



NBS Advisory Committee Meeting MINUTES

Wednesday, October 25, 2023
9:00 a.m. – 1:00 p.m.

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Attendees

Voting Advisory Committee Members Present	
Name	Position
Abdallah "Abe" Elias	Director of Medical Genetics and Clinical Geneticist, Shodair Children's Hospital (In Person)
Jennifer Banna, Vice Chair	Center Coordinator, Family to Family, Parent of child with rare metabolic disorder (Virtual)
Shelly Eagen, Chair	Nurse Practitioner, Pediatric Pulmonary, Billings Clinic (In Person)
Kotie Dunmire	High School Business and Special Ed Teacher, Butte High School Parent of child with Cystic Fibrosis and PKU (Virtual)
Miranda McCabe	EPSDT Program Specialist, DPHHS (Virtual)
Amanda Osborne	Licensed, Certified Professional Midwife, Helena Birth Studio (Virtual)
Allison Young	Pediatrician, Western Montana Clinic (Virtual)

Voting Advisory Committee Members Absent	
Name	Position
Sarah Sullivan	RN, Parent to two children with homocystinuria
Marion Rudek	Nurse Practitioner, Blackfeet Community Hospital

Non-Voting Advisory Committee Members	
Name	Position
Amber Bell	Newborn Screening Coordinator, Children's Special Health Services, DPHHS (In Person)
Miranda Reddig	Program Specialist, Newborn Screening, DPHHS (In Person)
Chelsea Pugh	Nurse Consultant, Newborn Screening, DPHHS (Virtual)
Debbie Gibson	Lab Services Bureau Chief, Montana Public Health Laboratory, DPHHS (In Person)
Jeanne Lee	Newborn Screening and Serology Supervisor, DPHHS (In Person)
Nikki Goosen	Newborn Screening Clinical Laboratory Science Lead, DPHHS (In Person)

Jacqueline Isaly	Family and Community Health Bureau Chief, DPHHS (In Person)
Margaret Cook-Shimanek	Acting State Medical Officer, DPHHS (Virtual)
Dani Lindeman	Laboratory System Improvement Manager, DPHHS (Virtual)

Facilitators	
Name	Position
Anna Schmitt	Co-founder, Yarrow (In Person)
Mikaela Miller	Public Health Specialist, Yarrow (Virtual)

Guests	
Name	Position
Anna Grantham	Hunter's Hope Foundation (Virtual)
Joanna Kurtzberg	Pediatric Transplant Physician, Duke University School of Medicine (Virtual)
Dieter Matern	Co-Director of Genetics Lab, Mayo Clinic (Virtual)

Public	
Name	Position
Miranda McAuliffe	ALD Foundation (Virtual)
Steve Shapero	MT Advocate, Family Member, Sponsor of Nomination Packet (Virtual)
Bruce Kirbo, Jr.	Attorney (Virtual)
Lesla Brackbill	Advocate for Newborn Screening for Krabbe (Virtual)

Welcome & Roll Call

(Yarrow Facilitators, Voting & Non Voting Committee Members, Ground Rules)

- Yarrow 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.
- Yarrow then led an Icebreaker by asking the participants “What is bringing you joy lately?”
- Yarrow provided an overview of the Agenda, Ground Rules, and the Public Comment Period to happen at the end of the meeting.
- Quorum was established.
 - All Conflict of Interest forms for X-ALD have been received.

Unfinished Business Review

- **Internal Committee Updates**
 - Updated Condition Nomination Form
 - This form was previously a static PDF. This format made it difficult to submit responses. It is proposed that we change the format to a fillable PDF and other changes to make the document more comprehensive and easier to follow.
 - AElias requests that instructions are added.
 - The committee agreed this would be helpful, along with adding a previously received form as a sample to follow.
 - Updated Bylaws and Vote
 - Absentee Voting - The committee discussed whether the following language may be added to the bylaws:

Quorum

A majority of the committee members constitutes a quorum to do business. *The quorum should be based on attendance at meetings in person or by proxy.* See Mont. Code Ann. § 2-15-124.

Voting members may vote upon a condition or any other voting matter, in absentia, if:

1. *Member is ill or has an emergency that otherwise prevents meeting attendance;*
2. *The voting in absentia member feels they are well enough informed to make a vote;*
3. *Votes on conditions must still be presented at an Advisory Committee meeting;*
4. *If voting by email, a form will be sent to members not present in order to collect their vote;*
5. *If voting by proxy, absentee members can assign another voting member the power to vote in their stead. Formal documentation of proxy must be presented to the committee by the absent member prior*

to the meeting where a vote is held.

