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Treating the Incurable: An Overview of Friedreich's Ataxia (FRDA)

The purpose of this presentation is to educate individuals about the progressive neurodegenerative disease, Friedreich's ataxia (FRDA) and to explain the underlying origin of the disease.This includes assessment and treatment, which will allow speech-language pathologists to have a better understanding of FRDA.

Friedreich's ataxia is the most common autosomal recessive ataxia that is caused by a lesion in the cerebellum and corticospinal tract. It is a hereditary disorder that is caused by a mutation of the frataxin gene. The typical onset is in puberty and advances into adulthood (Reetz et al., 2016). The symptoms of the disorder progress in severity as an individual with FRDA ages. A diagnosis of FRDA results in reduced life expectancy and premature death. Speech-language pathologists working with clients who have FRDA should prepare their treatment plan in preparation for the degeneration of their client's symptoms related to: reduced intelligibility, vocal dysphonia, altered prosody, articulatory imprecision, hypernasality, proprioception, higher-order cognition deficits , and depression (Vogel et al., 2017).

Treatments utilized by speech-language pathologists have been minimally researched. To target the loss of higher-order cognitive deficits, Ciancarelli, Coffini, & Carolei (2010) advocate for the use of cognitive rehabilitation that targets: executive functioning, global cognitive functioning, short-term and long-term memory, nonverbal reasoning, lexical fluency, information processing speed, selective attention, working memory, and depression. Reduced intelligibility is targeted through drill therapy that Murry (1983) claims to increase articulatory complexity by differentiating vowel duration as well as contrasting between plosive voiced and unvoiced phonemes. Symptoms associated with dysarthria are targeted using Lee Silverman Voice Treatment (LSVT). Sapir et al., (2013) states that LSVT improves short term and long term memory in phonatory and articulatory functions, intelligibility of speech and overall communication.

Although it is the most common hereditary ataxia, there is limited research targeting treatment and objectives towards delaying the progression of symptoms for Friedreich's ataxia. The role of a speech-language pathologist is to assist individuals with FRDA with their deficits and preserve their current abilities. This poster provides other speech-language pathologists an evidence based foundation that contains overall important information about Friedreich's ataxia, as well as speech therapy that can be implemented.

References

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