Texas Department of State Health Services
1100 W. 49th, Austin, Texas 78756
Moreton Building, M2-204
June 18, 2015 via conference call
10:00 a.m. – 2:00 p.m.
Minutes

Members via Conference Call
William Morris, LVN
Alice K. Gong, MD
Charleta Guillory, MD
Thomas M. Zellars, MD
Scott D. McLean, MD
Felicia M. Adams, MSN
Aida Gonzalez, RN

Staff
Beth Rider, Department of State Health Services (DSHS), Ombudsman, Committee Support
Karen Hess, DSHS, Newborn Screening Genetics Branch Manager
Patricia Hunt, DSHS Laboratory, Metabolic Screening Group
Brendan Reilly, Program Specialist, DSHS Laboratory, Biochemistry & Genetics Branch
Debra Freedenberg, MD, PhD, DSHS Newborn Screening Unit
Rachel Lee, PhD, Branch Manager, DSHS Laboratory, Biochemistry & Genetics Branch
Susan Tanksley, PhD, DSHS, Laboratory Operations Unit Manager
Eugenia Dunham, DSHS, Newborn Screening Support Group Manager
Lynette Borgfeld, DSHS, Laboratory, Endocrine, Hemoglobinopathies
D'Andra Luna, DSHS Laboratory, DNA Analysis Group
Michelle Shaffer, DSHS, Newborn Screening Endocrine Group
Sam Cooper, DSHS, Specialized Health Services Section

Guests
Shannon Lucas, March of Dimes
Daniela De Luna Olivares, Health and Human Services Commission (HHSC)
Sanjiv Harpavat, MD, PhD via conference call

Call to Order and Roll Call of Committee Members, Staff and Guests
Chairman Morris called to order the June 18, 2015 meeting of the Newborn Screening Advisory Committee at approximately 10:15 a.m. Introductions were made and Chairman Morris welcomed everyone. Members, staff and guests attending are listed at the beginning of these minutes. A GoToMeeting link was provided for the benefit of Committee members to allow them to visually follow along with each agenda topic and PowerPoint presentation.

Review and Approval of Minutes
Chairman Morris asked if everyone had received a copy of the minutes from the February 27, 2015 meeting. He asked the Committee if there were any concerns or changes that needed to be made. No changes or concerns were noted and Chairman Morris requested that a motion be made to accept the minutes. Dr. Guillory made a motion to approve the minutes as is. Motion passed.

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Timeliness of Newborn Screening – Rachel Lee, PhD, Brendan Reilly

Rachel Lee and Brendan Reilly provided updates on the timeliness of newborn screening. A PowerPoint presentation was presented, which included an Overview that focused on Pre and Post-analytical Measures, (including a national comparison, a current status, and cost estimates)

- Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) provided recommendations earlier this year. In the recommendations, they specified that the presumptive positive results for time-critical conditions should be communicated to the healthcare providers within five days of life. All abnormal results of the other conditions should be reported to the healthcare providers within seven days of life. All results should be reported within seven days of life. In order to meet these standards, the ACHDNC also recommends that specimens should be collected within 48 hours after birth and specimens should be received in the laboratory within 24 hours after collection. The recommendations also include a goal to achieve compliance of more than 95% by the end of 2017.
- The Association of Public Health Laboratories (APHL) conducted a national survey to gather data regarding timeliness for all 50 states. Data gathered between January and May 2014 revealed 87.5% of first screen specimens were collected within 24-48 hours of life, which was above the national median (82.2%). However, during this timeframe, only 20.5% of specimens were received at the DSHS Laboratory within 24 hours of collection (national median = 25%). More recent data (July to December 2014) show that 96.2% of first screens were collected within 48 hours after birth (meeting the 95% goal), and 23.8% of first screens were received within 24 hours of collection.
- A Transit Time Project was initiated in 2013 to decrease time from collection to receipt in the DSHS Laboratory. Transit time decreased during 2014, from approximately 70% of first screens received within three days of collection to about 90% of first screens received within three days of collection. Since late 2014, however, the results have plateaued.
- Receipt within 3 days of collection is still the goal being used in Texas.
- Gaps and Barriers to improvement?
    - Submitters –
        - Consistent poor performers
        - Inefficient use of courier service
        - Delayed delivery of specimens to lab from nurseries
        - Inadequate/weekend staffing
    - DSHS Couriers
        - Current couriers do not cover all submitters
        - Courier operating hours
        - Weather and holiday delays
        - Geographic size of state

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    - Laboratory
        - No receiving on Sundays or major holidays
        - Lack of resources to provide education to low volume problem sites
        - Lack of resources to monitor efficient use of courier service
        - Inefficient/incomplete escalation system to monitor worst performers
- New initiatives
    - Escalation team for persistent poor performers
    - Identify top 10 based on percentage delayed as opposed to volume delayed
    - Courier cost analysis
    - Investigate needs for additional staff to provide onsite education
- DSHS Courier
    - Study was conducted based on two Tiers of courier service [Lone Star Delivery and Processing (full-service courier) and FedEx]
- Analytical and post-analytical measures in Texas for July to December 2014 – 30.7% of presumptive positive results for time-critical conditions were reported within five days of life, 79% of all other presumptive positive results for non-critical conditions were released within seven days of life and 25.4% were reported within seven days of life.

