Newborn Screening Project Charter Report

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September 2005

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Executive Summary

Over the past year, there has been considerable attention and focus, nationally and at the state level, on proposals to increase the number of disorders screened in newborns. Newborn Screening (NBS) is best understood as a comprehensive system beginning with an initial collection of blood to screen newborns through a follow-up and tracking process with an eventual outcome of treatment and care of children confirmed with a disorder. Currently, each year in Texas approximately 375,000 children are born and about 96% of them are screened for genetic disorders. Of the 3,750,000 screens performed annually, Department of State Health Services (DSHS) case management staff follow-up on approximately 10,000 children who have abnormal screens and require additional testing. Ultimately, about 400 of the children screened are confirmed with a genetic disorder.

DSHS invited the National Newborn Screening and Genetic Resource Center (NNSGRC) to review Texas' NBS program in February 2005. The review identified multiple issues and recommendations for improvements to the NBS program. In April of 2005, DSHS Divisions of Family & Community Health, and Prevention and Preparedness partnered to establish a NBS Project Charter. This report briefly describes the activities, as well as, additional tasks undertaken by the Project Charter Team. The passage of House Bill (HB) 790, 79th Regular Session, charged DSHS with multiple activities, one of which was to review the NNSGRC report. This document includes brief descriptions of the parallel activities of the past year, as well as a summary of the NNSGRC Draft Report. The report of the NBS Project Charter Team also describes additional activities, literature reviews, and surveys undertaken by the workgroup to garner additional stakeholder input and a better understanding of trends in NBS. The NBS Project Charter team has presented recommendations for consideration based on the NNSGRC review and the Team's additional research that if accepted will enhance the NBS program in Texas.

Background/Issue
In April of 2005, the DSHS Divisions of Family & Community Health, and Prevention and Preparedness partnered to charter a NBS Project. The project's purpose was to improve the department's NBS program. Activities included: review of the NNSGRC Draft Report (Appendix 1), analysis, recommendations and implementation of a plan as approved by DSHS leadership. The work associated with this NBS Charter Project occurred in the context of parallel activities including national activities and recommendations regarding expansion of NBS, DSHS's approved exceptional item to expand NBS, and the passage of HB 790, 79th Legislature, Regular Session, 2005. Further explanation of these parallel activities, tasks of the NBS Project Charter Team and recommendations follow.

National Newborn Screening and Genetics Resource Center (NNSGRC) Review
From February 28, 2005 to March 2, 2005, the NNSGRC conducted a technical review of the NBS Program in Texas. NBS technical reviews are conducted through a cooperative agreement between the federal Maternal and Child Health Bureau Genetic Services Branch of the Health Resources and Services Administration (HRSA) and the University of Texas Health Science Center at San Antonio Department of Pediatrics. The process included an in-depth, onsite review, evaluation of program materials, interviews with staff, discussions with stakeholders and responses to specific questions posed by the program and external stakeholders.

The Texas review team was comprised of nine professionals from across the country, including a representative from the Centers for Disease Control and Prevention (CDC) and a HRSA representative. Internal and external stakeholder meetings were held in Austin, Houston, San Antonio, and Dallas. Approximately 51 external stakeholders representing 20 unique interest groups participated. A draft report from the NNSGRC review was received on April 20, 2005. The report provided comments from the review team and stakeholders, responses to specific questions, and recommendations. To date, a final report has not been received, but is anticipated in the near future.

Historical and Current Newborn Screening (NBS) in Texas
NBS began in Texas in 1963 as a Phenylketonuria (PKU) pilot program. In 1965, the Legislature adopted a statute (Chapter 262, Vernon's Ann. Civ. St.) requiring population-wide NBS for PKU. Over the next forty years, the program expanded to include additional disorders. Texas rules (Texas Administrative Code 25; Chapter 37.56) require two screenings per newborn; the first is recommended at 72 hours of life and the second at 1-2 weeks of age. Currently, DSHS receives approximately 750,000 newborn specimens annually, screening each specimen for seven disorders. This translates into more than 3,750,000 tests performed per year. The disorders currently screened include: Phenylketonuria, Galactose-1-Phosphate Uridyl Transferase Deficiency (Classical Galactosemia), Congenital Hypothyroidism, three Hemoglobinopathies, and Congenital Adrenal Hyperplasia. Annually, approximately 10,000 abnormal results require follow up and about 400 infants are diagnosed with one of the disorders. In addition to the seven blood specimen screenings done in Texas, newborns are screened for hearing loss if they are born in hospitals or in larger birthing centers in counties with populations over 50,000.

National Trends in NBS/Expansion
In March 2005, the American College of Medical Genetics (ACMG) recommended a uniform panel of NBS disorders and a uniform NBS system that included a recommendation that states mandate newborn screening tests for 29 disorders (http://mchb.hrsa.gov/screening/summary.htm 8-12-8005). In addition to those 29 "core" disorders, the ACMG identified an additional 25 disorders as "secondary targets" for NBS. Before the ACMG recommendation, the major initiative for a uniform NBS panel was from the March of Dimes, which recommended newborn screening for 10 disorders.

The March of Dimes now supports the ACMG recommendations. Additionally, in July of 2004, the Department of Health and Human Services, Maternal and Child Health (MCH) Bureau, recommended that states develop educational materials that included guidance to parents regarding options for screening additional conditions not mandated by their state.

Population screening of newborns began in the 1960s when testing for PKU, an amino acid metabolism disorder, was instituted in some states. Between 1960 and 1990, sickle cell, endocrine, and carbohydrate disorders were added to many state NBS programs. Currently all 50 states have a NBS program. As of August 2005, 14 states mandated screening for more than 30 NBS disorders, and 32 states require the use of tandem mass spectrometry (MS/MS) for newborn screening: (http://genes-r-us.uthscsa.edu/nbsdisorders.pdf 8-12-2005). In the year 2000, only seven states mandated screening using MS/MS: (http://genes-r-us.uthscsa.edu/resources/newborn/00/ch2_complete.pdf 8-12-2005).

By the early 1990s, MS/MS technology emerged to meet the need to quickly screen large numbers of newborns for disorders in amino acid metabolism, organic acidemias (http://www.msud-support.org/web11_2.htm 8-12-8005), and fatty acid oxidation. These disorders have been recognized since the 1930s, 1960s, and 1970s, respectively, as causes of infant morbidity and mortality. Twenty of the 29 core disorders are detectable by MS/MS, and the Texas NBS program could increase the number of recommended screened disorders by 19 with the use of MS/MS. Of the 25 secondary targets, MS/MS detects 22 of these disorders.

79th Regular Session, 2005- HB 790 and Legislative Appropriations Request

In its Legislative Appropriations Request (LAR) to the Legislature for Fiscal Years (FY) 2006 and 2007, DSHS proposed an exceptional item to expand the NBS program by adding four disorders: glutaric acidemia type I (GA1), maple syrup urine disease (MSUD), homocystinuria, and medium-chain acyl-CoA dehydrogenase deficiency (MCADD). A task force convened in 2002 by the legacy agency, Texas Department of Health, recommended this additional screening panel. The task force, comprised of medical experts, geneticists, physicians, parents, and legislative representatives, made this recommendation based upon the reliability of the screen, frequency of the disorder, available treatment, and the March of Dimes' recommendation. All four of the disorders can be detected by MS/MS.

In its LAR, DSHS proposed the following increase in appropriation and full time equivalents for the NBS program in order to expand it to perform tests for an additional four disorders.

Funding	2006	2007	Biennium
General Revenue	$ 3,217,146	$ 702,002	$ 3,919,148
Medicaid	$ 32,769	$ 34,227	$ 66,996
Interagency Contract	$ 32,769	$ 34,227	$ 66,996
Public Health Svc Fee		$ 1,486,218	$ 1,486,218
Medicaid Reimbursement		$ 1,857,912	$ 1,857,912
	$ 3,282,684	$ 4,114,586	$ 7,397,270

Personnel (Full Time Equivalents)

7 Laboratory Staff
2 Case Management Staff

During its Regular Session, the 79th Legislature passed HB 790, effective September 1, 2005, which amends Chapter 33 of the Texas Health and Safety Code, the statute that governs the Texas NBS

Program. The amendments provide a framework to govern the expansion of the NBS Program funded by Senate Bill (SB) 1, the General Appropriations Act.

Key provisions of HB 790 include:

- DSHS is required to review and study the assessment by the NNSGRC of the current Texas NBS Program. This report documents the methodology and recommendations of that review.
- DSHS is charged with consulting the report of the ACMG "or another report determined by the department to provide more appropriate newborn screening guidelines" in determining which tests are required under the program. The ACMG report envisions the expansion of NBS Programs to 29 tests (disorders). Texas currently requires eight of the recommended disorders, including hearing.
- The extent of the expansion of the program is based on available funding.
- DSHS is allowed to adjust the fees charged for the NBS Program.
- DSHS is required to conduct a cost-effectiveness study to determine whether the best means of performing newborn screening services is to use a private laboratory and case managers or continue to perform the services itself. The study, the results, and methodology will be submitted by March 1, 2006, to the Governor's Office.
- An expanded NBS program must be implemented by November 1, 2006.

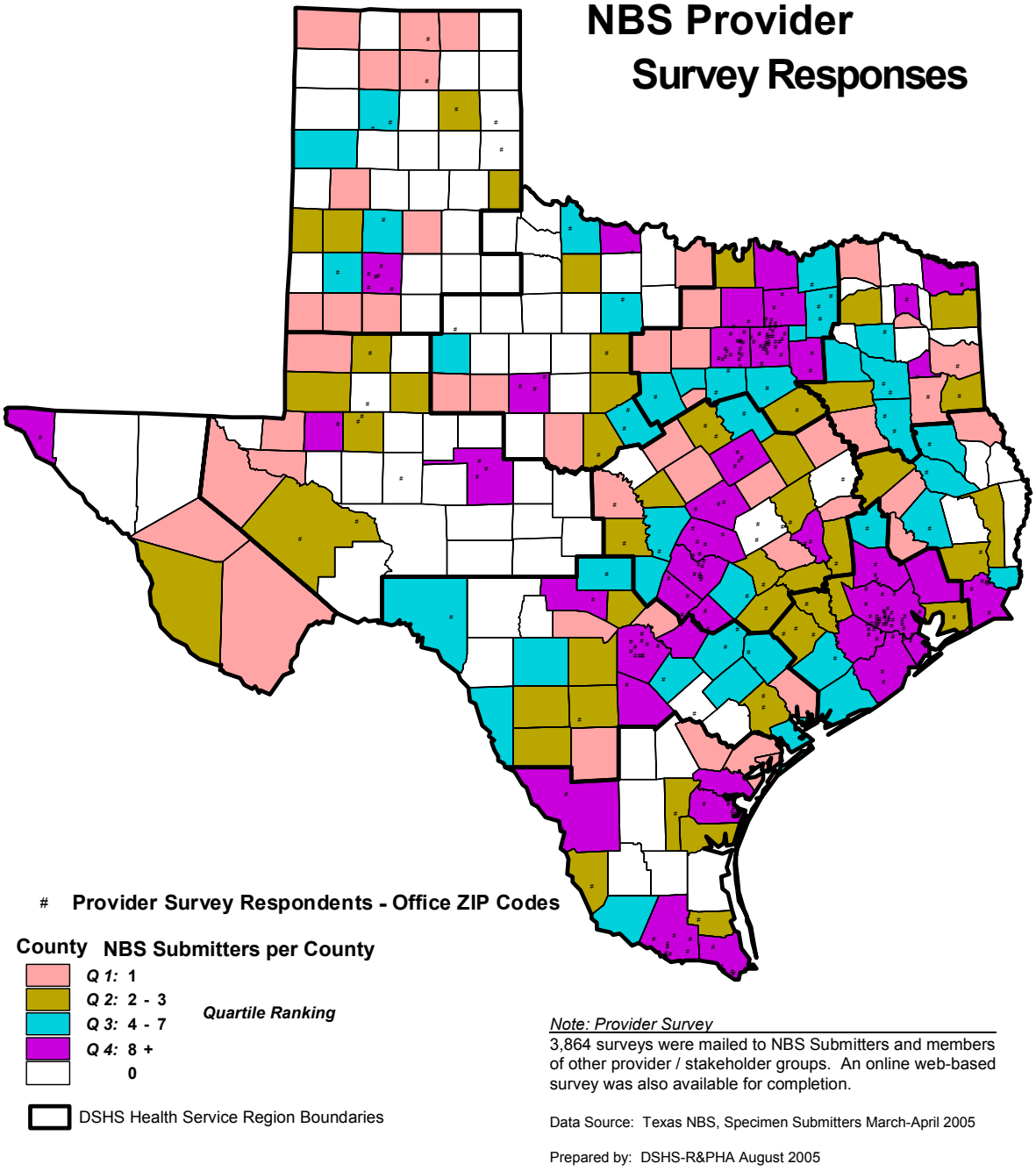
NBS Project Charter

In response to the NNSGRC review, DSHS formed a NBS Project Charter in April 2005 (Appendix 2). The Charter involved the review and analysis of the NNSGRC Draft Report and Recommendations. The NBS Project Charter Team was comprised of staff from the Divisions of Family & Community Health and Prevention and Preparedness, the Centers for DSHS Policy and Innovation, Consumer and External Affairs, and HHSC staff appointed by Commissioner Hawkins. This initiative and resulting report serves to meet the intent of one mandate of HB 790.

After an initial, thorough review of the NNSGRC draft report, the team developed an issues list and utilized it to structure subsequent research and analysis. The issues were grouped into the following categories: funding, stakeholder involvement, specimen collection and laboratory testing, follow-up, diagnosis/treatment and management, evaluation and education, and program management.

The NBS Project Charter Team conducted additional literature reviews on the issues identified in the NNSGRC review, and conducted surveys of both providers and parents. The provider survey was performed by DSHS in July of 2005. Surveys were distributed to 3,864 identified providers/stakeholders who interact regularly with the NBS Program. Additionally, members of organizations representing providers and affected professions, including the Texas Medical Association (TMA), Texas Hospital Association (THA), Board of Nurse Examiners (BNE), Texas Nurses Association (TNA), Texas Association of Family Practitioners (TAFP) and Texas Pediatric Society (TPS) were provided notice and invitation to participate in the survey by way of newsletter, web-links, banner messages, and email. A total of 403 provider surveys were received, entered, and analyzed. Figure 1 shows the response distribution.

Figure 1



In addition, a telephone survey was conducted, which interviewed parents whose infants had had a positive screen for a genetic disorder, resulting in a subsequent diagnosis in calendar year 2004. A subset, 10% of the total diagnosed cases, was identified as potential survey participants. NBS staff successfully contacted 36 or 3.7% of the total number of cases that were diagnosed in calendar year 2004.

Results of both surveys were compiled and analyzed by DSHS Research & Public Health Assessment (R&PHA) Office (Appendix 3& 4).

Summary of Issues Identified in the NNSGRC Draft Report, Project Charter's Additional Research, Survey Findings, and Actions to Date

NNSGRC Reported Funding Issues

- Significant discrepancies exist in funding between the laboratory and follow–up services.
- Increased funding is needed to ensure adequate infrastructure and follow-up.
- Grant funding to subspecialties should be considered.
- Fees in Texas are low in comparison to other states; need to evaluate fee structure and determine if continuum of services versus just the laboratory costs can be fee-based.
- Fee collection mechanism should be evaluated.
- Providers expressed concern regarding inadequacy of insurance reimbursement.
- Stakeholders expressed concern regarding un-funded community role in assuring appropriate follow-up.
- Consideration of a fee increase in advance of expansion.
- Consideration of fee incentives.

Funding- Additional Findings and Actions to Date

The source, cost, and breakdown of funding for NBS services vary widely between states. In Texas, one of the primary funding mechanisms for the laboratory component of the NBS Program consists of a basic charge (fee-for-service) to hospitals and providers, who collect the specimen from the newborn. The charge covers the cost of a supply of filter paper specimen collection devices (NBS cards), which are color-coded to indicate whether they have been used to collect from fee-for-service, Medicaid, or charity care newborns, and are shipped to the providers in bulk.

Texas' current fee-for-service charge of $19.50 per test panel is low in comparison to other states, but slightly more than the average $15 (Appendix 1). The average cost of $15, according to research conducted by PricewaterhouseCoopers LLP, represents, as in most states, the cost of processing specimens. Charges in a small number of other states, such as Wisconsin and Utah, may also cover the cost of initial and repeat testing, metabolic formula, short-term follow-up, and counseling. Although Texas has the authority to charge up to $38 for the filter paper/card, the fee has not been increased since April 2002. Currently, DSHS is proposing rules to allow for an increase in the fee up to $40 per test.

In Texas, roughly fifty percent of all test panel requests are for Medicaid-eligible newborns. The remaining fifty percent are for newborns eligible for charity care or who have private commercial insurance coverage. For FY 2005, Texas estimates that total revenue of $13.1 million will be generated from NBS laboratory services based upon historical records and the estimated number of newborns, born in the state.

The amount of funding for the case management follow-up components of the NBS Program in FY 2005 was approximately $700 thousand out of a total budget of $1.9 million for genetic services. The remaining $1.2 million was utilized for genetic evaluations and counseling, genetic testing, prenatal counseling, education, and outreach contracts serving pregnant women, children and fathers. The funding source included a mixture of Title V MCH Block Grant and Medicaid dollars.

The experience of other states demonstrates that expansions of state public health NBS programs are almost invariably accompanied by an increase in funding to support the expansion. When only considering the technical component, the best estimate for calculating the cost to add additional screening tests is to add the unit cost of the specific test. Screening for additional disorders would

produce more abnormal and positive screening test results and would require added follow-up activities. In the state of California, for example, $2.7 million was appropriated for expanded screening in 2005. The budget for that state's expansion was calculated to cover additional staff in the laboratories and on follow-up teams, as well as enhanced data capacity.

The Texas NBS program is also considering a proposal to increase funding and balancing the disparity between the laboratory and case management. Five additional positions have been allocated to case management for follow-up since June 2005. Additionally, the increase in funds appropriated by the Legislature will allow DSHS to add two positions, and develop educational materials for providers and the public.

Results of the provider survey described stakeholders' opinions and experiences with funding. Less than one-fifth of the respondents (15%) said that billing for NBS services, in comparison with other medical services, was easy or very easy. Likewise, less than a tenth (10%) said that reimbursement to hospitals and physicians for NBS from third party payers was adequate or very adequate, while 21.4% said it was inadequate or very inadequate. Over a quarter (25%) of respondents indicated NBS program services should be covered by fees: laboratory specimen collection (59%), follow-up of abnormal test results (48%), first and second specimen analysis (44% and 40%), transporting specimens to DSHS (38%), follow-up of unsatisfactory test results (36%), services of primary care physicians and pediatricians (36%), and education of parents and guardians (25%).

NBS Charter Team members met with the Texas Department of Insurance (TDI) on July 29, 2005 and established a dialogue regarding insurance reimbursement for NBS in Texas. The NBS Charter Team shared feedback from stakeholders as well as identified next steps to pursue in partnership with TDI to explore ways of addressing provider dissatisfaction with reimbursement.

NNSGRC Reported Stakeholder Issues

- DSHS should convene a formal broad-based, multi-disciplinary advisory committee with subcommittees
- Committee should consist of multiple stakeholders from within and outside of government including:
    - Other agencies: Texas Department of Insurance (TDI) and Health and Human Services Commission (HHSC)
    - Programs within DSHS: Children with Special Health Care Needs (CSHCN), Women Infants and Children (WIC), Birth Defects, Newborn Hearing Screening (NBHS) and Title V
    - Organizations: TMA, TPS, THS
    - Providers: family practice physicians, pediatricians, nurses, nutritionist, genetic counselors, subspecialty physicians, etc.
    - Consumers, family members, ethicists and advocacy groups
    - Insurance carriers- Health Maintenance Organizations (HMOs), Managed Care Organizations (MCOs) etc.
- Program decisions should be made by a formal committee process; appropriate decisions for committee to consider include:
    - determination of the adequacy of current infrastructure;
    - fees increases or structure changes; and
    - determination of disorders to be screened

- Consideration should be given to a NBS task force to define the elements of a comprehensive newborn screening system in addition to the over all broad based advisory committee

Provider and Parent Involvement and Education - Additional Findings and Actions to Date

Elizabeth Campbell, MA, and Lainie Friedman Ross, MD, PhD, in their study, *Incorporating Newborn Screening into Prenatal Care* (American Journal of Obstetrics and Gynecology (2004) 190, 876-877), explored parental attitudes about genetic testing and screening of infants and children for conditions that present through the life cycle, including NBS. The study included 13 semi-structured focus groups throughout the Chicago area, and participants were black and white parents representing diverse socioeconomic status. Participants were recruited through neighborhood institutions, elementary schools, churches, and a federally qualified health center. When parents were asked to recall the testing of their infants at birth, few were able to recall the information. After the purpose of NBS was explained, parents saw clear benefits of the screening, and wanted to have more information regarding the nature and purpose of the screening. Respondents suggested ways they could better be informed regarding NBS, including education about NBS. Parents indicated that education was not a viable option after labor and delivery as they were too preoccupied, but that information could be effectively understood during the prenatal period, and they could give informed consent, if required, or at least have the knowledge about what was being done to their infants in the postnatal period.

The Texas NBS parent survey conducted in July 2005, as well as the provider survey, also suggested a need for increased parent-focused education about NBS. Among parents whose infants had been diagnosed with a disease detected through NBS, many stated that they had not known much about newborn screening when their infants were first screened. When prompted, many parents suggested that information for the parents should be tailored specifically for their baby's diagnosis. A total of 89%, indicated the letter from NBS staff about their baby's positive screening result was easy to understand. Likewise, 83% said that the brochures and other information they had received after their baby's diagnosis was easy to understand. Taken together, these findings suggest that literature from NBS staff is generally effective but that general information could be made more widespread, and diagnosis-specific information would substantially help parents of infants with positive screens. The NBS program annually mails out the NBS brochure to all obstetricians to distribute to their prenatal patients. Once the NBS program is made aware of a child's diagnosis, specific disorder related brochures and booklets are mailed to parents.

The provider survey indicated that the majority of education to families regarding NBS occurs after delivery (68%). Almost half (43%) said they did not receive parent-focused NBS educational materials in the preferred languages of their families. The most commonly preferred languages were English and Spanish. Other preferred languages mentioned by respondents were: dialectical Texan Spanish, Vietnamese, Chinese, Arabic, French, Urdu, Korean, Tagalog, Hmong, and Japanese. Approximately a quarter of respondents (25%) said that they or a colleague on their staff would be available to help identify NBS needs and solutions for patient education.

Regardless of the need for materials, almost half (42%) of the provider survey respondents rated the parent-focused NBS educational materials as either effective or very effective, and 24% had a neutral opinion. Respondents' suggestions for improving the parent-focused educational materials revealed two primary audiences. The first audience suggested requires detailed information, especially about false positive results and downloadable fact sheets. The second audience suggested tends to have a lower educational level and/or lower literacy level, and respondents suggested simplification of medical terms and concepts, more pictures and other visual aids, a toll-free hotline

with a bilingual educator, videos and other multimedia formats, and expansion of the educational role of social workers. Respondents also advocated the use of NBS materials as part of prenatal care, parent-focused websites, and increased community awareness of NBS. Given these responses, it is recommended that the program identify mechanisms to influence the provider community to change the practice and provide education prior to delivery.

Currently, the NBS program convenes three *ad hoc* consultant groups annually. The groups are comprised of endocrinologists, hematologists, and metabolic specialists. The program plans to continue utilizing these groups and will identify avenues to enhance their role in reviewing protocols that guide the clinical aspects of follow-up, and parent and provider educational materials; furthermore, it is recommended that DSHS identify resources to compensate the specialists for their travel to and from Austin. Historically, Title V funds were utilized for this purpose, and participation has been negatively impacted since funds are no longer available to support travel costs for these specialists' services.

The provider survey indicated general concurrence with recommended members of an advisory group. The majority of respondents indicated that the areas they or their colleagues felt most able to advise in were patient education (25%) and patient follow-up, case management and systems of care (23%). Less than 10% of respondents stated that they would advise in more program-focused aspects of NBS, such as genetic counseling; infrastructure building; program evaluation and research; ethical, legal, and social issues; and cost-benefit analysis and funding. Together these findings might be related to providers' own desire and perceived need for training in NBS. This is supported by the majority (80%) of providers thought that professional organizations should include NBS as a continuing education topic, and approximately half wanted at least yearly training on new issues in specimen collection, expansion of newborn screening, and MS/MS technology (49%, 54%, and 48%, respectively).

On September 21, 2005, a health care summit will include NBS as a topic in order to obtain additional stakeholder feedback.

NNSGRC Reported Specimen Collection and Laboratory Testing Issues

- Recommend a formal education plan for providers to improve the inclusion of important demographic information
- Identify options for linking 1st and 2nd screens, as well as linking screens with birth certificates
- Recommend instituting an automated mechanism for providers to obtain results; ensure 24/7 access for providers
- Re-establish voice response system through a toll-free line
- Recommend consideration of courier services for timely transport of specimens
- Investigate ways to address delay in reporting certain conditions
- Recommend reporting preliminary results before weekends or holidays versus delaying
- Perform formal audit of 2nd test system

Specimen Collection and Laboratory Testing- Additional Findings and Actions to Date

Missing demographic information can cause laboratory specimens to be either delayed or rejected for testing. For example, on February 15, 2005, the date of specimen collection became a required field of information needed for testing. Though submitters had been notified of the new criteria, many specimens lacking this information were submitted. In order to educate providers and to prevent specimen rejection, NBS laboratory personnel began an intensive telephone campaign to all

submitters who had failed to include this newly required data. After three weeks, telephone calls for all specimens had to be discontinued due to the impact on laboratory staff workload. As a result, effort was then redirected to focus on submitters who have three or more unsatisfactory specimens rejected in a week due to no date of collection data and who have not recently been contacted. Additional methods currently and historically utilized to educate providers include newsletters, global comments listed on the bottom of all patient reports, regional site training, posters and videotapes.

Provider survey respondents indicated that their preferred training formats were newsletters (66%), brochures (57%), and toolkits (36%). The majority (63%) of respondents indicated the current newsletter and educational materials helpful or very helpful. The department will investigate additional methods for future training on laboratory issues. One of the new positions in case management will be an educator enhancing the case management organizational structure to create a team of three positions focused on provider and client education.

The NNSGRC report identified options for linking first and second screens, as well as linking screens with birth certificates. The current NBS Laboratory Information Management System (LIMS) performs only limited matching functions in order to identify second screens of previously abnormal results. For specimens to be linked in the system, it is essential to have accurate and complete patient demographic data submitted by the provider and accurately entered into the LIMS. One option for linking first and second screens is to use a two-part specimen collection form. The form would be separated at the hospital/birthing center when the first specimen is collected and sent to the laboratory for testing. The other half of the form could be given to the parent who would then take it to the baby's first doctor visit for second specimen collection. A similar procedure was pilot tested several years ago, and several problems were identified such as the second part being lost, contaminated, or forgotten, thereby not being available at the well-child visit.

Other options listed in the report for linking specimens include the following:

- Using specimen serial number, birth certificate number, or other unique identifier on the second and any subsequent screens to easily link specimens. This matching solution is used in some NBS programs.
- Linking the NBS forms with adjacent or the same serial numbers. Some NBS programs have noted problems with lost or damaged cards.
- Using a 'birth passport' where the parents are given a unique identifier (such as the first specimen card serial number), and this identifier is linked to other medical records.

Survey results indicated the majority of respondent's felt it would be helpful (36%) or very helpful (39%) if all information pertaining to the first screen and second screen, birth record, and follow-up records were linked. Use of a two-part form to link the first and second screens was a controversial idea, with only 10% stating that they liked the idea and did not foresee any problems. Therefore, linking the information through data management appears to be more feasible logistically. However, less than a fifth of the respondents stated the cost of data linking (19%), collecting, and maintaining a registry for long-term follow-up (19%), should be covered by fees.

