NEWBORN SCREENING IN NEBRASKA

Newborn Screening for

Metabolic and Inherited Disorders

AND

Early Hearing Detection & Intervention

2009 Annual Report

Department of Health & Human Services

DHHS

NEBRASKA

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NEBRASKA EARLY HEARING DETECTION AND INTERVENTION ANNUAL REPORT - 2009

NEWBORN SCREENING FOR INBORN ERRORS OF METABOLISM AND INHERITED DISORDERS

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

Newborn Screening is a system involving many elements including:

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

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

In 2009, newborn screening efforts resulted in successfully identifying and treating 55 newborns affected with conditions in time to prevent problems associated with them:

- 5 babies with partial (treated) biotinidase deficiency
- 1 baby with congenital adrenal hyperplasia
- 14 babies with congenital primary hypothyroidism + 1 with congenital hypothyroidism
- 8 babies with cystic fibrosis + 1 with non-classical CF
- 12 babies with hemoglobinopathies (1 sickle cell disease, 5 SC-disease, 3 sickle beta thalassemia, 1 beta thalassemia major, 1 C-disease, and 1 E-disease)
- 3 babies with MCAD (medium chain acyl-coA dehydrogenase deficiency)
- 3 babies with phenyketonuria + 1 hyperphenylalaninemia
- 1 baby with mild 3-MCC (3-methylcrotonyl carboxylase deficiency)
- 1 with VLCAD (very long chain acyl-co-A dehydrogenase deficiency)
- 3 with SCAD (short chain acyl-co-A Dehydrogenase deficiency)
- 1 hypertyrosinemia of infancy (treated)

The incidence rate of conditions in Nebraska based on the screened conditions identified from 2006 - 2009 and number of births screened those three years: 1:583 births

WHAT IS NEWBORN SCREENING?

Newborn screening programs have been around for over four decades in all 50 states and in several countries. The compulsory screening panel varies slightly from state to state but the overall goal is the same: prevent or minimize the serious effects of the conditions screened. In 2009 Nebraska's required screening panel included 28 metabolic, endocrine, hematologic and other conditions.

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 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 CF Center is always recommended.

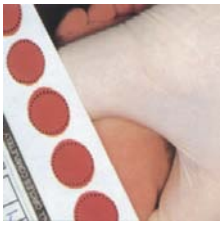
Conditions included in Nebraska's required blood-spot screening panel in 2009 were:

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
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
Isovaleric Acidemia

HOW THE NEWBORN SCREENING PROCESS WORKS

1: TESTING

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



Specimen shipped overnight to newborn screening laboratory, PerkinElmer

Specimen data entered into data system

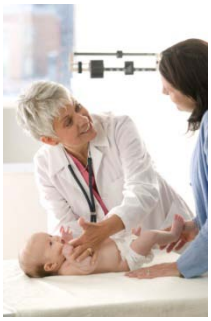
Specimen tested for multiple conditions

2: FOLLOW UP

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

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

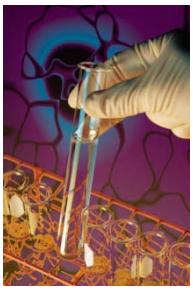
Parent's bring baby back in for evaluation and more testing



3: DIAGNOSIS/ INTERVENTION

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

Repeat or confirmatory testing occurs



Parent education on signs/symptoms to watch for

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

4: TREATMENT & MANAGEMENT

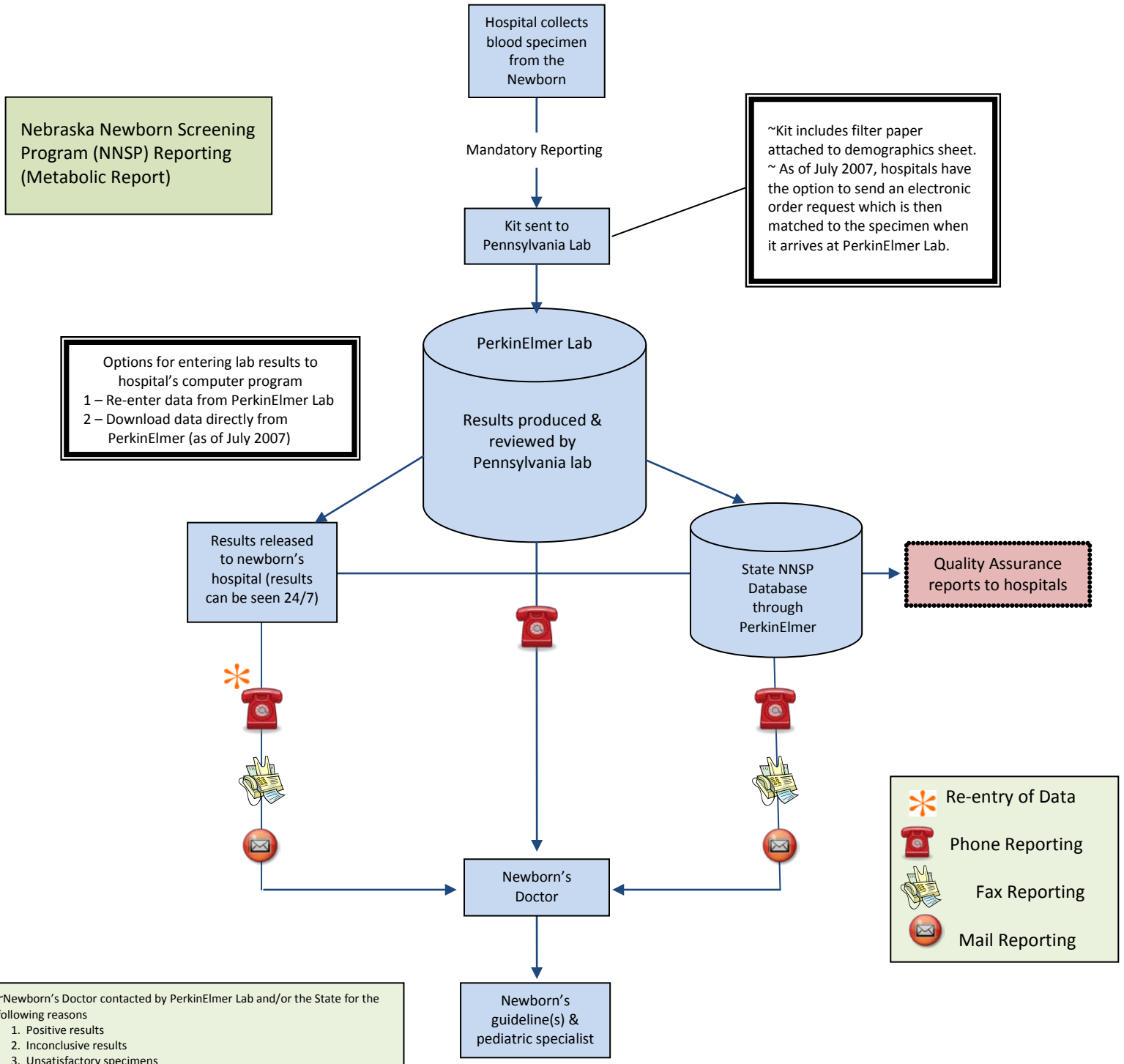
Once diagnosis is made, treatment begins. (For some life threatening conditions, treatment may occur prior to diagnosis- on recommendation of specialist.

Parents receive treatment guidelines / education.

Team Support services as appropriate, e.g.:

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

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



~Newborn's Doctor contacted by PerkinElmer Lab and/or the State for the following reasons
1. Positive results
2. Inconclusive results
3. Unsatisfactory specimens
4. Specimens drawn too early

~Reports from the State back to submitting hospitals:
1. Quarterly Quality Assurance Reports
   - Individual hospital's data compared to state data
2. Specific variable reports i.e.:
   -Turnaround time for lab results
   -Rates for unsatisfactory specimens

~For clarification, the data are located in the PerkinElmer Database and the state accesses it via a secure internet connection. Hospital (submitters) may also access their specimen's data this way. The state has some ability to access the data, but it is primarily entered, edited and maintained by the Pennsylvania lab. Also, the follow-up tracking reports and QA Reports are customized by the state.

System Overview

In 2009, 60 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.

Ongoing consultation with the laboratory, metabolic specialists Richard Lutz, M.D., William Rizzo M.D., and Jill Skrabal, R.D., Kathryn Heldt, R.D., and Rose Kreikemeier, RN, the Cystic Fibrosis Center Director John Colombo, M.D. and Dee Aquazzino, pediatric endocrinologist Kevin Corley, MD, and pediatric hematologist James Harper, M.D. ensured expert advice and assistance was available as needed throughout the year.

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

Treatment services received 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 2009 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.

- ❖ Local presentations by the program manager included a newborn screening update for the Physician Seminar Day at St. Elizabeth Regional Medical Center in Lincoln, an update to the Maternal and Child Health Committee of the Nebraska Medical Association, and at the 2009 Nebraska Public Health Conference.

- ❖ Internal staff development efforts included the Program Manager and follow-up staff attending the Association of Public Health Laboratory's National NBS & Genetics Symposium in San Antonio. Julie Luedtke presented on the status of the Clinical and Laboratory Standards Institute's "Guidelines for Newborn Screening for Premature, Sick and Low Birth weight Infants", and also co-chaired the committee. She also presented a poster on "Policy Analysis of Dried Blood Spot Testing for CMV and Genetic Causes of Hearing Loss" a joint project of the NBS and EHDI programs.

Policy

- Newborn Screening Program staff Krystal Baumert and Karen Eveans continued to serve on the Newborn Screening Committee of the Heartland Newborn Screening and Genetics Collaborative.

