



Newborn Screening FACT Sheet

Argininemia (ARG)

What is Argininemia?

Argininemia (ARG) is a condition that causes harmful amounts of arginine and ammonia to build up in the body. It is considered an amino acid condition because people affected with ARG are unable to break down an amino acid, a small molecule that makes up proteins, known as arginine. You may also hear ARG called a urea cycle condition. This name is used to describe conditions that cause ammonia to accumulate in the body. If untreated, argininemia can cause muscle problems and developmental delay. However, if the condition is detected early and proper treatment is initiated, individuals with argininemia can often lead healthy lives.

What Causes ARG?

When we eat food, enzymes help break it down. Some enzymes help break down proteins into their building blocks, called amino acids. Other enzymes break down these amino acids. In ARG, the enzyme arginase is not working correctly.

Arginase's job is to help break down the amino acid arginine and remove ammonia from the body through the urine. If your baby has ARG, then his or her body either does not make enough, or makes non-working arginase enzymes.

When arginase is not working correctly, the body cannot break down arginine correctly or get rid of ammonia in the blood. This causes high levels of ammonia and arginine in the blood, which can be dangerous. Everyone has some ammonia and arginine in his or her blood, but too much can be toxic.

ARG is an autosomal recessive genetic condition. This means a child must inherit two copies of the non-working gene for ARG, one from each parent, in order to have the condition. The parents of a child with an autosomal recessive condition each carry one copy of the non-working gene, but they typically do not show signs and symptoms of the condition. While having a child with ARG is rare, when both parents are carriers, they can have more than one child with the condition.

What Symptoms or Problems Occur with ARG?

[Symptoms are something out of the ordinary that a parent notices.]

Signs of ARG can begin any time from infancy to childhood. Usually, signs begin to show at around one to three years of age and include:

- delayed growth
- developmental delays
- balancing trouble
- tight, rigid muscles (called spasticity)
- irritability
- poor appetite
- sleeping longer or more often
- vomiting
- weak muscle tone (called hypotonia)
- trouble regulating body temperature (your baby might get cold easily)
- small head size
- hyperactivity

Many of these signs may occur when your baby eats foods that his or her body cannot break down. They can be triggered by long periods of time without eating, illness, and infections.

If your baby shows any of these signs, be sure to contact your baby's health care provider immediately.

What is the Treatment for ARG?

1. Your baby may need to be on a restricted diet in order to avoid the proteins that his or her body cannot break down. A dietician or nutritionist can help plan a healthy diet for your child. Your baby's doctor may recommend special foods or formulas for children with ARG. These formulas will likely need to continue through adulthood.

2. Medication – Your baby's health care provider may prescribe medications to help your baby's body get rid of excess arginine and ammonia.

3. Blood tests – Regular blood tests will check your child's amino acid and ammonia levels.

Things to Remember

Call your doctor right away if your child has any of the signs mentioned above. Children with high ammonia should be treated in the hospital.