

NEWBORN SCREENING IN NEBRASKA

Newborn Screening for Metabolic and
Inherited Disorders

AND

Newborn Hearing Screening



2006 Annual Report



Dedication

Nebraska has lost a legend. Hobart E. Wiltse, M.D., PhD passed away June 6, 2007, at the age of 75, after a courageous battle with cancer. Everyone who was blessed to have known Dr. Wiltse had their lives changed for the better. He was the consummate teacher, caring and knowledgeable clinician, dedicated researcher and unflappable advocate.

Every baby born in Nebraska benefits from the comprehensive newborn screening system we have, thanks to Dr. Wiltse. Dr. Wiltse made sure Nebraska participated in the early 1960s field trials when a simple blood spot was collected on newborn babies and tested for Phenylketonuria or PKU. He tirelessly advocated for strong legislation that would protect every baby, not just some.



Throughout the years Dr. Wiltse aided in the growth and improvement of the newborn screening system. Because of his efforts, 8 Disorders are now required to be screened and another 26 made available to every newborn. Nebraskan's benefit from a high quality, strong newborn screening infrastructure for education, testing, short term follow-up, treatment and management, and ongoing quality assurance, thanks in large part to the efforts and guidance of Dr. Wiltse.

Dr. Wiltse graduated from the University of Nebraska in 1948 and UNMC College of Medicine in 1953. He completed his pediatric fellowship training at Johns Hopkins University and the University of California at Los Angeles School of Medicine. The Wiltse Center for the Study of Metabolic Disorders at Children's Hospital in Omaha is named in his honor.

Earlier this year, Dr. Wiltse was presented with a lifetime achievement award from the Governor and the Nebraska Health and Human Services System in appreciation for his contributions to the treatment, education and advocacy of and for people with metabolic and inherited Disorders.

He was a mentor to many, and his grace and wisdom will be forever missed, but never forgotten.

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NEWBORN SCREENING FOR INBORN ERRORS OF METABOLISM AND INHERITED DISORDERS

The goal of newborn screening for metabolic and inherited Disorders is to identify newborns at risk for certain metabolic, endocrine, hematologic and other conditions that would otherwise be undetected until damage has occurred, and for which intervention and/or treatment can improve the outcome for the newborn.

Newborn Screening is a system involving many elements including:

- ❖ Education of health care professionals and parents and efforts to increase public awareness
- ❖ Proper and timely collection of quality specimens
- ❖ Appropriate and timely transmittal of specimens to the Newborn Screening laboratory
- ❖ Rapid quality testing methods
- ❖ Timely notification of the infant's parents
- ❖ Timely retrieval of the infant for confirmatory or repeat testing
- ❖ Appropriate referral of family to specialists for diagnosis, treatment and counseling
- ❖ Assuring access to needed specialized services and treatment
- ❖ Evaluation and Quality Assurance

Each of these components of the system requires ongoing monitoring to ensure quality.

In 2006, newborn screening efforts resulted in successfully identifying and treating 43 newborns affected with conditions in time to prevent problems associated with them:

- ❖ 4 babies with partial (treated) biotinidase deficiency
- ❖ 1 baby with congenital adrenal hyperplasia
- ❖ 10 babies with congenital primary hypothyroidism
- ❖ 13 babies with cystic fibrosis (11 classical, 2 atypical)
- ❖ 6 babies with hemoglobinopathies (3 sickle cell disease, 2 sickle hemoglobin- C disease, and 1 hemoglobin-E Disease)
- ❖ 5 babies with hyperphenylalaninemia (4 clinically significant)
- ❖ 1 baby with 3-MCC Deficiency
- ❖ 1 baby with IBCD (Isobutyryl-CoA Dehydrogenase Deficiency)
- ❖ 2 babies with transient tyrosinemia
- ❖ 1 with presumed cobalamin defect

WHAT IS NEWBORN SCREENING?

Newborn screening programs have been around for over four decades in all 50 states and in several countries. The compulsory screening panel varies from state to state but the overall goal is the same: prevent or minimize the serious effects of the conditions screened. In 2006, the conditions each state screened for became more uniform as an increasing number of states adopted screening by MS/MS (Tandem Mass Spectrometry). Nebraska has required this MS/MS supplementary screening to be offered to every newborn's parents since 2003.

The effects of screened conditions can range from brain and nerve cell damage resulting in severe mental retardation, to damage to the child's heart, kidney, liver, spleen, eyes, problems with physical growth, stroke and even death.

In 2006 Nebraska required screening for 8 conditions:

- **Biotinidase Deficiency**
- **Congenital Adrenal Hyperplasia**
- **Congenital Primary Hypothyroidism**
- **Cystic Fibrosis**
- **Galactosemia**
- **Hemoglobinopathies, (Sickle Cell, Sickle hgb. C & thalassemias)**
- **MCAD**
- **PKU**

The conditions for which screening is done are individually rare, so consultation with and/or referral to the appropriate pediatric specialist such as a geneticist, metabolic specialist, hematologist, endocrinologist or an Accredited CF Center is always recommended.

In addition to the 8 required tests, Nebraska required that every parent be offered the option of "supplemental testing" for amino, fatty and organic acid conditions by tandem mass spectrometry.

Individually each condition is quite rare. However, collectively as many as one in every 800-1000 babies are diagnosed each year in Nebraska with conditions from the current screening panel!

In 2006, the program began required screening for Cystic Fibrosis and Congenital Adrenal Hyperplasia.

The first year was successful in:

- **Implementing the laboratory screening algorithms such that the number of false positives was kept to a very acceptable minimum**
- **Parents had educational material about these conditions before screening, and information about what the results meant when they had an inconclusive or a positive screening result**
- **Physicians were assisted in connecting with pediatric sub-specialists to assist them in diagnosis and treatment of affected newborns**
- **Affected newborns were identified and entered into treatment in a timely manner**



HOW THE NEWBORN SCREENING PROCESS WORKS

1: TESTING

Baby is born.
Dried blood spot
specimen is collected
@ 24-48 hours of life



Specimen shipped
overnight to newborn
screening
laboratory, Pediatrix



Specimen data entered
into data system



Specimen tested for
multiple conditions



2: FOLLOW-UP

Inconclusive or positive
screen results reported
by phone/fax/letter from
lab and follow-up staff
to baby's physician.



Baby's physician or
health care provider
contacts baby's parents

Parent's bring baby back
in



for evaluation and more
testing



3: DIAGNOSIS/ INTERVENTION

Depending on the screen
result, and on the
condition screened:

Repeat or confirmatory
testing occurs



Parent education for
signs/symptoms to
watch for



Baby's physician
consults with and/or
refers baby to pediatric
sub-specialist
appropriate to the
condition



4: TREATMENT & MANAGEMENT

Once diagnosis is made,
treatment begins. (For
some life threatening
conditions, treatment
may occur prior to
diagnosis- on
recommendation of
specialist.



Parent's receive
treatment guidelines /
education.

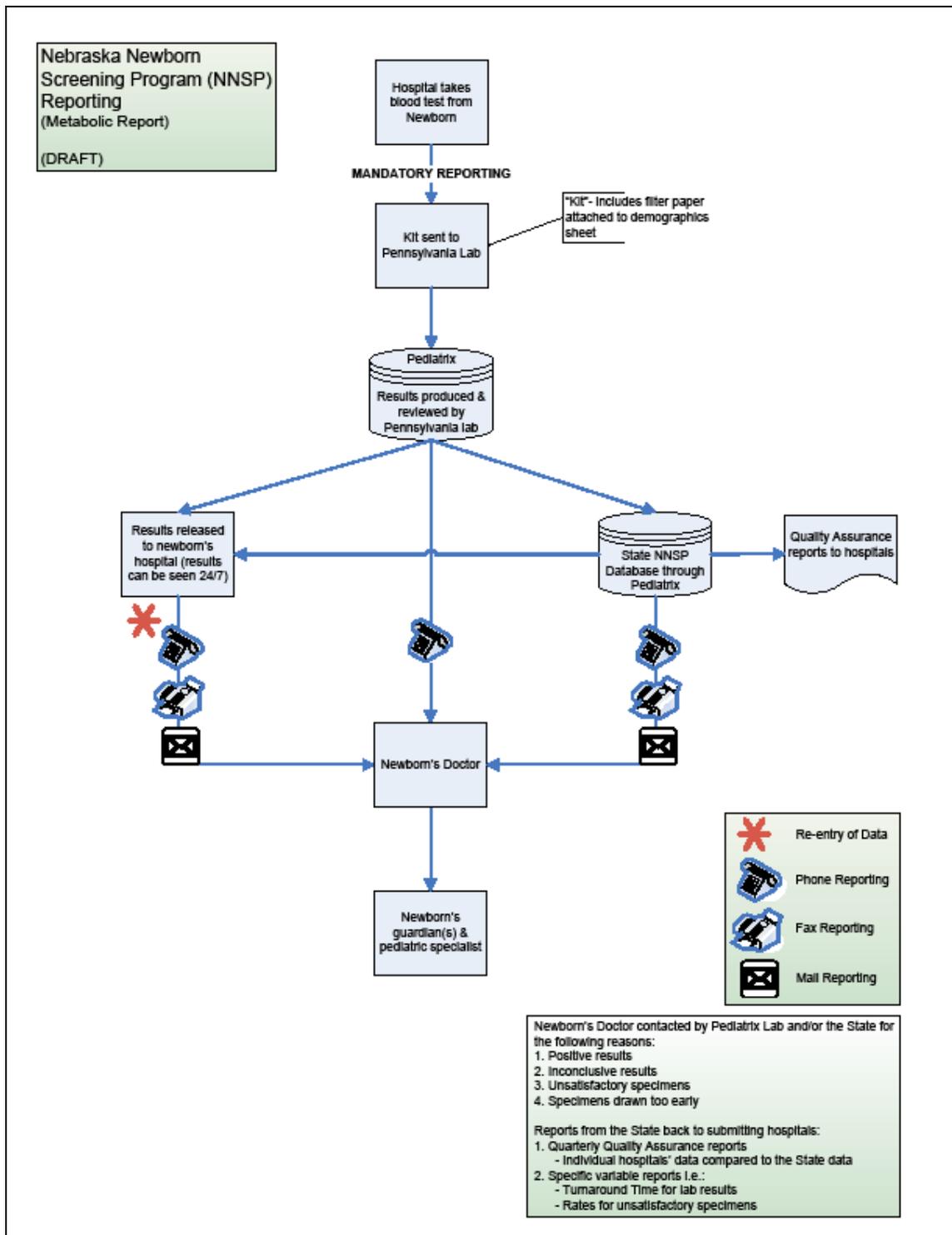


Team Support services
as appropriate, e.g.:

- metabolic dietitian
monitoring &
consultation
- ongoing blood
monitoring
- referral to early
intervention
services
- pulmonary/ CF
services
- ped endocrine
monitoring
- ped hematology
monitoring
- genetic counseling
& consideration of
family testing
- Other allied health
services as needed



Data flow: This chart demonstrates how data from newborn screening is produced, transmitted and utilized to facilitate the retrieval of newborns at risk for any of the conditions screened, so they can be evaluated, diagnosed and have treatment initiated.



System Overview

In 2006, 62 Nebraska hospitals sent specimens to Pediatrix Screening Laboratory. This laboratory is under contract with the State of Nebraska to conduct all of the newborn screens.



Supplemental screening (additional test results from Tandem Mass Spectrometry testing) was provided at no extra cost and required no extra blood. The supplemental screening provides results on fatty acid, amino acid and organic acid Disorders. Educational efforts by physicians and hospital staff using written materials from the Newborn Screening Program helped parents understand their options. More than 97% of parents consented to the supplemental screening for their newborns in 2006.

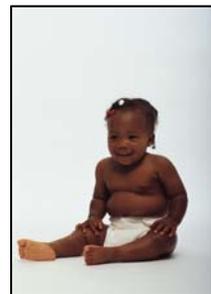
The Newborn Screening Program in the Nebraska Department of Health and Human Services was staffed by Mike Rooney, Administrative Assistant, Krystal Baumert, Follow-up Coordinator, Karen Eveans, Follow-up Specialist, and Julie Miller, Program Manager.

Ongoing consultation with the laboratory, metabolic specialists RicHard Lutz, M.D., William Rizzo M.D., and Jill Skrabal, R.D., the Cystic Fibrosis Center Director John Colombo, M.D. and Dee Aquazzino and pediatric hematologist James Harper, M.D. ensured expert advice and assistance was available as needed throughout the year.

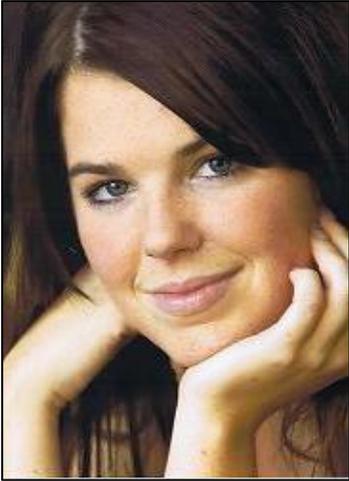
Quarterly meetings with the Newborn Screening Advisory Committee provided invaluable guidance to the program on several policy and quality assurance issues.

Treatment services received substantial support via the \$10 per infant screened fee, State General Funds and Title V Maternal and Child Health Block Grant funds. This included funding for special metabolic formulas, metabolically altered/pharmaceutically manufactured foods, and support for specialty dietitian services and sub-specialist M.D. consultation services.

Quarterly quality assurance reports were sent to every birthing hospital, as well as Children's Hospital of Omaha, a facility that completes a significant number of screens on babies transferred to them. In addition, the Advisory Committee reviewed several quality assurance reports at each quarterly meeting.



Living with PKU... Jenn's success story!



When I was three days old, I was diagnosed with Phenylketonuria. My treatment: A life-long, low-protein diet. Of course there are obvious disadvantages to this metabolic disease, but I wouldn't trade having it for the world.

As a child, I had to endure all the embarrassment of having my own "special" foods at birthday parties, sleepovers, and at school lunches. Like any kid, I just wanted to fit in and be normal. As I grew older, more and more people in the community (of about 4,000) started recognizing me and knew about my diet. Whenever I called Pizza

Hut, I just stated my name and they instantly knew to make a personal pan, no cheese, tons of veggies. I even got notes on the inside of my pizza box from the employees. The local coffee shop also started carrying Coffee Mate, which I use in place of milk in my lattes. My community made living with my diet a whole lot easier.

When I started high school, I used my PKU to stand out. I no longer wanted to be a normal part of the crowd. I embraced chances to explain my disease to people. It's a great conversation starter! Everyone is always fascinated and eager to learn about it. When it came time to apply for colleges and scholarships last year, I applied for the Rural Health Opportunities Program, a "health career" scholarship. In my mandatory essay, I wrote about my PKU and how it has affected my life and why I desire a career as a physician. Growing up with a metabolic disease and being in close proximity with doctors quite frequently has inspired me to seek a health career. I landed an interview for the scholarship and received it the next day.

I truly feel God gave me this disease for a reason. Without having PKU and such awesome role models at the Medical Center, I wouldn't have realized what my purpose in life was. This is a test that I'm going to pass. I'm using what I'm given to achieve my dream. My undergraduate tuition to Wayne State College is fully paid, and I have a guaranteed spot in medical school at the University of Nebraska Medical Center.

The Nebraska Newborn Screening Program would like to thank Jenn Harney for sharing her inspirational story of living with PKU.

National Attention on Newborn Screening in 2006

Secretary's Advisory Committee Policy Recommendations

The federal Health and Human Services Secretary's "Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children" (SACHDGDNC) continued its efforts to evaluate the state of the States and developed a policy and procedure for evaluating candidate conditions for appropriateness for newborn screening as well as other recommendations to the Secretary of Health and Human Services. The SACHDGDNC has three subcommittees established to assist in evaluating newborn screening systems and recommend priorities and strategies for insuring equity and quality amongst screening programs. The subcommittees are: Laboratory, Follow-up and Education. Nebraska's own Amy Brower, Ph.D., serves on the Committee and the Nebraska Newborn Screening Program Manager has the honor of serving on the Follow-up Subcommittee. The Follow-up subcommittee developed plans for a stakeholder meeting designed to produce a widely accepted white paper on the elements of long-term follow-up and management, to be presented to the SACHDGDNC to hopefully help guide the Committee's recommendations to the Secretary of Health & Human Services.

March of Dimes Advocacy

In 2006 the March of Dimes, Nebraska Chapter began advocating for every newborn in Nebraska to be screened for the 29 conditions in the core panel recommended by the American College of Medical Genetics (ACMG). In addition to March of Dimes, the SACHDGDNC as well as the American Academy of Pediatrics have endorsed this recommendation. Although Nebraska required screening for 8 conditions (10 if hemoglobinopathies are counted as SC, SS and Thalassemia), and universally offered the supplemental tandem mass spectrometry screen since 2006, March of Dimes continues to work with the Department to meet their national goal of ensuring every baby in every state receives mandatory screening for all 29 conditions in the core panel.

