

Table of Disorders Screened by Program

Condition	Incidence	Symptoms if no Detected	Treatment
Other Disorders			
<p>Biotinidase Deficiency:</p> <p>A condition in which the body is unable to use biotin, a B vitamin.</p>	<p>1 in 60,000 births (1 in 40,385 births in Hawaii)</p>	<p>Mental Retardation, seizures, skin rash, loss of hair, death</p>	<p>Supplement with biotin</p>
<p>Congenital Adrenal Hyperplasia (CAH):</p> <p>A condition in which the adrenal glands are unable to produce normal amounts of certain hormones.</p>	<p>1 in 13,700 births (1 in 25,699 births in Hawaii)</p>	<p>Salt wasting, dehydration, shock in infants</p> <p>Abnormal genital organs in females</p>	<p>Glucocorticoid and/or mineral corticoid</p>
<p>Congenital Hypothyroidism:</p> <p>A condition in which the thyroid gland cannot make enough thyroid hormone for normal body and brain growth.</p>	<p>1 in 4,485 births (1 in 2,501 births in Hawaii)</p>	<p>Mental retardation, other brain damage, growth delay</p>	<p>Thyroid hormone replacement</p>
<p>Cystic Fibrosis:</p> <p>A condition that causes thick, sticky mucus and fluids to build up in certain organs in the body, especially the lungs and the the pancreas.</p>	<p>1 in 3,000 births (1 in 19,959 births in Hawaii)</p>	<p>Pancreatic insufficiency and lung disease</p>	<p>Enzyme replacement and organ transplant</p>
<p>Galactosemia:</p> <p>A condition in which the body cannot break down a sugar (galactose) found in milk.</p>	<p>1 in 60,000 births (1 in 141,349 births in Hawaii)</p>	<p>Severe brain damage, kidney damage and eye abnormalities in neonates, death</p>	<p>Strict galactose-free diet</p>

<p>Hemoglobinopathies (including Sickle Cells):</p> <p>Conditions in which abnormal hemoglobin in red blood cells may cause anemia.</p>	<p>Sickle cell disease:</p> <p>1 in 15,000 births</p> <p>(1 in 40,385 births in Hawaii)</p>	<p>Sickle cell disease:</p> <p>Anemia, painful crises, death</p>	<p>Sickle cell disease:</p> <p>Penicillin</p>
<p>Severe Combined Immunodeficiency (SCID)</p> <p>A group of inherited primary immune disorders</p>	<p>1 in 54,000 births</p> <p>(Incidence in Hawaii TBD, anticipated to be similar to nationally observed incidence noted above)</p>	<p>Severe defect in T cells and altered B cell function leads to failure to thrive and recurrent serious and/or life-threatening infections, death</p>	<p>Short-term treatment of infections and preventive medications.</p> <p>Long-term bone marrow transplant, gene therapy and/or enzyme replacement therapy</p>
Amino Acid Disorders			
<p>Arginase Deficiency:</p> <p>A condition in which the body cannot get rid of a toxic substance called ammonia.</p>	<p>Rare</p> <p>(No cases in Hawaii)</p>	<p>Developmental delay, seizures, hyperactivity, ataxia</p>	<p>Restrict arginine and protein in diet</p> <p>Supplement with amino acids other than arginine</p> <p>Sodium benzoate therapy</p>
<p>Argininosuccinate Lyase Deficiency (ASA):</p> <p>A condition in which the body cannot get rid of a toxic substance called ammonia.</p>	<p>1 in 70,000 births</p> <p>(1 in 174,112 births in Hawaii)</p>	<p>Mental retardation, potential lethal coma, seizures, anorexia, vomiting, lethargy</p>	<p>Restrict protein in diet</p> <p>Supplement with arginine</p>
<p>Citrullinemia:</p> <p>A condition in which the body cannot get rid of a toxic substance called ammonia.</p>	<p>n/a</p> <p>(1 in 174,112 births in Hawaii)</p>	<p>Mental retardation, potential lethal coma, seizures, anorexia, vomiting, lethargy</p>	<p>Low protein diet</p> <p>Sodium benzoate, phenylacetate, arginine</p>
<p>Homocystinuria:</p> <p>A condition in which the body cannot break down several amino acids in protein foods.</p>	<p>1 in 200,000 births</p> <p>(No cases in Hawaii)</p>	<p>Heart disease, stroke, possible mental retardation, psychiatric problems</p>	<p>Low methionine diet</p> <p>Supplement with pyridoxine, L-cysteine, and betaine</p>

