MISSOURI NEWBORN SCREENING

2007 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, Sickle Cell Standing Committee, Cystic Fibrosis Standing Committee and Newborn Hearing 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 committees provide 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 Newborn Screening Program Staff

Newborn screening lab staff (above) work at the State Public Health Laboratory in Jefferson City.

Staff in the Bureau of Genetics and Healthy Childhood (right) provide the follow-up component for the newborn screening program.

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.

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 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 hearing screening are provided immediately. The results will tell the health care staff if further screening or an audiological assessment may be necessary.

Our son was born at the end of December 2007. We were so happy and excited that he was finally here. He was absolutely perfect!

After a week had passed, we received a phone call with unexpected news from Dr. Laurie Smith. She stated that our son's newborn screen results detected that he had MCAD (medium chain acyl-coA dehydrogenase deficiency). Our world stopped. We were devastated and numb upon learning this news. She continued to explain what MCAD was and she wanted to see us in a couple of days. We then went to Children's Mercy Hospital and met with Dr. Smith and her staff and spoke to them about MCAD. Dr. Smith and her staff further explained what MCAD was and what to expect. She wanted to have another blood test done to confirm that our son had MCAD. She called us a couple of days later to tell us the results. Not only did he have MCAD, but his labs showed the highest levels that the state of Missouri had ever seen.

Today, our son is doing great! Without the newborn screening, we do not know if we would be writing this to explain how grateful we are that the test, which revealed that our son has MCAD, was performed. We truly believe that the newborn screen saved our son's life! We are grateful that the newborn screen was performed and hope that it will someday become a law in every state so a parent does not lose their child due to a disorder that could have been caught in a screening. Our hope is that the state of Missouri continues to provide funding for the newborn screenings, so precious lives can be saved.

- Parents of a son diagnosed with MCAD
The family lives in western Missouri.

Missouri Newborn Blood Spot Screening

In 2007 several noteworthy accomplishments were made in Missouri's newborn screening program. Cystic fibrosis screening was added to the newborn panel on June 1, 2007, after a three month pilot program. CF is an inherited chronic disease that affects the lungs and digestive system of about 30,000 children and adults in the United States. A defective gene and its protein product cause the body to produce unusually thick, sticky mucus that clogs the lungs and leads to life-threatening lung infections. CF also obstructs the pancreas and stops natural enzymes from helping the body break down and absorb food.

It was anticipated that approximately 25 infants per year would be confirmed positive for CF in Missouri. During the three-month pilot program, nine infants were diagnosed with cystic fibrosis. For the remaining seven months of the year, fourteen infants were confirmed positive for a total of 23 cases confirmed during the 10 months of 2007 that screening was conducted for cystic fibrosis.

With the addition of CF to the newborn screening panel, Missouri's screening program now screens for 28 of the 29 conditions recommended by the American College of Medical Genetics and the March of Dimes. Only screening for biotinidase deficiency remains to be added in order to fully meet the recommendations. It is anticipated that screening for biotinidase will be added within calendar year 2008.

When considering secondary disorders detected through newborn screening, the State Public Health Laboratory currently performs screening for 66 genetic and metabolic disorders on all infants born in Missouri.

A new public health laboratory was completed and during July 2007, the State Public Health Laboratory moved into this state-of-the-art facility located at 101 N. Chestnut in Jefferson City. A dedication ceremony was held September 20, 2007. Compared to the 68,000 square feet of the old laboratory, the new laboratory's 117,402 square feet provides needed expansion for programs. A new biosafety level 3 laboratory area contains 11,505 square feet and is designed for processing the most serious infectious agents.

The rapidly expanding newborn screening laboratory, which had been located in southeast Jefferson City for the past nine years as a separate laboratory, moved into the new building the last week of July. Through careful planning, newborn screening staff moved the laboratory with minimal down time. Administrative staff moved over one week before laboratory staff, and two new tandem mass spectrometers were installed and validated a month before the move. The move began on a Thursday evening when all testing was stopped. All equipment and supplies were moved Friday, instruments were started up and validated on Saturday and Sunday, and testing was resumed on Monday.

