

## Opis choroby \*

### Definicja

A rare genetic glycogen storage disease characterized by either lactate dehydrogenase (LDH) M- or H-subunit deficiency. Main features of LDH M-subunit deficiency are exertional fatigue and muscle pain potentially accompanied by myoglobinuria. Some patients may develop pustular psoriasis-like skin lesions. Complications of pregnancy, such as frequent abdominal pains and increased uterine tone with a risk of dystocia have also been described. LDH H-subunit deficiency manifests with low serum LDH activity of unclear clinical relevance.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

GSD due to lactate dehydrogenase deficiency  
Glikogenoza z powodu niedoboru  
dehydrogenazy mleczanowej  
GSD z powodu niedoboru dehydrogenazy  
mleczanowej  
Niedobór LDH  
Glycogenesis due to lactate dehydrogenase  
deficiency  
LDH deficiency

#### Kod ORPHA

2364

#### Kod OMIM

614128

#### Kod ICD10

E74.4

#### Kod ICD11

5C51.3

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#### \*Źródło

orphanet