What disorders are screened for by the newborn screen?

Endocrine Disorders

The endocrine system is important to regulate the hormones in our bodies. Hormones are special signals sent to various parts of the body. They control many things such as growth and development. The goal of newborn screening is to identify these babies early so that treatment can be started to keep them healthy. To learn more about these specific disorders please click on the name of the disorder below:

English: Congenital Adrenal Hyperplasia
Esapnol Hiperplasia Suprarrenal Congenital

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- http://www.newbornscreening.info/Parents/otherdisorders/CAH.html
- http://www.newbornscreening.info/spanish/parent/Other_disorder/CAH.html

- Congenital Hypothyroidism **(Hipotiroidismo Congénito)**
- http://www.newbornscreening.info/Parents/otherdisorders/CH.html
- http://www.newbornscreening.info/spanish/parent/Other_disorder/CH.html

Hematologic Conditions

Hemoglobin is a special part of our red blood cells. It is important for carrying oxygen to the parts of the body where it is needed. When people have problems with their hemoglobin they can have intense pain, and they often get sick more than other children. Over time, the lack of oxygen to the body can cause damage to the organs. The goal of newborn screening is to identify babies with these conditions so that they can get early treatment to help keep them healthy. To learn more about these specific disorders click here (XXX).

- Sickle Cell Anemia (**Anemia de Célula Falciforme)**
- http://www.newbornscreening.info/Parents/otherdisorders/SCD.html
- http://www.newbornscreening.info/spanish/parent/Other_disorder/SCD.html

- SC Disease (See Previous Link)

- Sickle Beta Thalassemia (See Previous Link)

Enzyme Deficiencies

Enzymes are special proteins in our body that allow for chemical reactions to take place. Enzymes are necessary for proper body function and play an important role in growth and development. To learn more about disorders on the newborn screen caused by an enzyme problem click here (XXX)

- Galactosemia (**Galactosemia)**
- http://www.newbornscreening.info/Parents/otherdisorders/Galactosemia.html
  http://www.newbornscreening.info/spanish/parent/Other_disorder/Galactosemia.html

- Biotinidase Deficiency
- http://ghr.nlm.nih.gov/condition=biotinidasedeficiency

Cystic Fibrosis (Fibrosis Quística)

Cystic Fibrosis is a condition caused when the body has problems moving around salt and chloride ions (Ions are chemicals important for specific body functions, like moving water around in the body). These problems with ion movement can cause a baby to build up thick mucus in their lungs. This thickened mucus makes it hard for babies to breathe normally and puts them at increased risk for infections. The mucus can also cause blockages in the gut, which prevent a baby from being able to break down their food properly. When this happens, the baby cannot use the vitamins and nutrients properly and may have growth and weight gain problems. The goal of newborn screening is to identify babies with this condition early so they can get treatment to help with their lung function and to help them to break down their food. To learn more about this condition click here (XXX)

http://www.newbornscreening.info/Parents/otherdisorders/CF.html

http://www.newbornscreening.info/spanish/parent/Other_disorder/CF.html

Amino Acid Disorders-

Amino acids are the building blocks that make up the proteins in the food we eat, and the formula/breast milk babies drink. Some medical conditions can occur because the body cannot use these amino acids properly. This can cause them to build up to dangerous levels in our body. When this happens it can cause damage to a baby's brain and other organs. Sometimes babies with these conditions will get sick very fast, but other times newborn babies appear very healthy and they do not show symptoms until they are a little older. The goal of newborn screening is to identify babies with these conditions early, so that proper treatment can be started to help keep them healthy. To learn more about these specific disorders click here (XXX):

- **Argininosuccinate aciduria (Deficiencia de Argininosuccinato Liasa)**
  http://www.newbornscreening.info/Parents/aminoaciddisorders/ASAL.html
- **http://www.newbornscreening.info/spanish/parent/Amino_acid/ASAL.html**

  Citrullinemia (Citrulinemia)
  http://www.newbornscreening.info/Parents/aminoaciddisorders/ASAS.html
- **http://www.newbornscreening.info/spanish/parent/Amino_acid/ASAS.html**

