TEXAS Health and Human Services | Texas Department of State Health Services

BIOCHEMISTRY AND GENETICS BRANCH
LABORATORY SERVICES
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Newborn Screening Disorders Screened by Time Criticality

Time Critical Disorders

Screening Test	RUSP Category
CAH	
Congenital Adrenal Hyperplasia (CAH)	Core Condition
GALACTOSEMIA	
Classical Galactosemia (GALT)	Core Condition
AMINO ACID DISORDERS	
Argininosuccinic Aciduria (ASA)	Core Condition
Citrullinemia Type 1 (CIT)	Core Condition
Maple Syrup Urine Disease (MSUD)	Core Condition
Citrullinemia Type 2 (CIT II)	Secondary Condition
FATTY ACID DISORDERS	
Long Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)	Core Condition
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	Core Condition
Trifunctional Protein Deficiency (TFP)	Core Condition
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	Core Condition
Carnitine Acylcarnitine Translocase Deficiency (CACT)	Secondary Condition
Carnitine Palmitoyltransferase Type I Deficiency (CPT I)	Secondary Condition
Carnitine Palmitoyltransferase Type II Deficiency (CPT II)	Secondary Condition
Glutaric Acidemia Type II (GA2)	Secondary Condition
ORGANIC ACID DISORDERS	
3-Hydroxy-3-Methyglutaric Aciduria (HMG)	Core Condition
Beta-Ketothiolase Deficiency (BKT)	Core Condition
Glutaric Acidemia Type I (GA I)	Core Condition
Isovaleric Acidemia (IVA)	Core Condition
Methylmalonic Acidemia (Methylmalonyl-CoA mutase)	Core Condition
Holocarboxylase Synthase Deficiency (MCD)	Core Condition
Propionic Acidemia (PROP)	Core Condition
Methylmalonic Acidemia with Homocystinuria (Cbl C, D)	Secondary Condition
SPINAL MUSCULAR ATROPHY	
Spinal Muscular Atrophy (SMA) due to homozygous deletion of exon 7 in SMN1 and ≤ 3 copies of SMN2	Core Condition

Time Sensitive Disorders

Screening Test	RUSP Category
BIOTINIDASE DEFICIENCY	
Biotinidase Deficiency (BIOT)	Core Condition
HYPOTHYROIDISM	
Primary Congenital Hypothyroidism (CH)	Core Condition
Screening Test	**RUSP Category**

CYSTIC FIBROSIS	
Cystic Fibrosis (CF)	Core Condition
SCID	
Severe Combined Immunodeficiency (SCID)	Core Condition
T-cell related lymphocyte deficiencies	Secondary Condition
X-ALD	
X-linked Adrenoleukodystrophy (X-ALD)	Core Condition
HEMOGLOBINOPATHIES	
S,C disease	Core Condition
S,S Disease (Sickle Cell Anemia)	Core Condition
S, Beta-Thalassemia	Core Condition
Various Other Hemoglobinopathies	Secondary Condition
AMINO ACID DISORDERS	
Homocystinuria (HCY)	Core Condition
Classical Phenylketonuria (PKU)	Core Condition
Tyrosinemia Type I (TYR I)	Core Condition
Argininemia (ARG)	Secondary Condition
Benign hyperphenylalaninemia (H-PHE)	Secondary Condition
Biopterin defect in cofactor biosynthesis (BIOPT BS)	Secondary Condition
Biopterin defect in cofactor regeneration (BIOPT REG)	Secondary Condition
Hypermethioninemia (MET)	Secondary Condition
Tyrosinemia Type II (TYR II)	Secondary Condition
Tyrosinemia Type III (TYR III)	Secondary Condition
FATTY ACID DISORDERS	
Carnitine Uptake Defect (CUD)/Carnitine transport defect	Core Condition
2,4 Dienoyl-CoA Reductase Deficiency (DE-RED)	Secondary Condition
Medium Chain Ketoacyl-CoA Thiolase Deficiency (MCKAT)	Secondary Condition
Medium/Short Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHAD)	Secondary Condition
Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)	Secondary Condition
ORGANIC ACID DISORDERS	
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	Core Condition
Methylmalonic Acidemia (Cobalamin disorders Cbl A, B)	Core Condition
2-Methylbutyrylglycinuria (2MBG)	Secondary Condition
2-Methyl-3-Hydroxybutyric Aciduria (2M3HBA)	Secondary Condition
3-Methylglutaconic Aciduria (3MGA)	Secondary Condition
Isobutyrylglycinuria (IBG)	Secondary Condition
Malonic Acidemia (MAL)	Secondary Condition
SPINAL MUSCULAR ATROPHY	
Spinal Muscular Atrophy (SMA) due to homozygous deletion of exon 7 in SMN1 and ≥ 4 copies of SMN2	Core Condition

Core Conditions on RUSP not currently screened in Texas: Glycogen Storage Disease Type II (Pompe), Mucopolysaccharidosis Type I (MPS I), Mucopolysaccharidosis Type II (MPS II), and Guanidinoacetate methyltransferase deficiency (GAMT)

Secondary Conditions on RUSP not currently screened in Texas: Galactoepimerase deficiency, Galactokinase deficiency