

Opis choroby *

Definicja

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).

Dane

Klasyfikacja

Kategoria

Kod ORPHA

352

Kod OMIM

230400

Kod ICD10

E74.2

Kod ICD11

5C51.4Y

*Źródło

orphanet