Galactosemia

National Cancer Institute

Source

National Cancer Institute. Galactosemia. NCI Thesaurus. Code C84723.

An autosomal recessive inherited metabolic disorder caused by mutations in the GALE, GALK1, and GALT genes. It is characterized by deficiency of the enzymes responsible for the metabolism of galactose. Signs and symptoms include intellectual disability, hepatomegaly, hepatic failure, and renal failure.