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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, resulting in impaired glycogenolysis during exercise. It typically presents in young individuals with exercise-induced muscle pain, fatigue, and recurrent episodes of rhabdomyolysis, posing diagnostic and management challenges, 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 of exercise-induced hospitalizations. Physical examination was unremarkable, and the patient denied systemic symptoms.

Methods:
This study received Institutional Review Board approval (MSH-IRB-26-11— *Exertional Rhabdomyolysis and Myoglobinuria in a Young Male*). Patient consent was obtained. The principal investigator for the study was Sandhia Senthilnathan.

Findings:
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, creatine kinase (CK) was markedly elevated at 102,148 U/L. The patient was initiated on aggressive intravenous fluid resuscitation, resulting in a steady decline in CK levels over the course of hospitalization, decreasing to 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 admission, with no evidence of acute kidney injury.

Discussion:
In athletic and active individuals, recurrent rhabdomyolysis with myoglobinuria should raise suspicion for metabolic myopathies such as McArdle disease. Delayed diagnosis can lead to repeated episodes of muscle breakdown, renal complications, and significant impairment in physical performance and 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 modifications, adequate carbohydrate intake prior to exertion, and recognition of early symptoms of rhabdomyolysis.

Conclusion:
This case highlights the importance of maintaining a high index of suspicion for metabolic myopathies in young adults presenting with exertional rhabdomyolysis. Early recognition of McArdle disease can guide management strategies, support safe exercise practices, and help prevent recurrent muscle injury and long-term complications.