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

orphanet