Until the current LIMS was implemented in July 2004, providers were able to access patient results 24 hours, 7 days a week through a Voice Response System (VRS). A new interface between the LIMS and the VRS would allow this system to be reactivated. Laboratory personnel are currently working with a vendor to re-establish this system.

As early treatment of specific newborn screening disorders is critical to the patient's health outcome, and because analytes may begin to degrade as a specimen ages, it is essential that specimens are received at the laboratory and tested as quickly as possible after collection. Newborn screening specimens are currently shipped via the US Postal Service and rejected for testing ('specimen too old upon receipt') when received more than 13 days after specimen collection. The Laboratory Optimization Project, which seeks to improve and economize laboratory services for all DSHS laboratories, is considering implementing a statewide courier system to improve and expedite specimen-handling procedures. A Request for Proposal (RFP) for a statewide courier system is currently being drafted. Newborn screening specimens would be part of this system if a bid were awarded.

A total of 38% of the provider survey respondents indicated that the laboratory fee should cover the transport of specimens to DSHS. However, only 18% of the respondents indicated that the extra cost of courier service, in order to ensure timely transport of newborn screening specimens, was worth the benefit of the service.

In addition to expediting delivery of specimens to DSHS, NNSGRC recommended that the DSHS NBS laboratory seek ideas for decreasing turnaround time (TAT) for result reporting. One change to decrease TAT was implemented within days of the NNSGRC Review. The galactose1-phosphate uridyl transferase (GALT) assay results were previously released the morning after the retest assay was performed. These results are now released by 2:30 pm on the same day that the retest assay is performed. One suggestion for decreasing TAT in the NNSGRC review was to report preliminary results before a weekend or holiday versus delaying these results. For those assays that are considered time-sensitive due to the potential life-threatening nature of the disorders (GALT Deficiency and CAH), extra steps are taken during extended holiday periods and long weekends. However, at this time, the laboratory staff is hesitant to report preliminary results for all assays. Before weekends and holidays, the panic level used to report preliminary CAH results to case management is lowered from >185ng/mL to >120ng/mL. In addition, during extended holiday periods, personnel come in the morning after the overnight run to evaluate CAH screen assays, for potential panic values and notify case management if any are identified. Also for extended holiday periods, GALT retest assays are performed the same day as the initial screen, and any abnormal levels (panic value) are manually reported to case management. The NNSGRC Review Team has also suggested that the NBS laboratory consider a routine six or seven-day workweek, as well as, shift work.

Currently, every baby born in Texas is required by rule (Texas Administrative Code 25; Chapter 37.56) to have two newborn screens unless a parent objects on religious principles. The second screen benefits those babies whose first specimen was drawn 'too early' (typically prior to 24 hours after birth). The majority of the first newborn screen specimens in Texas are collected between 24 and 72 hours of birth (Gonzalez (1995) Conference Proceedings, Early Hospital Discharge: Impact on Newborn Screening, Washington, DC p155-166).

Scientific evidence of the utility of a second screen in the diagnosis of NBS disorders follows:

- Routine re-screening is recommended when the initial screen is taken before the baby is 24 hours of age. The chance of a false-negative result for PKU is greater when the sample is taken before the baby is 72 hours old. (Scriver *et al.* (1982) **Pediatrics**, 69: 104-106).
- In the Northwest Regional Screening Program, 19 of 182 infants with primary hypothyroidism were detected on the second newborn screen (10.4%). Note that in 1977, the Federal General Accounting Office estimated that the average lifetime cost of treating an

undiagnosed hypothyroid patient would be $330,000 (LaFranchi *et al.* (1985) **Pediatrics**, 76: 734-740).

- In the Texas NBS Program, approximately 75% of the non-classic CAH cases were identified by the second newborn screen, and as many as 4% of the salt-wasting cases and 52% of the simple-virilizing CAH cases were identified only by the second newborn screen (Gonzalez (1995). **Early Hospital Discharge: Impact on Newborn Screening**, Proceedings of a conference held in Washington, DC, p155-166).
- Between February 1, 1980 and January 1, 1985 in the Texas NBS Program, 21 of 414 (5%) of the hypothyroidism cases were detected from the second newborn screen (Levine and Therrell (1986) **Pediatrics**, 78: 375-376).
- In Texas in 1994, two of 15 (13.3%) cases of CAH were detected on the second newborn screen. The second newborn screen identified five additional cases of non-classic CAH (Brosnan *et al.* (1998) **Public Health Reports**, 113: 170-178).
- The Northwest Regional Screening Program has identified 21/133 MS/MS cases (18%) on the second screen since implementation of their expanded newborn screening program (Judi Tuerck, RN, MS, Assistant Professor, Oregon Health & Science University, Child Development & Rehabilitation Center).

NNSGRC Reported Follow-Up Issues

- Evaluate staffing patterns for necessary professional level and retention
- Evaluate mechanisms of communicating results to providers: telephone calls versus fax
- Evaluate effectiveness of letters to physicians and families

Follow Up- Additional Findings and Actions to Date

Case management follow-up staff should be appropriately trained – nurses, genetic counselors, social workers; others can be trained as recommended by the Council of Regional Networks for Genetic Services (CORN). The department has improved the organizational structure with the approval of additional staff to track abnormal result cases by disorder types, for example, metabolic, endocrinology, and hematology. The addition of nursing and public health staff will enhance the NBS case management program's follow-up activities.

Case management follow-up activities should include short and long-term components. It is critical to follow a positive test result to diagnosis and beyond to ensure needed services are received, as endorsed by the Association of State and Territorial Health Officials (ASTHO), the CORN, and the American Academy of Pediatrics (AAP). Additional recommendations of ASTHO, CORN and AAP include:

- Follow-up should include advice on appropriate interim measures in advance of confirmatory tests to avert a potentially lethal crisis.
- The NBS case management system must ensure follow-up of any positive, or potentially positive, result to the point of resolution.
- Specifically trained follow-up coordinators such as nurses, genetic counselors and social workers best accomplish follow-up activities.
- Data transfer should be timely, and protocols need to clearly define the processes to reach a conclusion of diagnosed, cleared or lost to follow-up.
- Documentation of contacts with physician and family is essential.
- Communication should be rapid in accordance with the CORN guidelines, and follow-up documentation should be regularly reviewed.

- CORN guidelines indicate the need for protocols to address problems such as: no Primary Care Physician (PCP); language difficulties; adopted babies; unable to locate family; refusal to take action by PCP or family; no insurance; or no funding.
- Ideally, long-term follow-up should include registries to which updated information, treatment compliance and outcomes could be added.

The current NBS program incorporates all the above-mentioned recommendations of ASTHO, CORN and AAP. To ensure that the processes are the most effective for Texas, the case management follow-up program has initiated a project to review letter and fax content to ensure consistency and understanding. The NBS Project Charter Team recommends re-evaluating the protocols that guide contact with providers regarding abnormal results, i.e., telephone call versus fax.

Provider survey results indicated that NBS should contact the physician directly for moderately abnormal results (68%) and borderline results (36%), in addition to the current policy of contacting the physician directly for very abnormal results. Additionally, respondents indicated NBS case management should contact physicians' staff for borderline results (53%). The majority of survey respondents indicated that the primary care physician or pediatrician should be responsible for follow-up (74%).

NNSGRC Reported Diagnosis/ Treatment and Management Issues

Sufficient capacity to diagnose and treat exists in Texas

- Possible exceptions include West Texas and Rio Grande Valley

Diagnosis/ Treatment and Management- Additional Findings and Actions to Date

CORN recommendations for diagnosis, treatment and management include the following:

- Time from birth to diagnosis should be as short as possible.
- Confirmatory testing should be performed before treatment.
- Treatment and management should be a partnership between primary physician/treatment center/ and the NBS Program.
- Data regarding treatment and management should be collected and reviewed to determine program effectiveness.
- Long-term tracking and outcome evaluation should be maintained to ensure health care is obtained.
- Diagnostic tests should be reported as a part of follow-up activities.

Although the NNSGRC review indicated that there appears to be sufficient capacity to diagnose and treat in Texas, with the possible exceptions of West Texas and the Rio Grande Valley, statewide survey results indicate room for improvement. Provider survey results indicated that the majority of respondents are capable or very capable (60%) of diagnosing patients with abnormal screens while slightly less than 50% (46%) indicted the capacity to treat. The majority of respondents indicated the capacity to coordinate care (67%) but less than 50% (41%) indicated ease in referring to subspecialists. Reasons for difficulty in referring ranged from transportation and scheduling issues to subspecialists being too busy to see patients. Further analyses indicated that neither public health region (PHR) nor rurality (i.e., rural, micropolitan, or metropolitan county status) significantly related to a provider's capability to diagnose, treat, coordinate care, or refer to subspecialists. Provider survey results indicated that the majority of respondents would like regional centers for referral (56%).

NNSGRC Reported Evaluation and Education Issues

- Consider providing feed back on quality of screens submitted to the submitter/provider
- Investigate current software capacity for providing necessary data for program evaluation
- Provide timely annual program data
- Consider targeted education and public relations efforts where indicated
- Evaluate protocols for 'Lost to Follow-up' scenarios and review mechanism to capture data
- Conduct a staffing analysis to include community level follow-up, as well as internal follow-up activities
- Encourage hospitals to adopt NBS as a quality assurance performance measure
- Ensure education of providers/ public regarding the importance of newborn screening and available options
- Ensure physicians understand their requirement to report confirmatory diagnoses to DSHS
- Maximize use of existing communication mechanisms: web, newsletters etc.
- Reevaluate mechanisms for distributing materials: active versus passive
- Re-evaluate communication tools for education and eliminate barriers, such as language, reading level, etc.
- Improve practitioner manual

Evaluation and Education- Additional Findings and Actions to Date

As mentioned, additional staff has been identified for NBS case management. Two of the new positions will have a role in quality assurance and the ability to target technical assistance and training to providers who have a high level of unsatisfactory specimens. An additional position will focus on overall education and training. The NBS Project Charter Team recommends providing submitters of newborn screens with 'report cards' to better understand their performance in relation to other submitters of screens.

Provider survey results indicated providers wanted several types of statistical feedback from the NBS program including general data, state-wide and regional comparisons to other states, as well as specimen unsatisfactory statistics (42%), specimen presumptive positive statistics (30%), number of screens submitted (34%), number of patients lost to follow up (38%), and prevalence of conditions (37%).

Public awareness coupled with professional and patient education is a significant NBS Program responsibility. Parent information should be at an elementary school level and ethnically and culturally sensitive. Professional literature may include protocols, manuals, videos, and slideshows, but personal contact and demonstrations are more effective per the CORN guidelines.

Provider survey respondents indicated that follow-up by case management staff was helpful; with only 1% of respondents saying that case management program staff were unhelpful or very unhelpful. Approximately half of the survey respondents indicated a need for additional follow-up training.

Currently, NBS program staff provide feedback and educational materials to submitters that have a significant percentage of unsatisfactory screens. The program sends a multitude of educational materials to providers annually including: collection guides and posters, compact disc on collection techniques, newsletters, practitioner guides and weight charts. As staff identify a need to communicate a high priority message to submitters, the newsletter, as well as urgent messages printed on brightly colored paper are mailed to submitters. All new submitters of NBS are provided with a comprehensive packet of information on all NBS literature available. The NBS website

receives approximately 300,000- 400,000 hits annually. Mail outs regarding PKU and pregnancy are sent annually to all girls identified with PKU who are turning 13 and of potential reproductive age. The NBS program has regular contact with identified medical residency programs and provides NBS materials for residency training. Regular mail outs offering NBS materials regarding collection techniques and disorders are sent to nursing and phlebotomy schools and programs.

NNSGRC Reported Program Management Issues

- Improve data and maintain registry for collection of long-term follow- up data and comparative data
- Consider regional designations for follow up and 'catchment areas' for responsibility to ensure confirmatory diagnosis and treatment
- Consider California model in reorganization of follow-up and case management
- Consider three options for implementation of expansion
    - , MS/MS testing as an option and contract out
    - , Mandate MS/MS testing and contract
    - , Delay MS/MS until state laboratory is ready
- Perform a thorough cost analysis of the system
- Clarify statutory or rule change to allow program elements other than laboratory testing to be included in the fees charged
- Evaluate current Texas use, retention and storage protocols and other written protocols including screens lost to follow-up

Program Management- Additional Findings and Actions to Date

Under Chapter 33, Health and Safety Code, DSHS is mandated to maintain a roster of children born in Texas who have been diagnosed as having one of the disorders for which the screening tests are required. NNSGRC recommended that DSHS combine the scientific and programmatic interests associated with learning more about rare conditions based on large population studies. NNSGRC recognized that a staffing requirement evaluation might be in order to maintain the roster at the level needed to provide adequate data for research efforts. Staffing for case management has been evaluated and will increase by five in FY 2006.

NNSGRC recommended that the department consider regional designations for follow-up and catchment areas for responsibility to ensure confirmatory diagnosis and treatment, potentially using the California model in reorganization of follow-up and case management. The California model provides for designations of distinct geographic regions in the state. Although the NBS Program is administered centrally, the case management staff are spread over the 7 regional NBS Area Service Centers (ASC). Each regional area has a Special Care Center (SCC), approved by California's Children's Services, where newborns identified with a disorder through the NBS Program have access to a diagnostic evaluation, and ongoing specialty care from a multi-disciplinary team. In this model, all newborns referred to the SCC by the program are eligible for a diagnostic evaluation regardless of income. The team recommends any consideration of regional approach be delayed until after the cost-effectiveness study is complete and a determination regarding outsourcing is made.

According to a report by the Association of State and Territorial Health Officials, Financing State Newborn Screening Systems in an Era of Change (2005), "45 states collect newborn screening fees as the primary source of program funding." This article states that the majority of funding goes to support the laboratory testing and on average, only about one-third of funds are allocated to follow-up in traditional NBS programs. DSHS legal has done a review of existing statutory language and

determined that increasing fees to cover services in addition to laboratory services is allowable (Appendix 5).

Laboratory and case management staff are currently in the process of reviewing protocols for children "lost to follow-up" and recommending necessary changes. In response to HB 790, the department will issue a Request for Letter of Interest (RLI) and a subsequent RFP to determine the most cost effective approach of providing NBS.

Summary of NBS Project Charter Team Recommendations

During the finalization of this report, development of an RFP to determine cost effectiveness of outsourcing or maintaining NBS in-house began. In light of this development, it is the Project Charter Team's understanding that many of these recommendations will be considered for inclusion in the RFP if they are consistent with stakeholder input and ultimate agency decisions.

The following actions are recommended for consideration in the design and ultimate implementation of an expanded NBS system:

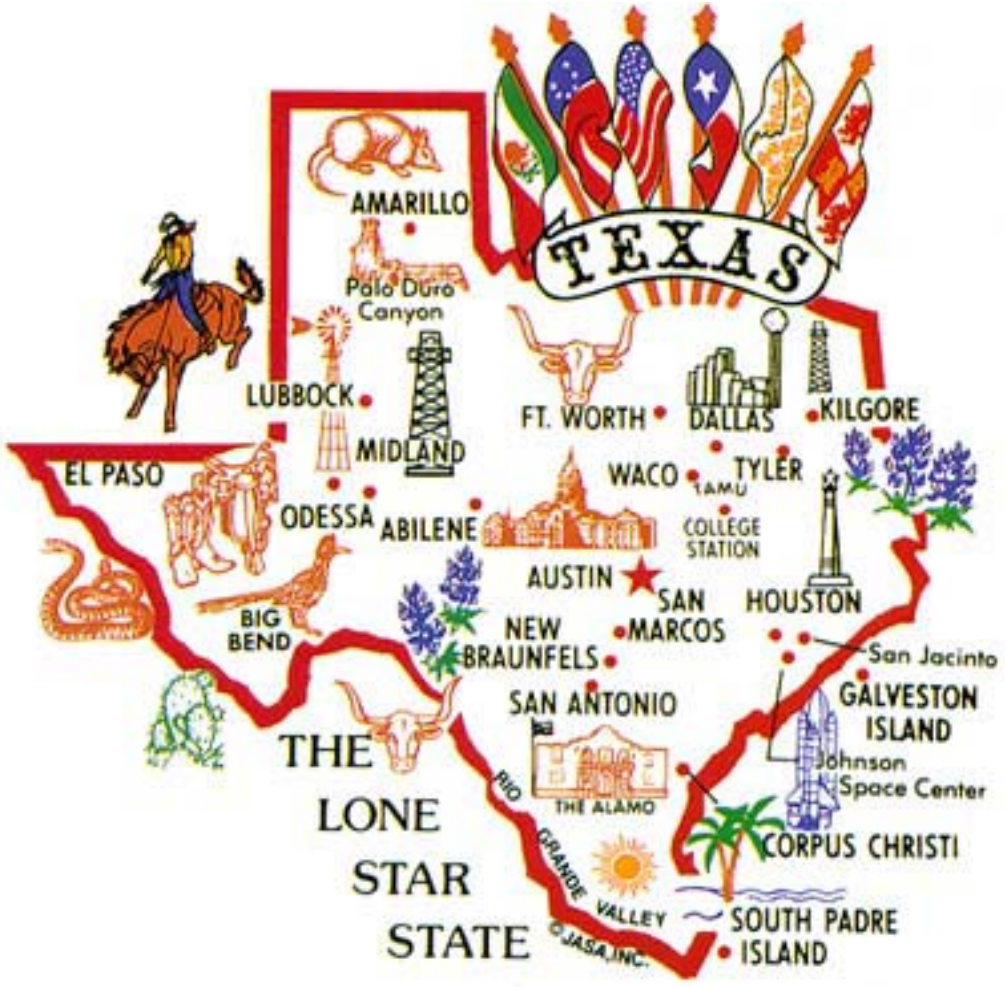
- Develop the budget necessary to finance laboratory and case management follow-up activities to achieve the enhancements and recommendations of the Project Charter Team.
- Increase distribution of general information on NBS and ensure diagnosis-specific information is available for parents of infants with positive screens.
- Develop educational materials in preferred languages identified by stakeholders.
- Identify mechanisms to provide education to expectant parents prior to delivery.
- Continue utilizing the three *ad hoc* consultant groups:
    - Identify avenues to enhance their role in reviewing protocols that guide the clinical aspects of follow-up
    - Identify avenues to enhance their role in the development of parent and provider educational materials; and,
    - Identify mechanism to increase participation in the adhoc meetings.
- Identify mechanisms and funding to link first, second and subsequent screen information
- Explore various avenues to ensure timely delivery of specimens to laboratory from submitter
- Provide submitters of newborn screens with 'report cards' to better understand their performance in relation to other submitters of screens.
- Enhance training to submitters, physicians and nurses.
- Evaluate systems of communication with submitters regarding abnormal NBS results
- Study regional approach to follow-up and treatment 'catchment areas' and submit recommendation to DSHS leadership

DRAFT
CONSULTATION REPORT

TEXAS

NEWBORN SCREENING PROGRAM

FEBRUARY 28 - MARCH 2, 2005



National Newborn Screening and Genetics Resource Center
Austin, Texas USA
NNSGRC

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CONSULTATION REPORT
TEXAS NEWBORN SCREENING PROGRAM

1.0.0 Introduction

1.1.0 Logistics Summary

On February 28 - March 2, 2005, a select Newborn Screening Technical Assistance Review Team (Review Team brief resumes in Appendix 1) reviewed the newborn screening program of the Texas Department of State Health Services (DSHS). The National Newborn Screening and Genetics Resource Center (NNSGRC) sponsored this review through a cooperative agreement with the Genetic Services Branch of the Maternal and Child Health Bureau (MCHB), Health Resources and Services Administration (HRSA). The review was at the request of Dr. Sanchez, Commissioner of the Department of State Health Services (see Appendix 2). Due to the physical size of the State of Texas and the short amount of time available to the team in which to gather sufficient information to adequately address the issues presented to the team, an additional four experts augmented the usual team of five so that the team could be divided into two groups. One group traveled to the Dallas area for meetings with local stakeholders there and the other traveled to Houston returning through San Antonio with meetings of local stakeholders in both locations. In this way, the two teams were able to meet with more hospital staff, physicians, nurses, parents and other interested parties.

Introductory sessions at the Austin headquarters of DSHS provided the Review Team with the opportunity to learn about the administrative details of the newborn screening program and to physically visit in the laboratory and the follow-up areas. Conference telephone lines were provided by DSHS at two different times on the first day so that interested parties who could not attend working sessions with the teams could interact by telephone. All 9 Review Team members visited the maternity section of Brackenridge Hospital in Austin, a public hospital serving the Travis County area. In Dallas, Team 1 met with staff and other parties at Zale Lipshy Hospital and at the Institute for Metabolic Disease at the Baylor University Medical Center. In Houston, Team 2 met with staff and interested parties at the Baylor College of Medicine and with staff and interested parties at the Santa Rosa Hospital in San Antonio. At the request of Commissioner Sanchez, a special 30-minute session was held with representatives of Pediatrix Screening, Inc., an affiliate of Pediatrix Medical Group, Inc., a for-profit healthcare services company in Pennsylvania, prior to the exit interview, which was held with DSHS administrative staff on the morning of March 2.

1.2.0 Logistics Details

The introductory meeting on February 28 included: from DSHS Administration Evelyn Delgado, Alexis Hathaway, MD, Jann Melton-Kissel and Susan Neill, Ph.D.; from Follow-up – Margaret Bruch, Margaret Drummond-Borg, M.D., Michele Goddard, Paula Geurin, Theresa Berru, and Daisy Johnson; from DSHS Laboratory – Eldridge

Hutcheson, Ph.D., Susan Tanksley, Ph.D., Lynette Borgfeld, Carl Danford, Particia Ramos, and Bill Blackburn; DSHS related programs - Joy O'Neal (Newborn Hearing Screening), Sam Cooper (CSHCN), Fouad Berrahou (Title V), Alison Smith (Legislative Liaison), and Mark Canfield, Ph.D. (Birth Defects); from Texas Medical Association (TMA) - Barbara James, R.N., and Leo Cigarroa, M.D.; and from Perkin Elmer Corporation - Brad Shields. Following introductions, Ms. Delgado reviewed the administration of the Texas Newborn Screening Program (TNSP) followed by Ms. Borgfeld providing a detailed discussion of the laboratory operations and Dr. Drummond-Borg discussing follow-up operations. Questions and answers were addressed throughout the session. Afterwards, the Review Team was given a tour of the screening laboratory and the follow-up area.

During a working lunch, DSHS made available telephone lines for previously identified interested persons to call in and speak with the Review Team. Two persons, Dr. Mueller and her colleague, Ms. Jacobs, from the Houston Sickle Cell Center, participated in the session. An informative exchange provided information to the Review Team about some of the issues identified by subspecialists who provide follow-up diagnostic, counseling and other linkage services. Following this session, multiple conference lines were made available at the request of TMA for a second session, but no participants responded. The reason for the low participation in these two conferences was not readily evident.

In the afternoon, the Review Team and Dr. Drummond-Borg visited at Brackenridge Hospital in Austin where local stakeholders were present. Attending this discussion session were: Teri McClain, RN, Brackenridge Hospital (perinatal case manager); Terry Cloud, MT, Brackenridge Hospital (laboratory - sending specimens to DSHS); Barbara James, RN, (TMA); Debbie Freedenberg, Austin (clinical geneticist); Lakshmy Vaidyanathan, MD, Seton Medical Center (hospitalist); Susan Crane, RNC, Seton Medical Center (director of neonatal services); Gwenn Gallagher, Children's Hospital of Austin (NICU clinical manager); Glenda Amato, Brackenridge Hospital (clinical manager); and Becky Roberson, RN, Seton Medical Center (case manager).

Team 1 (composed of Dr. Mann, Dr. Bartoshesky, Ms. Hermerath, and Ms. King - see listing in front of report and curriculum vitae in Appendix 1), accompanied by Dr. Drummond-Borg and Ms. Bruch from DSHS, journeyed to Dallas for meetings there. On March 1, this team met with local stakeholders for discussions at the Zale Lipshy Hospital, a part of the University of Texas Southwestern Medical Center. Attending the morning session were: Anna Barden, Parkland Hospital; Dave Talley, Parkland; Debbie Rifkin, parent of a child with tyrosinemia; Diane Milam, Baylor Hospital; Greg Beckham, Baylor Hospital; Zora Rogers, MD, University of Texas Southwestern Medical Center (Pediatric Hematologist); Michelle Bowen, Parkland (Phlebotomy Supervisor); Kevin Stuteville, Parkland; Angelique Ramirez, Parkland Hospital (COPC); Mary Bergman, Parkland Hospital (COPC); Dawn Raimer-Hall, Children's Medical Center; Sharon Turnley, City of Dallas; Pauletta Jones, City of Dallas; Linda Siddens, Parkland Hospital (Laboratory Manager); Greg Jackson, University of Texas Southwestern Medical Center; Perrin White, MD, University of Texas Southwestern Medical Center (Pediatric Endocrinologist), Vikki Yealls, Garland Health Department. In the afternoon,

Team 1 toured the Institute of Metabolic Disease at the Baylor Hospital campus and met with Dr. Charles Roe and other staff members in order to assess the capabilities of the Institute relative to supplemental screening with tandem mass spectrometry (MS/MS).

Team 2 (composed of Dr. Therrell, Dr. Hannon, Dr. Desposito, Ms. Neier and Mr. Hoffman - see listing in front of report and curriculum vitae in Appendix 1), accompanied by Dr. Hutcheson and Ms. Melton-Kissel from DSHS, traveled to Houston. On March 1, this team met with local stakeholders for discussions at the Baylor College of Medicine. Attending the morning meeting were: Roslyn Thomas, Ben Taub Hospital (Newborn Screening Clerk); Carolyn Fairchild, Ben Taub Hospital (Data Information Coordinator); and from Baylor College of Medicine - Lori Sielski (Pediatric Faculty); Suzanne Lennon, Texas Children's Hospital (Metabolic Dietician); Kerri Lamance, RN, (Genetics); William Craigen, MD, (Genetics); Reid Sutton, MD (Biochemical Genetics Laboratory); Brett Graham, MD (Genetics); David Horst, MD (Neonatalogy - representing TMA Maternal/Perinatal Committee); and from the March of Dimes, Amye Webster. In the afternoon, Team 2 continued its discussions with local stakeholders in San Antonio at the Santa Rosa Hospital. Attending the afternoon meeting were: Howard Britton, MD, UTHSCSA (Pediatric Hematologist); Alice Gong, MD, UTHSCSA (Neonatologist); Michele Maxwell, San Antonio Metropolitan Health District; Fernando Gurerra, MD, San Antonio Metropolitan Health District (Director); Donna Willey-Courand, MD, UTHSCSA (Cystic Fibrosis Center Director), and Raymond Lewandowski, MD, Corpus Christi (Clinical Geneticist - representing TMA).

Prior to the Exit Review on March 2 at DSHS headquarters in Austin, the combined Review Team met with representatives of Pediatrix Screening. Present in the meeting from Pediatrix were: Philip Vaughn, MD, Bill Slimak, Stan Grossman, and Jaime Capello. Team members were told by Pediatrix representatives that this meeting was necessary because they had been informed of the review and the review schedule too late to schedule attendance or participation of their representative in any of the scheduled discussion periods or telephone conferences. Immediately following the meeting with Pediatrix, the Review Team met with DSHS representatives to report tentative findings and responses to issues and questions presented to the Team in the initial invitation (see Appendix 2). This oral review was intended to discuss some of the answers to questions and issues to be addressed in the written report and to exchange clarifying information where needed. Present at this exit review, in addition to the Team, were: Evelyn Delgado, Jann Melton-Kissel, Margaret Bruch, Susan Neill, Eldridge Hutcheson, Lynette Borgfeld, Margaret Drummond-Borg, and Mark Canfield.

