

Georgia's Newborn Screening Panel and Disorder Specific Information

Organic Acid Disorders	 <u>Beta Ketothiolase (BKT)</u> <u>Cobalamin A and B Deficiency (Cbl A, B)</u> <u>Glutaric Acidemia type I (GA1)</u> <u>3-OH 3-CH Glutaric Aciduria (HMG)</u> <u>Isovaleric Acidemia (IVA)</u> <u>3 Methylcrotonyl-Co A Carboxylase Deficiency (3MCC)</u> <u>Multiple Carboxylase Deficiency (MCD)</u> <u>Methylmalonic Acidemia (MMA)</u> <u>Propionic Acidemia</u>
Fatty Acid Oxidation Disorders	 <u>Carnitine Uptake Defect</u> <u>Long Chain 3 hydroxyl acyl-CoA dehydrogenase</u> <u>Deficiency (LCHADD)</u> <u>Medium Chain acyl-CoA dehydrogenase Deficiency</u> <u>Trifunctional Protein Deficiency (TFP)</u> <u>Very Long-chain acyl-CoA dehydrogenase</u> <u>Deficiency (VLCADD)</u>
Amino Acid Disorders	 <u>Argininosuccinic Acidemia</u> <u>Citrullinemia</u> <u>Homocystinuria</u> <u>Maple Syrup Urine Disease (MSUD)</u> <u>Phenylketonuria (PKU)</u> <u>Tyrosinemia</u>
Lysosomal Storage Disorders	 <u>Mucopolysaccharidosis I (MPSI)</u> <u>Pompe Disease</u>



Endocrine Disorders	 <u>Congenital Adrenal Hyperplasia (CAH)</u> <u>Congenital Hypothyroidism (CH)</u>
Hemoglobinopathy Disorders	 Sickle Cell Anemia Sickle Beta Thalassemia Sickle C Disease Other Hemoglobin Variants
Other Disorders	 <u>Biotinidase Deficiency</u> <u>Cystic Fibrosis (CF)</u> <u>Galactosemia</u> <u>Krabbe Disease</u> <u>Severe Combined Immunodeficiency (SCID)</u> <u>Spinal Muscular Atrophy (SMA)</u> <u>X-linked Adrenoleukodystrophy (X-ALD)</u>
Point of Care Screening Disorders	 <u>Critical Congenital Heart Disease (CCHD)</u> <u>Hearing Impairment</u>

(rev 2/10/2022)