

## Opis choroby \*

### Definicja

A rare inborn error of metabolism characterized by abnormal urinary excretion of D-glyceric acid due to D-glycerate kinase deficiency. Reported manifestations are highly variable and include a severe encephalopathic picture, chronic metabolic acidosis, developmental delay, intellectual disability, microcephaly, seizures, behavioral abnormalities, as well as only mild speech delay and apparently normal development.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

D-glycerate kinase deficiency  
Kwasica D-glicerynowa  
Niedobór kinazy D-glicerynowej  
D-glyceric acidemia

#### Kod ORPHA

941

#### Kod OMIM

220120

#### Kod ICD10

E74.8

#### Kod ICD11

5C50.7Y

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#### [\\*Źródło](#)

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