American College of Medical Genetics ACT Sheets

The ACMG development of ACT (Action) sheets for all conditions included in their recommended core panel concluded with posting of these on their web-site. The Nebraska program provides these ACT sheets for all positive screening results from tandem mass spectrometry screening to the newborn's physician. For all other positive screen results, the program provides ACT sheets previously produced by the NNSP and its consultants and approved by the Nebraska Newborn Screening Advisory Committee.

Clinical Laboratory Standards Institute Guidelines

Guidelines for short-term follow-up published for the first time in 2006, were sponsored by the former National Committee on Clinical Laboratory Standards (NCCLS) group (now Clinical Laboratory Standards Institute or CLSI). The Nebraska Newborn Screening Program Manager participated on the work group that developed these guidelines.

As new knowledge becomes available and technology evolves, guidelines need to be periodically reviewed and revised. The Newborn Screening Program Manager participated on the work group that reviewed and revised the fifth edition of the CLSI guidelines for collection of dried blood spots for newborn screening. She also helped with the development of a new educational video to accompany the guidelines, expected to be available in 2008.

MAJOR INITIATIVES of 2006 in NEBRASKA

Education

- ❖ Mike Rooney of the Nebraska Newborn Screening Program successfully managed contracts to obtain additional translations of the patient education materials “Parent’s Guide to Your Baby’s Newborn Screening” and the “Supplemental Consent Form” in French and three Sudanese dialects (Dinka, Nuer and Anuak). Translations were completed and made available on request. The translations were intended for the immigrant and refugee populations from Africa and the Sudan. Nebraska Newborn Screening parent education materials are now available in 10 languages.
- ❖ The program continued to provide supplies of the “Parent’s Guide” and supplemental newborn screening consent forms to all birthing hospitals and upon request to pediatric and family physician clinics and childbirth educators.
- ❖ In January, an educational mailer on cystic fibrosis screening and reporting of meconium ileus (MI) was sent to all birthing hospitals. The Wisconsin Newborn Screening Program (the State with the most experience in screening for CF) had reported past experience with excessive MI reporting. In order to minimize the risk for a false negative, the screening algorithm for babies with MI or other bowel obstruction is different than for the rest of the newborn population. Therefore, this education piece was intended to proactively avoid the over-reporting.
- ❖ In December, education materials focused on helping hospital staff understand what circumstances including other bowel obstructions, qualify as reasons to check the MI box on the filter paper specimens. This was intended to address a concern with possible under-reporting of MI.
- ❖ Dr. James Harper, Pediatric Hematologist from the University of Nebraska Medical Center produced pod-casts about differential diagnosis of screened positive hemoglobinopathies and posted these to the UNMC web site and this was linked from the Newborn Screening Web pages.
- ❖ In preparation to distribute parent education newborn screening videos purchased under the Heartland Regional Newborn Screening & Genetics Collaborative, the program surveyed community based public health prenatal care and education providers as to their interest and preferred format. The videos are expected to be received in 2007.
- ❖ State-to-State collaboration and technical assistance was provided by the Nebraska Newborn Screening program to Kansas, via meeting in Topeka with the State’s Children’s Services Advisory Committee, which among other things was looking into models for newborn screening systems that could accommodate expansion into the use of tandem mass spectrometry for screening. In addition the Kansas Newborn Screening staff spent a day in Nebraska learning about Nebraska’s system of newborn screening, in particular focusing on education, short-term follow-up and quality assurance.

- ❖ Internal staff development included Krystal Baumert’s participation in a week-long training session on “Interpreting tandem mass spectrometry results for newborn screening”. The session held at the Institute of Metabolic Disease, Baylor University was co-sponsored by the Association of Public Health Laboratories and the National Newborn Screening & Genetics Resource Center. Karen Eveans participated in the Annual North American Cystic Fibrosis Conference held in Colorado.
- ❖ Newborn screening staff participated in the Annual meeting/conference of the Heartland Regional Newborn Screening & Genetics Collaborative held in September in Omaha. Julie Miller, and G. Bradley Schaefer, M.D. served on the planning committee for this conference.
- ❖ The Newborn Screening display and educational materials were exhibited at the Family Health Conference held in Kearney attended by more than 350 nursing, public health and social service professionals.
- ❖ Presentations/training sessions about newborn screening were provided on request to:
 - Creighton Pediatrics,
 - West Holt Community Hospital in Atkinson,
 - Spring meeting of Medical Technologists’ Scientific/Education meeting in Holdrege,
 - Six roundtable discussions at the June 2006 meeting of the Family & Consumer Science teachers in Kearney, Nebraska.

Policy

Regulations add CF and CAH

In 2006, regulation changes requiring screening for cystic fibrosis (CF) and congenital adrenal hyperplasia (CAH) were implemented. Within this first year of screening for these conditions Nebraska successfully identified one newborn with CAH and thirteen newborns with cystic fibrosis.

Screening algorithms for both CAH and CF were more complicated than what we were accustomed to in Nebraska because of various factors. For example normal reference ranges for 17 Alpha Hydroxy Progesterone (17 OHP) the analyte initially tested in the CAH screen, must be adjusted for birthweight ranges. In addition, there are “critical” cutoffs requiring urgent notification and action, vs. “inconclusive” cutoffs that are not reported out until a more sensitive reflex test (extracted 17OHP) is completed. Cystic Fibrosis screening algorithms are complicated because of reflex testing for the $\Delta F508$ CFTR gene mutation on a subset of elevated Immunoreactive Trypsinogen results, vs. reflex testing 36 mutation on a different subset. Results that indicate a lower suspicion of CF are recommended to initially be followed up with a repeat filter paper specimen. Results that indicate a higher risk of CF cause a recommendation for referral to an Accredited CF Center and sweat testing. Continuous education of providers and follow-up communication with the newborns’ physician and pediatric sub-specialists are essential to successfully ensuring diagnosis and treatment occur in a timely manner.

Revisions to Lab Screening Protocols

In an effort to reduce the burden of false positives yet retain sufficient reliability in screening, the lab contract was revised relevant to the screening algorithms and recommended actions for Biotinidase deficiency and Galactosemia. These changes incorporated intermediate cut-offs for which the result interpretation of “inconclusive” meant the results were slightly abnormal, but not alarmingly so, and collection of a “repeat” dried blood spot filter paper specimen was recommended. Those results for which the suspicion of disease is higher, are reported as presumptive positive, and recommendations for confirmatory testing and referral to a metabolic specialist are made. These revisions were made in the summer of 2006 with the approval of the Nebraska Newborn Screening Advisory Committee with the hope of reducing anxiety for families. When revising the contract, the program also had to revise parent education “fact sheets,” laboratory report language, letters and faxes used for notification of the baby’s physicians, and information on the Newborn Screening Web Site.



Pediatrix Screening laboratory staff punch blood spots to prepare for testing.

Consent Regulations Revised

A group of stakeholders conducted the annual review in September, 2006 of the regulations governing model consent for predictive genetic testing (one for supplemental newborn screening, one for prenatal predictive genetic testing, and one for other predictive genetic testing). As a result of this review, several revisions were initiated to improve clarity of the regulations. Revisions were expected to be promulgated sometime in 2007. The stakeholder group also identified problems with access to the model consent forms for predictive genetic testing. The forms had been distributed a few years earlier, and electronic access was available on the HHSS web page. However, navigation to find them was noted to be difficult. Revisions to the web page, and a searchable publications link was recommended and expected to be made in 2007.

Federal District Court Ruling

In response to the 2005 Federal District Court ruling in *Spiering vs. State of Nebraska*, the Newborn Screening regulations were revised to improve the clarity of the requirement for timing of specimen collection for babies not born in the hospital. The expectation was and is that every Nebraska newborn must be screened at between 24-48 hours, not just babies born in the hospital. The regulation revision now makes this explicitly clear. The requirement is intended to provide optimum timing for performing the screening in order to identify affected newborns and initiate treatment or intervention in time to prevent any damage. The requirement provides equal protection for every child born in Nebraska.

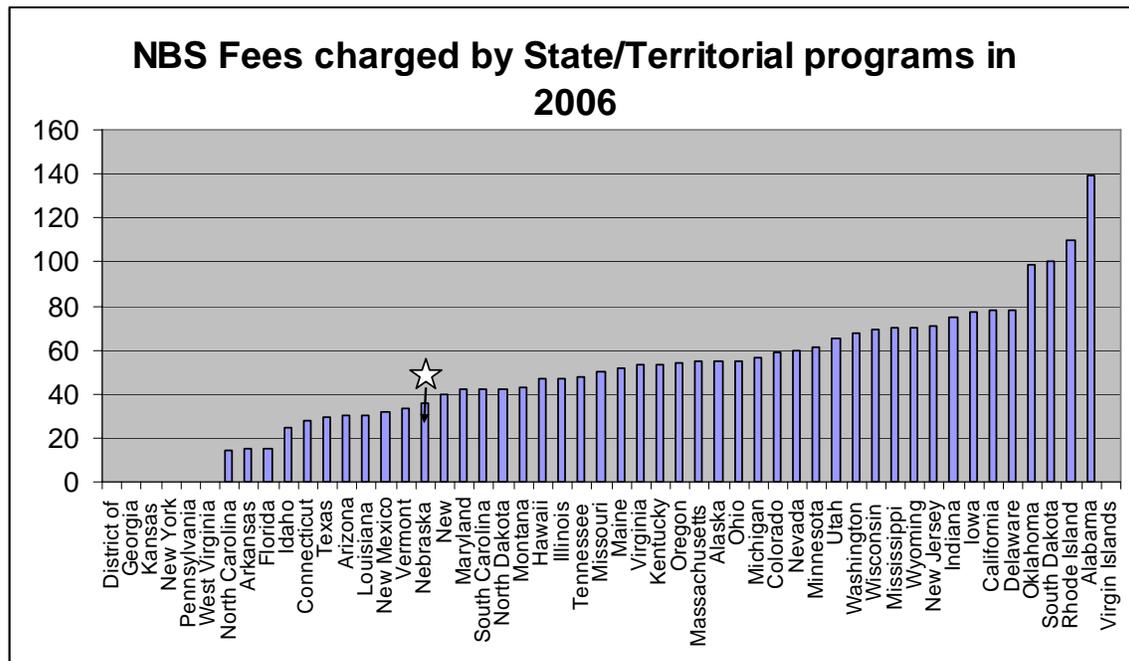
Legislation introduced in 2006

LB 1104, introduced by Senator Synowicki would have allowed for a religious exemption to the required newborn screening, allowing parents to dissent from having their newborns screened. The Legislature’s Health and Human Services Committee heard testimony on this bill, but no further action was taken on the bill in 2006.

Financing Newborn Screening

The Newborn Screening Advisory Committee developed a Position Paper on Fiscal Sustainability for Nebraska's Newborn Screening System and presented this to Dr. Joann Schaefer, Chief Medical Officer and Director of the Department of Health & Human Services Regulation & Licensure. Primarily the report identified these issues:

- Costs continued to rise while funding streams have been reduced or stayed flat.
- Nebraska does not have an insurance mandate requiring coverage of the treatments necessary to prevent the mental retardation, physical disability or death for the conditions screened, however the law does require the State to make the formula available, and up to \$2000/eligible individual each year toward the metabolic foods.
- Because it is successful, newborn screening continues to identify more people in need of these treatments, which are usually required for the person's lifetime.
- Laboratory charges were \$25.75 in 2006 in addition to the \$10 fee. The fee is returned to the State and used to provide the statutorily required formula and metabolic foods.
- According to the National Newborn Screening and Genetic Resource Center data reported by State in 2006, fees for screening ranged from \$0 to \$139.33. Those fees included support for a range of services from laboratory testing only, to laboratory, program administration/ follow-up, treatment, education and genetic services. The following tables show state to state comparisons of the 45 states that charged a fee for newborn screening based on the 2006 data:



Source: <http://genes-r-us.us.uthscsa.edu/>

Subsequent to the Advisory Committee's Position Paper, the Newborn Screening Program submitted a legislative bill proposal to the Health and Human Services System Policy Cabinet that would have provided for a fee structure to support the newborn screening system.

Quality Assurance

Quarterly quality assurance reports were sent to each birthing hospital and Children's Hospital in Omaha. These reports include the individual hospital's quarterly measures and a state-wide comparison on each measure. In addition, the publication "QI Hints" is sent out with each quality assurance report to the person designated by the birthing hospital Administrator.

Topics in 2006 included:

- To Consent or not consent...Parent Education Issues
- TESTS MISSING, how to avoid striking the panic button!
- Specimen collection timing, and
- KUDOs for quality specimen collection

Hospitals are encouraged to make the QI Hints available to all staff involved with parent education, specimen collection and handling, result reporting and tracking of screening results.

NEWBORN SCREENING ADVISORY COMMITTEE

The Newborn Screening Advisory Committee (NBSAC) provided technical expertise and policy guidance to the Nebraska Newborn Screening Program. Members commit at least a half a day every three months to advise the State Program. Several members provided extensive review and consultation beyond the committee meetings to help the program meet the recommendations of the larger Committee. The following summarizes this guidance:

Quality Assurance Reviews:

In 2006, the Committee continued to review quarterly quality assurance reports from the program. The Committee also monitored aggregate data received by the program on supplemental screening using Tandem Mass Spectrometry. Refer to Section III of this report for summaries of this data.

Quality and Technical Reviews:

The parent experts and medical experts of the Newborn Screening Advisory Committee were invaluable to the program in reviewing and making recommendations on newborn screening education, testing, follow-up, evaluation and financing.

Dried Blood Spot Testing for Genetic Causes of Hearing Loss:

External proposals were made to the Department to evaluate the feasibility of storing dried blood spots long-term to be retrieved for diagnostic testing of genetic causes of hearing loss. The Advisory Committee conducted an extensive evaluation of several options, made a recommendation, and the Department took this recommendation in late 2006 to the Newborn Hearing Screening Advisory Committee for their consideration. See Appendix C for more information.

Hemoglobinopathy Screening & Diagnosis Technical Reviews:

Committee Structure:

The Hemoglobinopathy committee (an off-shoot from the NBSAC) met to address Hemoglobinopathy screening and diagnostic issues. Members, James Harper M.D. and Elizabeth Thompson M.D., (Pediatric Hematologists), Jeanine Kean, R.N. and two parents, met in October with staff of the NBS program, Krystal Baumert, Karen Eveans, Julie Miller and the Medically Handicapped Children's Program, Dr. Jeanne Garvin. Their recommendations were endorsed by the full Advisory Committee, and implemented by the program. The recommendations:

- enhanced coordination of services with the Medically Handicapped Children's Program,
- altered the short-term follow-up procedures to send a letter to the baby's physician at 5 months of age for those needing additional testing at 6 months,
- implemented procedures for accessing the free (grant funded) DNA testing through the Children's Hospital Oakland Research Institute (CHORI) for difficult to diagnose newborns with hemoglobinopathies.

Emergency Preparedness Plan (Ad-Hoc Committee)

In the fall of 2006, the Newborn Screening Program Manager began development of a new all-hazaRDs emergency preparedness plan. The plan is designed to provide alternatives or back-up for the functions of newborn screening related to obtaining screening supplies, testing, ensuring short-term follow-up, and maintaining management and treatment services. A work group of local experts in emergency planning and implementation met in December to help refine and build on the plan. Dr. James Harper, Dr. Thomas Williams, and Dr. B.J. Wilson helped with recommendations regarding local community efforts, medical supply response teams, local communications infrastructure, and other aspects of the plan. At the October 2006 NBS Advisory Committee meeting the plan was discussed and Dr. Schaefer suggested Newborn Screening participate in the Health and Human Services System's table top or functional exercises as a means to evaluate the effectiveness of the plan.

