

Wisconsin Newborn Screen Positive Result Informational Handout Phenylketonuria or Phenylalanine hydroxylase deficiency (PKU)

What is the Wisconsin Newborn Screen?

All babies in Wisconsin have a test called the Newborn Screen (NBS) in their first 24 hours of life. A blood sample is taken from baby's heel for this test. This test looks for rare medical conditions that may not be caught or show up in your baby right away.

Newborn screening gets babies the help they need by:

- Finding out they have a condition before they have any medical problems
- Allowing treatment to be started right away
- Preventing medical emergencies

What was my child's NBS result?

Your baby's NBS has screened positive with a high level of a marker called "phenylalanine." This means there is a high chance that your baby has a condition called Phenylketonuria or Phenylalanine hydroxylase deficiency (PKU).

This test is a screening test. We will need more information to find out if he or she has this diagnosis.

What is PKU and why is it important?

PKU affects how protein is broken down in the body. Over time, a high level of phenylalanine is harmful to the brain. People with this disorder can develop developmental delay and seizures if they not treated for PKU at an early age. People with PKU who are treated from an early age may have no medical or developmental problems at all.

What are the next steps?

Your provider will give you instructions about when to have your baby seen either at their clinic or the emergency department. You should continue to feed your baby like you were (breastmilk or formula) until you are seen in the Genetics clinic and a dietitian helps you come up with a new feeding plan.

Tests and Doctor's Visits

This will help us know if your baby has PKU or not. Your baby's health care provider will order:

- An urgent visit to the primary care clinic and/or emergency department
- Referral to the Genetics Clinic
- Blood and urine (pee) tests

What if I have questions or concerns?

Contact your primary care provider right away: If your baby is not eating well, gets sick, or you have questions or concerns about how he or she is doing.

Call the Children's Wisconsin Genetics Center: If you have questions about Newborn Screen, PKU, or your baby's test results.

- Call 414-266-3347, option 3
- Leave a message and a nurse or genetic counselor from the Newborn Screen program will call back.
- Office is open Monday-Friday, 8 AM-4PM

Scan the QR codes below for links to more information

Medline Plus
Genetics



NewbornScreening.Info



National
PKU Alliance

