

NEWBORN SCREENING IN NEBRASKA

Newborn Screening for Metabolic & Inherited Disorders

and

Early Hearing Detection & Intervention

See new data
presented for the first
time on the outcomes
of Nebraska patients
with metabolic
conditions!



2011 Annual Report

Department of Health & Human Services

DHHS
NEBRASKA

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**NEBRASKA EARLY HEARING DETECTION AND INTERVENTION
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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 2011, newborn screening efforts resulted in successfully identifying and treating 35 newborns affected with conditions in time to prevent problems associated with them:

- ❖ 3 babies with partial (treated) biotinidase deficiency (BIO)
- ❖ 1 baby with congenital adrenal hyperplasia (CAH)
- ❖ 4 babies with congenital primary hypothyroidism (CPH), 3 with congenital hypothyroidism, and 1 hypothyroidism-prematurity
- ❖ 6 babies with cystic fibrosis
- ❖ 1 baby with classic galactosemia and 2 with Duarte galactosemia
- ❖ 7 babies with hemoglobinopathies (2 sickle cell disease, 3 SC-disease, 1 C-disease, and 1 hemoglobin E disease)
- ❖ 1 baby with isovaleric academia (IVA)
- ❖ 3 babies with phenylketonuria (PKU)
- ❖ 5 babies with transient tyrosinemia who responded to treatment

The incidence rate of conditions in Nebraska based on the screened conditions identified from 2007-2011 and number of births screened those five years:

1:620 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 2011, Nebraska's required screening panel included 28 metabolic, endocrine, hematologic and other conditions.

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

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 a screened 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/letter
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

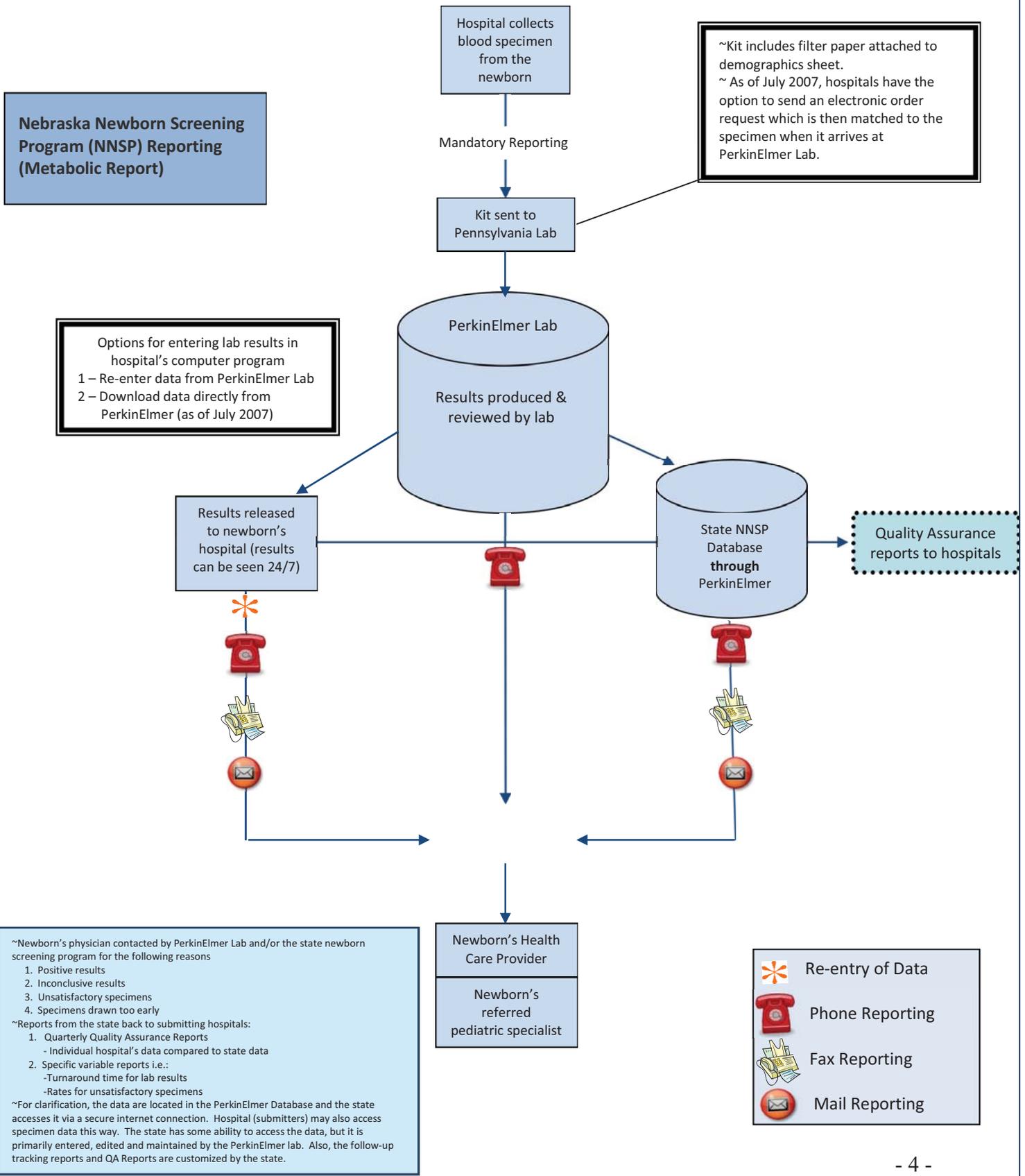


Team Support services as
appropriate, e.g.:

- 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



Data Flow: This chart demonstrates how data from newborn screening is produced, transmitted and used to facilitate the recall of newborns at risk for any of the screening panel conditions. They can then be evaluated, diagnosed and have treatment initiated.



System Overview

In 2011, 59 Nebraska hospitals 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 by Mike Rooney, Administrative Assistant, Krystal Baumert, Follow-up Coordinator, Karen Eveans, Follow-up Specialist, and Julie Luedtke, Program Manager.

Expert advice and assistance were available as needed throughout the year by consultation with the laboratory staff and other specialists. 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, RN. Consultation regarding Cystic Fibrosis were 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 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 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.



MAJOR INITIATIVES of 2011 in NEBRASKA

Education

- ❖ Mike Rooney of the Nebraska Newborn Screening Program continued to track and distribute the “Parents Guide To Your Baby’s Newborn Screening” to the 59 birthing hospitals, Children’s hospital and upon request to some Obstetric, Family Physician and Pediatric practices.
- ❖ The program surveyed obstetric care providers regarding their willingness to provide a one-page education piece developed by the program, to expecting women during their third trimester. Responses received overwhelmingly supported their willingness to provide this education. Supplies were sent in 2011 to 10 OB practices representing 21 obstetricians and 23 practices representing 72 family physicians providing obstetric services. A follow-up survey to determine ease-of-use also resulted in positive responses. All but one practice reported it was “very easy” and one practice reported it was “easy” to incorporate the one-pager into their routine parent education practices.
- ❖ Local presentations on newborn screening included Pediatric Grand Rounds at Children’s Hospital in June 2011 by Drs. John Colombo, Richard Lutz, James Harper, and newborn screening program manager. Julie Luedtke also presented on the Clinical and Laboratory Standards Institute’s (CLSI) guidelines for premature, low birth weight and sick newborns at the Latin American Congress for neonatal screening of inborn errors in metabolism in Cusco, Peru in September.
- ❖ Internal staff development efforts included the program manager and follow-up staff attending the Association of Public Health Laboratory’s (APHL) National Newborn Screening (NBS) & Genetics Symposium in November. The program manager also attended the Heartland NBS & Genetics meeting in Bismarck, ND in August.
- ❖ Educational mailings were sent out in March and June with FAQs to prepare pediatricians, family physicians, neonatologists, hospital laboratory and nursery personnel for Neonatal Intensive Care Unit (NICU) protocol changes effective in July 2011. A Health Care Provider’s update was sent out in October covering the impact of the NICU changes, evaluation of SCID for newborn screening, and quality assurance issues addressed during the year.

Policy

- Newborn Screening Program staff Krystal Baumert, Karen Eveans and Julie Luedtke continued to serve on the Newborn Screening Committee of the Heartland Newborn Screening and Genetics Collaborative. Karen Eveans served as an advisor to the CLSI workgroup developing guidelines for screening for cystic fibrosis. She also served as the Heartland’s representative to the NBS Clearinghouse’s education materials workgroup. The program manager continued to serve on: the APHL’s Newborn Screening & Genetics Committee and the Heartland Region’s Advisory Council.

