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GALACTOSEMIA MKB 10: E74

Abstract: In this article, we will consider galactosemia, a disease associated with impaired enzyme activity. Who identified galaztosemia. Let's look at the origin of the disease, its symptoms, heredity, prevention and diagnosis. It talks about the methods of treatment of the disease and prevention of the disease.

Keywords: Galactosemia, MKB 10, enzyme, chromosome, amino acid, autosomal chromosome, mutation, recessive, toxic poisoning, lactose, galactose, glucose, galactose-1 phosphaturidyltransferase, cataract, amniotic fluid, GALK, GALK1, osteoporosis, Diet, Mendelian inheritance, Liver cirrhosis, jaundice, hypotension, lethargy, hypogonadism, diarrhea, homozygous, heterozygous.

Introduction: Galactosemia (English galactosemia, Greek galactose + aima, galactose + blood, accumulation of galactose in the blood) is a rare genetic metabolic disease that affects a person's ability to properly metabolize the sugar galactose. For the first time information about galactesemia was discovered by the German doctor Friedrich Goppert (1870-1927) in 1917.

The incidence of the disease is from 1:20,000 to 1:120,000 in newborns. This disease is inherited in an autosomal recessive state. The inheritance of this disease is based on Mendelian laws. Reasons for its origin: in the normal metabolic process of the body, under the influence of lactose enzymes contained in food products, glucose is broken down into galactose. In galactosemia, the enzyme galactose 1-phosphaturidyltransferase, which carries out this process, is insufficiently produced or is not produced at all due to a mutation of the gene responsible for the production of this enzyme.

As a result, galactose 1-phosphate, which is toxic for the body, begins to accumulate in various tissues, and it causes the enlargement of the cataract, the development of cataracts, and similar pathological processes.

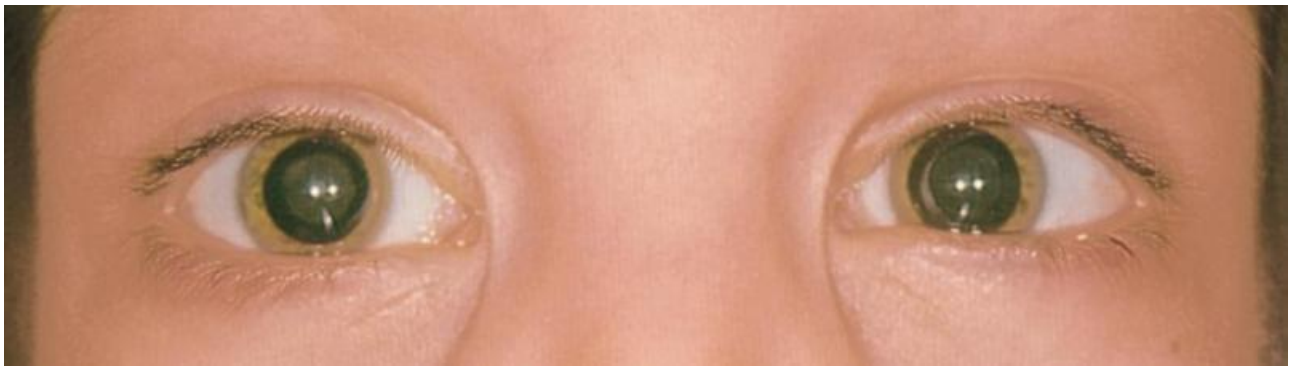
Symptoms: Babies may appear healthy at birth, but after a few days, after being breastfed or formula fed, children begin to develop life-threatening symptoms.

For example:

- ✓ Vomiting and diarrhea.
- ✓ Poor nutrition and weight gain.
- ✓ Hepatocellular damage.
- ✓ Lethargy, hypotonia.

The development of this acute neonatal toxicity syndrome can lead to the development of sepsis, cataracts and even brain pseudotumor.

Galactosemia in children can lead to the development of cataracts if left untreated.

