Olgu Sunumu
Case Report

Galactosemia: Not To Be Missed Cause of Neonatal Jaundice

GALAKTOZEMİ: ATLANMAMASI GEREKEN NEONATAL SARILIK NEDENİ

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ABSTRACT

A case representing neonatal jaundice that includes direct and indirect hyperbilirubinemia was diagnosed with classical galactosemia. Even though she was initially approached as a neonatal sepsis case, after the cessation of her milk consumption, jaundice resolved, and the diagnosis was made and supported by additional diagnostic tools. It has been shared to emphasize the importance of assessment and urgent management of the disease during the neonatal period.

Keywords: Neonatal Jaundice, Galactosemia, Milk

ÖZ

Sarılık nedeniyle başvuran direk ve indirek hiperbilirubinemi saptanan yenidoğan olgusunda klasik galaktozemi tanısı konulmuş ve genetik mutasyon sonucuyla tanı doğrulanmıştır. Bu olgu, yenidoğan döneminde saptanan sarılığın galaktozemi açısından acil değerlendirilmesinin ve tedavisinin önemini vurgulamak için paylaşılmıştır.

Anahtar Kelimeler: Galaktozemi, neonatal sarılık, anne sütü

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CASE REPORT

A female infant, born at the 38th week of gestation, 2400 grams, presented with jaundice and slight hemorrhage of the umbilicus at thirteen days of age. She had been fed with breastmilk exclusively and showed no signs of suboptimal feeding. Even though her jaundice that reached from top to toe was apparent, the physical examination revealed no abnormal vital signs, hepatosplenomegaly or neurologic signs referring to bilirubin encephalopathy.

She has healthy parents that have a consanguineous marriage. In addition, she has a healthy seven-year-old sister and a deceased brother due to hydrocephaly at 2,5 years of age. On presentation, her total bilirubin was 22 mg/dl, direct bilirubin 8,6 mg/dl, alanine transaminase 66

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U/L, aspartate transaminase 149 U/L, gamma glutamyl transferase 33 U/L. However, her stool had no discoloration; likewise, her prothrombin time and active partial thromboplastin time values were normal.

Since her total bilirubin level had exceeded the phototherapy limit, she received therapy for two days. Also, her blood, cerebrospinal fluid, and urine samples were collected for microbiological work. Due to the lack of equipment, the reducing agent in urine could not be checked, but her urine sample showed no signs of urinary tract infection. She was given ampicillin and gentamicin treatment until the exclusion of late neonatal sepsis. Moreover, her breastfeeding was cut in case of possible galactosemia, and she started feeding with the galactose-free formula. On follow, umbilical hemorrhage did not persist; total and direct bilirubin levels decreased to 5,6 mg/dl and 3,8 mg/dl, respectively.

During the hospitalization, possible etiologies of cholestasis and indirect hyperbilirubinemia were investigated. No immune or non-immune hemolysis was detected by the direct-anti-globulin test, peripherally smear test, or reticulocyte count. Also, possible galactose-6-phosphate dehydrogenase deficiency, TORCH infections, thyroid function disorders and biliary atresia were ruled out.

Her national screening test for phenylketonuria had been detected positively. Thus, her serum phenylalanine level was checked and resulted as usual. A blood sample to detect galactose-1-phosphate uridyltransferase (GALT) activity using the Beutler enzyme spot test was sent to rule out classic galactosemia, and no enzyme activities were detected. Therefore, she was diagnosed with classical galactosemia. In addition, her genetic tests were ordered to support the diagnosis and revealed a pathogenic homozygote c.425T>A (pMet142Lys) mutation in the GALT gene (1).

DISCUSSION

Galactosemia is an autosomal recessive inherited disorder of galactose metabolism. The incidence is 1/60000 per birth in the USA. The anticipated incidence of the disease in Turkey is more than that, approximately 1/10000 per birth. However, a cohort that presents the rate has not been reported yet (2).

Three different galactosemia types have been shown to date. Classical galactosemia (type 1) is caused by a deficiency of the galactose-1-phosphateuridyltransferase enzyme, which provides galactose-1-phosphate metabolism (3). The enzyme deficiency could arise due to several mutations detected in the GALT gene (4).

Although consuming mother's milk by breastfeeding is extremely important for infants' development, in this case, its imminent cutting down on a diet is vital to protect these infants from deterioration as the milk contains galactose. Otherwise, it would be inevitable for these infants to develop severe clinical symptoms, such as lethargy, prolonged jaundice, hypoglycemia, cataract, sepsis (primarily due to Escherichia coli), Fanconi syndrome, infertility and acute hepatic failure, which can progress to hepatocellular carcinoma.

There is no accuracy in observing the whole clinical signs together, but each requires special attention. In the study assessing 22 infants diagnosed with galactosemia during the period, 86% of the participants presented with jaundice that included indirect and direct hyperbilirubinemia (5).

The last published galactosemia case had presented with indirect hyperbilirubinemia that resolved after commencing lactose-free formula (6). The mean diagnosis time of these infants presented with jaundice was found at 2-3 weeks of life. Also, abnormal coagulation test results were found to be accompanied by jaundice. Although our case had an additional complaint of umbilical hemorrhage, no coagulation abnormality was found (7).

Since galactosemia is one of the metabolic diseases that needs cutting down on own mother's milk, it brings out the importance of the screening. Luckily, neonatal screening for galactosemia has already been applied in some developed countries successfully. This detects many cases at an early stage. Even though most countries do not have such an opportunity, physicians need to acknowledge the clinical signs of galactosemia to plan the management timely. Therefore, this report has been made to emphasize awareness.

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