

Clinical Management Guidelines for Friedreich Ataxia

Chapter 3.6. Dysphagia in Friedreich ataxia

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3.6 Dysphagia in Friedreich ataxia

Adam P. Vogel, Lisa Graf, Maggie-Lee Huckabee and Matthis Synofzik

This chapter describes the effects of Friedreich ataxia on swallowing, the functional consequences of swallowing impairment (dysphagia), and strategies for managing dysphagia. In making recommendations for management, the authors were tasked with answering the question:

For individuals with Friedreich ataxia, what management strategies could be implemented for dysphagia?

3.6.1 The effects of Friedreich ataxia on swallowing function

Dysphagia (swallowing impairment) impacts all individuals with Friedreich ataxia (FRDA) at some stage during the disease (1). It worsens over time (2) and is linked to disease severity (3). Dysphagia in FRDA is characterized by slow oral and pharyngeal transit, poor bolus control, and delayed initiation of swallowing (1, 2). Specifically, FRDA can lead to decreased strength and coordination of the musculature used during swallowing. These changes often result in timing-based deficits, leading to increased risk of penetration and aspiration of food or liquids. Upper limb ataxia and difficulty controlling cutlery and cups while eating and drinking also impact mealtime safety.

At mid to later stages of FRDA, coughing or choking while drinking or during mealtimes and difficulty managing secretions become more common. Approximately 25% of people with FRDA are thought to aspirate silently on thin liquids (1), highlighting the need for detailed monitoring of mealtime safety.

3.6.2 Functional consequences of dysphagia

In some cases, dysphagia increases the likelihood of hospitalization for preventable conditions, such as pneumonia, delirium, urinary tract infections, and dehydration (4), and in most cases, it reduces quality of life (5). Changes to swallowing-related quality of life include extended mealtime duration (potentially intensified by upper limb ataxia and the challenges associated with feeding independently); altered self-perception (linked to changing roles within family and social groups); as well as reduced participation in social events and gatherings (1).

3.6.3 Management of dysphagia

Dysphagia management needs to consider impairment, function and swallowing-related quality of life (6). An important first step in management is comprehensive identification of how the dysphagia affects the individual with respect to impairment, activity and participation:

- Impairment can be tested via imaging methods such as videofluoroscopy (VFSS) or fiberoptic endoscopic evaluation of swallowing (FEES). These tools can provide data on the characteristics of deficits as well as the presence of aspiration and penetration during swallowing.
- Activity can be measured by administering purposely designed clinical bedside examinations (CBE) such as the Clinical Assessment of Dysphagia in Neurodegeneration (CADN) (3). CBEs like CADN yield data on the day-to-day sequelae of the impairments, such as trialing liquids and varying food textures and consistencies and anamnesis (case history).

- Participation can be measured via standardized patient-reported outcomes or questionnaires that provide data on the impact of dysphagia on quality of life (7).

Four types of dysphagia management are potentially available for people with FRDA. These include behavioral swallowing intervention/swallowing rehabilitation, such as exercises; neuromuscular electrical stimulation; compensatory strategies, such as postural techniques and education on mealtime behaviors; and diet modification, including thickened fluids. There is currently no empirically documented population specific evidence on the benefit of swallowing therapy for individuals with FRDA. Evidence for the suggested therapies is based on diseases and disorders similar, but wholly distinct from FRDA. The presence of dysphagia in most individuals with FRDA highlights the need for effective and proven therapies for improving swallowing.

Best practice statement

Individuals with Friedreich ataxia who are experiencing difficulties in swallowing should be offered detailed expert counselling on dysphagia management strategies, with a focus on the specificities and mechanisms of dysphagia in degenerative ataxia, as well as their impact on the individual and their friends and families.

Recommendations

Grading for strength of recommendation and level of evidence

For the rating of the **strength** of the recommendation, in addition to evidence from studies in FRDA, evidence from like conditions, clinical experience and expert consensus are taken into account when published evidence is not available.

The **level of evidence** is based on published evidence from studies in FRDA. If there is no published evidence in FRDA, evidence from other like conditions or clinical expertise may have been used to make the recommendation – this is graded as ‘very low’ or in some cases ‘low’ level evidence. See the table below for an explanation of the symbols used to grade recommendations.

Strength of recommendation	Symbol	Level of evidence	Symbol
Strong for intervention	↑↑	High	⊕⊕⊕⊕
Conditional for intervention	↑	Moderate	⊕⊕⊕○
Neither intervention nor comparison	—	Low	⊕⊕○○
Conditional against intervention	↓	Very low	⊕○○○
Strong against intervention	↓↓		

Behavioral swallowing intervention

<i>Should behavioral swallowing intervention/swallow rehabilitation versus no intervention be used for all people with Friedreich ataxia?</i>	Strength	Level of evidence
For people with Friedreich ataxia, the guidelines panel suggests that behavioral therapies are <i>not</i> used as a treatment for dysphagia.	↓	⊕○○○

Justification: There are no trials or published evidence supporting the use of behavioral therapies to improve swallow function in Friedreich ataxia.

