



Texas Newborn Screening Performance Measures Project

DEPARTMENT OF STATE HEALTH SERVICES

Texas Newborn Screening Performance Measures Project

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Texas Department of State Health Services

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Table of Contents

Texas Newborn Screening Program	3
DSHS Texas NBS Program Organizational Structure	5
Texas Newborn Screening Program Process	6
Texas Newborn Screening System	7
Texas Newborn Screening Performance Measures Project	8
TNSPMP Purpose and Need	8
TNSPMP Goals and Objectives	10
TNSPMP Key Stakeholders and Project Team Members	13
Evidence-Based Approach to Quality Performance Measures	32
Evidence-Based Performance Measures for Newborn Screening ...	32
Types of Performance Measures	33
TNSPMP Meeting Agendas and Notes	35
Appendix	
Quick Reference Guide to Screening Disorders	
Texas Newborn Screening Panel	
2007 Newborn Screening Statistics	



Texas Newborn Screening Program

Newborn Screening (NBS) is a public health program that screens for a specific set of inherited disorders and hypothyroidism. In most instances, the parents are unaware of being “silent carriers” for the genetic condition, and the diagnosis in their newborn infant is unanticipated. If left untreated, the cost of these conditions is enormous in terms of human morbidity and mortality, suffering, and economic burden. Identification of these disorders and timely interventions can lead to the elimination or reduction of negative health effects. What began as a simple inexpensive screen for one metabolic disorder has grown into a six-part system involving screening, short term follow-up, diagnosis, treatment/management, and evaluation with an education process encompassing all stages. Using new technology and national guidance, screening programs now have the capability to test for more than 40 disorders.

Texas rules (Texas Administrative Code 25; Chapter 37.56) require two screenings per newborn; the first is recommended at 24-48 hours of age and the second at 1-2 weeks of age. Currently, the Texas Department of State Health Services (DSHS) laboratory receives approximately 800,000 newborn specimens annually and analyzes each specimen for 27 disorders.¹ This translates into more than 4.8 million tests performed each year making it the largest newborn screening laboratory in the country.

Approximately 16,000 abnormal results will be identified annually, and each will require that the Newborn Screening Follow-Up and Case Management staff provide initial contact and guidance to physicians. Newborn screening follow-up comprises short-term and long-term activities. Short-term follow-up includes notification of the abnormal screen by fax, letter, and phone to the medical providers and parent/legal guardian with suggestions on next steps. These notifications are then tracked until further laboratory test results are received. If necessary, phone calls are made to the provider to ensure that the infant has been seen by a healthcare professional and that a

¹ See appendix for the Quick Reference Guide to Screening Disorders discussing the current disorders tested at the Texas Department of State Health Services Laboratory.



determination has been made regarding management of the infant.

Newborn screening case management includes steps taken to locate hard-to-find infants and to provide the family with resources to ensure infants are seen by physicians. They also ensure completion of appropriate further testing as indicated, consultation with appropriate specialists, determination of diagnosis, and initiation of treatment. Case management staff may act as subject matter resource for the family, the primary care physician and specialist.

Long-term follow-up includes the tracking of diagnosed patients to monitor health outcomes. TNSP activities involve conducting routine physician questionnaires regarding continuity of care, gathering patient health and demographic information, and educating parents of diagnosed children.

From the 16,000 results identified as abnormal, over 600 hundred infants are expected to be diagnosed with one of the 27 disorders annually.² Early detection and management of these disorders caused by inherited genetic conditions (with the possible exception of Hypothyroidism) can prevent mental retardation and other catastrophic health problems in affected children. In most instances, the parents are unaware of being silent carriers for the genetic problem, and the diagnosis of a genetic disease in their newborn infant is unanticipated.

The goal of the TNSP is to decrease the morbidity and mortality of infants born in Texas through customer-oriented, high quality newborn screening laboratory analyses, follow-up, case management, and outreach education.

² For 2007 program statistics, see appendix 2007 Newborn Screening Statistics.



DSHS Texas Newborn Screening Program Organizational Structure

The TNSP is a program incorporated within two organizational divisions in the Texas Department of State Health Services. These are the Division of Prevention and Preparedness Services and the Division for Family and Community Health Services.

The Division of Prevention and Preparedness Services includes the Laboratory Services Section that conducts analytical screens or tests on the newborn screening specimens. The laboratory newborn screening website is <http://www.dshs.state.tx.us/lab/newbornscreening.shtm>.

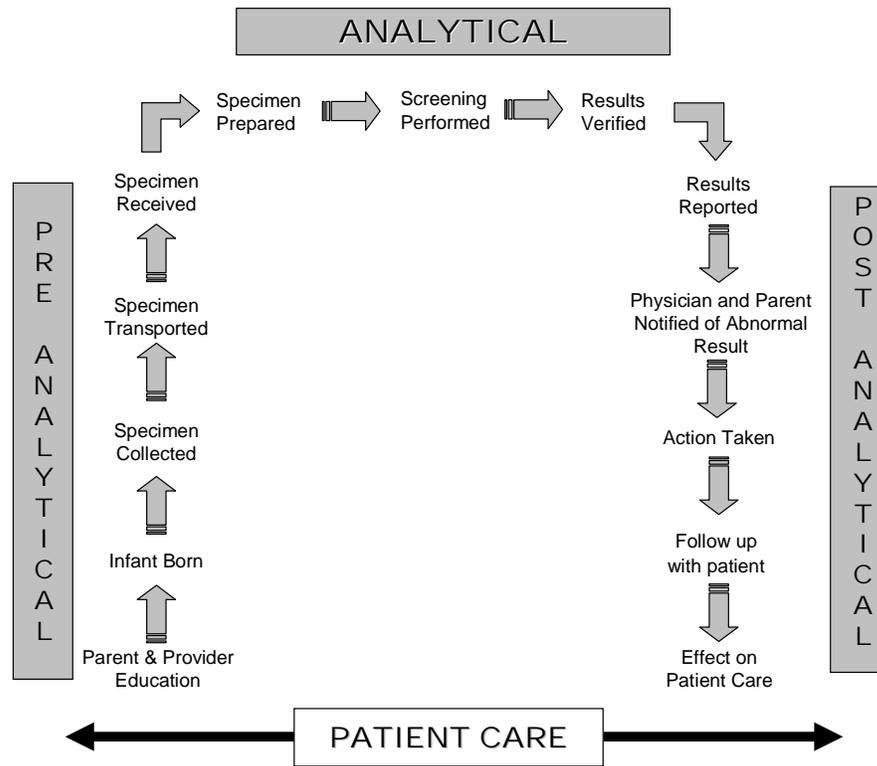
The Division for Family and Community Health Services includes the Specialized Health Services Section which provides newborn screening follow-up, case management and outreach education to parents, submitters, and providers. The case management newborn screening website is <http://www.dshs.state.tx.us/newborn>.



Texas Newborn Screening Program Process

The Texas Newborn Screening process encompasses several steps including: parent and provider education, collection of the blood specimens for the initial and second screen, specimen transport and receipt, specimen preparation, laboratory analysis of the blood specimens, case management or follow-up of abnormal screens to ensure diagnosis/treatment and long-term follow-up. Figure 1 depicts a *general* process involved with screening newborns.

