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RESEARCH LETTER

Very long-chain acyl-coenzyme A dehydrogenase deficiency —A rare monogenic steatotic liver disease presenting with acute liver injury in adulthood: A case report and brief review on nutrition management

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Dear Editor,

Very long-chain acyl-coenzyme A dehydrogenase (VLCAD) catalyzes the first step of mitochondrial beta-oxidation of long-chain fatty acids (LCFA), having a chain length of 14 to 20 carbons [1]. The deficiency of VLCAD (VLCADD, OMIM 201475) is an exceptionally rare disorder with autosomal-recessive inheritance, characterized by mutations in the ACADVL gene on chromosome 17p13 [1]. This rarity underscores the unique and specialized nature of the case.

The case report was prepared after obtaining consent and ensuring patient anonymity.

A 23-year-old lady from southern India presented with a history of non-projectile, non-bilious vomiting for 25 days. There was no history of abdominal pain, diarrhea, dysphagia, fever or headache. She is the firstborn to third-degree consanguineous parents. She had a history of recurrent episodes of vomiting in childhood lasting one to two days and requiring intravenous fluids following short febrile illnesses. There was no history of similar disease in the family. On examination, her body mass index was 21.6 kg/m^2. She was dehydrated at presentation but did not have generalized muscle tenderness with normal systemic examination.

The supplementary material includes her biochemical and hematological investigations at presentation. She had total bilirubin 3.2 mg% (direct 2.8 mg%), aspartate aminotransferase (AST) 243 U/L, alanine aminotransferase (ALT) 184 U/L and international normalized ratio (INR) 1.52, suggestive of acute liver injury (ALI). She had normal creatinine phosphokinase (CPK) levels. Computerized tomography (CT) of the abdomen showed diffuse fatty infiltration of the liver, normal biliary system and portal and hepatic veins. Etiological evaluation of ALI ruled out viral hepatitis, tropical infections such as scrub typhus and leptospirosis, drug/toxin intake, Wilson's disease and autoimmune hepatitis (AIH).

Percutaneous liver biopsy showed diffuse marked macrovesicular steatosis, minimal cholestasis, no significant fibrosis and no evidence of AIH (Fig. 1A). Further evaluation confirmed the diagnosis of VLCADD (Fig. 1B).

After stabilization, she was started on a VLCFA-restricted diet and advised to seek care during intercurrent illnesses. During the three-month follow-up after diagnosis, the bilirubin levels and INR normalized and the aminotransferases were in the improving trend (total bilirubin 0.7 mg%, AST 113 U/L, ALT 44 U/L and INR 1.2).

VLCADD belongs to disorders of intermediary metabolism and, hence, can present with life-threatening acute metabolic decompensation [2]. Triggers for such

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