Newborn Screening Advisory Committee
FINAL APPROVED Meeting Minutes
July 23, 2021
12:00 p.m.

Location: Microsoft Teams Live Event

Table 1: Newborn Screening Advisory Committee attendance on Friday, July 23, 2021.

MEMBER NAME	IN ATTENDANCE
Kaashif Ahmad, M.D., M.Sc.	Yes
Beryl (Pam) Andrews	Yes
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
Elizabeth (Kaili) Stehel, M.D.	Yes

Agenda Item 1: Welcome and Introductions
Dr. Alice Gong, Chair of the Newborn Screening Advisory Committee (Committee), convened the meeting at 12:00 p.m. and welcomed everyone in attendance.

Agenda Item 2: Committee Business Logistics
Dr. Gong introduced Mr. Eric Owens, Health and Human Services Commission (HHSC), Policy & Rules, Advisory Committee Coordination Office. Mr. Owens reviewed logistical announcements, called roll, asked members to introduce themselves, and determined a quorum was present.

Dr. Gong recognized Dr. Nancy Beck for her service and advised members that it would be her final meeting with the Committee. Dr. Gong asked David R. Martinez, Director, Department of State Health Services (DSHS) Newborn Screening (NBS) Unit, to introduce himself and other DSHS staff members. Mr. Martinez called on Aimee Millangue, Laura Arellano, Karen Hess, Dr. Susan Tanksley, and Steve Eichner to provide brief introductions. Mr. Martinez stated Dr. Debra Freedenberg and Ryan Hutchison will also be joining the meeting.

Agenda Item 3: Consideration of Meeting Minutes for April 16, 2021
Mr. Owens reminded members that the April 16, 2021 draft meeting minutes were sent to members via email for review and asked if members had any edits or changes. Hearing none, Mr. Owens requested a motion to approve the April 16, 2021 meeting minutes.

MOTION: Dr. Speer made a motion to approve the April 16, 2021 meeting minutes. Dr. Guillory seconded the motion. Mr. Owens conducted a roll call vote, and the motion carried with 12 approvals, with no objections or abstentions.

Agenda Item 4: Legislative Update
Dr. Gong introduced Mackenzie Spahn and Taj Sheikh, Government Relations Specialists, DSHS Government Affairs (GA). Ms. Spahn announced that Ms. Sheikh would not be joining. Ms. Spahn referenced the PowerPoint and handout, *87th Legislative Session Wrap-Up* and provided a legislative update.

Highlights of the presentation included:
- General statistics on number of bills filed, passed, and vetoed
- Bills that were not signed or vetoed by the end of the veto period on June 20th became law by default
- Number of bills filed for analysis or monitoring to ensure tracking of anything that had impact to public health or DSHS
- Newborn Screening Preservation Account
    - Budget bill rider needed to address $0 balance and for the ability for additional revenue to go into the account
    - Additional funding request necessary because the way budget rider was written, account would not have funds deposited to it to fund additional screens
    - Legislative Appropriations Request needed to fully fund Spinal Muscular Atrophy (SMA) testing implementation to include equipment and reagents
    - Budget rider that was passed was right-sized so additional revenue will go into the account
    - Funding request approved for a transfer of up to $12 million in revenue allowed for additional tests
    - SMA testing is fully funded with $4.2 million.

Members discussed:
- Clarification that the $12 million for funding additional tests is collected from the revenue above the appropriations to the account.
- If it can be identified where the money collected from newborn screening tests goes.

ACTION ITEM: Ms. Spahn will follow up with Budget program staff on questions that members send to her by email.

Agenda Item 5: 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 and Debra Freedenberg, M.D., Ph.D., Medical Director, DSHS NBS Unit.

Dr. Freedenberg referenced the PowerPoint and handout, *Texas NBS Spinal Muscular Atrophy (SMA) Case Update* and provided an update on three SMA cases identified from June 1, 2021 to July 19, 2021.

Members discussed:

- If second screen is done if condition is identified with first screen.
- Screening algorithm for first and second-tier testing for SMA.
- Expectation of milder clinical symptoms when there are more copies of SMN2.
- Clinical follow up and outcome for baby who had only 1 copy of SMN2.
- If expense of gene therapy is a barrier to treatment.
- DSHS Guidance and a pediatric neurologist referral list are available to clinicians so that they are aware of next steps and options.
- Babies with only one copy or zero copies of SMN2 are stronger candidates for gene replacement therapy.

