

Introduction

All infants born in South Carolina are required by law to be screened for metabolic disorders in accordance with the regulation promulgated by the Board of the Department of Health and Environmental Control. This regulation is further defined by Official Departmental Instructions that specify the roles and responsibilities of each entity involved in the newborn screening process. 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:

Amino Acid Metabolism Disorders

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

Carbohydrate Metabolism Disorders

Classical Galactosemia (GALT)
Galactokinase Deficiency (GALK)
Galactose Epimerase Deficiency (GALE)

Organic Acid Metabolism Disorders

Propionic Acidemia (PA)
Methylmalonic Acidemia—Co-A Mutase Deficiency (MUT)
Methylmalonic Acidemia—Vit B 12 Disorders (CBL A,B)
Methylmalonic Acidemia—Other (CBL C,D)
Malonic Acidemia (MA)
Isobutyryl coA Dehydrogenase Deficiency (IBCD)
Isovaleric Acidemia (IVA)
2-methylbutyryl coA Dehydrogenase Deficiency (2-MBCD)
3-methylcrotonyl coA Carboxylase Deficiency (3-MCC)
 β -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 Chain Ketoacyl CoA Thiolase Deficiency (MCKAT)
Short Chain acyl coA Dehydrogenase Deficiency (SCAD)
Medium and Short Chain 3-OH acyl coA Dehydrogenase Deficiency (M/SCHAD)
Dienoyl coA 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)
Carnitine Uptake/Transport Defect (CUD)
Carnitine Palmitoyltransferase I Deficiency (CPT I)
Carnitine Palmitoyltransferase II Deficiency (CPT II)
Carnitine/Acylcarnitine Translocase Deficiency (CACT)

Hormone and Enzyme Disorders:

Congenital Hypothyroidism
Congenital Adrenal Hyperplasia (CAH)
Biotinidase Deficiency

Other Genetic Disorders:

Cystic Fibrosis (CF)
Sickle Cell Disease
Sickle C Disease
Sickle β Thalassemia
Variant Hemoglobinopathy Disorders and Traits (including sickle cell trait)

Tests for other disorders may be added in the future.

All of these disorders can be detected during early infancy, and the infant can be treated to prevent devastating outcomes. South Carolina has screened for PKU since 1965 and for congenital hypothyroidism since 1978. The Neonatal Screening law was amended to add screening for Hemoglobinopathies, effective July 1987. The screening panel was expanded in October 1992 to include testing for galactosemia and CAH. Screening for MCAD began in August 2000. A major test panel expansion occurred in November 2004 when testing for cystic fibrosis, biotinidase deficiency and other disorders of amino acid, fatty acid and organic acid metabolism that can be found by tandem mass spectrometry was added.

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
Congenital hypothyroidism	10
CAH	3
Biotinidase deficiency	1
Hemoglobinopathy disorders (including sickle cell disease)	100
Hemoglobinopathy traits (including sickle cell trait)	2000
Cystic fibrosis	11

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.