Oregon Health Authority
Northwest Regional Newborn Bloodspot Screening Advisory Board

Meeting Summary **September 3, 2025**

Location

Videoconference

Quorum

Board attendees constituted a quorum for the meeting.

Board Members Attending

Marilyn Hartzell, M.Ed., Board Chair, Family Representative
Angela Douglas, MD Representative of a statewide association of pediatricians
Cheryl Grabham, Representative of advocacy association regarding newborns with medical or rare disorders
Andrea Keating, LDM, CPM, Representative of a statewide association of midwives
Sherly Paul, Representative of a statewide association of nurses
Elizabeth Powers, MD, FAAFP, Representative of birthing center or hospital
Kara Stirling, MD, Representative of a birthing center or hospital

Board Members Absent

Rusha Grinstead, Representative of Medicaid or insurance industry
Jill Levy-Fisch, Representative of advocacy association regarding newborns with medical or rare disorders
Mort Murry, MD, Representative of advocacy association regarding newborns with medical or rare disorders
Amy Yang, MD, Contracted medical consultant

NBS Program Staff

Patrice Held, Newborn Screening Program Manager
Kasfian Khan, Legislative and Community Engagement Coordinator
Sharon Willis, Newborn Screening Laboratory Manager

Guests

No guests were present

Members of the Public

John Powell
Kathie Fraser
SM Smith

Jensen Strategies Facilitation Team

Erik Jensen, Facilitator
Emily Rehder, Operations Manager
Laura Schneider, Project Associate

ACTION ITEMS

The Board took the following actions:

- Approved meeting summary for the May 28, 2025, meeting.

MEETING AGENDA ITEMS

1. Welcome / Introductions

Chair Dr. Elizabeth Powers opened the meeting and asked for the Board members, OHA staff, and facilitators to introduce themselves. She also expressed her appreciation to Marilyn Hartzell for her long service and expertise as the immediate past Board Chair. Dr. Powers shared that the position representing a statewide association of pediatricians is still vacant and if anyone knows of a potential candidate, please contact Patrice Held, NWRNBS Program Manager.

2. Meeting Overview & Approval of Meeting Summary

Erik Jensen, Board Facilitator, reviewed the agenda for the meeting. He also noted the Rules Committee would start at 11:30am after the Board meeting. Erik reminded everyone of the dates for the upcoming Board meetings noting varying times:

- Wednesday, December 3, 2025, 1:00pm – 4:00pm
- Tuesday, March 3, 2026, 9:00am – Noon
- Wednesday, May 27, 2026, 1:00pm – 4:00pm

All Board meetings, starting with this one, will be in a Zoom webinar format. Also, going forward, Board members will be sent two calendar invites – one to hold the date and the second within a month of the meeting with a link and an RSVP request. For assistance email Emily Rehder (emily@jensenstrategies.com).

Dr. Powers called for the approval of the May 28, 2025, Advisory Board meeting summary and asked for a formal vote.

Decision: The May 28, 2025, NWRNBS Advisory Board meeting summary was approved unanimously.

3. Program Updates

Patrice Held, NWRNBS Program Manager, provided updates on the Program.

2025 Legislative Session: Patrice reviewed items from the 2025 Legislative Session: HB2741 passed allowing families to decline screening; requires coordinated care organization (CCO) to cover costs of screening; establishes confidentiality of information and documents related to screening and subsequent care; directs OHA to implement a parent/guardian education program on medical conditions, screening, and resource organizations; requires OHA to establish a follow-up program for longer term patient care within available resources; and gives updates to language and definitions. The statute goes into effect January 1, 2026.

Questions from the Board included:

Q: How is the contact made to do the follow up with the family?
A: OHA connects the baby's primary care provider to specialists to conduct confirmatory testing and evaluation. If the testing is confirmed positive, the baby is cared for by a specialist, along with the primary medical provider. Whether OHA will have the ability to do some long-term follow-up to ensure babies identified with conditions are still getting access to care years later will depend upon resources. Some other states' newborn screening programs have long-term follow-up programs that Oregon can learn from. There may be other opportunities to help families stay connected, such as sending families information about advocacy groups or putting the advocacy organizations on the OHA website - which will fulfill the requirement of the statute. Suggestions from the Board of other approaches are welcome and may be discussed in future meetings.

Q: Did the bill allocate financial resources for the lab or OHA? Was this more of a mandate or just permission?
A: No funding was allocated to the program. There was a funding bill that did not pass. The statute wording says the long-term follow-up is dependent upon available resources.

