УДК [61+615.1](06)(043.2)
ББК 5+52.81
А 43
ISBN 978-985-21-1569-8

Sehajmeet S., Satyam P.

GALACTOSEMIA IN NEWBORNS. A CASE OF CLASSIC GALACTOSEMIA

Tutors: associate professor Uvarova E.V.,
senior lecturer Pivchenko T.P.
1st Department of Childhood Diseases
Belarusian State Medical University, Minsk

Relevance. Galactosemia is an autosomal recessive disease, a disorder of galactose metabolism. The prognosis of the disease largely depends on early diagnosis and timely pathogenetic therapy.

Aim: improve knowledge of differential diagnosis of hyperbilirubinemia using the example of a clinical case of galactosemia type 1 in a newborn.

Materials and methods. The newborn was treated in the Practical Center "Mother and Child" from the 8th to the 47th day of life. Analysis of medical record documents of patient and an analysis of literary data on the pathology under study has been carried out.

Results and discussion. In a clinical case we will talk about child (boy) was born from 2 pregnancies, 2 urgent births at a period of 284 days. Apgar score 8/9. Weight at birth was 3700 g, height - 54 cm. This pregnancy occurred against the background of polyhydramnios and mild anemia. Mother's age is 31 years. Blood type A(II) Rhesus D positive. The child was breastfed from the birth. From the 3rd day of life, the appearance of jaundice was noted, which was regarded as physiological. On the 4th day of life, the icterus of the skin increased, the level of total bilirubin was 362.8 μmol/l. Phototherapy and infusion therapy were carried out for the purpose of detoxification. On day 5, during treatment, the previous level of total bilirubin remained at 365.8 μmol/l (direct bilirubin level – 25.9 μmol/l). During the early neonatal period, the clinical picture was severe due to increasing hepatic failure. A newborn boy was admitted to the “Mother and Child” Practical Center on the 8th day of life. The maximum level of total bilirubin was 551 μmol/l. The examination of the patient began with the exclusion of the most likely diseases. After consultation with a geneticist, to exclude hereditary metabolic diseases, the child underwent selective blood screening. According to the results of screening, an increase in the level of galactose + galactose-1-phosphate was detected in the blood (3256 μmol/l when the norm is up to 250), and a positive test in the urine for reducing sugars and mucopolysaccharidosis. Results of molecular genetic diagnostics: the proband was found to be a homozygous carrier of the p.Q188R mutation in exon 6 of the GALT gene. Diagnosis: Hereditary metabolic disease. Classic galactosemia type 1. Autosomal recessive type of inheritance. Timely cancellation of enteral nutrition containing galactose and lactose made it possible to stabilize the child’s condition and avoid irreversible damage to organs and systems.

Conclusions. Neonatal screening is especially needed to detect classic galactosemia. Early diagnosis and treatment in galactosemia lead to a higher chance of a better prognosis, whereas infants and children that are left untreated suffer from major health consequences, such as intellectual disability, behavioral issues, poor growth, deficient development, nutritional deficiencies.