DISEASE:
Glycogen storage disease due to muscle glycogen phosphorylase deficiency

NAME:	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
DESCRIPTION:	Myophosphorylase deficiency (McArdle's disease), or glycogen storage disease type 5 (GSD5) , is a severe form of glycogen storage disease characterized by exercise intolerance.
ORPHACODE:	368
SYNONYMS:	GSD due to muscle glycogen phosphorylase deficiency GSD type 5 GSD type V Glycogen storage disease type 5 Glycogen storage disease type V Glycogenosis due to muscle glycogen phosphorylase deficiency Glycogenosis type 5 Glycogenosis type V McArdle disease Myophosphorylase deficiency

XREF(S):	Orphanet MedDRA ICD-10 MeSH OMIM MeSH
ANALYTE(S):	PYGM
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/56

RELATED CONTENT

Related Genetic Tests

- Mc Ardle disease, glycogene storage disease type V
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- glycogen phosphorylase, muscle associated

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB