Newborn Screening Advisory Committee

Meeting Minutes

April 29, 2022
12:00 p.m.

Locations:
Microsoft Teams

Robert Bernstein Building, Room K-100
1100 W. 49th St, Room K-100, Austin, Texas 78756

Table 1. Newborn Screening Advisory Committee member attendance Friday, April 29, 2022

MEMBER NAME	IN ATTENDANCE
Kaashif Ahmad, M.D., M.Sc.	Yes
Beryl (Pam) Andrews	Yes (Arrived after roll call)
Khrystal Davis, J.D.	Yes
Titilope Fasipe, M.D., Ph.D.	Yes
Melissa Frei-Jones, M.D.	Yes
Alice Gong, M.D.	Yes
Charleta Guillory, M.D., M.P.H.	Yes
Tiffany McKee-Garrett, M.D.	No
Barbra Novak, Ph.D., C.C.C.-A.	Yes
Joseph Schneider, M.D.	Yes
Fernando Scaglia, M.D.	Yes (Arrived during Agenda Item 4)
Michael Speer, M.D.	No
Elizabeth (Kaili) Stehel, M.D.	Yes (Arrived during Agenda Item 5)

Agenda Item 1 - Welcome, introductions, and logistical announcements

Dr. Melissa Frei-Jones, Vice-Chair of the Newborn Screening Advisory Committee (NBSAC), convened the meeting at 12:02 p.m. and welcomed everyone in attendance. She introduced Sallie Allen, Texas Health and Human Services Commission (HHSC), Policy & Rules, Advisory Committee Coordination Office. Ms. Allen reviewed logistical announcements, called roll, asked members to introduce themselves, and determined a quorum was present.

Ms. Allen announced that the Advisory Committee Coordination Office has a new staff member, Jacqueline Thompson, and asked her to introduce herself to the committee. Ms. Allen then turned the floor over Dr. Frei-Jones.

Dr. Frei-Jones asked Karen Hess, Texas Department of State Health Services (DSHS), Unit Director, Newborn Screening (NBS) Unit, to introduce herself and other DSHS staff members. Ms. Hess introduced Lori Gabbert Charney, DSHS, Section Director, Maternal and Child Health Section, and called on Dr. Debra Freedenberg, Dr. Susan Tanksley, Aimee Millangue, Laura Arellano, Brendan Reilly, and Steve Eichner to provide introductions. Dr. Frei-Jones thanked advisory committee members and DSHS staff for joining the meeting.

Agenda Item 2 - Consideration of October 8, 2021, meeting minutes

Dr. Frei-Jones reminded members that program staff emailed the October 8, 2021 draft meeting minutes for review and asked members and staff for any edits or changes. Dr. Frei-Jones requested a motion to approve the October 8, 2021 meeting minutes. Aimee Millangue, DSHS, Advisory Committee Liaison and Ombudsman, NBS Unit, requested an edit for Agenda Item 1 and Dr. Joseph Schneider requested an edit for Agenda Item 12.

MOTION: Dr. Alice Gong made a motion to approve the October 8, 2021 meeting minutes. Dr. Gong then amended the motion to approve the October 8, 2021 meeting minutes with the edits presented for Agenda Items 1 and 12. Dr. Schneider seconded the motion. Ms. Allen conducted a roll call vote, and the motion carried with 9 approves, no objections, or abstentions.

Agenda Item 3 - Whole genomic sequencing in newborn screening

Dr. Frei-Jones announced that Rachel Lee, Ph.D., DSHS, Microbiological Sciences Branch Manager and NBS Technical Advisor, Laboratory Operations Unit, is unable to join the meeting, so the topic will be tabled for the next meeting.

Agenda Item 4 - Status of Memorandum of Understanding and Data Use Agreement with Medicaid and Vital Statistics

Dr. Frei-Jones introduced Susan Tanksley, Ph.D., DSHS, Unit Manager, Laboratory Operations Unit, to provide the status on the Memorandum of Understanding (MOU) and Data Use Agreement with Medicaid and Vital Statistics.

