

Newborn Screening Mandated Screening Panel and Mandated Follow-up Panel

Conditions Mandated for Screening and Follow-up by NSFP

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) – Effective 2/1/17

X-ALD X-linked adrenoleukodystrophy – **Effective 4/1/17**

Conditions Mandated for Follow-up by NSFP

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

<u>Hemoglobinopathies</u>

Hb SC-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