List of Disorders Screened

Amino Acid Profile (AA) Disorders

Argininemia (Arginase Deficiency)
Carbamoylphosphate Synthethase I Deficiency
Citrullinemia
*Type I (Arginosuccinate Synthetase Deficiency)
Type II (Citrin Deficiency)
*Argininosuccinate Lyase Deficiency (Argininosuccinic Aciduria)
$Nonketotic Hyperglycinemia
due to Glycine Cleavage System H Protein Deficiency
due to Aminomethyltransferase Deficiency
due to Glycine Decarboxylase Deficiency
*Homocystinuria or variant forms of Hypermethioninemia
Hypermethioninemia
due to Glycine N-Methyltransferase Deficiency
due to S-Adenosylhomocysteine Hydrolase Deficiency
due to Methionine Adenosyltransferase Deficiency
Hyperornithinemia-Hyperammonemia-Homocitrullinuria
Hyperornithinemia-Hyperammonemia-Homocitrullinuria w/gyral atrophy
*Phenylketonuria
Benign Hyperphenylalaninemia due to:
Phenylalanine Hydroxylase Deficiency
GTP Cyclohydrolase I Deficiency
Pterin-4-Alpha Carbinolamine Dehydratase Deficiency
6-Pyruboyltetrahydropterin Synthase Deficiency
Biopterin defect in co-factor biosynthesis
Biopterin defect in co-factor regeneration
Tyrosinemia
*Type I (Hepatorenal)
Type II
Type III
*Maple Syrup Urine Disease
Type IA
Type IB
Type II
$Ornithine transcarbymylase deficiency (OTC)

Fatty Acid Profile (FA) Disorders

*Carnitine Uptake Deficiency (CUD)
Carnitine Palmitoyl Transferase Deficiency
Type I (CPT I)
Type II (CPT II)
Short Chain AcylCoA Dehydrogenase Deficiency (SCAD)
Glutaric Acidemia Type II (MADD or GAII)
*Medium Chain AcylCoA Dehydrogenase Deficiency (MCAD)
Medium/Short Chain AcylCoA Dehydrogenase Deficiency (M/SCHAD)
2,4 Dienyl CoA Reductase Deficiency
*Very Long Chain AcylCoA Dehydrogenase Deficiency (VLCAD)
*Long Chain Hydroxyl AcylCoA Dehydrogenase Deficiency (LCHAD)
Carnitine /Acylcarnitine Translocase Deficiency(CACTD)
*Trifunctional Protein Deficiency
*X-linked Adrenyleukodystrophy

Organic Acid Profile (OA) Disorders

*Mitochondrial Acetoacetyl CoA Thiolase (Beta Ketothiolase/SKAT) Deficiency
*Propionic Acidemia
*Methylmalonic Acidemia Cobalamin Disorder (CBL A, B)
Methylmalonic Acidemia Cobalamin Disorder (CBL C, D)
*Methylmalonyl-CoA Mutase Deficiency
*Multiple CoA Carboxylase Deficiency
Malonic Aciduria (MA)
Isobutyryl CoA Dehydrogenase Deficiency (IBCD)
*Isovaleric Acidemia (IVA)
2 Methylbutyryl Glycinuria (2MBG)
2 Methyl 3 Hydroxybutyric Aciduria (2M3HBA)
*3 Hydroxy 3 Methylglutaryl CoA Lyase Deficiency (HMG)
*3 Methyl Crotonyl CoA Carboxylase Deficiency (3 MCC)
3 Methylglutaconyl CoA Hydratase Deficiency (3MGA)
*Glutaric Acidemia Type I (GAI)
Medium/Short Chain 3 hydroxyacyl CoA dehydrogenase deficiency (M/SCHAD)

Lysosomal Storage Disorders(LSD)

Krabbe
*Pompe
Gaucher
Fabry
*Mucopolysaccharidosis I (MPSI)

Other Disorders

*Congenital Hypothyroidism
*Congenital Adrenal Hyperplasia
*Biotinidase Deficiency
*Classical Galactosemia
Galactokinase Deficiency
Galactose Epimerase Deficiency
*Cystic Fibrosis
*Sickle Cell Anemia
*Sickle Beta Thalassemia
*Hemoglobin S/C Disease
Other Variant Hemoglobinopathies
*Severe Combined Immunodeficiency
T-cell related lymphocyte deficiencies
*Spinal Muscular Atrophy

Screening Mandated/Information Collection Only

*Critical Congenital Heart Defect
*Hearing Defect

Legend

* = Core RUSP Conditions

$ = Other disease screened not core or secondary conditions

NBS Disorder/Profile Information

TRANSFUSIONS

Unless transfusion is marked, the assumption is that the infant has not been transfused.

TOTAL GALACTOSE

Galactose results are based upon the assumption that the infant has had lactose feeding.

CYSTIC FIBROSIS

The CF DNA Mutation analysis is only performed when there is an elevated IRT.

TANDEM MASS SPECTROMETRY

(AA): Arginine, Citrulline, Cit/Arg, Glycine, Leucine, Methionine, Ornithine, Orn/Cit, Phenylalanine, Tyrosine, Phe/Tyr, Valine, Succinylacetone, Argininosuccinic Acid, Asa/Arg

(FA):C0,C4,C5,C5:1,C5DC+C6OH,C6,C8,C8/C10,C10,C10:1,C10:2,C12:1,C14,C14:1,C14:2,C14-OH,C16,C16:1,C16-OH,C18,C18:1,C18:2,C18:1-OH, C0/C16, C0/C18

(OA): C3, C3DC+C4OH,C4, C4DC+C5OH, C5, C5:1,C6-DC, C3/C2,C4DC+C5OH/C8

LYSOSOMAL DISORDERS

(LD): GAA, GALC, GBA, GLA, IDUA