Introduction

All infants born in South Carolina are required by law to be screened in accordance with the regulation 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. A blood spot specimen is to be collected from each infant born in SC, preferably between 24 and 48 hours of age, and sent to the DHEC Bureau of Laboratories (BOL) within 24 hours of collection. At present infants are tested for certain metabolic, hormone/enzyme, and genetic disorders. The specific disorders included on the test panel are listed below. This list uses terminology consistent with the American College of Medical Genetics report "Newborn Screening: Towards a Uniform Screening Panel and System," Genet Med 2006; 8 (5) Suppl: S12-S252.

Metabolic Disorders:
<u>Amino Acid Metabolism Disorders</u>
Phenylketonuria (PKU)
Benign Hyperphenylalaninemia
Defect of Biopterin Cofactor Biosynthesis
Defect of Biopterin Cofactor Regeneration
Maple Syrup Urine Disease (MSUD)
Homocystinuria
Hypermethioninemia
Citrullinemia I
Citrullinemia II
Argininosuccinic Aciduria
Tyrosinemia I
Tyrosinemia II
Tyrosinemia III

<u>Carbohydrate Metabolism Disorders</u>
Classical Galactosemia (GALT)
Galactokinase Deficiency (GALK)
Galactose Epimerase Deficiency (GALE)

<u>Organic Acid Metabolism Disorders</u>
Propionic Acidemia (PA)
Malonic Acidemia (MA)
Methylmalonic Acidemia—Co-A Mutase Deficiency (MUT)
Methylmalonic Acidemia—Vit B 12 Disorders (CBL A,B)
Methylmalonic Acidemia—Other (CBL C,D)
Isovaleric Acidemia (IVA)
2-methylbutyryl coA Dehydrogenase Deficiency (2-MBCD)
3-methylcrotonyl coA Carboxylase Deficiency (3-MCC)
ß-ketothiolase Deficiency (SKAT)
3-methyl-3-OH-glutaryl coA Lyase Deficiency (HMGL)
3-methyl-glutaconyl coA Hydratase Deficiency

Multiple Carboxylase Deficiency (MCD)
Glutaric Aciduria I (GA I)
2-methyl-3-OH-butyric Aciduria (2M3HBA)

Fatty Acid Metabolism Disorders
Medium Chain Acyl coA Dehydrogenase Deficiency (MCAD)
Medium/Short Chain 3-OH acyl coA Dehydrogenase Deficiency (M/SCHAD)
Dienoyl co-A Reductase Deficiency
Long Chain 3-OH acyl coA Dehydrogenase Deficiency (LCHAD)
Trifunctional Protein Deficiency (TFP)
Very Long Chain acyl coA Dehydrogenase Deficiency (VLCAD)
Multiple acyl coA Dehydrogenase Deficiency (MAD/GA II)
Medium Chain Ketoacyl CoA Thiolase Deficiency (MCKAT)
Carnitine Uptake/Transport Defect (CUD)
Carnitine Palmitoyltransferase I Deficiency (CPT I)
Carnitine Palmitoyltransferase II Deficiency (CPT II)
Carnitine/Acylcarnitine Translocase Deficiency (CAT)

Hormone and Enzyme Disorders
Primary Congenital Hypothyroidism
Congenital Adrenal Hyperplasia (CAH)
Biotinidase Deficiency

Other Genetic Disorders
Cystic Fibrosis
Sickle Cell Disease
Sickle C Disease
Sickle ß Thalassemia
Variant Hemoglobinopathy Disorders and Traits (including sickle cell trait)
Severe Combined Immunodeficiency (SCID) and related disorders

Tests for other disorders may be added in the future. The table below shows an estimate of the number of infants born with a disorder detectable by newborn screening in SC each year.

Disorder	Projected Number of Infants Born with Disorder In SC Per Year
PKU	3
Galactosemia	1
MCAD	3
Other disorders of amino acid, fatty acid or organic acid metabolism	4
Primary congenital hypothyroidism	15
CAH	3
Biotinidase deficiency	1
Hemoglobinopathy disorders (including sickle cell disease)	100

Hemoglobinopathy traits (including sickle cell trait)	2000
Cystic fibrosis	11
SCID	1

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