Opis choroby *

Definicja

A rare disorder of ketone body transport characterized by recurrent episodes of ketoacidosis provoked by fasting or infections in the first years of life. The episodes are typically preceded by poor feeding and vomiting and are associated with dehydration, in severe cases also with decreased consciousness and insufficient respiratory drive. Hypoglycemia is observed only infrequently. Patients with homozygous mutations tend to present at a younger age, have more profound ketoacidosis, and may show mild to moderate developmental delay in addition.

Dane

Klasyfikacja
Choroba

Kod ORPHA
438075

Kod OMIM
616095

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet