

DISEASE:**Glycogen storage disease due to muscle phosphofructokinase deficiency**

NAME:	Glycogen storage disease due to muscle phosphofructokinase deficiency
DESCRIPTION:	Muscle phosphofructokinase (PFK) deficiency (Tarui's disease), or glycogen storage disease type 7 (GSD7), is a rare form of glycogen storage disease characterized by exertional fatigue and muscular exercise intolerance. It occurs in childhood.
ORPHACODE:	371
SYNONYMS:	GSD due to muscle phosphofructokinase deficiency GSD type 7 GSD type VII Glycogen storage disease type 7 Glycogen storage disease type VII Glycogenosis due to muscle phosphofructokinase deficiency Glycogenosis type 7 Glycogenosis type VII Tarui disease
XREF(S):	Orphanet OMIM MedDRA ICD-10

ANALYTE(S):	<u>PFKM</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/59>

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

- phosphofructokinase, muscle

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB

Source URL: <http://gentest.healthdata.be/disease/59>