

Niedobór mutarotazy galaktozowej

Kod Orpha: 570422 Kod OMIM: 618881

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

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.