Highlights of the update included:

- The DSHS Center for Health Statistics matched screens against vital statistics and Medicaid rolls to determine whether a child is enrolled in Medicaid or not, or is Medicaid eligible.
- The DSHS Laboratory had an initiative to move the Medicaid matching process from the Center for Health Statistics to the DSHS Laboratory by updating their Memorandum of Understanding (MOU) with Medicaid and putting in place a data use agreement with vital statistics.
- Effective November 2021, the DSHS Laboratory has an active data use agreement with vital statistics which allows them to obtain vital statistics information needed for matching against newborn screening data. The Laboratory uses that information to match with Medicaid eligibility information.
- The Laboratory is learning the matching process and has access to vital statistics information, but it is retrospective information because they need to match it against newborn screening specimen information and the Medicaid enrollment. The process takes about four months after screening is complete.
- The MOU with Medicaid is not updated yet, but is in the signature process.
- The good news about having access to vital statistics information is that the Laboratory will be able to do more matches to try to determine how many

births are actually being screened on a regular basis, which is a question Texas has not been able to answer in the past.

Members discussed:

- If the laboratory will have additional staffing and compensation for taking over duties from another department.
- The matching process takes four months not because of the coding used to do the linkage process but the availability and completeness of the vital statistics and Medicaid data.
- The Laboratory is able to match all babies born, not just babies that are considered not under Medicaid.
- If matching includes hearing screens or if it only includes blood spot data.
- The Laboratory will be able to say they know how many babies did not get a first newborn screen in the future.
- What actions can be taken once it is determined that a baby did not receive a newborn screen.
- If the Laboratory has an obligation to follow up with providers of babies who did not get a newborn screen.
- If the Newborn Screening Program would recommend a screening for a 16-month old baby who has not received a newborn screen.
- Under the MOU and data use agreement, if NBS data could be linked with other Medicaid data such as whether they received vaccines.

ACTION ITEMS:

- The Laboratory will investigate if it would be possible to follow up with providers about babies that have not been screened.
- Once data is available, the Laboratory will report on how many babies they have identified as not screened.

Agenda Item 5 - Screened conditions status updates

Dr. Frei-Jones introduced Debra Freedenberg, M.D., Ph.D., DSHS, Medical Director, NBS and Genetics, and Ms. Hess to provide an update on screened conditions. Dr. Frei-Jones announced they would provide updates for Agenda items 5a, 5b and 5c,

consecutively. Dr. Freedenberg and Ms. Hess referenced the PowerPoint and handout, *Newborn Screening Update*.

Agenda Item 5a - Diagnosed cases

Highlights of the presentation included:

- Chart with diagnosed case data for 55 Recommended Uniform Screening Panel (RUSP) conditions from 2018 - 2021
  - Newborn Screening can pick up 10%-20% more diagnoses than what is included on the RUSP and are not included in the chart
  - Spinal Muscular Atrophy screening began June 1, 2021
  - X-Linked Adrenoleukodystrophy screening began August 5, 2019
  - The program also reports to parents on their baby's sickle cell trait status
- Critical Congenital Heart Disease (CCHD) data on diagnosed cases
  - Only receive reports on diagnosed cases and not screens
  - 1123 cases have been reported since screening began in September 2014 through March 2022
  - 125 babies were identified post-natal with pulse oximetry, so they could have been missed without CCHD screening
- 1295 confirmed cases of CCHD in Texas from September 2014-March 2022 with primary and secondary target conditions, which is slightly higher than the 1123 cases reported since babies may have been diagnosed with more than one condition
- 80 CCHD Reporting Facilities and how many cases they have reported
- 2020 Texas Early Hearing Detection and Intervention (TEHDI) Program newborn hearing screening data
  - About 34% of babies' parents did not provide consent
  - About 2% of babies 359,770 screened did not pass their most recent or final screen and needed follow-up
- 2020 TEHDI Program diagnosis data
  - About 25% of babies screened are cleared without hearing loss
  - About 2% have transient hearing loss
  - About 7% have permanent hearing loss

