

# PGM1-CDG

**Kod Orpha: 319646 Kod OMIM: 614921**

## Opis choroby \*

### Definicja

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.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

CDG syndrome type 1t

CDG1t

CDG-1t

Zespół CDG typu 1t

CDG-1t

CDG1T

Congenital disorder of glycosylation type 1t

Congenital disorder of glycosylation type 1t

PGM1-related congenital disorder of glycosylation

Phosphoglucomutase-1 deficiency

#### Kod ORPHA

319646

#### Kod OMIM

614921

#### Kod ICD10

E77.8

#### Kod ICD11

5C54.0

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## **Rozszerzony opis choroby**

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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