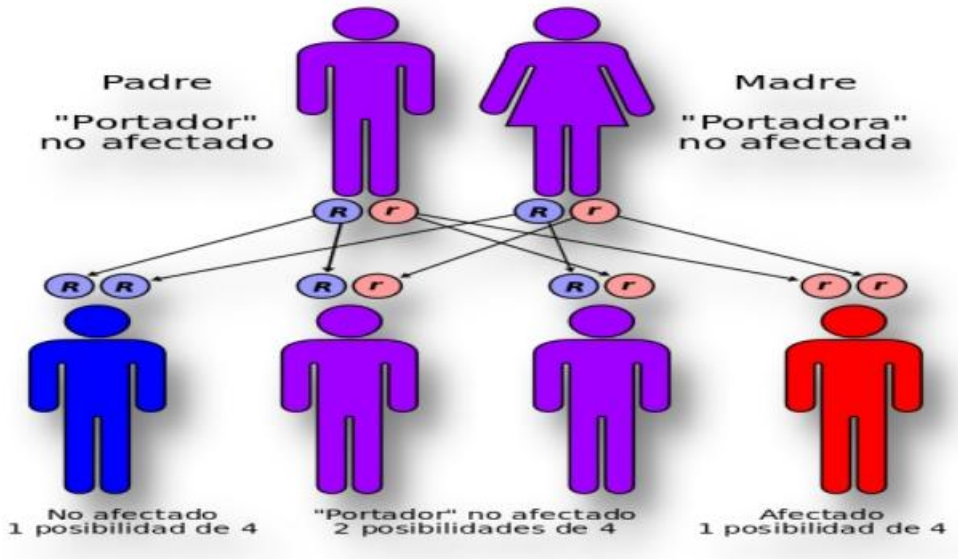


can cause speech difficulties, kidney disease, liver failure, sepsis, and ovarian failure.

Inheritance of the disease: Inheritance is an autosomal recessive disease (aa). If it is in the (AA) state, this gene is not affected by this disease, if it is in the (Aa) state, this gene is a carrier, but it does not show symptoms of the disease. But if two carrier genes form a family, their children have a 25% chance of having sick children.

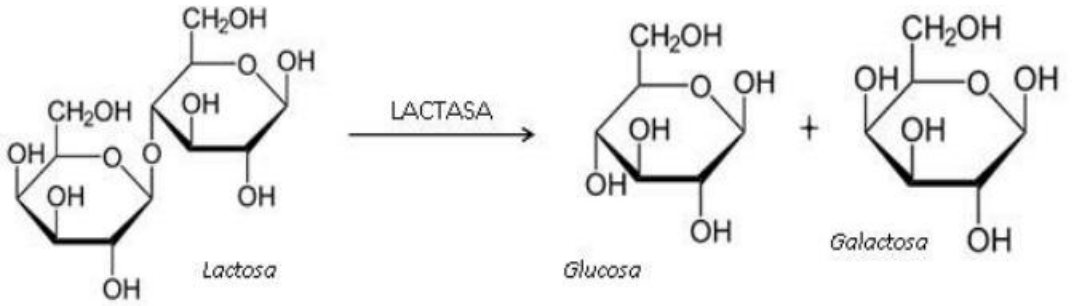
- ✓ aa× Aa: 50% of children will be homozygous for the mutation (i.e. affected), and the remaining 50% will be carriers.
- ✓ aa × AA: all her offspring will be carriers, but none of them will develop the disease.
- ✓ Aa × AA: 50% of children will be carriers, but none will show the disease.

It should be noted that cases of pregnancy of sick women are very rare, because they suffer from ovarian atrophy and hypogonadism.

