SWALLOWING FEATURES IN HUNTINGTON'S DISEASE
A comparison between safe versus unsafe swallowing

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BACKGROUND

Huntington's Disease (HD) is a neurodegenerative disorder caused by a CAG repeated expansion in the mutated HD gene on chromosome 4p16.3, which encodes the mutated protein huntingtin. Dysphagia has been reported in around 72-100% of patients with HD already in the early stage [1-2] and aspiration pneumonia is the main cause of death in this population [3]. Despite its high prevalence, at the present time only few studies have investigated dysphagia in HD. Moreover, many of these studies are limited to single case reports and did not include an instrumental assessment of swallowing nor a meticulous assessment of oral preparatory stage [4]. Hence, little is known regarding swallowing features of this population.

AIMS

This study attempt to improve knowledge on dysphagia in HD. In particular, it aims to:

1. Identify pharyngeal swallowing profiles in HD
2. Compare neurological and swallowing features in safe versus unsafe swallowers

METHODS

Patients were consecutively recruited between April 2016 and July 2017 during their first or follow-up neurological visit. Inclusion criteria were: clinical diagnosis of HD with familiarity and/or genetic confirmation, no enteral feeding, ability to express their informed consent. Patients with comorbidity with other neurological diseases, history of head neck cancer, self-reported or documented dysphagia prior to HD diagnosis were excluded.
Overall, 24 patients (10 males and 14 females) were included. Mean age at the time of the assessment was 60±12.3 (39-79), mean age at disease onset was 51.8 ±18.4 (32-76), mean disease duration was 8.3 ±4.3 years (3-20). All patients underwent a neurological assessment and an instrumental and clinical evaluation of swallowing.

NEUROLOGICAL ASSESSMENT

- *Unified Huntington's Disease Rating Scale* (UHDRS)

INSTRUMENTAL ASSESSMENT

Fiberoptic endoscopic evaluation of swallowing:
- 5-10-20 ml blue dyed water and pudding x 3
- ½ cracker x 3
- *Penetration-aspiration scale* (PAS) and *Yale pharyngeal residue severity rating scale* (YPRSRS)

CLINICAL ASSESSMENT

- *Mann Assessment Swallowing Assessment* (MASA)
- *Test of Masticating and Swallowing Solids* (TOMASS)
- *Functional oral intake scale* (FOIS)
- *Huntington's disease dysphagia scale* (HDDS)
- *Mealtime assessment scale* (MAS)

Patients with a PAS score ≥2 with at least one consistency were considered unsafe swallowers, while patients with a YPRSRS score ≥3 with at least one consistency were considered inefficient swallowers. Mann-Whitney test was used to compare deglutition variables between safe and unsafe swallowers.

RESULTS

Based on FEES analysis, 11 patients were considered safe swallowers and 13 patients unsafe swallowers. Swallowing efficacy was preserved in 7 patients, while a YPRSRS ≥3 was assigned 17 patients with at least one consistency. Prevalence of pharyngeal swallowing profiles is shown in the graph below.

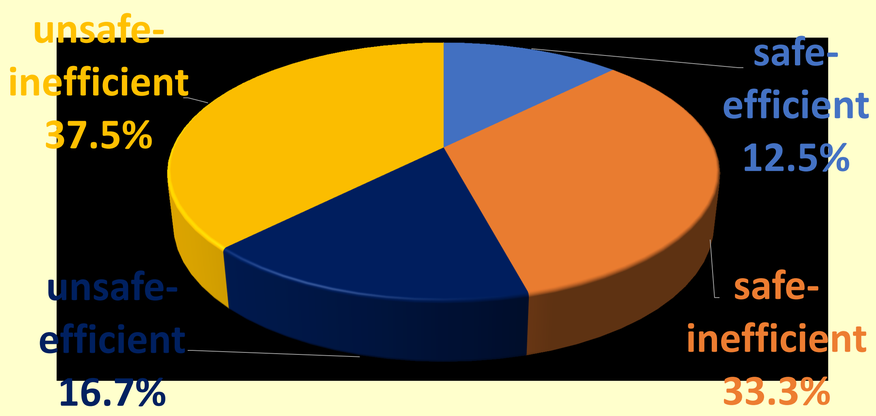
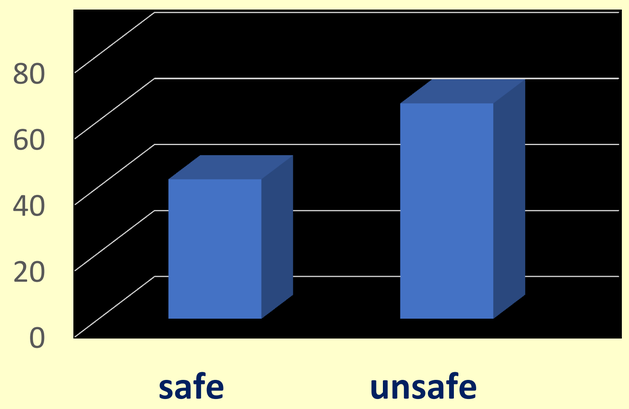


Fig 1: *HD's pharyngeal swallowing profiles*

When comparing safe versus unsafe swallowers, the two groups were comparable for age (p=0.424) and duration of the disease (p=0.089).



A statistical significant difference was found for the scores of the UHDRS I subscale, assessing motor function (p=0.028).
None of the investigated swallowing features was statistically different.

Fig 2: *UHDRS I in safe vs unsafe swallowers*

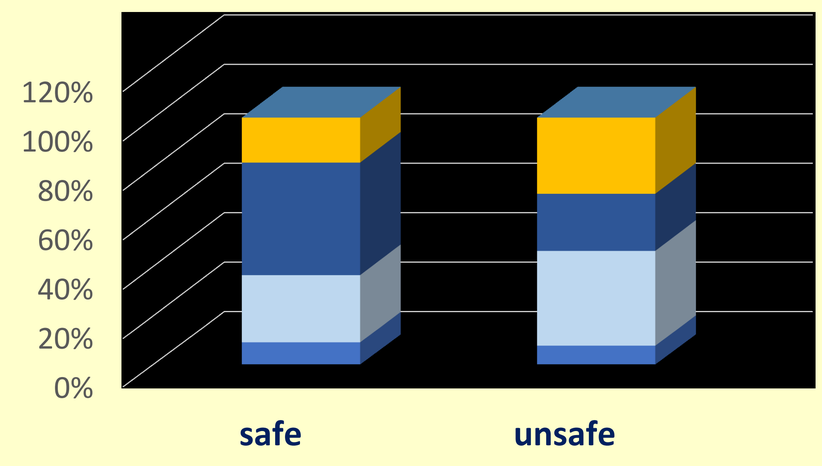


Fig 3: *YPRSRS in valleculae with solids in safe vs unsafe swallowers*

CONCLUSIONS

Present data suggest that efficacy is more impaired than safety in HD patients. Swallowing safety seems to be progressively reduced with disease progression and decline of motor function. HD patients unsafe swallowers show comparable pharyngeal residue, efficacy of oral phase, performance in meal consumption, oral intake and perception of swallowing impairment to HD safe swallowers. Lack of statistically significant differences may be due to the small sample size and the low severity of airway invasion in our unsafe swallowers group.

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