Pennsylvania Newborn Screening and Follow-Up Program (NSFP)

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)**

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**

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**
MPS I	**Mucopolysaccharidosis type I (Hurler Syndrome)**
GALC	**Globoid cell luekodystrophy (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**