

Positive Result:

Blood Spot Screen Result Notification



Absent/Reduced Citrulline

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has low levels of an amino acid called citrulline (CIT).

What does this mean?

Low 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 low levels of CIT are called proximal urea cycle disorders. A positive result does not mean your baby has a proximal urea cycle 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 are proximal urea cycle disorders?

Proximal urea cycle disorders are a group of disorders caused by the inability to change ammonia to urea. Ammonia is a waste product that is created when the body breaks down amino acids from protein. Urea is also a waste product from this break down and leaves the body in the urine. Because ammonia doesn't get changed to urea, dangerous amounts of ammonia build up in the body and causes health problems.

What health problems can it cause?

Possible health problems include:

- Lack of energy
- Poor feeding and 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 consists of a special diet that avoids protein. Certain medications and supplements may be prescribed. Sometimes dialysis (hospital procedure that removes toxins like ammonia from the blood) is needed. In severe cases, a liver transplant will be considered.

Early treatment can be life-saving. Even with treatment, some children still have high ammonia at times. 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