The members of the NBSAC in 2006 were:

- **Khalid Awad**, M.D., *Neonatologist*, Neonatal Care PC, Omaha
- **Lawrence Bausch**, M.D., *Neonatologist*, Saint Elizabeth Regional Medical Center, Lincoln
- **John Colombo**, M.D., *Pediatric Pulmonologist, Director*, Nebraska Cystic Fibrosis Center, UMC, Omaha
- **Kevin Corley**, M.D., *Pediatric Endocrinologist*, Children's Hospital, Munroe/Meyer Institute for Genetics and Rehabilitation, UNMC, Omaha
- **Jeanne Egger**, *Parent*, Hallam
- **David Gnarra**, M.D., *Pediatric Hematologist*, Children's Hospital, Omaha
- **VICE CHAIR, James L. Harper**, M.D., *Pediatric Hematologist*, UNMC, Omaha
- **Kathryn Heldt**, RD, *Dietitian*, Children's Hospital Metabolic Clinic, Omaha
- **Mary Kisicki**, RN, *Parent*, Papillion
- **CHAIR: RicHard Lutz**, M.D., specialist in *Pediatric Genetics, Endocrinology, Metabolism*, Munroe/Meyer Institute for Genetics and Rehabilitation, UNMC, Omaha

- **Bev Morton**, *Parent*, Lincoln
- **Samuel Pirruccello**, M.D., *Pathologist*, Regional Pathology Services, UNMC, Omaha
- **Christine Reyes**, M.D., *Pathologist*, Pathology Center, Omaha
- **William Rizzo**, M.D., specialist in *Pediatric Genetics, Endocrinology, Metabolism*, Munroe Meyer Institute for Genetics and Rehabilitation, UNMC, Omaha
- **Kathy Rossiter**, MSN, *Certified Pediatric Nurse Practitioner*, Children's Hospital Metabolic Clinic, Omaha
- **Jill Skrabal**, R.D., *Dietitian*, Munroe Meyer Institute for Genetics and Rehabilitation, UNMC, Omaha
- **Corri Stearnes**, *Parent*, Omaha
- **Douglas Stickle**, Ph.D., *Technical Director, Clinical Chemistry*, UNMC, Omaha
- **William Swisher**, M.D., *Pediatrician*, Lincoln Pediatric Group, Lincoln
- **B.J. Wilson**, M.D., *Neonatologist/Perinatologist*, Saint Elizabeth Regional Medical Center, Lincoln, March of Dimes Representative
- **Ex-officio Hobart Wiltse**, M.D., Ph.D., *Pediatric Metabolic Specialist*, UNMC, Retired, Omaha



(Pictured above members of the Newborn Screening Advisory Committee and staff: **Seated left to right:** Mary Kisicki, Karen Eveans, Krystal Baumert, Christine Reyes, M.D., Jeanne Egger. **Middle Row:** Mike Rooney, B.J. Wilson, M.D., Julie Miller, William Rizzo, M.D., Kathy Rossiter, MSN, APRN, David Gnarra, M.D.. **Back Row:** Lawrence Bausch, M.D., Khalid Awad, M.D., James Harper, M.D., Joann Schaefer, M.D., RicHard Lutz, M.D., Doug Stickle, Ph.D., and John Colombo, M.D.. **Not pictured:** Kevin Corley, M.D., Kathryn Heldt, R.D., Bev Morton, Samuel Pirruccello, M.D., Jill Skrabal, RD, Corri Stearnes, William Swisher, M.D., Hobart Wiltse, M.D.

Assurance of Treatment and Management of Conditions

How Treatment and Management Is Paid For:



Part of the public health assurance role of Newborn Screening is ensuring treatment availability and access. Toward that end, the state program manages several contracts to ensure provision of otherwise prohibitively expensive formulas, foods, and services not always reimbursed by insurers. In 2006, 59 individuals were served through these programs: five infants, 34 children < 22 years of age, 14 pregnant women or women of childbearing age, and 6 adult males > 21.

Insurance usually covers medical treatments for some screened conditions such as prophylactic penicillin for patients with sickle cell disease, or synthetic thyroid hormone for patients with congenital primary hypothyroidism. However, many do not cover the metabolic formulas, and none cover the pharmaceutically manufactured foods required for PKU and other metabolic conditions screened on the supplemental panel. Therefore the biggest funding source supporting the metabolic foods and formulas was revenue generated from the \$10 per infant screened fee (approximately \$260,000 per year). The State General Fund appropriation of \$42,000 also helped provide for these medically necessary formulas and foods and the associated nutritional counseling for patients identified with PKU or the other metabolic conditions identified on the supplemental screen. Title V Maternal and Child Health Block Grant funds then filled in the gaps for metabolic foods/formula and nutritional counseling. The Medically Handicapped Children's Program provides some assistance to eligible families with children who have a hemoglobinopathy such as sickle cell disease or those with cystic fibrosis.

Individuals affected with screened metabolic conditions can obtain the metabolic formula through the Nebraska Medical Center Adult Metabolic Clinic, or at the Children's Hospital Metabolic Clinic. Ongoing dietary consultation, pediatric metabolic specialty care and routine blood monitoring are also provided. Individuals can order the pharmaceutically manufactured foods from product lists provided by manufacturers/distributors who have contracts with the State. Prior to 2006, 5 companies had contracts. In 2006 two more were added. Families can order up to \$2,000 of the metabolically altered foods per year without having to pre-pay.

Nebraska's families:



In Federal Fiscal Year 2006, metabolic formula ordering and distribution and specialized nutritional counseling and monitoring were provided via a contract with the University of Nebraska Medical Center for \$287,640.34. Through this contract 59 patients with inherited metabolic conditions identified through screening were served at the metabolic clinics. During State Fiscal Year 2006, fifty individuals utilized the pharmaceutically manufactured foods program, ordering foods with a value totaling \$57,277.75.



Mike Rooney coordinates the day-to-day metabolic foods program helping families to understand the program and stay connected, and monitoring the vendors' compliance with the contracts. To help families keep track of their orders, in 2006 he developed a new tracking log and distributed copies to all families and individuals eligible to participate in the program.

Sustaining the obligation to ensure access to treatment:

The number of children identified with conditions requiring special formula is anticipated to increase. The metabolic diets are required for life, and so people do not “age-out” of the need for the special formulas or foods. State General Funds have remained flat and federal allocations to Nebraska of Maternal and Child Health Title V Block grant funds have been reduced or flat for several years. Therefore the program continues to look for sustainable ways to continue to assure access to needed services for people who have these conditions.

Research for alternative therapies and cures:

It is an exciting time, where momentum in research is building hope for effective alternative therapies and the search for cures. As reported in the “National PKU News,” Vol. 18, Number 2, Fall 2006, “A new technique pioneered by the laboratory of Dr. Savio Woo at Mount Sinai School of Medicine in New York is a potential breakthrough for gene therapy in PKU. This technique could also be used to cure other genetic metabolic diseases such as maple syrup urine disease (MSUD), various organic acidemias, and urea cycle Disorders among others.” (They report curing PKU in mice via an effective mode of delivering gene therapy). Other therapies showing promise include the use of large neutral amino acids to block the phenylalanine from crossing into the brain; phenylalanine ammonia lyase, an enzyme that may block gut absorption of phenylalanine from food; and tetrahydrobiopterin (BH4) which when given in high doses to people with select PKU gene mutations can cause the enzyme to work more effectively and thus lower blood phenylalanine levels.

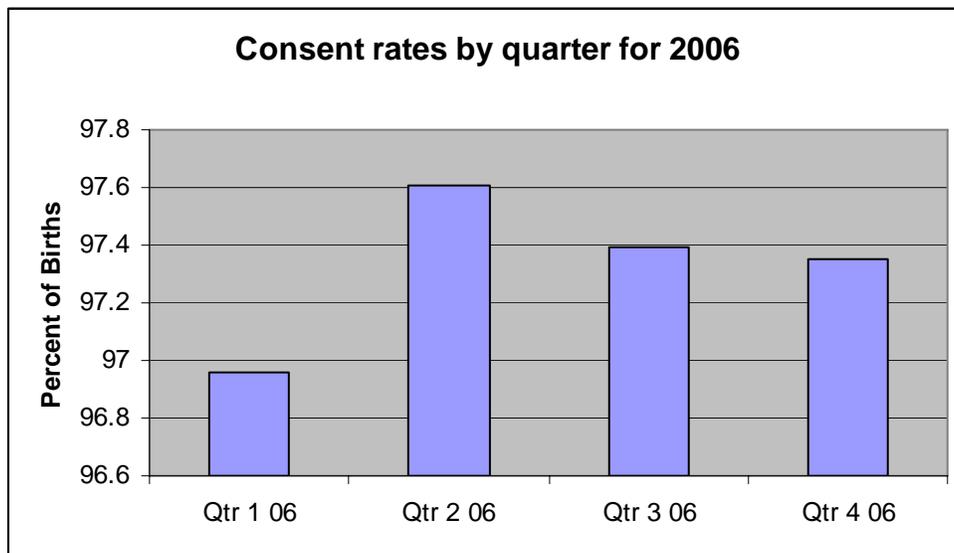
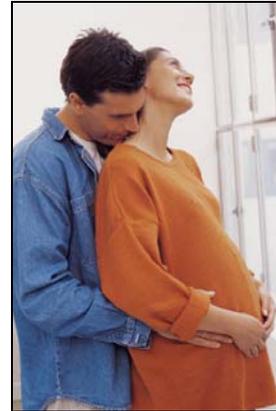
Nebraska’s pediatric metabolic specialists Dr. William Rizzo and Dr. Richard Lutz helped keep the Nebraska Newborn Screening Advisory Committee and Program staff apprised of progress in these areas.



PROCESS/OUTPUT DATA FOR 2006

PATIENT EDUCATION

Consent for supplemental screening



Overall for 2006, 97.32% of parents consented to the supplemental newborn screening panel from MS/MS. Hospital personnel reported that because it does not require any extra blood, and no additional cost, most parents are requesting it.

SPECIMEN
COLLECTION,
HANDLING AND
TRANSPORT



Age at Time of Specimen Collection (Initial Specimen) 2006

Age at time of collection	Number of births	Percent of births
0-12 hours	168	0.62
12-24 hours	114	0.42
Collected day 2 (24-48 hours of age)	24,290	89.58
Day 3	2,159	7.96
Day 4	201	0.74
Day 5	33	0.12
Day 6	19	0.07
Day 7	16	0.06
Over 7 days	116	0.43

Regulations require all specimens to be collected between 24-48 hours of birth, or prior to discharge, transfer or transfusion which ever comes first. Specimens collected past day two are at increased risk of a delayed diagnosis.

Unsatisfactory Specimens for 2006

Number of specimens unsatisfactory / Total # initial specimens	100 / 26,819	0.38% of initial specimens
REASONS specimens were UNSATISFACTORY	Number	% of unsats
Exposed to heat or humidity	9	9%
Heavily applied, layered or double spotted	17	17%
Blood spots not soaked through to the back of the filter paper	27	27%
Serum or fluid mixed with sample	12	12%
Specimen contaminated or diluted	6	6%
QNS (Quantity not sufficient)	26	26%
Multiple drops that are unevenly soaked through	1	1%
Clotted blood on surface of the filter paper	1	1%
Blood spots not allowed to air dry prior to shipping	1	1%

The art and science of correctly collecting and handling dried blood spots on filter paper requires trained health care professionals, who consistently follow the Clinical and Laboratory Standards Institute procedures for specimen collection. Every unsatisfactory specimen must be repeated, to ensure sufficiently reliable screening results.

Drawn Early
(less than 24 hour)
Specimens
for 2006



Reason specimen collected at less than 24 hours of age	Number / Percent
Baby to be transferred	108 or 44.45%
Baby to be transfused	26 or 10.7 %
Unable to determine reason from data received at NNSP	109 or 44.8%

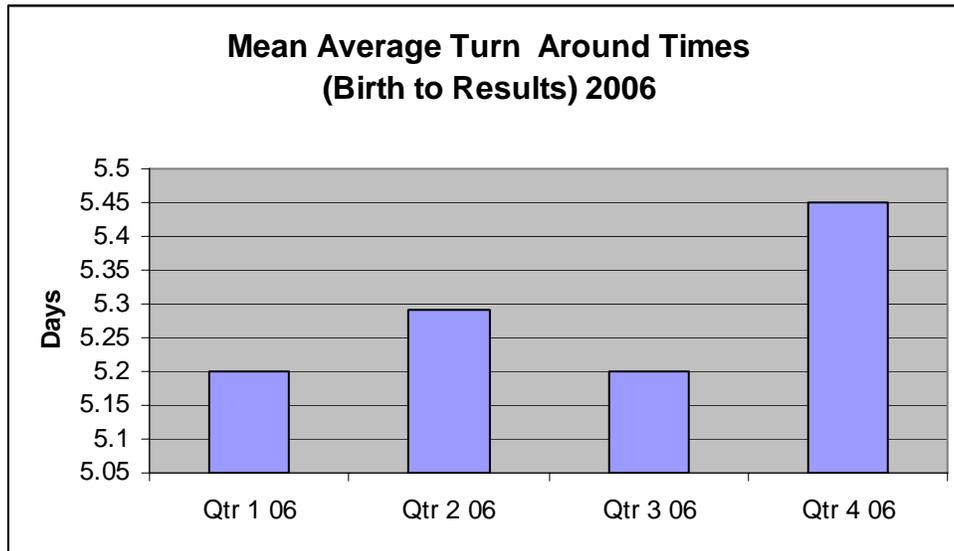
- ❖ Nine of the newborns whose specimens were drawn early did not get repeated as they expired.
- ❖ An additional 91 infants were reported as drawn early but when verifying with the birthing facilities they reported that they had made recording errors in these cases, and they submitted written corrections to the screening laboratory.



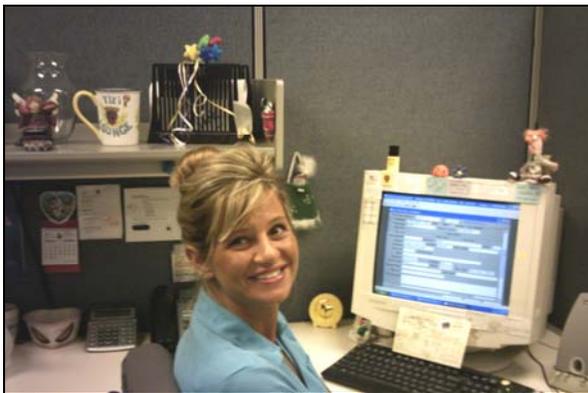
Denise Mihalik
of Pediatrix
Screening
Laboratory Inc.,
logs in
specimens
received via
overnight
shipping.

Specimen Turnaround Time

Regular monitoring of turnaround time between birth and reporting of results of the initial specimen is an important indicator for how well the newborn screening system is functioning. Specifically its ability to be able to identify affected infants in time to prevent the effects of the condition.



LABORATORY TESTING DATA



Heidi Oskamp, Pediatrix Laboratory Inc., enters data into the Pediatrix electronic data system off of the tear away sheets from the filter paper collection kits.



Pediatrix laboratory staff perform testing on a variety of laboratory instruments.

Presumptive Positive Screening Rates

Screening programs by their very nature are designed to find those at higher risk of a disease in order to facilitate their diagnosis and treatment to prevent morbidity and mortality. Screening tests were never designed to be diagnostic and so a small percent of screen results will be positive that upon repeat or confirmation are found to be normal. Nebraska and programs

across the country strive to minimize the number of newborns that require repeat or confirmatory testing (presumptive positive), and maximize the probability of identifying those affected. Nebraska continued to sustain a relatively low false positive rate for every condition screened.

Including only the conditions required to be screened (8), times the number of newborns screened (26,819), the number of tests completed for Nebraska newborns in 2006 were 212,152. Of this 212,152 there were 537 presumptive positive results requiring repeat or confirmatory testing. This is an overall presumptive positive rate of only 0.24%.

Over 97% of Nebraska newborns also received the supplemental Tandem Mass Spectrometry testing for the additional fatty acid, organic acid and amino acid Disorders, and only 212 of these required repeat testing. Nine of the 212 went on to confirmatory testing. Six received diagnoses of conditions identified initially through the tandem mass spectrometry screen.



Donald Chace, Ph.D, MSFS, Pediatrix Inc., next to one of the tandem mass spectrometers in the laboratory. Dr. Chace was one of the original developers of MS/MS testing for newborn screening and has been a leading researcher in the MS/MS field.

Specific presumptive positive rates by condition			
Condition	National rate 2006*	Nebr. 5 year mean average (2002-2006)**	Nebraska 2006 rates (mean average)***
Biotinidase Deficiency	0.06% 6:10,000	0.1% 10:10,000	0.06% 6:10,000
Congenital Adrenal Hyperplasia	0.46% 46:10,000	N/A Mandatory screening began January 2006	0.03% 3:10,000
Congenital Primary Hypothyroidism	0.80% 80:10,000	0.3% 30:10,000	0.19% 19:10,000
Cystic Fibrosis	0.42% 42:10,000	NA Mandatory screening began January 2006	0.25% 25:10,000
Galactosemia	0.78% 78:10,000	0.02% 2:10,000	0.05% 5 :10,000
MCAD	0.02% 2:10,000	N/A Mandatory screening began July 2003	0.02% 2:10,000
Phenylketonuria	0.05% 5:10,000	0.02% 2:10,000	0.03% 3:10,000

*National Rate 2006 is based on the sum of all reported presumptive positives divided by the sum of all the infants reported screened for the disease specified. This rate is converted from % to X:10,000 (rounded) for common reporting purposes. National data source: "2006 National Newborn Screening Report, Initial Screening Results," Biotinidase, Congenital Adrenal Hyperplasia, Congenital Hypothyroidism, Cystic Fibrosis, Galactosemia, MCAD, PKU newborns screened total column and newborns presumed with condition column. For Biotinidase Deficiency 22 states reported, CAH 26 states reporting, CH 29 states reporting, cystic fibrosis 15 states, galactosemia 29 states reporting, MCAD 24 states reporting and PKU 29 states reporting as of report run date of 7/23/07. Caution should be used in comparison of numbers.

**Nebraska's 5-year mean: is the mean of the 5 rates figured for each year individually for 2002 through 2006.

***Nebraska's rate 2006: is the number of presumptive positives divided by the total number of newborns screened in 2006.

CAVEAT: States use varying instruments, methodologies and cut-offs. In addition, the national data report identifies inconsistencies in reporting by some states which brings into question the validity of the data. Therefore, direct correlations cannot be made from the data that are available. However, from the summary of data on the next page, one can extrapolate that in general, Nebraska's chosen technology, methodologies and cut-offs have resulted in positive screening rates that are reasonable compared to other newborn screening programs across the country. Rates for hemoglobinopathies were not figured due to variances in reporting methods for the national report, and from states. The national database uses data submitted by individual states, and can be found at <http://www2.uthscsa.edu/mnsis/>.