The newborn screening staff is excited to work in the new facility and move forward with an expanding newborn screening program that places Missouri among the nation's leaders.

Next steps

Missouri is continuing to expand the conditions screened to reach the recommendations of the American College of Medical Genetics (ACMG) and the March of Dimes (MoD). Screening for biotinidase deficiency is expected to begin in December 2008. This will fulfill Missouri's goal of screening for all 29 core conditions recommended by the ACMG and the MoD. When considering secondary conditions, screening for these disorders actually allows for a total of 67 disorders to be detected through newborn screening.

Missouri Newborn Screening Disorders Screened and Reported

- **Classical galactosemia (GALT)**
- **Congenital adrenal hyperplasia (CAH)**
- **Congenital primary hypothyroidism (CH)**
- **Cystic fibrosis (CF)**
- **Amino Acid Disorders**
  - Argininemia (ARG, arginase deficiency)
  - Argininosuccinate acidemia (ASA, argininosuccinase)
  - Defects of biopterin cofactor biosynthesis (BIOPT-BS)
  - Defects of biopterin cofactor regeneration (BIOPT-RG)
  - Citrullinemia type I (CIT-I, argininosuccinate synthetase)
  - Citrullinemia type II (CIT-II, citrin deficiency)
  - Homocystinuria (HCY, cystathionine beta synthase)
  - Hyperphenylalaninemia (H-PHE)
  - Hypermethioninemia (MET)
  - Maple syrup urine disease (MSUD, branched-chain ketoacid dehydrogenase)
  - Phenylketonuria (PKU, phenylalanine hydroxylase)
  - Tyrosinemia type I (TYR-1, fumarylacetoacetate hydrolase)*****
  - Tyrosinemia type II (TYR-II, tyrosine aminotransferase)
  - Tyrosinemia type III (TYR-III, hydroxyphenylpyruvate dioxygenase)
- **Fatty Acid Disorders**
  - Carnitine acylcarnitine translocase deficiency (CACT)
  - Carnitine uptake defect (CUD, carnitine transport defect)*****
  - Carnitine palmitoyl transferase deficiency I (CPT-1a)
  - Carnitine palmitoyl transferase deficiency II (CPT-II)
  - Dienoyl-CoA reductase deficiency (DE-RED)
  - Glutaric acidemia type II (GA-II, multiple acyl-CoA dehydrogenase deficiency)
  - Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
  - Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
  - Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
  - Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
  - Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
  - Trifunctional protein deficiency (TFP)
  - Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

- **Organic Acid Disorders**
  - 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
  - 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG, SBCAD)
  - 3-Hydroxy 3-methylglutaric aciduria (HMG, 3-Hydrox 3-methylglutaryl-CoA lyase)
  - 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
  - 3-Methylglutaconic aciduria (3MGA, Type I hydratase deficiency)
  - Beta ketothiolase (BKT, mitochondrial acetoacetyl-CoA thiolase, short-chain ketoacyl thiolase)
  - Glutaric acidemia type I (GA-1, glutaryl-CoA dehydrogenase)
  - Isobutyryl-CoA dehydrogenase deficiency (IBG)
  - Isovaleric acidemia (IVA, Isovaleryl-CoA dehydrogenase)
  - Malonic acidemia (MAL, malonyl-CoA decarboxylase)
  - Methylmalonic acidemia (CBL A,B; vitamin B12 disorders)
  - Methylmalonic acidemia (CBL C,D)
  - Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)
  - Multiple carboxylase deficiency (MCD, holocarboxylase synthetase)
  - Propionic acidemia (PROP, propionyl-CoA carboxylase)

