

October, 2008

To persons interested in Newborn Screening:

You are receiving this Executive Summary of the Newborn Screening 2007 Annual Report as a stakeholder in the health of Nebraska's youngest citizens - our newborns. For the full report, access the Newborn Screening Web page at the Nebraska Department of Health and Human Services Web site at: <http://www.dhhs.ne.gov/nsp> under "reports and publications."

This report covers the Newborn Screening System in Nebraska for the calendar year 2007.

The achievements that have occurred in Nebraska for both newborn blood-spot and hearing screening are due to the tremendous efforts of a system of people across the state. The state programs provide a framework, and to a degree, a back-up system via tracking and follow-up. We believe the most important work, however, occurs where people live, where they get their care, and where their babies are born.

This executive summary highlights a few of the many achievements and the continuing challenges of newborn blood-spot and newborn hearing screening systems in Nebraska.

Sincerely,



Julie Miller, Program Manager
Newborn Screening and Genetics
Nebraska Department of Health and Human Services



Jeff Hoffman, MS, CCC-A, Program Manager
Early Hearing Detection and Intervention Program
Nebraska Department of Health and Human Services

Highlights from the 2007 Nebraska Newborn (Blood-spot) Screening Program, Annual Report:

- **In 2007, Nebraska required screening for 8 conditions:** Biotinidase deficiency, Congenital Primary Hypothyroidism, Congenital Adrenal Hyperplasia, Cystic Fibrosis, Galactosemia, Hemoglobinopathies, MCAD and PKU.
- All newborns were offered supplemental screening at no extra cost and which required no extra blood. Supplemental screening involved tandem mass spectrometry screening for additional fatty acid, organic acid and amino acid disorders.
- Supplemental screening was universally offered to all parents, but was not required. Parents had to decide whether to consent to, or dissent from, supplemental screening.
- **More than 97% of Nebraska newborn's parents consented to the supplemental screening.**
- The following babies were identified and entered into early treatment and intervention, most with specialty clinics specific to their type of condition:
 - 4 babies with partial biotinidase deficiency (treated)
 - 4 babies with hemoglobinopathies
 - * (1 with Sickle Hgb. C disease, 1 with Hgb.-C disease/C-Beta Thalassemia, 1 with Sickle Beta Thalassemia, and 1 Beta Thalassemia major)
 - 2 babies with transient tyrosinemia (only 1 did not require treatment)
 - 1 baby with congenital adrenal hyperplasia
 - 16 babies with congenital primary hypothyroidism
 - 12 babies with cystic fibrosis
 - 1 with glutaric acidemia type I
 - 1 baby with methylmalonic acidemia
 - 1 Duarte variant Galactosemia (tx'd)
- In 2007 the Nebraska Newborn Screening Advisory Committee recommended mandating the conditions screened on the supplemental panel. The public process to revise regulations was completed in 2007 and this was implemented on 7/1/08.

In 2007, newborn screening efforts resulted in successfully identifying and treating 42 newborns affected with conditions in time to prevent mental retardation, other chronic illness and disability, and infant death.

Individually each condition is quite rare. However in 2007, 1 in 643 babies were identified through newborn screening to have one of the clinically significant screened conditions.

The newborn screening fee of \$35.75 billed to hospitals for the mandatory or mandatory + supplemental screen was the 15th lowest in the Country.

- The contractor during 2007 was Pediatrix Screening Laboratory in Pennsylvania. In addition to providing quality testing services, courier service to ensure overnight specimen delivery was an important element of that contract. Hospitals received the lab reports via 24/7 electronic access or via hard copy receipt in the mail. All abnormal or out of range reports were reported by phone and fax. Pediatrix supported the program's follow-up and tracking as well as quality assurance monitoring by providing access to data reports designed by the state program from their electronic data system.
- Ensuring a quality specimen is collected and handled correctly the first time is important to ensuring rapid identification of newborns affected with screened conditions. Unsatisfactory specimens, specimens collected too early (at less than 24 hours of age), and specimens collected post transfusion can create delays in the timely identification of newborns. The Newborn Screening Advisory Committee monitored these and several other measures each quarter:
 - 265 or 0.98% of all newborns had a specimen collected before 24 hours. Babies to be transferred accounted for 68% of these and babies to be transfused accounted for 4.5%. The reason for early collection is unknown for the remaining 27.5%.
 - Only 114 or 0.42% of all newborns initial specimens were "unsatisfactory," requiring repeat collection.
 - The mean-average turn around time from birth to results ranged between 5.2 - 5.45 days. The time from birth to collection, and the in-lab turn around times continued trending downward for even more rapid turn around rates.

