

Opis choroby *

Definicja

A rare, genetic vacuolar myopathy characterised by mild myopathy or elevated levels of creatine kinase in the blood without associated symptoms.

Dane

Klasyfikacja

Choroba

Synonimy

Myopathy due to calsequestrin and SERCA1 protein overload
Vacuolar aggregate myopathy
Myopathy due to calsequestrin and SERCA1 protein overload
Vacuolar aggregate myopathy

Kod ORPHA

88635

Kod OMIM

616231

Kod ICD10

G71.8

Kod ICD11

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*Źródło

orphanet