Northwest Regional Newborn Bloodspot Screening Advisory Board
September 6, 2023
8:30am-12:30pm PST

In attendance:

- Marilyn Hartzell, family member of a person affected by a disorder on the Newborn Screening Panel
- Dr. Awe Lapcharoensap, Statewide Association of Pediatricians
- Dr. Dawn Mautner, Medicaid/Medical Director representing insurance
- Sherly Paul, Home Visiting Nurse
- Kara Stirlling, Providence, representing hospitals
- Dr. Amy Yang, Geneticist
- Patrice Held, Newborn Screening Program Manager
- Kasfian Khan, Program staff
- Akiko Saito, Administrative Director PHL
- Jacqui Umstead, Newborn Screening follow up team member/health educator
- Jess Scott Schwoerer, technical consultant on GAMT deficiency

FACILITATOR'S SUMMARY

Program Updates -

Patrice Held, Program Manager, provided the following updates*:*

- *Slides were shared and the PPT was provided as follow up to the Board and posted on the program website. Covered topics -*
    - Internal operations
    - 2023-24 Strategic planning with APHL
    - Dr. Charlene Lai welcomed as new Board member on behalf of Oregon Pediatrics Association

    - Still recruiting for advocacy organization and representative working with the OHPL newborn screening lab. Please recruit and send referrals our way!
    - End of Legislative session: fee ratification is complete, now in statute. Other bills did not move forward but may be revived in next legislative session.

GAMT Deficiency Review

Information was shared from technical consultant and expert in GAMT deficiency Jessica Scott Schwoerer. Read-ahead materials were provided to the Advisory Board prior to the meeting. Slides with a summary were shared that reflected a summary of the report materials. Some highlights from the overview included:

- Newborns screened/diagnosed through pilot studies indicate this is a rare disorder.
- Assay development would be needed.
- The technology is available and could work within the existing flow of the laboratory and the state of Oregon's short term follow up flow.
- Re: long term follow and access to treatment: it is available and expertise exists. In general treatments, medically necessary supplements distributed at a metabolic clinic, are covered by insurance.
- Oregon rural and frontier communities do present barriers to access to care. Telehealth can help some, but continued dietary management presents a potential barrier.

Questions/ comments from Board members included:

- Question re: treatment. Symptoms? Improvements:
    - Neurologic - developmental delays, seizures, movement disorders, behavioral concerns available evidence suggests improvements will occur if you start treatment particularly seizure control. But no reverse of all health concerns that have already occurred. Usually siblings identified earlier in life have better improvements (infancy and toddlers had better than later diagnosis)

- Numbers of diagnoses are pretty small. Impacts ability to identify demographics associated with higher risk (race/ethnicity e.g.). Not a clear sense of condition being more prevalent in any particular group?
    - Correct. No specific population appearing more at risk
- Other disorders in screening that are as rare as this disease?
    - Yes
- If we are going to test for this, does it overlap with any other disorders? Meaning will others potentially show up with this screening?
    - The tabulate they will use in screening would not detect other potential deficiencies in that pathway. So, no.
    - Any other pilots pick up disorders incidentally? No.
- How easy is it to catch the disorder with this test? How accurate is this test? False positives/negatives?
    - False positives from the collective of all pilots is about 2:100k screened (estimate) but we would have to monitor to see how this number changes with increased screening. In Oregon, 1.26 false positives/year.
    - No false negatives were reported.
- Confirmation test available? How accurate?
    - Yes confirmatory is available and is very good, through metabolic testing, DNA testing, and MRI.
- Effects from treating a false positive?
    - Treatment is mild protein restriction and then supplements, shouldn't be harmful to a baby, but you also have time to determine if there is a false positive before initiating diet therapy.

Board Deliberation of GAMT Deficiency

Using the established review protocol, the Board deliberated on the considerations for whether to recommend adding this disorder to the screening panel.

CATEGORY 1: The following criteria were responded to by the Program:

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, getting there but still early.**
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. **Laboratory can / is developing Assay, could put in pipeline for FDA approval in combination with other tests**.
5. Diagnostic and specialty testing is available. **Yes**
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. **No longer a relevant criteria as written. Dawn offered to assist with revising the criteria to reflect current rules re: the Prioritized List.**
9. The NWRNBS Program has sufficient information to perform a fiscal analysis. **Yes - and fiscal impacts look to be not a concern.**
10. The impact to the NWRNBS contracted partners has been assessed. **With New Mexico, yes. No perceived challenges. Additional contracted partners not reached are Saipan, Guam, Navajo Nations and some military bases. Fewer contracted partners potentially changes the weight of this criteria and may be revisited.**

