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

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*Źródło

orphanet