- The program manager continued to serve on: the APHL's Newborn Screening & Genetics Committee, the Heartland Region's Advisory Council, and the Newborn Screening workgroup, and the National Coordinating Centers Long Term Follow-up Work Group.

- The Newborn Screening Advisory Committee continued its quarterly review of quality assurance data of preanalytical (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 proposed changes from the newborn screening laboratory relative to unsatisfactory specimens due to overspotting, expanded capacity of the lab to screen for Tyrosinemia Type I through development of the assay for succinylacetone, and issues associated with reagent shortages.

- The Newborn Screening Advisory Committee (NBSAC) undertook evaluation of proposed regulations recommending revisions to sections addressing:
    - 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 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, and
    - clean-up language and clarifying definitions.

- The NBSAC in collaboration with the Advisory Committee for the EHDI program continued its evaluation of the multiple policy implications of various models of integrating dried blood spot testing for Congenital Cytomegalovirus (CMV) and other genetic causes of hearing loss.

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 2009 quarterly Quality Assurance Reports were sent to each birthing hospital and Children's Hospital in Omaha. These reports included the individual hospital's quarterly measures and a statewide comparison on each measure. In addition, the publication "QI Hints" was sent out with each quality assurance report to the person(s) designated by the birthing hospital administrator.

Topics in 2009 included:

-Blood spot quality issues
-Technical assistance visit availability to hospitals
-Progress report on improved unsatisfactory specimen rates
-New electronic tracking system for shipping specimens

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 (NBSAC) provided technical expertise and policy guidance to the Nebraska Newborn Screening Program. Members commit at least a half a day every three months to advise the state program. 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 of the NBSAC in 2009 were:

- **CHAIR, James L. Harper**, M.D., *Pediatric Hematologist*, UNMC, Omaha
- **VICE-CHAIR Khalid Awad**, M.D., *Neonatologist*, Neonatal Care PC, Omaha
- **Lawrence Bausch**, M.D., *Neonatologist,* Saint Elizabeth Regional Medical Center, Lincoln
- **John Colombo,** M.D., *Pediatric Pulmonologist, Director,* Nebraska Cystic Fibrosis Center, 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
- **Kathryn Heldt,** RD, *Dietitian*, Children's Hospital Metabolic Clinic, Omaha
- **Mary Kisicki,** RN, *Parent*, Papillion
- **Richard Lutz,** M.D., specialist in *Pediatric Genetics, Endocrinology, Metabolism,* Munroe/Meyer Institute for Genetics and Rehabilitation, UNMC, Omaha
- **Bev Morton,** *Parent,* Lincoln
- **Samuel Pirruccello**, M.D., *Pathologist*, Regional Pathology Services, UNMC, Omaha
- **Deborah Perry,** M.D., *Pathologist*, Pathology Center, Omaha
- **William Rizzo,** M.D.**,** specialist in *Pediatric Genetics, Endocrinology, Metabolism,* Munroe Meyer Institute for Genetics and Rehabilitation, UNMC, Omaha
- **Kathy Rossiter,** MSN, CPNP, JD, Omaha
- **Jill Skrabal,** R.D., *Dietitian*, Munroe Meyer Institute for Genetics and Rehabilitation, UNMC, Omaha
- **Corri Stearnes,** *Parent*, Omaha
- **Douglas Stickle**, Ph.D., *Technical Director, Clinical Chemistry*, UNMC, Omaha
- **William Swisher,** M.D., *Pediatrician,* Lincoln Pediatric Group, Lincoln
- **B.J. Wilson,** M.D., *Neonatologist/Perinatologist*, Saint Elizabeth Regional Medical Center, Lincoln

Assurance of Treatment and Management of Conditions

How Treatment and Management is Paid:



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. (Some patients move out of state/new patients move in or are born/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 on the supplemental panel. Therefore the biggest funding source supporting the metabolic foods and formulas was revenue generated from the $10 per infant screened fee (approximately $270,000 per year). The State General Fund appropriation of $42,000 also helped provide for these medically necessary formulas and foods and the associated nutritional counseling for patients identified with PKU or the other metabolic conditions identified on the supplemental screen. Title V Maternal and Child Health Block Grant funds then filled in the gaps for metabolic foods/formula and nutritional counseling. The Medically Handicapped Children's Program provides some assistance to eligible families with children who have a hemoglobinopathy such as sickle cell disease or those with cystic fibrosis.

Individuals affected with screened metabolic conditions can obtain the metabolic formula through the Nebraska Medical Center Adult Metabolic Clinic, or at the Children's Hospital Metabolic Clinic. Ongoing dietary consultation, pediatric metabolic specialty care and routine blood monitoring are also provided 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. Families can order up to $2,000 of the pharmaceutically altered foods per year without having to pre-pay.

Nebraska's families:



In Federal Fiscal Year 2009, metabolic formula ordering and distribution and specialized nutritional counseling and monitoring were provided via a contract with the University of Nebraska Medical Center for $349,233. The individuals eligible for the metabolic foods utilized the pharmaceutically manufactured foods program, ordering foods with a value totaling $64,513.



Mike Rooney coordinates the day-to-day metabolic foods program helping families to understand the program and stay connected, and monitoring the vendors' compliance with the contracts. 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.

Sustaining the obligation to ensure access to treatment:

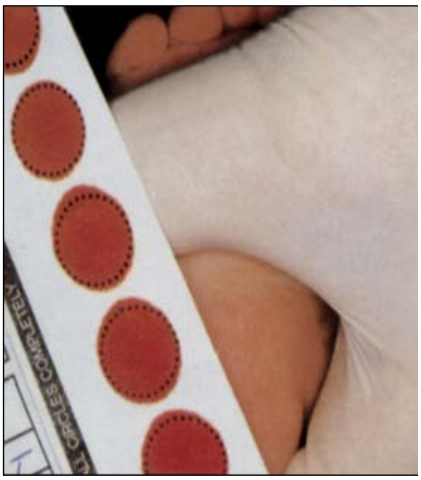
The number of children identified with conditions requiring special formula will always increase. The metabolic diets are required for life, so people do not "age-out" of the need for the special formulas or foods. State General Funds have remained flat and federal allocations to Nebraska of Maternal and Child Health Title V Block grant funds have been reduced or flat for several years. While a new drug received FDA approval, for which about 40% of patients with PKU are expected to be responsive, these medications are 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.

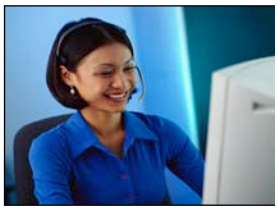
Nebraska's Newborn Screening Fees

In 2009 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). 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 2009 only 6 were lower (AZ, FL, ID, LA, NC, TX).

PROCESS/OUTPUT DATA FOR 2009

SPECIMEN COLLECTION, HANDLING AND TRANSPORT





Age at Time of Specimen Collection (Initial Specimen) 2009

Age at time of collection	Number of births	Percent of births
0-12 hours	212	0.77
12-24 hours	125	0.46
Collected day 2 (24-48 hours of age)	25,989	94.75
Day 3	855	3.12
Day 4	75	0.27
Day 5	21	0.08
Day 6	25	0.09
Day 7	12	0.04
Over 7 days	114*	0.42
Time of collection unknown	1	0.01

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

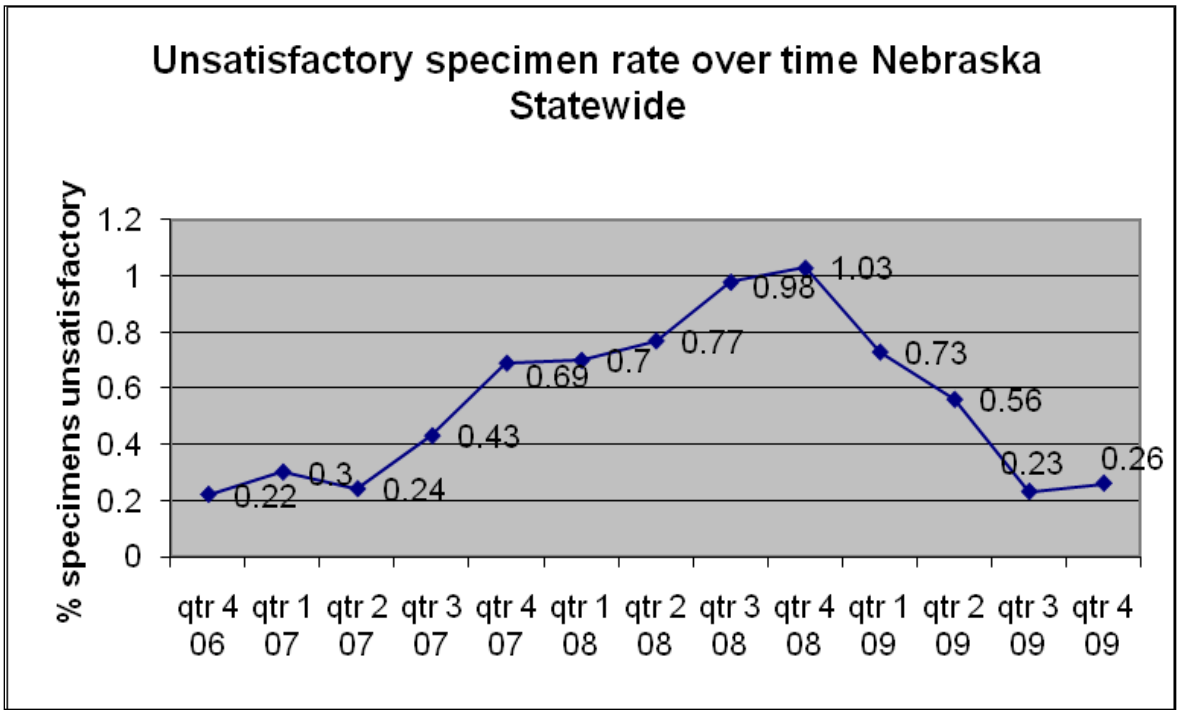
Regulations require all specimens to be collected between 24-48 hours of birth, or prior to discharge, transfer or transfusion whichever comes first. Specimens collected past day two are at increased risk of a delayed diagnosis.

