

Introduction

Newborn screening is recognized internationally as an essential, preventive public health program for early identification of genetic disorders in newborns that can affect their life and long-term health. The State of Texas mandates that each newborn receive screening soon after birth followed by a second screen between 7 and 14 days of age. Each year, the Texas Department of State Health Services (DSHS) newborn screening laboratory performs approximately 780,000 newborn screens making it the highest volume screening laboratory in the world. In February 2007, the DSHS Texas Newborn Screening Program (TNSP) added Tandem Mass Spectrometry (MS/MS) screening technology and increased its newborn screening panel from 7 to 27 disorders¹. The implementation of MS/MS screening has allowed for improved sensitivity, specificity, and scope of newborn screening services.

The goal of the TNSP is to identify infants that screen positive for newborn screening disorders. Each year approximately 15,000 abnormal screens are identified by the program. If a disorder is detected and treated early in a newborn, the result prevents serious problems such as mental retardation, illness or death and also saves on healthcare costs and institutional services. The TNSP encompasses several steps and processes including: collection of the blood specimens for the initial and second screen; laboratory analysis of the blood specimens; case management/follow-up of abnormal screens to ensure diagnosis/treatment, and; long-term follow-up.²

¹ For a quick reference to the newborn screening disorders, please see Appendix.

² For information on treatment of common disorders, please see Appendix.

Purpose and Need

The size and scope of the TNSP creates unique challenges in identifying affected infants, and in ensuring timely, appropriate care to infants in need. Texas is the largest state in the contiguous United States with the longest shared border in the country. Hispanic families often move back and forth across the Texas-Mexico border making it difficult to ensure all children born in Texas are actually screened. TNSP fiscal year 2006 reporting of Title V data indicated that 96.9% of newborns had at least one newborn screen in that fiscal year. This indicates that possibly 12,000 newborns were not screened, and possibly some of these could have one of the newborn screening conditions.

Access to care is a problem for families with an affected child that live in rural communities. Parents sometimes must travel several and sometimes hundreds of miles for appropriate care for their newborn by pediatric sub-specialists. With over 2,300 birthing facilities and healthcare practitioners submitting over 780,000 specimens each year, on-going educational efforts are always needed to address issues with specimen quality, timely submission and reporting, physician notifications and patient diagnosis, treatment and on-going evaluation.

The challenges noted above pose the greatest risk to children identified with one of the 14 disorders that can cause critical medical emergencies within the first week of life. Disorders in this category include salt wasting congenital adrenal hyperplasia (SW-CAH), galactosemia (GAL), medium chain acyl-CoA dehydrogenase deficiency (MCAD), and other disorders detectable by MS/MS technology. Data has shown that infants confirmed with these high-risk disorders do not always receive timely treatment. For example, data from August 2003 through June 2005 show 50% of infants identified with the salt-wasting form of congenital adrenal

hyperplasia (CAH) were not started on a treatment regimen until after two weeks of life.

Endocrinology specialists advocate treatment by two weeks of life to ensure positive outcomes for affected children. Treatment delays could result in serious consequences such as adrenal crises requiring hospitalization and even death.

The Texas Department of State Health Services is committed to improving performance standards for the Texas Newborn Screening Program (TNSP). To this end, the Texas Department of State Health Services proposes the Texas Newborn Screening Performance Measures Project (TNSPMP) in response to funding opportunity titled, Evidence-Based Laboratory Medicine; Quality/Performance Measure Evaluation. The performance goal of the TNSPMP is to identify tangible measures that link the quality of patient care with the quality of pre and post-analytical stages³ of the newborn screening process providing a self-sustaining model for systematic and continuous quality assessment. This goal will be attained through three phases of activities including initial investigation and study design to identify performance measures, data collection and analysis, and performance measures pilot-testing. The TNSPMP will create a Steering Committee that, in collaboration with a Newborn Screening Quality Improvement Team (NSQI), will be responsible for the main activities of the project. The purpose of the TNSPMP is to establish evidence-based best practices in the areas of pre- and post-analytical stages of the newborn screening process that will serve as a model for nationwide replication.

³ For a general definition of the pre and post analytical stages as it relates to this funding opportunity, see Appendix.

Goals and Objectives for the Texas Newborn Screening Performance Measures Project (TNSPMP)

Goal 1. Steering Committee and Charter: A formalized steering committee consisting of subject matter experts and newborn screening stakeholders will be in place to direct project activities and efforts of the three-year TNSPMP.

Objective 1.1. Within the first 3 months, a 16 member Steering Committee will be established which will include key stakeholders in the Texas Newborn Screening Program.

Objective 1.2. Within the first six months of Year 01, the Steering Committee will create a charter containing a vision and mission statement.

Objective 1.3. By the end of Year 03, the Steering Committee will have met at least quarterly during the three-year grant period.

Goal 2. Evidence-Based Performance Measures: Develop and define performance measures that reveal gaps in providing timely and effective treatment to infants diagnosed with newborn screening disorders.

