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		

<u>*Źródło</u>

orphanet