

Oregon Health Authority

Northwest Regional Newborn Bloodspot Screening Advisory Board

Meeting Summary

February 4, 2020

Location: Oregon State Public Health Lab, Hillsboro, Oregon

Attendees

Board attendees constituted a quorum.

Board Members

Silke Akerson, CPM, LDM, representative of a statewide association of midwives

Chris Biggs, MS, NWRNBS Program Manager (co-chair)

Dr. Philip Dauterman, MD, FCAP, entity that contracts with NWRNBS for newborn bloodspot screening

Anna Dennis, MS, CGC, advocacy association regarding newborns with medical or rare disorders

Cheryl Hanna, MD, representative of a statewide association of pediatricians—co-chair, subject matter expert X-ALD

Dana Hargunani, MD, MPH, Medicaid or insurance industry

Marilyn Hartzell, M.Ed., person or family member of a person affected by a disorder on the Newborn Screening Panel

Wannasiri (Awe) Lapcharoensap, MD, representative of a statewide association of pediatricians

Jill Levy-Fisch, representative of advocacy association regarding newborns with medical or rare disorders

Joanne Rogovoy, advocacy association regarding newborns with medical or rare disorders

Kara Stirling, MD, representative of birthing center or hospital

Amy Yang, MD, contracted medical consultant, vice chair

Cate Wilcox, MPH, honorary representative

Collette Young, PhD, honorary representative

Absent

Deb Wetherelt, RN

Visiting experts

Ladawna Gievers, MD, pediatric bio-ethicist

Laurel Boyd, MPH, contracted disorder researcher

Erika Finanger, MD, subject matter expert SMA

David Koeller, MD, subject matter expert X-ALD

Members of the public

Cheryl Grabham

Wren Grabham

Sarah Miller

Shelly Eisenberg

Don Stetcher

Maynard Friesz, Cure SMA (on phone)

David Randall

Staff

Chris Biggs, MS

Nicole Galloway, PhD

John Fontana, PhD, HCLD (ABB)

Oregon Consensus Facilitation Team

Robin Harkless, facilitator

Cat McGinnis, note-taker

ACTION ITEMS

- The program will send a meeting evaluation to the board for assisting with process adjustments moving forward.
- The program will send the revised charter, with additional details on roles and responsibilities and feedback mechanisms for chair, vice chair, to the board for review.
- For the next meeting, the program will gather additional information about how other states are testing for SMA and X-ALD, per the questions that arose during today's meeting.
- The program will revise the draft criteria for removal of disorders and redistribute it to the board in time for a June/July face to face meeting.

1. Introductions

Attendees introduced themselves.

2. Follow-up from last meeting

Evaluation of last meeting. Robin Harkless, Oregon Consensus (OC) facilitator, shared the high-level results of the first (July 19, 2019) board meeting evaluation, noting there were nine respondents. Responses on the questions were average to positive on information and presentations. Some felt the meeting was a little rushed and would like to be able to take time needed to understand the information and also have more dialogue with each other. Robin also reminded the board that her team are not subject matter experts, rather are providing collaborative process support via process design, meeting facilitation, and third-party documentation of the group's work. The board, comprised of many different experiences and perspectives across the system, will not always be at the same level of comprehension on a topic and will need to accommodate each other in shared learning. She asked that any process needs be raised with her team as they arise.

ACTION: The OC team will update an evaluation to be sent out for today's meeting and suggested the feedback is very helpful in helping the program and facilitation team make adjustments as this board is just getting established.

Legislative report. The program provided copies of the board's report that was sent to the legislature in December 2019. The next legislative report is due September 15, 2020. After that, reports will be due September 15th of every even-numbered year. Given OHA's internal draft review timeline, the board may or may not be able to have the results of the next in-person meeting (to be scheduled in June or July) included in the September report.

Rule changes. Nicole Galloway reported on two rule changes. The first was discussed at the July 2019 meeting and went into effect November 25, 2019. The second rule update was administrative and did not have an impact on stakeholders. This went into effect February 1, 2020.

