

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

*Źródło

orphanet