Hospitals improved from 91.92% in 2008 to 94.75% of births screened at the correct time (between 24-48 hours of birth) in 2009.

Unsatisfactory Specimens for 2009

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. After two and a half years of QA and aggressive educational effort, State averages finally returned to the good performance levels routinely seen prior to mid 2007.

A substantial amount of effort went into to reducing the unsatisfactory specimen rate that had been increasing since 2007. Finally in 2009 the rate decreased significantly. The effort was worth it because unsatisfactory specimens are 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 art and science of correctly collecting and handling dried blood spots on filter paper requires trained health care professionals, who consistently follow the Clinical and Laboratory Standards Institute procedures for specimen collection. <u>Every</u> unsatisfactory specimen must be repeated, to ensure sufficiently reliable screening results.

Drawn Early
(less than 24 hours)

Specimens for 2009

YEAR 2009
SUMMARY OF DRAWN EARLY DATA

January 1, 2009 – December 31, 2009

#DE's per month:

Jan 2009	8	(8 known transferred)	(0 expired)
Feb 2009	24	(14 known transferred)	(0 expired)
Mar 2000	26	(14 known transferred)	(1 expired – specimen not repeated)
Apr 2009	22	(15 known transferred)	(0 expired)
May 2009	28	(14 known transferred)	(3 expired – specimen not repeated)
June2009	24	(16 known transferred)	(2 expired – specimen not repeated)
July 2009	21	(12 known transferred)	(1 expired – specimen not repeated)
Aug 2009	29	(20 known transferred)	(1 expired – specimen not repeated)
Sep 2009	31	(19 known transferred)	(1 expired – specimen not repeated)
Oct 2009	18	(13 known transferred)	(0 expired)
Nov 2009	24	(14 known transferred)	(3 expired – specimen not repeated)
Dec 2009	30	(13 known transferred)	(0 expired)

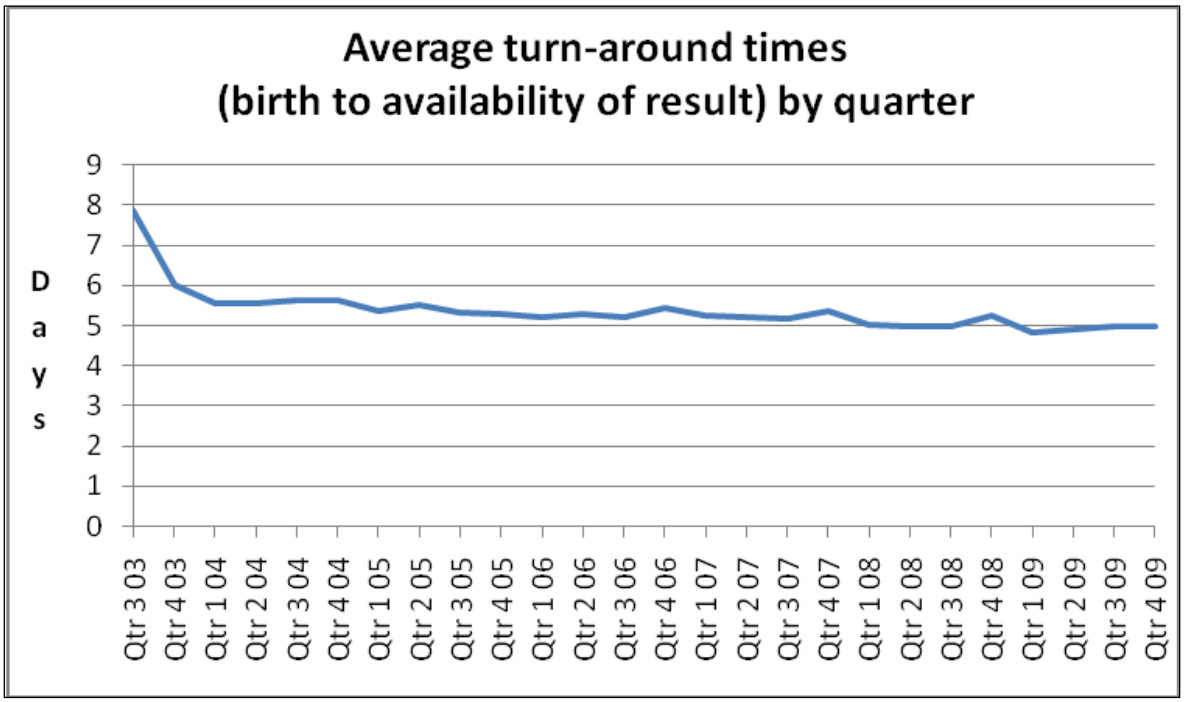
TOTAL: 290

****Note:** There were an additional 105 infants that were reported as drawn early and upon notification to the birthing facility it was reported and documented that there was a reporting error made.

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 2008 thanks to in-lab efforts at PerkinElmer Genetics, and hospital personnel responding to the quarterly quality assurance reports when turnaround times for collection were above the benchmark/average of 1.5 days of age.



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 percent of screen results will be positive that upon repeat or confirmation are found to be normal. Nebraska and programs across the country strive to minimize the number of newborns that require repeat or confirmatory testing (presumptive positive), and maximize the probability of identifying those affected. Nebraska continued to sustain a relatively low false positive rate for every condition screened.

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 a 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 **2009 Data**	**# Screened**	**# Presumptive Positive or inconclusive on initial screen**	**Presumptive Positive Rate**	**# lost to follow up**	**# confirmed Positive/ Diagnosed (classical or partial w tx/)**
Biotinidase deficiency	27,131	22	0.08%	0	5
Congenital Adrenal Hyperplasia	27,131	32	0.11%	0	1
Congenital Primary Hypothyroidism	27,131	114	0.42%	1 expired	15
Cystic Fibrosis	27,131	60	0.22%	2 not repeated 1- expired	9
Galactosemia	27,131	12	0.04%	0	0
Hypertyrosinemia of infancy (tx'd)	27,131	24	0.08%	0	1
MCAD	27,131	11	0.04%	0	3
PKU	27,131	5	0.01%		4
Sickle Cell Disease & other clinically significant hgbs	27,131	12	0.04%	0	12
SCAD	27,131	12	0.04%	0	2
VLCAD	27,131	6	0.02%	0	1
3-MCC	27,131	5	0.01%	0	1
All other abnormal Hgb's (carriers/variants)	27,131	450	1.65%		
All other abnormal MS/MS results	27,131	250	0.92%	0	See above

* babies lost to follow up expired before repeat or confirmatory testing could be completed

** 107 abnormalities from the MS/MS testing were elevations of multiple amino acids consistent with babies who were receiving parenteral nutrition

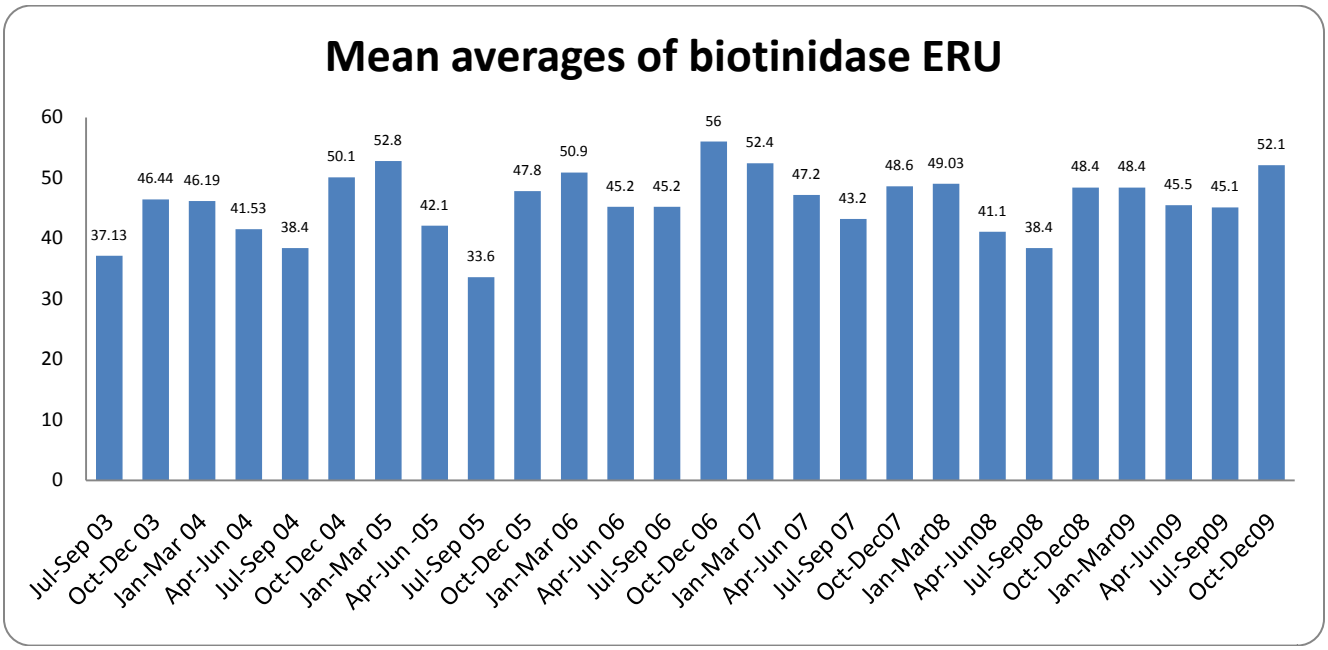
Mean Averages of Laboratory Test Measures

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.