Updated as of 2/2015

<p>Phenylketonuria (PKU):</p> <p>A condition in which the body cannot break down one of the amino acids found in protein foods.</p>	<p>1 in 15,900 births</p> <p>(1 in 47,116 births in Hawaii)</p>	<p>Severe mental retardation, seizures</p>	<p>Low phenylalanine diet</p>
<p>Tyrosinemia Types I and II:</p> <p>A condition in which the body cannot break down several amino acids in protein foods.</p>	<p>1 in 100,000 births</p> <p>(1 in 1,846 French Canadian births)</p>	<p>Liver disease, kidney problems, seizures, rickets</p>	<p>Low phenylalanine and tyrosine diet</p> <p>Liver transplant if necessary</p>
Organic Acid Disorders			
<p>Beta-Ketothiolase Deficiency:</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>Rare</p> <p>(No cases in Hawaii)</p>	<p>Recurrent, severe metabolic acidosis</p>	<p>Sodium bicarbonate, IV fluids</p> <p>Possible dialysis</p> <p>Supplement with carnitine</p>
<p>Glutaric Acidemia Type I:</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>1 in 30,000 live births</p> <p>(1 in 87,056 births in Hawaii)</p>	<p>Neurological deterioration, muscle weakness, seizures, possible dystonic cerebral palsy</p> <p>Some people may have no symptoms</p>	<p>Restrict lysine and tryptophan in diet</p> <p>Supplement with riboflavin and carnitine</p>
<p>Isobutyryl CoA Dehydrogenase Deficiency:</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>Very rare</p> <p>(No cases in Hawaii)</p>	<p>Heart problems</p>	<p>Carnitine supplementation</p>
<p>Isovaleric Acidemia:</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>1 in 50,000 births</p> <p>(1 in 43,528 births in Hawaii)</p>	<p>Vomiting, lack of appetite, lethargy, neuromuscular irritability, hypothermia</p>	<p>Protein-restrictive diet</p> <p>Supplement with carnitine and glycine</p>

<p>Malonic Aciduria:</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>Rare</p> <p>(No cases in Hawaii)</p>	<p>Developmental delay, vomiting, seizures, cardiomyopathy, hypoglycemia</p>	<p>Avoid fasting</p> <p>Restrict fats in diet</p>
<p>Maple Syrup Urine Disease (MSUD):</p> <p>A condition in which the body cannot break down several amino acids in protein foods.</p>	<p>1 in 150,000 births</p> <p>(1 in 70,674 births in Hawaii)</p>	<p>Neonatal coma, convulsions, mental retardation, death</p>	<p>Diet low in branched chain amino acids</p>
<p>Methylmalonic Acidemias:</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>1 in 50,000 to 1 in 100,000 births</p> <p>(No cases in Hawaii)</p>	<p>Lethargy, vomiting, dehydration, respiratory distress, muscle weakness, coma, seizures, developmental delay</p>	<p>Low-protein diet and/or restriction of isoleucine, valine, and threonine</p>
<p>Multiple Carboxylase Deficiency (MCD):</p> <p>A condition in which the body cannot break down and get rid of certain organic acids</p>	<p>1 in 87,000 births</p> <p>(1 in 87,056 births in Hawaii)</p>	<p>Seizures, immune system impairment, skin rashes, hair loss, hearing loss, mental retardation</p>	<p>Biotin supplementation</p>
<p>Propionic Acidemia:</p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>1 in 35,000 to 1 in 75,000 births</p> <p>(No cases in Hawaii)</p>	<p>Mental retardation, seizures, movement disorders, coma, sudden death</p>	<p>Avoid fasting, low protein diet</p> <p>Supplement with cornstarch, carnitine, and biotin</p> <p>Antibiotic and human growth hormone treatment</p>
<p>2-Methyl-3-Hydroxybutyryl CoA Dehydrogenase Deficiency:</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>Rare</p> <p>(No cases in Hawaii)</p>	<p>Developmental delay</p>	<p>In progress</p>