Mean Averages of Laboratory Test Measures

The program continues to provide lab testing data to the Newborn Screening Advisory Committee to monitor ongoing quality. The following tables depict the quarterly mean averages for biotinidase measures, T₄ the primary screen for Congenital Primary Hypothyroidism, 17-OHP for congenital adrenal hyperplasia, Immunoreactive trypsinogen for CF and GALT and total galactose used to screen for Galactosemia. Access to data for mean averages for PKU and MCAD is not available from the Tandem Mass Spectrometry results from Pediatrix Screening Laboratory. These means can tell us something about stability of the assay, reagents etc. over time. Health care providers familiar with the mean averages, might feel more comfortable explaining the "relative risk" to parents of newborns with positive screening results, by comparing how far out of range the result is from the mean average, and from the normal/expected range.

	Jan-Mar 2006	Apr-Jun 2006	Jul-Sep 2006	Oct-Dec 2006
Biotinidase mean averages	50.94	45.209	45.24	56.035

	Jan-Mar 2006	Apr-Jun 2006	Jul-Sep 2006	Oct-Dec 2006
17-OHP mean averages for CAH	17.708	17.195	15.847	17.415

	Jan-Mar 2006	Apr-Jun 2006	Jul-Sep 2006	Oct-Dec 2006
Extracted 17-OHP mean averages for CAH	29.267	12.467	16.019	15.3

	Jan-Mar 2006	Apr-Jun 2006	Jul-Sep 2006	Oct-Dec 2006
T ₄ mean averages for CH	16.266	16.045	16.389	17.273

	Jan-Mar 2006	Apr-Jun 2006	Jul-Sep 2006	Oct-Dec 2006
TSH mean averages for CH *	6.765*	6.312*	6.711*	6.652*

*Averages do not include TSH's > 220

	Jan-Mar 2006	Apr-Jun 2006	Jul-Sep 2006	Oct-Dec 2006
IRT mean averages for Cystic Fibrosis	24.187	21.251	21.691	25.658

	Jan-Mar 2006	Apr-Jun 2006	Jul-Sep 2006	Oct-Dec 2006
Galt mean averages for Galactosemia	334	303	292	352

	Jan-Mar 2006	Apr-Jun 2006	Jul-Sep 2006	Oct-Dec 2006
Total galactose averages for Galactosemia	3.14	3	2.9	3.086

Home Births

For 2006, there were 69 home births reported to the Department of Health and Human Services Regulation and Licensure's Newborn Screening Program (some reported later in 2007). All 69 reported births were screened.

NEWBORN SCREENING OUTCOME DATA

	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Total Births	23,631	23,862	24,209	24,958	25,109	25,515	26,067	26,443	26,349	26,898
Births Screened	23,627 99.9%	23,858 99.9%	24,118 99.9%	24,863 99.6%	25,043 99.7%	25,478 99.85%	26,008 99.77%	26,391	26,288	26,819
Total Births Lost to Follow-up	4	4	9	6 + (89 not screened -as expired @ <48 hours.)*	2 + (64 not screened as expired @ < 48 hours)	5 + (32 not screened as expired @ < 48 hours)	5 + (54 not screened as expired @ < 48 hours)	2 + (50 not screened as expired @ < 48 hours)	0 + (61 not screened as expired @ < 48 hours)	2 + (79 not screened as expired @ < 48 hours)
Total Births PP	1,140	547	357	412	432	456	415	499	503	537
Home Births	90	83	86	109	93	99	70	60	55	69
Home Births Screened	86	81	77	105	88	95	65	60	54	69
Home Births Lost to follow-up¹	4	2	9	4	2 + (3 expired)	2 + (2 expired)	3 + (2 expired)	0	0 + (1 expired)	0

*Began match with death records beginning in calendar year 2000, to more accurately report #s actually screened.

Biotinidase Deficiency	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Presumptive Positive	5	3	4	2	4	3	4	34*	78	14
Confirmed Negative	2	2	2	2	1	1	0	29	71	9
Confirmed Positive Profound	1	1	1	0	0	2	1	0	1	0
Confirmed Positive (Partial no tx)	0	0	0	0	0	0	0	0	0	0
Confirmed Positive (Partial tx)	2	0	1	0	3	0	3	6	5	4
Lost to follow-up	0	0	0	0	0	0	0	0	1**	1**

*Screening protocols identified most of these as “inconclusive,” for which repeat screening rather than confirmatory testing, ruled out the condition.

** lost to follow-up as newborn expired

Congenital Adrenal Hyperplasia	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Presumptive Positive	N/A	10								
Confirmed Negative	N/A	9								
Confirmed Positive	N/A	1								
Confirmatory Lost to follow-up	N/A	0								

Congenital Primary Hypothyroidism	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Presumptive Positive	771	274	108	114	115	129	89	63	58	51
Confirmed Negative	746	265	92	104	105	113	75	55	48	41
Confirmed Positive	10	6	13	8	7	15	11	8	9	10
Confirmatory Lost to follow-up	15	3	3	2*	3*	1*	3*	0	1*	0

*Lost to follow-up as babies expired.

Cystic Fibrosis	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Presumptive Positive on Initial Screen	N/A	7								
Inconclusive on Initial Screen (repeat recommended)	N/A	62								
CF suspected due to prenatal testing	N/A	1								
meconium ileus or other bowel obstruction	N/A	1								
Confirmed Positive for CF	N/A	11								

Cystic Fibrosis (Continued)	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Confirmed Positive for Atypical CF	N/A	2								
Confirmed Positive CF Carrier	N/A	10								
Confirmatory Lost to follow-up	N/A	0								
Confirmed Negative	N/A	47								
Pending diagnosis	N/A	1								

Galactosemia	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Presumptive Positive	43	9	13	12	15	5	3	9	1	8
Confirmed Negative	29	9	8	8	9	5	0	6	1	8
Confirmed Positive (Classical)	0	0	0	1	0	0	1	0	0	0
Confirmed Positive, Duarte (not treated)	6	0	3	1 Duarte Hmzgt	0	0	1	0	0	0
Confirmed Positive, Duarte (treated)	6	0	2	2 Duarte Mixed Htrzgt. (1 tx'd 1 year)	6 Duarte Mixed Htrzgt.	0	1	3	0	0
Confirmed Neg. Classical/CP carrier	1	0	0	8	0	0	0	0	0	0
Confirmatory testing not done¹	1	0	0	0	0	0	0	0	0	0

Hemoglobinopathies		1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
SICKLE CELL DISEASE FS Screened positive	3	1	3	2	4	4	5	0	1	3	
Confirmed Positive	3	1	3	2	4	4	5	0	1	3	
SICKLE CELL TRAIT FAS Screened positive	88	54	120	139	146	156	150	171	186	206	
Confirmed Positive	40	54	60	104	102	111 (+1 other variant)	102	81	115	184	
Diagnosis Unknown	48	0	60	35	44	45	48	90	71	22	
OTHER CLINICALLY SIGNIFICANT Screened positive	-	1	3	14	21	2	1	4	6	3	
Confirmed Positive	-	1	1	2	3	2	1	2	6	3	
OTHER HEMOGLOBIN VARIANTS Screened positive	-	30	228	106	145	150	153	205	162	163	

MCAD *	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Screened Positive	N/A	N/A	N/A	N/A	N/A	3*	3	5	10	5
Confirmed Negative	N/A	N/A	N/A	N/A	N/A	2	3	1	7	5
Confirmed Positive	N/A	N/A	N/A	N/A	N/A	1	0	4	3	0

*Mandatory screening for MCAD began 7/01/2002. Prior to that about 34% of newborns were voluntarily screened in Nebraska in 2000 and 2001.

Phenylketonuria (PKU)		1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Presumptive Positive	137	43*	3	6**	4	3	7**	7	3	6	
Confirmed Negative	106	40	0	2	2	1	1	1	1	1	
Confirmed Positive Classical PKU	3	2	1	1	1	1	2	1	2	0	
Confirmed Positive Hyperphe	4		2 (tx'd)	1 transient	1	1	3	5 (3 of these tx'd)	0	5 (4 of these treated)	

*1998: One confirmatory testing not done – residence in another state

**2000 and 2003: One each year for whom confirmatory testing was not done as the babies expired

Tandem Mass Spectrometry Supplemental Screening Results

SUMMARY OF MS/MS FINDINGS Jan 1 – Dec 31, 2006

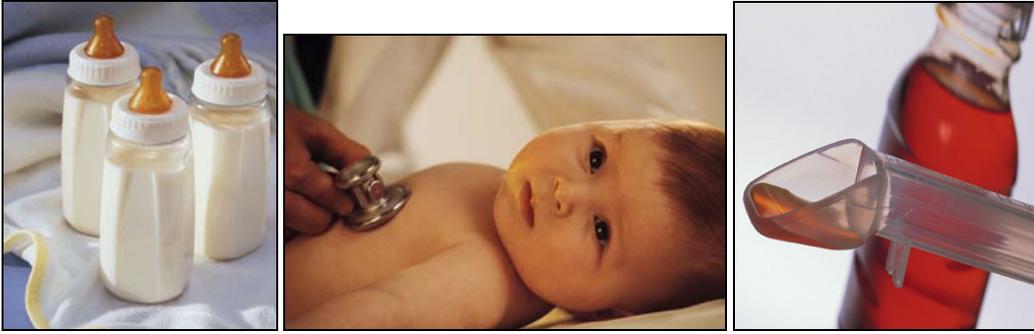
(Including MCAD and PKU, MS/MS screened conditions on the required screen)

Numbers include a few babies with one abnormality on screen and a different abnormality on repeat.

Initial findings	Out of Range on Screen	Repeated or Confirmed negative	Confirmed positive	Pending (P) or Lost to Follow-up (LTF)
C3	11	11		
C3 & C3/C2 & C3/C16	15	13	1 with presumed cobalamin defect	1 pending
C4	24	23	1 with IBCD (Isobutyryl co-A Dehydrogenase Deficiency)	
C6DC	1	1		
C8	5	5		
Several Amino Acids (hyperlimentation often)	55	51		1 pending, 3 expired before repeat completed
Methionine	57	56		1 pending
Methionine & Citrulline	1	1		
Methionine & Tyrosine	5	4		1 expired before repeat
Phenylalanine & Phe/Tyrosine ratio	6	1	4 hyperphe - tx'd 1 mild hyperphe - not tx'd	
Methionine and Methionine/Phe ratio	3	3		
Tyrosine	25	22	2 Transient tyrosinemia	1 pending
Leucine/Isoleucine & others	1	1		
Short & Medium Chain Acylcarnitines	4 + 2 on repeats	6		
C5OH	1		1 3-MCC (3-methylcrotonyl carboxylase deficiency)	
Free & Short Chain Acylcarnitines	1	1		
C3 & C3/C16	1	1		
C5 and C5/C4	1	1		
C10 OH	1	1		
C10 OH, C10, and C8	1	1		
Methionine & C3	1	1		
C12	1	1		
TOTALS	223	205	10	4 pending, 4 expired

*Lost to follow-up designated when the patient/parent can no longer be found and there is no medical home, or they have moved out of state to an unknown location.

**The vast majority of abnormal screens from MS/MS require only a repeat screen to rule out the condition. Confirmatory testing is recommended in a small percentage of cases where the concentration of analytes are "significantly" abnormal, or concentrations of analytes increase on repeat screens.



Intervention Data

Intervention data is one of the most important measures for determining how well we are doing as a system to ensure timely treatment of affected infants.

The following data is grouped by condition and shows Nebraska's averages/ranges for 2006. In some cases "intervention" (family consultation, evaluation, and monitoring of the newborn) occurred well before the age actual treatment was initiated, as treatment was pending confirmatory testing and diagnosis.

The data also includes national averages/ranges According to the most recent available data "National Newborn Screening Report -2006" available at the National Newborn Screening and Genetics Resource Center's Web site. Data for this section of the Annual Report was run on 7/24/07.

<http://genes-r-us.us.uthscsa.edu/resources/newborn/00chapters.html>

Comparisons should be made with extreme caution. States and territories included in the averages in this report have birth numbers from fewer than 2,000 per year to around 500,000 per year. Likewise, resources necessary to complete testing, follow-up, confirmation, diagnosis and treatment also vary from state to state. The intervention data is one kind of outcome data that can, over time, help identify how well a state's system is working in newborn screening. The mean average age at time of treatment can be an indicator of whether adequate resources are devoted to each of the components of a comprehensive newborn screening system: education, specimen collection handling and transportation procedures, laboratory procedures, follow-up and referral procedures, confirmation and treatment.

Biotinidase Deficiency

Nebraska 2006 Intervention Data	U.S. 2006 Intervention Data
Goal age for treatment initiation: Upon Diagnosis	35 States reported data.
# diagnosed/treated: 0 profound 4 partial deficiency's treated	30 cases of profound biotinidase deficiency reported
Mean avg. age at Initiation of treatment: 21 days	2 cases or 7% treated by 7 days of age 13 or 44% treated between 8-14 days of age 3 or 10% treated between 15-21 days of age 8 or 27% treated at > 21 days of age 4 or 14% age of treatment unknown/not reported
Range of ages at Initiation of treatment: 13-26 days	Range of ages at Tx initiation.: 4 - > 21 days

Source: <http://www2.uthscsa.edu/nnsis/> data entered by 7/24/07

Congenital Adrenal Hyperplasia

Nebraska 2006 Intervention Data	U.S. 2006 Intervention Data
Goal age for treatment initiation: Upon Diagnosis	42 States reported data.
# diagnosed/treated: 1	125
Mean average age at Initiation of treatment: 10 days	53 cases or 43% treated by 7 days of age 43 cases or 35% treated by 8-14 days of age 3 cases or 2% treated by 15-21 days of age 13 cases or 10% treated at > 21 days of age 13 or 10% age of treatment unknown/not reported
Range of ages at initiation of treatment (N/A)	Range of ages at initiation of treatment: <3 - > 21

Source: <http://www2.uthscsa.edu/nnsis/> data entered by 7/24/07

Congenital Primary Hypothyroidism

Nebraska 2006 Intervention Data	U.S. 2006 Intervention Data
Goal age for treatment initiation: As early as possible, upon diagnosis.	47 States reported data.
# diagnosed/treated: 10	# diagnosed/treated: 1521
Mean avg. age at Treatment initiation: 10 days	Age at initiation of treatment: 364 cases or 24% treated by 7 days of age 490 cases or 32% treated between 8-14 days of age 181 or 12% treated between 15-21 days of age 304 or 26% treated at > 21 days of age 88 or 6% age at tx. unknown or not reported
Range of ages at treatment initiation: 6-18 days	Range of ages at treatment initiation < 3 - > 21 days

Source: <http://www2.uthscsa.edu/nnsis/> data entered by 7/24/07

Cystic Fibrosis

Nebraska 2006 Intervention Data	U.S. 2006 Intervention Data
Goal age for treatment initiation: As early as possible, upon diagnosis.	15 States reporting data
# diagnosed/treated: 11 Classical CF 2 Atypical CF	# diagnosed/treated: 132
Mean average age at initiation of treatment: 29.33 days Excluding the outliers: one diagnosed prenatally and one delayed diagnosis at 226 days, the mean average age of intervention/treatment was 12.5 days.	Age at initiation of treatment: 28 cases or 21% treated by 15 days 21 or 16% treated at 16-30 days 10 or 8% treated at 31-45 days 2 or 1.5% treated at 46-60 days 2 or 1.5% treated at 61-75 days 2 or 1.5% treated at 76-90 days 9 or 7% treated at > 90 days 58 or 44% age of tx. unknown or not reported
Range in age at initiation of treatment: 1-226 days (1 @ 226 days -delay due to social situation and dx. requiring observation & DNA sequencing).	Range in age at initiation of treatment 1->90 days

Source: <http://www2.uthscsa.edu/nnsis/> data entered by 7/24/07

Galactosemia

Nebraska 2006 Intervention Data	U.S. 2006 Intervention Data
Goal age for treatment initiation: As early as possible, upon diagnosis. Diet intervention upon positive screening result	44 States reporting data
# diagnosed/treated: 0	46 cases of classical galactosemia identified
Mean avg. age at treatment initiation: N/A	Age at treatment: 25 or 54% treated at 7 days of age or less 6 or 13% treated between 8-14 days of age 2 or 4% treated between 15-21 days of age 3 or 6.5% treated at >21 days of age 10 or 22% age of treatment unknown or not reported
Range of ages at Tx. initiation: N/A	Range of age at treatment initiation: < 3 days - > 21

Source: <http://www2.uthscsa.edu/nnsis/> data entered by 7/24/07

MCAD - Medium Chain Acyl Co-A Dehydrogenase Deficiency

Nebraska 2006 Intervention Data	U.S. 2006 Intervention Data
Goal age for treatment / intervention initiation: As early as possible, upon positive screening result – parent education/consultation.	41 States reported data
# diagnosed/treated: 0	# diagnosed/treated: 167
Average age at intervention (avoid fasting): N/A	Age from birth to treatment: 59 or 35% 7 days or less 41 or 25% between 8-14 days of age 12 or 7% between 15-21 days of age 21 or 13% at > 21 days of age 34 or 20% age treatment unknown
Range in age at treatment: N/A	Range in age at treatment: <3 -> 21 days

