Methylmalonic Acidemia and Propionic Acidemia

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 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."

KEY POINTS:

- **You have just heard that your baby may have MMA or PPA. 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 be diagnosed.**
- **If treated early, children with organic acid disorders can have healthy growth and development.**

What if my baby needs more testing?

If you are told that your baby needs follow-up testing, it does not necessarily mean that your baby is at risk. 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

A positive newborn screen for MMA or PPA can, in some cases, be indicative of a vitamin B12 deficiency. Vitamin B12 deficiency may be due to maternal diet or abnormal absorption. In some cases, supplementation with vitamin B12 may be beneficial. Most babies who have follow up testing for MMA and PPA are healthy, and will not have either condition. 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.

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What are MMA and PPA?

Methylmalonic academia (MMA) and propionic academia (PROP) are inherited conditions in which the body is unable to break down certain fats and proteins. They are considered organic acid conditions because they can lead to a harmful amount of organic acids in the body. If left untreated, these conditions can result in serious health problems.

What does this mean?

Although these conditions cannot be cured, they can be treated. If further testing finds that your baby has MMA or PPA, he or she may need to be on a special diet. Certain medications and supplements may be prescribed. Early treatment can help to minimize or prevent health problems. If treated early, children with MMA or PPA can have healthy growth or 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. You will want to have these follow up tests done as soon as possible. In some cases, you may be asked to visit a specialist and/or dietician. The specialist and/or dietician will talk to you about the best plan for your baby.

What are the signs and symptoms of MMA and PPA?

Some babies with MMA or PPA never have serious signs or symptoms. Sometimes signs and symptoms may not develop until later in life. Signs and symptoms could include: excessive sleepiness, not eating well, not gaining weight, or frequent vomiting. If you become concerned about your baby's growth, feeding or activity, please talk to your 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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