

**Newborn Screening Advisory Committee
Meeting Minutes
January 31, 2020
10:00 a.m.**

**Moreton Building, Public Hearing Room M-100
1100 W. 49th Street, Austin, TX 78756**

Table 1: Newborn Screening Advisory Committee attendance Friday, January 31, 2020.

MEMBER NAME	IN ATTENDANCE
Beryl (Pam) Andrews	Yes
Nancy Beck, M.D.	Yes
Khrystal Davis	Yes
Titilope Fasipe, M.D., Ph.D.	By Phone
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
Scott McLean, M.D.	Yes
Joseph Schneider, M.D.	Yes
Michael Speer, M.D.	No
Elizabeth (Kaili) Stehel, M.D.	By Phone
VACANT	

Table 2: Newborn Screening Advisory Committee attendance Friday, January 31, 2020.

GUEST NAME and ORGANIZATION	IN ATTENDANCE
Terese Finitzo, OZ	Yes
Kelly Coggin	Yes
Kevin Coggin	Yes
Caroline Grossman, Mirepoix LLC	Yes
Joel de Jesus, Mirepoix LLC	Yes
Kevin Jaglinksi, Grifols	Yes
Barbra Novak, Texas Children's Hospital	Yes

Agenda Item 1: Welcome and Introductions

Dr. Alice Gong, Chair of the Newborn Screening (NBS) Advisory Committee, convened the meeting at 10:00 a.m. Dr. Gong greeted everyone, introduced herself, and requested members, staff, and the public in the room introduce themselves.

Agenda Item 2: Committee Business Logistics

Dr. Gong turned the floor over to Ms. Stephanie Gutierrez, HHSC, Policy & Rules, Advisory Committee Coordination Office. Ms. Gutierrez reviewed logistics, called roll, and determined quorum.

Agenda Item 3: Review and Approve Meeting Minutes for October 11, 2019

Ms. Gutierrez requested a motion to approve the October 11, 2019 meeting minutes.

MOTION: Dr. Joseph Schneider made a motion to approve the October 11, 2019 meeting minutes. Dr. Charleta Guillory seconded. A roll call vote was taken for members over the phone. A voice vote was taken for members in the room. The motion carried with no objections and no abstentions.

Agenda Item 4: Chair Appreciation

Dr. Gong introduced and turned the floor over to Mr. Jeremy Triplett, Director, DSHS Maternal and Child Health Section.

On behalf of DSHS Commissioner Dr. John Hellerstedt and Associate Commissioner of the DSHS Community Health Improvement Division, Dr. Manda Hall, Mr. Triplett extended appreciation to Dr. Guillory for her dedication and service as Chairwoman of the Newborn Screening Advisory Committee. Mr. Triplett presented Dr. Guillory with a plaque in recognition of her outstanding service to the Committee and the state of Texas.

Agenda Item 5: X-linked Adrenoleukodystrophy (X-ALD) testing update

Dr. Gong introduced and turned the floor over to Dr. Debra Freedenberg, Medical Director, DSHS Newborn Screening Unit. Dr. Freedenberg referenced the PowerPoint and handout, *X-linked Adrenoleukodystrophy (X-ALD) Testing Update*.

Members discussed:

- Females who are carriers for X-ALD may have milder symptoms when they are older. Some women may have more significant disease and neurologic compromise due to skewed X-chromosome inactivation where the X-chromosome that carries the pathogenic variant is active in more cells than not
- Siblings of children with confirmed diagnosis are being referred to centers that have genetic counseling and the expertise to evaluate family members
- The methodology for the DNA testing is Polymerase Chain Reaction (PCR) Amplification and Sanger sequencing

- A third of the individuals identified as having X-ALD have the childhood onset cerebral form
- The NBS program can accept requests for DNA testing for families of newborns identified with a pathogenic variant of X-ALD by the NBS program
- Once a year the laboratory will re-assess if variants of uncertain significance (VUS) have been reclassified. At this point there is a national effort to assess out how newborn screening data can be submitted for input into public databases
- A proposal has been submitted to the Texas Pediatric Society's annual state meeting to educate pediatricians on X-ALD screening with DSHS co-presenting

Agenda Item 6: Screened conditions status updates

Dr. Gong turned the floor over to Dr. Freedenberg. Dr. Freedenberg referenced the PowerPoint and handout, *Newborn Screening Clinical Care Coordination Diagnosed Cases 2014-2018*.