- **Homocystinuria http://ghr.nlm.nih.gov/condition=homocystinuria**

- **Maple syrup urine disease (branched-chain ketoacid dehydrogenase deficiency) (Enfermedad de la Orina con Olor a Jarabe de Arce)**
- **http://www.newbornscreening.info/Parents/aminoaciddisorders/MSUD.html**
- **http://www.newbornscreening.info/spanish/parent/Amino_acid/MSUD.html**

- **Phenylketonuria/ hyperphenylalaninemia (Fenilcetonuria)**
- **http://www.newbornscreening.info/Parents/aminoaciddisorders/PKU.html**
- **http://www.newbornscreening.info/spanish/parent/Amino_acid/PKU.html**

- **Tyrosinemia Type I (Tirosinemia, tipo 1)**
- **http://www.newbornscreening.info/Parents/aminoaciddisorders/Tyrosinemia.html**
- **http://www.newbornscreening.info/spanish/parent/Amino_acid/Tyrosinemia. html**

- **Tyrosinemia Type I/II/III**
- **http://ghr.nlm.nih.gov/condition=tyrosinemia**

- **Argininemia (Deficiencia de Arginasa)**
  http://www.newbornscreening.info/Parents/aminoaciddisorders/argininemia. html
- **http://www.newbornscreening.info/spanish/parent/Amino_acid/argininemia. html**

Organic Acid Disorders

Organic Acid disorders are caused when the body cannot break down the protein in our food correctly because there is a problem with an enzyme. (Enzymes help our body to perform chemical reactions, like breaking down our food). When a person cannot break down their protein correctly, harmful substances can build up in the blood and the urine. When this happens, it can cause damage to a baby's brain and other organs. Sometimes babies with these conditions will get sick very fast, but other times newborn babies appear vary healthy and they do not show symptoms until they are a little older. The goal of newborn screening is to identify babies with these conditions early, so that proper treatment can be started to help keep them healthy. To learn more about these specific disorders click here (XXX):

- **Malonic Acidemia**
- **http://ghr.nlm.nih.gov/condition=malonylcoenzymeadecarboxylasedeficiency**

- **Propionic Acidemia (Acidemia propiónica)**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/PA.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/PA.html**

- **Cobalamin C deficiency (Acidemia metilmalónica con homocistinuria)**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/MMA_HCU.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/MMA+HCU.html**

- **Methylmalonic Acidemia (Acidemia metilmalónica)**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/MMA.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/MMA.html**

- **Isovaleric Acidemia (Acidemia isovalérica)**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/IVA.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/IVA.html**

- **2- Methylbutyryl-CoA Dehydrogenase Def.**
- **Glutaric Acidemia (GA-1; Glutaryl CoA Lyase Deficiency) (Acidemia glutárica, tipo 1)**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/GA1.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/GA1.html**

- **2-Methyl-3Hydroxybutyryl-CoA Dehydrogenase Deficiency (Deficiencia de 2-Metilbutiril CoA Deshidrogenasa)**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/2MBC.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/2MBCD.html**

- **Multiple Carboxylase Deficiency (Deficiencia de Holocarboxilasa Sintetasa)**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/HCSD.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/HCSD.html**
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- **Beta Ketothiolase (Mitochondrial Acetyl-CoA Thiolase Deficiency**
- **Deficiencia de beta-cetotiolasa**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/BKD.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/BKD.html**

- **3-Hydroxy-3Methylglutaric aciduria**
- **Deficiencia de 3-hidroxi-3-metilglutaril-CoA liasa**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/HMGCoA. html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/HMG.html**
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- **3-MCC (3 Methylcrotonyl- CoA carboxylase deficiency)**
- **Deficiencia de 3-metilcrotonil CoA carboxilasa**
- **lhttp://www.newbornscreening.info/Parents/organicaciddisorders/3MCC.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/3MCC.html**

- **Isobutyryl-CoA Dehydrogenase Deficiency**
- **Deficiencia de isobutiril-CoA deshidrogenasa**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/ICoA.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/IBD.html**

- **Glutaric Acidemia Type II**
- **Acidemia glutárica, tipo 2**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/GA2.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/GA2.html**

