

## Disorders

### Amino Acid Disorders

- Phenylketonuria (PKU)
- Homocystinuria (HCY)
- Maple syrup urine disease (MSUD)
- Tyrosinemia (TYR)
- Citrullinemia (CIT)
- Argininosuccinic acidemia (ASA)

### Organic Acidemias

- Isovaleric acidemia (IVA)
- Glutaric acidemia type I (GAI)
- 3-Hydroxy 3-methyl glutaric aciduria (HMG)
- Multiple carboxylase deficiency (MCD)
- Methylmalonic acidemia - mutase deficiency (MUT)
- Methylmalonic acidemia - Cbl A, B
- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
- Propionic acidemia (PA)
- $\beta$ -Ketothiolase deficiency (BKT)

### Fatty Acid $\beta$ -Oxidation Defects

- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Long-chain 3-hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)
- Trifunctional protein deficiency (TFP)
- Carnitine uptake defect (CUD)

### Galactosemia (GALT)

### Biotinidase deficiency

### Endocrine Disorders

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

### Hemoglobinopathies

- Sickle cell disease (S/S)
- Sickle cell-hemoglobin C disease (S/C)
- Sickle cell- $\beta$ -thalassemia (S/beta-thal)

### Cystic fibrosis (CF)