Ms. Khrystal Davis, as an SMA parent, caregiver, and advocate, expressed appreciation for those who made SMA implementation in Texas possible.

Agenda Item 6: Screened conditions status updates

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

Dr. Freedenberg referenced the PowerPoint and handout, *Texas X-Linked Adrenoleukodystrophy (X-ALD) Case Update, August 2019 – June 2021* and provided an update on X-ALD cases identified from August 2019 to June 2021, and discussion of 23 males and 13 females affected

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

Highlights of the presentation included:

- Follow up on request for number of premature babies in the Neonatal Intensive Care Unit (NICU) by looking at data for low birth weight babies
- 32,165 total presumptive positive Congenital Hypothyroidism (CH) from 2016-2020 by birth weight
    - Data includes only one presumptive positive screen per baby
    - Low birth weight total, 48.5%
    - Normal weight total, 51.5%
- 1,719 CH diagnosed cases by birth weight from 2016-2020

Members discussed the high percentage of population of low-birth weight babies that are presumptive positives for CH compared to rate of presumptive positives for babies of normal birth weight.

Due to technical issues, the Committee proceeded to the next agenda item.

Agenda Item 7: Future condition implementation updates
Susan Tanksley, Ph.D., Laboratory Operations Unit Manager, DSHS Laboratory, and Debra Freedenberg, M.D., Ph.D., Medical Director, DSHS Newborn Screening Unit provided an update on implementation of future conditions.

Dr. Freedenberg stated:

- Two conditions on the Recommended Uniform Screening Panel (RUSP) for which Texas is not screening for due to funding are Mucopolysaccharidosis (MPS) Type 1 and Pompe disease (Glycogen storage disease type II).
- Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) is considering additional conditions, Mucopolysaccharidosis (MPS) Type 2 or Hunters Syndrome and Guanidinoacetate Methyltransferase Deficiency (GAMT)
- NBS Program anticipates that by the time MPS 1 and Pompe are implemented, Mucopolysaccharidosis (MPS) Type 2 will have made it through the ACHDNC review process and can be implemented with the same technology and similar methodology
    - Clinically, similar enough to MPS 1
    - Has an enzyme replacement therapy available
    - Different from MPS 1 in that it is an X-linked condition
    - Similar clinical manifestations as other Mucopolysaccharidoses

Dr. Tanksley stated:

- During the planning process to implement Pompe and MPS 1, also investigating how to add the screening for MPS 2 at the same time.
- The evidence review process requires that a condition be voted on within nine months of when it is voted to go to evidence review, so ACHDNC vote to recommend adding MPS 2 to the RUSP should be within nine months.
- Ultimately, the Secretary of Health and Human services determines whether a condition gets added to the RUSP or not.
- Texas does not currently have the funds to start implementation of Pompe and MPS 1.
- Hope to have money flow into NBS Preservation Account to begin implementation.
- Trying to determine how to incorporate bits and pieces of implementation in DSHS Laboratory's plans without cost or take advantage of other funding opportunities.
- DSHS Laboratory is building capacity for whole genome sequencing technology.

Members discussed:

- If GAMT screening could be implemented with the Laboratory's current platform.
- What new genomic technology might be available in the future that would impact newborn screening.
- Whether whole genomics screening could pick up all conditions for which Texas screens.
- Goals of a Rare Disease Subcommittee.
- Whether legislation allows Committee to recommend screening for conditions not on the RUSP.

MOTION: Ms. Khrystal Davis made a motion to form a Rare Disease Subcommittee to discuss future conditions, genomics, next generation screening, and other issues that impact the rare disease community. Dr. Guillory seconded the motion. Mr. Owens conducted a roll call vote, and the motion carried with 12 approvals, with no objections or abstentions.

