What is cystic fibrosis?
Cystic Fibrosis (CF) is a condition that affects breathing and digestion or the body’s use of food and liquids that the baby eats and drinks. People with CF have very thick mucus, which can clog the lungs and digestive system leading to a bad cough, life-threatening lung infections, and poor weight gain and growth. There are many kinds of treatments for CF that involve daily breathing therapy and medication. Early diagnosis and treatment can improve growth, lung function and add years to a child’s life.

What is Severe Combined Immunodeficiency (SCID)?
SCID is an inherited condition in which the baby’s immune system does not work well and the body is unable to fight off serious infections. If found and treated early, the baby can live and have a healthier life.

What are amino acid disorders, organic acid disorders and fatty acid oxidation disorders?
Each of these conditions affects the body’s ability to produce energy from food. These conditions cause the buildup of toxins or poisons in the body. Two of the most common conditions in this group are phenylketonuria (PKU) and medium chain acyl-CoA dehydrogenase deficiency (MCADD).

What is phenylketonuria (PKU)?
PKU is a disorder that affects the body’s ability to break down phenylalanine, which is part of the protein found in foods. The build up of phenylalanine in the blood can affect brain development and cause severe mental retardation and behavioral problems. Treatment places the baby on a special infant formula. The child will follow a special diet lifelong.

What is medium chain acyl-CoA dehydrogenase deficiency (MCADD)?
MCADD is a condition that affects the body’s ability to make energy from stored fats. Babies born with this condition seem normal at birth, but they can suddenly have seizures and go into a coma if they have not eaten for a few hours. Without emergency treatment they may die or be left with developmental problems. Treatment includes avoiding long periods without eating, especially during times of illness.

If you have any concerns about the results of the screening tests, please contact your baby’s doctor.
Newborn screening is not a diagnostic test. Although a normal result is very reassuring, it does not guarantee that a baby does not have one of these disorders. If a baby develops symptoms of one of these disorders, the baby should be further examined, even if the newborn screen was normal.

Children with Special Health Care Needs Help Line
1-800-737-3028

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Explaining the Disorders

For each of these conditions, early diagnosis, medical care and treatment can prevent or reduce serious medical problems and may even save your baby’s life.

What is primary hypothyroidism?
The thyroid gland, which is located in the neck, makes a hormone that is important for normal growth, development, and learning. Primary hypothyroidism occurs when a baby’s thyroid gland does not make enough thyroid hormone. Treatment is giving or replacing the hormone that is in short supply with a medicine the baby can take by mouth.

What is galactosemia?
Galactosemia is a condition that does not allow the body to use a sugar called galactose that is found in milk. A baby with galactosemia can become very ill after just a few days of drinking breast milk or formula that contains this sugar. Some early signs and symptoms may include vomiting, yellowing of the skin or failure to grow or thrive. Treatment involves changing to a galactose-free (soy-based) formula which is easy to get from the store.

What is congenital adrenal hyperplasia (CAH)?
CAH is a group of conditions that affect the amount of hormones produced by the adrenal glands, which are located above the kidneys. These hormones are needed for life and play an important role in sexual development. Symptoms of CAH can be life-threatening. Things to look for in your baby may include weakness, dehydration or even shock. If your baby has CAH, the baby’s body cannot make these hormones. Treatment is by giving or replacing the hormone to the baby which is usually a medicine you can take by mouth.

What is sickle cell disease?
Sickle cell disease is a problem with red blood cells. Sickle cell disease affects the part of the red blood cell that carries oxygen to all parts of the body. People with sickle cell disease can have serious health problems that can lead to anemia (low red blood cell count), severe pain, life-threatening infections, strokes and many hospitalizations. Penicillin taken by mouth helps prevent and treat some symptoms.

What is biotinidase deficiency?
Biotinidase deficiency is a condition that affects the body’s ability to recycle biotin, a common vitamin found in many foods and so there is not enough biotin in the body. Biotinidase deficiency can cause seizures, mental retardation, skin rash, hair and hearing loss and even death. Treatment consists of daily biotin which is a medicine taken by mouth.