Online publication ahead of print, AJMHS Vol 56, 2021

Rhabdomyolysis Due to Carnitine Palmitoyltransferase II Deficiency – a Common but Underrecognized Condition

Entela Greca[1*], Saimir Seferi[1], Anila Babameto[2], Ahmet Duraku[1], Bjeshkatore Selmani[1]

[1] Nephrology Department, "Mother Teresa" University Hospital, Tirana, Albania
[2] Genetic Department, "Mother Teresa" University Hospital, Tirana, Albania

Abstract

A young 33-year-old male goes to the emergency room with weakness, nausea, anuria, which started three days before admission. The symptoms appeared after a prolonged exercise. An acute kidney injury developed, and Hemodialysis treatment was needed. At the clinical presentation, he had a high plasma Creatine Kinase (CK) level and CK-MB level. The genetic testing confirmed the diagnosis of Inherited Rhabdomyolysis, a metabolic disorder of Carnitine Palmitoyl transferase II Deficiency.

Keywords: Carnitine Palmitoyltransferase II deficiency, Rhabdomyolisis, metabolic disorder, acute renal failure, genetic disorder

Address for correspondence: Entela Greca*, Nephrology Department, "Mother Teresa" University Hospital, Tirana, Albania. E-mail: entigreca@gmail.com