DISORDERS DETECTED BY THE WASHINGTON NEWBORN SCREEN (2020)

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 63,000)	Inability to break down galactose, a major sugar found in milk	Measure activity of enzyme needed to break down galactose; DNA test if indicated	Severe intellectual and developmental disability, liver disease, blindness, overwhelming infections and death	Dietary restriction of milk and other foods containing galactose	Prevent death, improve intellectual function, and reduce other morbidity
Congenital Adrenal Hyperplasia (CAH) (1 in 14,000)	Impaired production of cortisol and other adrenal hormones	Measure adrenal hormone: 17-hydroxyprogesterone (17-OHP) level	Salt loss and shock may result in early sudden death; virilization and abnormal growth	Cortisol and salt-retaining hormone replacement	Prevent death, reduce virilization and abnormal growth
Organic Acid Disorders (1 in 29,000) (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 and death	Dietary restriction of offending amino acid(s) and use of special metabolic formula	Prevent death, intellectual and developmental disability and other neurological damage
Fatty Acid Oxidation Disorders (1 in 11,000) (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 and muscles, and death	High carbohydrate, low-fat diet and 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, and death	Dietary restriction of offending amino acid(s) and use of 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

Questions? Please contact:
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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 4,700)	Production of abnormal hemoglobin	Separate and visualize hemoglobin proteins by isoelectric focusing, with confirmation by high performance liquid chromatography and DNA analysis, if indicated	Severe infections and possible death	Antibiotic prophylaxis to help prevent infections and parental education to recognize health crises	Prevent death, reduce infections and other morbidity
Congenital Hypothyroidism (1 in 950)	Inadequate production of thyroid hormone	Measure thyroid stimulating hormone (TSH) level	Intellectual and developmental disability, growth failure	Thyroid hormone replacement	Normal growth and intellectual development
Biotinidase Deficiency (1 in 86,000)	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 (CF) (1 in 5,500)	Defect in the cystic fibrosis transmembrane conductance regulator (CFTR) gene	Measure immunoreactive trypsinogen (IRT) level; DNA test if indicated	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 and possibly lung function
Severe Combined Immunodeficiency (SCID) (1 in 88,000)	Complete lack of immune system	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	Prevent death and cure the condition
X-linked Adrenoleukodystropy (X-ALD) (1 in 17,000)	Peroxisomal disorder caused by mutations in ABCD1 gene leading to accumulation of very long chain fatty acids (VLCFA) in tissues and organs	Measure very long chain fatty acid levels by tandem mass spectrometry	Severe debilitating sensorimotor, behavioral and cognitive functions that can lead to death 2-4 years after onset of symptoms for those affected with childhood cerebral ALD	Cortisol (hormone replacement), Stem cell transplant (HSCT)	Treatment with HSCT may prevent death and disability

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Glycogen Storage Disorder (Pompe disease) (1 in 28,000)	Inability to break down glycogen, (a complex sugar)	Measure activity of enzyme needed to break down glycogen	Muscle weakness, possible cardiac and respiratory failure, and possible death	Enzyme replacement therapy	Prevent death, reduce need for mechanical ventilation and other morbidity
Mucopolysaccharidosis type I (MPS-I) (1 in 36,000)	Inability to break down glycosaminoglycans (large sugar molecules)	Measure activity of enzyme needed to break down glycosaminoglycans	Progressive cognitive decline	Hematopoietic stem-cell transplantation and/or Enzyme replacement therapy	Slow or halt cognitive decline, reduce other morbidity
Spinal Muscular Atrophy (SMA) (1 in 15,000)	Genetic disorder that results in lack of survival motor neuron (SMN) protein, causing progressive death of nerve cells in the spinal cord	DNA test: detect the presence/absence of exon 7 of SMN1 by real-time PCR	Muscle weakness, possible difficulty walking, swallowing, breathing, or even death.	One-time gene therapy or regularly administered intrathecal or oral medications	Prevent death, slow or halt disease progression