementation of interventional programs in managing dysphagia in HD and report favorable results with some improvements reported to have persisted for as much as 3 years [4,6,7]. In general, there is a paucity of high level evidence to support the management of dysphagia in HD, with case study and expert opinion levels forming the main body of literature [8]. However, the Standards of Care Working Group has produced a consensus view reflecting the opinions of experts working in this field and using available evidence from the literature. It is hoped this will enable others to provide optimum intervention and care to the HD patient and their family.

The aims of this guideline are:

- To assist practitioners in reducing the risks of aspiration and airway obstruction and in applying appropriate methods to support food and fluid intake;
- To inform the optimal, individualized management of swallowing disorder in HD in order to enable uniformity of care internationally and to document speech and language therapy practice, and the different approaches around Europe to the management of HD;
- To review scientific literature to identify evidence-based practice in managing oral feeding difficulties in HD;
- To establish expert opinion and consensus regarding usefulness and efficacy of interventions;
- To make recommendations on effective practice, based on the literature, clinical data and expert opinion available, therefore, promoting standard methodologies.

Stages of HD

There have been several attempts to define the stages of progression of HD [9,10]. Current research may provide further options [10]. For these guidelines recommendations have been made for three stages of the disease; early, mid and late.

- Early stage

Early disease cannot be defined as the time of diagnosis since diagnosis can be made at variable times. Most individuals present with early neurological and psychiatric features that may have started to cause difficulties at work and home.

- Mid stage

Mid-stage disease is reached when the affected person has to cease work, or at least change employment to a post that is less challenging. Executive function declines and some cognitive deficit may be demonstrated. Involuntary movements are obvious but individuals may still be able to feed themselves.

- Late stage

Late-stage disease occurs when employment becomes impossible, the individual is no longer able to live independently, self-care ceases and cognitive decline is obvious. Some patients maintain the ability to feed themselves to a late stage, but swallowing difficulties and involuntary movements create increasing difficulties.

Methods

Role of the European Huntington's Disease Network

The European Huntington's Disease Network (EHDN) was formed in 2003, to provide a platform for clinicians and families to work together to find a cure for HD, and to coordinate research. Working groups were created to address various themes, such as Standards of Care. Care of HD varies widely throughout the world, and there are many HD management clinics worldwide. Some follow guidelines for care provision that they themselves have devised, but there is a scarcity of peer-reviewed evidence for any standard of care in HD.

We present a guideline for care of the speech and language therapy aspects of HD. The literature has been reviewed, and an expert consensus view for management of this aspect of care of HD has evolved. We expect this to enable others to provide best care for the HD patient, their family and their carers. We also expect these guidelines to evolve as evidence emerges, and we will welcome this.

- Literature review

A systematic literature search was performed electronically using OVID Medline (1988–2009), OVID Embase (1988–2009) and EBSCO Cinahl (1988–2009) to identify evidence within the themes of communication, language problems, dysarthria and dysphagia in HD. A search strategy was developed in collaboration with the members of the Standards of Care Speech Language Therapist (SLT) Group. The literature search was updated during the development of the guideline. All relevant publications were identified and categorized to evidence statements according to the Scottish Intercollegiate Guidelines Network (SIGN 50). Owing to the lack of scientific evidence recommendations were also formulated based upon clinical experience and expert consensus from within the EHDN Standards of Care SLT group.

Key priorities in assessment, management & treatment of swallowing disorders in HD

Referral

Early referral to Speech and Language Therapy is recommended and recognized as good practice in the management of long-term neurological conditions [11]. This facilitates the accumulation of baseline clinical data and aids clinical decision making on timely assessment and/or interventions. It is crucial in managing dysphagia in HD because of the multiple factors involved, including cognitive and behavioral changes. SLT referrals should be discussed as early as the person is identified at being at risk of developing the disease, as it is recognized that there is a group who will want to seek early support and information. There may be significant anxieties based on family experiences.

Objectives of referral

The Standards of Care Working Group in Speech and Language Therapy agreed that the following objectives are important to the person with HD and their family on being referred to speech and language therapy services:

- Seeking and receiving information
- Assessment of swallowing safety and identification of current risks
- Opportunity to discuss future management and options
- Receiving pertinent advice on relevant areas: appetite changes, dietary modifications and on strategies for optimizing swallow safety
- Reduction of patient and family/carers anxieties on swallowing problems
- Opportunity to begin forming an effective relationship with the appropriate professionals

Assessment

Pre-assessment information

Information should be gathered in accordance with standards recommended by SLT professional bodies as with other patient groups referred for dysphagia assessment [12]:

- Medical history
- Current medications
- Dietary history
- Feeding/swallowing history
- Patient-specific information

Some of this information should be obtained if possible from other members of the multidisciplinary team prior to patient interview. If possible partner/carers should also be interviewed to obtain a detailed history of the general course of the disease and current status. It is agreed that information gathered only from the patient, particularly as the disease progresses, may not provide a sufficiently adequate profile. This information will assist in determining the type, scope and timing of further assessments.

