North Carolina State Laboratory of Public Health

Newborn Screening List of Disorders

June 1, 2016

Disorders potentially detectable by Tandem Mass Spectrometry (MS/MS):

Amino Acid Disorders

Argininemia (ARG)

Argininosuccinic aciduria (ASA)

Citrullinemia, type 1 (CIT)

Homocystinuria (HCY)

Maple syrup urine disease (MSUD)

Phenylketonuria (PKU)

Tyrosinemia, type I (TYR I)

Tyrosinemia, type II (TYR II)

Tyrosinemia, type III (TYR III)

Defects of Biopterin cofactor biosynthesis (BIOPT-BS)

Defects of Biopterin cofactor regeneration (BIOPT-REG)

Citrullinemia, type II (CIT-II)

Hypermethioninemia (MET)

Organic Acid Disorders

2-Methylbutyrylglycinuria (2MBG)

3-Hydroxy-3-methylglutaric aciduria (HMG)

3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)

Beta-ketothiolase deficiency (BKT)

Glutaric acidemia, type 1 (GA 1)

Holocarboxylase synthetase deficiency (MCD)

Isobutyrylglycinuria (IBG)

Isovaleric acidemia (IVA)

Malonic acidemia (MAL)

Methylmalonic acidemia (MUT; Cbl A, B; Cbl C, D)

Propionic acidemia (PROP)

Fatty Acid Oxidation Disorders

Carnitine uptake defect/ Carnitine transport defect (CUD)

Carnitine acylcarnitine translocase deficiency (CACT)

Carnitine palmitoyltransferase type II deficiency (CPT II)

Glutaric acidemia, type II (GA 2)

Medium-chain acyl CoA dehydrogenase deficiency (MCAD)

Long-chain L-3 Hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)

Short-chain acyl-CoA dehydrogenase deficiency (SCAD)

Trifunctional protein deficiency (TFP)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Disorders detectable by biochemical and other technologies:

Biotinidase deficiency (BIO)

Classic Galactosemia (GALT)

Congenital adrenal hypoplasia (CAH)

Cystic Fibrosis (CF)

Primary congenital hypothyroidism (CH)

Hearing loss (HEAR)

Hemoglobin C disease (FC)

Hemoglobin E disease (FE)

Sickle cell disease (FS, Hb S/S)

Sickle/hemoglobin C disease (FSC, Hb S/C)

Sickle/hemoglobin E disease (FSE, Hb S/E)

Sickle/hemoglobin D disease (FSD, Hb S/D)

Hemoglobin D disease (FD, Hb D/D)

Sickle/Beta Thalassemia (FSa, Hb S/Beta Thal)