The Review Team was impressed with the cooperation of all personnel with whom it interacted, both at the DSHS and at the other facilities. The program staff appears dedicated and interested in maintaining a successful, effective newborn screening program as evidenced by their involvement in this review. Despite recent reorganizations within the Department, the program continues to perform its mission carefully and methodically. There are many questions to be answered concerning future directions, but staff dedication will contribute positively to any program changes.

1.3.0 Program Overview

Texas is a unique state and continues to be one of the fastest growing states in the nation. Nationally it ranks:

- Second in population.
- Second in number of births.
- Third in percent of residents under 18 years of age.
- Third in number of persons below the poverty level.
- First in number of persons without health insurance.
- Forty-fifth in government expenditures per capita.

In terms of healthcare related state demographics, Texas statistics include:

- Forty-seven percent of state residents are Hispanic, Black or other minority group.
- Eighty percent of the population lives inside metropolitan areas.
- Thirty-one percent of the state's population is between the ages of 25 and 44.
- Women of childbearing age make up about 20% of the population.
- More than 15% of state residents live below the poverty level.

In an average week in Texas, approximately:

- 6,600 babies are born.
- 1,000 babies are born to mothers who receive inadequate prenatal care.
- 500 low birthweight babies are born.
- 50 deaths occur to babies younger than 1 year of age.

Newborn screening began in Texas late in 1963, when a pilot program was implemented to screen for phenylketonuria (PKU). Following a successful pilot study, PKU screening was required by statute in 1965 and Texas joined some 30 other states in initiating population-wide newborn screening for PKU. In 1977, the screening statute was expanded to include testing for congenital hypothyroidism (CH), galactosemia (GAL), and homocystinuria (HCY), but funding availability prevented testing implementation. In 1978, with the introduction of automated punching equipment, inexpensive bacterial testing for GAL and HCY were added to the program, and on January 1, 1980 (with new appropriations), CH was also added. In 1983, after screening almost a million newborns without detecting a case of HCY, screening for HCY was discontinued in favor of screening for sickle cell diseases (SCD). On June 1, 1989, screening was expanded to include congenital adrenal hyperplasia (CAH). Screening for GAL (which included transferase, kinase, and epimerase deficient forms) was modified in 2004 to include screening for only galactose transferase deficiency (GALT). Currently the program requires screening for five conditions: PKU, CH, GALT, CAH, and SCD. Recent recommendations by the American College of Medical Genetics suggest a method of counting the conditions screened such that TNSP would now be counted as testing for eight conditions, including hearing screening, and screening for SC-disease, and S-beta thalassemia (both of which are identified in the screening process for SCD).

The current Texas Program Rules require that all babies born in the State receive two screens. This requirement is the result of significant numbers of cases of both CH and CAH having been identified through second screens following a normal initial screening test result. In 2004, the live births in Texas totaled approximately 375,000 and the TNSP reported 756,130 specimens received for testing (approximately 3,000/day). The number of specimens received includes initial screens, required second screens, and repeat screens requested because of out of range previous results. It is assumed that over 98% of newborns receive the first screen, with over 90% receiving both screens. Specimen data are not linked to birth records, nor to each other, so that an accurate accounting of compliance in meeting the testing requirements is not possible - whether it is compliance with the initial test or compliance in completing the two required tests. Similarly, it is not possible to accurately account for multiple specimens received on a particular infant.

The administrative/follow-up/educational arm of the TNSP resides in the Division for Family and Community Health Services and the screening laboratory resides in the Division for Prevention and Preparedness Services. This means that administratively the TNSP is split between the Specialized Health Services Section and the Laboratory Services Section. Each section is in a different organizational division of DSHS with separate division directors, each reporting to the Commissioner. Funding for the two program components also arises from different sources.

The funding for administrative/follow-up/educational activities (reported to be approximately $700,000 annually) comes from Medicaid and Title V Maternal and Child Health Block Grant funds, and laboratory funding (reported to be approximately $14,000,000 annually) with the exception of a small percentage of funds for uninsured patients comes from fee revenue (including private insurers and Medicaid). The amount of the fee is set by the Laboratory Services Section, in compliance with Program Rules, and includes a separate fee schedule for newborns that qualify for Medicaid. Currently the fees are $19.50/specimen for non-Medicaid and $16.20 for Medicaid patients (Medicaid fees are accounted for separately within DSHS, since it is also the Medicaid Agency within Texas). Fees are collected through the purchase of newborn screening collection kits from DSHS with payment due 120 days after the order is filled. Hospitals and physicians ordering collection kits may request kits at no charge for use with Medicaid patients and patients without insurance. There are no specific arrangements or requirements that insurers pay for the screening services (including test kits), although they are encouraged to do so. Data regarding insurance billing and payment practices are not available, so it is not possible to assess whether charges and payments resulting from insurance claims are uniform, but some anecdotal evidence points to non-uniformity and, perhaps non-coverage of the procedure. A national billing code does not currently exist; however, some insurers within the State appear to have created a local code for their use within Texas.

Currently all newborn screening laboratory testing is performed in the DSHS laboratory; however, the screening statute allows the DSHS to develop a program for other laboratories to perform the testing. The statute requires the DSHS to provide

laboratory testing and to use this laboratory for developing tests for detecting the conditions included in the screening panel, for developing methods for prevention or treatment of the conditions, and for other purposes defined by the DSHS. Likewise, the statute allows the program to provide certain services, within available funds, either directly or through approved providers, for patients of any age meeting eligibility criteria established by Program Rules for any of the conditions included in the screening panel. It is unclear whether funds for program follow-up activities can be included in the fee charged for the screening kits. It is also unclear whether funds for the other laboratory activities beyond routine testing [e.g.. test development (which might include equipment rental and reagent purchases for possible expansion)] can be included in the fee. Currently, neither are included.

The table below was created from information reported to the National Newborn Screening and Genetics Resource Center (NNSGRC) over the years and was validated by Dr. Drummond-Borg prior to the Review Team visit. These data give an indication of the success of the screening program in terms of its ability to detect cases. Legislative consideration is currently being given to requiring newborn screening for multiple other conditions including cystic fibrosis, biotinidase deficiency and many other metabolic conditions detectable with tandem mass spectrometry (MS/MS). An increase of 50-75 cases annually is anticipated from the an expanded MS/MS testing program, with an additional 75-100 cases/year of cystic fibrosis (CF) possible if testing for CF is added. In both cases, the anticipated increase in patients to be recalled for follow-up is expected to be approximately 2,000/year

Table 1. Texas Newborn Screening Summation of Diagnosed Cases: 1991-2000

Year	Births[1]	PKU	CH	GAL	CAH	FS	FSC	FSA	FAS
1991	322,065	9	113	1	12 (SW), 7 (SV)	78	34	2	4,505
1992	325,104	15	115	1	15 (SW), 6 (SV)	95	45	11	5,128
1993	326,267	10	136	2	17(SW), 6 (SV)	81	28	8	4,804
1994	325,521	10	124	3	11 (SW), 5 (SV)	92	27	5	4,079
1995	326,587	9	155	6	17 (SW), 6 (SV)	88	47	4	4,816
1996	334,197	7	145	2	12 (SW), 6 (SV)	94	40	3	4,817
1997	335,731	5	140	8	15 (SW), 4 (SV)	75	28	4	4,297
1998	346,101	11	159	5	16 (SW), 5 (SV)	92	42	15	4,544
1999	352,970	11	156	13	14 (SW), 2 (SV)	105	47	15	4,732
2000	371,676	11	157	3	17 (SW), 9 (SV)	82	37	15	4,868
Totals	3,366,219	98	1,400	44	202	882	375	82	46,590
Incidence	--	1:34,349	1:2,404	1:76,505	1:16,664	1:3,817	1:8,977	1:41,051	1:72

[1] From National Center from Health Statistics

1.4.0 Organization of Consultation Report

This Consultation Report is organized to first address the specific areas of concern raised in the January letter of invitation from Commissioner Sanchez. Initial responses concern the issues raised directly by DSHS and are followed by comments and answers to other questions raised by the Texas Medical Association (TMA) (see letter with accompanying questions in Appendix 2). Comments on these issues are followed by a discussion of other points considered important by the review team. Finally, an overview summation is given using a template in which strengths, weaknesses and possible future actions are enumerated. It is suggested that the possible actions be reviewed and developed into an action plan for strengthening the program. Persons reviewing this report are referred to Appendix 3 for published guidelines considered essential to the success of newborn screening systems. These guidelines, entitled *U.S. Newborn Screening System Guidelines: statement of the Council of Regional Networks for Genetic Services (CORN)*, were the result of findings from multiple state consultations similar to the one conducted in Texas. All members of the team are available for further consultation either collectively or independently if needed.

2.0.0 Issues from the Program

The following issues and questions were submitted to the Review Team by the DSHS prior to the Team's visit and formed the basis for the major focus of this review.

2.1.0 Provide comparison information of other state's mechanisms for funding newborn screening and pros and cons for alternate approaches.

Currently in the United States, there are two primary mechanisms for funding newborn screening programs - fees and legislative appropriations. Programs also receive funding either directly (direct funding transfers) or indirectly (service delivery) from other sources such as Title XIX (Medicaid), Title V [Maternal and Child Health Block Grant, including Children with Special Health Care Needs program (CSHCN)], Women Infants and Children's (WIC) program, and various other programs (including state appropriations). Further elaboration on current financing strategies can be found in the 2003 Report of the Government Accounting Office (http://www.gao.gov/new.items/d03449.pdf). Since this question appears to be aimed at primary funding aside from legislative appropriations, the answer will focus on direct funding mechanisms from fee collection.

Historically, newborn screening programs have relied heavily on tax revenues and the public health laboratory for the testing services considered essential to newborn screening. Over time, programs have expanded from a single screening condition, PKU to more conditions. These expansions were motivated in the 1970s and 1980s by the ability to screen for higher prevalence conditions such as CH. With expanded screening came increasing demands for related program services, screening for more conditions, and consequently the need for additional funding. State legislatures gradually shifted from a tax revenue based financial model to a fee model. Table 2 gives a comparative summation of the current financing schemes of the 51 U.S. newborn screening programs (50 states and the District of Columbia). Of the 51 programs, all but 5 currently obtain at least some of their funding through a fee-for-service.

Table 2. Tabulation of State Newborn Screening Program Fees

State	Births (Occurrence) In 2001	Percent Medicaid births (2000[a])	Number of screens required	Number of disorders currently mandated (1/2005)	Current Fee 1/2005	Notes
Alabama	59,766	45.0	1	14	$139.33	Two screens strongly recommended.
Alaska	9,907	52.0	1	>30	$55.00	Fee includes any repeats.
Arizona	85,757	44.0	2	8	$20.00	Separate fee for each mandated specimen.
Arkansas	36,301	43.7	1	4	$14.83	
California	528,539	42.4	1	>30	$78.00	
Colorado	67,100	32[b]	2	7	$53.25	Fee includes 2 mandated specimens (2-part form).
Connecticut	43,179	26.7	1	>30	$28.00	
Delaware	11,360	41.0	2	29	$64.00	Fee includes 2 mandated specimens and any repeats.
District of Columbia	15,037	28[b]	1	7	No Fee	
Florida	205,991	44.0	1	5	$15.00	
Georgia	134,402	44.0	1	10	No Fee	
Hawaii	17,127	25.0	1	>30	$47.00	
Idaho	20,161	34.2	1	>30	$23.00	$46 for double kits if screening occurs prior to 48 hrs.
Illinois	181,086	37.2	1	>30	$47.00	
Indiana	86,710	42.0	1	>30	$62.50	Includes $32.50 laboratory surcharge and all repeats.
Iowa	37,756	23.0	1	>30	$56.00	Fee includes any repeats.
Kansas	39,052	12[b]	1	4	No Fee	
Kentucky	53,227	38.8	1	4	$14.50	
Louisiana	65,620	41.0	1	5	$18.00	Fee expected to increase to $40.00 later in 2005.
Pennsylvania	13,567	20[b]	1	9	$44.00	
Maryland	68,663	29.0	1	>30	$42.50	Fee includes repeats; 2 screens strongly recommended.
Massachusetts	82,237	24.2	1	10	$54.75	
Pennsylvania	132,159	27.7	1	11	$55.72	Fee includes any repeats.
Minnesota	67,428	31.3	1	>30	$61.00	
Mississippi	41,145	53.7	1	40	$70.00	
Missouri	76,690	39.0	1	14	$25.00	
Montana	10,935	40.0	1	4	$39.34	
Nebraska	25,107	28.8	1	6	$30.75	
Nevada	31,007	27.6	2	>30	$60.00	Fee includes 2 mandated specimens (2-part form).
Pennsylvania	14,055	20.8	1	6	$18.00	Fee includes hemoglobinopathies when requested.
New Jersey	112,639	23[b]	1	20	$71.00	
New Mexico	26,808	49.6	2	6	$32.00	Fee includes 2 mandated specimens (2-part form).
New York	255,029	41.1	1	>30	No Fee	
North Carolina	119,132	40.5	1	26	$10.00	
North Dakota	8,839	28.0	1	29	$36.00	
Ohio	152,033	33.1	1	30	$33.75	
Oklahoma	48,895	46.0	1	7	$75.59	Fee includes hearing screening.
Oregon	46,200	32.2	2	26	$54.00	Fee includes 2 mandated specimens (2 -part form). Extra single forms are $27.
Pennsylvania	143,957	25.0	1	6	No Fee	Many hospitals offer extra tests for fee. Fees vary.
Rhode Island	13,319	35.4	1	9	$59.00	
South Carolina	53,255	47.0	1	30	$42.00	
South Dakota	10,784	32.8	1	3	$18.53	Fee does not include hemoglobinopathies if requested.
Tennessee	83,521	37.7	1	>30	$47.50	
Texas	370,482	45.1	2	5	$19.50	Separate fee for each mandated specimen.
Utah	49,041	25.8	2	4	$31.00	Fee includes 2 mandated specimens (2-part form).
Vermont	6,149	23.0	1	21	$33.30	
Virginia	96,535	22.7	1	9	$32.00	
Washington	79,078	42.5	1	9	$60.90	Fee includes repeats; 2 screens strongly recommended.
West Virginia	21,000	55.2	1	4	No Fee	
Wisconsin	68,006	35.5	1	26	$65.50	$30.00 laboratory surcharge included in fee.
Wyoming	5,758	38.0	1	7	$45.00	Fee implemented for first time August 1, 2004.
TOTAL	4,031,531	39[b] (Nationally)				

(a) From Kaiser State Health Facts Online, http://www.statehealthfacts.kff.org.
(b) 2000 Medicaid statistics unavailable so statistics are taken from Kaiser Commission on Medicaid and the Uninsured, 1995.

Texas was one of the later programs to move to a fee revenue program, initiating a fee only recently, in 1998. As initiated, the Texas newborn screening fee was solely to assist in paying for laboratory costs and as such, was attached to the cost of the laboratory specimen collection form. Other program services such as follow-up of abnormal and unsatisfactory testing results (with primary care physicians, subspecialists, parents, and others who might be associated with patient care), education of parents and healthcare professionals, treatment/medical management, and long-term outcome monitoring are supported from other funds (if they are included in the funding stream at all). As noted in the Section 1.3.0, the newborn screening laboratory budget is approximately $14,000,000, while the budget for follow-up/administration is approximately $700,000. The newborn screening follow-up budget provided to the Review Team indicates an additional $1,300,000 for genetic services, but clarification received during the review indicated that these funds were not specifically intended for use for follow-up of newborn screening.

The vast difference in the amount of funding between laboratory services and follow-up/administration appears consistent with complaints from the subspecialty community heard at the various "town hall" discussions in Austin, Dallas, Houston and San Antonio. In other newborn screening programs of which the Review Team has knowledge, the budget for follow-up/administration/education is generally closer to a 1:1 ratio than the 20:1 ratio present in Texas. The significant dissatisfaction expressed in the healthcare and subspecialty community about TNSP follow-up seems to relate directly to a lack of funding for these services. The Review Team encountered consistent complaints from all subspecialty personnel interviewed that no adequate funding or funding mechanism exists within the newborn screening program for the various follow-up and linking services required from healthcare providers and subspecialists. These concerns appear to tie directly to budgetary issues with the TNSP follow-up program and third party reimbursement issues, which do not appear to be adequately addressed within the current TNSP infrastructure. Since third party reimbursement directly impacts TNSP specimen submitters, and ultimately the services provided as part of the overall newborn screening system, it is incumbent on the TNSP administration to take an active interest in resolving reimbursement issues. In at least one other State where similar concerns from specimen submitters went unaddressed, the newborn screening laboratory is now facing the added responsibility of directly billing third party payers.

While all subspecialists interviewed expressed a continuing desire to assist the TNSP in ensuring that all presumptive positive newborn screening results resulted in appropriate follow-up actions, there were significant concerns about the manner in which anticipated program expansions might financially impact the healthcare system, including hospitals, community based clinical practices, subspecialty centers, and county clinics. When questioned about the availability of sufficient subspecialists to handle the anticipated caseload from expanded testing with MS/MS, the subspecialists who responded felt that sufficient capacity existed within the State. There were concerns, however, about the distribution of services, particularly the lack of services in West Texas and in the Rio Grande Valley. The lack of subspecialty services in these areas means that increased costs will be encountered when patients in these areas need follow-up services.

There were also concerns about payment for the linkage services that will be required by expansion of the program. In the past, few financial considerations appear to have been given by the TNSP for the resulting follow-up services that occur within the medical community, particularly in the subspecialty centers (assistance in locating patients, scheduling appointments, providing limited counseling and education, and other medical follow-up issues). The concern is that continued unavailability of financial support for these activities will result in even greater negative financial impact.

Additionally, there were significant concerns expressed about the manner in which the newborn screening fee and the fee collection process were initially implemented. While implemented accordance with appropriate Rules governing laboratory fees, there appears to have been involvement of physicians or other outside stakeholders in the actual deliberations about fees and the fee collection process. At the time of the fee implementation, there were significant issues raised by both the Texas Hospital Association (THA) and TMA. The THA opposed a single up front fee that would cover the entire screening cost for each newborn (i.e. both first and second specimens), since this fee would primarily affect hospital revenue flow. The TMA opposed a split fee, since the second fee would primarily affect the revenue flow at the physician's office. In the end, the split fee process was adopted.

Additional fee increases are expected as the program to improves and expands its services. Representatives of hospitals, county clinics, primary care practices expressed concern about perceived additional financial burdens on an already stretched system. There were reports of third party payers that either did not allow payment for newborn screening services or paid at a lower rate than billed. A lack of consistency in the amount of payments was also indicated. There did not appear to be any involvement of the TNSP or any other organized effort in resolving or mediating insurance issues. There were no data available either from DSHS, TMA, or other sources that would indicate the magnitude of the third party reimbursement issues and, in particular, their possible impact on whether newborns were receiving the required second screens.

Currently, the most popular method for collecting fee revenue across the country is through the sale of newborn screening collection kits, as in Texas. However, other program fee collection models exist that appear to have been less contentious in their implementation including: (1) a one-time charge for all collection cards for a particular newborn, (2) monthly billing for testing services based on records of specimens tested at the screening laboratory, and (3) birthing records submitted from hospitals to the screening program showing numbers of specimens submitted over a specified time interval. In all of these cases, the cost of screening is ultimately a hospital charge and is generally included in the global birthing fee/reimbursement that exists as a maternity expense.

The concept of a second specimen is present to some extent in all programs. Second testing is required on all newborns in 8 programs, including Texas, Arizona, Colorado, Delaware, Nevada, New Mexico, Oregon, and Utah, and strongly recommended to the extent that more than 80% of newborns receive second screening in

at least 3 other programs, Alabama, Maryland, and Washington. All programs require some newborns to undergo additional testing when the initial specimen is collected too early (as defined by the program - usually before 12 or 24 hours of age). Except for Arizona, all other programs that require second testing, as in Texas, obtain payment from the hospital providing the initial screening test and all include a sufficiently high initial testing fee to cover the expenses of any additional testing required. In Arizona, a bill is sent upon completion of screening tests to the submitting entity or the insurance company for the patient, if one exists. This model appears to be even more contentious than the model in place in Texas, and there are ongoing discussions about improving fee collections in Arizona by using a different model.

It is important to have a sound accounting basis on which to calculate the fee and it should cover all program expenses including education, follow-up, linkages to services, counseling, and other activities associated with the program including limited treatment/medical management where possible (see report from the American Academy of Pediatrics Newborn Screening Task Force - *Pediatrics* 2000;106:383-427). In establishing a fee, it is equally as important to have the support of the majority of those who might be affected including the physicians, hospitals, insurers, Medicaid administrators and others. In cases where an active advisory committee is functioning, this committee can serve as the venue for problem solving and advocacy among the stakeholders. Thus, the advisory committee and other stakeholders should be involved in financial and other decision-making processes so that they can feel a sense of ownership of the program and its decisions. It is important for the public and others involved in financing newborn screening to understand that newborn screening is a system and system finances MUST ultimately cover education, tracking, confirmation, medical management, and long-term outcome studies – items sometimes overlooked in an effort to lower costs. Failure to adequately consider overall system finances and services ultimately results in lower quality of the screening program.

In cases where program expansion requires significant increases in operating expenses, such as with expanded MS/MS screening, start-up costs may be significant due to the cost of new equipment, building renovation, laboratory personnel, follow-up personnel and services, and public/professional education. Of the programs represented on this review team (Delaware, New Jersey, Oklahoma, Oregon, Washington, and Wisconsin), all found it necessary to increase their fee-for-service in advance of offering the actual expanded testing program in order to finance some of the expenses associated with start-up. Additionally, California, which screens approximately 570,000 newborns annually (second to Texas, even though it has more births as a result of the two screen requirement in Texas) has recently implemented an increase of $18 so that the current fee is $78, despite an anticipated start-up date approximately 6 months in the future.

When the option of an early (6 months or more prior to program implementation) fee was discussed with TNSP personnel, the information obtained indicated that financing in this way is not currently an option under the Rules allowing the fee. Questions about the inclusion of other expenses in the current laboratory fee, or creating an additional program fee to finance other aspects of the newborn screening system, were met with responses that indicated that a fee structure including other program expenses

might be possible. Other innovative financing strategies for start-up of an expanded program included lease-purchase of testing equipment, reagent-rental plans, and outsourcing of part or the entire program.

Short of obtaining start-up funding from legislative appropriations, which is simplest for the program, the Review Team agrees that an increased fee sufficient to cover all program costs (including those not currently covered with other resources) must be considered. Additionally, implementation of the fee prior to program implementation should be considered. This may require clarifying wording in the statute that allows for a fee. The newborn screening fee should include necessary program cost increases for personnel (laboratory and follow-up), follow-up services, education, and equipment. In view of the apparent extensive dissatisfaction with the current fee collection system from the physician community, reevaluation of the fee collection mechanism is suggested. Because any fee increases or changes in collection mechanisms affect those who provide maternal and infant health care, all groups, including hospital and healthcare practitioners, should be involved in fee discussions. Additionally, it may be prudent to include potential payers including insurers and Medicaid administrators. Because much of the data that could answer questions about compliance (matching with birth records and with subsequent specimens on the same infant) and insurance issues (whether there is reimbursement and amount of reimbursement), are not currently available, any financial considerations should emphasize improvements in the data systems currently in use so that they can accommodate these data needs. Fee discounts may be useful to encourage better compliance with diagnostic and demographic reporting requirements (a question raised elsewhere in the review questions) and offers a potential mechanism for encouraging electronic information sharing for future considerations as computerized downloading of patient records becomes a consideration (such a system was previously in operation at Parkland Hospital in Dallas, but was recently discontinued as a result of computer system changes within the Texas Newborn Screening Program (TNSP).

2.2.0 Evaluate current utilization of advisory/stakeholder groups in Texas and provide recommendations for the most effective use of such expertise.

The TNSP currently has three ad hoc newborn screening advisory committees - metabolic, endocrine, and hematology. Each of these appears to be inclusive of all pediatric subspecialists within the State in that particular subspecialty, and the committee meetings include appropriate clinical staff members such as nutritionists, counselors, and others. There does not, however, appear to be committee membership beyond those directly involved in newborn subspecialty care. In their ad hoc capacity, the newborn screening advisory committees are convened approximately once annually, but members interviewed seemed to be unaware of particular committee missions. In recent years there have not been sufficient funds available to provide for travel reimbursement for committee members and this was noted as another indication of the program's lack of financial responsibility to those consulting with the program, although all who were interviewed noted that they were aware of the need for financial restraint on the part of DSHS programs. It was noted that in years past, reimbursements covered travel expenses for a limited contingency from each subspecialty center.

While the various newborn screening advisory committees appear to have been used for program advice at times, particularly when new conditions were being added to the screening panel, the committees currently appear to be used only as a means of sharing research activities among the subspecialty centers and as a sounding board for annual data reports on newborn screening activities. In discussions with Review Team members, persons serving as members of the newborn screening advisory committees reported that they did not feel that they had input into program decisions, nor did they feel that they were consulted on issues of importance to the medical or subspecialty community, including fee setting, fee collection, computerization, conditions on the newborn screening testing panel, testing methods, and follow-up protocols, among others. Whenever asked, there appeared to be universal agreement that a more formal advisory committee structure and charge were needed.

The Review Team agrees that a formal advisory system is important for all newborn screening programs. This is reinforced in other guidance about newborn screening (*Screening* 1992;1:135-47 and *Pediatrics* 2000;106:383-427). Programs have adopted various models for their advisory systems, most of which center around a single program advisory committee. The most effective committees have multi-disciplinary representation and usually include members both from within and outside of government. An example of government programs that might be represented on the program advisory committee include the Medicaid Program, the Texas Department of Insurance (TDI), the Birth Defects Program, the WIC Program, the CSHCN Program, and the Newborn Hearing Screening Program. Possible non-government members should include broad representation from newborn screening stakeholders including, for example, primary care physicians, obstetricians, the Medical Association, the Pediatric Society, the Hospital Association, nurses, nutritionists, genetic counselors, a representative from the insurance industry, community activists, subspecialty physicians with an interest in newborn screening (such as an endocrinologist, hematologist and/or metabolic disease specialist), business men or women, and may also include legal, ethical and religious representation. Most newborn screening advisory committees have found it essential to include several lay advocates - individuals with disorders detectable by newborn screening or members of families of affected individuals. It is generally agreed that committee staffing should be provided by the program and interested follow-up, administrative, and laboratory personnel should be encouraged to attend meetings to provide technical information. However, in order to achieve the goal of obtaining outside program advice, program personnel should not have a formal role in committee deliberations or voting. It may be also be useful to have an internal advisory committee that includes personnel and subspecialty consultants to guide program operations.

The Newborn Screening Advisory Committee should meet regularly and formally, with an appropriate agenda that includes brief descriptions of the issues to be discussed. The agenda should be available to members well in advance of committee meetings. Minutes should be a part of the formal process and should be widely and actively distributed to any interested party following each meeting. TNSP staff should assist with scheduling, agenda preparation, travel arrangements, etc. Teleconferencing is an option for some of the meetings in order to decrease costs, and all who were interviewed about the advisory committee process expressed interest in participating in

such conferences. However, there was general agreement that at least one face-to-face meeting of the committee annually was needed.

The Review Team understands that if a formal advisory committee is organized, the membership may have to be defined by statute. Whether or not this approach is taken or there is a less formal approach, the Review Team agrees that there is a need for a more formal committee structure (which includes a mission and defined operating protocol) than is currently in place. In whatever form this may take, it is essential that the committee understands and agrees to its role and its rules. There must be a clearly stated mission that includes a defined committee role and process for communication with the program. Most programs have found that a strong independent chair with standing in the medical or consumer community is helpful. Some programs have used committee co-chairs to help ensure that personal agendas do not compromise the committee's effectiveness.

