

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-ketoglutarrowej
--------------------------------	--

Kod ORPHA 31	Kod OMIM 203740	Kod ICD10 E88.8
------------------------	---------------------------	---------------------------

Kod ICD11
5C53.1

*Źródło

orphanet