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