

NEWBORN SCREENING INFORMATION MATRIX

Disorders Currently Screened in Washington – September 2008

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Disorder & (Prevalence in WA)	Definition	Screening Test	Impact Without early Treatment	Treatment	Benefits of Early Treatment
Biotinidase deficiency (1 in 60,000)	Deficiency of biotin, part of the Vitamin B complex	Enzyme assay: Measure Biotinidase activity	Seizures, damage to immune system, mental retardation, hearing loss	Oral biotin supplementation	Prevent all adverse consequences
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, virilization & abnormal growth	Cortisol & salt-retaining hormone replacement	Prevent death, reduce virilization & abnormal growth
Congenital hypothyroidism (1 in 1,600)	Inadequate production of thyroid hormone	Measure thyroid stimulating hormone (TSH) level	Mental retardation, growth failure	Thyroid hormone replacement	Normal growth and mental development
Cystic fibrosis (1 in 3000 expected)	Defect in the cystic fibrosis transmembrane conductance regulator (CFTR) gene	Measure immunoreactive trypsinogen (IRT) level	Thick, sticky mucus builds up in the lungs and digestive system	Pancreatic enzymes, vitamin supplements, chest physiotherapy, antibiotics	Improve physical growth, cognitive function & possibly lung function

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Galactosemia (1 in 40,000)	Inability to break down galactose, a major sugar found in milk	Enzyme assay: measure galactose-1-phosphate uridyl transferase (GALT) activity	Galactose accumulates in vital organs, leading to severe mental retardation, liver disease, blindness, overwhelming infections and death	Dietary restriction of galactose	Prevent death, improve mental function & reduce other morbidity
Sickle cell disease (1 in 10,000)	Production of abnormal hemoglobin	Separate and visualize hemoglobin proteins by electrophoresis	Severe infections and possible death	Antibiotic prophylaxis to help prevent infections & parental education to recognize health crises	Prevent death, reduce infections and other morbidity

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Disorder Group & (expected prevalence in WA)	Definition	Screening Test	Impact Without Early Treatment	Treatment	Benefits of Early Treatment
Amino acid disorders (1 in 10,000)	Inability to break down amino acids, found in all foods containing protein	Measure amino acid levels by MS/MS	Mental retardation, seizures, coma & death	Dietary restriction of offending amino acid(s) using a special metabolic formula	Prevent mental retardation and other neurological damage
Fatty acid disorders (1 in 13,000)	Inability to process or break down fats in the body due to missing or dysfunctional enzymes	Measure acylcarnitine levels by MS/MS	Serious damage to brain, liver, heart, eyes and muscles & death	High carbohydrate, low-fat diet & avoidance of fasting	Prevent mental retardation and other neurological damage
Organic acid disorders (1 in 25,000)	Inability to process or break down organic acids, byproducts of protein and fatty acid metabolism	Measure acylcarnitine levels by MS/MS	Severe nerve and physical damage & death	Dietary restriction of offending amino acid(s) and use of a special metabolic formula	Prevent mental retardation and other neurological damage

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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)
- Beta-Ketothiolase deficiency (BKT)
- Glutaric acidemia type I (GA-I)
- 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