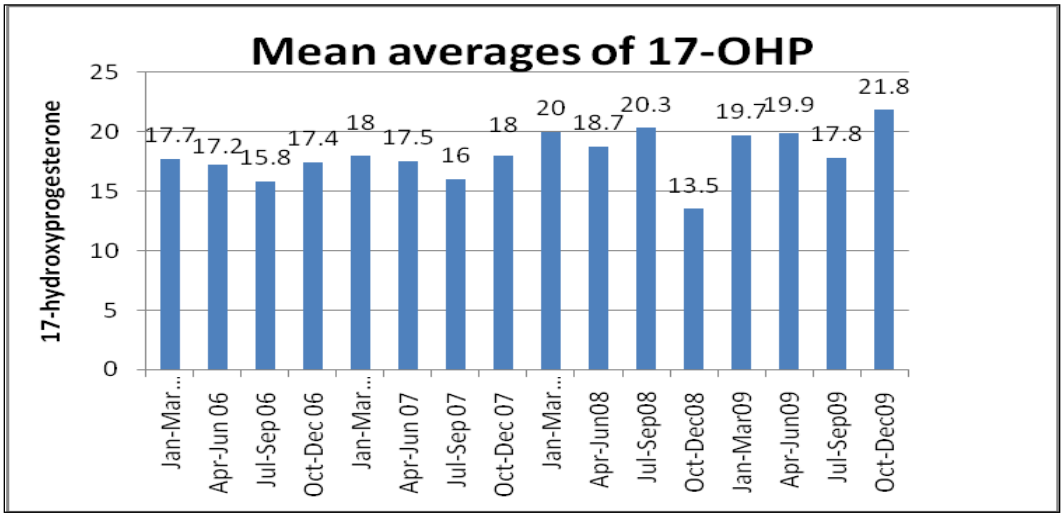
Health care providers familiar with the mean averages might feel more comfortable explaining the "relative risk" to parents of newborns with positive screening results, by comparing how far out of range the result is from the mean average, and from the normal expected range.

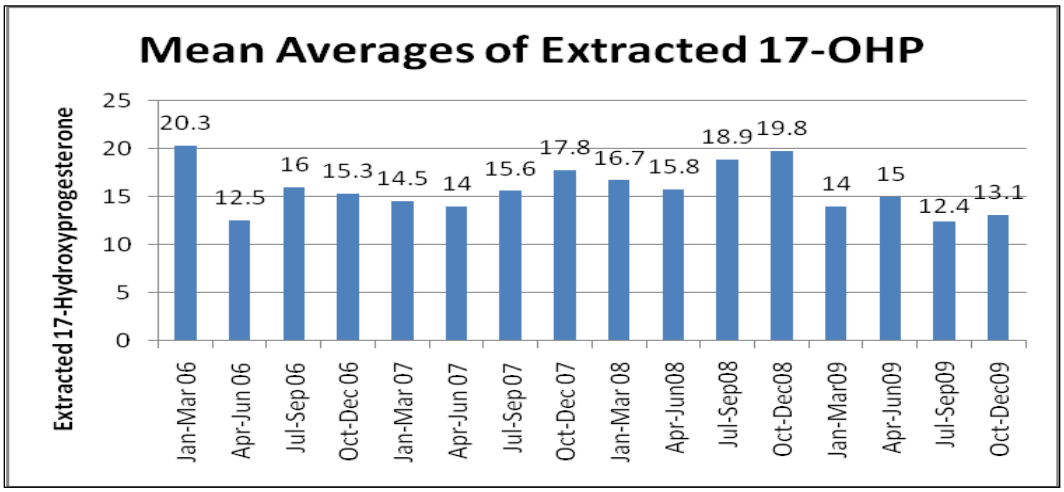
Expected seasonal differences can be seen each summer when heat exposure may impact the mean average enzyme levels detected in screening for biotinidase deficiency.

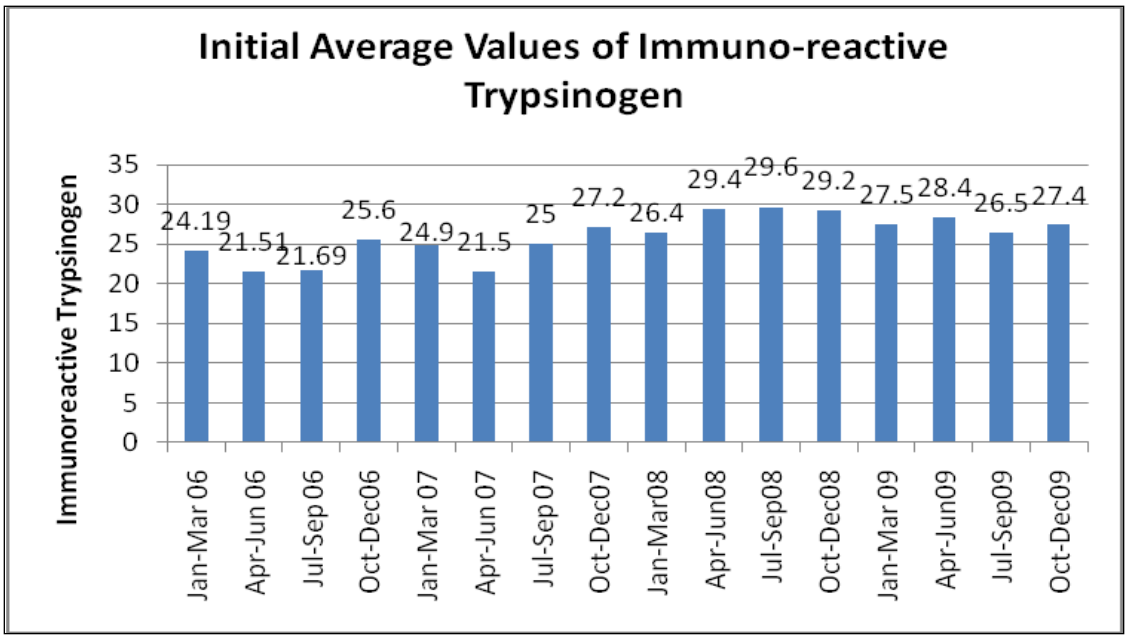
The Nebraska Newborn Screening Program sends a reminder each spring to hospital laboratories about specific practices to follow that will minimize the risk of specimens becoming heat denatured. This is intended to avoid the associated increase in the number of rejected specimens. In 2009, only 6 babies required repeat specimens 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 a significant improvement over 2008 when 29 babies required such repeat testing.



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.