Objective 2.1. By the end of Year 01, the Steering Committee will identify evidence-based deficiencies and barriers in the Texas Newborn Screening Program (TNSP) and develop the clinical question(s) to be used as the focus for the three-year project.

Objective 2.2. By the end of Year 01, performance evaluation and assessment tools will be developed to identify and measure pre and post-analytical deficiencies and gaps in the TNSP identified by the Steering Committee.

Objective 2.3. By the end of Year 02, individuals involved in pre and post-analytical stages (e.g. healthcare providers, DSHS laboratory staff, and DSHS TNSP case management staff) will be surveyed using Texas Performance Evaluation and Assessment Toolkit (TxPEAT) and other assessment strategies/tools created in Year 01 by the Steering Committee.

Objective 2.4. By the end of Year 02, the Project Coordinator with the assistance of the Project Specialist will analyze data collected from the survey tools. The Steering Committee and Newborn Screening Quality Improvement Team (NSQI) will develop and finalize performance measures for evaluating pre and post-analytical stages of newborn screening.

Goal 3. Pilot Performance Measures: Pilot key performance measures for effectiveness of improving time to treatment in order to improve quality of life for newborns diagnosed with newborn screening disorders.

Objective 3.1. Plan, design, and execute a pilot study.

Goal 4. Identify and Publish Specific Interventions: By the end of Year 03, the Steering Committee will identify, recommend, and document interventions which are likely to improve the performance and address quality issues identified.

Methods and Technical Approach for objectives of the Texas Newborn Screening

Performance Measures Project (TNSPMP)

Goal 1. Steering Committee and Charter: *A formalized steering committee consisting of subject matter experts and newborn screening stakeholders will be in place to direct project activities and efforts of the three-year TNSPMP.*

Objective 1.1. Within the first 3 months, a 16 member Steering Committee will be established which will include key stakeholders in the Texas Newborn Screening Program.

A Steering Committee will be established that represents stakeholders from all stages of the screening process including representatives from birthing facilities, primary care physicians, pediatric sub specialists, medical societies and associations, insurance carriers, parents and subject matter experts. Potential committee members expected to have leadership roles in the steering committee include:

- Brad Therrell, Ph. D., Director, National Newborn Screening and Genetics Resource Center (NNSGRC)
- Larry Sweetman, Ph.D., Director of the Mass Spectrometry Laboratory, Institute of Metabolic Disease at Baylor Research Institute.
- V. Reid Sutton, M.D., Assistant Professor of Molecular and Human Genetics, also serves as Director of the Medical Genetics Residency Program at Baylor College of Medicine. Dr. Sutton is certified by the American Board of Medical Genetics (Clinical Genetics) and by the American Board of Pediatrics.
- George R. Buchanan, M.D., Professor of Pediatric Oncology and Hematology, UT Southwestern Medical School; Children's Cancer Fund Distinguished Chair in Pediatric Oncology and Hematology; Director Southwestern Comprehensive Sickle Cell Center
- Zora R. Rogers, M.D., Associate Professor, Pediatrics, UT Southwestern Medical School; Clinical Core Director and Intercenter Research Study Principal Investigator, Southwestern Comprehensive Sickle Cell Center
- Don P. Wilson, M.D., Texas A&M Health Science Center College of Medicine

Professor of Pediatrics Chair, Department of Pediatrics; Chief, Section of Pediatric Endocrinology

- Jose L. Gonzalez, M.D., J.D., M.S.Ed., Vice Chair of Medical Education, Program Director for the Pediatric Residency Program, Internal Medicine-Pediatrics Combined Residency Program Associate Director, Associate Clerkship Director, and is an Associate Professor of Pediatrics in the Division of Endocrinology

These individuals have earned national and international recognition as leaders in the newborn screening community and will bring a wealth of expertise. The remaining steering committee members will be selected and invited based on their area of expertise and a demonstrated interest in building collaborations to advance newborn screening in Texas. Specialists in the fields of health economy, statistics, clinical study design, population screening, and program evaluation and assessment, will be identified and made available to the steering committee as needed.

The steering committee will be guided by formal roles with defined responsibilities and expectations. Responsibilities will include deciding on populations to be included in assessments, priorities for outcomes, providing important background material that reveals issues from different perspectives, approving assessment tools, interpreting findings of assessment, selecting performance measures for pilot tests, etc. All Steering Committee members will be asked to sign a letter of commitment (or memo of understanding) to serve for the three-year project period with provisions for the selection of an alternate with equivalent expertise if they are unable to participate.

Objective 1.2. Within the first six months of Year 01, the Steering Committee will create a charter containing a vision and mission statements.

The approach to create a charter will follow DSHS standard practices for project management charters. The Charter will clearly establish the committee's right to make decisions and lead the project. The Charter will be made available to all project stakeholders and everyone associated with the project, reaching as wide an audience as practical. The Charter will outline the direction and constraints of the project. The scope section of the Charter will describe the project objectives and deliverables, customers and their needs and requirements, and project stakeholders. The resources section of the Charter will name the project manager and other key project team members, the deadline, staff effort limit, budget, and other organizational constraints which the project must live within. The Charter will also outline a communication plan for involved entities for the project.