Q: It's nice the CCO must cover the cost of the screening, and maybe the Board can talk about options for outreach and follow-up in the future. The Board might propose a range of things and inform future legislative requests for additional funding. Is that kind of thing useful to the Board to discuss?
A: Absolutely, to figure out what options are for long-term follow-up within OHA resources.

Q: Now that the requirement is CCO's must cover the cost of the screening, will this impact community providers?
A: We need to understand that better. Rusha Grinstead is the Board's Medicaid representative and may have helpful information.

Preparations for Infantile Krabbe Disease Screening: Patrice provided an update regarding preparations for Infantile Krabbe Disease screening.

- OHA has validated the method to measure the GALC activity with babies who have low activity. Samples will be sent for psychosine testing at the Mayo Clinic. Reporting and communication templates for primary care providers are under development. Confirmatory tests and treatment plan protocols are being developed with the OHSU metabolic and transplant teams.
- OHA has notified providers, partners, and public that screening will commence November 1st.
- Oregon Administrative Rules (OAR) will be updated at the Rules Advisory Committee meeting directly following the Board meeting.

Infantile Krabbe Disease Screening: Patrice reviewed the Infantile Krabbe Disease (IKD) screening parameters related to psychosine values. She noted the national panel, from which the Advisory Board bases its screening recommendations, only approved IKD versus the entire disease spectrum that would include the later onset

forms. IKD is identified when pychosine levels are above 10 nmol/L and designated as abnormal. Test results that are between 2 nmol/L and 10 indicate a risk of the later onset or atypical form of Krabbe

After consultation with the Mayo Laboratories, who will be conducting psychosine testing, and the OHA legal team, it was decided that all babies with an elevated psychosine greater than 2 are going to be referred for additional testing and evaluation even though our target of the screening test is IKD.

Questions and comments from the Board included:

Q: Since some of the family advocates asked for the testing to look at these mid-range levels, we appreciate the additional review. How frequently is a positive test anticipated? Was it less than one a year? Also, how frequently would the mid-range 2-10 occur? Are the resources there to follow up with the 2-10 result?
A: It is estimated the frequency would be 2 to 3 times a year more than infantile. It is a higher frequency, but it is such a rare disease that they're probably only looking at one or two a year that would be greater than 2 nmol/L. There is reluctancy to include the full spectrum because it is not always clear when the later onset forms will appear requiring more monitoring and intervention for families at risk. There are also concerns from providers on how to best care and treat children with the later onset forms. National recommendations for monitoring and treatment, especially for the later onset forms, are expected to be published. At least 10 or 11 states right are now testing for the full disease spectrum of Krabbe.

Q: Is it possible and appropriate to contact the family advocates to make them aware of the change?
A: Yes, there has already been an opportunity to reach out to talk to one of the family members who will hopefully help get the word out to the community.

Comment: It seems fortuitous that this is what we have to do because it does seem like there's more evidence that earlier treatment before symptom onset is the most beneficial. We're not looking at huge number of people to follow so the risk is less than the potential benefits of disclosing the information and has my support.

Q: If the enzyme level is low and sent to Mayo for further testing, is the provider notified at that time pending the results from Mayo?
A: Mayo is poised to do this very, very quickly so we will not release the report until we have the psychosine values.

Q: You only anticipate a very small number needing to be sent for that confirmatory testing, the enzymes should be normal in most?
A: Yes, that is correct.

<u>Newborn Screening by Whole Genome Sequencing Collaboratory Initiative</u>

As shared at the last Board meeting, there is a research opportunity, administered by the National Institute of Health (NIH), to assess the feasibility of a collaborative, multi-state model for newborn screening (NBS) using whole genome sequencing as a first-tier assay for analysis of a select group of genetic conditions actionable in the first year of life. NIH is funding a primary recipient to organize all phases of the study, including

consenting, sequencing, and the return of the results. This primary recipient would be partnering with interested public health laboratories.

OSPHL has submitted its name for consideration to a research participant. If OSPHL is selected, it would work with identified hospitals to implement the consenting process for families to participate in whole genome sequencing on the newborn screening sample. For families who consented to the study, OSPHL would send the samples to the primary recipient to complete the sequencing and subsequently OSPHL would return the results to the family members along with OHSU and the appropriate specialists.

Questions and comments from the Board included:

Q: What is the timeline in learning if we were selected?
A: A decision is anticipated in the next month or two although that may be delayed with current events at NIH.

4. NWRNBS Program 2024 Annual Report

Patrice presented the 2024 NWRNBS Program Annual Report. This was the first year OHA created the report and intend to submit the 2025 report in the spring of 2026. Hopefully, it will be beneficial to the submitters as well as the public to help raise awareness of the benefits of screening. The 2024 metrics were reviewed by Patrice including screening numbers, collection statistics, sample transit times, turnaround time for results, number of babies identified with a treatable condition, most prevalent conditions identified, and false positive rates.

