

Positive Result:

Blood Spot Screen Result Notification



Elevated C3 Acylcarnitine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C3 acylcarnitine.

What does this mean?

High levels of C3 acylcarnitine can indicate that your child has a metabolic disorder. If your baby has a metabolic disorder, more testing is needed to find out which metabolic disorder it is. The most common metabolic disorders with high levels of C3 acylcarnitine are propionic acidemia (PA) and methylmalonic acidemia (MMA). A positive result does not mean your baby has PA, MMA, or a different metabolic disorder, 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 propionic acidemia (PA) and methylmalonic acidemia (MMA)?

PA and MMA are part of a group of disorders called organic acid disorders. With PA and MMA, the body is unable to break down certain fats and proteins. Using stored fat for energy is especially important between meals when the body is not getting new energy from food. During periods without food (fasting) or illness, health problems can begin.

What health problems can it cause?

Possible health problems of PA and MMA include:

- Poor feeding
- Sleepiness
- Vomiting
- Muscle weakness (hypotonia)
- Coma

Children with these disorders can benefit from prompt and careful treatment.

What treatment options are available?

Treatment for both PA and MMA consists of a special diet and avoidance of fasting. Certain medications and supplements may be prescribed. Early treatment can be life-saving. Even with treatment, however, some learning disabilities or developmental delays can still occur.

Children with PA or MMA should see their regular doctor and a doctor who specializes in metabolic disorders.

Resources

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

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org