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Disorders Screened in New Jersey

In accordance with Chapter 24 of Public Laws of 1988 (N.J.S.A. 26:2-110 and N.J.S.A. 26:2-111), New Jersey has expanded its statewide system of newborn biochemical testing to include a total of 54 disorders, which, if not detected early, can cause severe health problems, mental retardation, and even death.

New Jersey Newborn Biochemical Screening Program Disorders

Fatty Acid Oxidation Disorders: Babies with any of these fatty acid oxidation disorders have trouble using fat for energy. A special enzyme, which converts fat to energy, is either missing or not working correctly. This can lead to a medical crisis if the baby fasts (goes without eating) for very long. This crisis can include vomiting, low blood sugar, or more serious problems such as coma. Typically, a special diet and special precautions may need to be taken of the baby is sick.

- 2,4-Dienoyl-CoA reductase deficiency
- Carnitine palmitoyltransferase I deficiency
- Carnitine palmitoyltransferase II deficiency
- Carnitine/acylcarnitine translocase deficiency
- Carnitine uptake defect
- Glutaric academia type II
- Long chain 3-Hydroxyacyl-CoA dehydrogenase deficiency
- Long chain acyl-CoA dehydrogenase deficiency
- Medium/short chain 3-Hydroxl acyl-CoA dehydrogenase deficiency
- Medium chain acyl-CoA dehydrogenase deficiency
- Medium chain ketoacyl-CoA thiolase deficiency
- Short chain acyl-CoA dehydrogenase deficiency
- Trifunctional protein deficiency
- Very long chain acyl-CoA dehydrogenase deficiency

Organic Acidemia Disorders: Babies born with one of these disorders cannot remove certain waste products from their blood. This can lead to vomiting, low blood sugar, developmental delays, and coma. Treatment depends on the disorder the baby has, but may include low protein diet, vitamins, and avoiding fasting. A baby born with one of these disorders needs lifetime treatment and regular medical care.

- 2-Methyl-3-hydroxybutyric aciduria
- 2-Methylbutyrylglycinuria
- 3-Hydroxyl-3-methylglutaryl-CoA lyase deficiency

- 3-Methylcrotonyl-CoA carboxylase deficiency
- 3-Methylglutaconic aciduria
- Beta-Ketothiolase deficiency
- Glutaric academia type I
- Isobutyrl glycinuria
- Isovaleric acidemia
- Malonic academia
- Methylmalonic academia-Cobalamin A, B
- Methylmalonic academia-Cobalamin C, D
- Methylmalonic academia-Mutase
- Multiple carboxylase deficiency
- Propionic academia

Amino Acid Disorders: Caused by defects in the disposal of the carbon skeletons of the branched chain amino acids after the initial transamination step. With the excretion of ornithine transcarbamylase deficiency, which is X-linked, all amino acid disorders are autosomal recessive. The metabolic encephalopathy is often accompanied by respiratory depression, seizures, and hypoxic-ischemic brain injury.

Urea Cycle Disorders: Metabolic problems where excessive amounts of ammonia accumulate in the blood. These excessive amounts of ammonia can lead to behavioral problems, mental retardation, coma, and even death. Babies with these disorders need a special diet and/or medications with medical monitoring.

Amino Acid Disorders and Urea Cycle Disorders:

- Argininemia
- Argininosucciic academia
- Hyperphenylalanemia (Benign)
- Biopterin cofactor defect of biosynthesis
- Biopterin cofactor defect of regeneration
- Citrullinemia type I
- Citrullinemia type II
- Homocystinuria
- Hypermethioninemla
- Maple syrup urine disease
- Phenylketonuria
- Tyrosinemia Type I
- Tyrosinemia Type II
- Tyrosinemia Type III

Endocrine Disorders:

- Congenital Adrenal Hyperplasia (CAH): This is a group of disorders in which the adrenal glands do not produce normal amounts of certain essential hormones. If untreated, serious loss of

body salt and water, and even death, may occur. Lifetime treatment includes daily medication and close monitoring to prevent complications of the disease.

- Congenital Hypothyroidism: Congenital hypothyroidism results when the body does not make enough thyroid hormone. If untreated, this disorder can cause mental retardation, abnormal growth, deafness, and neurological problems. Treatment includes medication to replace the missing thyroid hormone. Early treatment can prevent the damage caused by the disease.

Metabolic Disorders: These diseases are related to metabolic disorders which can affect the normal body and brain development of an infant. Newborn screening is done by getting a few drops of blood by pricking the heel of the baby. Blood samples are then submitted for study and tested for these medical conditions. Newborn found positive with metabolic disorders are referred to a medical specialist for treatment. Normal growth and development of the baby can be facilitated by your doctor's treatment plan which can save your baby from lifelong health and development problems.

- Biotinidase deficiency
- Classical galactosemia
- Galactoepimerase deficiency
- Galactokinase defiency

Other Disorders:

- Cystic Fibrosis: this disorder causes thick mucus to collect in the lungs and intestines, which can result in breathing problems, lung infections, and poor digestion of food. These newborns need frequent medical monitoring, including specialist care. Early identification and treatment has been shown to improve growth and development in children with cystic fibrosis.
- Hemoglobinopathies, including Hemoglobin S/Beta-thalassemia, Hemoglobin S/C disease, an Sickle Cell Anemia: In this group of disorders, the red blood cells are malformed. Babies with these disorders are more likely to have anemia, episodes of severe pain, and life-threatening infections. Babies with any of the hemoglobinopathies could require a variety of treatments including antibiotics to prevent serious infections.