

**Newborn Screening Advisory Committee  
Final Approved Meeting Minutes  
April 16, 2021  
12:00 p.m.**

**Location: Microsoft Teams Live Event**

Table 1: Newborn Screening Advisory Committee attendance Friday, April 16, 2021.

| MEMBER NAME                     | IN ATTENDANCE                  |
|---------------------------------|--------------------------------|
| Kaashif Ahmad, M.D., M.Sc.      | Yes                            |
| Beryl (Pam) Andrews             | Yes (arrived at agenda item 2) |
| Nancy Beck, M.D.                | Yes                            |
| 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.     | Yes                            |
| Barbra Novak, Ph.D., C.C.C.-A.  | Yes                            |
| Joseph Schneider, M.D.          | Yes                            |
| Michael Speer, M.D.             | Yes (arrived at agenda item 4) |
| Elizabeth (Kaili) Stehel, M.D.  | Yes (arrived at agenda item 2) |

**Agenda Item 1: Welcome and Introductions**

Dr. Alice Gong, Chair of the Newborn Screening Advisory Committee (NBSAC), convened the meeting at 12:02 p.m., welcomed everyone in attendance and provided opening remarks.

**Agenda Item 2: Committee Business Logistics**

Dr. Gong introduced, Ms. Sallie Allen, HHSC, Policy & Rules, Advisory Committee Coordination Office (ACCO). Ms. Allen introduced and announced that Eric Owens, a new facilitator, will assume the role at the next meeting. Ms. Allen reviewed logistical announcements, called roll, and determined a quorum was present. Dr. Gong thanked Ms. Allen for her work and welcomed Mr. Owens.

Dr. Gong introduced David R. Martinez, Director, DSHS Newborn Screening (NBS) Unit and asked him introduce himself and other DSHS staff members. Prior to the

introductions, Mr. Martinez requested a moment of silence in memory of Ginger Scott, Lead Nurse, DSHS NBS Unit. Mr. Martinez then called on DSHS staff, Dr. Debra Freedenberg, Aimee Millangue, Laura Arellano, Ryan Hutchison, Karen Hess, Dr. Susan Tanksley, Brendan Reilly and Dr. Rachel Lee, to introduce themselves.

### **Agenda Item 3: Review and Approve Meeting Minutes for January 22, 2021**

Ms. Allen reminded members they received a copy of the draft of the January 22, 2021 meeting minutes and asked if members had any edits or changes. Dr. Gong asked for clarification on the multiplex Polymerase Chain Reaction (PCR) test for Spinal Muscular Atrophy (SMA); Dr. Tanksley provided her with an explanation. Dr. Joseph Schneider requested if the action items noted in the January minutes are not addressed at the April meeting, that they be carried forward and noted in the minutes of the April meeting as action items for discussion at future meetings.

**MOTION:** Dr. Schneider made a motion to approve the January 22, 2021 meeting minutes. Dr. Guillory seconded the motion. Ms. Allen conducted a roll call vote, and the motion carried with no objections or abstentions. One member was not present at the time of the roll call vote.

### **Agenda Item 4: Spinal Muscular Atrophy (SMA) screening implementation update**

Dr. Gong introduced and turned the floor over to Susan Tanksley, Ph.D., Laboratory Operations Unit Manager, DSHS Laboratory, Debra Freedenberg, M.D., Ph.D., Medical Director, DSHS NBS Unit and Karen Hess, Genetics Branch Manager, DSHS NBS Unit.

#### **Dr. Tanksley and Dr. Freedenberg stated:**

- Funding approval was received on July 2, 2020 to implement SMA
- Obtaining full-time employees and ordering equipment
- How the lab will be screening for SMA using a two-tiered approach
- Laboratory Information Management System (LIMS) has been modified
- Still on target for early June implementation
- Clinical care algorithms are being finalized so that providers have the guidance of where to refer the patients and which diagnostic tests are required, including the development of ACT and FACT sheets
- Lab equipment is being modified and configured
- Reporting procedures are being determined
- Held the first of two meetings with specialists in the care of SMA to help with laboratory and clinical care coordination protocols and algorithms
- Education brochure has been developed as well as online provider education module as part of Texas Health Steps
- Grand Rounds for health care providers has been scheduled
- Internal education for staff is planned
- Educational insert notices will be sent out with NBS kits
- Press Office will release information closer to the go-live date

