

Mission:

To protect, promote & improve the health of all people in Florida through integrated state, county & community efforts.



Rick Scott
Governor

Celeste Philip, MD, MPH
Surgeon General and Secretary

Vision: To be the Healthiest State in the Nation

April 27, 2018

Dear Colleague:

The Florida Department of Health, Newborn Screening Program, is pleased to announce that beginning May 1, 2018, all newborns screened will be tested for X-Linked Adrenoleukodystrophy (X-ALD). This addition to the panel of disorders screened in Florida was recommended in 2016 by the Florida Genetics and Newborn Screening Advisory Council and funding was authorized during the 2017 Legislative session.

It is important that each provider become familiar with the protocol following identification of a positive result for X-ALD. An X-ALD information sheet is enclosed. Infants with presumptive positive results will be referred to the nearest Genetic Specialty Center. The referral centers are located in Gainesville, Tampa and Miami. If the diagnosis is confirmed, the baby will then be referred to Endocrinology and Neurology for additional management.

Follow-up staff from the Newborn Screening Program will direct these procedures and advise you of actions needed. These procedures will only apply to X-ALD; no changes will be made to the protocols currently practiced for other conditions on the screening panel.

If you have any questions, please contact the Florida Newborn Screening Program at 850-245-4201. Thank you for your continued support of the Florida Newborn Screening Program.

Sincerely,

Celeste Philip, MD, MPH
Surgeon General and Secretary

Enclosure

Newborn Screening for X-Linked Adrenoleukodystrophy in Florida

Definition: It is a genetic disorder that occurs primarily in males. It mainly affects the nervous system and the adrenal glands. There are three types of X-ALD: a childhood cerebral form, an adrenomyeloneuropathy form, and a form called Addison's disease only.

Incidence: X-ALD affects 1 in 17,000 individuals (males and females) worldwide.

Florida Method of Screening:

First Tier Screening: Screening for X-ALD is completed by analysis of very long chain fatty acids (VLCFA) using mass spectrometry, specifically the concentration of C26:0. If the concentration is normal, the sample is deemed within normal limits. If abnormal, second tier screening is performed.

Second Tier Screening: Samples with VLCFA concentrations above a certain threshold will be tested a second time using a more specific assay. If the concentration is normal, the sample is deemed within normal limits. If abnormal, a repeat specimen will be collected.

- Repeat specimens will be specifically marked for X-ALD testing.
- Repeat specimens will have second tier screening completed. If concentration is normal, the sample is deemed within normal limits. If abnormal, third tier screening is performed.

Third Tier Screening: Sequencing of the ABCD1 gene. Persistent elevations of VLCFAs still warrant a referral regardless of whether an ABCD1 mutation is detected.

Referral to Genetic Specialty Center: Infants with a second abnormal newborn screen for X-ALD are referred to one of three Genetic Specialty Centers for diagnostic evaluation. If diagnosed with X-ALD, patients will be referred to Pediatric Endocrinology and Pediatric Neurology for clinical assessments of adrenal and neurological function and need for treatment.

For more information about X-ALD, please visit the following sites:

ALD Connect www.aldconnect.org/index.php

The Myelin Project www.myelin.org/ald-adrenoleukodystrophy

Gene Reviews <http://www.ncbi.nlm.nih.gov/books/NBK1315/>

Fight ALD (X-ALD information from the parent's perspective) www.fightald.org

Genetics Home Reference <http://ghr.nlm.nih.gov/condition/x-linked-adrenoleukodystrophy>

Genetic and Rare Diseases Information Center
<https://rarediseases.info.nih.gov/diseases/5758/x-linked-adrenoleukodystrophy>

National Institute of Neurological Disorders and Stroke
www.ninds.nih.gov/disorders/adrenoleukodystrophy/adrenoleukodystrophy.htm