LETTER TO EDITOR (VIEWERS CHOICE)

RECURRENT VOMITING IN A CHILD: A RARE CASE OF ISOVALERIC ACIDEMIA

Patil SV, Kalyanshettar SS, Naren SD, Kiran kumar

A 3 1/2 years old girl, born to 2nd degree consanguineous marriage was admitted with chief complaints of recurrent episodes of projectile non-bilious vomiting since 6 months of age each lasting for 3-4 days and recurs every 4 to 5 months and each time requires hospital admission. Child used to be normal in between episodes. She had a NICU admission on 5th day of life for vomiting. A sibling had died on 19th day of life with similar complaints. On examination, she was drowsy, had tachycardia, tachypnea and moderate dehydration. A clinical diagnosis of cyclical vomiting was made and child was started on antiepileptic for abdominal migraine. Vomitus used to have a peculiar odor and this made us suspect other causes for the condition of the child. The child was investigated further. Urine routine was normal except for ketone bodies. Blood gases showed metabolic acidosis with high anion gap. Urine screening (thin layer chromatography) revealed mild increase in leucine, isoleucine and valine, alanine and hydroxyproline. Blood tandem mass spectrometry revealed marginally elevated serum homocysteine level and acylcarnitine pattern which was suggestive of isovaleric acidemia. Patient was started on dietetic management, carnitine 100mg/kg/day & megavitamin therapy. The child improved subsequently and was advised to continue carnitine.

Isovaleric acidemia is a rare autosomal recessive disorder with an incidence of 1 in 2 lakhs (1). It is due to deficiency of Isovaleryl-CoA dehydrogenase required for the metabolism of the amino acid, leucine. It presents within a few days or weeks of life with vomiting and ketoacidosis progressing to lethargy, coma and death in greater than 50% of the patients (2). We report a rare case of isovaleric acidemia presenting as recurrent vomiting. Distinctive odor of "sweaty feet" during an illness due to accumulation of volatile isovaleric acid is classical (3). Diagnosis involves screening for isovaleric acidemia using tandem mass spectrometry analysis of dried blood spot specimen. Treatment involves reducing protein intake, particularly the branched-chain amino acid leucine. During an acute episode, aggressive use of glucose and electrolytes is necessary. Glycine supplementation has proven beneficial because this amino acid is conjugated to isovalerate, forming the less harmful isovalerylglycine. Carnitine treatment is similarly effective (4-6).

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From: Department of Pediatrics, BLDE University, Sri BM Patil Medical college, Bijapur, Karnataka.

Address for Correspondence: Dr SS Kalyanshettar, Department of Pediatrics, BLDE University, Sri BM Patil Medical college Bijapur, Karnataka, India. Email: ssk_dr@yahoo.co.in

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