



X-Linked Adrenoleukodystrophy

What is newborn screening?

Newborn screening is a blood test to check for conditions that might be hidden at birth. To do the screening, a nurse takes a few drops of blood from your baby's heel soon after birth. This blood sample is required for all newborn babies.



Newborn screening is not the same as diagnostic testing. A diagnostic test can tell with more certainty whether or not a child has a condition. On the other hand, a screening test simply indicates that a child *may* have a

condition. The purpose of a screening test is to find babies that should have diagnostic testing.

What if my baby needs more testing?

If you are told that your baby needs follow-up testing, it does not necessarily mean that your baby is at risk. An out of range result may occur because:

- The sample was too small
- The sample was collected too early
- The sample was collected too close to a feeding
- The baby was born too early or had a low birth weight

Most babies who have follow-up testing for X-ALD are healthy, and will not be diagnosed. However, out of range screening results CAN indicate a disorder, so it is important to follow your doctor's advice & get your baby tested quickly so that final results can be confirmed.

KEY POINTS:

- You have just heard that your baby may have X-ALD. Please understand that the newborn screening is just that--- a screening test. Further testing is required to confirm or rule out the diagnosis.
- Many babies who have out of range newborn screens for X-ALD are healthy, and will not be diagnosed.
- If treated early, children with X-ALD can have healthy growth and development.

What is x-linked adrenoleukodystrophy?

Adrenoleukodystrophy (ALD) occurs when certain fats cannot be broken down in the body. These fats build up and affect how the body normally functions. This disease largely affects the nervous system and adrenal glands. ALD is different for each child and depends on if the child is male or female. Only boys have X-Linked ALD. If your baby is a girl, she may just be a carrier for X-ALD. There are three types of X-ALD found in males. The health problems and when problems start vary widely, even among family members. Newborn screening cannot tell the difference between these types.



What does this mean?

Although X-ALD cannot be cured, it can be treated. If further testing finds that your baby has X-ALD, he or she may need to have special therapies and/or medications. This will help prevent serious health problems.

What happens next?

Your baby's doctor may ask for the newborn screen to be repeated or, for your baby to have more testing. This follow up testing is important to know if treatment is needed. In some cases, you may be asked to visit a healthcare specialist. Long term monitoring by specialty centers will allow for appropriate timing of interventions.

What are the signs and symptoms of X-ALD?

Signs and symptoms of X-ALD can be very different from one baby to another. Some babies do not show any symptoms. Other babies can have vomiting, fatigue, weakness, learning disabilities and serious health problems. If you become concerned about your baby's feeding or activity, please talk to your child's pediatrician.

What if I still have questions?

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns. The Connecticut Newborn Diagnosis and Treatment Network (the Network) is available to answer questions and put you in touch with the best resource. To reach the Network, you can call 860-837-7870, Monday-Friday, 8:30am-4:30pm. We also recommend the website www.babysfirsttest.org as an accurate and informative resource.

This fact sheet was written for information purposes only. It should not replace professional medical advice, diagnosis or treatment.

Connecticut Department of Public Health
Connecticut Newborn Screening Program • 860.920.6628
Connecticut Newborn Diagnosis and Treatment Network • 860.837.7870
Adapted, with permission, from the Minnesota Department of Public Health