Objective 1.3. By the end of Year 03, the Steering Committee will have met at least quarterly during the three-year grant period.

Meeting minutes and tangible deliverables are traditional methods of monitoring progress toward this objective. Activities involved will ensure coordination of pre-planned meetings to allow for 100% attendance for committee members or alternates. Incentive to encourage participation steering committee members will include the provision for travel expenses including air travel, lodging, and meals. Honorariums will also be awarded after the successful completion of each meeting. Quarterly meetings will be scheduled for a minimum of two days, and videoconferencing will be available for those who would like to participate, but are unable to travel to the meeting location. Monthly conference calls will be scheduled between formal

meetings to keep all involved apprised of progress and developments, and to ensure ongoing and timely evaluation of grant activities.

Goal 2. Evidence-Based Performance Measures: Develop and define performance measures that reveal gaps in providing timely and effective treatment to infants diagnosed with newborn screening disorders.

Objective 2.1. By the end of Year 01, the Steering Committee will identify evidence-based deficiencies and barriers in the TNSP and develop the clinical question(s) to be used as a focus for the three- year project.

Final methods and the technical approach for this objective will be determined through the work of the Steering Committee in cooperation with members of the NSQI and guidance from Centers for Disease Control and Prevention (CDC) staff. General resources, literature, and publications that focus on evidence-based medicine will be reviewed including: The Cochrane Library, Clinical Evidence (www.clinicalevidence.com), ACP Journal Club (www.acpjc.org), Bandolier (www.jr2.ox.ac.uk/bandolier), TRIP Database (www.tripdatabase.com), PubMed (www.pubmed.com), and Clinical Chemistry (www.clinchem.org). In addition to literature searches, the Steering Committee will review National Newborn Screening and Genetics Resource Center (NNSGRC) recommendations resulting from a March 2005 review of the TNSP. The review was at the request of Eduardo J. Sanchez, M.D., M.P.H., past Commissioner of Health of the DSHS, and was led by Dr. Therrell and a team of eight federal, state and private newborn screening subject matter experts and expert reviewers. The extensive three-day program assessment resulted in a document containing specific observations and suggestions for improvement. Since then, the TNSP has enacted procedures to address many of the review team

suggestions; however, outstanding issues will be reviewed for potential topics that could be pertinent to this grant activity.

Once the information is reviewed, a report on the literature search will be written to summarize findings including explanations for the lack of studies and suggestions of new studies to identify and define laboratory medicine gaps. The report will also summarize outstanding issues documented by the NNSCRC review team, and include discussions on whether or not addressing them would be within the scope of the CDC project.

Objective 2.2. By the end of Year 01, performance evaluation and assessment tools will be developed to identify and measure pre and post-analytical deficiencies and gaps in the TNSP identified by the Steering Committee.

Based upon issues identified through the literature review and programmatic review summary, assessment tools will be developed for evaluating the size and scope of pre and post-analytical gaps. An existing tool developed for newborn screening program assessment that could be utilized as a framework to assess specific aspects of pre and post-analytical processes is the NNSGRC Performance Evaluation and Assessment Scheme (PEAS). Examples of pre and post-analytical areas of focus in the PEAS include: prenatal education, specimen collection and transport, short-term follow-up, long-term follow-up (e.g. medical management of conditions and outcomes), and follow-up support activities such as interpretation of test results, and communications between DSHS and healthcare providers.

Steering committee and NSQI members will identify and select specific pre and post-analytical topics relating to performance problems identified during the assessment phase of the grant. With the assistance of DSHS statisticians experienced with medical data assessments, a Texas Performance Evaluation and Assessment Toolkit (TxPEAT) will be developed. The

toolkit will be organized into specific topic areas so appropriate assessment tools can be easily administered. For example, assessing pre-analytical issues may require data extractions to determine specimen transit time from date of birth to date of receipt in the laboratory, and post-analytical assessments may include surveys to document when a physician received notification of an abnormal screen, when confirmation tests were ordered, and when treatment began. Evaluating patient outcome over time would require tools to capture information from physician notes, patient charts, and possibly information from the family. The final TxPEAT will be officially approved by the steering committee to use as one of the surveying tools.

Objective 2.3. By the end of Year 02, individuals involved in pre and post-analytical stages (DSHS laboratory, healthcare providers, and TNSP case management) will be surveyed using TxPEAT and other assessment strategies/tools created in Year 01 by the Steering Committee.

The method and technical approach for surveying will involve both web-based and paper-based approaches followed by random audit site visits and follow-up calls. The DSHS laboratory management is actively seeking a contractor to implement a web-based system, which will allow remote entering of newborn screening demographic information from the newborn screening test request, and provide healthcare providers access to final newborn screening results via the internet. The system will also have additional functionality for communication with program stakeholders and will enable web-based access to the surveys developed through this grant opportunity. The different survey methods and approaches will be employed in order to effectively communicate with over 4,300 healthcare professionals associated with newborn screening specimen collection, follow-up, and/or treatment. Contact information will be extracted from the Newborn Screening Case Management database, which includes contacts for family practitioners, pediatricians, neonatologists, phlebotomists, nurses, social workers, genetic

counselors, metabolic specialists, endocrinologists, hematologists, geneticists, dieticians, nutritionists and midwives. Important stakeholders not represented in the database, but who will likely be surveyed include parents, consumers, newborn screening advocacy groups, obstetricians, insurance representatives, medical associations, and Texas public education staff. To increase participation in survey responses, providers may be offered incentives with a monetary value not to exceed 15% of the total budget. Offered incentives will be approved by the steering committee.

