

Disorders Included in the DSHS Newborn Screening Panel

December 1, 2009

AMINO ACID DISORDERS:

Argininosuccinic Acidemia (ASA)
Citrullinemia (CIT)
Homocystinuria (HCY)
Maple Syrup Urine Disease (MSUD)
Phenylketonuria (PKU)
Tyrosinemia Type I (TYRI)

FATTY ACID DISORDERS:

Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
Trifunctional Protein Deficiency (TFP)
Carnitine Uptake Deficiency (CUD)
Carnitine Palmitoyl Transferase Deficiency1 (CPT1)

ORGANIC ACID DISORDERS:

Glutaric Acidemia I (GA-I)
3-OH 3-Methyl Glutaric Aciduria (HMG)
Isovaleric Acidemia (IVA)
Multiple Carboxylase Deficiency (MCD)
3 -Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)
Methylmalonic Acidemia (MMA)
Propionic Acidemia (PA)
Beta-Ketothiolase Deficiency (BKT)

CYSTIC FIBROSIS

GALACTOSEMIA

BIOTINIDASE DEFICIENCY

ENDOCRINE DISORDERS:

Congenital Hypothyroidism (CH)
Congenital Adrenal Hyperplasia (CAH)

HEMOGLOBINOPATHIES including:

Hb S/S
Hb S/C
Hb S-Beta thalassemia