

## 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  
Glycogenesis due to GLUT2 deficiency

#### Kod ORPHA

2088

#### Kod OMIM

227810

#### Kod ICD10

E74.0

#### Kod ICD11

5C51.3

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#### \*Źródło

orphanet