

Newborn Screening Advisory Committee

**Texas Department of State Health Services
1100 West 49th Street, Austin, Texas
Moreton Building, M2-204
June 20, 2014 via Conference Call
Minutes**

Conference Call

William Morris, LVN
Charleta Guillory, MD
Nancy Beck, MD
Mark Lawson, MD

Staff

Debra Freedenberg, MD, PhD, Department of State Health Services (DSHS), Newborn Screening Unit
Michael Chisum, Program Attorney, DSHS, Office General Counsel
David R. Martinez, Manager, DSHS, Newborn Screening Unit
Susan Tanksley, PhD, DSHS, Laboratory Operations Unit Manager
Karen Hess, DSHS, Newborn Screening Genetics Branch Manager
Brendan Reilly, Program Specialist, DSHS Laboratory, Biochemistry & Genetics Branch
Monica Molina, Ombudsman, DSHS, Newborn Screening Unit

Call to Order

Chairman Morris called to order the June 20, 2014 meeting of the Newborn Screening Advisory Committee at approximately 10:10 am.

Roll call of committee members, staff and guests

David R. Martinez introduced Monica Molina as the new ombudsman for the Newborn Screening Program and as the new support for the committee. Other introductions were made and it was determined that a quorum was not present. Members, staff and guests attending are listed at the beginning of these minutes.

Review and Approval of Minutes

Minutes for the December 20, 2013 and March 6, 2014 meetings could not be approved due to a quorum not being present.

Newborn Screening Program Overview-Debra Freedenberg, Susan Tanksley

The overview was on the agenda but not presented; David R. Martinez said that this was prepared as an overview for the purposes of the new committee members and is similar to what the committee has heard before. Mr. Martinez stated that this overview will probably not be presented at today's meeting and will be saved for Dr. Zellers and Ms. Gonzalez when they join the committee at the first meeting.

Newborn Screening Program Updates-Debra Freedenberg

Dr. Freedenberg gave the committee an update on the Newborn Screening Program.

- Goals
 - Two screening tests for each baby born in Texas
 - 24-48 hours of age
 - 1-2 weeks of age

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- Infants testing positive receive prompt and appropriate confirmatory testing
- Diagnosed infants are maintained on appropriate medical therapy
- Newborn Screening Panel
 - Currently screen for 29 disorders by blood spot
 - Congenital Hypothyroidism
 - Congenital Adrenal Hyperplasia
 - 3 Hemoglobinopathies
 - Galactosemia
 - Biotinidase Deficiency
 - 6 Amino Acid Disorders
 - 5 Fatty Acid Oxidation Disorders
 - 9 Organic Acid Disorders
 - Cystic Fibrosis
 - SCID
- Newborn Screening Workload
 - In 2013, received ~753,000 specimens (~386,700 newborns)
 - Specimens assayed and reported - ~745,500
 - Test specimens Monday through Saturday
 - Average 2,450 specimens per day
 - ~ 7,400 unsatisfactory specimens (~0.98%)
 - In 2012, ~ 16,145 (~2%) specimens reported with presumptive positive results
 - ~ 800 cases diagnosed annually
 - Testing performed 6 days a week
- Timeline of a specimen in the laboratory
 - Critical results could be out as soon as day 3
 - Most results are out by day 5
 - Some of the second tier testing for Hemoglobinopathy and MCAD are performed in lengthy batches, so it may go slightly longer for the DNA
- Quality Improvement Activities
 - Revised submitter quality report cards-available online since July 2013
 - Expedited submitter fax notification on unsatisfactory specimens
 - Monthly submitter calls to consult providers with highest unsatisfactory rates
 - Complete redesign of NBS laboratory website
 - NBS transit time and courier services
 - Server Operating System and SQL and Lab LIMS upgrades
 - NBS Lean 6-Sigma projects
- New Report Cards
 - Produced monthly
 - Accessible to all enrolled individuals at that facility
 - Facility-specific data compared to state average
 - Available online since July 2013