Subcommittee membership:

- Ms. Khrystal Davis – Chair
- Dr. Alice Gong (Ex-officio member)
- Ms. Pam Andrews
- Dr. Barbra Novak
- Dr. Charleta Guillory
- Dr. Michael Speer (Ex-officio member)

ACTION ITEMS:

- Rare Disease Subcommittee will develop a charter, develop reasonable and achievable objectives, and report back to the full committee.
- For Dr. Gong to remain on the subcommittee, one member will need to step down from the Rare Disease Subcommittee once a new Chair is elected, since the new Chair will be an ex-officio member of the subcommittee.

The Committee returned to Agenda Item 6.

Agenda Item 6: Screened conditions status updates

Dr. Gong turned the floor over to Ms. Hess to resume her update on screened conditions.

Highlights of the presentation included:

- Thyroid-binding globulin (deficiency and access)
- Of the CH Diagnosed cases by birth weight from 2016 – 2020, three of the conditions are on the RUSP and others are differentials
- Diagnosed cases of CH by birth weight from 2016-2020
    - 75-76% diagnosed cases come from regular birthweight babies
- CH Summary by birth weight, 2016-2020
- Overview of diagnosed case numbers for other screened RUSP conditions, 2019-2020

Members discussed:

- Clarifying what is included in data of CH diagnosed cases and the data for CH presumptive positives.
- Data demonstrates a lot of false positive CH screens on low birth weight babies.
- Since the DSHS Laboratory is needing diagnoses to develop the new CH algorithm, it is helpful to know they can get diagnoses for about 50% of the presumptive positives in the NICU population a lot faster.
- The need to educate clinicians that blood draws for thyroid studies can be done on NICU babies since some clinicians delay confirmatory testing due to birth weight.
- It would be helpful to send out the numbers of diagnosed cases to all caregivers of newborn babies, since it is powerful information.
- The DSHS Laboratory publishes an annual report that includes diagnosed case numbers, but not at the same detail given in the presentation.

ACTION ITEM: Program staff will inform the Endocrine manager about the neonatologists' encouraging getting an earlier CH diagnoses for NICU babies and will put together an education piece to share with the NICUs about not delaying confirmatory testing for CH.

Since the presenters for Agenda Item 8 and Agenda 9 were not available, the Committee proceeded to Agenda Item 10.

Agenda Item 10: Critical Congenital Heart Disease Subcommittee Reporting

Dr. Gong introduced Dr. Michael Speer, Subcommittee Chair, to provide an update.

Dr. Speer did not have a report for this agenda item.

Agenda Item 11: Newborn Hearing Screening in Neonatal Intensive Care Unit (NICU) Subcommittee Reporting

Dr. Gong introduced Tiffany McKee-Garrett, M.D., Subcommittee Chair, to provide an update.

Dr. McKee-Garrett did not have a report for this agenda item.

Dr. McKee-Garrett informed the Committee that the subcommittee's algorithm was presented at a Texas Pediatric Society meeting. Dr. Speer added that there has been some discussion with the Texas Collaborative for Health Mothers and Babies (TCHMB) and Perinatal Advisory Council (PAC) about making Congenital Heart Disease one of the factors that are used in certifying NICUs across the state.

Agenda Item 12: Sickle Cell Subcommittee Reporting

Dr. Gong introduced, Titilope Fasipe, M.D., Ph.D., and Melissa Frei-Jones, M.D., Subcommittee Co-Chairs. Dr. Frei-Jones provided members with an update.

Dr. Frei-Jones stated:

- Subcommittee met and worked on updates to the list of hemoglobinopathy consultants, an adult list, and ACT and Fact sheets.
- Instead of including information on the hemoglobinopathy consultants lists about which centers were able to offer a minimum standard of care for sickle cell disease, including stroke screening, immunizations, and hydroxyurea therapy, subcommittee decided to include recommendations on standards of care on ACT and Fact sheets.
- A group is starting a national effort to identify sickle cell centers of excellence by developing a national alliance of sickle cell centers that are vetted for membership.
- A need to empower individuals with information regarding what to look for in sickle cell care centers.
- Putting together an adult list has been challenging since Texas does not really have adult centers.

Members discussed:

- There is a lot that needs to be done with transition to adulthood.
- Work on adult lists will continue to be a challenge because of the lack of providers caring for adults.

The Committee returned to Agenda Item 8.