Newborn screening to prevent mental retardation and other physical disabilities and death is one of the most cost-effective public health programs.

Total expenditures for metabolic foods and formula for 67 patients totaled \$359,283. If these same amounts were used to treat rather than prevent mental retardation and other disabilities, only 2.3 people per year would be served in an intermediate care facility for persons with mental retardation.

Fast Facts... Nebraska has an aggressive quality assurance monitoring system. In addition to data evaluation each quarter by the Newborn Screening Advisory Committee, each hospital receives a quarterly report of several performance measures and how their averages relate to the state-wide averages.

Measures such as age at time of specimen collection, turn-around times, unsatisfactory specimen rates, drawn early specimen rates, age at diagnosis, intervention and treatment are all critical to monitoring and measuring the success of the program. Monitoring presumptive positive rates and keeping them low keeps costs lower for all by reducing the number of babies requiring confirmatory testing.

- **Nebraska's rate for reporting "presumptive positive" (PP) screening tests was lower than the national averages for congenital adrenal hyperplasia (CAH), congenital hypothyroidism, and cystic fibrosis.** With CAH, only 5 states out of 35 States reporting, had a lower presumptive positive rate.
- **While screening algorithms successfully kept the PP rates low, Nebraska's detection rates for some diseases were consistent with national averages (Congenital Adrenal Hyperplasia and Congenital Hypothyroidism), and detection rates for others were actually HIGHER than national averages.** For example Nebraska has a higher incidence rate of biotinidase deficiency, particularly partial biotinidase deficiency (partials in 2007: US about 1 : 28,894, NE 1 : 6,753). We also had a higher rate of cystic fibrosis (classical and non-classical) found through newborn screening (US 1 : 5,841; NE 1 : 2,077).
- In screening for Galactosemia, only 7 state programs had initial PP rates lower than Nebraska. Three other states had the same PP rate of .03% as Nebraska. One might be concerned it was too low and risked missing affected babies. However, although no babies with classical GALT were detected, one baby with the milder Duarte Variant Galactosemia was detected. This is good evidence that the screening algorithm and reference ranges are appropriate for detection of severe cases of classical Galactosemia.
- The age at which intervention or treatment begins for affected infants can be critical to preventing mental retardation, other disability and even death. Two of the conditions with the shortest windows of opportunity to intervene because of the possibility of early onset of devastating symptoms are classical Galactosemia and MCAD deficiency. In 2007 no babies with classical Galactosemia or MCAD deficiency were identified in Nebraska however, the one newborn with Duarte Variant Galactosemia was treated by 6 days of age. In 2007 in the US, 70 babies were born with classical Galactosemia. Forty nine percent of these babies were treated by 7 days of age or less .

Highlights from the 2007 Nebraska Early Hearing Detection and Intervention Program Annual Report



The Infant Hearing Act of 2000 established newborn hearing screening in Nebraska. Birthing facilities are to educate parents about hearing screening, to include newborn hearing screening as part of the standard of care, and to conduct hearing screenings for at least 95% of the newborns born in Nebraska.

- All of the current birthing facilities conducted newborn hearing screening in 2007.

The hearing of 98.6% of the 27,117 newborns was screened during the birth admission.

- The “refer” (did not pass) rate for hearing screening during birth admission was 3.7%.
- The rate of completed follow-up screening and/or diagnostic evaluation has continued to improve, increasing from 63% in 2001 to 88% in 2007.
- Follow-up services occurred within one month of birth for 75% of the newborns.

