



# Newborn Screening Disorders: Categorized by Time Criticality

## Time Critical Disorders: Immediate action needed.

Screening Test	RUSP Category
<b>CAH</b>	
Congenital Adrenal Hyperplasia (CAH)	Core Condition
<b>GALACTOSEMIA</b>	
Classical Galactosemia (GALT)	Core Condition
<b>AMINO ACID DISORDERS</b>	
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
<b>FATTY ACID DISORDERS</b>	
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
<b>ORGANIC ACID DISORDERS</b>	
3-Hydroxy-3-Methylglutaric 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
<b>OTHER</b>	
Spinal Muscular Atrophy (SMA) due to homozygous deletion of exon 7 in SMN1 and $\leq 3$ copies of SMN2	Core Condition
Infantile Krabbe Disease (Krabbe)	Core Condition
Glycogen Storage Disease Type II (Pompe)	Core Condition

## Time Sensitive Disorders: Prompt treatment required.

Screening Test	RUSP Category
<b>BIOTINIDASE DEFICIENCY</b>	
Biotinidase Deficiency (BIOT)	Core Condition
<b>HYPOTHYROIDISM</b>	
Primary Congenital Hypothyroidism (CH)	Core Condition

Screening Test	RUSP Category
<b>CYSTIC FIBROSIS</b>	
Cystic Fibrosis (CF)	Core Condition
<b>SCID</b>	
Severe Combined Immunodeficiency (SCID)	Core Condition
T-cell related lymphocyte deficiencies	Secondary Condition
<b>X-ALD</b>	
X-linked Adrenoleukodystrophy (X-ALD)	Core Condition
<b>HEMOGLOBINOPATHIES</b>	
S,C disease	Core Condition
S,S Disease (Sickle Cell Anemia)	Core Condition
S, Beta-Thalassemia	Core Condition
Various Other Hemoglobinopathies	Secondary Condition
<b>AMINO ACID DISORDERS</b>	
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
<b>FATTY ACID DISORDERS</b>	
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
<b>ORGANIC ACID DISORDERS</b>	
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
<b>OTHER</b>	
Spinal Muscular Atrophy (SMA) due to homozygous deletion of exon 7 in SMN1 and $\geq 4$ copies of SMN2	Core Condition
Mucopolysaccharidosis Type I (MPS I)	Core Condition
Mucopolysaccharidosis Type II (MPS II)	Core Condition

Core Conditions on RUSP not currently screened in Texas: Guanidinoacetate methyltransferase deficiency (GAMT)

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