

Positive Result:

Blood Spot Screen Result Notification



Elevated Phenylalanine

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 phenylalanine (PHE).

What does this mean?

High levels of PHE indicate that your baby has too much PHE in their blood (hyperphenylalaninemia). There are a number of possible causes for this, but more testing is needed to find what is causing it.

What happens next?

Your baby's doctor or a metabolic specialist familiar with hyperphenylalaninemia will help arrange for more testing. Your baby will also be seen by a metabolic specialist.

What is hyperphenylalaninemia?

Hyperphenylalaninemia happens when there is too much PHE in the blood. These high levels can go away over time (transient), or they can be caused by a disorder called phenylalanine hydroxylase (PAH) deficiency. There are two forms of this disorder: classic PKU and hyperphenylalaninemia (H-PHE).

PAH deficiency is a disorder where the body is unable to break down proteins from the food we eat. This causes PHE to build up in the blood. Too much PHE can cause health problems.

What health problems can it cause?

The most severe form is called classic PKU, and it is a lifelong condition. If untreated, it can cause:

- Irritability
- Dry, scaly skin (eczema)
- Growth problems
- Seizures
- Intellectual disabilities

The less severe form is called H-PHE. Most people with H-PHE have little to no health problems.

What treatment options are available?

Children with classic PKU are treated with a low protein diet. Certain medications may also be prescribed to help lower the PHE levels in the blood. If treated before symptoms develop, children can have healthy growth and development.

Treatment is not always needed for H-PHE. If treatment is recommended, it would be a low protein diet.

Children with classic PKU or H-PHE should see their regular doctor and a metabolic specialist.

Resources

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

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org