Clinical assessment

The clinical assessment gives information about the presence, nature and severity of dysphagia [13]. This should define the physiological dysfunction and its effect on patient’s activity and participation, identify the requirement for further investigation, examine the effectiveness of particular interventional strategies and facilitate the development of a management plan. In assessing the patient with HD it is recommended that the SLT considers gathering information on the following areas, all of which can have particular relevance in HD (Box 1).

The SLT should use the information gathered to determine the ongoing assessment procedure and to determine the safety of food trials. It is recognized that in early stage patients will usually be taking full oral diet. The SLT should also observe the following factors, which are likely to have particular relevance in assessing and managing dysphagia in HD:

- Mealtime routines
- Positioning
- Bolus size
- Utensils/assistive devices
- Environment
- Supervision

A number of swallowing assessments are available (e.g., Northwestern Dysphagia Screening Check Sheet [14]) and some have been developed with specific attention to Huntington’s Disease, for example, Yorkston and colleagues [3] and Basmagi [Unpublished Data]. These can plot baseline information and serve as a monitoring tool for further assessment as the disease progresses. Standardized testing, for example, Timed Water Swallow Test [15] can also be used as a measure of presence or absence of dysphagia in the early stages of the disease.

Observation of a meal intake in the home environment is considered beneficial to provide information, which may be obscured/ameliorated in a clinic-based examination of the swallow. This may include adverse and positive effects of environment (seating, distractions, and cutlery), rate of intake and possible effects of fatigue.

Box 1. Recommendation for clinical speech and language therapy assessment in Huntington's disease.

- Oro-facial examination, including cranial nerve assessment
- Generalized moto skills, including coordination, posture and tone
- Respiratory status
- Secretion management
- Oral hygiene and dental health
- Effects of emotional status, mood and behavior
- Cognitive status/factors
- Alertness levels
- Communications status
- Nutrition and hydration
- Ability to participate and co-operate

Instrumental assessment

Studies with other neurological populations have shown that the bedside clinical examination lacks specificity and sensitivity in identification of aspiration [16]. In other progressive conditions, such as motor neuron disease/amyotrophic lateral sclerosis, instrumental assessment is recommended to answer specific questions [17]. Videofluoroscopic evaluation of swallowing is an instrumental dynamic assessment of swallowing involving visualization of the structure and functions of the oral, pharyngeal and upper esophageal stages. It allows assessment of the presence and cause of aspiration and residue. It facilitates the experimentation of techniques, textures, postures and manoeuvres in order to improve the safety and effectiveness of the swallow [101]. Videofluoroscopy examination to provide both, assessment and interventional information is reported by Hamaka and colleagues [7]. Their study underlined some of the improvements in swallow achieved through altering bolus sizes and consistencies. Kagel and Leopold [4,6] demonstrated improvements in swallow using modified head and body postures using videofluoroscopy assessment. There are some cases where videofluoroscopy may not be an appropriate tool. For example, if in the SLT's judgment, the instrumental examination would not change the clinical management of the patient or the patient is unable to cooperate or participate [101].

Fiberoptic endoscopic evaluation of swallowing is an alternative instrumental assessment method. However, this technique is not widely used with patients with HD as it does not allow visualization of the oral stage and choreic movements can make the examination difficult.

Instrumental tools

Pulse oximetry and cervical auscultation are instrumental tools that have been used to assist in evaluating swallow function in other neurological dysphagic populations but the evidence on the reliability of these is inconsistent [18,19].

Management

Early stage

In the early stages of the disease the patient with HD may have no symptoms of dysphagia. Patients and/or family may identify the following:

- Pharyngeal lodging
- Occasional coughing on eating/drinking
- Slight difficulties in mastication and reduction in lingual control

The SLT should note whether there are any specific requirements noted in the referral and that, with genetic conditions such as HD, affected individuals and their families are likely to have past experiences, which may mean they seek support early. Any program of intervention that is introduced should be systematically and regularly reviewed and should always take account of the individual's own circumstances and wishes. In the early stages both general and specific advice can be appropriate. Therapeutic interventions in the form of oral/respiratory/phonatory exercises may also be introduced if appropriate at this time.

Management approaches in early-stage HD

- Raise awareness of swallowing (both normal swallow and potential areas of difficulty) and advise on self-monitoring techniques;
- Advise on mood changes and that the presence of anxiety or depression can affect the person's appetite. Stress may also trigger choreic change and affect the swallow process;
- Advise on reducing anxieties on fear of choking/drooling as well as on the embarrassment of making a mess whilst eating;
- Advise on reducing distractions and creating a relaxed environment during mealtimes;
- Advise on conserving energy before and during mealtimes;
- Advise on potentially difficult foods and textures [4,20,21];
- Advise on posture and positional changes, specifically identified for HD in the literature [4] and also for dysphagia in other conditions [21];
- The SLT should identify and agree which interventions receive priority at any stage in time.

Mid stage

Mid-stage HD can coincide with the first signs of swallowing difficulty and/or much more prevalent signs, having a greater impact on the individual's nutritional and medical status (Box 2) [4,6,7,23].