- Due to the accuracy of the test, there will not be a lot of conditions in the differential diagnosis for the DNA findings that will be identified
- Will have some families and parents come speak to give parent and family perspective on SMA screening

**Members discussed:**

- If the baby has a genetic mutation that causes SMA other than the homozygous deletion on exon 7, the first-tier SMA test will not pick it up
  - If SMA is not picked up in the first tier, it will not go through second-tier testing
  - About 5% of all SMA cases will not be picked up through the SMA assay
- Whether committee members can volunteer to receive test reports to make sure they go through correctly and are understandable

**Agenda Item 5: Future condition implementation updates**

Dr. Tanksley and Dr. Freedenberg referenced the PowerPoint/handout, *How to determine which conditions to screen for and how to add them?*

**Dr. Tanksley and Dr. Freedenberg stated:**

- No updates on Pompe and Mucopolysaccharidosis Type 1 (MPS1)
- Mucopolysaccharidosis type 2 (MPS2)- Hunter Syndrome will be reviewed for sending to formal evidence review for adding onto the Recommended Uniform Screening Panel (RUSP) at the next Health and Human Services (HHS) Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) meeting
- Possible candidate conditions for NBS include Fragile X, Duchenne’s Muscular Dystrophy, Neuronal Ceroid Lipofuscinosis 2 (CLN2), Cytomegalovirus (CMV), GAMT deficiency, is a short version of the list, but there are several conditions being considered, as many individuals want them added
- Calls are routinely received from individuals with various conditions who want to know how to request conditions be added. They are referred where to go, what help is available and what advice is available if they wish to proceed with the nomination process
- Conditions that are on the panel that are not being screened for are Pompe and MPS1, and SMA is being added, and there are two secondary conditions that by law, Texas is exempted from, Galactokinase and Galactosephosphotransferase

**Members discussed:**

- Whether MPS2 is on a similar platform that can be multiplexed with Pompe and MPS1 when they are added to the Texas NBS panel
- Some states are doing multiple lysosomal storage diseases at the same time

**Agenda Item 6: Screened conditions status updates**

Dr. Tanksley and Dr. Freedenberg provided updates on screened conditions. Dr. Freedenberg also referenced the PowerPoint/handout, *Texas NBS X-Linked Adrenoleukodystrophy (X-ALD) Case Update, August 2019 – March 2021*.

**Dr. Tanksley and Dr. Freedenberg gave updates on:**

- February winter storm
  - How the weather impacted the amount of work that was able to be performed in the laboratory and ability to perform routine operations starting Thursday, February 11
    - Some days of limited testing and others of complete shut downs
    - Disruptions to courier services through Wednesday, February 17
    - Not being able to utilize back up testing services in other states to get specimens tested that were collected through February 18 due to weather conditions
    - Staff were able to go in on Saturday, February 20 and Sunday, February 21 to resume testing and do recovery
  - Updates shared with health care providers with through the listserv, Texas Pediatric Society (TPS), Texas Hospital Association (THA), and Texas Medical Association (TMA)
  - There were a large number of unsatisfactory specimens
  - All specimens were tested, even if collection was unsatisfactory
  - First time utilizing some of continuity of operations planning
  - Collecting information for an after-action report to improve continuity of operations planning
  - Challenges for Clinical Care Coordination staff, including loss of electricity and water due to storm, inability to travel, loss of lead nurse, COVID-19
- X-ALD cases
  - 22 males, 13 females
  - 1 CADD
  - Zellwegger
    - 5 cases
    - 1 carrier
- NBS Exchange Open Enrollment
  - Funding opportunity for health care providers to implement an HL7 interface to electronically order NBS testing of blood spot specimens and electronically receive reports back closed on April 1
  - In various stages of contracting process with six applications received
  - Additional providers will lead to a huge increase in electronic orders and not having to enter orders manually

Dr. Tanksley introduced and turned the floor over to Brendan Reilly, Business Analyst, DSHS Laboratory. Mr. Reilly addressed Dr. Schneider's question from the January 22, 2021 NBSAC meeting on the rate of first screens linked to a second screen sample and clarified the limitations to the data.

**Members discussed:**

- How power outages impacted testing and reporting
- How well the program did under the circumstances
- If the committee could provide support for future emergency weather events
- If the freezing temperatures affected testing results
- Follow up for babies that might have been missed during the winter storm
- Only being able to confirm 85% of first screens are linked to second screens
- If the Laboratory could share at the next NBSAC meeting what hospitals and large practices are enrolled in the NBS Exchange, such as showing who is covered and who is missing on a map
- Concerns with private practices and pediatricians not applying

Steve Eichner, Health Information Technology Lead, DSHS Center for Health Policy and Performance, offered clarification about Medicaid funding for the NBS Exchange Open Enrollment.

**Agenda Item 7: Laboratory testing cost-estimate**

**Dr. Tanksley provided the laboratory testing cost-estimate.**

**Dr. Tanksley stated:**

- Cost-estimate for operating seven days a week was provided in response to a January 2016 Legislative Budget Board (LBB) request
- Exceptional Item requests could be received at time of any Legislative session
- The DSHS laboratory is testing specimens Monday through Saturday and calling out time critical conditions on Saturdays
- LBB wanted to know what it would cost for and how could the lab be able to reach the 95% recommended goal of meeting the timeliness measure.
- Cost to fund courier service for 95% of specimens to arrive at the lab within two days of collection, not the recommended 24 hours (over \$1 million)
- Cost for increasing the staffing levels to decrease turn-around times for specimens and space constraints (33 laboratory staff at around \$2,300,000; 4 Clinical Care Coordination staff at around \$180,000)
- Issues for operating seven days a week include retention of staff, increasing courier services, lab testing space, Information Technology support, having an operational building, having time to make LIMS changes and vendor support.

**Members discussed:**

- If it would be helpful for the DSHS Laboratory to prepare an updated funding request for meeting the timeliness goal for the next Legislative session.
- Budget estimate should include follow-up care costs if babies are not screened and future screening considerations.

**Agenda Item 8: Hypothyroidism screening update**

Dr. Tankley provided an update on hypothyroidism screening.

**Dr. Tanksley stated:**

- In December 2020, switched screening platforms
- With switch, decided to screen for both thyroid stimulating hormone (TSH) and thyroxine at the same time instead of just screening for thyroxine
- Engaged in a two-year grant with the Centers for Disease Control and Prevention (CDC) for a pilot project to determine the best algorithm for picking up congenital hypothyroidism cases.
- It was expected and has been the case that there is a larger volume of results that have required retesting or confirmatory testing
- In evaluating thyroxine and TSH as analytes for detection of congenital hypothyroidism, need to be able to determine which cases have been diagnosed and which are cases cleared to have accurate information to base new algorithm upon
- Year two will begin October 1, 2021 and end September 2022
- Working with endocrinology experts, including NBSAC member Dr. Nancy Beck, to analyze specimen and case-level data to determine ideal processes to reduce the time to notification to Clinical Care Coordination
- Optimal algorithm will maximize the identification of primary congenital hypothyroidism cases and decrease the false positive rate
- False positive rate has increased with the reporting of both analytes
- Will develop a detailed report and project plan and implement algorithm

**ACTION ITEM:** Program staff will provide the percentage of premature babies that might be in NICUs.

**Agenda Item 9: Overview of subspecialist follow up for time critical conditions**

Dr. Gong turned the floor over to Dr. Freedenberg and Ms. Hess. Dr. Freedenberg and Ms. Hess referenced the PowerPoint/handout, *Diagnosed cases and time critical conditions*.

**Dr. Freedenberg stated:**

- Only time critical condition that is not metabolic is Congenital Adrenal Hyperplasia (CAH)
- When a test is reflexed to DNA as the second or third tier, it will delay the ultimate timeliness
- Babies can screen positive on first screen but not in second—prompts consultation with specialist
- Ratios are standard for NBS

**Members discussed:**

- Aim is to pick up all diagnoses with high numbers of presumptive positives
- If meeting timeliness measures affects reporting of time critical conditions
- If locale delays reporting, treatment or diagnosis

- Process for getting number of diagnosed cases and workload for specialists
- Laboratory recognizes the large number of presumptive positives and is working on reducing false positives and evaluating cutoffs to improve the specificity of laboratory testing.

### **Agenda Item 10: Newborn Hearing Screening Consent Form**

introduced and turned the floor over to Dr. Elizabeth Kaili Stehel, committee member. Dr. Stehel referenced the handout, *Proposed Revised Newborn Hearing Screening Consent Form*.

