ISN WORLD CONGRESS OF NEPHROLOGY 2025
FEBRUARY 6-9, 2025 | NEW DELHI, INDIA

Acute kidney injury from rhabdomyolysis revealing glycogen storage disease

Dahmane Rihem, Zmantar Yosra, Mahfoudh Olfa, Boukadida Raja, Ghenam Nehed, Farhat Roua,Ben Aicha Narjes, Sahtout Wissal, Guedri Yosra, Mrabet Sanda, Azzabi Awatef, Zellama Dorsaf

Introduction:

Glycogen storage disease due to a deficiency in phosphoglycerate mutase is characterized by exercise intolerance. It is caused by homozygous or compound heterozygous mutations in the PGAM2 gene, which codes for the muscle isoenzyme of phosphoglycerate mutase, an enzyme involved in the final steps of glycolysis.

This case report describes a patient who presented with acute kidney injury secondary to rhabdomyolysis. The underlying cause of the rhabdomyolysis was subsequently identified as glycogen storage disease.

Methods

This is a case of a 27-year-old female patient who presented to the emergency department with severe fatigue, diffuse myalgia, and uremic syndrome.

The case

This is a 27-year-old female patient with no significant past medical history. The family history shows Presumptive metabolic myopathy in her father and one of her two brothers. Her father passed away at the age of 58 due to a cardiac disease.

The onset of her illness occurred in March 2024, starting with nausea, vomiting, severe fatigue, and muscle pain and weakness. These symptoms developed following physical exertion and a day of fasting.

The patient presented to the emergency department. The initial examination revealed severe fatigue with difficulty walking.

Laboratory tests showed renal insufficiency with a creatinine level of 1090 micromol/L, urea at 30 mmol/L, hyperkalemia at 6.2 mmol/L, hepatic cytolysis at 10 times the normal level, and rhabdomyolysis with creatine kinase (CPK) at 31,600 IU/L and lactate dehydrogenase (LDH) at 22,140 IU/L. Urinalysis revealed leukocyturia and hematuria without proteinuria.

She was admitted to the nephrology department.

The renal ultrasound showed no abnormalities. An electromyogram (EMG) ruled out myogenic involvement.

Treatment was initiated with intravenous hydration and correction of hyperkalemia, leading to good improvement, including a reduction in CK and LDH levels and a decrease in creatinine to 100 micromol/L within three weeks.

The muscle biopsy revealed an excess accumulation of glycogen. Cardiac and liver function tests were within normal limits. Genetic analysis confirmed glycogen storage disease by detecting a mutation in the PHKA2 gene (Xp22.2-p22.1).

Conclusion :

This case highlights the importance of considering metabolic disorders in the differential diagnosis of rhabdomyolysis-induced AKI.

Early diagnosis through genetic testing allows for timely initiation of preventive measures, reducing the risk of severe complications associated with glycogen storage disease.