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<p>2-Methylbutyryl CoA Dehydrogenase Deficiency:</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>Rare (No cases in Hawaii)</p>	<p>Lethargy, irritability, coma</p>	<p>Dietary restrictions</p>
<p>3-Hydroxy-3-Methylglutaryl (HMG) CoA Lyase Deficiency:</p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>Rare (No cases in Hawaii)</p>	<p>Persistent vomiting, muscle weakness, lethargy, seizures, coma</p>	<p>Avoid fasting, low fat, low protein, high carbohydrate diet</p> <p>Supplement with carnitine and glucose</p>
<p>3-Methylcrotonyl CoA Carboxylase Deficiency (3MCC):</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>Rare (1 in 58,037 births in Hawaii)</p>	<p>Muscle weakness and atrophy, seizures, dermatological changes</p>	<p>Dietary restrictions</p> <p>Supplement with carnitine and/or biotin</p>
<p>3-Methylglutaconyl CoA Hydratase Deficiency:</p> <p>A condition in which the body cannot break down and get rid of certain organic acids.</p>	<p>Rare (No cases in Hawaii)</p>	<p>Delayed motor development, short attention span, delayed development of speech</p>	<p>Still in development</p>
Fatty Acid Oxidation Disorders			
<p>Carnitine Uptake/Transport Defects:</p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>Rare (1 in 19,345 births in Hawaii)</p>	<p>Developmental delay, muscle weakness</p> <p>Possible coma and death</p>	<p>Avoid fasting, low fat diet</p> <p>Supplement with carnitine</p>
<p>Glutaric Acidemia, Type II:</p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>Rare (1 in 174,112 births in Hawaii)</p>	<p>Muscle weakness, nausea, vomiting</p> <p>Possible seizures, coma, and death</p>	<p>Avoid fasting, low fat diet</p> <p>Supplement with carnitine</p>

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<p>Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD):</p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>Rare</p> <p>(More common in those with Finnish ancestry)</p>	<p>Developmental delay, muscle weakness, possible liver failure</p>	<p>Avoid fasting</p> <p>Supplement with carnitine, cornstarch, MCT, and DHA</p>
<p>Medium Chain acyl-CoA Dehydrogenase Deficiency (MCAD):</p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>1 in 15,000 births</p> <p>(More common in Northern Europeans)</p> <p>(1 in 43,528 births in Hawaii)</p>	<p>Development delay, seizures, coma, sudden death</p>	<p>Avoid fasting, low fat diet</p> <p>Supplement with carnitine and cornstarch</p>
<p>Short Chain acyl-CoA Dehydrogenase Deficiency (SCAD):</p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>Rare</p> <p>(1 in 174,112 births in Hawaii)</p>	<p>Developmental delay, muscle weakness</p> <p>Can have no symptoms or problems</p>	<p>Diet low in fats</p> <p>Supplement with carnitine</p>
<p>Very Long Chain acyl-CoA Dehydrogenase Deficiency (VLCAD):</p> <p>A condition in which the body cannot break down dietary fats to make energy.</p>	<p>Rare</p> <p>(1 in 29,018 births in Hawaii)</p>	<p>Heart problems, liver problems, sudden infant death</p>	<p>Avoid fasting, avoid certain fatty foods</p> <p>Supplement with cornstarch, MCT, and possibly carnitine</p> <p>IV glucose during illness</p>