Agenda Item 8: Vital Statistics Presentation

Dr. Gong introduced Tara Das, Ph.D., M.P.H., M.L.I.S., State Registrar, DSHS Vital Statistics. Dr. Das referenced the PowerPoint and handout, *An Overview of Vital Statistics.*

Highlights of the presentation included:

- Uses of vital statistics
- Lifecycle of a vital event
- Organizational structure of DSHS Vital Statistics Program
- Information collected from the birth certificate
- Birth certificate registration, issuance and data routing process
- Birth certificate data quality
- Information collected from the death certificate
- Death certificate registration, issuance and data routing process
- Death certificate - cause of death
- Death certificate data quality
- Texas Electronic Vital Events Registrar (TxEVER) System infrastructure and technology
- Addressing vital statistics data quality

Members discussed:

- Clarifying how information on the baby's mother's race and the mother's socioeconomic status are obtained.
- If there has been an increase in self-reporting of race as "other."
- Informing Vital Statistics about the initiative to have a single pipeline from hospitals, doctor's offices, and others for electronically reporting information to the State rather than individual pipelines.
- If there is a way to include if and where newborn and Critical Congenital Heart Disease screening was done as pieces of vital statistics data.

Agenda Item 9: Critical Congenital Heart Disease (CCHD) Birth defects Presentation

Dr. Gong introduced Charles Shumate, Dr.P.H., Research Specialist, DSHS Birth Defects Epidemiology and Surveillance Branch. Dr. Shumate referenced the PowerPoint and handout, *Critical Congenital Heart Disease Surveillance in Texas.*

Highlights of the presentation included:

- Overview of Texas Birth Defects Registry (TBDR) and NBS CCHD data
- British Pediatric Association (BPA) Codes used for analysis
- Comparing Texas TBDR and NBS data on frequency of CCHDs by birth year
- Reviewing the numbers or and percentage of NBS cases captured by TBDR by birth year
- The TBDR has not caught up to NBS data, as data starting with 2018 is incomplete
- Case fatality rate for the first year of life, any CCHD, 1999-2018
    - Drop in fatalities due to improvement in surgical management of single ventricle lesions such as Hypoplastic Left Heart Syndrome (HLHS)
    - Not enough data to show impact of implementation of House Bill 740 in 2014 for passive pulse oximetry newborn screening
- Provisional case fatality rate for the first year of life, Hypoplastic Left Heart Syndrome (HLHS), 1999-2018
- Improving data reporting with a look at the number of facilities reporting though TBDR compared to the number of facilities reporting through NBS.
- Indication of death is through medical record or vital statistics.
- Case fatality in Texas peaked late 1990s/early 2000s.
- Cases decreased due to pediatric cardiologists.

Members discussed:

- Setting up a mechanism to provide feedback to facilities that may have missed the greatest number of opportunities to report through NBS as shown through TBDR data.
- Presentation shows the need to do a better job of screening for CCHD.
- Working with the PAC to look at the data of babies being transferred to other centers, since almost a third have had ultrasounds that picked up conditions prenatally. Are babies being delivered at centers where they should be delivered or is there is a delay in treatment because they are delivered somewhere else?
- Working with TCHMB on a statewide project on screening and reporting.

ACTION ITEM: Dr. Shumate will review data on defects other than HLHS that are not as easily picked up by prenatal ultrasound to see if there is an impact from pulse oximetry screening.

The Committee proceeded to Agenda Item 13.

Agenda Item 13: Newborn Hearing Screening Consent Form Subcommittee Reporting

Dr. Gong introduced Dr. Elizabeth Kaili Stehel, Subcommittee Chair, who provided members with an update. Dr. Stehel referenced the handout, draft of the *Consent to Release Information* form.

Dr. Stehel stated:

- Subcommittee met to make further changes as discussed at the previous Committee meeting.
- Form was simplified down to one page.
- Many items were removed, such as information that could be found in other educational materials that are available.
- Language was simplified to make it easier for providers to use and easier for families to understand.

