

Rhode Island currently screens babies for the 28 conditions listed below and hearing loss. The conditions are grouped into categories of endocrine, hemoglobin, metabolic and other conditions. Many serious effects of these conditions can be prevented with early detection and treatment. This list is current as of 7/2/2008.

Type of Condition	Condition Description & Treatment	Conditions Included in Newborn Screening		Year Screening Started in RI*
		Full Name of Condition	Abbreviation	
ENDOCRINE CONDITIONS	Babies with endocrine disorders do not make enough of certain types of hormones. This affects their growth and development. These conditions are treated with special medications	Congenital Hypothyroidism	CH	1976
		Congenital Adrenal Hyperplasia	CAH	7/1/1994
HEMOGLOBIN CONDITIONS	Hemoglobin conditions affect a baby's blood. They can lead to anemia (not enough red blood cells), slowed growth, pain, and damage to vital organs. Babies with hemoglobin disorders are treated with antibiotics because they can get very sick and even die from common infections. Screening targets the three most common conditions, but other hemoglobin conditions may be identified.	Sickle cell anemia	SS	5/1/1990
		Sickle/Beta-Thalassemia	S/Th	5/1/1990
		Sickle/Hemoglobin C disease	S/C	5/1/1990
METABOLIC CONDITIONS	<b>Amino Acid Conditions:</b> Babies with these conditions cannot digest certain amino acids (parts of proteins in our food). This can affect health, growth, and learning. Medications and special diets can help prevent these problems.	Phenylketonuria	PKU	1962
		Maple Syrup Urine Disease	MSUD	1965
		Homocystinuria	HCY	1965
		Citrullinemia	CIT	7/1/2006
		Argininosuccinic academia	ASA	7/1/2006
		Tyrosinemia type I	TYR1	7/1/2006
	<b>Fatty Acid Oxidation Conditions:</b> Babies with these conditions have problems making energy from fat in the body or from food. Serious health problems can occur if these conditions are not treated. Medications, special low fat diets, and feeding a newborn at least every 4 hours can help prevent these problems.	Medium-chain acyl-CoA dehydrogenase deficiency	MCAD	7/1/2001
		Very long-chain acyl-CoA dehydrogenase deficiency	VLCAD	7/1/2006
		Long-chain 3-OH acyl-CoA dehydrogenase deficiency	LCHAD	7/1/2006
		Trifunctional protein deficiency	TFP	7/1/2006
		Carnitine uptake defect	CUD	7/1/2006
		Isovaleric acidemia	IVA	7/1/2006
		Glutaric acidemia type I	GAI	7/1/2006
<b>Organic Acid Conditions:</b> Babies with these conditions have problems digesting certain parts of protein from the food they eat. This can cause serious effects on health, growth, and	Hydroxymethylglutaric aciduria/HMG-CoA lyase deficiency	HMG	7/1/2006	
	Multiple carboxylase deficiency	MCD	7/1/2006	
	Methylmalonic acidemia due to mutase deficiency	MUT	7/1/2006	

	can. This can cause serious effects on hearing, growth, and learning. Early treatment with medication can help prevent these problems.	Methylmalonic acidemia cblA and cblB forms	CBLA,B	7/1/2006
		3-Methylcrotonyl-CoA carboxylase deficiency	3MCC	7/1/2006
		Propionic acidemia	PROP	7/1/2006
		Beta-Ketothiolase deficiency	BKT	7/1/2006
<b>OTHER CONDITIONS</b>	Cystic Fibrosis affects the salt or water content of certain body fluids such as sweat and mucus. This can lead to problems with breathing, digesting, and other body functions. Treatment may include medications, dietary supplements, and other therapies.	Cystic Fibrosis	CF	7/1/2006
	Babies with biotinidase deficiency can have problems with growth and development. This condition is treated with special medicine.	Biotinidase Deficiency	BIOT	7/1/2006
	Babies with galactosemia cannot use the sugars in milk, formula, and breast milk. If it is not treated, it can hurt the baby's eyes, liver, and brain. This condition is treated with special formula and diet.	Classical Galactosemia	GALT	7/1/2006

\* Effective date of screening is determined by the date the sample was received at the laboratory. For example, CF screening has been for specimens received at the laboratory starting

#### Important Notes:

Other conditions may be identified by screening for the targeted list of conditions noted above

All tests may not be performed on repeat specimens

All tests may not be valid if age > 30 days on blood drawn date

Results are available in KIDSNET and are mailed to the primary care provider listed on the newborn screening specimen collection slip.

To obtain a lab report, or for questions about results, please contact the Newborn Screening Coordinator at VNA of Care New England (401) 737-6050 x 1126

#### Additional Resources:

American College of Medical Genetics Newborn Screening ACTION Sheets:

<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>

National Newborn Screening and Genetics Resource Center

<http://genes-r-us.uthscsa.edu/>

STAR-G Fact Sheets for Parents

<http://www.newbornscreening.info/Parents/facts.html>