Dysphagia in Huntington's disease

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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.

References

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