GALACTOSEMIA DISEASE HAIDA SYMPTOMS AND PROPHYLAXIS

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ABSTRACT
In this article, we will consider galactosemia, its causes, symptoms, treatment and preventive measures.

The purpose of the work is to study galactosemia in depth, find treatment measures and apply it in practice.

Carbohydrates that enter the small intestine are digested and absorbed into the blood in the form of monosaccharides (glucose, fructose, galactose). Monosaccharides are transported in the liver in the bloodstream through the portal vein. Hepatocitlat converts fructose and galactose into glucose, and glucose is then transformed into glycogen or released into the blood when necessary.

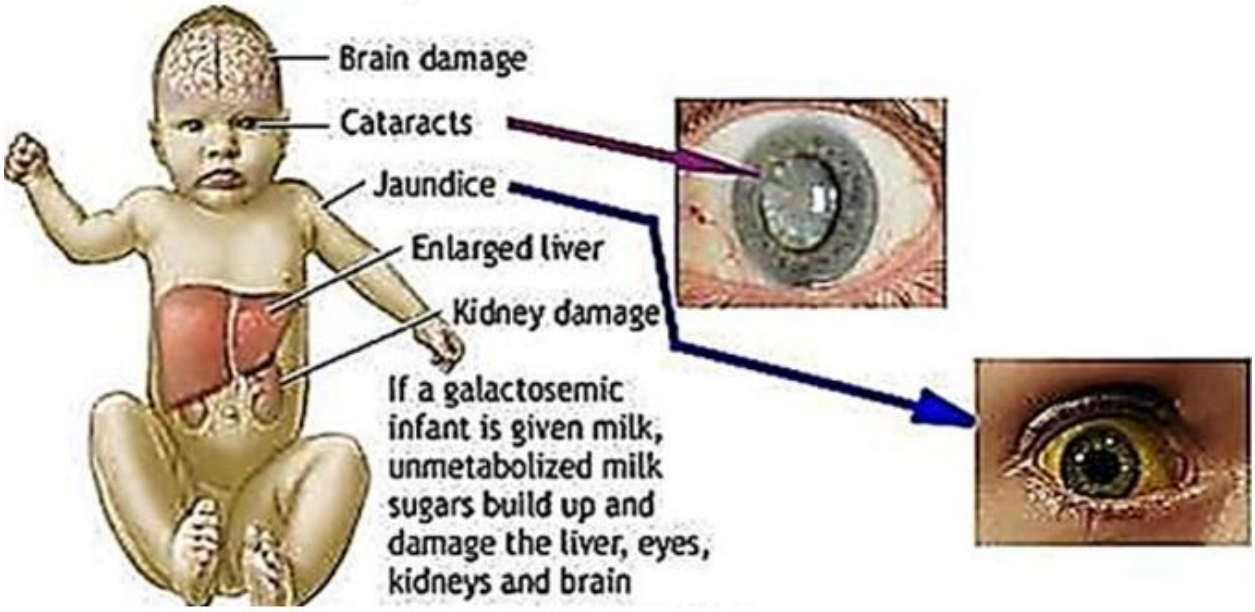
Thus, only one carbohydrate - glucose - enters the bloodstream from hepatocytes. The process of conversion of galactose to glucose, and the conversion of glucose to glycogen, is carried out biochemically in several stages: phosphorylation of galactose to galactose-1-phosphate, its transformation into uridinediphosphogalactose (UDF-galactose), and then uridinediphosphoglucose. Galactosemia is a hereditary disease based on a metabolic block in the conversion of galactose to glucose. It is a rare genetic disorder that is inherited in an autosomal recessive manner. Galactosemia is caused by the lack of an enzyme responsible for the absorption of galactose by the body.