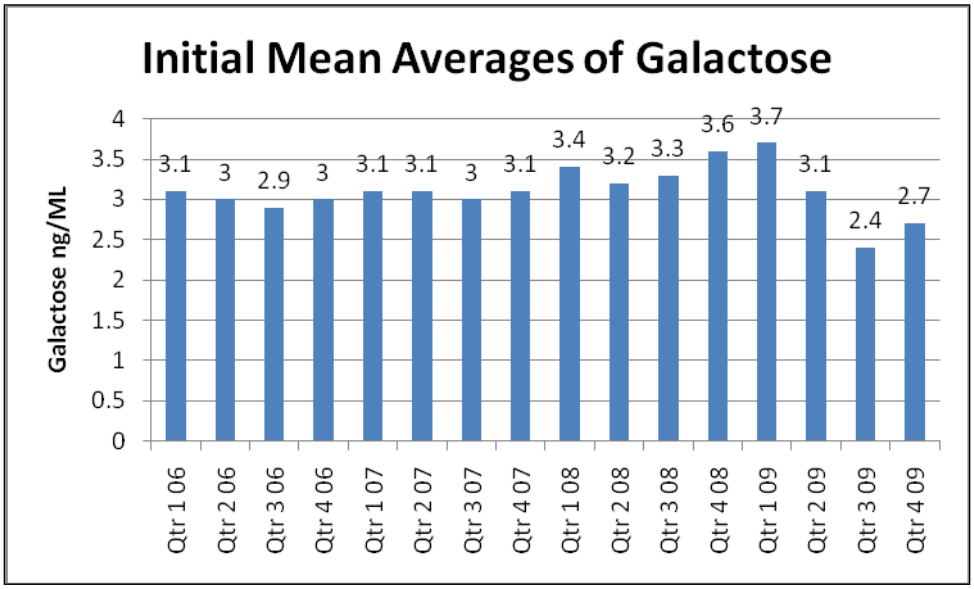


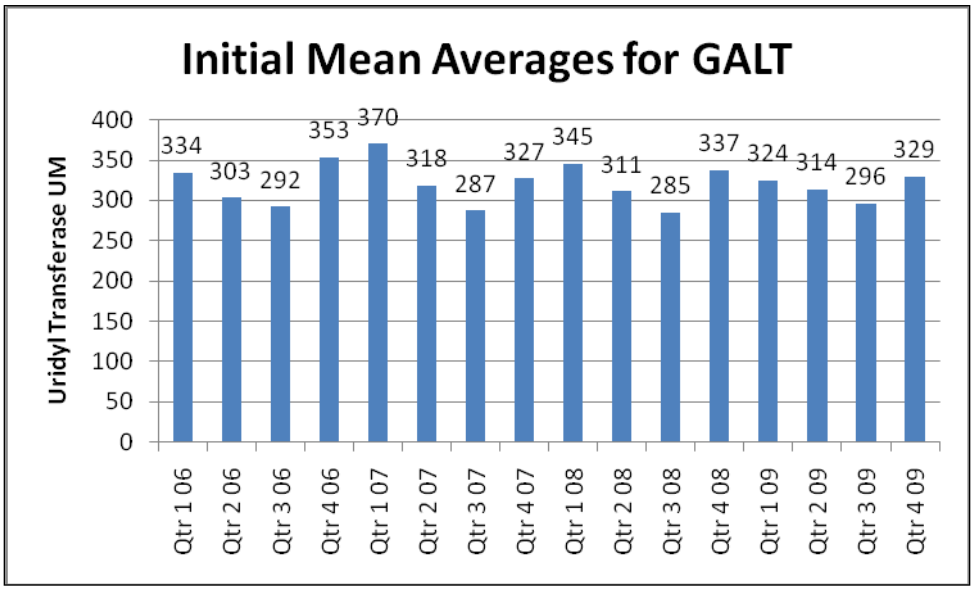
IRTs greater than 90 reflexed to test for ΔF508 the mutation most commonly associated with classical cystic fibrosis. (Those with one copy of the ΔF508) reflexed to testing for additional mutations on the INNOGENETICS 36 mutation panel). In Sept 09 the panel changed to a 39+ panel Xtag.

By looking at both elevations of galactose, and decreases in the enzyme activity of galactose phosphate uridyl transferase the laboratory can report with greater precision, those newborns at risk for classical Galactosemia who need immediate metabolic consultation/ referral and testing, vs. those whose findings are more consistent with a milder but potentially clinically significant form of Galactosemia.

Having this information can mean providing more parents with a bit more peace of mind since most will need only a repeat screen, vs. full confirmatory testing.

This also can translate into cost savings because doing a "requested repeat" screen at no charge may be all that is initially recommended, vs. more expensive confirmatory testing





Out of Hospital / Home Births

In 2009, there were 99 home births reported to the Department of Health and Human Services, Division of Public Health Services Newborn Screening Program (some reported later in 2010). All were screened, except for two of these babies who expired before they could be screened.

NEWBORN SCREENING DATA

	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009
Total Births	24,958	25,109	25,515	26,067	26,443	26,349	26,898	27,107	27,094	27,199
Births Screened	24,863 99.6%	25,043 99.7%	25,478 99.85%	26,008 99.77%	26,391	26,288	26,819	27,013	27,021	27,131
Total Births Lost to Follow up	6 + (89 not screened- as expired @ <48 hours.)*	2 + (64 not screened as expired @ < 48 hours)	5 + (32 not screened as expired @ < 48 hours)	5 + (54 not screened as expired @ < 48 hours)	2 + (50 not screened as expired @ < 48 hours)	0 + (61 not screened as expired @ < 48 hours)	2 + (79 not screened as expired @< 48 hours)	(94 not screened as expired @ < 48 hours)	1 (+ 73 not screened as expired @ < 48 hours)	3 (67 not screened as expired @<48 hrs, 1 discharged w/o screen & out of state)
Total Births PP**	412	432	456	415	499	503	537	511	553	99 + 912 (see foot note)
Home Births	109	93	99	70	60	55	69	80	86	99
Home Births Screened	105	88	95	65	60	54	69	78	85	97
Home Births Lost to follow up[1]	4	2 + (3 expired)	2 + (2 expired)	3 + (2 expired)	0	0 + (1 expired)	0	2 (both expired)	1 (expired)	2 (expired)

*Began 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.
Note: In 2010 began counting number of screens presumptive positive as those requiring confirmatory testing because of a more substantial out-of range result (99). The 912 out of range results listed required only repeat testing of a dried blood spot filter paper.

Biotinidase Deficiency	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009
Presumptive Positive	2	4	3	4	34*	78	14	5	4	5
Inconclusive								10	25	17
Confirmed Negative	2	1	1	0	29	71	9	11	23	17
Confirmed Positive Profound	0	0	2	1	0	1	0	0	1	0
Confirmed Positive (Partial no tx)	0	0	0	0	0	0	0	0	0	0
Confirmed Positive (Partial tx)	0	3	0	3	6	5	4	4	3	5
Lost to follow up	0	0	0	0	0	1**	1**	0	2***	0

*Screening protocols identified most of these as "inconclusive," for which repeat screening rather than confirmatory testing, ruled out the condition.
** lost to follow up as newborn expired

Congenital Adrenal Hyperplasia	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009
Presumptive Positive	N/A	N/A	N/A	N/A	N/A	N/A	10	3	17	7
Inconclusive								18	22	25
Confirmed/ repeated Negative	N/A	N/A	N/A	N/A	N/A	N/A	9	17	36	31
Confirmed Positive	N/A	N/A	N/A	N/A	N/A	N/A	1	1	1	1
Confirmatory or Repeat Lost to follow up	N/A	N/A	N/A	N/A	N/A	N/A	0	3*	2*	0

* expired before repeat or confirmatory testing could be done. Includes one set of twins.

Congenital Primary Hypothyroidism	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009
Presumptive Positive	114	115	129	89	63	58	51	39	57	56
Inconclusive (drawn early but low T4/high TSH								20	52	48
Confirmed Or repeated Negative	104	105	113	75	55	48	41	43	96	88
Confirmed Positive	8	7	15	11	8	9	10	16	12	15
Confirmatory or Repeat Lost to follow up	2*	3*	1*	3*	0	1*	0	3*	1	1*

*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.

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

Galactosemia	**2000**	**2001**	**2002**	**2003**	**2004**	**2005**	**2006**	**2007**	**2008**	**2009**
Presumptive Positive	12	15	5	3	9	1	8	0	0	2
Inconclusive repeat rec'd	n/a	n/a	n/a	n/a	n/a	n/a	n/a	9	9	10
Confirmed / repeated Negative	8	9	5	0	6	1	8	8	9	12
Confirmed Positive (Classical)	1	0	0	1	0	0	0	0	0	0
Confirmed Positive, Duarte (not treated)	1 Duarte Hmzg	0	0	1	0	0	0	0	0	0
Confirmed Positive, Duarte (treated)	2 Duarte Mixed Htrzgt. (1 tx'd 1 year)	6 Duarte Mixed Htrzgt.	0	1	3	0	0	1	0	0

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:

	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009
FS	2	4	4	5		1	3		3	1
FC					1	1		1*		1
FSC	1	2	2		1	1	2	1	1	5
FE	1						1			1
Sickle Beta Thal								1		3
Alpha Thal Major		1 (4-gene deletion)								
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 2009:

177 Sickle Cell Trait
9 Hgb. D Trait
12 Alpha Thal Trait
6 FAS + Alpha Thal Trait
150 diagnosis Unknown

27 Hgb. C Trait
39 Trait + other
8 miscellaneous trait
2 FAS + Alpha Thal silent carrier

15 Hgb. E Trait
16 Alpha Thal silent carrier
5 Gamma chain variants
1 FAC + Alpha Thal silent carrier

Diagnosis unknown for 151 positive hemoglobinopathy screens (not suspected for clinically significant conditions) (confirmatory results not reported back to program)

*MCAD **	*2000*	*2001*	*2002*	*2003*	*2004*	*2005*	*2006*	*2007*	*2008*	*2009*
Screened Positive	N/A	N/A	3*	3	5	10	5	0	4	3
Screened inconclusive (repeat only)**	N/A	N/A	N/A	N/A	N/A	N/A	N/A	2	2	8
Confirmed Negative or Repeated normal	N/A	N/A	2	3	1	7	5	2	2	8
Confirmed Positive	N/A	N/A	1	0	4	3	0	0	4	3

*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)	**2000**	**2001**	**2002**	**2003**	**2004**	**2005**	**2006**	**2007**	**2008**	**2009**
Presumptive Positive	6**	4	3	7**	7	3	6	0	0	4
Screened Inconclusive (repeat only)***	N/A	N/A	N/A	N/A	N/A	N/A	N/A	2	0	1
Confirmed Negative	2	2	1	1	1	1	1	2	0	1
Confirmed Positive Classical PKU	1	1	1	2	1	2	0	0	0	3
Confirmed Positive Hyperphe	1 transient	1	1	3	5 (3 of these tx'd)	0	5 (4 of these treated)	0	0	1

