DISEASE:
Glycogen storage disease due to liver phosphorylase kinase deficiency

NAME:	Glycogen storage disease due to liver phosphorylase kinase deficiency
DESCRIPTION:	Glycogen storage disease (GSD) due to liver phosphorylase kinase (PhK) deficiency is a benign inborn error of glycogen metabolism characterized by hepatomegaly, growth retardation, and mild delay in motor development during childhood.
ORPHACODE:	264580

SYNONYMS:	GSD due to liver phosphorylase kinase deficiency GSD type 9A GSD type 9C GSD type IXa GSD type IXc Glycogen storage disease type 9A Glycogen storage disease type 9C Glycogen storage disease type IXa Glycogen storage disease type IXc Glycogenosis due to liver phosphorylase kinase deficiency Glycogenosis type 9A Glycogenosis type 9C Glycogenosis type IXa Glycogenosis type IXc XLG
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	PHKA2 PHKG2
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RELATED CONTENT

Related Genetic Tests

- Glycogen storage disease type 9

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman

Related Analytes

- phosphorylase kinase regulatory subunit alpha 2
- phosphorylase kinase catalytic subunit gamma 2