#### **Members discussed:**

- Dr. Stehel's proposed edits to the consent form
- Clarifying consent to share information with Texas Early Hearing Detection and Intervention (TEHDI) and Texas School for the Deaf
- Length of form
- Wording might lead parents to think that they do not have to share results
- No consent is needed to enter results in TEHDI Management Information System (MIS), just for including identifying information
- Conveying that second step is needed only if baby does not pass
- Revised form's fifth-grade language level, bullet points and white space instead of paragraphs make it easier for the reader
- Having release signed by both parents, regardless if results are pass or fail
- Might be helpful to remove next steps, especially steps that would be applicable only if there is confirmed hearing loss
- Whether the identifying information that is entered in TEHDI is the birth mother's or baby's, and who could sign form
- If form is required to provide consent to notify providers
- Challenges with facilities having their own forms and processes even if the new Hearing Screening Rules require the consent form to be signed before the baby is screened
- Goal of form was to simplify consents across the continuum of care
- No mechanism for parents to fill out form again to change permissions after discharge from hospital
- Providing consent during birth screening allows for risk factor monitoring and updating TEHDI for late-onset hearing loss up to age five
- Providing form instructions for standardizing its use among providers

Dr. Gong called for the establishment of an ad hoc subcommittee to create a consent to release hearing screening and contact information form and appointed subcommittee members.

#### **Members of the subcommittee:**

- Dr. Elizabeth Stehel (Chair)
- Dr. Michael Speer (Ex-officio)
- Dr. Alice Gong (Ex-officio)
- Dr. Barbra Novak

- Dr. Tiffany McKee-Garrett

### **Agenda Item 11: Legislative Update**

Dr. Gong turned the floor over to Mr. Martinez.

#### **Mr. Martinez stated:**

- The 87<sup>th</sup> Session of the Texas Legislature began January 12, 2021
- ment Affairs (GA) staff have been working since November to review bills filed by legislators to identify legislation that will have an impact to DSHS
- Subject-matter experts from across the agency are then utilized to help in the analysis of these bills so that GA staff can provide meaningful feedback to legislators and prepare for potential legislative committee meetings.
- As of April 15, DSHS is tracking 657 bills. 132 have been assigned to the Community Health Improvement Division for further analysis.
- Legislation pertinent to Newborn Screening includes House Bill (HB) 2566/Senate Bill (SB) 580 - *Relating to newborn and infant hearing screenings*; this will increase reporting of newborn hearing screening results and managing parental consent.
- Reminder that committee members that they are free to participate in legislative advocacy during session as long as they represent themselves. Members must not engage in legislative advocacy of behalf of the committee or the department without prior authorization from DSHS.
- GA staff will attend the next meeting to give a final breakdown of the bills that passed and plans for implementation.

### **Agenda Item 12: Critical Congenital Heart Disease (CCHD)**

Dr. Gong turned the floor over to Ms. Hess. Ms. Hess referenced the PowerPoint/handout, *Newborn Screening CCHD Update*.

#### **Ms. Hess stated:**

- Targeting babies diagnosed post-natal with pulse oximetry (ox) since they would appear healthy at birth
- Of the babies diagnosed post-natal after a normal pulse ox, most had coarctation of the aorta and tetralogy of fallot.
- Doesn't appear that CCHD discriminates by ethnicity

#### **Members discussed:**

- Coarctation babies are not cyanotic.
- Babies who were not transported may have been delivered in a tertiary or quaternary center with or without a pre-natal diagnosis.
- Numbers of cases that were isolated cardiac anomalies and not syndromic
- Genetic testing can find things even if there are no physical abnormalities
- How the number of cases by reporting facility show there is underreporting
- Why there is underreporting

- Adding a new option to submit a diagnosis form online may help with the underreporting
- Having CCHD Subcommittee look at the data
- Status of recommending CCHD as a quality improvement project for the Texas Collaborative for Healthy Mothers and Babies (TCHMB)
- Status of CCHD Subcommittee's recommendation to educate hospital partners and clinicians on the importance of reporting by amending the CCHD Rules to clarify cardiologists are responsible for reporting

### **Agenda Item 13: Newborn Hearing Screening in Neonatal Intensive Care Unit (NICU)**

Dr. Gong introduced Dr. Tiffany McKee-Garrett, committee member. Dr. McKee-Garrett referenced the handout, *Newborn Hearing Screening in the NICU letter response from Dr. Hellerstedt*.

