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

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Backg ⸗ d & P e: 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 de ciency. 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 o en been misdiagnosed, due to large exclusions in the population when histopathologic diagnostic criteria required multiple ndings on muscle biopsy. Today the diagnosis relies on clinical presentation, including muscle imaging and is con rmed by genetic studies. GNE myopathy presents with unique patterns of muscle dominance-quadriceps vs. hamstrings, abductors vs. adductors, hip extensors vs. hip exors, plantar exors vs. dorsi exors, biceps vs. triceps-with subjective reports of tripping, di culty managing steps and rising from chairs. e authors have partook in data collection for