

MS/MS DISORDERS AND ANALYTES

Disorders	Primary Marker	Secondary Marker	Notes
	Acylcarnitines		
3MCC	C5OH	C5:1, C5-OH/C8	
BKT (SKAT)	C5OH	C5-OH/C0	
HMG	C5-OH	C6DC	
CPT-2 CAT	↑ C16 ↑C18	Low normal C0 ↑C14 ↑C18:1; C18:2 C16/C2 (C16+C18:1)/C2; C0/(C16+C18) low	
CPT-1	↓ C16↓C18 ↑C0 ↑C0/(C16+C18)	C18:1 low C18:2 low	
LCHAD TFP	C16-OH	C16:1-OH C18-OH C18:1-OH C18-OH/C16	
MMA	C3 Low Met	C3/C16 C3/C2 C3/Met	
MUT			
MCD			
PA			
MCAD	C8	C6; C10; C10:1 C8/C10 C8/C2	If C8:1 > C8 Not significant
VLCAD	C14:1	C12; C14; C14:2 C14:1/C12:1 C14:1/C16; C14:1/C2	If C14:2 < C14:1 Not significant
SCAD	C4	C4/C8; C4/C2	
GA-2 (MADD)	C12 C10	C4; C5; C8; C14; C14:1 or ↑C8 to C18:1	Looks like MCAD, VLCAD. Elevations of many carnitines
IVA	C5	C5/C0; C5/C2; C5/C3	
GA-1	C5DC	C5DC/C16; C5DC/C8 C5DC/C5-OH	
CUD	↓ C0	+ All ACs ↓ C2; C3; C16;C18; C18:1; C3/Met low	
	Aminoacids		
HCY	Met	Met/Phe	
MSUD	Leu	Val, L-alloisoleucine Leu/Phe; Val/phe	
PKU	Phe	Phe/Tyr	
Citrullinemia (ASD) OTC	Cit Low Cit	Cit/Arg; Cit/phe	
TYR-1 TYR-2	Tyr, SUAC Tyr		