MISSOURI NEWBORN SCREENING

2006 Annual Report



Missouri Department of Health and Senior Services

Jane Drummond, Director

Acknowledgments

The Missouri State Genetic Advisory Committee and its ancillary Newborn Screening Standing Committee play a vital role in supporting the activities of the Missouri Department of Health and Senior Services Newborn Screening Program.

The expertise the board provides is complemented by department staff who are motivated to help Missouri children receive the best care available when diagnosed with one of the serious medical conditions detectable through screening tests.

Missouri Department of Health and Senior Services
Division of Community and Public Health
Section for Healthy Families and Youth
Bureau of Genetics and Healthy Childhood
and
State Public Health Laboratory

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Missouri expanded its blood spot screening program in 2005 from 5 conditions to 27 conditions screened in newborns. In 2007, cystic fibrosis was added to the newborn screening panel.

In the coming year, biotinidase deficiency will be added to the panel. Missouri will be screening for all conditions recommended by the American College of Medical Genetics and the March of Dimes.

EXECUTIVE SUMMARY

The Missouri Department of Health and Senior Services (DHSS) is pleased to provide this summary of the annual report for newborn screening. The summary provides highlights of both blood spot and newborn hearing screening for calendar year 2006.

Highlights of Newborn Screening (blood spot)

- As of December 31, 2006, the Missouri newborn screening panel included 27 of the 29 nationally recommended screening disorders, which includes hearing. When considering secondary disorders detected through newborn screening, the State Public Health Laboratory actually performs screening for more than 50 genetic and metabolic disorders on all infants born in Missouri.

- All babies born in Missouri have screenings for these disorders completed from blood samples collected on one filter paper before the baby is discharged from the hospital or birthing facility.

- There were 90,276 blood specimens received in the laboratory from 81,353 reported births in Missouri. Records indicate that greater than 99.4% of babies born in 2006 had a suitable blood spot specimen submitted to the state laboratory; 8.6% of newborns also had a repeat blood spot screen.

- There were 135 newborns with confirmed disorders. All of these babies were followed by a genetic tertiary center or hemoglobinopathy center and are receiving care as part of a health care system with the exception of one with sickle cell disease. Babies were confirmed with the following conditions:

Hemoglobinopathies	62
Primary congenital hypothyroidism	36
Phenylketonuria	8
Amino acid disorders other than PKU	8
Congenital adrenal hyperplasia	7
Medium chain Acyl-CoA dehydrogenase deficiency (MCAD)	6
Fatty acid disorders other than MCAD	4
Organic acid disorders	3
Classical galactosemia	1

- Of the 62 hemoglobinopathies confirmed, 55 were cases of sickle cell disease.

- Collectively in Missouri, this means that one in every 599 babies screened was found to have a genetic or metabolic disorder from the newborn screening panel (excluding hearing screening) in 2006.

- A Cystic Fibrosis Task Force was assembled in 2006 to help the DHSS plan for and prepare to add cystic fibrosis to the newborn screening panel in 2007.

- Satisfaction surveys were conducted in 2006 for both families and physicians concerning their experience when abnormal screening results were reported. Family responses averaged 32% as very satisfied and 44% satisfied with their experience in the way the program was operating. Physician responses in total averaged 97% as very satisfied or satisfied for all of the survey questions, such as timeliness of notification of abnormal results and information contained in the newborn screen report.

Highlights of Newborn Hearing Screening

- There were 79,906 babies screened for hearing loss in 2006 out of a total of 81,353 births in Missouri, which equates to 98.2% of all births screened for hearing.

- A total of 3,010 babies did not pass the initial hearing screening (3.8% of babies screened) and were referred for further testing.

- Audiologists evaluated 635 infants and found that 44 (6.9%) had a permanent hearing loss.

- Of the 44 infants with permanent hearing loss, 28 (63.6%) of them received early intervention services by six months of age.

- A Newborn Hearing Screening Service Coordination Pilot Project was initiated in the Kansas City area in 2006 in collaboration with the Department of Elementary and Secondary Education, Division of Special Education. The project pairs a speech-language pathologist with the First Steps service coordinator when assisting a family who has had an infant diagnosed with severe or permanent hearing loss. The project provides an additional source of information to the family to help them in understanding options and making decisions concerning communication strategies and interventions for their infant.

- A satisfaction survey was conducted in 2006 of parents of babies requiring an audiological assessment as a result of not passing the newborn hearing screening. Approximately 89% of respondents were very satisfied or satisfied with the newborn hearing screening process.

