DISEASE:
Glycogen storage disease due to LAMP-2 deficiency

NAME:	Glycogen storage disease due to LAMP-2 deficiency
DESCRIPTION:	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.
ORPHACODE:	34587
SYNONYMS:	Danon disease GSD due to LAMP-2 deficiency Glycogenosis due to LAMP-2 deficiency Lysosomal glycogen storage disease with normal acid maltase activity
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	LAMP2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Cardiomyopathy, hereditary (gene panel)
- Cardiopathies, hereditary (gene panel)
- Hypertrophic cardiomyopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- lysosomal associated membrane protein 2

Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB
- Cardiopathies, hereditary (102 genes) - KUL
- Hypertrophic cardiomyopathy (75 genes) - IPG