    - About 66% of babies have not been able to be diagnosed
- According to Centers for Disease Control and Prevention, about 1.7 of every 1,000 babies will have hearing loss, which would be about 610 babies compared to the 404 identified

Members discussed:

- For CCHD Reporting facilities data, if some of the facilities are duplicated due to different spellings and names
- Clarifying that the lost to follow up (LTF) and lost to diagnosis (LTD) numbers are included in the numbers for no diagnosis
- 2020 numbers may have been affected by the COVID-19 pandemic, but Texas LTF and LTD percentage is usually around 50%, so keeping the numbers where they are was impressive
- CCHD cases are mostly reported to the state via fax.
- If CCHD can be reported more efficiently using an existing electronic reporting system.

Agenda Item 5b - Spinal Muscular Atrophy (SMA) screening implementation

Highlights of the presentation included:

- 16 cases identified from June 2021 – March 2022 – all confirmed with clinical and molecular diagnosis.
    - 1 – with 1 copy SMN2
    - 7 – with 2 copies of SMN2
    - 6 – with 3 copies of SMN2
    - 2 – with 4 copies of SMN2
- Current recommendation is for treatment options for any baby with up to 4 copies of SMN2.
- Expect numbers for this time period to go up with a number of cases still awaiting confirmation and clinical evaluation.

Agenda Item 5c - X-linked Adrenoleukodystrophy (X-ALD) screening implementation

Highlights of the presentation included:

- 37 males hemizygous affected and 20 female heterozygotes from August 2019-March 2022
- Heterozygote Klinefelter (XXY)– 1 Case
- CADD (Contiguous ABCD1 DXS1357E deletion syndrome) – 1 Case
- Zellweger Syndrome – 10 Cases, 1 Carrier
- Peroxisome Disorders – 6 Cases
  - 1-Peroxisomal Biogenesis Disorder
  - 2-D-Bifunctional Protein Deficiency
  - 1-PEX6
  - 1-TREX1
  - 1-Aicardi Gouiterres Syndrome
  - 1- NAXE leukoencphalopathy-1

Members discussed:

- Following a NAXE leukoencphalopathy case

Agenda Item 5d - Electronic ordering and reporting

Dr. Frei-Jones introduced Brendan Reilly, DSHS, Health Informaticist, Laboratory Operations Unit, Laboratory Services Section, to provide update on Agenda item 5d. Mr. Reilly referenced PowerPoint and handout, *Electronic Test Ordering and Reporting.*

Highlights of the presentation included:

- Update on NBS Exchange Open Enrollment opportunity and project
  - Grant funding expired September 30, 2021
  - DSHS investigating additional funding opportunities for submitting entities
  - Initiated six projects and completed three; other three in progress or on hold.

- One additional ongoing HL7 Interface Project with another hospital system started after the grant funding period closed
- Security and password reset improvements to NBS Web Portal
- Electronic Ordering and Reporting (EOR) Project status:
  - New system to replace for NBS Web Portal
  - Consolidation with all DSHS Lab testing
  - Supply ordering
- Current Electronic test ordering and reporting capability
  - Increase achieved through open enrollment opportunity
  - Close to 25% of samples reported electronically
  - 175,000 orders and results
  - At least 140 submitters participating electronically

Members discussed:

- Applicable to blood specimens only.
- Whether there has been any further activity toward creating a centralized gateway solution.
- If the laboratory is following a standard reporting format to report back to hospitals or are reports custom for each hospital.

