

Pompe Disease (PD)

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 baby's NBS result?

Your baby has screened positive on the Wisconsin newborn screen with a low marker called "acid alpha-glucosidase" or "GAA." This means your baby may have a condition called Pompe disease (PD), also called glycogen storage disease type 2.

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

What is PD and why is it important?

Pompe disease belongs to a group of conditions called glycogen storage disorders in which a complex sugar called glycogen builds up in the body and causes medical problems over time. Without treatment, babies with this condition can develop issues with their heart and muscles, but not all babies will have these problems right away.

What are the next steps?

Feed your baby every three hours, even overnight.

Do this while waiting for test results and more instructions.

Tests and Doctor's Visits

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

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

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.

Go to the emergency room right away: If your baby is not eating, throwing up, very sleepy and not waking up, has a seizure, or has other symptoms you are worried about.

Call the Children's Wisconsin Genetics Center: If you have questions about Newborn Screen, PD, 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:



Pompe & You:

