

## 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