

X-linked adrenoleukodystrophy (X-ALD)

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 high level of a marker called "C26:0-Lysophosphatidylcholine (C26:0-LPC)." This means that your baby may have a condition called X-linked adrenoleukodystrophy (X-ALD) or other conditions that are less common.

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

What is X-ALD and why is it important?

X-ALD belongs to a group of conditions called peroxisomal disorders. All babies with X-ALD will be healthy with no signs of the condition. Boys may develop problems with their nerves and development in childhood or may not develop any problems until adulthood. Girls may develop problems with their muscles and nerves in adulthood but many never have any medical problems.

The other conditions that can cause high C26:0-LPC can cause problems in newborns, including difficulty eating and weak muscles.

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 X-ALD 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

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, X-ALD, 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:



Navigating
ALD:

