

## 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

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#### \*Źródło

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