Public meeting law. Program staff attended training in public meeting laws. One take-away was that, during advisory board decision making, the consensus rating (“vote”) needs to be recorded for each board member by name.

Legislator tour. The program reported that they hosted a lab tour with Rep. McLain, Rep. Sollman, and Rep. Schouten.

Subcommittee on chair and vice chair roles and feedback mechanism. Chris Biggs, Marilyn Hartzell, and co-chair Cheryl Hanna worked on clarifying roles for the chair positions and reported their updates: board co-chair is a resource to the advisory board, is the voice of the board, and is the public face. The vice chair is to remain current on activities of the board and back-up the board co-chair should the board co-chair be unable to serve in the role. There was a change to value six—that all members have implicit bias, and recognition of that bias may have the effect of creating more equitable outcomes. The program will send the revised charter to the board for review. The group also began drafting questions for an anonymous survey as a mechanism for providing feedback to the chairs, and will send the initial questions out for board edits and additions. It was noted that when a feedback form is filled out, it should go to the facilitator not the program in order to comply with public meeting laws.

Experts. At the last meeting, the board requested that special experts attend this meeting, including an ethicist. Attending experts are listed above, including an ethicist, research consultant, and SMEs for the disorders discussed today.

Public comments outside of meetings. The program has an email address for the public to provide comments, which is located on a dedicated advisory board webpage. If board members know of someone with a comment, they should refer them there. The webpage address is: <https://www.oregon.gov/oha/PH/LABORATORYSERVICES/NEWBORNSCREENING/Pages/advisory.aspx> and the email address is: NBS.AdvisoryBoard@dhsosha.state.or.us

Work plan. The facilitator walked the group through the board’s work plan, including a timeline for the next legislative report.

3. Process for adding disorders to the panel

Chris Biggs, Program Manager, explained the process for adding a disorder to the screening panel once the board approves. The program will do the following:

- Finalize a fiscal analysis to inform their funding ask to the legislature
- Approach the legislature with fee increase request
- Initiate a rule change to add the disorder (requiring a rules advisory committee)
- Work to achieve *program readiness*, including validating that the testing method works correctly
- Make necessary changes to implement the screening

A board member asked whether, if the board votes to add a disorder, it will be added for sure. The program said there would have to be a really good reason not to add the disorder in such a case. A possible barrier would be lack of legislative funding.

4. General discussion

- *Funding for ACHDNC.* A board member pointed out that this year the Newborn Screening Saves Lives Act (signed by President Bush in 2006) reauthorization didn't pass and the Advisory Committee on Heritable Disorders in Newborns and Children Committee (ACHDNC) will not meet. A board member predicted that, if that committee does not reconvene, a national group will arise to fill the absence; however, some benefits will be lost.
- *Independent reports on SMA (spinal muscular atrophy) and X-ALD (X-linked adrenoleukodystrophy).* Laurel Boyd, the consultant who developed the independent disorder reports, discussed how the reports were built. Laurel has a background in epidemiology and medical subject review. She said the reports used ACHDNC methods. Major variations from ACHDNC format included: the reports looked extensively at context about the program, knowledge of experts, and feasibility of implementation from program perspective. Consultant looked at screening, treatment, and policy-related articles and screened for bias and discussed bias in the reports.
- Dana Hargunani pointed out that references to the Health Evidence Review Commission (HERC), who she represents, and coordinated care organizations (CCOs) in the reports were not completely accurate. She clarified that HERC has the authority to set benefits according to Medicaid. They set CCO coverage for Medicaid and fees for services. Broadly speaking, benefits must conform with HERC. Pharmacy and Therapeutics at OHA advises HERC on pharmacology and therapeutics.
- A board member asked if the reports look at FDA approved processes for treatments. The response was no, there is tangential discussion, but it's not a focus of the reports.

