NBS Advisory Committee Meeting
MINUTES

September 30, 2025
12:00 p.m. – 2:00 p.m.

Attendees

Voting Advisory Committee Members Present:

- **Jennifer Banna**, Committee Vice Chair - Center Coordinator, Family to Family; Parent of child with rare metabolic disorder; Representative of an advocacy association regarding newborns with medical conditions or rare disorders
- **Steve Shapero** - Family member of persons affected by a rare genetic disorder
- **Shawnalea Chief Goes Out** - Representative of the Medicaid insurance industry, Perinatal Health Program Officer at the Health Resources Division, DPHHS
- **E Lynne Wood** - Pediatric Neurologist, Billings Clinic
- **Abdallah "Abe" Elias** - Director of Medical Genetics and Clinical Geneticist, Shodair Children's Hospital
- **Shelly Eagen**, Committee Chair - Nurse Practitioner, Pediatric Pulmonary, Billings Clinic
- **Amanda Osborne** - Licensed, Certified Professional Midwife, Helena Birth Studio

Voting Advisory Committee Members Absent:

- **Marion Rudek** - Nurse Practitioner, Blackfeet Community Hospital, Representative of a tribal health care system
- **Kotie Dunmire** - High School Business and Special Education Teacher, Butte High School and Parent of child with Cystic Fibrosis and PKU

Non-Voting Advisory Committee Members Present:

- **Jeanne Lee** - Newborn Screening and Serology Supervisor, DPHHS
- **Douglas Harrington** - State Medical Officer, DPHHS
- **Chelsea Pugh** - Nurse Consultant, Newborn Screening, DPHHS
- **Miranda Reddig** - Program Specialist, Newborn Screening, DPHHS
- **Amber Bell** - Newborn Screening Coordinator, Children's Special Health Services, DPHHS
- **Nikki Goosen** - Newborn Screening Clinical Laboratory Science Lead, DPHHS

Non-Voting Advisory Committee Members Absent:

- **Jacqueline Isaly** - Family and Community Health Bureau Chief, DPHHS
- **Debbie Gibson** - Lab Services Bureau Chief, Montana Public Health Laboratory, DPHHS

Facilitators:

- **Mikaela Miller** - Public Health Specialist, Yarrow
- **Stephanie Burkholder** - Public Health Specialist, Yarrow

Public:

- **Justin Hopkin, MD** - Chief, Division of Hospital Medicine Strong Memorial Hospital School of Medicine and Dentistry
- **Kathy Crowley-Haywood**, CNM, Roots Birth (Missoula)
- **Josh Baker, MD,** Metabolic Geneticist, Chicago, IL

Welcome & Roll Call

- Chair Shelly Eagen, welcomed the group and did roll call while leading introductions so each person could introduce themselves by providing their organizations, roles, and a description of themselves.
    - Note: physical description is requested during introductions for those that might be seeing impaired.
- Co-chair, Jennifer Banna, read the acknowledgement.
- Yarrow provided an overview of the Agenda, Ground Rules, and the Public Comment Period.
- Seven of the nine voting members are present at the meeting; thus, a quorum was established.

Acid Sphingomyelinase Deficiency (ASMD) Nomination Packet Review

The Montana Newborn Screening Committee met to review a nomination packet for adding Acid Sphingomyelinase Deficiency (ASMD) to the state's newborn screening panel. The committee heard presentations on ASMD's two types, their symptoms, and treatment options, including enzyme replacement therapy. They discussed the condition's screening costs, which would add approximately $3 per sample. The current panel cost is $150.50 but with the addition of Pompe, the new panel cost will be $161.80 by January 2026. Thus, by adding ASMD, the new total panel cost will be $164.80. The committee will vote on whether to recommend adding ASMD to the screening panel, followed by a discussion of bylaws amendments and a public comment period.

Please note that this is a broad overview drawn from the nomination packet, but further details were provided by the Subject Matter Expert (SME) and family presenter at the previous (5/21/25) meeting.

- Two main types - A and B: The signs and symptoms between the two types are highly variable.
    - Type A:
        - Age of onset: early infancy

        - Signs/symptoms: Enlarged liver and/or spleen, accumulation of fluid in the abdomen, jaundice, feeding difficulties, constipation, nausea, vomiting, significant gastrointestinal reflux, failure to thrive, irritability, loss of reflexes, and progressive loss of muscle tone (hypotonia), and respiratory issues.
    - Type B:
        - Age of onset: infancy to adulthood
        - Signs/symptoms: Similar to Type A but not as severe. Enlarged liver and/or spleen, increased infections, prolonged bleeding, abdominal pain, liver disease, respiratory issues, neurological issues, delayed growth/puberty, and bone thinning.
- How is this disorder currently identified?
    - Symptomatic presentation followed by a blood test
- Why should it be screened at birth?
    - Early detection and management can help mitigate some of these serious health risks and improve quality of life
- How is this disorder treated?
    - Is there a treatment available?
        - Yes - FDA approved enzyme replacement therapy (ERT)
    - Is the treatment in the experimental phase?
        - No
- Proposed screening test method
    - Dried blood spot
- Status of the condition in the United States:
    - States currently screening for the condition: 2 (Illinois & New Jersey)
    - Condition has been reviewed by RUSP: Yes
    - Registries or databases currently established for the condition: 2