Next Steps

- The DHSS is working toward a web-based reporting system for birth records. Integration of vital records information with newborn screening information will enable the department to determine babies born in Missouri who have not been screened for hearing loss or other newborn disorders.

MISSOURI NEWBORN SCREENING ANNUAL REPORT 2006

What is Newborn Screening?

One of the great advances in preventive medicine has been newborn screening. Newborn screening is a public health program aimed at the early identification of conditions and the timely intervention by health care providers to eliminate or reduce associated mortality and morbidity. It is the goal that every newborn be screened for certain harmful or potentially fatal disorders that aren't otherwise apparent at birth. Newborn screening tests ideally take place before a newborn leaves the hospital. Babies are screened to identify serious or life-threatening conditions before symptoms begin. Individually, these conditions are rare. But collectively, in 2006, one in every 599 babies was found to have a disorder from the 2006 newborn screening panel (excluding hearing).

Many of these disorders are metabolic in nature, which means they interfere with the body's ability to use nutrients to produce energy and maintain healthy tissue. Other types of disorders that may be detected through newborn screening include problems with hormones or blood disorders. These metabolic and other inherited disorders can interfere with an infant's normal physical and mental development in a variety of ways. In some instances they can even lead to death.

With a simple blood test, doctors can often tell whether newborns have certain conditions that could eventually cause problems. The screening involves taking a few drops of blood by pricking the baby's heel and capturing the blood on a filter paper. The paper is sent to the newborn screening laboratory for testing and results are sent back to the hospital of birth and the physician of record. If results are considered to be abnormal, the family will be contacted for further testing of the baby's blood.

The other newborn screening test is a hearing test. This is usually done while the newborn is sleeping and involves placing a tiny earphone in the baby's ear and measuring his or her response to sound. The baby experiences no discomfort from this procedure. Results from the screening are provided immediately. The results will tell the health care staff if further screening or an audiological assessment may be necessary.

"Newborn screening could not have saved our baby boy, but it gave us answers when all we had were questions. Just two days after he was born, our baby's heart and breathing stopped. After he was revived and stabilized, doctors knew where to start to determine the cause. Thankfully results from the newborn screening were available within hours and identified the cause of our baby's illness."

Mother of a son diagnosed

Missouri Newborn Blood Spot Screening

The Newborn Screening Program is a joint effort between the State Public Health Laboratory (SPHL) and the Bureau of Genetics and Healthy Childhood (GHC). The testing of blood spots is performed at the SPHL and follow-up of "at risk" infants is the responsibility of GHC.
The Missouri Newborn Screening Program is mandated by state law to screen all infants born in Missouri for certain genetic and metabolic disorders (RSMo 191.331 and 191.332). The first newborn screening legislation in Missouri was passed in 1965. The original legislation required: "testing for phenylketonuria (PKU) and other metabolic disorders as are prescribed by the division of health."

During the 2001 Legislative Session, House Bill 279 passed, was signed by the Governor and became RSMo 191.332. This statute expanded the Missouri Newborn Screening Program from 5 conditions to over 20. Funding to implement this expansion was obtained and the necessary equipment, training, reagents and procedures were in place when expanded screening became a reality in Missouri on July 1, 2005. Consequently, from July 1, 2005 through 2006, Missouri screened for 27 of the 29 conditions recommended by the American College of Medical Genetics and the March of Dimes.

Newborn Blood Spot Screening Timeline

1967 phenylketonuria (PKU)

1979 congenital hypothyroidism

1985 galactosemia

1989 hemoglobinopathies

2002 congenital adrenal hyperplasia (CAH)

2002 newborn hearing screening

2005 expanded screening to cover tandem mass spectrometry (MS/MS) detectable conditions (21 core conditions were added to the above 6)

As of December 31, 2006, there are 27 of the 29 nationally recommended screening disorders screened in Missouri newborns. The 27 core conditions in the newborn screening panel in Missouri are:

Amino Acid Disorders

Argininosuccinic aciduria (ASA)
Citrullinemia (CIT)
Homocystinuria (HCY)
Hypermethioninemia (MET)
Maple Syrup Urine Disease (MSUD)
Phenylketonuria (PKU)
Tyrosinemias (TYR-I, TYR-II) *

