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Case report on an infant presenting with hypoglycemia, and milky serum

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ABSTRACT

A 4-month-old male baby who presented in a moribund condition with seizures was found to have hepatomegaly, hypoglycemia and milky serum. Serum triglycerides were markedly elevated (3 168 mg/dL) with cholesterol being 257 mg/dL and high density lipoprotein levels were low (19 mg/dL). The possibility of glycogen storage disease type I was considered in the diagnosis. Infants with glycogen storage disease type I may present like sepsis. The association of hepatomegaly, hypoglycemia and abnormal lipid profile stated above should alert the physician to consider glycogen storage disease type I in the diagnosis.

1. Introduction

Metabolic diseases caused by genetic mutations resulting in enzyme deficiencies in an intermediary metabolic pathway constitute a wide spectrum of diseases in clinical practice. Children with inborn errors of metabolism may present with variety of signs and symptoms, including metabolic acidosis, persistent vomiting, failure to thrive, developmental delay, elevated or decreased blood and urine levels of a particular metabolite, a peculiar odor or physical signs such as hepatomegaly.

The inborn errors of metabolism that lead to hypoglycemia in children may be due to carbohydrate metabolism disorders, fatty acid oxidation disorders, hereditary fructose intolerance, glycogen storage diseases, galactosemia, organic acidemias and very rarely due to phosphoenolpyruvate carboxykinase deficiency and primary lactic acidosis.

There are various paediatric illnesses associated with milky serum and hypertriglyceridemia, including glycogen storage disease type I, Niemann-Pick disease, Tay-Sachs disease, familial hypertriglyceridemia, hypothyroidism, red cell pyruvate kinase deficiency[1] and biliary atresia[1]. Hemolytic anaemia is known to have milky serum in children. Transient lipoprotein lipase activity impairment with hypertriglyceridemia and low HDL has been reported in 2 infants[2]. Glycogen storage disease type I or von Gierke's disease, is the most common form of the glycogen storage diseases. The deficiency of glucose-6-phosphatase impairs the ability of the liver to produce free glucose from glycogen and from gluconeogenesis. It causes severe hypoglycemia and results in increased glycogen storage in liver and kidneys, causing enlargement of both. Other metabolic derangements seen in this disorder include lactic acidosis, hypertriglyceridemia and hyperurecemia[3-5].

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2. Case report

A 4-month-old male infant was referred with respiratory distress and seizures. As the infant had a respiratory arrest, he was shifted with the ET tube *in situ* with bag and tube intubation. Child was born out of non-consanguinous marriage with birth weight of 2 570 g. There was a history of male sibling death due to similar complaints on the day 13 of life. On admission GSC was 6/15 (E_1, V_1, M_4) and he was detected to have hypotension (blood pressure 60 mmHg systolic), edema, soft hepatomegaly, pupillary asymmetry with poor reaction to light.

A complete blood count showed leucopenia total count (4 800/cumm) with neutropenia (N 17%, absolute neutrophil count-816/cumm). Arterial blood gas showed severe metabolic acidosis (pH-6.91, PCO_2-17.3 mmHg, HCO_3^--5.7 mEq/L, BE-26.6 mmol/L). Biochemical investigations showed hypoglycemia (RBS-32 mg/dL) or (1.2 mmol/L), elevated liver enzymes (AST-1 676 U/L, ALT-630 U/L) with deranged renal function tests (urea-119 mg/dL, creatinine-1.5 mg/dL) and hyperuricemia (uric acid-20 mg/dL). The serum was grossly lipemic. Lipid profile estimation showed markedly increased triglyceride levels (3 196 mg/dL), cholesterol levels (257 mg/dL) with low high density lipoprotein (HDL) (19 mg/dL). With above presentation and results of blood investigations, possibility of glycogen storage disorder type I was entertained. Genetic study was not done in this case.

Hypoglycemia and hypotention were managed with dextrose boluses and ionotropic support (dopamine, dobutamine). Child was put on IV antibiotics with continuation of ventilator support. Despite above management, child continued to deteriorate and succumbed within 24 hours of admission. An informed signed consent for publication of any patient identifiable material, including photographs was obtained.

3. Discussion

Disorders of intermediary metabolism occur as a result of three basic mechanisms: an enzyme deficiency with defective substrate conversion, a membrane transport defect resulting in failure of absorption or excessive excretion of a compound, and defects in receptors involved in mediating metabolism. Defects in intermediary metabolism result in a disruption of normal metabolic process and often produce an elevated urinary excretion of an abnormal metabolite.

Glycogen storage disease I is caused by deficiency of glucose-6-phosphatase enzyme which is required for glucose production from either glycogen breakdown or gluconeogenesis. Apart from hypoglycemia, plasma triglyceride and cholesterol concentrations are usually increased in this disorder[6].

Recurrent hypoglycemia in glycogen storage disease type I promotes increased synthesis and release of lipids into the blood compartment as well as decreased lipid clearance from the blood has been reported. A study showed two patients of glycogen storage disease type I with mean cholesterol levels of (580, 418 mg/dL) and mean triglyceride levels of (1 612, 1 054 mg/dL), respectively, and 40-fold increase in *de novo* lipogenesis in glycogen storage disease I patients compared to controls[7-9]. This case presentation was unique in the sense that it had thick milky serum which has provoked us to check lipid, and uric acid levles which showed marked hypertriglyceredemia, hyperuricemia with elevated serum transminases.

Thus even though there are various causes of milky serum in infants, a strong suspicion of glycogen storage disorder should be kept in mind whenever infants present with hepatomegaly, hypoglycemia and severe hypertriglyceridemia.

Conflict of interest statement

We declare that we have no conflict of interest.

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