Endocrine Disorders

Congenital adrenal hyperplasia (CAH)

In CAH, the lack of an enzyme may cause shock or death in infants because the kidneys lose too much salt and newborn girls may be incorrectly identified as boys. CAH is treated with steroids and special minerals to replace lost salt. CAH occurs in approximately one of every 15,000 newborns.

Congenital hypothyroidism

Inadequate production of the hormone thyroxine can lead to mental and growth retardation. It is treated with thyroxine tablets. The condition occurs in approximately one of every 2,200 newborns.

Hemoglobinopathies

Sickle cell disease (sickle cell anemia or homozygous S disease)

Sickle cell disease disorder of the hemoglobin (oxygen-carrying part) of red blood cells. Children with sickle cell disease have a high risk of developing infections or anemia at an early age making medical care is necessary. The disorder occurs in approximately one of every 1,800 newborns, with a higher frequency in African-Americans. Infants with other abnormal hemoglobins may also be detected by newborn screening.

Sickle cell trait (sickle cell carrier)

Testing for sickle cell disease also detects infants with sickle cell trait. Although an infant with sickle cell trait does not have the problems of sickle cell disease, his or her parents could have another child who does have sickle cell disease.

Inborn Errors of Metabolism - Fatty Acid Oxidation Disorders

Carnitine-acylcarnitine translocase deficiency (CAT)

Babies with CAT can have low blood sugar, and heart and liver problems which may lead to coma, liver damage and death during common illnesses such as colds and flu. Children with CAT should avoid certain foods, consume special formulas, eat small meals often and get treated in the hospital if they get sick. It is not known how many newborns have this condition.

Carnitine palmitoyltransferase I deficiency (CPT-I)

Classic infant CPT-I causes low blood sugar, sleepiness, vomiting and seizures, which can lead to coma and death. Treatment includes frequent feeding (special formulas are often given, especially during common illnesses such as colds and flu) and not eating certain foods. It is not known how many newborns have this condition.

Carnitine palmitoyltransferase II deficiency (CPT-II)

Classic CPT-II in older people causes muscle weakness and pain during fasting, illness or prolonged exercise. Neonatal CPT-II causes coma, seizures, unsteady heartbeats, muscle weakness and death. Treatment includes a high-carbohydrate, low-fat diet with special formulas and treatment in the hospital if a child gets sick. It is not known how many newborns have this condition.

Carnitine uptake defect (CUD)

CUD can be very different in each child and can cause vomiting, confusion, coma and death. It is also a cause of sudden infant death. Children who do not get sick in infancy can develop large hearts and muscle weakness. It is not known how many newborns have this condition.

2,4-Dienoyl-CoA reductase deficiency (2,4Di)

2,4Di causes muscle weakness and death despite treatment. It is not known how many newborns have this condition.

Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)

LCHAD causes many different problems in the heart, liver and lungs. It can vary from a severe infant form which causes death to a mild form in childhood which causes muscle weakness and loss of vision. Treatment includes avoidance of certain foods and addition of others. During the pregnancy with affected babies, the mother may develop complications. It is not known how many newborns have this condition.

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)

MCAD is caused by the lack of an enzyme that breaks down fats into sugars. The baby with MCAD can go into shock or die very suddenly. Treatment is making sure the baby eats on a very regular schedule, even if sick. If the baby does not eat, glucose may be given in the hospital. The disorder occurs in approximately one of every 23,000 newborns.

Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)

MCKAT causes vomiting, dehydration, problems with the acid-base balance in the blood, and liver problems. Effective treatment and incidence in the newborn population are unknown.

Medium/short-chain hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)

M/SCHAD can cause low blood sugar if the infant does not eat regularly. Treatment includes giving glucose during an illness. It is not known how many newborns have this condition.

Mitochondrial trifunctional protein deficiency (TFP)

TFP causes poor muscle tone, failure to thrive and feeding problems leading to low blood sugar. Death may be caused by liver, heart or respiratory failure. Treatment includes avoidance of certain foods, consuming a special formula and ensuring that the baby eats regularly. It is not known how many newborns have this condition.

Multiple acyl-CoA dehydrogenase deficiency (MADD)

MADD, also known as glutaric acidemia type II (GA-II), has three common forms. The two infant forms present with problems with the acid-base balance in the blood, which may lead to death. One of the neonatal forms includes congenital anomalies. Effective treatment for these forms has not yet been determined. The other form is different in each child and may include low blood sugar and liver problems. Treatment of the late onset form includes diet, special formulas and regular feeding. It is not known how many newborns have this condition.