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- Reports available for January 2013 forward
- Nationwide Attention-Transit Times
 - In 2009, baby with MCAD dies at 4 days of age in Colorado. Mother becomes active advocate
 - Media attention on transit times heightened-November, 2013
 - Milwaukee Journal Sentinel article “Deadly Delays”
 - Series of articles focused on timeliness in newborn screening
 - Primary criticisms
 - Hospitals-delays in transit to the laboratory
 - Laboratories-lack of weekend/holiday testing and lack of transparency (~ 20 states release data)
 - Texas noted as one of the poorest performers
 - Subsequent media
 - Minnesota Post, NPR, WOAI San Antonio, Houston Chronicle, Arizona Republic
 - Specimen Collection Rules and Instructions
 - Texas Administrative Code (Rules)
 - Blood specimens must be mailed to the department within 24 hours after collection
 - Instructions
 - Must ship dried specimen WITHIN 24 HOURS
 - DO NOT hold specimens for bulk mailing. Send within 24 hours of collection
 - Strategy for Improvement
 - Review and Revise Submitter Education
 - Expand Issue Awareness / Gather Information
 - Target Top 25 Sites
 - Identify Submitter Barriers and Issues
 - Implement New and Ongoing Outreach Initiatives
 - Pursue System Improvements
 - Enhance Monitoring
 - Expand Scope to Include Other Key Quality Measures
 - Overall Status - > 96 hours
 - Overall status for specimens that were received > 96 hours
 - 2013 averaged 14.6%
 - April 2014 averaged 3.7%
 - Electronic Data Transfer
 - Web-based demographic entry and reporting
 - Available to any healthcare provider, password protected
 - Users from 1,046 facilities submitting 75% of NBS specimens
 - ~2% of all demographics
 - ~12,000 result views per month

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- Monthly report cards available for all of 2013
- HL7 file transfer functions for LIMS
 - Direct transfer of demographics and results between computer systems
 - 3 large hospital systems fully implemented (~10% of all specimens)
 - Several facilities waiting to start implementation
 - New facilities on hold pending system reevaluation
- Second Tier Assay for CAH
 - Purpose is to dramatically cut false positive rate
 - Status
 - New LC/MS/MS installed
 - Method optimization is complete
 - Validation continues
 - 6 month pilot completed-analysis of results on-going
 - Preliminary finding
 - Reduce false positive rate by ~50%
- Second Tier Assay for VLCAD
 - Purpose is to provide additional information to metabolic specialists
 - Status
 - Control materials; e-mail sent out to metabolic specialists inquiring about genotypes for confirmed cases and already receiving information back
 - Validation plan proposal is almost complete and then will need appropriate approvals
 - Optimizing current primers before beginning validation testing
- Implementation of HB 411
 - HB 411 became law in September 2011 and made major changes to NBS specimen retention and residual use
 - Multiple internal processes, procedures and policies have been revised
 - Institutional Review Board policy has been revised
 - Opt-in for long term storage and possible research uses-effective June 1, 2012
 - Parent decision form and parent education form developed and distributed
 - ~49% of NBS have a parental decision form returned
 - 74% of those returned and valid give permission for public health research uses (36% of all newborns)
 - Survey of all submitters to identify challenges in returning parental decision forms
 - Worst performers tend to be large hospitals
 - Most common practice is to include Decision form in discharge packet with little or no explanation
 - Destruction process started in April 2014
- NBS Hearing Grant Activities
 - CDC-5 year \$162,000 grant, in year 3

