Abstract:

We report the case of a full term female newborn who had prolonged jaundice with urosepsis due to *Escherichia coli*. Her neonatal screening test was suggestive of galactosemia. Her abnormal test result alerted us which enabled us to prevent later complications of her metabolic disease especially neurological impairment by only milk modification.

Key words: Galactosemia, Jaundice, Metabolic Diseases, Milk, Neonatal Screening.

Introduction

Prolonged neonatal jaundice always warrant special attention to rule out secondary causes particularly urinary tract infection. Our aim was to first treat the urosepsis which was taught as original cause of prolonged neonatal jaundice. We ended up discovering a more important cause of both the jaundice and the sepsis that changed her prognosis.

Case Report

FH, a twenty day of life female newborn was born at 40 week of gestational age, by normal vaginal delivery, from a primiparous 23 year old lady with first degree consanguinity. She was exclusive breast feed child with birth weight of 4350 grams with smooth antenatal course. Her postpartum course was significant for prolonged jaundice, decreased oral intake and recurrent vomiting especially after the first week, giving her no weight gain. Baseline investigations revealed a total bilirubin of 12.7 mg/dL with direct bilirubin of 5.8 mg/dL and presence of leukocyturia, so she was referred as inpatient for management and further evaluation in the neonatal intensive care unit (NICU).

In NICU, she was found to be icteric, hypotonic and her weight was 4400 grams. She had flat anterior fontanel and no abdominal masses, no anemia, normal transaminase level, normal ammonia and lactate blood level. Her INR was 1.3, TSH normal and CRP was increased to 16 mg/dL. She was started on intravenous antibiotic and hydration therapy but a special call from the laboratory department alerted us about her abnormal neonatal screening test results that showed a galactose-1-phosphate uridyltransferase (GALT) enzyme below 1.2 (normal value > 1.5 U/g Hb). Low levels of GALT enzyme levels led...
to diagnosis of galactosemia. She was started on soy formula with quick improvement of her clinical conditions. Eyes examination was normal excluding the possibility of cataract. On serial clinical follow up, she has normal psychomotor and developmental milestones with good weight gain according to her age.

Discussion

This case emphasizes the importance of broadening the differential diagnosis in case of prolonged neonatal jaundice. Persistent jaundice in the neonate is defined as jaundice that lasts longer than 14 to 21 days [1]. It can occur in up to 15% of all newborns [2]. The difficult task facing primary care providers is discriminating between serious conjugated hyperbilirubinemia and benign unconjugated jaundice because in the early stage, the infants can look very well except for their jaundice. In our case, the baby was hospitalized for conjugated hyperbilirubinemia with urinary tract infection as the primary diagnosis but luckily, the early laboratory results of her neonatal screening test was available on the second day of her admission and revealed that she has low GALT enzyme level leading to diagnosis of classic galactosemia.

Galactosemia is an inherited metabolic disorder, due to the deficiency of the enzyme galactose-1-phosphate uridylyltransferase and with an estimated prevalence that ranges between 1 in 30,000 and 1 in 60,000 births [3]. Galactosemia due to galactose-1-uridylyl phosphate uridylyltransferase deficiency commonly causes hepatic dysfunction, but may not cause marked hypoglycemia. Its diagnosis in the newborn period is critical because of the associated liver and renal dysfunction, cerebral edema, cataract and the risk of gram-negative sepsis. When suspected, all intake of galactose (human milk and cow milk formulas) must cease. The diagnosis can be suspected on the basis of positive reducing substances in the urine and confirmed by metabolite or enzyme assay. Urine obtained more than 1 day after cessation of galactose intake may be negative for reducing substances [4].

Here, we should emphasize the importance of performing the universal neonatal screening tests for all neonates, to detect galactosemia and other life threatening metabolic diseases as early as possible and to be treated adequately as reported in our case which required only lactose free formula modification.

Conclusion

Prolonged neonatal jaundice could be a manifestation of a serious underlying disease especially if it is conjugated hyperbilirubinemia. The differential diagnosis must consider the possibility of an inborn error of metabolism. The association of conjugated hyperbilirubinemia with poor feeding and gram negative urosepsis in neonates should alert the physician to the diagnosis of classic galactosemia.

References

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