



Newborn Screening FACT Sheet

Tetrahydrobiopterin Deficiency (BIOPT-BS)

What is BIOPT-BS?

Biopterin defect in cofactor biosynthesis (BIOPT-BS) is a condition that causes high amounts of phenylalanine and other harmful substances to build up in the body. It is considered an amino acid condition because people with BIOPT-BS are unable to process certain amino acids, small molecules that make up proteins. Detecting the condition early and beginning treatment can often prevent the severe outcomes of BIOPT-BS.

BIOPT-BS is only one form of tetrahydrobiopterin deficiency.

What Causes BIOPT-BS?

When we eat food, enzymes help break it down. Some enzymes break down proteins into their building blocks, called amino acids. Other enzymes break down these amino acids. Enzymes sometimes need help from substances called co-enzymes. Co-enzymes attach to enzymes to help them do their jobs. Biopterin is an important co-enzyme for breaking down amino acids, especially the amino acid phenylalanine.

BIOPT-BS occurs when the body cannot make its own BH₄, the substance from which biopterin is made. Babies with BIOPT-BS either do not produce enough or produce non-working enzymes for making BH₄. BIOPT-BS affects either the GTP cyclohydrolase I enzyme or the 6-pyruvoyl tetrahydropterin synthase enzyme. If either of these enzymes does not work, then the body cannot make BH₄. Without BH₄ the body cannot make biopterin. When the body does not have biopterin, it cannot break down many amino acids, including phenylalanine. This causes phenylalanine levels to rise in the blood. Everyone has some phenylalanine in his or her blood, but high levels can be toxic.

What Symptoms or Problems Occur with BIOPT-BS?

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

Babies with BIOPT-BS usually seem healthy at birth.

Signs of BIOPT-BS may develop over time. These signs include:

- developmental delays
- seizures
- behavior troubles
- trouble regulating body temperature (your baby may get cold easily)
- poor sucking during feeding
- weak muscle tone (known as hypotonia)
- sleeping longer or more often

Many of these signs may occur when your baby eats foods that his or her body cannot break down. They can be triggered by long periods of time without eating, illnesses, and infections.

If your baby shows any of these signs, be sure to contact your baby's doctor immediately.

What is the Treatment for BIOPT-BS?

Dietary Treatment

Individuals with BIOPT-BS have trouble breaking down certain amino acids, including phenylalanine. This results in high levels of phenylalanine in the blood. Phenylalanine is found in many proteins and in artificial sweeteners. A dietician or nutritionist can help you plan a healthy diet for your child.

Medications and Supplements

Your baby's doctor may prescribe BH₄ supplements. BH₄ is a substance naturally made by the body, but individuals with BIOPT-BS may not make enough. BH₄ supplements can help reduce high phenylalanine levels. Your baby's doctor will need to write a prescription for these supplements.

Things to Remember

Children who receive early and on-going treatment for BIOPT-BS can have healthy growth and development. This is why newborn screening for BIOPT-BS is so important.