Assessments requiring data extraction and evaluation will be developed, conducted, evaluated, and documented by DSHS statisticians experienced with medical data assessments. *Objective 2.4. By the end of Year 02, the Project Coordinator with the assistance of the Project Specialist will analyze data collected from the survey tools. The Steering Committee and NSQI will develop and finalize performance measures for evaluating pre and post-analytical stages of newborn screening.*

The Steering Committee and the NSQI will meet to review select proposed performance measures. A report will be produced listing the proposed measures, definition, and significance of measures, the data to gather to calculate the measure, the data collection procedure, and the goal of the measure.

Goal 3. Pilot Performance Measure: Pilot key performance measures for effectiveness of improving time to treatment in order to improve quality of life for newborns diagnosed with newborn screening disorders.

Objective 3.1. Plan, design, and execute a pilot study.

The methods and technical approach for these objectives that are to be completed in year three will be determined by the Steering Committee. Steering Committee will agree on selection of a consulting firm to assist with planning, designing, and executing a pilot study.

Goal 4. Identify and Publish Specific Interventions: By the end of Year 03, the Steering Committee will identify, recommend, and document interventions which are likely to improve the performance and address quality issues identified.

The methods and technical approach for objectives to be completed in year three will be determined by the Steering Committee, with the assistance of the NSQI and CDC staff advisors.

Project Team and Staffing

Susan Tanksley Ph.D., Manager of the DSHS Biochemistry and Genetics Branch within the Laboratory Services Section, will act as principal investigator for the grant. Dr. Tanksley has managed the Newborn Screening Program since 2005, and successfully implemented the recent Newborn Screening expansion project that increased the number of disorders screened from 7 to 27.

Another key person who will be the liaison to Newborn Case Management is Margaret Bruch, LCSW, Manager of the Health Screening and Case Management (HSCM) Unit at the Department of State Health Services (DSHS). Ms Bruch provides general oversight of case management and follow up activities performed in the Newborn Screening Program. Within HSCM, Ms. Bruch oversees three Branches- Texas Health Steps (the Early and Periodic Screening, Detection, and Treatment Program in Texas), Case Management (for Medicaid and children with special healthcare needs) and Newborn Screening. There are 13 programs administered within HSCM all focusing on preventive health for children in Texas. Ms. Bruch's

oversight of the Newborn Screening Branch includes efforts of Unit staff in the central office to notify, educate and guide families and healthcare providers when an abnormal screen is detected as well as the efforts of regional case management staff to locate and ensure appropriate follow up care for hard to locate children across Texas.

Also included as a resource for this project is Margaret Drummond-Borg MD, FAAP, FACMG. She is the consultant physician for the HSCU, which includes the Newborn Screening Program. She has 12 years of experience with the TNSP as the Division Director for nine years, the Branch Manager for one year and the Consultant Physician for two years.

The grant-funding request will include salaries for a full time Project Coordinator (Program Specialist VI) who will oversee grant activities and a Project Specialist (Program Specialist IV) who will assist the Coordinator to fulfill the deliverables and objectives of the grant. The Program Specialist VI will serve as Project Coordinator and will create and execute project work plans and revise as appropriate to meet changing needs and requirements utilizing project management tools such as Microsoft Project or similar. The coordinator will be knowledgeable in the field of laboratory medicine; have methodological expertise in statistics, experience with literature review, critical appraisal and guideline development, and will also have a practical understanding of problems faced with the delivery of diagnostic services and care for newborns.

The Program Specialist IV will assist the Project Coordinator to fulfill the deliverables and objectives of the grant. The Program Specialist IV will be knowledgeable in survey deployment, data management, project evaluation, and data entry skills.

Additional DSHS laboratory personnel who will assist in grant activities include two Newborn Screening Program Group Managers, the Newborn Screening Program Specialist, a

statistician, the Laboratory Customer Service Representative, and one administrative staff. Other supporting staff will include the general research services provided by the DSHS Medical and Research Library.

Measures of effectiveness to demonstrate accomplishment of program activities.

The measures of effectiveness that will be described in the subsequent narrative are related to the performance goal stated in the “Purpose” and in sum, state that the Texas Newborn Screening Performance Measures Project (TNSPMP) will link quality patient care with the quality of pre and post-analytical stages⁴ of the newborn screening process providing a self-sustaining model for systematic and continuous quality assessment. Overall, the goal of the three-year TNSPMP is to enhance the quality of life for newborns diagnosed with a newborn screening disorder through an improved newborn screening program.

