Information on the 29 disorders the American College of Medical Genetics recommends for newborn screening.

Amino Acid Metabolism Disorders

Phenylketonuria (PKU) occurs in 1 in 25,000 newborns. Persons with PKU are unable to properly process the essential amino acid phenylalanine. PKU can result in severe mental retardation unless detected soon after birth and treated with a special formula.

Maple syrup urine disease (MSUD) incidence rate is less than 1 in 100,000. This inborn error of metabolism can be lethal if unrecognized and untreated. Rapid diagnosis and treatment are major factors in survival and outcome. Treatment consists of a special low-protein diet, which will vary depending on severity of symptoms, and sometimes, supplementation with the vitamin thiamin.

Homocystinuria (HCY) occurs in less than 1 in 100,000 babies. Individuals with this disorder lack an enzyme responsible for converting the amino acid homocysteine into cystathionine, which is needed for normal brain development. Treatment consists of a special diet and other supplements.

Citrullinemia (CIT) occurs in less than 1 in 100,000. CIT is a build-up in the body of citrulline and ultimately ammonia. With early diagnosis and treatment, normal development is possible. Treatment includes a low-protein diet, medications, and nutritional supplements.

Argininosuccinic academia (ASA) incidence is less than 1 in 100,000. Symptoms for ASA most often begin in the first few days of life, with build-up of argininosuccinic acid and ultimately ammonia. Treatment consists of a lowprotein diet, avoiding fasting, medications to prevent ammonia build-up, nutritional supplements, and in some cases, a liver transplant.

Tyrosinemia type I (TYR I) occurs in less than 1 in 100,000 infants. TYR I is due to absence of an enzyme, byproducts of the amino acid tyrosine, and a very toxic compound called succinylacetone that build up in the liver. Drug treatment, sometimes along with a low-protein diet, is very effective in preventing liver and kidney damage.

Organic Acid Metabolism Disorders

Isovaleric academia (IVA) occurs in less than 1 in 100,000. IVA is the inability to process the amino acid leucine. With early diagnosis and treatment, most children have normal development. Treatment includes a low-protein diet and nutritional supplements.

Glutaric acidemia type I (GA I) has an incidence of a little greater than 1 in 75,000. Babies may develop normally for up to 18 months until something affects a child's health, such as a mild viral illness, which may trigger the onset of symptoms. Some affected babies also are born with an enlarged head (macrocephaly). Treatment can vary, but may include dietary protein restriction and supplementation with a nutrient called Lcarnitine.

Hydroxymethylglutaric aciduria or HMG-CoA lyase deficiency or 3-OH 3-CH3 glutaric aciduria (HMG) occurs in less than 1 in 100,000 newborns. These babies have the inability to process the amino acid leucine. Avoiding fasting and following a diet low in protein and fat and high in carbohydrates, can lead to normal development.

Multiple carboxylase deficiency (MCD) has an incidence rate less than 1 in 100,000. This disorder is caused by a defect of an enzyme required to activate several biotin-dependent enzymes. Early diagnosis and treatment with biotin allows normal growth and development.

Methylmalonic acidemia due to mutase deficiency (MUT) occurs in greater than 1 in 75,000 newborns. MUT is a defect in the processing of four essential amino acids and other substances resulting in illness in the first week of life. Treatment includes a low-protein diet, vitamin B12 injections, and nutritional supplements.

Methylmalonic acidemia cblA and cblB forms (Cbl A, B) occurs in less than 1 in 100,000 babies. This inherited defect of vitamin metabolism can lead to build-up of acids in the blood. Treatment with vitamin B12 injections and a low-protein diet often prevents serious problems.

3-Methylcrotonyl-CoA carboxylase deficiency (3MCC) incidence is greater than 1 in 75,000. 3MCC is a defect in processing the amino acid leucine. Treatment with a low-protein diet and, in some cases, nutritional supplements may be helpful.

Propionic academia (PROP) occurs in greater than 1 in 75,000 babies. Even with treatment, including a low protein diet and nutritional supplements, some affected children suffer from developmental delays, seizures, abnormal muscle tone, and heart problems.

Beta-Ketothiolase deficiency (BKT) occurs in less than 1 in 100,000 newborns. BKT presents it's self with periodic episodes of acid build-up that can often be triggered by some childhood illness. With early diagnosis and prompt intravenous treatment to keep blood sugar levels up and blood acid levels down during an illness, children can develop normally. Additional treatments may vary, but can include avoidance of protein-rich diets and long-term treatment with bicarbonate.