- **Hemoglobinopathies**
  - Sickle cell anemia disease (Hb S/S)
  - Sickle hemoglobin-C disease (Hb S/C)
  - Sickle beta zero thalassemia disease
  - Sickle beta plus thalassemia disease
  - Sickle hemoglobin-D disease
  - Sickle hemoglobin-E disease
  - Sickle hemoglobin-O-Arab disease
  - Sickle hemoglobin Lepore Boston disease
  - Sickle HPFH disorder
  - Sickle “Unidentified”
  - Homozygous-C disease
  - Hemoglobin-C beta zero thalassemia disease
  - Hemoglobin-C beta plus thalassemia disease
  - Homozygous-E disorder
  - Hemoglobin-E beta zero thalassemia disease
  - Hemoglobin-E beta plus thalassemia disease
  - Homozygous beta zero thalassemia disease
  - Double heterozygous beta thalassemia disease
  - Hemoglobin-H disease

- **Others**
  - Hearing

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

The Missouri Newborn Screening Laboratory’s goal is to identify infants at risk and in need of diagnostic testing for the above disorders. A normal screening result does **NOT** rule out the possibility of an underlying metabolic/genetic disease.

On February 27, 2007 our daughter was born. She was a healthy, beautiful baby girl and we couldn't be happier that she was finally here. We took her home from the hospital and introduced her to our family and friends. Besides the lack of sleep that comes with having a newborn, we couldn't believe how perfect everything had gone for us. Our family had a healthy new member with no complications, and we couldn't be happier.

Ten days after bringing our daughter home from the hospital things finally started getting back to normal in our household. Then we received a phone call on that day that the newborn screening test showed a positive result for PKU. We were instructed to take our daughter to the genetics clinic at Children's Mercy Hospital where they would explain to us what was going on with our little girl. We were scared and unsure about what the future would hold for our new family.

Friday morning we were introduced to Dr. Laurie Smith and nutritionist Tarine Weihe. We received a crash course that morning on PKU and how it is treated. We were overwhelmed with emotions and questions that day. Questions we had were why did this happen to us? Would our child be normal? Should we have more kids? Can we afford to pay for the treatment? Over and over that day we tried to get a grip on the news that our daughter had PKU and how we would be directly responsible for her health.

Because of the newborn screening test, our daughter is reaching all her milestones and is a very intelligent little girl with nothing stopping her from reaching her full potential in life.

Over the next few weeks we asked a lot of questions and spent many hours researching everything about PKU. Children's Mercy Hospital has been wonderful in not only treating our daughter, but educating us and helping with any and all things that come with raising a child. We will do whatever it takes to keep her on track and in control of the special diet she needs to ensure a bright future with endless possibilities.

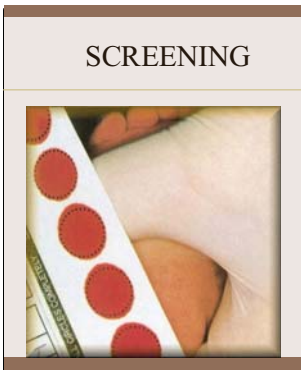
We are very thankful for the newborn screening test given to our child when she was born. Without the test we wouldn't have known anything was wrong until much later in her life when it would have been too late to help her. Because of the newborn screening test our daughter is reaching all her milestones and is a very intelligent little girl with nothing stopping her from reaching her full potential in life. It's all because of the newborn screening test. I'm sure we can speak for a lot of other families when we say that it was our daughter's savior to a high quality of life.

- Parents of a daughter diagnosed with PKU
The family lives in western Missouri.

The Newborn Screening Process

1: TESTING

- The baby's heel is pricked and a few drops of blood are collected on a filter paper 24 to 48 hours after birth.



- The dried blood spot specimen is shipped to the State Public Health Laboratory.
- Specimen is tested for multiple conditions.

2: FOLLOW-UP

- Positive screen results are reported by phone/ fax/letter from lab and follow-up staff to baby's physician. Results are also sent to the appropriate Genetic Tertiary Center in Missouri for follow-up.

- Specimen screening results are entered into data system.
- Baby's physician or health care provider contacts baby's parents.

- Parents bring baby back in for evaluation and more testing at the genetic center.

3: DIAGNOSIS/ INTERVENTION

- Depending on the screen result and the condition screened, repeat or confirmatory testing occurs at the genetic center.