A functional Advisory Committee can be a powerful advocate for the program. The Committee can be asked for advice on adding or subtracting conditions to the screening panel and other important cross-cutting issues such as financing. The Committee may also be useful in providing advice on many other program issues including legal and ethical issues, public relations, professional and consumer education, interactions with the health care community, and other program priorities. Program decisions made within the advice of outside advisors should lead to stronger support for their implementation. Without participation from the community that provides program support, namely clinicians, hospitals, parents and families, then the program faces a continuing uphill battle for its survival and effectiveness.

In Texas, a formal Newborn Screening Advisory Committee would likely oversee smaller working groups or subcommittees (such as the current ad hoc groups) with specific interests such as hemoglobinopathies, metabolic disease, endocrinopathies, parent and professional education, and community/consumer affairs. Other ad hoc or standing work groups can be formed as needed - for example for consideration of parent issues, screening for cystic fibrosis, biotinidase deficiency, or lysosomal storage diseases. Any funds needed to support the work of the Advisory Committee or its subcommittees should be included in fee considerations.

2.3.0 Recommend mechanisms for utilizing the existing network of physician specialists in the state for newborn screening consultation and ways to enhance/expand access. Identify any states utilizing telemedicine successfully.

The TNSP currently utilizes groups of consultants for each of the different classifications of disorders (metabolic, endocrine and hematology conditions) based on previous program experiences and interests expressed by various subspecialists and academic centers. These consultants assist the program by providing subspecialty expertise as a resource for the general clinical community. Where needed, they assist in conveying important information about the severity of the conditions to physicians and patients in order to ensure timely and accurate follow-up of presumptive positive screening results. In particular, the subspecialists advise on appropriate confirmatory

testing and/or clinical evaluation. In cases where a high probability of a disorder is suspected, consultants' names and contact information are provided to primary healthcare providers at the same time that result letters are sent further explaining positive findings. The healthcare providers are instructed to make contact with the subspecialists who will assist them with their follow-up testing. Subspecialists may also be sent testing results so that they are aware of possible cases in their catchment area. In all of the follow-up scenarios encountered by the Review Team, it appears that the subspecialists are expected to assist with follow-up activities, but there are no formal agreements in place nor is there any reimbursement available for any of the linkages and preliminary contacts that occur.

It has been the collective experience of the programs represented on the Review Team that a more formally organized mechanism for utilizing subspecialty input, advice, and program assistance provides a more effective way of handling follow-up. That is, a system of formally identifying and recognizing subspecialty service provision through grants and contracts seems to provide a better system of medical service to the families of patients identified with conditions included in the screening program. Indeed, the Review Team encountered dissatisfaction in the current follow-up procedures in almost all of its interactions with subspecialists. Examples of problems with communication between subspecialists and the TNSP were cited, including reports of test result information being faxed to physician's offices without first alerting the offices/clinics that such information was coming. This lack of coordination resulted in faxes lying unread for several hours or even days. Additionally, examples of delays in contact coming from local healthcare providers who had received result information but had not contacted subspecialists in an expeditious manner were also cited. These delays were also attributed to a lack of coordination in result transmittal to the primary healthcare provider.

The Review Team suggests that the TNSP reevaluate the way in which information is transmitted to the clinician community, both the primary care practitioner and the subspecialty consultant. Because the information is sometimes complex, depending on the condition suspected, it is important that a person knowledgeable about the basic information relating to a condition convey the follow-up request. Thus, additional nursing or program specialists may be needed in order to adequately transmit the proper information. This type of follow-up will likely require a budget increase since persons with the technical backgrounds needed (nurses or equivalent) may require higher salaries, and these considerations must be factored into the overall financial scheme. Ultimately, it is the recommendation of the Review Team that all program components be included in the fee such that adequate services can be provided. At the current time, the comprehensiveness of the follow-up system in the TNSP appears to be less than that desired and this relates directly to the perceived inadequacy of follow-up financing. The Review Team noted that follow-up nursing staff turnover has been significant and this likely relates to some of the difficulties being experienced in the field, but a system to provide adequate follow-up in circumstances where turnover is being experienced should also be a part of the follow-up and funding system. A plan for retention of employees should be developed.

2.4.0 Recommend mechanism for DSHS to improve physicians compliance with reporting of newborn disorder diagnosis. How does Texas' experience compare with other states?

Initially the Review Team was confused by this question since the word 'compliance' seemed to be out of place. However, clarification was given that there is a requirement for physicians to report diagnoses resulting from newborn screening to the TNSP. The physician community may not understand this requirement since the Review Team had difficulty understanding it. The Review Team agrees with the principle of the request and feels that this provides an excellent mechanism for obtaining needed follow-up information from the physician community. Education about this requirement should be highlighted in any communication from the program to the clinician community, including transmittals of test results, program newsletter, etc. Involving physicians and nurses in the decision-making processes suggested elsewhere in this report might also lead to improved cooperation throughout the clinician community if this issue is given visibility. For example, there may be vocal opposition or nonresponsiveness among the clinician community because of a perceived unfunded mandate relative to this and certain other program requirements. Some of those interviewed indicated that the present fee collection process already places an undue financial hardship on the practitioner who must collect the fee from uncooperative insurers with little assistance offered by the DSHS. The New Mexico program is employing discounted fees for certain aspect of program compliance and it may be appropriate to contact that program to learn if the incentives being used are working. If so, then perhaps some sort of discount for submitting timely and accurate diagnostic information would be appropriate for consideration in Texas. If there continue to be problems with diagnosis reporting, then the advisory committee(s) or the various medical professional societies and lay advocacy groups should be consulted for assistance and advice. A short message in *Texas Medicine* might also help.

2.5.0 Is the second newborn screen effective?

Ever since programs began requiring second screens, there have been discussions about the effectiveness of the second test. Initially, a second screen was recommended whenever there was a question about whether the first test was collected too early for physiologic processes to properly affect the analyte of interest. Thus, programs have suggested almost from the beginning of newborn screening that a specimen taken before sufficient protein intake might cause the test for PKU to be falsely negative. In 1979, a second newborn screening test was recommended by the Texas Newborn Screening Advisory Committee, and subsequently by the Department of Health, for all newborns tested before 36 hours of age and 24 hours of protein intake, (*Ligand Review* 1980;2:12-14). The original intent was to detect cases of phenylketonuria (PKU) that might not be detected by routine screening procedures in babies tested too early (before sufficient protein intake) and was in accordance with recommendations from the American Academy of Pediatrics (*Pediatrics* 1982;69:104). The recommendation for a second test led to an immediate increase in specimen testing volume of approximately 40% as physicians began to react. When CH screening began in 1980, occasional cases of CH were detected on the second screen when the initial screen showed normal results. These

findings were supported by other programs performing second screens, most notably the Northwest Regional Newborn Screening Program in Oregon (*Pediatrics* 1985;76:734-40). Because these findings were considered significant (~7% of all CH cases detected on second test or an overall incidence of ~1:40,000), the recommendation that all babies receive a second screen at 1-2 weeks of age was changed to a requirement in 1983 for babies screened prior to 36 hours of age and 24 hours of protein intake (*Pediatrics* 1986;78:375-6).

Over the years, the Texas program and others have continued to detect cases of CH and certain other conditions through the analysis of a second specimen. There are currently 8 programs that require a second screen on all babies and several more that strongly recommend it to the extent that compliance exceeds 80%. All programs require a second specimen if the first was taken 'too early' (the definition of 'too early' varies but is generally considered to be before 24 hours of age). Multiple reports from programs with second testing in place have been compiled [*Early Hospital Discharge: Impact on Newborn Screening, Proceedings of a conference held in Washington, D.C., March 31-April 1, 1995*. (Available from the National Maternal and Child Health Clearinghouse)] When screening for congenital adrenal hyperplasia was introduced in Texas in 1989, even more cases were detected from second screens. In particular, an in-depth survey of 1.9 million Texas newborns screened showed that 61% (17 of 28) of the classical simple virilizing cases of CAH detected through screening were detected solely on the basis of the second screen along with 87% (46 of 53) of non-classical cases (*Pediatrics*1998;101:583-90). This means that the detection rate from second testing for clinically significant simple virilizing CAH is about 1:110,000 or 3 - 4 cases in Texas per year.

Testing with tandem mass spectrometry is relatively recent and is available only in a few places where second testing is routine. Within the first year of operation in the Oregon program, several cases of metabolic disorders were reported from the second screening test following a normal initial test. These data were reported in preliminary form at the 2004 National Newborn Screening Symposium and are currently being prepared for peer-reviewed publication. This would indicate that second screening in Texas is likely to find more cases from second testing when tandem mass spectrometry is added to the program.

The Review Team included representatives from several programs that utilize a second screen including Delaware, Oregon, and Washington. In all cases, there was general satisfaction with the second screening experiences relative to program accomplishments. These programs pointed to several advantages of the required second screening, some of which have already been mentioned:

- Maximizes disease detection, especially for congenital hypothyroidism and CAH (and offers occasional benefits with some of the metabolic conditions, including experiences in Oregon with MS/MS testing).
- Helps to verify results of the first newborn screen. If there was a procedural error resulting from misidentification of patient, laboratory punching mistake, a clerical

error, or other system failing, the second specimen provides a timely check on the accuracy of the first test.

- Provides a mechanism for the primary care physician to ensure testing on all patients, in cases where initial testing results may not have been forwarded from the birthing facility, since the second specimen will usually occur at the primary care physician's office. *[It should be noted that some professionals who were interviewed reported that testing results from the initial specimen were often not available by the time the patient reported for their second screen at 1-2 weeks.]*

Given the experience of the Texas program when second testing was started and the experiences of some of the other programs that recommend a second screen, it is likely that if the requirement for a second screen were modified to a recommendation, there would continue to be a significant number of specimens collected as a second test 'safety net.' Likewise, experiences in Washington (*Am J Med Genetics* 1995;59:417-20) indicate that when second testing is not required on all infants, there is selective testing based on factors such as insurance and race.

2.6.0 Is linking all first and second screens effective and efficient?

In order to confirm that all newborns receive newborn screening, it is desirable to link the newborn screening experiences to birth certificates, which provide the ultimate record of babies born within the State. To have the most impact in improving child health, access to public health information, including all screening results and service encounter information, should be easily accessible to the child's medical home. Currently, only a limited number of public health systems are attempting to provide comprehensive program integration of child health information so that this can occur, and some privacy issues remain to be addressed before open and timely access to child specific public health information at the medical home can occur easily. Nonetheless, integrated information systems are being developed in many public health departments in order to minimize duplication of data maintained within the public health system, and to make basic client information (including linked first and second newborn screening information) available to multiple programs from a single information source. Ultimately, these patient public health records might be integrated into a child's electronic medical record maintained at the medical home. Thus, secure medical information may eventually be conveniently available to healthcare providers through the Internet or other electronic means (e.g. personal information devices) as it is needed.

Several states have integrated newborn screening testing records with patient demographic information so that automated voice response and fax reporting of newborn screening test results is available at the physician's office (with appropriate security considerations) upon request 24 hours a day. Such a system existed at one time in Texas but does not appear to be operational at the present time. The Review Team encourages the TNSP to reestablish the voice response service and to consider its availability through a toll free line. Programs that have used a toll free voice response system with faxing capability to make test results available to clinicians on a 24/7 basis have generally been pleased by the reception and response from the healthcare community.

The linking issue in Texas, while important for program efficiency and effectiveness, seems to relate to some issues that surrounded fee enactment in 1998. At that time, requiring primary care physicians to participate in the fee collection process seems to have left a negative impression on some physicians that continues to persist. A fee was required to obtain the specimen collection cards for both the initial and the second specimens for patients with an ability to pay (i.e. insured). There was no charge for specimen collection cards if the patient was uninsured or covered by Medicaid. The birthing facility was the usual payer for the initial specimen, and the primary care physician was the usual payer for the second specimen. In order to lessen the financial burden to both parties, the fee collection process extends to 120 days after shipment of the newborn screening blood collection forms (fees are collected based on forms ordered).

At the time the fee was initially implemented, it was reported that some physicians indicated a reluctance to perform the second screen, choosing instead to ask the patient to return to the birthing facility for testing. These actions were thought by some to have decreased compliance with the second testing requirement in the State. While the TNSP monitored the numbers of newborns receiving second testing for the first months following fee implementation (in order to ensure that the fee collection procedures did not have a negative impact on the program), it appears that collection of these data were discontinued (even on a periodic basis), and this issue is still a concern in the physician community. There is also a feeling that because of a lack of (or no) uniform third party payment for second testing, more physicians may have elected to send their patients back to the birthing facility as time has progressed. Thus, it would appear prudent for the TNSP to more formally evaluate whether or not all babies are receiving the required second testing, at least periodically. Comparison should be made to the second testing compliance prior to and immediately after fee implementation to see if there has been any significant change since 1998.

The perception by some in the Texas medical community is that DSHS was moving towards specimen matching with a newly purchased computer system. It was anticipated that this system would be able to address issues related to compliance with second screening requirements. The computer system currently in operation does not appear to have the capacity to efficiently (without extended manpower) match first and second screens on a routine basis. Although, the system performs limited (not fully automated and requires some manual interventions) matching to assist the follow-up group in matching repeat screens with those awaiting retesting as a result of initial presumptive positive testing results. Additionally, there is delayed matching process performed through a complex matching algorithm to accommodate Medicaid reimbursement requirements, but these matches occur at least 6 months following receipt of the specimens. The effectiveness of matching algorithms of this type are improved by increasing the number of matching fields, but this also slows the process. While effective for a billing process such as the one described for Medicaid, an extended matching algorithm process is usually not effective where speed is important such as in laboratory specimen check-in. A real time, efficient matching system is needed to evaluate whether all Texas newborns are receiving the required screens and to address potential barriers for those who are not.

Some programs have used various types of matching algorithms, such as discussed above, but they have usually required overnight matching in order to accommodate the processing time requirements. With such systems, data entry accuracy is essential. For this reason, automated download of data from the submitting facility (previously reported to be used by Parkland Hospital in submitting data to the TNSP, but no longer available) is a useful newborn screening data collection option. The use of a specimen serial number, birth certificate number or other unique identifier on the second or subsequent specimen forms that can easily link to the initial specimen form is the matching solution used in a number of programs.

While linking all specimens tested is not an easy task to accomplish, there are several examples of successful linking systems in other programs that may be of use in Texas. Some of the programs that require all newborns to be tested twice use linked specimen forms using either the same or adjacent newborn screening collection card's serial numbers. Thus, at the time the first specimen is collected, the second form is given to the parents for submission to their primary care physician for the second test. The success of this technique has been mixed. The primary problems are loss of the form and the possibility of form invalidation due to the method of storage until the second specimen is collected. A pilot test of this type of linking appears to have been tried in Texas at the time the second test was initially required and found to be unsuccessful for this reason. However, this pilot testing was performed many years ago and it may be worth repeating in an effort to improve the current linking process.

Another mechanism for linking (which was previously tried in Texas as part of the immunization program) involves a "birth passport." In this model, the newborns parents are given a birth passport at the time of birth that contains an identification number that is the same as the newborn screening collection card serial number. The passport may contain multiple tear off copies of the bar coded serial number that can be applied to other records, if desired. The serial number provides a unique linkage between other child health data systems as long as the number is captured in each system. Thus, for example the identification number may be listed on the birth certificate, the immunization record, etc., and the physician obtaining the second newborn screen may use the number as an identifier when submitting the specimen to DSHS.

In summary, the Review Team agrees with the concept of linking first and second specimens as a means of ensuring that all newborns receive the required testing. Further, the Team is aware of several programs that either have developed, or are developing, mechanisms for accomplishing such linkages. Linkage can be aided by use of serial numbers on the initial form and their inclusion in some manner as part of the second specimen form - either by linked forms, by inclusion of the initial specimen number in the data accompany the second specimen (in which case the parent or birth attendant will likely need to provide this number to the primary care physician - as with a " birth passport"), through linked electronic records available at the office of the primary care provider, or by some other innovative procedure. As an interim step, consideration should be given to confirming periodically that the numbers and percentages of initial

and follow-up specimens currently received is approximately the same as at the time of fee implementation in 1998.

2.7.0 Assess the current staffing for DSHS newborn screening case management activities. Assess existing protocols and practices and recommend ways to improve, enhance and gain efficiencies.

The case management activities are part of an overall system that includes program administration, short-term follow-up, education, and quality assurance. The staff must be adequate to administer all of the policies, procedures and educational activities associated with case management. Short-term follow-up and case management begin when a laboratory result is generated indicating a presumptive positive test result, receipt of unsatisfactory specimen or test. In either case, contact with the submitter is necessary. The submitter may be requested to validate or complete necessary demographic information, to submit another dried blood spot specimen, and/or to expedite clinical evaluation and serum confirmatory testing. Once the initial contact has been made, it is necessary to follow-up by checking to see if an additional sample has arrived and been analyzed or if the clinical confirmation has been accomplished. Thus, the case management staff must be continually making contact with submitting birth facilities and clinician's offices, and must be continually monitoring completed laboratory processing. In addition, there is a continuing need for education of healthcare professionals and the public, and for program evaluation through data collection and review. Currently there are three full-time nurses assigned to handle the telephone calls regarding rapid follow-up of extremely abnormal testing results. Each of these in turn oversees 3 public health technicians assisting with other follow-up activities, including reporting of results to clinicians who call in. Information on the number of calls received by the program appears to be lacking. Additionally, there is a medical director and an educator who is responsible for the program's website content and development of other educational materials.

Reviews of the existing protocols and practices for follow-up found that the follow-up practices were generally sound and in keeping with that performed in other programs. There appeared to be an excessive number of prepared letters in use and the Team suggests that all letters be reevaluated for appropriateness and consolidation. Although the program aims to have parent letter at the fourth grade reading level, some letters to parents appeared to be of a reading level higher than desired for information of this type. All letters should be evaluated for reading level and adjusted to meet the needs of parents and families. In particular, there were words describing the consequences of some of the screening conditions that seemed to be too scientific and not parent friendly.

During the program review process, the two teams that visited with various medical personnel associated with the TNSP were advised of apparent shortcomings of the follow-up process. Several clinicians indicated that results requiring immediate follow-up were faxed to them without a corresponding telephone call to alert them to the incoming faxes. Some also reported that healthcare personnel seeking subspecialty assistance also indicated receiving reports in a similar manner and their decision to seek subspecialty assistance was slow due to a lack of emphasis on the urgency of the needed

follow-up. There were also reports of difficulty in contacting a knowledgeable follow-up person at the case management office. While these difficulties may be the result of staff turnover and a lack of back-up capacity, these repeated reports led the Review Team to question whether or not an appropriate number of staff currently exist in the case management program and whether or not they have sufficient technical expertise to convey the necessary follow-up information. Adequacy of phone coverage during regular working hours (7:00 a.m. - 5:30 p.m.), after hours, and on weekends/holidays needs to be assessed.

The Review Team also noted that while there appears to be a legal requirement for the TNSP to maintain a registry of cases, this is only being done in a superficial way. There did not appear to be a formal mechanism for maintaining a case registry, nor was there an active effort to maintain any long-term follow-up data. The Texas program, because of its size, has an opportunity to learn more about the history of the conditions included in the newborn screening program. Long-term case management can be combined with a case registry to accomplish both the legal requirement for maintaining the registry and the scientific and programmatic interests associated with learning more about rare conditions on the basis of large population studies.

Because of the size of the Texas program, the Review Team did not feel that it had adequate information to address the question regarding adequate case management staff. Instead the Team suggests that a detailed staffing analysis be undertaken with particular emphasis on adherence to the follow-up procedures written in the operating manual. Additionally, this review should extend to the subspecialty centers and should include community-based clinics (local health departments) and primary care practices in order to evaluate whether or not proper and timely follow-up is occurring. It may be the case that additional staffing assistance at the subspecialty clinics is necessary in order to adequately address the needs of the program. If this is found to be the case, it may also be a more efficient use of funds to subcontract some of the follow-up activities regionally to ensure adequate and equitable follow-up and case management. In fact, it was noted that at one time hemoglobinopathy follow-up assistance was provided in some regions through a federal grant, but once the grant ceased the State did not sustain the program (even though it was considered to provide effective follow-up services). As a comparison, the Review Team noted that the California program, which obtains about 570,000 newborn screening specimens annually from 280 birthing facilities (compared to 750,000 newborn screening specimens from 200 birthing facilities and hundreds of primary care providers submitting second specimens in Texas), utilizes some 25 case management staff spread over 7 regional centers - significantly more than in the Texas program. Additional staffing also exists in the California program to cover program evaluation and education.

A summary of possible actions that may be appropriate to assess the capacity of the case management office include:

- Evaluate staffing and workload to ensure appropriate short-term follow-up services.
- Evaluate nurses' workloads and task assignments; consider redistribution of tasks or addition of staff to ensure nurses are available to make required phone calls,

field incoming phone calls from health care providers and perform other short-term follow-up tasks.

- Review Public Health Technicians' workload and evaluate appropriateness of tasks.
- Review current methods utilized for fielding consumers calls (i.e., hospitals, health care provider and parents) and consider methods to improve access to follow-up personnel such as pagers for nurses, caller ID displays, personalized voice mail messages that provide options to the caller for immediate assistance (i.e., nurse's page number or option to "dial 0" to reach a receptionist).
- Consider reinstitution/institution of a 24-hour Voice Response (and Fax) System that may assist in fielding some calls from physician offices, particularly those seeking reassurance that the results were properly transmitted.
- Consider an Internet-based reporting system with similar access capabilities to the Voice Response System, including security issues, and community support/availability issues.
- Assess turnover of follow-up staff and develop a long-range plan for recruitment and retention.
- Prioritize DSHS staffing to administer and assist with TNSP case management and education activities, and consider providing long term follow-up services in collaboration with other programs such as Children with Special Health Care Needs (i.e., care coordination services), Birth Defects, and Early Intervention.
- Evaluate staffing requirements that might be needed in order to comply with legal requirements to maintain a case registry.

2.8.0 Recommend ways to ensure the efficient expansion of NBS in Texas based on lessons learned from other states and their applicability to Texas' system, size, etc.

Currently, newborn screening programs expanding to include tandem mass spectrometry (MS/MS) have taken various approaches to deciding which disorders to include in the screening panel. One approach has been to mandate only the disorder(s) of higher prevalence or major interest (e.g. only MCAD or only a limited group of disorders -- MCAD, MSUD, GA-I, HCY, etc.). Most programs choosing this approach collect data on the other disorders with the intent of adding them at a later date when, and if, a significant prevalence is demonstrated. Another approach has been to mandate a limited number of disorders and offer the others as optional testing. In this approach, there is an ethical consideration to inform the parents of their rights to refuse the optional testing and to have a system of identifying the samples that should or should not be tested. Using a graded approach to adding rare metabolic disorders allows time for the program (laboratory, follow-up, and treatment) personnel to become familiar with the testing, follow-up and diagnosis process. A third approach has been to mandate (add) the disorders to be screened according to a biochemical classification system mirroring the analytical profiles that are possible [i.e. all of the fatty acid oxidation (FAO) and organic acid (OA) disorders and/or all of the aminoacidopathies]. Although many of the FAO and OA disorders may not individually meet traditional newborn screening prevalence criteria, when combined with all possible disorders observable with the technique, the combined prevalence is significant and meets the prevalence test. Justification for treating the disorders as a group is valid since the analytical technique, including sample

preparation and analysis, is a single procedure that prepares and analyzes the sample for all of the disorders (analogous to isoelectric focusing for multiple hemoglobins). This is essentially the approach recommended in the recent report of the American College of Medical Genetics (ACMG) to the Health Resources and Services Administration (HRSA).

There are many questions that must be answered within the TNSP before MS/MS testing is integrated into the screening system. In the April 13, 2001 issue of the CDC publication *Morbidity and Mortality Weekly Report (MMWR)*(see Appendix 8), recommendations were provided regarding the implementation, follow-up, and diagnosis/treatment of screening disorders currently detected by MS/MS newborn screening. These recommendations resulted from a meeting held in June of 2000 in San Antonio, Texas at which an invited working group of public and private MS/MS screening programs reported their experiences. It is strongly suggested that the TNSP review the recommendations given in the MMWR as part of their considerations concerning MS/MS implementation in Texas.

Thus far, there are two different options that have been instituted by state governments as an alternative to mandating expanded testing with MS/MS. In South Dakota, Montana, and Nebraska, supplemental testing is offered as an option, with the parent responsible for the testing cost, and the sample is sent to a testing laboratory out of state. Summary data from these tests are returned to the state newborn screening program so that they may be followed up and used in assessing the value of the program. In New Jersey, a law now requires that information about supplemental testing availability outside of the State mandated testing be given to the parents so that they might be aware of other options. A legislative resolution suggesting a similar approach was passed in Illinois several years ago and a law (like the New Jersey law) existed in Mississippi prior to the current law that requires the expanded testing. In both of these alternatives, testing cost is an issue and those without the ability to pay might not be able to take advantage of such testing. This is one of the major differences between optional and mandated testing programs - in the mandated program, all newborns must receive the screening without regard to their ability to pay.

Depending on the manner in which expanded MS/MS screening is implemented, the increased laboratory testing cost alone (without the follow-up/education component) would likely be in the neighborhood of $5-10 per newborn based on the experiences of other public health laboratories. The increased cost of follow-up/administration/education is a separate issue but would be expected to be a similar amount. The laboratory cost would presumably be included in the newborn screening fee, and the follow-up costs would be covered by other means unless a program change allowed its inclusion in the fee. To assist with start-up costs, many programs have found it necessary to increase their screening fee in advance of offering routine testing in order to pay for pilot testing and other start-up expenses including instrumentation and education. While it might be possible to obtain external grant support for program start-up, the funding available for this approach has been meager with only one or two laboratories receiving grants for this purpose. Other external funding sources might also be considered. For example, the March of Dimes (MOD) local chapter in Georgia agreed

to assist the state in funding four tandem mass spectrometers (including one for confirmatory testing at the university) for expanded newborn screening. While this is not a national MOD policy, it serves to illustrate the potential innovative funding sources that might be available to assist the program.

When a program decides to expand with MS/MS testing, as with Texas, program implementation can be a long process due to factors such as funding, instrument acquisition, operator training, pilot studies, educational activities, follow-up planning, etc. While there are ways to decrease the phase-in time, the fact remains that some lead-time is necessary before embarking on expanded MS/MS testing. Selected information from deliberations in Massachusetts are included in Appendix 7 as an example of their process for deciding on the way in which to expand their testing program. Several implementation options exist for expanding screening services if the decision is made to expand. While mandated program expansion with an appropriate fee increase and full coverage of all conditions described in the ACMG report (Executive Summary in Appendix) is the preferred mechanism for program expansion, other options exist. The most popular options for phased-in implementation are outlined below in order of acceptability to the reviewing team:

1. *Offer MS/MS testing as option and contract with an outside laboratory (public or private) for the laboratory portion of the program.*

   The main advantage of this option is that it allows the TNSP to maintain control over outside laboratory involvement in the screening system while it develops it own comprehensive screening system. Through a contract process, the TNSP can control the manner in which samples are collected and submitted, the way in which results are reported, and the way in which follow-up is handled. This option also allows for easier transition to the state laboratory once testing is available. Logistical difficulties that must be considered include the manner in which samples will be submitted to the testing laboratory and mechanism and amount of payment. If testing under this protocol is at the option of the parent, some will elect not to have the testing. Thus it will likely be necessary to have a separate billing procedure for those opting not to participate in the program. A contract mechanism whereby billing and payment for optional testing is the responsibility of the contracting laboratory may simplify the process. The screening programs in South Dakota (using the Institute for Metabolic Disease in Dallas) and Nebraska (using Pediatrix Screening, Inc.) have experience with this option and should be contacted for further information and advice if it is considered. Other public laboratories that might be interested in such a contract arrangement include the Oregon, Iowa, Delaware and Massachusetts newborn screening laboratories.

