

What will my baby be screened for?

AMINO ACIDEMIAS

- Argininosuccinic aciduria (ASA)*
- Citrullinemia, type 1 (CIT)*
- Homocystinuria (HCY)*
- Maple syrup urine disease (MSUD)*
- Classic phenylketonuria (PKU)*
- Tyrosinemia, type I (TYR-1) *

ORGANIC ACIDEMIAS

- Glutaric acidemia type I (GA-1)*
- 3-Hydroxy 3-methylglutaric aciduria (HMG)*
- Isovaleric acidemia (IVA)*
- 3-Methylcrotonyl-CoA carboxylase (3-MCC)*
- Methylmalonic acidemia - cobalamin disorders (Cbl-A,B) & methylmalonyl-CoA mutase deficiency (MUT) *
- β -Keto-thiolase (β KT)*
- Propionic acidemia (PROP)*
- Holocarboxylase synthetase deficiency (MCD) *

ENDOCRINE

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

FATTY ACID OXIDATION DISORDERS

- Carnitine uptake defect & Carnitine transport defect (CUD) *
- Long-chain L-3 hydroxyacyl-CoA dehydrogenase (LCHAD)*
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD) *
- Trifunctional protein deficiency (TFP)*
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD) *

HEMOGLOBINOPATHIES

- Sickle cell anemia (Hb SS)*
- Hemoglobin SC disease (Hb SC)*
- Sickle beta-thalassemia (Hb S β)*

OTHER

- Biotinidase deficiency (BIOT)*
- Cystic fibrosis (CF)*
- Classic galactosemia (GALT)*
- Severe combined immunodeficiencies (SCID)*