

Student Names: Jaclyn Forbes & Madison O'Brien
Faculty Sponsor Name: Yvonne Rogalski

Friedreich's Ataxia: A Comprehensive Overview

The intent of this presentation is to provide a comprehensive review of Friedreich's ataxia, including its etiology, pathophysiology, diagnostic signs, and potential treatments for speech-language pathologists to use in their clinical practice.

Friedreich's ataxia (FRDA) is a chronic, progressive disorder that affects multiple systems within the body, including the central nervous system (CNS) and the peripheral nervous system (PNS). The disorder was founded and named after Nikolaus Friedreich, who was a German physician in the late 1800s. Friedreich's ataxia is an inherited recessive disorder caused by a mutation of the homozygous triplet GAA expansion in the FXN gene on chromosome 9q13, which is responsible for producing the frataxin protein; a protein that is involved in iron metabolism and neuronal mitochondrial execution. The disorder affects approximately 1 in every 50,000 people that reside in the United States and is considered the most common form of recessive ataxia in the U.S. and Europe. Primary characteristics of the disorder include incoordination of movement, dysarthria, and gait and limb apraxia.

Friedreich's ataxia is characterized by a decrease in size of the dorsal root ganglia in addition to degeneration in the dorsal spinocerebellar tract and the lateral corticospinal tract. Severe degeneration is also found in the dentate nucleus located within the cerebellum, ultimately causing atrophy of the superior cerebellar peduncle. The neuropathophysiology results in ataxic dysarthria, altered nasality, voice quality, respiration, articulation, and phonation. Non-speech symptoms include uncoordinated gait, inability to balance or coordinate movements, weakness in the legs, sensory loss, and scoliosis. Other symptoms associated with Friedreich's ataxia are hearing loss, diabetes, dysphagia, dysmetria, chewing difficulty, abnormal eye movements, and loss of vision, which are likely the result of extensive degeneration of associated structures. At this point in time, there is no cure for Friedreich's ataxia and no known treatment that reduces the progression of the disorder. Medical treatment includes pharmacological usage, with an emphasis on antioxidants, though there have not been significant findings on the effectiveness of these treatments.

The role of the speech-language pathologist is to treat motor speech disorders that occur in individuals with Friedreich's ataxia in addition to providing therapy for dysphagia. Treatment is primarily behavioral and efforts are focused on problems related to motor control and coordination, which interfere with systems responsible for speech rate and prosody. This presentation will provide a comprehensive overview of Friedreich's ataxia, and provide speech-language pathologists with evidence-based treatment approaches.