

Time Critical Disorders:

The following table is from the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children’s (ACHDNC) recommendations on timeliness in newborn screening and was created based on the Society for Inherited Metabolic Disorders (SIMD) position statement and expert opinion from metabolic geneticists, hematologists, endocrinologist, and pulmonologists.

Organic Acid Conditions	Fatty Acid Oxidation Disorders	Amino Acid Disorders	Endocrine Disorders	Other Disorders
Propionic acidemia - PROP	Medium-chain acyl-CoA dehydrogenase deficiency - MCAD	Argininosuccinic aciduria - ASA	Congenital adrenal hyperplasia - CAH	Classic galactosemia - GALT
Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT	Very long-chain acyl-CoA dehydrogenase deficiency - VLCAD	Citrullinemia, type I - CIT		Glycogen Storage Disorder, type II- Infantile Pompe
Isovaleric acidemia - IVA	Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency - LCHAD	Maple syrup urine disease - MSUD		Infantile Krabbe Disease
3-Hydroxy-3-methylglutaric aciduria - HMG	Trifunctional protein deficiency - TFP			
Holocarboxylase synthase deficiency - MCD	Glutaric acidemia, type II- GA II			
β-Ketothiolase deficiency - BKT	Carnitine acylcarnitine translocase deficiency—CACT			
Glutaric acidemia, type I – GA 1	Carnitine palmitoyltransferase, type II deficiency – CPT II			