

Fatty Acid Oxidation Disorders				
Condition	Method	Tier	Biomarker	Value
2,4 Dienoyl-CoA Reductase Deficiency	MS/MS	1st	C10:2	≥ 0.12 μMol/L
Carnitine Palmitoyltransferase Deficiency Type 1	MS/MS	1st	C0/(C16+C18)	≥ 70
	MS/MS	1st	C0	≥ 60 μMol/L
Carnitine Palmitoyltransferase Deficiency Type 2 and Carnitine/Acylcarnitine Translocase Deficiency	MS/MS	1st	(C16+C18:1)/C2	≥ 0.40
	MS/MS	1st	C16	≥ 8.7 μMol/L
Carnitine Uptake Deficiency	MS/MS	1st	C0	< 4.0 μMol/L
Glutaric Acidemia (Type 2)	MS/MS	1st	C4	≥ 1.2 μMol/L
	MS/MS	1st	C5	≥ 0.60 μMol/L
	MS/MS	1st	C6	≥ 0.30 μMol/L
	MS/MS	1st	C8	≥ 0.50 μMol/L
	MS/MS	1st	C10	≥ 0.34 μMol/L
Long-Chain 3-Hydroxylacyl-CoA Dehydrogenase Deficiency and Trifunctional Protein Deficiency	MS/MS	1st	C16OH	≥ 0.25 μMol/L
	MS/MS	1st	C18OH	≥ 0.12 μMol/L
Medium-Chain 3-Ketoacyl-CoA Thiolase Deficiency	MS/MS	1st	C8	≥ 0.50 μMol/L
	MS/MS	1st	C8/C10	≥ 3.0
Medium-Chain Acyl-CoA Dehydrogenase Deficiency	MS/MS	1st	C6	≥ 0.30 μMol/L
	MS/MS	1st	C8	≥ 0.50 μMol/L
	MS/MS	1st	C10:1	≥ 0.40 μMol/L
	MS/MS	1st	C8/C10	≥ 3.0
Medium/Short-Chain Hydroxyacyl Dehydrogenase Deficiency	MS/MS	1st	C4OH	≥ 3.03 μMol/L
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	MS/MS	1st	C14:1	≥ 1.0 μMol/L
	MS/MS	1st	C14	≥ 0.80 μMol/L
	MS/MS	1st	C14:1/C16	≥ 0.25