Fiscal Year 2016 Blood Spot Newborn Screening
Follow Up Report
Short Term and Long Term Follow Up Activities

The Iowa Newborn Screening Program (INSP) is administered by the Iowa Department of Health (IDPH) in collaboration with the University of Iowa State Hygienic Laboratory (SHL) to provide testing and the Stead Department of Pediatrics at the University of Iowa Stead Family Children's Hospital to provide follow up services.

Iowa Newborn Screening Dried Blood Spot Program Report
Short Term and Long Term Follow Up Activities

The following report describes the purpose, processes and activities of the short term and long term follow up program component of the Iowa Newborn Screening Program. There is an appendix listing terms and definitions that readers may wish to refer to while reviewing this document. Program staff members are willing to answer any questions the reader might have. Contact information is provided at the end of the report.

Why Do Blood Spot Newborn Screening?

Blood spot newborn screening can detect disorders that are life threatening or life changing before an untoward event occurs. Babies can look and act perfectly healthy but still have one of these disorders. Sometimes, it's literally a matter of a few hours to a few days before tragedy can occur without newborn screening. It is estimated that 12,000 babies are positively impacted by newborn screening efforts in the United States each year. Newborn blood spot screening saves babies lives – it's as simple as that.

An Overview of the Laboratory and Clinical Process of Newborn Screening in Iowa

Local Hospital - At 24-48 hours of age, a few drops of blood are taken from a baby's heel to perform the newborn screening test. These drops are placed on a card that contains information about the baby, the mother, and the blood sample. This is called the dried blood spot card.

Courier - The card is picked up at the local hospital by a courier service and is driven to our newborn screening laboratory in Ankeny. Cards from throughout the state arrive in the laboratory around 9:00 pm each night (seven days a week).

Newborn Screening Laboratory - Once the card arrives in the lab, quality checks are performed and the data from the card is entered into a database by data entry staff. Laboratory staff start the testing process soon after the card arrives at the lab. A laboratory staff member calls and emails the short term follow up staff with any abnormal endocrine or metabolic results so that

immediate (and sometimes life-saving) action can occur. These results, along with testing results of other screened disorders, are entered into a database.

Short Term Follow Up/Medical Consultant – Short term follow up staff are informed of abnormal testing results. This is called a "presumptive positive" or "borderline" result. Presumptive positive means that the screening test for the disorder is abnormal and requires further action. It does not mean that the baby has that disorder. That is why the short term follow up component of the newborn screening program is crucial. Follow up staff help local care providers through the process of determining if a screening result is real (i.e., a "true positive") or a "false positive" (baby is not affected with a disorder/disease). Follow up staff inform the local hospital (if baby is still an inpatient) or local provider (if baby has gone home) of the abnormal results. Recommendations are provided to the primary care provider (PCP) verbally and then followed by a fax and/or email with the same information as per protocol. Education regarding the disorder that screened positive is also provided. A medical consultant (a MD who specializes in one of the disorders that we screen for) will also review abnormal results and assist staff and local providers when necessary. Sometimes it is recommended to repeat the newborn screen and/or to get additional specialized testing called confirmatory testing. The follow up staff review the tests recommended with local providers (and sometimes local laboratories too) and keep in touch with the PCP to make sure that these tests are obtained. Once the tests are obtained, follow up staff remain in communication with the local hospital or provider to obtain the results of further testing. Once these results are in, follow up staff review the results with the medical consultant to see if further action is necessary. Sometimes no further action is necessary and the case is closed as a "false positive". If the specialized testing is reviewed and does not appear to be normal, then a referral is made to a specialist so the baby can be further evaluated.

Long Term Follow Up – Referrals are made to specialized physicians and allied health care providers when a newborn screen is abnormal and/or a disorder is confirmed. Sometimes, the long term follow up staff recommend additional testing or decide to start treatment. For instance, if a baby is "presumptive positive" for PKU, a referral is made to a metabolic genetics center. Confirmatory testing is performed if not yet completed or the results of the confirmatory tests are reviewed. If it is determined that the baby has PKU, the parents are educated about the disorder and how to care for the child. This includes information not only about the disorder, but extensive education on how to manage the special diet required to treat this condition is given by the metabolic dietitian. The baby diagnosed with PKU will need to follow this special diet and will need to been seen by metabolic specialists for their lifetime.

Disorders Screened for in Iowa

AMINO ACIDEMIAS AND UREA CYCLE DISORDERS

- (ASA) Argininosuccinic aciduria*
- (CIT) Citrullinemia, type 1 or ASA Synthetase Deficiency*
- (HCY) Homocystinuria (cystathionine beta synthetase)*
- (MSUD) Maple Syrup Urine Disease*
- (PKU) Classic Phenylketonuria*
- (TYR-1) Tyrosinemia, type I*
- (ARG) Argininemia**
- (BIOPT-BS) Defects of biopterin cofactor biosynthesis**
- (CIT-II) Citrullinemia, type II**
- (BIOPT-REG) Defects of biopterin cofactor regeneration**
- (H-PHE) Benign hyperphenylalaninemia**
- (MET) Hypermethioninemia**
- (TYR II) Tyrosinemia, type II**
- (TYR III) Tyrosinemia, type III**

ORGANIC ACIDEMIAS

- (GA-1) Glutaric acidemia type I*
- (HMG) 3-Hydroxy 3-methylglutaric aciduria *
- (IVA) Isovaleric acidemia*
- (3-MCC) 3-Methylcrotonyl-CoA carboxylase*
- (Cbl-A,B) Methylmalonic acidemia (cobalamin disorders, vitamin B12 disorders)*
- (βKT) βeta-Ketothiolase*
- (MUT) Methylmalonic Acidemia (methylmalonyl-CoA mutase)*
- (PROP) Propionic acidemia*
- (MCD) Holocarboxylase synthase*
- (2M3HBA) 2-Methyl-3-hydroxybutyric aciduria**
- (2MBG) 2-Methylbutyrylglycinuria**
- (3MGA) 3-Methylglutaconic aciduria**
- (Cbl-C, D) Methylmalonic acidemia with homocystinuria**
- (MAL) Malonic acidemia**

FATTY ACID OXIDATION DISORDERS

- (CUD) Carnitine uptake defect (Carnitine transport defect)*
- (LCHAD) Long-chain L-3 hydroxyacyl-CoA dehydrogenase*
- (MCAD) Medium chain acyl-CoA dehydrogenase*
- (TFP) Trifunctional protein deficiency*
- (VLCAD) Very long-chain acyl-CoA dehydrogenase*
- (CACT) Carnitine acylcarnitine translocase**
- (CPT-Ia) Carnitine palmitoyltransferase type I**
- (CPT-II) Carnitine palmitoyltransferase type II**
- (GA2) Glutaric acidemia type II**
- (MCAT) Medium-chain ketoacyl-CoA thiolase**
- (M/SCHAD) Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase**

ENDOCRINE

- (CAH) Congenital adrenal hyperplasia *
- (CH) Primary Congenital hypothyroidism *

HEMOGLOBINOPATHIES

- (Hb SS) S,S Disease (Sickle Cell Anemia)*
- (Hb S/C) S,C Disease*
- (HB S/βTh) S, βeta-thalassemia*
- (Var Hb) Variant hemoglobinopathies **

OTHER

- (BIOT) Biotinidase deficiency *
- (CF) Cystic Fibrosis *
- (GALT) Classic Galactosemia *
- (GALE) Galactoepimerase deficiency **
- (HEAR) Hearing loss*

* Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) Recommended Uniform Screening Panel - Core Panel

** SACHDNC Recommended Uniform Screening Panel - Secondary Targets - Screening for the Core Panel of disorders may show information about secondary conditions (by-products of mandatory screening)

Disorders on the SACHDNC recommended panel that we do not screen for:
(DE-RED) 2,4 Dienoyl-CoA reductase deficiency**;(GALK) Galactokinase deficiency**; (SCAD) Short-chain acyl-CoA dehydrogenase**; (IBG) Isobutyrylglycinuria**, Pompe, Mucopolysaccharidoses I (MPS 1) and Adrenoleukodystrophy (ALD).

Screens Submitted (Calendar 2015)

There were 41,276 newborn screening cards submitted to the newborn screening laboratory for testing (39,303 initial screens and 1,973 repeat screens). This number includes more than one screen on some babies (repeat screens requested by follow up staff). The number of births would be slightly less than the number of screens submitted.

Borderline and Presumptive Positive Cases for CY 2015

Disorder	Borderline/ Indeterminate	Presumptive Positive
Biotinidase	N/A	16
Cystic Fibrosis	37	7
Endocrine Disorders		
Congenital Adrenal Hyperplasia	240	10

Congenital Hypothyroidism	456	17
Galactosemia	3	3
Hemoglobinopathies	N/A	7
Metabolic Disorders	N/A	445
Severe Combined Immunodeficiency	N/A	6

Confirmed Cases for Calendar 2015

Confirmed cases are counted in the year that they were confirmed, not necessarily the year the baby was born.

Primary Conditions

Newborn screening is designed to find disorders that are designated as primary disorders on the Recommended Uniform Screening Panel (RUSP). The confirmed cases listed below are only those disorders that have been designated as primary disorders on the RUSP. Most metabolic disorders are identified through tandem mass spectrometry, but two metabolic disorders are enzyme based tests. These are biotinidase deficiency and galactosemia. These two disorders are listed separately in this table. It should be pointed out that a very rare disorder, Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase (LCHAD), was detected in 2015. This is the first time that this disorder has been identified in the INSP. The incident rate is unknown because of the rarity of the disorder.

Disorder	**Cases Confirmed**
Biotinidase Deficiency (metab)	2
Cystic Fibrosis (CF)	6
Endocrine Disorders	
Congenital Adrenal Hyperplasia	0
Congenital Hypothyroidism	30
Galactosemia (metab)	3
Hemoglobinopathies	
Hgb S/vs Hgb S Beta Thalassemia	2
Hgb SC	1
Metabolic Disorders	
2 MBG	1
Argininosuccinic Aciduria (ASA)	1
Cobalamin C	1
LCHAD	1
MCAD	5
PKU	6
SCID	0
TOTAL	**59**

Secondary Conditions or Incidental Findings

Disorder	Number Confirmed
22q11.2 Deletion Syndrome	1
Carriers of Various Metabolic Disorders	4
Cystic Fibrosis Carriers	20
Cystic Fibrosis Related Metabolic Syndrome (CRMS)	5
Duarte Galactosemia	2
Hemoglobin EE	2
Hemoglobin H	1
Hemoglobinopathy Trait	477
Hyperphenylalinemia	2
Idiopathic T-Cell Lymphopenia	1
Transient Tyrosinemia	1
TOTAL	**516**

False Negative Case

When a baby is diagnosed with a disorder that we screen for, but the screening was reported as negative (normal), we refer to it as a "false negative". The program has to rely on individuals to report false negatives to the program because there is no other way to ascertain them. Often, we hear of these cases from the baby's PCP or through specialists. Although we strive to not have any false negatives, it does occur, especially in cystic fibrosis. The medical community knew before we started screening for cystic fibrosis that there would be a 10% false negative rate. The incidence of a false negative in cystic fibrosis increases when the baby has meconium ileus. This points out a very important fact; newborn screening is a *screening* test, not a diagnostic test.

We had 1 false negative case in 2015 for cystic fibrosis in a baby with meconium ileus.

Missed Screens – 1

One birthing facility failed to get a baby screened prior to discharge. The missed screen was discovered by the baby's PCP when she went to review NBS results with the parents at the 2 week routine check up. Baby was finally screened at about 4 weeks of life. The birthing facility was contacted and technical assistance and education was provided.

Lost to Follow Up/Against Medical Advice - 18

There were 18 babies who were categorized as lost to follow up or did not follow through with recommendations given to them by medical professionals and/or the newborn screening program. Thirteen babies were lost to follow up. Lost to follow up is when a baby cannot be

found by the baby's PCP or by the program (such as when a phone is disconnected and/or certified mail is returned) or when parents are contacted but do not bring the baby in for a recommended repeat screen or confirmatory testing despite multiple attempts to get them to do so. Seven of the 13 babies lost to follow up had an initial poor quality screen. See "Poor Quality Screens" later in this section. There were 5 babies that were "AMA" – against medical advice. These are situations where the PCP or program personnel have had an informed, educational conversation with the family about why they are recommending further testing and the parents still refuse after being appropriately counseled about what might happen if they do not do further testing.

Poor Quality Screens - 408 – 1.0%

There are various reasons that newborn screening samples are considered to be "poor quality". The sample could be contaminated, there isn't enough blood within the circles on the card, the blood spot is layered or clotted, the blood spot card has expired, etc. Screens are rejected when lab staff determines that the accuracy of the test results would be compromised for any of the reasons listed above.

Currently, the state percentage for poor quality samples is 1.0%. Compared to many other state programs, 1.0% is an acceptable number. However, the INSP would like the state average to be < 1.0%. The program provides technical assistance/education to reduce the percentage of poor quality screens in our state. It is imperative that a good quality screen be collected the first time. When it isn't, the baby's health could be impacted. Poor quality screens also increase the number of babies who never get a valid screen (see "lost to follow up" above). Finally, poor quality screens negatively impact timeliness in newborn screening.

Refusals -23 – Started tracking July 1, 2015

On July 1, 2015, the Iowa Department of Public Health instituted a new rule in the Iowa Code that reads as follows: that ITEM 6. Amend paragraph **4.3(2)"b"** as follows:

b. Refusal of screening. Should a parent or guardian refuse the screening, said refusal shall be documented in the infant's medical record, and the parent or guardian shall sign the refusal of screening form. The birthing facility or attending health care provider shall submit the signed refusal of screening form to the central laboratory within six days of the refusal. The birthing facility or attending health care provider may submit refusal forms via the courier service established for the transportation of newborn screening specimen collection forms.

It was decided that the refusal forms would be sent to NBS follow up staff for review because of other tracking activities done by the follow up program. There were 23 refusals from July 1, 2015 – December 31, 2015. The reasons listed on the form for refusals include: "don't see the necessity in doing the screen"," religious reasons", "cost" (for self pay patients), "my other kids are fine", and "too many false positives". We continue to do educational outreach to parents and providers so that they understand the importance of screening and the implications of not screening.

See "Quality Improvement" for information on a project with refusals.

Genetic Counseling

Genetic counseling is offered to all patients who have abnormal test results through the newborn screening process. The counseling is completed either face to face or over the phone. We hope to offer more genetic counseling through telehealth (like Skype) in the future.

Quality Improvement Activities (Fiscal Year 2016)

QI Officer

A new position for a Quality Improvement Officer was filled in 2015 by Ashley Comer, one of the NBS lab technologists. Although housed in the NBS lab, Ashley works with NBS personnel at the IDPH, SHL Coralville and follow up staff at UIHC to improve the quality of our program. One of the position's primary duties Ashley has is to interface and provide technical assistance to birthing centers and midwives across the state of Iowa.

Timeliness in Newborn Screening

The program's timeliness activities started in late 2014 when our NBS program was selected to participate in a non-funded Collaborative Improvement and Innovation Network (COIIN) to Improve Timeliness in Newborn Screening sponsored by the Health Resource Services Administration (HRSA)/Association of Public Health Laboratories (APHL). Participants in the COIIN included Stan Berberich from the SHL NBS lab; Kristen Ernsperger, nurse from Des Moines Mercy Medical Center; Carol Johnson, NBS follow up; Laura Malone, Iowa Hospital Association; and Kim Piper from IDPH. We determined our timeliness goal would be that 95% of NBS samples would arrive in the NBS laboratory =< 60 hours after birth. Throughout 2015, we developed and implemented a timeliness report that is sent to all birthing facilities and midwives. Program staff has also participated in several hospital visits and have spoken at various meetings to provide general NBS education as well as education on timeliness in NBS. In 2015, the Iowa Newborn Screening Program applied for and was granted funding from APHL/NewSTEPs 360 to continue work on timeliness in newborn screening.

Baby Matching

Baby matching is the process of "matching" a newborn screening result or an official NBS refusal form with birth certificates to make sure that all babies born in the state have been screened. Matching is completed by the IDPH and then a lists of babies who do not appear to have a NBS are sent to follow up staff to reconcile. If NBS results cannot be found, the birthing hospital is contacted to see if a screen was collected. If not, the baby's PCP is notified and asked to obtain the screen as soon as possible. Often we find that a baby has been screened in a bordering state. On occasion we find that a baby did not get screened. This process reduces the number of babies who do not get screened.

Refusals

The follow up staff member who does baby matching worked with our genetic counselor on a project to follow up on NBS refusals. The counselor would call the parents to talk with them about the NBS refusal. She would answer any questions that they had. We wondered if this one on one interaction where the parents could be fully educated about NBS would make parents reconsider and get their baby screened. Several admitted that they hadn't gotten any NBS education (whether this was the first baby or the fifth baby). Some said that if they would have received the education on NBS that was provided by the counselor, they probably would have made a different choice. However, only one parent changed their mind and got their baby screened. We stopped having the counselor make these calls as the benefit was not greater than the time and effort being put into the project.

Severe Combined Immunodeficiency

1) Newborn screening personnel from the laboratory and follow up continue to closely monitor results from SCID NBS. Staff meets quarterly to go over laboratory data as well as clinical data to make sure things continue to go well for the newest disorder added to our NBS panel.

2) Normative Values for Flow Cytometry – The University of Iowa is the only Flow Cytometry Laboratory in the state. When a baby is presumptive positive for SCID, the next step is to obtain a whole blood sample and send it to a flow cytometry laboratory for confirmatory testing. Unfortunately, there are no normative values established in flow cytometry for preterm or full term infants in the world. Therefore, the SCID medical consultant, Dr. Mary Beth Fasano, and our Flow Cytometry SCID medical consultant, Dr. Sergei Syrbu, are conducting research with colleagues in the University of Iowa Hospitals and Clinics Labor and Delivery Suite and the University of Iowa Blood Bank to determine what a normative value is on newborns at various gestational ages. Parents are consented to be a part of this study. Emily Phillips is the research coordinator for the project.

Case Closure-Metabolic

The metabolic short and long term follow up staff, as well as laboratory staff. meet once per month to go over all borderline/presumptive positive metabolic cases that have been identified during the month. During this meeting, cases are reviewed and discussed. We find that some cases can be closed and that some cases are still pending. Education on metabolic disorders is also provided from the clinical staff. This process allows us to ensure that cases are closed in a timely manner and that NBS cases are not lost during the follow up process.

Protocols and Educational Materials

Protocols and educational materials for each disorder are reviewed on a yearly basis, but can also be reviewed and altered on an as needed basis. Often, case experience brings an issue to light and changes to the protocols need to be made. Because this is a large undertaking (over 120 protocols and related educational material), follow up staff work on protocols all year long.

Database

The INSP is in the middle of switching over to an integrated database for the three newborn screening programs called the Iowa Newborn Screening Information System (INSIS). We continue to work on identifying needs as OZ Systems continues to build the blood spot newborn screening module. Weekly meetings have been occurring throughout this fiscal year. We have done one round of User Acceptance Training. We anticipate going live with this new database in December 2016.

Projects

NewSTEPs Repository

Confirmed cases are entered into this national repository by our follow up staff. At this time, NewSTEPs is still working on case definitions for hemoglobinopathies and SCID, so any confirmed cases of these disorders are not currently in this repository.

Region 4 Repository

Any confirmed metabolic or SCID cases are entered into this international repository by our follow up staff. This repository is also used as a tool for all presumptive positive metabolic cases. It gives a case score and lists all the possible metabolic conditions that could be associated with the metabolic analyte results.

Multidisciplinary Sickle Cell Comprehensive Clinic

Dr. Anjali Sharathkumar, along with Dr. Vasu Kailasnath, social worker Abby Yoder and psychologist Amanda Grafft from University of Iowa Children's Hospital have been working to establish a comprehensive sickle cell clinic for patients with hemoglobinopathies in the state of Iowa. The clinic was started mid FY 16 and takes place at the Iowa River Landing Clinic in Coralville, Iowa. The response has been positive so far. This multidisciplinary clinic is held once per month and includes MD medical consultation, genetic counseling (patient and group counseling and education), social work, and psychological services.

Education Presentations and Technical Assistance

The INSP provides education and technical assistance to NBS stakeholders throughout the state in a variety of ways. These include on site presentations, postings on the IDPH NBS list serve, webinars, mailings, phone calls, etc. We believe that education is the core activity for all other quality improvement initiatives. For a more in depth look at INSP educational activities provided by follow up staff, please see the educational activities list later in this report.

New Dried Blood Spot Card

Program staff has been working on revising the dried blood spot card. It is our hope that the changes being made will enhance the quality of the data collected on the card so that test interpretation will be more specific and identifying the baby's PCP will be easier. Changes planned include: 1) changing verbiage from "mother" to "guardian" in the next of kin field; adding a check box for meconium ileus (if present, baby likely has CF and the lab will go directly to DNA testing); revising the feeding method section, and changing/adding the physician information field to include the ability to list both the attending physician (person who ordered the NBS) and identifying the PCP who will care for the baby after discharge. Often, the name of the ordering physician is listed on the blood spot card, but the follow up staff need to interact with the baby's PCP for any abnormal screening results. We anticipate the new card will begin to be used approximately February 2017.

New Brochure

The Iowa Department of Public Health developed a new newborn screening brochure that encompasses the three newborn screening programs: blood spot screening, critical congenital heart disease screening, and hearing screening. The feedback has been positive from staff in birthing facilities; they like having all the newborn screening information in one brochure.

Facility Data Report

The INSP is working on revising the facility data report. This is a quality report sent to all birthing facilities. It includes information such as their poor quality rate, how many early collections there were (< 24 hours), and other quality indicators. We are working to include information on how well the card was filled out upon arrival in the NBS lab and also sharing information in a generic way about the confirmed cases that have been identified through NBS.

Federal Express vs Certified Letter

Sometimes, local PCP's and the INSP are unable to reach a family when there are outstanding recommendations because of abnormal newborn screening test results. When this happens, a certified letter is sent to the mother explaining that she needs to contact her baby's PCP and make arrangements to come in for further testing. Follow up staff had received feedback that families tend to ignore certified letters as certified letters have the connotation of containing "bad news", usually financial or legal issues. Follow up staff tried an experiment to see if using Federal Express to send these letters resulted in better follow through from families in general, and if that follow through was more timely than what is observed with a certified letter. Overall, we only had one case where sending the letter via Federal Express made things happen in a more timely fashion. Therefore, we are still using certified mail.

Other Projects and Relevant Information

Inborn Errors of Metabolism Collaborative Research Project – this is a natural history, consented patient registry for those patients with metabolic conditions. Emily Phillips is the research coordinator.

Clinical Trials/Patient Registries – the metabolic and lysosomal storage clinical teams participate in various clinical trials being conducted at the University of Iowa Children's Hospital for these disorders.

Educational Activities

Name	Date(s)	Presentation Title	Type	Name of Meeting	Location of Meeting	Attendee Only
John Bernat	2/29-3/4/16			WORLD 2016 Lysosomal Meeting	San Diego, CA	X
Natalie Dennler	11/4/15		Vendor	University of Iowa Health Fair	Iowa City, IA	
	2/8/16			Pregnancy Centering Group	Iowa River Landing Clinic, Coralville, IA	
	2/29-3/3/16			Newborn Screening and Genetic Testing Symposium	St. Louis, MO	X
	4/25/16			Pregnancy Centering Group	Iowa River Landing Clinic, Coralville, IA	
Fasano, Mary Beth	11/5-9/15			American College of Allergy, Asthma, and Immunology Annual Meeting	San Antonio, TX	X

	1/22/16	"When Eczema Isn't Just Eczema: How Not to Miss Immune Disorders" (included SCID)	Presenter	Clinical Symposia – Advances in Dermatology	Naples, FL	
	3/4-7/16			American College of Allergy, Asthma, and Immunology Annual Meeting	Los Angeles, CA	
	5/19/16			North Dakota Newborn Screening Advisory Committee	Bismarck, ND	X
	5/20/16	"Newborn Screening for SCID – What Iowa Has Learned So Far"	Presenter	North Dakota Newborn Screening SCID Conference	Bismarck, ND	
Hatland, Tammy	10/7/15		Vendor	Neonatal Update Conference	Marriott, Coralville, IA	
	11/4/15		Vendor	University of Iowa Health Fair	Iowa City, IA	
	2/29-3/3/16			Newborn Screening and Genetic Testing Symposium	St. Louis, MO	X

Hobert-Mellecker, Melody	10/6/15		Vendor	Children and Women's Services Fall Conference, sponsored by the University of Iowa	Holiday Inn, Coralville, IA	
	11/6/15	"Together-Saving Babies One Foot at a Time"	Presenter with Ramirez	Northwest Iowa Regional Best Practices in Newborn Screening Conference	Sheldon, IA	
	11/30/15	"Together – Saving Babies Lives One Foot at a Time"	Presenter	Great Rivers Medical Center	Burlington, IA	
	12/17/15		Presenter	Pregnancy Centering Group	Iowa River Landing Clinic, Coralville, IA	
	2/29-3/3/16			Newborn Screening and Genetic Testing Symposium	St. Louis, MO	X

	5/19/16			North Dakota Newborn Screening Advisory Committee	Bismarck, ND	X
	5/20/16			North Dakota Newborn Screening SCID Conference	Bismarck, ND	X
	5/22-29/16		Trainee	APHL Tandem Mass Spectrometry Workshop	Silver Spring, MD	X
Holida, Myrl	2/29-3/4/16			WORLD 2016 Lysosomal Meeting	San Diego, CA	X
	3/25/2016	"Lysosomal Storage Diseases"	Presenter	Grand Rounds, Dept. of Pediatrics	UIHC, Iowa City, IA	
Johnson, Carol (National)	10/7/2015	"Barriers to Timeliness in Newborn Screening"	Presenter	Quality Improvement Special Interest Group, North American Cystic Fibrosis Foundation Symposium	Phoenix, AZ	

	11/2/2015	"Barriers to Timeliness in Newborn Screening"	Presenter	Advisory Committee to HHS Secretary on Newborn Screening and Genetic Testing, Subcommittee on Timeliness in Newborn Screening	Webinar	
	2/29-3/3/16	"Collaborative Improvement and Innovation Network for Timeliness in Newborn Screening – the Iowa Experience to Date"	Poster	Newborn Screening and Genetic Testing Symposium	St. Louis, MO	
	2/29/2016	Roundtable Discussion "Short Term Follow Up Processes in Newborn Screening"	Facilitator	Newborn Screening and Genetic Testing Symposium	St. Louis, MO	
	3/1/2016	Short Term Follow Up Mixer – Facility Reports	Facilitator	Newborn Screening and Genetic Testing Symposium	St. Louis, MO	

	6/21-22/16	"Barriers to Timeliness in Newborn Screening"	Presenter	Cystic Fibrosis Newborn Screening Quality Improvement and Timeliness Meeting, APHL/NACFF	Denver, CO	
	6/23/2016	"Improving Efficiency in Newborn Screening Collection and Test Results"	Presenter	Robert Wood Johnson Foundation	Webinar	
Regional	5/1/15	"Timeliness in Newborn Screening and the COIIN Initiative"	Presenter with Kim Piper	Heartland Regional Genetics Collaborative Annual Meeting	Kansas City, MO	
	2/24/16	"Heartland COIIN for Timeliness in Newborn Screening"	Presenter with Kim Piper and Ashley Comer	Heartland Regional Collaborative Newborn Screening Subcommittee	Webinar	
	5/5-6/16	Heartland Timeliness Workshop	Presenter/ Facilitator with Berberich, Comer, Piper	Kansas City Airport Marriott	Kansas City, MO	
	5/19/16			North Dakota Newborn Screening Advisory Committee	Bismarck, ND	X

