

Kwasica oksoglutarowa

Kod Orpha: 31 Kod OMIM: 203740

Opis choroby *

Definicja

A rare, genetic, inborn error of metabolism disorder characterized by neonatal-onset of developmental delay, hypotonia, hepatomegaly, lactic acidemia, increased creatine kinase levels, elevated alpha-ketoglutaric acid in urine, and a decreased plasma beta-hydroxybutyrate-to-acetoacetate ratio. Pyruvate dehydrogenase deficiency can be associated, leading to hypoglycemia and neurologic anomalies, including seizures.

Dane

Klasyfikacja

Choroba

Synonimy

Alpha-ketoglutarate dehydrogenase deficiency
Niedobór dehydrogenazy alfa-ketoglutarowej

Kod ORPHA

31

Kod OMIM

203740

Kod ICD10

E88.8

Kod ICD11

5C53.1

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.