PUBLIC HEALTH DIVISION
Oregon State Public Health Laboratory
Northwest Regional Newborn Bloodspot Screening Program
Kate Brown, Governor

Oregon Health Authority

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To: Oregon birth facilities and health care providers

From: Patrice Held, Newborn Screening Program Manager

Subject: Addition of X-Linked Adrenoleukodystrophy to the Newborn Bloodspot Screening Panel

Beginning on January 1, 2023, the Northwest Regional Newborn Bloodspot Screening (NWRNBS) Program will add X-Linked Adrenoleukodystrophy (X-ALD) to its screening panel and initiate testing for all newborns.

What is changing?
Beginning on January 1, 2023, all specimens, including first and second screens, received for newborn screening at the OSPHL will be tested for X-Linked Adrenoleukodystrophy (X-ALD), in addition to the other disorders already on the screening panel.

The practitioner's manual containing information about X-ALD and other disorders can be found at www.bitly.com/or-nbs-manual (pdf).

How will the result report change?
For all specimens, the result table on the report will include X-Linked Adrenoleukodystrophy. The screening test measures the amount of a specific very long chain fatty acid, C26:0-LPC. Below are examples of potential results.

- If C26:0-LPC is within the normal range, this is a **normal screening result** and the baby is NOT at an increased risk for X-ALD.
- If C26:0-LPC is elevated, the baby is at an increased risk for X-ALD and will require either a repeat screen (**borderline screening result**) or diagnostic testing and follow-up with medical genetics (**abnormal screening result**).

Normal Screening Result

Screening Test	Analyte Result	Disorder Evaluation	Reference
X-Linked Adrenoleukodystrophy	Normal	Normal	Normal

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Borderline Screening Result

Screening Test	Analyte Result	Disorder Evaluation	Reference
X-Linked Adrenoleukodystrophy	C26:0=0.12	Borderline	C26:0-LPC<0.1 µM

Action Needed: Send a routine 2nd or repeat bloodspot within 7 days of receiving this result. Contact Newborn Screening Follow Up at 503-693-4174 with questions.

Abnormal Screening Result

Screening Test	Analyte Result	Disorder Evaluation	Reference
X-Linked Adrenoleukodystrophy	C26:0=0.16	Abnormal	C26:0-LPC<0.1 µM

Action Needed: This infant has been referred to a medical consultant. Send a repeat bloodspot immediately and order diagnostic testing. Contact Newborn Screening Follow Up at 503-693-4174 with questions.

How do birth facilities and health care providers implement this change?
There is no change to the process for collection of the first and second newborn bloodspot specimens. Birth facilities and health care providers should continue to collect specimens and review result reports for each infant.

If there is an abnormal result, the result report will detail additional actions you must take immediately to care for the infant. A phone call by the newborn bloodspot screening program will also accompany the abnormal result report.

What is X-Linked Adrenoleukodystrophy?
X-Linked Adrenoleukodystrophy (X-ALD) is a genetic disorder that occurs when very long chain fatty acids (VLCFA) cannot be broken down. The build-up of VLCFA damages the nervous system (brain and spinal cord) and adrenal glands which produce hormones. The newborn bloodspot screening will identify all three forms of X-ALD: childhood cerebral, adrenomyeloneuropathy (AMN), and adrenal-insufficiency. Symptoms may include progressive impairment of cognition, behavior, vision, hearing and motor function. Treatment includes corticosteroid therapy, hematopoietic stem cell transplant and gene therapy.

X-ALD is inherited in an X-linked manner. Heterozygous females may also be identified through screening and may develop symptoms later in life.

Please refer to the ACMG fact sheet for more information about newborn screening for X-ALD.
https://www.acmg.net/PDFLibrary/X-ALD-ACT-Sheet.pdf

For more information about the Oregon Newborn Bloodspot Screening Program, please visit www.healthoregon.org/nbs.

Why did the NWRNBS Program add this test to the screening panel?
The U.S. Secretary of Health and Human Services approved the addition of X-Linked Adrenoleukodystrophy to the Recommended Uniform Screening Panel for Newborn Bloodspot Screening. The Northwest Regional Newborn Bloodspot Screening Advisory Board, who advises the OSPHL on their screening panel, reviewed the condition and unanimously decided to add X-ALD. In

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order to meet national standards and comply with the recommendations of the advisory board, the OSPHL will perform testing for X-ALD starting on January 1, 2023.

If you have any questions regarding this announcement, please contact the newborn bloodspot screening program by phone at: 503-693-4173 or by email at: nwregional.nbs@odhsoha.oregon.gov

Sincerely,

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