- Parent education for signs/symptoms to watch for is conducted.

- Baby's physician consults with the specialist appropriate to the condition.

4: TREATMENT & MANAGEMENT

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

- Parents receive treatment guidelines/ education. Team support services as appropriate, include:
  - Metabolic dietitian monitoring and consultation
  - Ongoing blood monitoring
  - Referral to early intervention services
  - Pulmonary/CF services
  - Pediatriac endocrine monitoring
  - Pediatric hematology monitoring
  - Genetic counseling and consideration of family testing
  - Other allied health services as needed

Missouri Newborn Hearing Screening

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.

Provisional 2007 data for Missouri shows:

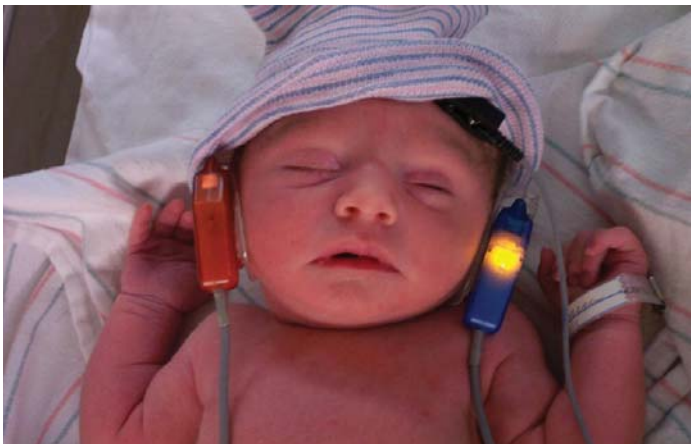
- 81,879 live births
- 79,980 (97.0%) infants screened by one month of age
- 1,925 (2.0%) infants screened after one month of age
- 1,489 infants required audiologic evaluation
- 356 (23.9%) infants received audiologic evaluation by three months of age
- 49 infants diagnosed with a permanent hearing loss
- 18 (36.7%) infants received early intervention services by six months of age

Note: This data was obtained June 20, 2008, and is subject to change because the process of collecting and analyzing the data is ongoing.

The Missouri Newborn Hearing Screening Program (MNHSP) Service Coordination Pilot Project continued into 2007. This project, a collaboration between the DHSS and the Department of Elementary and Secondary Education (DESE), Division of Special Education, pairs an expert in the unique needs of a newborn with hearing loss with the First Steps service coordinator for family interactions and service planning. The project focuses on families in the Kansas City area who have an infant diagnosed with severe to profound permanent hearing loss. All parent feedback received on the pilot in 2007 was excellent. However, the low number of newborns with diagnosed severe to profound hearing loss in the Kansas City area led the MNHSP to extend the pilot for another year. This was necessary in order to obtain more information about the services provided prior to expanding into the eastern part of the state.

Informational parent brochures about newborn hearing screening have been revised and are available to hospitals free of charge. Brochures should be given to all families upon admission into the hospital. They can also be used during childbirth classes and pre-natal visits. Call the MNHSP at 573-751-6266 to order the new brochures.

The MNHSP developed Audiology Cards in 2004 to provide parents information about the diagnostic process and intervention choices. In 2007, the Audiology Cards were revised based upon input from the Genetic Advisory Committee's Newborn Hearing Screening Standing Committee members. Each set is made up of thirteen different subject cards. Specific cards may be given to parents based upon the needs of individual families. Audiology Cards may be ordered by calling the MNHSP at: 573-751-6266.



Next steps

In an effort to reduce lost-to-follow-up after failure to pass the newborn hearing screening, the MNHSP planned a pilot project to begin in 2008 with three Missouri hospitals. The three pilot hospitals agreed to use a script to inform parents of non-passing results and to explain the importance of returning for another screening or for an audiologic evaluation. Additionally, the hospitals will make follow-up appointments for these families. The MNHSP will make reminder phone calls to the families prior to the appointment date and send a letter of notification to the baby's physician. In 2009, the MNHSP will meet with regional stakeholders and the hospitals involved in the lost-to-follow-up pilot project to review project results.

