



## Oregon Newborn Screening Program

What conditions are tested for Oregon babies?

### Amino Acid Disorders

- Argininemia (ARG)
- Argininosuccinic aciduria (ASA)
- Citrullinemia, type I (CIT)
- Citrullinemia, type II (CIT II)
- Phenylketonuria (PKU)
- Benign hyperphenlalaninemia (H-PHE)
- Homocystinuria (HCY)
- Hypermethioninemia (MET)
- Maple syrup urine disease (MSUD)
- Tyrosinemia, type I (TYR I)
- Tyrosinemia, type II (TYR II)
- Tyrosinemia, type III (TYR III)
- Biopterin defect in cofactor biosynthesis (BIOPT BS)
- Biopterin defect in cofactor regeneration (BIOPT REG)

### Endocrine Disorders

- Congenital adrenal hyperplasia (CAH)
- Primary congenital hypothyroidism (CH)

### Hemoglobin Disorders

- Hemoglobinopathies (Var Hb)
- S, Beta-thalassemia (Hb S/ $\beta$ Th)
- S, C disease (Hb S/C)
- Sickle cell anemia (Hb SS)

### Fatty Acid Oxidation Disorders

- Carnitine acylcarnitine translocase deficiency (CACT)
- Carnitine palmitoyltransferase I deficiency (CPT-IA)
- Carnitine palmitoyltransferase type II deficiency (CPT-II)
- Carnitine uptake defect (CUD)

- Glutaric acidemia, type II (GA-2)
- Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
- Trifunctional protein deficiency (TFP)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Medium/short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (M/SHAD)
- Medium-chain ketoacyl-CoA thiolase deficiency (MCAT)
- 2,4 Dienoyl-CoA reductase deficiency (DE RED)

### **Organic Acid Conditions**

- 2-Methyl-3-hydroxybutyric acidemia (2M3HBA)
- 2-Methylbutyrylglycinuria (2MBG)
- 3-Hydroxy-3-methylglutaric aciduria (HMG)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- 3-Methylglutaconic aciduria (3MGA)
- Beta-ketothiolase deficiency (BKT)
- Glutaric acidemia type I (GA1)
- Holocarboxylase synthetase deficiency (MCD)
- Isobutyrylglycinuria (IBG)
- Isovaleric acidemia (IVA)
- Malonic acidemia (MAL)
- Methylmalonic acidemia (cobalamin disorders) (Cbl A,B)
- Methylmalonic acidemia (methylmalonyl-CoA mutase deficiency) (MUT)
- Methylmalonic acidemia with homocystinuria (Cbl C, D, F)
- Propionic acidemia (PROP)

### **Lysosomal Storage Disorders (Oregon Only)**

- Pompe
- MPS-I
- Fabry
- Gaucher

### **Other Disorders**

- Biotinidase deficiency (BIOT)
- Galactosemia (GALT)
- Galactoepimerase Deficiency (GALE)
- Galactokinase Deficiency (GALK)
- Cystic fibrosis (CF) (IRT testing with reflex to a DNA panel)
- Severe combined immunodeficiency (SCID)