

EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding X-linked adrenoleukodystrophy (X-ALD) to the Recommended Uniform Screening Panel (RUSP) in 2015.

About the condition

X-ALD is a rare disorder caused by a change in a single human gene. Studies of patients with symptoms suggest that about 2-3 out of every 100,000 people have X-ALD. People with X-ALD do not have enough of a protein that helps the body break down certain types of fats. Babies with X-ALD look normal. There are different types of X-ALD that can cause problems with the adrenal glands, brain, and spinal cord. Without treatment, these problems can worsen quickly and cause death during childhood. X-ALD usually affects boys more severely than girls.

Treatment for X-ALD

There is no cure for X-ALD. Early diagnosis allows early monitoring and treatment for babies with X-ALD. Available treatments include cortisol replacement and human stem cell transplant. The treatment a patient needs depends on many factors, like the type of X-ALD.

Detecting X-ALD in newborns

Newborn screening for X-ALD can happen along with routine newborn screening for other conditions in the first few days of life. Newborn screening for X-ALD measures levels of a certain fatty acid. This process uses the same dried blood spots already collected to screen for other disorders. Newborns with high fatty acid levels are at higher risk for X-ALD. They need more testing and evaluation to diagnose the condition.

Public health impact

Based on what is known about screening and the risk of being born with X-ALD, experts think that screening all newborns in the United States for X-ALD would find about 143 babies with the condition each year (about 3.6 out of every 100,000 children born). It would prevent up to 64 cases of serious disability/death and up to 44 deaths due to the disease each year.

Committee decision

The Committee voted in 2015 to recommend adding X-ALD to the RUSP. As of 2016, the RUSP recommends that state newborn screening programs include X-ALD.