

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

Kod ORPHA	Kod OMIM	Kod ICD10
5	609016	E71.3

Kod ICD11
5C52.01

*Źródło

orphanet