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 SC Department of Public Health (DPH). 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 tests, a blood spot specimen shall 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 DPH Public Health Laboratory (PHL) within 24 hours of collection. At present, infants are tested for multiple 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 commonly found newborn screening conditions in South Carolina in 2023:

Select Conditions	# Of Infants Diagnosed with a Newborn Screening Finding in 2023
Hemoglobin Traits & Carriers	2163
Hemoglobin Diseases	55
Congenital Hypothyroidism (CH)	30
Spinal Muscular Atrophy (SMA)	7
Cystic Fibrosis (CF)	4
Congenital Adrenal Hyperplasia (CAH)	3
Classic Galactosemia	3
Medium Chain Acyl CoA Dehydrogenase Deficiency (MCAD)	2
Phenylketonuria (PKU)	2
Very Long Chain Acyl CoA Dehydrogenase Deficiency (VLCAD)	1
Isovaleric Acidemia (IVA)	1
Propionic Acidemia (PA)	1
3-MCC	1

Table of Contents

- 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 I)
  - Carnitine Palmitoyl transferase II Deficiency (CPT II)
  - Carnitine/Acylcarnitine Translocase Deficiency (CACT)
  - Carnitine Uptake/Transport Defect (CUD)

- Endocrine (Hormone) and Enzyme Disorders
  - 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)
  - Hearing Loss (HL)*
  - Critical Congenital Heart Defects (CCHD)*

- DPH Newborn Screening Contact Information
- Newborn Screening Brochures and Forms
- Instructions for Completing NBS Collection Form - DPH 1327
- Best Specimen Collection Timing by Disorder
- Criteria for Notification of Abnormal Results
    - Weekdays
    - Saturdays/Select holidays

- Referral Sources and Medical Specialists Contact Information

- Newborn Screening Law and Regulation
    - Appendix A - Religious Objection Form - DPH 1804
    - Appendix B - Information Release Form - DPH 1878
    - Appendix C - Blood Sample Storage Options Form - DPH 1812

- Instructions to Complete DPH 1804 Religious Objection form
- Copy of Parental Statement of Religious Objection Form - DPH 1804
- Resources and Acknowledgments