

READ THIS FIRST

Abnormal Newborn Screen: Adrenoleukodystrophy (ALD or X-ALD)

This is a pilot screening project for ALD. This child's screen for ALD was found to be abnormal. This will not be reported on the newborn screening lab report.

What do I do?

1 - Newborns with ALD are asymptomatic. However, the screen can also detect children with other serious disorders. Assess the child for signs and symptoms of the Zellweger Spectrum of diseases including hypotonia, hepatomegaly, poor feeding, seizures, hearing loss, vision loss, distinctive facial features, or skeletal abnormalities.

2 - Collect diagnostic testing:

-Very Long Chain Fatty Acids

You can choose from any of these labs. Contact the labs directly for sample, shipping and billing requirements.

-Quest Diagnostics

Test Code: 90559 Test Name: Very Long Chain Fatty Acids

-Mayo Medical Laboratories

Test Code: POXP Test Name: Fatty Acid Profile, Peroxisomal (C22-C26), Plasma

-ARUP Laboratories

Test Code: 2004250 Test Name: Very Long-Chain and Branched-Chain Fatty Acids Profile

Fax all test results to the NBS Follow Up Team at 404-778-8564.

3 - For routine questions, please call the NBS follow up team at 404-778-8560.

4 - If there are clinical concerns, page genetics at 404-785-7778.

What is ALD?

ALD is an X-linked disorder that affects the nervous system white matter and also the adrenal cortex. There are three different presentations that can affect boys. Girls can also be affected later in life with a milder form of adrenomyeloneuropathy.

What do I tell this family?

Their child has had an abnormal newborn screen suggestive of ALD. This does not mean their child has this disorder. Diagnostic testing will tell us if their child is affected and needs treatment from specialists.

More information can be found at www.babysfirsttest.org. Search for ALD.