5. Presentation on SMA

Dr. Erika Finanger, SMA pediatric expert, presented information about SMA. Highlights of presentation follow:

- SMA type 1 has been known about since 1891. Gene was not identified until 1995. After that, therapies moved quickly. Type 1 is the most common.
- SMA is the most common cause of genetic death among infants.
- There is generally a long delay between onset of symptoms and diagnosis (four month delay for type 1 and over a year for type 3). It's a long and stressful diagnostic journey for parents.
- The sooner SMA is diagnosed, the better the outcomes for the patient.
- Current treatments include nusinersen and onasemnogene abeparvovec (gene therapy). Studies of nusinersen show significant improvement in outcomes for patients.

- Issues related to equity: There is access to pediatric neurologists in Portland, Eugene, and Medford. Treatment is available only in Portland. Testing is free in two labs in the state. So far Dr. Finanger has been able to treat all patients with nusinersen. There is a battle to get coverage and some patients are still on free drugs with no insurance coverage.
- Seventeen states are currently screening for SMA. Fifteen more have adopted the screen, but have not yet implemented it. Six are piloting the test.

Q&A—

- Are there any ethnic groups at higher risk for SMA? No.
- Any idea why false positives are so different in different programs? Maybe the technology in screening. There may also be differences in how they define false positives.
- Based on what you've seen, would it be better to cast a wider net and catch carriers as well? Yes. The American College of Medical Genetics presses for carrier testing.
- What are the side effects of treatment? Nusinersen is pretty well tolerated; however, there are risks related to sedation and for administration of the lumbar injections. Some patients have a drop in platelets, but don't need action. We check for protein in the urine. Have had no complications. With gene therapy, patients need steroids for a month. There can be a reaction in the liver; one child needed hospitalization. Families help decide treatment.
- Do both treatments continue indefinitely? Gene therapy is a one-time treatment. Nusinersen is used for life.

7. Public comment

- Cheryl Grabham: I'm a long-time Oregon resident and mother of an SMA patient. I strongly recommend adding SMA to the newborn screening panel. Our daughter's symptoms didn't appear for two years. She began falling a lot. Daughter can no longer walk in the woods as she loved to do. SMA treatment has increased her strength. We've connected with other SMA families. It's an exciting time for treatment. It's most effective when diagnosed at birth. Hundreds of families are watching for swift action by the board.
- Wren Graham: I'm fourteen years old and have SMA type 3. Newborn screening for SMA is important. They have a longer chance of walking if they get treatment.
- Sarah Miller: I've lived a decade in Oregon. My child has SMA type one. She turned five in December. I urge the advisory board to add SMA quickly. Our child was having trouble swallowing, breathing, and moving. The child was diagnosed at six months. In a blind clinical trial, she got the drug. She gets around by wheelchair. She's tube fed and can't swallow. If it had been caught earlier, perhaps she could swallow and walk. It makes a lifetime of difference. Twenty-three states already screen—Oregon needs to be next. Time is of the essence, so babies can stay strong.

- Dave Randall: Shared personal story about his child being diagnosed in 2012 with SMA. There was no treatment at the time (and no screening). He started a foundation to help fight the cause and fund research. The landscape is different now.
- Maynard Friesz, Cure SMA, discussed national work on SMA. He discussed the nature and incidence of SMA. He urged the board to add SMA to the screening panel and pointed out that it is an important time for SMA; many new treatments are being identified and approved.
- Shelly Eisenberg: I urge you to support SMA screening. More treatment early is essential. My niece has SMA and has been in a motorized wheelchair since the age of two. She's almost eighteen now, is going to PSU, and wants to be a teacher. Her parents have concerns most parents of college students don't have to face.

8. Board review of SMA for potential addition to screening panel

Stage one: Disorder has been added to the RUSP.

