



www.labor-blessing.de

Institut für Laboratoriumsmedizin
Prof. Blessing, Dr. Fritjof Blessing und Kollegen
Virchowstr. 10c
78224 Singen
Tel.: 07731 - 995 60
Fax: 07731 - 982 683 1
email: info@labor-blessing.de

Mikrobiologie, Infektionsepidemiologie, Virologie,
Immunologie, Molekulare Genetik und Humangenetik
SGZ: Stoffwechsel- und Genzentrum + Laborärzte Singen

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 Medelian 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?Inq=EN&Expert=157