Agenda Item 6 - Congenital Hypothyroidism Pilot Project

Dr. Frei-Jones introduced Mr. Reilly to provide an update and referenced PowerPoint and handout, *Congenital Hypothyroidism Pilot Project.*

Highlights of the presentation included:

- Primary Thyroid-stimulating hormone (TSH) screening methods
- Pilot project goals with grant funding from the Centers for Disease Control and Prevention:
  - Collect Thyroxine (T4) and TSH levels on all specimens for 10 months
  - Evaluate primary screen T4 and TSH levels

    - Determine the optimal Congenital Hypothyroidism (CH) screening algorithm.
- Comparison of legacy/pilot algorithms by parameters
- Comparison of future algorithm options by diagnosed cases, possible missed cases and false positives over 10 months and estimated yearly DSHS cost
    - Option 1 - T4 only (based on 10% population cutoff by age at collection)
    - Option 2 - TSH only
    - Option 3 - T4 and TSH
    - Option 4 - TSH only – first screen and T4 and TSH – second screen
- Received feedback from endocrine specialists
- Program's preference is to move forward with Option 4 hybrid model
- Revised timeline since original timeline conflicts with delayed Laboratory Information Management System upgrade
- Considered questions to determine the option most appropriate for CH screening in Texas

Members discussed:

- Clarifying what pending cases meant on slide for legacy/pilot algorithm comparison

Agenda Item 7 - Future condition implementation updates

Dr. Frei-Jones introduced Dr. Tanksley and Dr. Freedenberg to provide an update on future condition implementation. They referenced the PowerPoint and handout, *Future conditions implementation update.*

Highlights of the presentation included:

- Status of conditions that have recently been approved or are under consideration to be added to the RUSP
    - Approved – Pompe and Mucopolysaccharidosis type I (MPS I)
    - Recommended for approval - Mucopolysaccharidosis type II (MPS II)
    - Evidence Review - Guanidinoacetate Methyltransferase Deficiency (GAMT)

    - Nominated for Review – Krabbe Disease and Congenital Cytomegalovirus (cCMV)
    - Not yet submitted – Other conditions such as Duchenne muscular dystrophy and other MPSs
- DSHS is working on plans and a timeline for implementing Pompe, Mucopolysaccharidosis type I (MPS I) and MPS II with an estimated go live date around May 2024
- Funding status:
    - NBS Preservation Account
    - Centers for Disease Control and Prevention (CDC) Building Capacity Grant
    - Other lab grant for building retrofit
    - Fee increase- Ongoing testing starting January 1, 2025

Members discussed:

- The maximum that could be collected from Medicaid for the NBS preservation account is $12 million per biennium and how much is currently in the account.
- If there are plans to take out from the statute the language about a maximum that could be collected for the NBS preservation account for the next legislative session.
- Whether Texas will be testing for all the conditions on the RUSP after implementing testing for Pompe and MPS I.
- Doctors always think what could have happened if a child could have been screened for a condition through newborn screening instead of being diagnosed later, so seeing other conditions coming down the pipeline like GAMT is good.

Since he would not be available later, Dr. Joseph Schneider requested to proceed to Agenda Item 10.

Agenda Item 10 - Health Information Technology (HIT) Subcommittee Reporting

Dr. Frei-Jones introduced Dr. Joseph Schneider, Subcommittee Chair. Dr. Schneider provided an update on HIT Subcommittee activities.

Dr. Schneider stated that the subcommittee did not meet since the last NBSAC meeting, but one of the initial purposes of the committee was to try to push forward the connection with vital statistics. He added that he would like to meet with Mr. Reilly and others during the coming months to see what's next. He then opened the floor for questions or comments.

Members did not have a discussion for this agenda item.

Returned to Agenda Item 8.

Agenda Item 8 - Sickle Cell Subcommittee Reporting

Dr. Melissa Frei-Jones introduced Dr. Titilope Fasipe, Subcommittee co-chair. Dr. Frei-Jones and Dr. Fasipe referenced the handout, *Sickle Cell Subcommittee Meeting Minutes.*

Highlights of report included:

- Subcommittee last met December 7
- Worked on updating list hemoglobinopathy consultants and creating an adult list of hemoglobinopathy consultants.
- Discussed updates to a survey created by Dr. Fasipe that the DSHS staff was going to send out to all of the individuals on the pediatric list, as well as the people who participate in the Hemoglobinopathy consultants group, to identify the adult providers they send patients to when their patients age out of the pediatric program
- Will review survey results at the next subcommittee meeting
- Reviewed Action (ACT) and FACT sheets
- Future task as a standing subcommittee is to review and update the lists and information sheets annually
- Will discuss long-term follow up forms and submitting process next and the possibility of electronic reporting

Members did not have a discussion for this agenda item.