Selection Criteria:

1. It can be identified at a period of time (24 to 48 hours after birth) at which it would not ordinarily be clinically detected. - True
2. A test with appropriate sensitivity and specificity is available. - True
3. There is a significant risk of illness, disability, or death if babies are not treated promptly (within the recommended time frame for the condition). - True
4. Effective treatment is available and access to follow-up care and counseling is generally available. - True
5. There are demonstrated benefits of early detection, timely intervention, and efficacious treatment. - True
6. The benefits to babies and to society outweigh the risks and burdens of screening and treatment. - True
7. There are minimal financial impacts on the family. - True
8. There is a public health benefit to conducting the test. - True
9. There exist responsible parties who will follow up with families and implement necessary interventions. - True
10. The condition’s case definition and spectrum are well described. - True

Updated cost analysis

- ASMD can be multi-plexed with Pompe disease (both tests can be assayed from the same sample), adding ASMD would increase the NBS test cost by approximately $3.00 per sample.
- Note: When Pompe was initially approved, the test cost was around $11.00, which will increase to $11.30 when testing theoretically begins in January.
    - Note from Jeanne: Current cost of newborn screening test is $150.50 per test.

ASMD Packet Discussion

Chair Shelly Eagan opened it up for comments/questions on the selection criteria. The group discussed the implementation of ASMD screening, noting that since it can be run on the same blood spot sample as Pompe disease, there is no concern about “real estate” (space) issues on newborn screening cards. DPHHS has ordered new cards and they can add some additional circles to the filter paper so they will soon (several months from now) be collecting 8 drops of blood instead of 5. Jeanne explained that Wisconsin does not currently screen for ASMD on their panel but they have a nomination to their panel to add it to their testing. Additional confirmatory testing processes would need to be developed. Dr. Elias and Shelly inquired about the process of determining second-tier processing for ASMD, and Jeanne acknowledged the need for further investigation on potential testing options, possibly other states or Mayo Clinic. The group agreed to consult with Dr. Justin Hopkin and Dr. Josh Baker are present at this meeting and the group agreed to consult with them on secondary testing. Dr. Justin Hopkin noted that there is a global clinical guideline statement on this. For positive tests, they run lysosphingomyelin and send it to Mayo Clinic, then do genetic sequencing and those are the two confirmatory tests. They have not had any false positives yet and have had 11 or so positives.

The group then discussed costs, testing procedures, and implications. They clarified that confirmatory testing would likely be covered by the patient's insurance or Medicaid, and Jenn recalled that false positives have not been an issue in Illinois.

Shelly inquired if enzyme replacement therapy (ERT) is accessible to families in Montana through infusion centers? Dr. Elias confirmed that, yes, they have other patients who get ERT here in Montana; it does take some coordination and might take a bit more time to organize but there is not the urgency that there is with Pompe, for example. Dr. Elias raised questions about the effectiveness of ERT and the challenges of predicting disease onset based on newborn screening results. ERT modifies the visceral manifestations and not the neurological, which can be beneficial. But the screening, alone, is difficult in predicting the phenotype and future treatment plan. The group also considered the limited experience with screening for this condition and debated the benefits of early screening/diagnosis versus clinical diagnosis later if a child presents with classic symptoms like hepatosplenomegaly, hypotonia, etc. Considerations around clinical progression between when they could have been screened/diagnosed with NBS versus being diagnosed later in infancy or childhood.

The group discussed the diagnosis and treatment of a rare genetic disorder, focusing on ERT and its limitations in reversing irreversible organ damage. Dr. Josh Baker shared unpublished data from Illinois, highlighting that while ERT effectively treats visceral manifestations, it cannot reverse damage in some cases, particularly in severe phenotypes. It is different from Gaucher where you can give ERT and it may result in damage reversal. The discussion also touched on the challenges of early diagnosis and the potential use of biomarkers like lysosphingomyelin to identify severe phenotypes earlier. ERT treats the visceral manifestations, not so much the neurological. Dr. Wood noted that even though ERT can help with hepatic and pulmonary complications it is still a major cause of morbidity and mortality, especially in kids under age 18 who do have this disease. Dr. Baker noted that those kids with hepatosplenomegaly (even who are not classic Type A) who receive treatment early on might have better quality of life and live longer because they aren't succumbing to the disease as quickly when we treat the organs.