The Program's 2024 accomplishments included: 60 collection trainings, 24 site visits, resources for submitters created, updated NICU guidelines, redesigned QA report, paperless reporting, and a redesigned NBS collection card.

Questions and comments from the Board included:

Q: How is a false positive determined?
A: The false positive rate varies by disease. The specificity of the measured analyte and the testing algorithm for a condition has a large impact on the false positive rate. For example, in lysosomal storage disorders, such as Krabbe, we measure the enzyme activity. If the activity is low, a second-tier test is performed to measure a more specific analyte, psychosine. Since we are looking at specific target analyte, the false positive rate for Krabbe disease is expected to be very low.

However, in the case of cystic fibrosis (CF), immunoreactive trypsinogen (IRT) is measured and the top 4% of IRT values each day are reported with a request to repeat the screen. If the IRT is elevated on the second screen, then molecular analysis of a variant panel is performed. The higher false positive rate for CF is due to the molecular analysis being performed only after the baby has two samples with elevated IRT. If sequencing was performed on the first sample with an elevated IRT, the false positive rate would drop dramatically.

Q: Do the families know the false positive rate going into the test?
A: The information should be communicated to them by their providers.

Comment: The report was well written, and it had a lot of valuable information. With so

much information that people either want from the Program or would benefit from knowing captured in one document it is very accessible to read.

Patrice asked for help in brainstorming whether it's accessible for the public beyond being on the website and how the information could be more bite-sized not only for expecting parents but also for the public.

5. Post ACHDNC Dissolution: Disorder Review Process

Patrice shared an update regarding the dissolution of the national Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) and the approaches being considered or implemented to fill the role including:

- A bill is being introduced in Congress to reauthorize the newborn screening program as the U.S. Health and Human Services (HHS) Secretary's advisory committee.
- The American College of Medical Genetics and Genomics has proposed to take up the work of the ACHDNC by establishing a committee to review conditions and make recommendations. The recommendations wouldn't get added to the Recommended Uniform Screening Panel (RUSP) but could provide states with some guidance.
- In lieu of the ACHDNC, the Health Resources and Services Administration (HRSA) is seeking comments on both Metachromatic Leukodystrophy and Duchenne Muscular Dystrophy that support or oppose their additions to the RUSP. The portal is open till mid-September.

Questions and comments from the Board included:

Q: Have they said whether the information that they use to make their determination will be shared? If so, how?
A: The scientific evidence for Duchenne Muscular Dystrophy screening was prepared and reviewed by the ACHDNC. However, the board decided not to recommend DMD to the RUSP. Scientific evidence report for Metachromatic Leukodystrophy screening was also generated, but the disease was not reviewed by the Committee, due to its termination. The scientific evidence for both conditions can be found on the website.

Patrice shared four conditions that OSPHL has been approached by advocates in support of adding to the Oregon screening panel including: Duchenne Muscular Dystrophy (DMD); Congenital cytomegalovirus (CMV); Metachromatic Leukodystrophy (MLD); and Cobalamin synthesis related defects.

DMD and CMV have been discussed but not reviewed by the Advisory Board because they are not on the RUSP. MLD has not been discussed by the Board, however, advocates recently met with OHSU medical consultants and it appears they are in favor of MLD screening but are waiting for it to be added to the RUSP or by another approach.

Erik facilitated a Board discussion on what alternative approaches OHA and/or the Program could take for condition review, if the Recommended Uniform Screening Panel (RUSP) is no longer an active resource. Currently, the Board's disorder evaluation protocol requires a condition to be listed on the RUSP before it can be considered for inclusion on the NWRNBS screening panel. For discussion purposes, the following

options were offered:

1. Cease all NWRNBS disorder evaluations until a national level review process is established to replace ACHDNC's role.
2. In lieu of Step 1 of the Board's disorder review protocol (addition to the RUSP):
    A. Establish a process to assess whether a disorder evaluation is appropriate – consulting with appropriate medical specialists.
    B. If evidence suggests screening is the best practice, medical specialists or advocates may bring the condition to the Board for consideration.
    C. The Board determines whether to conduct a formal NWRNBS program evaluation, starting with Step 2 of the Board's disorder review protocol. (Program Evaluation).
3. Other Option?

Erik asked if anyone would advocate for ceasing disorder reviews at this stage or would they advocate for an alternative? And if an alternative is to be considered, what does that looks like?

