

## Opis choroby \*

### Definicja

A rare mild form of galactosemia characterized by early onset of cataract and an absence of the usual signs of classic galactosemia, i.e. feeding difficulties, poor weight gain and growth, lethargy, and jaundice.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

GALK deficiency

Galaktozemia typu 2

Galaktozemia z niedoboru galaktokinazy

GALK-D

Niedobór GALK

GALK-D

Galactokinase deficiency galactosemia

Galactosemia type 2

#### Kod ORPHA

79237

#### Kod OMIM

230200

#### Kod ICD10

E74.2

#### Kod ICD11

5C51.41

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#### \*Źródło

orphanet