

Opis choroby *

Definicja

Glycogen storage disease due to aldolase A deficiency is an extremely rare glycogen storage disease (see this term) characterized by hemolytic anemia with or without myopathy or intellectual deficit. Myopathy can be severe enough to result in fatal rhabdomyolysis in some patients. A family with episodic rhabdomyolysis (triggered by fever) without hemolytic anemia has recently been reported.

Dane

Klasyfikacja

Choroba

Synonimy

GSD due to aldolase A deficiency
Choroba spichrzania glikogenu typu 12
Glikogenoza typu 12
Glikogenoza z powodu niedoboru aldolazy A
GSD typu 12
GSD z powodu niedoboru aldolazy A
GSD type 12
GSD type XII
Glycogen storage disease type 12
Glycogen storage disease type XII
Glycogenesis due to aldolase A deficiency
Glycogenesis type 12
Glycogenesis type XII

Kod ORPHA

57

Kod OMIM

611881

Kod ICD10

E74.0

Kod ICD11

5C51.3

[*Źródło](#)

orphanet