Questions and comments from the Board included:

Comment: It is such a hard time to be doing this work and appreciate all the information provided. This feels like a few years ago when there was a bill in the Oregon Legislature that failed, suggesting that rather than requiring addition to the RUSP, the board would have to evaluate any condition. The concern at the time was the massive amount of time and scientific resources that would be necessary to implement that bill. That said, you can't just ignore it until something is re-established but there would need to be a way to limit what conditions the Board evaluates.

Q: Are there other conditions that the Board has approved but not implemented yet?
A: Yes, GAMT and MPS2 have been approved. Implementing a new condition takes a lot of time and resources. Krabbe was easiest to implement and had the most advocacy support, so the Program opted to go with that disease first.

Comment: The first option sounds like the Board is sort of giving up or just holding, but we already have a couple of things in the queue that will need time to get implemented. So even if we aren't evaluating new disorders, we're not putting things on hold, we are moving forward with the resources that we have with what we've already approved.

Q: States often advance policies before there's anything consistent at the federal level, to what extent does Oregon coordinate with other states?
A: There are advocacy groups that sometimes approach the legislature directly to introduce bills to add conditions.

There is a group called the Everyday Life Foundation, and their goal was RUSP alignment legislation to be introduced in all states, but now we don't have the RUSP.

Comment: I often find great benefit to having government-only calls with colleagues in other states, where we can look for that alignment and kind of understand where the science is leading in advance of any sort of guidance or direction from any external party and help bridge the gaps sometimes.

Comment: Balancing science with desire and resources, we could consider new conditions depending on resources available. Is there a way to say that it is our goal or intent, but the resources required at a state level are so much greater without the national support and the infrastructure behind it? Can we communicate our desire to consider and move forward as the science support allows, and treatment is available while also recognizing reality of resource allocation.

Comment: If we have conditions the Board has approved and not implemented on the screening panel, it seems irresponsible to consider additional conditions until we've figured out how to move those forward.

While Krabbe was a lower lift, and had the most advocacy, but just being the most vocal doesn't mean it's necessarily the right one to move forward first. If we have other ones that have gone through the formal process and been approved but are not implemented yet, then I do think we should actuate the ones we've approved first. It is a balance between option 1 and 2. We might need to cease and desist right at this moment until we can actuate the disorders we have already approved and then evaluate our resources and decide. After that, it kind of depends how national and other state organizations address this issue.

Comment: In terms of sort of the hybrid between options 1 and 2, it would still take time to figure out what that looks like and have the Board go through and put together a new protocol or a substitute protocol for the time being, so there is that lapse in time, when the other disorders may be still be moving through the operational side of the process.

Comment: If we decide to go with any version of option number 2, we can't move forward, so we basically are doing option number 1 until we figure out something else.

Q: Are there other states who don't follow the RUSP and what is their process like to review disorders? How do you do that in an efficient, not resource intensive way, and are there ways we can partner with other states or state labs who are looking at this to pool our limited resources? What are other state labs are doing who are following the RUSP, are they coming up with a whole new process?
A: As noted, before, it would be beneficial to gather some additional information, and we will look into getting it.

Comment: It would be best to move forward with considering conditions, but only if the structure was developed in a meaningful and intentional way. Also appreciate the comment around being mindful of the resources, because our decision to add a condition is not where things end. A concern is if we make decisions to add multiple conditions and then find out through additional scientific and/or medical information that maybe the order we chose isn't ideal or doesn't work best for our program or for the people in our state.

Erik summarized the Board's direction to conduct research on what programs are doing in other states and come back to the Board with that information to have a more informed discussion.

Questions and comments from the Board included:

Comment: There is a concern that the information gathered from other states may be

too varied and not helpful considering each state has their own amalgamation of their legislature and groups.

Patrice added that the NWRNBS Program can pull together different protocols that states with advisory boards are using in making decisions.

Erik asked if there is particular information the Board feels would be helpful from these other states for an informed discussion.

Questions and comments from the Board included:

Comment: Always interested in how other programs are funding their screening.

Comment: There are different processes for filtering or screening the potential conditions. How they do their scientific review if they don't follow the RUSP and, if they do use RUSP, how they are managing it.

Q: What was the process in Oregon before the Board was put in place? How did conditions end up on the panel then?
A: The lab just decided without input.

6. Public Comment

No comments were presented.

7. Wrap-up

Dr. Powers commented that the meeting was efficient and effective and really appreciated the conversation. She asked the Board members to reach out to Erik or Patrice with any other thoughts or input. The next meeting will be on December 3rd from 1pm to 4pm. The meeting was adjourned.