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Urea cycle disorder misdiagnosed as multiple sclerosis: A case report

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Urea cycle disorders (UCD) are a collection of inborn errors of metabolism caused by dysfunction of any of the six enzymes or two transport proteins involved in urea biosynthesis. Urea cycle is the final pathway for nitrogen metabolism and dysfunction of this important pathway cause UCD, which are more common during the neonatal period. The suspicion for this metabolic disorder arises when patients present with elevated blood ammonia level and neurological manifestation without underlying hepatocellular dysfunction. In this article, we report a patient who presented with neurological dysfunction and coma in the immediate postpartum period. She was misdiagnosed for many years as a case of demyelinating disorder. The diagnosis was confirmed based on the presence of urine orotic acid and elevated certain serum amino acid levels. Hemodialysis was performed to the patient to correct the hyperammonemic-related dysfunction which was unresponsive to conventional measures. She improved gradually with repeated hemodialysis and made a full recovery. Her clinical and radiological status has not changed for five years since diagnosis was made. The importance of reporting this case is to illustrate that wrong diagnosis of patients as being affected with multiple sclerosis for many years due to MRI abnormalities rather than classical relapsing remitting nature of the disease may lead to grave consequences. In addition, the patient was treated with intravenous steroids several times which is contraindicated in patients with UCD as it may precipitate acute hyperammonemic attacks. We believe that the presence of symmetrical hyperintense insular cortical changes is pathognomonic for UCD. This radiological sign is extremely rare and seldom reported in the literature.

Biography

Hussein Algahtani is the Associate Dean of clinical affairs and the Head of the simulation Center in the College of Medicine at King Saud bin Abdulaziz University for Health Sciences in Jeddah, Saudi Arabia. He is also an Assistant Professor in Neurology and the neurosciences block coordinator. In addition, he is the Neurology section Head and the Head of Neurophysiology laboratory at King Abdulaziz medical city in Jeddah, Saudi Arabia. He is a well-known researcher with more than 50 publications in the literature.

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