

# X-Linked Adrenoleukodystrophy Fact Sheet for Parents

All babies born in North Carolina are screened at birth to look for certain medical conditions that can be treated if caught early. The newborn screening result showed it is possible your baby has X-linked adrenoleukodystrophy (X-ALD). Your baby will be referred to a specialist for additional testing to know for sure.

*There are usually no signs of X-ALD at birth.*

## What is X-ALD?

X-ALD is a rare genetic condition that can affect the brain and adrenal glands. People with X-ALD are unable to breakdown very long chain fatty acids (VLCFA). If VLCFAs are not broken down, they build up in the body and cause health problems. Males are more affected than females. Females are carriers of X-ALD and can pass it along to their children.

## What are the symptoms of X-ALD?

There are three different types of X-ALD, with symptoms ranging from mild to severe. The type of symptoms and the timing of their onset vary widely from person to person, even among members of the same family.

- **Cerebral ALD:** This type of X-ALD affects the brain and adrenal glands. Without early detection and treatment, it can cause blindness, loss of muscle control, and death.
- **Adrenomyeloneuropathy (AMN):** AMN affects the spinal cord. Patients may experience walking and balance problems, leg numbness or stiffness, and urinary disturbances. Females with ALD may also experience these symptoms in adulthood.
- **Addison Disease:** In this type, the body produces insufficient levels of the adrenal hormone, which can be life threatening if not treated.

## What happens next?

Your baby's primary care doctor will refer you to specialists at UNC Chapel Hill who are familiar with X-ALD and will work with your baby's health care provider. During a visit, the specialists will examine your baby and order additional testing. This testing will include taking a sample of blood from your baby.

Although the additional tests will confirm whether your baby has X-ALD, it is not possible to know which type your baby has until symptoms develop. There are other rare conditions that can cause a positive newborn screen result for X-ALD. If needed, your baby will also be tested for these.

## How is X-ALD monitored and treated?

X-ALD is treatable if detected early. If X-ALD is confirmed, your child will need regular visits with a team of specialists to monitor for disease progression. This monitoring will include routine bloodwork and MRIs of the brain. Corticosteroid therapy can treat adrenal problems. A bone marrow or stem cell transplant can slow or stop the progression of changes in the brain.

Your child's team of specialists will provide you with information about treatment options and help coordinate care through your child's health care provider.

## Where do I go for more information?

Use your phone's camera to scan the QR codes below.



**UNC Genetics:** <https://www.uncchildrens.org/uncmc/unc-childrens/care-treatment/genetic-disorders/#Newborn-Screening>



**Baby's First Test:** <https://www.babysfirsttest.org/newborn-screening/conditions/adrenoleukodystrophy>



**ALD Newborn Screening:** <https://aldnewbornscreening.org/>



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