Figure 1 Newborn Screening General Process Flow

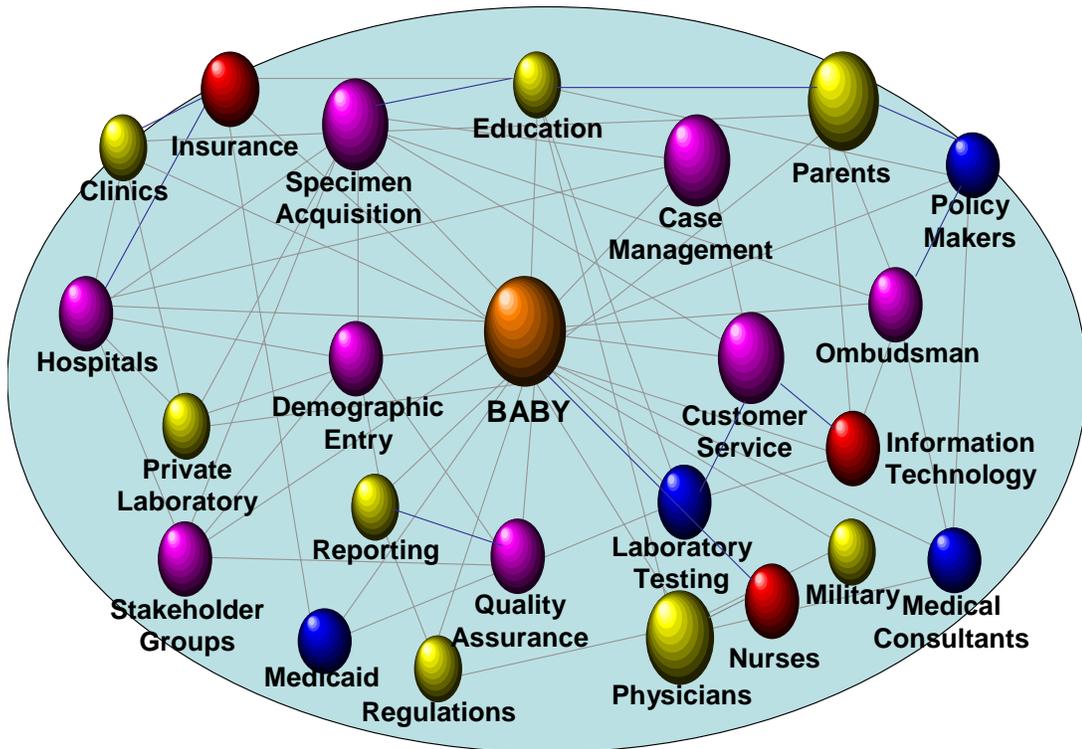




Texas Newborn Screening System

Newborn screening is more than laboratory testing. It is a coordinated and comprehensive system consisting of education, screening, follow-up, diagnosis, treatment and management, and program evaluation. The Texas Newborn Screening System consists of all the people, organizations, and entities that participate in serving all infants born in Texas and providing services to those who have screened positive for newborn screening disorders. Figure 2 is a *general* depiction of the various entities of the newborn screening system. This community includes parents, physicians, nurses, state laboratorians, case management and follow-up staff, prenatal educators, advocacy organizations, insurance carriers, state and federal funding partners, and researchers exploring new tests and treatments.

Figure 2 Newborn Screening System



Texas Newborn Screening Performance Measures Project

The Texas Newborn Screening Program (TNSP) was awarded a Centers for Disease Control and Prevention (CDC) three-year grant, titled “Evidence-Based Laboratory Medicine: Quality/Performance Measure Evaluation”, to support a project on developing evidence-based performance measures for newborn screening. This project is the Texas Newborn Screening Performance Measures Project (TNSPMP).

TNSPMP Purpose and Need

The size and scope of the TNSP creates unique challenges in identifying affected infants, and in ensuring timely, appropriate care to these infants. Texas is the largest state in the contiguous United States with the longest shared border in the country. Families often move back and forth across state lines and the Texas-Mexico border making it challenging to ensure that all children born in Texas are actually tested for the initial and second screen. TNSP fiscal year 2006 reporting of Title V³ data indicated that 96.9 percent of newborns had at least one newborn screen in that fiscal year. This indicates the possibility that roughly 12,000 newborns were not screened, and therefore, some with a newborn screening condition may have been missed.

Access to care can be a problem for families who live in rural Texas communities if they have an affected child. Parents sometimes travel hundreds of miles to pediatric sub-specialists for appropriate care for their newborn. In addition, with over 2,300 birthing facilities and healthcare practitioners submitting over 800,000 specimens each year, on-going educational efforts are always needed to address issues with specimen quality, timely submission and reporting, physician notifications, patient diagnosis, treatment, and on-going evaluation.

³ Title V is a Federal program initiated in 1935 that focuses on improving the health of all mothers and children. Title V has been amended over the years, though the underlying goal has remained constant: continued improvement in the health, safety, and well being of mothers and children.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT

The challenges previously noted increase risks to children identified with one of 14 disorders known to 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). Data has shown that infants confirmed with these high-risk disorders do not always receive timely treatment. For example, DSHS data from August 2003 through June 2005 show 50 percent of infants identified with SW-CAH were not started on a treatment regimen until after two weeks of life. Endocrinology specialists advocate treatment within two weeks of life to ensure positive outcomes for affected children. Treatment delays could result in serious consequences such as adrenal crises resulting in hospitalization and sometimes resulting in death.

The Texas Department of State Health Services is committed to improving performance standards for the Texas Newborn Screening Program (TNSP) and therefore proposed the Texas Newborn Screening Performance Measures Project (TNSPMP).

The goal of the TNSPMP is to develop a self-sustaining model for systematic and continuous quality assessment of pre and post analytical stages of the NBS system to improve health outcomes of children identified with a NBS disorder.



TNSPMP Goals and Objectives

GOAL 1 - TNSPMP PROJECT TEAM AND CHARTER

A formalized project team, consisting of subject matter experts and newborn screening stakeholders, will be in place to direct project activities and efforts of the three-year TNSPMP. The team consists of the TNSPMP Integrated Project Team, which includes DSHS NBS staff, and a select team of external stakeholders.

Objectives 1 - The project team will consist of 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, state and federal funding agencies, parents and subject matter experts.

Objective 2 - The project team will create a charter containing a vision and mission statement.

Objective 3 - The project team will meet face to face at least quarterly during the project period.

GOAL 2 - EVIDENCE-BASED PERFORMANCE MEASURES

Develop and define performance measures that reveal gaps/barriers to providing timely and effective treatment to infants diagnosed with newborn screening disorders.

Objective 1 - The project team will identify evidence-based deficiencies and barriers in the Texas Newborn Screening System and determine the direction and focus of the project.

General resources, literature, and publications that focus on evidence-based processes will be researched and reviewed by the project team. In addition to literature searches, the project team will review the National Newborn Screening and Genetics Resource Center (NNSGRC) recommendations resulting from a March 2005 review of the TNSP. A report will be produced summarizing the literature search findings and identifying outstanding issues documented by the NNSGRC review that are within scope of the TNSPMP.

Objective 2 - Based upon issues identified through the literature review and programmatic review summary, the TNSPMP team will develop assessment tools for evaluating the size and scope of pre and post analytical gaps/barriers. These tools will comprise the Texas Performance Evaluation and Assessment Toolkit (TxPEAT).

Examples of pre and post analytical areas of focus include prenatal education, specimen collection, specimen transport, short-term follow-up, medical management of conditions and outcomes, and follow-up support activities such as interpretation of test results, and communications between DSHS and healthcare providers.

The TNSPMP team will first identify and select specific pre and post analytical topics. The toolkit will then be tailored to these topics so that 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 physician notification of an abnormal screen, when confirmation tests were ordered, and when treatment commenced. Evaluating patient outcome over time may require tools to capture information from physician's notes and patient charts, and possibly information from the family.

Objective 3 - Individuals involved in pre and post analytical stages (e.g. health educators, healthcare professionals, medical specialists, and DSHS NBS staff) will be surveyed using the TxPEAT.

The survey will include web-based and paper-based approaches followed by random audit site visits and follow-up calls. Different survey methods and approaches will be employed to effectively gather input from approximately 4,000 healthcare professionals associated with newborn screening specimen collection, follow-up, and/or treatment. To increase participation in survey responses, providers may be offered monetary compensation for their efforts.

Objective 4 - Data collected from the survey tools will be analyzed. Results will be used to develop and propose performance measures for evaluating pre and post analytical stages of the newborn screening system.

A report will be produced listing the proposed performance measures, definition and significance of measures, the data needed to calculate the necessary measures, and data collection procedures.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT



GOAL 3 - PILOT PERFORMANCE MEASURES

The project team will plan, design, and execute a key performance measures pilot to test for effectiveness. The focus will be on evaluating performance, and how it ultimately relates to time-to-treatment and improved quality of life for newborns.

GOAL 4 - INTERVENTION RECOMMENDATIONS

The project team will identify, recommend, and document interventions that are likely to improve the performance of the newborn screening system and that address quality of care issues for infants diagnosed with newborn screening disorders.



TNSPMP Key Stakeholders and Project Team Members

The TNSPMP External Stakeholder Project Team was selected for its collective diverse perspective and technical expertise in order to provide input towards the development of performance measures for the newborn screening system. The diverse group of team members will provide varied perspectives in order to shape the development of sound performance measures used to help determine if the needs of diagnosed newborns and their families are being met.

It is a privilege to have influential and empowering stakeholders as part of this project. A warm “Thank you” is extended to the following individuals listed alphabetically below for their participation.

SANDRA BILLINGS

Sandra Billings became involved with the Congenital Adrenal Hyperplasia (CAH) community after her third son, Jackson, was diagnosed with Simple Virilizing CAH (SVCAH) in 2001. The initial newborn screen tested positive for Salt Wasting CAH (SWCAH). However, not entirely convinced, she sought help from renowned pediatric and adult endocrinologist Dr. Maria I. New, Professor of Pediatrics at the Mount Sinai School of Medicine in New York. Dr. New is credited for scientific breakthroughs in the study of CAH.

