**Wisconsin Division of Public Health**
**Metabolic Subcommittee Meeting**
**Minutes**
**Friday, October 17, 2025**
**10:00 am -2:00 pm**
Zoom: https://dhswi.zoomgov.com/j/1618839019?pwd=b4JirWyygxwgkIw4S7igJ6NgMaplXm.1
Or call: +1 669 254 5252 or +1 646 828 7666 or +1 551 285 1373 or +1 669 216 1590
Meeting ID: 161 883 9019

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| **Meeting Members:** | | | | | |
| X | Dr. Mei Baker | | Dr. Sam Huang | X | Lisa Obernolte |
| X | Dr. Donald Basel (Chair) | | Dr. Philip James | X | Stephanie Offord |
| X | Beth Boyd | | Dr. Kelly Jones | X | Matthew Rasberry |
| X | Therese Breunig | X | Jessica Kopesky | X | Dr. William Rhead |
| | Anna Cisler | X | Ashley Kuhl | X | Dr. Jessica Scott-Schwoerer |
| X | Nicoletta Drilias (Nikki) | X | Dr. Jennifer Kwon | X | Emily Singh |
| X | Dr. Michael Finkel | X | Wanda Meeteer | X | Dr. Bob Steiner |
| | Gretchen Heckel | | Dr. Roberto Mendez | X | Dr. Julie Thiel |
| X | Sonja Henry | X | Kaegan Mestel | X | Tammi Timmler |
| | Caitlin Hessenthaler | | Susana Morphis | X | Mary Marcus Walters |
| X | Tami Horzewski | | Dr. Mike Muriello | X | Dr. Katie Williams |
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| **Meeting Guests:** | | | | | |
| X | Amy White | X | Dr. Sheldon Garrison | X | Dr. Justin Hopkin |
| X | Dr. Joshua Baker | X | Mandy Quainoo | X | Leah Ricci |
| X | Kelly Gill | X | Travis Henry | X | Tamara Thompson |
| X | Kari Lato (Rare and Ready Alliance, focused on NBS) | X | Aviva Rosenberg (Gaucher Community Alliance) | | |

**Purpose of Meeting:**

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:

Expert Knowledge in Condition-Specific Clinical Care and Research
- Advises on condition-specific clinical care including ongoing changes in treatment options for affected individuals, emerging newborn screening technology, and condition-specific research.

Quality Assurance
- 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.
- Recommends changes in NBS practice to the Umbrella Committee and NBS Program.

Education for Families and Providers

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| | • Reviews and provides feedback on information and education materials for general public, families, and providers to assure accuracy of health information related to the specific condition(s).<br><br>Addition/Deletion of Conditions<br>• Recommends and/or reviews nominations for addition and deletion of conditions related to the subcommittee<br><br>**Facilitator/Chair:** Dr. Donald Basel **Recorder:** Tami Horzewski |

| Info Decision Discussion | Time: | Topic(s) | Follow-up Items Decision/action: Name: Date due: | Content-focused minutes |
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| Decision | 10:00-10:10 | Welcome, Review and Approve Minutes - Dr. Basel (Chair) | | • Introductions were made for newcomers.<br>• Jess Kopesky made a motion to approve the minutes. Dr. Kwon seconded. Motion passed to approve the minutes from the 4.11.25 Metabolic Subcommittee meeting. |
| Info | 10:10-10:30 | Acid Sphingomyelinase Deficiency (ASMD)/Niemann-Pick Disease Review of New Information<br>(Dr. Basel/Dr. Justin Hopkin/Dr. Joshua Baker) | | • Review of acid sphingomyelinase deficiency (ASMD) / Niemann-Pick Disease. We have voted on the NBS criteria in the past. Three criteria needed more information (#3, 6, 8), so new information related to these three criteria was discussed.<br>• Dr. Justin Hopkin discussed the studies related to the three criterion that needed more information for ASMD. Enzyme replacement theory: Infantile neurovisceral (type A) (unpublished) study of 3 patients ages 4, 5, and 6. Improved nutritional status and feeding, sleeping, and respiratory symptoms, Sibling study of two brothers after starting enzyme replacement therapy. Study in France of 271 patients for type B (chronic visceral form) and type AB (chronic neurovisceral form) and type A (infantile neurovisceral)<br>• Dr. Joshua Baker presented from Lurie Children's in Illinois. 9 patients are being monitored for ASMD because of genetic testing, mostly type B. 3 of those are receiving treatment.<br>• Dr. Hopkin discussed clinical protocol used at Lurie Children's and reported no patients had been unable to access the treatment they desired.<br>• Incidence is about 1 in 140,000 births. One of the rarest of rare diseases.<br>• From Lurie's patient population: One family expressed they wished they didn't know about ASMD from screening because it's stressful, |

