

## 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

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#### \*Źródło

orphanet