

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

[*Źródło](#)

orphanet