Management approaches in mid-stage HD

- Assessment and re-assessment should be a regular feature during this stage in the disease and will include bedside clinical evaluation, imaging and biofeedback assessments as appropriate;
- Safe swallowing recommendations and information to HD individuals and their family/ carers should be presented in both verbal and written format. Limitations should be explicit;
- Consideration should be given to training, in particular involving family and carers. Caregivers need to be able to facilitate optimally safe and pleasurable eating and drinking. In other dysphagic populations (including learning disability), patient and carer adherence to recommendations has been shown to be influenced by their understanding of the condition, and this can be improved by SLT training [24,27];
- Recommendations for a safe swallowing/eating and drinking plan may also be presented in picture/accessible information format to facilitate the eating/swallowing routine;
- Information should be provided on how to support the individual with HD safely, on how to implement the eating and drinking plan and also to identify when further input from the SLT is required;
- Therapeutic intervention to maintain and/or improve oro-motor function within agreed optimal timeframes may be implemented. These may include breathing exercises, oral and chewing exercises and individualizing swallow sequence, for example, cough postswallow [6];
- Consideration should be given to the individual’s level of cognition, alertness, receptive abilities and behaviors so that the interventional plan is aimed at optimum circumstances;
- The SLT should make recommendations to modify food and liquid consistencies, textures and temperatures as well as the viscosity in order to achieve improved swallow safety [4]. The modifications should link directly to the ongoing systematic assessment in order to increase the safety and efficiency of the swallow;
- The placement of the bolus, its size and the pacing of the feeding are also important in managing the safety and efficiency of the swallow. It is recognized that some individuals with HD will overfill the mouth increasing the risk of choking and forward spillage;
- The SLT should advise on posture and positioning to achieve optimum body and trunk position to support safe swallow function. This should include chin tuck positions to protect the airway and may include strategies to minimize hyperextension of the neck and trunk to minimize swallow effort and assist in pharyngeal clearance [4,22]. Joint working with colleagues in physiotherapy is recommended to achieve optimal positioning;

- Adaptive feeding equipment should be considered [4] and it is recommended that the SLT work with colleagues in occupational therapy to provide appropriate equipment;
- The SLT should advise on optimizing the individual's environment as this can have a significant influence on the mealtime and the aim should be to make it a safe and pleasurable experience. Social interaction, distraction and noise levels should be considered [26];
- The SLT will contribute to the discussions and multidisciplinary decision making regarding nonoral feeding/support feeding. This discussion and decision regarding the provision of nutrition and hydration by a non-oral route should include the individual with HD and their significant carers [27]. Discussion should be introduced while the patient's cognitive abilities allow full participation and decisions should be clearly documented.
- It is agreed that the cough reflex is often well preserved in HD and the protection afforded to the airway means oral feeding is often possible until the advanced stages of the disease.

Box 2. Swallowing features in mid-stage Huntington's disease.

- Hyperextension of neck and trunk
- Reduced mastication and lingual control
- Darting lingual chorea
- Drooling – spillage from mouth
- Tachyphagia/premature transfer particularly of liquids
- Intraoral retention following initial transfer
- Delayed and repetitive swallow
- Prolonged laryngeal elevation
- Coughing on liquids
- Choking on foods and liquids
- Reduced/disrupted breath control during the swallow
- Phonation during swallowing
- Belching
- Aerophagia
- Vomiting

End stage

It is agreed that there is significant overlap of presenting symptoms of oral feeding difficulties in HD in the mid and late stages. As the disease progresses, the challenge of maintaining adequate nutrition and hydration becomes greater due to the complexities involved (Box 3). Several of these areas have been shown to influence risk of aspiration pneumonia, for example, dependence on others for feeding [28] and oral hygiene [29].

Management approaches in late-stage HD

- Dietary modifications, the use of compensatory swallowing techniques, the manipulation of head or body postures and manoeuvres to support safe and efficient swallowing should continue. All of these factors must be reviewed regularly;
- Therapy techniques such as exercises or strategies to facilitate or stimulate the swallow should continue;
- Good oral hygiene should be maintained. This should ensure pathogenic organisms are not allowed to proliferate in the mouth and should reduce the risk of aspiration pneumonia [29];
- Training packages ranging from raising awareness of good practice to specific training in all aspects of dysphagia should be considered vital at this stage. All caregivers should possess the knowledge and skills to feed/support the feeding of patients with dysphagia safely;
- All caregivers should be in receipt of specific instructions on:
    - Food placement
    - Dietary modifications
    - Positioning and postures
    - Management of behavioral and environmental factors
    - Management of choking
- Regular monitoring of the individual with HD's weight, hydration, nutrition and occurrences of aspiration pneumonia is key at this stage in the disease;
- Increased risk relating to all or any of these areas should initiate multidisciplinary discussion regarding alternatives in future management;
- If this discussion has taken place with the individual with HD at an earlier stage when the ability to understand the options and the implications was clear, then the management decision should comply with the patient's wishes and rights.

Box 3. Complex aspects of late-stage Huntington's disease.

- Cognitive difficulties
- Communication difficulties
- Behavioral changes
- Mood disorder
- Appetite changes
- Weight loss/gain
- Increased aspiration risks
- Medication
- Oral health/dentition
- Feeding dependency
- Non-oral feeding/support feeding

Conclusion

This guidance document on the management of oral feeding in HD has been developed using a systematic review of all the available evidence as well as expert consensus on best clinical practice.