6. *Votes in absentia will count towards a quorum.*

- Voting members were asked whether they approve the language in the update to the bylaws with the following voting options:
 - *Yes, approve*
 - *No, do not approve*
 - *I do not have enough information to make a decision at this time*
- Vote Count
 - Jenn Banna
 - Shelly Eagen
 - Allison Young
 - Abdallah Elias
 - Miranda McCabe
 - Amanda Osborne

All 6 of the committee members present at the time of vote voted: “Yes, approve” to the bylaw updates. This passed with a quorum.

- Additional Members Determination
 - At the last meeting it was suggested that we may want to have a larger number of Committee voting members to more easily enable a quorum.
 - After reviewing the legislation, it was deemed that it is not possible to add more members at this time due to the current language within the legislation.
 - AYoung requested clarification on the legislation criteria.
 - It was responded that the legislation is written very specifically on the 9 people and their role types. To change this, the bill would have to be rewritten.
 - The background on these 9 are from the way the bill was originally written, to include those with various backgrounds.
 - Other professionals in the field have expressed interest in joining the committee. They can join as members of the public, or become members as current member terms end.
 - Voting members are on staggered terms to allow opportunity for new members to join the committee to allow for more diversity of expertise.
- External Communications
 - The committee also discussed the topic surrounding communications after an advocate reached out to voting committee members after receiving permission to do so. Going forward, the committee members would like their emails hidden from calendar invites.
 - In the future, all communications will be directed to the general NBS email (HHSNewbornAdvisoryCommittee@mt.gov)
 - This will ensure that all communications may be filtered and tracked through the general NBS email due to the public nature of this committee.

Newborn Screening Advisory Committee Vote on x-ALD

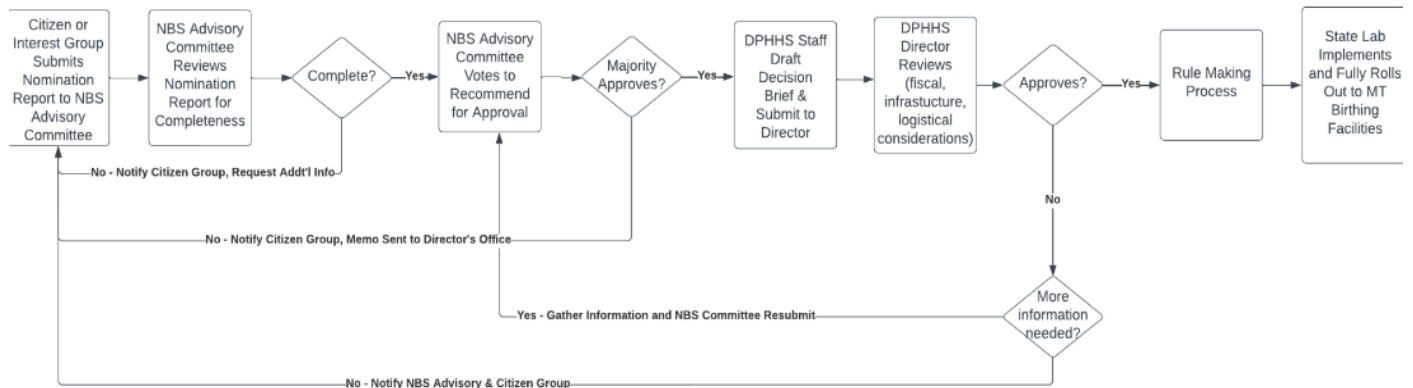
- Voting members were asked “Do you recommend including x-ALD on the Montana Newborn Screening Panel?” with the following voting options:
 - Yes, recommend
 - No, do not recommend
 - I do not have enough information to make a decision at this time
- Vote Count
 - Jenn Banna
 - Shelly Eagen
 - Allison Young
 - Abdallah Elias
 - Miranda McCabe
 - Amanda Osborne

All 6 of the committee members present at the time of vote voted: “Yes, recommend” to include x-ALD on the Montana Newborn Screening Panel. This passed with a quorum.