- 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 Newborn Screening Advisory Committee reviewed and evaluated several technical issues and changes proposed by the newborn screening laboratory relative to screening algorithms for cystic fibrosis and congenital adrenal hyperplasia. The committee also approved new information to be placed on newborn screening test results for babies who had abnormal screen results and were low birth weight, to provide physicians additional information based on birth weight ranges.
- With the implementation of admission screening prior to treatment for low birth weight, premature and sick newborns to NICUs in July of 2011, the program experienced a significant increase in drawn early specimens resulting in many more inconclusive results for CAH, CPH and CF. While this was anticipated and the repeat screens often resolved the findings as normal, the burden on the system was substantial, and the concern with unnecessarily alarming parents increased. As a result of ongoing analysis of this situation, the Newborn Screening Advisory Committee approved a proposal to no longer screen specimens collected at less than 24 hours for CAH, CPH and CF. This was implemented in October 2011 for CAH and CPH, and in January 2012 for CF.
- The committee continued its evaluation of Severe Combined Immune Deficiency (SCID) as a candidate condition for screening and in July 2011 recommended it for screening in Nebraska's panel. Meetings were held with Medicaid and private insurers to discuss implications and address questions. The Secretary of Health and Human Services had endorsed the recommendation by the Secretary's Advisory Committee on Heritable Diseases in Newborns and Children to make SCID part of the Recommended Universal Screening Panel (RUSP) in 2010.
- The regulation revisions submitted in 2010 addressing these issues were signed by the Governor in June, effective July 2, 2011:
 - the storage, use and disposal of residual newborn dried blood spots
 - the quality of information to be included on confirmatory laboratory test results
 - adoption of the current edition of the Clinical and Laboratory Standards Institute (CLSI) standards for blood collection which would allow (with certain precautions) the collection of blood spots via umbilical catheter for newborns in the neonatal intensive care unit
 - adoption of CLSI guidelines for premature, low birth weight and sick newborns admitted to NICU's for serial screening
 - clean-up language and clarifying definitions
- 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. Title V Block

Grant funds support 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.

Quality Assurance

In 2011 quality assurance reports were sent to each birthing hospital 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. The ability to produce the quality assurance reports of other measures such as turnaround time and unsatisfactory specimen rates for each hospital, were made available the last two quarters of the year.

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 provided input, presentations and proposals to the advisory committee. Several members provided extensive review and consultation beyond the committee meetings to help the program meet the recommendations of the larger committee.

The members in 2011 were:

- **Chair, William Rizzo**, M.D., specialist in *Pediatric Genetics, Endocrinology, 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 Nebraska
- **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
- **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**
- **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



Assurance of Treatment and Management of Conditions

How the Costs of Treatment and Management are Covered:

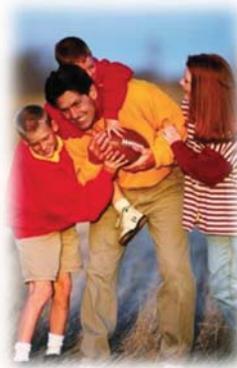
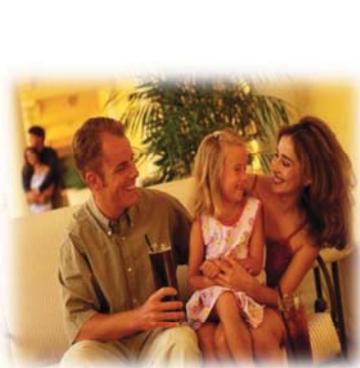


Part of the public health assurance role of newborn screening is ensuring treatment availability and access. Toward that end, the state program manages several contracts to ensure provision of otherwise prohibitively expensive formulas, foods, and services not always reimbursed by insurers. Approximately 65 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 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. 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 6 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 2011, metabolic formula ordering and distribution and specialized nutritional counseling and monitoring were provided via a contract with the University of Nebraska Medical Center for \$574,769. The individuals eligible for the metabolic foods utilized the pharmaceutically manufactured foods program, ordering foods during State Fiscal Year 2011 with a value totaling \$57,173.



Mike Rooney coordinates the day-to-day metabolic foods program helping families understand the program and stay connected, and monitoring vendors' compliance with the contracts. He provides a tracking log to families for their use in monitoring their orders and expenses and provides a mid-year spending report to each family. He also works closely with Jill Skrabal, RD 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.

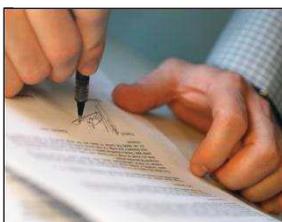
Nebraska's Newborn Screening Fees

In 2011 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 2011 only 5 were lower (FL, ID, LA, NC, TX).



PROCESS/OUTPUT DATA FOR 2011

SPECIMEN COLLECTION, HANDLING AND TRANSPORT



Age at Time of Specimen Collection (Initial Specimen) 2011

Age at time of collection	Number of births*	Percent of births
0-12 hours	1314	5.01
12-24 hours	196	.75
Collected day 2 (24-48 hours of age)	24075	91.85
Day 3	374	1.43
Day 4	44	0.17
Day 5	23	0.09
Day 6	6	0.02
Day 7	10	0.04
Over 7 days**	165	0.63
Time of collection unknown	0	0

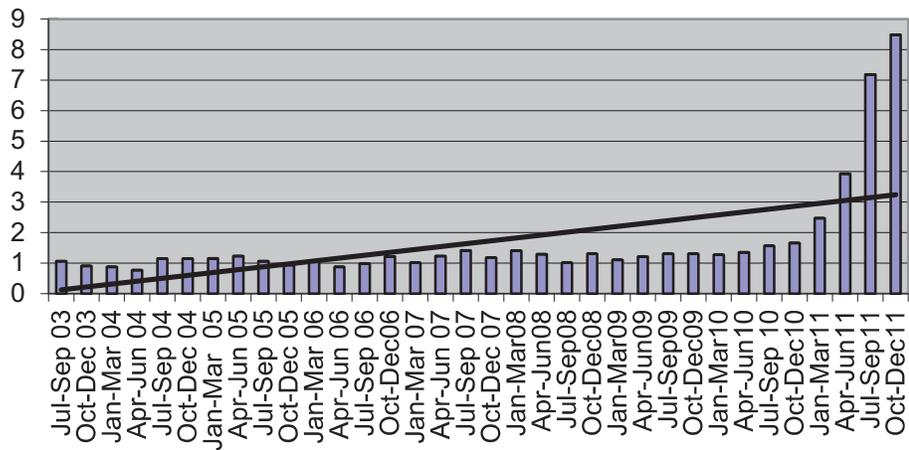
*Number of births listed includes babies transferred in from other states that Nebraska's lab screened for the first time.

**Initial specimens collected at greater than 7 days were from out-of-hospital births or hospital errors.

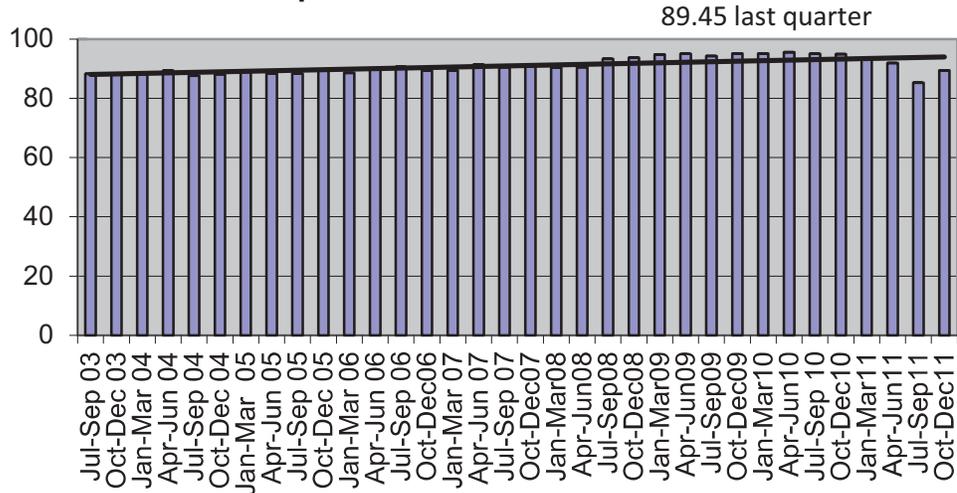
Regulations require all specimens to be collected between 24-48 hours of birth, or prior to discharge, transfer or transfusion whichever comes first. Specimens collected past day two are at increased risk of a delayed diagnosis. Premature, low birthweight and sick newborns admitted to NICU's should have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

In July of 2011, Nebraska implemented revised regulations requiring serial screening of all NICU admissions prior to any treatments (other than respiratory). Education about this policy change began late in 2010, and FAQs were distributed to hospitals and posted on the program's website in the spring and summer of 2011 to help prepare hospitals to implement. The dramatic increase in specimens collected at less than 24 hours from an average around 1 to 1.5% to greater than 8% reflects the adoption of the regulation in some facilities even before it became official.

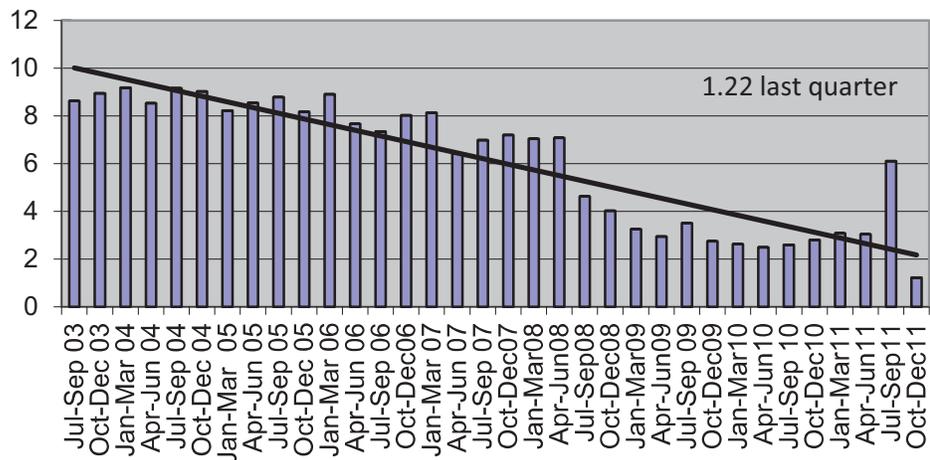
% Initial Specimens Collected at < 24 hours



% Initial Specimens Collected at 24-48 hours



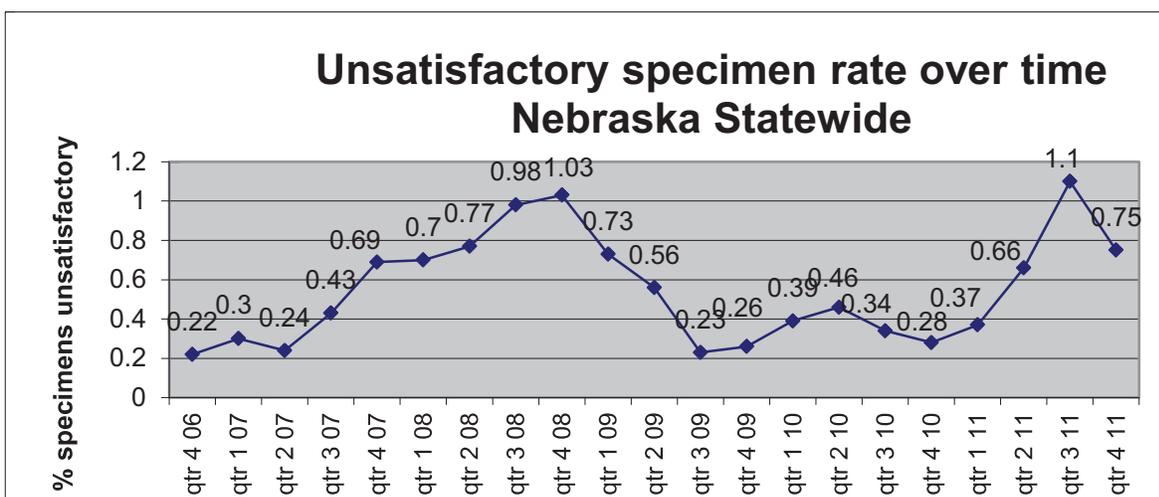
% Initial Specimens Collected on Day 3



Unsatisfactory Specimens for 2011

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.

The effort to reduce unsatisfactory specimens is valuable because they can be costly on many levels. Repeat screens must be done requiring 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. Maintaining low unsatisfactory specimen rates is a high priority goal of the Nebraska Newborn Screening Program. The peak in unsatisfactory rates is believed to be partially due to the increased number of specimens being collected overall related to the adoption of the NICU serial screening requirements.



Reasons specimens were declared unsatisfactory in 2011

	Number	Percent
Not soaked through to the back of the filter paper	77	37
Heavily applied, layered or double spotted	48	23
Serum or fluid mixed with specimen	22	11
Expired filter paper	15	7
Quantity not sufficient	13	6
Exposed to heat or humidity	12	6
Unclear patient identity or conflicting information on filter paper	6	3
Contaminated or diluted	5	2
Specimen older than 30 days when arrived at lab	3	1
Specimen was scratched or abraded	2	1
Inconsistent results on testing	1	1
Interfering substance present	1	1
Sample was wet when mailed	1	1
	206	100

**Drawn Early Specimens for 2011
(less than 24 hours of age)**



SUMMARY OF DRAWN EARLY DATA

January 1, 2011 – December 31, 2011

Month	Total Drawn Early	Pre-Transfer/ NICU Admit	Pre-Transfusion	Early Discharge	Reason Unknown*	Number Expired	Hospital Reporting Errors
January	32	14	2	1	15*	0	5
February	33	23	0	0	10*	0	4
March	71	63	0	1	7*	1**	9
April	78	69	0	0	9	0	4
May	56	44	0	0	12	3**	9
June	102	86		1	15	3**	8
New CLSI Guidelines Adopted July 2, 2011	Total Drawn Early	Drawn Early >2000 grams at birth	Drawn Early < 2000 grams at birth			Number Expired (not in total)	Hospital Reporting Errors
July	182	140	42			1	13
August	181	132	49			0	7
September	156	125	31			1	7
October	160	117	43			3	3
November	168	122	46			1	5
December	187	138	49			1	9

*Some of the unknowns may be related to early implementation of CLSI guidelines in neonatal intensive care units. If there is not a transfer between hospitals and the same physician is taking care of baby in NICU, it is not always possible to tell that the baby is in NICU.

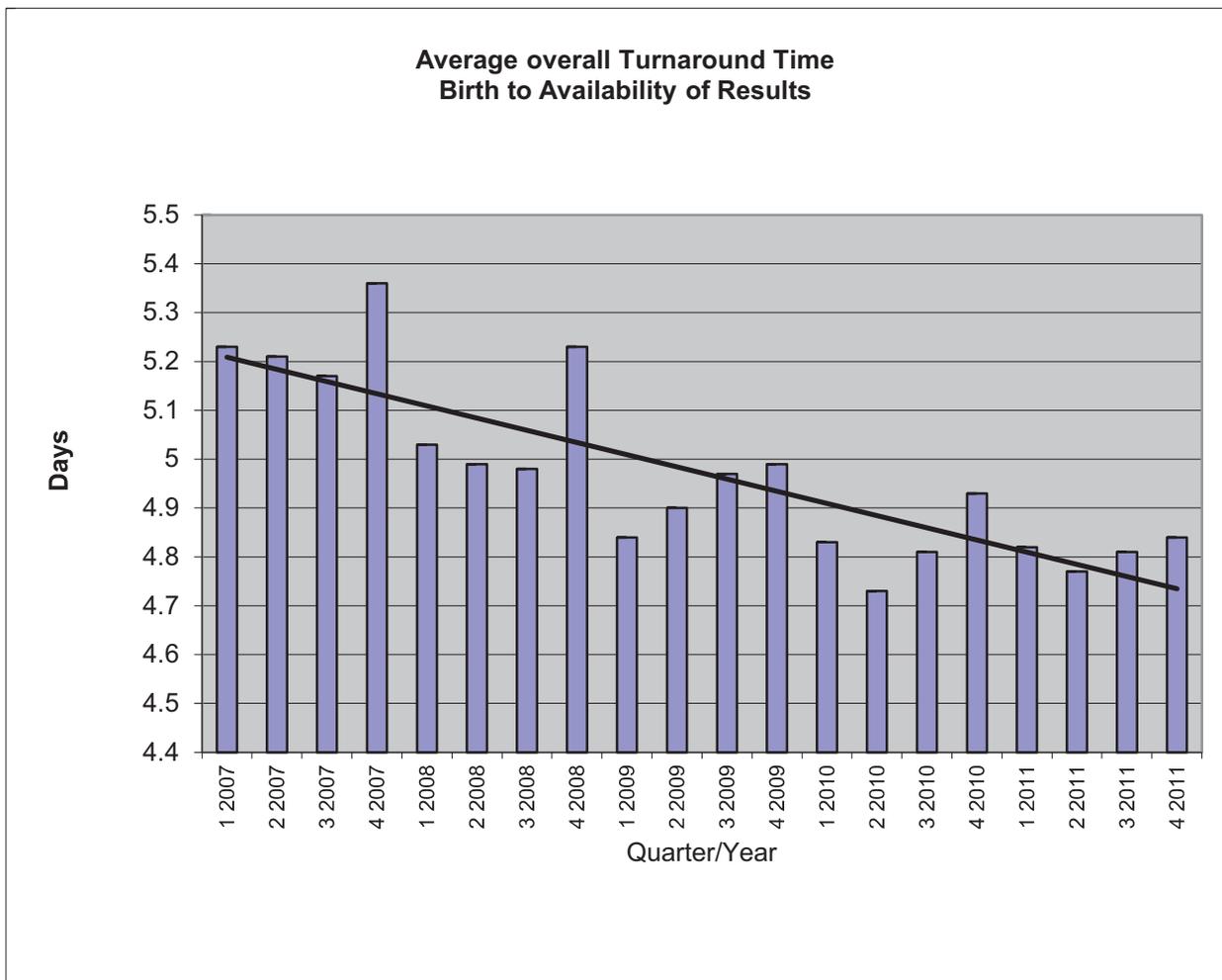
**Not added to total Drawn Early for Month.

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.

On the positive side, overall turnaround times continued to decline in the first half of 2011 thanks to in-lab efforts at PerkinElmer Genetics, and hospital personnel responding to the quality assurance reports when turnaround times for collection were above the benchmark/average of 1.5 days of age.

The Newborn Screening Advisory Committee reviews the quarterly results of average times from: birth to collection, collection to receipt in the lab, in-lab turnaround time, and overall turnaround time from birth to reporting out of results as in the following graphic in which the last quarter of 2011 ended up with average of 4.84 days.



LABORATORY 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 2011 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	26,029	62 (55 of these were inconclusive)	0.236%	3 (2 of these expired)	3 Partial deficiencies (1 not determined if partial or profound)
Congenital Adrenal Hyperplasia	26,029	37 (30 of these were inconclusive)	0.14%	2 (both expired)	1
Congenital Primary Hypothyroidism	26,029	443 (370 of these were inconclusive)	1.7%	8 (all expired)	4 + 3 congenital hypothyroidism and 1 hypothyroidism-prematurity
Cystic Fibrosis	26,029	290 (284 of these were inconclusive)	1.11%	7 (all expired)	6
Galactosemia	26,029	4	0.01%	0	1 Classical + 2 Duarte galactosemia
Isovaleric Acidemia	26,029	1	0.003%	0	1
PKU	26,029	9 (6 of these inconclusive)	0.03%	0	3 Classical PKU
Sickle Cell Disease & other clinically significant hemoglobinopathies (hgbs)	26,029	7	0.02%	0	7
All other abnormal hgbs (carriers/variants)	26,029	436	1.6%	161 no dx but 102 of these had confirmatory testing**	268 various carriers/trait
Transient Tyrosinemia	26,029	54	0.2%	0	5 (4 of these were treated)

* All but one baby lost to follow up expired before repeat or confirmatory testing could be completed.

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

Mean Averages of Laboratory Test Measures

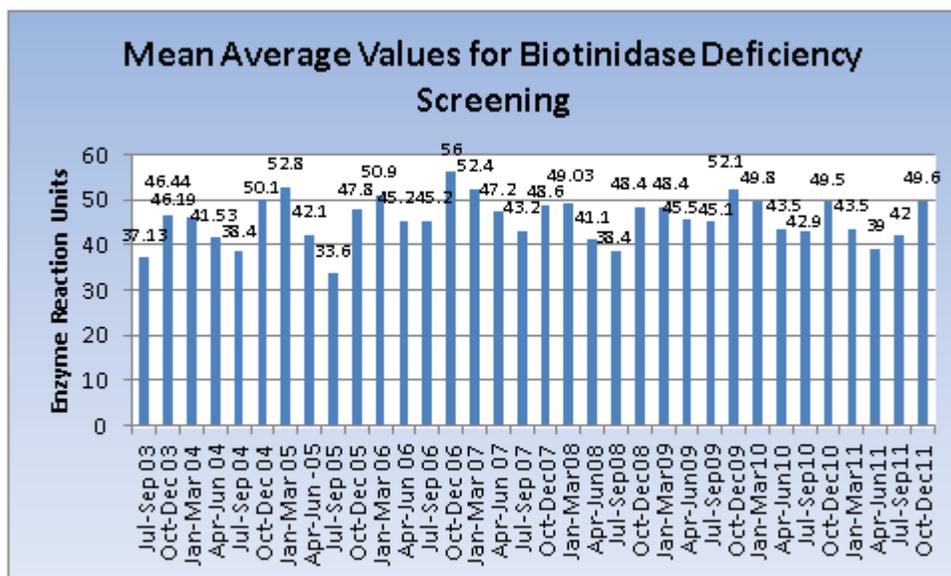
The program continues to provide lab testing data to the Newborn Screening Advisory Committee to monitor ongoing quality. The following tables depict the quarterly mean averages for biotinidase measures, 17-OHP for congenital adrenal hyperplasia, immunoreactive trypsinogen for CF, GALT, and total galactose used to screen for galactosemia. Access to data for mean averages for the amino acids and acylcarnitines used to screen for the fatty acid, amino acid and organic acid disorders are not available from the tandem mass spectrometry results from the screening laboratory. The T4 and TSH results are not included because some results were beyond the linearity of the assay prior to 2010 and would affect the accuracy of this data. 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.

Expected seasonal differences can be seen each summer when heat exposure may impact the mean average enzyme levels detected in screening for biotinidase deficiency.

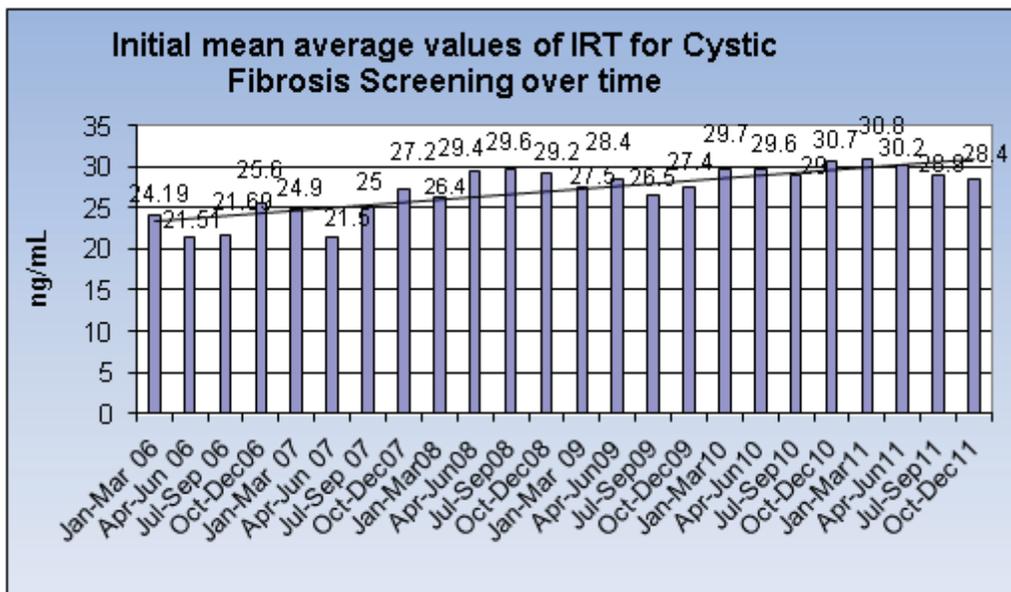
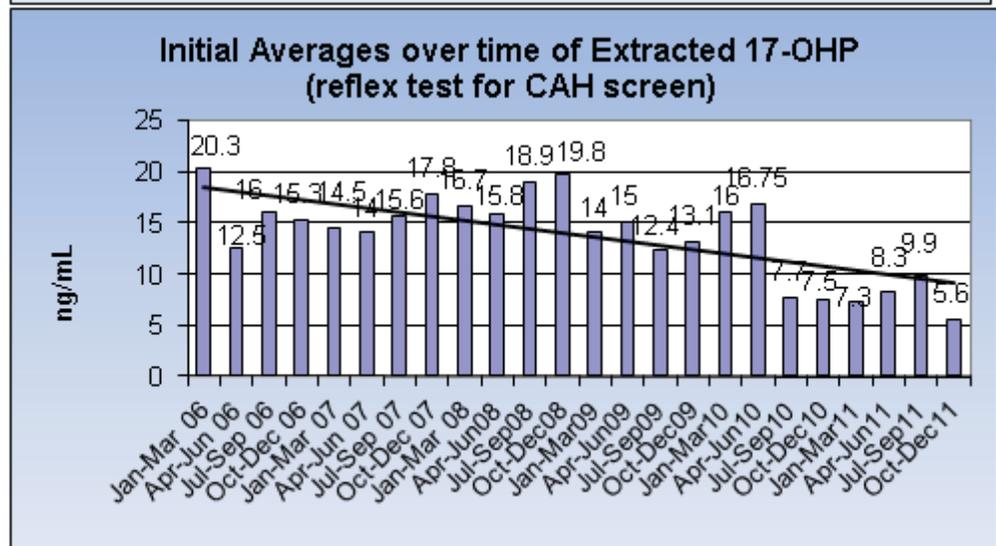
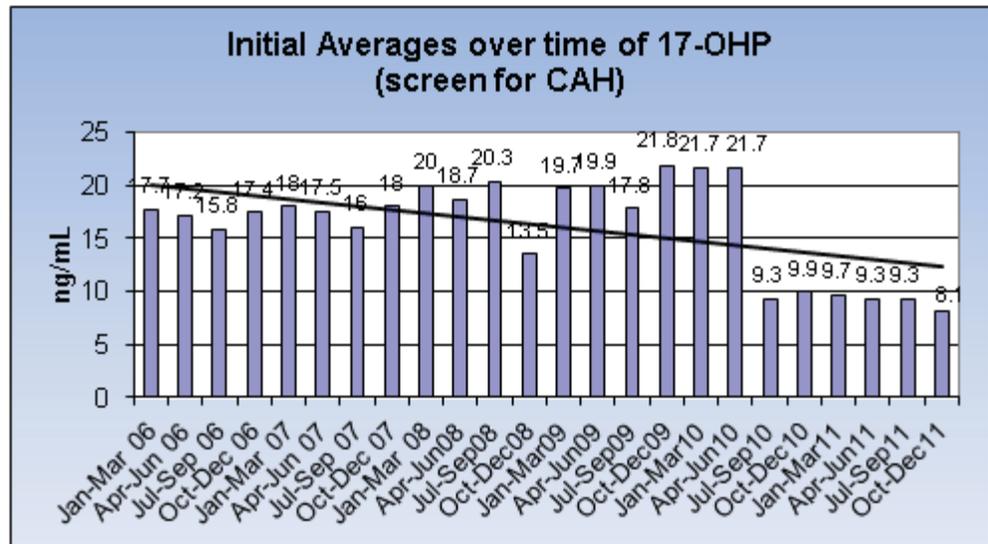
The Nebraska Newborn Screening Program sends a reminder each spring to hospital laboratories about specific practices to follow that will minimize the risk of specimens becoming heat denatured. This is intended to avoid the associated increase in the number of rejected specimens.

In 2011, 15 specimens were rejected and needed repeats to re-test enzyme assays used to screen for conditions such as biotinidase deficiency and galactosemia because the initial specimen had been exposed to heat/humidity. This was an increase from the prior 2 years.



Reflex testing of abnormal CAH screens using an extracted 17-OHP reduces the number of positive screens reported and needing confirmatory testing. The extracted 17-OHP is thought to minimize the effects of interfering substances.

Testing of < 24 hour specimens for 17-OHP was discontinued in October of 2011.



IRTs greater than 90 reflexed to test for ΔF508 the mutation most commonly associated with classical cystic fibrosis. (Those with one copy of the ΔF508 reflexed to testing for additional mutations on the Luminex 39 + mutation panel).

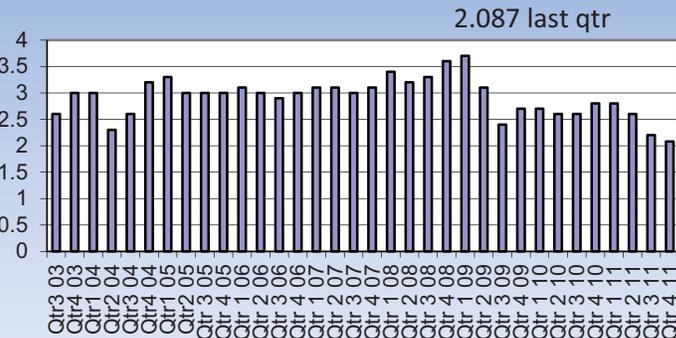
Mean Averages T4



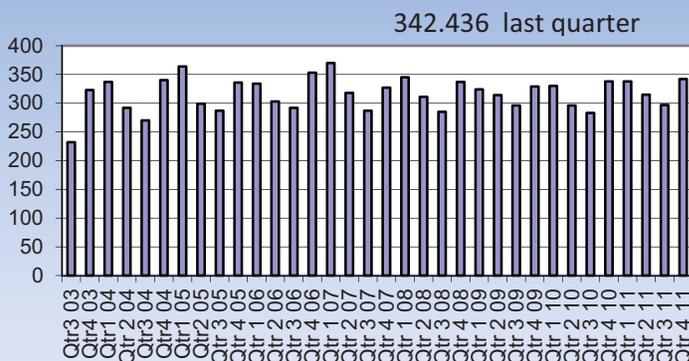
Mean Averages TSH



Total Galactose Values mg/dL by quarter



GALT Values uM by quarter



Screening for congenital primary hypothyroidism is completed for Nebraska births by testing thyroxine (T₄) on every baby, and those whose T₄s fall in the lowest 10th percentile of the run, reflex to test thyroid stimulating hormone (TSH). Babies with low T₄s and TSHs greater than 20 get reported at as presumptive positive.

Starting in October of 2011 specimens from babies collected at less than 24 hours of life are no longer tested for CPH. Repeat specimens are required at greater than 24 hours of life, and these are tested for CPH.

The galactosemia screen is highly sensitive. Both total galactose and GALT are measured. Elevated galactose in the presence of low enzyme activity is highly suggestive of galactosemia. In 2011 we identified one baby with classic galactosemia and two with Duarte galactosemia.

As is evident from Nebraska's historical data, the enzyme assays are subject to seasonal effects. Generally enzyme activity is lower in the hot summer months.

NEWBORN SCREENING DATA

	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
Total Births	25,515	26,067	26,443	26,349	26,898	27,107	27,094	27,199	26,243	26,094
Births Screened*	25,478 99.85%	26,008 99.77%	26,391	26,288	26,819	27,013	27,021	27,131	26,176	26,029
Total Births Lost to Follow up	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)	(94 not screened as expired @ < 48 hours)	1 (+ 73 not screened as expired @ < 48 hours)	3 (67 not screened as expired @ < 48 hrs, 1 discharge d w/o screen & out of state)	12 (10 of these expired @ < 48 hrs. of age)	65 (expired @ < 48 hours before screened)
Total Births pp**	456	415	499	503	537	511	553	1011	949	1646
Home Births	99	70	60	55	69	80	86	99	96	114
Home Births Screened	95	65	60	54	69	78	85	97	96	109
Home Births Lost to follow up	2 + (2 expired)	3 + (2 expired)	0	0 + (1 expired)	0	2 (both expired)	1 (expired)	2 (expired)	0	5 (3 of these expired)

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

** PP = Presumptive Positive. Includes all initial screen results requiring either a repeat dried blood spot or another confirmatory specimen and test. Nearly all MS/MS PP results require only repeat screens.

<i>Biotinidase Deficiency</i>	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
Presumptive Positive	3	4	34*	78	14	5	4	5	1	7
Inconclusive						10	25	17	17	55
Confirmed Negative	1	0	29	71	9	11	23	17	17	3
Confirmed Positive Profound	2	1	0	1	0	0	1	0	0	0
Confirmed Positive (Partial no tx)	0	0	0	0	0	0	0	0	0	0
Confirmed Positive (Partial tx)	0	3	6	5	4	4	3	5	1	5
Lost to follow up	0	0	0	1**	1**	0	2**	0	0	*3 (2 expired)

*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. Note: In 2011 one patient undetermined if Profound or Partial until several months later.

<i>Congenital Adrenal Hyperplasia</i>	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
Presumptive Positive	N/A	N/A	N/A	N/A	10	3	17	7	12	7
Inconclusive						18	22	25	26	30
Confirmed/repeated Negative	N/A	N/A	N/A	N/A	9	17	36	31	32	34
Confirmed Positive	N/A	N/A	N/A	N/A	1	1	1	1	2	1
Confirmatory or Repeat Lost to follow up	N/A	N/A	N/A	N/A	0	3*	2*	0	4*	2*

* Expired before repeat or confirmatory testing could be done.

<i>Congenital Primary Hypothyroidism</i>	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
Presumptive Positive	129	89	63	58	51	39	57	56	65	73
Inconclusive (drawn early but low T4/high TSH)						20	52	48	71	370
Confirmed Or repeated Negative	113	75	55	48	41	43	96	88	124	427
Confirmed Positive	15	11	8	9	10	16	12	15	11	8
Confirmatory or Repeat Lost to follow up	1*	3*	0	1*	0	3*	1	1*	1*	8*

*Lost to follow up as newborn expired.

Data for cystic fibrosis and hemoglobinopathies are presented in a different format because screening for CF is inherently more complex, and diagnosis for hemoglobinopathies can be more protracted and complex. Although the goal is to detect clinically affected newborns to initiate early treatment and prevent infant mortality and morbidity, the screening test can detect some carriers or people who have the trait for these conditions.

<i>Cystic Fibrosis:</i>		Year	2006	2007	2008	2009	2010	2011
Total Screened Positive			8	4	9	4	1	6
Of those:	Confirmed CF		4	8	9	1	3	6
	Confirmed Atypical CF		0	0	0	0	0	0
	CRMS (CF related metabolic syndrome)		0	0	0	0	1	0
Total Screened Inconclusive			62	54	53	50	122*	280
Of those:	Confirmed CF		3	2	5	1	0	0
	Confirmed Atypical CF		0	2	9	0	0	0
	CF Related Metabolic Syndrome					0	4	0
	Confirmed Carriers		12	10	6	10	9	18
	Found to be within normal limits on repeat		35	46	30	95	32	248
	Expired before confirmation could be done		4	1	6	9	2	6
	Lost to follow up		0	0	0	0	1	2
	Pending		0	1	0	2	0	0
Total with Meconium Ileus or Bowel Obstruction			4	13	1	8	3	4
Of those:	Confirmed CF		5	1	1	2	2	0
	Found to be within normal limits		7	3	0	1	6	3
	Pending diagnosis		1	0	0	0	0	1

* Includes 3 that were within normal limits upon follow-up with sweat chlorides.

<i>Galactosemia</i>	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
Presumptive Positive	5	3	9	1	8	0	0	2	1	4
Inconclusive repeat rec'd	n/a	n/a	n/a	n/a	n/a	9	9	10	7	0
Confirmed / repeated Negative	5	0	6	1	8	8	9	12	7	1
Confirmed Positive (Classical)	0	1	0	0	0	0	0	0	0	1
Confirmed Positive, Duarte (not treated)	0	1	0	0	0	0	0	0	0	0
Confirmed Positive, Duarte (treated)	0	1	3	0	0	1	0	0	1	2

Hemoglobinopathy Follow-up Changes:

Since 2006, follow-up procedures included sending a reminder letter to the baby's physician before the 6 month checkup when the initial confirmatory report indicated a possible alpha, beta or gamma chain variant or combination in the heterozygous state. These typically require additional blood work to diagnose, which previously was not usually reported back to the program. Often these were hemoglobin patterns that had Bart's present on the initial screen and the concern was a possible alpha Thalassemia. This has resulted in a significant increase of diagnosed and closed cases. Ultimately the goal is to provide families with better information about their child's hemoglobinopathy.

Abbreviation Key (Likely diagnosis associated with screening results)

FS: Sickle Cell Disease	FAS: Sickle Cell Trait
FC: Hemoglobin C Disease	FAC: Hemoglobin C Trait
FSC: Sickle Hemoglobin C Disease	FAD: Hemoglobin D Trait
FE: Hemoglobin E Disease	FAE: Hemoglobin E Trait
FSA: Sickle Beta Thalassemia	FAV: Hemoglobin Trait - unknown variant
HPFH: Hereditary Persistence Fetal Hemoglobin	FA+Barts: Possible alpha Thalassemia

Clinically Significant Hemoglobinopathies Confirmed Positive:

	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
FS	4	5		1	3		3	1	5	2
FC			1	1		1*		1	2	1
FSC	2		1	1	2	1	1	5	1	3
FE					1			1		1
Sickle Beta Thal						1		3	1 C-beta thal	
Alpha Thal Major										
Beta Thal Major						1		1		
HPFH				1						
FAE + possible Beta Thal					6					
FAS + possible Beta Thal					11					
FAS + Alpha Thal					9					
FAC + Alpha Thal					3					

Dx. = Sickle Hemoglobin C Disease or Hemoglobin C beta Thalassemia

Other Hemoglobinopathies Confirmed Positive in 2011:

135 Sickle Cell Trait	37 Hgb. C Trait	10 Hgb. E Trait
9 Hgb. D Trait	53 Trait + other	3 Alpha Thal silent carrier
6 Alpha Thal Trait	10 miscellaneous traits	

155 Confirmatory diagnoses unknown. (Confirmatory testing done for 104 of those, but no final diagnosis. None known to be clinically significant.)

MCAD *	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
Screened Positive	3*	3	5	10	5	0	4	3	2	2
Screened inconclusive (repeat only)**	N/A	N/A	N/A	N/A	N/A	2	2	8	0	0
Confirmed Negative or Repeated normal	2	3	1	7	5	2	2	8	1	2
Confirmed Positive	1	0	4	3	0	0	4	3	1	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.

**Inconclusive screen: Abnormal screen result requiring only a repeat screen, not confirmatory testing.

Phenylketonuria (PKU)	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
Presumptive Positive	3	7*	7	3	6	0	0	4	4	9
Screened Inconclusive (repeat only)**	N/A	N/A	N/A	N/A	N/A	2	0	1	0	0
Confirmed Negative	1	1	1	1	1	2	0	1	0	6
Confirmed Positive Classical PKU	1	2	1	2	0	0	0	3	2	3
Confirmed Positive Hyperphe	1	3	5 (3 of these tx'd)	0	5 (4 of these tx'd)	0	0	1	2	0

2003: One for whom confirmatory testing was not done as the baby expired.

**Inconclusive screen: Abnormal screen result requiring only repeat screen, not confirmatory testing.

Tandem Mass Spectrometry Screening Results (MS/MS)

Initial findings	# abnormal On screen	# confirmed negative	# pending or lost to follow up*	# confirmed positive
Methionine	149	146	3 expired	0
Several Amino Acids**	99	93	5 expired	0
A generalized elevation of short-chain & medium-chain acylcarnitines	21	19	2 expired	0
Propionylcarnitine (C3)	5	5	0	0
C3 & C3/C2 & C3/C16	27	25	1	0
Alanine	1	1	0	0
Arginine	1	1	0	0
C4 Butyrylcarnitine	4	4	0	0
C4OH 3-hydroxybutyrylcarnitine	1	1	0	0
Formiminoglutamic Acid	4	3	1 expired	0
Glycine	1	1	0	0
C5 Isovalerylcarnitine and other indices such as C4 Butyrylcarnitine	1	0	0	1 IVA
Methionine /Phenylalanine	1	0	1 pending	0
C14 Myristoylcarnitine	1	1	0	0
C8 Octanoylcarnitine	2	2	0	0
Phenylalanine & Phe/Tyrosine ratio	9	6	0	3 PKU
Tetradecenoylcarnitine (C14:1)	1	1	0	0
Tetradecenoylcarnitine (C14:1) to C16 & other long chain acylcarnitines	3	3	0	0
Tyrosine	54	49		5 Transient tyrosinemia (4 responded to tx)
2011 Totals (NE Infants)	384	361	3 pending 11 expired	9

*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.

**Multiple amino acid elevations generally indicate baby is on hyper alimentation, not presumptive positive for a condition.

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.

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.
3 Biotinidase Deficiency	15	12-17
1 Congenital Adrenal Hyperplasia	12	12
8 Congenital Primary Hypothyroidism (includes CH and CH-prematurity)	31	10-71
Cystic Fibrosis	10	3-18
3 Galactosemia (includes 2 Duarte)	6	4-8
1 Isovaleric Acidemia	5	5
3 Phenylketonuria	6	4-8
1 Hgb. C Disease	47	47
2 Sickle Cell disease	17	13-21
5 Transient Tyrosinemia (4 treated)	13	6-25
3 Hgb SC Disease	25	9-36
1 Hgb E Disease	17	17

2011 Outcome Data for Newborn Screening

As part of the oversight of state contracts with the metabolic clinic for the ordering and distribution of metabolic formula and with the CF Center for assistance with follow-up and consultation, data is collected to look at different outcomes.

Metabolic patients

The measures looked at for Nebraska's population of patients with metabolic conditions covered by Nebraska's Newborn Screening Panel are:

- The percent of time blood phenylalanine (or other appropriate metabolite) levels are within the optimal therapeutic range for patients ages 0-6, > 6-12, pregnant women and all other groups.
- The percent of patients ages 0-12 who are meeting all developmental milestones or all but one or two developmental milestones as assessed/determined by the pediatric metabolic specialist or who are receiving any special education services.
- The percent of patients greater than 18 years of age, who have graduated high school, attained a bachelor's degree or attained a master's level degree or higher.

Phenylalanine levels in 2011:

Birth to 6 year old group:

-83% had average phe levels in the optimal therapeutic range

6-12 year old group:

-80% had average phe levels in the optimal therapeutic range

13-18 year old group:

-56% had average phe levels in the optimal therapeutic range

19 and older group:

-54% had average phe levels in the optimal therapeutic range

Pregnancies:

-67% had average phe levels in the optimal therapeutic range

Developmental Milestones in 2011:

Birth to 6 years:

-100% meeting developmental milestones

6-12 year old group:

-100% meeting developmental milestones

Education level attainment in 2011:

-96.8% of patients > 18 years had a high school diploma

-6.25% had earned Associates degrees

-37.5% had earned Bachelor's degrees

-9.4% were in college working towards a Bachelor's degree

-15.6% had earned Master's degrees

-3% are currently attending medical school

State high school graduation rates compare as follows: In 2007-2008, the most recent data available from the National Center for Education Statistics, Wisconsin had the highest graduation rate at 89%, with 17 other states > 80% including Nebraska.

This data should eliminate any doubt about the effectiveness of newborn screening for early identification and treatment of metabolic conditions, and long term follow-up and management.

CF patient satisfaction summary

In 2011, 35 patients were referred to and received evaluations. Eighteen of these patients completed patient satisfaction surveys. Results were very positive and suggest the newborn screening system and diagnostic/follow-up care received from the Accredited Cystic Fibrosis Center in Nebraska is working hard to meet the needs of families.

100% of respondents answered “yes” to:

- “I understand the meaning of my baby’s test results.”
- “The questions I had about cystic fibrosis or cystic fibrosis carrier status were answered satisfactorily.”

Of parent respondents whose child’s outcome was non-CF:

- 100% answered “Yes” to: “The anxiety I may have felt about the possibility of my child having CF or about he/she’s carrier status was relieved.”
- 100% answered “No” when asked if they needed further information or help.

Of parent respondents whose child’s outcome was CF, 100% responded:

- Yes, when asked if the information received about their child’s diagnosis made them feel comfortable that he/she will receive appropriate care.”
- Yes, when asked if they needed help with resources, information for others in the family, and additional resources for support.

99% of respondents responded “excellent” and 1% responded “good” about their overall satisfaction of the screening and follow-up process.



NEBRASKA EARLY HEARING DETECTION AND INTERVENTION ANNUAL REPORT - 2011

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 required 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 required that regulations be promulgated to mandate newborn hearing screening if less than 95% of newborns in the state received 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. Picture 1 shows an infant receiving an OAE hearing screening.



Picture 1 - 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 2011

Birthing Facility Screening Programs

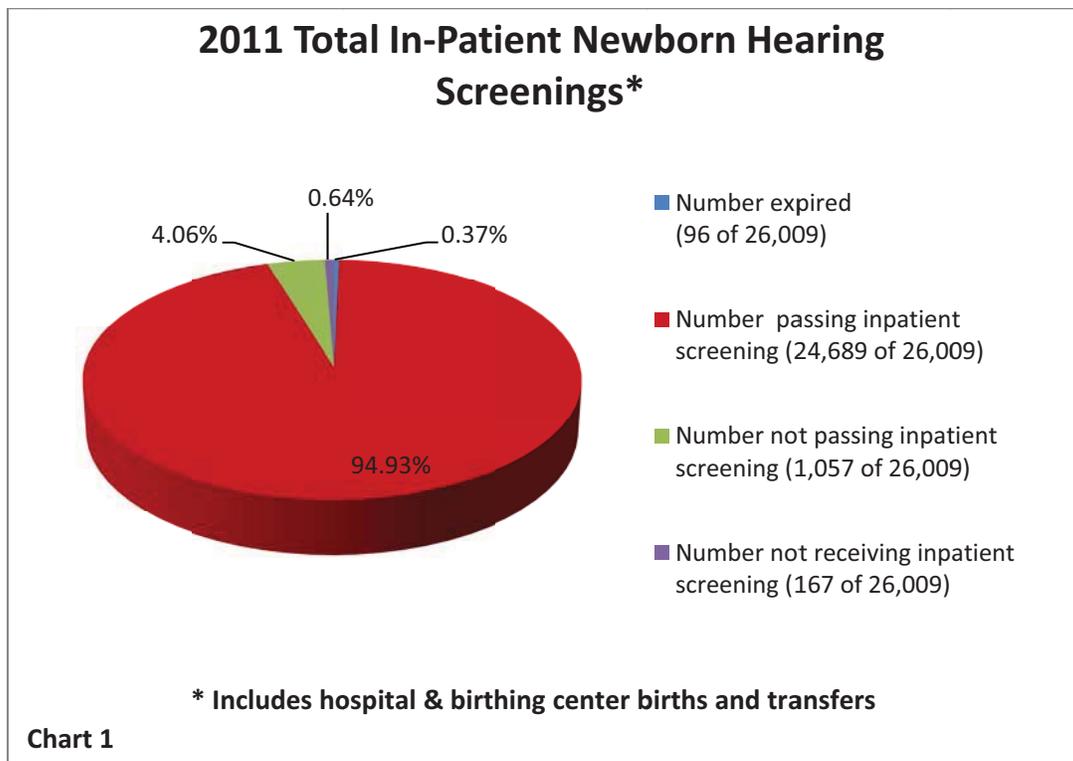
Since 2003, 100% of the birthing facilities in Nebraska have been conducting hearing screenings, consistent with the Neb. Rev. Stat. §71-4742 requirement that a hearing screening test be included as part of the standard of care for newborns. In 2011 there were 57 birthing facilities using OAE, ABR or both screening methods and one hospital with a visiting audiologist who performed post-discharge screening.

Hearing Screening at Birthing Facilities and Birthing Centers

In 2011 hearing screenings were reported on 25,746 newborns, or 98.98% of the 26,009 hospital births, prior to discharge from the 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.

Chart 1 (next page) shows the percentage passing the inpatient hearing screening, the percentage not passing the inpatient screening, the percentage not screened prior to discharge from the hospital and the percentage of the infants expiring prior to screening. In Nebraska, 24,689 (94.92%) newborns passed the inpatient hearing screening for 2011.

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 99.37% of parents about newborn hearing screening, hearing loss, and normal speech and language development in 2011. 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. Parent education material is sent to the parents of the babies who were not born in a hospital.

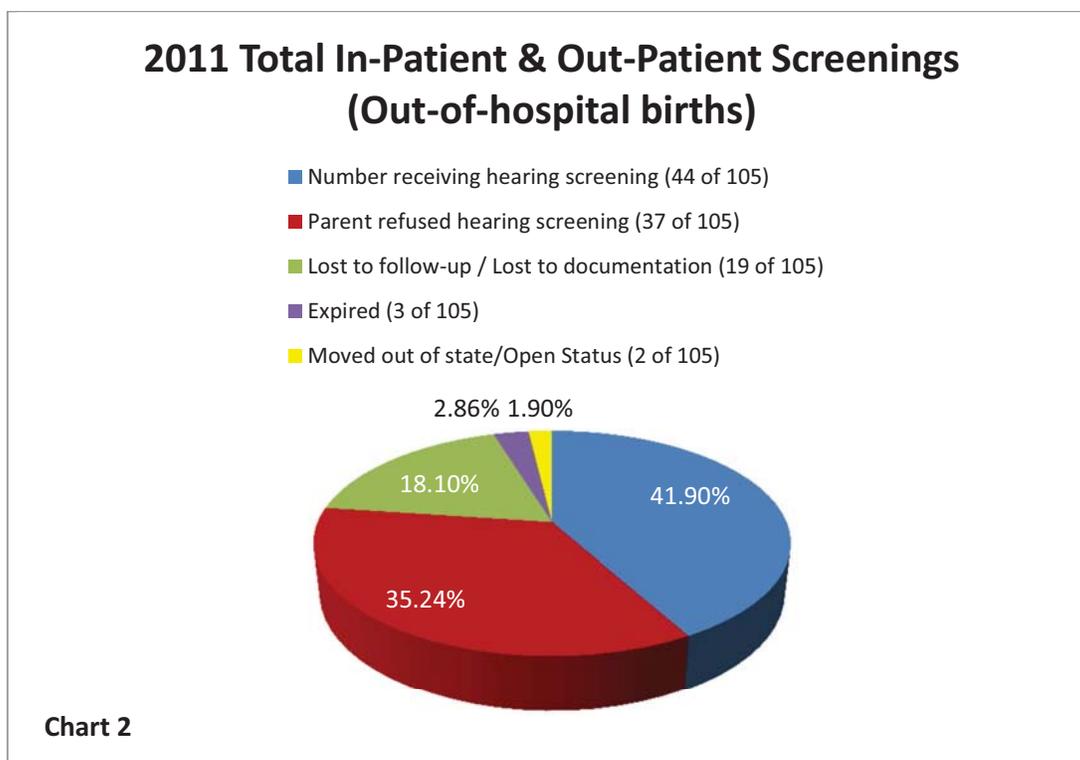
Monitoring, Intervention, and Follow-up Care

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 2011, a total of 1,403 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. Fifty-four (3.8%) of these

children are in the category of lost to follow-up/lost to documentation. A total of 55 children did not receive an outpatient screening or diagnostic evaluation due to the parent(s) refusing further action. Other cases where the hearing status has not been established include: 13 families not residing in Nebraska and 82 cases of further testing needed due to middle-ear problems or other medical conditions.

Non-Birthing Facility Births (Home Births)

There were 105 births that occurred outside of a birthing facility in 2011. Twenty-two of these births were unplanned home births where the infant was transferred within 24 hours to a birthing hospital. Of these 105 births, 44 (41.90%) received an inpatient or outpatient hearing screening. Thirty five percent (35.24%) did not receive a screening since the parent(s) refused to have the infant screened. See Chart 2 for the number and percentages of in-patient and out-patient screenings for out-of-hospital births.



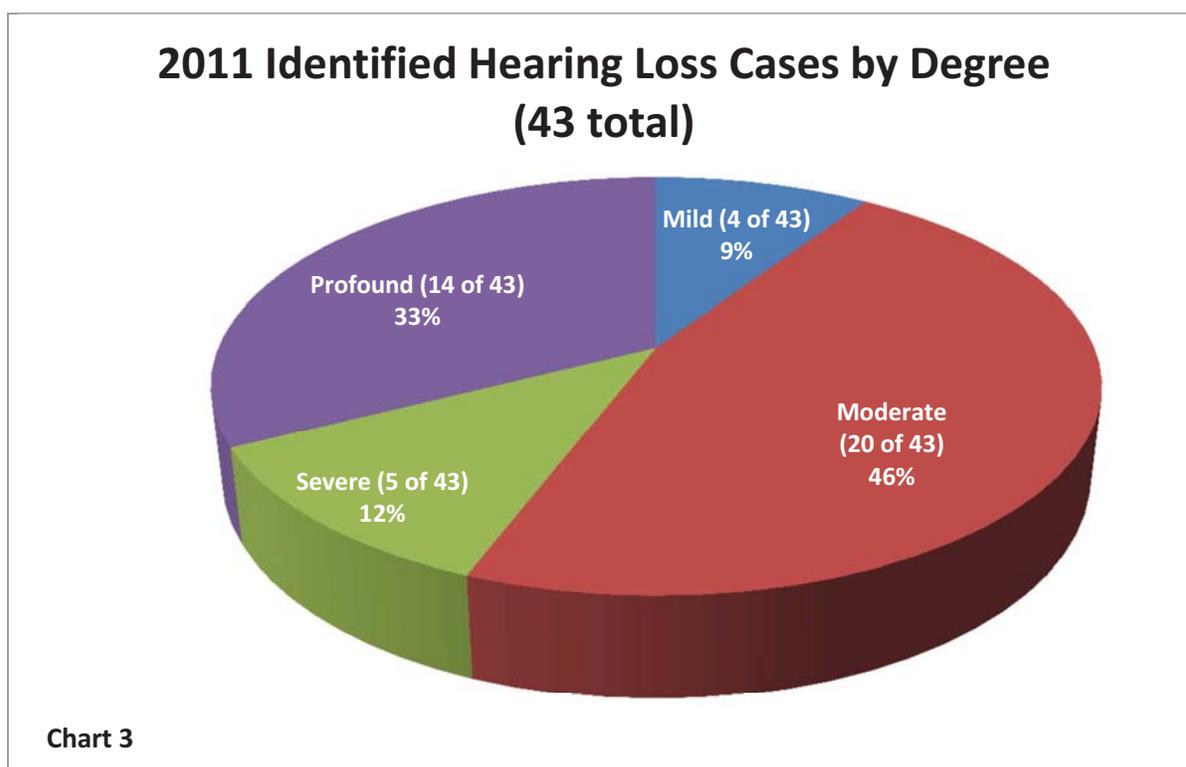
Confirmatory Testing/Audiologic Data Reported for 2011

The Advisory Committee for the NE-EHDI Program identified the initial level of follow-up hearing tests for many newborns 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 114 infants received a complete audiologic evaluation in 2011.

Degree of Hearing Loss

Neb. Rev. Stat. §71-4739 requires confirmatory testing facilities to report newborns and infants who are shown to have hearing loss based upon the follow-up hearing test.

According to the individual results reported by audiologists to the NE-EHDI Program, 43 infants born in 2011 have been diagnosed with hearing loss in one or both ears. Thirty-four of the infants were identified with bilateral hearing loss and nine infants were identified with unilateral hearing loss. The incidence of permanent congenital hearing loss in Nebraska for babies screened in 2011 is 1.7 per thousand births. Chart 3 (below) shows the severity of hearing loss for the 43 infants identified with hearing loss.



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 98% of infants received an inpatient screening within one month of age. For the newborns who were recommended for an audiologic diagnosis, 59% 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 32 (74.4%), out of the 43 infants born in 2011 and identified with a hearing loss, were referred to EDN within six months. So, the "1-3-6" national guideline for timeliness was not met for almost one-fourth of this group of 43 infants. Four (9.3%) children were referred to EDN after six months and seven (16.3%) infants were not referred directly to EDN by the audiologist or medical home.

ACTIVITIES – 2011

Funding

The NE-EHDI Program received 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. Also, in 2011, the NE-EHDI Program was awarded a contract with the CDC for a pilot program called iEHDI which is discussed in further detail below.

Advisory Committee

The NE-EHDI Program was developed based on the requirements identified in the Nebraska Infant Hearing Act of 2000 and the recommendations by the NE-EHDI Program Advisory Committee. Specific tasks to be accomplished by the Advisory Committee are as follows: 1) to review and, as necessary, revise the existing protocols to incorporate the electronic data system, 2) to develop new reporting, tracking, and follow-up protocols to effectively link the NE-EHDI Program and the early intervention systems, 3) to increase the program's responsiveness to the expanding cultural and linguistic communities in the state, 4) to support the development of an effective professional development system, and 5) to guide the long-term planning and evaluation of the NE-EHDI Program system in the state. The Advisory Committee of the NE-EHDI Program consists of no more than 20 voting members representing medical, audiology, parents, public health, family support, and education stakeholders. The Advisory Committee met four times during 2011.

Also in 2011, the Advisory Committee approved a charter for the committee and revised its mission statement to read: *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.* Members also elected a chair and vice-chair.

Projects

Hearing Screening Equipment for Birthing Facilities

Opportunities to contract for partial funding of new hearing screening equipment were offered to rural audiology facilities in Nebraska and to early childhood facilities. New equipment should reduce the number of babies who refer due to the use of aging or

inappropriate hearing screening equipment. A total of nine contracts were awarded in 2011.

Electronic Data System

The NE-EHDI Program was awarded another five years of CDC funds effective July 1st of 2011. A portion of those funds will support another enhancement to the Nebraska electronic data system for the NE-EHDI program.

This next enhancement will focus mainly on 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. Work has been started on this enhancement and is expected to be completed in 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.

iEHDI CDC Contract

The Centers for Disease Control (CDC) released an announcement soliciting proposals from EHDI Programs to participate in a two-year pilot project entitled iEHDI. 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 a limited data set is not directly identifiable and excludes direct identifiers, such as name of the individual or of relatives, employers, or household members of the individual. Nebraska and one other state were awarded a two-year contract in 2011.

Roots and Wings Parent Weekend

The fifth *Roots and Wings* parent weekend was held October 21 – 23, 2011 in Kearney, Nebraska. 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. Survey results on the sessions and activities were shared in a presentation to the NE-EHDI Program Advisory Committee.

This year the program also enlisted the participation of “host families” to provide interaction and to answer questions during the weekend’s informal times (evenings/breaks). A total of 36 parents or guardians and 34 children attended.

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 NE-EHDI Program provided funds to administer the NCHALB and to purchase loaner hearing aids. Twenty-two children were fitted with 40 hearing aids (18 children had two aids and four had one) in 2011 and 24 children returned their hearing aids (40) (16 children had two aids and eight had one during 2011).

With the support and agreement from the Nebraska Children’s Hearing Aid Loaner Bank and the NAEYC, the purchasing of children’s hearing aid loaners and the repair of children’s hearing aids was moved to the Nebraska Children’s Hearing Aid Bank/HearU Nebraska. This allows the NE-EDHI Program to re-allocate funds used for administrative costs to increase the number of hearing aids purchased and repair of additional infant and children’s hearing aids.

OAE Screeners

The NE-EHDI Program purchased two OAE hearing screeners. One unit is available for loan to birthing facilities to continue to provide an initial hearing screening on babies before they leave the facility when equipment is in need of repair. The second one is on loan to *Clinic with a Heart* which is a volunteer organization sponsored by seven different churches and two hospitals that serve low-income and non-English speaking individuals with health care needs. A volunteer audiologist performs the hearing screens.

Better Hearing and Speech Month

Initiated by the Community Health Educator II as part of May is *Better Hearing and Speech Month*, the NE-EHDI Program produced and distributed educational packets for third year resident doctors and second year nursing students. These packets contained educational materials regarding the hearing screening process, EHDI purpose, guidelines to follow with hearing services, and resources for families. Over 500 packets were delivered by hand, mail, or email.

Amish Births

An effort to reach out to the Amish community in Nebraska, using a new protocol for follow-up on out-of-hospital births was implemented. Through a community outreach initiative, contact was made with Amish families who were educated about the hearing screening process and the necessity to have screenings conducted on their children. Some of the families expressed interest in hearing screening. One issue faced by families in the Amish community was lack of insurance and the resources to pay for a hearing screening at the local community medical center and the lack of electricity in the homes. In partnership with the Early Development Network an audiologist agreed to visit this Amish community in Nebraska to conduct the hearing screenings at no charge. Efforts continue with other families and this relationship is at least now in place, building capacity for this community.

E-Fax and Electronic Communication; Scanning Documents to Records

Improvements to the NE-EHDI Program electronic communication and record keeping systems were implemented in 2011. Previously, outgoing and incoming correspondence with Primary Health Care Providers (PHCP), other medical providers, and parents was printed, faxed, and stored in hard copy. In an effort to improve efficiency and conserve resources, all outgoing correspondence to PHCPs is now generated in electronic format, faxed electronically via E-fax, and stored with the child's electronic record in the Electronic Registration System (ERS). Any incoming correspondence is scanned and the electronic document is attached to the child's ERS record.

Summary

- All the current birthing hospitals in Nebraska were conducting newborn hearing screening in 2011. All but one had conducted the hearing screenings prior to discharge from the hospital or birthing center.
- In 2011, birthing hospitals reported screening the hearing of almost 99% of newborns during birth admission or prior to discharge for those babies who were transferred to another hospital.
- The overall "refer" rate during 2011 for initial hearing screening during birth admission was 4.1%.
- In 2011, audiologic evaluations were initiated within three months of age for 59% of newborns when this type of evaluation was recommended.
- There were 204 babies born in 2011 whose hearing status was not objectively established, not including the 96 who expired before receiving or completing a hearing screening.
- The incidence of Permanent Congenital Hearing Loss identified and reported to the NE-EHDI Program (1.7 per thousand screened in 2011) is within the anticipated range of one to three per thousand.
- Almost 75% of the infants with hearing loss were referred to the Early Development Network and special education services within six months of birth.

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 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

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

Metabolic foods program, translation & distribution of patient education materials

WEB PAGE: <http://dhhs.ne.gov/publichealth/Pages/nsp.aspx>

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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

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

Jessie Shives, Community Health Educator, NE-EHDI program 402-471-6746

Follow-up, complex diagnostics, special projects

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

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Advisory Committee scheduling and support

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This report was prepared and published by the Nebraska Department of Health and Human Services, Newborn Screening Program, P.O. Box 95026, Lincoln, NE 68509-5026. Funding for this report was made possible through the Maternal and Child Health, Title V Block Grant.

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