

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	Synonimy
Choroba	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