Source: <http://www2.uthscsa.edu/nnsis/> data entered by 7/24/07

PKU - Phenylketonuria (Classical PKU)

Nebraska 2006 Intervention Data	U.S. 2006 Intervention Data
Goal age for treatment initiation: As soon as possible but no later than 7-10 Days after birth.*	46 States reported data
# classical PKU: 0 # hyperphenylalaninemia (treated): 4	131 Cases of classical phenylketonuria
Avg. age at treatment (hyperphe): 9 days	56 or 43% treated by 7 days of age 46 or 35% treated between 8-14 days of age 9 or 7% treated between 15-21 days of age 13 or 10% treated at > 21 days of age 7 or 5% age at treatment unknown or not reported Data on age at treatment for clinically significant cases of hyperphenylalaninemia not collected nationally.
Range ages at treatment (hyperphe): 4-16 days 50% treated by 7 days of age	Ranges in ages at treatment: < 3 -> 21 days 43% Treated by 7 days of age

*NIH Consensus Statement October/25/2000: Phenylketonuria: Screening and Management

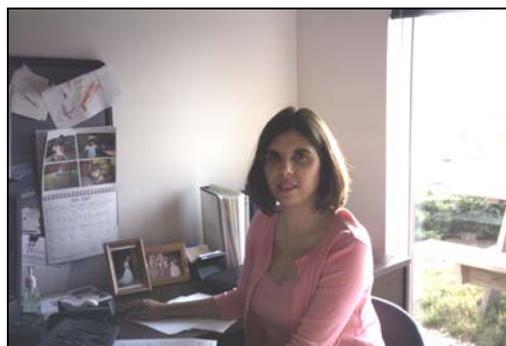
Source: <http://www2.uthscsa.edu/nnsis/> data entered by 7/24/07

Hemoglobinopathies

Nebraska 2006 Intervention Data	U.S. 2006 Intervention Data ¹
Goal age for treatment initiation: ² 60 days of age or less	46 States reported data
# cases diagnosed/treated Sickle Cell disease (S/S) 3 Sickle Hgb. C disease (S/C) 2 E Beta Thalassemia (E/Beta-thal) 1	# cases diagnosed/treated Sickle Cell Disease (S/S) 680 Sickle Hgb. C Disease (S/C) 384 E-Beta Thalassemia (E/Beta-thal) 22
Mean/Average age (days) at treatment: Sickle Cell Disease (S/S): 36 days Sickle Hgb. C disease (S/C): 19.5 days Hgb. E Beta-thalassemia (E/Beta-thal): 18 days	154 or 23% S/S treated between 0-30 days 135 or 20% S/S treated 31-60 days 99 or 15% S/S treated 71-90 days 69 or 10% S/S treated > 90 days 223 or 33% S/S age at treatment unknown or not reported 81 or 21% S/C Disease treated between 0-30 days 78 or 20% S/C Disease treated 31-60 days 68 or 18% S/C Disease treated 71-90 days 42 or 11% S/C Disease treated > 90 days 115 or 30% S/C Disease age treatment unknown or not reported 1 or 4.5% E/Beta Thal treated between 0-30 days 21 or 99.5% of E/Beta Thal age at treatment unknown or not reported
Range of ages (days) at treatment: 9-84 days	Range of ages (days at treatment) 0 - > 90
5/6 or 83% treated by 60 days of age.	449/1086 or 41% of S/S, S/C and E-Beta thal cases treated by 60 days of age.

Source: <http://www2.uthscsa.edu/nnsis/> data entered as of 7/24/07

² Treatment guideline from A Clinical Practice Guideline #6, Sickle Cell Disease: Screening, Diagnosis, Management and Counseling in Newborns and Infants, U.S. Dept. Of Health and Human Services, Public Health Service, Agency for Health Care Policy and Research.



Helping to ensure rapid turn-around times are James DiPerna MS/MS Laboratory Supervisor (above left) , and Bethy Sgroi M.S.(above right) one of the three genetic counselors from Pediatrix Screening Laboratory Inc., who quickly phone the positive or abnormal screening results to the Nebraska Newborn Screening Program, submitters and newborns' physicians.

PLANS



Screening Panel Expansion: Nebraska now screens nearly 100% of newborns for eight conditions and greater than 97% of newborns for the additional organic acid, fatty acid and amino acid Disorders that can be detected on Tandem Mass Spectrometry screening. The Newborn Screening Advisory Committee and the Department will consider the recommendation of the March of Dimes to require screening for those conditions on the supplemental screening panel that are screened by tandem mass spectrometry.

Other System Planning Efforts: The Nebraska Newborn Screening Advisory Committee will advise the Department of Health and Human Services Regulation and Licensure on implementation of elements of the National Newborn Screening & Genetics Resource Center's "Performance Evaluation and Assessment Scheme." The program will work to enhance the disaster preparedness/contingency plan. Also there are plans to develop a minimum data set for long term follow-up data collection and analysis of a sub-set of patients identified through newborn screening will occur. Collection and analysis of long term follow-up data can inform the system on the effectiveness of newborn screening, successful strategies for positive outcomes, and provide a conduit of communication to assure continuity of services for children with special health care needs due to metabolic or other screened conditions.

CONTINUING ACTIVITIES

Education: Educational activities by the NNSP will continue via publication of the Annual Report, and as needed through hospital and physician mailings. Opportunities for on-site education are always available upon request from hospitals. The program will distribute additional videos to public health and community-based prenatal care and prenatal education providers so expecting parents can be exposed to newborn screening during their third trimester.

Laboratory Testing: The contract with Pediatrix Screening laboratory is a one-year contract, renewable for five years. Annual renewals are dependent on the Department's assessment of contractor's performance. The laboratory will continue to pursue improved efficiencies for screening and collaborate with the Nebraska Newborn Screening Program to determine appropriateness of any proposed strategies. A new laboratory request for proposal process will begin in 2007 to competitively bid the contract for newborn screening laboratory testing services, to begin July 2008.

Follow-up, Tracking and Referral: The NNSP will continue to track every newborn to be sure they received an appropriate screen; to follow up on all transferred, drawn early, transfused, unsatisfactory, inconclusive and presumptive positive specimens; and to facilitate confirmatory testing and referral for diagnostic and treatment services. Ongoing and annual review and updating of short-term follow-up procedures will be completed.

Confirmatory Testing: The program will continue to work with specialists and the Newborn Screening Advisory Committee to ensure procedures recommended for confirmatory testing are communicated effectively to practitioners. The program will continue to use the ACMG ACT (Action Sheets) for all positive results from tandem mass spectrometry, to help physicians in Nebraska know what "next steps" to take when faced with a positive screening result for any of these rare conditions.

Diagnosis: Practitioners are strongly urged to consult with the pediatric specialist appropriate to the condition for which a newborn has a positive or abnormal screening result. The program will help link the newborn's primary care provider with specialists when needed.

Treatment: Access to treatment will continue to be an issue the program will monitor. The statutorily required payment for metabolic foods and formula will continue. The Program will continue to monitor the issues associated with access to treatment and seek ways to ensure funding is sufficient to meet affected individuals' needs.

Quality Assurance Monitoring: The Program and Advisory Committee will continue to review and act on quarterly quality assurance plan data as well as respond to trends identified with any problems in the interim periods. Quarterly QA reports and Quality Improvement Hints publications will continue to be sent to individual hospitals for their own evaluation and comparison with statewide numbers.

NEWBORN HEARING SCREENING

What is Newborn Hearing Screening?

Significant hearing loss is the most common birth defect with an estimated incidence rate of one to three per thousand live births. Before newborn hearing screening, many hearing losses were not diagnosed until 2 ½ to 3 years of age. Left undetected, hearing loss in infants can negatively impact speech and language acquisition, academic achievement, and social and emotional development. If detected early, however, the negative impacts can be diminished, and even eliminated, through early intervention.

Newborn hearing screening is an essential preventative public health program. It meets the following prerequisites for a population screening program –

- Condition is sufficiently frequent in the screened population
- Condition is serious or fatal without intervention
- Condition must be treatable or preventable
- Effective follow-up program is possible

In 2000, the Infant Hearing Act established newborn hearing screening in Nebraska. The statute requires birthing facilities to educate parents about newborn hearing screening, encouraged hospitals to voluntarily begin screening newborns for hearing loss and, by December 2003, to include hearing screening as part of the standard of care and to establish a mechanism for compliance review. The Act also required that regulations be promulgated to mandate newborn hearing screening if, by December 2003, less than 95% of newborns in the state were receiving a hearing screening. This report presents the status of newborn hearing screening in Nebraska during 2006 (see Nebraska Newborn Hearing Screening Data for 2006).

Newborn hearing screening requires objective physiologic measures to detect hearing loss in newborns and young infants. There are two basic techniques that birthing facilities in Nebraska use to screen newborns for hearing loss. Both are easily recorded in newborns and are noninvasive measures of physiologic activity that underlie normal auditory functioning.

The most frequently used screening technique is measurement of otoacoustic emissions, or OAEs. A miniature earphone and microphone are placed in the newborn's ear canal, low intensity sounds are presented, and responses produced by the inner ear are measured. The second screening technique, Auditory Brainstem Response, or ABR, uses small electrodes to detect certain brainwaves in response to sounds that are presented by a miniature earphone. For both methods, the response of each ear is measured. OAE and ABR are both reliable and accurate. Screening can occur as early as 12 hours of age, preferably with the newborn sleeping, and averages from five to 20 minutes to complete.

If a response is not detected for one or both ears, the result is a "refer" (did not pass). A "refer" to the screening test indicates that a hearing loss *may* exist but there are also other factors that may have contributed. A "refer" does indicate that a second screening is necessary to determine if the other factors, such as vernix in the ear canal, fluid in the middle ear cavity, movement,

equipment failures, or inexperience of the tester, contributed to the initial result. A “refer” on the second screening indicates the need for a diagnostic audiological evaluation to confirm or rule out a hearing loss and, if hearing loss is present, to begin to identify the type and degree of the loss.

Each birthing facility has established a newborn hearing screening protocol that identifies how the screening will be administered, the recording and reporting procedures, how refers will be handled, i.e., re-screen as an inpatient with the same or different screening technique or re-screen as an outpatient, and quality assurance measures.

EARLY HEARING DETECTION AND INTERVENTION SYSTEM

System Elements

The Newborn Hearing Screening Program in Nebraska strives to fulfill the four purposes of the Infant Hearing Act (Neb. Rev. Stat. §71-4735):

- “To provide early detection of hearing loss in newborns at the birthing facility, or as soon after birth as possible for those children born outside of a birthing facility,
- to enable these children and their families and other caregivers to obtain needed multidisciplinary evaluation, treatment, and intervention services at the earliest opportunity, and
- to prevent or mitigate the developmental delays and academic failures associated with late detection of hearing loss; and
- to provide the state with the information necessary to effectively plan, establish, and evaluate a comprehensive system for the identification of newborns and infants who have a hearing loss.”

Newborn hearing screening is one aspect of a comprehensive, integrated Early Hearing Detection and Intervention (EHDI) system. The first three principles of the Year 2000 Position Statement: Principles and Guidelines for Early Hearing Detection and Intervention Programs (Joint Committee on Infant Hearing, 2000) are:

1. All infants have access to hearing screening using a physiologic measure. Newborns who receive routine care have access to hearing screening during their hospital birth admission. Newborns in alternative birthing facilities, including home births, have access to and are referred for screening before 1 month of age. All newborns or infants who require neonatal intensive care receive hearing screening before discharge from the hospital. These components constitute universal newborn hearing screening (UNHS).
2. All infants who do not pass the birth admission screen and any subsequent re-screening begin appropriate audiologic and medical evaluations to confirm the presence of hearing loss before 3 months of age.
3. All infants with confirmed permanent hearing loss receive services before 6 months of age in interdisciplinary intervention programs that recognize and build on strengths, informed choice, traditions, and cultural beliefs of the family.

These three major principles serve as the foundation for the screening, referral, and audiological evaluation protocols developed by the Advisory Committee of the Nebraska Newborn Hearing Screening Program (NNHSP) in 2001. The guidelines established by the NNHSP Advisory Committee are for hearing screening to be completed during birth admission, audiological diagnostic evaluation to begin prior to six weeks of age to minimize the need for sedation, and appropriate early intervention activities to be initiated by six months of age. The logic model of the NNHSP (see Appendix A) describes the resources and activities needed to produce the projected results of the program.

The Early Hearing Detection and Intervention system in Nebraska is composed of five functional elements: Hearing Screening at Birth, Confirmatory Testing, Medical Evaluation, Early Intervention, and Tracking and Surveillance. One or more groups of professionals in a variety of settings assume responsibility of each element of the system. An overview of each of the elements and the primary activities are presented below. Included in this discussion are the Nebraska Revised Statute citations and the recommended protocols established by the Department of Health and Human Services through the Nebraska Newborn Hearing Screening Advisory Committee.

Hearing Screening at Birth

Birthing facilities in Nebraska have five primary activities related to screening the hearing of newborns:

1. The parent(s) of newborns are educated about the hearing screening, the likelihood of hearing loss in newborns, the importance of follow-up, community resources (including early intervention services), and normal auditory, speech and language development (Neb. Rev. Stat. §71-4740). If risk factors are present, hospital personnel educate parents to evaluate hearing every six months. *Note:* The Department of Health and Human Services is responsible for educating the parent(s) for newborns not born in a birthing facility (Neb. Rev. Stat. §71-4740).
2. A hearing screening test is part of each birthing facility's standard of care for newborns, effective 12/1/03 (Neb. Rev. Stat. §71-4742). Following hospital protocol for the procedure, each newborn's hearing in each ear is screened during birth admission using OAE and/or ABR screening techniques. A second inpatient screening is conducted within one to three weeks if the baby "refers" on the first screening. The outpatient re-screening for those that "refer" during birth admission may occur at the birthing facility or at a confirmatory testing facility.
3. A mechanism for compliance review is established for each birthing facility (Neb. Rev. Stat. §71-4742).
4. Results of the hearing screening for each newborn are reported to the newborn's Primary Care Provider. Weekly tracking reports are submitted to the NNHSP that identify newborns who "refer," transfer, or discharge without a hearing screening.
5. Annual reports are submitted to the NNHSP that indicate the following numbers: born in the birthing facility, recommended for screening, received screening during birth

admission, passed screening, did not pass screening, and recommended for monitoring and follow-up (Neb. Rev. Stat. §71-4739).

Confirmatory Testing

Newborns who have referred for one or both ears on the second hearing screening should receive an audiological diagnostic evaluation prior to reaching three months of age. The purpose of this evaluation is to confirm the presence of a hearing loss and to determine the type and degree of the hearing loss. The primary recommended activities that comprise the confirmatory testing component are:

1. An initial diagnostic evaluation using either OAE or ABR conducted as early as possible after referral, preferably before the infant is six weeks old. If the infant “passes” this initial part of the evaluation (outpatient re-screening), no further evaluation is usually needed.
2. If the infant “refers” on the initial part of the evaluation, the testing often proceeds immediately to a comprehensive diagnostic evaluation. This evaluation minimally includes measures of middle ear function (high frequency tympanometry), auditory sensitivity (air- and bone-conducted ABR), confirmatory measures (parent observations) and, depending upon the developmental age, behavioral audiological assessment (Visual Reinforcement Audiometry). Other measures may be included, as indicated.
3. Depending upon a variety of factors, referrals are made for further evaluation, diagnosis, treatment, and services. These referrals may be made to medical specialists and/or early intervention services.
4. Results of the initial and comprehensive audiological diagnostic evaluation are provided to the Primary Care Physician and NNHSP.
5. Annual reports are submitted to the NNHSP that indicate the number of newborns: who return for follow-up testing, who do not have a hearing loss and who do have a hearing loss (Neb. Rev. Stat. §71-4739).

Medical Evaluation

The infant’s Primary Care Provider (PCP) has the key role in the follow-up for those who “refer” on the initial hearing screening during the birth admission. Building on the concept of a Medical Home (Guidelines for Pediatric Medical Home Providers, American Academy of Pediatrics), the PCP has the primary role in identifying and accessing the medical and non-medical services needed to help children and their families achieve their maximum potential. The primary activities that comprise the medical element of the newborn hearing screening system are:

1. Birthing hospital notifies PCP of the newborn’s hearing screening results.
2. NNHSP notifies PCP about the hearing screening status and need for follow-up evaluation for those that did not pass the inpatient hearing screening or were discharged without a screening.
3. PCP or designee per hospital procedure informs parents of hearing screening results and need for re-screening.
4. PCP (or staff), hospital, or test provider schedules re-screen appointment to be completed

- in one to three weeks and notifies parents.
5. Provider of outpatient re-screening notifies PCP of results.
 6. PCP notifies NNHSP of outpatient hearing re-screening results.
 7. If “refer,” PCP makes referral for comprehensive diagnostic evaluation, educates parents about need for evaluation, and makes referral to Early Intervention services.
 8. If hearing loss is confirmed, PCP or diagnostic evaluator refers newborn/infant for complete medical and/or neuro-sensory evaluation and Early Intervention Services.

