Dysphagia in Huntington's disease

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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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