Niedobór syntetazy 3-hydroksy-3-metyloglutarylo-CoA
Kod Orpha: 35701 Kod OMIM: 605911

Opis choroby *

Definicja
3-hydroxy-3-methylglutaryl-CoA synthase deficiency (HMG-CoA synthase deficiency) is a rare autosomal recessively inherited disorder of ketone body metabolism (see this term), reported in less than 20 patients to date, characterized clinically by episodes of decompensation (often associated with gastroenteritis or fasting) that present with vomiting, lethargy, hepatomegaly, non ketotic hypoglycemia and, in rare cases, coma. Patients are mostly asymptomatic between acute epidodes. HMG-CoA synthase deficiency requires an early diagnosis in order to avoid hypoglycemic crises that can lead to permanent brain damage or death.

Dane

Klasyfikacja
Choroba

Synonimy
HMG-CoA synthase deficiency
Niedobór syntetazy HMG-CoA

Kod ORPHA
35701

Kod OMIM
605911

Kod ICD10
E71.3

Kod ICD11
5C52.02

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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