Opis choroby *

Definicja

Medium chain acyl-CoA dehydrogenase (MCAD) deficiency (MCADD) is an inborn error of mitochondrial fatty acid oxidation characterized by a rapidly progressive metabolic crisis, often presenting as hypoketotic hypoglycemia, lethargy, vomiting, seizures and coma, which can be fatal in the absence of emergency medical intervention.

Dane

Klasyfikacja
Choroba

Synonimy
ACADM deficiency
MCADD
Niedobór MCAD
Niedobór ACADM
Niedobór dehydrogenazy średnich łańcuchów acetylo-koenzymu A
Niedobór karnityny wtórny do niedoboru dehydrogenazy średnich łańcuchów acetylo-CoA
Carnitine deficiency secondary to medium-chain acyl-CoA dehydrogenase deficiency
MCAD deficiency
MCADD
Medium chain acyl-coenzyme A dehydrogenase deficiency

Kod ORPHA
42

Kod OMIM
201450

Kod ICD10
E71.3

Kod ICD11
5C52.01

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*Źródło

orphanet