

Descriptions of Disorders Proposed to Add to the Newborn Screening Panel

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All are disorders of metabolism. Early detection and treatment can prevent most or all of the consequences described.

AMINO ACID DISORDERS:

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.

Proposed New

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

FATTY ACID OXIDATION DISORDERS:

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.

Proposed New

- 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:

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.

Proposed New

- 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)