Goal 1 and its three related objectives focus on the creation of a formalized Steering Committee. A 16-member Steering Committee will be established within the first three months of Year 01. The committee will create a charter with a mission and vision statement meeting at least quarterly for the life of the project. The measures of effectiveness for this goal and objectives include collection and documentation of all 16 signed agreements of Steering Committee members, documentation of all meetings including meeting minutes that coincide with the required quarterly schedule, and submission of the charter with mission and vision statements to the CDC.

⁴ For a general definition of the pre and post-analytical stages as it relates to this funding opportunity, see Appendix.

Quantifiable measures of effectiveness for Goal 2 and its four objectives focus on the data collection and analysis phase of the project. Goal 2 states that performance measures that reveal gaps in providing timely and effective treatment to infants diagnosed with disorders will be developed. The first two objectives will be met by the end of Year 01, while the latter two are scheduled for Year 02. The measures of effectiveness that will be produced and submitted to the CDC by the end of Year 01 include a report on available research on pre and post-analytical newborn screening activities providing evidence of best measures of efficient and effective treatment, and a performance evaluation and assessment toolkit that will be used to conduct a statewide assessment of the TNSP for the pre and post-analytical stages of newborn screening. The two other objectives under this goal include surveying individuals involved in the pre and post-analytical stages (to determine the level of adherences to performance standards in the Texas Performance Evaluation and Assessment Tool (TxPEAT) created in Year 01 by the Steering Committee and finalization of the performance measures that will be used to assess the pre and post-analytical stages of newborn screening. The quantifiable measures of effectiveness will be a report submitted to the CDC indicating the number of surveys sent out in the State of Texas, the types of locations they were sent to, and the response and rejection rate. Finally, a report on the final performance measures that will be piloted will also be sent to the CDC as evidence of accomplishment of this goal and its related objectives.

The measures of effectiveness for Goal 3 - Pilot Performance Measures include thorough documentation of the activities related to planning, designing, and executing the piloting of key performance measures for effectiveness of such measures on quality of life for newborns diagnosed with newborn screening disorders. These will be included in a report to the CDC that will include step-by-step documentation of the planning process, the schedule by which this goal

followed, the successful completion of milestones or benchmarks at each point in the schedule, the rationale for the design and listed alternatives, and the timeline for piloting the performance measures. The latter will include the number and types of contacts made with the pilot sites, the names of the pilot sites, and documentation of any audits that occur.

Finally, Goal 4 focuses on identification and publication of the specific interventions that are likely to improve the performance and address quality issues identified. The measure of effectiveness for this goal will be a written report to the CDC that outlines the interventions, identifies the selection/omission process, details evidence-based literature used to make the decisions, and enumerates the number of publications in preparation, under review, and accepted into appropriate peer-reviewed journals.

Timeline

The Texas Newborn Screening Performance Measures Project (TNSPMP) will engage in activities centered on the assessment of individual performance measures in three distinct phases over the course of three years: (Phase I) initial investigation and study design; (Phase II) data collection and analysis; and (Phase III) pilot-testing performance measures. The goals and related objectives of TNSPMP are organized to correspond with these phases. The timeline presented below describes the schedule for accomplishing the activities to be carried out to achieve the related goals and objectives by the end of Year 03.

2007	Sept	Oct	Nov	Dec	Jan	Feb	Mar	Apr	May	Jun	Jul	Aug
Year 1 Project Begins September	2007				2008							
Phase I – Initial												

Investigation and Study Design												
Start-Up												
Hire Project Staff												
OBJ 1.1. Steering Members												
OBJ 1.2. Charter												
OBJ 1.3. Meetings												
OBJ 2.1. Deficiencies												
OBJ 2.2. Tx PEAT												

2008	Sept	Oct	Nov	Dec	Jan	Feb	Mar	Apr	May	Jun	Jul	Aug
Year 2 Project Begins September	2008				2009							
Phase II – Data Collection and Analysis												
OBJ 1.3. Quarterly Meetings												
OBJ 2.3. Conduct Survey												
OBJ 2.4. Data												

Analysis													
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2009	Sept	Oct	Nov	Dec	Jan	Feb	Mar	Apr	May	Jun	Jul	Aug
Year 3 Project Begins September	2009				2010							
Phase III – Pilot-test Performance Measures												
OBJ 1.3. Quarterly Meetings												
OBJ 3.1. Pilot Study												
OBJ 4.1. Intervention												

Evaluation Plan

The Evaluation Plan described here will systematically determine the efficacy of the measures of effectiveness designed to demonstrate that program activities have been successfully accomplished. The evaluation plan includes specific methods that will be used to evaluate the accomplishment of the aforementioned goals and objectives over the course of three years.

