

## Opis choroby \*

### Definicja

A rare metabolic myopathy characterized by exercise-induced cramp, myoglobinuria, and presence of tubular aggregates in the muscle biopsy. Serum creatine kinase (CK) levels are increased between episodes of myoglobinuria.

### Dane

#### Klasyfikacja

##### Choroba

#### Synonimy

GSD due to phosphoglycerate mutase deficiency  
Glikogenoza z powodu niedoboru mutazy fosfoglicerynianu  
GSD typu 10  
GSD z powodu niedoboru mutazy fosfoglicerynianu  
Miopatia z powodu niedoboru mutazy fosfoglicerynianu  
Niedobór mięśniowej mutazy fosfoglicerynianu  
GSD typu 10  
Glycogenosis due to phosphoglycerate mutase deficiency  
Muscle phosphoglycerate mutase deficiency  
Myopathy due to phosphoglycerate mutase deficiency

#### Kod ORPHA

97234

#### Kod OMIM

261670

#### Kod ICD10

E74.0

#### Kod ICD11

5C51.3

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#### [\\*Źródło](#)

orphanet