Methylmalonic Acidemia: Uncommonly Missed Diagnosis in Adults. Usefulness of doing MMA for seizures/neuropathy/weakness/ataxia

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Abstract
Purpose: To understand the presentation of Methylmalonic Acidemia in adults and prevent future missed diagnoses.

Introduction: Methylmalonic acidemia is a rare, autosomal recessive, condition that affects cobalamin metabolism leading to elevated levels of serum and urine methylmalonic acid, despite adequate levels of vitamin B12. It typically presents in the neonatal period with ketoacidotic hyperammonemic coma and elevated methylmalonic acid in the serum and the urine. Most patients die shortly after birth or have long-term neurological problems.

Methods: We followed a patient diagnosed with Methylmalonic Acidemia in adulthood to understand how the condition manifests in adults and how best to treat him. We completed neuropsychological testing, electromyography (EMG), and a variety of laboratory examinations to understand the full extent of the impact of the diagnosis on the patient.

Outcomes: The patient was referred to hematology and university of Miami medical center. He also started participating in physical therapy to regain his strength. He is currently on a modified diet and a physical therapy regimen and is just beginning to show signs of improvement. We are following closely.

Discussion: The presentation of Methylmalonic Acidemia adults can be extremely variable but can be life threatening. There is a reported case in literature where a patient developed muscle paralysis and had to be intubated for respiratory failure and improved almost immediately after diagnosis and treatment. Here, we present a case of a 31 year old man who presented with seizures, polyneuropathy, ataxia and memory loss who was found to have undiagnosed Methylmalonic Acidemia.