The methods for evaluating the accomplishment of Goal 1 and its related three objectives address the initial investigation and study design phase; and include detailed documentation of who, what, when, where, and how often specific related activities occur. For example, Goal 1 states that a formalized steering committee be formed to direct the Texas Newborn Screening Performance Measures Project (TNSPMP) activities over three years. Related objectives of Goal

1 include setting up the Steering Committee with 16 members (and dedicated alternates) that will create a charter with a vision and mission statement. Both activities are to occur within the first six months of Year 01. The methods to evaluate these activities include detailed documentation of the following to be included in a report to the CDC: (1) the method by which potential Steering Committee members were chosen; (2) number of potential Steering Committee members to be contacted; (3) production of writing scripts including protocols and procedures for type and frequency of contacts made; (4) number refusals/accepted invitations to be on the Steering Committee; (5) number of contacts (e.g. e-mail, telephone, fax, voice-messages, face-to-face) made to potential Steering Committee members across the State of Texas and the date on which they occurred; (6) receipt of at least 16 letters of commitment from Steering Committee members with identified alternates; (7) list of relevant literature and other materials given to the Steering Committee for creation of a charter; (8) meeting agendas outlining activities conducted or steps taken by the Steering Committee to create vision and mission statements; (9) and final copy of the charter including a vision and mission statement. These methods will be employed to ensure that these activities are completed within the first six months of Year 01, as stated in the goals and objectives. Finally, additional methods used to evaluate the accomplishment of Goal 1, Objective 1.3. (i.e. By the end of Year 03, the Steering Committee will have met a minimum of quarterly throughout the life of the project) include completion of a sign-in and contact sheet by the Steering Committee members, documentation of meeting agenda items and meeting minutes, and creation of a schedule of meetings to occur over the course of three years that mandates quarterly meetings and documentation that meetings occurred according to this schedule.

The evaluation plan for Goal 2 and its four objectives includes methods related to data

collection and analysis. Goal 2 states that performance measures will be developed that reveal gaps in providing timely and effective treatment to infants diagnosed with disorders. The related objectives are split into activities to be accomplished by the end of Year 01 and those to be accomplished by the end of Year 02. Specifically, by the end of Year 01, a report on available research on pre and post-analytical newborn screening activities providing evidence of best measures on efficient and effective treatment will be produced. Methods to evaluate the accomplishment of this activity include, but are not limited to, inclusion of a comprehensive literature review, listing of resources used to determine literature review, inclusion of a narrative summary of results of literature review, and determination of best measures or practices based on the review of the literature. Documentation that the report has been delivered by the end of Year 01 through online document delivery service tracking (e.g. FedEx/Kinko's or the United Parcel Service of America UPS online tracking system) and confirmation of delivery to CDC will be the final method utilized to establish successful accomplishment of this activity. An additional activity to be completed by the end of Year 01 is refinement and completion of the Texas Performance Evaluation and Assessment Toolkit (TxPEAT). Methods used to determine successful completion of this activity include documentation of meeting minutes that describe salient topics covered by the Steering Committee as it uses the Performance Evaluation and Assessment Scheme (PEAS) as a conceptual framework for the TxPEAT. Meeting minutes will outline the process by which the Steering Committee arrived at the final TxPEAT instrument including rationale and alternatives considered. Recommendations on the final TxPEAT will be documented in the report sent to CDC by the end of Year 01.

Two additional activities to be completed by Year 02 are included in Goal 2. These include collecting data (i.e. surveys or data extractions) from individuals involved in the pre and

post-analytical stages (e.g. healthcare providers, DSHS laboratory staff and DSHS TNBP case management staff) to determine the level of adherence to performance standards in the TxPEAT. Methods to evaluate this activity include documentation of the following: (1) the number of paper-surveys sent and to whom they were sent; (2) the number of web-based surveys completed and who completed them; (3) number, type, and results of random audits of individuals providing a survey; and (4) number of rejections to complete survey and individual responsible for rejection. The final method of evaluation of this activity will be submission and confirmation of receipt of delivery of a narrative summary report to CDC. The second activity (i.e. the Steering Committee will finalize performance measures that will be used to assess the pre and post-analytical stages of newborn screening) will be evaluated using similar methods to those previously described including documentation of meeting minutes, rationale on the decision-making process, alternatives considered by the Steering Committee, finalized decision, and submitted and confirmed receipt of summary report to CDC.

The next component of the evaluation plan is the measurement of Goal 3 that corresponds to the pilot-testing phase. Goal 3 states that a pilot-test of key performance measures (i.e., the TxPEAT) will be conducted. Related to this, the Steering Committee will determine the policies and procedures by which the pilot test will be conducted (i.e. to whom, during what timeframe, and standards for monitoring implementation). The methods to evaluate this activity include documentation of meeting minutes where policies and procedures for conducting the pilot test are determined, creation of written procedures for conducting the pilot test, schedule for pilot test completion including primary activities and timeline, and documentation by program staff of all major activities related to implementing the pilot test (i.e. internal program meetings, key program decisions made, documentation of activities conducted toward implementation). Finally,

all activities and accomplishments will be included in the report to the CDC.

The final component of the evaluation plan is the identification and publication of the specific interventions that are likely to improve performance and address quality issues identified. A written report to the CDC will include the selection process of the evidence-based intervention (i.e. literature searches conducted, alternatives, rationale for final decision) and will include the number of publications in preparation, under review, and accepted into appropriate peer-reviewed journals.

Performance measures

Program Goal – To identify affected infants with newborn screening disorders and ensure timely, appropriate care to infants in need.

