## Opis choroby *

Definicja

A rare, hereditary inborn error of metabolism characterized by an acute onset of encephalopathy in infancy or early childhood. Apart from these episodic acute events, the disorder shows a relatively benign course. Multiple metabolic abnormalities are present, including metabolic acidosis, respiratory alkalosis, hypoglycemia, increased serum lactate and alanine.

Dane

**Klasyfikacja**
Choroba

Synonimy
CA-VA deficiency
Niedobór CA-VA

**Kod ORPHA**
401948

**Kod OMIM**
615751

**Kod ICD10**
E74.8

**Kod ICD11**
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*Źródło

orphanet