Newborn Screening Mandated Screening Panel and Supplemental Mandated Screening Panel

Conditions Mandated for Screening and Follow-up by PA Newborn Screening Program

PKU	Phenylketonuria
MSUD	Maple Syrup Urine Disorder
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

Supplemental Conditions Mandated for Screening by PA Submitters and Follow-up by PA Newborn Screen Program

Acylcarnitine Disorders

Organic Acid Disorders

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

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

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Amino Acid Disorders

HCY	Homocystinuria
CIT	Citrullinemia
ASA	Argininosuccinic Acidemia
TYR	Tyrosinemia Type I

Hemoglobinopathies

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

Lysosomal Storage Disorders

GALC	Globoid Cell Leukodystrophy (Krabbe Disease)
MPS II	Mucopolysaccharidosis Type II (Hunter Syndrome)

Others

BIO	Biotinidase Deficiency
CF	Cystic Fibrosis
SCID	Severe Combined Immunodeficiency
GAMT	Guanidinoacetate Methyltransferase Deficiency

Point of Care Testing

CCHD	Critical Congenital Heart Defects
HEAR	Newborn Hearing Screening

Medical Disorders that can be Detected in the Differential Diagnosis of a Core Disorder

Organic Acid Disorders

CBL C,D	Methylmalonic Acidemia With Homocystinuria
MAL	Malonic Acidemia
IBG	Isobutyrylglycinuria
2 MBG	2-Methylbutyrylglycinuria
3 MBA	3-Methylglutaconic Aciduria
2M3HBA	2-Methyl-3-Hydroxybutyric Aciduria

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Fatty Acid Oxidation Disorders

SCAD	Short-Chain Acyl-CoA Dehydrogenase Deficiency
M/SCHAD	Medium/Short-Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency
GA2	Glutaric Acidemia Type II
MCAT	Medium-Chain Ketoacyl-CoA Thiolase Deficiency
DE RED	2,4 Dienoyl-CoA Reductase Deficiency
CPT IA	Carnitine Palmitoyltransferase Type I Deficiency
CPT II	Carnitine Palmitoyltransferase Type II Deficiency
CACT	Carnitine Acylcarnitine Translocase Deficiency

Amino Acid Disorders

ARG	Argininemia
CIT II	Citrullinemia Type II
MET	Hypermethioninemia
H-PHE	Benign Hyperphenylalaninemia
BIOPT (BS)	Biopterin Defect in Cofactor Biosynthesis
BIOPT (REG)	Biopterin Defect in Cofactor Regeneration
TYR II	Tyrosinemia Type II
Tyr III	Tyrosinemia Type III

Hemoglobinopathies

HGB D, E, O	Hemoglobin Point Mutation Testing Detects Hemoglobin D, E, and O

Other Disorders

GALE	Galactoepimerase Deficiency
GALK	Galactokinase Deficiency

*T-Cell related lymphocyte deficiencies may be identified by screening for SCID.

PLEASE NOTE: Birthing submitters may opt to screen for additional conditions. Please consult with your birthing hospital or midwife for the specific conditions that will be screened.