2. *Mandate testing for disorders detectable by MS/MS and contract with an outside laboratory (public or private) for this portion of the program.*

   This option allows for mandatory population screening without the need to expand the testing laboratory services in Austin. The advantages and

disadvantages described in "A" above apply here with the exception that it would not be necessary to have a billing option for those opting not to have the testing since testing would be part of the comprehensive newborn screening program. There would still be the need to increase the screening fee to offset expansion follow-up/administration/education costs. Because there are legal requirements regarding testing at the DSHS or an approved laboratory, it will be necessary to consult with general counsel before this (or other) outside option is pursued. Likewise, should the outside laboratory become unable to continue its testing obligations, then an alternative laboratory would need to be quickly located, which might be difficult unless it is addressed in advance. As an aid to fee deliberations, Pediatrix Screening, Inc. indicated in conference with the Review Team that its fee of approximately $29 in Mississippi is of the order of magnitude for providing comprehensive screening in Texas on a per specimen basis. Given the current $19.50 cost of the 5-test mandate in Texas, it is reasonable to assume that another $10 would be an approximate laboratory testing increase. Information available to the Review Team indicates that an additional cost of approximately the same amount is used for follow-up in the Mississippi program.

3. *Delay testing until it is available at the state laboratory meanwhile allowing primary care providers (or parents) to send testing to other laboratories apart from the state program.*

   The advantage of this option is that parents can immediately have access to expanded screening since it is already available. This option maintains the current payment scheme since parents would have to submit payment to testing laboratories as samples are submitted. It has the disadvantage that not all patients can afford testing and the data would not necessarily be available for the state to use in evaluating the utility of the testing. Additionally, duplication of some of the tests performed by the state might occur since some private laboratories offer these tests as part of their testing panel. It is also likely that samples will occasionally be mixed up with the wrong one being sent to the state laboratory and vice versa. A possible solution for maintaining the data would be for the DSHS to direct the primary care providers to a single outside laboratory with which the DSHS negotiates a contract (as in Option A above).

The current staffing experience among state public health laboratories is that medical technologists and chemists are effective MS/MS operators as long as they possess the technical abilities outlined below. Although instrument software is advancing to the point where interpretation of routine MS/MS spectra can be performed by the operator and/or manager, there is still a need for access to a qualified expert (MD, Ph.D., Geneticist, etc.) to assist in interpreting and reporting unusual MS/MS output. The MMWR Report recommends that MS/MS operators have a minimum of a Bachelor's degree in an appropriate science, and that operators should have: (1) good mechanical aptitude, (2) strong computer skills, and (3) a fundamental interest in mass spectrometry technology. Managers should have background experience in mass spectrometry.

Operational newborn screening laboratories with MS/MS experience have done extensive pilot testing to establish abnormal acylcarnitine and amino acid profiles for the disorders they are reporting. Preliminary data shared between programs seems to show fairly good agreement in spite of different instrumentation used. Sharing information of this type should help minimize the start-up period for new programs starting MS/MS testing. Due to the possible unfamiliarity of primary care providers with these disorders, the program should have an expert(s) (preferably a board-certified biochemical geneticist) available to assist with interpretation inquiries. Inquiries by the Review Team seemed to indicate that sufficient biochemical expertise is available in the State.

As with all newborn screening, abnormal analytical findings reported from the newborn screening laboratory must be followed up by a competent diagnostician with access to competent confirmatory laboratory services. There will always be testing results that do not clearly indicate whether or not the newborn has a medical condition requiring diagnosis and treatment. Screening is designed to reduce the number of patients that might go undiagnosed by identifying those at risk for the disorder. This invariably means that some will be identified that do not have the disorder suspected. The idea of improving screening techniques through program evaluation is to reduce these 'false positive' findings while eliminating the 'false negatives.' The amount of follow-up required will not truly be known until the program is implemented, but data from other programs should be helpful in making these determinations for planning purposes. It is suggested that reports from other states be closely analyzed in this regard. Most programs appear to be experiencing follow-up of less than 0.5% of patients screened, but this follow-up is a major concern of the subspecialty centers who already view their role as an unfunded mandate. Subspecialists interacting with the Review Team expressed a continuing concern that expanded screening further will further exacerbates their funding issues relative to newborn screening follow-up.

Given the fees charged in other programs for screening and the support system that is necessary, it is the opinion of the Review team that the current fee structure and other funding sources within the DSHS would have to be significantly increased to support an expanded program in a manner sufficient to ensure an adequate full-service newborn screening system. Further, it is also noted by the Review Team that the budget currently supporting the follow-up/administrative/education system of the TNSP is extremely low and does not adequately fund the services necessary for these aspects of the program.

The Review Team recommends that the elements of a comprehensive newborn screening system sufficient to adequately meet the current needs of Texas newborns be defined by a task force that includes, at a minimum, representatives of: the DSHS laboratory, the DSHS follow-up/administration /education program, each of the three ad hoc advisory groups (hematology, endocrinology, metabolics/genetics), the CHSCN program, parents with children having conditions representing by each of the three advisory groups, the TMA, the Texas Hospital Association (THA), the TDI, the nursing association, the insurance industry, and the Medicaid program. Once the appropriate system to meet current needs has been defined, a thorough cost analysis of the defined system should be completed and fee adjustments considered for its implementation.

In order to consider program expansion, a definition of the desired expanded system should be completed along with a thorough cost analysis. This task force (which could be the task force described above), in addition to the potential members previously listed, should also include parents of children with conditions to be included in the expansion and subspecialists with expertise in the expansion conditions. This would include, for example, a representative of the cystic fibrosis centers, a biochemical geneticist involved with metabolic conditions detectable by MS/MS, and parents of a child with a condition representative of the condition(s) under consideration. Rule changes should be effected as the system is defined and changes are needed. In the interim, steps should be taken to share information with the public about alternatives available to supplement the universal newborn screening requirements currently in effect. Given the current statute and program rules, it appears that a clarifying statutory or rule change may be necessary to allow program elements other than laboratory testing to be included in the fee. Likewise, a fee increase in advance of actually providing testing for the entire population (e.g. for test development and pilot testing) would facilitate financing program improvements (including expansion), and statutory or rule changes may be necessary for this to occur. Should testing in another laboratory be the preferred alternative, the current statute and rules already allow for this to occur. Under the current statute, a Program Rule change would be necessary to increase fees above $30 and to facilitate any changes in the method of fee collection, should this be a consideration (i.e. to charge a single fee for both required screening tests).

2.9.0 Recommend mechanism for DSHS to improve submitters' completion of patient demographic information on the NBS specimen collection card.

The most obvious mechanism for improving the completion of the demographic information portion of the newborn screening collection card is education. Educational In Service training at birthing facilities is a system component that is often overlooked or given diminished recognition due to budgetary constraints. However, many programs have staff dedicated to providing periodic training to birthing facilities in an ongoing effort to ensure quality throughout the entire screening process. These education and training needs could be met with a system of regional follow-up coordinators, The professional education provided often focuses on proper specimen collection techniques, which includes completion of demographic information. This is a critical part of the CLSI (formerly NCCLS) standard on blood collection on filter paper (Standard CLSI/ LA4-A4) and is graphically demonstrated in the accompanying video. This video or something similar is often made available to birthing facilities as one part of the education process and provides the potential for improving data completion.

Many programs now use “screening practice profiles” as a way of informing specimen submitters of their problems and successes. A computer surveillance system can be easily developed to monitor errors in specimen collection, demographic data entry, and handling. Such systems are available from software vendors on request or are easily developed from software system using common report generating software such as Crystal Reports. The TNSP can decide which errors to monitor and can use the profiles not only as a quality performance measure for birthing facilities and physician

offices, but also as a way of determining where educational interventions are most needed. Care must be taken in defining “errors” so that the errors identified are appropriate to improve the system.

The New Mexico newborn screening program reported using a reduced fee incentive program for birthing facilities to improve their sampling technique, and it would appear that this could be extended to demographic data. The Review Team suggests that the New Mexico program be contacted to see how this system has worked.

Some programs have reported success in having follow-up personnel or educators as members of birthing facility QA committees and in this way, improving screening practices. Another consideration is encouragement of hospitals to adopt newborn screening as a quality assurance performance measure. These committees usually address specific topics over a short period of time, typically 6-12 months, and have the potential to address specific issues such as the facility's newborn screening system. In addition to looking at ways to ensure the testing of every newborn, they would also concern themselves with accurate data transmittal and proper specimen collection, record keeping and transmittal to the screening laboratory. Accurate and complete data transcription and transmittal are also part of a hospital's responsibilities under the licensing requirements and are subject to review during the inspection process by their licensing authority.

Since the TNSP publishes a periodic newsletter and an active website, both of these present a possible mechanism to spread the word about the need for more complete data. Likewise, emphasizing this point in the Practitioner Manual should also be considered. For example, the section on page 6 regarding "Unsatisfactory Specimens" might be expanded to include demographic information. By using different colored ink or conversation boxes to emphasize particular points, the busy reader may have his(her) attention directed to the issues deemed important by the program.

For repeat offenders, personal contact by telephone or e-mail may provide increased compliance, and as a last resort, the hospital administrator might be a useful contact emphasizing increased liability for the facility if the information is not accurate and complete. Offer concrete suggestions to facilitate completion of the demographic information. For example, a clerk might complete the majority of the information ahead of time leaving only the collection date and time to be filled in at time of specimen collection. In such cases positive identification of the newborn with the information on the card must be part of the procedure.

2.10.0 Recommend mechanism for DSHS to improve/decrease NBS specimen arrival time at the DSHS laboratory.

Some states have contracted with private couriers (UPS, Fed Ex, DHL, etc) to pick up and deliver specimens to the newborn screening laboratory. The use of these private couriers has improved the transportation time for specimens by 24-36 hours in

comparison to routine specimen submissions through the U.S. Mail. On average programs have reported a cost of approximately $1 per specimen for these services. Utilization of courier service, at least at the larger birthing facilities, would likely result in some significant improvements in transport time in Texas, particularly in areas where mail service is known to be slow. Couriers routinely operate 6 days a week and the potential for decreases in specimen delivery times leads to consideration of a 6- or 7-day workweek. The need for an extended workweek is already an issue in the TNSP, since death can occur during the first two weeks of life for galactosemia and congenital adrenal hyperplasia, and rapid testing and result turnaround is critical.

Unfortunately, most courier services of the type described above only guarantee delivery by 10:30 a.m., which may raise timing issues in the laboratory. Some programs have been successful in making special arrangements for earlier delivery. Similar considerations may be available with the local mail service. It may be possible, for example, to routinely obtain mail before working hours, if this is not currently being done, in order to facilitate laboratory check-in earlier in the day, which can lead to earlier start times for the various analyses. Courier contracts should also include back-up arrangements in case of emergency loss of service, as for example, with a worker strike.

The Review Team noted that there might be other ways to improve turnaround times besides quicker transport. For example, the laboratory does not currently operate in any fashion on weekends. Thus, some analyses that would routinely be completed and reported on the day following check-in and testing are delayed for 2 days, when part or all of the procedure could be completed over the weekend. A routine 6- or 7-day workweek might also be considered along with shift work. If initial testing for some conditions is considered more important than the follow-up test (e.g. galactosemia and CAH), it might be possible to split the initial tests from the follow-up tests and begin one a day earlier. The Team also noted that reporting of some assays is delayed an extra day for no apparent reason other than the mechanics of reaccessing specimens for second level analyses. Consideration of other ways to reaccess specimens with less confusion might result in ways to perform all testing with a similar start time rather than delaying some for a day, as is currently the procedure with hemoglobinopathies.

The Review Team also noted that the more time sensitive tests, galactosemia and CAH, have preliminary results available for newborns on Fridays or the day before a holiday, but these are not currently reported. Because of the necessity for rapid treatment of these conditions in order to realize maximum benefit from screening and to prevent unnecessary deaths, the Team recommends that preliminary results be reported before a weekend or holiday in order to alert the healthcare provider and parents of the possibility of a serious condition. While this procedure may occasionally result in unnecessary 'alarm' when the final results prove to be 'normal,' failure to alert the proper authorities with a preliminary result could result in an unnecessary medical emergency that might have been prevented if someone were aware of preliminary screening results. This protocol will also be important for some of the conditions detected in an expanded screening program utilizing MS/MS techniques.

3.0.0 Issues from the Texas Medical Association

The following issues and questions were submitted to the Review Team as issues raised by the Texas Medical Association. Many of these questions require responses directly from the TNSP, and in those cases the Review Team provides comments as to how the answers to these questions might be obtained.

3.1.0 Using the ASTHO 8/04 issue brief as a base, how does Texas currently stand in regards to the 5 main components of an effective NBS program, i.e., screening, follow-up, diagnosis, treatment and management, and evaluation? What are the implications for each of the 5 components when Texas expands newborn testing?

The TNSP is a part of the overall newborn screening system described in the ASTHO brief. It has the responsibility to administer and coordinate all of the other components including education, which may be envisioned as either a sixth component of the screening system or a part of each of the five components listed above.

Screening - Information provided to the Review Team as part of the review process tends to indicate that a formal education plan for healthcare professionals interacting with the TNSP is not present. While some professional education is available through the practitioner manual, the website and various other program activities (e.g. newsletters) the lack of a methodical was of providing, evaluating and improving professional education may be contributing to some of the deficiencies implied in questions from the DSHS regarding ways to improve submitter compliance with demographic data, and ways to improve compliance of physicians in reporting case findings. While unsatisfactory specimens are currently not a problem, periodic submitter education about proper collection procedures, including providing accurate and complete demographic information, will help keep the unsatisfactory specimen rate low. The educational information contained on the program's website is exemplary and there are significant educational materials available of good quality. Distribution and usage of the materials appears to be left to the discretion of submitters and prenatal care providers, with limited personal interactions from the program, perhaps due to a lack of staffing resources. Education has a direct bearing on whether or not the program has an effective and efficient screening component.

Likewise, there appears to be no accurate information available regarding the numbers of newborns actually receiving either first or second screens as previously noted in Section 2.6.0. This information is critical to measuring compliance with the screening process, and the newly purchased computer system does not seem to be designed to provide efficient linkage information that might indicate the percentage of newborns receiving their mandatory second screen, nor does it seem to be part of any integration efforts with birth registration in order to ascertain compliance with the initial screen. Information provided by DSHS to the TMA (see Section 3.6.0 for actual data) indicated that only about 95% of newborns were thought to have obtained at least one newborn screen in 2004, which means that some 20,000 newborns may not have received a screening test. Given an overall expected incidence of the combined conditions included in the Texas screening panel of about 1:1200, this might mean that some 16 newborns had conditions that were not detected by the program. Although the Review Team did

not request specificr information about concerted educational or public relations activities that might improve the screening coverage rate, the Review Team did not identify any such ongoing activities (e.g. targeted education of midwives, targeted activities in border communities encouraging return for second testing on babies that might have been born in Texas and returned shortly after birth to Mexico, etc.). The possible impact of third party reimbursement problems on screening, particularly second screens at physicians' offices, was noted as another issue that might be negatively impacting the screening process. There were no indications of program activities that might address reimbursement issuesed, nor was there any data to indicate whether this was a real or perceived problem.

Follow-up - As noted in other parts of this report, the Review Team found that, while there are no known cases of late diagnosis that have caused any medical consequences to a newborn, members of the healthcare provider community that met with Team members identified a number of deficiencies in the way the follow-up system is working. In particular, there were several instances noted where follow-up results were faxed without accompanying telephone information and the subspecialty centers appear to be expected to assist with follow-up activities, despite no formal mechanism for reimbursement or accountability. It is particularly noteworthy that the respective budgets discussed by laboratory and follow-up/administration/education showed a difference of approximately 20:1 (laboratory: follow-up). All Review Team members noted that this large difference was significant and reflected the apparent follow-up difficulties noted. Based on their own experiences, Review Team noted that it is not uncommon for the ratio of funds required for laboratory and follow-up activities to be more evenly matched, as high as 1:1 in some programs. A large amount of staffing turnover was also noted in the follow-up program, and this led to the suggestion that recruitment/retention might need to be more formally addressed in program planning activities. The Review Team also noted that the follow-up protocols themselves seemed sound with appropriate documentation of actions apparent. On the other hand, program compliance with the registry requirement in the newborn screening Program Rules appeared to be relaxed, with little or no formal long-term outcome data being systematically collected.

Diagnosis - The TNSP is fortunate to have sufficient subspecialty consultants available to the program, so that appropriate diagnoses should not be a problem. There appear to be significant diagnostic service delivery issues in the Rio Grande Valley and in West Texas where few or no subspecialists reside, but the program is aware of this, as are the subspecialists, and there are mechanisms in place to provide subspecialty services wherever needed. The TNSP noted that there is a problem in obtaining diagnostic information from primary care providers, and this is an issue that can probably best be dealt with through education and better professional awareness. The lack of formal arrangements, including financial reimbursements and information accountability, to and from the subspecialty centers seems to be a problem that is causing unrest among the consultant community. Because of specialty consultants no longer receive direct patient referrals from TNSP, there is a perception that not all affected infants are receiving appropriate referral and care. This weakness in the diagnostic follow-up system is an issue that needs to be addressed in any financial planning, and has particular relevance to any fees collected for comprehensive program financing. The Team was not provided

information on the costs involved at the subspecialty centers for linking presumptive positive patients to diagnostic activities and this is important accounting data that is needed if the program is to address this concern. Most Review Team members noted that within their newborn screening system, contracts with subspecialty centers were providing necessary funds to cover these services and that the programs in turn receive timely and accurate information about various program outcome measure as part of the contracts. In one program, a surcharge was added with the addition of new tests to accommodate some of the additional follow-up services required.

Management - While it is likely that the management of patients identified by the screening program is continuous and appropriate, there is little data available to evaluate this aspect of the program. Because of the relatively large number of subspecialists available within the State, diagnostic and medical management appear to be available to all identified patients. One of the ideas of long-term case management and the case registry is the ability to collect program evaluation data on the long-term outcome of individuals identified by screening. These data provide a mechanism for determining the effectiveness of newborn screening and should provide information on which to base program changes. Long-term follow-up data is a critical need for most newborn screening systems. Without outcome data, it is impossible to accurately assess the program performance. These data can be accumulated through annual inquiries either to the treating physician, to the consulting specialist (if one exists), or to the parent. Since long-term outcome follow-up will invariably require funding if it is to be done correctly, it should be a part of any financing discussions.

Also included in management issues are considerations about record retention and specimen use, retention, and storage. Specimens and information collected for newborn screening are obtained for the specific purpose of screening for specified disorders. Their use for other than the purpose for which they were obtained presents both a legal and an ethical question. Specimen storage beyond six months implies a use for some purpose other than analytical validation of the screening tests since many of the analytes are not stable beyond that time. Because of the potential to provide patient DNA for future testing options, there are legal questions as to who truly owns the blood on the filter paper – the baby, the parents, or the State? Some programs are attempting to address the research potential offered by specimens though notification on the newborn specimen collection card. In some cases, permission to save and use the specimens is requested. If specimen cards are retained, their retention should comply with CLSI/NCCLS LA4-A4. Testing records, however, pose a different problem since they relate to genetic information and since a newborn with a late diagnosed condition may wish to pursue legal remedies after he(she) becomes of age. Many programs store testing records for longer periods, commonly the age of majority plus the statute of limitations for filing a lawsuit on behalf of the newborn after he(she) reaches majority (approximately 21 years). Both of these issues should be discussed with legal counsel and the advisory committee, and a written protocol established that satisfies concerns about ethical and legal issues.

Evaluation - There appear to be minimal activities within the program aimed at program evaluation. While there are procedures in place within the laboratory to actively

ensure the quality of testing, the quality of follow-up appears to be evaluated only passively, that is, by listening for complaints. For example, there is no mechanism for checking to see how the telephone process occurs and there appears to be no mechanism for validating that faxed results were received in a timely way, if the feedback from the healthcare providers interviewed is correct. Support functions such as accounting, data entry, and computer system functions should also be part of an evaluation plan. While records of cases detected are maintained and reported to the national newborn screening database, other requested information concerning length of time to diagnosis, cases lost to follow-up, etc. are not routinely reported. Some of these issues may relate to personnel turnover, but their persistence over time raises questions about a valid evaluation system to ensure that the program is accomplishing its goals. Questions related to data reports seemed to indicate that the internal computer system being used might not adequately meet the evaluation demands of the program or the computer system administrator is not cognizant of the need for timely evaluation reports. Some programs provide quarterly newsletters to the public with updated program evaluation information and some publish annual reports of this information. Both are excellent ways to update the community on the successes and areas of change within the program. Examples of the Iowa newsletter and the Nebraska annual report are given in the Appendix. Examples are also given of quality reviews of specimen submitters being used for evaluation of the screening process in Washington, also an evaluation activity not presently part of the TNSP.

3.2.0 How will the situation be resolved where physicians are inadequately reimbursed or not reimbursed at all for performing the second screen?

The Review Team noted that this concern was present during its meetings with the healthcare community. Since no meetings were held with representatives of the insurance community, it is not possible to answer this question directly. There appear to be two issues that must be dealt with before this issue will be solved. The first is whether or not there is an alternative to having physicians pay for the second screen. Based on the experiences and knowledge of the Review Team members, it appears that the TNSP is one of two programs requiring two screens that does not charge a single fee up front to cover the cost of both screens (the other program is Arizona). Additionally, it does not appear that this up front payment for the two screens in other programs has caused a significant problem in program financing even though this means that the financing is provided through charges to the birthing facility. The first suggestion, therefore, is that the program considers a single fee for the initial screening test that will cover the cost of repeat testing. Given the concern of at least one birthing facility that any additional newborn screening cost would have to be absorbed by the birthing facility without increase in third party reimbursement, representatives of birthing facilities and insurers should be actively engaged in the discussions described below.

It does not appear that the TNSP, TMA, or any other group has not investigated or addressed claims of inadequate or lack of third party reimbursement for newborn screening services. In view of the concerns raised over reimbursement issues, it is suggested that a more formal investigation of these claims be made. The quickest and easiest way may be through a survey of TMA members. Any survey should include a

request for specific information about the nature of the problem. Anecdotal information is not sufficient. If there is sufficient documentation of difficulties, then the root problems should be addressed. This may mean a collaborative effort involving DSHS, third party payers, the TDI and others. Because financing issues have a direct impact on the TNSP, DSHS should work to resolve any third party reimbursement issues. Adequate payment should include sufficient reimbursement for administrative costs at the physician or hospital collection site. In order to address this situation in the future, it is suggested that representatives of the insurance industry be invited to participate in any plans for program expansion. Additionally, representatives of the Medicaid Program should also be included since nearly half of the births in Texas qualify for Medicaid support. The Review Team considers appropriate reimbursement to be part of the overall newborn screening system and, therefore, encourages active involvement of payers in any discussions concerning system financing.

3.3.0 What is the cost/benefit analysis for Texas to continue doing two screens?

Very few cost benefit analyses exist in newborn screening. In Texas, there have been several documented reports of cases detected from second screening results when initial screening results were within normal limits. In particular see reports by Levine and Therrell (*Pediatrics* 1986;78:375-6.), Gonzalez (In: *Early Hospital Discharge: Impact on Newborn Screening, Proceedings of a conference held in Washington, D.C., March 31-April 1, 1995*, pp.155-66.), and Therrell, et.al. (*Pediatrics*1998;101:583-90.) concerning both CH and CAH. Other programs have also experienced cases detected on a second specimen when the first was reported as normal - most with endocrinopathies although occasional metabolic conditions have also been reported. More recently, the Oregon program has reported MS/MS detectable conditions detected on second screening following an initial normal finding. A cost study of CAH case detection on second screening in Texas has also been reported in the peer-reviewed literature (Brosnan, et.al., *Public Health Reports* 1998;13:170-8.). Nonetheless, mandated second testing on all newborns remains controversial. Currently there are 8 programs that require universal second testing, several that suggest it to the extent that over 80% compliance is reported, and all programs require a second test if the first is taken "too early" (although the definition of "too early" varies). Eliminating the second test would definitely result in program cost savings, but the potential litigations that might result when conditions are diagnosed late after an initial negative newborn screening test may negate any savings to the program and, more importantly, may result in tragic medical consequences to some who might have been detected on a specimen obtained at 1-2 weeks of age.

3.4.0 What is the status of parental consent in the current process?

In the current program, as in all other U.S. programs except three (WY, MD, DC), there is no special parental consent necessary for newborn screening. Instead parents have the option to refuse the test (refusal is not an option in at least 5 other U.S. programs). This issue was also addressed in the 2000 report of the AAP Newborn Screening Task Force as follows [see *Pediatrics* 2000;106(suppl):pp.410-411 (Executive Summary in Appendix)]:

- *Parents should receive information (on behalf of their children) about newborn screening.*
- *Prospective parents should receive information about newborn screening during the prenatal period. Pregnant women should be made aware of the process and benefits of newborn screening and their right of refusal before testing, preferably during a routine third trimester prenatal care visit.*
- *Parent knowledge should be reinforced after delivery by educational materials and discussion as needed by the infant's primary care health professional and/or knowledgeable hospital staff.*
- *Prenatal health care professionals as well as the infant's primary care health professional should be knowledgeable about their state's newborn screening program through educational efforts coordinated by the state's newborn screening program in conjunction with a newborn screening advisory body.*
- *Written documentation of consent is not required for the majority of newborn screening tests, for example, those tests of proven validity and utility.*
- *Parents should always be informed of testing and have the opportunity to refuse testing.*
- *If after discussions about newborn screening with health professionals, parents refuse to have their newborn tested, this refusal should be documented in writing and honored.*
- *If a newborn screening test is investigational or in the process of being developed, the benefits or potential risks have yet to be demonstrated, and identifiers are not removed from the specimen, informed consent should be obtained from parents and documented.*

Ultimately, it is essential that parents are adequately educated to make informed decisions on behalf of their newborn**.**

3.5.0 What is the difference between the annual report data posted on the DSHS web site and the data for reporting on the Federal Performance Measure of Title V of the Social Security Act for the Fiscal Year? Why are the reports different? What are the reasons for the 3-4 year delay in getting data on the web site?

The only difference that might exist concerns the time period of the defined report. That is, the Title V "year" may be defined differently that the TNSP "year." Nevertheless, the data should be quite close and over time the differences in overall program statistics should disappear. In the past a delay in reporting national data was partially due to the delay in reporting vital statistics data regarding births, which may be 6 months to a year late. Delays in reporting program data would have to be explained by the program, but are usually related to staffing and time constraints in assimilating the appropriate data. Currently there is a new On Line national newborn screening data system that allows cases to be reported at the time they are diagnosed. Automated downloading is possible. In this system, numerous reports are available such that the program can generate comparative evaluations based on specific time periods in order to

consider program improvements. The TNSP, as with other programs, is encouraged to link its newborn screening data to other child health programs in order to better evaluate program coverage and to provide more efficient data sharing with the medical home.

3.6.0 "Number and Percentage of Newborns and Others Screened, Confirmed, and Treated – FY 03" report implies no patient was lost to follow-up. DSHS does an additional report to the DSHS metabolic consultants with the number of newborns lost to follow-up. Should these reports be combined? Who exactly are the "Others" as stated in the title of the report?

This question seems to refer to the following table taken from a DSHS written response by Dr. Drummond-Borg in May 2004 in response to inquiries about program issues from the TMA.

NUMBER AND PERCENTAGE OF NEWBORNS AND OTHERS SCREENED, CONFIRMED AND TREATED
Sect. 506(a)(2)(B)(iii)

Total Births by Occurrence: 381,088* **Reporting Year: FY 03 (Sept 2002 thru Aug 2003**

Types of Screening Tests	(A) Receiving at least one Screen No.	%	(B) Presumptive Positive Screens	(C) Confirmed Cases	(D) Needing Treatment that Received Treatment No.	%
Phenylketonuria (Classical)	364,212	95.57	344	7	7	100
Congenital Hypothyroidism (Primary)	364,212	95.57	7087	196	196	100
Galactosemia (Classical)	364,212	95.57	328	7	7	100
Sickle Cell Disease	364,212	95.57	202	202	202	100
Congenital Adrenal Hyperplasia (Classical)	364,212	95.57	1924	14	14	100

* The 2003 occurrence births number is a preliminary number and is subject to change.

