



Homocystinuria/ Hypermethioninemia

What is newborn screening?

Newborn screening is a blood test to check for conditions that might be hidden at birth. To do the screening, a nurse takes a few drops of blood from your baby's heel soon after birth. This blood sample is required for all newborn babies.



Newborn screening is not the same as diagnostic testing. A diagnostic test can tell with more certainty whether or not a child has a condition. On the other hand, a screening test simply indicates that a child *may*

have a condition. The purpose of a screening test is to find all babies that should have diagnostic testing. When a child with an out-of-range newborn screening result has a follow-up test result within the normal range, it is sometimes called a "false positive."

What if my baby needs more testing?

If you are told that your baby needs follow-up testing, it does not mean that your baby has the health condition. An out of range result may occur because:

- The sample was too small
- The sample was collected too early
- The sample was collected too close to a feeding
- The baby was born too early or had a low birth weight

Most babies who have follow up testing for homocystinuria or hypermethioninemia are healthy, and will not have either of these conditions. However, out of range screening results CAN indicate a disorder, so it is important to follow your doctor's advice & get your baby tested quickly so that final results can be confirmed.

KEY POINTS:

- **You have just heard that your baby may have homocystinuria or hypermethioninemia. Please understand that the newborn screening is just that---a screening test. Further testing is required to confirm or rule out the diagnosis.**
- **Most babies who have out of range newborn screens are healthy, and will not have either of these conditions.**
- **If treated early, children with HCY/MET can have healthy growth and development.**

Connecticut Department of Public Health

Connecticut Newborn Screening Program • 860.920.6628

Connecticut Newborn Diagnosis and Treatment Network • 860.837.7870

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What is homocystinuria/hypermethioninemia?

Homocystinuria (HCY) is a treatable disorder affecting the way the body uses protein. Our bodies change proteins into smaller pieces called amino acids. Children with HCY cannot use the amino acid methionine. If HCY is not treated, homocysteine can build up in the blood and cause medical problems or disabilities.

Hypermethioninemia (MET) is a condition that is caused by having too much methionine in the body. Many people with MET do not show any signs of the condition. However, if MET is not treated, it can cause health problems with health or learning.



What does this mean?

Although these conditions cannot be cured, they can be treated. If further testing finds that your baby has HCY or MET, he or she may need to be on a special diet. Sometimes, a doctor will recommend vitamin supplements. This will help to prevent health problems. If treated early, children with HCY or MET can have healthy growth and development.

What happens next?

Your baby's doctor may ask for the newborn screen to be repeated or, for your baby to have more testing. This follow up testing is important to know if treatment is needed. In some cases, you may be asked to visit a specialist and/or a dietician. The specialist and/or dietician will talk to you about the best plan for your baby.

What are the signs and symptoms of HCY and MET?

Babies with HCY/MET usually look healthy at birth. Babies may have trouble gaining weight. If you become concerned about your baby's growth, feeding or activity, please talk to your child's pediatrician.

What if I still have questions?

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns. The Connecticut Newborn Diagnosis and Treatment Network (the Network) is available to put you in touch with the best resource. To reach the Network, you can call 860-837-7870, Monday-Friday, 8:30am-4:30pm. We also recommend the website www.babysfirsttest.org as an accurate and informative resource.

This fact sheet was written for information purposes only. It should not replace medical advice, diagnosis or treatment.

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