Early Intervention

Early intervention is an individualized program of services and supports based on the needs of the individual and family. Part C of the Individuals with Disabilities Education Act (IDEA) authorizes the creation of early intervention programs for infants and toddlers with disabilities. In Nebraska, the Early Development Network (EDN) provides services coordination for eligible families to identify and link with needed services, to work with multiple providers to ensure that services are provided, and to become Coordinators of services in the future. The recommended protocols for the primary early intervention activities within the Early Hearing Detection and Intervention system are:

1. PCP or diagnostic evaluator makes referral to Early Development Network (EDN).
2. EDN reviews for eligibility.
3. If eligible, EDN may provide assistance with diagnostic evaluation and treatment.
4. Services Coordinator may facilitate obtaining services from otologists, audiologists, community services, and others.

Tracking and Surveillance

The Nebraska Newborn Hearing Screening Program has been developed based on the requirements identified in the Infant Hearing Act (Neb. Rev. Stat. §71-4735 - §71-4744) and the NNHSP Advisory Committee’s recommended protocols to “...determine and implement the most appropriate system...to track newborns and infants identified with a hearing loss” and “...to effectively plan and establish a comprehensive system of developmentally appropriate services for newborns and infants who have a potential hearing loss or who have been found to have a hearing loss and shall reduce the likelihood of associated disabling conditions” (Neb. Rev. Stat. §71-4737). Activities of the NNHSP include:

1. Develop, implement, and monitor statewide systems to track newborns with or at-risk of hearing loss (Neb. Rev. Stat. §71-4737) and adopt and promulgate rules and regulations to implement the Infant Hearing Act (Neb. Rev. Stat. §71-4742 and §71-4744).
2. Gather required data and generate annual reports (Neb. Rev. Stat. §71-4739 and §71-4741).
3. Establish guidelines for referral to early intervention services (Neb. Rev. Stat. §71-4743).
4. Educate parents with out-of-hospital births about newborn hearing screening (Neb. Rev. Stat. §71-4740).
5. Apply for all available federal funding to implement the Infant Hearing Act (Neb. Rev. Stat. §71-4738).

NEWBORN HEARING SCREENING DATA FOR 2006

Birthing Facilities Data for 2006

Birthing Facility Screening Programs

The number of birthing facilities conducting newborn hearing screening increased rapidly since 2000 when only 11 hospitals were conducting either targeted or universal newborn hearing screening. In 2006, 100% of the birthing facilities in Nebraska were conducting hearing screenings, consistent with the Neb. Rev. Stat. §71-4742 requirement that a hearing screening test be included as part of the standard of care for newborns. Sixty two (62) of the 63 birthing hospitals conducted the hearing screening during the birth admission and one conducted the screening on an outpatient basis following discharge.

Birthing Facilities Conducting Newborn Hearing Screenings (2000-2006)

Year	Number of Birthing Facilities in Nebraska	Number Conducting Newborn Hearing Screening	Percentage Conducting Newborn Hearing Screening
2000	69	11	16%
2001	69	24	35%
2002	69	57	83%
2003	67	67	100%
2004	67	67	100%
2005	65	65	100%
2006	63	63	100%

Table 1

Annual Birthing Facilities Reports

Birthing facilities are required to annually report specific information about their newborn hearing screening programs to the Department of Health and Human Services Regulation and Licensure (Neb. Rev. Stat. §71-4739).

Birthing Facility Reports of Required Aggregate Data (2006)

Number of newborns born	26,854*
Number of newborns and infants recommended for a hearing screening test	26,303
Number of newborns who received a hearing screening during birth admission	26,615**
Number of newborns who passed a hearing screening during birth admission, if administered	25,657**
Number of newborns who did not pass a hearing screening test during birth admission, if administered	999**
Number of newborns recommended for monitoring, intervention, and follow-up care	961**

*Aggregate data reported by hospitals; number of births registered with Vital Records is 26,898.

**Figures include hearing screenings for babies transferred to Children's Hospital

Table 2

The data in Table 2 are based on annual aggregate data reported by the birthing facilities. Individual screening results and demographic data are not reported for all births. The NNHSP only receives specific information about newborns that “refer” on the initial hearing screening and about those that were discharged to home without receiving a hearing screening. The opportunity for error exists within the manual tracking system due to reporting errors, recording errors, duplicated entries because of newborn name changes, transfers from birth hospitals to NICUs, and whether newborns who expire are included in the weekly and/or annual reports. Without a system to accurately determine the status of each newborn’s hearing screening results, errors will be present in spite of the best efforts of everyone involved to provide accurate information.

Parent Education

Recommending a hearing screening test has been operationally defined as educating parents about newborn hearing screening, as required by Neb. Rev. Stat. §71-4740. The NNHSP provides print and video education materials free of charge to hospitals to help fulfill this requirement. Almost all parents (26,303 or 97.9%) received education about newborn hearing screening in 2006.

Newborns Receiving a Hearing Screening

The Infant Hearing Act requires that rules and regulations be adopted and promulgated if the annual percentage rate of newborns who receive a hearing screening during birth admission is less than 95% by December 1, 2003, or at any time thereafter. The annual aggregate reports submitted by the hospitals in 2006 show that 98.9% of the 26,898 births registered with Vital Records were screened during birth admission or prior to discharge to home. The numbers of newborns screened during birth admission increased dramatically since reporting began in 2000, when only slightly more than one third of newborns received a hearing screening during birth admission (see Table 3). This increase in the numbers of newborns receiving a hearing screening corresponds to the increase in the number of hospitals adopting newborn hearing screening as the standard of care for newborns and the support of sub-grants through the Nebraska Health Care Cash Fund to purchase screening equipment in 2002 and 2003.

Newborns Receiving a Hearing Screening (2000-2006)

Year	2000	2001	2002	2003	2004	2005	2006
Number Receiving a Hearing Screening during Birth Admission	8,978	15,272	22,615	25,275	25,966	26,179	26,615
Percent Receiving a Hearing Screening during Birth Admission	36%	61%	89%	97%	98%	99%	99%

Table 3

Newborns Discharged Without a Hearing Screening

During 2006, the annual aggregate hospital reports to NNHSP indicated that there were 243 newborns who did not receive a hearing screening during birth admission because of parent refusal (8), expired prior to screening (103), or discharge to home prior to screening (132). The NNHSP also tracked 693 newborns who were transferred to Neonatal Intensive Care Units in Nebraska and surrounding states prior to receiving a hearing screening.

Birth Admission Refer Rates

The annual aggregate reports received from the birthing facilities indicated that 999 newborns did not pass (“refer”) the hearing screening during birth admission. Of the newborns with hearing screenings conducted during the birth admission, the refer rate for all birthing facilities was 3.8% during 2006, which compares favorably with refer rates for previous years at Nebraska birthing facilities (see Table 4).

Birth Admission Refer Rates (2002-2006)

	2002	2003	2004	2005	2006
Refer rate for all birthing facilities	3.7%	3.6%	3.5%	3.4%	3.8%

Table 4

As discussed previously in this report, there are two measurement techniques used to conduct newborn hearing screening: Otoacoustic Emissions (OAE) and Auditory Brainstem Response (ABR). Almost half of the birthing hospitals in Nebraska are using OAE-only, almost one third are using ABR-only, and the remaining birthing hospitals are using a 2-step method (OAE, followed by ABR if the initial screening is a “refer”). The “refer” rates differ for the three approaches, with the OAE-only having the highest refer rate (see Table 5).

Refer Rates for Hearing Screening Techniques (2006)

	OAE-only	ABR-only	2-Step
Number of Birthing Facilities	30	23*	11
Number of Newborns Screened	2,914	9,465	14,236
Number of “Refers”	322	355	322
Refer Rate	11.1%	3.8%	2.3%

*includes Children’s Hospital

Table 5

Monitoring, Intervention, and Follow-up

The final aggregate data reported by the birthing facilities is the number of newborns recommended for monitoring, intervention, and follow-up care: 709 (85% of refers) in 2002, 676 (74% of refers) in 2003, 793 (86% of refers) in 2004, 863 (95% of refers) in 2005, and 961 (96% of refers) in 2006.

Audiological/Confirmatory Test Provider Data for 2006

Neb. Rev. Stat. §71-4739 requires confirmatory testing facilities to report the following:

- Number of newborns and infants who return for a follow-up hearing test
- Number of newborns and infants who do not have a hearing loss based upon the follow-up hearing test
- Newborns and infants who are shown to have a hearing loss based upon the follow-up hearing test

The Advisory Committee for the NNHSP identified the initial level of the follow-up hearing test as an outpatient re-screening of the newborn’s hearing. For those newborns and infants who pass this initial level of the follow-up hearing test, no further audiological evaluation would be needed, unless there are risk factors present that would warrant periodic monitoring. The Advisory Committee recommends that the re-screening occur within the first six weeks to minimize the need to sedate the infant to obtain reliable results and so that intervention can begin early if a hearing loss is identified.

Since the majority of newborns will pass this second screening, considerable cost savings can result by using either the OAE and/or ABR screening technique rather than proceeding directly to a complete diagnostic audiological evaluation. The Advisory Committee’s Audiological Diagnostic Protocol recommends that the referral center should be prepared to provide comprehensive audiological diagnostic procedures if the outpatient re-screening results indicate a “refer” status. However, some communities that do not have pediatric audiology services readily available have opted to have the initial re-screening occur at the birthing facility on an outpatient basis.

Annual Confirmatory Testing Facility Reports

Each year data regarding the follow-up hearing tests at confirmatory testing facilities have been gathered by surveying the audiologists in Nebraska. Twenty seven (27) of 28 confirmatory testing facilities responded, representing 65 licensed audiologists. The results of those surveys for 2006 are included in Table 6.

Required Follow-up Hearing Test Data Reported by Audiologists

	Re-screenings	Diagnostic Evaluations
Number of newborns/infants receiving a follow-up hearing test	656	174
Number of newborns/infants without a hearing loss	500	71
Number of newborns/infants with a hearing loss	155 (“refer”)	103

Table 6

Rate of Follow-up Outpatient Screening and Confirmatory Testing

Since 37 birthing hospitals conducted initial outpatient re-screenings, those figures are important to present a comprehensive view of the follow-up services being provided in Nebraska. In aggregate reports, the birthing facilities indicated that 343 newborns had received outpatient hearing screenings and confirmatory testing facilities indicated that 656 newborns received screenings for a total of 999 outpatient screenings. With aggregate reporting, it is not possible to determine an unduplicated count, since some infants, especially those with middle ear dysfunction and an accompanying transient conductive hearing loss, may be screened multiple times at one or more sites.

Diagnosis of Hearing Loss

The number of infants diagnosed with a hearing loss in Nebraska is reported in two ways: 1) aggregate reports submitted by audiologists of the number of infants shown to have a hearing loss based on follow-up tests (required by Neb. Rev. Stat. §71-4739) and 2) the individual diagnostic reports submitted to NNHSP by audiologists or Primary Care Providers. Statutory authority to require audiologists to report on all newborns and infants that receive audiological evaluations does not exist, so a one-to-one correspondence between the individual results reported to NNHSP and the required annual aggregate reporting does not exist. Audiologists reported conducting 174 diagnostic evaluations of infants born in 2006, identifying 103 infants with transient or permanent hearing loss.

Type and Degree of Hearing Loss

Analysis of the aggregate confirmatory testing reports submitted to NNHSP indicates that 72 of the infants with hearing loss meet the criteria for a Permanent Hearing Loss (PHL) (Note: aggregate reports may include duplicate entries). Fifty eight (58) of the infants were identified with a bilateral hearing loss, 69% in the mild to moderate range and 31% in the severe to profound range. The remaining 14 infants were identified with a unilateral hearing loss, 86% of which were in the mild to moderate range. The aggregate reports indicated that 35 infants had been fit with amplification.

Type and Degree of Permanent Hearing Loss, 2006 (n = 72)

Degree ► Type ▼	Bilateral Mild–Moderate	Bilateral Severe–Profound	Unilateral Mild–Moderate	Unilateral Severe–Profound
Sensorineural	34	17	8	2
Conductive	0	-	3	-
Mixed	6	1	1	0

Table 7

The estimates of incidence of permanent hearing loss in newborns range between 1 to 3 per thousand births nationally. Based on the birth rate in Nebraska during 2006 (26,898), an estimated 27 to 81 newborns would be identified with PHL. The incidence of PHL in Nebraska for young children born in 2006 reported in aggregate reports is no more than 2.7 per thousand.

Analysis of Individual Reports Received by NNHSP

The data presented in the two previous sections are based on mandated annual aggregate reports from birthing facilities and confirmatory testing facilities. The individual patient-specific reports received by the NNHSP provide information for additional analysis of the status of the Early Hearing Detection and Intervention system in Nebraska. Unduplicated patient-specific information was reported to NNHSP for 954 newborns. Of those, 834 newborns referred on the birth admission hearing screening and 120 newborns were discharged to home prior to receiving a hearing screening. These were the newborns who were tracked through follow-up re-screening, diagnostic evaluations and early intervention.

Follow-up Services

Follow-up screening and/or diagnostic evaluations were initiated for 871 of the 954 newborns who either did not pass the birth admission hearing screening or were discharged without a hearing screening. Of those 871 newborns, follow-up was completed for 833, initiated but not completed for 27, and is still in progress for 11 of them. There were 83 newborns needing follow-up for whom initiation of follow-up services were not initiated or reported to NNHSP.

In 2006, based on individual reports submitted to NNHSP, there were 86 newborns who needed confirmatory testing beyond the initial outpatient re-screening to determine the status of their hearing. The results of the 86 who referred on the first outpatient hearing screening were:

- 42 passed on a second re-screening
- 14 were lost to follow-up
- 3 are currently in follow-up for transient conductive hearing loss due to middle ear dysfunction
- 24 referred and then received a diagnostic evaluation
- 3 expired prior to re-screening or diagnostic evaluation

Sixty six (66) newborns received a diagnostic evaluation as the initial step for outpatient follow-up, bringing to 90 the total number of children for whom diagnostic reports were received. The results of the 90 audiological diagnostic evaluations are:

- 46 had normal hearing established either initially or following medical management for middle ear dysfunction
- 8 are currently in follow-up for transient conductive hearing loss due to middle ear dysfunction
- 1 expired prior to the follow-up diagnostic evaluation
- 9 were lost to follow-up
- 26 were diagnosed with a Permanent Hearing Loss (PHL). The average age at identification was 55.4 days and 23 of those infants with PHL were identified prior to 3 months of age.

Diagram 1 tracks the progress of the 954 newborns needing follow-up services through the newborn hearing screening system to the point of hearing status established (normal hearing, permanent hearing loss), results pending, or lost to follow-up.

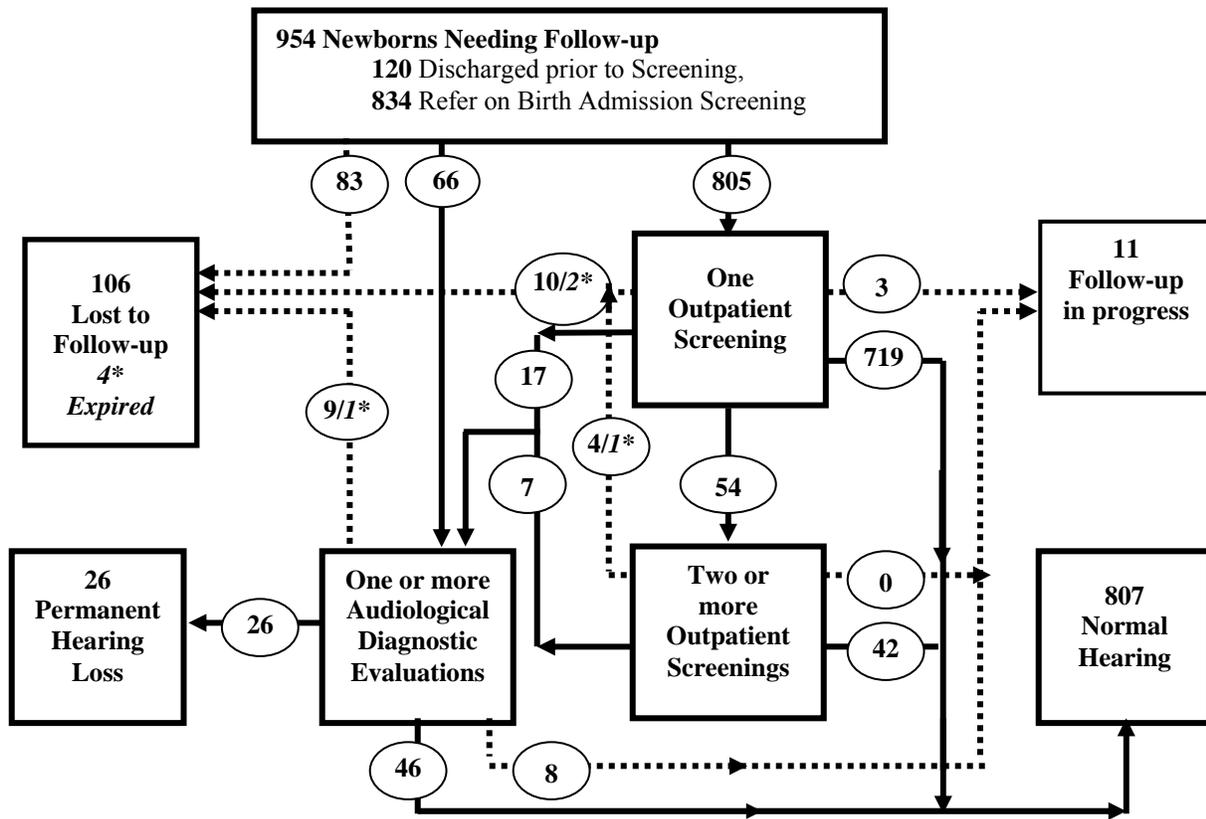


Diagram 1

Out-of-Hospital Births

Neb. Rev. Stat. §71-4740 requires the Department of Health and Human Services Regulation and Licensure to educate parents of newborns who are not born in a birthing facility about the importance of newborn hearing screening and to provide information to assist them in having the screening performed within one month after the child's birth. Although parent education was provided to the parents of all reported out-of-hospital births during 2006, only 44.1% (30 of 68) of out-of-hospital births were screened (see Table 8).

