

# Galaktozemia

**Kod Orpha: 352 Kod OMIM: 230400**

## 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

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[\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.