DNA tests proved both Ms. Billings and her husband were carriers of CAH. It was also diagnosed that her son Jackson had SVCAH instead of SWCAH, which was the original diagnosis from the initial newborn screening.

Two years later, Ms. Billings gave birth to another baby, Nolan, who also was diagnosed with CAH. Ms. Billings’ determination to seek the proper help, which required repeated visits to New York with her 3-month child, Jackson, better prepared her to handle Nolan’s case two years later.

Ms. Billings has authored and contributed articles to the CAH Organization of Texas (CAHOOT) newsletter, which is published by the Texas Department of State Health Services. The newsletter is designed to increase understanding among parents of children with CAH, physicians, and the public.

She has been actively involved with the CARES (Congenital Adrenal hyperplasia Research,

Education and Support) Foundation, Incorporated. She has served as a board member and on their research committee to find new and better treatments and she has also organized fund raisers to support the CARES Foundation.

Currently, Ms. Billings is a CARES Foundation support group contact for the Texas area, providing assistance and encouragement to parents who have just learned about their newborn's CAH diagnosis.

She lives in Houston, Texas with her husband of 15 years, Scott Billings, and their four sons, Braedon, 13; Rylan, 9; Jackson, 6; and Nolan, 4. At present, she is a stay-at-home mom working part-time in real estate. She would like to return to the workforce full-time when Nolan enters kindergarten and eventually open a real estate office with plans to expand it into a franchise.

GEORGE R. BUCHANAN, MD

George R. Buchanan, M.D., is Professor of Pediatrics at The University of Texas Southwestern Medical Center at Dallas. He holds the endowed Distinguished Chair in Pediatric Oncology and Hematology at UT Southwestern, and he is Medical Director of the Center for Cancer and Blood Disorders at Children's Medical Center of Dallas.

Dr. Buchanan received his M.D. degree at the University of Chicago and did his pediatric hematology-oncology training at Children's Hospital in Boston. He has been in Dallas since 1977. His interests include sickle cell disease and other anemias and bleeding disorders. He has served as Chair of the Subboard of the American Board of Pediatrics and as President of the American Society of Pediatric Hematology/Oncology. He is a member of the Executive Committee of the American Society of Hematology. He is also Director of the Southwestern Comprehensive Sickle Cell Center.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT

DONNA CLAEYS, BSN, RN

Donna Claeys, B.S.N., R.N., has extensive experience in the nursing field spanning over three decades. Ms. Claeys has worked with State, Military (federal), and City hospitals, Home Health, and Medicaid as both staff nurse and manager.

Her direct patient care experience includes neonatal intensive care, newborn care, pediatric oncology, obstetrics, and home health. Her indirect patient care experience includes Texas Health Steps–Comprehensive Care (THSteps-CCP) Medicaid Home Health prior authorizations, medical policy development with the Medicaid vendor, and medical policy development with the HHSC Medicaid/CHIP Office of the Medical Director. Ms. Claeys recently served as a member of an investigational review board for a new medical device designed to record heart rate variability. The research associated with this device is being used to develop a data base for standardizing recorded data.

Ms. Claeys received a Bachelor of Science in Nursing from The University of Texas Medical Branch in Galveston, Texas.

L. MARGARET DRUMMOND-BORG, MD

L. Margaret Drummond-Borg, M.D., Consultant Physician, Health Screening and Case Management Unit, has been involved with the Texas Newborn Screening Program since 1996. She has also worked with the Newborn Screening Program in New Zealand for five years. Dr. Drummond-Borg is board certified in pediatrics and clinical genetics.

Dr. Drummond-Borg received her medical degree from the University of Otago Medical School, New Zealand. Her genetic fellowships were completed in New Zealand and at the University of Washington, Seattle, and she did her pediatric residency in the Hospital for Sick Children, Toronto, Canada. Dr. Drummond-Borg has worked as a clinical geneticist in Texas, Washington, Arkansas, and Auckland, New Zealand. She has worked as a Primary Care Physician in New Zealand, England and Saudi Arabia.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT



A L I C E K G O N G , M D

Alice K Gong, M.D., is a Professor of Pediatrics at The University of Texas Health Science Center at San Antonio (UTHSCSA), Department of Pediatrics.

Dr. Gong received her M.D. degree at the University of Mississippi Medical Center and did her neonatal-perinatal medicine training at the State University of New York in Buffalo.

Dr. Gong has received many honors and awards during the course of her career, which spans over 30 years. Recently, she received a special achievement award, presented by the American Academy of Pediatrics for distinguished service and dedication to the mission and goals of the academy for her tireless efforts related to expanding the newborn screening program. She was also recently given the J.T. “Lamar” McNew Award by the Texas Medical Association to honor her outstanding service to residents and fellows in Texas.

J O S E L . G O N Z A L E Z , M D

Jose L. Gonzalez, M.D., serves as Vice-Chair for Pediatric Medical Education at The University of Texas Medical Branch in Galveston, overseeing all Department medical education activities. Dr. Gonzalez completed his pediatrics residency, including Chief Residency, at UTHSC-Southwestern and fellowship training in Pediatric Endocrinology and Diabetes at the University of Iowa in 1982. He received a J.D. degree from SMU School of Law in 1997 and a Masters of Science in Medical Education from the University of Southern California in 2002.

C H A R L E T A G U I L L O R Y , M D

Charleta Guillory, M.D., F.A.A.P., is an Associate Professor of Pediatrics in the Section of Neonatology at Baylor College of Medicine, Associate Director of Level II Nurseries at Texas Children’s Hospital and Director of Texas Children’s Hospital Neonatal-Perinatal Public Health Program. She earned her Bachelor of Science degree from the University of Southwestern Louisiana, her doctor of medicine degree from the Louisiana State University Medical School, completed her pediatric residency at Louisiana State Medical Center and the University of Colorado, and received her post-doctoral fellowship training in neonatal-perinatal medicine at Baylor College of Medicine. Board certified in both pediatrics and neonatal-perinatal medicine, Dr. Guillory is completing a Master of Science degree in public health at the University of Texas School of Public Health.

Her main research focus addresses the issue of decreasing infant mortality and morbidity. Her leadership in advocacy efforts with the State Children's Health Insurance Program and in community-based initiatives led to her receiving the National March of Dimes Award of Distinction and, in 2001, she received the Mary Owen Greenwood award, the chapter's most prestigious volunteer award, for her dedication and commitment to improving the lives of babies. In addition to her membership in the Texas Gulf Coast Chapter of the March of Dimes Birth Defects Foundation Board of Directors and Executive Committee, she is co-chair of the Texas Pediatric Society Fetus and Newborn Committee and appointed to the Legislative Committee, the Academy of Pediatrics Perinatal Section, and named a Fellow of the American Academy of Pediatrics. Dr. Guillory has written numerous textbook chapters in Contemporary Diagnosis and Management of Neonatal Respiratory Diseases, co-edited several issues of Infant Mortality Update, participated in numerous radio and television programs related to the health of women and children, and was chairperson of the March of Dimes Perinatal Health Needs Assessment (1995-1997). ABC/Channel 13 selected her for Houston's Top Women of Distinction Award, and received the City of Houston's Mayor Award for Outstanding Volunteer Service in Health. Dr. Guillory was one of six physicians to receive the 1999-2000 Robert Wood Johnson Health Policy Fellows Award from The Institute of Medicine of the National Academy of Sciences for Outstanding Health Science Professionals. As a recipient of the award, she served as a legislative assistant in the United States Senate (office of Senator John B. Breaux D-LA) promoting both health policy legislation and programs. Dr. Guillory received the 2003 Baylor College of Medicine Fulbright and Jaworski Faculty Excellence Award for Teaching and Evaluation and in 2004; she received the Fulbright and Jaworski Faculty Excellence Award for Educational Leadership. The following year she was named the 2005-2006 March of Dimes Leadership Volunteer of the Year for the State of Texas. In 2006, Dr. Guillory was named as one of "America's Top Pediatricians" by Consumers' Research Council of America and listed in their, "Guide to America's Top Pediatricians" and, she is also listed in The Best Doctors in America. This year she was selected by the One-Star Foundation to receive the Governor's Volunteer Award in the "Community Connector – Individual" Category.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT



C H E R Y L H E R M E R A T H , M B A , D L M (A S C P) , R M (N R M)

Cheryl Hermerath, M.B.A., D.L.M. (ASCP), R.M. (NRM), is the Manager for the Northwest Regional Newborn Screening Program for the Oregon Department of Human Services, Public Health Laboratory, recently in Hillsboro, Oregon.

