Table of Disorders Screened by Program

Condition	Incidence	Symptoms if not Detected	Treatment
Other Disorders			
Biotinidase Deficiency: A condition in which the body is unable to use biotin, a B vitamin.	1 in 60,000 births (1 in 37,767 births in Hawaii)	Intellectual disabilities, seizures, skin rash, loss of hair, death	Supplement with biotin
Congenital Adrenal Hyperplasia (CAH): A condition in which the adrenal glands are unable to produce normal amounts of certain hormones.	1 in 13,700 births (1 in 22,660 births in Hawaii)	Salt wasting, dehydration, shock in infants Abnormal genital organs in females	Glucocorticoid and/or mineral corticoid
Congenital Hypothyroidism: A condition in which the thyroid gland cannot make enough thyroid hormone for normal body and brain growth.	1 in 4,485 births (1 in 2,634 births in Hawaii)	Intellectual disabilities, other brain damage, growth delay	Thyroid hormone replacement
Cystic Fibrosis: A condition that causes thick, sticky mucus and fluids to build up in certain organs in the body, especially the lungs and the pancreas.	1 in 3,000 births (1 in 14,409 births in Hawaii)	Pancreatic insufficiency and lung disease	Enzyme replacement and organ transplant
Galactosemia: A condition in which the body cannot break down a sugar (galactose) found in milk.	1 in 60,000 births (1 in 169,952 births in Hawaii)	Severe brain damage, kidney damage and eye abnormalities in neonates, death	Strict galactose-free diet

Hemoglobinopathies (including Sickle Cell): Conditions in which abnormal hemoglobin in red blood cells may cause anemia.	Sickle cell disease: 1 in 15,000 births (1 in 19,994 births in Hawaii)	Sickle cell disease: Anemia, painful crises, death	Sickle cell disease: Penicillin
Severe Combined Immunodeficiency (SCID): A group of inherited primary immune disorders.	1 in 54,000 births (1 in 16,844)	Severe defect in T cells and altered B cell function leads to failure to thrive and recurrent serious and/or life-threatening infections, death	Short-term treatment of infections and preventive medications Long-term bone marrow transplant, gene therapy and/or enzyme replacement therapy
Amino Acid Disorders			
Arginase Deficiency: A condition in which the body cannot get rid of a toxic substance called ammonia.	Rare (No cases in Hawaii)	Developmental delay, seizures, hyperactivity, ataxia	Restrict arginine and protein in diet Supplement with amino acids other than arginine Sodium benzoate therapy
Argininosuccinate Lyase Deficiency (ASA): A condition in which the body cannot get rid of a toxic substance called ammonia.	1 in 70,000 births (1 in 116,241 births in Hawaii)	Intellectual disabilities, potential lethal coma, seizures, anorexia, vomiting, lethargy	Restrict protein in diet Supplement with arginine
Citrullinemia: A condition in which the body cannot get rid of a toxic substance called ammonia.	n/a (1 in 232,482 births in Hawaii)	Intellectual disabilities, potential lethal coma, seizures, anorexia, vomiting, lethargy	Low protein diet Sodium benzoate, phenylacetate, arginine
Homocystinuria: A condition in which the body cannot break down several amino acids in protein foods.	1 in 200,000 births (No cases in Hawaii)	Heart disease, stroke, possible intellectual disabilities, psychiatric problems	Low methionine diet Supplement with pyridoxine, L-cysteine, and betaine

Phenylketonuria (PKU): A condition in which the body cannot break down one of the amino acids found in protein foods.	1 in 15,900 births (1 in 56,650 births in Hawaii)	Severe intellectual disabilities, seizures	Low phenylalanine diet
Tyrosinemia Types I and II: A condition in which the body cannot break down several amino acids in protein foods.	1 in 100,000 births (1 in 1,846 French Canadian births) (No cases in Hawaii)	Liver disease, kidney problems, seizures, rickets	Low phenylalanine and tyrosine diet Liver transplant if necessary
Organic Acid Disorders			
Beta-Ketothiolase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids.	Rare (No cases in Hawaii)	Recurrent, severe metabolic acidosis	Sodium bicarbonate, IV fluids Possible dialysis Supplement with carnitine
Glutaric Acidemia Type I: A condition in which the body cannot break down and get rid of certain organic acids.	1 in 30,000 live births (1 in 116,241 births in Hawaii)	Neurological deterioration, muscle weakness, seizures, possible dystonic cerebral palsy Some people may have no symptoms	Restrict lysine and tryptophan in diet Supplement with riboflavin and carnitine
Isobutyryl-CoA Dehydrogenase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids.	Very rare (No cases in Hawaii)	Heart problems	Carnitine supplementation
Isovaleric Acidemia: A condition in which the body cannot break down and get rid of certain organic acids.	1 in 50,000 births (1 in 38,747 births in Hawaii)	Vomiting, lack of appetite, lethargy, neuromuscular irritability, hypothermia	Protein-restrictive diet Supplement with carnitine and glycine