CATEGORY 2: The following criteria were discussed by the Board:

Criteria	NOTES
The population level public health benefits of screening outweigh the risks and harms.	Yes, enthusiastic. Science and practice suggests that yes the benefit is strong. To identify harms - potentially- false positive screenings effects, resources utilized for those screens. But the benefit of early intervention outweighs risks and harms. What would happen to older siblings or family members who were identified with the disorder given a new screening available? That too would be a benefit to older family members. Lower seizures, mobility improvements, etc. Adding to the screen could potentially help improve population level public health benefits.
There is adequate capacity and expertise in the NWRNBS program to implement and maintain testing and reporting.	Yes
There is adequate capacity and expertise in the NWRNBS program to implement and maintain follow-up and education for providers and parents.	Yes
The NWRNBS Program has adequate fiscal resources for implementing the test, performing the test and conducting follow-up and education.	Yes. Added costs for this would be low. (no additional instrumentation or staff needed). Low number of referrals to clinicians would likely keep increased costs to a minimum - but would need to be negotiated at a later time.

	RE: education and outreach - we have within-program PH nurse, follow up team, and contract with OHSU for health educators already connected to the program. Consider fee increases that are not currently covering new disorders added to the panel in recent/near future. *GAMT deficiency may get rolled into a combined FDA approved treatment and absorbed into kit costs. Small increased cost anticipated (Operating expense.)
The population level incidence, prevalence and disease burden are significant enough to merit screening.	Yes. Although a rare disease - burden is significant and interventions would have dramatic positive results.
Diagnostic and specialty testing is available and accessible that allows a definitive diagnosis to be made.	Yes
An effective treatment that is proven to result in clinically significant benefits is available and accessible.	Yes
There is equitable care and treatment for the disorder.	Challenge for all families with rare genetic conditions. Difficult for families to implement - education and procedure. Families have to be hands on with the treatment which can be challenging. Access to counseling, in - person visits. Any new condition screened for and treated for becomes a challenge for families, with ease over time. Knowledge and ability to take action is

	important. Re: coverage for counseling? Within OHSU, supplements distribution infrastructure is set up as a charge to insurance, no charge to Medicaid. Otherwise this would be more challenging. Potential equity challenge getting insurance to cover medical foods and supplements. Challenge with certain private insurance providers. Cost barrier could present an equity challenge. Alternatives, e.g. health cooperative coverage - challenging to work with given no appeals mechanisms for denial of coverage. Different from standard insurance, different rules apply.
Addition of the disorder is not prohibitive to NWRNBS contracted partners.	New Mexico was contacted, no barrier. Other contracted partners would need to be contacted to be sure. Guam, Saipan, Navajo Nations, and some military bases. That evaluation has not been completed. Considerations are: do they have a clinical geneticist? What would care look like? Where and how? Geographic spread:# of geneticists. Program can make a recommendation about confirmatory test needed and course of treatment with confirmed cases; long term care would be up to the individual entity and related health care system.

Conclusion: The Advisory Board reached a strong consensus to recommend adding GAMT deficiency to the Newborn Screening Panel. **All 1's from participating Board members on the call today.**

Public Comment: No public comment was provided during today's meeting.

Protocol and Process Review

The Board began a discussion of this topic and will engage in a more intentional, structured review to update the protocol before the next round of disorder reviews. Comments shared today included:

- Category 2 Criteria #8: Change or remove, is not reflective of current requirements.
- Review of technical information by consultants has been really helpful and accessible to non-technical board members.

The Board also offered the Program suggestions to fill open board representation:

- Every Life advocacy organization
- Navajo Nation area nurse consultant. Amy Yang provided a contact to the Program.
- Other general nurse practitioners who work with Navajo Nation. Awe will share contacts with the Program.
- Dr. Roberta Hunt - professor of sociology at PSU - good perspective on equity.
- End of term in 2024 - the current Charter suggests the terms end in July.
- For Board members who terms are ending, the Program welcomes referrals for replacement but this is not a requirement.

General discussion:

- Please refer black families who have had screening and treatment to me, Sherley, to help with health literacy.
- Board schedule going forward: put a regular meeting together? Vs. polling for the meeting every time?
    - Group generally agreed on the “5th” Monday cadence to avoid other carved out times for standing meetings.
    - To accommodate childcare/drop off - 9 am start and shorter meetings (9am-noon preferred by many).
    - Avoid Monday’s and Friday’s.

- **ACTION**: The group on the call agreed to meet on the 5th Tuesday, starting **Tuesday January 30th, 9am-noon**. A calendar invitation will go out.

The meeting adjourned at 12:30pm.