Secondary Panel Implementation – Debra Freedenberg, MD, Rachel Lee, PhD

Dr. Debra Freedenberg informed the Committee of the screening for the secondary conditions as implemented on May 26, 2015. The Texas Newborn Screening statute requires newborn screening for disorders listed on the Recommended Uniform Screening Panel (RUSP) to the extent that funding allows. It exempts screening for galactokinase and galactose epimerase deficiencies. The DSHS Newborn Screening Laboratory performs testing for all laboratory-based, core disorders on the RUSP except for Pompe disease which was added to the RUSP in March 2015. Hearing and Critical Congenital Heart Disease (CCHD) screening are point-of-service tests typically performed at the birthing facility. There are 24 additional secondary conditions that were added. Some of the secondary conditions may have an unclear natural history or lack appropriate medical therapy that affects long-term outcome. They are detected during screening for core conditions and no additional blood spots will need to be collected. The additional conditions will be detected from the same specimen using existing assays. Of the 24 conditions, we detected 18 of them and so we are really only adding six new conditions. There were three new Fatty Acid Oxidation Disorders, two Organic Acid Disorders and one Amino Acid Disorder added. ACT/FACT sheets for each condition on the secondary panel have been created and are available on line. At this time, there will not be a change in the newborn screening fee. However, we plan to conduct a work load unit study to re-evaluate the newborn screening fee after finalizing a new newborn screening IT contract in 2015.

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Dr. Lee informed the Committee that everything went pretty well during the implementation. There were a few bumps as expected, but the Laboratory staff are working through them. In the first three weeks of testing, there were about 30 actual specimens that were reported out as presumptive positive for the new secondary disorders.

X-ALD Public Health Impact Assessment – Debra Freedenberg, MD, Susan Tanksley, PhD

Dr. Freedenberg gave a summary of X-ALD from a clinical perspective to the Committee. X-ALD stands for X-Linked Adrenoleukodystrophy. X-Linked means the gene is located on the X chromosome. It is a metabolic disorder that affects the central nervous system and adrenal glands. It is due to a mutation in the ABCD1 gene. ABCD1 stands for ATP Binding Cassette, sub-family D, first member 1. It is the only gene in that particular sub-family that has been identified. It affects metabolism with very long-chain fatty acids, and it is a peroxisomal disorder. It can present as spectrum of disease, typically with progressive neurological decline which is the cerebral ALD. Seventy percent of boys affected also have an adrenal insufficiency, called Addison's disease. It can present across a life span. Neurologic involvement can also occur later in adolescence or adulthood. Females can be identified as being carriers or heterozygous for this mutation, and up to 20% can present neurologic symptoms in later adulthood. The typical onset of this condition in terms of clinical symptoms would not be during the neonatal period. The treatment is stem cell transplantation. Usually that is not done until there is MRI evidence of some brain damage that occurs with the continued lack of ABCD1 gene expression. Estimates are about 1 per 20,000 male births. There are some states that are screening. Clinically, about 35% to 40% of patients have adult onset of cerebral adrenoleukodystrophy. There has also been a gene trial that is ongoing and/or has been concluded, but hasn't been thought to be the answer. This condition is currently undergoing evidence review for the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) as part of the assessment to determine if the condition should be added to the Recommended Uniform Screening Panel (RUSP).

A public health system impact assessment is now conducted for all conditions that are under consideration for addition to the RUSP. All states are asked to provide feedback on the impact to their state if the condition is added as well as barriers to implementation and timing for implementation. A summary of the nationwide data is shared with the ACHDNC as part of the evidence review. Texas Newborn Screening Advisory Committee members and metabolic specialists were asked to provide input from their perspectives. The public health system impact assessment to the Association of Public Health Laboratories was due June 17, 2015. Dr. Tanksley gave a summary of the Texas responses to the survey. It would take less than one year to receive authorization as we have authorization once a condition is on the RUSP. We would need more staff. To get funding, we estimated that it would take one to three years after it was added to the RUSP. Our primary challenges would be to provide the screening tests and then clinical coordination for X-ALD as well as long term follow-up for carriers and individuals with

peroxisomal disorders. By adding a new condition, it impacts other improvement activities and other things that we are trying to do. The cost per specimen to conduct the screening is unknown right now, as is the cost of treatment for newborns diagnosed with X-ALD. This is a public health system impact assessment - not a newborn screening program assessment. So when you think about the cost of treatment from a system perspective, it would be very high.

