Carnitinpalmitoyltransferase II

Biochemisch: Serum-Quotient (C16+C18:1)/C2 (+)

Bestätigung: http://www.labor-blessing.de/files/pdf/HG.....-16.02.12.pdf

Metagene: http://www.metagene.de/appl/index.html

Stand: 25. Juli 2016

CARNITINE PALMITOYL TRANSFERASE DEFICIENCY (II)

▾ Disease

Disease	CARNITINE PALMITOYL TRANSFERASE DEFICIENCY (II)
Synonym	MYOPATHY WITH DEFICIENCY OF CARNITINE PALMITOYLTRANSFERASE II, CPT2
OMIM	255110 OMIM = Online Medalian Inheritance of Men
Orphanet	157
Gene locus	1p32.3
Summary	rare autosomal recessive 1) classical milder form in adults 2) severe form in infants unusual type of neonatal form (Hug et al. 1989, 1991; North et al. 1995) CPT-1 deficiency can be missed when analysis is performed in plasma, whereas CPT-2 deficiency can be missed when analysis is performed in DBS [de Sain-van der Velden MG 2013]

OMIM: http://omim.org/entry/255110

Orphanet: http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=157