Stage two, category 1 criteria: The program reported on its review:

1. The condition is well-defined in newborns. Yes
2. Earlier intervention results in improved outcomes compared to later identification. Yes
3. The population level incidence and prevalence are known. Yes
4. There is a Federal Drug Administration (FDA) approved testing method available using dried blood spots or an accurate testing method is available that meets clinical laboratory requirements for validation and testing by the laboratory using dried blood spots? Yes. (Mostly molecular-based tests that are not FDA approved, but they can be validated.) Note—program analysis assumes there would be screening for homozygotes only and not carriers because of the complexity of the latter.
5. Diagnostic and specialty testing is available. Yes—free and quick (if doctor knows to test for it).
6. A treatment is available. Yes
7. The contracted NWRNBS medical consultants have been consulted and appropriate specialized medical consultation is available or can be obtained by the program. Yes
8. The specific condition appears in the funded region of the prioritized list as determined by the Oregon Health Evidence Review Commission. Yes
9. The NWRNBS program has sufficient information to perform a fiscal analysis. Yes
10. The impact to NWRNBS contracted partners has been assessed. Yes (Contacted other programs, and Saipan responded.)

Stage three, category 2 criteria: The board discussed its review:

Q&A—

- Does HERC support treatment coverage under Oregon Health Plan (OHP)? OHP has adopted coverage for both treatments with some prior authorization. OHP sets the foundation for CCO coverage. However, Dr. Finanger pointed out that a lot of patients are not authorized by the CCO. This may limit the treatment options available to each individual patient.

- Is there coverage for administering the drug and related procedures as well as for the drug itself? Dr. Finanger reported that coverage of the administering treatment is spotty, but quickly evolving.
- Is there any information about when free drugs will end? According to Dr. Finanger, no, but it's always a worry.
- If a patient doesn't have OHP and has poor insurance what do they do? What about the timing of treatment while they deal with red tape? Dr. Finanger asks insurers for a forty-eight-hour turnaround. It's tricky. They want to treat type 1 babies within days. The provider has to have an individual financial agreement with parents before treating given the costs.
- Is there any cost-benefit analysis regarding the burden on the state of early detection versus treatment without early detection? Dr. Finanger reports there is an article on the cost of delayed treatment.

Board analysis of criteria—

Criterion 1: *The population level public health benefits of screening outweigh the risks and harms.*

Board discussion, summarized below:

***Facilitator's note:** *While some of the dialogue was aimed at evaluating the disorder for criterion 1, the group spent a lot of time discussing the ethicality and costs of doing carrier screening and second tier testing. The comments and questions are separated into these two discussions below.*

Carrier screening:

- A board member raised concerns with identifying carriers, for the reason that the process if done by the program doesn't adequately allow for informed choice about carrier screening. There may be a lot of people ethically or religiously opposed to carrier screening who will turn down the entire SMA test or blood spot screening as a result.
- A board member pointed out that one in twenty infants with SMA will be missed if there is no screen for carriers. Essentially, every five years one would be missed. Is that acceptable given the impact on families?
- What about detecting but not reporting carriers? Currently the lab doesn't do that. There is an obligation to report findings.
- The principle of Children's Right to an Open Future is in question if a child doesn't have the ability to make a decision themselves about finding out if they're a carrier. How many children is it right to miss in order to preserve that right to an open future?
- Might you hold carrier information until the child is eighteen? Program response: A public health screening program inevitably misses some cases so even this approach would not ensure we would catch everyone.

- Are there different modalities and costs for identifying people with the disorder vs. identifying carriers as well? Program response: Program can adapt the modality to either, but cost difference is not in the testing, it's in the other program pieces.
- If we attempt to capture carrier status, we would need a lot of genetic counselors. One in thirty infants tested are carriers.
- Yes, but is there a way to capture as many sick babies as possible?
- Lab is currently sending out second-tier testing for lysosomal storage disorders, which also identifies carriers. Program response: lysosomal storage disorders are a special case since there are pseudo-deficiencies as well as carriers. Second- tier testing was implemented to reduce the number of false positive results reported.
- A couple other tests incidentally identify carriers. Primary care physicians have to follow up with families. It does harm to families who are carriers—it's very stressful. They have to understand complicated information.
- Comment to Dr. Finanger—if you develop a test to identify carriers, they would need follow-up sequencing and follow-up surveillance. Yet, only one in one thousand is going to develop the disease. What is the logistical impact to your clinic? There are 800–1,300 carriers each year. Clinic couldn't see these new patients. There is not enough genetic counselor support to handle this.
- Informed choice is critical—everyone should be consented. If carriers tested for, there could be significant damage to the testing program. It would reduce screening overall. It's a very serious concern.
- What are other states doing regarding carrier status? Program response: New York was doing a pilot which included identifying carriers for SMA. Other programs are not identifying carriers. Program will find more information on this.

