



**Missouri Department of Health and Senior Services**

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## **X-ALD Newborn Screening in Missouri Goes Live**

### **Dear Practitioner, Laboratory Manager, or Newborn Nursery Manager:**

The Missouri Department of Health and Senior Services' Newborn Screening Program launched a full population pilot in December 2021 to screen all Missouri newborns for X-linked Adrenoleukodystrophy (X-ALD). While screening results for X-ALD have not appeared on the routine NBS laboratory reports during this pilot/implementation period, all newborns were tested for this disorder, and any displaying high risk abnormal results were referred for further confirmatory testing.

Effective December 1, 2022, the Missouri State Public Health Laboratory (MSPHL) will be going live with the routine reporting of X-ALD on the standard NBS laboratory reports. All infants with high-risk abnormal results for X-ALD will be phoned and faxed to the health care provider of record and to our contracted genetic referral centers who will coordinate confirmatory testing and next steps.

Adrenoleukodystrophy is an X-linked genetic disease that affects males most severely. ALD primarily affects the nervous system and the adrenal glands. ALD may cause progressive loss of the myelin sheath, which acts as an insulator for the nerves in the brain and spinal cord. This can result in a variety of neurological problems including cognitive, mobility, and sensory issues. ALD may also cause a deficiency of certain hormones due to damage to the adrenal glands. Adrenal insufficiency may cause weakness, weight loss, skin changes, vomiting, and coma. If left untreated, the severe form of ALD can lead to critical and irreversible disabilities that can ultimately lead to death. Early diagnosis through newborn screening allows for proactive care, consistent monitoring, and lifesaving treatment.

As the MSPHL continues screening, there is an increased likelihood for providers to have newborns in their care with high-risk newborn screening results for ALD. In preparation of caring for these families, the Missouri Newborn Screening Program would like to encourage you to review the following resources.

#### **Provider Education**

- American College of Medical Genetics (ACMG) ACT Sheet at [www.acmg.net/PDFLibrary/X-ALD-ACT-Sheet.pdf](http://www.acmg.net/PDFLibrary/X-ALD-ACT-Sheet.pdf).
- National Coordinating Center for the Regional Genetics Networks, NCC Knowledge Nugget Series: X-ALD ACT Sheet at [www.acmgeducation.net](http://www.acmgeducation.net).

#### **HEALTHY MISSOURIANS FOR LIFE**

The Missouri Department of Health and Senior Services will be the leader in promoting, protecting and partnering for health.

AN EQUAL OPPORTUNITY / AFFIRMATIVE ACTION EMPLOYER: Services provided on a nondiscriminatory basis.

- Information for pediatric health care providers at [www.babysfirsttest.org/newborn-screening/pediatric-providers](http://www.babysfirsttest.org/newborn-screening/pediatric-providers).

#### Parent Resources

- ALD Alliance, at [www.aldnewbornscreening.org](http://www.aldnewbornscreening.org), helps families across the United States that are newly diagnosed with ALD by giving them the resources they need to fight this rare and devastating disease.
  - *ALD Brochure: Our Parent's Guide to Adrenoleukodystrophy* – available in English and Spanish at [www.aldnewbornscreening.org/ald-brochure](http://www.aldnewbornscreening.org/ald-brochure).
  - *Care Packages for Newborn Screening Families* – families who have a baby diagnosed through newborn screening can register for a free care package at [www.aldalliance.org/family-support](http://www.aldalliance.org/family-support).
- The STAR-G Project, funded through HRSA, developed a website, [www.newbornscreening.info](http://www.newbornscreening.info), and parent fact sheets, which are now maintained and updated by the Western States Regional Genetics Network. There are downloadable PDF fact sheets designed specifically for males and females with ALD, available in English and Spanish.
  - [www.newbornscreening.info/x-ald\\_female-x-linked-adrenoleukodystrophy/](http://www.newbornscreening.info/x-ald_female-x-linked-adrenoleukodystrophy/)
  - [www.newbornscreening.info/x-ald\\_male-x-linked-adrenoleukodystrophy/](http://www.newbornscreening.info/x-ald_male-x-linked-adrenoleukodystrophy/)
- Knockout ALD is a non-profit dedicated to raising awareness of and funds for ALD research, and is the only ALD organization in the Midwest. Knockout ALD can provide parent-to-parent resources for families with a new diagnosis through newborn screening. Parents can contact the organization through the website, [www.knockoutald.com](http://www.knockoutald.com), or via email at [runforald@yahoo.com](mailto:runforald@yahoo.com).

For more information about Missouri's newborn screening program, please contact [tracy.klug@health.mo.gov](mailto:tracy.klug@health.mo.gov) at the Missouri State Public Health Laboratory or [lori.swartz@health.mo.gov](mailto:lori.swartz@health.mo.gov) with the Missouri Newborn Screening Follow-up Program.