

## 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](#)

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