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

NBS Provider Resources

Time-Sensitive Newborn Screening Disorders

Time-Sensitive Disorders: Prompt treatment required.

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

Screening Test	RUSP Category
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
OTHER	
Spinal Muscular Atrophy (SMA) due to homozygous deletion of exon 7 in SMN1 and **≥ 4 copies of SMN2.** SMN2 copy number directly correlates with severity.	Core Condition
Mucopolysaccharidosis Type I (MPS I)	Core Condition
Mucopolysaccharidosis Type II (MPS II)	Core Condition
Guanidinoacetate Methyltransferase Deficiency (GAMT)	Core Condition

Core Conditions on RUSP not currently screened in Texas: Duchenne muscular dystrophy, Metachromatic leukodystrophy.
Secondary Conditions on RUSP not currently screened in Texas: Galactoepimerase deficiency, Galactokinase deficiency.