

Parent Information

Inconclusive Newborn Screening Test Result for X-LINKED ADRENOLEUKODYSTROPHY (X-ALD) a-dree-no-luke-o-dis-tro-fee

What is Newborn Screening?

After babies are born, five drops of blood are collected from the baby's heel to test for certain diseases. These conditions usually cannot be found simply by looking at or examining a baby.

What is X-Linked Adrenoleukodystrophy (X-ALD)?

X-linked adrenoleukodystrophy is a genetic disorder that occurs primarily in males. It mainly affects the nervous system (brain) and the adrenal glands, which help the body fight stress.

A baby with X-ALD cannot break down certain fats (very long chain fatty acids) to be used for energy. These fats build up and cause health problems.

There are three types of X-ALD: a childhood cerebral form (most severe type), an adrenomyeloneuropathy type (AMN), and a form called Addison's disease. Newborn screening cannot tell the difference between these types. X-ALD is different for each child. Males and females are affected differently. The age when problems start varies widely, even among family members. X-ALD is a lifelong condition that may result in serious health problems.

What does an inconclusive result mean?

An inconclusive test result means that your baby needs to have further evaluation. Your baby's doctor may arrange for more testing, a consultation with a doctor who has expertise in X-ALD, and/or counseling regarding X-ALD and your baby.

What do I need to do now?

Your baby's doctor will tell you about the specific plan for your baby. If further testing is prescribed, it should be done without any delays even though your baby appears well. If your baby does have X-ALD, finding out quickly will allow treatment to be started promptly. It is also important to follow all of your baby's doctor's instructions for evaluation, testing, and treatment.

What problems can X-ALD cause?

In males, the childhood cerebral form of X-ALD causes hyperactive behavior, learning disabilities, seizures, blindness, paralysis, and death if untreated.

AMN is an adult onset type of X-ALD that affects the spinal cord causing leg weakness, bladder problems, and adrenal gland problems.

Males with Addison's disease will only develop adrenal symptoms if untreated which may include: vomiting, fatigue, low blood pressure, weakness, and skin darkening.

Females who are carriers for X-ALD have milder symptoms that start in adulthood. Some women may never show any symptoms.

How is X-ALD treated?

X-ALD can be treated. Possible treatments include: support therapies, adrenal hormone replacement and for those with cerebral disease, stem cell transplant.

Resources for parents

Nebraska Newborn Screening Program (402) 471-0374
www.babysfirsttest.org
www.dhhs.ne.gov/nsp/