

Opis choroby *

Definicja

Multiple acyl-CoA dehydrogenation deficiency (MADD) is a disorder of fatty acid and amino acid oxidation and is a clinically heterogeneous disorder ranging from a severe neonatal presentation with metabolic acidosis, cardiomyopathy and liver disease, to a mild childhood/adult disease with episodic metabolic decompensation, muscle weakness, and respiratory failure.

Dane

Klasyfikacja

Choroba

Synonimy

Glutaric acidemia type 2
MADD
Niedobór MAD
Kwasica glutarowa typu 2
Acyduria glutarowa typu 2
Glutaric aciduria type 2
MAD deficiency
MADD

Kod ORPHA

26791

Kod OMIM

231680

Kod ICD10

E71.3

Kod ICD11

5C52.01

[*Źródło](#)

orphanet