Arkansas Department of Health
Newborn Screening Result: Elevated C26:0 LCD
Infant may have X-Linked Adrenoleukodystrophy (X-ALD) Disorder
Information Sheet for Parents

You have just heard that your infant may have X-Linked Adrenoleukodystrophy (X-ALD) Disease. **Please understand that the newborn screening is just that – a screening test. Not all cases that are screened positive will be confirmed to have the diagnosis. The information below is meant to keep you informed while further testing is done to evaluate the newborn screening result.**

Overview:
X-ALD is a rare genetic disorder found primarily in males. This occurs when the body cannot break down very long chain fatty acids known as VLCFA which are found in foods. The ABCD1 gene is either missing or not working properly causing the VLCFAs to build up in the body. The ABCD1 gene is located on the X chromosome. A mutation in the ABCD1 gene will cause X-ALD.

What is X-ALD?
X-ALD is a single peroxisomal protein disorder that affects the nervous system and adrenal glands. This disorder is caused by mutations or changes in the ABCD1 gene which is located on the X chromosome. The mutations cause a deficiency of the ALDP protein. This will cause a very long chain fatty acid (VLCFA) to accumulate in the body. Increased levels of VLCFAs affect the adrenal cortex and the nervous system. X-ALD is primarily found in males. Females that have the same ALD mutation are carriers. There are three forms of X-ALD:
a) childhood cerebral form (which is primarily found in males)
b. adult onset (adrenomyeloneuropathy – referred to as AMN)
c. Addison disease.

Why is newborn screening done for X-ALD disease?
Newborn screening for X-ALD disease offers early detection so that treatment can begin earlier. Earlier detection for X-ALD is important for preventing many of the complications that may arise should the disorder go untreated during a child's infancy. Specialist may be able to improve the quality of life for individuals with X-ALD.

Does a positive newborn screening result mean that a baby has X-ALD?
Some babies who are screened will be identified as "positive" on screening, but later found to not have the disorder. Further testing needs to be done in order to determine if the baby has the disorder, or to rule it out as a "false positive".

How common is X-ALD?
X-ALD occurs in approximately 1:17,000 estimated for males and females.

What are the signs and symptoms of X-ALD?
No clinical signs are expected in newborns. In infancy you may see poor feeding, bony abnormalities, abnormal liver function testing, hypotonia, renal cysts, sphincter paraparesis and increased skin pigmentation.

How is X-ALD diagnosed?
After receiving a positive newborn screen, the most important thing parents can do is to be sure that their baby has confirmatory testing as soon as possible.

Is there a cure for X-ALD?
There is no cure for X-ALD. There are treatment options which can help reduce the risk of complications when started early.

How is X-ALD treated?
Adrenal steriod replacement for adrenal insufficiency and hemotopoietic stem cell transplantation may be helpful. Seeing specialists in neurology and endocrinology to help with evaluation and management of this disease is very important.

Where can I get additional information?
http://www.arnewbornscreening.com
https://www.huntershope.org/family-care/leukodystrophies/adrenoleukodystrophy/
https://rarediseases.org/rare-diseases/adrenoleukodystrophy/
https://www.babysfirsttest.org/newborn-screening/conditions/adrenoleukodystrophy