Benefit of Inpatient Rehabilitation in Patients with Paramyotonia Congenita: a Case Report

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Abstracts will be considered for both poster and platform presentations

Movement disorders

Case Description
Paramyotonia congenita (PMC) is a rare, non-progressive genetic disorder with a reported prevalence of 1 in 100,000 within the United States. The autosomal dominant disorder is caused by a mutation of muscle sodium channel gene SCN4A and is characterized by episodic myotonia. Inability of voltage gated sodium channels to maintain appropriate sodium ion influx results in increased propagation of action potentials to the sarcoplasmic reticulum and a subsequent rise in calcium ion release. This in turn causes muscle myotonia through the muscle contraction cascade. Myotonia can be exacerbated by extreme physical activities or cold temperatures.

Discussions
We present the case of a 43 year old Caucasian female with a known history of PMC confirmed via genetic testing and electromyography. She presented to the emergency department as a stroke alert with a sudden onset of right sided hemiparesis. Extensive stroke evaluation was negative for an ischemic event and the patient was diagnosed with a “PMC exacerbation. ” Given her functional limitations at the time, she was transferred to an acute inpatient rehabilitation facility. Initial therapy evaluations showed significant functional deficits in all activities of daily living including ambulation that was present for a period of years prior to admission. Therapy emphasized endurance as well as energy conservation techniques. Additionally, multiple medication changes were completed including dosing adjustment and initiation of new medications. Upon completion of her acute rehabilitation and discharge to home, she was independent for all activities of daily living including household ambulation without an assistive device. She was also able to manage her daily activities without myotonia exacerbations.

Conclusions
Appropriate therapy techniques and medication management in an inpatient rehabilitation setting can provide substantial benefit in patients with functional deficits related to paramyotonia congenita.