

Positive Result:

Blood Spot Screen Result Notification



Elevated Arginine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of an amino acid called arginine.

What does this mean?

High levels of arginine can indicate that your child has argininemia. A positive result does not mean your baby has argininemia, but more testing is needed to know for sure.

What happens next?

Your baby's doctor or a metabolic specialist will help arrange for more testing. Your baby will also be seen by a metabolic specialist.

What is argininemia?

Argininemia is part of a group of disorders called amino acid disorders. Children with argininemia are unable to process ammonia, a waste product that is created when the body breaks down amino acids from protein. This causes dangerous amounts of ammonia to build up in the body.

What health problems can it cause?

Argininemia is a lifelong condition. A child with argininemia usually has no symptoms in the newborn period. If left untreated, it can cause:

- Lack of energy
- Poor feeding
- Poor growth
- High levels of ammonia in the blood (hyperammonemia)
- Tight, rigid muscles (spasticity)
- Intellectual disabilities
- Seizures

Children with argininemia can benefit from prompt and careful treatment.

What treatment options are available?

Treatment for argininemia consists of a special diet that avoids protein. Certain medications and supplements may be prescribed. Early treatment can be beneficial. Even with treatment, some children still have episodes of high ammonia. This can result in long-term intellectual disabilities and spasticity.

Children with argininemia should see their regular doctor and a doctor who specializes in metabolic disorders.

Resources

Genetics Home Reference:
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org