Fatty Acid Oxidation Disorders

Our body gets energy from the food we eat; it breaks down sugar, protein, and fat to make energy. The body typically breaks down sugar and protein first, and then starts to breakdown the fat. When a person/baby is not eating (fasting), the body must use the fat the body has stored up for energy. Individuals who have a fatty acid oxidation disorder have problems with an enzyme that help to break down their fat. (Enzymes help our body to perform chemical reactions, like breaking down our food). When the body cannot use our fat stores for energy, the body has no other way of getting energy. Without this energy a person cannot survive. Babies who have a fatty acid oxidation disorder can get very sick very fast if they are not given some type of food or nutrient for energy. If your baby is not fasting, he/she may look very healthy and you may not know your baby has one of these conditions until it is too late. The goal of newborn screening is to identify babies with these conditions early, so that proper treatment can be started to help keep them healthy. To learn more about these specific disorders click here (XXX):

- **SCAD- Short chain acyl-CoA dehydrogenase deficiency**
- **Deficiencia de acil-CoA deshidrogenasa de cadena corta**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/SCADD.html**
- **http://www.newbornscreening.info/spanish/parent/Fatty_acid/SCADD.html**

- **SCHAD- Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency**
- **Deficiencia de 3-hidroxiacil-CoA deshidrogenasa de cadena corta**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/SCHADD.**
  http://www.newbornscreening.info/spanish/parent/Fatty_acid/SCHADD.html

- **MCAD- Medium chain acyl-CoA dehydrogenase deficiency**
- **Deficiencia de acil-CoA deshidrogenasa de cadena media**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/MCADD.html**
- **http://www.newbornscreening.info/spanish/parent/Fatty_acid/MCADD.html**

- **LCHAD- Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency**
- **Deficiencia de acil-CoA deshidrogenasa de cadena media**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/LCHADD.ht ml**
- **http://www.newbornscreening.info/spanish/parent/Fatty_acid/LCHADD.html**

- **VLCAD- Very long chain acyl-CoA Dehydrogenase**
- **Deficiencia de Acil-CoA deshidrogenasa de cadena muy larga**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/VLCADD.html**
- **http://www.newbornscreening.info/spanish/parent/Fatty_acid/VLCADD.html**

Carnitine Uptake Deficiency

- **Deficiencia de la proteína transportadora de carnitina**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/Carnitine.html**
- **http://www.newbornscreening.info/spanish/parent/Fatty_acid/CTD.html**

- **CPT I (Carnitine Palmitoyltransferase Deficiency Type 1)**
- **Deficiencia de carnitina palmitoiltransferasa, tipo 1A**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/CPT1.html**
- **http://www.newbornscreening.info/spanish/parent/Fatty_acid/CPT1A.html**

- **CPT II (Carnitine Palmitoyltransferase Deficiency Type 1)**
- **Deficiencia de carnitina palmitoiltransferasa, tipo 2**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/CPT2.html**
- **http://www.newbornscreening.info/spanish/parent/Fatty_acid/CPT2.html**

- **Trifunctional Protein Deficiency**
- **Deficiencia de proteína trifuncional**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/TFP.html**
- **http://www.newbornscreening.info/spanish/parent/Fatty_acid/TFP.html**
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- **Carnitine Acyl-Carnitine Translocase Deficiency**
- **Deficiencia de carnitina-acilcarnitina translocasa**
- **http://www.newbornscreening.info/Parents/fattyaciddisorders/CAT.html**
- **http://www.newbornscreening.info/spanish/parent/Fatty_acid/CAT.html**
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- **2,4-dienoyl-CoA Reductase**

- **Isobutyryl-CoA Dehydrogenase Deficiency**
- **Deficiencia de isobutiril-CoA deshidrogenasa**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/ICoA.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/IBD.html**

- **Glutaric Acidemia Type II**
- **Acidemia glutárica, tipo 2**
- **http://www.newbornscreening.info/Parents/organicaciddisorders/GA2.html**
- **http://www.newbornscreening.info/spanish/parent/Organic_acid/GA2.html**

Urea Cycle Disorders

The urea cycle is an important function that the cells in the body use to get rid of a chemical called ammonia; ammonia builds up as a result of the food we eat. Normally, the body uses the cycle to remove this extra ammonia and we get rid of it when we use the bathroom. Children that have urea cycle disorders cannot get rid of the extra ammonia. When this happens it can cause damage to a baby's brain and other organs. Sometimes babies with these conditions will get sick very fast, but other times newborn babies appear very healthy and they do not show symptoms until they are a little older. The goal of newborn screening is to identify babies with these conditions early, so that proper treatment can be started to help keep them healthy. Newborn screening cannot detect all urea cycle disorders. To learn more about these specific disorders we can screen for click here (XXX):