Short-chain acyl-CoA dehydrogenase deficiency (SCAD)

Infants with SCAD may have seizures, breathing problems, problems with the acid-base balance in the blood, poor feeding, vomiting or failure to thrive. Treatment includes diet and special formulas. The disorder occurs in one of every 40,000 to 100,000 newborns.

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

About half of all infants with VLCAD develop low blood sugar, and liver and heart problems, which often result in death. Others may not get sick unless they are not eating regularly and then there may or may not be liver or heart problems. Treatment includes a high carbohydrate, low fat diet, special formula, regular feeding and avoiding strenuous exercise. It is not known how many newborns have this condition.

Inborn Errors of Metabolism - Organic Acid Disorders

Cobalamin A,B cofactor deficiency (CBl A,B)

Cbl A, B, one of the methylmalonic acidemias, may cause problems with the acid-base balance in the blood. This causes sleepiness, coma and death in some children, though others will not have any problems. Treatment includes a special diet, antibiotics, special formula and liver and renal transplants. It is not known how many newborns have this condition.

Cobalamin C,D cofactor deficiency (CBl C, D)

Cbl C, D, one of the methylmalonic acidemias, may cause poor muscle tone; failure to thrive; seizures; developmental delay; and problems in the eye, bone marrow and kidneys. Treatment includes diet restrictions and supplementation. It is not known how many newborns have this condition.

Glutaric acidemia type I (GA-I)

Most infants with GA-I are born with or develop large heads, but the level of illness varies widely. Cerebral palsy and muscle weakness may develop. Prompt treatment at first signs of illness may prevent brain damage. A special diet may improve health. The disorder occurs in approximately one of every 40,000 Caucasian newborns.

3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)

HMG is caused by the lack of an enzyme that breaks down some proteins. It may cause failure-to-thrive or coma. HMG is treated with a special diet and giving additional glucose in case of a common illness such as cold or flu. It is not known how many newborns have this condition.

Isobutyryl-CoA dehydrogenase deficiency (IBCD)

IBCD can cause heart problems and anemia. Treatment includes a special diet, special formula and regular feedings. It is not known how many newborns have this condition.

Isovaleric acidemia (IVA)

Newborns with acute IVA can get sick in the first few days of life, having poor feeding, vomiting and problems with the acid-base balance in the blood. This can lead to coma and death. Those with chronic IVA may present later in infancy or childhood with similar symptoms. Treatment includes a special diet, special formula and regular feeding. The disorder occurs in approximately one of every 230,000 newborns.

Malonic acidemia (MA)

MA causes developmental delay and other symptoms including poor muscle tone, low blood sugar, problems with the acid-base balance in the blood, large heart, diarrhea, vomiting and seizures. Treatment includes special diet and formulas. It is not known how many newborns have this condition.

2-Methylbutyryl-CoA dehydrogenase deficiency (2-MBCD)

2-MBCD causes poor muscle tone, sleepiness and low blood sugar, leading to mental retardation, cerebral palsy and vision problems. Treatment includes a special diet with special formula. It is not known how many newborns have this condition.

3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)

3-MCC is caused by the lack of an enzyme that breaks down some proteins. Many children with this condition do not get sick, but some can go into coma or develop weak muscles. Treatment with the vitamin biotin or a special diet can help. The disorder occurs in approximately one of every 50,000 newborns.

3-Methylglutaconic acidemia (3-MGA)

3-MGA can cause mild to severe developmental and physical problems. Treatment includes special diet and formula. It is not known how many newborns have this condition.

2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (MHBD)

MHBD is usually diagnosed in boys. Symptoms include progressive loss of motor skills, and mental retardation with epilepsy may also develop. Treatment includes special diet with special formula. It is not known how many newborns have this condition.

Methylmalonyl-CoA mutase deficiency (MUT)

MUT, one of the methylmalonic acidemias, causes tiredness, failure to thrive, vomiting and dehydration, which can lead to profound problems with the acid-base balance in the blood, breathing problems, poor muscle tone and death in most children. Treatment includes special diet and formula. Most children eventually develop liver cancer making liver and kidney transplants necessary. The disorder occurs in approximately one of every 48,000 newborns.

Mitochondrial acetoacetyl-CoA thiolase deficiency (BKT)

Also known as beta-ketothiolase deficiency, BKT can cause vomiting, diarrhea, coma, mental retardation and death. Treatment involves a special diet and formula. It is not known how many newborns have this condition.

Multiple carboxylase deficiency (MCD)

Infants with MCD may refuse food leading to vomiting, breathing problems, poor muscle tone, seizures and tiredness, which can lead to coma and death. Treatment includes biotin supplementation. The disorder occurs in approximately one of every 87,000 newborns.

