Abstract Form

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Project Title:	A Unique Presentation of a Well-Functioning Adult with Methylmalonic Acidemia

Research Category (please check one):

☐	**Original Research**	☒	**Clinical Vignette**	☐	**Quality Improvement**	☐	**Medical Education Innovation**

Abstract

INTRODUCTION

Methylmalonic acidemia (MMA) is a lethal autosomal recessive disorder diagnosed in the infants involving inborn defects in amino acid metabolism. Mortality is reported at a median age of death at 2.2-years in patients with the *mut0* enzymatic subtype. We present a case of a 29 year old female with MMA mut0 subtype, or complete deficiency of methylmalonyl coA mutase, presenting with intellectual deficit without severe neurological dysfunction. Her clinical picture represents a rare presentation of MMA, as patients with her disease typically do not survive past the first few years of life.

CASE PRESENTATION

A 29-year-old Caucasian female with MMA subtype mut0, status-post liver and kidney transplant, splenectomy, type II diabetes, scoliosis, and developmental delay secondary to neonatal trauma who presented to the emergency department with one episode of new onset witnessed generalized tonic-clonic seizure. At baseline, she ambulates short distances without use of DME, completes ADLs with assistance, independently feeds self and brushes her teeth, receives nightly tube feeds through G tube, but eats small meals. Prior to this, patient had recent head trauma of unwitnessed fall; without loss of consciousness, but mildly confused and somnolent. She was initiated on Levetiracetam. A 24 hour EEG showed no seizure activity. CT Brain without contrast was negative for acute infarct, bleed, or mass. MRI Brain without contrast showed focal cortical and subcortical white matter signal abnormality involving the high left frontal lobe concerning for a glioma. During the hospital course patient had no further seizure activity and has been stable while on Levetiracetam.

DISCUSSION

MMA presents within the neonate period as a sepsis-like picture with poor feeding, vomiting, hypotonia, ketosis, acidosis, and seizures. Patients will experience multisystemic disease, involving neurologic, renal, intellectual, and ocular deficits. Isolated bilateral infarcts have only been reported in MMA. Developmental delay was also found to be present in all MMA patients with cobalamin-C metabolism error[7].

Kidney transplantation provides enough enzyme activity to allow for methylmalonic acid excretion. Kidney transplantation is recommended often in combination with a liver transplant as it can improve survival and quality of life and prevent further neurological deterioration. The metabolic conversion of propionate occurs in the liver, therefore a transplant can compensate to avert metabolic decompensation[10]. It has been concluded that patients who have had a liver transplant lived 1.5 years longer with 7.9 quality-adjusted life years than patients who were treated with nutritional management alone[11].

Mortality in patients with the mut0 subtype has changed dramatically over time. One study demonstrated that 100% of patients died at a median age of 1.6-years-old in the 1970, 50% died at 7.6-years-old in the 1980s, and 20% died at 2.2-years-old in the 1990s[12]. Our patient is a well-functioning adult with liver and kidney transplant with intellectual disability but without severe neurological dysfunction. She has considerably exceeded mortality expectations.

CONCLUSION

In the last few decades, there have been breakthrough studies that show treatment options can prolong life expectancy and improve quality of life in patients with the mut0 subtype. Both kidney and liver transplants have proved beneficial, as evidenced by the decrease in mortality rates. As research in this area expands, both parents and patients with MMA can be hopeful about leading a life with minimized complications.