

Newborn Screening Update

November 2009



In May 2008 the State Board of Health added 15 disorders to the list of conditions in Chapter 46-650 WAC.

Amino acid disorders

These disorders are characterized by the body's inability to correctly process amino acids or the inability to process the ammonia that is released during the break down of amino acids. The accumulation of amino acids, ammonia or other by-products may cause severe complications including mental retardation, coma, seizures, and possibly causing death.

- Argininosuccinic acidemia (ASA)
- Citrullinemia (CIT)
- Tyrosinemia type I (TYR I)

Fatty acid oxidation disorders

These disorders are characterized by the body's inability to efficiently use stored fat to make energy. During times of extra energy need such as prolonged fasting or acute illness, affected infants can suffer dangerously low blood sugar and metabolic crises resulting in serious damage affecting the brain, liver, heart, eyes, muscle, and possibly causing death.

- Carnitine uptake defect (CUD)
- Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHADD)
- Trifunctional protein deficiency (TFP)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD)

Organic acid disorders

These disorders are characterized by errors in processing amino acids resulting in the accumulation of non-amino organic acids and toxic intermediates. This may lead to metabolic crisis with increases in acid and ammonia in the blood, and dangerously low blood sugar resulting in severe nerve and physical damage and possibly causing death.

- 3-OH 3-CH₃ glutaric aciduria (HMG)
- Beta-Ketothiolase deficiency (BKT)
- Glutaric acidemia type I (GA 1)
- Isovaleric acidemia (IVA)
- Methylmalonic acidemia (Cbl A, B)
- Methylmalonic acidemia (mutase deficiency) (MUT)
- Multiple carboxylase deficiency (MCD)
- Propionic acidemia (PROP)