	5/20/16	"Newborn Screening Follow Up – What's it All About?"	Presenter	North Dakota Newborn Screening SCID Conference	Bismarck, ND	
State/Local	7/15/15 7/20/15	"Together – Saving Babies Lives One Foot at a Time"	Presenter	St. Luke's Unity Point Medical Center	Cedar Rapids, IA	
	9/22/2015	"COIIN Timeliness in Newborn Screening – The Iowa COIIN Initiative"	Presenter with Stan Berberich	St. Luke's Unity Point Medical Center COIIN Team	Cedar Rapids, IA	
	11/10/15		Vendor	Iowa Healthcare Collaborative Meeting	Altoona, IA	
	11/30/15	"Together – Saving Babies Lives One Foot at a Time"	Presenter with Melody Hobert-Mellecker	Great Rivers Medical Center	Burlington, IA	
	1/13/16	"Timeliness in Newborn Screening for Birthing Centers – Iowa's COIIN Project"	Presenter with Berberich, Comer, Piper	Birthing Facilities and Midwives throughout Iowa	Webinar	

	4/6/16		Vendor	Iowa Statewide Perinatal Conference	Des Moines, IA	
	4/12/16	"COIIN Timeliness in Newborn Screening "	Presenter with Berberich, Comer, MPH student	Mercy Medical Center	Dubuque, IA	
	4/12/16	"COIIN Timeliness in Newborn Screening "	Presenter with Berberich, Comer, MPH student	Finley Medical Center	Dubuque, IA	
	4/15/16	"COIIN Timeliness in Newborn Screening "	Presenter with Berberich, Comer, MPH student	Mercy Hospital	Iowa City, IA	
	4/21/16	"The Iowa Newborn Screening Program – Saving Babies One Foot at a Time"	Presenter	Iowa Leadership Education in Neurodevelop-mental and Related Disabilities	Iowa City, IA	

Marcy, Jennifer	7/1/2015-6/30/2016	Genetic counseling for patients/families being seen for CF sweat testing	Counselor	Cystic Fibrosis Center	Pediatric Specialty Clinic, University of Iowa Children's Hospital	Iowa City, IA
	7/1/2015-6/30/2016	Genetic counseling for patients/families being seen for positive confirmatory testing for SCID	Counselor	Pediatric Immunology Clinic	Pediatric Specialty Clinic, University of Iowa Children's Hospital	Iowa City, IA
	7/1/2015-6/30/2016	Genetic counseling for patients with a confirmed disorder screened for by the INSP	Counselor	Telephone	Office	Iowa City, IA
	February 2016	Genetic counseling and disorder education for patients with hemoglobinopathies; single counseling and educational sessions along with group counseling and educational sessions	Presenter/ counselor	Multidisciplinary Sickle Cell Clinic	Iowa River Landing clinic, University of Iowa, Coralville, IA	

	2/29-3/3/16			Newborn Screening and Genetic Testing Symposium	St. Louis, MO	X
Miller, Judy	July 2015	"The Use of Kuvan in the Treatment of PKU"	Organizer, presenter	PKU Low Protein Cooking Event	Hotel Kirkwood, Cedar Rapids, IA	
	8/4-5/15			Heartland Telemedicine Training	Little Rock, AK	X
	4/6/2016		Organizer	Low Protein Grocery Store Event	Hy-Vee Grocery Store, East Des Moines, IA	X
	4/27-30/16			Genetics Metabolic Dietitians International	Phoenix, AZ	X
	5/14/2016		Organizer	PKU Family Event (sponsored by BioMarin)	Blank Park Zoo, Des Moines, IA	X
	6/21/2016		Organizer	Low Protein Grocery Store Event	Hy-Vee Grocery Store, Cedar Rapids, IA	X
Norris, Andrew		Nothing to report				

Phillips, Emily	7/20/15	"Together – Saving Babies Lives One Foot at a Time"		St. Luke's Unity Point Medical Center	Cedar Rapids, IA	X
	8/2/15		Vendor – Importance of NBS	Corridor Parent Expo	Hotel Kirkwood, Cedar Rapids, IA	
	8/19/15 11/18/15 6/1/16	"NICU Nursing Orientation for Newborn Screening"	Presenter	UIHC NICU	Iowa City, IA	
	8/19/15 8/31/15 10/23/15 11/19/15 1/13/16 2/10/16 3/9/16 4/7/16 5/4/16	"The Importance of Newborn Screening and Timeliness and How to Collect a Screen"	Presenter	Newborn Screening Education for Residents Prior to NICU/Nursery Rotations	Iowa City, IA	
	8/22/15		Vendor	Iowa State Fair	Des Moines, IA	
	9/9/15 11/9/15		Presenter	Pregnancy Centering Group	Iowa River Landing Clinic, Coralville, IA	

	9/21/15 11/2/15 1/11/16 2/22/16 4/4/16 5/16/16	"The Importance of Newborn Screening and Timeliness and How to Collect a Screen"	Presenter	Newborn Screening Education for Medical Students Prior to NICU/Nursery Rotations	Iowa City, IA	
	10/26/15		Vendor	Association of Women's Health, Obstetrics and Neonatal Nurse State Conference	Des Moines, IA	
	11/4/15		Vendor	University of Iowa Health Fair	Iowa City, IA	
	4/5/16		Vendor	Iowa Statewide Perinatal Conference	Des Moines, IA	
	5/19/16			North Dakota Newborn Screening Advisory Committee	Bismarck, ND	X
	5/20/16			North Dakota Newborn Screening SCID Conference	Bismarck, ND	X

Serrano Russi, Alvaro	10/8/15	"Metabolic Disease and Emergencies"	Presenter	Department of Pediatrics Residency Program	UIHC Iowa City, IA	
	12/17/15	"Inborn Errors of Metabolism Associated with Critical Illness"	Presenter	Pediatric Intensive Care-Fellow Core Curriculum	UIHC Iowa City, IA	
	1/21/15; 3/25/15; 4/15/15; 1/26/16	"Newborn Screening Overview"	Presenter	Educator Early Clinical Experience for First Year Medical Students	UIHC Iowa City, IA	
	2/29-3/3/16	"Glutaric Acidemia Type II and LCHAD Deficiency Cases Identified by Newborn Screening Short Term Outcome"	Poster	Newborn Screening and Genetic Testing Symposium	St. Louis, MO	X
		"Two Year Clinical Follow Up of a Patient with Methylbutyrylglycinuruia Identified by Newborn Screening"	Poster	Newborn Screening and Genetic Testing Symposium	St. Louis, MO	X
	4/3-6/16	"Incidental Medulloblastoma in Adolescent with Known Glutaric Acidemia Type 1"	Poster	Society of Inborn Errors of Metabolism	Ponte Vedra Beach, FL	X