Members discussed:

- The number of cases of Severe Combined Immunodeficiency (SCID) being 34 on the core, with various genes being identified
- Reconciling missed cases found clinically that had not been previously identified though NBS testing by re-testing specimens that meet timeframes and going through Quality Assurance (QA) processes, such as reviewing cut-off values
- There are metabolic geneticists in Texas who treat patients with Short-chain Acyl-CoA Dehydrogenase (SCAD) and want the information.
- The NBS Program considers input from biochemical geneticists
- The NBS Program does not have a mechanism for removing a screen from the Texas newborn screening panel
- The law requires the NBS Program to screen for core and secondary conditions
- Secondary target conditions are reported as a consequence of screening for core conditions
 - When the lab adopted the secondary target conditions, analyte levels were changed to find more of them

Agenda Item 7: Spinal Muscular Atrophy (SMA) screening implementation update

Dr. Gong introduced and turned the floor over to Dr. Susan Tanksley, DSHS, Laboratory Operations Unit Manager. Dr. Tanksley referenced the handout, *Special Provision 16, Limitation: Expenditure and Transfer of Public Health Medicaid Reimbursements, letter to the Governor and Legislative Budget Board*.

Dr. Tanksley stated:

- The laboratory is evaluating screening assays to be used to identify SMA
- The molecular assay will be multiplexed with SCID screening therefore reducing equipment and staffing needed to add SMA to the panel
- The lab has identified an assay that works well. Additional testing is being conducted before a validation study begins
- The lab has purchased reagents and are waiting for them to arrive to begin the evaluation of a second-tier assay. This assay can be used with existing equipment so additional purchasing is not needed, which would require further planning and lengthen the timeframe for implementation
- NBS Clinical Care Coordination has obtained grant funding to develop educational materials
- DSHS sent a letter to the Governor and Legislative Budget Board (LBB) to seek funding for the implementation of SMA
 - The Governor and LBB replied with questions to which DSHS responded

Members discussed:

- The funding request to the LBB is slightly lower than the original request
- The initial screen the lab is intending to implement is to pick up the DNA deletion and not point mutations, as recommended by national standards
- Implementation can begin 18 months after receiving funding, so the expected go live date has been pushed from January 2021 to a later date
- Language in a response letter should include how many children will not get diagnosed, the benefits of early treatment, and possible death may help expedite the funding request

Agenda Item 8: Future condition implementation updates

Dr. Gong turned the floor over to Dr. Tanksley. Dr. Tanksley referenced the handout, *Special Provision 16, Limitation: Expenditure and Transfer of Public Health Medicaid Reimbursements, letter to the Governor and Legislative Budget Board*.

Dr. Tanksley stated:

- The lab is requesting funding to conduct a space utilization study to identify additional space within the existing laboratory facilities to implement testing beyond SMA and to develop bioinformatics capabilities and speed up data analysis for genetic sequencing
- The lab is in the process of switching out equipment to change the technology and instruments used for the assays for Congenital Adrenal Hyperplasia (CAH), Hypothyroidism and Cystic Fibrosis to free up space for additional testing

- The lab is looking at the algorithms for screening CAH and hypothyroidism due to high false positive rates
- A second-tier test has been added to the CAH assay
- The lab is researching the best way to screen for hypothyroidism

Dr. Freedenberg updated the committee on the Newborn Screening Saves Lives Act, which has not been renewed. One consequence of this is that the authorization for the Advisory Committee on Heritable Disorders in Newborns and Children has expired.

Agenda Item 9: Proposed Newborn Hearing Screening Rules

Dr. Gong introduced and turned the floor over to Mr. David Martinez, DSHS, Director, Newborn Screening Unit. Mr. Martinez referenced the handout, *Draft of Title 25 Health Services, Part 1 Department of State Health Services, Chapter 37 Maternal and Infant Health Services, Subchapter S Newborn Hearing Screening*.