Criterion 1

- Casting a wide net to not miss a baby is worth much to a family. Missing one baby is too many.
- Facilitator to the group: much of the discussion so far is around carrier testing. Back to criterion 1. Assuming as the program has, that only testing for homozygote deletions would be conducted, does anyone disagree that criterion 1 has been met? No dissent. Affirmative response around the room that criterion 1 is met.

Criterion 2: *There is adequate capacity and expertise in the NWRNBS program to implement and maintain testing and reporting.*

Facilitator's note: *From this point forward, the group agreed to assume, as the program had in its evaluation of the initial, tier one criteria, that only a test to identify individuals with the disorder would be done, not a test which would also identify carriers. The group agreed to follow up at a future meeting after the program gathered additional information from other states to inform the carrier testing discussion.*

Discussion:

- The program reports that the test is easy—everything else is complex. They do have the expertise and are doing a policy option package for the legislature based on the assumption that they will add the test without the carrier portion. They will need more people to perform follow-up and report results.

Criterion 3: *There is adequate capacity and expertise in the NWRNBS program to implement and maintain follow-up and education for providers and parents.*

Discussion: The board generally agreed that this criterion has the same evaluation as criterion two.

Criterion 4: *The NWRNBS program has adequate fiscal resources for implementing the test, performing the test and conducting follow-up and education.*

Discussion:

- How feasible is it to implement without carrier status and later add a test for carrier status? Program response: Program would need to revalidate the methodology, which is not difficult. They would have to add follow-up staff. They would need good data to make the change, since there's a huge education piece.
- Would the board have to do this analysis again for carrier testing if we were to consider adding it to the screen? Program response: Yes.
- With no carrier test, what is the fiscal impact to the program? Program response: Need personnel for testing and follow-up. Need to amend consultant contracts. Would have to take need for additional position authority and fees to the legislature in 2021.
- If no carrier testing, would need education so providers know there could be a case of SMA even though newborn screening had been done.

Criterion 5: *The population level incidence, prevalence, and disease burden are significant enough to merit screening.*

Discussion: All board members said yes.

Criterion 6: *Diagnostic and specialty testing is available and accessible that allows a definitive diagnosis to be made.*

Discussion: All board members said yes.

Criterion 7: *An effective treatment that is proven to result in clinically significant benefits is available and accessible.*

Discussion: All board members said yes.

Criterion 8: *There is equitable care and treatment for the disorder.*

Discussion:

- Is it equitable for rural people? If you live in eastern Oregon and the provider submits a bad sample, it could be three weeks before there are test results. What can we do to make it more equitable? Courier service? Program response: We did not assume courier service in our evaluation of tier one criteria.
- Equity of timing is not unique to this condition. There is a concern about making that a criterion when other conditions are also time sensitive and the same rural burdens apply.
- Program comment: courier service is not in the policy option package. Chris acknowledged that the board asked for a cost analysis of courier service at the July 19, 2019 meeting, but the program has not been able to do this yet in preparing for the current meeting.
- So many tests are time-sensitive. Need to work hard on educating hospitals in rural areas about the importance of screening. As far as courier service, some states are dealing with it case by case.
- Suggestion that discussion about equity—in regards to all conditions—be continued at another meeting in a broader context.
- All board members agree that SMA testing is as equitable as newborn screening can be.