#### **Members discussed:**

- If the NBS Medical Director has reviewed and shared the protocol with TCHMB and the Regional Advisory Councils
- If protocols are not accepted as a NICU standard of care, at least they could be shared with NICUs as a quality improvement project

### **Agenda Item 14: Health Information Technology**

Dr. Gong introduced Dr. Joseph Schneider, committee member. Dr. Schneider referenced the handout *Health information technology letter response from Dr. Hellerstedt*.

#### **Members discussed:**

- Since Dr. Hellerstedt's response regarding the Integration and Data Exchange Center of Excellence (iCOE) was not specific to NBS, the committee should continue to push toward linkage with vital statistics.
- Dr. Schneider was invited to an American Public Health Laboratory call on NBS reporting standards and got to see some of the variability in reporting that exists across the country.
- Reviewing examples of some of the reporting differences and getting update of where the department is in terms of moving forward at a future subcommittee meeting.

### **Agenda Item 15: Sickle Cell Subcommittee Reporting**

Dr. Alice Gong introduced Dr. Melissa Frei-Jones, M.D., Subcommittee Co-Chair. Dr. Frei-Jones referenced the handout *Sickle Cell Subcommittee Meeting Minutes, March 31, 2021*.

#### **Dr. Frei-Jones stated:**

- Dr. Titilope Fasipe and Dr. Frei-Jones worked on editing the ACT and FACT sheets to address inconsistencies between the forms.
- There is one location on the pediatric list of hemoglobinopathy providers that Dr. Fasipe is trying to confirm with Galveston.
- They intended to verify with locations included on list that they provided the standard of care as listed in the National Institutes of Health (NIH) guidelines
- There is a movement within the sickle cell community to develop standards and set up Sickle Cell Disease Centers of Excellence
- A Centers for Medicare & Medicaid Services (CMS) report was shared with NBSAC members that gives a glance at data related to sickle cell patients who receive Medicaid and CHIP services.

Dr. Gong announced that another bill was filed, SB 1952, with a section that opens up opting out of newborn screening for any reason and takes out a religious reason for opting out. Dr. Tanksley stated an update that includes bills of interest will be prepared and provided to Dr. Gong, Dr. Speer, TMA, THA, TPS and March of Dimes.

**Members discussed:**

- Sickle cell ranks second in number of positive tests for NBS
- If there is a summary of bills related to NBS
- If there is a companion house bill to SB 1952
- Determining the number of sickle cell patients in Texas
- Percentage of sickle cell patients on Medicaid and CHIP and if it covers treatment
- Sickle cell legislation out of Representative Jarvis Johnson's office in Houston include a bill for a sickle cell registry (HB 3673, 87<sup>th</sup> Session) and a bill on Medicaid expansion to include sickle cell patients.
- If Medicaid bill will extend to sickle cell patients beyond childhood

**Agenda Item 16: Public Comment**

No public oral comment was received.

**Written public comment was received from:**

Cure SMA supports the newborn screening of SMA. Cure SMA and Texas supporters are grateful for the leadership of the Committee and for the steady work of the state laboratory, especially given the challenges of 2020.

**Agenda Item 17: Future Agenda Items/ Next Meeting Dte/ Adjournment**

Dr. Gong opened the floor for discussion of future agenda items and stated the next meeting was scheduled for July 23, 2021.

**Members discussed:**

- Legislative update from Government affairs office
- CCHD birth defects data

- Program staff will look into how Committee could provide support for future weather emergencies and disasters
- Action Item follow-up list:
  - Data linking for vital statistics
  - Medicaid funding
- Subcommittee updates from:
  - Hearing Screening consent form
  - Critical Congenital Heart Disease
  - Health Information Technology
  - Sickle Cell
  - Hearing Screening in the NICU
- Updated cost estimate for seven-day working lab
- Standing items:
  - Screened conditions
  - Future conditions
  - SMA update

Mr. Martinez informed members that the NICU Hearing Screening protocol was posted on the TEHDI and NBSAC Business websites.

Dr. Gong thanked members and adjourned the meeting at 3:43 p.m.

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Below is the link to the archived video of the April 16, 2021 Newborn Screening Advisory Committee (NBSAC) 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://texashsc.swagit.com/play/04172021-504/2/>