



**Wisconsin Division of Public Health  
Ad Hoc Metabolic Subcommittee Meeting**

**Thursday, December 18, 2025**

**12:00 pm -1:00 pm**

Zoom: <https://dhs.wi.zoomgov.com/j/1616847912?pwd=HSYXrasKhhRiUaya5RLuaidhiamaVf.1>  
Or call: +1 669 254 5252 or +1 646 828 7666 or +1 551 285 1373 or +1 669 216 1590

Meeting ID: 161 684 7912

Minutes

| <b>Meeting Members:</b> |                          |   |                    |   |                             |
|-------------------------|--------------------------|---|--------------------|---|-----------------------------|
| X                       | Dr. Mei Baker            |   | Dr. Philip James   | X | Stephanie Offord            |
| X                       | Dr. Donald Basel (Chair) | X | Jessica Kopesky    | X | Matthew Rasberry            |
| X                       | Beth Boyd                | X | Ashley Kuhl        |   | Dr. William Rhead           |
|                         | Therese Breunig          |   | Dr. Jennifer Kwon  | X | Dr. Jessica Scott-Schwoerer |
|                         | Nicoletta Drilias        |   | Wanda Meeteer      | X | Emily Singh                 |
| X                       | Dr. Michael Finkel       |   | Dr. Roberto Mendez | X | Dr. Bob Steiner             |
| X                       | Gretchen Heckel          | X | Kaegan Mestel      | X | Dr. Julie Thiel             |
| X                       | Sonja Henry              |   | Susana Morphis     |   | Mary Marcus Walters         |
|                         | Caitlin Hessenthaler     | X | Dr. Mike Muriello  |   | Dr. Katie Williams          |
| X                       | Tami Horzewski           |   | Lisa Obernolte     |   |                             |
| <b>Meeting Guests:</b>  |                          |   |                    |   |                             |
| X                       | Zoe Culshaw-Klein        |   | Kelly Gill         | X | Travis Henry                |
|                         | Mandy Quainoo            |   | Leah Ricci         | X | Errin Rider                 |
|                         | Tamara Thompson          | X | Amy White          | X | Lesa Brackbill              |

