

Opis choroby *

Definicja

Carnitine palmitoyltransferase 1A (CPT-1A) deficiency is an inborn error of metabolism that affects mitochondrial oxidation of long chain fatty acids (LCFA) in the liver and kidneys, and is characterized by recurrent attacks of fasting-induced hypoketotic hypoglycemia and risk of liver failure.

Dane

Klasyfikacja

Choroba

Synonimy

CPT1A deficiency

Niedobór CPT1A

Niedobór L-CPT1

Niedobór L-CPTI

Niedobór palmitylotransferazy 1 karnityny wątrobowej

Niedobór palmitylotransferazy I karnityny wątrobowej

Niedobór palmitylotransferazy IA karnityny

Carnitine palmitoyl transferase IA deficiency

Hepatic carnitine palmitoyl transferase 1 deficiency

Hepatic carnitine palmitoyl transferase I deficiency

L-CPT1 deficiency

L-CPTI deficiency

Kod ORPHA

156

Kod OMIM

255120

Kod ICD10

E71.3

Kod ICD11

5C52.00

*Źródło

orphanet