Mr. Martinez stated:

- The informal public comment period is intended for the program to receive feedback on the first draft rules
- The rules revisions are based on House Bill (HB) 2255
- The internal Texas Early Hearing Detection and Intervention (TEHDI) team is working with DSHS legal staff
- The rules have been streamlined, re-organized, and restated succinctly in plain language
- Added a section on consent at the beginning
- Emphasized all stakeholders on the continuum of care should utilize the TEHDI Management Information System
- The rules are scheduled to be presented at the May 2020 Executive Council meeting, published in July 2020 for formal public comment, then effective by October 2020
- The program is in the process of adjusting the rule based on feedback received during the informal public comment period

Members discussed:

- The best way the committee will view and provide feedback to the rule before it is published
- §37.503 Consent.
 - (F)(2) is not within the state's authority to determine
 - Mr. Martinez clarified that the 'center' referenced is the Statewide Primary Resource Center, not the location of a doctor's medical records

- (A) should be revised to state; No consent is needed to have a hearing screening but then explain why the consent is necessary
 - The verbiage is confusing when describing consent for written consent or releasing information in the event a screening is failed
 - Mr. Martinez clarified that the program will provide consent forms for hearing screening and sharing individually identifiable information to capture data over time, so providers can see that the family did consent to have their information identified in the system for providers to view
 - The draft of the DSHS consent form and revised rules will be posted for public comment
- Members requested changing the title of §37.503 “Consent” to state “Consent to Release Information”
- Members requested (b) be revised to state “for consent of information, birthing centers are responsible”

MOTION: Dr. Joseph Schneider made a motion that DSHS will provide the consent form and rules that incorporates the revisions based on the comments to be submitted for formal comment by the April 24, 2020 advisory committee meeting. Dr. Tiffany McKee-Garrett seconded the motion. A roll call vote was taken for members over the phone. A voice vote was taken for members in the room. The motion carried with no objections nor abstentions.

Agenda Item 10: Break

Dr. Gong announced a 15-minute break.

Agenda Item 11: Laboratory Testing/Newborn Screening Kit Fee Increase Impacts

Dr. Gong reconvened the meeting at 12:00 p.m.

Dr. Gong introduced and turned the floor over to Mr. Brendan Reilly, DSHS, Program Specialist, Laboratory. Mr. Reilly referenced the handout, *Impact of NBS Fee Increase*.

Members discussed:

- Physicians identifying more with clinics and being less likely to set up their own private practices

Agenda Item 12: Interoperability of Newborn Screening Programs

Dr. Gong turned the floor over to Mr. Reilly. Mr. Reilly referenced the PowerPoint and handout, *Interoperability for Newborn Screening Programs*.

Members discussed:

- Getting the detailed plan of interoperability to members for review
- No progress has been made with integrating data with vital statistics

- A single source of relaying information across the state with consideration of Clinical Laboratory Improvement Amendments (CLIA) and College of American Pathologists (CAP) laboratory accreditation requirements
- Creating a subcommittee

MOTION: Dr. Joseph Schneider made a motion to create a subcommittee to work with DSHS to develop a plan to get closer to the vision presented by Mr. Reilly. Dr. Scott McLean seconded the motion. A roll call vote was taken for members over the phone. A voice vote was taken for members in the room. The motion carried with no objections nor abstentions.

Subcommittee membership:

- Dr. Joseph Schneider – Chair
- Ms. Pam Andrews
- Dr. Elizabeth (Kaili) Stehel
- Ms. Khrystal Davis
- Dr. Alice Gong
- Dr. Michael Speer

Agenda Item 13: Break

Dr. Gong announced a 5-minute break.

Agenda Item 14: Sickle Cell Subcommittee Reporting

Dr. Gong reconvened the meeting at 1:02 p.m.

Dr. Gong introduced and turned the floor over to Dr. Titilope Fasipe, Subcommittee Co-Chair. Dr. Fasipe referenced the handout, *Sickle Cell Subcommittee Meeting Minutes December 13, 2019*.

There was no member discussion.

Agenda Item 15: Newborn Hearing Screening in Neonatal Intensive Care Unit (NICU) Subcommittee Reporting

Dr. Gong introduced and turned the floor over to Dr. Tiffany McKee-Garrett, Subcommittee Chair. Dr. McKee-Garrett referenced the PowerPoint and handout, *Best Practices for Hearing Screening in Texas NICUs*.

