Arizona Department of Health Services
Newborn Screening Program

MSMS Pilot Project – Secondary Markers

Amino Acid Disorders	Analytes	Reference Ranges	Ratios	Reference Ranges
Phenylketonuria	Phe	< 181.80 µmol/L	Phe/Tyr	N/A
Maple syrup urine disease	Leu	< 280 µmol/L	Leu/Phe	< 4.70
	Val	< 340 µmol/L	Val/Phe	< 5.20
Homocystinuria	Met	< 75 µmol/L	Met/Phe	< 1.40
Citrullinemia type I & Argininosuccinic acidemia	Cit	≤ 5 days < 91.36 µmol/L > 5 days < 148.46 µmol/L	Cit/Arg	N/A
Tyrosinemia type I	Tyr	< 552.00 µmol/L	Tyr/Cit	N/A
Organic Acidemia Disorders				
Isovaleric acidemia (IVA)	C5	< 0.60µmol/L	C5/C0	< 0.025
			C5/C2	< 0.031
			C5/C3	< 0.460
Glutaric acidemia type I (GA 1)	C5DC	< 0.25µmol/L	C5DC/C5OH	< 2.00
			C5DC/C8	< 3.75
			C5DC/C16	< 0.20
Methylmalonic acidemia , Methylmalonic acidemia (mutase), Propionic acidemia , Multiple carboxylase deficiency (MCD)	C3	< 6.00µmol/L	C3/C2	< 0.21
			C3/C16	< 2.25
3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)† & 3-Hydroxy-3-methylglutaric acidemia (HMG)*	C5-OH	≤ 5 days <0.78 µmol/L > 5 days < 0.92 µmol/L	C5-OH/C8	N/A
	C6DC*	N/A	C5-OH/C0†	N/A
Beta-ketothiolase deficiency (BKT)	C5:1	< 0.25 µmol/L	C5-OH/C8	N/A
	C5-OH	N/A		
Fatty Acid Oxidation Disorders				
Carnitine uptake defect	C0 (Low)	> 8 µmol/L	(C0+C2+C3+C16+C18+C18:1)/Cit	N/A
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)	C8	< 0.60 µmol/L	C8/C2	N/A
	C6	N/A	C8/C10	N/A
	C10	N/A	C5-OH/C8	N/A
	C10:1	N/A		
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	C14:1	< 0.80 µmol/L	C14:1/C16	N/A
	C14	N/A		
	C14:2	N/A		
Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (LCHAD) & Trifunctional protein deficiency (TFP)	C16-OH	< 0.25 µmol/L	C16-OH/C16	N/A
	C16:1-OH	N/A		
	C18-OH	N/A		
	C18:1-OH	N/A		

Effective 1/1/13