Opis choroby *

Definicja

Glycogen storage disease due to muscle and heart glycogen synthase deficiency is characterised by muscle and heart glycogen deficiency. It has been described in three siblings (two brothers and their younger sister). The older brother died at 10.5 years of age as a result of sudden cardiac arrest and the younger brother presented with hypertrophic cardiomyopathy, abnormal heart rate and blood pressure during exercise, and muscle fatigability. The sister showed no symptoms but a lack of glycogen was identified through muscle biopsy. The syndrome is caused by homozygous missense mutations in the gene encoding muscle glycogen synthase.

Dane

Klasyfikacja
Choroba

Synonimy
GSD due to muscle and heart glycogen synthase deficiency
Choroba spichrzania glikogenu typu 0b
Glikogenoza typu 0b
Glikogenoza z powodu niedoboru mięśniowej i sercowej syntazy glikogenu
GSD typu 0b
GSD z powodu niedoboru mięśniowej i sercowej syntazy glikogenu
GSD type 0b
Glycogen storage disease type 0b
Glycogenosis due to muscle and heart glycogen synthase deficiency
Glycogenosis type 0b

Kod ORPHA
137625

Kod OMIM
611556

Kod ICD10
E74.0

Kod ICD11
5C51.3

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*Źródło

orphanet