Parental Consent Form Changes for Newborn Screening – Susan Tanksley, PhD

In 2009, new legislation required DSHS to provide disclosure that the newborn screening specimen would be saved and could be used for research. If parents wanted to have the specimens destroyed, they could fill out a form and send it to us or have the physician send it to us, and then we would destroy that specimen within 60 days. In the next legislative session, HB 411 changed the law to require consent for retention and external research use of the specimens. The form was changed to reflect this change. In June 2012, the language was changed to reflect this opt-in process and is currently part of the existing newborn screening collection kits. In December, 2014, a federal law passed which essentially makes any federally-funded research using newborn screening specimens into human subject research, thus requiring informed consent in order to utilize the specimens for research. We have determined that our existing forms do not meet the requirements of informed consent. National discussions suggest that broad consent will be allowed in the next revision of the Common Rule. We are trying to reach a level we feel meets the elements of broad consent. Dr. Tanksley asked the Committee to email Beth Rider if they have any feedback regarding the forms.

Medicaid Managed Care (MCO) Case Management – Daniela De Luna Olivares

Daniela De Luna Olivares from HHSC Program Operations gave an overview of Medicaid Managed Care (MMC) to the Committee. In addition to explaining the concept of managed care, Ms. De Luna Olivares gave a PowerPoint presentation with some of the components she thought would be of interest with regards to newborn screening, such as:

- Provider Network
- Prior authorization requirements
- MSHCN and service management coordination
- Challenges, barriers and recommendations

Ms. De Luna Olivares reported that approximately 85% of Medicaid recipients are enrolled in Managed Care Organizations (MCOs). She discussed the different programs in MMC, such as STAR, STAR+PLUS, and STAR Health. Currently, the STAR MCOs are required to identify Members with Special Health Care Needs (MSHCN). All STAR+PLUS members are considered MSHCN. Members with STAR MCOs receive service management, which is the same as case management. An individualized plan is developed by the MCO which includes access to specialty care providers with experience serving MSHCN. Service coordination is provided by STAR+PLUS and STAR Health MCOs for all members enrolled.

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TxPOP2 Project Update – Charleta Guillory, MD, Alice Gong, MD

Dr. Guillory and Dr. Gong gave updates to the Committee on the TxPOP2 Project. Dr. Guillory stated that TxPOP1 was a joint regional, educational, and quality improvement project initiative of the University of Texas Health Science Center of San Antonio, Baylor College of Medicine, and DSHS. The project was initiated to provide screening for apparently healthy newborns to identify Critical Congenital Heart Disease (CCHD). The topics discussed with the Committee were presented in a PowerPoint presentation.

- Quality improvement – February to July
  - 12,946 births in the 13 facilities
  - 96% of babies admitted received a CCHD screening in the recommended time frame
  - 11 positive screens
    - One had severe CCHD from secondary target
    - Two were in the <90% group
    - Seven were the indeterminate of 90-95%
    - One had > greater 3% difference
- HB 740 mandated that all Texas babies be screened for CCHD, not just apparently healthy babies in newborn nurseries
- CCHD screening using pulse oximetry

Dr. Guillory and Dr. Gong discussed several cases of interest.

Biliary Atresia Follow-up – Sanjiv Harpavat, MD

Dr. Harpavat gave the Committee an update on biliary atresia. He gave a PowerPoint presentation which will not be repeated in the minutes. Dr. Harpavat reported that early intervention results in better outcomes. The statistics show the procedure Kasai Hepatoportoenterostomy performed earlier following diagnosis improves the patient's outcome. However, the Biliary Atresia is being diagnosed later, negatively affecting patient outcome. The studies are ongoing to determine ways to make screening for Biliary Atresia more acceptable.

Legislative Session Summary – Karen Hess

Karen Hess gave the Committee an update on the Legislative session. The Committee was provided a hand-out with the summary. Ms. Hess discussed SB 791 which will require DSHS to develop materials for Cytomegalovirus (CMV) education in conjunction with the Texas Medical Board. The provision that was removed by the House from this bill was the provision for CMV testing for newborns who are deaf at birth. The reason for testing is so that CMV diagnosis can be made early and babies and their families can be educated through appropriate case management.

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The Sunset bills initially recommended the consolidation of all five agencies. The legislature decided to take a more graduated approach to consolidation and passed bills that would restructure the agencies over a longer period of time. Ms. Hess pointed out that the Department of Family and Protective Services (DFPS) and DSHS would remain as independent agencies with a 2023 Sunset review. Dr. Freedenberg informed the Committee that the Interagency Council for Genetic Services (IACGS) was abolished by SB 219, 84th Regular Legislative Session.

Public Comments
None

Future Agenda Items
1) Celebration of Secondary Panel
2) Pompe
3) CCHD NICU screening

Adjournment
The next meeting will be in October, 2015; however, the final date has not been determined. This will be a meeting to be held at DSHS in the Moreton Building. There being no further business, the meeting was adjourned.