

# X-linked Adrenoleukodystrophy (X-ALD)

## General Overview

### Q. What is X-ALD?

A. X-ALD is a treatable, inherited disorder that can show up in early childhood, adolescence or adulthood. It causes a buildup of very long-chain fatty acids in tissues and organs of the body that can affect the brain and adrenal glands.

### Q. How many forms of X-ALD are there?

A. There are three main forms of X-ALD: an adrenal form (the early form), a childhood cerebral form (the most severe) and a mild form that shows up in adulthood (the late form). X-ALD can present with different symptoms, even within the same family.

### Q. What does it mean if a baby has an abnormal X-ALD screen?

A. An abnormal screen means that a baby may have X-ALD. Diagnostic tests are needed to rule out or confirm X-ALD. If a person is diagnosed, there is no way to predict which form of X-ALD they will have.

### Q. How do I find out if a baby with an abnormal screen has X-ALD?

A. A blood test can detect the buildup of very long-chain fatty acids. Additional tests may be recommended by doctors who take care of people with X-ALD.

### Q. What are the effects of having X-ALD if it is not treated?

A. If the adrenal or cerebral forms of X-ALD are not treated, the buildup of very long-chain fatty acids can affect the adrenal glands and brain, leading to behavioral problems, muscle weakness, hearing loss, blindness, and potentially death.

### Q. What is the treatment for X-ALD?

A. The type of treatment depends on the form of X-ALD. Steroids are given to treat adrenal insufficiency. A stem cell transplant can stop X-ALD from getting worse for patients with the cerebral form. Doctors called biochemical geneticists take care of babies with X-ALD and can tell you about tests and treatments.

### Q. Why would a baby have X-ALD?

A. X-ALD is an inherited disorder, caused by a mutation in the ABCD1 gene. This gene is on the X chromosome, so boys are mainly affected. Newborn screening programs identify some girls who are carriers of an X-ALD mutation; they may develop mild symptoms as adults.

### Q. How common is X-ALD?

A. X-ALD occurs in about 1 in every 14,000 boys. The most severe form of X-ALD occurs in about 1 in 40,000 boys.

For more information: <http://www.ezrocks.org/ald.html> and <http://www.babysfirsttest.org> (search for ALD)