Fatty Acid Disorders

Carnitine Uptake Defect (CUD) *
Carnitine/acylcarnitine translocase deficiency (CACT)
Carnitine palmitoyl transferase deficiency (CPT-II)
3-Hydroxy-hexadecanoyl disorders (LCHAD, TFP)
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
Multiple acyl-CoA dehydrogenase deficiency (GA-II)
Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Disorders

Glutaric acidemia, type 1 (GA-I)
Isovaleric acidemia (IVA)
Methylmalonic acidemias (MUT, Cbl A, B)
Propionic acidemia (PROP)
3-Hydroxy-isovaleryl disorders (3MCC, HMG, BKT, MCD)

Hemoglobin Disorders

Hb S/Beta-thalassemia
Hb S/C disease
Hb S/S disease (Sickle Cell Anemia)

Others

Congenital Adrenal Hyperplasia (CAH)
Congenital Hypothyroidism (CH)
Galactosemia (GALT)
Hearing deficiency

*There is a lower probability of detection of this disorder during the immediate newborn period.

The Missouri Newborn Screening Program's goal is to identify infants at risk and in need of diagnostic testing for the disorders and, if confirmed with the condition, to get them into a system of health care which includes a specialist for management of their condition. A normal screening result does not rule out the possibility of an underlying metabolic/genetic disease. See Appendix 1 for diagnoses and prevalence rates.

Method of Collecting Blood Specimens

1. In the hospital newborn nursery or in a physician's office, infant blood specimens are collected on Newborn Screening Specimen Collection Forms purchased from the SPHL. Instructions are provided on the back of the form to assist in collecting a completed specimen. Initial Forms allow for submission of blood spots and hearing screening information on the first infant specimen. Repeat Forms are available and are designed only for submission of blood spots on any specimen collected after the initial specimen.

2. A specimen will be collected from all infants, regardless of age, before being discharged from the hospital or birthing facility. See Appendix 2 for specimens received.

    - A specimen collected between 24-48 hours of life is considered optimum for newborn screening.

    - The INITIAL newborn screening specimen form is used for the FIRST specimen collected from an infant; for all subsequent specimens collected from the infant, the REPEAT newborn screening specimen form is used.

    - A repeat specimen is required within 14 days of life if the initial specimen was collected before 24 hours of life.

    - INITIAL specimens from sick or premature infants should be collected before a blood transfusion or between 24-48 hours of life. All sick or premature infants should have a REPEAT specimen collected between 7-14 days of life.

    - All specimens should be sent to the SPHL within 24 hours of collection.

Reporting Newborn Screening Blood Spot Results

Once testing is completed, the SPHL sends laboratory reports to the hospital of birth providing the results of newborn screening. See Appendix 3 for a summary of abnormal results.

- "HIGH RISK" screening results are telephoned and faxed immediately to the health care provider of record and to the appropriate follow-up center followed by a mailed copy of the lab report. High risk results will include referral to a genetic tertiary center.

- "BORDERLINE RISK" screening results are faxed to the healthcare provider of record followed by a mailed copy of the lab report.

- "UNSATISFACTORY" specimen results are faxed the day of receipt to the health care provider collecting the specimen followed by a mailed notice. The number of unsatisfactory blood spot specimens is of concern to the program. See Appendix 4 for information concerning unsatisfactory results.

- "NORMAL" screening results are mailed within one week of receipt in the laboratory to the healthcare provider of record and the submitting facility.

Follow-up of Newborns

If a high risk result is obtained on a screening, in addition to the provider of record indicated on the filter paper, the appropriate genetic tertiary center (Cardinal Glennon Memorial Hospital in St. Louis, Children's Mercy Hospital in Kansas City, St. Louis Children's Hospital, or University Hospital and Clinics in Columbia) is also notified by the Department of Health and Senior Services (DHSS) of the results. The appropriate hemoglobinopathy center at St. Louis Children's Hospital, Children's Mercy Hospital, or University Hospital and Clinics is notified of abnormal newborn hemoglobin results. These genetic and hemoglobinopathy centers have responsibility for tracking and follow-up of newborns with abnormal results and they provide consultation with the primary care physician on the care of the infant.

- To insure that all children needing follow-up are appropriately and expeditiously treated, the laboratory report will include the name and telephone number of the designated referral center contact who will provide detailed information on confirmatory testing and treatment of the infant.

- Staff in GHC and the SPHL consult with the centers on a regular basis concerning the status of newborns requiring follow-up to assure that newborns are being found and are receiving further evaluation and care for their condition.

