

Quick Reference To Newborn Screening Disorders



Biotinidase Deficiency (BIO) BIO is an enzyme deficiency that occurs in about 1 in 60,000 newborns and can result in seizures, hearing loss, and death in severe cases. Treatment is simple and involves daily doses of biotin.

Congenital Adrenal Hyperplasia (CAH) CAH is caused by decreased or absent production of certain adrenal hormones. The most prevalent type is detected by newborn screening in about 1 in 15,000 newborns. Early detection can prevent death in boys and girls and sex misassignment in girls. Treatment involves lifelong hormone replacement therapy.

Congenital Hypothyroidism (CH) Inadequate or absent production of thyroid hormone results in CH and is present in about 1 in 3,500 newborns. Thyroid hormone replacement therapy begun by 1 month of age can prevent mental and growth retardation.

Congenital Toxoplasmosis (TOXO) Infection of the fetus with a parasite ingested by the mother during pregnancy can result in TOXO in the newborn. The transmission rate is about 30% and the national incidence is approximately 1 in 10,000 newborns based on limited screening. Early diagnosis and drug therapy reduces the risk of blindness, mental retardation or other serious complications.

Cystic Fibrosis (CF) CF occurs in about 1 in 4,000 U.S. newborns and is characterized by progressive lung disease, pancreatic dysfunction and other organ failures. Confirmatory testing usually involves sweat testing and treatment leads to decreased hospitalizations and better nutritional and pulmonary outcomes. Caucasians are at an increased risk.

Galactosemia (GAL) Failure to metabolize the milk sugar galactose results in GAL and occurs in about 1 in 50,000 newborns. The classical form detected by newborn screening can lead to cataracts, liver cirrhosis, mental retardation and/or death. Treatment is elimination of galactose from the diet usually by substituting soy.

Homocystinuria (HCY) HCY is caused by an enzyme deficiency that blocks the metabolism of an amino acid that can lead to mental retardation, osteoporosis and other problems if left undetected and untreated. The incidence is approximately 1 in 350,000 U.S. newborns. Treatment may involve dietary restrictions and supplemental medicines.

Maple Syrup Urine Disease (MSUD) MSUD is a defect in the way that the body metabolizes certain amino acids and is present in about 1 in 200,000 U.S. newborns. Early detection and treatment with dietary restrictions can prevent death and severe mental retardation. There is an increased risk in Mennonites.

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Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency The most common disorder in the way the body metabolizes fatty acids is called MCAD deficiency. Undetected, it can cause sudden death. Treatment is simple and includes ensuring regular food intake. The incidence from newborn screening is not yet known, but is thought to be approximately 1 in 15,000 newborns.

Phenylketonuria (PKU) An enzyme defect that prevents metabolism of phenylalanine, an amino acid essential to brain development, is known as PKU and occurs in approximately 1 in every 19,000 U.S. newborns. Undetected and untreated with a special diet, PKU leads to irreversible mental retardation. Persons of European descent are at increased risk.

Sickle Cell Disease (SCD) Sickle cell anemia (Hb-SS-Disease) is the most prevalent SCD and causes clogged blood vessels resulting in severe pain and other severe health problems. Other common SCDs include Hb-SC-Disease and various thalassemias. Newborn screening detects about 1 in 2,500 newborns with SCD annually. Persons of African or Mediterranean descent are at an increased risk.

Other Fatty Acid Oxidation (FAO) Disorders Besides MCAD deficiency, other FAO disorders may be detected through newborn screening. They are usually described in categories based on the length of the fatty acid involved. Undetected and untreated they can cause seizures, coma, and even death. The incidences of various FAO disorders are not known since it is only recently that early detection through newborn screening has occurred.

Organic Acid (OA) Disorders Organic acidemias are a group of metabolic disorders that lead to accumulation of organic acids in the blood and urine and may be detected in newborn screening through analysis of acylcarnitine profiles. Symptoms can be diminished by restricting protein in the diet and supplementation with vitamins and/or carnitine. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known.

Urea Cycle Disorders (UCD) A UCD is a genetic disorder caused by a deficiency of one of the enzymes responsible for removing ammonia from the blood stream. Some UCDs may be detected as a part of newborn screening. They are characterized by seizures, poor muscle tone, respiratory distress, and coma, and result in death if left undetected and untreated. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known.

