Carnitintransporterdefekt

Biochemisch: freies Carnitin (--), oft alle Acylcarnitine (-),
geringer Anstieg der Serumkonzentration unter Substitution von L-Carnitin

Bestätigung: http://www.labor-blessing.de/files/pdf/HG/Carnitin-Defizienz-14.03.12.pdf

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

Stand: 25. Juli 2016

CARNITINE TRANSPORTER DEFECT. PRIMARY SYSTEMIC CARNITINE DEFICIENCY

▾ Disease

Disease	CARNITINE TRANSPORTER DEFECT. PRIMARY SYSTEMIC CARNITINE DEFICIENCY
Synonym	CARNITINE DEFICIENCY, SYSTEMIC, DUE TO DEFECT IN RENAL ABS.; MYOPATHY-METABOLIC, CARNITINE DEFICIENCY, PRIMARY AND SECON
OMIM	212140 OMIM = Online Medalian Inheritance of Men
Orphanet	158
Gene locus	5q31.1
Summary	rare autosomal recessive mutation in the SLC22A5 gene

OMIM: http://omim.org/entry/212140

Orphanet: http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=DE&Expert=51208