

Table 1

<u>Disorders and Deficiencies</u>	<u>Primary Markers</u>	<u>Secondary Markers</u>	<u>Possible Symptoms</u>	<u>Rate of Occurance</u> Per every live birth	<u>Method of Analysis</u>
Amino Acid Disorder/ Deficiencies					
Phenylketonuria (PKU)	PHE	Phe/Tyr	Untreated: Impaired cognitive development and neurophysiological functions	>1/25000	Tandem Mass Spectrometry
Maple Syrup Urine Disease (MSUD)	LEU	Val, Leu/Phe, Val/Phe, Leu/Ala	Neurological symptoms, urine smells like maple syrup	<1/100,000	Tandem Mass Spectrometry
Homocystinuria (HCY)	MET	Met/Phe	Mental retardation, eye complications, stroke, bone abnormalities	<1/100,000	Tandem Mass Spectrometry
Citrullinemia (CIT)	CIT	Cit/Tyr, Cit/Arg	Seizures, coma, brain damage, death	<1/100,000	Tandem Mass Spectrometry
Argininosuccinic Aciduria (ASA)	ASA	Asa/Arg, Cit, Cit/Tyr, Cit/Arg	brain swelling, coma, death	<1/100,000	Tandem Mass Spectrometry
Tyrosinemia I, II, III (TYR-I, II, III)	TYR		Liver and kidney failure, nerve damage, painful cornea erosions with photophobia, death	<1/100,000	Tandem Mass Spectrometry
Fatty Acid					

Disorders					
Carnitine Uptake Defect (CUD)	Low C0	Low C2, CUD ratio, C0/C16+C18	Hypoglycemia, death	<1/100,000	Tandem Mass Spectrometry
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)	C8	C8/C10, C8/C2, C6, C10:1	Seizures, liver failure, death	>1/25,000	Tandem Mass Spectrometry
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	C14:1	C14:1/C12:1, C14:1/C16, C14, C14:2, C14-OH	Heart and liver failure death by first of year	>1/75,000	Tandem Mass Spectrometry
Long Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (LCHAD)	C16-OH	C18-OH, C18:1-OH, C16-OH/C16	Infant: Heart/lung/liver failure, death. Child: low muscle tone, developmental delay, heart/lung/liver failure may develop later in infancy or childhood, most likely following an illness.	>1/75,000	Tandem Mass Spectrometry
Trifunctional Protein Deficiency (TFP)	C16-OH	C18-OH, C18:1-OH, C16-OH/C17	Low muscle tone, seizures, heart failure, and coma, often following an illness. or instant death	<1/100,000	Tandem Mass Spectrometry
Organic Acid Disorder/ Deficiencies					
Propionic Acidemia (PROP)	C3	C3/C2	Brain damage, coma, death	>1/75,000	Tandem Mass Spectrometry
Methylmalonyl-CoA Mutase Deficiency (MUT)	C3	C3/C2, C3/C16	Death in the first month or brain damage for survivors	>1/75,000	Tandem Mass Spectrometry

Methylmalonic Acidemia/ Vitamin B12 Disorder (Cbl-A,B)	C3	C3/C2, C3/C16, C4DC	Brain damage, coma, seizures, paralysis, death	<1/100,000	Tandem Mass Spectrometry
Isovaleric Acidemia (IVA)	C5	C5/C3, C5/C2, C5/CO	Permanent brain damage, death, coma	<1/100,000	Tandem Mass Spectrometry
Glutaric Acidemia Type I (GA-I)	C5DC	C5DC/C5-OH, C5DC/C8, C5DC/C16	Brain damage, seizures, low muscle tone, cerebral-palsy like symptoms and death within the first decade of life.	>1/75,000	Tandem Mass Spectrometry
3-Hydroxy-3- methylglutaryl-CoA Lyase Deficiency (HMG)	C5-OH	C5-OH/C8, C5-OH/CO, C6DC	Brain damage, mental retardation, coma, and death	<1/100,000	Tandem Mass Spectrometry
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)	C5-OH	C5-OH/C8, C5-OH/CO	Brain damage, seizures, liver failure, and death in infancy or no symptoms at all until adulthood	>1/75,000	Tandem Mass Spectrometry
Multiple Carboxylase Deficiency (MCD)	C5-OH	CC5-OH/C8, C5-OH/CO, C3	Skin rashes, hair loss, brain damage, and death	<1/100,000	Tandem Mass Spectrometry
Beta Ketothiolase Deficiency (BKT)	C5-OH	CC5-OH/C8, C5-OH/CO, C5:1	Progress to a coma, brain damage, and death	<1/100,000	Tandem Mass Spectrometry