Sharathkumar, Anjali	October 2015			Heartland Sickle Cell Network Annual Meeting	St. Louis, MO	X
	October 2015			Meeting with the Washington University Sickle Cell Team and Clinic	St. Louis, MO	X
Starner, Timothy	10/8-10/15			North American Cystic Fibrosis Foundation Meeting	Phoenix, AZ	X
	6/21-22/16			Cystic Fibrosis Newborn Screening Quality Improvement and Timeliness Meeting	Denver, CO	X
Stimson, Cheryl	July 2015		Organizer	PKU Low Protein Cooking Event	Hotel Kirkwood, Cedar Rapids, IA	X

	4/6/2016		Organizer	Low Protein Grocery Store Event	Hy-Vee Grocery Store, East Des Moines, IA	X
	4/27-30/16			Genetics Metabolic Dietitians International	Phoenix, AZ	X
	5/14/2016		Organizer	PKU Family Event (sponsored by BioMarin)	Blank Park Zoo, Des Moines, IA	X
	6/21/2016		Organizer	Low Protein Grocery Store Event	Hy-Vee Grocery Store, Cedar Rapids, IA	X
	6/30/2016		Co-Author, Chapter	Simplified Diet Manual on PKU		

Committees

Name	Committee Name	Role
Bernat, John	Management of the Iowa Newborn Screening Panel Subcommittee	Member
	Center for Congenital and Inherited Disorders Advisory Committee (CIDAC)	Ex-Officio

Hatland, Tammy	Newborn Screening Information System Subcommittee	Member
Hobert-Mellecker, Melody	North Dakota Newborn Screening Advisory Committee	Member
	Management of the Iowa Newborn Screening Panel Subcommittee	Member
	Heartland Genetics Collaborative Newborn Screening Workgroup	Member
Holida, Myrl	Center for Congenital and Inherited Disorders Advisory Committee	Ex-Officio
Johnson, Carol	APHL/New STEPs Short Term Follow Up Workgroup	Co-Chair
	NewSTEPs/Cystic Fibrosis Foundation Special Interest Group on Improving NBS and Timeliness for Cystic Fibrosis	Member
	APHL NBS Steering Committee	Member
	Heartland Genetics Collaborative Newborn Screening Workgroup	Member
	North Dakota Newborn Screening Advisory Committee	Member
	Center for Congenital and Inherited Disorders Advisory Committee	Ex-Officio
	Management of the Iowa Newborn Screening Panel Subcommittee	Member
	Newborn Screening Information System Subcommittee	Chair
	CIDAC Informed Consent Subcommittee	Member
	CIDAC Use and Retention of Dried Blood Spot Subcommittee	Member

	Newborn Screening COIIN for Timeliness Team	Member
	Iowa NBS Executive Committee	Member
Marcy, Jennifer	Management of the Iowa Newborn Screening Panel Subcommittee	Chair
Phillips, Emily	CIDAC Informed Consent Subcommittee	Member
	Management of the Iowa Newborn Screening Panel Subcommittee	Member
Serrano-Russi, Alvaro	Center for Congenital and Inherited Disorders Advisory Committee	Ex-Officio
	Management of the Iowa Newborn Screening Panel Subcommittee	Member
Sheffield, Val	Center for Congenital and Inherited Disorders Advisory Committee	Member
	Iowa NBS Executive Committee	Member
Starner, Timothy	NewSTEPs/Cystic Fibrosis Foundation Special Interest Group on Improving NBS and Timeliness for Cystic Fibrosis	Member

Program Personnel – Follow Up Services (Fiscal Year)

Name	Title	Disorder	Location
Bernat, John	MD Medical Consultant	Lysosomal Storage Disorders	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
DeBoer, Pamela		Metabolic Disorders	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Dennler, Natalie	Case Manager, Short Term Follow Up	Iowa	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Endocrinologist On Call – Vanessa Curtis, Katie Larsen Ode, Andrew Norris, Liuska Pesce, Michael Tansey, Eva Tsalikian	MD On Call	Endocrine Disorders (Congenital Adrenal Hyperplasia; Congenital Hypothyroidism)	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Fasano, Mary Beth	MD Medical Consultant	SCID	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Ferguson, Polly	MD Medical Consultant (back up to Dr. Fasano)	SCID	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA

Genetics On Call – John Bernat, Alvaro Serrano-Russi, Val Sheffield, Pamela Trapane	MD On Call	Primarily metabolic disorders, but the geneticist on call triages all incoming NBS calls after hours, weekends, and holidays	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Hatland, Tammy	Case Manager, Short Term Follow Up	Iowa	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Hobert-Mellecker, Melody	Case Manager, Short Term Follow Up	North Dakota	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Holida, Myrl	PA, Long Term Follow Up	Lysosomal Storage Disorders	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Johnson, Carol	Program Administrator and Short Term Follow Up	Iowa	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Kremer, Courtney	ARNP, Long Term Follow up	SCID	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Marcy, Jennifer	Genetic Counselor, Short Term and Long Term Follow Up	Counselor for all disorders; Short Term Follow Up for Cystic Fibrosis and Hemoglobinopathies along with Case Managers	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA

Miller, Judy	ARNP, Long Term Follow Up	Metabolic Disorders	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Norris, Andrew	MD Lead Medical Consultant	Endocrine Disorders (Congenital Adrenal Hyperplasia; Congenital Hypothyroidism)	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Phillips, Emily	Case Manager, Short Term Follow Up	South Dakota	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Ramsey, Laura	ARNP, Long Term Follow Up	Cystic Fibrosis	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Rumelhart, Stephen	PA, Long Term Follow Up	SCID	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Serrano-Russi, Alvaro	MD Medical Consultant	Metabolic Disorders	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Sharathkumar, Anjali	MD Medical Consultant	Hemoglobinopathies	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA

Sheffield, Val	Medical Director		Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Starner, Timothy	MD Medical Consultant	Cystic Fibrosis	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Stimson, Cheryl	Dietitian, Long Term Follow Up	Metabolic Disorders	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA
Syrbu, Sergei	MD Medical Consultant	SCID, Flow Cytometry	Stead Family Department of Pediatrics, University of Iowa Hospitals and Clinics Iowa City, IA

Although Short Term Follow Up Case Managers have a primary state assignment, they are cross trained to do follow up activities in all three states. Case Managers cover all three states when they are on call on weekends and holidays. In addition, the Case Mangers work on Iowa NBS projects, serve on Iowa NBS committees, and provide education for the Iowa NBS program.