Ms. Hermerath has been actively involved in the Newborn Screening community for over 10 years. She currently holds the Chair position for the Association of Public Health Laboratories (APHL) Newborn Screening and Genetics in Public Health Committee. Since 2006, she has been participating in a workgroup with the National Newborn Screening and Genetics Resource Center (NNSGRC) and Association of Public Health Laboratories (APHL) organization to address protocol development for routine second testing in newborn screening. She is a member of the Western States Genetic Services Collaborative Committee and the APHL Newborn Screening & Genetics in Public Health Committee. Ms. Hermerath contributed to a NNSGRC March 2005 review of the Texas Newborn Screening Program that provided observations and recommendations to the program.

Ms. Hermerath earned a Masters in Business Administration at the University of Phoenix and a Bachelors of Science in Microbiology at the Northern Arizona University in Flagstaff, Arizona.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT

SCOTT MCLEAN, MD

Scott McLean received his M.D. degree at Uniformed Services University of the Health Sciences and did his pediatric training at the Madigan Army Medical Center in Tacoma, Washington. He completed a Clinical Genetics Fellowship at Children's National Medical Center in Washington, D.C., in 1993.

Over the course of his career, Scott has served on a number of newborn screening committees and work groups. He has been an advisor to the Texas Newborn Screening Program since 1993. In 2002, Scott participated in the Texas Department of Health Tandem Mass Spectrometry Advisory Group. He served as an ex officio representative on the American College of Medical Genetics Expert Group on Newborn Screening. He currently is a member of the Newborn Metabolic Screening Integrated Project Team for the Department of Defense. He is also a member, ex officio, of the Secretary's Advisory Committee on Genetics, Health, and Society—representing the Department of Defense. For the Subcommittee on Follow-up and Treatment of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, he served as an advisor to the development of a 2007 white paper titled, "The Road Map to Long-Term Follow-Up and Treatment in Newborn Screening."

He has held several assignments with the military. In 2004 and 2005, Scott served as the DISCOM Surgeon, 1st Cavalry Division in Taji, Iraq. Since 1997, he has been on the medical staff at Brooke Army Medical Center and Wilford Hall Medical Center in San Antonio. Dr. McLean is the Clinical Genetics Consultant to the Surgeon General of the United States Army, Clinical Associate Professor at The University of Texas Health Sciences Center in San Antonio, and Assistant Professor of Pediatrics for the Uniformed Services University of the Health Sciences, F. Edward Hebert School of Medicine at Bethesda, Maryland.

FRANCISCO RAMIREZ

Francisco Ramirez is currently a clinical social worker/supervisor for Service Access at Border Region Community MHMR. As certified through the licensing board of Texas, he is a Licensed Master Social Worker Advanced Practitioner (LMSW-AP). He is a graduate of St. Edwards University and has over 30 years experience as a social worker in a border community.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT



JOHN SAITO, MD

John Saito, M.D., is the Chief of Pediatric Pulmonology and Director of Pediatric Cystic Fibrosis at the Children's Hospital at Scott & White in Temple, Texas.

Dr. Saito earned his medical doctorate at the Temple University School of Medicine in Philadelphia, Pennsylvania. He conducted his pediatric residency at the Orlando Regional Health System in Florida and completed his pediatric pulmonology fellowship at the University of North Carolina at Chapel Hill.

Dr. Saito's area of special expertise involves infant pulmonary function test and flexible bronchoscopy. He also has special knowledge in longitudinal evaluation of lung physiology in infants with bronchopulmonary dysplasia. His current research project at Scott and White involves neonatal sweat testing for early diagnosis of cystic fibrosis.

EILEEN SHERIDAN-SHAYEB, MD

Eileen Sheridan-Shayeb, M.D., has been the Director of Ambulatory Pediatrics at the Medical Center Hospital Family Health Center since 1996. Previously, Dr. Sheridan was the Director of Ambulatory Pediatrics at the Texas Tech Health Science Center in Odessa. Dr. Sheridan earned her M.D. at the Universidad Autonoma de Neuvo Leon and her Masters in Public Health at The University of Texas at Houston.



REID SUTTON, MD

Reid Sutton, M.D., is board certified in both clinical genetics and clinical biochemical genetics and provides medical care for individuals with inborn errors of metabolism in the Texas Children's Hospital Metabolic Clinic in Houston, Texas. Dr. Sutton is Assistant Professor of Molecular and Human Genetics at Baylor College of Medicine in Houston, Texas and also serves as the Medical Director of the Biochemical Genetics Diagnostic Laboratory. He is the Director of both the Medical Genetics Residency Program and the American Board of Medical Genetics Diagnostic Laboratory training programs in clinical biochemical genetics, clinical cytogenetics and clinical molecular genetics. Dr. Sutton was raised in Kentucky and attended Transylvania University and the University of Kentucky College of Medicine. He did his pediatric residency at Washington University/St. Louis Children's Hospital in St. Louis and received training in Medical Genetics and Clinical Biochemical Genetics at Baylor College of Medicine. Dr. Sutton is a member of the Society of Inherited Metabolic Diseases, American College of Medical Genetics, American Society of Human Genetics and the International Skeletal Dysplasia Society.

LARRY SWEETMAN, PHD

Larry Sweetman, Ph.D., serves as the Director for the Mass Spectrometry Unit at the Institute of Metabolic Disease, Baylor University Medical Center/Baylor Research Institute in Dallas.

Over the course of his extensive career, Dr. Sweetman has authored and contributed to numerous peer-reviewed publications related to newborn screening disorders. He was a consultant to the Oklahoma State Department of Health to facilitate the provision of quality follow-up services for newborn tandem mass spectrometry screening. He was also a consultant to the Texas Department of State Health Laboratory Services during the Texas Newborn Screening Expansion.

Dr. Sweetman earned a Bachelor of Arts degree in Chemistry from the University of Colorado and a doctorate in Biochemistry from the University of Miami.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT



BRADFORD THERRELL, JR., PHD

Bradford Therrell, Jr., Ph.D., is the director of the National Newborn Screening and Genetics Resource Center (NNSGRC) and a professor in the Department of Pediatrics at The University of Texas Health Science Center at San Antonio. He previously worked at the Texas Department of Health Bureau of Laboratories for over 28 years, where he was in charge of newborn screening and other biochemical screening programs. Dr. Therrell was one of the founding members of the International Society for Neonatal Screening serving as its first Secretariat and later as President.

Dr. Therrell was awarded the prestigious Guthrie Award by the Society for outstanding international contributions in newborn screening. He has been actively involved in providing reviews and expert advice to newborn screening programs in the U.S. as part of the activities of the Health Resources and Services Administration and in assisting developing countries as part of the activities of the International Atomic Energy Agency.

SISTER MARY NICHOLAS VINCELLI, BS, RN, MA

Sister Mary Nicholas Vincelli, B.S., R.N., M.A., is a member of the Sisters of St. Joseph of Carondelet in St. Paul, Minnesota. Sister Nicholas earned her Master of Arts in Medical-Surgical Nursing from State University of Iowa and her undergraduate degree from the College of St. Catherine in St. Paul, Minnesota.

Sister Mary Nicholas's advocacy efforts and dedication to serving her community has led her to receiving numerous awards over the course of her extensive career. Among many, she was a recipient of the Distinguished Service Award for Exceptional Service to the Health and Welfare of Texas Children by the Texas Pediatric Society in 1987, the "Outstanding U.S./Mexico Border Health Professional" Award by the U.S./Mexico Border Health Association in 1986, the National Perinatal Award for individual contribution to Maternal Child Health in 1997, the Sounds of Texas Excellence Award to find babies with hearing loss in 1998, and the Community Service Helping Hands Award by the Texas Department of Health in 1999. She was also named Outstanding Public Health Nursing Leader of the Year in 2000 for the US Health and Human Services Region VI.

Sister Nicholas has also served in countless appointments as board member and chairperson of committees and advisory boards for organizations that serve the public health of children in Texas. She was a Signer of Articles of Incorporation and Board Member of Rio Grande Health and

Medical Services, Inc in 1989. She served as an Advisory Committee Member for the Early Childhood Intervention Program from 1981 to 1994. She was also a member of the Child Fatality Review Committee and the Migrant Medical Advisory Board from 1996 to 2002. Sister Nicholas is currently and has been a long standing Board Member of Texas Perinatal Association since 1996. In 2003, she served as Vice President for the organization.

Sister Nicholas was the Director of Nursing Services for the Texas Department of Health Public Health Region 11 in Harlingen, Texas from 1990 to 2002. She is currently retired.

MORGAN WALTHALL, MSW

Morgan Walthall, M.S.W., is the Texas State Director of Public Affairs for the March of Dimes. As director, Ms. Walthall works with March of Dimes volunteers to develop and implement the Texas chapter's advocacy efforts on issues related to birth defects, newborn screening, immunization and access to affordable health care for children and pregnant women.

