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

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*Źródło

orphanet