

Galactosemia is a rare, hereditary disorder of carbohydrate metabolism that affects the body's ability to convert galactose (a sugar contained in milk, including human mother's milk) to glucose (a different type of sugar). The disorder is caused by a deficiency of an enzyme galactose-1-phosphate uridylyl transferase (GALT) which is vital to this process. Early diagnosis and treatment with a lactose-restricted (dairy-free) diet is absolutely essential to avoid profound intellectual disability, liver failure and death in the newborn period. Galactosemia is inherited as an autosomal recessive genetic condition. Classic galactosemia and clinical variant galactosemia can both result in life-threatening health problems unless treatment is started shortly after birth. A biochemical variant form of galactosemia termed Duarte is not thought to cause clinical disease due to lactose consumption.