Since joining the organization in 2005, Ms. Walthall has been involved with the implementation of House Bill 790, related to the newborn screening expansion. Ms. Walthall was instrumental in ensuring that biotinidase was included in the expanded panel.

Recently, Ms. Walthall lead March of Dimes advocacy efforts to add language to point of sale warning signs posted where tobacco products are sold—specifically to warn pregnant women of the risks of smoking. Ms. Walthall also worked closely with the Texas CHIP Coalition, successfully advocating for policy changes to CHIP that will improve access to the program by both eliminating the 90-day waiting period and allowing for 12 months of continuous eligibility.

Ms. Walthall also works as a state liaison for the March of Dimes Office of Government Affairs. Her responsibilities include establishing relationships with congressional district offices and responding as needed to ensure members of Congress are aware of March of Dimes advocacy efforts in Washington, D.C. Ms. Walthall worked hand-in-hand with the March of Dimes Office of Government Affairs to ensure the passage of the PREEMIE Act, which was signed by the President in late 2006. Ms. Walthall has also worked with Texas congressional members to secure their support of the Newborn Screening Saves Lives Act.

Prior to working for the March of Dimes, Ms. Walthall was the Legislative Aide for State Representative John E. Davis during the 2005 Texas Regular Legislative Session. Ms. Walthall worked on issues related to both public and mental health.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT



Ms. Walthall completed her undergraduate studies in psychology and counseling at Southwest Baptist University in Bolivar, Missouri. She then received her Master's of Social Work from the University of Texas at Austin.

Ms. Walthall lives in Austin, Texas and enjoys playing tennis and soccer and spending time with her friends and family.

DON P. WILSON, MD

A native of Newton, Mississippi, Don P. Wilson, M.D., attended the University of Southern Mississippi, receiving a degree in pre-medical education. He earned a medical degree from the University of Mississippi before completing residency training and post-graduate fellowship in Pediatric Endocrinology at Baylor College of Medicine in Houston, Texas. He is board certified in Pediatrics and Pediatric Endocrinology.

Throughout his medical career, Dr. Wilson has devoted himself to clinical service, professional education, and medical research. Most of his professional focus has been in the area of childhood diabetes. His clinical skills and knowledge have brought hope and support to hundreds of children and their families affected by diabetes.

In 1997, Dr. Wilson was selected as Chairman of the Department of Pediatrics, Texas A&M University College of Medicine. In addition to a busy, comprehensive clinical service, the Department has an active medical research program and a post-graduate educational program in general pediatrics.



JERALD L. ZARIN, MD

Jerald L. Zarin, M.D., is an experienced managed care medical director and board certified pediatrician. He received his medical degree from the Albert Einstein College of Medicine, completed his pediatric training at Rainbow Babies and Children's Hospital/Case Western Reserve University, and earned his Master in Business Administration degree from Houston Baptist University.

In addition to his longtime pediatric practice experience in solo and small and large group environments, Dr. Zarin has been a managed care medical director for for-profit and non-profit HMOs with both commercial and Medicaid/SCHIP products. He is currently regional medical director for Blue Cross Blue Shield of Texas—a division of Health Care Services Corporation.

He has served on many Texas Health and Human Services Commission committees: SB1165 Work Group for Complex Children with Special Healthcare Needs; Medicaid/CHIP Medical Directors Committee; Medical Home Workgroup; and both Harris and Travis Service Areas Medicaid/CHIP Regional Advisory Committees.

Dr. Zarin is the longtime editor of the newsletter of the Section on Administration and Practice Management of the American Academy of Pediatrics and was a member of the Executive Committee of the Section. He is a member of the Editorial Board of the AAP Pediatric Practice Management Online website. He is also a member of the Managed Care Committee of the Harris County Medical Society, and the Practice Management and Children with Special Needs Committees of the Texas Pediatric Society.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT



The TNSPMP Integrated Project Team (DSHS staff) is listed alphabetically below.

MARGARET BRUCH, LCSW

Margaret Bruch, L.C.S.W., is the Manager of the Health Screening and Case Management Unit at the Department of State Health Services. In this role, she manages various programs relating to health care and benefits for children such as: vision, hearing, spinal screening; genetics; newborn screening and newborn hearing screening, amplification services for children, case management; public health dental and THSteps medical and dental benefits', outreach and informing and administrative services.

Ms. Bruch earned her graduate degree in social work and Bachelor of Arts degree at the University of Texas at Austin.

SHERRY CLAY, MA

Sherry Clay, M.A., serves as the Quality Assurance Unit Manager for the Laboratory Services Section of the Department of State Health Services. The Quality Assurance Unit is responsible for the pre and post analytical aspects of laboratory testing (specimen acquisition and result reporting), as well as quality assurance, employee safety and environmental compliance and management of the unique mechanical systems required for a laboratory building. Early in her career she worked in Pediatric Endocrinology at Baylor College of Medicine, worked as part of a research group at the Veterans Administration Hospital in Houston, Texas, served as Quality Assurance Manager for companies manufacturing medical devices and investigational new drugs. She has been worked in the DSHS laboratory for the past 21 years.

MIRSA DOUGLASS, MBA

Mirsa Douglass, M.B.A., is the Project Manager for the Texas Newborn Screening Performance Measures Project. In 2006, she was appointed as the liaison between newborn screening laboratory staff and PerkinElmer to ensure that proper software and hardware upgrades were installed for the Newborn Screening Expansion project.

Ms. Douglass was a nominee for the Joseph N. Murphy Award, which recognizes excellence in customer service for employees within the Department of State Health Services Laboratory Services Section. During her five years at the public health laboratory, she worked as an analytical

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT

chemist and as a project specialist. Ms. Douglass contributed to various projects, including the Texas Influenza Outreach project, Texas Public Health Laboratory System Assessment project, Newborn Screening Expansion project, and Public Health Laboratory Information Management System implementation project.

Ms. Douglass earned a Bachelor of Arts in Chemistry from Austin College in Sherman, Texas and a Masters in Business Administration from the University of Dallas in Arlington.

ELDRIDGE HUTCHESON, PHD

Eldridge Hutcheson, Ph.D., serves as the Laboratory Operations Manager for the Texas Department of State Health Services, where he is responsible for all testing in the Austin laboratory, including newborn screening. Previously, he was Director of Biochemistry and Genetics, which included the Newborn Screening Laboratory at the former Texas Department of Health.

Dr. Hutcheson received a Ph.D. in Chemistry from The University of Texas at Austin and did his postdoctoral work at the University of Tennessee Medical School where he subsequently became Assistant Professor of Biochemistry. He has been with Veterans Administration Medical Centers, both performing research and as Chief of Biochemistry in the pathology laboratory, and with National Health Laboratories (now LabCorp).

DAVID MARTINEZ

David Martinez serves as the Branch Manager for the Newborn Screening Case Management program which is responsible for follow-up of abnormal newborn screens for genetic disorders and hearing loss. Mr. Martinez successfully administered the expansion of the Newborn Screening Case Management/Follow-up program in 2006. Through the expansion, the program increased the panel of disorders requiring follow up from seven genetic disorders to 27 disorders.

Previously, Mr. Martinez managed programs for the Texas Workers' Compensation Commission. This included the Medical Quality Review program that is responsible for reviewing the quality of care provided to injured workers in the workers' compensation system. Mr. Martinez also managed the Medical Dispute Resolution program, designed to help medical providers seek resolution of disputes regarding the medical necessity of treatment provided, preauthorization disputes, and fee-related disputes in the workers compensation system. Mr. Martinez successfully

implemented the use of Independent Review Organizations in the Medical Dispute Resolution program. Mr. Martinez has 17 years of state government experience.

Mr. Martinez earned a Bachelor of Arts degree in Sociology from The University of Texas at Austin.

JANN MELTON-KISSEL, R.N., M.B.A

Jann Melton-Kissel, R.N., M.B.A., is the Director of Specialized Health Services Section in the Division for Family and Community Health Services at the Texas Department of State Health Services (DSHS). In this role, she oversees the following programs: Children with Special Health Care Needs Services Program, Kidney Health Care Program, Hemophilia Assistance Program, Donor Education and Awareness Registry Program, Texas Health Steps – Medical and Dental, Oral Health, Case Management, Newborn Screening Case Management and Follow-Up, Newborn Hearing Screening, Vision, Hearing and Spinal Screening (in schools), Genetics, and the Program for Amplification for Children in Texas. Ms. Melton-Kissel is also one of the sponsors for the Texas Newborn Screening Performance Measures Project.