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| | | | | but most families are grateful to know the answer and not have such a long diagnostic odyssey. Even if asymptomatic, families can watch for symptoms and act as quickly as possible once symptoms arise.<br>• No biomarker validated yet to determine if patients will be symptomatic or when to start treatment. |
| Decision | 10:30 - 10:45 | Review and Vote<br>(Dr. Basel/Tami Horzewski/Voting Members) | | Voting – 11 members<br>• Criterion 3 – 11 meets<br>• Criterion 6 – 10 meets, 1 needs more information<br>• Criterion 8 – 11 meets<br><br>Emily Singh made a motion to take the recommendation to add ASMD to the Wisconsin newborn screening panel to the Umbrella Committee. Dr. Mei Baker seconded the motion. Motion approved by 10. 1 voter abstained because we may find individuals that are not going to be symptomatic for a very long time. Motion passed. |
| Info | 10:45– 11:10 | Gaucher Nomination Background Information<br>(Dr. Garrison/ Dr. Finkel) | | Dr. Michael Finkel, DO, Children's Wisconsin & Medical College of Wisconsin. Dr. Finkel gave an overview of Gaucher Disease.<br>• Gaucher Disease (GD)<br>o Deficiency in glucocerebrosidase<br>o Autosomal recessive due to biallelic variants in *GBA1* gene<br>o Broad span of age at onset of clinical symptoms<br>o Variable signs and symptoms<br>o Variable severity and rate of disease progression<br>• Cellular Dysfunction<br>o Misfolding of lysosomal membrane-bound GCase proteins leads to cellular dysfunction<br>o Accumulation of GL1 and other glycolipids<br>o Storage of GL1 in macrophage lineage of reticuloendothelial system<br>o Accumulated Gaucher cells cause<br>▪ Hepatosplenomegaly<br>▪ Bone marrow infiltration leading to bone pain and cytopenias<br>▪ CNS symptoms in some due to neuronal cell death<br>• Clinical subtypes<br>o 3 subtypes |

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| | | | | ▪ Type 1 non-neuronopathic<br>▪ Type 2 acute neuropathic<br>▪ Type 2 subacute neuronopathic<br>o Delineated by the involvement of the central nervous system and its progression<br>• Additional features/risks<br>o Growth restriction and puberty delay in untreated children<br>o Increased risk for multiple myeloma<br>o Pulmonary infiltration or hypertension<br>o Neurodegeneration in neuronopathic types (GD2 and 3)<br>o Manifestations are irreversible in 25% of GD patients without timely ERT<br>o Association with Parkinson Disease<br>• Phenotypic variation in GD<br>o Type 1<br>▪ Early Dx: Childhood<br>▪ Late Dx: Adulthood<br>o Type 2<br>▪ Perinatal/Lethal: Perinatal/Newborn<br>▪ Classical: Infancy<br>o Type 3<br>▪ 3a: Childhood<br>▪ 3b: Childhood<br>▪ 3c: Childhood<br>• Prevalence in US is 1 in 50,000 to 1 in 100,000<br>• More common in Ashkenazi Jewish population<br>• Enzyme replacement therapy (ERT) was first used to treat GD.<br>• Screening<br>o First tier screen: Glucocerebrosidase enzyme activity on DBS by MS/MS<br>o Optional second tier screen<br>o Confirmatory diagnosis<br>• Treatment<br>o Enzyme Replacement Therapy<br>o Substrate Reduction Therapy<br>• Why should GD be added?<br>o Detection of GD1 and GD3 during an asymptomatic period<br>o Reliable screening and diagnosis |