Members discussed:

- Needing clarity on what information parents are consenting to share and to whom the information will be shared
- Some facilities may be interpreting the law differently and only providing the form to parents when babies do not pass their hearing screen.
- Facilities have the responsibility documenting intake of the form in the Texas Early Hearing Detection and Intervention Program (TEHDI) Management Information System (MIS) and maintaining the consent form as part of the medical record.
- By law, all results must be reported and TEHDI needs consent to provide personal information for follow up on cases of late onset hearing loss.
- Changes that can be made so that the language used is more specific about what information is being shared.
    - Revise the first paragraph so that it reads "By law, your child's hearing screening results, pass or fail."
    - Clarifying consent language
        - Add "private" to "information"
        - Results only go to Statewide Outreach Center at Texas School for the Deaf if the baby fails the hearing screen

MOTION: Dr. Stehel made a motion to accept the revised version from June 17, 2021 subcommittee meeting with three corrections and to send the corrected Consent Form to Mr. Martinez, who will send it to Legal and Communications. Dr. Speer seconded the motion. Mr. Owens conducted a roll call vote, and the motion carried with 12 approvals, with no objections or abstentions.

ACTION ITEM: Run revised form through language checker to verify level of language has not been upgraded.

Agenda Item 14: Health Information Technology Subcommittee Reporting
Dr. Gong introduced Joseph Schneider, M.D., Subcommittee Chair, who provided members with an update.

Dr. Schneider stated:

- Subcommittee met on June 18th and discussed the formation an interoperability collaborative by the Health Services Authority that is intended to work on specific issues of interoperability.
- First topic covered by collaborative was newborn naming conventions.
- Brendan Reilly, DSHS Laboratory, gave an update on newborn screening lab orders and results interoperability through the grant to connect doctor's offices and hospitals to the State.
- Funding has started for a Texas HHS Interoperability platform.
- Discussed the 21st Century Cures Act as it relates to newborn screens.

Members discussed:

- Relevance of inviting Dr. Das from Vital Statistics to present to the Committee and introducing idea of including newborn screening as a vital event.
- Primitiveness of Texas' birth certificate data collection efforts relative to other states, where information flows electronically from the Electronic Medical Record.

ACTION ITEM: Subcommittee will focus on connections to the birth certificate registry and the next steps for linking NBS and the registry.

Agenda Item 15: Review of Bylaws
Dr. Gong provided an update to members on the bylaws.

Dr. Gong stated:

- There is a new department template for the bylaws, so there will be changes to put the bylaws on the new template.
- Bylaws be voted on by the Committee at the next meeting on October 8.
- There will be an election for a new Chair and Vice Chair at the next meeting, so this meeting is her last meeting.

ACTION ITEM: Members should bring up anything they want addressed or changed in the bylaws by the next meeting.

Due to technical issues, the Committee proceeded to Agenda Item 17.

Agenda Item 17: Future Agenda Items/ Next Meeting Date/ Adjournment
Dr. Gong opened the floor for discussion of future agenda items and stated the next meeting was scheduled for October 8, 2021.

Members discussed:

- Reviewing the coordinated response from the NBS Laboratory and NBS Clinical Care Coordination during the ice storm and the continuity of operations plan
- Action Item follow-up list:
    - Data linking for vital statistics
    - Medicaid funding
- Standing items:
    - Screened conditions updates
    - Future conditions implementation updates
    - Subcommittee updates from:
        - Critical Congenital Heart Disease Subcommittee
        - Health Information Technology Subcommittee
        - Sickle Cell Subcommittee
        - Hearing Screening in the NICU Subcommittee
        - Rare Disease Subcommittee
- Updated funding request for meeting timeliness goals estimate for 7-day working lab to include follow-up care cost if babies are not screened
- Review of the bylaws
- Officer Election

The Committee returned to Agenda Item 16.

Agenda Item 16: Public Comment
Mr. Owens read the public comment logistical announcements and called on Ms. Lisa Otto to address the Committee.

Ms. Otto stated that stated that she would like to request adding Noonan Syndrome to Newborn Screening in the State of Texas and informed members about the disorder.

Dr. Gong thanked Ms. Otto for her comments.

Dr. Gong thanked members and adjourned the meeting at 4:08 p.m.

==========

Below is the link to the archived video of the July 23, 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://texashhsc.swagit.com/play/07232021-806/2/