

Descriptions of Disorders on the Current Newborn Screening Panel

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AMINO ACID DISORDERS: 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. Current panel:

- Homocystinuria (HCY)
- Maple syrup urine disease (MSUD)
- Phenylketonuria (PKU)

Biotinidase deficiency: deficiency of an enzyme (biotinidase) that facilitates the body's recycling of biotin. The result is biotin deficiency, which if undetected and untreated, may result in severe neurological damage or death.

Congenital adrenal hyperplasia: a severe disorder of adrenal steroid metabolism which may result in death of an infant during the neonatal period if undetected and untreated.

Congenital hypothyroidism: a disorder of thyroid function during the neonatal period causing impaired mental functioning if undetected and untreated.

Cystic fibrosis: a life-shortening disease caused by mutations in the gene encoding the cystic fibrosis transmembrane conductance regulator (CFTR), a transmembrane protein involved in ion transport. Affected individuals suffer from chronic, progressive pulmonary disease and nutritional deficits. Early detection and enrollment in a comprehensive care system provides improved outcomes and avoids the significant nutritional and growth deficits that are evident when diagnosed later.

Galactosemia: a deficiency of enzymes that help the body convert the simple sugar galactose into glucose resulting in a buildup of galactose and galactose-1-PO₄ in the blood. If undetected and untreated, accumulated galactose-1-PO₄ may cause significant tissue and organ damage often leading to sepsis and death.

Hemoglobinopathy: a hereditary blood disorder caused by genetic alteration of hemoglobin which results in characteristic clinical and laboratory abnormalities and which leads to developmental impairment or physical disabilities.

FATTY ACID OXIDATION DISORDERS: 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. Current panel:

- Medium chain acyl-CoA dehydrogenase deficiency (MCADD)