Members discussed:

- The subcommittee needing to discuss the proposed Hearing Screening Rules to make sure the hearing screening best practice addressed in the Rules does not conflict with the work of the subcommittee
- The ideas of the subcommittee not being placed in the proposed Rules since it has not yet been evaluated for feasibility

- The program's processes and the proposed Rules are for well babies, not NICU babies, and hearing screening at 1, 3, 6 months is recommended
- Adding a line in the proposed Rules that all babies should be screened
- Making recommendation to Commissioner Dr. John Hellerstedt to provide the process map algorithm the subcommittee developed and send it to Texas Collaborative of Healthy Mother and Babies and Perinatal Advisory Council members to test the algorithm's functionality

MOTION: Dr. Scott McLean made a motion for the Chair and NICU Subcommittee Chair draft a letter to Commissioner Dr. John Hellerstedt asking him to recommend to the Perinatal Advisory Council and other agencies involved in the care of newborns especially those involved with sick and preterm babies that a new algorithm be considered for enhanced detection of hearing loss in preterm infants as outlined by the subcommittee for purpose of quality improvement for the NICU. Dr. Joseph Schneider seconded the motion. A roll call vote was taken for members over the phone. A voice vote was taken for members in the room. The motion carried with no objections nor abstentions.

Agenda Item 16: Critical Congenital Heart Disease (CCHD) Subcommittee Reporting

Dr. Gong introduced and turned the floor over to Dr. Scott McLean, Subcommittee Chair. Dr. McLean referenced the PowerPoint and handout, *Report of the Critical Congenital Heart Disease (CCHD) Subcommittee to the Texas Newborn Screening Advisory Committee (NBSAC)*.

Members discussed:

- Birth defects can do case level information for babies that have been screened for CCHD
- Individual hospital systems reporting to a repository
- Manual input for electronic medical records by midwives

ACTION ITEM: Dr. Speer will provide follow up on collaboration with the TCHMB.

Agenda Item 17: Newborn Screening Consultant (NSC) Fees Subcommittee Reporting

Dr. Gong turned the floor over to Dr. Scott McLean, Subcommittee Chair. Dr. McLean referenced the handout and PowerPoint, *Report from the NBS Consultants Subcommittee 31 January 2020*.

Members discussed:

- Consultants identified by the NBS program
- Drafting a letter to Commissioner Dr. Hellerstedt
- NBS Benefits Program is in place to ensure equity for people screened can receive treatment. It's an open enrollment program for laboratory testing of all conditions.

MOTION: Dr. Joseph Schneider made a motion for the chair and subcommittee chair of the NBS Consultant Fees to write a letter to Commissioner Dr. Hellerstedt based on language that is presented in the Recommendation DRAFT 2 of the NBS Consultant Fees presentation. Dr. Charleta Guillory seconded the motion. A roll call vote was taken for members over the phone. A voice vote was taken for members in the room. The motion carried with no objections nor abstentions.

Agenda Item 18: Public Comment

Ms. Gutierrez read public comment logistical announcements and called Ms. Kelly Coggin and Dr. Barbra Novak to address members.

Ms. Coggin, SMA mother, informed members of her experience as a parent of someone with SMA.

Dr. Novak, audiologist at Texas Children's Hospital, offered clarification to discussions that took place earlier in the meeting regarding the 1, 3, 6 rule not being able to be applied to NICU babies.

Agenda Item 19: Future Agenda Items/ Next Meeting Date/ Adjournment

Dr. Gong stated the next meeting is April 24, 2020 then opened the floor for discussion of future agenda items.

Members discussed:

- Informatics subcommittee report
- Medicaid funding from newborn screening reimbursements distributed outside of the program
- X-ALD family testing covered by insurance
 - Included in genetics testing standard of care
- Action item follow-up list
- Vital Statistics data linking
- Even-year annual review of bylaws
- Revised Proposed Hearing Screening Rules
- Follow up with subject matter expert on biliary atresia presentation

Dr. Gong adjourned the meeting at 2:38 p.m.

Webcast: <https://texashhsc.swagit.com/play/01312020-668>