**GALACTOSEMIA**

In Lithuania, newborn screening for galactosemia (GAL) has been implemented since 2015. All neonates are tested for total galactose levels in dry blood samples.

**What is galactosemia?**

Galactosemia is a rare hereditary metabolic disorder characterized by impaired metabolism of carbohydrate galactose. Galactose is not broken down or is broken down unusually slowly, causing serious health problems.

**What is the incidence rate of galactosemia?**

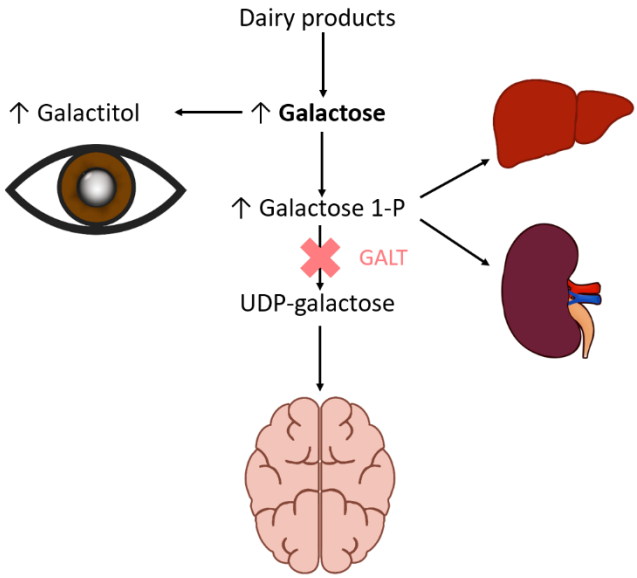
There are several forms of the disease and incidence of each varies. Most common form - classical galactosemia – affects on average 1 in 40,000 – 60,000 newborns, other two forms are very rare. In Lithuania, incidence of galactosemia is 1 in 42,000 births.

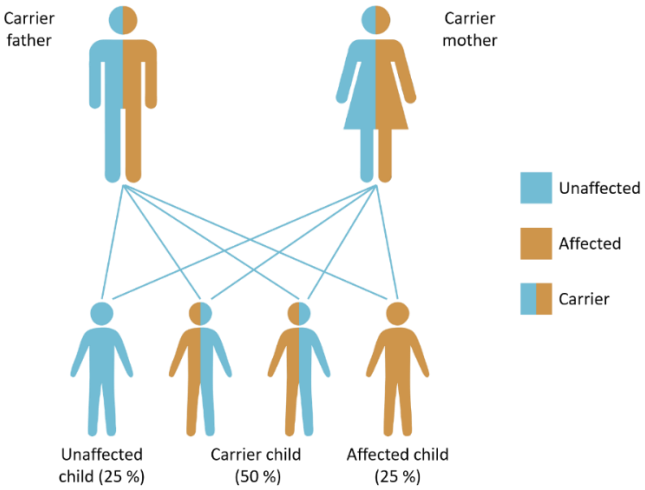
**What causes galactosemia?**

Galactose is a carbohydrate produced by digestion of lactose. Galactose is abundant in breast milk and all dairy products.

Three enzymes are known to be involved in galactose metabolism and their dysfunction can cause galactosemia. Classical galactosemia is most common and related to deficiency of enzyme GALT (galactose-1-phosphate uridyltransferase).

When galactose metabolism is impaired, galactose and by-products of galactose metabolism accumulates in blood or tissues and have toxic effect. Lack of important galactose metabolism products develops as well. Elevated levels of galactose are found in blood samples.




**How is galactosemia inherited?**

Galactosemia is inherited in autosomal recessive manner. Usually both parents are healthy carriers of a disease-causing mutation. If child receives an altered gene from both parents, he or she is born with GAL. Each pregnancy holds 25 percent chance of having a healthy child who will not be a carrier of the disease, 50 percent probability that a child will be a healthy carrier of the disease and 25 percent chance of having a child with GAL.

**How does galactosemia manifest?**

First symptoms appear when breastfeeding or formula-feeding starts.

Classical galactosemia causes vomiting and diarrhea, decreased muscle tone, and later jaundice. Liver and kidney are damaged, cataracts develop. Hepatic and renal impairment and susceptibility to *E. coli* sepsis endanger life of the baby.

Sometimes course of the disease is slower, as mental retardation, liver enlargement, cataracts gradually progress. Approximately 80 % of affected girls under 20 years of age develop ovarian failure.

Other rare disorders of galactose metabolism are similar in clinical picture.

**How is galactosemia treated?**

Immediate dietary treatment without lactose and galactose is prescribed. Breastfeeding is discontinued, and lactose-free formula is administered. Most of the symptoms caused by galactosemia usually disappear if treatment is started on time. However, some children with classical galactosemia develop long-term complications.