

When Baby Needs a Second Test for Elevated MET (Hypermethioninemia or Homocystinuria)

A small sample of your baby's blood was collected soon after birth and sent to the DHEC laboratory for testing. This testing is called Newborn Screening. In SC, newborns are tested for several genetic and biochemical disorders. Sometimes, a second test is needed to help your doctor decide if your baby has one of these disorders. In many cases, the second test will be normal. However, if your baby does have a newborn screening disorder, early treatment will give him or her the best chance to grow up healthy.

Because a compound called Methionine was elevated in your baby's first test, he or she could possibly have a disorder called Hypermethioninemia or Homocystinuria.

What is Hypermethioninemia (MET)?

Hypermethioninemia (MET) is a condition that occurs when there is a high amount of methionine in the body. It is considered an amino acid condition because people with MET are unable to break down an amino acid, a building block of proteins, known as Methionine. Many people with MET do not show signs of the condition.

What is Homocystinuria?

Homocystinuria is a genetic disorder that is found in a few babies born each year. Homocystinuria can be identified when compounds called amino acids are measured in a baby's blood. Amino acids are the "building blocks" of protein. When a baby has homocystinuria, he or she cannot use one of the amino acids that are found in foods including breast milk and infant formula.

This amino acid, MET, builds up in the baby's blood and can cause the baby to have problems with the eyes and bones. Dangerous blood clots can form. Homocystinuria can also cause severe intellectual disability.

How will I know if my baby really has Homocystinuria?

If your baby's newborn screening result showed very high MET levels, he or she may have homocystinuria. The newborn screening test will be repeated, and additional tests will be done to help the doctors figure out if your baby has homocystinuria. Usually, the results of these tests take a few days to come back. You will also be referred to a doctor who specializes in these kinds of disorders called a metabolic specialist.

What do I need to do until I know the final results?

Your baby may not have any symptoms at first, but you will need to follow your doctor's instructions very carefully. If your baby seems to be getting sick, call your doctor right away.

How is Homocystinuria treated?

Homocystinuria is usually treated with a special diet. At first, babies must be fed a special metabolic formula. When they begin to eat solid foods, the protein in their diet will need to be controlled. They will still need to drink metabolic formula to make sure they get everything they need to grow properly. A dietitian will help the family learn which foods the baby can safely eat.

A few babies with homocystinuria only need to take extra amounts of Vitamin B6. The doctors will run additional tests to see if that is all that is needed for your baby.

What else should I do to keep my baby as healthy as possible?

Don't forget to keep all of your well baby check-ups! Seeing the doctors regularly and following your baby's diet plan carefully are the best things you can do to help your baby grow and develop.

Internet Resources: <http://www.babysfirsttest.org/>