



## β-Ketothiolasemangel

**Biochemisch:** Tiglylcarnitin (C5:1), Hydroxyisovalerylcarnitin (C5OH) (+)  
Org. Säuren (Urin): Tiglylglycin (+), 2-Methyl-3-Hydroxybuttersäure (+),  
2-Methylacetoessigsäure (+)

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

**Stand:** 25. Juli 2016

BETA-KETOTHIOLASE DEFICIENCY	
▼ Disease	
<b>Disease</b>	BETA-KETOTHIOLASE DEFICIENCY
<b>Synonym</b>	BETA-KETOTHIOLASE DEFICIENCY; ACIDEMIA, 3-KETOTHIOLASE DEFICIENCY, 3-OXOTHIOLASE DEFICIENCY, T2 DEFICIENCY, MITOCHONDRIA
<b>OMIM</b>	<a href="http://omim.org/entry/203750">203750</a> OMIM = Online Medalian Inheritance of Men
<b>Orphanet</b>	<a href="http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&amp;Expert=134">134</a>
<b>Gene locus</b>	11q22.3
<b>Summary</b>	rare autosomal recessive mutation in the acetyl-CoA acetyltransferase-1 gene clinical, biochemical and genetic heterogeneity 30 mutations of T2

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**Orphanet:** [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?lng=EN&Expert=134](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=134)