Criterion 9: Addition of the disorder is not prohibitive to NWRNBS contracted partners.

Discussion:

- One of the program partners (Saipan) reported that, with a population of 50,000, they do see SMA cases.
- Program comment: Only one partner responded to the survey—Saipan. So, impact to partners is somewhat unknown. There's a need to hear from other partners or we can assume that no response equates to no prohibitive impact.

Consensus check on proposal to add SMA to the screening panel with no carrier screening at this time.

(1=enthusiastic agreement, 2 = agreement, 3 = on the fence/neutral, 4 = serious questions or concerns but not going to block from moving forward, 5=no agreement, would block action)

Votes:

Amy Yang 1
Anna Dennis 1
Jill Levy-Fisch 1
Kara Stirling 1
Joanne Rogovoy 1
Marilyn Hartzell 1
Wannasiri (Awe) Lapcharoensap 1
Cheryl Hanna 1
Silke Akerson 1
Phillip Dauterman 1

Strong consensus to add SMA to panel.

Discussion:

- Program next steps: This consensus recommendation will be included in the legislative report, and the program will seek legislative approval in 2021.
- Facilitator: There is an unresolved question about whether to add carrier screening. At this time, is there a recommendation from the board about adding this to the panel? (No proposals were offered at this time.)
- **Action:** Board members requested information from other states that are identifying carrier status. Board questions include: What percent do and don't do carrier status? What known impacts are there to the system, families, and the medical community? Have states that screen for carriers found any reduction in overall screening for SMA? For those with opt-in requirements, what are the rates?
- **Action:** Follow-up information gathering on options available for carrier testing? Cost analysis of different methods as current models, including costs to primary care doctors.

9. Preliminary discussion of X-ALD

Since some board members needed to leave, the facilitator invited their comments about X-ALD.

Discussion:

- Silke Akerson had to leave, but reported that if polled now, she would vote 4 on the consensus scale because she doesn't feel clearly informed or convinced about whether early identification of X-ALD improves outcomes.
- Kara Stirling - how often is early intervention missed, per criterion 2?
- A subject matter expert available for this disorder suggested that, if a bone marrow transplant is done on an X-ALD patient before the brain is involved, it improves survival. The challenge is that symptoms appear between three and ten years of age. Early intervention is the only way to catch X-ALD before the brain is involved.

10. Presentations about X-ALD

Dr. David Koeller, subject matter expert for X-ALD and Cheryl Hanna, MD, representative of a statewide association of pediatricians—co-chair, subject matter expert X-ALD, each gave presentations on X-ALD. Highlights of the presentations follow:

- People with X-ALD have trouble processing very long chain fatty acids. It affects membranes.
- Men will pass on an X-ALD gene to all daughters, who will then be carriers of the disease. They will pass the gene on to no sons. Female carriers put either sex offspring at 50/50 risk. Females who are carriers of X-ALD develop mild symptoms with no central nervous system or adrenal impacts.
- Pretty rare condition and there is no way to predict phenotype.

- X-ALD can be treated with bone marrow transplant, and gene therapy is coming. Bone marrow transplant can't be done until a change shows on MRI. So, if you know a child has X-ALD, MRIs can be used to catch progression of the disease before symptoms surface.
- The largest institutional cohort report is "Outcomes after Allogeneic Hematopoietic Cell Transplantation for Childhood Cerebral X-ALD."
- Regarding equity—historically there has been insurance coverage for clinical symptoms. If it becomes part of the newborn screening, there should be no problem with coverage. There are potential equity issues due to limited imaging capacity in rural areas. If a patient needs stem cell transplant, it is unclear whether it will be covered in Oregon. (The group was reminded and passed on to David the comment from earlier in the meeting that treatment is on the HERC priority list, but may be interpreted differently by CCOs. Coverage may be discretionary at the CCO level.) Patients would have to come to Portland for a bone marrow transplant, some are sent to Minnesota.
- Diagnostic testing will get some positives that are not related to X-ALD. Treatments have high morbidity rates and high cost. Testing will identify untreatable disorders and adult onset for which there is no treatment.