Outcome Data - Newborn Screening Specimens and Results

- In 2006 there were 80,891 babies tested in the state newborn screening laboratory. There were 90,276 blood specimens received in the laboratory. Specimens received included:

Initial	Repeat	Unsatisfactory
80,891	7,769	1,616

- Abnormal test results from laboratory screening of these specimens were:

High Risk	Moderate Risk	Borderline Risk
297	103	2,042

- One hundred thirty-five (135) confirmed disorders were diagnosed from these abnormal results.

Disorder Confirmed	Number	Receiving Care in a Health Care System
Hemoglobinopathies	62	61
Primary Congenital Hypothyroidism	36	36
Phenylketonuria (PKU)	8	8
Amino Acid Disorders other than PKU	8	8
Congenital Adrenal Hyperplasia (CAH)	7	7
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	6	6
Fatty Acid Disorders other than MCAD	4	4
Organic Acid Disorders	3	3
Classical Galactosemia	1	1
Total	**135**	**134**

- Average laboratory turnaround times from receipt of specimen to reporting are:

Results	Turnaround Times
High Risk Result*	1.5 days
Borderline Risk**	4.5 days
Normal Result ***	6 days

* the result is telephoned and faxed to the physician of record
** the result is faxed to the physician of record
*** hard copy reports are mailed to the physician of record and the submitting facility; final abnormal results are also included in this category

- Refer to the appendices for additional 2006 newborn blood spot screening information.

Major Initiatives of Newborn Screening (blood spot) for 2006

- An issue with specimen collection for premature, sick, and low birth weight infants was identified and new collection guidelines were prepared by program and laboratory staff and shared with hospitals and birthing centers.

- Several training programs were presented around the state during 2006 to hospital staffs concerning drawing of blood for filter paper specimens and state public health laboratory procedures for reporting to hospitals.

- Planning for adding cystic fibrosis screening began with the Cystic Fibrosis Task Force in preparation for adding this condition to the newborn screening panel in 2007.

- A parent survey and a physician survey were developed and mailed in 2006. Results of the surveys are found in Appendix 6.

Next Steps

Plans for adding biotinidase deficiency to the newborn screening panel are underway. Consultation with the Genetic Advisory Committee will be done throughout the planning phase and during the pilot program for the condition. It is hoped that biotinidase deficiency will be added in calendar year 2008.

When considering secondary disorders detected through newborn screening, scientists at the State Public Health Laboratory actually perform screening for more than 50 genetic and metabolic disorders on all infants born in Missouri.

Missouri Newborn Hearing Screening

As a result of legislation passed in 1999 (RSMo 191.925 through 191.937), on January 1, 2002, every infant born in Missouri is required to have their hearing screened prior to discharge from an ambulatory surgical center or hospital.

Hospital and birthing center staffs, as well as some physicians' offices, perform initial and repeat hearing screenings on newborns. Hearing screening results are sent to the SPHL and then sent to GHC for entry into the tracking system. Follow-up of infants who missed or did not pass a final hearing screening is the responsibility of GHC. Surveillance activities are designed to assure that newborns either repeat the screening or have a diagnostic audiological assessment.

Program Procedures

1. The Missouri Newborn Hearing Screening Program (MNHSP) strives to assure all babies born in Missouri receive a hearing screen and appropriate follow-up. The goal of this program is to have hearing screens completed on all newborns by three months of age and referral of infants with hearing loss to early intervention services by six months of age. The MNHSP contacts parents whose newborns missed or did not pass the newborn hearing screening and encourages them to schedule a screening or diagnostic evaluation, as appropriate. Parents of those infants diagnosed with a permanent hearing loss are encouraged to seek appropriate early intervention.

2. At the hospital or place of birth, prior to blood collection, demographic information is first completed on the Newborn Screening Specimen Collection Form. The hearing screening portion of the form is then removed and newborn hearing screeners write the results of the final, inpatient newborn hearing screening in the remaining spaces. The results of an outpatient rescreen are placed on a Hearing Only form.

3. Newborn hearing screeners provide a valuable service as they:

    - Give parents the DHSS Newborn Hearing Screening Program pamphlet.
    - Screen for hearing loss prior to discharge to home.
    - Inform parents or guardians of the results.
    - Give parents instructions on how to obtain a rescreen or audiologic evaluation if the infant does not pass the screening.