Out-of-Hospital Births (2001 – 2006)

	2001	2002	2003	2004	2005	2006
Out-of-hospital births	93	99	70	60	55	68
Number screened	5	16	12	13	15	30
Percentage screened	5.4%	16.2%	17.1%	21.7%	27.3%	44.1%

Table 8

Timeliness of Follow-up Re-screening/Testing

To meet the state and national guidelines of "1-3-6" (hearing screening completed by 1 month,

audiological diagnostic evaluation initiated by 3 months, early intervention initiated by 6 months), the timeliness of initiation of follow-up activities is an important aspect of the quality of services. For the newborns who did not pass the initial hearing screening during birth admission and who received follow-up services, 75.6% received an initial outpatient re-screening or diagnostic evaluation prior to 1 month of age. The peaks of follow-up activity occurred at approximately 1 week, 2 weeks, and 3 weeks of age (see Chart 1). The average age of follow-up service initiation was 28.0 days.

Of the 120 newborns who were discharged prior to screening and received follow-up services, the average time to the initial outpatient hearing screening was 23.2 days with 85.5% of those newborns receiving the initial screening prior to 1 month of age. Although an initial outpatient newborn hearing screening by 1 month of age does not meet the intent of Neb. Rev. Stat. §71-4739 for each newborn to be screened during birth admission, it does meet the national Early Hearing Detection and Intervention goal of every newborn having a hearing screening by 1 month of age.

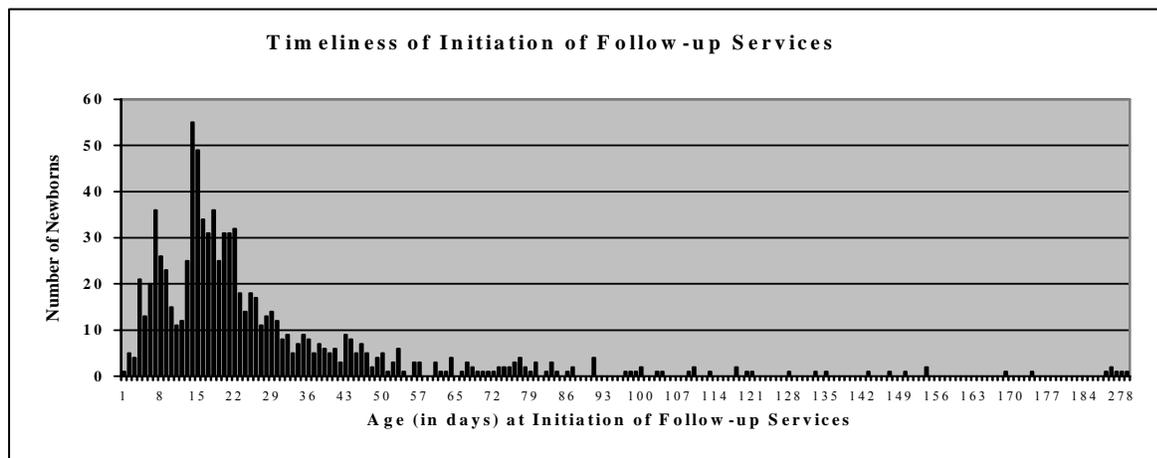


Chart 1

Neb. Rev. Stat. §71-4742 states: "...it is the goal of this state to achieve a one-hundred-percent screening rate." While Nebraska has made great strides in developing a comprehensive newborn hearing screening system, there are also infants for whom the status of their hearing is not known. In 2006, there were at least 442 newborns whose hearing status has not been established:

- 11 were identified with hearing problems associated with middle ear dysfunction but additional follow-up evaluations have not yet been completed.
- 23 newborns had a follow-up re-screening or diagnostic evaluation that was inconclusive with no additional follow-up received or reported.
- 83 infants had no outpatient follow-up initiated or reported
- 165 newborns with "refer" results were not reported to NNHSP (difference between aggregate birthing facility reports and individually-identifiable reports)
- 12 newborns with birth admission hearing screenings were not reported to NNHSP (difference between aggregate birthing facility reports and individually-identifiable reports)
- 33 of the out-of-hospital births were not screened or the results were not submitted to NNHSP

- 103 newborns expired prior to receiving a hearing screening
- 4 expired after follow-up was initiated but before it was completed
- 8 parents refused the hearing screening during birth admission

Based on the analysis of the aggregate hospital reports and actual file counts, the hearing status of 98.4% of the 26,898 newborns was confirmed as either normal or with a permanent hearing loss present.

Early Intervention

The purpose of the Infant Hearing Act (Neb. Rev. Stat. §71-4735) is to “obtain needed multidisciplinary evaluation, treatment, and intervention services at the earliest opportunity and to prevent or mitigate the developmental delays and academic failures associated with late detection of hearing loss.” The Early Development Network, Nebraska’s Part C Early Intervention Program, verified that 16 (62%) of the 26 infants with PHL were eligible for Early Intervention services. Services for 14 of the 16 infants were begun prior to 6 months of age and two (2) were verified after 6 months of age. Eight (8) infants were not referred to EDN. Fourteen (14) of the infants diagnosed with a PHL have a medical home.

Summary

- All the current birthing hospitals in Nebraska were conducting newborn hearing screening in 2006. All but one were conducting the hearing screenings during the birth admission.
- The benchmark of 95% of newborns having a hearing screening during birth admission by December 1, 2003 established by Neb. Rev. Stat. §71-4742 has been met. In 2006, birthing hospitals reported screening the hearing of 98.9% of newborns.
- The overall refer rate during 2006 for initial hearing screening during birth admission was 3.8%.
- In 2006, follow-up re-screening occurred within one month of birth for 74.3% of those newborns for which follow-up activities were initiated. The average age at the time of the initiation of follow-up re-screening or diagnostic evaluation was 28.0 days.
- The average age at diagnosis of hearing loss was 55.4 days for those reported to NNHSP in 2006 and 88.5% of the evaluations occurred within 3 months of birth.
- The incidence of Permanent Hearing Loss identified and reported to NNHSP (1 per thousand in 2006) appears to be within the anticipated range of 1 to 3 per thousand.

ACTIVITIES – 2006

Funding

Health Resources Services Administration/Maternal and Child Health Bureau

The Nebraska Newborn Hearing Screening Program received grant funds from the Health Resources Services Administration/Maternal and Child Health Bureau to fund the basic operations of the NNHSP. Funding amounts were \$38,750 for the 3 month period from 4/1/06 to 6/30/06 and \$98,750 for the 9 month period from 7/1/06 through 3/31/07. The goals of the NNHSP are:

System Goal 1 – The hearing of all newborns in Nebraska will be screened during the birth admission or, if born out-of-hospital, by one month of age.

System Goal 2 – All newborns who “refer” on the initial outpatient hearing re-screening will complete an audiologic diagnostic evaluation prior to 3 months of age.

System Goal 3 – All infants with confirmed hearing loss will begin receiving early intervention services prior to six months of age.

System Goal 4 – All infants with a confirmed hearing loss will have a medical home.

System Goal 5 – Families of young children with a confirmed hearing loss will have access to a family-to-family support system.

System Goal 6 – The hearing of young children in Nebraska will be screened at various times prior to age 3.

System Goal 7 – Hearing health professionals will increase their capacity to provide appropriate services to young children.

System Goal 8 – NNHSP will provide an effective structure for the newborn hearing screening and intervention system in Nebraska.

Centers for Disease Control and Prevention

The NNHSP also received \$145,850 from the Centers for Disease Control and Prevention for the second year of a three year Early Hearing Detection and Intervention Tracking, Surveillance and Integration cooperative agreement. The goals of the cooperative agreement are:

Goal 1 – Hearing screening results will be electronically reported to NNHSP for all occurrent births in Nebraska.

Goal 2 – Pediatric audiologic evaluations and risk factors will be electronically reported to NNHSP for young children identified with a hearing loss.

Goal 3 – The NNHSP data system, integrated with electronic birth certificate registry, will be electronically linked with related child data systems.

Goal 4 – A formative and summative evaluation of the NNHSP tracking, surveillance and integration project will be conducted and the results disseminated.

Advisory Committee

The NNHSP was developed based on the requirements identified in the Infant Hearing Act of 2000 and the protocols recommended by the Advisory Committee. Specific tasks to be accomplished by the Advisory Committee are 1) to continue to increase the representation of stakeholders, 2) to review and, as necessary, revise the existing protocols to incorporate the electronic data system, 3) to develop new reporting, tracking and follow-up protocols to effectively link the NNHSP and the early intervention systems, 4) to increase the program's responsiveness to the expanding cultural and linguistic communities in the state, 5) to support the development of an effective professional development system, and 6) to guide the long-term planning and evaluation of the EHDI system in the state. The Advisory Committee of the Nebraska Newborn Hearing Screening Program consists of 22 members representing medical, audiology, parents, family support, and education stakeholders (see Appendix B). The Advisory Committee met quarterly during 2006 and provided the following guidance to the NNHSP:

- Approved the formation of audiology, evaluation, and family support sub-committees.
- Recommended approaches to retrieve the dried blood spots from the newborn screening laboratory to assist with the identification of the etiology of confirmed hearing loss (see Appendix C).
- Approved pursuing the creation of a hearing aid loaner bank for children recently identified with a hearing loss in partnership with other organizations.
- Approved the "Investing in Family Support" work plan to organize parent-to-parent support systems and parent leadership opportunities.
- Refinement of the criteria for the "Lost to Follow-up" category in instances where re-screening or diagnostic results are not accessible or available.
- Provided input into the ongoing development of three aspects of the NNHSP: 1) parent education materials, 2) identification of the initial point of contact for parents of infants recently diagnosed with a hearing loss, and 3) annual parent survey.

Projects for 2006

Electronic Data System

Development of the Newborn Hearing Screening Module by QS Technologies/Netsmart for integration with the Electronic Vital Records System continued during the first ten months of 2006. To meet the target implementation date of January 1, 2007, selected staff of the largest

birthing facilities were trained in November and December, with the rest of the birthing facilities scheduled for training in January and February, 2007. The integrated system is based on the birth records and provides for the reporting of hearing screening results for all occurrent births in Nebraska. This will eliminate the need to manually record, transmit, and track demographic information on each newborn who “refers” or is discharged without a hearing screening. The reporting system will allow for better follow-up due to increased accuracy, consistency, and timeliness of newborn hearing screening information provided to the NNHSP by birthing hospitals. It will also provide better quality assurance and descriptive statistics than the current reporting of aggregate information by the birthing facilities.

Early Head Start ECHO Project

To begin the process of implementing periodic early childhood hearing screening in Nebraska, the ECHO project, developed by National Center for Hearing Assessment and Management and funded by the Head Start Bureau, trains Early Head Start programs to conduct OAE hearing screenings. A team, consisting of four audiologists, an educator of the deaf, an early childhood training Coordinator, and the AAP EHDI Chapter Champion, were trained to conduct the ECHO trainings. OAE screening equipment was provided as part of this project. Four additional Early Head Start programs were trained to conduct OAE screenings with the infants and toddlers enrolled in their programs in 2006. Discussions and technical assistance by phone and in-person occurred to overcome difficulties in following-up with re-screenings and in timely submission of paperwork.

Educational Materials

Parent education materials (brochures, videotapes) were provided at no charge to all birthing facilities. The new parent education brochure “Can Your Baby Hear?” and the new follow-up brochure “Your Baby Needs Another Hearing Screening” were written at an average health literacy level and are now available in ten languages (English, Spanish, Arabic, Russian, Vietnamese, Chinese, French, Nuer, Dinka, and Anuak).

Resource Guide

A Parent Resource Guide is provided to the Primary Care Provider to give to the family of each infant identified with a Permanent Hearing Loss (PHL). The resource guide was updated with new materials about educational and health services, unbiased information about communication options, and the [Funding Toolkit](#), a resource developed by Laura Barrett of the Central/Western Nebraska Partnership for Children Who are Deaf and Hard of Hearing.

Family-to-Family Support Plan

Representatives from the Regional Program for Students Who are Deaf and Hard of Hearing, PTI-NE and NNHSP attended the “Investing in Family Support” conference in Salt Lake City. The workgroup developed a work plan to initiate a collaborative approach to strengthen the formal and informal types of family support in the state. The proposed work plan was approved at the December, 2006, NNHSP Advisory Committee. The initial action was to create a Family Support Sub-committee of the Advisory Committee.

Together for Kids and Families

Nebraska's State Early Childhood Comprehensive Systems (SECCS) grant program Together for Kids and Families seeks to achieve optimum outcomes for Nebraska's young children and their families through comprehensive system planning and collaborative effort among stakeholders. The NNHSP program manager serves on workgroups to implement the following strategies:

- Develop and implement a collaborative initiative to promote the medical home approach as a standard of care for all children
- Establish a comprehensive program to promote regular recommended pediatric visits for children, following the American Academy of Pediatrics and Bright Futures guidelines
- Integrate parent to parent peer support systems into existing and new programs and services for families
- Develop capacity for an early childhood data monitoring system through creation of an ECCS data agenda

National Initiative for Children's Healthcare Quality (NICHQ) Learning Collaborative

Nebraska was one of eight states selected to participate in a Learning Collaborative, funded by the Health Resources Services Administration/Maternal and Child Health Bureau and developed by NICHQ. The purpose of the project is to reduce the number of babies who are lost to follow-up by developing strategies that are shown to be effective through small tests of change.

Partners in the NICHQ Learning Collaborative included the Nebraska Medical Center, Fred LeRoy Health and Wellness Clinic, Boys Town Pediatric Primary Care, Boys Town National Research Hospital, Early Development Network (Part C), Medically Handicapped Children's Program, Metro Regional Program for Children Who are Deaf or Hard of Hearing, Medicaid/Primary Care Unit, Together for Kids and Families and the NNHSP.

Boys Town National Research Hospital – special project

Boys Town National Research Hospital, through a contract with NNHSP supported by HRSA supplemental funds, developed Early Hearing Detection and Intervention professional development materials for nurses, a web-portal for physicians on the www.babyhearing.org website, and a Spanish Public Service Announcement.

Hearing Aid Loaner Bank

During the summer of 2006, a joint partnership was developed between the University of Nebraska – Lincoln and the NNHSP to study the feasibility of establishing a hearing aid loaner bank in Nebraska for young children identified with a PHL. Two audiology graduate students at UNL explored the funding, administration, and selection criteria for states with hearing aid loaner banks, the capacity of audiologists licensed in Nebraska to fit hearing aids for newborns and infants, hearing aid manufacturers' programs to provide discounts and support for hearing aid loaner banks, funding sources through private and non-profit organizations, and cost estimates to develop and maintain a hearing aid loaner bank.

Projects Planned for 2007

Electronic Data System

Full implementation of the integrated reporting system will occur beginning January 1, 2007. Reports to monitor the reporting will continue to be created, technical assistance will be provided to data input clerks, and specifications for revision and enhancement, including reporting of audiologic diagnostic reports and linkages with other child health data systems, will be developed.

Periodic Early Childhood Hearing Screening

Training and support to implement an OAE hearing screening program during well-child visits will be offered to several child health clinics. The training materials developed by the National Center for Hearing Assessment and Management will be used.

Initial Point of Contact

A multidisciplinary workgroup will continue to determine how best to establish an initial point of contact for parents of newborns/infants identified with a PHL. The anticipated outcome of this work is that the parents will be able to access timely and appropriate early intervention services through a recognized point of entry that is knowledgeable about hearing loss, its effects on young children, and available resources.

Family-to-Family Support

The “Investing in Family Support” work plan will be implemented. The four goals of the plan are: Goal 1 – Develop an advisory and oversight mechanism for parent-to-parent support. Goal 2 – Determine existing and desired parent-to-parent supports. Goal 3 – Develop leadership capacities of families. Goal 4 – Enhance the capacity of professionals to link families with parent-to-parent support.

Hearing Aid Loaner Bank

Partnerships will be fostered to establish a hearing aid loaner bank for young children recently identified with a Permanent Hearing Loss.