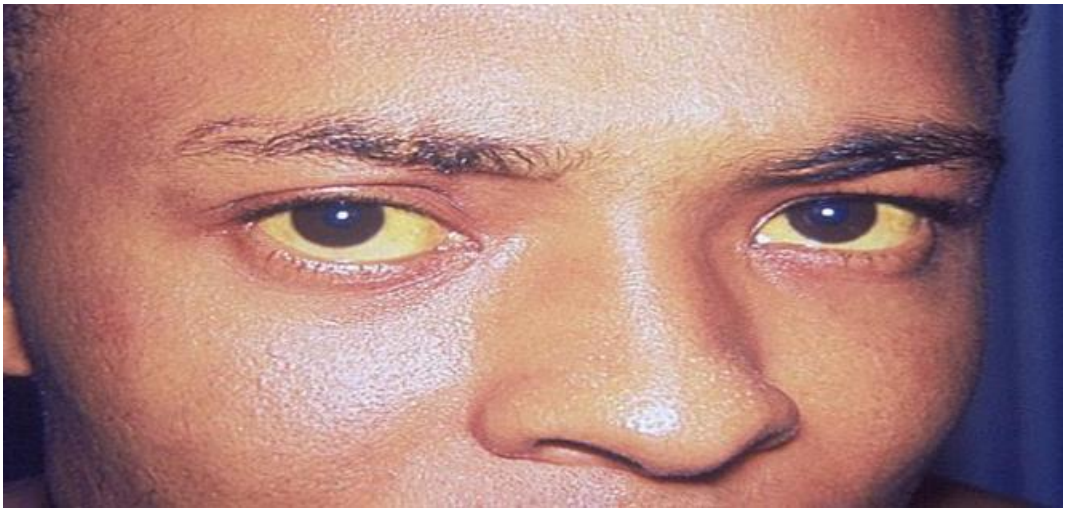


Causes of galactosemia: In the process of lactose digestion, the lactase enzyme breaks down the molecule into glucose and galactose. People with galactosemia have a very low level or complete absence of enzymes necessary for the further metabolism of galactose, which leads to the accumulation of galactose 1-phosphate in various tissues. This accumulation produces toxic levels

of galactose, which can lead to hepatomegaly (pathological enlargement of the liver), cirrhosis, kidney failure, cataracts, brain damage, and ovarian dysfunction in women, as in the classic form of the disease. Treatment of this disease is very important, because without treatment, mortality in children with galactosemia is 75%. The treatment of this disease will be discussed later.



Here is the breakdown reaction of lactose.



Diagnosis of galactosemia: Babies are routinely tested for galactosemia and diagnosed with the disease. In children with galactosemia, it is mainly manifested by jaundice.

In addition, diarrhea is observed in large quantities. It is difficult to make a final diagnosis because these processes are not considered to be the processes characteristic of galactosemia.

To check for galactosemia, blood is taken from the heel of babies or their urine is tested. It looks for three enzymes needed to convert galactose into glucose in the blood. Glucose is the sugar that the body uses for energy.

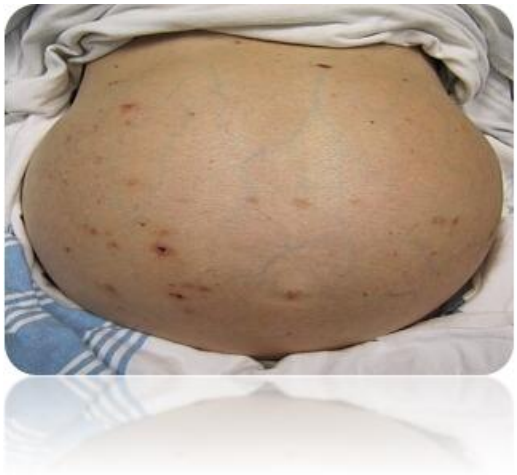
A person with galactosemia lacks one of these enzymes, which causes high levels of galactose in the blood or urine. It is important to test newborns for metabolic diseases without delay, because they can have serious, irreversible effects or even die in the first days of life.