"The overwhelming thing I've noticed about newborn hearing screening here at Boone Hospital is that hearing loss and all its risk factors are finally on the radar screens of all primary care physicians. It has made so many more health care providers and parents aware of the necessity for children's hearing to be normal in order to achieve academic success."

Audiologist at Boone Hospital, Columbia, Missouri

4. Newborn hearing screening results are reported, including missed and refused, to GHC. All rescreens and outpatient results, including missed and refused, are also reported.

   Diagnostic audiological assessments are reported to GHC using the form found on the DHSS website (http://www.dhss.mo.gov/NewbornHearing/) or audiologists can report electronically. Audiologists who wish to submit results electronically may contact Catherine Harbison at Catherine.Harbison@dhss.mo.gov or at 573-751-6266 to register. Reports are requested to be sent within seven days of the screening or diagnostic assessment.

5. GHC staff follow-up with families by sending parent notification letters to all parents of newborns who failed or missed the newborn hearing screening. If no result is received within three months of the baby's date of birth, staff attempts a phone call to the parents. Staff also contacts birthing hospitals and physicians' offices in an attempt to obtain missing results. GHC sends parents of children identified with permanent hearing loss information about early intervention services provided through First Steps and is a resource for questions pertaining to childhood hearing loss.

Newborn Hearing Screening Results

The Centers for Disease Control and Prevention (CDC) recommends that all infants be screened for hearing loss by one month of age; infants who screen positive for hearing loss receive an audiologic evaluation by three months of age; and infants with confirmed hearing loss receive early medical and intervention services by six months of age.

Hospitals and birthing centers, as well as physicians' offices, screened 98.2% (79,906) of all babies born in Missouri (81,353) in 2006 for hearing loss. Of those 79,906 babies:

96.2% (76,896) passed
3.8% (3,010) did not pass

Additionally:

- 94.9% (75,847) were screened before one month of age.
- Audiologists evaluated 635 infants. Of those 635: 22.8% (145) were evaluated by three months of age.
- Forty-four (44) were identified with a permanent hearing loss. Of those 44: 63.6% (28) received early intervention by six months of age.

This data was obtained November 13, 2007, and is subject to change because the process of collecting and analyzing the data is ongoing. Obtaining results sometimes takes several months.

Major Initiatives for Newborn Hearing Screening for 2006

The Newborn Hearing Screening Service Coordination Pilot Project was initiated in 2006. This project is a collaboration between the DHSS and the Department of Elementary and Secondary Education (DESE), Division of Special Education and has been implemented in the Kansas City area. An audiologist, an educator of the deaf and hard-of-hearing or a speech-language pathologist with experience with deaf or hard-of-hearing children, is paired with the First Steps service coordinator for family interactions and service planning related to an infant diagnosed with severe to profound permanent hearing loss. The pilot service coordinator is referred to as the MO-Hear. The MO-Hear assists the First Steps service coordinator and the family in the following activities:

1) Understanding the child's hearing loss and the implications for education
2) Understanding amplification options
3) Understanding communication options
4) Obtaining additional resources on hearing loss
5) Locating services
6) Providing guidance to families regarding communication strategies (language stimulation) to use with the child prior to enrollment in an intervention program
7) Responding to parents emotional needs as they work through the grieving process

All of the above activities will help to educate parents and empower them to make informed decisions for their child.

A parent survey of infants who went through the hearing screening and audiological assessment was mailed to parents. Survey results are found in Appendix 6.

Next Steps

The DHSS is working toward a web-based reporting system for birth records. When this becomes a reality, newborn hearing screening information will be an addendum to the birth record. The integration of vital records information with newborn screening information will enable the department to determine babies born in Missouri who have not been screened for hearing loss or other newborn disorders. The DHSS is hopeful this will become operational in calendar year 2009.

The DHSS would like to implement a MO-Hear Pilot Program in the St. Louis region that would emulate the program in the Kansas City area. Plans and options will be reviewed in hopes of implementing this in calendar year 2009.

Contact Information for Newborn Screening

Telephone Contacts

573-751-2662	Newborn Screening Laboratory main number
573-751-3334	Order newborn screening specimen forms; person
573-522-4991, Ext 3226	Order newborn screening specimen forms; automated attendant
573-751-6266	Genetics and Healthy Childhood, for follow-up information

Web Addresses

Newborn Screening Laboratory - http://www.dhss.mo.gov/Lab/Newborn/index.html

Newborn Screening Program - http://www.dhss.mo.gov/Genetics/index.html

Newborn Hearing Screening Program - http://www.dhss.mo.gov/NewbornHearing/

"Saint Luke's East in Lee Summit, Missouri, did a great job following up after our daughter did not pass her initial test. They brought us back in for a rescreening and when she didn't pass, they ensured that we were referred to a specialist. Great job!"

