

DD SIG Episode 47: Friedreich Ataxia with Jeannie Stephenson & Staci Shearin

In this installment of our series on rare neurologic diseases, Christina Burke talks with Jeannie Stephenson and Staci Shearin about Friedreich's Ataxia, a progressive, autosomal recessive hereditary multi-system neurodegenerative disease that impacts approximately 1 in 40,000 people in the U.S. Jeannie and Staci cover assessment, PT treatment and prognosis for people with this unique diagnosis that causes significant balance and gait challenges.

The Degenerative Diseases Special Interest Group is part of the Academy of Neurologic Physical Therapy – www.neuroPT.org

Guest Information

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Associate Professor, School of Health Professions
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Key Moments:

02:20 – Jeannie explains the genetic basis of Friedreich's Ataxia, its clinical presentation, and how it differs from other ataxias.

09:30 – Staci discusses performing a robust assessment of patients with FA and considerations for helping these individuals stay as active and engaged as possible throughout their lifespan.

17:50 – Jeannie discusses her longitudinal research study on gait, balance, and neurological status in individuals with FA.

23:25 – Chris asks Staci about interventions for patients with FA. Staci discusses evidence-based interventions for FA, including balance activities, and why a focus on the trunk and hips is essential for these individuals.

For More Information on FA:

Friedreich's Ataxia Research Alliance (FARA): <https://www.curefa.org/>

Clinical Management Guidelines for Friedreich Ataxia: frdaguidelines.org

National Ataxia Foundation: <https://www.ataxia.org/>

National Institutes of Health (NIH): <https://www.ninds.nih.gov/health-information/disorders/friedreich-ataxia>

National Organization for Rare Disorders (NORD): <https://rarediseases.org/rare-diseases/friedreichs-ataxia/>

Related resources:

Scale for the Assessment and Rating of Ataxia (SARA): <http://www.ataxia-study-group.net/html/about/ataxiascales/sara/SARA.pdf>

International Co-operative Ataxia Rating Scale (ICARS): <https://www.sralab.org/sites/default/files/2017-06/ICARS.pdf>

Modified Friedreich's Ataxia Rating Scale (mFARS): <https://www.curefa.org/pdf/Instructions-for-administering-the-mFARS.pdf>

Related Articles:

Buckley E, Mazzà C, McNeill A. A systematic review of the gait characteristics associated with Cerebellar Ataxia. *Gait Posture*. 2018;60:154-163. doi:10.1016/j.gaitpost.2017.11.024

Ilg W, Synofzik M, Brötz D, Burkard S, Giese MA, Schöls L. Intensive coordinative training improves motor performance in degenerative cerebellar disease. *Neurology*. 2009;73(22):1823-1830. doi:10.1212/WNL.0b013e3181c33adf

Keller JL, Bastian AJ. A home balance exercise program improves walking in people with cerebellar ataxia. *Neurorehabil Neural Repair*. 2014;28(8):770-778. doi:10.1177/1545968314522350

Milne SC, Kim SH, Murphy A, et al. The Responsiveness of Gait and Balance Outcomes to Disease Progression in Friedreich Ataxia. *Cerebellum*. 2022;21(6):963-975. doi:10.1007/s12311-021-01348-2

Milne SC, Corben LA, Georgiou-Karistianis N, Delatycki MB, Yiu EM. Rehabilitation for Individuals With Genetic Degenerative Ataxia: A Systematic Review. *Neurorehabil Neural Repair*. 2017;31(7):609-622. doi:10.1177/1545968317712469

Siler, M., & Andrews, B. A Multi-modal Treatment Approach for a Young Adult with Friedreich's Ataxia: A Case Report. *JSPTR*. 2014b, February 24. <http://www.ptstudentjournal.org/assets/JSPTR-Volume7-Number1-Article3.pdf>

Stephenson J, Zesiewicz T, Gooch C, et al. Gait and balance in adults with Friedreich's ataxia. *Gait Posture*. 2015;41(2):603-607. doi:10.1016/j.gaitpost.2015.01.002

Zesiewicz TA, Stephenson JB, Kim SH, et al. Longitudinal gait and balance decline in Friedreich's Ataxia: A pilot study. *Gait Posture*. 2017;55:25-30. doi:10.1016/j.gaitpost.2017.03.019

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