Newborn Screening Advisory Committee
Meeting Minutes
January 22, 2021
12:00 p.m.

Location: Microsoft Teams Live Event

Table 1: Newborn Screening Advisory Committee attendance Friday, January 22, 2021.

MEMBER NAME	IN ATTENDANCE
Kaashif Ahmad, M.D., M.Sc.	Yes
Beryl (Pam) Andrews	No
Nancy Beck, M.D.	Yes
Khrystal Davis, J.D.	Yes (Arrived at Item #4)
Titilope Fasipe, M.D., Ph.D.	No
Melissa Frei-Jones, M.D.	Yes (Arrived at Item #6)
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 (Arrived at Item #5)
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 (NBS) Advisory Committee, convened the meeting at 12:00 p.m. and welcomed everyone in attendance.

Agenda Item 2: Committee Business Logistics
Dr. Gong introduced, Ms. Sallie Allen, HHSC, Policy & Rules, Advisory Committee Coordination Office and she reviewed logistics announcements, called roll, and determined a quorum was present.

Agenda Item 3: Review and Approve Meeting Minutes for October 2, 2020
Ms. Allen requested a motion to approve the October 2, 2020 meeting minutes.

MOTION: Dr. Charleta Guillory made a motion to approve the October 2, 2020 meeting minutes. Dr. Michael Speer seconded the motion. Ms. Allen conducted a roll call vote, and the motion carried with no objections or abstentions. Five members were not present at the time of the roll call vote.

Due to presenter connectivity, proceeded to Agenda Item 5.

Agenda Item 5: ClinVar Genetic Variants Database update
Dr. Gong introduced, Rachel Lee, PhD, Microbiological Sciences Branch Manager, DSHS Laboratory. Dr. Lee referenced the PowerPoint handout, *ClinVar Genetic Variants Database.*

Highlights of the presentation:

- ClinVar welcomes submissions from clinical testing labs, research labs, genetics clinics, patient registries, locus-specific databases, expert panels and groups establishing practice guidelines
- There are specific data requirements for each submission
- Required Evidence is observant in three variants
- Collection Method for ClinVar is specific to each setting
- Affected Status relates to the condition for the interpretation
- ClinVar collects variant data, but is working with Centers for Disease Control (CDC)
- All data submitted is de-identified

Returned to Agenda Item 4.

Agenda Item 4: Health Information Technology Subcommittee Reporting
Dr. Gong introduced, Joseph Schneider, M.D., Subcommittee Chair, and Max Solodky, Director, Strategy and Planning, HHSC, Chief Technology Officer (CTO) Office. Dr. Schneider stated the subcommittee met on December 22, 2020 and noted Steve Eichner, Brendan Reilly and Max Solodky gave a presentation on interoperability and data exchange. Mr. Solodky presented a revised version of that presentation to the committee members and referenced the powerpoint/handout, *Integration and Data Exchange Capabilities.*
Dr. Schneider also noted, that after Mr. Solodky's presentation, he would present a draft recommendation letter related to interoperability and HHS gateway, for review and approval to be submitted to the DSHS Executive Commissioner.

Highlights of the presentation included:

- Overview of the organizational challenges of HHSC data exchange.
    - Limited timely and accurate information across systems
    - Many point-to-point integrations
    - Inability to service comprehensive data requests
    - No comprehensive Source of Truth
    - Limited dedicated support for data exchange services
- Texas does not have a centralized data exchange
- Programs like Newborn Screening have direct interfaces with some hospitals, but do not have interfaces with other directly related programs, such as Newborn Hearing, Birth Defects, and Vital Statistics
- Laboratory orders and results are heavily dependent on physical forms, mail, and faxes
- Data exchange resources and funding are siloed, creating redundant and overlapping systems (people, technology, processes)
- Provided several business case scenarios which outlined how a data exchange strategy would be beneficial to agency.

- The Texas State Legislature has passed legislation related to interoperability, privacy, and supportive infrastructure.
- The Integration and Data Exchange Center of Excellence (iCOE) business model could address the data exchange challenges and provide a solution by:
    - Establishing a dedicated technical team
    - Outline the business values that could be achieved
    - Design data exchange platform between entities to streamline connectivity and standardize exchange protocols, formats
- Interoperability of a Future Ecosystem allows for connection between the external partners and internal HHS agencies
- Referenced Gateway examples related to:
    - *North Carolina Health Information Exchange Authority* (NC HIEA) (https://hiea.nc.gov/)
    - *Michigan Health Information Network* (MiHIN) (https://mihin.org/)
    - Washington State Department of Health – *OneHealthPort HIE* (https://www.onehealthport.com/)
- Benefits of utilizing iCoE platform include:
    - Centralized Data Exchange, including Medicaid point data
    - A path to a comprehensive source
    - Reduction of the communication lag between agencies and providers
    - Improved quality of patient care across providers through targeted data sharing
    - Enhanced response capability to national and statewide medical emergencies (ex: COVID)
    - Reducing siloed resources and customized integration cost to external partners
    - Recognize Texas as a leader in data interoperability technology and promote the lease of this exchange platform to other states
- Next Steps
    - Leadership review
    - Cross-program funding options
    - Define initial roadmaps and partners
    - Management committee prioritization
    - Execute on first project
    - Recruit initial team

Next, Dr. Schneider presented the draft letter regarding interoperability and HHS gateway and members concurred with the intent of the Newborn Screening program serving as a pilot program.

