LIST OF THE 56 NEWBORN SCREENING DISORDERS SCREENED

Arginase Deficiency (ARG)

Argininosuccinic Acid (ASA)

Beta-kethothiolase Deficiency (BKT)

Biotinidase Deficiency (BIOT)

Carnitine Uptake Defect (CUD)

Carnitine Acylcarnitine Translocase Deficiency (CACT)

Carnitine Palmitoyl Transferase 1 Deficiency (CPT-1)

Carnitine Palmitoyl Transferase 2 Deficiency (CPT-2)

Citrullinemia (CIT-1)

Citrullinemia Type 2 (CIT-2)

Congenital Adrenal Hyperplasia (CAH)

Congenital Hypothyroidism (TSH and T4)

Cystic Fibrosis (CF)

Ethylmalonic Encephalopathy (EE)

Galactosemia (GAL)

Glutaric Acidemia Type 1 (GA-1)

Glutaric Acidemia Type 2 (GA-2)

Hb S/Beta-thalassemia (HbS/Th)

Hb S/C Disease (Hb S/C)

Sickle Cell Anemia (Hb S/S)

Various Hemoglobinopathies (includes Hb E)

Hydroxymethylglutaric Aciduria (HMG)

Hypermethioninemia (MET)

Hyperphenylalaninemia (H-PHE)

Homocystinuria (HCY)

Isobutyryl-CoA Dehydrogenase Deficiency (IBG)

Isovaleric Acidemia (IVA)

Krabbe

Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHADD)

Malonic Acidemia (MAL)

Maple Syrup Urine Disease (MSUD)

Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCADD)

Methylmalonic Acidemia (MMA Cbl A form and MMA Cbl B form)

Methylmalonic Acidemia (MMA Cbl C form and MMA Cbl D form)

Methylmalonyl-CoA Mutase Deficiency (MUT)

2-Methyl-3-Hydroxybutyric Aciduria (2M3HBA)

2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBDH)

3-Methylglutaconic Aciduria (3MGA)

3-Methlycrotonyl-CoA Carboxylase Deficiency (3MCC)

Mucopolysaccharidosis (MPS-1, Hurler's Disease)

Mucopolysaccharidosis (MPS-II, Hunter Syndrome)

Multiple Carboxylase Deficiency (MCD)

Non-Ketotic Hyperglycinemia (NKHG)

Phenylketonuria (PKU)

Pompe

Propionic Acidemia (PA)

Severe Combined Immunodeficiency (SCID)

Short-chain Acyl-CoA Dehydrogenase Deficiency (SCADD)

Spinal Muscular Atrophy (SMA)

Trifunctional Protein Deficiency (TFP)

Tyrosinemia Type 1 (TYR-1)

Tyrosinemia Type 2 (TYR-2)

Tyrosinemia Type 3 (TYR-3)

Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)

X-linked adrenoleukodystrophy (X-ALD)

Guanidinoacetate Methyltransferase Deficiency (GAMT)

Also part of the newborn screen but not listed is Critical Congenital Heart Disease (CCHD) and Hearing Screen