Malonic Aciduria: A condition in which the body cannot break down and get rid of certain organic acids.	Rare (No cases in Hawaii)	Developmental delay, vomiting, seizures, cardiomyopathy, hypoglycemia	Avoid fasting Restrict fats in diet
Maple Syrup Urine Disease (MSUD): A condition in which the body cannot break down several amino acids in protein foods.	1 in 150,000 births (1 in 84,976 births in Hawaii)	Neonatal coma, convulsions, intellectual disabilities, death	Diet low in branched chain amino acids
Methylmalonic Acidemia: A condition in which the body cannot break down and get rid of certain organic acids.	1 in 50,000 to 1 in 100,000 births (No cases in Hawaii)	Lethargy, vomiting, dehydration, respiratory distress, muscle weakness, coma, seizures, developmental delay	Low-protein diet and/or restriction of isoleucine, valine, and threonine
Multiple Carboxylase Deficiency (MCD): A condition in which the body cannot break down and get rid of certain organic acids.	1 in 87,000 births (1 in 116,241 births in Hawaii)	Seizures, immune system impairment, skin rashes, hair loss, hearing loss, intellectual disabilities	Biotin supplementation
Propionic Acidemia: A condition in which the body cannot break down dietary fats to make energy.	1 in 35,000 to 1 in 75,000 births (No cases in Hawaii)	Intellectual disabilities, seizures, movement disorders, coma, sudden death	Avoid fasting, low protein diet Supplement with cornstarch, carnitine, and biotin Antibiotic and human growth hormone treatment
2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids.	Rare (No cases in Hawaii)	Developmental delay	In progress

2-Methylbutyryl-CoA Dehydrogenase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids.	Rare (No cases in Hawaii)	Lethargy, irritability, coma	Dietary restrictions
3-Hydroxy-3-Methylglutaryl (HMG)-CoA Lyase Deficiency: A condition in which the body cannot break down dietary fats to make energy.	Rare (No cases in Hawaii)	Persistent vomiting, muscle weakness, lethargy, seizures, coma	Avoid fasting, low fat, low protein, high carbohydrate diet Supplement with carnitine and glucose
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC): A condition in which the body cannot break down and get rid of certain organic acids.	Rare (1 in 77,494 births in Hawaii)	Muscle weakness and atrophy, seizures, dermatological changes	Dietary restrictions Supplement with carnitine and/or biotin
3-Methylglutaconyl -CoA Hydratase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids.	Rare (No cases in Hawaii)	Delayed motor development, short attention span, delayed development of speech	Still in development
Fatty Acid Oxidation Disorders			
Carnitine Uptake/Transport Defects: A condition in which the body cannot break down dietary fats to make energy.	Rare (1 in 232,482 births in Hawaii)	Developmental delay, muscle weakness Possible coma and death	Avoid fasting, low fat diet Supplement with carnitine
Glutaric Acidemia, Type II: A condtion in which the body cannot break down dietary fats to make energy.	Rare (1 in 232,482 births in Hawaii)	Muscle weakness, nausea, vomiting Possible seizures, coma, and death	Avoid fasting, low fat diet Supplement with carnitine

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD): A condition in which the body cannot break down dietary fats to make energy.	Rare (More common in those with Finnish ancestry) (No cases in Hawaii)	Developmental delay, muscle weakness, possible liver failure	Avoid fasting Supplement with carnitine, cornstarch, Medium-Chain Triglycerides (MCT), and Docosahexaenoic Acid (DHA)
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD): A condition in which the body cannot break down dietary fats to make energy.	1 in 15,000 births (More common in Northern Europeans) (1 in 58,120 births in Hawaii)	Development delay, seizures, coma, sudden death	Avoid fasting, low fat diet Supplement with carnitine and cornstarch
Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD): A condition in which the body cannot break down dietary fats to make energy.	Rare (1 in 232,482 births in Hawaii)	Developmental delay, muscle weakness Can have no symptoms or problems	Diet low in fats Supplement with carnitine
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD): A condition in which the body cannot break down dietary fats to make energy.	Rare (1 in 33,211 births in Hawaii)	Heart problems, liver problems, sudden infant death	Avoid fasting, avoid certain fatty foods Supplement with cornstarch, MCT, and possibly carnitine IV glucose during illness