MOTION: Dr. Charleta Guillory made a motion to approve and grant Dr. Gong authority to edit the letter as needed and submit to DSHS program staff for submission to DSHS Commissioner Hellerstedt. Dr. Michael Speer seconded the motion. Ms. Allen conducted a roll call vote, and the motion carried with no objections or abstentions. Three members were not present at the time of the roll call vote.

Agenda Item 6: 2019 Newborn Screening Annual Report
Dr. Gong introduced, Dr. Susan Tanksley**,** Laboratory Operations Unit Manager, DSHS Laboratory, and announced that Dr. Tanskley would present agenda items, 6, 7, 8 and 9, consecutively. Dr. Tanksley referenced two handouts, *Newborn Screening Annual Statistics* and *Newborn Screening Annual Report 2019* and provided an overview of the information.

Dr. Tanksley stated:

- The one-pager outlines the summary of all the statistics for 2019
- Comparative statistics for 2017, 2018 and 2019 annual reports are on the website
- Compiling 2020 numbers entails feedback on diagnosed cases to ensure report contains correct number of cases for the year
- Total number of screens annually has declined slightly from 2018
- Timeliness measures are calculated monthly on specimens
- so that specimens are received in a timely fashion and so critical newborn screening results go out to the healthcare providers quickly so families are aware and additional testing is obtained.
- Transit time is calculated from collection to receipt in the laboratory; numerous factors attest to meeting timeliness goals, i.e. time of birth, collection of specimen, scheduled courier pickup and delivery time. Although a low percentage of specimens arrive within 24 hours of collection, the percentage does improve significantly with those received within 48 hours.

Members discussed:

- Number of first screens is not equal to the birth rate, and the number of babies not screened is not known since there is no data linking with Vital Statistics
- Even with 97.3% of initial screen specimens collected within 48 hours of birth, that shows over 10,000 babies are being missed
- Number of first screens and second screens do not match
- Performance measures for time to diagnosis or intervention
- Challenge of meeting timeliness goal of receiving specimens within 24 hours of collection with the DSHS lab operating on a 6-day work week

ACTION ITEM: DSHS Lab will provide the matching rate of first screens to second screens and the cost-estimate for operating 7 days a week.

Agenda Item 7: Spinal Muscular Atrophy (SMA) screening implementation update
Dr. Tanksley provided an update on SMA screening implementation.

Dr. Tanksley stated:

- Funding approval was received on July 2, 2020.
- New Full-time Employees (FTEs) have been obtained and are being hired, and equipment has been ordered.

- There will be a two-tiered approach to screening for SMA.
    - In the first-tier assay, SMA and Severe Combined Immunodeficiency (SCID) will be tested together using a real-time Polymerase Chain Reaction (PCR) assay to detect absence of a deletion that causes SMA and low/no T-cell receptor excision circles which indicates possible SCID.
    - The first-tier test validation has been completed, and the data is being analyzed. A report will be written soon and submitted for approval.
    - The validation study will start soon for the second-tier test to detect copy number of SMN2 gene; the number of copies of SMN2 correlates well with disease severity and is used to determine if a child needs treatment.
- Specifications are in development for the Laboratory Information Management System (LIMS), which is being updated to include testing, reporting and follow-up functions for SMA.
- Clinical care algorithms are being developed so that providers know where to refer patients, which diagnostic tests are required, etc.
- Two meetings with specialists in the care of SMA patients are being planned prior to June 2021 to ensure appropriate guidance can be provided to healthcare providers who care for infants with abnormal screening results for SMA.
- For education purposes,
    - a brochure has been developed,
    - an online provider education module is in final stages of development,
    - Grand Rounds has been scheduled for healthcare providers to learn about SMA.
- Three new NBS staff will be hired.
- On target to implement screening for SMA in June 2021.
- Plan is to increase the newborn screening fee effective January 1, 2022

Agenda Item 8: Future condition implementation updates
Dr. Susan Tanksley**,** Laboratory Operations Unit Manager, DSHS Laboratory, referenced the handout *Pompe-MPS 1 Cost estimate Summary* and provided an overview of the cost estimate information.

