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
Glycogenosis due to aldolase A deficiency
Glycogenosis type 12
Glycogenosis type XII

Kod ORPHA
57

Kod OMIM
611881

Kod ICD10
E74.0

Kod ICD11
5C51.3

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*Źródło

orphanet