

Newborn Screening Mandated Screening Panel and Mandated Follow-up Panel

Conditions Mandated for Screening and Follow-up by NSEP

PKU	Phenylketonuria
MSUD	Maple Syrup Urine Disease
Hb SS-Disease	Sickle Cell Anemia
CH	Congenital Hypothyroidism
CAH	Congenital Adrenal Hyperplasia
GALT	Transferase Deficient Galactosemia (Classical)
GAA	Glycogen Storage Disease Type II (Pompe Disease)
MPS I	Mucopolysaccharidosis type I (Hurler Syndrome)
X-ALD	X-linked adrenoleukodystrophy
SMA	Spinal Muscular Atrophy – Effective 3/1/2019

Conditions Mandated for Follow-up by NSEP

Acylcarnitine Disorders

Organic Acid Disorders

IVA	Isovaleric Acidemia
GA I	Glutaric Acidemia Type I
HMG	3-Hydroxy 3-Methyl Glutaric Aciduria
MCD	Multiple Carboxylase Deficiency
MUT	Methylmalonic Acidemia (Mutase Deficiency)
3MCC	3-Methylcrotonyl-CoA Carboxylase Deficiency
Cbl A,B	Methylmalonic Acidemia (Cbl A,B)
PROP	Propionic Acidemia
BKT	Beta-Ketothiolase Deficiency

Fatty Oxidation Disorders

MCAD	Medium-Chain Acyl-CoA Dehydrogenase Deficiency
VLCAD	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency
LCHAD	Long-Chain L-3-Hydroxy Acyl-CoA Dehydrogenase Deficiency
TFP	Trifunctional Protein Deficiency
CUD	Carnitine Uptake Defect

Amino Acid Disorders

HCY	Homocystinuria
CIT	Citrullinemia
ASA	Argininosuccinic Aciduria
TYR I	Tyrosinemia Type I

Hemoglobinopathies

Hb SC-Disease	Sickle-C Disease
Hb S	S-Beta Thalessemia



Lysosomal Storage Disorders

GLA	Fabry Deficiency
ASM	Niemann-Pick A/B
GBA	Gaucher Deficiency
GALC	Globoid cell leukodystrophy (Krabbe Disease)

Others

BIO	Biotinidase Deficiency
CF	Cystic Fibrosis
SCID	Severe Combined Immunodeficiency

Point of Care Testing

CCHD	Critical Congenital Heart Defects
HEAR	Newborn Hearing Screening