Medical science of Uzbekistan
published: 13 March 2025
doi.org/10.56121/2181-3612-2025-2-51-53
Received: 10 February 2025
Accepted: 18 February 2025
Published: 13 March 2025

Article/Review

GALACTOSEMIA IN NEWBORNS

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Abstract.

Relevance. Galactosemia is a rare inherited disease that occurs as a result of a genetic mutation blocking the enzymes involved in galactose metabolism. Clinical manifestations are directly related to the accumulation of intermediate metabolites in the body and impaired function of internal organ cells. **The aims and objectives of the study:** to study the frequency and clinical features of galactosemia in children. **Materials and methods of the study.** To solve the set tasks, we conducted a study of clinical, anamnestic and laboratory and instrumental examination parameters in children with galactosemia. **Research results.** The study revealed that the frequency of galactosemia health of the mother, her obstetric and gynecological history, the peculiarities of the course of pregnancy and childbirth do not play a significant role in the development of galactosemia in newborns. **Conclusion.** We have determined that the age, health of the mother, her obstetric and gynecological history, the peculiarities of the course of pregnancy and childbirth do not play a significant role in the development of galactosemia in newborns.

Key words: galactosemia, newborns, clinical features, complications.

Introduction

Various metabolic disorders are observed quite often in childhood. One of them is galactosemia - a pathology in which carbohydrates from food are not absorbed. The disease is based on mutations of genes involved in the metabolism of galactose - a monosaccharide included in lactose. In this case, the conversion of galactose into glucose is blocked.

Depending on the enzyme block, there are 3 types of galactosemia: type I - a defect of galactose-1-phosphate uridyltransferase (GALT), type II - a defect of galactokinase (GALC), type III - anomaly of UDP-galactose-4-epimerase (GALE). Hypergalactosemia, accumulation of galactitol and galactose-1-phosphate in the body lead to the development of intracellular edema, dysfunction of red blood cells, liver cells, brain and other organs

The aims and objectives of the study: to study the frequency and clinical features of galactosemia in children.

Material and methods

To solve the set tasks, we conducted a study of clinical, anamnestic and laboratory and instrumental examination parameters in children with galactosemia.

Results and discussion.

In the study, according to the literature, galactosemia occurs equally often in both boys and girls [1,5]. Among the patients in our study group, boys predominated 7 (71,2%). All the studied patients were born full-term, without signs of asphyxia at a gestational age of 37-39 weeks, with normal parameters of physical development. Average indicators: body weight – 3,295±181,61 g, height – 51,8±1,44 cm, head circumference – 33,6±0,51 cm, chest circumference – 33,5±0,21 cm.

When studying the pre- and perinatal history of children with galactosemia, it was determined that the average age of mothers was 20,5±28,5 years; of these, 6 (45,5%) women were somatically healthy; 4 (30,3%) had chronic pathology of the urinary and cardiovascular systems and two women suffered from immune-inflammatory diseases. Sick newborns were more often (4; 66,6%) born to women who were pregnant again and giving birth again with a complicated gynecological and obstetric history (abortion - 3; stillbirth - 1; colpitis, adnexitis - 6, myoma - 1 case). Complicated family history of genetic pathology was not found.

Pathological pregnancy was detected in 4 (60%) of the eleven examined mothers: intrauterine infection (IUI) - in 4, preeclampsia - in 2, threatened miscarriage, chronic fetoplacental insufficiency (CFPI) and isthmic-cervical insufficiency (ICI) - in 2.

Thus, we were unable to identify any characteristic features of the pre- and perinatal history in children with hypogalactia.