- **Argininemia (Arginase Deficiency)**
- **http://ghr.nlm.nih.gov/condition=arginasedeficiency**
- **Argininosuccinic Aciduria**
- **http://ghr.nlm.nih.gov/condition=argininosuccinicaciduria**
- **Citrullinemia Type I and II**
- **http://ghr.nlm.nih.gov/condition=citrullinemia**

Severe Combined Immunodeficiency Disorder (Inmunodeficiencia combinada grave e)

Severe Combined Immunodeficiency Disorder (SCID) is the name for a group of disorders that cause babies to be born without a working immune system. Newborns with SCID may seem healthy at first because they are getting protection from the mother's immune system for the first few weeks of life. However, without treatment, common infections and vaccines can be life threatening for these infants. Further testing is needed to determine if your baby may have trouble fighting infections. To learn more about SCID, please click on the link below

https://www.newbornscreening.info/Parents/otherdisorders/SCID.html
https://www.newbornscreening.info/spanish/parent/Other_disorder/SCID.html

Spinal Muscular Atrophy (SMA) (atrofia muscular espinal) is a genetic disease that effects the part of the nervous system that controls voluntary muscle movement. There are many types of SMA. The type differ in age of onset and severity of muscle weakness This disease in it's severe form robs people of physical strength by affecting the motor nerve cells in the spinal cord, taking away the ability to walk, eat, or breathe. Without any treatment, affected children usually die before the age of two years. To learn more about SMA, please click on the link below

https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Spinal-Muscular-Atrophy-Fact-Sheet

Lysosomal Storage Disease (LSD) (enfermedad de almacenamiento lisosomal) are a group of approximately 50 rare inherited metabolic diseases resulting from an abnormal build-up of various toxic materials in the body's cells due to defects or absence of specific enzymes. These enzymes are responsible for breaking down complex fats and sugars. These disorders may affect different parts of the body, including the skeleton, brain, skin, heart, and central nervous system.

MD NBS Laboratory screens for three LSDs: Pompe Disease. Fabry Disease and Hurler Syndrome.

- **Pompe disease (enfermedad de Pompe**) is an inherited disorder caused by the buildup of a complex sugar called glycogen in the body's cells. This disease occurs when there isn't any or enough enzyme alpha-glucosidase. This enzyme is responsible for breaking down complex sugars The accumulation of glycogen in certain organs and tissues, especially muscles, impairs their ability to function normally. To learn more about Pompe, please click on the link below:

- **https://www.newbornscreening.info/Parents/otherdisorders/Pompe.html**
- **https://www.newbornscreening.info/spanish/parent/Other_disorder/Pompe.html**

- **Fabry disease** (**Enfermedad de Fabry)** is a rare genetic disorder caused by a buildup of particular type of fat in the body's cells. This disease occurs when there isn't any or enough enzyme alpha-glucosidase A. This enzyme is necessary for breakdown of particular lipid in the body. When the metabolism of this lipid does not occur, particular lipid accumulates in body cells. The cell damage causes multiple symptoms including potentially life-threatening consequences. To learn more about Fabry, please click on the link below:

- **https://newbornscreening.info/Parents/otherdisorders/Fabry.html**
- **https://www.newbornscreening.info/spanish/parent/Other_disorder/Fabry.html**

- **Hurler syndrome (síndrome de hurler**), also known as mucopolysaccharidosis type I (**MPS I**), is a rare genetic disorder that results in the buildup of mucopolysaccharides due to a deficiency or absence of enzyme alpha-L iduronidase. This enzyme is responsible for the degradation of Glycosamineoglycans (GAGs). Accumulation of GAGs may cause skeletal abnormalities, cognitive impairment, heart disease, respiratory problems, enlarged liver and spleen, characteristic facies and reduced life expectancy.
  To learn more about Hurler (MPS I), please click on the link below:

- **https://newbornscreening.info/Parents/otherdisorders/MPSI.html#1**