Northwest Regional Newborn Bloodspot Screening Advisory Board

Wednesday, May 31, 8:30 am–12:30 pm PST
and
Thursday, June 8, 1:30 pm–3:00 pm PST
Videoconference

FACILITATOR SUMMARY: May 31, 2023 MEETING

Program and Legislative Updates

Patrice Held, Program Manager, shared relevant program updates with the board and status of any legislation related to the program. Highlights include:

- John Fontana has retired from his position as the NWRNBS lab director. The interim director is Dr. Luedtke, and Akiko Saito will serve as interim business director.
- The program implemented screening for X-ALD as of January 1, 2023, per the advisory board's direction and program approval.
- Overall, a lot of work is being accomplished within the program and there is a lot going on!
    - A question was asked about community outreach to consumers about specimen tracking and the LIMS system.
- Re: legislation related to the program, the following bills were still being tracked at the time of this meeting:
    - SB 5526, which would ratify fee changes

- HB 2617, which would put new terms and conditions on the operation and scope of the advisory board as well as waive the fee for families paying out-of-pocket costs for screening
- HB 2608, which would draw from the General Fund to cover costs
- HB 2927, which would establish a statewide steering committee on sickle cell disease

RUSP Updates and Implications for Advisory Board Work
Sarah Viall, clinical consultant for Oregon Newborn Screening Program, shared information about the RUSP process, rounds of review, and specifically the status of Krabbe in that national system. Sarah shared that the Krabbe discussion was controversial and difficult for the panel to reach a conclusion on whether to add it to the panel. In the end, the vote was split 7-yes and 7-no, and the disorder was not added to the RUSP.

Board Business—Disorder Review: MPS II (Hunters Syndrome)
The board was reminded of the protocol and process for adding and removing disorders from the NWRNBS screening panel. The board heard an overview of MPSII and was able to ask questions of technical consultant, Emily Singh. Materials had been sent in advance of today's session to allow the Board time to review the technical information.

Board questions about MPS II (Hunters Syndrome) included:
Q: No definitive treatment, but partial?
A: Yes, correct. There is nothing 100 percent curative or preventative. But working on treatments in research. Nothing approved yet by FDA.

Q: Chance of death with transplant is 12 percent?

A: Yes

Q: Heterogeneity of outcomes with same genetic defect. Why do some have different responses to the same gene defect?

A: Testing can help but there is not perfect clarity. There is a large number of variants within IDS (the gene) that can lead to the clinical phenotype. Detection of all variants is not guaranteed with current approaches. Even then, variants of "uncertain significance" of the clinical phenotype will occur..

Q: Pseudo-defect: More common than actual condition?

A: Yes, correct.

Q: Wouldn't this still be good information for the families to have?

Q: Equity issues you noted re: geographical, financial barriers to access to treatment, sex disparity. You also mentioned small numbers. What about race/ethnicity data?

A: Gap appears to be closing, but previously, Asian ancestry was associated with a higher risk for disease (but does not seem to be borne out as we continue to look at newborn screening data). No ethnic breakdowns readily available or documented at this time.

Q: Oregon is a duplicate/triplicate state for testing. How does that compare to or factor in when looking at other states?

A: Not able to find data that would suggest any implications for MPS II screening.

Q: What about preemies?

A: The screening algorithm would not need to be modified for preemies.

Q: ERT is lifelong treatment?

A: Yes. Until/unless a new treatment becomes available.

Note: Illinois and Missouri are testing.

Q: Cost of enzyme treatment for uninsured patients. Is funding available?

A: (Board member response) Yes, usually you can get Medicaid to cover, especially if it's on the screening panel. Some organizations have care management that helps facilitate access/coverage for treatment.

Q: Treatment for stem cell—is that included in consideration of treatments?

A: (board member response) The funding line for the prioritized list no longer applies to children on OHP. So this is no longer a barrier. Healthier Oregon program—kids without immigration status will qualify for full coverage.

Patrice reviewed the Stage II, Category 1 criteria on behalf of the program and determined that all criteria were met with a "yes" response; however the first criteria related to the condition being well defined in newborns, which was not as conclusive as the other criteria.

The advisory board reviewed several of the State II, Category 2 criteria. The following bullets reflect the dialogue:

- Concerned with fiscal burden to the program for requiring this test. Do we face a tradeoff with other needs?
- Alternatively, the opportunity to support quality of life for even one child is worth pursuing. The aim is an easier outcome for all.
- Positive predictive value—is that high or low? Five referrals leading to 1 positive and 4 false positives is pretty low.
- Certainty for developing symptoms is low.
- Informed/self-determination of families is important.
- Comparison to other disorders on the panel: it falls in the middle of these disorders on the existing panel.
- Re: expertise and capacity for testing—yes to both.
- If fee the fee increases, long term costs should be considered, including small business impacts

Action: The Advisory Board ran out of time to conclude its review of the disorder and agreed to schedule a follow-up meeting in June to complete the work. The meeting was scheduled for June 8 at 1:30 pm PST.

FACILITATOR SUMMARY: June 8, 2023 meeting

(Continued) Review of MPS II Disorder

The advisory board continued its review, with the following summary of comments / dialogue:

- Adding this to the panel—does this create risks associated with cost to the program over time?
    - Program response: Fee increases over time are a concern, but this issue is not just linked to MPS II. It is a broader system issue.
- Small business concerns—pre-purchase of screening kits is prohibitive for some birthing practices.
- Concern for sustainability on fee for service.
- What conditions will be important for us to say “yes” to recommending adding this to the screening panel?
- Is diagnostic and specialty testing available—yes, and usually it is covered by insurance, in addition to sponsoring programs that will often offer this genetic testing for little or no cost if not covered by insurance.
- Parents/families—most often would rather know vs. not know.
- Are there clinically significant benefits from treatment? Does improve the situation but does not halt the disease. It does minimize impact and extends mobility time.
- Equitable care and treatment? Likely treatment is in the form of infusions and this can create equity challenges related to transportation and for access for rural families. That said, this treatment could be accessed via mobile stations.

- Is there an impact to contracted partners of the Oregon lab? Not known at this time, but likely not a major impact given fewer partners than previously.
- Enzyme replacement is considered experimental—Is this a concern related to OHP? Could this be a barrier?

Conclusion
The advisory board reached a strong consensus to recommend adding MPSII to the Newborn Screening Panel. Votes were as follows on a consensus scale of 1 (full agreement) to 5 (no agreement):

Cheryl Hannah—2
Marilyn Hartzell—2
Andrea Keating—2
Awe Lapcharoensap—2
Liz Powers—1
Joanne Rogovoy—2
Kara Stirling—3
Amy Yang—2

Public Comment
There was no public comment offered during today's meeting.