11. Board review of X-ALD for potential addition to screening panel

Stage two, category 1 criterion: The program reported on its review (based on an assumption that it would not be performing second-tier testing):

1. The condition is well-defined in newborns. Hard to answer yes because spectrum is so broad.
2. Earlier intervention results in improved outcomes compared to later identification. Hard to answer yes—though there is evidence that earlier is better, it's not necessarily in the first 28 days after birth.
3. The population level incidence and prevalence are known. Yes
4. There is a Federal Drug Administration (FDA) approved testing method available using dried blood spots or an accurate testing method is available that meets clinical laboratory requirements for validation and testing by the laboratory using dried blood spots? Yes, but will pick up other conditions with no treatment.
5. Diagnostic and specialty testing is available. Testing is suggestive, but need follow-up to confirm.
6. A treatment is available. Yes
7. The contracted NWRNBS medical consultants have been consulted and appropriate specialized medical consultation is available or can be obtained by the program. Yes
8. The specific condition appears in the funded region of the prioritized list as determined by the Oregon Health Evidence Review Commission. Yes
9. The NWRNBS program has sufficient information to perform a fiscal analysis. Yes
10. The impact to NWRNBS contracted partners has been assessed. Yes (Contacted other programs, and only Saipan responded.)

The program gave X-ALD a qualified "yes" to move on to stage 3 review by the board.

Board analysis of category 2 criteria—

Criterion 1: *The population level public health benefits of screening outweigh the risks and harms.*

Discussion:

- Adrenal insufficiency is very common in childhood with the youngest affected kids diagnosed in the first six months of life. The 80 percent getting adrenal insufficiency outweighs the harm to families of getting potential information about other disorders.
- It's important to treat cerebral X-ALD early.
- It destroys a family if they don't know to watch for symptoms.
- It being X-linked means a female carrier will be identified with a high likelihood of adult-onset symptoms and no known treatment.
- Is it ethical to identify female carrier tangentially to the newborn screen? Mother alone will feel responsible.
- If child is diagnosed, mother is the carrier and faces possibility of devastating adult disease. Adult can plan for it if they know.
- Mothers of boys with the X-ALD gene are almost always carriers. They will be identified by default through the detection of their sons.
- If we have a boy with X-ALD who has a ten-year-old sister, we're not likely to recommend testing her. Would wait until she's eighteen and can decide on her own about testing. But we wouldn't be giving the mother the right not to know.
- If you do testing, at what point does carrier cease to have a choice?
- Never want to not test a child over collateral information about the mother.
- If son has X-ALD 95 percent of mothers will have it. Are we more concerned about females being incidentally identified without consent of individual?
- Any way to screen just male babies? Program response: Gender is sometimes not identified on specimens so this would be difficult to perform with accuracy. Logistically, this would be very difficult given the high volume.
- Does the carrier information create a risk that insurance will say X-ALD is a prior condition? (It has implications for life and disability insurance, some businesses, and the military.)

Criterion 2: *There is adequate capacity and expertise in the NWRNBS program to implement and maintain testing and reporting.*

Discussion: Program requires more resources.

Criterion 3: *There is adequate capacity and expertise in the NWRNBS program to implement and maintain follow-up and education for providers and parents.*

Discussion: More resources for this capacity and expertise would be needed.

Criterion 4: *The NWRNBS program has adequate fiscal resources for implementing the test, performing the test and conducting follow-up and education.*

Discussion: See above.

Criterion 5: *The population level incidence, prevalence, and disease burden are significant enough to merit screening.*

Discussion: Some felt that one to five cases per year is frequent enough to justify addition to the panel.