Ms. Melton-Kissel earned a Bachelor of Science in Nursing Degree at The University of Texas at Austin and a Master of Business Degree at the University of Central Texas (Farlton State).

Before coming to the DSHS, Ms. Melton-Kissel worked for 13 years at an urban teaching hospital (Austin, Brackenridge Hospital) in obstetrical nursing. Her proudest achievement is when she co-opened the first hospital-based alternative birthing center in Texas in 1979. Ms. Melton-Kissel's TDH/DSHS career began in the mid-1980's in the regulatory arena, surveying free standing birthing centers for compliance with state regulations and quality of service. She also surveyed hospitals, end stage renal disease facilities, home health agencies, abortion facilities, and ambulatory surgery centers.

Ms. Melton-Kissel currently serves as a Major in the Texas State Guard Medical Brigade (Medical Reserve Corps).

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT

SUSAN NEILL, PHD, MBA

Susan Neill, Ph.D., M.B.A., is the Director of the Laboratory Services Section at the Texas Department of State Health Services (DSHS). Dr. Neill oversees and directs the activities of the laboratories including Microbiological Sciences, Environmental Sciences, Biochemistry & Genetics, Emergency Preparedness, Quality Assurance, Women's Health Laboratory, and the South Texas Laboratory. During her 21-year career with DSHS and the legacy Texas Department of Health, she has served in various management roles in the laboratory including managing the newborn screening area. Dr. Neill is one of the sponsors for the Texas Newborn Screening Performance Measures Project. She also was instrumental in securing newborn screening expansion services for the State of Texas which was officially launched in February 2007.

Dr. Neill earned her Ph.D. in Microbiology at the University of Illinois at Urbana-Champaign in 1982. She also earned her M.B.A at St. Edward's University in Austin, Texas in 1998. She received her undergraduate degree in Microbiology from The University of Texas at Austin.

Dr. Neill is actively involved in various organizations. Currently, she is the Secretary-Treasurer for the Association of Public Health Laboratories. Dr. Neill is also a member of the American Board of Bioanalysts American Society for Microbiology and the Pan American Society for Clinical Virology.

SHARON NEWCOMB-KASE, BA, LBSW, MSW

Sharon Newcomb-Kase B.A., L.B.S.W., M.S.W., serves as the Ombudsman in the Newborn Screening Support Group, Texas Department of State Health Services. She received a Bachelor of Arts degree with a major in Sociology from The University of Texas, Austin, and a Master's of Social Work degree from Western Michigan University.

Ms. Newcomb-Kase recently served as the Technical Coordinator for the Community Resource Coordination Groups (CRCGs) and the Texas Integrated Funding Initiative (TIFI) in the Office of Program Coordination for Children and Youth, Texas Health and Human Services Commission. Ms. Newcomb-Kase has worked for the Michigan Department of Community Health in the Mental Health Services to Children and Families division.

Ms. Newcomb-Kase has provided genetic counseling and genetic case management services in the western region of the state of Texas, having worked for Cook Children's in Ft. Worth and the legacy agencies of TX Mental Health and Mental Retardation and the TX Department of Health.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT



SUSAN TANKSLEY, PHD

Susan Tanksley, Ph.D., is the Biochemistry and Genetics Branch Manager in the Laboratory Services Section of the Texas Department of State Health Services in Austin. She manages the day-to-day operations of the Biochemistry and Genetics Branch which encompasses the state newborn screening and clinical chemistry laboratories. These high-volume testing areas process 4,500-5,000 specimens per day.

Dr. Tanksley's leadership and direction were instrumental in the successful expansion of newborn screening testing and database management from seven to 27 disorders. The expansion was implemented on an aggressive timeline and officially launched in February 2007.

Previously, Dr. Tanksley served as the DNA Diagnosis Team Lead, for the Texas Department of Health/Texas Department of State Health Services. Using her technical expertise, she managed a human genetic testing laboratory and developed new tests to identify mutations causing newborn diseases.

Dr. Tanksley has 14 years of hands-on laboratory testing experience and seven years of newborn screening experience. Dr. Tanksley earned her doctorate in Genetics from Texas A&M University.

TEXAS NEWBORN SCREENING
PERFORMANCE MEASURES PROJECT



DONNA WILLIAMS

Donna Williams is a Program Specialist for the Newborn Screening Laboratory at the Texas Department of State Health Services. Recently, she created an internal integrated team between laboratory and case management personnel known as the Texas Newborn Screening Continuous Quality Improvement team. This team is charged with improving the Newborn Screening Program.

As Project Manager for the DSHS Newborn Screening Expansion Project, Ms. Williams was instrumental in the successful launch of the expansion in February 2007.

Previously, Ms. Williams served as the Newborn Screening Project Coordinator for the National Newborn Screening and Genetics Resource Center (NNSGRC). In this role, Ms. Williams facilitated communication and coordination of national newborn screening advisors, coordinated the collection, analysis and distribution of information regarding newborn screening policies, procedures, and practices, managed various NNSGRC subcontracts, and assisted with the design and upkeep of the NNSGRC website.

She has also been a member on various HRSA sponsored projects including projects focused on integration of child health systems, developing a newborn screening self assessment checklist, and evaluating and developing culturally and literacy level appropriate newborn screening parent education material.

Prior to joining NNSGRC in 2000, Ms. Williams worked for over 15 years for the Texas Department of Health Bureau of Laboratories. There she held many positions within the Newborn Screening Program, including five years as supervisor of the hypothyroidism screening laboratory and five years as Quality Assurance Officer for both the Newborn Screening and Texas Health Steps Programs. Ms. Williams is also trained and has years of experience in meeting facilitation and conflict management.

Ms. Williams has an undergraduate degree in microbiology and also holds a specialist certification in Microbiology from the American Society of Clinical Pathologists.



Evidence-Based Approach to Quality Performance Measures

Performance measurement is a key step in any program management and forms the basis of continual quality improvement. If proper measures do not exist, the effectiveness of any program cannot be assessed. Performance measurement results can be used to identify areas where intervention and action is required. It also provides feedback and motivation for continual improvement.

Evidence-Based Performance Measures for Newborn Screening

Studies have shown that early screening and early intervention for newborns diagnosed with newborn screening disorders have prevented child mortality and morbidity.

Performance measures exist in newborn screening programs. An example established under Title V Maternal and Child Health Programs (Title V) is the percent of newborns that are screened and confirmed with condition(s) as mandated by their State-sponsored newborn screening programs that receive appropriate follow -up as defined by their State.

The TNSPMP effort will review existing and develop additional performance measures with defined criteria used to evaluate the health care benefits and/or health care delivery to infants diagnosed with newborn screening disorders.



Types of Performance Measures

Types of performance measures used can include structure, process, and outcomes measures.⁴

Structure - These performance measures reveal the adequacy of resources and organizational arrangements currently in place to deliver care. Examples could include the number of providers with quality improvement programs relating to newborn screening collection and the percentage of nurses who have received formal training on specimen collection and transport.

Process - The appropriate provider and case management and follow-up activities are carried out to deliver care. Examples could include: Percent of unsatisfactory specimens and time-to-treatment measurements at different stages of the process.

Outcomes - These performance measures relate to the patient outcomes resulting from health care services provided to newborns. Examples could include the percentage of diagnosed newborns successfully treated, percentage of avoidable complications and deaths, and the level of the family's satisfaction with provided health care.

⁴ Reference: Performance Measurement in Clinical Practice: Performance Measures, From ACP Medicine Online, Posted 06/07/2006, Stephen D. Persell, MD, MPH; David W. Baker, MD, MPH, FACP; Kevin B. Weiss, MD, MPH, FACP. <http://www.medscape.com/viewarticle/535458?rss>



TNSPMP Meeting Agendas and Notes

This section is a placeholder to collect meeting agendas and notes for upcoming quarterly meetings and workgroup webinar sessions.

See the following page to view the agenda for the first meeting.

