

DISORDERS DETECTED BY THE WASHINGTON NEWBORN SCREEN (2014)

Table 1: Disorders on this page can be deadly if not detected and treated within days following birth.

Disorder (Prevalence in WA)	Definition	Screening Test	Impact without Early Treatment	Treatment	Benefits of Early Treatment
Galactosemia (1 in 40,000)	Inability to break down galactose, a major sugar found in milk	Measure activity of enzyme needed to break down galactose	Severe intellectual and developmental disability, liver disease, blindness, overwhelming infections and death	Dietary restriction of milk and other foods containing galactose	Prevent death, improve mental function & reduce other morbidity
Congenital Adrenal Hyperplasia (CAH) (1 in 16,000)	Impaired production of cortisol and other adrenal hormones	Measure adrenal hormone: 17-hydroxyprogesterone (17-OHP) level	Salt loss & shock may result in early sudden death, also virilization & abnormal growth	Cortisol & salt-retaining hormone replacement	Prevent death, reduce virilization & abnormal growth
Organic Acid Disorders (1 in 25,000) (see list below)	Inability to process or break down organic acids, byproducts of protein and fatty acid metabolism	Measure acylcarnitine levels by tandem mass spectrometry	Severe nerve and physical damage & death	Dietary restriction of offending amino acid(s) & needs special metabolic formula	Prevent death, intellectual and developmental disability and other neurological damage
Fatty Acid Oxidation Disorders (1 in 13,000) (see list below)	Inability to process or break down fats in the body	Measure acylcarnitine levels by tandem mass spectrometry	Serious damage to brain, liver, heart, eyes, muscles & death	High carbohydrate, low-fat diet & avoidance of fasting	Prevent death, intellectual and developmental disability and other neurological damage
Amino Acid Disorders (1 in 10,000) (see list below)	Inability to break down amino acids, found in all foods containing protein	Measure amino acid levels by tandem mass spectrometry	Intellectual and developmental disability, seizures, coma & death	Dietary restriction of offending amino acid(s) & needs special metabolic formula	Prevent death, intellectual and developmental disability and other neurological damage

Amino Acid Disorders

*Argininosuccinic acidemia (ASA)
*Citrullinemia (CIT)
Homocystinuria (HCYS)
*Maple Syrup Urine Disease (MSUD)
Phenylketonuria (PKU)
Tyrosinemia type I (TYR-I)

Organic Acid Disorders

3-OH 3-CH3 glutaric aciduria (HMG)
Glutaric acidemia type I (GA-I)
Beta-Ketothiolase deficiency (BKT)
*Isovaleric acidemia (IVA)
*Methylmalonic acidemia (Cbl A, B)
*Methylmalonic acidemia (*mutase deficiency*) (MUT)
Multiple carboxylase deficiency (MCD)
*Propionic acidemia (PROP)

Fatty Acid Oxidation Disorders

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

* Not all amino acid, organic acid and fatty acid oxidation disorders are life-threatening within days of birth. The disorders noted by an asterisk can be deadly if not detected and treated within days of birth

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Table 2: Disorders on this page are not deadly within days of birth, but delay in treatment may result in later death or profound, permanent disability

Disorder (Prevalence in WA)	Definition	Screening Test	Impact without Early Treatment	Treatment	Benefits of Early Treatment
Sickle Cell Diseases and Hemoglobinopathies (1 in 10,000)	Production of abnormal hemoglobin	Separate and visualize hemoglobin proteins by isoelectric focusing, with confirmation by high performance liquid chromatography and DNA analysis	Severe infections and possible death within 2-3 months following birth	Antibiotic prophylaxis to help prevent infections & parental education to recognize health crises	Prevent death, reduce infections and other morbidity
Congenital Hypothyroidism (1 in 1,600)	Inadequate production of thyroid hormone	Measure thyroid stimulating hormone (TSH) level	Intellectual and developmental disability, growth failure	Thyroid hormone replacement	Normal growth and mental development
Biotinidase Deficiency (1 in 60,000)	Deficiency of biotin, part of the Vitamin B complex	Measure activity of enzyme needed to recycle biotin	Seizures, damage to immune system, intellectual and developmental disability, hearing loss	Oral biotin supplementation	Prevent all adverse consequences
Cystic Fibrosis (1 in 5,000)	Defect in the cystic fibrosis transmembrane conductance regulator (CFTR) gene	Measure immunoreactive trypsinogen (IRT) level	Significant nutritional deficits due to thick, sticky mucus in the digestive system. Severe lung infections due to mucus	Pancreatic enzymes, vitamin supplements, chest physiotherapy, antibiotics	Improve physical growth, cognitive function & possibly lung function
Severe Combined Immunodeficiency (1 in 45,000)	Complete lack of immune system in the baby	DNA test: measure number of T-cell excision circles (TRECs) by real-time PCR	Severe life-threatening infections that complicate treatment and possible death	Stem-cell transplant or gene therapy, depending on the genotype of the baby	Prevent death and cure the condition



Questions? Please contact:

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To submit a request, please call 1-800-525-0127 (TDD/TTY 1-800-833-6388).