Galactosemia can also be detected before receiving breast milk or formula containing galactose. Almost all cases of classic galactosemia can be detected by newborn screening or NBS. This test

does not depend on the consumption of protein or lactose, and therefore, if the child has not had a blood transfusion, the disease is detected in the first examination. Therefore, the sample should be taken before transfusion. Enzyme damage can occur if sample analysis is delayed or exposed to high temperatures.

For all those who test positive, called the Florida test, another type of test is performed and diagnosed.

- ✓ Determination of the amount of galactose and galactitol in plasma.



- ✓ Accumulation of fluid in the abdominal cavity
- ✓ Difficulties in growth and rest
- ✓ Enlargement of the liver or other affected organs such as the brain, eyes, and kidneys

Treatment of galactosemia: Treatment of galactosemia is divided according to the type of enzyme deficiency and is mainly based on strict dietary control.

ü GALK deficiency: milk should be excluded from the diet (other small sources of galactose can be tolerated). Cataract surgery is often necessary if the diagnosis is made late.

ü GALE deficiency: there is great variability in clinical manifestations. Some forms do not require treatment (only control). Severe forms should be treated with a galactose-restricted diet.

ü GALT deficiency: treatment in this case consists of eliminating all dietary intake of galactose before confirming the diagnosis. Treatment should continue throughout life

A normal average daily intake of galactose is about 6.5 g, and a restricted diet containing about 40 mg of galactose is recommended in patients with classic galactosemia. It is not known exactly

how much galactose is consumed by patients with galactosemia, so the minimum intake should not exceed 125 mg per day.

The main source of galactose in the diet is almost exclusively lactose from mammalian milk and dairy products, although lactose is also present in excipients and in various manufactured and industrial products.

It is recommended to consume milk with soy-based proteins. With the introduction of complementary feeding, maintenance of a completely galactose-free diet is somewhat more complicated due to difficulties in determining the actual content of free or bound galactose in foods.

The diet of patients with galactosemia is aimed at adding external galactose (exogenous) in the least amount to that produced by the body itself (endogenous synthesis). For this purpose, foods are divided into 3 groups:

• Foods that contain almost no galactose (<5 mg/100 g)

• Foods that should be used under supervision (5 mg-20 mg / 100 g): soy formulas with soy flour, pumpkin, Brussels sprouts, peppers, leeks, tomatoes, cocoa, yeast, sunflower seeds, watermelon, kiwi...

• Foods prohibited due to high galactose content (>20 mg/100 g): milk and any dairy products, organ meat, peas, dates, dried figs, raisins...

In addition to diet, two additional treatments should be considered.

The first is a calcium supplement, because the diet for galactosemic children does not provide the necessary amount of calcium. Women with GALT deficiency (classic galactosemia) may need hormonal treatment for ovarian failure.

In 3 types of galactosemia, it is necessary to control the treatment and the general control of the evolution of patients. In each case, it depends on the characteristics of each of the 3 disadvantages and individual needs. Given the complications and treatment methods, this control is more complete and frequent in the case of GALT deficiency (classical galactosemia).

Conclusion: In order to prevent this disease, before starting a family, they should go to genetic centers and get tested. Because a woman or a man knows that he is not sick, but he can be a carrier. Since genes are in the heterozygote (Aa) state in the carrier organism, they cannot cause any pathological conditions. Therefore, it is not recommended to start a family if it is known that both of them are heterozygous after the examination. If an organism in a heterozygous state builds a family with a healthy organism (AA), their children will not have any anomalies or pathological processes.

If a family starts a family without undergoing examinations, if their children feel sick, they should immediately consult a doctor, because the earlier the treatment is started, the more it will be possible to treat the disease before it reaches a serious level. Otherwise, this disease can end in death. Girls with this disease may have problems with the development of the ovaries, and because of this, many women with this disease cannot have children. Patients with this disease must follow a strict diet. They should exclude dairy products from their diet. They should always

take calcium supplements, because the amount of lactose in the body decreases due to the lack of consumption of dairy products. That's why he should always take calcium supplements.\

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