



X-LINKED ADRENOLEUKODYSTROPHY (X-ALD)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from a baby's heel 24 to 36 hours after birth. The blood is tested for rare disorders that may affect a baby's health and development. A **positive newborn screen** means that a baby may have one of these disorders. More testing is needed to know for sure.

Your baby had a positive newborn screen for X-linked adrenoleukodystrophy (X-ALD). Your baby's primary care provider or the Newborn Screening Program will notify you to arrange for more testing.

What is X-ALD?

X-ALD is a genetic disorder that is passed on (inherited) from a parent to a child. X-ALD typically affects boys. Girls can be carriers and may develop mild symptoms as adults.

Boys with X-ALD cannot break down certain fats called very long chain fatty acids (VLCFAs). These fats build up in the body and can harm the brain, spinal cord, and adrenal glands. In the brain and spinal cord, VLCFAs damage the covering that protects the nerves. In the adrenal glands, they affect the way the body makes chemicals called hormones. Hormones help the body to work properly.

There are three main forms of X-ALD: childhood cerebral, adrenomyeloneuropathy (AMN), and Addison disease.

What problems can X-ALD cause?

It is not possible to know which form of X-ALD a boy has until he develops symptoms. Symptoms can be mild or severe, beginning in childhood or adulthood.

A boy with X-ALD may develop:

- Behavioral problems
- Learning difficulties
- Muscle weakness and difficulty walking
- Fatigue
- Nausea and vomiting

It is very important to follow the doctor's instructions for regular testing and early treatment.

What is the treatment for X-ALD?

There is no cure for X-ALD. Treatment is life-long and can include:

- Blood stem cell transplant (for childhood cerebral form)
- Hormone replacement therapy
- Physical therapy

Boys who have X-ALD should see their regular doctor. They should also see a doctor who specializes in X-ALD as part of a special care team.

Specialized treatment helps boys with X-ALD live the healthiest lives possible.

It is very important to get follow-up testing as soon as possible.

Resources and Support

Michigan Newborn Screening

Nurse Consultant

Toll-free: 1-866-673-9939

newbornscreening@michigan.gov

X-ALD Newborn Screening Coordinating Center

734-647-8938

Children's Special Health Care Services

Toll-free: 1-800-359-3722

Online Resources

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition/x-linked-adrenoleukodystrophy>

National Organization for Rare Disorders

<https://rarediseases.org/rare-diseases/adrenoleukodystrophy/>

Baby's First Test

www.babysfirsttest.org



Michigan Newborn
Screening

Phone 1-866-673-9939

www.michigan.gov/newbornscreening

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