Abstract Form

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Project Title:	A Case Study of Exertional Rhabdomyolysis Complicated by Myotonia Congenita

Research Category (please check one):							
☐	**Original Research**	☒	**Clinical Vignette**	☐	**Quality Improvement**	☐	**Medical Education Innovation**

Abstract

Introduction: Myotonia congenita is a rare congenital neuromuscular channelopathy characterized by an inability of the muscles to relax quickly following voluntary contraction. Patients often experience muscle stiffness, transient weakness, and occasional muscle pain triggered by sudden exertion or exercise after rest. These patients are at higher risk for developing exertional rhabdomyolysis due to impaired muscle relaxation. This case report describes a patient with a known diagnosis of myotonia congenita who developed exertional rhabdomyolysis following a spin class session.

Case Report: 27-year-old female with a past medical history of myotonia congenita and post-operative hypothyroidism who presented with severe bilateral anterior thigh pain, swelling, weakness, muscle stiffness, difficulty ambulating, and dark urine after participating in a spin class session following a 2-month hiatus from the gym. Her myotonia congenita condition was diagnosed via genetic testing during childhood, is characterized by intermittent stiffness in her hands/wrists and lower extremities, with symptoms typically well managed with mexiletine. Notably, her father also carries a diagnosis of myotonia congenita. Initial evaluation in the emergency department revealed hypertension (BP 149/92) and tachycardia (HR 107); the rest of her vital signs were within normal limits. Physical exam was remarkable for mild bilateral anterior quadriceps tenderness without bruising or firm compartments, and normal power in the lower extremities. Laboratory findings were notable for a hemoglobin of 14.3, ALT 355, AST 1024, and a markedly elevated CK of 117,123, with normal renal function. Urinalysis revealed red-colored urine with 3+ blood, 2+ protein, and 6-10 RBCs per high-power field. Right upper quadrant ultrasound findings were unremarkable. The patient received aggressive IV fluid hydration with normal saline. Although her symptoms improved and CK decreased to 45,139 on day 2 of hospitalization, it subsequently increased and peaked at 164,022 later that same day. IV sodium bicarbonate was thus added to the fluid regimen to aid CK clearance. The patient maintained good urine output and stable renal function throughout the hospital course. Due to the persistent CK level elevation and fluctuations, coupled with suspected delayed recovery from rhabdomyolysis related to myotonia congenita, physical therapy was suspended, and the patient was advised to avoid strenuous physical activity until her CK levels improved. MRI of bilateral thighs revealed diffuse quadriceps muscle edema and no abscess or necrosis. CK levels began to steadily decrease by day 4 of hospitalization, reaching a nadir of 7,993 by discharge on day 7 of hospitalization. The patient was instructed to follow-up with her PCP and neurologist to obtain a specialized referral to a neuromuscular specialist for ongoing management of myotonia congenita.

Discussion: Although patients with congenital myopathies represent a small fraction of rhabdomyolysis cases, they are at higher risk for developing exertional rhabdomyolysis. In the context of myotonia congenita, this patient was likely predisposed to developing muscle injury due to impaired muscle relaxation following intense exercise. Despite the similarities in management of rhabdomyolysis compared to patients without congenital myopathies, this case highlights a fluctuating and prolonged recovery course of CK levels in a patient with myotonia congenita, likely due to impaired muscle relaxation inherent in this condition. Post-discharge follow-up with a neurologist who specializes in neuromuscular disorders is important for long term management of myotonia congenita and prevention of future rhabdomyolysis episodes.