IDPH
ILLINOIS DEPARTMENT OF PUBLIC HEALTH

NEWBORN SCREENING DISORDER PANEL
INFORMATION FOR PARENTS

WHAT DISORDERS ARE INCLUDED IN THE NEWBORN SCREEN?

All babies born in Illinois should be screened for the disorders listed below. Following is a short description of the disorders included in Illinois newborn screening. If you live in Illinois, but deliver your baby in another state, your baby may not be screened for these same disorders. You may want to discuss with your baby's doctor, how to obtain additional screening if necessary. This screening is most accurate soon after your baby is born, so it is important to make arrangements before your baby's birth, or as soon as possible after birth.

ADRENOLEUKODYSTROPHY

Adrenoleukodystrophy (ALD) is a rare disorder that affects the brain, nervous system, and adrenal glands. A baby born with adrenoleukodystrophy cannot break down certain fatty acids. These fatty acids build up within the body and can cause serious medical problems or even death. There is no cure for ALD, but early diagnosis can ensure babies receive treatment during the early stages of the disease.

AMINO ACID AND UREA CYCLE DISORDERS

PKU is one of the more familiar of the amino acid disorders. Babies with amino acid and urea cycle disorders cannot properly process amino acids, the building blocks of proteins. Without treatment, the buildup of these chemicals can cause serious medical problems affecting the baby's health and development, including damage to vital organs, seizures and coma. Treatment depends on the disorder the baby has, but may include special low protein diets, medical treatments and medications. A baby with an amino acid or urea cycle disorder must have regular medical care.

BIOTINIDASE DEFICIENCY

Biotinidase deficiency may occur in one of every 150,000 births. Babies with this problem cannot use a vitamin called biotin. Without treatment, the baby's growth and development will not be normal. A baby with biotinidase deficiency needs medicine containing biotin and regular medical care.

CONGENITAL ADRENAL HYPERPLASIA

Congenital adrenal hyperplasia (CAH) may occur in one of every 15,000 births. Most babies with this problem cannot produce enough of a hormone called cortisol. Sometimes, they also cannot produce enough of a salt-retaining hormone. Babies with CAH may develop medical problems including severe fluid loss (dehydration). Without enough cortisol, a baby's growth and development will not be normal. A baby with CAH is given medicine to replace these important hormones and needs regular medical care.

CONGENITAL HYPOTHYROIDISM

Congenital hypothyroidism may occur in one of every 3,000 newborns. A baby born with this problem does not make enough thyroid hormone, which is needed to help the baby grow normally and stay healthy. Without thyroid hormone, a baby's growth and mental development will not be normal. A baby with congenital hypothyroidism is treated with medicine to replace the missing thyroid hormone and needs regular medical care.

CYSTIC FIBROSIS

Cystic fibrosis (CF) may occur in one of every 4,000 newborns. A baby with CF produces thick, sticky mucus that causes respiratory and digestive problems. CF affects growth and damages a baby's lungs and other organs. A baby with CF is treated with special diet supplements and vitamins to improve growth. In addition, breathing treatments, medications and special exercises can help maintain respiratory function. A baby with CF needs regular medical care.

FATTY ACID OXIDATION DISORDERS

Babies with one of these disorders have trouble burning fat for energy. Without treatment, these disorders can lead to serious medical problems affecting the baby's health and development, including seizures, liver damage and coma. Treatment depends on the disorder the baby has, but may include special diets, supplements and medications. Babies with fatty acid oxidation disorders must have regular medical care.

GALACTOSEMIA

Galactosemia may occur in one of every 60,000 newborns. A baby with galactosemia cannot digest a certain part of milk sugar called galactose. The resulting buildup of galactose in the baby's body can cause serious medical problems, including liver and brain damage. Babies with galactosemia are treated with a special lactose-free diet and need regular medical care. Early treatment can help prevent physical and mental damage.

LYSOSOMAL STORAGE DISORDERS

Babies with lysosomal storage disorders cannot break down certain waste products. These waste products build up within the body and cause serious medical problems affecting the baby's health and development. Treatment depends on the specific disorder, and can include medications, enzyme replacement therapy, or may require stem cell or cord blood transplant very early in life. A baby with a lysosomal storage disorder needs immediate and on-going medical care.

ORGANIC ACID DISORDERS

Babies with organic acid disorders cannot remove certain waste products from their blood. Without treatment, the buildup of these waste products causes serious medical problems affecting the baby's health and development, including seizures, coma and brain damage. Treatment depends on the disorder the baby has, but may include special diets, supplements and medications. A baby with an organic acid disorder must have regular medical care.

PHENYLKETONURIA

Phenylketonuria (PKU) may occur in one of every 12,000 births. Babies with PKU, an amino acid disorder, cannot properly use a substance found in proteins, phenylalanine. If this problem is not found and treated early, PKU can cause developmental delays and mental retardation. A baby with PKU is given a special formula and diet and needs regular medical care.

SEVERE COMBINED IMMUNE DEFICIENCY

Severe combined immune deficiency (SCID) is a rare, genetic disorder affecting the white blood cells responsible for fighting off infections. Most babies with SCID appear healthy after birth because they still have their mother's antibodies protecting them. But without treatment, a mild infection can be life threatening. The most effective treatment for SCID is a bone marrow transplant with regular medical care.

SICKLE CELL DISEASE, TRAIT AND RELATED CONDITIONS

Each year it is estimated that more than 80 babies in the state will be diagnosed with sickle cell disease, and 40 more babies will be diagnosed with another kind of sickling disorder. Sickling disorders can cause pain, damage vital organs and lead to serious infections. Babies with these disorders are treated with antibiotics and need immunizations and regular medical care to help prevent some of these medical problems. The newborn screen for sickle cell disease also may find other related conditions that may or may not need treatment.

SPINAL MUSCULAR ATROPHY

Spinal muscular atrophy (SMA) is a genetic disease that affects the body's nervous system and affects muscle strength and movement. There is a loss of motor neurons in the spinal cord and the brainstem which leads to atrophy of the muscles used in breathing, swallowing, crawling, walking, sitting up, and head control. Symptoms of the disease depends on the type of SMA the baby has. Although there is no absolute cure for SMA, currently available treatments and supportive therapies have been shown to improve development and quality of life. Starting treatment with drug therapy earlier has been shown to improve your child's long-term outcome.

Rev. 6/2020