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 It
CDG1t
CDG-It
Zespół CDG typu 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

Kod ORPHA
319646

Kod OMIM
614921

Kod ICD10
E77.8

Kod ICD11
5C54.0

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*Źródło

orphanet