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GNE myopathy: Recognizing key features to optimize physical therapy treatment in a rare myopathy

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Background & Purpose: GNE myopathy, a rare autosomal recessive adult-onset disorder with progressive muscle atrophy and weakness, is due to a missing GNE/MNK enzyme, causing a sialic acid deficiency. Progressive distal limb weakness with unique quadriceps sparring presentation is common. Investigational drug trials exist, but the disease currently has no cure. GNE myopathy has often been misdiagnosed, due to large exclusions in the population when histopathologic diagnostic criteria required multiple findings on muscle biopsy. Today the diagnosis relies on clinical presentation, including muscle imaging and is confirmed by genetic studies. GNE myopathy presents with unique patterns of muscle dominance-quadriceps vs. hamstrings, abductors vs. adductors, hip extensors vs. hip flexors, plantar flexors vs. dorsiflexors, biceps vs. triceps-with subjective reports of tripping, difficulty managing steps and rising from chairs. The authors have partook in data collection for a GNE myopathy IRB approved drug trial for 4-years and are now seeing this population in the clinic. There is no literature available on GNE myopathy and physical therapy at this time. This report will identify the clinical characteristics of GNE myopathy and highlight the role of physical therapy (PT) in improving physical function, decreasing falls risk and improving quality of life in this patient (pt) population.

Case Description: Patient is a 42-year old-female, noted a 6 year progressive decline in distal BLE weakness with increased falls. She was referred to PT for strengthening, balance and gait training and also transition from soft over the counter AFOs to custom AFOs. She was not enrolled in a drug trial. Pt presented on evaluation with impaired strength, balance, endurance and increased fear of falls. Pt received 30-60 min individual PT sessions 1-2 times per week for 32 sessions.

Result: At onset of care, pt was evaluated per standard outcome measures to assess for baseline function and falls risk. Her plan of care was established with goals of strengthening dominant muscle groups to optimize function, balance training to decreased falls risk and improve confidence with mobility and progressing high level mobility, with recommendations to appropriate AFOs. Pt demonstrated the below improvements in standard outcome measures from her start to end of care: 5 Time Sit to Stand: 9 seconds to 6 seconds, Timed Up and Go: 7.8 seconds to 6.6 seconds, Gait Speed (GS) self-selected: 1.21 m/s to 1.49 m/s, GS fast: 1.56 m/s to 1.79 m/s, Mini-BESTest: 20/28 to 27/28 and Hi-MAT assessment: 27/54 to 29/54. Pt improved her fall rate from in 3 months.

Conclusion & Discussion: Knowledge of GNE myopathy presentation and prognosis enabled PT to develop targeted strengthening programs to improve functional strength, decrease risk of falls, and improve quality of life. Focused strengthening of dominant muscles in moderate intensity to prevent fatigue is essential in a population with difficulty generating new muscle fibers. Education on appropriate bracing to decrease falls risk and improve high level mobility added to pt quality of life. More research is warranted as treatment options for pts with GNE myopathy progress.

Biography

Jenna DeSimone is a Specialist in Neurological Recovery and is a board certified Neurological Clinical Specialist, and a Senior Staff Physical Therapist at NYU Langone Health. She is a graduate of Villanova University (BS) and received her Doctorate in Physical Therapy from Sacred Heart University in 2012 and joined NYU Langone Health the same year. After spending three years rotating through inpatient care, acute rehab, and outpatient care, she began her specialization in community-based rehab in the neurological spectrum of care. She has since risen to the senior level as a non-rotating member of the Neurological Outpatient Department and is a certified clinical instructor, serving as a Lead Instructor for physical therapy students. Her prior involvement in research includes work with Ultragenyx Pharmaceuticals on Phase II-IV research programs for GNE myopathy and participation in IRB approved research on brain injury, aphasia and physical therapy.

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