



## **ARG 1 Deficiency** **(Argininemia/Arginase Deficiency)**

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### **What is it?**

Arginase deficiency is one type of amino acid disorder. People with this condition have problems removing ammonia from the body. Ammonia is a harmful substance. It is made when protein and its building blocks (amino acids) are broken down for use by the body.

### **What causes it?**

This is one of a small number of conditions called urea cycle disorders (UCD).

It occurs when an enzyme called arginase is either missing or not working properly. This enzyme's job is to help break down the amino acid arginine and to help remove ammonia from the body.

When arginase is not working, arginine, along with ammonia, can build up in the blood. This can have serious effects on a child's growth, learning and health.

### **If arginase deficiency is not treated, what problems occur?**

The effects of this condition vary from person to person. Symptoms can start right at birth or not until later in childhood. Many children have their first symptoms around age 1. Effects in infants can include:

- 1) Poor growth.
- 2) Learning delays.
- 3) Poor coordination and balance.
- 4) Fussiness or illness after eating high protein food.

Episodes of illness caused by high ammonia levels in the blood can sometimes occur in

young infants but are not common. Some of the first symptoms of high ammonia are:

- 1) Poor appetite.
- 2) Excess sleepiness or lack of energy.
- 3) Irritability.
- 4) Vomiting.

If untreated, other symptoms can follow:

- 1) Muscle weakness
- 2) Decreased or increased muscle tone
- 3) Breathing problems
- 4) Problems staying warm
- 5) Seizures
- 6) Swelling of the brain
- 7) Coma, and sometimes death

Sometimes, symptoms of arginase deficiency do not begin until later in infancy or childhood. Common effects in older infants and children can include:

- 1) Poor growth.
- 2) Small head size.
- 3) Hyperactivity.
- 4) Behavior problems.
- 5) Learning disabilities.
- 6) Avoidance of meat or other high-protein foods.
- 7) Vomiting and excessive sleepiness.

Episodes of high ammonia, described above, happen rarely. If they occur, they are more likely to happen:

- 1) After going without food for a long period of time.
- 2) During an illness or infection.
- 3) After high-protein meals.

## **What is the treatment for arginase deficiency?**

Your baby's primary doctor will work with a metabolic doctor and a dietician to care for your child.

Prompt treatment is needed to prevent the build up of arginine and ammonia. You should start treatment as soon as you know your child has the condition.

The following are treatments often recommended for babies and children with arginase deficiency.

### ***Low-Protein Diet and/or Special Medical Foods and Formula***

Most children need to eat a diet made up of very low protein foods and special medical foods. Your dietician will create a food plan that contains the right amount of protein, nutrients and energy to keep your child healthy. The food plan should be continued throughout your child's life.

#### ***Low-Protein Diet***

One of the main treatments is a low-protein diet. Foods to avoid or strictly limit include:

- 1) Milk, cheese and other dairy products.
- 2) Meat and poultry.
- 3) Fish.
- 4) Eggs.
- 5) Dried beans and legumes.
- 6) Nuts and peanut butter.

Eating foods high in protein can cause ammonia and arginine to build up, resulting in the symptoms described above. Many vegetables and fruits have only small amounts of protein and can be eaten in carefully measured amounts.

Do not remove all protein from the diet. Your child still needs a certain amount of protein for normal growth and development. Any changes in the diet should be made under the guidance of a dietician.

### ***Medical Foods and Formula***

There are medical foods such as special low-protein flours, pastas and rice that are made especially for people with amino acid disorders.

Your child may be given a special formula that contains the correct amount of amino acids and nutrients. Your metabolic doctor and dietician will decide whether your child should use this formula and how much to use.

Your child's exact food plan will depend on many things, such as his or her age, weight and general health. Your dietician will fine-tune your child's diet over time.

### ***Medication***

Other medications can help the body get rid of excess arginine and ammonia. Your metabolic doctor will decide which medications your child should take.

### ***Blood Tests***

Your child will need to have regular blood tests to measure ammonia and amino acid levels. Your child's diet and medication may need to be adjusted based on blood test results.

### ***Call Your Doctor at the Start of Any Illness***

Illness or infection can sometimes lead to high arginine and ammonia levels. In order to prevent problems, call your doctor right away when your child has any of the following:

- 1) Loss of appetite
- 2) Low energy or extreme sleepiness
- 3) Vomiting
- 4) Fever
- 5) Infection or illness
- 6) Behavior or personality changes
- 7) Difficulty walking or balance problems

Ask your metabolic doctor if you should carry a special travel letter with medical instructions for your child's care.

### **What happens when arginase deficiency is treated?**

With prompt and lifelong treatment, children with arginase deficiency may be able to live healthy lives with typical growth and learning.

Even with treatment, some children still have effects from high blood levels of arginine and ammonia. This can result in permanent learning problems or mental retardation.

