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