Introduction

What is Newborn Screening?

Newborn Screening is a comprehensive public health system designed to prevent severe and potentially lethal outcomes from a variety of congenital disorders. The newborn screening system includes:

– Education to Hospitals, Providers, and Families
– Screening of all newborn infants
– Follow-up of abnormal findings
– Evaluation, Diagnosis, and Treatment
– Lifelong Management of identified conditions

All infants born in this state are required by the *South Carolina Code of Laws* (sections 44-30-37 and 44-37-35) to be screened in accordance with SC Regulation 61-80, promulgated by the Board of the Department of Health and Environmental Control (DHEC). This regulation is further defined by *Official Departmental Instructions* that specify the roles and responsibilities of each entity involved in the newborn screening process.

For the lab portion of the screening test, a blood spot specimen is to be collected from each infant born in SC. The specimen should ideally be collected between 24 and 48 hours of age and sent to the SC DHEC Public Health Laboratory (PHL) within 24 hours of collection. At present, infants are tested for select metabolic, hormone/enzyme, immune, and genetic disorders. The specific disorders on the test panel are included in this manual.

Purpose

The purpose of newborn screening is to identify infants at risk and in need of more definitive testing. **As with any laboratory screening test, both false positive and false negative results are possible. Initial screening test results are insufficient information to base definitive diagnosis or treatment.**

This manual uses terminology consistent with the American College of Medical Genetics (ACMG) report "*Newborn Screening: Towards a Uniform Screening Panel and System*," Genetic Med 2006; 8 (5) Supple: S12-S252. Tests for other disorders may be added in the future.

Select Conditions Table

The table below shows the number of infants born with the most common detectable newborn screening conditions found in South Carolina in 2022:

Select Conditions	# Of Infants Diagnosed with a Newborn Screening Finding in 2022
Hemoglobin Traits & Carriers	2085
Hemoglobin Diseases	65
Congenital Hypothyroidism (CH)	54
Cystic Fibrosis (CF)	15
Congenital Adrenal Hyperplasia (CAH)	5
Biotinidase Deficiency	2
Classic Galactosemia	2
Phenylketonuria (PKU)	2
Late Onset Pompe Disease	2
Spinal Muscular Atrophy (SMA)	2
Medium Chain Acyl CoA Dehydrogenase Deficiency (MCAD)	1
Carnitine Palmitoyl Transferase type 1 Deficiency (CPT 1A)	1
Glutaric Acidemia type 1 (GA-1)	1

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- Metabolic Disorders - Amino Acid Metabolism
    - Phenylketonuria (PKU)
        - Benign Hyperphenylalaninemia (H-PHE)
        - Defect of Biopterin Cofactor Biosynthesis (BIOPT-BS)
        - Defect of Biopterin Cofactor Regeneration (BIOPT-BS)
    - Homocystinuria (HCY)
        - Hypermethioninemia (MET)
    - Maple Syrup Urine Disease (MSUD)
    - Citrullinemia I (CIT I)
        - Citrullinemia II (CIT II)
    - Argininosuccinic Aciduria (ASA) - *PENDING*
    - Argininemia aka Arginase deficiency (ARG) - *PENDING*
    - Tyrosinemia I (TYR I)
        - Tyrosinemia II (TYR II)
        - Tyrosinemia III (TYR III)

- Carbohydrate and Lysosomal Metabolism Disorders
    - Classic Galactosemia (GALT)
    - Duarte variant Galactosemia
    - Galactokinase Deficiency (GALK)
    - Galactose Epimerase Deficiency (GALE)
    - Glycogen Storage Disease Type II (Pompe)
    - Mucopolysaccharidosis Type 1 (MPS 1)
    - Krabbe Disease - NEW

- Organic Acid Metabolism Disorders
    - Propionic Acidemia (PROP)
    - Malonic Acidemia (MAL)
    - Methylmalonic Acidemia - CoA Mutase Deficiency (MUT)
    - Methylmalonic Acidemia - Vitamin B 12 Disorders (Cobalamin A, B)
    - Methylmalonic Acidemia with Homocystinuria - (Cobalamin C, D, F)
    - *Iso-butyryl-CoA dehydrogenase deficiency (IBG)*
    - Isovaleric Acidemia (IVA)
    - 2-Methylbutyrylglycinuria (2MBG)

- 3-methylcrotonyl CoA Carboxylase Deficiency (3-MCC)
- Beta-ketothiolase Deficiency (βKT)
- 2-methyl-3-OH-butyric Aciduria (2M3HBA)
- 3-hydroxy-3-methylglutaric aciduria (HMG)
- 3-methylglutaconic aciduria (3MGA)
- Multiple Carboxylase Deficiency (MCD)
- Glutaric Acidemia Type I (GA I)

- Fatty Acid Oxidation Disorders
  - Medium Chain Acyl CoA Dehydrogenase Deficiency (MCAD)
    - Medium Chain ketoacyl CoA Thiolase deficiency (MCAT)
  - *Short Chain acyl CoA Dehydrogenase Deficiency* (*SCAD*)
  - Medium/Short Chain 3-OH Acyl CoA Dehydrogenase Deficiency (M/SCHAD)
  - Dienoyl co-A Reductase Deficiency (DE RED)
  - Long Chain 3-OH Acyl CoA Dehydrogenase Deficiency (LCHAD)
  - Trifunctional Protein Deficiency (TFP)
  - Very Long Chain Acyl CoA Dehydrogenase Deficiency (VLCAD)
  - Glutaric acidemia type II (GA II)
  - Carnitine Palmitoyl transferase I Deficiency (CPT IA)
  - Carnitine Palmitoyl transferase II Deficiency (CPT II)
  - Carnitine/Acylcarnitine Translocase Deficiency (CACT)
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  - Congenital Hypothyroidism (CH)
  - Congenital Adrenal Hyperplasia (CAH)
  - Biotinidase Deficiency (BIOT)

- Hemoglobin Disorders
  - Sickle Cell Disease (Hgb SS)
  - Sickle C Disease (Hgb S/C)
  - Sickle Beta Thalassemia (Hgb S/ß Th)
  - Various Hemoglobin Disorders and Traits
  - Sickle Cell Foundation Contacts in South Carolina

- Other Genetic Disorders
  - Cystic Fibrosis (CF)
  - Spinal Muscular Atrophy (SMA)
  - Severe Combined Immunodeficiency (SCID)
    - T cell related immune disorders
  - X-linked Adrenoleukodystrophy (X-ALD) - *PENDING*
  - Hearing Loss (HL)*
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- Copy of Parental Statement of Religious Objection Form - DHEC 1804
- Resources and Acknowledgments