All newborns were immediately put to the mother's breast after birth and were breastfed until the diagnosis of galactosemia was established): up to 9-16 days - 4 children, up to 32 days and up to 4 months - 7 cases each. In the study, in all sick children, the GAO indicators in the blood taken on the 4th day of life were significantly higher than the normal values and ranged from 13,4 to 106,7 ng / ml (58,7±4,05). When repeating the GAO study (retest) against the background of ongoing breastfeeding, an increase in this indicator in all patients was noted by an average of 12,84±7,6% (1,9-31,3%) and the manifestation of clinical manifestations of galactosemia. The most frequent and first symptoms of galactosemia, appearing in newborns in the first days after milk intake, are vomiting (37%) and diarrhea, early developing cataracts are typical (42%). In addition, hepatomegaly (90%), jaundice (78%), anorexia and weight loss (54%), abdominal distension (20%), lethargy (16%), ascites (14%), splenomegaly, dark urine, pallor (7%), hemorrhagic syndrome (5%), edema, cholelithiasis, acholic stool, dysuria (2%) are typical.

The basis of pathogenetic therapy of galactosemia is lifelong diet therapy with lactose-free products for newborns - the use of therapeutic lactose-free breast milk substitutes. This normalizes galactose levels in the blood and relieves acute symptoms of the disease. Without treatment, death is possible from sepsis and multiple organ failure during the first days of life, before the diagnosis of galactosemia is established [1,3,4]. If the diet is not followed, residual damage to the central nervous system (CNS) develops: delayed psychomotor development, impaired motor skills. Cataracts and blindness may develop, and the function of the sex glands may be impaired. With late diagnosis and late initiation of diet therapy, severe and irreversible damage to the liver and brain is possible [2].

Conclusion

Thus, the data we obtained on the clinical course of galactosemia are fully consistent with the literature data. We have determined that the age, health of the mother, her obstetric and gynecological history, the peculiarities of the course of pregnancy and childbirth do not play a significant role in the development of galactosemia in newborns. All sick infants were born on time, in satisfactory condition, with optimal indicators of physical development. In all the children studied, against the background of breastfeeding during the first week - first months of life, increasing dyspeptic disorders were noted (vomiting, regurgitation, refusal of the mother's breast with the formation of postnatal hypotrophy), as well as varying degrees of severity of signs of depression of the central nervous system (hypotension, hyporeflexia), then - a delay in neuropsychic and speech development. Jaundice syndrome with clinical, laboratory and instrumental signs of liver damage was recorded much less often. The most severe and rare manifestation of galactosemia in newborns is sepsis with an unfavorable outcome due to the development of multiple organ failure.

List of references

[1] Welling L., Boelen A., Derks T.G., Schielen P.C., de Vries M., Williams M., et al. Nine years of newborn screening for classical galactosemia in the Netherlands: Effectiveness of screening methods, and identification of patients with previously unreported phenotypes. Mol Genet Metab. 2017 Mar; 120 (3): 223–228.

[2] Shaw K.A., Mulle J.G., Epstein M.P., Fridovich-Keil J.L. Gastrointestinal health in classic galactosemia. JIMD Rep. 2017; 33: 27–32.

[3] Frederick A.B., Zinsli A.M., Carlock G., Conneely K., Fridovich-Keil J.L. Presentation, progression, and predictors of ovarian insufficiency in classic galactosemia. J Inherit Metab Dis. 2018 Sep; 41 (5): 785–790.

[4] Walter J.H. Galactosemia. Valle D., Beaudet A.L., Vogelstein B., Kinzler K.W., Antonarakis S.E., Ballabio A., Gibson K. Ed. The Online metabolic and molecular bases of inherited disease. New York, NY: McGraw-Hill; 2014.

[5] Kisljuk G.I., Kononenko V.V., Kubanskij E.K., Vjalyh E.K. Neonatal'nyj skrining, kak metod jekspress-diagnostiki nasledstvennoj patologii u novorozhdennyh detej. Innovacii v medicine. Materialy VII mezhd. nauch. konf., posvjashhennoj 82-letiju Kurskogo Gosudarstvennogo Medicinskogo Universiteta. 2016; 56–59.