Subgroup considerations: This recommendation is for individuals with Friedreich ataxia with swallowing problems.

Compensatory strategies

<i>Should compensatory strategies (postural techniques, mealtime behaviors) versus no compensatory strategies be used for all people with Friedreich ataxia?</i>	Strength	Level of evidence
For people with Friedreich ataxia, the guidelines panel suggests employing some compensatory strategies for improving swallowing safety in people with dysphagia.	↑	⊕○○○
Justification: There is some limited evidence supporting the use of compensatory strategies for improving dysphagia outcomes in neurological conditions other than Friedreich ataxia. This management approach is low impact and not likely to lead to any adverse effects.		
Subgroup considerations: This recommendation is for all individuals with Friedreich ataxia, to improve swallowing safety.		

Modified diet/thickened fluids

<i>Should modified diet and thickened fluids versus no diet/fluid modification be used for all people with Friedreich ataxia?</i>	Strength	Level of evidence
For people with Friedreich ataxia, we cannot recommend either modified diets/thickened fluids or no diet modification to improve swallow safety.	—	⊕○○○
Justification: There are no data supporting the use of modified diet in Friedreich Ataxia.		
Subgroup considerations: This recommendation is considered appropriate for people with Friedreich ataxia with moderate to severe dysphagia.		

Neuromuscular electrical stimulation

<i>Should neuromuscular electrical stimulation versus no swallowing intervention be used for all people with Friedreich ataxia?</i>	Strength	Level of evidence
For people with Friedreich ataxia, the guidelines panel suggests neuromuscular electrical stimulation is <i>not</i> used as a treatment for dysphagia.	↓	⊕○○○
Justification: There are no trials or published evidence supporting the use of neuromuscular electrical stimulation to improve swallow function in Friedreich ataxia.		
Subgroup considerations: This recommendation is for individuals with Friedreich ataxia with dysphagia.		

Lay summary

Lay summary of clinical recommendations for dysphagia in Friedreich ataxia

Swallowing difficulties (dysphagia) happen in almost all people with Friedreich ataxia and can worsen over time. Aspiration (food, liquid and/or saliva entering the lungs) is a problem with dysphagia, but is difficult to predict and warrants regular monitoring of swallowing function. Aspiration can lead to frequent coughing when eating or drinking. However, aspiration can also happen without coughing, which increases the risk of chest infection. Dysphagia can also lead to longer mealtimes and other changes to the quality of life of a person with Friedreich ataxia.

Why these recommendations?

Possible management strategies for dysphagia include:

- compensatory approaches, like controlled flow drinking containers or changing the way a person sits during mealtimes
- diet modification, such as avoiding foods known to cause coughing or using thickened fluids
- behavioral interventions designed to help swallowing, and
- neuromuscular electrical stimulation.

There are no randomized controlled trials or other high-quality research studies supporting any management strategies for dysphagia in Friedreich ataxia. The strategies used currently are based on studies in people with other neurological conditions as well as clinical experience.

There is some limited evidence to support the use of compensatory approaches to help swallowing problems in people with Friedreich ataxia. Although benefits of modified diets have not yet been shown, they are unlikely to be harmful. At this time, behavioral techniques or neuromuscular electrical stimulation are not recommended as treatments for dysphagia.

What does this mean for you as a person living with Friedreich ataxia or caring for someone living with Friedreich ataxia?

It is important for all people with Friedreich ataxia to discuss their swallowing with a healthcare professional. There are some strategies to help compensate for swallowing for people with Friedreich ataxia that may help to minimize dysfunction and optimize swallowing safely.

Who are these recommendations specifically for?

These recommendations are for all individuals with Friedreich ataxia with minor to severe swallowing difficulties.

Author details

Lisa Graf, MSc

University Hospital of Tübingen, Tübingen, Germany

Maggie-Lee Huckabee, PhD

Distinguished Professor, University of Canterbury, Christchurch, Canterbury, NZ

Email: maggie-lee.huckabee@canterbury.ac.nz

Matthis Synofzik, MD

Professor, Hertie-Institute for Clinical Brain Research & Center for Neurology, Tübingen, Germany

Email: matthis.synofzik@uni-tuebingen.de

Adam P. Vogel, PhD

Professorial Fellow, The University of Melbourne, Melbourne, Victoria, Australia

Email: vogela@unimelb.edu.au

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