Measure	Data Collection Method	Data Collection By
The percent of screen positive newborns who received timely follow-up to definitive diagnosis and clinical management for condition(s) screened at the Texas Newborn Screening Laboratory.	Data extraction from newborn screening database.	Program Specialist VI, Program Specialist IV, and assistance by Case Management personnel
The number of days treatment started after a definitive diagnosis of a newborn screening disorder.	Data extraction from newborn screening database.	Program Specialist VI, Program Specialist IV, and assistance by Case Management

		personnel
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Goal 1. Steering Committee and Charter: A formalized steering committee consisting of subject matter experts and newborn screening stakeholders will be in place to direct project activities and efforts of the three-year Texas Newborn Screening Performance Measures Project (TNSPMP).

Measure	Data Collection Method	Data Collection By
Number of committee members who signed a letter of commitment.	Count letters of commitments.	Program Specialist VI, Program Specialist IV
Number of committee members who qualify and fit specified qualification criteria.	Gather resumes of each committee member and compare criteria with defined credentials and background qualifications.	Program Specialist VI, Program Specialist IV

Goal 2. Evidence-Based Performance Measures: Develop performance measures that reveal gaps in providing timely and effective treatment to infants diagnosed with disorders.

Measure	Data Collection	Data Collection By
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	Method	
Report produced discussing evidence of best measures for efficient and effective treatment where measures relate to pre and post-analytical newborn screening activities.	Collect and post on web for review by all stakeholders.	Program Specialist VI, Program Specialist IV
A finalized evaluation and assessment toolkit for pre and post-analytical newborn screening activities.	Collect and post on web for review by all stakeholders.	Program Specialist VI, Program Specialist IV
The number of Tx PEAT survey responses.	Use auto data transfer of survey data from web database to Microsoft Access. Manual entries into Microsoft Access database for written surveys. Count number of records in Microsoft Access Database.	Program Specialist VI, Program Specialist IV
The number of performance measures for pre and post-analytical newborn screening activities.	Count number and list proposed performance	Program Specialist VI, Program Specialist IV

	measures in Performance Measures Report.	
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Goal 3. Pilot Performance Measure: Pilot test key performance measures for effectiveness of improving time to treatment in order to improve quality of life for newborns diagnosed with newborn screening disorders.

Measure	Data Collection Method	Data Collection By
Contract with consulting firm to assist with pilot study.	DSHS Bidding Process	Program Specialist VI

Goal 4. Identify and Publish Specific Interventions: By the end of Year 03, the Steering Committee will identify, recommend, and document interventions which are likely to improve the performance and address quality issues identified.

Measure	Data Collection Method	Data Collection By
Number of documented interventions.	Count recommendations.	Program Specialist VI

Appendix

A. Quick Reference to 27 Newborn Screening Disorders

- Biotinidase Deficiency (BIOT) - BIOT is an enzyme deficiency that occurs in about 1 in

60,000 U.S. newborns and can result in seizures, hearing loss, and death in severe cases.

Treatment is simple and involves daily doses of biotin. (1)

- Congenital Adrenal Hyperplasia (CAH) – 21-Hydroxylase Deficiency - CAH is caused by decreased or absent production of certain adrenal hormones. The most prevalent type is detected by newborn screening in about 1 in 9,000 Texas newborns. Early detection can prevent death in boys and girls and sex misassignment in girls. Treatment involves lifelong hormone replacement therapy. (1)
- Congenital Hypothyroidism (CH) - Inadequate or absent production of thyroid hormone results in CH and is present in about 1 in 2,000 Texas newborns. Thyroid hormone replacement therapy begun by 1 month of age can prevent mental and growth retardation.(1)
- Galactosemia (GAL) – Galactose-1-Phosphate Uridyltransferase (GALT) Deficiency - Failure to metabolize the milk sugar galactose results in GAL and occurs in about 1 in 50,000 U.S. newborns. The classical form detected by newborn screening can lead to cataracts, liver cirrhosis, mental retardation and/or death. Treatment is elimination of galactose from the diet usually by substituting soy for milk products. (1)
- Homocystinuria (HCY) - HCY is caused by an enzyme deficiency that blocks the metabolism of an amino acid that can lead to mental retardation, osteoporosis and other problems if left undetected and untreated. The incidence is approximately 1 in 350,000 U.S. newborns. Treatment may involve a special restricted protein diet and supplemental medicines, including Vitamin B6. (1)
- Maple Syrup Urine Disease (MSUD) - MSUD is a defect in the way that the body metabolizes certain amino acids and is present in about 1 in 200,000 U.S. newborns.

