

## Opis choroby \*

### Definicja

A rare disorder of galactose metabolism characterized by persistent congenital galactosemia due to deficiency of the enzyme galactose mutarotase. Patients may present bilateral cataract, while gastrointestinal symptoms or severe liver dysfunction are absent. The natural history of the disease is unknown. Severe complications, such as neurological symptoms, have not been reported under early treatment with a galactose-restricted diet.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

GALM deficiency

Galaktozemia typu 4

Galactosemia type 4

#### Kod ORPHA

570422

#### Kod OMIM

618881

#### Kod ICD10

E88.8

#### Kod ICD11

5C51.4Y

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#### \*Źródło

orphanet