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
Glycogenosis 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