NORTH CAROLINA NEWBORN SCREENING PROGRAM PANEL
CORE AND SECONDARY CONDITIONS

Amino acid disorders

- Argininemia[§]
- Argininosuccinic Aciduria*
- Benign Hyperphenylalaninemia[§]
- Biopterin Defect in Cofactor Biosynthesis[§]
- Biopterin Defect in Cofactor Regeneration[§]
- Citrullinemia, Type I*
- Citrullinemia, Type II[§]
- Classic Phenylketonuria*
- Homocystinuria*
- Hypermethioninemia[§]
- Maple Syrup Urine Disease*
- Tyrosinemia, Type I*
- Tyrosinemia, Type II[§]
- Tyrosinemia, Type III[§]

Endocrine disorders:

- Primary Congenital Hypothyroidism*
- Congenital Adrenal Hyperplasia*

Fatty acid oxidation disorders

- Carnitine Acylcarnitine Translocase Deficiency[§]
- Carnitine Palmitoyltransferase Type I Deficiency[§]
- Carnitine Palmitoyltransferase Type II Deficiency[§]
- Carnitine Uptake Defect/Carnitine Transport Defect*
- Glutaric Acidemia Type II[§]
- Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency*
- Medium-chain Acyl-CoA Dehydrogenase Deficiency*
- Medium-chain Ketoacyl-CoA Thiolase Deficiency[§]
- Medium/short-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency[§]
- Short-chain Acyl CoA Dehydrogenase Deficiency[§]
- Trifunctional Protein Deficiency*
- Very Long-chain Acyl-CoA Dehydrogenase Deficiency*

Hemoglobin disorders:

- S, βeta-Thalassemia*
- S,C Disease*
- S,S Disease (Sickle Cell Anemia)*
- Various other hemoglobinopathies including[§]**:**
    - Hemoglobin C Disease[§]
    - Hemoglobin E Disease[§]
    - Sickle/hemoglobin C Disease (FSC, Hb S/C)[§]
    - Sickle/hemoglobin E Disease (FSE, Hb S/E)[§]
    - Sickle/hemoglobin D Disease (FSD, Hb D/D)[§]

Lysosomal Storage Disorders:

- Mucopolysaccharidosis Type I*
- Glycogen Storage Disease Type II (Pompe)*

Organic acid conditions

- 2-Methylbutyrylglycinuria[§]
- 2-Methyl-3-Hydroxybutyric Aciduria[§]
- 3-Hydroxy-3-Methyglutaric Aciduria*
- 3-Methylcrotonyl-CoA Carboxylase Deficiency*
- 3-Methylglutaconic Aciduria[§]
- β-Ketothiolase Deficiency*
- Glutaric Acidemia Type I*
- Holocarboxylase Synthase Deficiency*
- Isobutyrylglycinuria[§]
- Isovaleric Acidemia*
- Malonic acidemia[§]
- Methylmalonic Acidemia (Cobalamin Disorders)*
- Methylmalonic Acidemia (Methylmalonyl-CoA Mutase)*
- Methylmalonic Acidemia with Homocystinuria[§]
- Propionic Acidemia*

Other conditions

- Biotinidase Deficiency*
- Classic Galactosemia*
- Critical Congenital Heart Disease[¥]
- Cystic Fibrosis*
- Galactoepimerase Deficiency[§]
- Galactokinase Deficiency[§]
- Hearing Loss[¥]
- Severe Combined Immunodeficiencies*
- T-cell Related Lymphocyte Deficiencies[§]
- Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1*
- X-linked Adrenoleukodystrophy*

*Core condition on the RUSP, screening conducted at NCSLPH

[¥]Core condition on the RUSP, screening conducted at point of care

[§]Secondary condition on the RUSP, screening conducted at NCSLPH