Opis choroby *

Definicja

A rare glycogen storage disease due to a deficiency in solute carrier family 2, facilitated glucose transporter member 2 and characterized by hepatorenal glycogen accumulation leading to severe renal tubular dysfunction and impaired glucose and galactose metabolism.

Dane

Klasyfikacja
Choroba

Synonimy
GSD due to GLUT2 deficiency
Choroba Fanconiego i Bickela
Glikogenoza Fanconiego i Bickela
Glikogenoza z powodu niedoboru GLUT2
GSD z powodu niedoboru GLUT2
Glycogen storage disease due to GLUT2 deficiency
Glycogenosis due to GLUT2 deficiency

Kod ORPHA
2088

Kod OMIM
227810

Kod ICD10
E74.0

Kod ICD11
5C51.3

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*Źródło

orphanet