Galactosemia

WHAT IS GALACTOSEMIA?

Galactosemia is a genetic disorder causing too much galactose in the blood. Every person is born with a number of enzymes, or proteins, that are needed to break down fats and sugars for energy. One of those enzymes, galactose-1-phosphate uridyltansferase or GALT, is important in the break down of a specific sugar called galactose. Galactose is one of the main components of lactose, the primary sugar found in milk and dairy products. People with galactosemia have a deficiency of the GALT enzyme. This means that their GALT enzyme does not adequately break down galactose, which accumulates in the body as galactose-1-phosphate and can lead to health problems.

NORMAL WAY OF MAKING ENERGY FROM LACTOSE IN THE BODY

Milk ⇒ Lactose ⇒ Galactose ⇒ Glucose ⇒ Energy

The baby with galactosemia cannot change galactose into glucose because the body is missing an enzyme to do this. If the baby with galactosemia is untreated, galactose goes in to the blood and starts to collect. The blood then carries the galactose to different organs of the body, causing damage to the tissue.

WHAT ARE THE SYMPTOMS OF CLASSICAL GALACTOSEMIA?

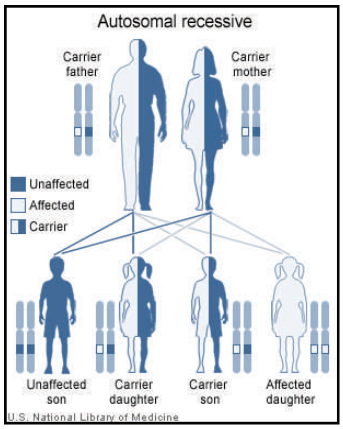
The baby fails to grow and gain weight. The baby may cry frequently, have vomiting and stomach bloating. Symptoms begin to appear within days after milk feeding is started. Milk feeding includes breast milk, most commercial formulas and milk from cows or goats.

If the condition is not discovered in the first few days, other problems may develop from the build up of galactose. These include enlarged liver with cirrhosis, eye cataracts, infections and mental retardation. Most of these problems can be reversed if galactosemia is discovered and treated early.

Babies who are carriers of galactosemia or have a variant form may not have any symptoms. This is because they have enough of the enzyme to break down galactose.

WHAT CAUSES GALACTOSEMIA?

Galactosemia is an inherited recessive genetic condition. Both parents must carry a altered gene for galactosemia if an infant is to have the condition. A carrier parent has one normal gene and one altered gene. The infant who has inherited the altered gene from each parent has the condition and he or she is not able to convert galactose to glucose. The one normal gene in a carrier parent provides enough enzyme capability so this person does not show any signs of galactosemia. Thus, people with an altered gene for the galactosemia enzyme rarely know that they are carriers unless they have a child with the condition.



TESTING FOR GALACTOSEMIA

Classical galactosemia has been found to occur about once in 50,000 births. If the screening test is abnormal, further testing is necessary to confirm the diagnosis. A sample of blood is taken from the baby to test for the GALT enzyme level and the specific gene(s). A low level of enzyme may indicate galactosemia, a carrier of the galactosemia gene, or a variant of galactosemia. If too low, treatment should begin at once.

TREATMENT FOR CLASSICAL GALACTOSEMIA

To treat classical galactosemia, all sources of galactose must be removed from the baby's diet. Most galactose in food is a part of lactose, the sugar found in milk, but other foods may contain related substances, which must also be avoided. The baby will be put on a soy-based milk substitute (ProSobee® or Isomil®) that does not contain lactose or galactose. Galactose and lactose are also found in many other foods and in some medicines. As a baby gets older, all dairy products and certain other foods will need to be restricted.

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