We had two new medical consultants join us during FY16. Dr. Anjali Sharathkumar has replaced Dr. Natalie Kamberos as the medical consultant for hemoglobinopathies. Dr. Timothy Starner has replaced Dr. Miles Weinberger as the medical consultant for cystic fibrosis. Dr. Weinberger retired in FY 16 after serving as the medical consultant for cystic fibrosis since it was added to the NBS panel in Iowa. Laura Ramsey, ARNP, replaces Elizabeth Dowd, ARNP, as the long term follow up nurse for cystic fibrosis. Elizabeth retired in FY16. Like Dr. Weinberger, Elizabeth served as the long term follow up nurse for cystic fibrosis since cystic fibrosis was added to the Iowa NBS panel.

Challenges

The biggest challenge facing newborn screening nationally and in Iowa is the rapid addition to disorders on the RUSP. In addition, the newest disorders added to the RUSP have both congenital and late onset presentations (anywhere from 3 months of life to 50 years of age). Finding the right balance between the public health perspective vs an individual perspective is a challenge.

Another challenge is educating the stakeholders in newborn screening about newborn screening. Many of the stakeholders, such as parents, don't realize that they have a role to play in the newborn screening process. A goal is to educate parents in the prenatal period rather than in the post-delivery setting or when an abnormality is found. Primary care providers are also stakeholders in the newborn screening process. As we know, PCPs are very busy people. What is the best way to educate PCPs throughout the state? How do you educate their staff, who are often the person you talk to when calling with abnormal NBS results? Midwife education is a particular challenge. Midwives who are not affiliated with a birthing hospital are out there on their own, doing their own thing. Many do not realize important issues related to NBS, particularly how important timeliness in NBS is.

Contact Information

Iowa Newborn Screening Follow Up	319-384-5097 iowanewbornscreening@uiowa.edu
Carol Johnson Iowa Newborn Screening Follow Up Coordinator	319-356-7248 carol-johnson@uiowa.edu

Appendix A

Terms and Definitions Used in Newborn Screening and in this Report

Against Medical Advice – Refers to a situation where medical advice is not followed by a patient/parent/guardian despite being educated about why it is important and the ramifications of not following medical advice

Amino Acid Disorders – Babies born with one of these disorders cannot process certain amino acids in their body. The amino acids, along with other toxic substances, build up in the body and cause serious effects on health, growth and learning. Treatment may include a special diet for life, close monitoring and/or vitamin and amino acid supplements. An example of an amino acid disorder is phenylketonuria (PKU). Babies with PKU cannot process a substance called phenylalanine. Left untreated, phenylalanine builds up in the bloodstream and causes brain damage, intellectual disability, depression, and other problems. If PKU is detected early and the special diet is started by Day 10 of life, these problems can be greatly reduced or prevented. PKU occurs in about 1 in every 12,000 births.

Baby Matching – A term used in newborn screening where a birth certificate is "matched" with a newborn screening result or an official NBS refusal form to make sure that all babies born in the state have been screened.

Beta Thalassemia – *Beta thalassemia major* usually causes severe anemia that can occur within months after birth. If left untreated, severe anemia can result in insufficient growth and development, as well as other common physical complications that can lead to a dramatically decreased life-expectancy. Fortunately, in developed countries beta thalassemia is usually identified by screening in the newborn period, before symptoms have developed. Children who are identified early can be started on ongoing blood transfusion therapy as needed. Although transfusion therapy prevents many of the complications of severe anemia, the body is unable to eliminate the excess iron contained in the transfused blood. Over time, the excess iron deposits in tissues and organs, resulting in damage and organ failure. Another medication must be administered to help the body eliminate the excess iron and prevent iron-over-load complications. *Beta thalassemia intermedia* describes the disease in individuals who have moderate anemia that only requires blood transfusions intermittently, if at all

Biotinidase Deficiency – Babies with biotinidase deficiency cannot reuse the vitamin biotin. Biotin helps maintain the normal body functioning. Without treatment, this disorder can lead to seizures, developmental delay, eczema and hearing loss. Biotin has to be added to the diet for treatment of this disorder. This disorder occurs in about 1 in every 60,000 births.

Borderline - a term used for some newborn screening disorders where the results are not normal, but are not high enough to be considered presumptive positive. A repeat screen on the baby is requested when the results are "borderline".

Card – a card/form that contains circles with filter paper to deposit the blood from the baby's heel on. This card also contains demographic information regarding the baby, mother, and sample information. Also called the "dried blood spot card".

Carrier – a person that has inherited a genetic trait or mutation but has no symptoms of the disease

Confirmatory/Second Tier Testing – specific testing that is recommended and performed post newborn screening to determine if a baby has a specific disorder or not, e.g. a sweat test for cystic fibrosis.

Confirmed – used to convey that the newborn screen and/or confirmatory testing determined that a baby had a disorder.

Congenital – a condition or problem present at birth.

Congenital Adrenal Hyperplasia – Babies born with this disorder have adrenal glands that cannot make enough of the hormone cortisol, and sometimes not enough of the hormone aldosterone. Sometimes this disorder affects the development of the genitals. You treat this disorder by taking medication that replaces the hormones that are deficient or eliminating the source of excess hormones. Without treatment, severe cases of this disorder can cause death. This disorder occurs in about 1 in every 16,000 births.

Congenital Hypothyroidism – Babies with this disorder are born with a thyroid gland that does not make enough thyroid hormone. This can lead to poor growth and abnormal brain development. If it is detected in time, a baby can be treated with medication. This disorder occurs in about 1 in every 4,000 births.

Courier – the contractual entity that travels to Iowa birthing facilities on a daily basis to pick up newborn screening cards and delivers them to the newborn screening laboratory.

CRMS - When a person has a sweat test that gives an intermediate (borderline) result or a genetic test that shows only one CF gene, he or she is said to have CFTR-related metabolic syndrome (CRMS). People with CRMS can be at a higher risk of having problems in the airways and sinuses; the intestines and pancreas; or the reproductive tract.

Cystic Fibrosis – Cystic fibrosis (CF) is the most common inherited (genetic) disorder, affecting about 30,000 children and adults in the US. A defective gene causes lung infections and digestive problems with malnutrition. CF can be life-shortening[5]. It's important to diagnose CF early, so that CF health care providers can help parents learn ways to keep their child as healthy as possible and delay problems related to CF. Research shows that children who receive CF care early in life have better nutrition and are healthier than those who are diagnosed later. Good nutrition in CF is important for overall health and well-being.

Early Collection – the newborn screen was obtained prior to 24 hours of age. The newborn screen is not valid if collected before 24 hours of age. A repeat screen will be requested on the baby by program staff.

False Negative – a term used when the newborn screen was negative, but a baby is found to have a disorder that we are screening for. As stated above, the newborn screen is a screening test, not a diagnostic test. Every attempt is made to reduce the number of false negatives, but it is understood that some cases will be missed. It is an inherent part of newborn screening.

False Positive – the newborn screen was positive for a particular disorder, but further testing was negative for the disorder.