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| | | | | ○ Approved treatments are available and accessible<br><br>Sheldon Garrison, PhD Rogers Behavioral Health<br>• GD is a multi-system disease that leads to a spectrum of disease severity<br>• Lengthy diagnostic delay<br>• 2019 Healthcare Cost and Utilization study<br>○ Annually 380 inpatient discharges<br>○ Annually 473 emergency department discharges<br>• States currently screening for GD<br>○ Missouri<br>○ Illinois<br>○ Tennessee<br>○ New Jersey<br>○ New York (pilot)<br>○ Oregon<br>• Currently no states are doing second tier testing.<br>• Availability of second tier testing<br>○ Mayo Clinic offers second tier testing. No states have been using this service yet.<br>○ Right now, the states screening have different call out rates. New Jersey is the highest and Oregon is the lowest.<br>• Is there information on if the biomarker can be used for phenotype correlation?<br>○ No direct phenotype/genotype correlation or biomarker /phenotype correlation at this time.<br>○ Second tier may miss some homozygous (adult onset/late onset) cases.<br>○ Mayo is going to be working with Oregon (blind sample exchange) to further develop second tier testing.<br>• After 8 years of screening in Illinois:<br>○ 21 screened positive<br>○ 8 were recommended for treatment. 5/8 of these patients were 3 years or younger at the time of treatment.<br>▪ Type 1: 5 cases<br>▪ Type 2: 2 cases<br>▪ Type 3: 1 case<br>• Type 3 neurologic outcome |

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| | | | | ○ ERT does not have a proven direct effect on the neurologic outcome. Still learning more about whether this can stabilize the neurologic side of the disease as well.<br>• Additional questions to consider:<br>○ What proportion of those needing treatment will be children?<br>○ How is timing of treatment determined?<br>• Is Gaucher in consideration for the recommended uniform screening panel (RUSP)?<br>○ Submitted in late 2024. Was on the list for review Feb 2025, but this meeting never occurred. Currently, no process to move forward on a federal level.<br>○ The American College of Medical Genetics and Genomics (ACMG) is moving forward with setting up a coalition on an interim basis to provide a substitute for the ACHDNC that maintained the RUSP and evaluate conditions.<br>• Can you be identified with Type 1 in newborn screening and not show symptoms until adulthood? Yes.<br>○ Standard of care is when biomarkers rise, treat before symptoms appear.<br>• Cost is around $2-$3 per infant to add this screening onto the Wisconsin panel, without considering cost for second tier testing. |
| Decision | 11:10-11:30 | Review and Vote on Nine Criteria (Dr. Basel/Tami Horzewski/Voting Members) | • Dr. Baker will pull together information on cost and logistics for GD at the WSLH.<br>○ Potential to do DNA testing on children with specific variants that can cause false negatives. | Voting<br>• Criterion 1<br>○ Meets: 10<br>○ Does not meet: 0<br>○ More info needed: 1<br>• Criterion 2<br>○ Meets: 10<br>○ Does not meet:<br>○ More info needed: 1 (Genotype/Phenotype)<br>• Criterion 3<br>○ Meets: 11<br>○ Does not meet: 0<br>○ More info needed: 0 (Type 2 same level of diagnosis)<br>• Criterion 4<br>○ Meets: 10<br>○ Does not meet: 0 |

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| | | | | ○ More info needed: 1<br>• Criterion 5<br>○ Meets: 11<br>○ Does not meet: 0<br>○ More info needed: 0<br>• Criterion 6<br>○ Meets: 3<br>○ Does not meet: 1<br>○ More info needed: 7 (2nd tier)<br>• Criterion 7: N/A<br>• Criterion 8<br>○ Meets: 11<br>○ Does not meet: 0<br>○ More info needed: 0<br>• Criterion 9<br>○ Meets: 0<br>○ Does not meet: 0<br>○ More info needed: 11<br><br>• More information is needed on Criterion 6 and Criterion 9.<br>• Motion to move Gaucher to Umbrella Committee: Dr. Rhead<br>○ No second<br>• Motion to recommend addition of GD to the WI NBS panel with the condition of obtaining more information on 2nd tier screening regarding turnaround time and cost: Dr. Rhead<br>○ Second: Dr. Baker<br>○ 10 voted yes<br>○ 1 abstained<br>○ Motion passed |
| | 11:30-<br>11:40 | Break | | (Skipped) |
| Info | 11:40-<br>11:50 | Department of Health Services (DHS) Update<br>(Dr. Julie Thiel/Dr. Bob | | Updates from Department of Health Services:<br><br>• DHS has moved location. New address is 201 E. Washington Ave.<br>• XALD and MPS1 added 8/1/25 by rule, as well as change in blood card fee. |