Amanda raised a question about the inclusion of this disorder in newborn screening. Why would we not include this and why have only two states included it so far? Dr. Hopkin explained this is most likely influenced by the availability of effective therapy and other state-based programs. ERT was only approved in 2022 so it's a relatively new treatment. This has been nominated for the RUSP (almost 15 years ago) and it was shot down at the time because it did not have an effective therapy like it does now. Other states also like to see an effective state-based program, which there are not many (other than Illinois) so once HRT came about and Dr. Baker published his paper it opened up more avenues for the advocacy groups to go to the states to ask for this to be put on the screening panel. Dr. Elias assumes in the next few years that other states will adopt it, especially those with larger populations which will provide more data too. A question to ask ourselves is: Do we want to pioneer that and be one of the early adopters or wait and see what other states do?

Dr. Harrington supported adding the disorder, citing its relatively low cost and potential benefits. The committee considered the feasibility of implementation and follow-up, noting that while there are uncertainties, the condition is relatively straightforward to manage. They agreed that the disorder fits with other conditions previously approved for screening, and Dr. Elias expressed confidence that approving this would not significantly impact resources for future applications.

The committee discussed the financial implications of recommending additional programs, with Jennifer raising questions about funding limitations. They reviewed the status of newborn screening for ASMD in Montana. The group confirmed that while there are local resources available for ASMD care in Montana, some patients may need to travel for specialized treatment, though telemedicine and local outreach clinics could be sufficient for certain consultations or to manage/coordinate the therapies. Dr. Elias highlighted the growing challenge of organizing care for rare disorders, particularly as more patients are diagnosed earlier through newborn screening, and expressed concerns about future healthcare funding for children's care in general.

Dr. Hopkin explained that while there is no approved treatment for severe neurological symptoms, there are experimental therapies in development in various stages. Neurological

symptoms are rare but it's important to remember that these symptoms appear on a spectrum, and it can be difficult to tell early on in life. The infusions can often be provided in-home after a period of receiving them in a clinic setting. The committee also considered the cost implications, with Jeanne noting that adding ASMD would increase the screening fee to $164.80 per screen.

Committee Vote on ASMD

- Seven of the nine voting members are present today (quorum was met):
    - Jenn Banna
    - Shelly Eagen
    - Abe Elias
    - Amanda Osborne
    - Steven Shapero
    - E Lynne Wood
    - Shawnalea Chief Goes Out

- Explanation of voting options:
    - Voting members were asked *"Do you recommend including ASMD / Niemann-Pick disease on the Montana Newborn Screening Panel?"* with the following voting options:

        - *Yes, recommend*
        - *No, do not recommend*
        - *I do not have enough information to make a decision at this time*

- The vote was held on whether to recommend the addition of ASMD to the Montana Newborn Screening panel.
    - Result of vote:
        - **7 members: *Yes, recommend***
        - 0 members: *No, do not recommend*
        - 0 members: *I do not have enough information to make a decision at this time*
    - Mikaela announced the results: With a majority in favor, the proposal of adding ASMD to the MT NBS panel was recommended by the committee. Thank you all for your participation. Your input helps guide our direction and ensures transparency in our decision-making process.

Bylaws Amendments

- Chair Shelly Eagen acknowledged that this is Amanda Osborne's last meeting and thanked her for her time and commitment to the committee over the past several years.
- Mikaela indicated we have found a replacement for Amanda's position and that she attended as a member of the public today.
- The NBS Advisory Committee bylaws were emailed to all voting and non-voting members for review on Aug 21. Members were asked to review the bylaws and complete a form indicating their review.
- The form included a free-text space for members to propose an amendment(s) to the bylaws.

    - Reminder emails were sent over the next several weeks.
    - NO amendments to the bylaws were proposed by the time of this meeting.
- The group also discussed potential bylaws amendments, including the possibility of adding an abstention option for votes. Dr. Elias wonders about having another voting option: One to abstain from voting. You're forced to make a decision but (regardless of a conflict of interest) maybe not everyone is ready to vote or wants to vote. The concern would be that there would be a quorum but no decision. Perhaps needing an option that leans more towards "I do have enough information I'm just not ready to cast a vote and bring it back to us again in a year."
    - This was not suggested prior to the meeting. Some discussion ensued and it was agreed that members would think about this more and it would be brought up for further discussion at the next committee meeting. Mikaela asked Dr. Elias and/or Shawnalea to put something in writing that we can use moving forward.

Public Comment Period

- Additional comments via email were accepted up to 3:40 pm MT on September 30, 2025.
    - No additional comments were received.

Newborn Screening Advisory Committee Next Steps

- Follow up email will be sent soon and will include:
    - Meeting minutes
    - Recording
    - Presentation slides
- A doodle poll will be sent out to Committee members to schedule the next meeting.
    - The next meeting will occur in the spring 2026.
- Please email if you have questions, comments, or need anything.

This meeting was concluded by Mikaela Miller at 2:40 pm MT on September 30, 2025.