

Carnitinpalmitoyltransferase I

Biochemisch: langkettige AC (-), Serum-Acylcarnitine < 20%

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

Stand: 25. Juli 2016

CARNITINE PALMITOYL TRANSFERASE DEFICIENCY (I)	
▼ Disease	
Disease	CARNITINE PALMITOYL TRANSFERASE DEFICIENCY (I)
Synonym	HYPOGLYCEMIA, HYPOKETOTIC, WITH DEFICIENCY OF CARNITINE PALMITOYLTRANSFERASE I, CPT1
OMIM	255120 OMIM = Online Medalian Inheritance of Men
Orphanet	156
Gene locus	11q13.3
Summary	rare autosomal recessive recurrent attacks of fasting hypoketotic hypoglycemia, normal or high serum carnitine 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/255120>

Orphanet: http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=156