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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 a unique quadriceps sparing 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 in managing steps and rising from chairs. The authors have partook in data collection for a GNE myopathy IRB approved drug trial for four 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: Pt is a 42 year old female, noted a six year progressive decline in distal BLE weakness with increased falls. She was referred to PT for strengthening, balance and gait training and to transition from soft over the counter ankle foot orthosis (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. Treatment emphasized strengthening dominant muscle groups to optimize function, balance training, and progressing high level mobility with appropriate AFOs.

Outcomes: First and final outcome measures: 5 time sit to stand was 9 sec to 6 sec, timed up and go 7.8 sec to 6.6 sec, 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 27/54 to 29/54. Fall rate from x1 weekly to x1 in 3 months.

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 patients with GNE myopathy progress.

Recent Publications

1. Brady S, Squier W and Hilton-Jones D (2013) Clinical assessment determines the diagnosis of inclusion body myositis independently of pathological features. *Journal of Neurology, Neurosurgery and Psychiatry* 84(11):1240-1246.
2. Haghigi A, Nafissi S, Qurashi A et al. (2016) Genetics of GNE myopathy in the non-Jewish Persian population. *European Journal of Human Genetics* 24(2):243-51.
3. Kazamel M I, Sorenson E J and Milone M (2016) Clinical and Electrophysiological Findings in Hereditary Inclusion Body Myopathy Compared With Sporadic Inclusion Body Myositis. *Journal of Clinical Neuromuscular Disease* 17(4):190-6.
4. Monies D, Alhindi H N, Almuhaizea M A et al. (2016) A first-line diagnostic assay for limb-girdle muscular dystrophy and other myopathies. *Human Genomics* 10(1):32.
5. Urtizberea J A and Behin A (2105) GNE Myopathy. *Med Sci (Paris)* doi:10.1051/medsci/201531s306.

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Biography

Jenna DeSimone, a specialist in neurological recovery, is a board certified Neurological Clinical Specialist and a Senior Staff Physical Therapist at NYU Langone Health. A graduate of Villanova University (B.S.), Jenna 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 III-IV year physical therapy students. Jenna has presented case studies on CIDP and GNE Myopathy on the national level, most recently at the American Physical Therapy Association's Combined Sections Meeting in San Antonio, Texas (2017) and in New Orleans, Louisiana (2018)

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