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 type 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