DISEASE:
Glycogen storage disease due to muscle phosphorylase kinase deficiency

NAME:	Glycogen storage disease due to muscle phosphorylase kinase deficiency
DESCRIPTION:	Glycogen storage disease due to muscle phosphorylase kinase (PhK) deficiency is a benign inborn error of glycogen metabolism characterized by exercise intolerance.
ORPHACODE:	715
SYNONYMS:	GSD due to muscle phosphorylase kinase deficiency GSD type 9D GSD type 9E GSD type IXd GSD type IXe Glycogen storage disease type 9D Glycogen storage disease type 9E Glycogen storage disease type IXd Glycogen storage disease type IXe Glycogenosis due to muscle phosphorylase kinase deficiency Glycogenosis type 9D Glycogenosis type 9E Glycogenosis type IXd Glycogenosis type IXe

XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PHKA1 PHKG1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- phosphorylase kinase regulatory subunit alpha 1
- phosphorylase kinase catalytic subunit gamma 1

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB