



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 the baby's heel soon after birth.



Newborn screening is a screening test. A screening test is used to find which babies should have diagnostic testing before they become sick. Diagnostic testing is used to learn with more certainty whether or not a child has a condition.

What if my baby's newborn screen was reported as out of range and more testing is needed?

If you are told that your baby needs follow-up testing, it does not always mean that your baby has the disorder. An out of range result could 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 with this condition. However, out of range screening results CAN lead to a diagnosis, so it is important to follow your doctor's advice & get your baby tested quickly.

What happens next?

Your baby's doctor might ask for the newborn screen to be repeated or for your baby to have more testing. In some cases, you may be asked to visit a healthcare specialist.

KEY POINTS:

- **You have just heard that your baby might have X-ALD. Please understand that the newborn screening is just that: a screening test. More testing is needed 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 with X-ALD.**
- **If diagnosed early, children with X-ALD can have healthy growth and development.**

What is x-linked adrenoleukodystrophy?

X-linked Adrenoleukodystrophy (X-ALD) occurs when the body cannot break down certain fats (very long chain fatty acids). These fats build up and affect how the body functions. This disease can affect the nervous system (how body systems talk to each other) and adrenal glands (an organ in the body that makes hormones important for the body). The symptoms of X-ALD can be different for each person with the diagnosis. Most people with X-ALD have a slowly progressive neuromuscular disease that starts in adulthood (men can start in 30s and women can start around post-menopausal time). Some males (boys) can develop more severe symptoms at an earlier age. There are three main types of X-ALD among males. The health problems and when problems start vary widely, even among family members. Newborn screening cannot tell the difference between these types.



What are the signs and symptoms of X-ALD?

Signs and symptoms of X-ALD can be very different from one baby to another. Most babies do not show any symptoms. Others can have vomiting, fatigue, or muscle weakness. If you notice a change in your baby's behavior or feeding, talk to your child's pediatrician.

What treatment is available?

Although X-ALD cannot be cured, it can be treated. If further testing finds that your son has X-ALD, he will need to have his health monitored by specialists in neurology and endocrinology. Some people with X-ALD need to have special therapies and/or medications. If your daughter is confirmed to have X-ALD, then your healthcare team would discuss testing any brothers, and possibly the parents, to see if they might also have X-ALD. Since females with X-ALD are not expected to have symptoms until mid to late adulthood, medical interventions would be very unlikely in childhood.

What if I still have questions?

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns after they hear that their child's newborn screen has found that their child might benefit from more testing. The Connecticut Newborn Diagnosis and Treatment Network (the Network) is available to answer questions and/or put you in touch with the best resource(s). 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 informative resource on health conditions screened for with newborn screening.

**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

