Opis choroby *

Definicja

A mitochondrial disorder of long chain fatty acid oxidation characterized in most patients by onset in infancy/ early childhood of hypoketotic hypoglycemia, metabolic acidosis, liver disease, hypotonia and, frequently, cardiac involvement with arrhythmias and/or cardiomyopathy.

Dane

Klasyfikacja
Choroba

Synonimy
LCHAD deficiency
LCHADD
Niedobór LCHAD
Niedobór dehydrogenazy długich łańcuchów 3-hydroksyacetylo-koenzymu A
LCHADD
Long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency

Kod ORPHA
5

Kod OMIM
609016

Kod ICD10
E71.3

Kod ICD11
5C52.01

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*Źródło

orphanet