Fatty Acid Oxidation Disorders

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) occurs in 1 in 25,000 babies. Seemingly well infants and children can suddenly develop seizures (caused by low blood sugar), liver failure, coma, and death. Treatment includes avoidance of fasting and nutritional supplements.

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD) has an incidence greater than 1 in 75,000. Symptoms can first appear at any age from the newborn period through adulthood, but tend to be most severe in infants. Treatment includes a high-carbohydrate/low-fat diet, nutritional supplements, avoidance of fasting and prolonged exercise.

Long-chain 3-OH acyl-CoA dehydrogenase deficiency (LCHAD) occurs in greater than 1 in 75,000 infants. Symptoms can begin soon after birth, resulting in heart, lung or liver failure and death. In other cases, symptoms such as low muscle tone, developmental delay, heart, lung or liver failure may develop later in infancy or childhood. Treatment includes a high-carbohydrate/low-fat diet, nutritional supplements, and avoidance of fasting.

Trifunctional protein deficiency (TFP) has an incidence of less than 1 in 100,000. A seemingly healthy infant can die suddenly of what appears to be sudden infant death syndrome. Other infants may develop low muscle tone, seizures, heart failure and coma, often following an illness. Treatment is based on strict avoidance of fasting, a low-fat diet and nutritional supplements.

Carnitine uptake defect (CUD) occurs in less than 1 in 100,000 babies. These infants are missing a transporter that brings in carnitine from the blood. Early diagnosis and treatment with carnitine permits normal development.

Hemoglobinopathies

Sickle cell anemia (Hb SS) incidence rate is greater than 1 in 5,000 and has a much higher incidence among African-Americans at 1 in 400. Hb SS is a blood disease that can cause severe pain, damage to the vital organs, stroke, and sometimes death in childhood. Affected babies should receive all regular childhood vaccinations and additional treatments may include intermittent pain medications and regular blood transfusions.

Hb S/Beta-Thalassemia (Hb S/Th) occurs in greater than 1 in 50,000 infants. In this form of sickle cell anemia, the child inherits one sickle cell gene and one gene for beta thalassemia, another inherited anemia. Routine treatment with penicillin may not be recommended for all affected children.

Hb S/C disease (Hb S/C) occurs in greater than 1 in 25,000 infants. Hb S/C is another form of sickle cell disease, in which the child inherits one sickle cell gene and one gene for another abnormal type of hemoglobin called HbC. As with Hb S/Th, this form is often milder the Hb SS and routine penicillin treatment may not be recommended.

Others

This mixed group of disorders includes some diseases that are inherited and others that are not genetic. This group of disorders varies greatly in severity, from mild to life-threatening.

Congenital hypothyroidism (CH) occurs in greater than 1 in 5,000 babies. CH is a thyroid hormone deficiency which severely affects both growth and brain development. If detected soon after birth, the condition can be treated simply with oral doses of thyroid hormone to permit normal development.

Biotinidase deficiency (BIOT) has an incidence rate of greater than 1 in 75,000. BIOT is an inherited deficiency of Biotinidase. Undiagnosed and untreated, the deficiency can lead to coma and death. If the condition is detected soon after birth, these problems can be completely prevented with daily oral doses of biotin.

Congenital adrenal hyperplasia (CAH) occurs in greater than 1 in 25,000 newborns. CAH refers to a set of inherited disorders resulting from defects in the synthesis of hormones produced by the adrenal gland. Treatment includes salt replacement and hormone replacement.

Classical galactosemia (GALT) occurs in greater than 1 in 50,000 babies. Affected babies are missing the liver enzyme needed to convert galactose. Milk and other dairy products must be eliminated from the baby's diet for life.

Though treatment dramatically improves the outlook for affected infants, there is still some risk of developmental delays.

Hearing loss (HEAR) has an incidence of greater than 1 in 5,000. Without early testing, most babies with hearing loss are not diagnosed until 2 or 3 years of age. Early diagnosis allows use of hearing aids by 6 months of age, helping prevent serious speech and language problems.

Cystic fibrosis (CF) occurs in greater than 1 in 5,000 babies. Abnormalities in the cystic fibrosis protein result in lung and digestive problems, and death at an average age of 30-35 years. Treatment varies depending on severity of symptoms, but may include a high-calorie diet supplemented with vitamins and medications to improve digestion, respiratory therapy to help clear mucus from the lungs, and medications to improve breathing and prevent lung infections.

Source:
http://www.babysfirsttest.org/sites/default/files/A%20guide%20for%20prenatal%20educators.pdf