Dried Blood Spot

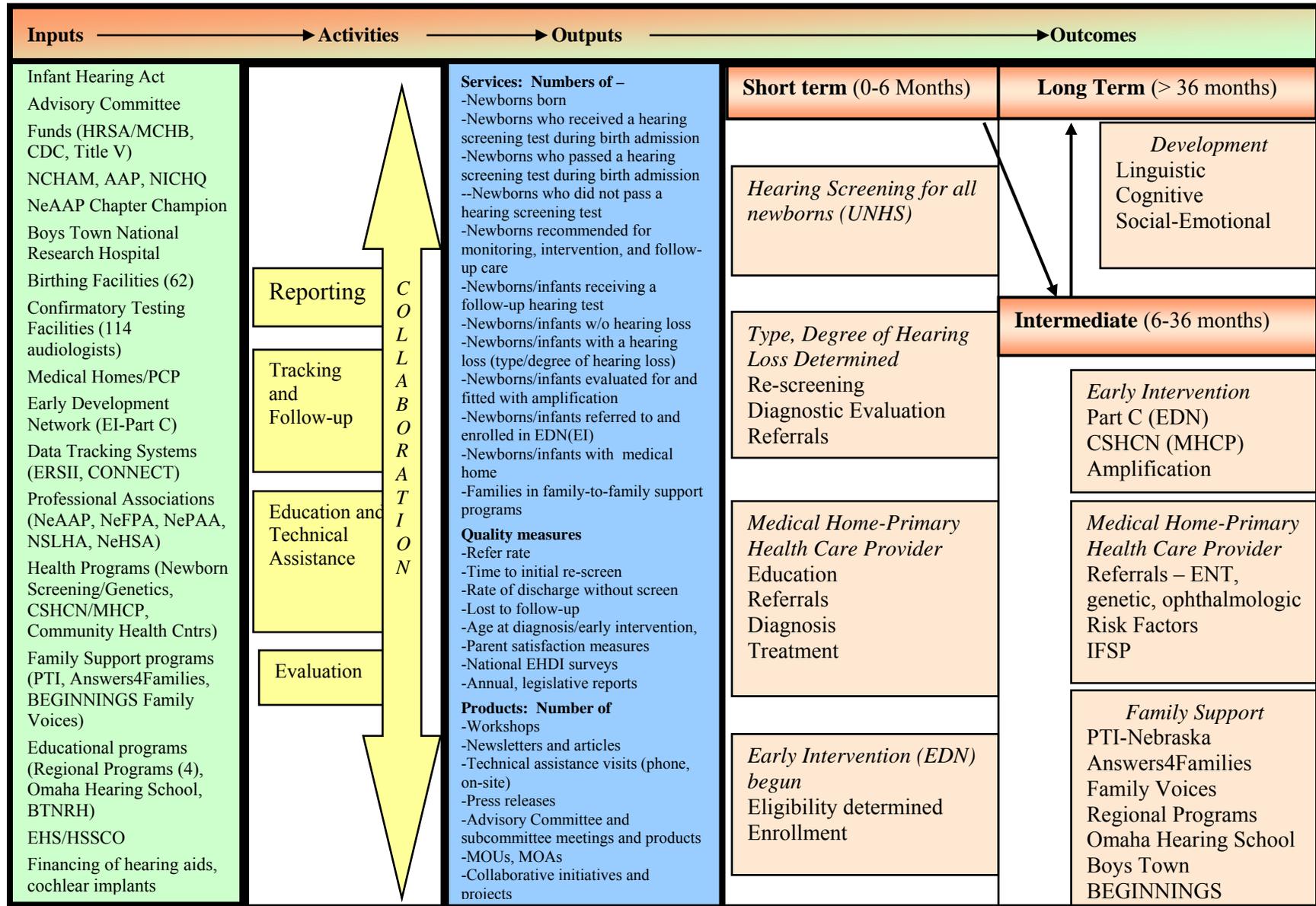
The feasibility of retrieving the newborn dried blood spot for identification of congenital cytomegalovirus (CMV), Connexin 26 and 30, mitochondrial, and Pendred syndrome will continue to be evaluated and prioritized. Identification of these factors, which are risk factors for later-onset hearing loss, can also assist in establishing the etiology of a congenital hearing loss.

Website

A website for the NNHSP will be developed as part of the Newborn Screening Program website.

Appendix A.

Nebraska Newborn Hearing Screening Program – Logic Model (2006)



Appendix B.

Advisory Committee Members

Committee Member	Group/Facility Represented
Steve Boney, PhD	Professor, Audiology University of Nebraska - Lincoln
Margaret A. Coleman	Nebraska Commission for the Deaf and Hard of Hearing
Lora Langley	Ponca Tribe of Nebraska
Regina Watson	Hearing Screening Coordinator Tri County Area Hospital, Lexington
Mary Pat Moeller, PhD	Director, Center for Deafness Boys Town National Research Hospital
Stacie Ray	Parent, Clinical Practice Supervisor – Audiology University of Nebraska - Lincoln
G. Bradley Schaefer, M.D.	Geneticist, Munroe-Meyer Institute, Nebraska Medical Center
Monica Seeland	Vice President, Nebraska Hospital Association
Britt Thedinger, M.D.	Otologist, Ear Specialists of Omaha
Donald M. Uzendoski, M.D.	Nebraska Chapter, American Academy of Pediatrics Early Hearing Detection and Intervention Chapter Champion
Robert Wergin, M.D.	Nebraska Academy of Family Physicians
Dawn Peters	Parent Training and Information-Nebraska
Eleanor Kirkland	Head Start State Collaboration Office (NDE)
Audrey Isaacson	Parent
Jennifer Hales	Parent
Kenny Johnson	Parent
Rhonda Fleischer	Liaison, Regional Programs for Deaf and Hard of Hearing Students (NDE)
Jeanne Garvin, M.D.	Medical Director Medically Handicapped Children’s Program (CSHCN) (HHS)
Charlie Lewis	Answers4Families
Micaela Swigle	Co-Lead, Early Development Network (Part C) (HHS)
Julie Miller	State Genetics Coordinator, Newborn Screening Program (HHS)
Krystal Baumert	Follow-up Coordinator, Newborn Screening Program (HHS)
Jeff Hoffman	Manager, Newborn Hearing Screening Program (HHS)

Appendix C: Summary of Analysis of Ways to Address Genetic Causes of Hearing Loss

Summary of discussions on risk/benefit of storing and retrieving or testing dried blood spots for genetic causes of hearing loss:

Options	(+’s)	(-’s)
<p>1) Require screening of all newborns for CMV, Connexin 26, Pendred, Mitochondrial. (26,500 births/year) \$21.00/baby for testing (estimated)</p>	<p>- Alert physicians of need for more frequent hearing screening due to congenital CMV for those who pass the initial hearing screen. This allows for the earliest intervention for acquired hearing loss due to CMV. (e.g. re-screen q 6 months v. at 3 years of age). Since may develop neuromuscular, vision & vestibular pathologies as well, elevates need for referral to early intervention.</p> <p>-“May” reduce risk of developing hearing loss if CMV and treated with ganciclovir®...but see (-)</p> <p>-If DFNB1 mutation found, can inform parents of 20-30% chance for progressive hearing loss.</p> <p>-“May” prevent hearing loss due to mitochondrial <i>A1555G</i> by avoiding use of aminoglycoside antibiotics ...but see (-)</p> <p>-Serves as reflex hearing screen, and helps define etiology of hearing loss for those who fail the OAE/ABR and are diagnosed. MAY reduce/eliminate the diagnostic odyssey if clear DNA results are obtained.</p> <p>Strongest argument for: best opportunity to catch acquired hearing loss at its earliest point.</p>	<p>-Unnecessarily alert/alarm physicians and parents of children who have congenital CMV who won’t develop hearing loss, excessive cost for repeated screening cost for these children. May harm parent/child bond due to parent’s disability expectations.</p> <p>-Significant increase in cost to the current \$35.75 screen.</p> <p>-Ganciclovir® not recommended as a prophylactic to prevent acquired hearing loss due to CMV, but when used to treat serious viral infections, has been found to have some preventive effect against hearing loss.</p> <p>-May have potential for progressive hearing loss if mitochondrial (<i>A1555G</i>) regardless of gentomycin® (aminoglycoside antibiotic) exposure.</p> <p>-Mutation analysis may give unclear picture...could be possible carrier vs. a compound heterozygote – but one mutation might not be on panel screened, and may or may not know phenotype association.</p> <p>---concern with any DNA screening as a “primary” screen given the uncertainty mentioned above</p> <p>-Must have data system capacity to transmit results from DNA lab on to NBS laboratory report, and report that can be run daily identifying abnormal results. (Report easy, in-lab transmittal more of an issue).</p> <p>-New contract (can’t amend).</p>
<p>2) Universally offer (optional) to all parents of newborns the same panel (anticipate 95% or 25,175 tests / year) \$21.00/baby for testing (estimated)</p>	<p>Same (+’s) as above, but should expect significantly lower uptake than 95% given the \$21.00 additional cost. May be similar to uptake when supplemental cost was \$25 (< 50% of parents opted for the testing, and there were easier to understand/clearer benefits to that testing). Therefore benefits will be incrementally lower based on the % of newborns getting the “optional” screen.</p>	<p>-Same (-’s) as above</p> <p>-Likely to miss children with any of the conditions</p> <p>-Reduces cost/benefit if children are missed</p> <p>-New contract required. Can’t amend.</p>

<p>3) Store specimens for 5 years to be retrieved upon request from physician (ENT usually) maybe 100-150 x's / year) (Storage and retrieval costs...could pass these on as billable for the testing) For five years of storage/retrieval -- Year 1 \$0.10 per specimen -- Year 2 \$0.20/specimen -- Year 3 \$0.30/specimen -- Year 4 \$0.40/specimen and Year 5 and greater \$0.50/specimen. (estimated)</p>	<p>-May help with diagnosis/management/diagnostic odyssey if the acquired loss is found to be due to CMV. Doesn't eliminate need to test for Connexin 26/ Pendred and Mitochondrial <i>A1555G</i>, but is the only specimen that will help with identifying if hearing loss could be due to congenitally acquired CMV.</p>	<p>-Actual annual costs would be about \$2,650 the first year; \$5,300 the 2nd year; \$7,950 the 3rd year, \$10,600 in year 4, and \$13,750 the 5th and every year after the 5th year. How would this be paid? (Increase per infant screened cost by .10 per year for 5 years. -Does nothing to help with early identification of acquired hearing loss for those with CMV -New contract required, can't amend (can not change scope of services under contract, so would have to publish a new RFP and go through the competitive bidding process).</p>
<p>4) Screening of all newborns that refer on the OAE/ABR screen, for the same panel (900-1000/year) (we usually have these results weekly, and could send a list once a week for retrieval/testing) \$55.00/baby for retrieval & testing (estimated)</p>	<p>-May serve as a compromise option. -Since may develop neuromuscular, vision & vestibular pathologies as well, elevates need for referral to early intervention. -If DFNB1 mutation found, can inform parents of 20-30% chance for progressive hearing loss. -Serves as reflex hearing screen, and helps define etiology of hearing loss for those who fail the OAE/ABR and are diagnosed. MAY reduce/eliminate the diagnostic odyssey if clear DNA results are obtained. -If DFNB1 mutation found, can inform parents of 20-30% chance for progressive hearing loss. -"May" prevent hearing loss due to mitochondrial <i>A1555G</i> by avoiding use of aminoglycoside antibiotics ...</p>	<p>-Does nothing to help with earlier identification of newborns who pass the screen but will acquire hearing loss due to CMV. -Challenging to set up a system administratively to accomplish this routinely for every baby. How can we ensure this happens? -May have potential for progressive hearing loss if mitochondrial (<i>A1555G</i>) regardless of gentomycin (aminoglycoside antibiotic) exposure. -Mutation analysis may give unclear picture...could be possible carrier vs. a compound heterozygote – but one mutation might not be on panel screened, and may or may not know phenotype association. -New contract required, can't amend.</p>
<p>5) Universally offer (optional) the blood-spot test for CMV only to all parents of newborns (anticipate 95% or 25,175 / year) (Testing costs) \$3.50/baby for testing (estimated). The CMV assay is a qualitative assay designed for the detection of the presence of circulating CMV viral DNA in the blood as an indicator for CMV infection. The assay was developed to target conserved regions of two viral genes. Reference will be provided. (Additional cost could affect uptake of this optional testing).</p>	<p>- Alert physicians of need for more frequent hearing screening due to congenital CMV for those who pass the initial hearing screen. This allows for the earliest intervention for acquired hearing loss due to CMV. (e.g. re-screen q 6 months v. at 3 years of age). Since may develop neuromuscular, vision & vestibular pathologies as well, elevates need for referral to early intervention. -"May" reduce risk of developing hearing loss if CMV and treated with ganciclovir...but see (-) -If DFNB1 mutation found, can inform parents of 20-30% chance for progressive hearing loss. -May serve as a compromise option – or perhaps a way to incrementally implement a plan to eventually</p>	<p>-Likely to miss children with CMV due to refusal of the optional screen. -should expect somewhat lower uptake than 95% given the \$3.50 additional cost (but problem less of an impact than the \$21 cost would have). Therefore benefits will be incrementally lower based on the the % of newborns getting the "optional" screen -Probably does not eliminate the need for further DNA testing for Connexin 26, Pendred and Mitochondria <i>A1555G</i>. -Reduces cost/benefit if children are missed -New contract required. Can't amend existing contract.</p>

	accomplish the ideal option	
6) Pull all the specimens (for potential later testing) of all newborns confirmed with hearing loss (25-75/year...would require longer storage of specimens - perhaps 6 months to a year, but reduce the # of specimens needing retrieval) NO CHARGE. Cap of 100 specimens/year -- return shipping not included.	-Least expensive -Benefit mostly to etiology diagnosis which then informs management protocols, at minimum cost - No need to change contract	-Does nothing to help children who pass the hearing screen but will acquire hearing loss due to CMV
7) Pull all specimens for potential later testing of all newborns who refer on screening (900-1000) (We have all results of babies who refer on screening within a week or two of birth). These could be stored there, or returned to the state for longer term storage. \$5.00/specimen (estimated). Includes retrieval and return shipping.		

The above options were considered in detail by a work group of the Newborn Screening (blood-spot) Advisory Committee, and ultimately the recommendation was to implement procedures to encourage physicians to get the audiological evaluation and diagnosis completed by less than 90 days, and recommend for those diagnosed with sensorineural hearing loss that they request the dried blood spot be returned so it could be tested for genetic causes of hearing loss, or at a minimum, CMV.

The Committee also recommended that concurrently the programs should continue to work towards a system in which we could store specimens for 5 years to be retrieved upon request while universally offering blood-spot screening for CMV only to all newborns.

Subsequently, the Committees and the State Program should consider universally offering the CMV/Pendred/Connexin 26/ Mitochondrial testing to all newborns, and do screening all newborns that refer on the initial hearing screen.

These recommendations have been discussed by both the Newborn Screening Advisory Committee and the Newborn Hearing Screening Advisory Committee.

The staff of the **Nebraska Newborn Screening (Blood-spot) Program** are available to help with your questions at the numbers listed below. General areas of responsibilities are listed:

Julie Miller, Newborn Screening/Genetics Program Manager (402) 471-6733

Program planning, evaluation and management, professional and patient education, metabolic formula

Krystal Baumert, NBS Follow-up Coordinator (402) 471-0374

Metabolic and Endocrine conditions, Transfusions, Home Births

Karen Eveans, NBS Follow-up Specialist (402) 471-6558

Hemoglobinopathies and Cystic Fibrosis, Drawn Early and Unsatisfactory Specimens

Mike Rooney, Administrative Assistant (NBS & EHDI) (402) 471-9731

Metabolic foods program, Patient Education materials translations and distribution

WEB PAGE: www.dhhs.ne.gov/nsp

E-mail contact: newborn.screening@hhs.ne.gov

E-FAX: (402) 742-2332

Regular Fax: (402) 471-1863

Nebraska Newborn Screening Program
Department of Health and Human Services
P.O. Box 95026
Lincoln, NE 68509-5026

Pediatrics Screening Laboratory Director, Joseph Quashnock, PhD (412) 220-2300 (Pennsylvania)

The staff of the **Nebraska Early Hearing Detection & Intervention Program** are available to help with your questions at the numbers listed below. General areas of responsibilities are listed:

Jeffrey Hoffman, MS, CCC-A, Early Hearing Detection & Intervention (EHDI) Program Manager

(402) 471-6770 Program planning, evaluation and management, systems development

Claire Covert, EHDI program Staff Assistant (402) 471-3579

Follow-up, patient education materials distribution, data management, special projects

Mike Rooney, Administrative Assistant (NNSP & EHDI) (402) 471-9731

Follow-up, patient education materials translations

Jim Beavers, Business Analyst, EHDI Program (402) 471-1526

Data system planning and testing, development of reports, system security, training and technical assistance.

Nebraska Early Hearing Detection & Intervention Program
Lifespan Health Services, Division of Public Health, DHHS
P.O. Box 95026
Lincoln, NE 68509-5026

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