****2000 and 2003**: One each year for whom confirmatory testing was not done as the babies 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	106 (+11 on repeats)	104 + 11	2-expired	
Several Amino Acids	95 (+12 on repeats)	91 +12	4-expired	
Several Amino Acids & A generalized elevation of short-chain & medium-chain acylcarnitines	1	1		
Propionylcarnitine (C3)	18 (+2 on repeats)	18 +2		
C3 & C3/C2 & C3/C16	23 (+1 on repeat)	23		
C3 & C3/C2 & C3/C16 + Mild Elevations of Amino Acids	1	1		
Glutarylcarnitine (C5DC)	3	3		
Citrulline	1 (+1 on repeat)	1 +1		
Butyrylcarnitine (C4)	6	4		2 Confirmed Positive for SCAD
Butyrylcarnitine (C4) and other indices such as the relation ratio of C4 to Propionylcarnitine (C3)	3 (+ 3 on repeats)	2 +3		1 Confirmed Positive for SCAD
Octanoylcarnitine	8 (+1 on repeat)	5		3 Confirmed Positive for MCAD
Generalized Elevation of Short-Chain & Medium-Chain	8 (+26 on repeats)	8 +26		
Methionine & Tyrosine	15	15		
Methionine & Propionylcarnitine	1	1		
Methionine & C 3 & C3/C2 & C3/C16	1	1		
Myristoylcarnitine (C14) & C14:1 & C16	1			1 Confirmed Positive for VLCAD
Tetradecenoylcarnitine (C14:1) to C16 & other long chain acylcarnitines	5	5		
3-Hydroxydecanoylcarnitine (C10OH), C8 & C10	2	2		
3-Hydroxyisovalerylcarnitine (C50H)	1 (+1 on repeat)	1		1 Confirmed Positive for 3-MCC Deficiency
Free Carnitine & Short Chain Acylcarnitines	1	1		
Tyrosine	19 (+5 on repeats)	17 +5	2	1 Confirmed Positive for Transient Tyrosinemia 1 Confirmed Positive for Self Limited Hypertyrosinemia of Infancy
Phenylalanine/ Tyrosine ratio	5	1		3 Confirmed Positive for PKU 1 Confirmed Positive for Hyperphenylalanemia
Forminioglutamic Acid (FIGLU)	1	1		
2009 Totals (NE Infants)	**326**	**306**	**6 expired**	**14 Confirmed Positive**

*Lost to follow up designated when the patient/parent can no longer be found and there is no medical home, or they have moved out of state to an unknown location.
**The vast majority of abnormal screens from MS/MS require only a repeat screen to rule out the condition. Confirmatory testing is recommended in a small percentage of cases where the concentration of analytes are "significantly" abnormal, or concentrations of analytes increase on repeat screens.
***Total babies less than # of abnormal screens as some that had more than one abnormal screen, e.g. methionine on initial screening, and multiple amino acids on a repeat screen.



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 at intervention/tx.	Range in ages at intervention/tx.
5 Biotinidase Deficiency (partial) (3 treated)	15	10-20 days
1 Congenital Adrenal Hyperplasia* (classical)	2 days	
14 Congenital Primary Hypothyroidism	11 days	5-27 days
1 Congenital Hypothryoidism	85 days	
6 Cystic Fibrosis	10 days	2-13 days
1 Non-Classical Cystic Fibrosis	16 days	
3 MCAD	7 days	4-12 days
3 PKU	7 days	All tx'd @ 7 days
1 Hyperphenylalaninemia	5 days	
6 Sickle Cell Disease	10 days	6-15 days
1 Hgb. C Disease	33 days	
1 Hgb. E Disease	24 days	
3 Sickle Beta Thalassemia	13 days	8-22 days
2 SCAD	19.5 days	15-24 days
1 VLCAD	7 days	
1-3MCC Deficiency	12 days	
1-Hypertyrosinemia of Infancy	20 days	

One infant with 11-ß-Hydroxylase Deficient CAH identified outside the screening program. The screen was normal for this infant. This form of CAH is unlikely to be picked up by the screening test for classical CAH in which the 17-hydroxyprogesterone is typically elevated.

NEBRASKA EARLY HEARING DETECTION AND INTERVENTION ANNUAL REPORT - 2009

Introduction

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

In 2000, the Infant Hearing Act established newborn hearing screening in Nebraska. The Nebraska Early Hearing Detection and Intervention (NE-EHDI) Program strives to fulfill the four purposes of the Infant Hearing Act (Neb. Rev. Stat. §71-4735):

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

The Act required birthing facilities to educate parents about newborn hearing screening, to include hearing screening as part of the standard of care and to establish a mechanism for compliance review by December 2003. 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.

Newborn hearing screening requires objective physiologic measures to detect hearing loss in newborns and young infants. There are two basic techniques available to screen newborns for hearing loss. Both are easily recorded in newborns and are non-invasive measures of physiologic activity that underlie normal auditory functioning.

The most frequently used screening technique is measurement of otoacoustic emissions, or OAEs. A miniature earphone and microphone are placed in the newborn's ear canal, low intensity sounds are presented, and responses produced by the inner ear are measured. The second screening technique, Auditory Brainstem Response, or ABR, uses small electrodes to detect certain brainwaves in response to sounds that are presented by a miniature earphone. For both methods, the response of each ear is measured. OAE and

ABR are both reliable and accurate. Screening can occur as early as 12 hours of age, preferably with the newborn sleeping, and averages from five to 20 minutes to complete.

If a response is not detected for one or both ears, the result is a "refer" (did not pass). A "refer" to the screening test indicates that a hearing loss *may* exist but there are also other factors that may have contributed. A "refer" does indicate that a second screening is necessary to determine if the other factors, such as vernix in the ear canal, fluid in the middle ear cavity, movement, equipment failures, or inexperience of the tester, contributed to the initial result. A "refer" on the second screening indicates the need for a diagnostic audiologic evaluation to confirm or rule out a hearing loss and, if hearing loss is present, to identify the type and degree of the loss and to begin intervention services.

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

Newborn Hearing Screening Data Reported for 2009

Birthing Facility Screening Programs

The number of birthing facilities conducting newborn hearing screening increased rapidly from 2000 when only 11 hospitals were conducting either targeted or universal newborn hearing screening (see Table 1). 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. Fifty seven (57) of the 58 birthing hospitals conduct the hearing screening during the birth admission and one conducts the screening on an outpatient basis following discharge.

Birthing Facilities Conducting Newborn Hearing Screenings (2000-2009)

Year	Number of Birthing Facilities in Nebraska	Number Conducting Newborn Hearing Screening	Percentage Conducting Newborn Hearing Screening
2000	69	11	16%
2001	69	24	35%
2002	69	57	83%
2003	67	67	100%
2004	67	67	100%
2005	65	65	100%
2006	63	63	100%
2007	63	63	100%
2008	61	61	100%
2009	58	58	100%

Table 1

Annual Birthing Facility Reports

Birthing facilities are required to annually report specific information about their newborn hearing screening programs to the Department of Health and Human Services (Neb. Rev. Stat. §71-4739). The ERS-II data system, an integrated module of the State's Vital Record system, automatically calculates these figures for each birthing facility.

Birthing Facility Reports of Required Aggregate Data (2009)

Number of newborns born in birthing facilities	27,104
Number of newborns and infants recommended for a hearing screening test	26,523
Number of newborns who received a hearing screening during birth admission	26,806*
Number of newborns who passed a hearing screening during birth admission, if administered	25,655*
Number of newborns who did not pass a hearing screening during birth admission, if administered	1151*
Number of newborns recommended for monitoring, intervention, follow up care	596

*Includes babies transferred to Children's Hospital and Medical Center, a non-birthing facility *Table 2*

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 education materials free of charge to hospitals to help fulfill this requirement. Print materials are available in 10 languages. Birthing facilities reported educating almost all parents (26,523 or 97.9%) about newborn hearing screening, hearing loss and normal speech and language development in 2009. Neb. Rev. Stat. §71-4740 requires the 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. There were 95 babies recorded as having been born out-of-hospital in 2009. Parent education material was sent to the parents of the 79 babies who were not admitted to a hospital immediately following birth.

Newborns Receiving a Hearing Screening

The Infant Hearing Act requires that rules and regulations be adopted and promulgated if the annual percentage rate of newborns who receive a hearing screening during birth admission is less than 95% by December 1, 2003, or at any time thereafter. Hearing screening results reported for occurrent births in 2009 show that 26,806 newborns, or 98.9% of hospital births, were screened during birth admission or prior to discharge to home. The number 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 (see Table 3). This increase in the numbers of newborns receiving a hearing screening corresponds to the increase in the number of hospitals adopting newborn hearing screening as the standard of

care for newborns and the support of sub-grants through the Nebraska Health Care Cash Fund to purchase screening equipment in 2002 and 2003.

Newborns Receiving a Hearing Screening Prior to Discharge to Home (2000-2009)

	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009
Number Receiving a Hearing Screening during Birth Admission	8,978	15,272	22,615	25,275	25,966	26,179	26,615	26,737	26,772	26,806
Percent Receiving a Hearing Screening during Birth Admission	36%	61%	89%	97%	98%	99%	99%	99%	99%	99%

Table 3

Newborns Discharged Without a Hearing Screening

During 2009, the ERS-II reports available for each birthing facility indicated that there were 303 newborns who did not receive a hearing screening during birth admission because the newborn expired prior to screening (89), was discharged to home prior to screening (209) or the parent(s) refused (5).

Birth Admission "Refer" Rates

The ERS-II reports available for each birthing facility indicated that 1,150 newborns did not pass the hearing screening during birth admission or prior to discharge to home for those babies who were transferred to another hospital. Of the hearing screenings conducted during birth admission, the "refer" rate for all birthing facilities was 4.3% during 2009 (see Table 4). In the last two years, the "refer" rate has increased by 0.3% annually after staying relatively steady for the preceding six years.

Birth Admission "Refer" Rates (2002-2009)

	2002	2003	2004	2005	2006	2007	2008	2009
"Refer" rate for birthing facilities	3.7%	3.6%	3.5%	3.4%	3.8%	3.7%	4.0%	4.3%

Table 4

There are two measurement techniques used to conduct newborn hearing screening: Otoacoustic Emissions (OAE) and Auditory Brainstem Response (ABR). Half of the birthing hospitals in Nebraska are using OAE-only, almost one third are using ABR-only, and the remaining birthing hospitals are using a 2-step method (OAE, followed by ABR if the initial

screening is a "refer"). The "refer" rates differ for the three techniques with the OAE-only having the highest "refer" rate (see Table 5).

"Refer" Rates for Hearing Screening Techniques (2009)

	OAE-only	ABR-only	2-Step
Number of Birthing Facilities	29	19	10
"Refer" Rate	9.8%	2.1%	3.8%

Table 5

Monitoring, Intervention, and Follow up Care

Another ERS-II report available for each birthing facility is the number of newborns recommended for monitoring, intervention, and follow up care. In 2009, 596 (51.8% of the babies who did not pass) were recommended for monitoring, intervention and follow up care by the birthing facilities. Regardless of whether the hospital indicated a recommendation had been made to the parent(s), the NE-EHDI Program's tracking and follow up processes were followed for each baby who did not pass the hearing screening during birth admission.

The NE-EHDI Program also tracked 1,533 newborns who were transferred to neonatal intensive care units or to hospitals with a higher level of care in Nebraska and surrounding states prior to receiving a hearing screening.

Out-of-Hospital Births

Although parent education material was provided by the NE-EHDI Program to the parents of all reported out-of-hospital births during 2009 who were not immediately admitted to a hospital, only 35.8% of out-of-hospital births were screened (see Table 6). The remainder was not screened or the results were not submitted to NE-EHDI Program.

Out-of-Hospital Births (2001 – 2009)

	2001	2002	2003	2004	2005	2006	2007	2008	2009
Out-of-hospital births	93	99	70	60	55	68	77	82	95
Number screened	5	16	12	13	15	30	34	39	34
Percentage screened	5%	16%	17%	22%	27%	44%	44%	48%	36%

Table 6

Confirmatory Testing/Audiologic Data Reported for 2009

The Advisory Committee for the NE-EHDI Program identified the initial level of the follow up hearing test for many newborns as an outpatient screening of the newborn's hearing. For those newborns and infants who pass this initial level of follow up, no further audiologic evaluation would be needed, unless there are risk factors present that would warrant periodic monitoring.

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

Annual Confirmatory Testing Facility Reports

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

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

Each year data regarding the follow up hearing tests at confirmatory testing (audiologic evaluation) facilities have been gathered by surveying the audiologists in Nebraska. Thirty seven (37) confirmatory testing facilities responded in 2009, representing 80 licensed audiologists. The results of those surveys are included in Table 7.

Required Follow up Hearing Test Data Reported by Audiologists

	Re-screenings	Diagnostic Evaluations
Number of newborns/infants receiving a follow up hearing test	794	236
Number of newborns/infants without a hearing loss	676	142
Number of newborns/infants with a hearing loss	118 ("refer")	80

Table 7

Diagnosis of Hearing Loss

The number of infants diagnosed with a hearing loss in Nebraska is reported in two ways: 1) aggregate reports submitted by audiologists with the number of infants shown to have a hearing loss based on follow up tests (required by Neb. Rev. Stat. §71-4739) and 2) the individual diagnostic reports submitted to NE-EHDI Program by audiologists or Primary Care Providers. Aggregate reports may include duplicate entries. Statutory authority to require audiologists to report on all newborns and infants who receive audiologic evaluations does not exist, so a one-to-one correspondence between the individual results reported to NE-EHDI Program and the required annual aggregate reporting does not exist. As shown in Table 7, the aggregate reports indicated that 80 infants were identified with either transient or permanent hearing loss.

Type and Degree of Hearing Loss

Analysis of the individually-identifiable confirmatory testing reports submitted to the NE-EHDI Program indicates that 46 of the infants with hearing loss meet the criteria for a Permanent Congenital Hearing Loss (PCHL) and two who had passed the newborn hearing screening were later identified with a permanent hearing loss, either acquired or later-onset. Thirty two (32) of the infants with a PCHL were identified with a bilateral hearing loss, 31% in the mild to moderate range and 69% in the severe to profound range. Eleven (11) infants were identified with a unilateral hearing loss, all of which were in the mild to moderate range. The remaining five (5) infants were identified with auditory neuropathy spectrum disorder. Individually-identifiable records indicated that amplification was recommended for 26 (per ERS) of the babies with PCHL and 22 (19 through NCHLB plus 3 ERS) were reported to have been fit with a hearing aid. However, the aggregate reports indicated that 22 infants had been fit with amplification.

Type and Degree of Permanent Congenital Hearing Loss, 2009 (n = 48)

Degree ► Type ▼	Bilateral Mild–Moderate	Bilateral Severe–Profound	Unilateral Mild–Moderate	Unilateral Severe-Profound
Sensorineural	7	18	3	0
Conductive	0	-	7	-
Mixed	0	2	0	0
Undetermined	3	2	1	0
Auditory Neuropathy Spectrum Disorder		4	1	

Table 8

The estimates of the incidence of PCHL in newborns range between one to three per thousand births nationally. Based on the birth rate in Nebraska during 2009 (27,199 including both birthing facility and out-of-hospital births), an estimated 27 to 82 newborns would be identified with PCHL. The incidence of PCHL in Nebraska for babies screened in 2009 reported in individual reports is 1.7 per thousand births.

Tracking and Follow up Results for 2009

The NE-EHDI Program tracked 1,360 newborns who were reported as not passing a newborn hearing screening during birth admission. Of those, 1,151 newborns had a birth admission hearing screening "refer" status and 209 newborns were discharged to home prior to receiving a hearing screening. These were the newborns who were tracked through follow up outpatient screenings, diagnostic evaluations and early intervention services.

Rate of Follow up Outpatient Screening and Confirmatory Testing

Follow up services include outpatient hearing screenings, audiologic diagnostic evaluations, or a combination of the two, depending upon clinical findings. The aggregate reports from the confirmatory testing facilities indicated that 794 newborns received screenings and 236 received audiologic diagnostic evaluations. With aggregate reporting, it is not possible to determine an unduplicated count, since some infants, especially those with middle ear dysfunction and an accompanying transient conductive hearing loss, may be screened or evaluated multiple times at one or more facilities. The data in Diagram 1 below provides another view of outpatient events based on the ERS-II Data System.

Follow up Services and Outcomes

Based on individual reports submitted to the NE-EHDI Program, follow up screening and/or diagnostic evaluations were completed for 1,173 infants with 1,127 having normal hearing and 46 being diagnosed with a PCHL. The evaluation process is still in progress for 27 infants, the parents of 14 infants refused to complete the recommended follow up, 20 families moved with no further contact possible, and 4 infants expired before completion of the follow up services. There were 122 newborns needing follow up for whom follow up services were not initiated, were initiated but not completed, or were not reported to NE-EHDI Program. These are designed as "Lost to System."

Diagram 1 tracks the services and outcomes of the 1,360 newborns needing follow up services through the EHDI system and indicates the results for those infants.

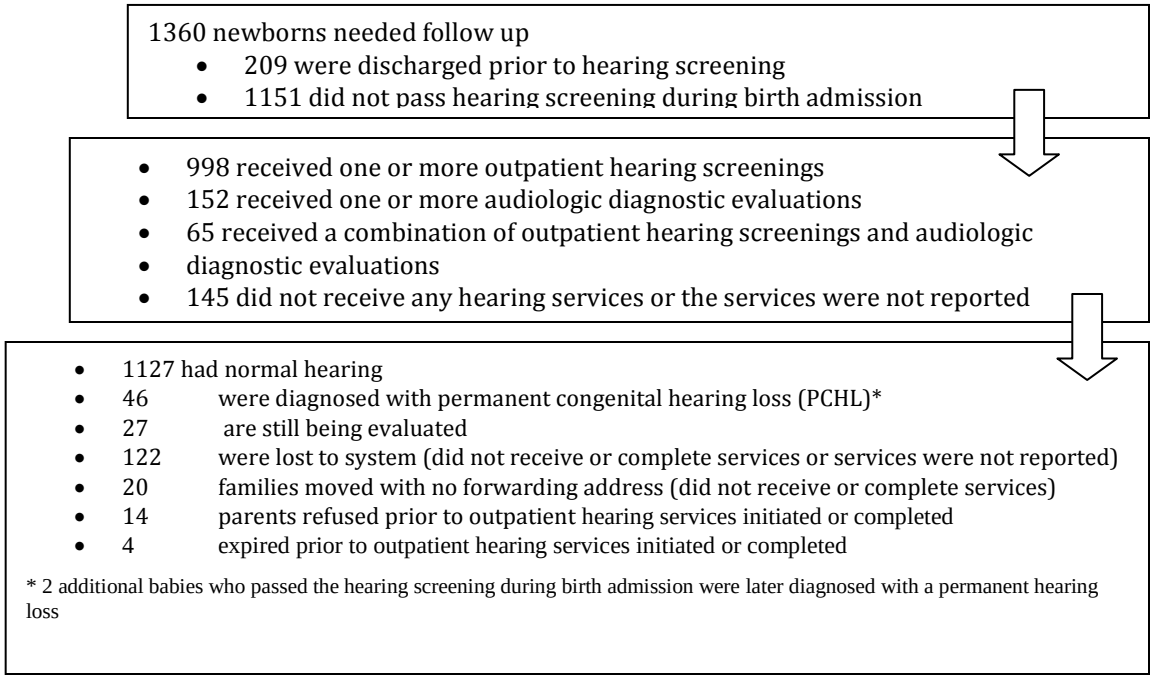


Diagram 1

Timeliness of Follow up Screening/Testing

To meet the state and national guidelines 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. For the newborns who received follow up services, 65.7% received an outpatient screening or diagnostic evaluation prior to 1 month of age. The peak of follow up activity occurred at approximately 2 weeks of age (see Chart 1). The average age of follow up service initiation was 33.9 days.