The guidance considers the progression of the disease over time and the importance of the involvement of Speech and Language Therapy Services to support food and fluid intake and manage changes in the safety and efficiency of the swallowing function. It underlines the need for flexibility and responsiveness in addressing the changing clinical picture of the affected individual and the value of involving the family and care givers in the exchange of information and in particular interventions to minimize the devastating effects of the disease. These clinical guidelines are an important tool in attempting to provide equity and quality of service provision and are also work in progress which will require review and revision as new research becomes available.

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Chapter 7

Is adopting the chin tuck posture when swallowing effective in patients with Huntington's disease?

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Abstract

Objective

Patients with Huntington's disease suffer from dysphagia leading to aspiration. In order to prevent aspiration, it is often recommended that the patient adopts the chin tuck posture when swallowing. Whether this method is effective in preventing aspiration in these patients has, however, not been described. The goal of our study was to investigate whether the chin tuck intervention is a technique that should be recommended in HD patients in order to reduce spilling, prevent aspiration, and to reduce residue.

Methods

Forty-five patients underwent a videofluoroscopic swallowing study, which involved presenting them with 10cc of thin liquid to be swallowed with the head in normal, upright position, and 10cc thin liquid to be swallowed with the head in chin tuck position. The following features were analyzed: spilling before and during the swallow; penetration and aspiration; residue in the valleculae, and piriform sinus. The Wilcoxon signed ranks test and the Mc Nemar test were performed to investigate the differences between the two postures.

Results

All 45 HD patients underwent videofluoroscopy. Of these, 31 patients (68.9%) performed a technically good chin tuck posture, and 14 patients (31.1%) were unable to adopt the chin tuck posture when swallowing. No significant differences between the postures were found with regard to spilling, aspiration, and residue.

Conclusion

In our study population of HD patients, the chin tuck intervention was not found to be effective. We, therefore, conclude that the chin tuck procedure should no longer be advised in patients with HD.

Introduction

Huntington's disease (HD) is a neurodegenerative disease; the most prominent characteristics are disturbed movements, disturbed behaviour, and cognitive decline [1]. Almost all patients with HD suffer from dysphagia [2]. Aspiration is one of the serious consequences of dysphagia, as well as weight loss, malnutrition and dehydration. Up to 86% of HD patients die of pneumonia due to aspiration [3]. An intervention often recommended by speech-and-language therapists to prevent aspiration is to adopt the chin tuck posture during swallowing. This involves tucking the head into the neck while swallowing (Figure 1) [4], with the aim of reducing the chance of aspiration due to the change in head position [5]. When the chin is tucked to the front of the neck, the tongue-base is close to the pharyngeal back wall, the epiglottis inverts into a more protective position over the entrance to the trachea, and the size of the valleculae increases [4,6]. It has not, however, been shown that this method is effective in preventing aspiration in these patients. The aim of our study was, therefore, to investigate whether the chin tuck posture is a useful recommendation for HD patients in order to reduce spilling, prevent penetration and aspiration, and also to reduce residue after swallowing.

Figure 1 Chin tuck posture and normal posture
Image left: chin tuck position. Image right: normal position. The left image demonstrates that when the chin is tucked the tongue-base is close to the pharyngeal back wall, the epiglottis inverts into a more protective position over the entrance to the trachea, and the size of the valleculae increases.

Materials and Methods

Subjects

This study was part of a larger dysphagia study in HD [7] which included 45 HD patients (Table 1). Patients with a CAG repeat size ≥36 and without other diseases that could affect swallowing were included. Included patients were enrolled from all three clinical stages described by Roos [8]. In our study, the stages were defined as: stage I, patients living at home (n=13); stage II, patients living at home and having daycare (n=18); and stage III, patients living in a nursing home (n=14). Patients in stage I were consecutively selected from April 2012 until June 2012 from the outpatient Department of Neurology of the Leiden University Medical Centre (LUMC), and patients in stages II and III were consecutively selected during the same period from a Huntington Care Centre. The study was approved by the medical ethics committee of the LUMC. All patients gave their informed consent.

Table 1 Clinical and demographic information of the 45 patients with Huntington's disease

Patients	
N (males/female)	45 (23/22)
Age, yrs (SD)	53.7 (11.8)
Age at onset, yrs (SD)	43.8 (10.1)
Disease duration, yrs (SD)	9.8 (5.2)
CAG repeat length (mean ,SD)	44.2 (3.2)
N Stages of HD	
Clinical stage I	13
Clinical stage II	18
Clinical stage III	14

Values are means (SD), except for the distribution across stages.
Abbreviations: SD, standard deviation; HD, Huntington's Disease; CAG, repeat length triplets number (≥36, pathological range)

Data collection

All patients underwent a videofluoroscopic swallowing study (VFSS), which involved presenting 10cc thin liquid to be swallowed with the head in normal, upright position, and 10cc thin liquid to be swallowed with the head in chin tuck position. Water-soluble liquids were used to reduce risks when aspiration occurred. VFSS were recorded on a Toshiba Ultimax-I at 15 frames per second. Each video was analyzed in slow motion to define any swallowing disorder. The following features were analyzed: the ability to perform the chin tuck posture, spilling before and during the swallow; penetration and aspiration; residue in the valleculae, and piriform sinus. For penetration and aspiration, the Penetration-Aspiration-Scale (PAS) was used. A score between 2-5 was defined as penetration, and 6-8 as aspiration [9]. Two raters independently analyzed the images of the VFSS: a radiologist experienced in dysphagia, and a speech and language scientist experienced in dysphagia and HD. Conclusions were reached by consensus.

