Galactosemia with Rubella Infection

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Galactosemia is a well described inborn error of carbohydrate metabolism. The deficiency of enzymes Galactose-1-uridyl transferase, Galactokinase or Uridyl diphosgalactose-4-epimerase alters the normal galactose metabolism (l). The resultant hypoglycemia and accumulation of abnormal metabolites are responsible for the clinical manifestations of this metabolic disorder. The disease is believed to commence *in-utero (2,3)* and the clinical features are expressed by early infancy. If untreated, the disease is fatal. The clinical features of cataract, hepatosplenomegaly and jaundice are similar to the manifestations of intrauterine infections. We are presenting a case of galactosemia with evidence of congenital rubella infection. To the best of our knowledge such a presentation has not been reported earlier.

Case Report

A 22-day-old male infant weighing 3420 g was admitted with complaints of persistent jaundice, progressive abdominal distension and lethargy. It was the third infant born of non-consanguineous marriage. His previous two siblings had died in the neonatal period. This baby who was delivered normally and was put on breastfeeds developed jaundice on day 3. The jaundice progressively deepened, accompanied by abdominal distension and lethargy.

He was also noticed to hepatosplenomegaly, ascites and bilateral cataracts. Investigations revealed hypoglycemia, raised transaminases, hyperbilirubinemia, prolonged prothrombin time and prolonged APTT. Other investigations revealed serum total protein and albumin concentrations of 4.1 g/dl and 2.6 g/dl, respectively and an elevated blood ammonia level of 145 mg/dl. The ascitic tap revealed a transudate. Renal functions and blood counts were normal. Galactosemia was confirmed on the basis of urine being positive for reducing subtances but negative for glucose, and urine chromatography demonstrating the presence of galactose. Incidentally, both baby and mother tested positive for anti-Rubella IgM antibodies. Echocardiography was normal. The blood culture grew Klebsiella species.

The infant was treated with anti-hepatic failure measures, intravenous antimicrobials, diuretics, Vitamin K and lactose and galactose free formula. Subsequently, his activity improved. He remained euglycemic, showed a decrease in jaundice and improvement of liver functions. Though the ascites persisted, he was discharged at request after 2 weeks of therapy.

Discussion

The incidence of classical galactosemia has been reported to be 1:60,000(1). The primary diagnosis is often made on the basis of the clinical presentation and the detection of non glucose reducing substances in the urine. Variability in dietary intake could influence the detection of urinary galactose(3,4). Therefore, enzyme estimation

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becomes mandatory for the early diagnosis of classical galactosemia.

The presence of ascites, low serum proteins, elevated liver enzymes and prolonged prothrombin time reflects significant and chronic hepatocellular dysfunction. Hsia and coworkers (3) have attributed the uncommon presentation of ascites to poor dietary intake. The clinical features of hepatosplenomegaly, jaundice and cataract are also common presentations of congenital rubella infection. In the present case, both the baby and the mother had repeatedly tested positive for rubella IGM antibody. It is uncommon for rubella IgM antibody to persist over 6 weeks after infection in adults (5). It is, therefore, inferred that the infant had been infected beyond the first trimester, at a period too late for the teratogenic effects of the virus to manifest. The normal birthweight of the infant also supports this inference. However, the possibility of rubella infection contributing to the severity of the liver disease in a galactosemic infant, can not be ruled out.

Despite being a well described entity, galactosemia, a treatable metabolic disorder continues to be misdiagnosed as septicemia. The well described association of Gram negative infections in galactosemics(4,6), further complicates the differentiation. The absence of screening facilities for inborn errors of metabolism further enhances this difficulty. The association of rubella makes us feel that again another better recognized disease has crept up to encourage the misdiagnosis of a treatable inborn error of metabolism. Early and strict dietary restriction of lactose in the diet can result in the reversal of most clinical featurs of galactosemia. Optimally, homozygous mothers must be placed on galactose free diet during pregnancy (6). Hsia had observed (3) that the degree of restriction rather than the age of commencement of dietary restriction had a greater influence on the IQ of the patient.

The pleotropism of galactosemia, its resemblance to intrauterine infections and association with septicemia increases the difficulty in diagnosing this disease. The task is even more daunting for clinicians in a developing country where investigative facilities like enzymes assays are limited. The positive response to early and strict dietary modification, however makes it mandatory to screen for galactosemia despite the availability of a more familiar diagnosis.

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