

## **Argininosuccinic Aciduria (ASA)**

A urea cycle disorder

### ***What is it?***

Argininosuccinic Aciduria (also known as ASA) is an inherited urea cycle disorder. People with urea cycle disorders, like ASA, cannot properly break down and get rid of certain components of protein. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down the protein, certain amino acids and a chemical called ammonia build up in the blood and urine and cause problems when a person eats normal amounts of protein, or becomes sick.

### ***What are the symptoms?***

People with ASA may appear normal at birth. After a few days of life, a newborn with ASA may develop poor feeding, lack of energy, vomiting, problems breathing, or seizures. If left untreated, brain damage, coma, and death will occur. Many symptoms of ASA can be prevented by immediate treatment and lifelong management. People with ASA typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

### ***Inheritance and frequency***

ASA is inherited in an autosomal recessive manner. This means that for a person to be affected with ASA, he or she must have inherited two non-working copies of the gene responsible for causing ASA. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have ASA. Typically, there is no family history of ASA in an affected person. About 1 in 70,000 babies born have ASA.

### ***How is it detected?***

ASA may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

### ***How is it treated?***

ASA is treated by eating a diet low in protein and drinking a special formula, and sometimes medication, as recommended by a genetic metabolic medical professional.

**DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.**

### **For more information:**

#### **Genetics Home Reference**

Website: <http://www.ghr.nlm.nih.gov>

**Save Babies Through Screening Foundation**

4 Manor View Circle  
Malvern, PA 19355-1622  
Toll Free Phone: 1-888-454-3383  
Fax: (610) 993-0545  
Email: [email@savebabies.org](mailto:email@savebabies.org)  
Website: <http://www.savebabies.org>

**ASA Kids – A Support Group for Kids With Argininosuccinic Aciduria**

Phone: (409) 735-4332  
Website: <http://www.asakids.org>

**National Urea Cycle Disorders Foundation**

4841 Hill Street  
La Canada, CA 91011  
Email: [info@nucdf.org](mailto:info@nucdf.org)  
Website: <http://www.nucdf.org>

**American College of Medical Genetics**

Newborn Screening ACT Sheets and Confirmatory Algorithms  
<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>

**Cardinal Glennon Children’s Hospital**

St. Louis, Missouri 314-577-5639  
Website: <http://pediatrics.slu.edu/index.phtml?page=geneticsdiv>

**Children’s Hospital at University Hospital and Clinics**

Columbia, Missouri 573-882-6991  
Website: <http://www.genetics.missouri.edu/>

**Children’s Mercy Hospital**

Kansas City, Missouri 816-234-3290  
Website: <http://www.childrens-mercy.org/content/view.aspx?id=155>

**St. Louis Children’s Hospital**

St. Louis, Missouri 314-454-6093  
Website: <http://www.peds.wustl.edu/genetics/>