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| | | Steiner/Tami Horzewski/Leah Ricci) | | • GAMT was approved by the Secretary.<br>• MPS II and Infantile Krabbe are being recommended to the Secretary to be added to the Wisconsin newborn screening conditions panel. If approved by the Secretary, it will move through rulemaking process.<br>• ACHDNC was a federal committee that advised on newborn screening conditions. They set the RUSP. The ACHDNC was disbanded earlier this year. ACMG has offered to take over some of the duties of the ACHDNC in the interim.<br>• WI legislature is reviewing a bill that WI should be aligned with the RUSP. There is flexibility in the wording of that bill. Not sure where/when it will move forward.<br>• Research workgroup has completed its work and has made a recommendation to the Secretary. The report is currently under review. More will be shared once the report is approved.<br>• NBS Publications – four new 1-pagers: one discussing all 3 parts of NBS, 1 for hearing screening, 1 for heart screening, and 1 for blood screening. The work group created a Plain community brochure as well. Also have postcards for birth units/midwives to use with families about following up with audiology on a refer hearing screening result. NBS swag was also ordered to table at events when sharing these new publications with the public. |
| Info | 11:50–12:00 | WI State Lab of Hygiene (WSLH) Update (Dr. Baker) | | No WSLH updates.<br><br>Changes to Metabolic Services (Nikki Drilias & Tammi Timmler)<br>• Discontinued PKU program at Marshfield Medical Center. Will continue to follow a few other metabolic patients. Lab folks are moving formula over to Beaver Dam, closest site to Madison. Mary Marcus Walters will be able to pick it up for clinic in Madison. PKU patients at Marshfield are transitioned to Madison or Milwaukee or Mayo Clinics.<br>• 9 PKU patients transferred; 7 have established care and remaining 2 have appts next week.<br><br>Update from Amy White:<br>• Mayo Clinic is screening for Infantile Krabbe disease (psychosine 2.0 and up) and MPS II for babies born at the two facilities in Wisconsin, because they are already on Minnesota's NBS program. Have |

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| | | | | guidance documents already that Mayo Clinic could share to help other hospitals understand protocols when Krabbe disease is added to the Wisconsin blood screening panel. |
| Info | 12:00 – 12:25 | Metabolic Screening Summary (Dr. Baker) | | 2024 Report from WSLH<br><br>Aminoacidopathies and Urea Cycle Disorders<br>• PKU – 5 reported, 5 confirmed<br>• Tyrosinemia Type II/III – 3 reported, 0 confirmed<br>• Homocystinuria – 4 reported, 1 confirmed<br>• ASA – 1 reported, 0 confirmed<br>• MSUD – 3 reported, 0 confirmed<br><br>Fatty Oxidation Disorders<br>• VLCADD – 3 reported, 1 confirmed<br>• CUD – 2 reported, 1 confirmed<br>• MCADD – 2 reported, 2 confirmed<br>• DE RED – 1 reported, 0 confirmed (infant deceased)<br>• M/SCHAD – 1 reported, 0 confirmed<br><br>Organic Acidurias<br>• Propionic acidemia – 4 reported, 3 confirmed<br>• Methylmalonic acidemia – 2 reported, 0 confirmed<br>• MMA/HCY – 6 reported (one maternal case), 2 confirmed (Cbl C)<br>• Isovaleric acidemia – 1 reported, 1 confirmed<br>• SBCADD – 1 reported<br>• 3-MMC/MCD/3MGA – 12 reported, 1 confirmed<br>• Glutaric acidemia type 1 – 6 reported, 1 confirmed<br><br>Other Disorders<br>• Biotinidase Deficiency – 1 reported, 1 confirmed<br>• Pompe – 6 reported, 5 confirmed, mainly Late Onset type<br>• X-ALD – 13 reported, 9 confirmed (2 false positives)<br><br>Diet Monitoring<br>• 2024<br>  ○ 2,061 total specimens<br>  ○ 259 individuals |

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| | | | | ○ 7 specimens rejected |
| Discussion | 12:25 - 12:30 | Plan Next Meeting/Agenda Items | | Meet in Spring 2026 again. Tami will send out a poll. |

**Next meeting date: Spring 2026**

**"Parking Lot" Items:**