LCHAD-Mangel (long chain 3-hydroxyacyl-CoA-dehydrogenase)

Biochemisch: Hydroxyacylcarnitine C14OH – C18OH (+),

Org. Säuren (Urin): Hydoxydicarbonsäuren (+)

Bestätigung: http://www.labor-blessing.de/files/pdf/HG/LCHAD-Defizienz-17.12.12.pdf

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

Stand: 25. Juli 2016

LONG-CHAIN-3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY (LCHAD)

▾ Disease

Disease	LONG-CHAIN-3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY (LCHAD)
Synonym	LCHAD DEFICIENCY
OMIM	609016 OMIM = Online Medalian Inheritance of Men
Orphanet	5
Gene locus	2p23.3
Summary	rare (1:500000 in Sweden) autosomal recessive

OMIM: http://omim.org/entry/609016

Orphanet: http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=5