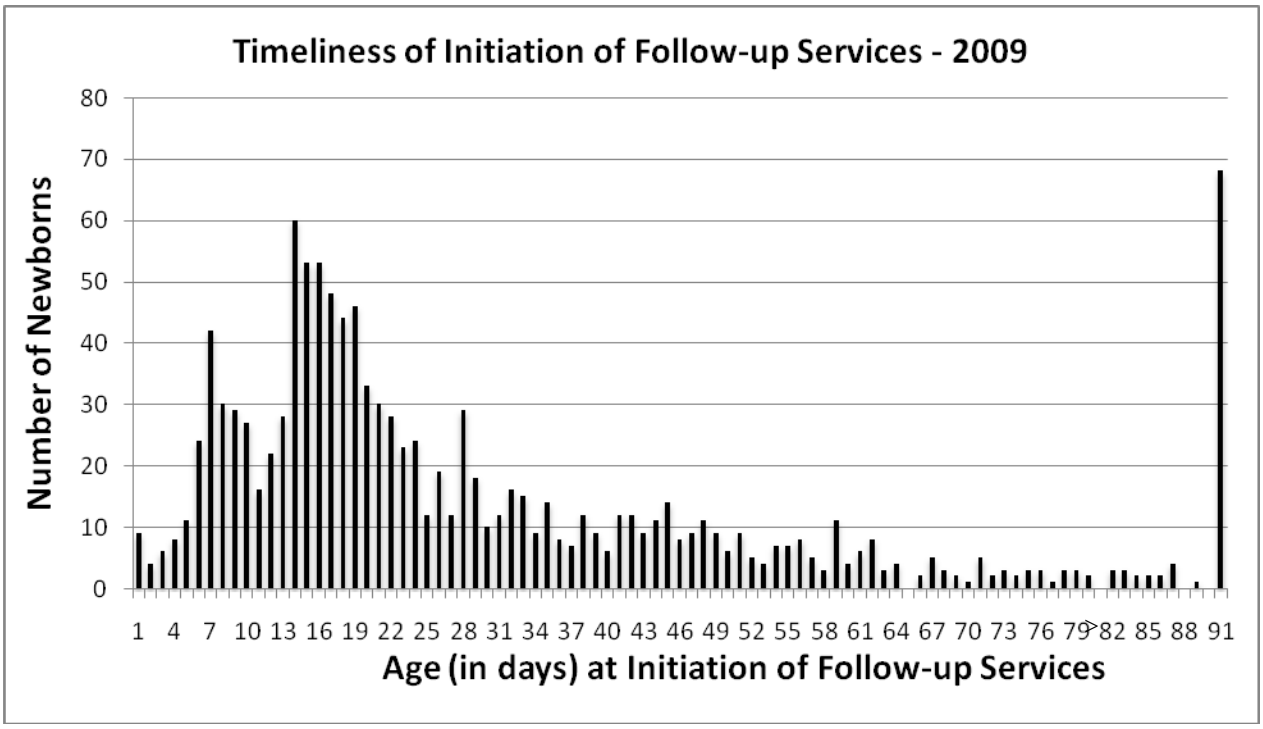


Chart 1

Individual reports were received by the NE-EHDI Program for 46 infants diagnosed with a permanent hearing loss. Two of these were newborns who had passed the newborn hearing screening during birth admission but were diagnosed with a later-onset or progressive hearing loss. The average age at confirmation of hearing loss was 138.3 days with 56% having a hearing loss confirmed before 90 days of age, the recommended benchmark.

Incomplete Results

Neb. Rev. Stat. §71-4742 states: "…it is the goal of this state to achieve a one-hundred-percent screening rate." While Nebraska continues to make very good progress in developing a comprehensive early hearing detection and intervention system, there are also infants for whom the status of their hearing is not known. Overall in 2009, there were 337 babies whose hearing status has not been objectively established:

- 122 infants with no outpatient follow up initiated, completed or reported after

not passing or receiving the inpatient newborn hearing screening.
- 27 infants were identified with hearing problems associated with middle ear dysfunction but additional follow up evaluations have not yet been completed.
- 61 of the out-of-hospital births were not screened or the results were not submitted to NE-EHDI Program.
- 20 families moved before hearing services were initiated or completed.
- 14 parents refused the hearing screening during birth admission or refused to complete the hearing screening after discharge to home.
- 93 newborns expired prior to receiving an inpatient hearing screening or completing outpatient follow up services.

Based on the analysis of the hearing screening and follow up records, the hearing status (normal hearing or permanent hearing loss) of 98.8% of the 27,199 newborns born in birthing facilities and out-of-hospital has been established.

Early Intervention

The purpose of the Infant Hearing Act (Neb. Rev. Stat. §71-4735) is to "obtain needed multidisciplinary evaluation, treatment, and intervention services at the earliest opportunity and to prevent or mitigate the developmental delays and academic failures associated with late detection of hearing loss." Forty-eight records for the Early Development Network (EDN), Nebraska's Part C Early Intervention Program, indicate that 42 (88%) of the 48 infants born in 2009 and diagnosed with a PHL (46 PCHL) were referred to EDN. Of those, 39 verified for special education services. Verification for 31 of the 39 infants was completed prior to 6 months of age and 8 (21%) infants were verified after 6 months of age. Four (8%) infants were not referred to EDN. Three (6%) were referred but not verified (1 (2%) moved or lived out-of-state and the parents of 1 (2%) infants withdrew prior to verification). Eighteen (38%) of the infants diagnosed with a PCHL have an identified medical home.

ACTIVITIES – 2009

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. A new three-year competitive grant from HRSA/MCHB was awarded to continue improving the rate of follow up to newborn hearing screening. 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 Infant Hearing Act of 2000 and the recommendations by the NE-EHDI Advisory Committee. Specific tasks to be accomplished by the Advisory Committee are 1) to continue to increase the representation of stakeholders, 2) to review and, as necessary, revise the existing protocols to incorporate the electronic data system, 3) to develop new reporting, tracking and follow up protocols to effectively link the NE-EHDI Program and the early intervention systems, 4) to increase the program's responsiveness to the expanding cultural and linguistic communities in the state, 5) to support the development of an effective professional development system, and 6) to guide the long-term planning and evaluation of the EHDI system in the state. The Advisory Committee of the NE-EHDI Program consists of 22 members representing medical, audiology, parents, public health, family support, and education stakeholders. The Advisory Committee met three times during 2009. There are three official sub-committees of the NE-EHDI Advisory Committee: Audiology, Evaluation and Family Support.

Projects

Hearing Screening Equipment for Birthing Facilities

Opportunities to contract for partial funding of new hearing screening equipment were offered to birthing facilities to reduce the number of babies who referred during birth admission hearing screening due to use of aging or inappropriate hearing screening equipment. The funding was made available to small birthing facilities with less than 500 births, large birthing facilities with more than 500 births that were using OAE hearing screening technology, and hospitals with NICUs that did not have dedicated ABR hearing screening equipment.

Electronic Data System

The ERS-II data system was revised to improve functionality and to incorporate additional reports for the birthing facility users. The administrative tracking and follow up system was also further developed.

Family-to-Family Support

The Family Support Work Group of the NE-EHDI 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.

Loss and Found DVD

With funding from the HRSA/MCHB supplemental grant, the NE-EHDI Program joined seven other states in contracting with the national chapter of Hands & Voices to develop an educational DVD for birthing facilities to show to parents of babies who did not pass the newborn hearing screening to reduce the number of babies who are lost to system. The

DVD, available in English and Spanish, is based on experiences of families with following up to failed newborn hearing screenings. It includes Nebraska-specific information.

Nebraska Children's Hearing Aid Loaner Bank (NCHALB)

The NCHALB began providing loaner hearing aids to young children in January, 2008. The NCHALB is a partnership between the University of Nebraska - Lincoln Barkley Center, Nebraska Association for the Education of Young Children and the NE-EHDI Program. The NE-EHDI program provides funds to administer the NCHALB and to purchase loaner hearing aids. Requests for additional funding were sent to potential funders. There were 42 hearing aids loaned to 24 young children (18 with a binaural fitting (two hearing aids) and six (6) with a monaural fitting (one hearing aid). Three children are waiting for cochlear implantation surgery.

Summary

- All the current birthing hospitals in Nebraska were conducting newborn hearing screening in 2009. All but one had conducted the hearing screenings during the birth admission.
- The benchmark of 95% of newborns having a hearing screening during birth admission by December 1, 2003 established by Neb. Rev. Stat. §71-4742 continues to be met. In 2009, birthing hospitals reported screening the hearing of 98.9% of newborns during birth admission or prior to discharge to home for those babies who were transferred to another hospital.
- The overall "refer" rate during 2009 for initial hearing screening during birth admission was 4.3%.
- In 2009, follow up hearing screenings or audiologic evaluations were initiated within one month of birth for 65.7% of those newborns for which follow up activities were provided.
- The average age at the time of the initiation of follow up hearing screening or diagnostic evaluation was 33.9 days.
- For the 48 infants identified with a permanent hearing loss, including congenital, later-onset and progressive hearing loss, the average age at confirmation of hearing loss was 138.3 days.
- There are 244 babies (0.9% of those born in 2009) whose hearing was not objectively established and there are 93 who expired before receiving or completing a hearing screening.
- The incidence of Permanent Congenital Hearing Loss identified and reported to NE-EHDI Program (1.7 per thousand screened in 2009) is within the anticipated range of 1 to 3 per thousand.
- Over 81% of the infants with a permanent congenital hearing loss were verified for early intervention and special education services.

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:

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Metabolic foods program, Translation & Distribution of Patient Education materials

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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:

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Ann Hirschfeld, Staff Assistant, EHDI Program (402) 471-3579
Follow up, patient education materials distribution, data management
Mike Rooney, Administrative Assistant (NNSP & EHDI) (402) 471-9731
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