Condition Nomination Process Review

1. Nomination packet is sent to the NBS joint email at HHSNewbornAdvisoryCommittee@mt.gov.
 - Within 48 hours the sender will be notified that the packet was received.
2. CSHS & Lab (and potentially Chair and Vice Chair) decide if the nomination packet is complete. Additional information may be requested.
 - The sender will be notified whether the packet was complete or incomplete within 2 weeks.
3. The completed nomination packet will then get sent to the full Advisory Committee for review.
 - The nominated condition will be put on the agenda for the next Advisory Committee meeting at least 1 month prior to the meeting where it will be reviewed.
4. The designated person (or Chair) leads the Advisory Committee through the nomination packet during the meeting. Additional information will be presented from SME, Lab, and Family Story as appropriate.
 - A vote on the nominated condition will occur during a Committee meeting once the process is complete. This may occur within a varied number of meetings depending on the number of conditions that are already in the queue to be reviewed.
5. Lastly, a vote will be held for the nominated condition at a Committee Meeting.
 - Within 1 week a report will be sent to the DPHHS director for review.

Nomination Flow Chart



Krabbe Presentation and Background Information

- Joanna Kurtzberg with Duke University, Dieter Matern with Mayo clinic, and Anna Grantham with Hunter's Hope foundation were requested by the Lieutenant Governor's office to present at this committee meeting.
- This does not follow our standard nomination process, however, we intend to follow this process if Krabbe is to be reconsidered.
- When Krabbe was last considered by this committee in December 2022, it resulted in a vote to not add the condition, which was agreed upon by the Governor. This occurred prior to the condition being reconsidered by RUSP in February 2023.
- Dr. Joanna Kurtzberg, Pediatric Transplant Physician with Duke University, and Dr. Dieter Matern, co-director of the biochemical genetics lab at Mayo Clinic, provided two presentations on Krabbe.
- Presentation slides are attached.

Krabbe Discussion

- AElia: where are the RUSP recommendations currently?
 - JKurtzberg's response: At their last meeting they decided to proceed with the accelerated pathway for reconsideration of the disease within 9 months, which means it will likely be voted on within the first quarter of 2024.
 - This reconsideration is limited to the early infantile or infantile form of the disease.
 - DMatern states that the difficulty getting Krabbe approved by RUSP is because the committee focuses only on previously published evidence. Until they reviewed the Kentucky paper, this had caused the RUSP committee to focus concerns on false positives and the past treatment outcomes.

- DMatern states that if you are already doing other RUSP screenings, then the Krabbe screening is not difficult to add.
 - DMatern states that there are additional papers in the works that provide evidence for the outcomes of psychosine testing in various states. These are to help provide awareness of the issues that were previously created when only doing GALC activity and genotype tests and to minimize false positives.
- AElias: What are the outcomes in other centers? Can all of the outcomes nationwide be compiled?
 - JKurtzberg states that the paper she wrote contains data that reflected outcomes from all 4 centers, even though it was only 6 patients.
 - This is difficult since the disease is not screened for in all states and has low incidence/is very rare so the numbers are small.
- AElias: It would make sense to extract the outcomes from all 11 states screening for Krabbe during the ongoing RUSP reconsideration, is this being considered?
 - JKurtzberg and DMatern agree that all the data from all transplanted newborns diagnosed at screening is being tracked from all 11 states.
 - The data from the paper published in 2005, represents 14 babies that were diagnosed in utero due to a family history of another family member having Krabbe disease. These were all transplanted within their first month or so of life.
 - JKurtzberg states that these numbers are also low because Krabbe is only being screened in about 1/3 of the births in the U.S.
 - Families that decide against treatment must also be considered when weighing the amount of data available.
 - Denver, Salt Lake City, or Seattle will likely be the transplant centers used for MT newborns.
- NGoosen: If you get an initial low GALC, and then have a normal psychosine, at that point, you're confident it's negative or would you want to get a second screen to be sure?
 - DMatern states "I'm confident it's negative. And I'm confident it's negative for infantile and late-infantile forms of Krabbe disease. I'm fairly confident it's not juvenile. But, I think one of the things that is also very important is that you're very transparent in terms of making clear what you actually, what your target is. So if you think in RUSP terms, the core condition in my opinion is infantile and late infantile Krabbe disease and the secondary targets are the later onset forms."
 - Infantile related Krabbe disease is prioritized, secondary targets are the later onset forms of the disease.
 - When SMA got on the RUSP, it was for a very specific form of the disease
 - This is similar to how the RUSP is considering proceeding with Krabbe - to not identify ALL cases, but to identify very specific Krabbe cases.
- ABell: We don't screen for 49 conditions right?
 - Correct, we screen for 33.
- ABell: The cost estimate for adding Krabbe screening is \$1-2 if the lab is already screening for other lysosomal disorders, which we do not, so the cost could be more?