Parent from Kansas City

Appendix 1: Prevalence Rates of Disorders

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED PREVALENCE RATE
Amino Acid Disorders	**16**	**1/13,000***
Argininosuccinic aciduria		
Citrullinemia		
Homocystinuria		
Hypermethioninemia		
Maple syrup urine disease		
Phenylketonuria (PKU)	8	1/15,000**
Maternal PKU	3	
Hyper Phenylalaninemia	5	
Tyrosinemias		
Congenital Adrenal Hyperplasia (CAH)	**7**	**1/13,000****
Congenital Hypothyroidism	**36**	**1/2,400****
Fatty Acid Oxidation Disorders	**10**	**1/10,000*****
Carnitine uptake defect	1	
Carnitine acylcarnitine translocase deficiency		
3-Hydroxy-hexadecanoyl disorders	1	
Medium-chain acyl-CoA dehydrogenase deficiency	6	
Multiple acyl-CoA dehydrogenase deficiency		
Short-chain acyl-CoA dehydrogenase deficiency	1	
Very-long chain acyl-CoA dehydrogenase deficiency	1	
Galactosemia	**1**	**1/40,000****
Hemoglobinopathy	**62**	
Sickle cell anemia disease	55	**1/1,700** Total population; **1/400** African-American population
Other disease conditions	7	

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED PREVALENCE RATE
Organic Acid Disorders	**3**	**1/50,000******
Glutaric acidemia, type I		
Isobutyrylglycinuria (IBG)	1	
Isovaleric acidemia		
Methylmalonic acidemias	1	
Propionic acidemia		
3-hydroxy-isovaleryl disorders	1	

* Combined prevalence of all Amino Acid Disorders
** Missouri incidence
*** Combined prevalence of all Fatty Acid Oxidation Disorders
**** Combined prevalence of all Organic Acid Disorders

Appendix 2: Newborn Screening Laboratory Report Specimens Received

	Number Babies Tested	Specimens Received			Total Infant Specimens
		Initial	Repeat	Unsatisfactory	
Jan	6881	6881	657	148	7686
Feb	6177	6177	614	128	6919
Mar	6691	6691	690	149	7530
Apr	6144	6144	574	110	6828
May	7021	7021	651	95	7767
Jun	6627	6627	658	107	7392
Jul	6624	6624	641	97	7362
Aug	7784	7784	726	158	8668
Sep	6839	6839	635	92	7566
Oct	7238	7238	696	185	8119
Nov	6430	6430	586	151	7167
Dec	6435	6435	641	196	7272
Y.T.D.	**80891**	**80891 (89.60%)**	**7769 (8.61%)**	**1616 (1.79%)**	**90276**

Appendix 3: Newborn Screening Laboratory Report Abnormal Results

ABNORMAL RESULTS

Disorder		Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec	Y.T.D.
CAH	Confirmed	0	0	1	0	0	1	2	0	2	0	1	0	**7**
	High Risk	29	28	11	8	7	9	4	6	8	4	5	3	**122**
	Borderline Risk	84	71	57	27	33	26	24	31	20	25	29	31	**458**
CH	Confirmed	2	2	4	1	5	1	5	3	3	7	1	2	**36**
	High Risk	4	5	8	3	6	7	10	4	2	6	3	4	**62**
	Borderline Risk	71	67	84	52	44	48	57	37	47	62	60	69	**698**
GAL	Confirmed	0	0	0	0	0	1	0	0	0	0	0	0	**1**
	High Risk	1	1	0	0	1	5	0	0	3	1	0	0	**12**
	Borderline Risk	5	4	8	11	12	18	22	18	9	7	7	3	**124**
PKU	Confirmed	0	0	2	0	0	1	1	1	2	0	0	1	**8**
	High Risk	2	0	2	0	0	3	2	1	3	0	0	0	**13**
	Moderate Risk	0	1	1	0	0	2	1	0	0	0	0	1	**6**
	Low Risk	9	11	17	8	3	2	4	0	0	0	0	0	**54**
OTHER AA	Confirmed	3	0	1	0	0	3	1	0	0	0	0	0	**8**
	High Risk	2	0	0	1	0	1	0	0	0	0	1	0	**5**
	Moderate Risk	1	2	5	5	2	3	4	6	3	2	4	0	**37**
	Low Risk	0	0	0	0	20	40	29	42	28	32	14	13	**218**
OA	Confirmed	1	0	0	0	2	0	0	0	0	0	0	0	**3**
	High Risk	1	0	1	1	3	0	0	1	0	0	0	0	**7**
	Moderate Risk	3	2	1	0	8	2	4	2	3	3	7	5	**40**
	Low Risk	17	16	23	8	52	29	24	21	21	19	20	27	**277**
MCAD	Confirmed	2	1	0	0	1	0	0	1	0	1	0	0	**6**
	High Risk	2	1	0	0	1	1	0	1	0	1	0	0	**7**

