

Opis choroby *

Definicja

Glycogen storage disease (GSD) due to phosphorylase kinase deficiency is a group of inborn errors of glycogen metabolism that is clinically and genetically heterogeneous. This group comprises GSD due to liver phosphorylase kinase (PhK) deficiency, GSD due to muscle PhK deficiency and GSD due to liver and muscle PhK deficiency (see these terms).

Dane

Klasyfikacja

Grupa fenomenów

Synonimy

GSD due to phosphorylase kinase deficiency
Choroba spichrzania glikogenu spowodowana niedoborem PhK
Choroba spichrzania glikogenu typu 9
Choroba spichrzania glikogenu typu IX
Glikogenoza spowodowana niedoborem kinazy fosforylazy
Glikogenoza spowodowana niedoborem PhK
Glikogenoza typu 9
GSD spowodowana niedoborem kinazy fosforylazy
GSD typu 9
GSD typu IX
GSD type 9
GSD type IX
Glycogen storage disease due to PhK deficiency
Glycogen storage disease type 9
Glycogen storage disease type IX
Glycogenesis due to phosphorylase kinase deficiency
Glycogenesis type 9
Glycogenesis type IX
Gycogenesis due to PhK deficiency

Kod ORPHA

370

Kod OMIM

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Kod ICD10

E74.0

Kod ICD11

5C51.3

[*Źródło](#)

orphonet