

Opis choroby *

Definicja

Glutaryl-CoA dehydrogenase (GCDH) deficiency (GDD) is an autosomal recessive neurometabolic disorder clinically characterized by encephalopathic crises resulting in striatal injury and a severe dystonic dyskinetic movement disorder.

Dane

Klasyfikacja

Choroba

Synonimy

GA1

Acyduria glutarowa typu 1

GA1

GCDHD

Kwasica glutarowa typu 1

Niedobór dehydrogenazy glutarylo-koenzymu A

GCDHD

Glutaric acidemia type 1

Glutaric aciduria type 1

Glutaryl-coenzyme A dehydrogenase deficiency

Kod ORPHA

25

Kod OMIM

231670

Kod ICD10

E72.3

Kod ICD11

5C50.E1

*Źródło

orphanet