



## Newborn Screening FACT Sheet

# Biopterin-4alpha-carbinolamine dehydratase deficiency (BIOPT-REG)

### What is BIOPT-REG?

Biopterin defect in cofactor regeneration (BIOPT-REG) 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-REG 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-REG.

Biopterin defect in cofactor regeneration is only one form of tetrahydrobiopterin deficiency.

### What Causes BIOPT-REG?

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-REG occurs when the body cannot recycle and reuse BH<sub>4</sub>, the substance from which biopterin is made. Babies with BIOPT-REG either do not produce enough or produce non-working enzymes for recycling BH<sub>4</sub>.

BIOPT-REG affects either the 4 $\alpha$ -carbinolamine dehydratase enzyme or the dihydropteridine reductase enzyme. If either of these enzymes is not working, then the body cannot reuse BH<sub>4</sub>. When the body cannot recycle and reuse BH<sub>4</sub>, the body cannot make enough 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-REG?

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

Babies with BIOPT-REG usually seem healthy at birth. Signs of this condition may develop over time.

Signs of BIOPT-REG include:

- developmental delays
- seizures
- behavior problems or 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-REG?

#### Dietary Treatment

Your baby may need to be on a restricted diet to avoid the amino acid (a building block of proteins) phenylalanine.

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

#### Medications and Supplements

Your baby's doctor may prescribe BH<sub>4</sub> supplements. BH<sub>4</sub> is a substance naturally made by the body, but individuals with BIOP-REG may not have enough. BH<sub>4</sub> 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-REG can have healthy growth and development.