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