	Moderate Risk	0	0	0	1	1	0	0	0	1	0	0	0	**3**
	Low Risk	2	4	5	1	0	6	9	2	6	4	4	1	**44**
OTHER FA	Confirmed	0	0	0	1	2	0	0	0	0	0	1	0	**4**
	High Risk	2	2	0	0	2	1	0	0	0	0	0	0	**7**
	Moderate Risk	3	0	2	3	1	0	0	2	0	0	2	4	**17**
	Low Risk	18	15	19	9	18	13	15	15	10	14	8	15	**169**

CAH = congenital adrenal hyperplasia
GAL = galactosemia
PKU = phenylketonuria
FA= fatty acid
CH = congenital hypothyroidism
AA= amino acid
OA= organic acid
MCAD= medium chain acyl-CoA dehydrogenase deficiency

Confirmations may still be received and consequently affect year-to-date totals.

8/13/2007

Appendix 4: 2006 Unsatisfactory Samples

INCOMPLETE SATURATION: Uneven saturation; blood did not soak through the filter paper. Possible causes: Removing filter paper before blood has completely filled circle or before blood has soaked through to opposite side; improper capillary tube application; allowing filter paper to come in contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood specimen collection.	482
LAYERED CLOTTED OR SUPERSATURATED: Possible causes: Touching the same circle on filter paper to blood drop several times; filling circle on both sides of filter paper; application of excess blood; clotted swirl marks from improper capillary application. Use of unheparinized capillary tube.	335
BLOOD ON OVERLAY COVER: Overlay cover came in contact with wet blood specimen. Sample is unsatisfactory for testing because blood soaked from back of filter onto the gold colored backing of the form. The filter circles are designed to hold a specific quantity of blood. If the wet filter is allowed to come in contact with the paper backing of form, blood can be drawn out of filter making the quantitative tests performed by the Newborn Screening Laboratory invalid. Allow blood spots to thoroughly air dry for at least 2 hours in a horizontal position, away from direct heat and sunlight. Do not allow the blood to touch any surface during drying, including other parts of the form.	310
DILUTED, DISCOLORED OR CONTAMINATED: Possible causes: squeezing or milking of area surrounding the puncture site; allowing filter paper to come in contact with gloved or ungloved hands, or substances such as alcohol, formula, antiseptic solutions, water, hand lotion, powder, etc., either before or after blood specimen collection; exposing blood spots to direct heat; allowing blood spots to come in contact with tabletop, etc. while drying the sample.	214
QUANTITY NOT SUFFICIENT: Quantity of blood on filter not sufficient for testing. Possible causes: Removing filter paper before blood has completely filled circle; not allowing an ample sized blood drop to form before applying to filter; inadequate heel stick procedure.	136
SPECIMEN ABRADED: Filter scratched, torn or abraded. Possible causes: Improper use of capillary tubes. To avoid damaging the filter paper fibers, do not allow the capillary tube to touch the filter paper. Actions such as "coloring in" the circle, repeated dabbing around the circle, or any technique that may scratch, compress, or indent the paper should not be used.	57
OLD SPECIMEN: Specimen greater than 15 days old when received at State Public Health Laboratory. The collection card should be transported or mailed to the Newborn Screening Laboratory within 24 hours after specimen collection. Avoid the practice of holding onto specimens to wait for more to accumulate before mailing, also referred to as "batching" the specimens. Although batching may seem more efficient, it's not worth it in the long run because a delay in screening and treatment can cause irreparable damage to a child with metabolic disease.	35
OLD FORM: Sample received on out-of-date form. The current newborn screening specimen collection form has a printed form number located next to the form bar code that begins with **"B05"**. If a form number begins with any other letters or digits and does not have a printed bar code, then it is an outdated form and should not be used.	22
SERUM RINGS: Serum separated into clear rings around blood spot. Possible causes: Card dried vertically (on side) instead of flat; squeezing excessively around puncture site; allowing filter paper to come in contact with alcohol, hand lotion, etc.	10
OTHER UNSUITABLE	6
REFUSAL: Parents refused to have Newborn Screening. Parents who object to testing on religious grounds shall state those objections in writing. The written objection shall be filed with the attending physician, certified nurse midwife, public health facility, ambulatory surgical center or hospital. It is important that the signed refusal of testing be kept with the baby's medical records. Upon receipt, the attending physician, certified nurse midwife, public health facility, ambulatory surgical center or hospital shall send a copy of the written objection to the Department of Health and Senior Services to the attention of Genetics and Healthy Childhood, 930 Wildwood Dr., Jefferson City, MO 65109.	5
NO BLOOD: Filter submitted without blood.	4
TOTAL UNSATISFACTORY SPECIMENS RECEIVED	1,616 (1.79%)

