Public Health Division

Oregon State Public Health Laboratory
Northwest Regional Bloodspot Screening Program

Date: September 8, 2025

To: Oregon birth facilities and health care providers

From: Northwest Regional Newborn Bloodspot Screening (NWRNBS) Program

Subject: Addition of Infantile Krabbe Disease to the Newborn Bloodspot Screening Panel

Beginning on November 1, 2025, the NWRNBS Program will add Infantile Krabbe Disease (IKD) to its screening panel and initiate screening for all newborns.

What is changing?

On November 1, 2025, the first valid specimen received for newborn screening at the OSPHL will be screened for Infantile Krabbe Disease in addition to the other disorders already on the screening panel.

How will the result report change?

For all specimens, the result table on the report will include Infantile Krabbe Disease. The screening test measures GALC enzyme activity. If GALC enzyme activity is low, the sample will be sent for second tier testing to measure psychosine concentrations in the dried blood spot sample. Below are examples of potential results.

- If GALC enzyme activity is within the normal range, this is a normal screening result and the baby is NOT at an increased risk for IKD.
- If GALC enzyme is low and psychosine concentration is markedly elevated (>10nmol/L), this is a time critical abnormal screening result, and the baby is at high risk for IKD.
- If GALC enzyme is low and psychosine concentration is mildly elevated (between 2-10nmol/L), the baby may be at risk for later onset forms of Krabbe disease.

7202 NE Evergreen Pkwy, Suite 100, Hillsboro, OR 97124
Voice: 503-693-4174 | Fax: 503-693-5600 | All relay calls accepted | www.healthoregon.org/nbs

Example Reports:

Normal

Screening Test	Analyte Result	Disorder Evaluation	Reference
Infantile Krabbe Disease	Normal	Normal	Normal

Abnormal Time Critical

Screening Test	Analyte Result	Disorder Evaluation	Reference
Infantile Krabbe Disease	GALC Low Markedly elevated PSY	IKD Abnormal	Normal

Abnormal

Screening Test	Analyte Result	Disorder Evaluation	Reference
Infantile Krabbe Disease	GALC Low Mildly elevated PSY	See Comments	Normal

How do birth facilities and health care providers implement this change?

There is no change to the process for collection of the 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 actionable abnormal result, the comment section of the result report will detail additional immediate actions you must take for the baby.

What is Infantile Krabbe Disease?

Infantile Krabbe Disease is caused by a deficiency of the lysosomal enzyme, galactocerebrosidase (GALC), which leads to increased psychosine (PSY) levels. Psychosine is toxic at high concentrations. Babies with IKD will appear normal at birth, but quickly develop symptoms including extreme irritability, feeding difficulties, failure to thrive and spasticity. These symptoms can worsen rapidly and cause death by age 2. Hematopoietic stem cell transplantation (HSCT) is the recommended treatment for newborns confirmed to have IKD. Transplant should occur by six weeks of life for the best

outcomes. IKD is the screening target, but babies with later onset forms of the disease may be detected.

Please refer to the ACMG fact sheet for more information about newborn screening for Infantile Krabbe https://www.acmg.net/PDFLibrary/Krabbe-Infantile.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 Infantile Krabbe Disease to the Recommended Uniform Screening Panel for Newborn Bloodspot Screening. The advisory board for the NWRNBS Program reviewed the condition and unanimously recommended its addition to the Oregon panel March 24, 2025.

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