

# Positive Result:

## Blood Spot Screen Result Notification



### Elevated Methionine

#### 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 methionine.

#### What does this mean?

High levels of methionine can indicate that your child has liver problems, received a special type of feeding called TPN, or has a metabolic disorder.

More testing is needed to find out what caused the high methionine level. If your baby has a metabolic disorder, the most common metabolic disorder with high levels of methionine is homocystinuria. A positive result does not mean that your baby has homocystinuria, 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 homocystinuria?

Homocystinuria is part of a group of disorders called amino acid disorders. With homocystinuria, the body is unable to break down protein from the food we eat. This causes methionine to build up in the blood that can lead to health problems.

#### What health problems can it cause?

Homocystinuria is a lifelong condition. If untreated, it can cause:

- Eye problems
- Blood clots
- Bone abnormalities
- Intellectual disabilities
- Behavioral difficulties

Children with homocystinuria can benefit from prompt and careful treatment.

#### What treatment options are available?

Although homocystinuria cannot be cured, it can be treated. Treatment includes eating a low protein diet. Certain medications may be prescribed. If treated before symptoms develop, many children can have healthy growth and development.

Children with homocystinuria should see their regular doctor, an eye specialist, and a doctor who specializes in homocystinuria.

### Resources

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

**Save Babies Through Screening Foundation:**  
[www.savebabies.org](http://www.savebabies.org)

**Baby's First Test:**  
[www.babysfirsttest.org](http://www.babysfirsttest.org)