Opis choroby *

Definicja

Glycogenosis due to glucose-6-phosphatase (G6P) deficiency or glycogen storage disease, (GSD), type 1, is a group of inherited metabolic diseases, including types a and b (see these terms), and characterized by poor tolerance to fasting, growth retardation and hepatomegaly resulting from accumulation of glycogen and fat in the liver.

Dane

Klasyfikacja
Choroba

Synonimy
G6P deficiency
Choroba spichrzania glikogenu spowodowana niedoborem G6P
Choroba spichrzania glikogenu typu 1
Choroba Von Gierke
Glikogenoza typu 1
Glikogenoza wątrobowo-nerkowa
GSD spowodowane niedoborem G6P
GSD typu 1
Niedobór G6P
GSD due to G6P deficiency
GSD type 1
GSD type I
Glycogen storage disease due to G6P deficiency
Glycogen storage disease type 1
Glycogen storage disease type I
Glycogenosis type 1
Glycogenosis type I
Hepatorenal glycogenosis
Von Gierke disease

Kod ORPHA
364

Kod OMIM
232240

Kod ICD10
E74.0

Kod ICD11
5C51.3

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*Źródło

orphanet