

Why Does My Baby Need More Testing for PKU?



State law requires that all babies have the newborn screening test before leaving the hospital. A few drops of blood were taken from your baby's heel and tested for certain diseases. Your baby now needs more testing as soon as possible. Not all babies with an initial "positive" result will have phenylketonuria (PKU).

California
Department of
Public Health



Newborn Screening Program
Genetic Disease Screening Program
www.cdph.ca.gov/nbs

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Why Does My Baby Need More Testing?

Your baby's test showed a "positive" result for PKU. More testing is needed to find out if your baby has this disease. Not all babies with an initial "positive" result will have this disease.

Babies can look healthy at birth and still have PKU. If untreated, this disease can cause severe health problems.

What Is PKU?

PKU is a disease that causes phenylalanine (an amino acid found in protein) to build up in the blood and body. Babies with this disease do not have enough of an enzyme called phenylalanine hydroxylase, which is needed to break down phenylalanine. Over time, this buildup of phenylalanine can cause mental retardation, seizures or convulsions, and eczema, a skin problem.

How Does a Baby Get This Disease?

A baby with PKU inherited a gene from each parent. Both parents must carry the gene for a baby to inherit this disease.

Is PKU Common?

About one in 28,000 babies in California (about 25 babies a year) is born with PKU.

Other babies can have a milder form of PKU. They may not need treatment. Newborn Screening only finds some of the babies with a milder form.

What Should I Do Right Now?

- Return to the doctor's office or hospital lab as instructed to have your baby tested again.
- Continue breast milk or milk-based formula feedings as directed by the doctor while waiting for the test result.
- Do not use any PKU formula product until the diagnosis of PKU has been made.

How Will I Know My Baby's Result?

The Newborn Screening staff reports the result to the doctor. The baby's doctor will let you know the results.

What If the Test Is Negative?

Your baby does not have PKU. You can breastfeed or use a milk-based formula. Further testing is not needed.

What If the Test Is Positive?

Your baby will be put on a special formula product right away. The doctor will refer you to a metabolic clinic for treatment of your baby.

How is PKU treated?

Babies with PKU must drink a special formula product. They cannot have regular infant formulas. When children with PKU start eating solid foods, they can't have foods that are high in protein.

Milk, eggs, meats, fish, cheese and other high protein foods are not allowed.



Foods that are low in protein will gradually be added to the diet. They will continue to take the special formula product every day. This formula is needed for growth.

Prompt and continuous dietary treatment helps the baby to grow and to develop normally and prevents mental retardation.

Where Can I Get More Information?

Call the doctor or the Newborn Screening staff listed below for any questions or concerns. For a complete list of diseases screened for by the California Newborn Screening Program visit our website at www.cdph.ca.gov/nbs