

**Recommended Uniform Screening Panel
Core Conditions
(DRAFT March 2016)**

ACMG Code	Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
		Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder			
PROP	Propionic Acidemia	X					
MUT	Methylmalonic Acidemia (methylmalonyl-CoA mutase)	X					
Cbl A,B	Methylmalonic Acidemia (Cobalamin disorders)	X					
IVA	Isovaleric Acidemia	X					
3-MCC	3-Methylcrotonyl-CoA Carboxylase Deficiency	X					
HMG	3-Hydroxy-3-Methylglutaric Aciduria	X					
MCD	Holocarboxylase Synthase Deficiency	X					
βKT	β-Ketothiolase Deficiency	X					
GA1	Glutaric Acidemia Type I	X					
CUD	Carnitine Uptake Defect/Carnitine Transport Defect		X				
MCAD	Medium-chain Acyl-CoA Dehydrogenase Deficiency		X				
VLCAD	Very Long-chain Acyl-CoA Dehydrogenase Deficiency		X				
LCHAD	Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency		X				
TFP	Trifunctional Protein Deficiency		X				
ASA	Argininosuccinic Aciduria			X			
CIT	Citrullinemia, Type I			X			
MSUD	Maple Syrup Urine Disease			X			
HCY	Homocystinuria			X			
PKU	Classic Phenylketonuria			X			
TYR I	Tyrosinemia, Type I			X			
CH	Primary Congenital Hypothyroidism				X		
CAH	Congenital adrenal hyperplasia				X		
Hb SS	S,S Disease (Sickle Cell Anemia)					X	
Hb S/BTh	S, βeta-Thalassemia					X	
Hb S/C	S,C Disease					X	
BIOT	Biotinidase Deficiency						X
CCHD	Critical Congenital Heart Disease						X
CF	Cystic Fibrosis						X
GALT	Classic Galactosemia						X
GSD II	Glycogen Storage Disease Type II (Pompe)						X
HEAR	Hearing Loss						X
SCID	Severe Combined Immunodeficiencies						X
MPS I	Mucopolysaccharidosis Type 1						X
X-ALD	X-linked Adrenoleukodystrophy						X