After food enters the intestine, the galactose contained in it is transferred to the liver, where it must be metabolized. Due to the liver's innate inability to transform galactose into glucose, the process is disrupted, and the substance is distributed throughout the body with the blood stream, and in this an increase in the concentration of galactose in the blood is shown, and the excretion of galactose in the urine (galactosuria) occurs. Thus, with the presence of galactose in the blood, it is characterized by a dislike for milk (milk lactose is an alimentary source of galactose). Cell damage with liver dystrophy and hepatomegaly - brain dystrophy with mental disorder, oxygen deficiency in body tissues, kidney, eye dysfunction are noted. For the first time, galactosemia was described in 1917, but at that time scientists and doctors did not yet

understand the true causes of this pathology. Almost 40 years later, in 1956, a group of researchers led by scientist Hermann Kelker discovered that galactosemia develops as a result of a violation of galactose metabolism.

Currently, galactosemia can occur in 1 in 60,000 newborns. It is worth noting that, unlike a number of other genetic diseases, the frequency of occurrence of galactosemia in the world is not the same.

Symptoms of galactosemia are shown in Figure 1



Many clinical signs indicate multiple tissue damage. Symptoms of galactosemia manifest themselves immediately from the first days of life.

1. The first symptom of galactosemia is a disorder of the gastrointestinal tract.
2. On the part of the central nervous system, along with visual impairment, there is a delay in psychomotor development, which becomes noticeable some time after the onset of the disease.
3. Changes in the size of the liver

One of the main symptoms that we pay attention to when diagnosing this disease is jaundice, gastrointestinal diseases, liver enlargement and hemorrhagic syndromes appear in newborns from the age of 4-5 days.

Depending on the severity of the disease, the following types of the disease are distinguished:

Lightweight form

If galactosemia is mild, newborns vomit after drinking milk, babies refuse the breast. Frequent watery diarrhea and flatulence also develop. A blood test reveals hypoglycemia (lack of glucose).

Medium heavy form

The mild form is accompanied by the following clinical signs: liver enlargement, severe jaundice. He lags behind in mental development. Anemia is noted in the blood analysis.

Heavy form

Children experience severe vomiting and loss of appetite after eating milky food. In newborns, the disease is manifested by hyperexcitability, watery diarrhea and gag reflexes. Usually, these babies weigh more than 5 kg at birth.

After 1-2 months, kidney failure develops, the lens of the eye is affected, signs of liver damage are added to the following symptoms: the size increases, cirrhosis develops. Also, the work of the brain is disturbed, ascites may appear. Treatment should be started in time, because the severe level can end in death due to the rapidly growing failure of vital organs.

Prevention

• Screening tests for the development of pathology among all newborns.
• Early detection of galactosemia (prenatal studies).
• If you suspect this disease, you should immediately switch the child to a milk-free diet.
• Minimizing the consumption of dairy products by women during pregnancy if there is a risk of developing galactosemia.

Treatment of galactosemia

Treatment is very long and is carried out by therapists and pediatricians in an outpatient setting.

Treatment is based on a lactose-free diet. Today, a wide range of special nutritional formulas can be purchased without milk sugar. As the child grows, the diet expands. Introducing new foods into the diet is done very carefully. A lactose-restricted diet is observed until the child is five years old, after which the enzyme malfunction can be partially compensated. If galactosemia in a newborn is a severe form, the diet is followed for life. At the same time, almost all foods containing starch and lactose are removed from the diet. In this case, fructose-containing products become an alternative source of carbohydrates. Medical treatment of galactosemia, as a rule, is symptomatic, aimed at improving metabolic processes in the body, as well as eliminating disorders in the functioning of damaged organs. For this purpose, drugs that improve metabolism are prescribed. With galactosemia, it is forbidden to take any medication without consulting a doctor.

Conclusion: So, if we conclude from this article, galactosemia is considered a genetic disorder and is passed from father or mother to child. There is currently no specific treatment for this condition. We must control the disease through diet. That is, the food we eat should not contain starch and lactose. Of course, you should not try to treat this condition on your own. In consultation with our pediatricians and therapists, it will be appropriate to consume drugs or food products that improve metabolism.

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