A Rare Cause of Episodic Vomiting: Late-onset Multipl Acyl-CoA Dehydrogenase Deficiency As a Treatable Metabolic Myopathy

Ayşe Yasemin Çelik[1], Çiğdem Seher Kasapkara[2], Burcu Civelek Ürey[2], Esra Kılıç[3], Nesrin Ceylan[1], Bekir Furkan Yalçın[4], Didem Ardıçlı[1]

[1] Ankara City Hospital, Children's Hospital, Department of Pediatric Neurology

[2] Ankara City Hospital, Children's Hospital, Department of Pediatric Metabolism

[3] Ankara City Hospital, Children's Hospital, Department of Pediatric Genetics

[4]Ankara City Hospital, Children's Hospital, Department of Pediatrics

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Introduction:

Multiple acyl-CoA dehydrogenase deficiency (MADD) is a rare inherited metabolic disorder of fatty acid, amino acid, and choline metabolism, caused by recessive mutations in the ETFA, ETFB, and ETFDH genes. Herein, we present a case of late-onset MADD, who presented with neck weakness and episodes of vomiting.

Case Report:

An 11-year-old boy presented with vomiting attacks, easy fatigue, and weight loss for the last 6 months. He was referred to our hospital for further investigation with a suspicion of a possible malignancy. Prenatal, natal, and developmental history was normal. Family history revealed parental consanguinity and a deceased sibling at the age of 3 years following a febrile infection. On neurological examination, he had prominent cervical and proximal weakness, nasal speech, dysphagia, and absent deep tendon reflexes (Figure 1). Laboratory workup showed elevated transaminase (AST/ALT: 1645/895 U/L) and serum creatinine kinase levels (2389 U/L). The patient's eye examination, cardiological evaluation, and abdominal USG were normal. The magnetic resonance imaging of the head and spine was normal. Metabolic tests showed a characteristic acylcarnitine profile suggestive of MADD. Molecular analysis of the *ETFDH* gene confirmed a homozygous pathogenic variant c.1130T>C (p.Leu377Pro). He showed dramatic improvement to high-dose riboflavin (2x100 mg), coenzyme Q 10 (5 mg/kg/day), and carnitine (100 mg/kg/day) supplementations. The patient was discharged 5 days after the treatment with normal muscle tone and muscle strength (Figure 1). On the sixth month follow-up, he had a normal systemic and neurological examination without any symptoms.

Figure 1: A-B: Patient photography before treatment showing prominent cervical and axial weakness. C: Dramatic improvement of muscle strength after treatment.

Conclusion:

MADD may be misdiagnosed as a different type of lipid storage myopathy, a glycogen storage disease, progressive muscular dystrophy, or other muscular or gastrointestinal diseases. Since it is a treatable disorder, the diagnosis of MADD deficiency should be considered especially in patients presenting with fluctuating proximal/axial weakness, episodic vomiting, and easy fatigue. Early treatment with dietary regulations, high-dose riboflavin, carnitine supplementation in those with carnitine deficiency, and coenzyme Q_{10} supplementation is life-saving.

References:

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