



**NBS Advisory Committee Meeting
MINUTES**

Tuesday, November 19, 2024
9:00 a.m. – 10:00 a.m.

Attendees	2
Welcome & Roll Call	3
Unfinished Business Review	4
Gaucher Discussion	4
Newborn Screening Advisory Committee Vote on Gaucher	5
Thanks and Next Steps	6

Attendees

Voting Advisory Committee Members Present	
Name	Position
Shelly Eagen, Chair	Nurse Practitioner, Pediatric Pulmonary, Billings Clinic
Jennifer Banna, 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
Kotie Dunmire	High School Business and Special Education Teacher, Butte High School Parent of child with Cystic Fibrosis and PKU
Amanda Osborne	Licensed, Certified Professional Midwife, Helena Birth Studio
Sarah Sullivan	RN, Parent to two children with homocystinuria
Miranda McCabe	Early and Periodic Screening, Diagnosis, and Treatment (EPSDT) Program Specialist, DPHHS
Abdallah "Abe" Elias	Director of Medical Genetics and Clinical Geneticist, Shodair Children's Hospital
Shawnalea Chief Goes Out	(observer)
Lynne Wood	(observer)

Voting Advisory Committee Members Absent	
Name	Position
Allison Young	Pediatrician, Western Montana Clinic, Physician who is board-certified in obstetrics, pediatrics, family medicine, or neonatology
Marion Rudek	Nurse Practitioner, Blackfeet Community Hospital, Representative of a tribal health care system

Non-Voting Advisory Committee Members	
Name	Position
Amber Bell	Newborn Screening Coordinator, Children's Special Health Services, DPHHS
Debbie Gibson	Lab Services Bureau Chief, Montana Public Health Laboratory, DPHHS
Jeanne Lee	Newborn Screening and Serology Supervisor, DPHHS

Jacqueline Isaly	Family and Community Health Bureau Chief, DPHHS
Dani Lindeman	Laboratory System Improvement Manager, DPHHS
Douglas Harrington	Acting State Medical Officer, DPHHS
Miranda Reddig [absent]	Program Specialist, Newborn Screening, DPHHS
Nikki Goosen [absent]	Newborn Screening Clinical Laboratory Science Lead, DPHHS
Chelsea Pugh [absent]	Nurse Consultant, Newborn Screening, DPHHS

Facilitators	
Name	Position
Anna Schmitt	Co-founder, Yarrow
Mikaela Miller	Public Health Specialist, Yarrow

Guests	
Name	Position
None	None

Public	
Name	Position
Aviva Rosenberg	Co-Founder of Gaucher Community Alliance

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.

- Yarrow provided an overview of the Agenda, Ground Rules, and the Public Comment Period.

Unfinished Business Review

- X-ALD Update
 - X-ALD Screening began on September 3, 2024.
- Pompe Update
 - Adding Pompe to the Montana Newborn Screening Panel was approved by the Montana Department of Health and Human Services (DPHHS) Director
 - Procedure has begun for making the rule change necessary to add Pompe. Another update will be sent once testing begins.
- Membership Update
 - 4 Committee members agreed to end their terms early so that we may stagger the positions.
 - 3 positions have been filled thus far:
 - Representative of the Medicaid / Insurance industry: Miranda McCabe will be replaced with Shawnalea Chief Goes Out.
 - Physicians or Nurse Practitioners who are board-certified in obstetrics, pediatrics, family medicine, or neonatology: Dr. Allison Young will be replaced by Dr. E. Lynne Wood.
 - Person /Family Member affected: Sarah Sullivan will also be replaced. An offer letter was sent out to a new representative and we anticipate the position will be filled soon.
 - Thank you to the Committee members who are being replaced for their service and dedication to the Committee.
 - The Committee is still taking applications for the representative of a birthing center position.
- Committee Feedback
 - After the last meeting concluded, Committee members voiced their concern surrounding the lack of Committee discussion during the meetings. It was proposed that we would hold an additional meeting strictly surrounding how we may facilitate and encourage discussion and engagements by the Committee members during voting meetings.
 - Voting Meeting options included:
 - Holding an additional 1 hour meeting in February
 - Delaying the ASMD presentation until the Fall 2025 and discuss at regular Spring 2025 meeting
 - Discuss before the ASMD presentation during the Spring 2025 meeting
 - Discuss after the ASMD presentation during the Spring 2025 meeting
 - Other
 - Committee members voted and decided to have an additional 1 hour meeting in February.

Gaucher Discussion

- Gaucher Disease has not been nominated or reviewed by the RUSP.
- Dr. Abe Elias: Gaucher Disease results in a variable spectrum of severity of symptoms. Gaucher Disease Type 1 is the most common form of the disease. This form is often very mild in symptoms and may also be asymptomatic. There is also some concern for overdiagnosis.
- Dr. Douglas Harrington: It is relevant to consider treatment options for each type. Enzyme replacement therapy (ERT) is primarily successful in Type 1. It is only used in Type 3 for the amelioration of non-central nervous system issues and Type 2 is not. However, there is the possibility of stem cell transplants or gene therapy on the horizon, which makes this issue more complex.
- It is currently being screened in 7 states.
- Gaucher Disease can be multiplexed with Pompe disease, meaning the test can be run on the same blood spot test as Pompe disease.

Newborn Screening Advisory Committee Vote on Gaucher

- Voting members were asked *“Do you recommend including Gaucher 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
 - Miranda McCabe
 - Amanda Osborne
 - Kotie Dunmire
 - Abe Elias
 - Sarah Sullivan

4 of the voting committee members present at the time of vote voted: “No, do not recommend” to include Gaucher disease on the Montana Newborn Screening Panel. This did not pass quorum.

Public Comment Period

- Aviva Rosemberg: She states that she is very disappointed with the outcome, however, she thanks the committee for their time. ERT is the standard of treatment care for all types of Gaucher Disease. There is currently an abstract being published in February that demonstrates that every patient in the United States diagnosed with Gaucher Disease with Type 2 or Type 3 is receiving ERT as soon as they have the diagnosis, so there is no palliative care anymore. That is not the standard of care for any form of the disease which makes it crucial to have the babies

tested and on treatment as quickly as possible to prevent the suffering of the infant. She will continue her work to get babies around the country screened for the disease, so that their families don't have to suffer through unbearable pain and watching their child suffer. The quality of life for a Type 3 patient can be vastly improved with the introduction of ERT within weeks of their birth.

- Abe Elias: Although this was not approved, there is still opportunity for it to be re-reviewed in the future.

- Additional comments via email were accepted up to 12:00 pm MT on November 19th.
 - No additional comments were sent.

Thanks and Next Steps

- Follow up email will be sent soon and will include:
 - Meeting minutes
 - Recording
 - Presentation slides
 - Feedback survey
- A decision memorandum will be written to the Montana Department of Health and Human Services regarding the Committee's recommendation.
- A doodle poll will be sent out to Committee members to schedule the next meeting.
 - The next meeting will occur in February.
- Please email if you have questions, comments, or need anything.

This meeting was concluded by Anna Schmitt at 10:15 am on November 19, 2024.