Appendix 5: Hemoglobinopathy Report

Specimens Received:	**91,023**
Initial:	80,891 (88.9%)
Repeat:	7,769 (8.5%)
Unsatisfactory:	1,616 (1.8%)
Whole Blood:	747 (.8%)
Analyses (Tests) Performed:	**118,847**
First Tests:	90,751 (76.4%)
Retests:	9,595 (8.1%)
Controls/Standards:	17,766 (15.0%)
Proficiency Testing:	130 (.1%)
Whole Blood Tests:	605 (Record beginning July 1) (.5%)

Significant Results = 1,664					
Sickle Cell Disease		**Other Disease Conditions**		**Trait Conditions**	
FS	37	FC	1	FAS	1,047
FSC	16	FCA	3	FAC	314
FSA	2	FE	1	FAX	150
		Highly Elevated Barts	2	FAE	44
				FAD	36
				FASX	2
				FACX	1
				Slightly Elevated Barts	8
Total	**55 (3.3%)**	**Total**	**7 (.4%)**	**Total**	**1,602 (96.2%)**

Geographic Follow-up of Significant Disease and Trait Conditions					
Significant Disease Conditions			**"S" Trait Conditions (includes repeats)**		
St. Louis Area	33	53%	St. Louis Area	653	59%
Kansas City Area	18	29%	Kansas City Area	305	28%
Remainder of MO	11	18%	Remainder of MO	146	13%
Total	**62**	**100%**	**Total**	**1,104**	**100%**

Note: Because of rounding, percentages will not necessarily add to exactly 100%.

Appendix 6: Newborn Screening Satisfaction Surveys

A satisfaction survey of parents and physicians was conducted for families of babies having abnormal newborn screening results reported in 2006. Key findings:

Newborn Screening Parent Satisfaction Survey - Parent Response*			
	Very Satisfied	Satisfied	Not Satisfied
Explanation of abnormal MS/MS results	33%	40%	27%
Timeliness on notification of abnormal MS/MS screen results	47%	40%	7%
Number of follow-up tests or newborn screen results done to determine diagnosis	20%	60%	13%
Timeliness of follow-up tests and/or newborn screen	27%	40%	20%
Answers to parents' questions about the disorders screened and testing methodology	33%	40%	27%

* Some categories will not total to 100% because of no response.

Newborn Screening Physician Satisfaction			
	Very Satisfied	Satisfied	Not Satisfied
Timeliness on notification of abnormal MS/MS newborn results	79%	21%	0%
Method of receiving abnormal MS/MS results	82%	14%	4%
Information contained in the newborn screen report	87%	11%	2%
Result interpretation of newborn screen report	79%	18%	3%
Ease on contacting a genetic tertiary center for consultation	79%	18%	3%
Recommendations of the genetic tertiary center	82%	11%	7%

A Satisfaction Survey of parents of children born in 2006 who went through the newborn hearing screening and audiologic assessment process was completed in June of 2007.

Key findings:

- 89% of respondents were very satisfied or satisfied with the newborn hearing screening process
- 7% of respondents were somewhat satisfied
- 4% of respondents were not satisfied

In addition:

- 95% of the respondents reported that the birth hospital notified them of the screening result
- 86% of the respondents reported that the birth hospital provided them with the newborn hearing screening program brochure