TNSPMP MEETING AGENDA AND NOTES

AGENDA

TNSPMP Kick-Off Meeting

DSHS Austin, M739 (Boardroom)

Friday, February 1, 2008

8:30a	Arrival, Registration, Coffee	
9:00a	Welcome and Purpose Introduction of Facilitator	Dr. Susan Neill
9:15a	Attendee Introductions Goals for the Day	Robin Scott; All
9:30a	Brief Overview of Project Charter Remarks from Co-Sponsor Remarks from CDC Grant Sponsor	Dr. Susan Neill Jann Melton-Kissel Dr. Susan Snyder
10:10a	Overview of Newborn Screening and Q and A	Dr. Susan Tanksley David Martinez
10:45a	Break	
11:00a	Overview of Project Process Q and A	John Scott, PMP
11:30a	Assessing the Current System What's Good and Not So Good?	Robin; All
12:00	Break for Lunch	
12:30p	Continue Lunch and Discussions of Current System Priorities for Fixes Success Vision	Robin; All
2:00p	Project Roles and Responsibilities Team Working Methods & Ground Rules	Robin; All
2:30p	Review Timelines; Determine Next Steps	Robin; All
3:15p	Meeting Plus/Delta (Feedback)	Robin; All
3:30p	Closing Comments & Adjourn	Dr. Susan Tanksley

TNSPMP MEETING AGENDA AND NOTES

CHARTER PROJECT TEAM MEETING
 DSHS AUSTIN, M739 (BOARDROOM)
 FRIDAY, FEBRUARY 1, 2008

MEETING NOTES

ATTENDEES

<i>Sandra Billings</i>	√	<i>Sister Mary Nicholas Vincelli</i>	√
<i>George R. Buchanan</i>	√	<i>Morgan Walthall</i>	
<i>Donna Claeys</i>	√	<i>Don P. Wilson</i>	√
<i>Margaret Drummond-Borg</i>		<i>Jerald L. Zarin</i>	√
<i>Alice Gong</i>	√	<i>Margaret Bruch</i>	√
<i>Jose L. Gonzalez</i>	√	<i>Sherry Clay</i>	√
<i>Charleta Guillory</i>	√	<i>Mirsa Douglass</i>	√
<i>Cheryl Hermerath</i>	√	<i>Eldridge Hutcheson</i>	√
<i>Scott D. McLean</i>		<i>David R. Martinez</i>	√
<i>Francisco Ramirez</i>	√	<i>Jann Melton-Kissel</i>	√
<i>John Saito</i>	√	<i>Susan Neill</i>	√
<i>Eileen Sheridan-Shayeb</i>		<i>Sharon Newcomb-Kase</i>	√
<i>Reid Sutton</i>	√	<i>Susan Tanksley</i>	√
<i>Larry Sweetman</i>	√	<i>Donna Williams</i>	√
<i>Brad Therrell</i>	√	<i>Susan Snyder</i>	√

TIMELINE

- 02/08/08 Distribute TNSPMP Kick-Off meeting notes for review.
- 02/15/08 Distribute the TNSPMP plan and work breakdown structure for review.
- 02/22/08 Hold a conference call to review the TNSPMP plan.
- 02/27/08 Deadline to provide comments and input on TNSPMP plan.
- 02/29/08 Obtain official sign-off and approval to move forward with TNSPMP plan.
- 04/11/08 Distribute a draft of a summary report discussing gaps and barriers. The summary report will include an internal assessment of DSHS Newborn Screening program using PEAS, a short discussion on the National Newborn Screening and Genetics Resource Center's (NNSGRC) review and information found from literature searches.
- 04/17/08 Hold TNSPMP Quarterly Meeting to review summary report, determine the area of focus for performance measures, and to review proposed performance measures.
- 09/28/08 End of first year project period

GOALS FOR MEETING

After opening statements and introductions, participants commented on the expectations of the meeting.

- ❖ Learn process of newborn screening in its entirety from drawing the specimen, screening, through notification of physicians and parents, to treatment and care of infants.
- ❖ Learn the relationship of this project to the Program Evaluation and Assessment Scheme (PEAS).
- ❖ Learn what can we do differently? How will it impact the system?
- ❖ Gain a better understanding of follow-up mechanisms including initial notification or call to parent.
- ❖ Move agenda forward to screen for 28 disorders including Cystic Fibrosis (CF).
- ❖ Understand the newborn screening process and address the timing of first screen (access). Screens are not done at the appropriate time.
- ❖ Learn procedures of initial call and parent contact.
- ❖ Understand method for educating health care providers.
- ❖ Concerned with laboratory technique, specifically false-positives.
- ❖ Network

BRIEF OVERVIEW OF PROJECT CHARTER

Dr. Neill briefly discussed the goal and scope of the TNSPMP.

The goal is to provide evidence-based performance measures to the medical community in Texas that can be used nationwide to identify opportunities to improve patient care for newborns identified with congenital and heritable disorders. The scope of the performance measures is limited to pre and post analytical activities, not testing activities.

ASSESSING THE CURRENT SYSTEM

Participants shared points on the pros and cons of the current newborn screening system.

WHAT'S NOT SO GOOD?

- ❖ Newborn screening program is not valued by parents and providers. The lack of understanding and awareness across the system may influence the lack of value for the newborn screening program.
- ❖ Adequate financing is needed to cover newborn screening activities including education, case management and follow-up activities.
- ❖ Cystic Fibrosis screening is not in place. (Texas has the largest population of affected children with this inherited disease.)

- ❖ Lack of advocacy efforts with the legislature. Research and findings are needed to support and influence policy. A strategic process is needed to make decisions for the newborn screening system.
- ❖ Not every infant is screened for newborn disorders (including newborn hearing screening). Approximately 90 to 95 percent of infants receive a first screen. Of those, approximately 90 percent receive a second screen. Need a method to coordinate with birth records to determine who has/has not been screened.
- ❖ Lack of access to confirmatory testing.
- ❖ Hospitals need a way to know how they are performing.
- ❖ Lack of an effective courier system to ensure timeliness of specimen transport and delivery to the state laboratory.
- ❖ Providers are unaware of how to access DSHS or national information on newborn screening.
- ❖ Lack of adequate training for regional staff coupled with their multiple responsibilities outside of newborn screening program.
- ❖ Lack of specialists and lack of access to specialists for newborn screening disorders.
- ❖ Psycho-social barriers exist for families which lead to diagnosed infants not receiving proper follow through on treatment and care.
- ❖ Input from advisory committees and advisory boards for DSHS is diminished. Input from subject matter experts and external entities is lacking or non-existent. DSHS has discontinued travel reimbursements for advisory members.
- ❖ Information on diagnosis of affected infants is not communicated back to DSHS in a timely manner.
- ❖ Not enough information is made available to providers about disease specific guidelines.
- ❖ Sickle cell and other newborn screening disorders lack proper follow-up, especially for long term. “What is the condition of the baby 10 years from now?”
- ❖ Mobility of families poses a challenge to track screening.
- ❖ Lack of available information on disorders that are not screened in Texas and where families can obtain that screening.
- ❖ Lack of referrals to Early Childhood Intervention Services (ECI) which is the statewide program for families with children, birth to three, with (or at risk for) disabilities and developmental delays.
- ❖ The size and demographics of Texas poses many challenges for the newborn screening program.
- ❖ Texas has a high percentage of children (per capita) relative to other states.
- ❖ It is difficult to track and monitor children transferring across state lines and the Mexican border.

- ❖ There are many undocumented children, which causes problems when trying to track abnormal results, ensure screens are obtained, etc.
- ❖ Lack of information regarding the impacts on screening results of premature babies, those on total parenteral nutrition (TPN), those who had blood transfusions, etc.
- ❖ Too many repeated screens due to TPN, premature births, blood transfusions, etc.
- ❖ Not doing full scan for tandem mass spectrometry – could pick up more disorders
- ❖ Lack of health care provider education.
- ❖ Lack of coordination at birth to identify every child. Need to have linkage between birth certificates and newborn screening records. Every birth certificate should have newborn screening report.
- ❖ Hospital turnaround times to send specimens to the laboratory are not timely. Hospitals may be batching specimens before sending off.
- ❖ DSHS needs adequate or better feedback/notification from physicians when diagnosed infants are treated. Processes in place lack standardization.
- ❖ Eligibility issues with special health care needs: Medicaid, Children with Special Health Care Needs (CSHCN) - an insurance provider, Texas Health Steps Comprehensive Care Program (THSteps CCP) which provides extra benefits for Medicaid clients, etc.

WHAT'S GOOD?

- ❖ DSHS processes a high volume of specimens, approximately 800,000 per year. The laboratory screening turnaround time is very good considering the volume of specimens.
- ❖ DSHS staff shows dedication and enthusiasm.
- ❖ Excellent website which nicely outlines material, is easily accessible, organized presentation, and addresses a diverse audience.
- ❖ Dedication of providers to screen and follow through with infants.
- ❖ DSHS commitment to improve newborn screening program and the overall system.
- ❖ Having a world class, state-of-the-art laboratory making pre and post analytical issues easier to address.
- ❖ Willingness of DSHS staff to outreach and educate.
- ❖ Free continuing education credits (CE) and continuing medical education (CME) credits.

VISION OF SUCCESS

Participants shared their vision of success for the newborn screening system.

