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Compromised therapeutic value of pediatric liver transplantation in ethylmalonic encephalopathy: A case report

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Abstract

BACKGROUND

Ethylmalonic encephalopathy (EE) is a rare autosomal recessive metabolic disorder caused by impaired mitochondrial sulfur dioxygenase. Due to poor therapeutic effect of current conventional treatments, progressive psychomotor regression and neurological impairment usually contribute to early death in the first decade of life. Liver transplantation (LT) is emerging as a novel therapeutic option for EE; however, worldwide experience is still limited.

CASE SUMMARY

An 18-month-old patient with the diagnosis of EE received a living donor liver transplant in our institution, which, to our knowledge, is the first case in Asian-Pacific countries. During 20 mo of follow-up, the longitudinal metabolic evaluations revealed a wild fluctuation in urinary EMA levels, still far beyond the normal range. Urinary 2-methylsuccinic acid levels gradually restored to normal, whereas the concentrations of urinary isobutyrylglycine and plasma C4- and C5-acylcarnitines fluctuated markedly

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Cited by: 19 **Author:** Carlo Dionisi-Vici, Daria Diodato, Giuliano T...
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By reading this **case report** on a patient with an unusually mild form of **ethylmalonic encephalopathy** (EE), readers will learn about the acute and chronic management of this rare condition and about the importance of keeping metabolic causes, such as EE, in mind in patients presenting with a purely neurological phenotype.

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Frontiers | Novel Compound Heterozygous Variants of ETHE1

Ethylmalonic encephalopathy

Rare Autosomal Recessive Inborn Error

Ethylmalonic encephalopathy is a rare autosomal recessive inborn error of metabolism. Patients affected with EE are typically identified shortly after birth, with symptoms including diarrhea, petechiae and seizures. The genetic defect in EE is thought to involve an impairment in the degradation of sulfide intermediates in the body. Hydrogen sulfide then builds up to toxic levels. EE was initially described in 1994. Most cases of EE have been described in individuals of Mediterranean or Arabic origin.

Wikipedia

Data from: Wikipedia

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