

Benign Hyperphenylalaninemia (H-PHE)

What is Benign Hyperphenylalaninemia (H-PHE)?

Benign hyperphenylalaninemia (H-PHE) is a mild form of phenylketonuria. It is considered an amino acid condition because people with H-PHE have problems breaking down an amino acid, a building block of proteins, known as phenylalanine. Most people with this condition experience mild symptoms or no symptoms. Children with H-PHE have more phenylalanine in their bodies than is typical, but they have a lower amount of phenylalanine in their bodies than do children with the condition known as classic phenylketonuria. Measuring the amount of phenylalanine in your baby's body can help doctors determine if your baby has this condition.

What Causes H-PHE?

When we eat food, enzymes help break it down. Some enzymes break down protein into its building blocks, called amino acids. Other enzymes break down these amino acids. In H-PHE, the enzyme phenylalanine hydroxylase (PAH) is not working correctly.

PAH's job is to break down the amino acid phenylalanine. Babies with H-PHE make less PAH than babies without H-PHE. They can break down phenylalanine, but not as quickly as babies without H-PHE. If the body cannot break down phenylalanine quickly enough, phenylalanine can build up in the blood. Everyone has some phenylalanine in his or her blood, but high levels could be harmful. Babies with H-PHE have elevated levels of phenylalanine, but these levels are usually not dangerous.

H-PHE is an autosomal recessive genetic condition. This means that a child must inherit two copies of the non-working gene for H-PHE, one from each parent, in order to have the condition. The parents of a child with an autosomal recessive condition each carry one copy of the non-working gene, but they typically do not show signs and symptoms of the condition. While having a child with H-PHE is rare, when both parents are carriers, they can have more than one child with the condition.

What Symptoms or Problems Occur with H-PHE?

[Symptoms are something out of the ordinary that a parent notices.]

Benign H-PHE is a form of phenylketonuria (PKU). Different forms of PKU have varying severity of signs. Because H-PHE is a less severe type of PKU, babies with H-PHE typically do not show any signs. Babies with H-PHE typically have no complications. They can have healthy growth and development. However, some babies with H-PHE do have a small risk of brain damage without treatment. This is why newborn screening for H-PHE is important.

What is the Treatment for H-PHE?

Dietary Treatment: Babies with H-PHE usually do not require any treatment.

Some individuals with more severe forms of H-PHE may need to limit the amount of phenylalanine in their diet and drink a special medical formula that contains no phenylalanine. Phenylalanine is a substance found in many proteins and also in artificial sweeteners. Babies with H-PHE cannot break down phenylalanine as quickly as babies without H-PHE. Your baby's doctor can help determine how much phenylalanine is safe for your baby.

Things to Remember

Your child's phenylalanine (Phe) blood levels and diet should be checked periodically to make sure the Phe is kept at certain levels.

The lifelong treatment goal is to maintain blood Phe levels in the range of 120-360 $\mu\text{mol/l}$ (2-6 mg/dl) in patients of all ages for life.

A woman with H-PHE should be checked at a PKU center when planning a pregnancy and during pregnancy to prevent birth defects.