The Review Team can only comment on the report it is aware of, which is the one above. Information shared with the metabolic consultants was not available. If there was a different report made to the program's consultants showing cases with "Presumptive Positive Screens" that were "Lost to Follow-up," it would seem that the two reports should be combined into a more comprehensive program report so that the full extent of the program can be evaluated. In this way continuous program improvements can be implemented and their results monitored. This same reporting deficiency has been noted in the National Data Report for the TNSP, that is, the numbers "Lost to Follow-up" are usually not reported. It is also interesting to note in the report above that there is a number reported for newborns receiving at least one screen. Information given to the Review Team indicated that a definitive number was not available since there is not matching between specimens received and birth certificate information. If the number given in the table is an approximation, then a footnote explaining the way the approximation was obtained seems appropriate. Even more interesting is the fact that approximately 17,000 newborns appear to have not been screened in 2003.

3.7.0 Re: newborns lost to follow-up, what are the DSHS standards for "all reasonable steps to contact the physician and locate the family. DSHS has no evidence of poor outcome or diagnosis in these newborns"?

It is both necessary and acceptable for a newborn screening program to include a protocol by which it classifies a newborn as "Lost to Follow-up." It is probable in a program as large as the Texas program that occasionally it will not be possible to locate a newborn needing additional testing to resolve a newborn screening test result. In such cases, a written protocol outlining the procedures to be followed should exist and these should be followed. The written protocols in the follow-up operations manual delineate such processes. While it is true that there appears to be no evidence of poor outcome or diagnosis in newborns who may be lost to follow-up, there also appears to be no evidence to the contrary.

3.8.0 What are the protocols used for confirmatory testing on presumptive positive screen patients? What is the clinical evidence base for such protocols? Which newborns have a second newborn screen rather than confirmatory testing? Why does DSHS, for presumptive positive patients, only separate first screens from second screens for hypothyroid data and not other disorders?

The Review Team did not investigate the individual confirmatory testing protocols that have been developed by the TNSP for each type of presumptive positive test result. It is assumed that the subspecialty consultants have had sufficient control of the confirmatory testing process over the years, so that the TNSP does not make specific confirmatory testing recommendations apart from those approved by subspecialty consultants. General confirmatory testing algorithms have been published [*J Pediatrics* 2000;137 (suppl 4);S1-S47] and contain some of the general recommendations for confirmatory follow-up found in the TNSP Practitioner Manual. The reason for separating first screens from second screens for congenital hypothyroid testing resides in the fact that the primary screen, thyroxin, is expected to be lower in older newborns. The approximate breakpoint is at about 7 days of age. Since a low thyroxin is an indication of possible congenital hypothyroidism, the older newborns are usually batched together so that their results will be judged against a different (higher) normal range. Similarly, newborns that were born prematurely may also show elevated 17-OHP results for CAH, and they are usually compared to a different expected range adjusted on the basis of birth weight.

3.9.0 DSHS NBS Program has "the disorder occurs with significant frequency" as the number one criterion for inclusion in the testing program; yet DSHS does not have a formal definition of "significant frequency". Please clarify.

The frequency with which a condition occurs is only one of several variables traditionally considered in deciding on which conditions should be included in the testing program. At least one newborn screening program, Wisconsin, defined significant frequency at one point as 1:100,000 (*Arch Pediatr Adolesc Med* 1997;151:561-4.) but subsequently revised this definition when MS/MS testing was implemented. Since many

conditions can be simultaneously detected using MS/MS testing, some have advocated for use of the cumulative frequency of all conditions detectable in deciding on program expansion. Recently, the ACMG reported its investigation into a decision-making algorithm that might be useful to all U.S. programs and included a recommendation of conditions to be included in a core panel of tests using national data and more comprehensive criteria reflecting the changes in technology and science in recent years. This report, *Newborn Screening: Toward a Uniform Screening Panel and System*, is extensive (over 350 pages) and includes detailed discussions of its research process. Ultimately, the recommendation is for programs to routinely screen for 29 listed conditions that give rise to an additional 26 conditions as part of the screening process and differential diagnosis. Using the counting procedure present in this report, Texas includes eight conditions (including hearing screening) in its current mandate. The majority of the other conditions would be part of the screening panel available if MS/MS technology is adopted, with biotinidase deficiency and cystic fibrosis remaining to be included by other screening techniques. The Review Team recommends using a formal decision making process such as that outlined in the ACMG report for addressing additions to the TNSP. Complete adoption of the recommended screening panel is also recommended as early as the program can feel confident of accomplishing its mission of effectively detecting and providing diagnostic and medical management to all newborns identified with the conditions recommended.

3.10.0 Is the calculation of positive predictive value used in Texas a commonly used calculation elsewhere?

Calculation of positive predictive value is performed according to a standard mathematical formula. In newborn screening, the positive predictive value for a particular procedure may not reflect the predictive value for the testing procedure itself. Cutoff values used for various tests are often determined by factors other than the statistics of the testing procedure itself. For example, programs may determine that it is reasonable to follow-up on a certain percentage of children with results indicating the possibility of a particular condition in order to decrease the risk of a false negative test result. While statistically a certain testing procedure may give a relatively tight standard deviation of analytical test results, the biochemistry of the condition may dictate a different cutoff (or cutoffs) if the statistical cutoff causes too many cases to be overlooked. Thus, programs may decide to use percentages of babies outside of an expected range to define their follow-up caseload rather than strict mathematical formulation related to means and standard deviations. For this reason, use of positive predictive values sometimes defines the system being used rather than the strict analytical procedure in use and must be applied carefully. Nonetheless, the nature of screening is such that relatively low positive predictive values are usually the case and improvements in screening techniques through such strategies as second-tier testing are constantly under evaluation to improve the positive predictive values of the screening procedure.

3.11.0 DSHS does not match all first to second screens and therefore "cannot exactly determine the number of first and second specimens". How is data/reporting validated? How do other states doing 2 screens handle this issue?

Overall validation of the number of newborns receiving newborn screening in Texas is not currently possible because there is no matching between birth certificates and newborn screens. A match of Medicaid patients is performed according to a complex algorithm and occurs several months after the newborn screen occurs due to time delays in ascertaining Medicaid eligibility. The results of these matches were not given to the Review Team but presumably would give some indication as to the number of Medicaid patients receiving both initial and follow-up tests. Some approximations for overall coverage could likely be made from these numbers. Alternatively, the program must base its approximations on total number of specimens received for testing and the indication on the form as to whether the specimen was provided for initial or second testing. As noted in Section 2.6.0, other states performing two screens have developed mechanisms for matching or linking the two screens so as to better evaluate screening coverage and these may be applicable to the TNSP. Innovative computerized linkages are also possible and are partially employed in the TNSP as a means of linking initial presumptive positive samples with specimens submitted for second tests.

3.12.0 Are the three consultant committees which have replaced the TDH Genetics Advisory Committee the most effective/efficient means for clinical oversight of the NBS Program? To alleviate possible conflict-of-interest issues, should an objective, external advisory group have a role in addition to an internal advisory group? How broad a committee of clinicians would an optimal clinical advisory group have for a state as large and diverse as Texas?

The Review Team understands that program expansion led to the development of three separate advisory groups with the Genetics Advisory Committee eventually assuming the role of the Metabolic Advisory Committee. An extensive discussion of the Review Team's view of advisory committees (both external and internal) and their structure is given in Section 2.2.0. The Review Team endorses the concept of a broad based multi-disciplinary Newborn Screening or Genetics Advisory Committee with subcommittees for specific considerations such as the three current committees. In the broader scheme of issues, newborn screening is actually a component of ongoing genetics activities in the public health system and in some jurisdictions, the newborn screening program is advised by a subcommittee of a larger Genetics Advisory Committee. The Review Team also feels that program decisions affecting multiple stakeholders should have involvement from those stakeholders in the decision-making process, and there are a variety of ways in which this can be accomplished.

3.13.0 What are the national norms for timeframes for screening and confirmatory testing? How do Texas timeframes compare?

Because the conditions included in screening panels require immediate attention if they are to be appropriately managed for maximum benefit to the patient, newborn screening programs work to have all patients diagnosed and managed within the first month of life - preferably within three weeks or earlier. The exceptions are conditions that can cause death more quickly such as CAH, galactosemia, and certain metabolic conditions not currently included in the Texas screening panel. Guidance developed by CORN [*J Pediatrics* 2000;137 (suppl 4):S3-S4 and again on S11] recommended that, "*A*

laboratory report on every infant should be sent to the infant's PHCP and/or birthing facility within 7 days of receipt of the specimen." It further noted that, "*The rapid follow-up of the infant with a screen-positive test result is the highest priority and the primary responsibility of the follow-up component of the system*." The Texas program appears to generally meet the technical recommendations of reporting out results within 7 days (which includes weekends and holidays), but there were questions about the rapidity with which the more time critical results for galactosemia and CAH presumptive positives are reported given the lack of a working schedule beyond the 5-day workweek and a routine holiday working schedule (both within the laboratory and with follow-up support). The lack of notification of preliminary results when they are available prior to the weekend or an extended holiday was also noted as a problem area. Other suggestions related to improved turnaround times are given in Section 2.10.0. Comparative data for times to diagnosis for all programs may be found online at http://genes-r-us.uthscsa.edu. A review of the Texas data reported indicated that the times from birth to diagnosis for all conditions reported is reported as "not known." Thus, from the data available, it would appear that the TNSP may not monitor the final diagnostic and treatment times, and may, therefore, be unable to evaluate how quickly patients are diagnosed and/or treated, which the Review Team considers to be a critical outcome measure.

3.14.0 Are the protocols used by Case Management re: timeframes, follow-up activities, closing cases, etc., effective and efficient? How is the effectiveness of the Case Management function determined?

While the protocols available from the Case Management Section appear to reflect appropriate procedures for ensuring rapid, efficient, and effective follow-up, the lack of data available regarding actual times to diagnosis and treatment make these activities difficult to evaluate. It may be assumed that case detection is proceeding in a timely manner based on the lack of complaints from patients or physicians regarding delayed diagnosis, but this is not the preferred way to evaluate the follow-up system. The Review Team encountered complaints about slowness in contacting subspecialty centers versus historical experiences, which may indicate reporting changes due to staffing shortages, but collection of program evaluation data at the time of diagnosis and periodically surveying the subspecialty centers would be a better method for obtaining information about case management effectiveness.

Program evaluation was addressed by the AAP Newborn Screening Task Force [*Pediatrics* 2000; 106 (suppl 2): p. 413] in the following way:

> *Ideally, the information obtained by a newborn screening program would allow the description of:*
> - *The number and percent of children*
> - *adequately screened,*
> - *with appropriate follow-up,*
> - *with false-positive and false-negative results,*
> - *with specific diagnoses, and*
> - *with appropriate care.*
> - *The time between the newborn screen and the initiation of treatment.*

- *The long-term improvement in health status occurring as a result of screening, follow-up, diagnosis, and treatment.*
- *The number of children diagnosed with a condition missed by the screening programs and, where possible, an assessment of the reasons they were missed.*
- *The number and percentage of children lost to follow-up.*
- *Defining reporting procedures (e.g., what reports will be made, who will receive them).*
- *Ensuring commitment to maintaining systems.*
- *Ensuring that procedures for maintaining, transmitting, analyzing, and disseminating data conform to ethical guidelines and legal standards.*

3.15.0 How can an ongoing DSHS role as a liaison with TDI (re: nbs reimbursement issues from health plans and coverage issues for formula/treatment) be implemented? Inconsistent/inadequate reimbursement has resulted in some physicians declining doing any screenings and sending the patients to the hospitals for such screening and in difficulty obtaining treatment services for patients.

Little information was provided to the Review Team regarding interactions between DSHS and the TDI. Reports of inconsistent/inadequate reimbursement were obtained anecdotally but documented problems did not appear to have been systematically collected or assessed. In order to determine actions needed, it would appear that some data on the problems would be helpful. This could be simply done by TMA querying its members in a more structured way in order to ascertain documentation of actual problems (see Section 2.1.0). Once problems are identified, then DSHS could partner with TDI to resolve the issues to the benefit of the program. Some payment difficulties may exist because there are not standard insurance payment codes across the country. Some insurers who have developed their own insurance codes for the screening process - which includes the cost of the collection card plus office expenses - as a remedy. Once there is a more standardized screening system across the country, then it is more likely that the AMA will adopt a newborn screening payment code, but this is not expected any time soon.

3.16.0 Are there any hospitals in Texas that are refusing to do newborn screening, first and/or second screen?

The Review Team had only limited experiences in obtaining information from hospitals and no information was given that would indicate that hospitals are refusing to perform newborn screening. However, no systematic study was provided that would substantiate the presumption that all are participating. A periodic study of submitting facilities should answer this question.

3.17.0.0 What efforts are done to ensure newborns who are delivered by lay midwives (not certified nurse midwives) are screened?

Lay midwives were not a focus of the newborn screening review. Certified nurse midwives appear to be aware of newborn screening through their certification

requirements. A survey of the newborn screening database might provide information about lay midwife submitters who are not certified, provided they did not submit specimens through another mechanism (such as a health clinic) and that the identification number of the submitters is captured and includes a mechanism for identifying the category of submitter. The Review Team is aware that a system was in place in the TNSP at one time that identified the type of submitter (physician, hospital, clinic, etc.) by virtue of an identifying code number within the submitter's broader identification number. Such systems do not appear to be in place currently, and there is no information on the number of babies delivered by midwives who receive screening.

Plan for expansion of testing

3.18.0 Are there adequate physician/metabolic experts/other clinician resources in the state, especially in underserved areas of the state, for appropriate follow-up? Does the follow-up need to be centralized? Is there adequate funding for treatment of disease detected where treatment is proven effective? Is additional State funding needed to address this issue? Should expanded testing be implemented if adequate funding for treatment is not available? Also, TMA recommends that interaction with physicians and hospitals located in border cities be included in the state assessment.

In its brief visits to facilities in Austin, Dallas, Houston, and San Antonio, the Review Team attempted to obtain information to address questions about program expansion, particularly for conditions detectable by MS/MS, which are currently limited to metabolic conditions. It appears that there are sufficient subspecialty resources available to handle the projected caseload, particularly in Houston and Dallas. To a large extent, the follow-up of metabolic patients appears to be concentrated in a few places at present and this is expected to continue. There also appears to be adequate biochemical genetics laboratories available to assist with the diagnostic workups. Particular areas that appear to be problematic relative to diagnostic services include the Rio Grande Valley and West Texas (both of which have border issues complicating follow-up considerations). The shortage of subspecialty services in these areas is apparently well known and steps have been and are being taken to provide the needed coverage in these areas. Financing diagnostic and management services appears to be a major concern of the subspecialists and reimbursement issues have been addressed in other areas of this report. As noted previously, the Review Team was surprised to see the large budget differences between the laboratory and follow-up parts of the TNSP, and based on experiences in the programs of Review Team members and in other program reviews, considers TNSP follow-up activities to be severely under funded given the information available. Funding for treatment was not cited as an issue. Rather, funding for support follow-up activities required at the clinics (linking patients to services, providing basic information about the conditions identified, tracking patients, and obtaining test results, etc.) were the unrecognized and "unfunded mandates" at issue.

The issues raised in this question are important considerations for the program. They are perhaps best handled by deliberations between program advisors (advisory committees) or a task force of stakeholders, including parents, who are aware of the local

difficulties, resources, and opportunities for problem solving. Parent advocates point to the fact that babies with these conditions will be born with or without the screening program and that newborn screening presents a proven way to decrease the morbidity and mortality that results. Any expansion must be addressed in a logical, sensible and cooperative way if the screening program is to be effective, and this will take a team effort of all of the system stakeholders. The Review Team questions the extent of the "team effort" to date, particularly since questions about major planning efforts during the last two years (since the Legislature failed to act on expansion) were answered by "none."

3.19.0 What are the anticipated false positive rates and call (re-sampling) rates?

Recall rates and false positive rates vary somewhat from program to program based on the experience and training of those who are tasked with implementing screening and result interpretation. General experiences point to an expected recall of approximately 0.5% of patients screened. This rate also varies with prevalence of the conditions within the population and includes PKU, which is currently part of the TNSP. This number may be half of the follow-up currently required for either CH or CAH, both of which are approximately 1% in Texas. Thus it would be anticipated that 1500-2000 newborns would require follow-up services annually. This number should not be significantly impacted by the fact that two screens are required, since theoretically the same newborns recalled on the first test should comprise the majority of newborns recalled by the second test.

3.20.0 What primary care education program is necessary before and during implementation of expanded screening as well as on an ongoing basis?

Education of primary care practitioners concerning the conditions included in expanded screening will be an important part of any expansion. The ACMG is currently completing ACT Sheets for all conditions in its recommended panel of tests so that model information will be available for newborn screening programs to use when reporting results to the primary care practitioner. These "just in time" information sheets are designed to contain only the essential details necessary for immediate follow-up actions on the part of the clinician. Further FACT Sheets with more detailed information about the conditions are also under development and many programs already have such resources. In particular, the Massachusetts, Oregon and California programs have extensive information available. Because some of the medical management issues may be considered differently in different subspecialty settings, any information shared with the physician community should first be reviewed and agreed to by the subspecialty consultants in Texas. A HRSA-funded project at the LSU Health Science Center in Shreveport is also preparing model primary educational pamphlets for parents and for healthcare providers. These materials have been prepared utilizing focus groups of parents and physicians and are in the final preparatory phase prior to dissemination. Some programs have also utilized other educational opportunities to assist in educational activities including presentations at professional meetings, publication of informational articles in the local medical journal, information on program websites, newsletters, webcasts, and videotapes.

3.21.0 What additional public/patient education steps need to be taken?

One of the major educational efforts will be preventing dissemination of misinformation. It is essential for the public, policy makers and expectant parents to have accurate information concerning any program changes. Just as important is information about other testing options if TNSP cannot provide expanded testing. A recent (July 2004) correspondence from HRSA encouraged all state health departments to inform parents of the availability of other newborn screening tests outside of program mandates if such tests were routinely available. At the time of this review, the TNSP was considering whether and how this information might be added to its program brochure.

3.22.0 There is concern regarding issues such as the impact on families of false positives. How will that be addressed in the plan for expanding NBS?

The nature of newborn screening programs is to identify newborns at increased risk for certain congenital conditions that may not be apparent at or near birth and, therefore, may not be easily detected. As with any screening program, newborn screening tests are designed to identify newborns with no or minimal false negative results (a newborn who may have the condition in question, but whose test results are interpreted as normal), and as few false positive results (a newborn who does not have the condition in question, but whose screening results are identified for further follow-up) as possible. While false positive results can have an impact on the parents who must deal with the repeat testing and concern about whether their newborn might be affected, newborn screening programs work hard to minimize negative impacts by continually trying to improve the testing procedures and by working to provide follow-up information in a sensitive way. One of the problems identified in some programs arises in the manner in which information is transmitted, particularly if the transmitter is not familiar with the conditions in question or does not understand the possibility for false positive testing results. These issues will be important points to address in any program expansion. While the MS/MS technology is much more specific and sensitive than traditional screening methods, there will still be anomalous results that will need to be followed. Elimination of false negative results is a more significant issue, since such results can result in more severe medical consequences.

3.23.0 What does the public think about expanding NBS?

In general the public appears to support the concept of expanded screening, particularly parent advocacy groups for rare medical conditions. Recent press activity has focused on the benefits to families from newborn screening tests, often without regard to the infrastructure that might be necessary within a state-mandated newborn screening program. There have been occasional negative comments in the press by both consumers and professionals, but the vast majority of reactions encountered by Review Team members have supported expansion. The March of Dimes has announced its intention to annual report cards on how States are complying with the ACMG recommendations for 54 conditions (including both core and secondary targets). There is

currently a public comment period on the recently released ACMG report on expanded screening, and it will be interesting to see what comments are received.

3.24.0 How will DSHS amend the criteria for the disorders to be included in the NBS Program, i.e., "an effective treatment/intervention exists", when MS/MS is implemented?

The Review Team was not provided with information about criteria used by the DSHS for deciding on conditions for inclusion in the newborn screening panel. Anecdotal information indicated that no formal decision-making process exists apart from acceptance of condition(s) by the DSHS Advisory Council as part of Program Rule changes. The historical Wilson and Jungner criteria have traditionally been used for support or arguments against certain conditions, and the recent ACMG Report includes more extensive criteria and a potential scoring system to be used in such decisions, but there is no requirement that this system be used. Experiences of Team members have shown that the terms "effective," "treatment," and "intervention" are defined differently depending on who may be arguing the point. The ACMG Report argues in favor of reporting all test findings of clinical significance from the technology being used. Further information on this argument may be found in the Report.

3.25.0 What is more cost effective and implemental in a reduced time frame: enhanced screening by DSHS or utilizing the services of The Institute for Metabolic Diseases?

Arguments exist on both sides of the issue of implementing screening at the DSHS screening laboratory or utilizing the services of another laboratory such as The Institute for Metabolic Disease, Pediatrix Screening, Inc. or another outside laboratory. It must be emphasized that laboratory issues are only part of the considerations necessary in expanding the newborn screening program. Follow-up issues are equally important.

There will indeed be a start-up period of several months required if the TNSP is given an expansion mandate. Other programs have found this period of time to extend to at least 6 months. In addition to obtaining equipment, modifying computer software, developing an educational program, and developing familiarity with the conditions, there are also issues of personnel and policy that must be addressed. On the other hand, no other laboratory appears to currently have the capacity to instantaneously analyze the number of specimens received daily by the TNSP. The Pediatrix laboratory reported currently analyzing approximately 1500 specimens daily at its facility (approximately half of the current Texas workload) and the Institute for Metabolic Disease processes even fewer specimens on a daily basis. Whether or not either of these facilities could gear up to perform the required number of tests on a significantly faster schedule than DSHS is not known since any other testing facility would also require additional equipment, space and personnel. On the other hand, both of these facilities have expertise with the conditions included in the screening panel that is not available at DSHS. If either were to be a part of the testing considerations in Texas, care would also have to be taken to implement a sufficient follow-up system to handle the anticipated workload of

cases that will require further study. Likewise, a more efficient and effective way of interfacing with subspecialty providers will likely be required.

Alternatively, it may be possible to obtain the assistance of an outside laboratory during the start-up period. For example, either or both of the facilities mentioned could be utilized as contracted testing facilities to analyze specimens by MS/MS until the TNSP is adequately prepared. A model for such a shared system of testing currently exists in Minnesota where a single blood spot is separated from the blood collection form at the State laboratory and forwarded by overnight courier to the Mayo Clinic for MS/MS testing. Follow-up issues would still need to be addressed and the laboratory service contracts would need monitoring by DSHS to ensure appropriate quality of testing for Texas newborns. Additionally, the testing contract would need to be configured to allow for transition of methods and activities to the DSHS when local laboratory services are ready. If longer-term contracts are considered, then consideration should also be given to service issues if company policies, issues or ownership, or changes in program emphasis at the contract facilities were changed such that the anticipated services were no longer available. That is, without a functioning public health laboratory available, DSHS might find itself in a difficult position relative to continuing its screening program.

3.26.0 How would single screening by The Institute for Metabolic Diseases affect false negative rates and programmatic costs?

The laboratory costs for testing half as many newborns by switching to a single MS/MS test should result in a laboratory budget roughly reduced by half, whether this occurs at DSHS or outside. Such a decision should be carefully considered, since at least one program performing required second screening is reporting preliminary data indicating that up to 10% of the total cases detected by MS/MS in that program are detected on second screens following a normal first screen. Additionally, the second screen provides a natural safety net for newborns whose first specimen was unsatisfactory or not done, and has the potential for confirming a presumptive positive finding more rapidly that might otherwise occur if there were no routine requirement for a second screen between 7-14 days. It may also be of interest that in California, where 540,000 newborns are screened annually using the single test model for all conditions in the screening panel, the fee for a single screen was recently set at $78, exclusive of screening for CF and biotinidase deficiency (similar to the screening panel under consideration in Texas).

3.27.0 What are the funding requirements in the community (not at DSHS) for case management/follow up? Is the budget request adequate for this?

The issue of adequate funding in the community has been addressed in several Sections previously. The bottom line is that the Review Team cannot answer this question and it does not appear that it can be answered by anyone at the present time. Additional and complete data are needed relative to program expectations, community participation in the process and financing requirements. This issue should be discussed in more detail in advisory committee meetings or in deliberations of a task force considering program and system issues. The Review Team was unclear about what

specific items are included in any budget requests currently under consideration. Given the information about funding that was available, it is likely that funding for follow-up services needs to be increased along with the follow-up services provided. The extent of service and funding increases should be carefully analyzed using sound accounting practices and comparisons with other programs already experienced in the expansions being considered.

3.28.0 How will the parental consent process function?

In the current system, parents are allowed the option of refusing newborn screening. The Review Team did not receive any information indicating an anticipated change in the current practice. In some programs where expanded testing is given as an option (i.e. not required), then a consent process may be present, but this process would not be usual in cases where the testing is mandated. See Section 3.4.0 for information from the AAP Task Force Report on consent issues. The Missouri newborn screening program added a consent option to its specimen collection to allow data sharing between health programs. If consent for testing or for data sharing is a concern of the TNSP, then contact should be made with another states involved in such a practice to obtain additional information and suggestions.

3.29.0.0 What data will be stored? Where? What are the ethics standards applied to the data repository?

Information about data storage was not shared with the Review Team. Data storage is an important issue since confidential genetic information is involved, and care should be taken to ensure privacy of all information related to newborn screening. If electronic records are retained, they must be maintained in a way that ensures their validity, accessibility, and confidentiality. Statutory requirements regarding electronic record retention should be followed and, all record interactions should be appropriately documented. Similarly, where paper records are retained, they should adhere to State record keeping requirements.

Generally, records related to testing of newborn screening specimens are retained for a period sufficient to meet the legal requirement associated with the newborn's right to challenge parental decisions that might have caused damage to the newborn. In this way, should parents refuse a newborn screening test that might have identified a severely debilitating condition, they may be subject to legal challenge from the newborn after (s)he reaches the age of majority. Often the statute of limitations for such legal actions is 3 years past the age of majority (which in many jurisdictions is 18 years of age), so that records are retained for 21 years. Residual specimens remaining after analysis are a special form of genetic record and should be treated with equal attention to privacy. Residual specimen issues are discussed in more detail in Section 3.1.0.

3.30.0.0 What is a realistic and reasonable time frame for annual report data to be on the DSHS web site?

Annual program data should be maintained in a manner such that periodic reports can be obtained with minimal effort and within a relatively short period of time. A new Online national reporting system supported with HRSA funds is now available through the NNSGRC. This system allows real time entry of cases as they are diagnosed and requires relatively little entry time to record the case data. The data entry process was designed so that minimal time is required and electronic downloads from local system databases are possible. The system in return allows for comprehensive data reports on individual programs or on all programs across the country. In this way, if data are entered as they are obtained, then annual program reports or other periodic reports can be available within minutes. Recommendations elsewhere in this report encourage a written annual report for wide dissemination as a public relations/information exercise.

3.31.0.0 What is the specific plan for identifying and tracking those patients with diagnoses of disorders being screened for through MS/MS but were not detected through the NBS Program?

The Review Team was not apprised of any current or future activities planned relative to active ascertainment of cases diagnosed outside of the newborn screening program. This is an issue of interest to most newborn screening programs that is not generally pursued, usually due to of a lack of personnel time. A simpler way to determine such cases might be to include them in reportable conditions required by the DSHS.