Statistics

To investigate any significant difference between the normal posture and the chin tuck posture when swallowing, the Wilcoxon signed ranks test was applied for ordinal variables, and the Mc Nemar test was performed for dichotomous variables. For the statistical analyses, SPSS 22 was used.

Results

Of the 45 patients, 31 (68.9%) were able to perform the chin tuck posture, while 14 patients (31.1%) were not. On dividing the patients into the three different disease stages, it was found that in stage I, 2 (15.4%), in stage II, 4 (22.2%) and in stage III, 8 (57.1%) patients were unable to perform a good chin tuck posture. The outcomes of the measured features: spilling, penetration and aspiration, residue, and the differences between normal posture and chin tuck posture are shown in Table 2. No significant differences were found for any of the features measured.

Table 2 Videofluoroscopic findings of swallowing 10cc thin liquid with the normal posture and the chin tuck posture

	All stages				Stage I		Stage II		Stage III	
	Normal posture N=31 (%)	Chin tuck N=31 (%)	Z	p	Normal posture N=11 (%)	Chin tuck N=11 (%)	Normal posture N=14 (%)	Chin tuck N=14 (%)	Normal posture N=6 (%)	Chin tuck N=6 (%)
Spilling*										
1. before	6 (19.4)	2 (6.5)		.219	0	0	5 (35.7)	1 (7.1)	1 (16.7)	1 (16.7)
2. during	14 (45.2)	7 (22.6)		.118	4 (36.4)	1 (9.1)	7 (50.0)	4 (28.6)	3 (50.0)	2 (33.3)
PAS**	8 (25.8)	6 (19.4)	-0.690	.490	1 (9.1)	1 (9.1)	4 (28.6)	5 (35.7)	3 (50.0)	0 (0)
Residue**										
- valleculae										
no	15 (48.4)	12 (38.7)	0.000	1.000	8 (72.7)	6 (54.5)	4 (28.6)	4 (28.6)	3 (50.0)	2 (33.3)
yes	16 (51.6)	19 (61.3)			3 (27.3)	5 (45.5)	10 (71.4)	10 (71.4)	3 (50.0)	4 (66.7)
- piriform sinus										
no										
yes	17 (54.8)	18 (58.1)	-0.714	.475	10 (90.9)	8 (72.7)	4 (28.6)	7 (50.0	3 (50.0)	3 (50.0)
	14 (45.2)	13 (41.9)			1 (9.1)	3 (27.3)	10 (71.4)	7 (50.0	3 (50.0)	3 (50.0)

*Mc Nemar test (2-tailed); **Wilcoxon signed ranks test (2-tailed); Abbreviations: vs, versus; PAS, Penetration Aspiration Scale

Discussion

A videofluoroscopic study was carried out to evaluate the widely used recommended strategy in HD patients, i.e. to swallow with the chin tuck intervention. Our results show that no significant differences were found for aspiration, residue or spilling when swallowing with the head in chin tuck position compared to swallowing with the head in normal upright position. Analysis of our patient group, therefore, shows that the chin tuck intervention is not helpful. In our patient group, 14 (31%) could not perform a good chin tuck posture, especially in the last stage of the disease (n=8) when patients suffer from severe dysphagia. In our study group, the most prominent problem was that patients in the last stage did not have the ability to maintain the head position due to choreatic movements (n=4). Four other patients suffered from severe cognitive decline and therefore could not properly understand the instructions. No significant difference was found for aspiration when changing the position of the head. It is, therefore, very unlikely that the chin tuck intervention is able to reduce aspiration in this particular group of patients. Whether the chin tuck intervention reduced the risk of aspiration was also investigated in a group of Parkinson’s disease (PD) patients, using VFSS [10], and comparing them to a reference group. The authors hypothesized that the chin tuck intervention should eliminate aspiration in PD, but when compared to the reference group, they found no elimination of aspiration. In fact, the PD patients even had an increased risk of aspiration when adopting this posture. Regarding the presence of residue in the valleculae and piriform sinus, our study found no significant change with the chin tuck posture. Since residue in the sinuses of the pharynx is a potential risk factor for aspiration after the swallow [6], it is of interest to know that the chin tuck posture does not reduce the risk of residue. In summary, the chin tuck intervention while swallowing is a not recommended intervention in HD patients. It does not significantly decrease the risk of spilling, penetration, aspiration, or residue.

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Chapter 8

Summary and concluding remarks

Huntington's disease (HD) is a progressive neurodegenerative disease with an autosomal, dominant mode of inheritance. The chromosomal abnormality is a CAG repeat expansion in the *HTT* gene on chromosome 4. The mutant protein, huntingtin, causes neurodegeneration in the brain, particularly in the caudate nucleus and putamen. Characteristic of HD are movement abnormalities (chorea, hypokinesia), cognitive decline and psychiatric disturbances [1]. Patients with HD also suffer from dysphagia which can have serious consequences, such as weight loss, dehydration, and pneumonia leading to death. Many patients with HD die of aspiration pneumonia.

