

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

[*Źródło](#)

orphanet