

Opis choroby *

Definicja

Carnitine palmitoyltransferase II (CPT II) deficiency is an inherited metabolic disorder that affects mitochondrial oxidation of long chain fatty acids (LCFA). Three forms of CPT II deficiency have been described: a myopathic form, a severe infantile form and a neonatal form (see these terms).

Dane

Klasyfikacja

Choroba

Synonimy

CPT2

CPT2

CPTII

Niedobór palmitylotransferazy karnityny 2

CPTII

Carnitine palmitoyltransferase deficiency type 2

Kod ORPHA

157

Kod OMIM

608836

Kod ICD10

E71.3

Kod ICD11

5C52.00

*Źródło

orphanet