The act of swallowing is a complex motor program involving a sequence of activation and inhibition of muscles in the mouth, pharynx, larynx and esophagus. Swallowing is divided into four phases: the *preparatory oral phase*, the *oral phase*, the *pharyngeal phase* and the *esophageal phase* [2]. It is known that patients with HD have disturbances in all phases of ingestion.

The first aim of this thesis was to gain insight into the specific dysphagia problems in HD, and also to investigate if indeed most patients die of pneumonia due to aspiration. The introduction *(chapter 1)* of this thesis poses the problem that knowledge about dysphagia in HD is lacking and describes the steps required to improve understanding of the specific swallowing problems in patients with HD.

The findings in the literature were not conclusive and did not include information about the relation between dysphagia and disease stage *(chapter 2)*. Furthermore, the most frequent cause of death, pneumonia, was not further specified according to type of pneumonia. We therefore studied 224 records of the Leiden University Medical Center (LUMC) Brain Bank of deceased HD patients and investigated the type of pneumonia *(chapter 3)*. Our data confirmed that 87% of the patients who died of pneumonia, the pneumonia was due to aspiration.

In order to start monitoring dysphagia in HD patients systematically during different stages of the disease, a new measurement instrument was needed. We developed the Huntington's Disease Dysphagia Scale (HDDS) *(chapter 4)* especially for HD patients. The HDDS reflects all phases of ingestion and can be used in all stages of HD. The scale is found to be a reliable and valid measure of the severity of dysphagia in HD. The availability of this new scale now allows prospective monitoring of dysphagia in patients with HD.

Another aim was to investigate specific dysphagia features in HD. We started a study to identify these and to establish when the dysphagia manifests in the course of the disease *(chapter 5).* Based on videofluoroscopic swallowing studies (VFSS), we found that 78% of our patients suffered from dysphagia, and that dysphagia occurred with liquids as well as solid food. Swallowing difficulties start already in the first stage of HD, and become more severe as the disease progresses. Many types of swallowing disturbance were found at every phase of ingestion, the most prominent being: spilling before and during the swallow, penetration into the trachea and aspiration, and residue in the valleculae and piriform sinus. Based on our results, we formulated the following recommendations: decrease volume and increase viscosity of liquid intake, and trigger a dry swallow after each swallow for all food consistencies. Whether these recommendations significantly decrease aspiration can be a subject of future research.

The final aim of this thesis was to initiate intervention studies on dysphagia in HD. Because no guideline or effective intervention for dysphagia in patients with HD existed, we began by developing a guideline based on best practice *(chapter 6)*. A group of European speech and language therapists (SLT), experienced in HD, was formed within the working group Standards of Care of the European Huntington's Disease Network (EHDN). The guideline describes dysphagia problems per stage of HD, and makes recommendations for intervention and clinical advice. Like every guideline, this one is also developing continuously and will require review and revision on a regular basis as new information becomes available. One of the recommendations in the European guideline is the chin tuck position while swallowing, a commonly used intervention with the idea of preventing aspiration and reducing spilling and residue in patients. It was our impression that the chin tuck method was often recommended in HD by speech and language therapists. But as there is no evidence for improvement with this intervention for HD patients, we carried out a videofluoroscopic swallowing test with and without the chin tuck test procedure (*chapter 7*). No significant differences were found for spilling, aspiration or residue in swallowing with the head in normal upright position or the chin tuck posture. We therefore recommend that the chin tuck intervention should not be used in HD patients.

Concluding remarks

In the literature on HD little attention has been paid to swallowing disturbances and the sometimes severe consequences of choking, although it is known that the main cause of death in HD patients is aspiration pneumonia due to choking. More in depth knowledge on dysphagia in HD leads to greater insights into this specific problems, so that treatment advice can be developed and systematically studied with standardized methods of assessment.

Assessment

Although most studies use the Unified Huntington's Disease Rating Scale (UHDRS) to quantify HD severity, we considered the UHDRS less appropriate for our study. The UHDRS contains domains, such as cognitive assessment, behavioral assessment, and functional assessment. Alas, no item in the UHDRS, not even in the motor assessment, deals with swallowing. We developed the dysphagia scale, the HDDS, which provides a tool for standardized assessment and therefore would allow for comparative or multicentric studies. It would be of interest to investigate the relation between the progression of the disease and the outcomes of the HDDS in a larger group of patients.

Future perspectives

Looking forward, we will translate the HDDS into English and German after which it will be validated, so that larger groups of patients around the world can be monitored for dysphagia, in order to gain greater insight into the severity of the swallowing problems in HD. Of further interest and importance is to initiate more intervention studies on dysphagia in HD. A few procedures involving physical manoeuvers to improve swallowing do exist and have been validated in different populations [3], such as neurological patients with different etiologies, for example: patients with cricopharyngeal dysfunction [4], patients suffering from ischemic infarctions, hemorrhages, tumors and trauma [5]. These procedures focus on a different, specific part of the physiology of swallowing. One of these is the Mendelsohn manoeuver [2] which triggers a voluntary prolongation of laryngeal excursion at the midpoint of the swallow, with the aim of increasing the extent and duration of laryngeal elevation in order to extend the duration of cricopharyngeal opening. This manoeuver also achieves a better coordination of the timing of all swallowing phases. Based on the findings described in chapter 5, we now know that the timing of the swallowing in HD is disturbed, especially in the oral and pharyngeal phases of the swallow. We therefore recommend starting an intervention study on the effect of this Mendelsohn manoeuver in HD patients.