Dr. Tanksley stated:

- A cost estimate was developed for implementation of Pompe and Mucopolysaccharidosis Type I (MPS I) and submitted to the Legislative Budget Board in November 2020.
- These costs include a building retrofit, equipment, reagents, consumables, staff, changes to the laboratory information management system and education/training.
- There are two methods that can be used to test for these disorders; these methods are being investigated.
- The DSHS Laboratory in Austin, where newborn screen testing is performed, does not have adequate space to add this testing, but we are switching some existing equipment currently, which will create more space in the laboratory.

- This should be adequate for the equipment needed to screen for Pompe and MPS-I.
- If funds are received, the building retrofit before new equipment can be obtained/installed could begin in Fall 2021, and implementation is estimated to be January 2024.

Agenda Item 9: Screened conditions status updates
Dr. Susan Tanksley**,** Laboratory Operations Unit Manager, DSHS Laboratory, and Dr. Debra Freedenberg, Medical Director, DSHS Newborn Screening Unit, referenced PowerPoint/handouts, *DSHS Notice regarding Funding Opportunity for Newborn Screening Submitters,* and *Texas NBS X-linked Adrenoleukodystrophy (X-ALD) Case Summary, August 2019-January 2021.*

Dr. Tanksley stated:

Congenital Hypothyroidism screening –

- DSHS obtained a CDC grant to fund a project to improve the screening method for primary congenital hypothyroidism.
- DSHS changed the equipment and testing algorithm in December 2020.
    - Prior to December 7, 2020, lab was screening for thyroxine levels as a first-tier test.
    - Measuring both thyroxine and thyroid-stimulating hormone in every specimen.
    - Expected and have experienced a larger volume of results requiring either retest or confirmatory testing.
    - Need timely information from healthcare providers when cases are diagnosed or cleared so that we can refine the screening algorithm to reduce false positives.
    - It's also critical to know if any disorder that screened for is not detected during screening (a "missed" case). CCC and the lab keeps track of cases not detected.

Dr. Freedenberg provided case summary results by category for X-ALD:
- 17 males hemizygous affected
- 9 female heterozygotes
- CADD (Contiguous ABCD1 DXS1357E deletion syndrome) – 1 Case
- Zellweger Syndrome - 4 cases and 1 carrier

Agenda Item 10: COVID-19 Newborn Screening Protocols
Dr. Gong introduced, Mr. Brendan Reilly, Business Analyst, Laboratory Services, DSHS. Mr. Reilly referenced the PowerPoint/handout, *COVID-19 Impact on Newborn Screening.*

Highlights of the presentation:
- Since the beginning of the pandemic, there has been a decrease of approximately 4% in screens received
- Drop in specimen collection is consistent with nationwide trend
- Unclear whether decline is due to declined birth rates or due to declining submission of newborn screening specimens

- There has also been an equivalent decrease in follow-up screens received
- For babies born in March, that was a 50% increase in missed 2nd screens (approximately 900 babies)

Proceeded to Agenda Item 14.

Agenda Item 14: Newborn Hearing Screening in the Neonatal Intensive Care Unit (NICU) Subcommittee Reporting
Dr. Gong introduced Tiffany McKee-Garrett, M.D., Subcommittee Chair. Dr. McKee-Garrett referenced the handouts *Newborn Hearing Screening in the Neonatal Intensive Care Unit (NICU) Meeting Minutes, December 3, 2020, Letter to Dr. Hellerstedt* and *Protocol for Neonatal Intensive Care Unit (NICU) Hearing Screens).*

Dr. McKee-Garrett stated:

- Subcommittee met on December 3, 2020 and discussed finalizing a letter regarding recommendations for newborn hearing screening and Texas NICU. The letter was sent to DSHS Commissioner Dr. John Hellerstedt in December 2020.
- Response will be shared with committee once it is received.

Returned to Agenda Item 11.

Agenda Item 11: Newborn Hearing Screening Rule Reporting
Dr. Gong introduced Mr. David Martinez, DSHS, Director, Newborn Screening Unit, and advised that Mr. Martinez will present agenda items 11 and 12, consecutively. Mr. Martinez provided updates to members and referenced the PowerPoint handout, *Newborn Screening Hearing Rules.*

Mr. Martinez provided members with links to the Texas Secretary of State website and the DSHS Rules and Regulation webpage. Members may access these links to review the NBS rule in detail. Members should send their comments regarding the consent form to him and Ms. Millangue.

Mr. Martinez stated:

- Revised Newborn Hearing Screening rules are effective as of December 23, 2020
- All formal and informal stakeholder comments were reviewed and considered.
- The consent form was reviewed by Government Affairs, Legal counsel and DSHS Communications for plain language.
- The consent form is a separate document and is referenced in the newborn screening rule. Although it is part of the rule, having it as a separate document allows revisions to be made without having to through the entire rulemaking process.