|  |   |
|--|---|
|  | <p><b>Purpose of Meeting:</b></p> <p>The Metabolic Subcommittee serves in an advisory capacity to the Umbrella Committee and NBS Program regarding the specific screening-related condition(s) in the following areas:</p> <p>Expert Knowledge in Condition-Specific Clinical Care and Research</p> <ul style="list-style-type: none"> <li>• Advises on condition-specific clinical care including ongoing changes in treatment options for affected individuals, emerging newborn screening technology, and condition-specific research.</li> </ul> <p>Quality Assurance</p> <ul style="list-style-type: none"> <li>• Reviews de-identified screening and confirmatory testing data to monitor screening test performance, such as screening positive predictive value, screening false positive rate and false negative rate.</li> <li>• Recommends changes in NBS practice to the Umbrella Committee and NBS Program.</li> </ul> <p>Education for Families and Providers</p> <ul style="list-style-type: none"> <li>• Reviews and provides feedback on information and education materials for general public, families, and providers to assure accuracy of health information</li> </ul> |
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|            |  |   |  | PSY10  |            |  |            |  |           |  |
|------------|--|---|--|--|------------|--|------------|--|-----------|--|
|            |  |   |  | Code   |            |  |            |  |           |  |
|            |  |   |  | Comment  |            |  |            |  |           |  |
|            |  |   |  | <table border="1"> <tr> <td>PSY Normal</td> <td>*NEGATIVE* In this sample, the concentration of psychosine was normal.</td> </tr> <tr> <td>PSY10 2-10</td> <td>In this sample, the concentration of psychosine is not indicative of infantile Krabbe disease. However, a later onset form of Krabbe disease or atypical Krabbe disease due to saposin A cofactor deficiency (due to pathogenic variants in the prosaposin gene, PSAP) cannot be excluded. We recommend confirmatory testing to include determination of GALC activity in leukocytes (MCL test ID: GALCW), measurement of psychosine in erythrocytes (MCL test ID: PSYR), and molecular genetic analysis of the GALC gene (MCL test ID: GALC).</td> </tr> <tr> <td>PSY10 &gt;10</td> <td>*POSITIVE* In this sample, the concentration of psychosine is consistent with infantile Krabbe disease due to galactocerebrosidase (GALC) deficiency. Less likely is atypical Krabbe disease due to saposin A cofactor deficiency caused by pathogenic variants in the prosaposin gene, PSAP. We recommend immediate follow up and referral to an experienced transplant center, as well as confirmatory testing to include determination of GALC activity in leukocytes (MCL test ID: GALCW), measurement of psychosine in erythrocytes (MCL test ID: PSYR), and molecular genetic analysis of the GALC gene (MCL test ID: GALC).</td> </tr> </table> | PSY Normal | *NEGATIVE* In this sample, the concentration of psychosine was normal. | PSY10 2-10 | In this sample, the concentration of psychosine is not indicative of infantile Krabbe disease. However, a later onset form of Krabbe disease or atypical Krabbe disease due to saposin A cofactor deficiency (due to pathogenic variants in the prosaposin gene, PSAP) cannot be excluded. We recommend confirmatory testing to include determination of GALC activity in leukocytes (MCL test ID: GALCW), measurement of psychosine in erythrocytes (MCL test ID: PSYR), and molecular genetic analysis of the GALC gene (MCL test ID: GALC). | PSY10 >10 | *POSITIVE* In this sample, the concentration of psychosine is consistent with infantile Krabbe disease due to galactocerebrosidase (GALC) deficiency. Less likely is atypical Krabbe disease due to saposin A cofactor deficiency caused by pathogenic variants in the prosaposin gene, PSAP. We recommend immediate follow up and referral to an experienced transplant center, as well as confirmatory testing to include determination of GALC activity in leukocytes (MCL test ID: GALCW), measurement of psychosine in erythrocytes (MCL test ID: PSYR), and molecular genetic analysis of the GALC gene (MCL test ID: GALC). |
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| Share      | 12:20-12:40  | Share State Survey Information and Related Data (Zoe Culshaw-Klein) |  | <p>State Survey data on psychosine result report levels was shared. Data included the number of states that have changed from screening at Psychosine levels equal to or greater than ten to Psychosine levels between two and ten.</p> <p>Data was also shared on clinical versus newborn screening for Krabbe Disease.</p>   |            |  |            |  |           |  |
| Disc/Dec   | 12:40-12:55  | Discussion & Vote (Dr. Basel/Tami Horzewski)                        |  | <p>Motion proposed by Dr. Jess Scott -Schwoerer to withdraw the nomination and go back through the committee process to reassess for full Krabbe Disease. There was not a second on the motion.</p> <p>Motion proposed by Emily Singh to amend the current nomination of Infantile Krabbe Disease with Psychosine levels equal to or greater than ten to</p>   |            |  |            |  |           |  |



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|      |            |   |  | <p>Krabbe with Psychosine levels equal to or greater than two.<br/>The motion was seconded by Mei Baker.<br/>The motion was approved 7-1.</p> <p>The subcommittee discussed that the initial nomination submitted was for full Krabbe and the subcommittee decided at the time to follow the RUSP and go with Infantile Krabbe with Psychosine levels equal to or greater than ten. There was much discussion about full Krabbe at the initial Metabolic Subcommittee meeting.</p> <p>Since the SACNBS already sent their report to the DHS Secretary and the condition was approved, if the Metabolic Subcommittee's recommendation/motion to amend the nomination were to be accepted by the SACNBS, a letter to the DHS Secretary noting the change would be needed.</p> |
| Disc | 12:55-1:00 | Summarize Next Steps<br>(Dr. Basel/Dr. Steiner) |  | <p>Dr Basel will prepare a letter for the Umbrella Committee and SACNBS chairs providing some background information, and the motion proposed at the Ad Hoc Metabolic Subcommittee meeting.</p> <p>Tami will email Dr. Fost and work on scheduling an Ad Hoc SACNBS meeting for early-mid January.</p>  |

Next meeting date: TBD

“Parking Lot” Items:



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