Criterion 6: *Diagnostic and specialty testing is available and accessible that allows a definitive diagnosis to be made.*

Discussion:

- Yes, if the program administers only first-tier testing and primary care physicians do second-tier testing.
- DNA tests are complicated to order. And insurance often doesn't cover out-of-house testing. Would be better to keep second-tier testing with the program. Program response: If the program keeps it in-house for five positive tests per year, is it worth raising the fee for all screenings?
- The program would have to have staff do follow-up to track down primary care doctors to see if the infant got the second-tier test.
- There are too many mistakes with doctors ordering the wrong DNA tests.

Criterion 7: *An effective treatment that is proven to result in clinically significant benefits is available and accessible.*

Discussion: Group agrees—Yes.

Criterion 8: *There is equitable care and treatment for the disorder.*

Discussion:

- Same equity concerns as with SMA. Also, if lab were to screen only males, that would be an equity concern. Equity concerns are not unique to this disorder.
- Some families will not be able to pay for genetic testing if they have to ask insurers to pay for it versus having the lab do it.
- Is this criterion important enough on its own to determine whether to add the condition to the panel?

- If condition is not symptomatic in the future, low income families might not do second-tier testing. A board member added that many of these families have Medicaid.
- Program: Program can estimate costs of second-tier testing. Would require a request to the legislature. Have to be reasonable about what they can ask primary care doctors to do.
- We're considering adding the condition because it has been added to the RUSP. Twenty-three states are already testing for it. Oregon generally gets insurance coverage.
- Program: Test kit now cost's \$80. Adding two more tests (SMA and X-ALD with no second-tier testing) along with other components will increase and perhaps nearly double the cost of the kit.

Criterion 9: *Addition of the disorder is not prohibitive to NWRNBS contracted partners.*

Discussion: No X-ALD cases in Saipan. No information from other partners.

Consensus check on proposal to add X-ALD to the screening panel with no second-tier screening by the program at this time.

(1=enthusiastic agreement, 2 = agreement, 3 = on the fence/neutral, 4 = serious questions or concerns but not going to block from moving forward, 5=no agreement, would block action)

Votes:

Amy Yang 2

Anna Dennis 2

Jill Levy-Fisch 1

Joanne Rogovoy 2

Marilyn Hartzell 2

Wannasiri (Awe) Lapcharoensap 2

Cheryl Hanna 1

Silke Akerson 4 (not present for vote, but offered her vote before leaving)

Phillip Dauterman 2

Consensus to add X-ALD to the screening panel.

Discussion:

- Program next steps—finish fiscal analysis and give to the legislature. Develop rule. Work with consultants.
- Are any states controlling for testing only males? The program will get information for the next meeting.

12. Next steps

- Reminder of questions raised today for further discussion:
 - How to address SMA carrier question?

- What about testing only males for X-ALD?
 - Who should do second-tier testing for X-ALD?
 - What are the issues around equity for all screenings?
 - What are the broader impacts (programmatic) of follow-up tests?
- The board suggested they would like to complete their work on X-ALD and SMA in time for the 2021 legislative session.
 - A phone meeting of the board will be convened in **late April** to finish SMA and X-ALD discussions informed by the program's information gathering.
 - A doodle poll will be sent out with limited date options.
 - These minutes will be reviewed/adopted by the board at the April conference call meeting.
 - The board will look to finalize disorder removal protocol and criteria at their **June/July in-person** meeting and if time allows, evaluate Gaucher and Fabry disorders for potential removal from the screening panel.
 - Chris Biggs has a conference in April that should provide more information from other states about X-ALD and SMA for the board consideration at the June/July meeting.
 - Chris noted one correction to the draft protocol distributed to the board: item 6 should be "not result in harm." The program will revise the draft criteria for removal of disorders and redistribute it to the board.
 - The program will send out a doodle poll with three potential dates for this meeting.
 - The board will need to provide feedback on the draft legislative report by early July in order for OHA to do its internal review process and meet the September 15 deadline for submittal to the legislature. Oregon Consensus will develop an early draft in spring for the board to review after the next conference call.

Adjourned