**Classical Galactosemia (GALT)**

Classical galactosemia is caused by mutations in the GALT gene. This gene encodes for the enzyme galactose-1-phosphate uridyltransferase, which is one of the enzymes that break down galactose. If the enzyme fails to function, increasing amounts of galactose-1-phosphate build up in the body, causing damage to tissues. The symptoms usually appear in the first few days of life. Babies suffer from vomiting, diarrhea, liver damage, jaundice, and fail to thrive. They are more susceptible to infection from bacteria such as *E. coli* than normal. Untreated babies usually die, or have severe brain damage. Feeding babies from birth on lactose-free formula milk is necessary. As they get older, a special diet absent of galactose and lactose is necessary. Even so, treated children are still at risk of poor growth, eye and speech problems, and mild intellectual disability. Women tend to suffer from premature ovarian insufficiency, so may not be able to have children. A “clinical variant” galactosemia, with slightly milder symptoms and without the increased risk of bacterial infection, has been described. This is also caused by defects in the GALT gene. Other types of galactosemia are caused by defects in other genes.

The incidence of classical galactosemia has been estimated as 1 in 10,000 to 1 in 48,000 in the general population. The disease is particularly common among Irish travelers and their descendants, where up to 1 in 14 may be carriers, compared to about 1 in 125 in the general population. The “clinical variant” form is mainly found in African Americans. The disease is autosomal recessive, typically requiring both parents to be asymptomatic carriers of the faulty gene. If a sufferer has children with a partner who is not a carrier for the disease, the children will be asymptomatic carriers.

**Sources**

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