

Rhode Island Newborn Screening Program List of Conditions

Each baby born in Rhode Island is screened for the conditions listed below and for hearing loss. Other similar conditions may also be detected through this screening. This list is correct as of June 1, 2007 but may change as conditions are added to or removed from the testing panel. If you have any questions, please contact the Rhode Island Newborn Screening Program at (800) 942-7434.

- 3-Methylcrotonyl-CoA carboxylase deficiency
- Argininosuccinic acidemia
- Beta-ketothiolase deficiency
- Biotinidase deficiency
- Carnitine uptake defect
- Citrullinemia
- Congenital adrenal hyperplasia
- Congenital hypothyroidism
- Cystic fibrosis
- Galactosemia
- Glutaric acidemia type I
- Homocystinuria
- Hydroxymethylglutaric aciduria/HMG-CoA lyase deficiency
- Isovaleric acidemia
- Long-chain hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
- Maple syrup urine disease
- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
- Methylmalonic acidemias
- Multiple carboxylase deficiency
- Phenylketonuria (PKU)
- Propionic acidemia
- Sickle cell disease/hemoglobin disorders
- Trifunctional protein deficiency
- Tyrosinemia type I
- Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency