New Outcome Data and Shorter/Abbreviated Report

NEWBORN SCREENING IN NEBRASKA

Newborn Bloodspot Screening for Metabolic & Inherited Disorders

and

Early Hearing Detection & Intervention

2012 Annual Report

Department of Health & Human Services

DHHS

NEBRASKA

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I know a father who lost his first child to an undetected disorder that a few years later likely would have been detected through newborn screening. He once told me that most things in life that other people find stressful, don't phase him, because he knows he's "already experienced the worst day of my life."

The worst day of my life was the day I learned my child was born with life-threatening critical congenital heart defects (CCHD), and yet the worst day of my life was a painful but beautiful blessing. Many times in my son's journey we nearly lost him, but each time he was saved by the best possible medical care. Had I not been blessed with that terrible day of reckoning, I would have found out the hardest way.

I advocate for newborn screening because I want more people to have worst days like mine that contain an undercurrent of hope and the possibility of action.

I want no parent to have worst days like my friend, who will never see his child grow up.

We can change the world so that the worst thing that happens to a parent is learning that their child has a chance instead of losing that child without a fight. People often feel sorry for me that my son has had so many heart surgeries, but I know we're the lucky ones. Even if I lose my son, at least I know we had a chance to fight, and all the years that opportunity afforded us. Everyone should be so blessed.

Contributed by and with permission of Amanda Rose Adams

Author of "My Son Is Not His Disease and Neither Am I"

http://www.babysfirsttest.org/newborn-screening/blog/my-son-is-not-his-disease-and-neither-am-i

NEWBORN SCREENING FOR INBORN ERRORS OF METABOLISM AND INHERITED DISORDERS

The goal of newborn blood spot screening 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 physician and parents
- Timely recall 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 2012, newborn screening efforts resulted in successfully identifying and treating 53 newborns affected with conditions in time to prevent problems associated with them:

- 5 babies with partial (treated) biotinidase deficiency (BIO)
- 2 babies with congenital adrenal hyperplasia (CAH)
- 13 babies with congenital primary hypothyroidism (CPH), 2 with primary hypothyroidism not congenital, and 2 with hypothyroidism
- 6 babies with cystic fibrosis and 3 with CF related metabolic syndrome (CRMS)
- 7 babies with hemoglobinopathies (3 sickle cell disease, 1 SC-disease, 1 D/Beta-0 Thalassemia disease, 1 hemoglobin C Disease, and 1 hemoglobin E disease)
- 4 babies with MCAD - medium chain acyl-coA dehydrogenase deficiency
- 4 babies with phenylketonuria (PKU) and 1 with hyperphenylalaninemia
- 2 babies with SCAD – short chain acyl-coA dehydrogenase deficiencey
- 2 babies with transient tyrosinemia who responded to treatment
  (Plus 1 mild hypermethioininemia that did not require treatment)

The incidence rate of conditions in Nebraska based on the screened conditions identified over the last 5 years from 2008-2012 and number of births screened those five years:

1 in 529 births

ABOUT 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 slightly from state to state but the overall goal is the same: prevent or minimize the serious effects of the conditions screened. In 2012, Nebraska's required screening panel included 28 metabolic, endocrine, hematologic and other conditions.

- Arginino Succinic Acidemia
- Beta-ketothiolase Deficiency
- Biotinidase Deficiency
- Carnitine Uptake Defect
- Citrullinemia
- Congenital Adrenal Hyperplasia
- Congenital Primary Hypothyroidism
- Cystic Fibrosis
- Galactosemia
- Glutaric Acidemia Type I
- Hemoglobinopathies
  (Sickle Cell, Hgb. C & Thalassemias)
- Homocystinuria
- Isovaleric Acidemia
- Maple Syrup Urine Disease
- Long Chain Hydroxy Acyl-CoA Dehydrogenase Def.
- Medium Chain Acyl-CoA Dehydrogenase Deficiency
- Methylmalonic Acidemia (Mutase)
- Methylmalonic Acidemia (Cbl A & B)
- Multiple Carboxylase Deficiency
- Phenylketonuria
- Propionic Acidemia
- Tyrosinemia
- Trifunctional Protein Deficiency
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency
- 3-Hydroxy 3-Methyl Glutaric Aciduria
- 3-Methylcrotonyl-CoA Carboxylase Deficiency

The effects of screened conditions if not detected and treated can range from brain and nerve cell damage resulting in severe intellectual disability, to damage to the infant or child's heart, kidney, liver, spleen, eyes, problems with physical growth, stroke and even death.

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 Cystic Fibrosis (CF) Center is always recommended when an infant is identified with a positive screen to be at higher risk of having one of these conditions.

So, just how common are these conditions in Nebraska's population?

Year Screening Began*	Condition	Incidence rate
1967	PKU	1: 11,538
1979	Congenital Primary Hypothyroidism	1: 2,317
1987	Biotinidase Deficiency (Profound & Partial)	1: 10,069
1997	Galactosemia (Classical)	1:122,766
	(Classical plus Duarte tx'd)	1: 23,019
	Hemoglobinopathies (Sickle Cell Disease)	1: 16,142
	Sickle Hemoglobin C Disease	1: 31,131
	Hemoglobin C Disease	1: 87,166
	Hemoglobin E Disease	1:145,277
	Sickle Beta Thalassemia	1:217,916
	Beta Thalassemia Major	1:217,916
	Hereditary Persistence Fetal Hgb.	1:435,832
2002	MCAD	1: 14,529
	(2003 MS/MS universally offered, ≅97% opt in)	
2006	Cystic Fibrosis	1: 2,868
	Congenital Adrenal Hyperplasia	1: 20,712
2008	(MS/MS became mandatory)	
	Conditions detected so far by MS/MS beyond MCAD & PKU):	
	Arginino Succinic Acidemia	1:265,101
	Glutaric Acidemia Type I	1:132,551
	Homocystinuria	1:265,101
	Isovaleric Acidemia	1:265,101
	Long Chain Acyl-CoA Dehydrogenase Deficiency	1:265,101
	Methylmalonic Acidemia	1: 53,020
	Transient Tyrosinemia (tx'd)	1: 13,255
	Very Long Chain Acyl-CoA Dehydrogenase Def.	1:132,551
	3-methyl crotonyl Co-A Carboxylase Def.	1: 44,184
	Hypermethioninemia	1:132,551
	Short Chain Acyl-CoA Dehydrogenase Deficiency	1: 66,275
	Carnitine deficiency due to maternal GAI	1:265,101
	Isobutyrl Co-A Dehydrogenase Deficiency	1:265,101

**Data used to determine incidence rates was from 1991 through 2012, or from the year screening began after 1991 for each listed condition.*

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 lab, PerkinElmer

Specimen data entered into data system

Specimen tested for multiple conditions

2: FOLLOW UP

Inconclusive or positive screen results reported by phone/fax from lab and state program staff to baby's health care provider

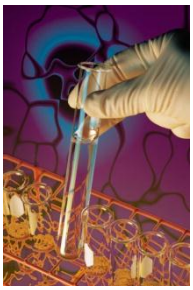
Baby's health care provider contacts parents

Parents bring baby in for confirmatory testing, and further evaluation as needed

3: DIAGNOSIS/ INTERVENTION

If screening results indicate a need:

Repeat or confirmatory testing occurs



Parent education on signs/symptoms to watch for

Baby's health care provider consults with and/or refers baby to pediatric 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 the recommendation of the pediatric specialist)

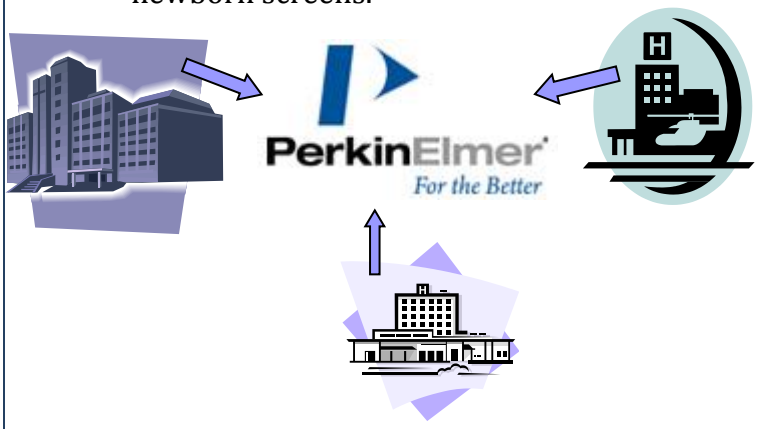
Parents receive instructions and education about treatment

Team Support services as appropriate, e.g.:

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

System Overview

In 2012, 62 birthing facilities in Nebraska sent specimens to PerkinElmer Screening Laboratory. This laboratory is under contract with the State of Nebraska to conduct all of the newborn screens.



The Newborn Screening Program in the Nebraska Department of Health and Human Services was staffed in 2012 by Mike Rooney, Administrative Assistant, Krystal Baumert, Follow-up Coordinator, Karen Eveans M.D., Follow-up Specialist, and Julie Luedtke, Program Manager.

Expert advice and assistance are available as needed throughout the year by consultation with the laboratory staff and other specialists. In 2012, the specialists in metabolic diseases were Richard Lutz, M.D., William Rizzo M.D., Jill Skrabal, R.D., Kathryn Heldt, R.D., and Rose Kreikemeier, MSN, CPNP. Consultation regarding Cystic Fibrosis was with the CF Center Director John Colombo, M.D. and Dee Aquazzino CF Center Coordinator. Pediatric endocrinologist Kevin Corley, M.D., and pediatric hematologist James Harper, M.D. were also frequently consulted.

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

Treatment services received 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 facility, 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.

MAJOR INITIATIVES of 2012 in NEBRASKA

Education

- The Nebraska Newborn Screening Program continued to track and distribute the "Parents Guide To Your Baby's Newborn Screening" to the 62 birthing facilities, Children's hospital and upon request to some Obstetric, Family Physician and Pediatric practices.

- The "QI Hints" newsletter was sent with each hospital's quality assurance reports quarterly.

- With funding assistance from the Heartland NBS & Genetics Collaborative, replacement parent education DVD's were purchased and redistributed to birthing facilities that no longer had working copies, and who committed to making these available to new parents.

- An updated electronic version of the "Practitioner's Manual" was placed on the Newborn Screening Web-page at www.dhhs.ne.gov/nsp. The program alerted appropriate health care providers about the availability of this resource.

- The "Healthcare Provider's Update" also included information on changes to the Cystic Fibrosis Screening algorithm to adopt a floating cut-off to adjust for what appears to be some seasonal fluctuation of the mean as well as lot-to-lot reagent variability. Other changes and policy issues were also discussed in that issue which can be found on the website (above).

- The spring meeting of the American Society Clinical Laboratory Science NE –Clinical Laboratory Management Association, and American Society for Clinical Pathology featured a presentation on "What's New in the Evolving World of Newborn Screening" attended by approximately 25 laboratorians from across Nebraska.

- Following publication of national guidelines on screening for cystic fibrosis from the Clinical and Laboratory Standards Institute, Nebraska modified a few practices and provided updated educational materials to health care providers. Primarily the change was for babies whose initial or repeat screens are collected at 12 days of age or older, for whom a lower Immuno-reactive Trypsinogen cut-off was implemented.

- Birthing facility nursery and NICU education coordinators were notified of the availability of a free parent education video "One Foot at a Time" available from the Save Babies Foundation. This video is an excellent resource including parent testimonials and great explanations about what newborn screening is and why it is so important. The link to this video is also available on Nebraska's NBS webpage.

- "Early Discharge and your Baby's Newborn Screen" is a new brochure to help birthing facilities explain why specimens must be collected before discharge. This brochure was made available to all birthing facilities. It is particularly beneficial for new parents who choose to leave the hospital before their baby is 24 hours old.

Policy

- The Newborn Screening Advisory Committee continued its quarterly review of quality assurance data of pre-analytical (e.g. unsatisfactory specimen rates and types), analytical (e.g. statistical performance of assays over time) and post-analytical (e.g. age at time of intervention or treatment for diagnosed patients) performance measures for the system.

- The SCID sub-committee with NBS personnel further developed screening protocols for the population of newborns in the NICU expected to have higher rates of abnormal screens for SCID. This was in preparation for the possible addition of Severe Combined Immune Deficiency (SCID). SCID has been part of the Secretary of HHS's endorsement to be included in the Recommended Universal Screening Panel since 2010.

- Financing Newborn Screening: The program uses state general funds, the newborn screening fee ($10/infant) and Title V Maternal and Child Health Block Grant funds to support access to treatment for the metabolic foods and formula. The Title V Block Grant also funds the administrative aspects of the program (education, follow up, program management and quality assurance). The state general fund appropriation has stayed the same since 1997, and the Title V Block Grant appropriation to the state is below 1997 levels. The program continues to look for creative ways to make shrinking funds go further as costs increase.

- Under the auspices of the NBS Advisory Committee, an expert subcommittee on screening for critical congenital heart disease met in 2012 and developed recommendations for policy endorsed by the larger NBS Advisory Committee. Those recommendations were shared with the leadership of DHHS as well as the Senator who introduced LB 225 requiring hospitals to provide screening for CCHD.

Quality Assurance

In 2012 quality assurance reports were sent to each birthing facility and Children's Hospital in Omaha. These reports included the individual hospital's quarterly measures on missing demographic information from the filter paper and a statewide comparison.

Special QI Initiatives

In response to increasing rates of unsatisfactory specimens, especially due to "blood spots not soaked through", a special initiative was begun. Hospitals with rates above the benchmark maximum of 0.5% unsatisfactory, were invited to voluntarily participate. A quality indicator and strategy were established that required a supervisor to quality inspect specimens before the baby was discharged, and if needed they were authorized to order repeat specimens. This resulted in almost universal reduction in unsatisfactory specimens in participating hospitals, and substantially reduced the age by which newborns with unsatisfactory specimens were recollected and had reliable screening results. Results of this initiative were shared in a poster at the 2013 National/International NBS Symposium. The poster received recognition as 2nd place in the "best poster" category judged by the Association of Public Health Laboratories.

Another quality improvement initiative undertaken in 2012 addressed the substantially longer average turnaround times for confirming babies with positive hemoglobinopathies. These efforts are described in more detail later in this report, and were reported on during an oral presentation at the 2013 National/International NBS Symposium.

NEWBORN SCREENING ADVISORY COMMITTEE

A huge debt of gratitude is owed to the dedicated members of the Newborn Screening Advisory Committee who commit their time and expertise to the Nebraska Newborn Screening Program. Much of Nebraska's success can be directly tied to their recommendations and guidance!

The Newborn Screening Advisory Committee provided technical expertise and policy guidance to the Nebraska Newborn Screening Program. Members commit at least a half day every three months to advise the state program. Representatives from PerkinElmer Genetics laboratory regularly provide input, presentations and proposals to the advisory committee. Several members provide extensive review and consultation beyond the committee meetings to help the program meet the recommendations of the larger committee.

The members in 2012 were:

- **Chair, William Rizzo,** M.D., specialist in *Pediatric Genetics,, Metabolism,* Munroe Meyer Institute for Genetics and Rehabilitation, UNMC, and Children's Hospital, Omaha
- **Vice Chair, Richard Lutz,** M.D., specialist in *Pediatric Genetics, Endocrinology, Metabolism,* Munroe Meyer Institute for Genetics and Rehabilitation, UNMC, & Children's Hospital, Omaha
- **Khalid Awad**, M.D., *Neonatologist*, Methodist Women's Hospital, Omaha
- **Lawrence Bausch**, M.D., *Neonatologist,* Lincoln
- **Angela Brennan**, M.D., *Family Physician*, St. Paul
- **John Colombo,** M.D., *Pediatric Pulmonologist, Director,* Nebraska Cystic Fibrosis Center, UNMC, 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
- **James Harper,** M.D., *Pediatric Hematologist,* UNMC, Omaha
- **Kathryn Heldt,** R.D., *Dietitian*, Children's Hospital Metabolic Clinic, Omaha
- **Mary Kisicki,** R.N., *Parent*, Papillion
- **Rose Kreikemeier,** M.S.N., C.P.N.P, *Pediatric Nurse Practitioner*, Children's Hospital Metabolic Clinic, Omaha
- **Bev Morton,** *Parent,* Lincoln
- **Samuel Pirruccello**, M.D., *Pathologist*, Regional Pathology Services, UNMC, Omaha
- **Deborah Perry,** M.D., *Pathologist*, Pathology Center, Omaha
- **Kathy Rossiter,** M.S.N, C.P.N.P., J.D., Omaha
- **Monica Seeland,** RHIA, Nebraska Hospital Association, Lincoln
- **Steven Sindelar,** M.D., *Pediatrician,* Omaha
- **Jill Skrabal,** R.D., *Dietitian*, Munroe Meyer Institute for Genetics and Rehabilitation, UNMC, and Children's Hospital, Omaha
- **Corri Stearnes,** *Parent*, Omaha
- **Leisha Suckstorf,** *Parent,* Norfolk
- **B.J. Wilson,** M.D., *Neonatologist/Perinatologist*, Saint Elizabeth Regional Medical Center, Lincoln

In response to Federal recommendations to screen newborns for critical congenital heart disease the Advisory Committee requested a subcommittee of experts in screening and treating critical congenital heart disease examine the issues and make recommendations. This subcommittee included:

- Khalid Awad, MD, Neonatology, Methodist Women's Hospital, Omaha
- Tamara Dolphen, MPAS, PA-C, Pediatric Cardiology, Children's* Omaha
- Carman DeMare, APRN Cardio-thoracic surgery, Children's* Omaha
- Cristopher Erickson, MD, Cardio-thoracic surgery, Children's* Omaha
- Bonnie Hentzen, RN, Critical access hosp, Memorial Community Health System Seward
- Howard Hsu, MD, Pediatric Cardiology, Children's* Omaha
- Ameeta Martin, MD, Pediatric Cardiology, Lincoln Pediatric cardiology
- B.J. Wilson, MD, Neonatology, Saint Elizabeth Regional Medical Center, Lincoln

*Children's Hospital and Medical Center

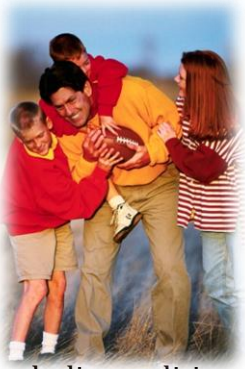
Assurance of Treatment and Management of Conditions

How the Costs of Treatment and Management are Covered:

Part of the public health assurance role of newborn screening is ensuring treatment availability and access. The state program manages several contracts for provision of otherwise prohibitively expensive formulas, foods, and services not always reimbursed by insurers. Approximately 72 patients received services through these contracts. (During any given year, some patients move out of state/new patients move in or are born/ newly diagnosed with metabolic conditions).

Insurance often 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. Therefore a large 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 tandem mass spectrometry screen. Title V Maternal and Child Health Block Grant funds (MCH funds) then provided the most

substantial support for the metabolic foods and formula exceeding $300,000 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 and necessary for proper management. Individuals can order the pharmaceutically manufactured foods from product lists provided by the manufacturers/distributors that have contracts with the State Newborn Screening Program. Families can order up to $2,000 of the pharmaceutically altered foods per year without having to pre-pay.

In Federal Fiscal Year 2012, metabolic formula ordering and distribution and specialized nutritional counseling and monitoring were provided via a contract with the University of Nebraska Medical Center for $486,301. The individuals eligible for the metabolic foods utilized the pharmaceutically manufactured foods program, ordering foods during State Fiscal Year 2012 with a value totaling $61,196.

The newborn screening program coordinates the day-to-day metabolic foods sub-program helping families understand the program and stay connected, and monitoring vendors' compliance with the contracts. Families receive a tracking log for their use in monitoring their orders and expenses and they receive an annual spending report. The program works closely with the metabolic clinic to ensure timely contract amendments of appropriate metabolically altered food products as manufacturers continue to expand their offerings. The contract for the ordering and distribution of metabolic formula is managed by the program manager and carried out by the metabolic clinic physicians and a dietitian.

Sustaining the obligation to ensure access to treatment:

The number of people with conditions requiring special formula will always increase. The metabolic diets are required for life, 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 MCH funds have been reduced or flat for several years. The Newborn Screening Program then requires a higher proportion of the MCH funds to help meet the statutory mandate. While a relatively new drug is available to which about 40% of patients with PKU are expected to respond positively, this medication is expensive as well. Therefore the program continues to look for sustainable ways to continue to assure access to needed services for people who have these conditions.

Nebraska's Newborn Screening Fees:

In 2012 the charge for newborn screening continued to be $38.50. The laboratory testing fee was $28.50 and the state fee (per statute and regulation) was $10.00 per infant screened. (State fee used only to help pay for treatment services). These fees are billed to the hospital and then are part of the hospital's charges. Hospital charges are separate and not regulated by the program. Based on the National NBS & Genetics Resource Center data, of the 47 states that charged a fee for newborn screening in 2012 only five were lower (FL, ID, LA, NC, TX).

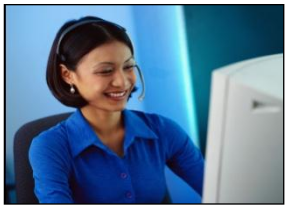
Special recognition for Nebraska's Treatment Center of Inborn Errors of Metabolism

Nebraska's Inborn Errors of Metabolism Treatment Center (Nebraska Medical Center & Children's) was one of only 5 clinics in North America (four in the U.S., one in Canada) selected to participate in a national pilot program to provide support to women with PKU who are pregnant or want to become pregnant. The maternal PKU Mentoring program will address the danger of high Phe levels in women with PKU and provide additional social support. Congratulations to the interdisciplinary team at the metabolic clinic!

The pediatric metabolic specialty treatment team from left to right: Eric Roth M.D., William Rizzo M.D., Jill Skrabal, R.D., L.M.N.T., C.D.E., .Richard Lutz M.D.

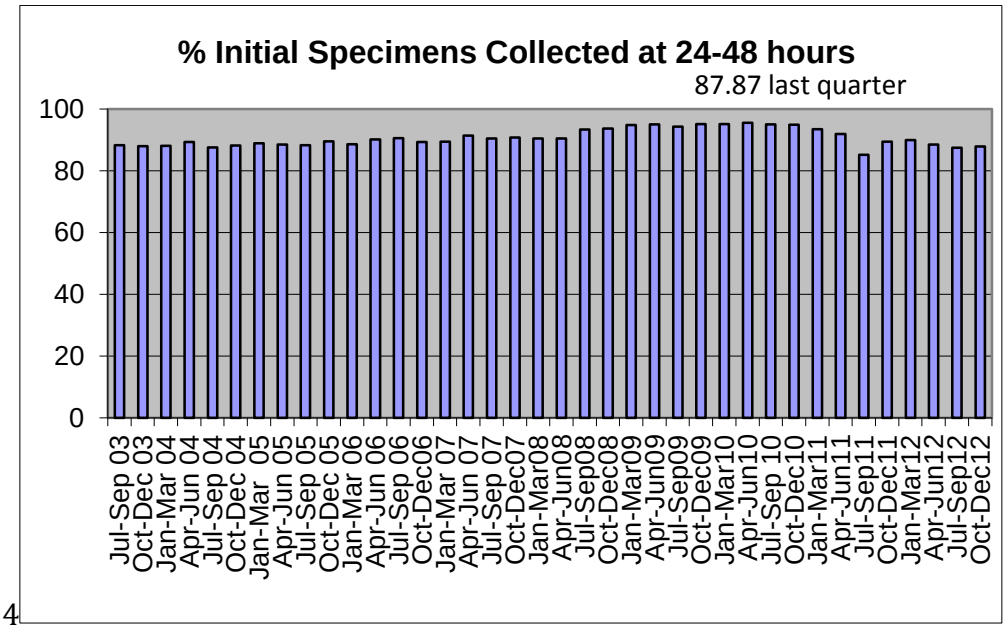
PROCESS/OUTPUT DATA FOR 2012

SPECIMEN COLLECTION, HANDLING AND TRANSPORT



***Number of births listed includes babies transferred in from other states that Nebraska's lab screened for the first time.**
****Initial specimens collected at greater than seven days were from out-of-hospital births or hospital errors.**

AGE AT TIME OF COLLECTION
Regulations require all specimens to be collected between 24-48 hours of birth, or prior to discharge, transfer or transfusion whichever comes first. Specimens collected past day two are at increased risk of a delayed diagnosis. Premature, low birthweight and sick newborns admitted to NICU's should have an admission screen collected before any treatments (other than respiratory).



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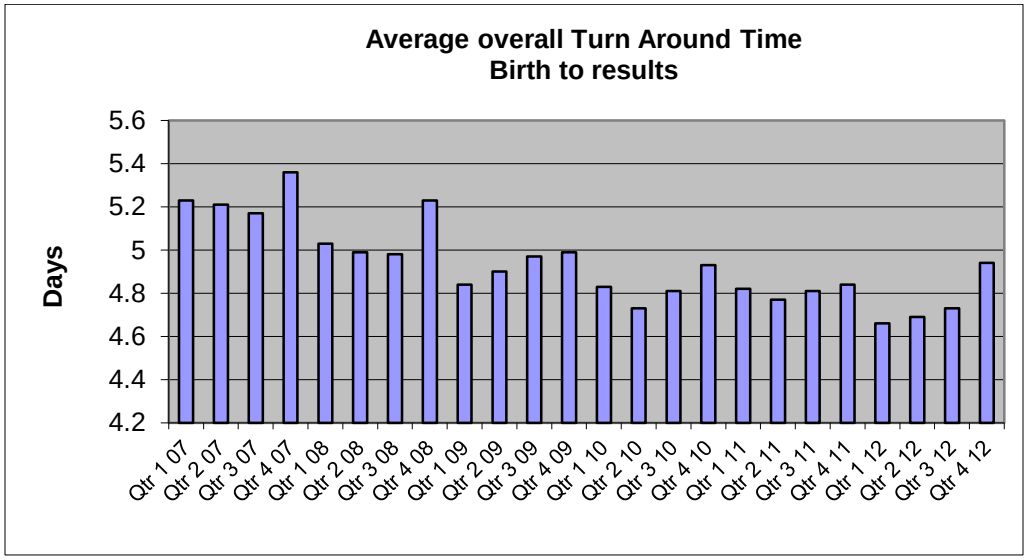
Note: Approximately 10% of specimens are collected on day one at < 24 hours of age, mostly due to NICU admission and need to collect prior to interfering treatments, but some are due to early discharge.

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.

Turnaround time's averaged below five days throughout 2012. The fourth quarter increase reflects the typical annual trend, partially due to holidays with no overnight deliveries, and partially due to winter storms that delayed shipments a couple of times in 2012.



Unsatisfactory Specimens for 2012 (Benchmark No Greater than 0.5%)

Although Nebraska's unsatisfactory specimen rate was increasing, it was still among the lowest of unsatisfactory rates in the U.S. However, because every unsatisfactory specimen requires the baby to have another specimen collected, and creates the potential for a delayed diagnosis, the program takes this issue very seriously.

It's important to reduce unsatisfactory specimens because these specimens can be costly on many levels. Repeat screens must be done requiring extra effort on the part of newborn screening follow up, hospital, screening lab and physician office personnel, plus the effort and inconvenience to families to have to return to the hospital for the repeat heel stick procedure on their infant. Although the screening laboratory does not charge for requested repeat specimens, hospital phlebotomy charges may apply. The biggest cost however could be to the newborn affected with one of the screened conditions. He/she may have a later-than-desireable age at treatment if the initial specimen is unsatisfactory delaying the age by which reliable results are available. Maintaining low unsatisfactory specimen rates is a high priority goal of the Nebraska Newborn Screening Program.

Reasons specimens were declared unsatisfactory in 2012

	Number	Percent
Not soaked through to the back of the filter paper	82	42%
Expired Filter Paper	34	18%
Quantity Not Sufficient	29	15%
Heavily applied, layered or double spotted	20	10%
Serum or Fluid mixed with specimen	12	6%
Contaminated or Diluted	7	3.5%
Exposed to Heat or Humidity	5	3%
Sample got wet	2	1%
Scratched or Abraided	1	.5%
Conflicting demographic data	1	.5%
Interfering Substance	1	.5%
Total:	194	100%

Quality Improvement for Unsatisfactory Specimens

The Program developed a quality improvement indicator and strategy with the assistance of Dennis Freer, PhD from PerkinElmer Laboratory and Samuel Pirruccello, MD from the University of Nebraska Medical Center. This Quality Indicator was shared with birthing facilities which had > 0.5% unsatisfactory specimens. Fifteen facilities were invited to participate initially and in the second quarter of implementation.

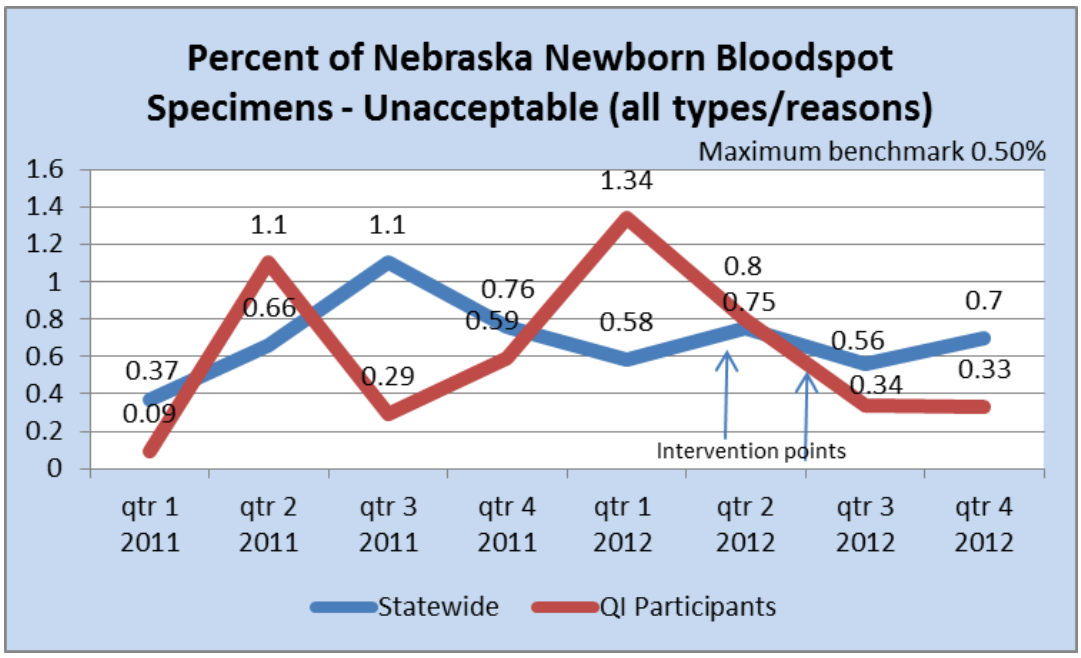
The strategy called for birthing facilities to assign a person or persons to inspect each specimen to look for blood spots "not soaked through to the other side". They were to reject those specimens and be authorized to order or collect a repeat. This was intended to not only reduce the number of rejected specimens from the newborn screening lab but to reduce the amount of time it took to get a repeat screen.

Of the 15 invited hospitals, eight voluntarily participated and almost universally every facility's performance improved or reduced the unsatisfactory specimen rate to zero!

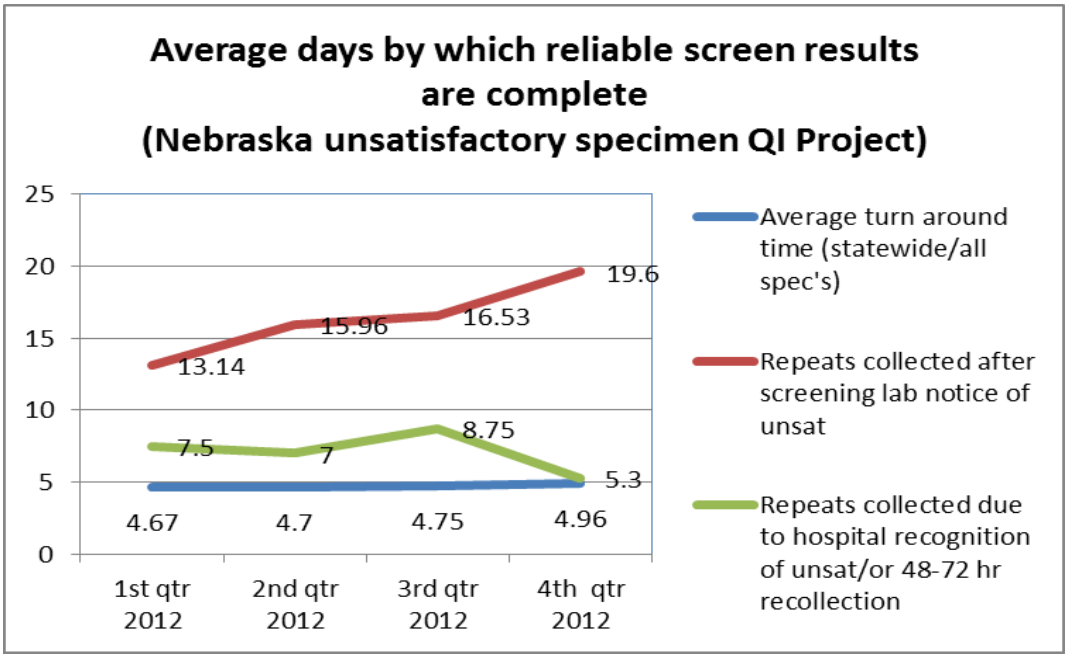
The data in table 1 and graphs 2 & 3 below support that this evidence-based strategy is now proven, and will be recommended to all birthing facilities.

Hospital	Unsat rate 4th qtr 2011	Unsat rate 1st qtr 2012	Unsat rate 2nd qtr 2012	Unsat rate 3rd qtr 2012	Unsat rate 4th qtr 2012	Unsat rate 1st qtr 2013	Unsat rate 2nd qtr 2013
Facility A	4.08%	3.92%	0%	0%	0%	8.82%	0%
Facility B	0%	5.88%	0%	4.65%	0%	0%	3.1%
Facility C	0.13%	0.69%	0.13%	0.13%	0.12%	0.13%	0%
Facility G	0%	4.35%	0%	0%	0%	0%	0%
Facility D	2.67%	6.9%	2.5%	0%	0%	1.28%	0%
Facility E	1.34%	2.04%	2.94%	1.42%	1.36%	0.81%	0.7%
Facility F	0%	0.46%	0.41%	0%	0.71%	0.79%	0%
Facility H	3.03%	0%	10.0%	0%	0%	0%	0%

Table 1



Graph 2



Graph 3

There is clear benefit in hospitals recognizing an unacceptable specimen ***before*** the baby is discharged, and collecting a second acceptable specimen. This practice reduces the days by which a complete reliable screen result is obtained. Increasing the number of repeat specimens collected earlier to replace unacceptable / unsatisfactory specimens is one way to improve turnaround time.

TESTING DATA

PerkinElmer Genetics Inc. Laboratory uses several instruments to complete the testing. While tandem mass spectrometry provides the screening for 20 of the required conditions, other methods are used for the other 8.

Presumptive Positive, Inconclusive, & Confirmed Positive Numbers & Rates

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 percentage 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.

Most of the babies requiring any follow up for abnormal results in Nebraska require only a repeat dried blood spot specimen which usually has a normal result.

- When an initial screening result is reported out as "inconclusive" the recommended follow up is a repeat dried blood spot specimen. (Most of these will be normal on repeat).
- When a screening result is reported out as "presumptive positive," the follow up is treated more urgently and usually a confirmatory test by a different method or on a different kind of specimen (serum, whole blood, urine etc.) is necessary.

Often the results are abnormal primarily because the baby was premature, sick, low birth weight, or receiving special treatment such as parenteral nutrition which can interfere with newborn screening results. These babies account for a disproportionate amount of the follow up needed. However this is not an argument to delay screening on these babies as they are at equal or possibly higher risk of having one of the screened conditions.

Condition Screened 2012 Data	**# Screened**	**# Presumptive Positive or inconclusive on screen**	**Presumptive Positive Rate**	**# lost to follow up***	**# confirmed Positive/ Diagnosed (classical or partial w tx/)**
Biotinidase deficiency	26,221	34 (29 of these were inconclusive)	0.12%	0	5 Partial deficiencies
Congenital Adrenal Hyperplasia	26,221	36 (27 of these were inconclusive)	0.13%	0	2 - Classical Congenital Adrenal Hyperplasia
Congenital Primary Hypothyroidism	26,221	96	0.36%	0	13 CPH + 2 primary hypothyroidism (not congenital); 2 hypothyroidism, and 1 transient hypothyroidism
Cystic Fibrosis	26,221	163 (127 of these were inconclusive)	0.62%	2 (1 expired, 1 parent refused confirmatory sweat test)	7 Cystic Fibrosis, 2 CF Related metabolic syndrome (CRMS)
Galactosemia	26,221	2	0.007%	0	0
PKU	26,221	5	0.01%	0	4 PKU Classical 1 Hyperphenylalaninemia
Sickle Cell Disease & other clinically significant hemoglobinopathies (hgbs)	26,221	7	0.02%	0	3 Sickle Cell Disease 1 SC-Disease 1 Hgb D/Beta 0 Thal 1 Hgb C Disease 1 Hgb E Disease
All other abnormal hgbs (carriers/variants)	26,221	401	1.5%	117 no dx but 89 of these had confirmatory testing**	157 Sickle trait 40 Hgb C trait 19 Hgb E trait 4 Hgb D trait 28 Misc. traits 4 Alpha Thal Silent Carrier 12 Alpha Thal Trait
MS/MS Screened Disorders (20 primary targets of amino acid, fatty acid and organic acid conditions):	26,221	436 (all MS/MS results)	1.66%	12 (expired before confirmed or repeated)	(See PKU above) plus 4 MCAD 2 SCAD 2 Transient Tyrosinemia 1 Mild (Not tx'd) Hypermethioininemia

* None of these were suspected of clinically significant conditions in infancy, but screen results suggested various traits.

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.

Several factors can conspire to create delays in treatment, so speed and persistence in follow up are essential. Some examples of these factors include babies with prolonged treatment in NICUs, parental resistance to confirmatory testing, problems in locating parents because contact information provided to the hospital or recorded on the filter paper collection cards was incorrect or no longer accurate.

Condition & number of babies diagnosed	Average age (days)at intervention/tx.	Range in ages at intervention/tx.
5 Biotinidase Deficiency (Partials)	18	6-31
2 Congenital Adrenal Hyperplasia	9	6-13
18 Congenital Primary Hypothyroidism (includes other forms of hypothyroidism)	10	6-42
9 Cystic Fibrosis	14	4-23
4 MCAD	4	0-11
4 Phenylketonuria + 1 Hyperphe	5	3-7
1 Sickle Hgb. C Disease	57	n/a
3 Sickle Cell Disease	28	16-52
1 Hgb. C Disease	16	n/a
1 Hgb D / Beta) Thal Disease	52	n/a
1 Hgb E Disease	16	n/a
2 SCAD	52.5	46-59
2 Transient Tyrosinemia	12.5	11-14
1 Mild Hypermethioninemia	Not tx'd	n/a

2012 Outcome Data for Newborn Screening

2012 Nebraska PKU
Long-term follow-up measures of patient outcomes:

Measures to evaluate patient outcomes are important to evaluating the effectiveness of the intervention, and to support the need for continued funding of the contract for metabolic formula to support patients identified with metabolic conditions via newborn screening who require the formula.

Three measures are agreed upon between the Nebraska Newborn Screening Program which administers the metabolic formula contract, and the University of Nebraska Medical Center, Metabolic Clinic in collaboration with the Children's Metabolic Clinic. Here is a breakdown of phenylalanine levels in the state of Nebraska for the calendar year 2012:

1) **Percent of patients maintaining their average phenylalanine levels within the optimal range for their age group/demographic.**

   In the birth to 6 years of age group, the average phenylalanine level was 2.8 mg/dL All (100%) had an average phenylalanine level in the optimal 2-6 mg/dl range.

   In the 6 to 12 year age range, the average phenylalanine level for 2012 was 5.3 mg/dl. (91%) had an average phenylalanine level in the optimal 2-8 mg;dl range.

   In the 13 to 18 year age range, the average phenylalanine level was 9.6 mg/dL. (50%) had an average phenylalanine level in the optimal 2-10 mg/dL range.

   In the 19 years and over age category, the average phenylalanine level was 11.8 mg/dL (48%) had an average phenylalanine level in the optimal 2-10 mg/dL range.

   There were 3 pregnancies in the 2012 calendar year. One hundred percent of these pregnancies kept their average pregnancy phenylalanine level in the optimal 2-6 mg/dl range, with an average phenylalanine level during pregnancy of 4.7 mg/dl among the group.

1) **Percent of patients age 0-12 who are meeting all developmental milestones as assessed/determined by the pediatric metabolic specialist MD"**

In the year 2012, all children ages 0-12 were assessed by the pediatric metabolic specialist as "meeting developmental milestones."

Several children have been diagnosed with ADHD and are currently being treated with medication. One patient has an unrelated neurological disorder which is requiring special education, and one other patient was requiring special education for school difficulties.

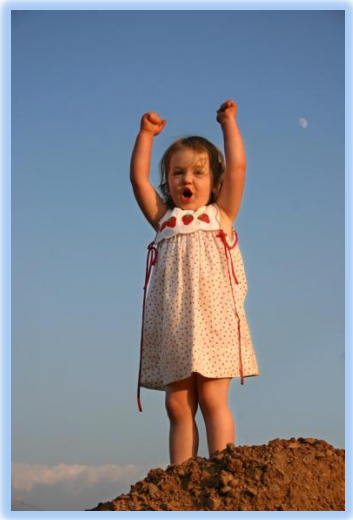
2) Number and percent of patients > 18 years of age who have graduated high school and college:

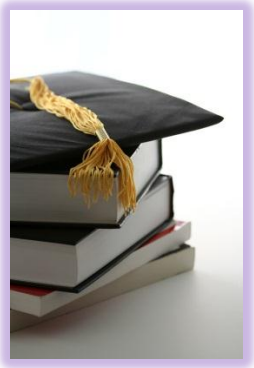
In 2012, 97% of patients ages >18 had a high school diploma
In 2012, 6% had earned Associates Degrees, 39% had earned Bachelor's degrees, with another 6% currently working towards a Bachelor's Degree
15% had earned Master's Degrees
3% currently attending medical school

State high school graduation rates compare as follows: In 2010-11, the most recent data available from the US Department of Education:

Nebraska (all students) had a graduation rate of 86%. Only three other states had higher rates (Vermont and Wisconsin at 87% and Iowa at 88%). The substantially higher rate of graduation in our population of people with PKU speaks very highly of the individuals, their families and the health care and education professionals working with them long term!

This data supports the outstanding success of newborn screening for metabolic conditions, when long term management and treatment is maintained.





NEBRASKA EARLY HEARING DETECTION AND INTERVENTION ANNUAL REPORT - 2012

The Nebraska Early Hearing Detection and Intervention Program develops, promotes, and supports systems to ensure all newborns in Nebraska receive hearing screenings, family-centered evaluations, and early intervention as appropriate.

Introduction

Approximately one to three in 1,000 babies are born with permanent hearing loss, making hearing loss one of the most common birth defects in America. Before newborn hearing screening, children who were deaf or hard of hearing sometimes were not identified until 2-½ to 3 years of age. Left undetected, this delayed identification can negatively impact the child's speech and language acquisition, academic achievement, and social and emotional development. If detected soon after birth, the negative impacts can be reduced and even eliminated through early intervention.

The Infant Hearing Act became a state law in Nebraska in 2000 and required the hearing screening of newborns in birthing facilities in Nebraska as a standard of care. Also in 2000, the Nebraska Department of Health and Human Services started the Nebraska Newborn Hearing Screening Program. Today the program is known as the Nebraska Early Hearing Detection and Intervention (NE-EHDI) Program and is funded through federal grants. This program strives to fulfill the following four main 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.
- To prevent or mitigate the developmental delays and academic failures associated with late detection of hearing loss.
- 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.

The act also requires birthing facilities to educate parents about newborn hearing screening and any necessary follow-up care. The education includes the hearing screening test, the likelihood of the newborn having a hearing loss, follow-up procedures, and community resources, including referral for early intervention and a description of the normal auditory, speech, and language developmental process in children. The Act also

requires that regulations be promulgated to mandate newborn hearing screening if less than 95% of newborns in the state receive a hearing screening.

There are two basic techniques available to screen newborns for hearing loss. Both are easily performed on newborns and are non-invasive measures to determine auditory functioning.

The most frequently used screening technique is measurement of otoacoustic emissions, or OAE. A miniature earphone and microphone are placed in the newborn's ear canal, low intensity sounds are emitted, and responses produced by the inner ear are measured. The second screening technique, Auditory Brainstem Response (ABR), uses small electrodes to detect certain brainwaves in response to sounds from the miniature earphone. For both methods, the response of each ear is measured. Equipment using either technology is 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. The picture below shows an infant receiving an OAE hearing screening.

OAE Hearing Screening

If a response is not detected for one or both ears, the result is a "refer" (did not pass). A "refer" on the screening test indicates possible hearing loss in one or both ears but there are also other factors that may have contributed. A "refer" indicates 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 or equipment failures contributed to the initial result. A "refer" on the outpatient (second) screening indicates the need for a diagnostic audiologic evaluation to confirm or rule out hearing loss. Early intervention services are an option for families in the event of confirmed hearing loss.

Each birthing facility has established a newborn hearing screening protocol. In the event of a "refer" inpatient screening, the outpatient screening will usually be performed by the hospital, an audiologist or physician.

Newborn Hearing Screening Data Reported for 2012

Birthing Facility Screening Programs

Since 2003, 100% of the birthing facilities in Nebraska have been 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. In 2012 there were 55 birthing facilities using OAE, ABR or both for inpatient screening methods and one hospital with a visiting audiologist who performed post-discharge OAE screening.

Hearing Screening at Birthing Facilities and Birthing Centers

In 2012 inpatient hearing screenings were reported on 26,021 newborns, or 99.34% of the 26,193 newborns available for an inpatient screening. There were 90 home births with 23 of these transferring to a Nebraska hospital. The percentage of newborns screened during

birth admission has increased dramatically since reporting began in 2000, when only slightly more than one-third of newborns received a hearing screening during birth admission.

In Nebraska, 25,179 (96.76%) newborns passed the inpatient hearing screening. An outpatient screening or audiology evaluation is recommended for infants who do not pass the inpatient screening or who do not receive the inpatient screening.

Parent Education

Recommending a hearing screening test has been operationally defined as educating parents about newborn hearing screening, hearing loss, and normal communication development as required by Neb. Rev. Stat. §71-4740. The NE-EHDI Program provides print and video educational materials free of charge to hospitals to help fulfill this requirement. Print materials are available in 10 languages.

Birthing facilities reported educating over 99% of parents about newborn hearing screening, hearing loss, and normal speech and language development in 2012. The statute also requires the Nebraska Department of Health and Human Services 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. This is accomplished through letters and printed materials sent to the parents, along with phone calls.

Monitoring, Intervention, and Follow-up Care

The NE-EHDI Program's tracking and follow-up processes are followed for each baby who is reported as not passing the hearing screening during birth admission and for infants not receiving the inpatient hearing screening. In 2012, a total of 1,013 infants (hospital and non-hospital births) were tracked to encourage the parent(s) to have the infant receive an outpatient hearing screening or audiologic diagnostic evaluation.

The following shows the outcome of the 1,013 infants tracked by the NE-EHDI program:

833 Passed outpatient screening and/or diagnostic testing (Closed)
52 Parent(s) refused to complete the screening/testing process
46 Lost (no response to NE-EHDI letters and phone calls)
31 Diagnosed deaf or hard of hearing
30 Middle-ear problems or inconclusive diagnostic testing
9 Moved out of Nebraska
7 Expired outpatient
5 Pending diagnostic testing

A total of 890 infants in the above group received an outpatient screening. Some of infants skipped the outpatient screening and had an audiologic evaluation.

Confirmatory Testing/Audiologic Data Reported for 2012

The Advisory Committee for the NE-EHDI Program identified the initial level of follow-up hearing tests as an outpatient screening of the newborn's hearing. Since the majority of newborns will pass this outpatient screening, considerable cost savings can result by using either the OAE and/or ABR screening technique rather than proceeding directly to a complete audiologic diagnostic evaluation. According to the individual results reported by audiologists to the NE-EHDI Program, a total of 86 infants received a complete audiologic evaluation in 2012. Thirty-one were diagnosed as deaf or hard of hearing after not passing on the inpatient screening and one identified with a late-onset hearing loss due to meningitis at about four months old.

Timeliness of Follow-up Screening / Evaluations / EDN Services

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."

To meet the state and national guidelines, established by the Joint Committee on Infant Hearing (JCIH) of "1-3-6" (hearing screening completed by 1 month, audiologic diagnostic evaluation completed by 3 months, early intervention initiated by 6 months), the timeliness of initiation and completion of follow-up activities is an important aspect of the quality of services. Over 99% of infants received an inpatient screening within one month of age. For the newborns who were recommended for an audiologic diagnosis, 58.1% received the evaluation by three months of age according to individual data received by the NE-EHDI Program from audiologists.

Records for the Early Development Network (EDN), Nebraska's Part C Early Intervention Program, indicate that 23 (85.2%) out of the 27 infants residing in Nebraska in 2012 and identified with a hearing loss, were referred to EDN within six months. Four were not verified for EDN within six months and four resided outside of Nebraska and were not eligible for Nebraska EDN services.

ACTIVITIES – 2012

Funding

The NE-EHDI Program continued to receive funding from the Health Resources Services Administration/Maternal and Child Health Bureau (HRSA/MCHB) and the Centers for Disease Control and Prevention (CDC). The HRSA/MCHB grant funded the basic operations of the NE-EHDI Program. The CDC cooperative agreement funding supported the development and implementation of the integrated electronic data reporting and tracking system.

Advisory Committee

The NE-EHDI Program was developed based on the requirements identified in the Nebraska Infant Hearing Act of 2000 and the recommendations of the NE-EHDI Program Advisory Committee. The purpose of the advisory committee, according to its Charter, is to provide direction and guidance to the NE-EHDI Program regarding the newborn hearing screening system. Specific advisory committee activities include, but are not limited to, the following:

- To discuss and advise on the goals for the NE-EHDI Program.
- To advise on the improvement of reporting, tracking, and follow-up protocols to effectively link the NE-EHDI Program and early intervention systems.
- To assist in increasing the Program's responsiveness to the expanding cultural and linguistic communities in the state.
- To guide the long-term planning and evaluation of the NE-EHDI system in the state.
- To review the quarterly newborn screening statistics and make recommendations for program improvements.

The advisory committee of the NE-EHDI Program consists of no more than 20 voting members representing the following:

- Audiologists
- Deaf/Hard of Hearing community
- Early Intervention Services
- Ears, Nose and Throat Specialist/Otorhinolaryngologists or Otologist
- Family Support
- Hospitals (preferably hearing screening coordinator)
- Parents
- Pediatrics
- Public Health

Advisory committee meetings are held four times a year and open to the public.

Projects

Hearing Screening Equipment for Birthing Facilities

Opportunities to contract for partial funding of new hearing screening equipment were offered to the Early Development Network, Early Head Start, Community Health Clinics, Regional Programs for Students who are Deaf/Hard of Hearing, and Educational Services Units. New equipment should reduce the number of babies who refer due to the use of aging or inappropriate hearing screening equipment. A total of five contracts were awarded in 2012.

Hospital Site Visits

In the fall of 2012, the program manager and business analyst traveled to western Nebraska to visit four hospitals. The purpose of these visits was to determine what assistance the NE-EHDI Program could provide, how to lower refer rates, and to establish relationships with the hospitals. Two of the four hospitals that were visited showed improvement on their inpatient refer rates after the site visit.

Clinic With a Heart

In 2011, the NE-EHDI Program purchased two OAE screeners, one to loan to birthing facilities whose equipment was not working and one for community use which was loaned to Clinic With a Heart in 2012. The University of Nebraska-Lincoln Student Academy of Audiology (UNL SAA) provided hearing screenings at no charge once a month at Clinic With a Heart. Clinic With a Heart is a way for individuals who are either underinsured or have no insurance to get health care.

If an individual refers on the screening, he or she is referred to the Sharing Clinic which occurs four times a year at the Barkley Center on East Campus. At the Sharing Clinic the individual is given a full diagnostic evaluation and if they qualify for hearing aids attempts are made to set them up with donated hearing aids. In April, UNL SAA awarded Clinic With a Heart a Community Outreach Award for its dedication and hard work to provide services for these individuals.

Children's Hearing Aid Loaner Bank

The Nebraska Children's Hearing Aid Loaner Bank (NCHALB) began providing loaner hearing aids to young children in January 2008. The NCHALB was a partnership between the University of Nebraska-Lincoln Barkley Center, Nebraska Association for the Education of Young Children (NAEYC), and the NE-EHDI Program. The program is now known as the Nebraska Children's Hearing Aid Bank/HearU Nebraska. The NE-EHDI Program continued to provide funds for administration of the program and to help purchase loaner hearing aids in 2012. Twenty-seven children were fitted with 44 hearing aids in 2012 with the total number of 131 children fitted with hearing aids from 2008 through 2012.

Family-to-Family Support

The Family Support Work Group, a subcommittee of the NE-EHDI Program Advisory Committee, provided input regarding parent education materials and planning for family support activities. Partnership with the Nebraska chapter of Hands and Voices continued, including exploration of establishing a mentoring program to provide parent-to-parent support when a young child is identified with a permanent hearing loss. They also reviewed changes being proposed to the NE-EHDI Program parent education brochures and plan to explore the needs of Hispanic populations related to hearing loss.

The NE-EHDI Program also provided support and assistance to Hands and Voices to successfully apply for and receive approval to establish a Nebraska Guide By Your Side program. Funding from Part C (known in Nebraska as the Early Development Network) to support those efforts was also received.

Roots and Wings Parent Weekend

The sixth *Roots and Wings* parent weekend was held September 28-30, 2012 in Nebraska City at the Lied Lodge. The parent weekend targets families with children up to three years old through a contract with the Boys Town National Research Hospital. The goal of this workshop was to provide: 1) families basic information on hearing loss, 2) an overview of current hearing technology, 3) knowledge on the various ways to communicate with deaf or hard of hearing individuals, 4) emotional support during the difficult period after a family receives the diagnosis, and 5) an opportunity to network with other families. The positive survey results on the sessions and activities were shared in a presentation to the NE-EHDI Program Advisory Committee.

Parent Workshops

Hispanic Panel – Outreach to the Hispanic community has been a focus of the Family Support Work Group. In collaboration with Nebraska Hands and Voices, the Metro Regional Program, and Parent Training and Information (PTI) Nebraska held a panel discussion with Spanish-speaking families. The University of Nebraska-Omaha's Spanish Club and three students from the ASL Club assisted along with students volunteers from UNO who provided child care. The Nebraska Commission for the Deaf and Hard of Hearing was also represented and EduCare Omaha provided the facility at no cost.

There were four interpreters, two ASL and two Spanish. The families truly benefited from the presentation and said they sincerely appreciated the effort that went into planning the event. Translating the three languages was a challenge, but everyone worked together the make it happen. Eight families attended, consisting of 12 adults and 23 children.

Parent Advocacy Workshop – The NE-EHDI Program worked with Hands and Voices to organize a workshop for parents of deaf or hard of hearing children in the Hastings, Nebraska area. It was provided in collaboration with the Central/Western Nebraska Regional Program for Students who are Deaf or Hard of Hearing and PTI-Nebraska with child care provided by the Regional Program. The Nebraska Commission for the Deaf and Hard of Hearing was also represented. The purpose of the workshop was to help parents build skills for effective parental advocacy for their child. Seven adults and 13 children attended the event.

CDC Site Visit

In August, 2012, four representatives, including the Nebraska Centers for Disease Control (CDC) liaison, visited the NE-EHDI Program. The CDC sought to see how the data compiled by the NE-EHDI Program was being used, not just in how the data was being collected and stored. The CDC representatives were very interested in the NE-EHDI Program's data system and quite impressed with how well the data is being utilized. They encouraged the

NE-EHDI Program to think about their five-year plan and the overall goal for the data system. They also suggested that the NE-EHDI Program staff consider what it wants to accomplish and what improvements are needed.

Electronic Data System

The NE-EHDI Program is partially funded by the CDC. A portion of the CDC funds paid for a 2012 enhancement to the Nebraska electronic data system for the NE-EHDI Program.

This enhancement corrected three weaknesses in the current system: 1) creating hearing information records when there is not a birth certificate - the current system creates the hearing record from the birth record and sometimes the birth certificate is not created for several months after the inpatient or outpatient hearing screening, 2) logging all incoming and outgoing communication between the NE-EHDI Program and all other people involved in the care of children tracked by the NE-EHDI Program (e.g., parents, physicians, audiologists, medical staff, early head start, and early intervention services) along with planned follow-up communication and, 3) recording services received by those children identified as deaf or hard of hearing as well as recording outcomes. This enhancement was in the final testing phase at the end of 2012.

iEHDI CDC Contract

The NE-EHDI Program provided data to the Centers for Disease Control (CDC) as a partner in the iEHDI Data Project in 2012. The purpose of the contract was to "…obtain a limited set of existing, individual level data from a minimum of three states." This data will be used to determine ways to improve the quality and completeness of Early Hearing Detection and Intervention (EHDI) data at the national level and help address questions related to assessing progress towards national EHDI benchmarks. The information in the data set excludes direct identifiers, such as name of the individual or of relatives, employers, or household members of the individual. The iEHDI project will end in 2013.

Summary

- All the current birthing hospitals in Nebraska were conducting newborn hearing screening in 2012. All but one had conducted the hearing screenings prior to discharge from the hospital or birthing center.

- In 2012, birthing hospitals reported screening the hearing of over 99% of newborns prior to discharge from the hospital.

- The overall "refer" rate during 2012 for hearing screening during birth admission was 3.2%.

- In 2012, audiologic evaluations were initiated within three months of age for almost 60% of newborns when the newborn did not pass the inpatient screening.

- There were 81 babies born in 2012 whose hearing status was not objectively established, excluding the 97 who expired before receiving or completing screening.

- The incidence of Permanent Congenital Hearing Loss identified and reported to the NE-EHDI Program (1.2 per thousand screened in 2012) is within the anticipated range of one to three per thousand.

- Almost 86% of the infants with hearing loss and residing in Nebraska were verified for the Early Development Network and received special education services within six months of birth.

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

Julie Luedtke, 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, drawn early specimens
Karen Eveans, NBS Follow up Specialist 402-471-6558
Hemoglobinopathies and cystic fibrosis, unsatisfactory specimens
Susie Lyness, Administrative Assistant 402 471-9731
Metabolic foods, patient education materials, advisory committee and staff support

WEBPAGE: http://dhhs.ne.gov/publichealth/Pages/nsp.aspx
E-mail contact: dhhs.newbornscreening@nebraska.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

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

PerkinElmer Genetics Screening Laboratory Vice President and General Manager, Bill Slimak 412-220-2300

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

Kathy Northrop, Early Hearing Detection & Intervention (NE-EHDI) Program Manager 402-471-6770
Program planning, evaluation and management, systems development

Jim Beavers, Business Analyst, NE-EHDI Program 402-471-1526
Data system planning and testing, development of reports, system security, training and technical assistance

MeLissa Butler, Community Health Educator, NE-EHDI Program 402-471-3579
Follow up, patient education materials distribution, data management

Debie Seiler, Community Outreach Coordinator, NE-EHDI Program 402-471-1440
Follow up, community outreach and education

Kelci Kilthau, Community Health Educator, NE-EHDI program 402-471-6746
Follow up, complex diagnostics, special projects

Nebraska Early Hearing Detection & Intervention Program
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