

Positive Result:

Blood Spot Screen Result Notification



Elevated Tyrosine with Normal Succinylacetone

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 tyrosine but normal levels of a by-product of poor tyrosine breakdown called succinylacetone.

What does this mean?

High levels of tyrosine with normal levels of succinylacetone is often seen in newborns, especially in those that are born before their due date. However, it can also indicate that your child has a type of tyrosinemia. A positive result does not mean your baby has tyrosinemia, but more testing is needed to know for sure.

What happens next?

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

What is tyrosinemia?

Tyrosinemia is part of a group of disorders called amino acid disorders. With tyrosinemia, the body is unable to break down protein from the food we eat containing tyrosine. This causes tyrosine to build up in the blood (tyrosinemia). There are three types of tyrosinemia. However, tyrosinemia types 2 and 3 are most likely to have normal succinylacetone levels.

What health problems can it cause?

Tyrosinemia types 2 and 3 are lifelong conditions.

If untreated, tyrosinemia type 2 can cause:

- Eye sensitivity to light (photophobia)
- Eye redness
- Skin problems
- Poor coordination

Tyrosinemia type 3 is very rare and the symptoms are not well known. Symptoms may include poor coordination and seizures.

Children with tyrosinemia types 2 or 3 can benefit from prompt and careful treatment.

What treatment options are available?

Although tyrosinemia types 2 and 3 cannot be cured, it can be treated. Children with either type 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 types 2 or 3 should see their regular doctor and a doctor who specializes in tyrosinemia.

Resources

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

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org