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Exertional Rhabdomyolysis in a Young Male with Suspected McArdle Disease

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Abstract

Introduction: McArdle disease is a rare inherited disorder of skeletal muscle metabolism caused by deficiency of myophosphorylase, leading to impaired glycogenolysis during exercise. It typically presents in young individuals with exercise-induced muscle pain, fatigue, and recurrent episodes of rhabdomyolysis. Early recognition is essential, as delayed diagnosis can result in repeated muscle injury and complications, particularly in physically active populations.

Case Description: A young male presented for evaluation of persistently elevated creatine kinase (CK) levels approaching 100,000 U/L and dark, blood-tinged urine following a moderate-intensity workout. He denied medication use, supplement intake, or substance exposure, and urine toxicology screening was negative. Family history was notable for a brother with similar episodes requiring hospitalization after exercise. Physical examination was unremarkable, and the patient denied systemic symptoms.

The patient was treated with aggressive intravenous fluid resuscitation, including an initial bolus followed by maintenance fluids, with close monitoring of CK levels and renal function. On admission, CK was markedly elevated at 102,148 U/L. Levels trended downward with treatment: 90,157 U/L on hospital day 2, 42,462 U/L on day 3, 14,155 U/L on day 4, and 6,162 U/L by day 5. Renal function remained stable throughout hospitalization, with no evidence of acute kidney injury.

This study received Institutional Review Board approval (MSH-IRB-26-11: Exertional Rhabdomyolysis and Myoglobinuria in a Young Male), and informed patient consent was obtained.

Discussion: In athletic and physically active individuals, recurrent rhabdomyolysis with myoglobinuria should raise suspicion for underlying metabolic myopathies such as McArdle disease. The patient's markedly elevated CK levels, exercise intolerance, and positive family history are highly suggestive of this diagnosis. Delayed recognition can lead to recurrent muscle breakdown, renal complications, and reduced quality of life.

Further diagnostic evaluation is planned, including genetic testing for mutations in the PYGM gene, non-ischemic forearm exercise testing, and possible muscle biopsy if indicated. The patient was referred to a neuromuscular specialist and counseled on exercise modification, pre-exercise carbohydrate intake, and early recognition of rhabdomyolysis symptoms.

Conclusion: This case highlights the importance of maintaining a high index of suspicion for metabolic myopathies in young adults presenting with exertional rhabdomyolysis. Early diagnosis of McArdle disease allows for targeted management strategies, safer exercise practices, and prevention of recurrent muscle injury and long-term complications.