In 2008, the MNHSP will consider expanding the Service Coordination project with DESE into the St. Louis region after evaluating the data from the Kansas City pilot program.

Contact Information for Newborn Screening

Telephone Contacts:

Newborn Screening Laboratory main number	573-751-2662
Order newborn screening specimen forms; person	573-751-3334
Order newborn screening specimen forms; automated attendant	573-522-4991, Ext. 3226
Genetics and Healthy Childhood, for follow-up information	1-800-877-6246

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/

Appendix 1: Projected Incidence Rates – 2007 Births

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Amino Acid Disorders	**9**	**1/8,000***
Arginemia		
Argininosuccinate acidemia		
Citrullinemia type I		
Citrullinemia type II		
Defects of biopterin cofactor biosynthesis		
Defects of biopterin cofactor regeneration		
Homocystinuria	1	
Hypermethioninemia		
Hyperphenylalaninemia	2	
Maple syrup urine disease		
Maternal PKU		
Phenylketonuria (PKU)	6	1/15,000
Tyrosinemia type I		
Tyrosinemia type II		
Tyrosinemia type III		
Classical galactosemia (GALT)	**2**	**1/50,000**
Congenital adrenal hyperplasia (CAH)	**2**	**1/13,000**
Congenital primary hypothyroidism (CH)	**32**	**1/3,000**
Cystic fibrosis (CF)	**23**	**1/4,000**
Fatty Acid Oxidation Disorders	**9**	**1/10,000***
Carnitine acylcarnitine translocase deficiency		
Carnitine uptake defect		
Carnitine palmitoyl transferase deficiency I		
Carnitine palmitoyl transferase deficiency II		
Dienoyl-CoA reductase deficiency		
Glutaric academia type II		
Long-chain hydroxyacyl-CoA dehydrogenase deficiency		
Medium-chain acyl-CoA dehydrogenase deficiency	5	
Medium-chain ketoacyl-CoA thiolase deficiency		
Medium/Short chain L-3 hydroxy acyl-CoA dehydrogenase deficiency		
Short-chain acyl-CoA dehydrogenase deficiency	2	
Trifunctional protein deficiency		
Very-long chain acyl-CoA dehydrogenase deficiency	1	
Unknown fatty acid oxidation disorder	1	

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Organic Acid Disorders	**6**	**1/25,000***
2-Methyl-3-hydroxybutyric aciduria		
2-Methylbutyryl-CoA dehydrogenase deficiency		
3-Hydroxy 3-methylglutaric aciduria		
3-Methylcrotonyl-CoA carboxylase deficiency	1	
3-Methylglutaconic aciduria		
Beta ketothiolase		
Glutaric acidemia, type I	1	
Isobutyryl-CoA dehydrogenase deficiency	1	
Isovaleric acidemia	2	
Malonic acidemia		
Methylmalonic acidemia (CBL A,B; vitamin B12 disorders)		
Methylmalonic acidemia (CBL, C,D)	1	
Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)		
Multiple carboxylase deficiency		
Propionic acidemia		
Hemoglobinopathies	**28**	**1/1,700***
Sickle cell anemia disease (Hb S/S)	12	**1/3,000** Total population; **1/400** African-American population
Sickle hemoglobin-C disease (FSC)	7	
Sickle beta zero thalassemia disease		
Sickle beta plus thalassemia disease (FSA)	3	
Sickle hemoglobin-D disease		
Sickle hemoglobin-E disease		
Sickle hemoglobin-O-Arab disease		
Sickle hemoglobin Lepore Boston disease		
Sickle HPFH disorder		
Sickle “Unidentified”		
Homozygous-C disease (FC)	3	
Hemoglobin-C beta zero thalassemia disease		
Hemoglobin-C beta plus thalassemia disease		
Homozygous-E disorder (FE)	1	
Hemoglobin-E beta zero thalassemia disease		
Hemoglobin-E beta plus thalassemia disease		
Homozygous beta zero thalassemia disease		
Double heterozygous beta thalassemia disease		
Hemoglobin-H disease (Highly Elevated Barts)	1	
Other (FCD)	1	