Agenda Item 12: Legislative Update
Mr. David Martinez, DSHS, provided members with a legislative update.

Mr. Martinez stated:

- As of January 12, 2021, DSHS is tracking 173 bills and 17 of those have been assigned to Community Health Improvement division for further analysis
- Currently no bills directly related to newborn screening services at DSHS have been filed.
- The newborn screening lab may be impacted by some bills, but to Program's knowledge they have not been any specific to newborn screening identified to date.
- As a reminder, committee members are free to participate in legislative session as long as it conducted as independent representation and not in any way on behalf of the committee or the department.
- Program will request to have a representative from Government Affairs to provide legislative updates at the next meeting, or a summary of updates will be provided to the Committee.

Agenda Item 13: Critical Congenital Heart Disease (CCHD) Subcommittee Reporting

Dr. Gong introduced Dr. Michael Speer, subcommittee chair. Dr. Speer referenced the handout *Critical Congenital Heart Disease (CCHD) Subcommittee Meeting Minutes, December 29, 2020.*

Dr. Speer stated:

- Subcommittee met on December 29, 2020 and noted they had received a response to the CCHD recommendation letter that was sent to DSHS Commissioner Hellerstedt. A copy of the letter was emailed to subcommittee and NBSAC members on September 4, 2020.
- Based on response letter, following are action items to be addressed:
    - A follow-up letter sent to Dr. Catherine Eppes, Chair, Texas Collaborative for Healthy Mothers and Babies (TCHMB), stating the NBSAC would like to suggest that CCHD screen be taken up by the Neonatology group of TCHMB as a proposed quality improvement project.
    - The NBS program will initiate the rulemaking process to amend the rule to clarify which providers have responsibility for CCHD Reporting.
    - Karen Hess, Genetics Branch Manager, DSHS NBS Unit, will report on the CCHD data through 2020 at the April 2021 NBSAC meeting**.**
    - The program will invite Birth Defects to present their CCHD data at the April 2021 NBSAC meeting.
    - Add an annual report of CCHD data and reporting status as a standing agenda item for the NBSAC to see if improvements have happened.

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, December 4, 2020*.

Dr. Frei-Jones stated:

Sub-committee met on December 4, 2020 and provided following updates:

- Dr. Fasipe and Dr. Frei-Jones worked on updating the pediatric consultants list and developed an adult consultants list. Finalization of list to be discussed further.
- A sickle cell diagnosis alone does not qualify a patient for Social Security Supplemental Security Income (SSI), and applicants are often denied without another serious medical complication such as stroke or renal failure. This issue is being investigated by a subcommittee on the Sickle Cell Task Force.
- The Sickle Cell Task Force is looking into SSI denials for sickle cell patients who do not have co-morbidities.
- There are areas in Texas, especially in rural areas, where it is not known where sickle cell patients receive care and may only see their PCPs.
- Finalize Pediatric and Adult Hemoglobinopathy Consultant lists and post to the Newborn Screening (NBS) website, and possibly the Genetics Providers and Sickle Cell Committee Resource Page. These will be updated annually.
- Propose to the full committee that the Sickle Cell subcommittee become a standing committee as opposed to an ad hoc committee. Chair and Committee agreed to make the Sickle Cell Subcommittee a standing subcommittee.
- Continue to work on document review of ACT/FACT Sheets for sickling conditions, Parent and Primary Care Provider letters, and Long-term care form.
- Develop a process to enable the different centers and programs on the Hemoglobinopathy Consultants lists to collaborate on sickle cell-related issues under the purview of the Sickle Cell Subcommittee.

Agenda Item 16: Public Comment

Ms. Allen read the public comment logistical announcements and called on those registered for public comment.

Oral public comment was received from:

- Ms. Beth Moore, a parent of two children with SMA, expressed her support of the newborn screening of SMA in Texas.

Written public comment was received from:

- Mr. Maynard Friesz, Vice President, Advocacy and Policy, Cure SMA, supports the newborn screening of SMA. He commented that Cure SMA and our 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 Date/ Adjournment

Dr. Gong would like for the Sickle Cell Subcommittee to investigate public comments received from Mirepoix, LLC.

Members discussed future agenda items:

- CCHD data - update on numbers reported
- Action Item follow-up list:
    - Data linking for vital statistics
    - Medicaid funding
- Funding update
- HIT recommendation letter
- Cost estimate for 7-day working lab
- Overview of subspecialist follow up for time-critical conditions
- NBS consent form – TEHDI information
- Hematologists – participate as Subject Matter Experts (SMEs) for subcommittee
- Legislative updates

Dr. Gong thanked members and adjourned the meeting at 4:21 p.m.

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To listen to the webcast recording of the January 22, 2021 meeting go to:
https://texashhsc.swagit.com/play/01222021-825/2/