MT Newborn Screening (NBS) Advisory Committee Meeting
MINUTES
Monday, July 27, 2026
10:00 p.m. – 11:30 p.m.

Attendees

Voting Advisory Committee Members <u>Present</u> (Name - Position)

- **Abdallah "Abe" Elias** - Director of Medical Genetics and Clinical Geneticist, Shodair Children's Hospital
- **Shelly Eagen**, Committee Chair - Nurse Practitioner, Pediatric Pulmonary, Billings Clinic
- **Steven Shapero** - Family member of persons affected by a rare genetic disorder
- **Bridget Evans** - Representative of the Medicaid insurance industry, Health Resources Division, DPHHS
- **E Lynne Wood** - Pediatric Neurologist, Billings Clinic

Voting Advisory Committee Members <u>Absent</u> (Name - Position)

- **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
- **Rose LaPine** - Nurse Practitioner, Blackfeet Tribal Public Health
- **Kotie Dunmire** - High School Business and Special Education Teacher, Butte High School and Parent of child with Cystic Fibrosis and PKU
- **Kathy Crowley-Haywood,** Certified Nurse Midwife, Roots Birth & Women's Health

Non-Voting Advisory Committee Members <u>Present</u> (Name - Position)

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

Non-Voting Advisory Committee Members <u>Absent</u> (Name - Position)

- **Debbie Gibson** - Lab Services Bureau Chief, Montana Public Health Laboratory, DPHHS
- **Kayla Cummins** - Administrative Assistant ECFSD, DPHHS

Facilitators (Name - Position)

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

Guests (Name - Position)

- None

Public (Name)

- None

Welcome & Roll Call

- Committee 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.
- Shelly Eagen read the acknowledgement: “We thank the families, caregivers, committee members, and advocates for their contributions to the Montana Newborn Screening Program. We recognize that each condition reviewed affects children and families in Montana, and we strive to balance the emotions and vulnerabilities shared with the need for careful, sometimes difficult, discussions on logistics and finances. Our goal is to ensure the process is publicly accessible, transparent, and carefully examined.”
- Yarrow provided an overview of the Agenda, Ground Rules, and the Public Comment Period. The agenda for the day was outlined, including lab updates, metachromatic leukodystrophy (MLD) discussion time, and a public comment period. The format for the public comment period was explained, with a 2-minute time limit per speaker and instructions for raising hands or using *9 to participate.

Lab Updates

- Jeanne Lee provided an update on Mei Baker's development of an MLD assay, which is expected to be ready by the end of this year. Mei is exploring the possibility of multiplexing the assay with X-ALD, which could simplify testing by using a single blood spot. If MLD is added, the current

pricing is estimated to increase by $3.00 per screen, though this could be lower if multiplexing is implemented.

MLD Discussion

- Dr. Elias asked if the development by Mei Baker includes both the first tier (sulfatides detection) and second tier (enzyme detection). He mentioned several other states are working on development of a test. Most labs that are developing are working on the second tier to increase specificity.
    - Jeanne does not have the answer to this question. She assumes it is just the first tier right now but can ask Mei Baker if she has plans for the second tier. Mei Baker was invited to join the call today but she was unable to attend. Jeanne will plan to get clarification on this and possibly invite her to our next meeting in the fall.
    - Dr. Elias noted that many labs are still working on developing both first and second-tier assays due to the complexity of sulfatide analysis and the need to reduce false positives.
- Bridget Evans asked about the Administrative Rules of Montana (ARM) - if testing for MLD is ready by the end of the year, do we have to wait to begin screening newborns?
    - Jeanne explained that due to the administrative rule process and legislative session constraints, if the committee votes to add MLD, screening would likely begin by the end of 2027 or early January 2028. Jeanne noted that the ASMD screening process is currently facing similar delays, with implementation scheduled for January 2027.
- Dr. Wood asked to clarify the process after screening. If a child screens positive, the result will go to Shodair and then they'll get referred out to get a brain MRI?
    - Dr. Elias explained that initial screening includes mass spectrometry for sulfatides, followed by confirmatory testing including ARSA enzyme activity, urine sulfatides, and molecular testing. Once it's confirmed (ideally within 2 weeks) then they would do the referral to neurology with MRI. He reminded the group that New York has a universal pilot right now. Although it's a test that can be multi-plexed, the sulfatides are finicky, so we need to think about the development but also validation (testing on a sufficient number of people to confirm) so he is curious about whether this will actually take longer than the end of the year.
- Mikaela shared a question that was submitted ahead of the meeting: What treatment centers are available in Montana for MLD, and if there are none, where is the closest treatment center?
    - Nikki: Montana does not have a qualified treatment center. There are 8 total centers, but Primary Children's Hospital in SLC is the closest. Minnesota and San Francisco are other close options. The Lenmeldy website has a map of treatment centers here: https://www.lenmeldy.com/treatment-centers/
- Jeanne wonders if it is difficult for children to get an MRI?
    - Dr. Wood clarified that while MRI scans can be performed locally, SPECT scanning may have limited interpretation capabilities, and suggested researching available scanners and labs for follow-up testing.

- Bridget raised a question about providers being educated not to refer patients for MRI when certain gene therapy treatments are not appropriate, which Dr. Wood clarified would be based on MRI **findings** rather than the presence of an MRI itself.
- Dr. Elias: Might be good to invite someone, such as Dr. Bonkowsky from University of Utah, to discuss the treatment process. He provided an explanation of the ex vivo therapy process, which involves using bone marrow, ablating certain cells, and introducing a functional copy of an enzyme using a lentivirus-like vector.
- Dr. Wood feels we have gathered the information that we wanted to know for this interim meeting.
- Dr. Harrington: Most labs he is aware of are doing a 2 or 3 tiered blood spot test (first sulfatides, second ARSA enzyme, third tier genetic confirmation from sequencing) and this is all done from the one blood spot. That system has 99-100% sensitivity and specificity, so it's really good.
    - From prior research, 50-60% of kids with late infantile onset need to be treated with the FDA-approved product before symptoms develop to have a good outcome.
- Nikki shared a reminder that they did recently increase blood spots from 5 spots to 8 spots, so we have more blood to work with.

Public Comment Period

- No members of the public attended this meeting.

Thanks and Next Steps

- Follow up email will be sent soon and will include:
    - Meeting minutes
    - Recording
    - Presentation slides
- Please remember to fill out the scheduling poll so that we can confirm a date for the next meeting.
    - The next meeting will occur in Fall 2026.
- Please email if you have questions, comments, or need anything.

This meeting was concluded by Mikaela Miller at 10:46am on July 27, 2026.