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Maple Syrup Urine Disease – A Rare Case Report

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Abstract: Maple syrup urine disease (MSUD) is an inborn error of metabolism. It is a rare autosomal recessive inheritance disorder.It occurs in 1 in180, 000 infants. It is due to the defect in the metabolism of branched chain amino acids such as valine, leucine, and isoleucine. The enzyme deficient is mitochondrial branched-chain alpha-ketoacid dehydrogenase complex (BCKAD).we have presented a case report of a new born baby girl with classical symptoms of maple surup urine disease. It was confirmed by high performance liquid chromatography (HPLC). Dietary management was done by pediatrician. Baby responds well to the treatment.

Keywords: MSUD, HPLC, alpha ketoacid dehydrogenase.

INTRODUCTION

Maple syrup urine disease is an inborn error of metabolism. It is a rare autosomal recessive inheritance disorder.It occurs in 1 in180, 000 infants [1]. It is due to the defect in the metabolism of branched chain amino acids such as valine, leucine, and isoleucine. The enzyme deficient is mitochondrial branched-chain alpha-ketoacid dehydrogenase complex (BCKAD). The BCKAD is a large complex with four subunits (E1a, E1b, E2 and E3) and is necessary for decarboxylation of branched-chain ketoacids (BCKAs), which is the second step in the degradation pathway of branched-chain amino acids (BCAAs) leucine, isoleucine and valine.

Mutations in both alleles encoding any subunit can result in the accumulation of BCAAs, all isoleucine and their corresponding BCKAs measured in blood, urine and cerebrospinal fluid. The clinical manifestation of MSUD depends on the severity of BCKAD deficiency. Patients with classic MSUD typically have enzyme activity less than 2%. They present with poor feeding, lethargy, irritability, maple syrup-like or burnt sugar smell and ketonuria in first week of life. Without treatment, they develop progressive neurological deterioration due to cerebral edema, culminating in coma, central respiratory failure and death. In patients with non-classical MSUD, residual enzyme activity varies from 2% to 30%, resulting in delayed clinical presentation to infancy or childhood as feeding problems, poor growth, developmental delay and behavioural problems. Non-classical patients may also experience severe metabolic intoxication and encephalopathy if sufficiently stressed by infectious illness, dehydration or prolonged fasting [2].

CASE REPORT

A newborn girl baby brought to the casualty department of Sree Balaji medical college &hospital with complaints of poor feeding, vomiting lethargy and generalized convulsions. The mother complains of abnormal odour in the baby's urine.

The baby was absolutely normal during thefirst week of postnatal life. From the8th day of postnatal period, the baby presented with all the above symptoms. After taking detailed history, it is noted that the parents had a consanguineous marriage. But no significant family history noted. Mother's obstetric history was normal. It is noted that the birth history of the baby was normal. On examination, we observed a burnt sugar odour in the baby's urine and also the baby girl presented with decreased muscle tone and neonatal reflexes.

Routine biochemical evaluation was done for serum electrolytes were within normal limits. Plasma samples were collected in heparin tube and urine samples was collected in a sterile container. Analysis was performed using High performance with TQ detector with a C18 column. A gradient elution was used for the separation; with mobile phase A (organic) comprising Acetonitrile with 1 mL/L Formic acid and 0.2 mL/L Hepatoflurobutyric acid ; and mobile phase B (aqueous)comprising water with 1 mL/L Formic acid

and 0.2 mL/L Hepatoflurobutyric acid at a flow rate of 400 mL/min. The injection volume was 5 microlitre. Standards were run in dilution (0, 250, 500 and 1000 mmol/L) of Val, Leu and Ile.. Controls were used for the reference. Test sample was run simultaneously. Chromotogram was obtained. Data analysis was done using SPSS software.

DISCUSSION

Serum electrolytes were within normal limits. Sodium 140 mEq/L (133-145 mEq/L), potassium 3.8 mEq/L (3.5-5.3 mEq/L), chloride100 mEq/L (98-106 mEq/L), calcium 8.3mg/dL (7.6-10.4 mg/dL), and magnesium levels 1.58 mg/dL (1.40-2.55 mg/dL) Chromatograph showed elevated levels of branched chain amino acids leucine 3,684, isoleucine 284, valine 426.

This baby was diagnosed with classical MSUD based on elevated BCAAs with the presence of all isoleucine in plasma and/or increased BCKAs in urine organic acids analysis.

CONCLUSION

MSUD is probably not uncommon in India especially among the South Asian subpopulation. The majority of patients has classical MSUD and presented with acute neurological symptoms within first week of life. Although we are able to diagnose and manage MSUD, we recognise that the clinical outcome remains to be optimised. We should aim towards earlier diagnosis through improving accessibility to diagnostic facilities, increasing awareness among physicians and general public and establishing a newborn screening programme.

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