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Case Report

Recurrent metabolic acidosis: is it by chance?

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ABSTRACT

Succinyl CoA 3 oxoacid CoA transferase deficiency (SCOTD) is an autosomal recessive disorder of ketone body utilization which present with recurrent episodes of metabolic acidosis. We have a 23-month old child who presented with 3 episodes of metabolic acidosis. Each time the toxicology profile came positive for ethyl alcohol. We couldn't get any history suggestive of poisoning. So we were in a great dilemma and considered the possibility of Munchausen syndrome by proxy and a metabolic cause. Even though the initial genetic result was nonspecific when we reanalyzed the sample for genetic analysis it came positive as Succinyl CoA 3 oxoacid CoA transferase (SCOT) deficiency. The child required intensive care support in all 3 times. We were able to manage the child with supportive measures each time. Now the child is under regular follow up and doing well. SCOTD should be considered in any children presenting as recurrent episodes of metabolic acidosis. The rarity of this disorder will lead to the diagnostic dilemma that we face as like in this case.

Keywords: Metabolic acidosis, SCOT deficiency, Ketone body, Toxicology

INTRODUCTION

Succinyl CoA 3 oxoacid CoA transferase deficiency (SCOTD) is a rare autosomal recessive disorder characterized by severe potentially fatal intermittent episodes of ketoacidosis due to defect in the pathway of ketone body utilization.[1]

We report a case of SCOTD deficiency in a 23-month-old male child presenting as recurrent episodes of metabolic acidosis requiring intensive care each time. On literature search only 30 cases have been reported to date.

CASE REPORT

A 23-month-old boy developmentally normal and immunized for age with uneventful history presented to our quaternary care center with fever and vomiting of 3-days duration with labored breathing of 1-day duration.

At admission child was tachypneic, drowsy with acidotic breathing. He was tachycardic (heart rate-170 /min), respiratory rate-40/ min, and blood pressure (100/60 mm hg). Systemic examination was normal. NG aspirate showed a brownish fluid with an alcoholic odor.

Blood gas showed high anion gap metabolic acidosis (6.8/22/195/3.6, lactate: 0.7, anion gap: 18). Blood sugars (98 g/dl) and serum electrolytes (S. Na: 141, S. K: 4.7, ionized Ca: 1.18) were normal. Liver (SGOT: 20, SGPT: 74) and renal function tests (BU/S Cr: 42/0.8) were also normal. Urine acetone was positive, but serum acetone was negative. Echo ruled out any structural heart disease. In this background we have narrowed our differentials to poisoning (alcohol) and inborn errors of metabolism.

On further enquiry revealed history of consumption of ayurvedic medicine at home by parents. Samples from the NG aspirate and the ayurvedic preparation revealed an alcoholic content. A tandem mass spectrometry was

performed which showed elevated malonyl carnitine (5.36 MOM (0.00-4.55)) which was nonspecific. Magnetic resonance imaging (MRI) brain and nerve conduction study were normal. Hence, an initial diagnosis of poisoning with ethyl alcohol from indigenous medicine was considered. Child required mechanical ventilation for stabilization and the metabolic acidosis was refractory to multiple boluses of bicarbonate which eventually ended in peritoneal dialysis. He also developed aspiration pneumonia with ARDS requiring high frequency oscillatory ventilation (9 days of ventilation). Post extubation he had complete head lag with muscle weakness (power-3/5) with depressed DTR that later improved with physiotherapy and he was discharged.

Second admission was within 1 week of previous episode, with complaints of vomiting of 1-week duration and lethargy. This time also blood gas showed a high anion gap metabolic acidosis. He needed intubation and mechanical ventilation due to impending cardiorespiratory failure. Repeat samples for toxicology was positive for ethyl alcohol. So, the possibility of poisoning and Munchausen syndrome by proxy was considered. But even after taking a detailed history from parents we could not find anything abnormal. Hence, a gene panel for IEM was sent which showed DLD gene mutation which was a variant of unknown significance. Child improved with symptomatic measures and was discharged on vitamin supplements.

Third admission was after 1 year with similar presentation. Our suspicion of Munchausen syndrome by proxy become stronger and considered informing police. This time toxicology quantitative analysis was sent which showed very less (<10 mg/dl) of ethyl alcohol. So, reanalyzed the gene panel for IEM and it came positive for ketone body synthesis defect (OXCT-1), and succinyl CoA oxoacid transferase deficiency. Child was discharged with oral bicarbonate, riboflavin, carnitine, vitamin supplements and dietary restriction. On follow up child was developmentally normal and doing well.

DISCUSSION

SCOTD is a rare genetic disorder in ketone body utilization characterized by severe potentially fatal intermittent episodes of ketoacidosis. It is an autosomal recessive disorder with a prevalence of <1/1000000. Age of onset is 1st week of life to 22 months of age.

It is precipitated by febrile illness, fasting, stress and prolonged physical exertion. Usually, patients will be asymptomatic between ketoacidosis episodes. Clinical manifestations include tachypnea, vomiting, coma, hypotonia with a normal psychomotor development. There will be episodic ketoacidosis without hypoglycemia and dehydration.

Lab investigations will show hyperketonemia, ketonuria, and low to normal carnitine. Enzyme assay will show SCOT deficiency and genetic analysis will show mutation in 3 oxoacid CoA transferase 1 gene mutation.

Differential diagnosis of this includes physiological ketosis, beta ketothiolase deficiency, fatty acid oxidation defects and monocarboxylate transporter 1 deficiency.[1]

These children require moderate protein restriction with avoidance of prolonged fasting. Metabolic acidosis requires oral bicarbonate supplementation.[2] All the episodes require management in intensive care setting. People with SCOT deficiency usually have a permanently elevated level of ketones in their blood (persistent ketosis). The frequency of ketoacidosis attacks varies among affected individuals.[3]

Risk of premature death during neonatal and infantile period is high. Usually, frequency decreases after 10 years of age. Life expectancy is as good as general population. These children usually have normal growth and development when proper treatment and diet is followed.[1]

CONCLUSION

Recurrent metabolic acidosis is always challenging in terms of determining an etiology and preventing another attack. Though the history of ayurvedic medicine exposure, alcohol in the toxicological profile, initial nonspecific genetic mutation in clinical exome were pointers for malingering but repeated reviewing of history, analysis of clinical features and investigations made us to think strongly of a defect in the metabolic pathway involving ketone body metabolism that made us to reanalyze the clinical exome and arrive at the diagnosis.

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