DISEASE:
PGM1-CDG

NAME:	PGM1-CDG
DESCRIPTION:	A rare, genetic, congenital disorder of glycosylation and glycogen storage disease characterized by a wide range of clinical manifestations, most commonly presenting with bifid uvula with or without cleft palate at birth, associated with growth delay, hepatopathy with elevated aminotransferase serum levels, myopathy (including exercise-related fatigue, exercise intolerance, muscle weakness), intermittent hypoglycemia, and dilated cardiomyopathy and/or cardiac arrest, due to decreased phosphoglucomutase 1 enzyme activity. Less common manifestations include malignant hyperthermia, rhabdomyolysis, and hypogonadotropic hypogonadism with delayed puberty.
ORPHACODE:	319646
SYNONYMS:	CDG syndrome type It CDG-It CDG1T Congenital disorder of glycosylation type 1t Congenital disorder of glycosylation type It PGM1-related congenital disorder of glycosylation Phosphoglucomutase-1 deficiency

XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PGM1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)
- 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
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- phosphoglucomutase 1

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL
- Neuromuscular disorders (166 genes) - VUB