

# Pennsylvania Newborn Screening and Follow-Up Program (NSFP)

## Conditions Mandated for Screening and Follow-up by NSFP

- 1. Congenital Adrenal Hyperplasia (CAH)** (Kon-JEN-i-tal Ah-DRE-nal Hi-per-PLA-zhah)  
Babies born with this condition have a defect in an important substance (enzyme) the body needs. CAH can cause dehydration, shock and even death within a few days of birth. Medical problems can be prevented when treatment is started soon after birth.
- 2. Congenital Hypothyroidism (CH)** (Kon-JEN-i-tal Hi-po-THI-roid-ism)  
Babies born with this problem do not have a thyroid hormone. They may look healthy. If not detected, the condition can cause poor growth and mental retardation. Giving the baby special medicine every day can prevent these results.
- 3. Galactosemia (GALT)** (Gah-LAK-toe-SEE-mee-ah)  
Babies with this disorder cannot digest galactose. Galactose is a simple sugar found in breast milk, many formulas and milk products. This condition can harm the baby's eyes. It can cause serious liver and brain damage. Giving the baby a special milk-free diet as soon as the condition is found can prevent problems.
- 4. Maple Syrup Urine Disease (MSUD)**  
Babies born with MSUD cannot digest part of a food protein. Without treatment, MSUD can cause severe mental retardation or even death shortly after birth. To prevent these results, babies are given a special formula and diet.
- 5. Phenylketonuria (PKU)** (FEN-nil-KEE-tone-u-REE-ah)  
Babies who are born with PKU cannot digest a different part of a food protein. Untreated, PKU causes nerve and brain cell damage, which can result in mental retardation. This damage can be prevented when a baby gets a special formula and diet.
- 6. Sickle Cell Anemia (Hb SS-Disease)**  
Sickle cell disease is an inherited blood disorder that affects red blood cells. Sometimes these red blood cells become sickle-shaped (crescent shaped) and have difficulty passing through small blood vessels. Health maintenance for patients with sickle cell disease starts with early diagnosis, preferably in the newborn period and includes penicillin prophylaxis, vaccination against pneumococcus bacteria and folic acid supplementation.

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### Acylcarnitine Disorders

#### Organic Acid Disorders (OAs)

Babies with these inherited conditions are unable to break down the proteins they ingest. If untreated, OAs may cause breathing problems, seizures, brain swelling, stroke and coma, sometimes leading to death. To prevent these problems, babies are given a special formula, diet and sometimes medication.

Isovaleric Acidemia	(IVA)
Glutaric Acidemia Type I	(GA I)
3-Hydroxy 3-Methyl Glutaric Aciduria	(HMG)
Multiple Carboxylase Deficiency	(MCD)
Methylmalonic Acidemia (Mutase Deficiency)	(MUT)
3-Methylcrotonyl-CoA Carboxylase Deficiency	(3MCC)
Methylmalonic Acidemia	(Cbl A,B)
Propionic Acidemia	(PROP)

## **Beta-Ketothiolase Deficiency**

**(BKT)**

## **Fatty Acid Oxidation Disorders (FAODs)**

Babies with these inherited conditions are unable to break down fat. If untreated, FAODs may lead to serious complications affecting the liver, heart, eyes and general muscle development and possibly to death. To prevent these problems, treatment may vary and could include medications, diet modifications and avoiding long periods without food.

**Medium-Chain Acyl-CoA Dehydrogenase Deficiency**

**(MCAD)**

**Very Long-Chain Acyl-CoA Dehydrogenase Deficiency**

**(VLCAD)**

**Long-Chain L-3-Hydroxy Acyl-CoA Dehydrogenase**

**(LCHAD)**

**Deficiency**

**Trifunctional Protein Deficiency**

**(TFP)**

**Carnitine Uptake Defect**

**(CUD)**

## **Amino Acid Disorders (AAs)**

Babies with these inherited conditions are unable to process certain amino acids. If untreated, AAs can result in muscle weakness, breathing problems, seizures, swelling of the brain, coma and sometimes death. To prevent these problems, babies are given a special formula, diet and sometimes medication.

**Homocystinuria**

**(HCY)**

**Citrullinemia**

**(CIT)**

**Argininosuccinic Acidemia**

**(ASA)**

**Tyrosinemia Type I**

**(TYR I)**

## **Hemoglobinopathies (Sickle Cell Disease and other Hemoglobin Diseases)**

Sickle cell disease and other hemoglobin diseases are a group of genetic conditions that cause abnormalities with the blood. This leads to problems with blood circulation and anemia. Infants and children with sickle cell disease can die from lung and brain infections. Early treatment to prevent infections greatly reduces the chance of sickness or death. Sickle cell trait may also be identified through the screening.

**Sickle-C Disease**

**(Hb SC-Disease)**

**S-Beta Thalessemia**

**(Hb SA)**

## **Others**

**Biotinidase Deficiency (BIOT)** (Bio-tin-a daze de-fish-en-see)

Babies with this inherited condition have a lack of an enzyme called biotinidase. Without treatment, this disorder may lead to seizures, developmental delays, eczema and hearing loss. With early diagnosis and treatment, all symptoms can be prevented.

**Cystic Fibrosis (CF)**

Babies with this inherited condition have thick, sticky mucus and fluid build-up in certain organs, especially the lungs and pancreas. Some symptoms include repeated lung infections, poor weight gain and growth. If untreated, CF may cause serious health problems that could lead to early death. Many symptoms of CF can be controlled with medication and treatments.

**Severe Combined Immunodeficiency (SCID)**

SCID is a primary immune deficiency. The defining characteristic is usually a severe defect in both the T and B lymphocyte systems, resulting in the onset of one or more serious infections within the first few months of life. These infections may include pneumonia, meningitis or bloodstream infections. Many hospitals in Pennsylvania are now offering SCID testing.