3.32.0 Re: first and second screens, what is the most optimal option for matching the screens once MS/MS is implemented?

Matching considerations are not related to MS/MS implementation and a number of options are discussed in more detail in Section 2. The simplest solution would seem to be a computer solution within the current system available. There are a number of ways in which this could be accomplished and it appears that a partial matching system, requiring some manual intervention when exact matches are not present, is already in place for linking follow-up tests to initial tests for presumptive positive specimens. Some systems perform matching processes at the time of final data entry following a preliminary data entry process that allows for specimen processing. This two-step data entry process is necessary since the matching process may slow the process, which is usually not acceptable when certain laboratory functions rely on these data. Linking records through the unique serial number of the first specimen, if it is either a part of or included in the data of the second specimen, appears to be the most effective linking method with which the Review Team is familiar.

3.33.0 The operational timeline for correction of or enhancement of state infrastructure must be established prior to determining dates for implementation of MS/MS.

This question remains to be answered by the DSHS but comments on various aspects of the question are contained in Section 3.25.0.

3.34.0 Please clarify statements made at 12/02/04 Stakeholders meeting regarding DSHS estimating $3.5 million to implement MS/MS in Texas versus these written comments from DSHS on 12/01/04: "The proposed budget for expanded newborn screening in Texas is $6 million to $12 million for initial start-up costs and an additional cost per specimen of $2 - $24."

This question cannot be addressed by the Review Team and must be addressed instead by the TNSP. The only cost estimates provided to the Review Team were consistent with the $6-12 million statement above. In view of the infrastructure issues related to follow-up/administration/education, the cost estimates should likely be revised to address these system's issues. The understanding of the Review Team is that the current fee covers only laboratory costs. Whether other program costs can be included in the fee, and whether the fee can be increased for start-up and pilot testing prior to offering routine testing services are an issue that needs to be resolved. Because of apparent conflicting legal opinions on these issues within DSHS, it is likely that statutory clarification will be necessary for complete resolution.

3.35.0 The 1/3/05 edition of the St. Louis Post-Dispatch has an article regarding Missouri implementing MS/MS with comments that start-up costs (staff, equipment, and software) were funded by a federal grant. What similar options are available to Texas?

Federal grant support is available from time to time that might apply to various parts of the newborn screening system. The Review Team was not familiar with specifics of federal funding in support of the Missouri implementation of MS/MS but was familiar with various federal grants that were obtained to support aspects of theirs and other newborn screening programs. Most notable were grants for data system integration and genetics planning activities. While the grant application process is often not simple and the funds may not be significant to a program the size of the Texas program, they are nonetheless available and many programs have used them to improve their newborn screening systems. Particular attention should be paid to grants available from HRSA and CDC, with growing interest in newborn screening coming from NIH.

<u>Specific to cystic fibrosis screening</u>

3.36.0 What cut off level should be used for the serum trypsin level? Is there some variability between states currently doing the testing?

The October 15, 2004 MMWR (Vol. 53, No. RR-13) is devoted to newborn screening for cystic fibrosis. Laboratory methods used by the screening programs are summarized in that document which is included in the appendix of this report.

3.37.0 How will patients with positive tests and their PCPs be notified? How will patients get to a facility where reliable screening can be performed, i.e., an accredited CF Center?

There are many issues that need to be addressed before cystic fibrosis screening will be ready for addition to the Texas newborn screening panel. Given the recent endorsement of newborn screening for CF by a CDC working group and the CF Foundation, the Review Team recommends that a task force or advisory committee be organized to address whether and how CF screening should be incorporated into the Texas program. Items that should be formally addressed by this group include the CF screening infrastructure, the reporting process to parents, providers, and specialists, referral of presumptive positive screening cases to a CF Foundation approved sweat chloride testing center, and data collection. Experiences and advice should be actively accumulated from other programs that have already gone through the CF screening implementation process.

3.38.0 How will the CF centers be notified of patients referred to their facilities for newborn screening?

This issue should be one of those addressed by the advisory committee considering implementation of CF newborn screening in Texas (see Section 3.37.0 above). Further details about CF newborn screening processes will soon be available in a special supplement to the Journal of Pediatrics. Experiences of other programs should prove vital to notification and referral considerations.

3.39.0 How will DSHS involve CF Center Directors across Texas, along with individuals involved in newborn screening from other states, to help formulate the methods of screening patients? Will the center at Wilford Hall Medical Center be included?

As noted in Section 3.37.0 above, an advisory group is recommended to address all of the questions related to implementation of newborn screening for cystic fibrosis. This group should include adequate representation from the CF Centers and from the Military so that questions such as this one may be appropriately answered.

TEXAS Department of State Health Services

NEWBORN SCREENING PROJECT CHARTER

Date:	April 18, 2005		
Project Name:	Newborn Screening Project		
Project Manager:	Margaret Bruch, LCSW	**Phone:** 458-7111, ext. 3045	
Laboratory Liaison:	Eldridge Hutcheson, Ph.D.	**Phone:** 458-7111, ext. 2468	
Project Sponsor(s):	Alex Hathaway, M.D., M.P.H. Assistant Commissioner Prevention & Preparedness Evelyn Delgado Assistant Commissioner Family & Community Health	**Phone:** 458-7111, ext. 7729 **Phone:** 458-7111, ext. 2114	
Project Start Date:	April 18, 2005	**Target End Date:**	Phase I: 08-01-05 Phase II: To be determined.
Team Members:	Susan Neill, Ph.D., MBA, Susan Tanksley, Ph.D., Jann Melton-Kissel, RN, MBA, Margaret Drummond-Borg, M.D., Angie Estwick, Nick Dauster		

PURPOSE:
The purpose of the project is to improve the Newborn Screening (NBS) Program at DSHS by 1) reviewing recommendations of the National Newborn Screening and Genetics Resource Center (NNSGRC), 2) developing a plan based on the recommendations, and 3) implementing the plan as approved.

SCOPE:
The scope of the project includes both laboratory testing and case management services in the NBS Program at DSHS.

Phase I:
- Develop schedule for Phase I.
- Review/analyze NNSGRC recommendations.
- Identify additional stakeholders/team members as needed.
  Identify and evaluate assumptions, constraints, and risks.
- Develop plan based on NNSGRC recommendations.
- Submit plan for approval.
- Closeout Phase I.

Phase II:
- Based on approved plan, develop schedule for Phase II.
- Implement plan.
  Closeout Phase II.

KEY STAKEHOLDERS/DEPARTMENTS:
- DSHS Laboratory
- DSHS Case Management Branch
- NNSGRC Review Team

- Other external stakeholders
  - Texas Medical Association
  - Texas Hospital Association
  - Texas Nurses Association
  - March of Dimes
  - Cystic Fibrosis Foundation
  - Texas Pediatric Society
  - Texas Family Practice Association
  - Children's Hospitals and Related Institutions of Texas (CHARIOT)
  - Children's Hospital Association of Texas
  - Texas Department of Insurance
  - Texas Health and Human Services Commission
  - Interagency Council for Genetic Services
  - Newborn screening consultant groups
  - Sickle Cell Association of Texas
  - Parents/consumers
  - Laboratories performing newborn screening
  - Genetic counselors

MAJOR DELIVERABLES/MILESTONES and HIGH-LEVEL SCHEDULE:

Phase I:

- Preliminary schedule for Phase I by April 29, 2005.
- Plan with recommendations by August 1, 2005.

Phase II: Dates to be determined upon approval of plan from Phase I.

ASSUMPTIONS, CONSTRAINTS, AND RISKS:

Assumptions:

- Public interest around newborn screening in Texas will continue to be high.
- Team will be able to complete Phase I with sufficient stakeholder input by August 1, 2005.
- Phase I will be complete before DSHS begins study on cost effectiveness of the NBS Program.
- Efforts will result in improved newborn screening in Texas.

Constraints:

- DSHS has limited internal resources because current screening and all associated activities must continue.
- DSHS is not able to provide funding for stakeholder travel.
- Stakeholders have limited time available.

Risks:

- Various newborn screening initiatives may have conflicting schedules/deliverables, making it difficult for team members and stakeholders to schedule and prioritize work.

Approved by:

Project Sponsor: Alex Hathaway, M.D., M.P.H.
Assistant Commissioner
Prevention & Preparedness

4-19-05
Date

Project Sponsor: Evelyn Delgado
Assistant Commissioner
Family & Community Health

4-20-05
Date

Appendix 3: Provider Survey

Purpose: This survey was designed to assist the Texas Department of State Health Services (DSHS) Newborn Screening Program (NBS) and health professionals in enhancing the quality and effectiveness of screening activities for Texas newborns.

Participants: The common characteristic of all participants is involvement in the screening of Texas newborns.

Please feel free to disregard those questions that do not pertain to your expertise.

Initial Management and Building Infrastructure

1. What are your credentials or title? Please check all that apply.

199, 49.4% MD
72, 17.9% RN
31, 7.7% LVN
23, 5.7% Nurse practitioner
1, 0.2% Genetic counselor
9, 2.2% Midwife
1, 0.2% Case manager
7, 1.7% Health educator
1, 0.2% Receptionist
58, 14.4% Medical technician/laboratory staff
2, 0.5% Nutritionist
3, 0.7% Social worker
28, 6.9% Other:

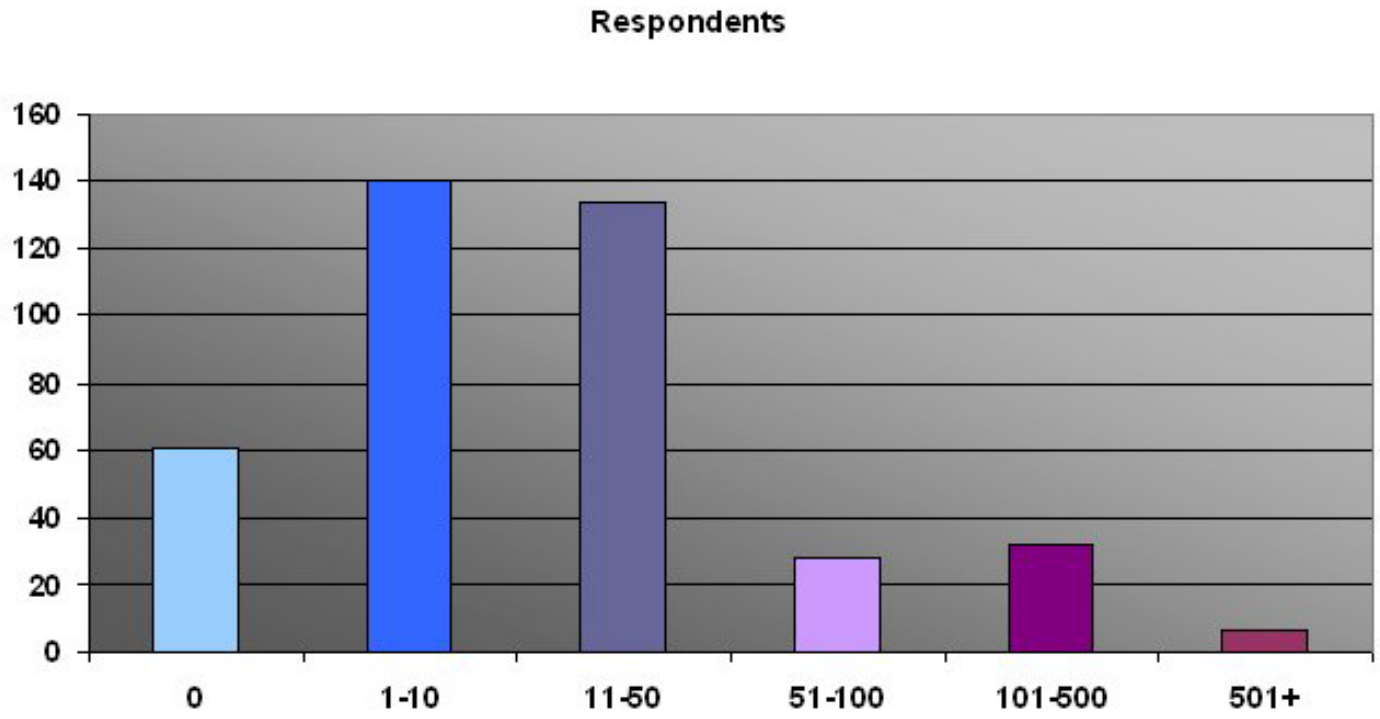
Other credentials included: DO, administrative assistant, clinical manager/nurse manager/office manager/ practice manager, director/lab director, family nurse practitioner, lab staff, medical assistant, medical technologst, nurse-midwife, phlebotomist, PA, and radiology tech

2. At what type of facility are you employed or associated with? Please check all that apply.

158, 39.2% Doctor's office
55, 13.6% Clinic
83, 20.6% Group practice
67, 16.6% Private practice
80, 19.9% Hospital
51, 12.7% Hospital NICU
7, 1.7% Birthing center
39, 9.7% Public health clinic/CHC
3, 0.7% Specialty center (metabolics, endocrine, hematology, etc.)
17, 4.2% Laboratory
11, 2.7% Other

Other facilities included: breastfeeding support center, DSHS regional office, family practice, FQHC, home birth practice, medical school & affiliated hospital, midwife private practice, newborn nursery, outpatient hospital facility, radiology practice, and rural health clinic

3. Professionally, approximately how many newborns (0-1 month old) do you see per month?



4. What is your office's ZIP code?

5. Overall, how would you describe the adequacy of the current process of submitting specimens and receiving results?

Very adequate	Adequate	Neutral	Inadequate	Very inadequate	Not applicable
76, 18.8%	221, 54.8%	44, 10.9%	19, 4.7%	8, 2.0%	20, 5.0%

Comments:

Comments included:

- Need to increase efficiency in:
  - Collecting specimens, especially with a recent increase in rejected specimens
  - Receiving results more quickly, especially between the first and second screen
  - Followup of positive screens and case management, especially keeping track of patients who move
- Concern with the high rate of false positives for low T4 in low birthweight and preterm infants

6. What potential pros and cons do you see in the implementation of tandem mass spectrometry (MS/MS) testing to expand the number of metabolic disorders that are screened?

Comments included:

- Pros:
  - Increased number of diseases screened and infants identified, earlier diagnoses
  - Fewer false positives with MS/MS
  - Increased overall quality of care and increased parental confidence

- Helps to address medical research concerns, especially with the increased research into causes of autism

- Cons:
  - Increased logistical concerns for training, specimen collection, and reporting of results
  - Currently inadequate infrastructure, especially for the increased need for followup and case management
  - Expensive, especially compared to reimbursement rates
  - Increased logistics due to false positive results in preterm infants
  - Need for immediate results and quicker turnaround due to the critical timing of disease treatment
  - Cost-benefit and cost-effectiveness concerns

Stakeholder Involvement and Communication

7. How often do you contact the DSHS laboratory?

40, 9.9% Never
164, 40.6% Rarely
79, 19.6% Once or twice a year
39, 9.6% Quarterly
40, 9.8% Monthly
15, 3.7% Weekly
3, 0.7% Daily
14, 3.4% Other

8. When you have contacted the DSHS laboratory, what kind of response have you received?

Very helpful	Helpful	Neutral	Unhelpful	Very unhelpful	Not applicable
106, 26.3%	162, 40.2%	39, 9.7%	5, 1.2%	3, 0.7%	18, 4.5%

9. How often do you contact the DSHS NBS Case Management Program staff for follow-up issues?

123, 30.5% Never
167, 41.4% Rarely
46, 11.4% Once or twice a year
17, 4.2% Quarterly
10, 2.5% Monthly
5, 1.2% Weekly
1, 0.2% Daily
12, 0.3% Other

10. When you have contacted the DSHS NBS Case Management Program staff for follow-up issues, what kind of response have you received?

Very helpful	Helpful	Neutral	Unhelpful	Very unhelpful	Not applicable
74, 18.4%	130, 32.3%	34, 8.4%	2, 0.5%	2, 0.5%	84, 20.8%

11. Would you or someone on your staff be available to help to identify NBS program needs and solutions? If so, for which activities? Please check all that apply.

100, 24.8% Patient education
91, 22.6% Patient follow-up, case management, and systems of care
31, 7.7% Genetic counseling
58, 14.4% Testing
11, 2.7% Infrastructure
32, 7.9% Program evaluation and research
21, 5.2% Ethical, legal, and social issues
19, 4.7% Cost-benefit analysis and funding
9, 2.2% Other
97, 24.1% Not available to help
56, 13.9% Not applicable

12. Which of the following stakeholder groups would you consider critical or necessary to serve as representatives to a broad-based, multidisciplinary NBS ad hoc committee to provide input to DSHS concerning the expansion and enhancement of the program? Please check all that apply.

316, 78.4% Family practice physicians, pediatricians
163, 40.4% Sub-specialty physicians

187, 46.4% Nurses
205, 50.9% Nutritionists, genetic counselors, social workers, and other relevant health professionals
44, 10.9% Representatives from these professional associations
158, 39.2% Consumers, parents, family members, individuals with the disorders
74, 18.4% Ethicists and advocacy groups:
90, 22.3% Texas Department of Insurance
12, 30.0% Major insurance carriers
111, 27.5% HHSC (Medicaid) staff
9, 2.2% Other:

Suggested professional associations included: American Association of Pediatrics (AAP), American Hospital Association (AHA), American College of Medical Genetics (ACMG), American Medical Association (AMA), American Society of Clinical Pathologists, Association of Texas Midwives, Association of Women's Health, Obstetric, and Neonatal Nurses (AWHONN), Board of Nursing Examiners, College of American Pathologists, Texas Academy of Family Practice (TAFP), Texas Medical Association (TMA), Texas Pediatric Society (TPS), Texas Hospital Association (THA), and Texas Rehabilitation Association (TRA)

Other professionals and advocacy groups included: geneticists, laboratory professionals, faculty-level physicians in metabolic research, March of Dimes, nurse practitioners, nurse midwives, direct entry midwives, and pediatricians

Education and Training

13. How many of your staff do you have that would benefit from NBS education and training?

79, 19.6% Less than 2 staff
135, 33.5% 2-5 staff
55, 13.6% 6-10 staff
35, 8.7% 11-20 staff
16, 4.0% 21-50 staff
13, 3.2% Greater than 50 staff
53, 13.2% Not applicable

14. Describe your staff's current need for education on the following topics. Please use a scale of 1 = never needed; 2 = rarely needed, 3 = needed yearly, 4 = needed more often than yearly.

Never needed	Rarely needed	Needed yearly	Needed more often than yearly	Topic
1: 74, 18.4%	**2**: 103, 25.6%	**3**: 78, 19.4%	**4**: 20, 5.0%	How to collect a NBS blood specimen
1: 63. 15.6%	**2**: 92, 22.8%	**3**: 99, 24.6%	**4**: 23, 5.7%	What is an unsatisfactory specimen
1: 52, 12.9%	**2**: 85, 21.1%	**3**: 109, 27.0%	**4**: 30, 7.4%	What is an abnormal specimen result
1: 42, 10.4%	**2**: 75, 18.6%	**3**: 132, 32.8%	**4**: 41, 10.2%	What is appropriate follow-up on abnormal results
1: 75, 18.6%	**2**: 87, 21.6%	**3**: 79, 19.6%	**4**: 20, 5.0%	How to complete the NBS patient demographic information form
1: 37, 9.2%	**2**: 51, 12.7%	**3**: 151, 37.5%	**4**: 45, 11.2%	New issues in NBS specimen collection
1: 29, 7.2%	**2**: 54, 13.4%	**3**: 162, 40.2%	**4**: 57, 14.1%	Expansion of newborn screening
1: 47, 11.7%	**2**: 40, 9.9%	**3**: 134, 33.3%	**4**: 60, 14.9%	Impact of tandem mass spectrometry (MS/MS) technology on NBS
1: 10, 2.5%	**2**: 1, 0.2%	**3**: 5, 1.2%	**4**: 6, 1.5%	Other
			33, 8.2%	Not applicable

15. In what kinds of formats would you prefer to receive educational information? Please rank these, using 1 = most effective for you and 14 = least effective for you. ***Responses that ranked these formats in the top 5:***

266, 66.1% Newsletter
229, 56.9% Brochures
145, 36.0% Toolkit
115, 28.6% Posters
97, 24.1% Borrowed videotape
36, 8.9% Purchased videotape
111, 27.6% CD-ROM
162, 40.2% Web site

108, 26.8% Regional site training
36, 8.9% Training workshop in Austin
121, 30.0% One-on-one training in facility
48, 11.9% Telephone/teleconference training
47, 11.7% Webcasts
6, 1.4% Other

16. How helpful do you find the DSHS *Newborn Screening News* newsletter?

Very helpful	Helpful	Neutral	Unhelpful	Very unhelpful	Not applicable
80, 19.8%	176, 43.6%	75, 18.6%	3, 0.7%	3, 0.7%	52, 12.9%

17. What kinds of articles in the DSHS *Newborn Screening News* newsletter would assist you in performing your job more effectively?

Wider circulation was requested, including some respondents who said they were not on the mailing list.

Suggested topics for articles included: disorders and their symptoms, case studies, tips on counseling and followup with parents, referral algorithm for abnormal values, specimen collection tips, accuracy of testing (e.g., statistics on true and false positives in Texas), new updates, handling specimens, parent education, professional continuing education, follow-up studies, lists of specialist/referral numbers, review of procedures, tips for screening preterm infants, and resources for Spanish speakers

18. In general, how helpful do you find education materials from DSHS (e.g., Web, newsletters)?

Very helpful	Helpful	Neutral	Unhelpful	Very unhelpful	Not applicable
64, 15.8%	185, 45.8%	81, 20.1%	6, 1.5%	0, 0.0%	47, 11.6%

19. What other kinds of resource materials would assist you in performing your job more effectively?

Suggested kinds of materials included: specific material for different health professionals and consumers, website with resources and referral algorithm for abnormal values, resources for Spanish speakers, handouts for parents, workshops, automated voice response system, quarterly or annual updates, toll-free hotline in Spanish and English, updated yearly practitioners guide/training manual and other training materials, patient teaching videos/DVDs, NBS information linked to immunization records and/or on TWICES, and information on other states' screening programs

20. Should professional organizations consider newborn screening as a potential topic area for which they provide continuing education for credit (CMEs, CEUs, CNEs, CHES, etc.)?

Definitely yes	Yes	Neutral	No	Definitely no	Not applicable
146, 36.2%	175, 43.4%	42, 10.4%	9, 2.2%	0, 0.0%	4, 1.0%

21. When do you educate the family about newborn screening? Please check all that apply.

109, 27.0% During the prenatal period
272, 67.5% After delivery
13, 3.2% Never
77, 19.1% Other
28, 6.9% Not applicable

Other times included: first doctor visit, 2-week checkup, when abnormal results were received, after NICU hospitalization, as needed, at the time of newborn screening, before the second screen, months and years after

delivery, childbirth class, at delivery, if the family comes in to check on immunization schedules, in the prenatal clinic, as needed/PRN, as a part of WIC classes, and when children are brought in for Texas Health Steps

22. Do you receive parent-focused NBS educational materials in the preferred languages of your families?

161, 40.0% Yes
173, 42.9% No
51, 12.7% Not applicable

If not, in which languages do you need materials? Please provide languages, title, and/or subject.

Languages: English, Spanish with simplified language and local Texan Spanish terminology and phrasing, Vietnamese, Hmong, Tagalog, Arabic, French, Japanese, Chinese, Korean, and Urdu

Titles and subjects: Fact sheets on each of the screened diseases that can be printed as needed in several languages, and importance of NBS testing

23. How effective are parent-focused NBS educational materials?

Very effective	Effective	Neutral	Ineffective	Very ineffective	Not applicable
32, 7.9%	138, 34.2%	95, 23.6%	7, 1.7%	1, 0.2%	67, 16.6%

What would make them more effective?

General: community awareness, prenatal education, and use of social workers for education

Lower literacy level subgroup: bilingual, cartoons and other pictures, 2 pages maximum, video, toll-free hotline with a bilingual educator, multimedia, and 4^{th}-5^{th} grade reading level

Higher literacy level subgroup: attractive website, explanation of false positives, and downloadable fact sheets and brochures for all conditions screened and why those conditions were selected

Screening

24. In general, how soon after specimen collection do you receive <u>abnormal</u> test results?

16, 3.9% 1-3 days
57, 14.1% 4-6 days
123, 30.5% 7-10 days
62, 15.3% 11-15 days
5, 13.6% Longer than 15 days
45, 11.0% Unknown
26, 6.5% Never / not applicable

25. In general, how soon after specimen collection do you receive <u>normal</u> test results?

3, 0.7% 1-3 days
10, 2.5% 4-6 days
61, 15.1% 7-10 days
94, 23.2% 11-15 days
160, 39.7% Longer than 15 days
32, 7.8% Unknown
24, 6.0% Never / not applicable

26. Do you require access to NBS information, other than test results, outside regular business hours (outside Mon-Fri, 8 am-5 pm)?

43, 10.6% Yes
300, 74.4% No
26, 6.5% Unknown
21, 5.2% Not applicable

What NBS information do you need outside regular business hours?

Information included: all results 24/7, abnormal screens and what labs to draw to confirm, access to the voice response system, help finding outside results if NBS performed at hospitals or other facilities, emergency referrals, handouts and other patient information, and receipt status of test results

27. Which of the following would be preferable ways for you to obtain <u>normal</u> NBS specimen result reports? Please rank these from 1=most preferred to 8=least preferred. ***Responses that ranked these formats as most preferred:***

94, 23.3% Web site
26, 6.5% Telephone system
20, 5.0% Toll-free telephone line
29, 7.2% Real-person telephone customer service
165, 40.9% First-class US mail
30, 7.4% Express mail service or other courier
119, 29.5% Fax
11, 2.7% Other
18, 4.5% Not applicable

Other ways included: automated phone system, email, computer interface with vendor LIS system and/or electronic health record (EHR), fax, lab corporation subcontractor, regular mail, and telegram

28. Which of the following would be acceptable ways for you to obtain <u>abnormal</u> NBS specimen result reports? Please rank these from 1=most preferred to 8=least preferred. ***Responses that ranked these formats as most preferred:***

49, 12.2% Web site
75, 18.6% Telephone system
27, 6.7% Toll-free telephone line
116, 28.8% Real-person telephone customer service
59, 14.6% First-class US mail
56, 13.9% Express mail service or other courier
149, 37.0% Fax
16, 4.0% Other:
12, 3.0% Not applicable

Other ways included: genetic counselors, computer interface with vendor LIS system and/or electronic health record (EHR), fax and telephone as a double check, direct notification from the lab, registered mail, and email with a verification system to ensure receipt

29. In your opinion, where should the second specimen be collected? Please check all that apply.

272, 67.5% Primary care physician's (PCP) or pediatrician's office
45, 11.2% Delivering physician's office
79, 19.6% Clinic
146, 36.2% Hospital/birthing center
72, 17.9% Commercial laboratory
24, 6.0% Other
19, 4.7% Not applicable / no opinion

30. Using courier service (e.g., express mail service) to transport specimens from your facility to the DSHS laboratory is a more expensive option than current mail delivery. Is the benefit of courier service worth the additional cost?