Early detection and treatment with a special restricted protein diet can prevent death and severe mental retardation. There is an increased risk in Mennonites. (1)

- Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency - The most common disorder in the way the body metabolizes fatty acids is called MCAD deficiency. Undetected, it can cause sudden death. Treatment is simple and includes ensuring frequent food intake. The incidence from newborn screening is not yet known, but is thought to be approximately 1 in 15,000 U.S. newborns. (1)
- Phenylketonuria (PKU) - An enzyme defect that prevents metabolism of phenylalanine, an amino acid essential to brain development, is known as PKU and occurs in approximately 1 in every 23,000 Texas newborns. Undetected and untreated with a special restricted protein diet, PKU leads to irreversible mental retardation. Persons of European descent are at increased risk. (1)
- Sickle Cell Disease (SCD) – includes Sickle Cell Anemia (Hb SS), Sickle Beta Thalassemia (Hb S/?Th) and Sickle-Hemoglobin C Disease (Hb S/C) - Sickle cell anemia (Hemoglobin-SS-Disease) is the most prevalent SCD and causes clogged blood vessels resulting in severe pain and other severe health problems. Other common SCDs include Hemoglobin-SC-Disease and various thalassemias. Newborn screening detects about 1 in 2,500 Texas newborns with SCD annually. Persons of African or Mediterranean descent are at an increased risk. Early treatment with daily penicillin prevents death in the first few years of life. (3)
- Tyrosinemia Type I (TYR 1) - TYR is caused by a deficiency in the liver of one enzyme that breaks down tyrosine. If not treated, the condition causes severe liver disease and other serious health problems. Treatment consists of medication including vitamin D

and nitisinone, and a special restricted protein diet. The estimated incidence is 1 case in every 100,000 live births. (1)

- Other Fatty Acid Oxidation (FAO) Disorders include Carnitine Uptake Defect (CUD), Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD), Trifunctional Protein Deficiency (TFP) and Very-Long-Chain Acyl-Co A Dehydrogenase Deficiency (VLCAD) - Disorders besides MCAD deficiency, other FAO disorders may be detected through newborn screening. They are usually described in categories based on the length of the fatty acid involved. Undetected and untreated they can cause seizures, coma, and even death. Treatment may include a low fat diet, frequent food intake, supplementation with L-Carnitine (Carnitor) and medium chain triglycerides. The incidences of various FAO disorders are not known since it is only recently that early detection through newborn screening has occurred. (4)
- Organic Acid (OA) Disorders include 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC), Beta-Ketothiolase Deficiency (BKD), Glutaric Acidemia Type I (GAI), Hydroxymethylglutaric Aciduria (HMG), Isovaleric Acidemia (IVA) Methylmalonic Acidemia(MMA) (Cbl A and Cbl B forms) (Cbl A,B), Methylmalonic Acidemia (mutase deficiency form) (MUT), Multiple Carboxylase Deficiency (MCD) and Propionic Acidemia (PROP) - Organic acidemias are a group of metabolic disorders that lead to accumulation of organic acids in the blood and urine and may be detected in newborn screening through analysis of acylcarnitine profiles. Symptoms can be diminished by restricting protein in the diet and supplementation with vitamins and/or L-Carnitine. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known. (9)

- Urea Cycle Disorders (UCD) include Argininosuccinic Acidemia (ASA) and Citrullinemia (CIT) - A UCD is a genetic disorder caused by a deficiency of one of the enzymes responsible for removing ammonia from the blood stream. Some UCDs may be detected as a part of newborn screening. They are characterized by seizures, poor muscle tone, respiratory distress, and coma, and result in death if left undetected and untreated. Treatment is by a special restricted protein diet and medications including phenylbutyrate to remove ammonia. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known. (2)

B. Information on Treatment for Disorders Most Commonly Included in State Newborn Screening Programs (Sources: National Newborn Screening and Genetics Resource Center and newborn screening literature)

Disorder	Description	Potential Outcome	Treatment
Phenylketonuria	Deficiency of an enzyme needed to break down the amino acid phenylalanine	Mental retardation, seizures	Low-phenylalanine diet
Congenital hypothyroidism	Inability to produce adequate amount of thyroid hormone	Mental retardation, stunted growth	Thyroid hormone
Galactosemia	Deficiency of an enzyme needed to break down the milk sugar galactose	Brain damage, liver damage, cataracts, death	Galactose-free diet
Sickle cell diseases	Inherited blood disorder causing hemoglobin	Organ damage, delayed growth, stroke	Penicillin, vaccinations

Disorder	Description	Potential Outcome	Treatment
	abnormalities		
Congenital adrenal hyperplasia	Deficiency of an adrenal enzyme needed to produce cortisol and aldosterone	Death due to salt loss, reproductive and growth difficulties	Hormone replacement and salt replacement
Biotinidase deficiency	Deficiency of the enzyme biotinidase, needed to recycle the vitamin biotin	Mental retardation, developmental delay, seizures, hearing loss	Biotin supplements
Maple syrup urine disease	Deficiency of the enzyme needed to metabolize leucine, isoleucine, and valine	Mental retardation, seizures, coma, death	Dietary management and supplements
Homocystinuria	Deficiency of the enzyme needed to metabolize the amino acid homocysteine	Mental retardation, eye problems, skeletal abnormalities, stroke	Dietary management and vitamin supplements

C. Total Testing Process from, “Total testing process applied to therapeutic drug monitoring: impact on patients' outcomes and economics, Schumachera, Barr, 1998

Regulatory Environment

