



Newborn Screening FACT Sheet

Benign Hyperphenylalaninemia (H-PHE)

What is Benign Hyperphenylalaninemia?

Benign Hyperphenylalaninemia is a mild form of phenylketonuria. It is an amino acid condition where affected people have trouble breaking down phenylalanine. Most people with this condition experience mild or no symptoms. Children with Benign Hyperphenylalaninemia have more phenylalanine in their bodies than is typical, but less than children with classic phenylketonuria. Measuring the amount of phenylalanine in your baby's body can help determine if your baby has this condition.

What causes Benign Hyperphenylalaninemia?

H-Phe is caused by an enzyme that does not work typically. It is a form of phenylketonuria, also known as PKU. When we eat, enzymes help break down food. Some enzymes break protein into its building blocks, called amino acids. Other enzymes break down these amino acids. In Benign Hyperphenylalaninemia, the enzyme phenylalanine hydroxylase (PAH) is not working correctly.

Babies with Benign Hyperphenylalaninemia have less PAH. They can break down phenylalanine, but not as quickly as normal. If the body cannot break down phenylalanine quickly, it can build up in the blood. It can be harmful if too much phenylalanine builds up but babies with Benign Hyperphenylalaninemia do not have harmful levels.

What are signs and symptoms of Benign Hyperphenylalaninemia?

Because Benign Hyperphenylalaninemia is a mild form of phenylketonuria (PKU), affected people typically do not show any signs. Babies with Benign Hyperphenylalaninemia typically have no complications. They can have healthy growth and development. However, some babies with Benign Hyperphenylalaninemia do have a small risk of

developmental delay without treatment. This is why newborn screening is important.

What is the treatment for Benign Hyperphenylalaninemia?

Babies with Benign Hyperphenylalaninemia usually do not require treatment. Some individuals may need to limit phenylalanine in their diet and drink a special medical formula with no phenylalanine as infants. Phenylalanine is in many proteins and artificial sweeteners. Your baby's doctor can help determine how much phenylalanine is safe.

Things to Remember

Your child's phenylalanine blood levels should be checked periodically to keep Phe levels within the target range.

A woman with Benign Hyperphenylalaninemia should visit a PKU center when planning or during pregnancy to prevent birth defects because elevated levels during pregnancy can harm the unborn baby. Overall, people with this condition can live healthy lives.

Resources:

Medline Plus

medlineplus.gov/genetics/condition/phenylketonuria

National PKU Alliance npkua.org/

Health Resources & Services Administration

newbornscreening.hrsa.gov/conditions/non-pku-hyperphenylalaninemia

Navigate Life Texas navigatelifetexas.org/en/diagnosis-healthcare/children-with-multiple-disabilities-rare-conditions-undiagnosed