74, 18.3% Yes
210, 52.0% No
66, 16.4% Unknown
29, 7.2% Not applicable
5, 1.2% Other

Funding, Billing, and Reimbursement

31. Whom do you bill for NBS services? Please check all that apply.

158, 39.2% Family's private pay/self-pay
196, 48.6% Family's insurance
139, 34.5% Publicly funded insurance
33, 8.2% Other:
44, 10.9% Unknown
85, 21.1% Not applicable

Other responses included: free for some or all patients/absorbed costs, billed through lab, CHIP, family members, grant funds, Medicaid, metropolitan health district, and Title V

32. Which CPT or HCPCS codes do you use to bill for NBS services?

Codes included: 34616, 36415, 53620, 82760, 82775, 83020, 83021, 83498, 84030, 84437, 90471, 90472, 99000, 99001, 99201-99213, 99381, 99391, v20.0, v20.2, v30.00, v77.3, A4774, S3620, Y99381, and CDM #60840333

33. How would you rate the difficulty of billing for NBS services, in comparison with other medical services?

Very easy	Easy	Neutral	Difficult	Very difficult	Not applicable
18, 4.5%	42, 10.4%	94, 23.3%	32, 7.9%	21, 5.2%	114, 28.3%

34. How would you rate the adequacy of the reimbursement you receive for NBS, in comparison with other medical services?

Very adequate	Adequate	Neutral	Inadequate	Very inadequate	Not applicable
5, 1.2%	35, 8.7%	77, 19.1%	45, 11.2%	41, 10.2%	112, 27.8%

35. In your opinion, which NBS program services should be covered by fees? Please check all that apply.

237, 58.8% Laboratory specimen collection
152, 37.7% Transporting specimens to DSHS
176, 43.7% First specimen analysis
163, 40.4% Second specimen analysis
192, 47.6% Follow-up of abnormal test results
145, 36.0% Follow-up of unsatisfactory test results
145, 36.0% Services of PCPs/pediatricians
90, 22.3% Services of subspecialists
80, 19.9% Services of other healthcare professionals
102, 25.3% Education of parents/guardians
52, 12.9% Education of physicians
51, 12.7% Education of other healthcare professionals
68, 16.9% Education of laboratory staff
58, 14.4% Education of the public (e.g., public awareness and other campaigns)
98, 24.3% Immediate treatment
89, 22.1% Case management/care coordination
73, 18.1% Short-term medical management
66, 16.4% Long-term outcome monitoring
78, 19.4% Data management—linking of first and second screenings, birth record, etc.
75, 18.6% Data management—collecting and maintaining a registry for long-term follow-up and comparative data
8, 2.0% Other

Additional comments regarding services and coverage by fees:

Comments were summarized into 3 main types of responses:

- This should be a free service to everyone, funded by the state.
- Insurance should cover fees and increase reimbursement rates.
- Supplies, such as collection cards, should only be paid for if they are used, and not prepaid.

Follow-up after Screening

36. What barriers exist to your timely reporting to the NBS Case Management Program of confirmatory testing, final diagnoses, and treatment plans?

45, 11.2% NBS reporting forms are too cumbersome
31, 7.7% Excessive number of requests for information
55, 13.6% Confusion as to what information is being requested
64, 15.9% Staff are too busy
21, 5.2% Unable to contact NBS Program staff
15, 3.7% Other:
187, 46.4% Not applicable

Other barriers included: baby not found, parents not found, phone number not found, and baby's surname changed

37. Which method would be the most effective for communicating <u>abnormal</u> newborn screening results to the responsible physician/submitter? Please rank these from 1=most effective to 6=least effective.

Results for "most effective":

272, 67.5% Telephone call directly to physician and then fax
49, 12.2% Telephone call to hospital/birthing center staff and then fax
23, 5.7% Secure email
95, 23.6% Fax to physician
18, 4.5% Fax to hospital/birthing center staff
21, 5.2% Other:
17, 4.2% Not applicable

Other methods included: computer interface, call to physician's nurse, fax to physician, US mail followup, genetic counselor, mail a hard copy, registered mail, phone call, DSHS Central or other health department representative, and telegram

38. Under which circumstances should DSHS contact <u>the physician directly</u>? Please check all that apply.

272, 67.5% Contact of physician by DSHS for moderate level (less than panic level) abnormal screening laboratory results
144, 35.7% Contact of physician by DSHS for borderline level abnormal screening laboratory results
66, 16.4% Contact of physician by DSHS for carrier status laboratory results
29, 7.2% Other:
50, 12.4% No opinion; not applicable

Other circumstances included: all abnormal results or findings, any time followup needs to be done, and private practice patients. Physicians are already contacted for panic levels.

39. Under which circumstances should DSHS contact <u>the physician's staff</u>? Please check all that apply.

230, 57.1% Contact of physician's staff by DSHS for moderate level (less than panic level) abnormal screening laboratory results
213, 52.9% Contact of physician's staff by DSHS for borderline level abnormal screening laboratory results
121, 30.0% Contact of physician's staff by DSHS for carrier status laboratory results
24, 6.0% Other:
45, 11.2% No opinion; not applicable

Other circumstances included: never unless that person is trained to deal with the results, all abnormal results, all inconclusive and/or unacceptable findings, any time followup needs to be done, and mother lacks a primary care physician. Physicians are already contacted for panic levels.

40. Who, in your opinion, should be responsible for following up on an <u>abnormal</u> first screen performed by the hospital/birthing center? Please check all that apply.

297, 73.7% Primary care physician (PCP)/pediatrician
53, 13.2%% Delivering physician
38, 9.4% Subspecialist physician
21, 5.2% Other healthcare professional:
166, 41.2% Hospital/birthing center staff
34, 8.4% Clinic staff
18, 4.5% Other:
15, 3.7% No opinion; not applicable

Other healthcare professionals included: social worker, pediatrician, physicians who saw the patient and/or the mother, subspecialty MD and RD, genetic counselor, hospital personnel, lab personnel, midwife, neonatologist, NP, PA, DSHS staff, and whoever received the report

Other persons included: the patient/family, city/county NBS, DSHS, hospital staff, lab, public health department, public health nurse

41. Which activities do you perform to ensure proper follow-up? Please check all that apply.

304, 75.4% Call parent
68, 16.9% Call hospital medical records department
106, 26.3% Send a letter via first class US mail
83, 20.6% Send a certified letter
30, 7.4% Home visit
66, 16.4% Request assistance from DSHS NBS Program staff
27, 6.7% Other:
51, 12.7% Not applicable

Other activities included: send a report to the physician, ask parents at office visit, call local police department or sheriff's office, call primary care providers, certified letter to the parents, reminder card, and contact hospital neonatology department

42. Which of the following are barriers to your ability to provide adequate follow-up after screening? Please check all that apply.

158, 39.2% Never saw the patient
274, 68.0% Cannot locate viable phone number for parent
220, 54.6% Cannot locate address for parent
240, 59.6% Parent does not keep appointment
67, 16.6% Parent refuses treatment
24, 6.0% Other:
69, 17.1% Not applicable

Other barriers included: family in Mexico, expired insurance, name changes, changes of primary care providers, incomplete records, do not receive records but are responsible for followup, wrong telephone number or address, and parents are noncompliant with followup

43. What communication methods are most effective for communicating <u>abnormal</u> newborn screening results to your patients' families?

Effective methods included: telephone call, email, letter, certified letter, direct face-to-face discussion, scheduling an appointment in primary care physician's office to discuss results, handouts, and call to parents from DSHS Central office

44. What follow-up needs do your patients have?

Patients' needs included: parent education, financial needs, transportation, immigration status, access to PCPs and specialists, cost-efficient treatment from a specialist, referrals and transportation to other health services and dietitians/nutritionists, timely updates on confirmatory test results, repeat thyroid screens for preterm infants, care coordination, counseling, and physician treatment after a home birth or birthing center birth

Diagnosis, Treatment, and Coordination of Care

45. What activities do you do to ensure coordination of confirmatory diagnostic testing? Please check all that apply.

268, 66.5% Contact parent to schedule lab work
210, 52.1% Schedule follow-up to ensure appointment was kept
23, 5.7% Other
73, 18.1% Not applicable

Other activities included: calling the physician, calling the court or police department to help with contacting the parents, home visits, and in-house testing with NICU patients

46. What activities do you do to inform the DSHS NBS Program about patient care? Please check all that apply.

82, 20.3% Call DSHS NBS Program to let them know labs were scheduled
161, 40.0% Fax lab results to NBS
53, 13.2% Phone lab results to NBS
25, 6.2% Other
120, 29.8% Not applicable

Other activities included: contact DSHS to inform them that lab specimens were drawn, call to check if the specimen was acceptable, mail confirmatory information and report forms, send lab results if requested, send the repeat NBS if abnormal, and nothing

47. How would you describe your capacity to diagnose patients with abnormal screens?

Very capable	Capable	Neutral	Incapable	Very incapable	Not applicable
85, 21.1%	158, 39.2%	54, 13.4%	8, 2.0%	4, 1.0%	62, 15.4%

48. How would you describe your capacity to treat patients with abnormal screens?

Very capable	Capable	Neutral	Incapable	Very incapable	Not applicable
63, 15.6%	122, 30.2%	82, 20.3%	29, 7.2%	6, 1.5%	64, 15.9%

49. How would you describe your capacity to coordinate care for patients with abnormal screens?

Very capable	Capable	Neutral	Incapable	Very incapable	Not applicable
113, 28.0%	156, 38.7%	39, 9.7%	9, 2.2%	2, 0.5%	54, 13.4%

50. How would you describe the difficulty of referring your patients to subspecialists?

Very easy	Easy	Neutral	Difficult	Very difficult	Not applicable
47, 11.7%	18, 29.2%	80, 19.8%	52, 12.9%	17, 4.2%	58, 14.4%

51. Which of the following are barriers to referring your patients to subspecialists? Please check all that apply.

72, 17.9% Unaware of who subspecialists are
127, 31.5% Subspecialists are too busy to see patients
137, 34.0% Patient transportation issues
130, 32.3% Patient scheduling issues
57, 14.1% Other
10, 2.5% Unknown
94, 23.3% Not applicable

Other barriers included: subspecialists' not accepting Medicaid, other financial issues, access to care/transportation issues, lack of subspecialists in the area, parents' not understanding why their child needs to see subspecialists, and subspecialists' refusing to see home birth or birthing center patients.

52. Would it be beneficial to establish regional centers with defined "catchment areas" to ensure confirmatory diagnosis and treatment?

Definitely yes	Yes	Neutral	No	Definitely no	Not applicable
77, 19.1%	148, 36.7%	73, 18.1%	18, 4.5%	4, 1.0%	31, 7.7%

Why or why not?

Benefits of regional centers with defined "catchment areas" included: less travel distance, availability of resources for local physicians, more information for parents, known location for treatment, decreased referral time, quicker followup, easier coordination of parental education/ referrals/ patient care, known source of care regardless of ability to pay, easier to standardize care, reduced risk of missed results, potential for centralized multidisciplinary evaluations, standardized laboratory quality for handling unusual tests, easier to establish parent support groups, and reliable funding

Drawbacks of regional centers with defined "catchment areas" included: decreased choice of subspecialists, another layer of bureaucracy, possible difficulties if patients have care outside of the catchment area, inability to provide continuity of care when patients move, inability to meet individual needs, and low cost-benefit of establishing the centers for the small number of patients

Data Management

53. What parts of the laboratory report could be improved for ease of use? Please check all that apply.

26, 6.5% Submitter/Facility address
73, 18.1% Patient information
71, 17.6% Result information
74, 18.4% Unsatisfactory result information
75, 18.6% Informational comments at the bottom of the result report
16, 4.0% Other
114, 28.3% Not applicable

Other parts included: addresses of PCPs, better highlighting of abnormal results, electronic transmission, recommendations/guidelines for abnormal result follow-up, identification (bolding) of required information, test number (sequence), secure online access to results, clearer interpretation of the test, known contact information for the patient, and accompanying parent information

Comments:

Comments included: patient contact information should go to both the inpatient provider and the PCP doing the outpatient care, abnormal results should be bolded, "notice" comments are unnecessary, and more room for doctor notes and parental notification comments

54. Which data do you routinely need or collect?

Comments included: patient and guardian names in full, race/ethnicity, dates of birth, telephone numbers, addresses, alternate contact numbers, patient ID number to link to hospital patient records, insurance information, family history, birthweight, formula, medicaid number, patient's and mother's SSNs, when second screen has not been done, positive screens, followup data, hypothyroid bloodwork results, NBS screening card number, and physician's orders

55. Would it be helpful to you if the first screen, second screen, birth record, and follow-up data from subspecialty centers were linked?

Very helpful	Helpful	Neutral	Unhelpful	Very unhelpful	Not applicable
156, 38.7%	146, 36.2%	38, 9.4%	2, 0.5%	2, 0.5%	22, 5.5%

56. In order to link the first and second screens, would you be willing to consider using a 2-part form? The first part would be used for the first (hospital) screen, and the second part would be used for the second (2 week) screen. Please check one.

52, 12.9% We would not use a 2-part form. **Why?**
135, 33.5% We would use a 2-part form if required
80, 19.9% We like the idea, but foresee problems. **What problems?**
42, 10.4% We like the idea and do not foresee any problems
37, 9.2% Not applicable

Reasons to not use a 2-part form included: first and second screens are performed in different sites, providers, hospitals, and/or areas, the forms would be lost or misplaced, increased possibility of miscommunication and error, increased paperwork, and the form should be electronic instead

Problems if a 2-part form were used included: the second part of the forms would be lost or misplaced, increased paperwork, noncompliance for the second screen, and the first and second screens are performed in different sites, providers, hospitals, and/or areas

Program Evaluation and Feedback

57. What types of feedback about your operations do you <u>currently</u> receive from DSHS? Please check all that apply.

148, 36.7% Assistance with unsatisfactory specimens
106, 26.3% Assistance with NBS follow-up protocols
107, 26.6% Assistance with specimen collection techniques
93, 23.1% Information on NBS disorders that are screened
76, 18.9% Assistance with interpretations of abnormal results
3, 0.7% Other
76, 18.9% No feedback received from DSHS
63, 15.6% Not applicable

Comments:

Comments included: feedback currently received only on hearing screens, would like more information about why specimens are rejected, need handouts about conditions that NBS screens for, need information about local dietary consults, need assistance for referrals to specialists, and need normal values for abnormal screens and guidelines

58. What types of feedback about your operations are you currently <u>not</u> receiving from DSHS, but you know would be useful for your QI and evaluation goals?

Types of feedback included: location of available subspecialists, what confirmatory tests are appropriate and where they can be obtained, annual report about each facility as compared to critical indicators, turnaround time for reports, assistance with lab results, number of screens and recollects that are sent per month or per quarter, alerts when incidence of positive screens changes, NBS followup protocols, how to bill properly, educational information for parents, incidence statistics for NBS conditions, verifying and updating education for collectors, and status of specific cases upon request

59. How would you rate the ease of providing feedback to DSHS?

Very easy	Easy	Neutral	Difficult	Very difficult	Not applicable
15, 3.7%	111, 27.5%	126, 31.2%	16, 4.0%	5, 1.2%	39, 9.7%

60. What types of statistical feedback would you find useful in the future? Please check all that apply.

170, 42.2% Specimen Unsatisfactory statistics
120, 29.8% Specimen Presumptive positive statistics
137, 34.0% Number of screen specimens submitted
153, 38.0% Number of "Lost to Follow-up" patients, based on the first screen
148, 36.7% Prevalence of conditions
128, 31.8% Data for your Health Service Region (Public Health Region), as a whole
128, 31.8% Data for Texas, as a whole
88, 21.8% Comparative data from other states
1, 0.2% Other
53, 13.2% Not applicable

61. How often would you like to receive statistical feedback?

13, 13.2% Never
111, 27.5% Once a year
65, 16.1% Twice a year
106, 26.3% Quarterly
19, 4.7% Monthly
12, 3.0% Upon request
0, 0.0% Other
29, 7.2% Not applicable

Thank you very much!
If you wish to comment further, please attach your comments and return them with this survey.

Comments included comments about the survey itself and continuation of comments about specific questions.

Appendix 4: Parent Survey

Note: Percentages might not sum to 100.0% because of missing responses. Percentages reflect the number of people answering each question according to the survey's skip patterns.

Opening paragraphs

Hello, my name is [name]. I am a [job title] with the Newborn Screening Program of the Texas Department of State Health Services. We're conducting a statewide study to find out more about the experiences of parents whose children have been diagnosed with a condition that was detected through newborn screening. Your telephone number was chosen at random from our records to be included in the study.

Would you be willing, at this time, to answer a few questions? This survey should take approximately fifteen (15) minutes of your time.

Education of the public

1) What information did you already know about newborn screening when your baby was first screened?

Effectiveness of letters to families

2) After your baby was screened, did you receive a letter from the Newborn Screening staff about your need to take your baby to a doctor for further testing?
   Yes --86.1% No--0.0% Don't know--2.8% Don't remember--11.1%
3) [If **yes**, they received a letter] Was the letter written in a language that you use every day?
   Yes--93.5% No--3.2% Don't know--3.2% Don't remember--0.0%
4) [If **yes**, they received a letter] Was this letter easy to understand?
   Yes--80.6% **No**--6.5% Don't know--3.2% Don't remember--0.0%
   - [If **no**, it was not easy to understand] What made the letter hard to understand?
5) [If **yes**, they received a letter] How could the letter have been improved?

Targeted education and PR efforts

6) At the time of the testing, did you have questions or concerns about the results of the testing?
   Yes--50.0% No--38.9% Don't know--2.8% Don't remember--5.6%
7) [If **yes**, they had questions or concerns] What were they?

8) [If **yes**, they had questions or concerns] Did you call the DSHS Newborn Screening staff in Austin?
   Yes--33.3% No--61.1% Don't know--0.0% Don't remember--5.6%
9) [If **yes**, they called the NBS staff] Did you have trouble reaching a person who would explain things to you?
   Yes--33.3% No--50.0% Don't know--0.0% Don't remember--0.0%
10) [If **yes**, they called the NBS staff] How could your call to Newborn Screening staff have been improved?

Active/passive mechanisms for distributing materials; eliminating barriers in communication tools

11) Were you given brochures or other information to read about your baby's condition after s/he was diagnosed?
    Yes--86.1% No--11.1% Don't know--2.8% Don't remember--0.0%
12) [If **yes**, they received information] Were the brochures written in a language that you use every day?
    Yes--90.3% No--6.5% Don't know--0.0% Don't remember--0.0%
13) [If **yes**, they received information] Was this information easy to understand?
    Yes--77.4% **No**--3.2% Don't know--3.2% Don't remember--3.2%
    - [If **no**, it was not easy to understand] What made this information hard to understand?
14) [If **yes**, they received a letter] How could the information have been improved?

Sufficient capacity for diagnosis and treatment

15) Did you have problems finding a specialty doctor in your area?
    Yes--16.7% No--83.3% Don't know--0.0% Don't remember--0.0%
16) Are you currently having any problems obtaining treatment for your baby's condition?
    Yes--16.7% No--80.6% Don't know--0.0% Don't remember--0.0%
17) [If **yes**, they are currently having problems] What kind of problems are you having?

End of survey

These are all the questions I have. I'd like to thank you on behalf of the Newborn Screening Program for your time and effort. If you have any questions or concerns, please feel free to contact [name] at [NBS phone number]. Thank you again

Appendix 5: Statutory Legal References – Newborn Screening

HB 790

HB 790 requires by March 1, 2006: (1) DSHS to conduct study to determine most cost-effective method of conducting NBS; (2) determine disorders to be studied; (3) obtain proposals or information on the conduct of NBS and compare costs of DSHS performing to costs of outsourcing to lab with at least two years experience in NBS tests; and (4) provide report to governor on (1).

By October 1, 2005, DSHS to study assessment of TX NBS program. HHSC executive commissioner may adopt rules to implement a NBS program.

If DSHS lab is more cost-effective than outsourcing DSHS shall obtain tandem mass spectrometers. Implement by November 1, 2006.

If outsourcing is more cost-effective, DSHS shall contract using competitive process. Implement by November 1, 2006.

DSHS may adjust amounts charged for NBS fees.

Newborn screening program

Health and Safety Code, Chapter 33, § 33.002 authorizes NBS program to combat morbidity and adoption of rules (now HHSC executive commissioner adopts rules).

§ 33.002 requires DSHS to establish and maintain lab to conduct activities to develop screening and diagnostic tests, develop ways to prevent or treat listed diseases/disorders, and serve other programs necessary to carry out program.

§ 33.003 says DSHS may invite all physicians, hospitals, and other health care providers that provide maternal and infant care to cooperate and participate. Other boards, agencies, departments, and political subdivisions may cooperate and are encouraged to furnish services and facilities to program.

§33.011 requires physicians to subject newborn child to screening tests approved by DSHS. DSHS may prescribe the screening tests procedures and standards. Tests must be performed by DSHS lab or a lab approved by DSHS under 33.016. To extent of available funding, DSHS shall require NBS tests for disorders listed in 2005 ACMG report. (as amended by HB 790)

§ 33.014 says if DSHS suspects child may have disorder/disease, DSHS shall notify person who submitted test and may notify parents and listed others and recommend further testing when necessary. DSHS shall recommend high risk child be placed under medical care for diagnosis and provide name of specialist in child's geographic area. DSHS, local health authority, and specialist may follow up a positive test with attending physicians and parents. (as amended by HB 790)

§ 33.015 each physician, health authority or other individual who has info on confirmed case detected by mechanism other than identification through screening by DSHS's lab shall report confirmed case to DSHS. DSHS may collect data from screening specimen form. DSHS shall

maintain roster of children born in TX who have been diagnosed with disorder/disease. DSHS may cooperate with other states in development of national roster.

§ 33.016 says DSHS may develop program to approve any lab that wishes to perform required tests. Board of Health (HHSC executive commissioner) may adopt rules prescribing procedures and standards for the conduct of the program. DSHS may prescribe test procedure to be used and standards for each test. DSHS may for good cause and after notice and an APA hearing if requested restrict, suspend, or revoke any approval of a lab.

§ 33.031 requires all newborns and others under 21 who have been found to be positive and who may be financially eligible to be referred to CSHCN. If eligible, then child shall be given services through CSHCN. If not eligible, then child shall be referred to NBS program for a determination of eligibility for NBS program services. (as amended by HB 790)

§ 33.032 is re program services. Within funding limits, DSHS may provide services directly or through approved providers to positive individual of any age who meets eligibility criteria specified by rule. Board (HHSC executive commissioner) may adopt rules. Board may charge fees for the provision of services except the services may not be denied because of inability to pay the fees. (as amended by HB 790)

§ 33.033 requires consent of parent of minor for services.

§33.034 requires fair hearing if DSHS denies, modifies, suspends, or revokes provider's status.

§ 33.035 says individual is not eligible for services at no or reduced costs if person with legal obligation to support is eligible for some other benefit that would pay for all or part of services. DSHS may waive eligibility. Rules shall provide criteria. § 33.036 requires fair hearing if eligibility is denied, modified, suspended, or revoked. §33.037 says board may require individual to reimburse DSHS for services. § 33.038 authorizes DSHS to recover expenditures if person does not reimburse.

25 Texas Administrative Code (TAC) Chapter 37, Subchapter D, §§ 37.51-37.67 are DSHS' rules on NBS.

Lab fees - generally

Health and Safety Code, Chapter 12, §§ 12.031 defines "public health services" to include lab services; personal health promotion, maintenance, and treatment services; and administrative services.

§ 12.032 authorizes Board of Health (HHSC executive commissioner) by rule to charge fees to persons who receive public health services from DSHS. Board may require DSHS contractors to charge fees for public health services provided by contractors. Contractor shall retain a fee collected and shall use the fee in accordance with the contract. Amount of fee may not exceed cost to DSHS of providing the service. Board may establish a fee schedule, shall consider person's ability to pay, and may not deny for inability to pay.

§ 12.034 says board shall establish collection procedures that shall be used by DSHS and DSHS contractors that are required to charge fees. Fees may be collected before or after services. Board may waive by rule collection procedures if administrative cost exceeds fees to be collected. If

board elects to require cash payments by program participants, money shall be deposited at end of each day and to state treasury within seven days.

S 12.035 requires DSHS to deposit all money collected for fees and charges under 12.032 to credit of public health services fee fund.

§ 12.036 in furnishing public health services, DSHS is subrogated to person's right to recover from insurance, etc. and DSHS may waive right to subrogation.

§ 12.037 says DSHS may modify, suspend, or terminate public health services to persons for nonpayment of billed services after notice and right to fair hearing. Board by rule shall prescribe criteria for action.

§ 12.038 says board may adopt rules to implement this law.

25 Texas Administrative Code (TAC) Chapter 73 are DSHS' rules on lab fees. § 73.21 concerns NBS. Test kits for Medicaid-eligible or charity care newborns are provided at no costs. The test kit fee for all other newborns is set at a maximum of $38.00 under § 73.54 (1) (F). A proposed rule will be published for comment shortly to increase the fees to a maximum of $40.

OAG opinions (multitude of such) say that state agencies can only charge a fee if there is a statute that allows the agency to charge the fee.

Health and Safety Code, § 12.0122 pertains to sale of lab services. It authorizes DSHS to contract for sale and provision of lab services with government entities or freestanding, nonprofit clinics. DSHS by rule may establish charges for sale. "Lab services" means services performed by DSHS lab. DSHS rule on this is at 25 TAC § 73.41 that says the charges for the sale of lab services shall be the charges negotiated in the contract.

Lab fees – bond debt service

DSHS rider 27 says all receipts from lab fees for biennium are appropriated to DSHS for transfer to TPFA for payment of debt services on the revenue bonds. Appropriations for indirect administration may be transferred for bond debt service payments only if lab fees generated by lab are insufficient to support debt service.

DSHS rider 45 says DSHS is appropriated any additional lab revenues generated in GR Dedicated Account 524, above the amounts identified for this account in the Comptroller's Biennial Revenue Estimate, for purpose of lab operations. This appropriation does not include amounts in Revenue Object 3561. The dollars above are contained in Comptroller's Biennial Revenue Estimate 2006-07 for Revenue Objects deposited into account 524 and exclude amounts estimated for Revenue Object 3561. Lab revenues deposited into Revenue Object 3561 are statutorily dedicated for lab debt service and may not be used for any other purpose.

HB 3050, 74th Legislature, 1995, Section 1 exempts public health services fund from sweep at end of biennium, recreated fund as special account in GR fund, and says fund may be used only for purpose designated by law.

HB 3050, 74th Legislature, 1995, Section 11 says revenue in excess of 1994 levels, as determined by comptroller, from receipts of lab analysis deposited to account 524 may be used only for purpose of financing TDH's lab facility.

HB 3050, 74th Legislature, 1995, Section 12 says revenue in excess of 1994 levels, as determined by comptroller, from EPSDT fee deposited to account 273 may be used only for purpose of financing TDH's lab facility.

DSHS's last annual report to TPFA (required by bond documents) says projected pledged revenues (for debt service) in FY 2006 are $1.2 million beginning fund balance, $2.1 million from non-Medicaid NBS fee, $0.6 million from safe drinking water fees, and $0.4 million from EPSDT blood lead fees for total debt service of $4.4 million. Footnote says NBS fee and other lab fees are projected to be about $6.0 million annually in FY 2006 and fees in excess of need for debt service are appropriated for lab operations.

Official Statement of TPFA Special Revenue Bonds (TDH Lab Project bonds were issued in 1998 and refunding bonds were issued in 2004) establishes pledge of revenues for debt service (pledged revenue). Obligation to make payments is subject to appropriations by the legislature. Pledged revenues (in order of application) are dedicated receipts generated by DSHS from lab fees or any other money available now or in the future to make debt service payments, transferred revenues (if legally available and necessary to make debt service payment), and any other source of legally available funds of DSHS designated for such purpose. DSHS on behalf of TBPC is obligated to make payments for debt service. Funds available to DSHS must first be applied to debt service payment.

No assurances are given as to future source and availability of pledged revenues. Obligations of TBPC and DSHS are absolute and unconditional and not subject to diminution, abatement, set-off, or counterclaim for any reason but are subject to appropriations of pledged revenues. DSHS shall seek the necessary biannual appropriations necessary to meet debt service obligations. As long as bonds are outstanding, DSHS shall levy, assess, charge and collect dedicated revenues and other money or fees legally available in amounts sufficient to provide money to pay debt service. DSHS agrees to refrain from action that would adversely affect tax-exempt status of the bonds.