Propionic acidemia (PA)

Infants with PA refuse to feed leading to vomiting, tiredness, poor muscle tone and seizures. Treatment is complicated and includes special diet, formula, antibiotics and frequent feedings. The disorder occurs in approximately one of every 100,000 newborns.

Inborn Errors of Metabolism - Urea Cycle Disorders

Argininemia (ARG)

ARG causes irritability, poor feeding, vomiting and failure to thrive progressing to seizures, psychomotor retardation, hyperactivity, mental retardation and growth failure. ARG is treated with a low-protein diet and arginine-free protein supplements. The disorder occurs in approximately one of every 300,000 newborns.

Argininosuccinic acidemia (ASA)

ASA causes vomiting, tiredness, mental and physical retardation, convulsions and coma. ASA is treated with a low-protein diet and arginine supplements. The disorder occurs in approximately one of every 70,000 newborns.

Citrullinemia (CIT)

CIT can cause progressive tiredness, poor feeding, vomiting, "grunting" breathing, seizures, coma and death if not treated. Treatment consists of diet, supplementation and aggressive treatment of acute symptoms. The disorder occurs in approximately one of every 57,000 newborns.

Hyperammonemia/hyperornithinemia/homocitrullinemia (HHH)

HHH may cause muscle movement problems, occasional confusion, seizures, and mental retardation. Treatment includes dietary restrictions with special supplementation. It is not known how many newborns have this condition.

Infectious Disease

HIV (Human Immunodeficiency Virus)

HIV is the virus that causes AIDS. The test checks the infant's blood for the presence of HIV antibodies. If a baby has HIV antibodies, the mother is infected with HIV. In all positive cases, the baby has been exposed to the virus. HIV-exposed newborns need repeat testing to see if they are infected. Their mothers also need special medical care.

Inborn Errors of Metabolism

Amino Acid Disorders

Homocystinuria (HCY)

In HCY lack of an enzyme in the liver can produce mental retardation and eye and circulation problems. It is treated with a special diet. The disorder occurs in approximately one of every 391,000 newborns.

Hypermethioninemia (HMET)

HMET causes delayed movement development with muscle weakness that leads to death. Treatment includes a special diet with special formula. It is not known how many newborns have this condition.

Branched-chain ketonuria

This is called maple syrup urine disease (MSUD) because the urine smells like maple syrup. A person with MSUD cannot break down some food proteins because they are missing a certain enzyme. Treatment with a special diet can prevent life-threatening complications. The disorder occurs in approximately one of every 270,000 newborns.

Phenylketonuria

A kind of food protein (phenylalanine) cannot be broken down by the body due to lack of an enzyme. Brain damage, which would normally result, can be prevented by a special diet low in phenylalanine. The disorder occurs in approximately one of every 19,000 newborns.

Tyrosinemia (TYR)

Infants with TYR develop liver, heart and movement problems. Treatment is very complicated and includes diet and a special formula. Since liver damage begins before birth, liver transplant is recommended if possible before the baby is two years old. TYR occurs in approximately one of every 250,000 newborns.

Other Genetic Conditions

Biotinidase deficiency

Lack of an enzyme (biotinidase) prevents the body from properly absorbing or recycling the vitamin biotin. Severe skin rashes, vision and hearing impairment, and brain damage can be prevented by special biotin pills taken daily. The disorder occurs in approximately one of every 95,000 newborns.

Cystic fibrosis (CF)

CF is caused by a defect in the way salts travel through cell walls creating thicker mucus and saltier sweat in the body. CF results in many infections in the lungs. It also interferes with absorption of foods and nutrients, and often results in poor growth and poor weight gain. Medical treatment of CF is complicated. The disorder occurs in approximately one of every 4,000 newborns, with a higher frequency in Caucasians.

Galactosemia (GALT)

GALT is a type of galactosemia in which a milk sugar (galactose) cannot be broken down by the body due to the lack of an enzyme. A diet low in galactose can prevent life-threatening complications. The disorder occurs in approximately one of every 55,000 newborns.

Krabbe disease

Krabbe disease is caused by a defect in an enzyme (galactocerebrosidase). Krabbe disease affects the nervous system. Treatment consists of a bone marrow transplant early in life. The disorder occurs in approximately one of every 100,000 newborns.

The New York State Newborn Screening Program

A service provided by the State Department of Health to families with newborn babies.

Newborn Screening Program
Wadsworth Center
NYS Department of Health
Empire State Plaza
P.O. Box 509
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http://www.wadsworth.org/newborn/index.htm