Galactosemia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Galactosemia. ORPHA:352

Galactosemia is a group of rare genetic metabolic disorders characterized by impaired galactose metabolism resulting in a range of variable manifestations encompassing a severe, life-threatening disease (classic galactosemia), a rare mild form (galactokinase deficiency) causing cataract, and a very rare form with variable severity (galactose epimerase deficiency) resembling classic galactosemia in the severe form (see these terms).