- The average age at initiation of follow-up was 29 days.
- The average age at diagnosis of hearing loss was 122 days for those reported to NE-EHDI in 2007.

In 2007, 52 infants with permanent hearing loss were identified, an incidence of 2 : 1000 births.

- Early Intervention (Part C, IDEA) services were initiated for 77% of the infants diagnosed with a permanent hearing loss. Over 87% were verified prior to 6 months of age.
- There were 131 babies who either did not receive the recommended follow-up hearing services or the results were not reported to the NE-EHDI Program.

Funding - The Nebraska Early Hearing Detection and Intervention Program received grant funds from the Maternal and Child Health Bureau and the Title V/Maternal and Child Health Block Grant to fund the basic program operations. The NE-EHDI Program also received funding from the Centers for Disease Control and Prevention to continue development of the ERS-II integrated data system through the Early Hearing Detection and Intervention Tracking, Surveillance and Integration cooperative agreement.

Advisory Committee - The Advisory Committee of the NE-EHDI Program consists of 23 members representing medical, audiology, parents, family support, and education stakeholders. The Advisory Committee met quarterly.

Electronic Data System - The hearing screening module of the Vital Records Electronic Reporting System (ERS-II), developed by Netsmart Technologies, Inc., was implemented on January 1, 2007. The integrated reporting system is based on the birth records and provides for the reporting of hearing screening results for all occurrent births in Nebraska. All birthing facilities were granted access to the hearing screening module and selected staff was trained to create records and report hearing screening result. Ongoing training and technical assistance was provided to birthing facility staff.

National Initiative for Children's Healthcare Quality 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. Program activities during the Learning Collaborative, which concluded in July, 2007, included reduction of the literacy level of parent materials; development of a parent checklist/roadmap; inclusion of parent phone number, primary language, and primary health care provider name on hospital reports; and development of a 1-page audiologic evaluation reporting form.

Nebraska Children's Hearing Aid Loaner Bank - Based on a feasibility study conducted during 2006, the Nebraska Children's Hearing Aid Loaner Bank (NCHALB) was organized during 2007. The purpose of the NCHALB is to provide immediate access to amplification for children when identified with a permanent hearing loss for an initial period of six months. Partners in the NCHALB are the University of Nebraska – Lincoln Barkley Center audiology department, the Nebraska Association for the Education of Young Children (NeAEYC) and the NE-EHDI Program. Contracts were signed to provide funding to NeAEYC for fiscal management and to UNL-Barkley Center for administration of the NCHALB. An organizing committee was formed and funding proposals, a brochure and the Web site (www.unl.edu/barkley/nchalb/index.shtml) were developed. One hearing aid manufacturer, GN ReSound, donated 30 hearing aids to the NCHALB.

Dried Blood Spot Retrieval - The feasibility of retrieving the newborn dried blood spot for identification of congenital cytomegalovirus (CMV), Connexin 26 and 30, mitochondrial, and Pendred syndrome continued 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. A work group of the Newborn Screening (blood-spot) Advisory Committee recommended the implementation of procedures to encourage physicians to get the audiological evaluation and diagnosis completed before 90 days so that they could request that the dried blood spot be returned so it could be tested for genetic causes of hearing loss, or at a minimum, CMV for those infants diagnosed with sensorineural hearing loss. Educational materials were developed and mailed to physicians, physician assistants and nurse practitioners. Beginning in October 2007, materials for parent consent and physician retrieval of the dried blood spots for babies identified with sensorineural hearing loss before three months of age were provided to the primary health care provider.

Nebraska Department of Health and Human Services
Division of Public Health, Lifespan Health Services Unit
Newborn Screening & Genetics Program
301 Centennial Mall South, Box 95026
Lincoln, NE 68509-5026

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EXECUTIVE SUMMARY OF NEBRASKA'S 2006 ANNUAL REPORT ON NEWBORN SCREENING
INCLUDING
SCREENING FOR METABOLIC AND INHERITED DISORDERS
AND
NEWBORN HEARING SCREENING

For full report see:
Web-site at <http://www.dhhs.ne.gov/nsp/>
or contact the Newborn Screening Program at 402 471-9731.

