

## **Amino Acidemias**

### **What are Amino Acidemias?**

Amino acidemias are inherited conditions that affect the way a person's body uses a part of food called amino acids. A person with an amino acidemia cannot breakdown a specific amino acid in food. Amino acids are needed for proper growth and development, but too much can cause serious health problems. In the case of an amino acidemia, a specific amino acid(s) builds up in the blood, and may penetrate and damage the brain and other organs of the body. The high levels of amino acid(s) ultimately cause serious health problems.

### **How Do Amino Acidemias Affect a Child?**

The symptoms of these conditions vary and depend on the type of amino acidemia.

### **What Causes Amino Acidemias?**

The amino acidemias Tennessee screens for are genetic conditions caused by changes in certain genes. These genes are responsible for making enzymes. These enzymes are responsible for breaking down amino acids. When there is an alteration in these genes, enzyme levels go down and amino acids build up in the blood.

Amino acidemias are inherited in an autosomal recessive pattern, which means two copies of the same gene must be changed for a person to be affected with an amino acidemia. Most often, the parents of a child with an autosomal recessive condition are not affected because they are "carriers," with one copy of the changed gene and one copy of the normal gene.

When both parents are carriers, there is a one-in-four (or 25%) chance that both will pass a changed gene on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25%) chance that they will each pass on a normal gene, and the child will be free of the condition. There is a two-in-four (or 50%) chance that a child will inherit a changed gene from one parent and a normal gene from the other, making that child a carrier like the parents. These chances are the same in each pregnancy for the same parents.

### **Is There a Test for Amino Acidemias?**

Yes. Babies are tested through newborn screening for amino acidemias before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to find out if it has more than a normal amount of certain amino acids.

There are various types of amino acidemias. The following is a list of amino acidemias that can be screened for:

- 5-Oxoprolinuria
- Argininemia (ARG)
- Argininosuccinate Lyase deficiency (ASA)
- Carbamoylphosphate Synthetase deficiency (CPS)
- Citrullinemia (CIT) Type I or II
- Homocystinuria
- Hyperammonemia/Orthithinemia/Citrullinemia (HHH)
- Maple Syrup Urine Disease (MSUD)
- Nonketonic Hyperglycinemia
- Phenylketonuria (PKU)
- Tyrosinemia Type I
- Tyrosinemia Type II

### **Can Amino Acidemia Symptoms Be Prevented?**

In many cases, most symptoms of an amino acidemia can be prevented by diet restrictions, replacement formula and/or prescription medication. Children and adults with amino acidemias

require follow-up care at a medical center or clinic that specializes in these types of metabolic conditions. Each treatment depends on the particular diagnosis. In addition, regular blood tests are used to monitor an individual's health.

**DISCLAIMER:** The information contained on this page is not intended to replace the advice of a genetic metabolic medical professional.

## Resources:



PO Box 1244  
Mansfield, MA 02048  
Phone: 877-996-2723  
[www.pku-allieddisorders.org/home.htm](http://www.pku-allieddisorders.org/home.htm)

**LOW-PROTEIN.COM**

Email: [lopro@webuniverse.net](mailto:lopro@webuniverse.net)  
[www.lowprotein.com](http://www.lowprotein.com)

## References:

- American Academy of Pediatrics (1996): Newborn Screening Fact Sheets (RE9632). Pediatrics 98:473-501.  
<http://aappolicy.aappublications.org/cgi/reprint/pediatrics;98/3/473.pdf>
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- Scriver, C.R. and Kaufman, S (2001) Mitochondrial Fatty Acid Oxidation Disorders. In: Scriver, C.R., Kaufman, S., Eisensmith, E., Woo S.L.C., Vogelstein, B. Childs, B. (eds) The Metabolic and Molecular Bases of Inherited Disease, 8th ed. McGraw-Hill, New York, Ch.101 pg. 2297-2326.