Another topic for future research is the influence of medication on the swallowing function in HD. It is known, for example, that antipsychotic medication influences the swallowing function, possibly resulting in dysphagia and asphyxia [6]. An association with worse swallowing function was also found in hospitalized, elderly patients who were receiving antipsychotics [7]. The effect on dysphagia of antipsychotic medication has been incidentally reported, e.g. in a case reports of a patient suffering from schizophrenia [8], and a patient with Parkinson's disease [9]. The influence of antipsychotic medication on swallowing has not been systematically examined in HD patients, while many patients receive these drugs.

It is therefore of interest to investigate the swallowing function in patients with HD who also receive antipsychotic medication.
The last aspect that needs to be mentioned is the psychological aspect of dysphagia in HD. It is our clinical experience that swallowing disorders are a frightening issue for HD patients, whose partners and family members have seen them choke on food or beverages. Patients do complain of swallowing difficulties and some are afraid of choking and possible suffocation. It is therefore important, especially now we do know the specific dysphagia features in HD, not only to focus on the clinical aspects of dysphagia, but also to investigate the influence of anxiety on dysphagia, or vice-versa, if patients develop swallowing disorders due to their anxiety about swallowing.

This thesis has been written with the aim of gaining more insight into specific swallowing problems in patients with HD. When we started our research, it was not known from which dysphagia features HD patients suffer, nor the severity of the dysphagia in HD. Our research revealed that up to 78% of the patients suffer from dysphagia, and that the swallowing disturbances already start in the first stage of the disease. It also showed that patients with HD have swallowing disturbances in all phases of ingestion, and that spilling, aspiration and residue are frequent findings. These aspects do explain the most frequently encountered primary cause of death in HD, namely aspiration pneumonia. It is our hope that this study will stimulate professionals to continue to look systematically at this clinically important problem. Not only the clinical aspects, but also the therapeutic, and psychological aspects are of importance. All are essential for the quality of life of patients with HD. The most important aims of this thesis were: create valid tools and develop evidence-based treatment for dysphagia in HD, in order to finally prevent aspiration pneumonia and, most importantly, to improve quality of life of patients with HD.

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Nederlandse samenvatting

De ziekte van Huntington (ZvH) is een progressieve neurodegeneratieve aandoening met een autosomale, dominante wijze van overerving. De chromosomale afwijking is een expansie van het CAG repeat op het *HTT* gen op chromosoom 4. Het mutante eiwit, huntingtin, veroorzaakt neurodegeneratie in de hersenen, vooral in de nucleus caudatus en het putamen. Kenmerken van de ZvH zijn afwijkingen als ongewilde bewegingen (chorea, hypokinesie), cognitieve achteruitgang en psychiatrische stoornissen. Patiënten met de ZvH hebben ook last van dysfagie hetgeen ernstige gevolgen kan hebben, zoals gewichtsverlies, uitdroging en een longontsteking die tot de dood kan leiden. Veel patiënten met de ZvH overlijden aan een aspiratiepneumonie.

Slikken is een complex motorische functie met een opeenvolging van het activeren en remmen van spieren in de mond, de keelholte, het strottenhoofd en de slokdarm. Slikken bestaat uit vier fasen: de voorbereidende orale fase, de orale fase, de faryngeale fase en de slokdarm fase. Het is bekend dat patiënten met de ZvH stoornissen hebben in alle fasen van het slikken.

De eerste doelstelling van dit proefschrift was om inzicht te krijgen in de specifieke slikproblemen bij de ZvH en ook om te onderzoeken of de meeste patiënten inderdaad sterven aan een longontsteking ten gevolge van aspiratie. De inleiding (hoofdstuk 1) van dit proefschrift beschrijft dat kennis over dysfagie bij de ZvH grotendeels ontbreekt en beschrijft de stappen die nodig zijn om het inzicht in de specifieke slikproblemen bij patiënten met de ZvH te verbeteren.

De bevindingen over slikproblemen bij de ZvH in de literatuur geven onvolledig inzicht en bevatten geen informatie over de relatie tussen dysfagie en het ziektestadium (hoofdstuk 2). Bovendien is de meest voorkomende doodsoorzaak, longontsteking, niet verder gespecificeerd naar het soort longontsteking. We onderzochten daarom 224 statussen van de Leids Universitair Medisch Centrum (LUMC) Brain Bank van overleden patiënten met de ZvH en onderzochten de aard van de longontsteking (hoofdstuk 3). Onze gegevens bevestigen dat 87% van de patiënten die stierven aan longontsteking, de longontsteking het gevolg van aspiratie was.

