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 disorder*
- 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 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[§]
- 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)[§]

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 1*
- 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 immunodeficiency (SCID)*
- T-cell related lymphocyte deficiencies§

*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