### **What causes the arginase enzyme to be absent or not working correctly?**

Genes tell the body to make enzymes. People with arginase deficiency have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the arginase enzyme does not work properly or is not made at all.

### **Is arginase deficiency inherited?**

This inherited condition affects both boys and girls equally.

Everyone has a pair of genes that makes the arginase enzyme. In children with arginase deficiency, neither of these genes works correctly. These children inherit one nonworking gene for the condition from each parent.

Parents of children with arginase deficiency rarely have the condition themselves. Instead, each parent has a single nonworking gene for arginase deficiency. They are called carriers. Carriers do not have the condition because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25 percent chance in each pregnancy for the child to have arginase deficiency. There is a 50 percent chance for the child to be a carrier, just like the parents. And, there is a 25 percent chance for the child to have two working genes.

### **Can other members of the family have arginase deficiency or be carriers?**

#### ***Having Arginase Deficiency***

If they are healthy and growing normally, older brothers and sisters of a baby with arginase deficiency are at low risk of having the condition. However, finding out whether other children in the family have the condition may be important because early treatment can prevent serious health problems. Ask your metabolic doctor whether your other children should be tested.

#### ***Arginase Deficiency Carriers***

Brothers and sisters who do not have arginase deficiency still have a chance to be carriers like their parents. Except in special cases, carrier testing should be done only in people older than 18.

Each of the parents' brothers and sisters has a 50 percent chance to be a carrier. It is important for other family members to be told that they could be carriers. There is a small chance they are also at risk to have children with arginase deficiency.

When both parents are carriers, newborn screening results are not sufficient to rule out the condition in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

### **Can other family members be tested?**

#### ***Diagnostic Testing***

Brothers and sisters of a child with arginase deficiency can be tested using blood, urine or skin samples.

#### ***Carrier Testing***

Carrier testing may be available. Your metabolic doctor or genetic counselor can answer your questions about carrier testing.

### **How may people have arginase deficiency?**

About one in every 300,000 babies in the United States is born with this condition.

### **Does arginase deficiency happen more frequently in a certain ethnic group?**

No, it does not happen more often in any specific race, ethnic group, geographical area or country.

### **Does arginase deficiency go by any other names?**

Arginase deficiency is sometimes also called hyperargininemia.

### **Where can I find more information?**

National Urea Cycle Disorders Foundation  
[www.nucdf.org](http://www.nucdf.org)

Children Living with Inherited Metabolic Diseases (CLIMB)  
[www.climb.org.uk](http://www.climb.org.uk)

National Coalition for PKU and Allied Disorders  
[www.pku.allieddisorders.org](http://www.pku.allieddisorders.org)

Genetic Alliance  
[www.geneticalliance.org](http://www.geneticalliance.org)

### **Children's Special Health Services (CSHS)**

State Capitol Judicial Wing  
600 E. Boulevard Ave., Department 301  
Bismarck, ND 58505-0269  
Toll Free: 800.755.2714  
701.328.2436  
Relay TDD: 701.328.3975  
CSHS website: [www.ndhealth.gov/CSHS](http://www.ndhealth.gov/CSHS)  
North Dakota Department of Health website:  
[www.ndhealth.gov](http://www.ndhealth.gov)

Family support resources available from CSHS:

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

### ***Family Resources***

Family to Family Network  
Center for Rural Health  
University of North Dakota  
School of Medicine and Health Sciences  
P.O. Box 9037  
Grand Forks, ND 58202-9037  
Toll Free: 888.434.7436  
701.777.2359  
Fax: 701.777.2353  
E-mail: [NDF2F@medicine.nodak.edu](mailto:NDF2F@medicine.nodak.edu)  
[www.medicine.nodak.edu/crh](http://www.medicine.nodak.edu/crh)

Pathfinder Services of ND  
Pathfinder Family Center  
1600 2<sup>nd</sup> Ave. SW, Ste. 19  
Minot, ND 58701  
Toll Free: 800.245.5840  
701.837.7500  
Relay TDD: 701.837.7501  
E-mail: [ndpath01@ndak.net](mailto:ndpath01@ndak.net)  
[www.pathfinder.minot.com](http://www.pathfinder.minot.com)

Family Voices of North Dakota, Inc.  
P.O. Box 163  
Edgeley, ND 58433  
Toll Free: 888.522.9654  
701.493.2634  
Fax: 701.493.2635  
[www.geocities.com/ndfv](http://www.geocities.com/ndfv)

\*\*This fact sheet has general information. Every child is different and some of these facts may not apply to your child specifically. Certain treatments may be recommended for some children but not others. All children should be followed by a metabolic doctor in addition to their primary-care provider.

### **Acknowledgement**

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**Disclaimer**

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[www.ndhealth.gov/familyhealth](http://www.ndhealth.gov/familyhealth)