

## Save Babies Through Screening Foundation's Recommendations for Newborn Screening

All the following diseases and conditions are currently detectable through newborn screening. At the present time, the same filter paper blood spot specimen can test for more than 50 diseases (using several different screening methodologies/equipment).

### Core Panel as recommended by the American College of Medical Genetics (ACMG)

3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)  
3-OH 3-CH<sub>3</sub> glutaric aciduria (HMG)  
Argininosuccinic acidemia (ASA)  
Beta-Ketothiolase deficiency (BKT)  
Biotinidase deficiency (BIOT)  
Carnitine uptake defect (CUD)  
Citrullinemia (CIT)  
Classical galactosemia (GALT)  
Congenital adrenal hyperplasia (*21-hydroxylase deficiency*) (CAH)  
Congenital hypothyroidism (CH)  
Cystic fibrosis (CF)  
Glutaric acidemia type I (GA 1)  
Hb S/C disease (Hb S/C)  
Hb S/ $\beta$ -thalassemia (Hb S/ $\beta$ Th)  
Homocystinuria (*due to CBS deficiency*) (HCY)  
Isovaleric acidemia (IVA)  
Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHADD)  
Maple syrup disease (MSUD)  
Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)  
Methylmalonic acidemia (*Cbl A,B*) (Cbl A, B)  
Methylmalonic acidemia (*mutase deficiency*) (MUT)  
Multiple carboxylase deficiency (MCD)  
Phenylketonuria (PKU)  
Propionic acidemia (PROP)  
Sickle cell anemia (*Hb SS disease*) Hb (SS)  
Trifunctional protein deficiency (TFP)  
Tyrosinemia type I (TYR I)  
Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD)

### Secondary Targets as recommended by the American College of Medical Genetics (ACMG)

2-Methyl 3-hydroxy butyric aciduria (2M3HBA)  
2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)  
3 Methylglutaconic aciduria (3MGA)  
Argininemia (ARG)  
Biotin cofactor biosynthesis, defects of (BIOPT BS)

Biopterin cofactor regeneration, defects of (BIOPT REG)  
Carnitine palmitoyltransferase I deficiency (*liver*) (CPT IA)  
Carnitine palmitoyltransferase II deficiency (CPT II)  
Carnitine: acylcarnitine translocase deficiency (CACT)  
Citrullinemia type II (CIT II)  
Dienoyl-CoA reductase deficiency (DE RED)  
Galactokinase deficiency (GALK)  
Galactose epimerase deficiency (GALE)  
Glutaric acidemia Type II (GA 2)  
Hypermethioninemia (MET)  
Hyperphenylalaninemia, benign (H-PHE)  
Isobutyryl-CoA dehydrogenase deficiency (IBG)  
Malonic acidemia (MAL)  
Medium/short-chain L-3-OH acyl-CoA dehydrogenase deficiency (M/SCHADD)  
Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)  
Methylmalonic acidemia (*Cbl C,D*) (Cbl C,D)  
Short-chain acyl-CoA dehydrogenase deficiency (SCADD)  
Tyrosinemia type II (TYR II)  
Tyrosinemia type III (TYR III)  
Variant Hb-pathies (*including HB E*) (Var Hb)

#### **Additional Disorders**

Unclassified Fatty Acid Oxidation Disorders (**U-FAOD**)

#### **Other Abnormal Profiles**

Hyperalimentation (TPN)  
Liver Disease  
Medium Chain Triglyceride (MCT) Oil Administration  
Presence of EDTA Anticoagulants in Blood Specimen  
Treatment with Benzoate, Pyvalic Acid, or Valproic Acid