

DISEASE:
3-hydroxy-3-methylglutaryl-CoA synthase deficiency

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| NAME: | 3-hydroxy-3-methylglutaryl-CoA synthase deficiency |
| DESCRIPTION: | 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 episodes. HMG-CoA synthase deficiency requires an early diagnosis in order to avoid hypoglycemic crises that can lead to permanent brain damage or death. |
| ORPHACODE: | 35701 |
| SYNONYMS: | HMG-CoA synthase deficiency |
| XREF(S): | Orphanet ICD-10 OMIM |
| ANALYTE(S): | HMGCS2 |
| CREATED: | 13 May 2019 - 01:02 |
| CHANGED: | 22 Jun 2023 - 16:14 |

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