Om dysfagie in de verschillende fasen van de ziekte stelselmatig te monitoren was een nieuw meetinstrument nodig. We ontwikkelden de Huntington's Disease Dysphagia Scale (HDDS) (hoofdstuk 4) speciaal voor patiënten met de ZvH. De HDDS reflecteert alle fasen van het slikken en kan in alle stadia van de ZvH gebruikt worden. De schaal blijkt betrouwbaar en valide om de ernst van dysfagie bij de ZvH te meten. De beschikbaarheid van deze nieuwe schaal stelt ons in staat om prospectief dysfagie bij patiënten met de ZvH te monitoren en het effect van interventies te beoordelen.

Een ander doel van dit proefschrift was om de specifieke kenmerken van het slikken bij de ZvH onderzoeken. We begonnen met een onderzoek om de slikstoornissen te identificeren en vast te stellen wanneer in de loop van de ziekte de dysfagie zich manifesteert (hoofdstuk 5). Gebaseerd op videofluoroscopische beelden vonden we dat 78% van onze patiënten slikstoornissen had en dat dysfagie voorkwam bij zowel het slikken van vloeistoffen als vaste voeding. Slikproblemen beginnen al in de eerste fase van de ZvH en zijn meer uitgesproken naarmate de ziekte vordert. Veel stoornissen in het slikken werden gevonden en ook in elke fase van het slikken. Het meest prominent was: spilling voor en tijdens het slikken, penetratie in de luchtpijp en aspiratie, en residu in de valleculae en sinus piriformis. Op basis van onze resultaten kwamen we tot de volgende aanbevelingen: het verkleinen van de bolusintake, het verhogen van de viscositeit van vloeistoffen en het inzetten van een droge slik na iedere hap of slok. Of deze aanbevelingen aspiratie sterk doen verminderen zal een onderwerp van toekomstig onderzoek zijn.

Het laatste doel van dit proefschrift was om interventie studies te starten. Omdat er geen richtlijn of bewezen effectieve interventie voor dysfagie bij patiënten met de ZvH bestond, zijn we begonnen met het ontwikkelen van een richtlijn op basis van best practice (hoofdstuk 6). Een groep Europese logopedisten met ervaring met de ZvH werd samengesteld binnen de werkgroep Standards of Care van het Europese Huntington's Disease Network (EHDN). De richtlijn beschrijft dysfagieproblemen per fase van de ziekte en doet aanbevelingen voor interventie en geeft klinisch advies. Zoals bij elke richtlijn, wordt ook deze richtlijn voortdurend verder ontwikkeld en zal er regelmatig toetsing en herziening nodig zijn als nieuwe informatie beschikbaar komt. Een van de aanbevelingen in de Europese richtlijn is de kin-op-de-borst methode tijdens het slikken, een veel gebruikte interventie met het idee dat spilling, aspiratie, en residu bij patiënten hierdoor aanzienlijk verminderd. Onze indruk was dat door logopedisten de kin-op-de-borst methode vaak werd aanbevolen bij patiënten met de ZvH. Maar omdat er geen bewijs voor verbetering van het slikken is door deze interventie specifiek voor patiënten met de ZvH, hebben wij een videofluoroscopisch slikonderzoek gedaan waarbij patiënten vloeistof slikten met en zonder de kin-op-de-borst methode (hoofdstuk 7). Er werden geen significante verschillen gevonden voor spilling, aspiratie en residu bij het slikken met het hoofd in de normale verticale positie of de kin-op-de-borst houding. Wij raden daarom aan de kin-op-de-borst methode niet meer te gebruiken bij patiënten met de ZvH.

Slotopmerkingen

In de literatuur over de ZvH is weinig aandacht besteed aan slikstoornissen en de soms ernstige gevolgen van verslikking, hoewel het bekend is dat de belangrijkste oorzaak van overlijden bij patiënten met de ZvH een aspiratiepneumonie is, te wijten aan verslikking. Kennis over dysfagie bij de ZvH leidt tot meer inzicht in deze specifieke problematiek, zodat de behandelingsadviezen kunnen worden ontwikkeld en systematisch bestudeerd met gestandaardiseerde methoden voor onderzoek.

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Curriculum Vitae

Anne-Wil Heemskerk-van den Berg was born on February 19, 1979 in Leiden, the Netherlands. From 1998 until 2002 she attended the study Speech and Language Therapy at the Hogeschool Rotterdam. Thereafter she started working as a Speech and Language Therapist in Topaz Overduin, Katwijk. During this period she developed a keen interest in working with Huntington's disease patients, especially in dysphagia. In 2007 she commenced her study "Clinical Language, Speech and Hearing Science" at the University Utrecht and passed her doctoraal in 2010. In that year she started her research to dysphagia in Huntington's disease as PhD student in the Dept of Neurology at the Leiden University Medical Centre. Anne-Wil became an active member of the European Huntington's Disease Network, were she was co-lead facilitator of the working group Speech and Language Therapy on behalf of the Standards of Care. Anne-Wil regularly gives lectures about dysphagia in Huntington's disease in national and international symposia. After passing her PhD, Anne-Wil will continue her research on dysphagia in Huntington's disease.
Anne-Wil is married with Klaas Heemskerk and they have three children: Anton (2006), Anna (2008) and Bart (2010).