- DMatern states that yes, there are 2 companies that sell reagents at different costs.
- Cost varies whether the lab is already testing for other lysosomal disorders, if not, the upfront cost is greater.
- JLee: Is the psychosine a mass spec test?
 - DMatern: Yes, a liquid chromatography and it requires very sensitive equipment.
 - The 4 labs that offer this test are Mayo, Pittsburgh, Greenwood in SC, Nationwide Children's in OH
- AYoung: What was the lab that may potentially be able to be used for MT lysosomal storage disorder tests?
 - JLee: Wisconsin currently performs the Pompe test but they do not have Krabbe.
 - DMatern: They may be awaiting the RUSP position prior to adding Krabbe.
- DMatern states the importance of considering the risks of false positives with broad screening, as opposed to missing those born with Krabbe. This can be mediated by a more specific screening process.
- Thank you to the presenters. End of presentation discussion.

Open Voting Member Discussion

- The committee feels that there seems to be a general lack of awareness about newborn screening.
 - A majority of pediatricians do not know what is included in the NBS panel.
 - The committee discussed that it may be worth looking into educational materials for the public that may be accessible from the website.
 - It was stated that DPHHS has a brochure.
 - This committee is made up of volunteers and there is no additional funding to be provided for resources spent on an awareness and education campaign for NBS.
- We seem to be seeing reactive nominations as opposed to proactive screening based on the conditions that Montana should be adding.
- The committee has the ability to meet more than the minimum requirement for 2 meetings per year, however, there is often difficulty with meeting quorum.
 - The nomination and review process could be more efficient, but it may require reviewing the legislation behind the committee and the number of members allowed.
- Who decides which things are important to test on in Montana, for example a public health entity?
 - There does not seem to be a person or entity in Montana who oversees this.
 - This is where the advisory committee comes in.
 - An entity that may be able to aid in getting the word out is the Montana chapter of the American Academy of Pediatrics.
- Guanidinoacetate methyltransferase deficiency (GAMT) was recently added to the RUSP
 - An email about GAMT was received at the end of September, they were asked to follow the standard nomination process.

- It is part of 3 different bio-deficiency disorders, mostly affecting boys, there is currently no screening or treatment.
- Why did the Advisory Committee go back to Krabbe?
 - The legislation called out Krabbe to be the first condition to be heard, it was reviewed outside of the standard nomination process.
 - The advisory committee made a recommendation not to add.
 - The individuals that presented today were asked to present by the Governor's office.
 - The committee needs to be given the flexibility to review conditions according to the process set in place, in order to prevent inequities in disease nominations.
 - It may be worth communicating that this did not feel productive and this should be voiced to the higher powers that requested this.
 - The committee would like to draft a memo to be sent to leadership on the matter of agenda setting and process following.
 - This should come from the committee as opposed to the contracted facilitator.
 - Yarrow will draft a memo on this based on information provided in the feedback surveys, and send it to the committee for review.
- AYoung: Would like to know more about a Rare Disease Advisory Council for Montana
 - Where do nominations come from if the public is not aware about NBS?
 - Do we need to re-evaluate the nomination process?
 - The nomination process is based off of the process of Wisconsin, Minnesota, and other states.

Newborn Screening Advisory Committee Public Comment Period, & Wrap Up

Public Comment Period

- SShapero: resident in Montana who has been impacted by x-ALD throughout his life, wants to thank the committee for their thoughtful consideration.
- MMcAuliffe: Here on behalf of the ALD Alliance, also the mother of a son who has x-ALD, based on the process in her home state of New York, she would like to make a suggestion for the nomination process, since they are only meeting twice per year, she recommends the committee replies back to the nominator to ask for any updated data for the review process. She would like it to be stated for the record that the Wisconsin lab tests for ALD.
- Anna Grantham, Hunter's Hope: they did not request to attend the meeting, they were asked to so they were unaware of the standard procedure but moving forward, they will use the nomination form.
 - They have a newborn screening council that meets monthly and is open to any state that is considering Krabbe screening.
- Additional comments can be sent to the email up to 1:30 MT on October 25th.

Thanks and Next Steps

- Follow up email will be sent soon and will include:

- Meeting minutes
- Recording
- Transcription
- Presentation slides
- Feedback survey
- The public website will be updated to reflect today's determination.
- A memo will be drafted and sent to the DPHHS Director for review, Director's decision will be posted on the website.
- A doodle poll will be sent out to schedule the next meeting.
 - The next meeting will occur in April.
- Please email if you have questions, comments, or need anything.

This meeting was concluded by Anna Schmitt at 12:40 pm on October 25, 2023.