



## Amino Acid Disorders Information for Parents

### ➤ Overview

Amino acid disorders are a group of rare, inherited conditions that affect infants from birth. They are caused by enzymes that do not work properly. Protein is made up of smaller building blocks, called amino acids. A number of different enzymes are needed to process these amino acids for use by the body. Because of missing or non-working enzymes, people with amino acid disorders cannot process certain amino acids. These amino acids, along with other toxic substances, then build up in the body and cause problems.

### ➤ Kansas Newborn Screening for amino acid disorders

The newborn screening program in the State of Kansas is designed to screen for six different amino acid disorders. These disorders include:

|  |                                  |
|--|----------------------------------|
| Phenylketonuria (PKU)                    | Maple Syrup Urine Disease (MSUD) |
| Homocystinuria (HCY)                     | Tyrosinemia, type I (TYR I)      |
| Argininosuccinate Lyase Deficiency (ASA) | Citrullinemia (CIT)              |

### ➤ Why is newborn screening done for amino acid disorders?

Newborn screening is done for amino acid disorders so that babies with these conditions can be diagnosed and treated quickly. Immediate diagnosis and treatment of amino acid disorders is important for normal development and health. Without prompt diagnosis and treatment, infants with amino acid disorders will develop varying degrees of developmental delay or intellectual disabilities, medical complications and may even experience death.

### ➤ Does a positive result from the Kansas Newborn Screening Lab mean that my baby has an amino acid disorder?

**No, not necessarily.** Newborn screening identifies babies at increased risk for having one of these disorders. Additional tests will need to be done to determine if the baby actually has an amino acid disorder.

### ➤ What are the signs and symptoms of amino acid disorders?

The age that symptoms start and the types of symptoms that a person has vary. Many babies with these conditions will appear normal at birth. Some of the disorders will cause developmental delay or intellectual disabilities if not treated promptly. Other newborns may develop symptoms such as poor appetite, sleepiness, vomiting, or irritability. If the condition is not treated promptly, babies can develop more serious problems including breathing problems, seizures, swelling of the brain, or even coma or death.

### ➤ Is there a cure for amino acid disorders?

No, there is no cure for amino acid disorders. However, the outcome is best in infants who are treated early and continue with life-long treatment. Treatment usually consists of a special diet and sometimes medications or supplements.

### ➤ Where can I get additional information?

Kansas Newborn Screening at [www.kdheks.gov/newborn\\_screening/info\\_parents.htm](http://www.kdheks.gov/newborn_screening/info_parents.htm)  
Save Babies Through Screening Foundation at [www.savebabies.org](http://www.savebabies.org)  
Screening, Technology and Research in Genetics at [www.newbornscreening.info](http://www.newbornscreening.info)