

Positive Result:

Blood Spot Screen Result Notification



Elevated Succinylacetone

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of succinylacetone (SUAC). SUAC is a by-product from poor protein breakdown.

What does this mean?

High levels of SUAC can indicate that your baby has tyrosinemia type 1. A positive result does not mean your baby has tyrosinemia type 1, 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 tyrosinemia type 1?

Tyrosinemia type 1 is part of a group of disorders called amino acid disorders. Tyrosine is an amino acid. Amino acids are the building blocks to make protein. With tyrosinemia type 1, the body is unable to break down protein from the food we eat. This causes tyrosine to build up in the blood (tyrosinemia). There are three types of tyrosinemia. However, elevated succinylacetone is mostly seen in tyrosinemia type 1.

What health problems can it cause?

Tyrosinemia type 1 is a lifelong condition. If untreated, it can cause:

- Diarrhea
- Vomiting
- Poor weight gain
- Nosebleeds
- Lack of energy
- Kidney and liver problems
- Trouble breathing
- Seizures
- Coma, sometimes leading to death

Children with tyrosinemia type 1 can benefit from prompt and careful treatment.

What treatment options are available?

Although tyrosinemia type 1 cannot be cured, it can be treated. Children with tyrosinemia type 1 are treated with a low protein diet. Certain medications may be prescribed to help lower the tyrosine levels in the blood. If treated before symptoms develop, children can have healthy growth and development.

Children with tyrosinemia type 1 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