

Opis choroby *

Definicja

Glycogen storage disease due to LAMP-2 (Lysosomal-Associated Membrane Protein 2) deficiency is a lysosomal glycogen storage disease characterised by severe cardiomyopathy and variable degrees of muscle weakness, frequently associated with intellectual deficit.

Dane

Klasyfikacja

Choroba

Synonimy

Danon disease

Choroba Danona

Choroba spichrzania glikogenu z prawidłowym poziomem kwaśnej maltazy

Glikogenoza z powodu niedoboru LAMP-2

GSD z powodu niedoboru LAMP-2

GSD due to LAMP-2 deficiency

Glycogenosis due to LAMP-2 deficiency

Lysosomal glycogen storage disease with normal acid maltase activity

Kod ORPHA

34587

Kod OMIM

300257

Kod ICD10

E74.0

Kod ICD11

5C51.3

*Źródło

orphanet