Newborn Screening Laboratory Methodologies

Metabolic Disorders

Disorders of Amino Acid Metabolism	**Laboratory Methodology**
Citrullinemia (CIT) Homocystinuria (HCY) Maple Syrup Urine Disease (MSUD) Phenylketonuria (PKU) Tyrosinemia I and II (TYR)	Tandem mass spectrometry

Disorders of Fatty Acid Metabolism	
Carnitine Acylcarnitine Translocase Deficiency (CACT) Carnitine Palmitoyl Transferase Deficiency I and II (CPT I AND II) Carnitine Uptake Deficiency (CUD) Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) Medium-Chain Acyl-CoA Dehydrogenase Deficiency Trifunctional Protein Deficiency Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	Tandem Mass Spectrometry

Disorders of Organic Acid Metabolism	
Beta-Ketothiolase Deficiency (BKD) Glutaric Acidemia I (GA I) Isovaleric Acidemia (IVA) Malonic Aciduria Methylmalonic Acidemia (MMA) Proprionic Acidemia (PPA) 3-Methylcrotonyl-CoA Carboxylase Deficiency	Tandem Mass Spectrometry

Enzyme Deficiencies	
Biotinidase	Qualitative Colorimetric Assay
Galactosemia	Fluorometric Assay for Galt Enzyme
Endocrime Disorders	
Congenital Hypothyroidism	Time Resolved Fluoroimmuno Assay
Congenital Adrenal Hyperplasia	Time Resolved Immuno Assay
Hemoglobinopathies	
Hemoglobinopathies	High Pressure Liquid Chromatography If abnormality, Gel Isoelectric Focusing to confirm
Pulmonary/Digestive Disorders	
Cystic Fibrosis	Immunoreactive Trypsinogen: Second tier: DNA