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- Tracking and data integration
- electronic health records
- HL7 messaging
- Enhanced interoperability of management information system
- Educational materials and training on enhancements
- HRSA-3 year \$300,000 grant, in year 3
 - Lost to follow-up
 - Multiple pilots on most effective follow-up procedures
 - Parent Support Group Projects utilizing quality improvement strategies
 - Early Childhood Hearing Outreach (ECHO) Project to better connect with Head Start
- Critical Congenital Heart Disease (CCHD)
 - US Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC)
 - In 2010, recommended that CCHD be added to the newborn Recommended Uniform Screening Panel (RUSP) to identify newborn with structural heart defects
 - 2011, endorsed by Secretary of HHS Kathleen Sebelius
 - 2013, Texas HB 740 added CCHD to core panel in 83(R) session
 - Implementation planned for Fall, 2014 when rules are finalized
 - The seven defects classified as CCHD are:
 - Hypoplastic Left Heart Syndrome (HLHS)
 - Pulmonary Atresia with intact septum (PA/IVS)
 - Tetralogy of Fallot (TOF)
 - Total Anomalous Pulmonary Venous Return (TAPVR)
 - Transposition of the Great Arteries (TGA)
 - Tricuspid Atresia (TA)
 - Truncus Arteriosus Communis (TAC)
 - CCHD Activities
 - DSHS distributed survey to all birthing hospitals and birth facilities to determine current readiness to implement CCHD screening
 - Survey developed by D'Andra Morin, Debbie Freedenberg and Dorothy Mandel
 - 60/96 (62.5%) perform CCHD screening
 - 48/60 (80%) of those performing CCHD screening use recommended algorithm
 - CCHD in Texas
 - Funded Texas Pulse Oximetry Project (TxPOP) an Educational Initiative
 - Dr. Alice Gong (UT San Antonio Health Science Center) and Dr. Charleta Guillory (Baylor College of Medicine) co-principal investigators
 - Developed CCHD tool available on DSHS website as well as Texas Pediatric Society website

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- Tool Kit contains multiple educational materials
 - Algorithm brochures for parents and health care providers, nurse information, technical information
 - DSHS funded TxPOP2 related to NICU protocols and rural hospitals
- NBS Education Efforts
 - NBS Grand Rounds 2013-2014
 - Dr, Elana O’Campo-CCHD
 - Dr. Priya Kishnani-Pompe Disease
 - Tales from the Crib
 - NBS Journal Club Educational Outreach
 - Overview of NBS-available both as webinar and in person presentation
 - CF
 - SCID-in development
- Genetics
 - Activities
 - Sponsored through MCH funds
 - Two Genetics Centers funded for Clinical Care (other facilities requested non-renewal)
 - Fee for Service
 - Teratogen Information Service contract awarded to UT Houston
 - Community based genetics seminars-Baylor Evening with Genetics
 - Three medical provider educational conferences planned, Tales from the Crib, Newborn Screening
 - Conference sites
 - San Antonio 7/26/2014
 - Dallas 8/2/2014
 - Austin 8/9/2014
 - Projects
 - Funding of project to explore how changes in the health care system impact genetic services-UNT
 - Funding of project to update educational content of the DSHS Genetics website-UT Houston
 - Funding of 12 clinical genetics medical student summer 2014 internships
 - UT Southwestern/Dell Children’s Hospital
 - Baylor/Children’s Hospital San Antonio
 - UT Houston
 - UTMB
- Potential New Conditions
 - Pompe

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- The Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC) voted to add Pompe disease to the RUSP on May 17, 2013
- HHS Secretary Sebelius requested the Interagency Coordination Council (ICC) to review with recommendation by July, 2014
- Mucopolysaccharidosis Type 1 (MPS1)
 - DACHDNC sent to formal evidence review
- X-Linked Adrenoleukodystrophy (X-ALD)
 - Next condition to have formal evidence review

Transit Time for Bloodspots-Brendan Reilly

Brendan Reilly discussed with the committee the details regarding the progress that has been made on transit time for bloodspots.

- Presented graph that outlines monthly delay in transit times on screens received > 96 hours
 - Big spike in 12/13 due to the Thanksgiving and Christmas holidays
 - 3/14/14 - courier service added for facilities that submit first screen specimens and have the capacity to collect specimens for Sunday pick-up
 - 4/8/14, Spotlight Newsletter; Clinical Care Coordination Newsletter issue focused on the importance of decreasing transit times.
 - 4/14/14, initiated Tier 2 Courier pilot; Pilot courier project initiated under existing FedEx and Lone Star Overnight contracts to assess the efficacy of a delivery service for newborn screening specimens only. Fifty-four submitters added as of 5/16/14
 - 4/22/14, Dr. Lakey/THA Letter; letter developed by DSHS and signed jointly with THA; letters were mailed out 4/22/14.
 - 5/17/14, monthly Spotlight expanded; Additional recognition added for lower volume and 2nd screen only submitting facilities who meet specimen transit time and satisfactory specimen quality measures 100% of the time.
 - 5/31/14, 73 sites were participating in Tier 2 Pilot.