Fatty Acid Oxidation Disorders – Babies with fatty acid disorders are unable to breakdown stored fats for energy. People who have this disorder cannot fast, and need prompt medical intervention when they have the stomach flu, fevers, etc. One example of a fatty acid disorder is Medium Chain acyl-CoA Dehydrogenase Deficiency (MCAD). Babies born with MCAD cannot break down fat into energy because an enzyme is missing or does not work correctly. People with MCAD should not fast (go without food) for very long or they can experience low blood sugar, seizures, coma and even death. MCAD occurs in about 1 in every 12,000 births.

Galactosemia – Babies with this disorder cannot convert galactose, a sugar present in milk, into glucose, a sugar the body uses as an energy source. Galactosemia can cause death in infancy, or blindness and intellectual disability. A baby will this disorder is not able to drink milk and/or eat other dairy products. They have to drink special formula and follow a special diet for their lifetime. This disorder occurs in about 1 in every 70,000 births.

Hemoglobin E Disease - is an inherited blood disorder characterized by an abnormal form of hemoglobin, called hemoglobin E. People with this condition have red blood cells that are smaller than normal and have an irregular shape. It is thought to be a benign condition. The mutation that causes hemoglobin E disease has the highest frequency among people of Southeast Asian heritage (Cambodian, Laotian, Vietnamese and Thai). However, it is also found in people of Chinese, Filipino, Asiatic Indian, and Turkish descent.

Hemoglobin H Disease - Hemoglobin H disease is a relatively mild form of thalassemia that may go unrecognized. It is not generally considered a condition that will reduce one's life expectancy. Transfusions are rarely needed in this disorder, except in a variant of this disorder called constant Spring. Occasionally additional medication is required for treatment.

Hemoglobinopathies – Hemoglobinopathies are inherited red blood cell disorders. Hemoglobin is the protein in the blood that carries oxygen from the lungs to the body. The most common hemoglobin disorder is sickle cell disease. When sickle cell shaped cells block small blood vessels, less blood can reach that part of the body. Sickle cell anemia occurs in about 1 in every 375 African Americans.

Iowa Department of Public Health – state agency that administers and oversees newborn screening processes in Iowa.

Long Term Follow Up - fundamentally, long-term follow-up comprises the assurance and provision of quality chronic disease management, condition-specific treatment, and age-appropriate preventive care throughout the lifespan of individuals identified with a condition included in newborn screening. Integral to assuring appropriate long-term follow-up are activities related to improving care delivery, including engagement of affected individuals and their families as effective partners in care management, continuous quality improvement through the medical home, research into pathophysiology and treatment options, and active surveillance and evaluation of data related to care and outcomes.

Lost to Follow Up – refers to situations where the baby cannot be located (moved with no forwarding address, guardian doesn't respond to phone calls or certified letters) or when the guardian is contacted about further testing but doesn't bring the baby in for further work up.

Medical Consultant – A physician who makes medical recommendations for a specific disorder to the newborn screening program, state health department, and health care providers throughout the state. They may also assist with development of protocols and provide education.

Newborn Screening Laboratory - The newborn screening laboratory is part of the State Hygienic Laboratory at the University of Iowa (Ankeny campus). This is the laboratory where the testing is performed.

Organic Acidemia – Babies born with organic acid disorders have a chemical imbalance in their bodies which can be toxic. Organic acids play an important role in the breakdown of fats, sugars and protein for the body's use and storage. Muscle wasting, seizures, developmental delays and even death can occur if untreated. Treatment may include a special diet, monitoring and medications.

Outcome – the final determination of a newborn screen, such as "confirmed, false positive, false negative, etc.

Poor Quality – a term used to describe that the sample was not able to be tested. A sample is called "poor quality" when the blood does not soak through the filter paper layers, when the sample is clotted, when too much blood is placed on the card, etc. A repeat screen will be requested by program staff.

Presumptive Positive – a term used by the laboratory and follow up personnel to identify a screen that was positive. The term "presumptive" is used because until further testing is done,

the result is considered positive until the disorder is confirmed or determined to be a false positive.

Primary Care Provider/Local Care Provider – also known as "PCP". The physician who is taking care of the baby, or is listed as the baby's physician.

Rejected Sample – similar to early collection and poor quality determinations. This term is usually used in association with a screen that was submitted after the 30 day cut off time frame. It is also used when the screening card does not have enough information recorded on it to determine who the baby really was.

Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) - The committee advises the Secretary, U.S. Department of Health and Human Services on the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and standards. Specifically, the committee provides to the Secretary, the following: Advice and recommendations concerning grants and projects authorized under the Heritable Disorders Program administered by the Health Resources and Services Administration; technical information to develop Heritable Disorders Program policies and priorities will enhance the ability of the state and local health agencies to provide screening, counseling and health care services for newborns and children who have or are at risk for heritable disorders; and recommendations, advice and information to enhance, expand or improve the ability of the Secretary to reduce mortality and morbidity from heritable disorders in newborns and children. The committee was chartered in February 2003.

Short Term Follow Up - refers to the process of ensuring that all newborns are screened, that an appropriate caregiver is informed of results, that repeat testing on a new specimen or confirmatory testing has been completed, and that the infant has received a diagnosis and, if necessary, treatment.

Sickle Cell Disease/Trait – Sickle cell anemia is caused by an abnormal type of hemoglobin called hemoglobin S. Hemoglobin is a protein inside red blood cells that carries oxygen. Hemoglobin S changes the shape of red blood cells. The red blood cells become shaped like crescents or sickles.The fragile, sickle-shaped cells deliver less oxygen to the body's tissues. They can also get stuck more easily in small blood vessels, as well as break into pieces that can interrupt healthy blood flow. These problems decrease the amount of oxygen flowing to body tissues even more. Sickle cell anemia is inherited from both parents. If you inherit the sickle cell gene from only one parent, you will have sickle cell trait. People with sickle cell trait do not have the symptoms of sickle cell anemia. Sickle cell disease is much more common in people of African and Mediterranean descent. It is also seen in people from South and Central America, the Caribbean, and the Middle East. About 90,000-100,000 residents of the US have sickle cell disease. One in every 500 blacks/African American's have disease. One in every 36,000 Hispanics have sickle cell disease. One in every 12 blacks/African American's have sickle cell trait.

Tandem Mass Spectrometry (MS/MS) - Tandem mass spectrometry, also known as MS/MS or MS2, involves multiple steps of mass spectrometry selection, with some form of fragmentation occurring in between the stages. In a tandem mass spectrometer, ions are formed in the ion source and separated by mass-to-charge ratio in the first stage of mass spectrometry (MS1). Ions of a particular mass-to-charge ratio (precursor ions) are selected and fragment ions (product ions) are created by collision-induced dissociation, ion-molecule reaction, photodissociation, or other process. The resulting ions are then separated and detected in a second stage of mass spectrometry (MS2). This is the technology used for most metabolic disorders.

Trait – a distinct, observable change in a person that might be inherited, such as sickle cell trait which can possibly be determined by newborn screening. It is not true sickle cell disease.

Unsatisfactory Specimen – a term used to state that there was not enough blood placed on the card to perform the newborn screen.