

Positive Result:

Blood Spot Screen Result Notification



Elevated Citrulline

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 citrulline (CIT).

What does this mean?

High levels of CIT can indicate that your child has a metabolic disorder. If your baby has a metabolic disorder, more testing is needed to find out which metabolic disorder it is. The most common metabolic disorders with high levels of CIT are citrullinemia type I (CIT-I) and argininosuccinic acidemia (ASA). A positive result does not mean your baby has CIT-I, ASA, or a different metabolic disorder, 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 citrullinemia type I (CIT-I) and argininosuccinic acidemia (ASA)?

CIT-I and ASA are part of a group of disorders called amino acid disorders. Children with these disorders 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?

Possible health problems of CIT-I and ASA include:

- Lack of energy
- Poor feeding
- Poor growth
- High levels of ammonia in the blood (hyperammonemia)
- Intellectual disabilities
- Seizures
- Coma, sometimes leading to death

Children with these disorders can benefit from prompt and careful treatment.

What treatment options are available?

Treatment for both CIT-I and ASA consists of a special diet that avoids protein. Certain medications and supplements may be prescribed. Early treatment can be life-saving. Even with treatment, some children still have episodes of high ammonia. This can result in brain damage causing lifelong learning problems and intellectual disabilities.

Children with these disorders 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