- ❖ A system that works for everyone
- ❖ A way or ways to identify success
- ❖ Setting the standard by which others measure
- ❖ Positive role model for other states
- ❖ Identify and treat diagnosed infants
- ❖ A knowledgeable community (consumers, health care providers, nurses, physicians, etc.)
- ❖ Understand barriers and have developed strategies to address and/or overcome barriers
- ❖ System that “picks up” diagnosed infants prior to onset
- ❖ A system where every player knows what to do, when, and how to do it. Players know how the whole system works.
- ❖ A screen for every birth certificate
- ❖ An information system that works

CATEGORIZING THE ISSUES AND CHALLENGES

Participants categorized the issues and challenges through an exercise that involved writing the topic areas on separate pieces of papers and silently grouping these on the wall as a group. Not listed in any particular order, the following categories were established

SIZE AND DEMOGRAPHICS OF TEXAS – OVERARCHING CHALLENGE

BUDGET AND FINANCE

- Include CF in screen
- overall financing of program
- legislative advocacy
- access to confirmatory testing
- external advisors to DSHS are lacking
- ratio/percent of kids in Texas

SYSTEMS PROCESSING

- Human medical resource
- undocumented children
- coordinate with birth record
- border issues (Mexico & other border states)
- not every baby screened
- lack of courier system
- hospital turnaround time (sample to lab time)
- where do families go for diseases not screened in Texas
- opportunity for outside professional organization input

FOLLOW-UP

- Family psychosocial and economic issues to follow through
- mobility of families vs. screening
- sickle cell & other disorders lack of follow-up (where is baby 10 years from now)
- untimely information for diagnosis and outcome
- lack of specialists
- multiple responsibilities of regional staff
- lack of referrals to ECI
- lack of access to specialists
- eligibility for Medicaid/CSHCN/THSteps CCP

EDUCATION

- Lack of parent awareness
- parents and provider care physicians (PCP) don't value NBS
- providers don't know how to access info from DSHS/national resources
- lack of regional staff training
- lack of health care provider education
- not enough info to providers about disease specific guidelines
- impacts on screening (premature babies, tpn, transfusions, etc.)

PROJECT ROLES AND RESPONSIBILITIES

Mirsa Douglas briefly commented on the roles and responsibilities of the external stakeholder group.

Roles and responsibilities will be outlined in the project plan to be delivered several weeks after the meeting. Examples are attending quarterly TNSPMP Team meetings, participating in conference calls, reviewing documents written by the internal DSHS team, providing overall input, providing access to pertinent information, prioritizing proposed performance measures, providing recommended uses of performance measures, etc.

TEAM WORKING METHODS AND GROUND RULES

Participants agreed on a basic method for making group decisions as the project moves forward.

- ❖ 80 percent consensus method – When participants are 80 percent “OK” with the decision, it is acceptable to move forward with the decision.
- ❖ Decisions are made in person.
- ❖ There is only one meeting; No side conversations or side meetings.

PARKING LOT ISSUES

Topics of financing and funding of the newborn screening program was placed in the parking lot issues. Discussion points are shown below.

- ❖ A huge variance exists between the fees from state to state.
- ❖ Texas legislature provides funding for under-insured and uninsured. 60 percent of the births are Medicaid funded.
- ❖ Start-up funding is necessary because revenue streams are collected on a reimbursement method. Newborn screening cards marked as ‘Medicaid/Charity’ are processed quarterly to validate eligibility. ‘Paid’ cards are paid by the ordering physician. Payment for newborn screening ‘Paid’ cards is due 120 days after shipment.
- ❖ Fees were determined based on the cost of the testing, education efforts, and case management activities within DSHS. The fee changed from \$19.50 to \$29.50 per card where \$3.00 is now allocated to case management activities.
- ❖ To take advantage of funding synergies, the newborn screening program is funded through other sources such as Title V. Although Title V funding is decreasing, some of these funds are used to support laboratory, case management and follow-up activities.
- ❖ Barriers exist to increase fees: Provisions and interpretation in the law do not allow increases and TMA and THA stakeholder input would not tolerate fees over \$30.00
- ❖ A cost analysis would be beneficial to understand the true costs between the laboratory and case management activities. Typically, there is a 50-50 split of allocated costs between laboratory and case management activity. Fees are as high as \$175.00 (ex. Alabama). Texas fees are on the low end of the national range.



MEETING PLUS/DELTA (FEEDBACK)

Participants shared thoughts about what they liked and didn't like about the meeting.

CHANGES

Did not like the table cluster layout
 Would have preferred a room arrangement such that tables are situated in a circular layout so all seats have an equally good view of all participants.

POSITIVE FEEDBACK

Appreciated overview of the newborn screening program
 Good orchestration and involvement of meeting
 Good movement and activity for participants
 Delicious cakes
 Effective facilitator
 Well-planned meeting by staff, good preparation
 Nice selection of room and location

Appendix

Quick Reference Guide to Screening Disorders⁵

BIOTINIDASE DEFICIENCY (BIOT)

Biotinidase Deficiency or 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.

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.

CONGENITAL HYPOTHYROIDISM (CH)

Congenital Hypothyroidism or CH results from inadequate or absent production of thyroid hormone. It 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.

GALACTOSEMIA (GAL)

Galactose-1-Phosphate Uridyltransferase 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.

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

⁵ Formatting or grouping of disorders recommended by American Academy of Pediatrics.

protein diet and supplemental medicines, including Vitamin B6.

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.

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.

PHENYLKETONURIA (PKU)

Phenylketonuria is an enzyme defect that prevents metabolism of phenylalanine, an amino acid essential to brain development. It 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.

SICKLE CELL DISEASE (SCD)

Sickle Cell Disease or 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.

TYROSINEMIA TYPE I (TYR 1)

Tyrosinemia or TYR1 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.

OTHER FATTY ACID OXIDATION (FAO) DISORDERS

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.

ORGANIC ACID (OA) DISORDERS

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.

UREA CYCLE DISORDERS (UCD)

Urea Cycle Disorders (UCD) includes 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.

Texas Newborn Screening Panel

Amino Acid Disorders
Argininosuccinic Acidemia (ASA)
Citrullinemia (CIT)
Homocystinuria (HCY)
Maple Syrup Urine Disease (MSUD)
Phenylketonuria (PKU)
Tyrosinemia Type 1 (TYR1)
Fatty Acid Disorders
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
Trifunctional Protein Deficiency (TFP)
Carnitine Uptake Deficiency (CUD)
Organic Acid Disorders
Glutaric Acidemia 1 (GA-1)
3-OH 3-Methyl Glutaric Aciduria (HMG)
Isovaleric Acidemia (IVA)
Multiple Carboxylase Deficiency (MCD)
3 -Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)
Methylmalonic Acidemia (MMA) mutase deficiency (MUT)
Methylmalonic Acidemia (MMA) cobalamin A and B deficiency (CblA and CblB)
Propionic Acidemia (PA)
Beta-Ketothiolase Deficiency (BKT)
Galactosemia
Biotinidase Deficiency
Endocrine Disorders
Congenital Hypothyroidism (CH)
Congenital Adrenal Hyperplasia (CAH)
Hemoglobinopathies including:
Hb S/S
Hb S/C
Hb S-Beta Thalassemia

2007 Newborn Screening Statistics

Below are the newborn screening statistics reported for 2007.

General Specimen Statistics	
Number of Specimens Reported	791,784
Number of Unsatisfactory Specimens (Not Tested)	14,324
Number of Presumptive Positives	16,596

Below are the 2007 newborn screening disorders and reported cases. Numbers indicated in parenthesis are disorders reported but are not on the core panel for screening.

Biotinidase Deficiency (BIOT) Cases	
Profound BIOT	4
Partial BIOT	8

Congenital Adrenal Hyperplasia (CAH) Cases	
Salt Wasting CAH	14
Simple Virilizing CAH	2
Other	(10)

Congenital Hypothyroidism (CH) Cases	
Primary CH	158

Galactosemia(GAL) Cases	
Classical Gal	5
Other	(71)

Homocystinuria (HCY) Cases	
Homocystinuria	2

Maple Syrup Urine Disease (MSUD) Cases	
MSUD	1

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency Cases	
MCAD	16

PKU Cases	
Classical PKU	12
Variants	(13)

Sickle Cell Disease (SCD) Cases	
SS	90
SC	40
S/ β Thalassemia	10
SD	(1)
Other (Non-Sickling)	(37)

Tyrosinemia (TYR) Cases	
TYR	0
Other	(8)

Other Fatty Acid Oxidation Disorders Cases	
LCHAD	2
VLCAD	6
Other	(1)

Organic Acid Disorders Cases	
3 MCC	7
GA-1	7
IVA	1
MMA	3
MCD	1
PROP	1
Other	(2)

Urea Cycle Disorders (UCD) Cases	
ASA	1
CIT	1
