



Time-Critical Newborn Screening Disorders

Time-Critical Disorders: Immediate action needed.

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-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
OTHER	
Spinal Muscular Atrophy (SMA) due to homozygous deletion of exon 7 in SMN1 and ≤ 3 copies of SMN2. SMN2 copy number directly correlates with severity.	Core Condition
Infantile Krabbe Disease (Krabbe)	Core Condition
Glycogen Storage Disease Type II (Pompe)	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.