Agenda Item 9 - Rare Diseases Subcommittee Reporting

Dr. Frei-Jones introduced Ms. Khrystal Davis, Subcommittee chair. Ms. Davis referenced the handout, *Rare Diseases Subcommittee Meeting Minutes.*

Highlights included:

- Subcommittee last met on December 28
- Dr. Amy Brower, Associate Project Director of the Newborn Screening Translational Research Network (NBSTRN) and Co-Principal Investigator, presented to the subcommittee and provided an overview NBSTRN's work
- NBSTRN operates under contract from the National Institute of Child Health and Human Development (NICHD)
- Dr. Brower stated there is one project looking at genomics utilization in NBS, including policy and implementation considerations.
- Dr. Brower recognized the subcommittee in its ability to be future facing and looking at new capabilities to screen and diagnose as well as new technologies to treat rare conditions and invited participation in the NBSTRN forum, which is open to the public
- Subcommittee discussed that there were not changes in the criteria in nominating and recommending conditions for RUSP inclusion but a clarification of the questions that would be asked during evidence review.
- Program staff will send enabling statute for the NBS program and NBS preservation account to the subcommittee and identify speakers who could give more direction on what the statute allows
- Subcommittee future plans include inviting presenters such as Dr. Sanjiv Harpavat and members of the public who provided public comment on Noonan syndrome and discussing logistics for a pilot study of biliary atresia and making a recommendation in support of the pilot study.

Members did not have a discussion for this agenda item.

Agenda Item 11 - Hearing Screening Consent Form

Dr. Frei-Jones introduced Ms. Karen Hess and Dr. Debra Freedenberg. They referenced the PowerPoint and handout, *Consent to Release Information Form*.

Highlights of the presentation included:

- The *Consent to Release Information Form* was finalized earlier this year and is now posted on the NBS website under newborn hearing.
- Hearing screening works with OZ Systems, and they have outreach programs. Every other month, they meet with the certified hearing sites.
- Meetings are mandatory and help with newborn hearing screening in the state.
- At the January meeting, they announced the release of the new form.
- At the May meeting, they will announce the final deadline for adopting the form, December 31, 2022.

Members discussed:

- If consent can be tracked now and compared with the numbers after the form is adopted to see if the new form makes a difference.
- Who is getting the consent and at which time are they getting the consent.
- Enabling legislation for hearing is different than that for dried blood spots.

Agenda Item 12 - Public Comment

Ms. Allen read the public comment logistical announcements and called on Ms. Miranda McAuliffe, ALD Alliance, to address members. Ms. McAuliffe shared information about a quarterly consortium for state lab screening and stated that ALD Alliance now runs a care package program for families receiving a diagnosis of ALD through newborn screening.

Agenda Item 13 - Future agenda items, next Meeting date, and adjournment

Dr. Frei-Jones opened the floor for discussion of future agenda items and stated the next meeting was scheduled for July 8, 2022.

Members discussed:

- Whole genomic sequencing in newborn screening
- NBS preservation account
- Distribution of Medicaid funding received from newborn screening kits
- Standing items
  - Screened conditions updates
  - Future conditions implementation updates
  - Subcommittee reports
    - Sickle Cell
    - Rare Diseases
    - Health Information Technology
- Action item follow up list:
  - Continuity of operations
  - Updated funding request for meeting timeliness goals and the estimated cost of a 7-day working lab, to include follow up care costs if babies are not screened
- New member solicitation
- Continuing hybrid meetings

Dr. Frei-Jones thanked members and adjourned the meeting at 3:49 p.m.

Below is the link to the archived video of the April 29, 2022 NBSAC meeting that will be available for viewing approximately two years from date meeting was posted on website and based on the DSHS records retention schedule.

https://texashhsc.new.swagit.com/videos/170910