Secondary Panel-David R. Martinez

David R. Martinez gave the committee an update on the secondary panel. Mr. Martinez informed the committee that Executive management is still evaluating the secondary panel and looking internally for the funding. The Department was evaluating whether to go out with an exceptional item; however, Dr. Lakey would like to look internally for the funding. Currently, there are several system related items that need decisions prior to going forward with the secondary panel. These include a major Laboratory Information Management System (LIMS) upgrade and the current Request for Proposals (RFP) for the testing reagents, kits, equipment and LIMS; and an operating system hardware upgrade.

Newborn Screening Rules Update-CCHD, Hearing-Karen Hess, David R. Martinez

Karen Hess gave the committee an update on the newborn screening rules. The CCHD rules are moving forward and making good progress. A 30 day public comment period ended May 11, 2014. A few

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comments were received and have been incorporated into the rules where possible. The rules are going through the DSHS/HHSC approval processes. Hopefully, these will be implemented by September 2014. Ms. Hess presented the most current reporting form available for the committee's comments.

David R. Martinez gave the committee an update on the newborn screening hearing rules. The rules went out for informal comments which were shared with the committee. The informal comment period went for 10 days ending on May 10, 2014, and the relevant comments were incorporated. We are moving forward with the rules. We are on the agenda to propose the rules to the State Health Services Council on August 14, 2014. We hope to have these rules published in the Texas Register around the beginning of October 2014. Then we will go into a 30 day public comment period which should end around the beginning of November 2014. Mr. Martinez reported that previous rules were repealed. The rules have been restructured with the important parts of the process put right up front. The committee will see that we are addressing confidentiality of data up front, our information management tracking system and screening follow-up and reporting. We really condensed the rules and what helped us also to do that, we took the entire newborn screening certification process out of the rules and put them in a policy, which we are posting on our website. This allows for updates to the certification process without the need for rule-making.

Department of State Health Services Sunset Review Impact on NBSAC-David R. Martinez

David R. Martinez gave the committee an update on the Sunset Review activities. The Sunset Commission has issued their report on DSHS. The report is posted on their website and DSHS has responded to the report. You can see the agency's response on the DSHS home page. In the Sunset report, *Issue 8: DSHS' Numerous Advisory Committees Lack Strategic Purpose, Limiting Their Effectiveness and Wasting Resources* lists the Newborn Screening Advisory Committee, identifying it as an active committee. They are making recommendations to eliminate some inactive committees. The recommendation is also to put advisory committees into rules because the department does have authority to create advisory committees. Mr. Martinez suggested that the committee take the time to read the report. During the last newborn screening rules update the NBS Advisory Committee was added to the rules, which is one of the Sunset Commission's recommendations.

Other Business

David R. Martinez gave an update on the NBS Advisory Committee. The committee had some members whose terms had expired. We did solicit during the month of May for 3 positions; 2 of the 3 are the new positions that were added by the Legislature during the last session. Solicitation for 1 hospital representative, 1 health care provider involved in newborn screening and 1 physician was publicized. Thirteen applications have been received and those applications are currently under review by the internal review team. The review will be completed during the first week of July. The reviewers' results will be compiled and submitted to Dr. Lakey with their recommendations. Committee members are expected to be in place by the end of July.

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Public Comments

None

Proposed Agenda Items

Discuss Sunset Review recommendations and updates

Discuss NBS 50th Anniversary

Adjournment

The next meeting is tentatively scheduled for October 2, 2014. The time and location to be determined. There being no further business, the meeting was adjourned at approximately 12:00 p.m.