*Combined incidence of all disorders in this disorder category

Appendix 2: Newborn Screening Laboratory Report – Specimens Received 2007

	Number Babies Tested	Specimens Received			Total Infant Specimens
		Initial	Repeat	Unsatisfactory	
Jan	6974	6974	729	207	7910
Feb	6229	6229	596	175	7000
Mar	6469	6469	736	183	7388
Apr	6249	6249	660	184	7093
May	7180	7180	662	140	7982
Jun	6679	6679	604	155	7438
Jul	7536	7536	683	181	8400
Aug	7219	7219	670	163	8052
Sep	6467	6467	599	202	7268
Oct	7322	7322	686	204	8212
Nov	6326	6326	616	235	7177
Dec	6450	6450	621	267	7338
Y.T.D.	**81,100**	**81,100 (88.87%)**	**7,862 (8.62%)**	**2,296 (2.52%)**	**91,258**

Appendix 3: Newborn Screening Laboratory Report – Abnormal Results 2007*

Disorder		Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec	Y.T.D.
CAH	Confirmed	0	0	0	0	0	1	0	0	0	0	1	0	**2**
	High Risk	8	7	0	2	9	8	11	9	9	8	12	7	**90**
	Borderline Risk	58	42	33	37	30	40	36	38	30	50	28	32	**454**
CF	Confirmed	0	0	3	3	3	2	1	2	2	4	2	1	**23**
	Referred	11	5	6	6	7	9	7	4	6	7	7	5	**80**
CH	Confirmed	5	1	8	4	0	0	3	2	2	5	2	0	**32**
	High Risk	13	7	13	6	1	3	5	7	9	7	4	2	**77**
	Borderline Risk	68	43	64	73	82	63	67	45	41	59	73	63	**741**
GAL	Confirmed	0	0	0	1	0	1	0	0	0	0	0	0	**2**
	High Risk	2	0	4	3	3	4	3	6	6	1	1	1	**34**
	Borderline Risk	5	3	6	3	7	7	15	21	10	6	6	2	**91**
PKU	Confirmed	0	1	1	0	1	2	0	0	0	0	0	1	**6**
	High Risk	0	1	2	0	1	2	0	0	0	0	0	2	**8**
	Moderate Risk	0	0	0	0	0	0	1	0	0	1	1	1	**4**
	Low Risk	0	1	2	2	0	0	3	5	2	3	1	0	**19**
OTHER AA	Confirmed	0	0	0	0	0	0	0	1	0	0	0	2	**3**
	High Risk	0	2	0	0	0	0	0	0	2	1	1	1	**7**
	Moderate Risk	4	0	3	1	0	0	3	0	0	2	2	2	**17**
	Low Risk	19	18	24	14	18	21	38	42	35	32	25	35	**321**
OA	Confirmed	0	0	0	0	1	1	0	1	1	0	2	0	**6**
	High Risk	0	2	1	0	2	1	0	3	2	0	3	1	**15**
	Moderate Risk	6	1	0	2	2	1	2	0	0	2	3	3	**22**
	Low Risk	41	49	52	67	21	42	19	21	16	23	24	14	**389**

continued

Disorder		Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec	Y.T.D.
MCAD	Confirmed	0	1	0	0	0	0	1	0	0	1	1	1	**5**
	High Risk	0	1	0	0	0	0	2	0	0	2	1	2	**8**
	Moderate Risk	0	0	0	0	0	0	0	0	0	0	0	0	**0**
	Low Risk	9	5	3	7	3	1	3	6	10	7	5	6	**65**
OTHER FA	Confirmed	1	0	1	0	1	1	0	0	0	0	0	0	**4**
	High Risk	0	0	0	1	1	0	0	0	0	0	0	0	**2**
	Moderate Risk	3	1	3	0	0	2	2	1	0	1	2	4	**19**
	Low Risk	17	21	20	24	22	21	20	33	30	29	15	14	**266**

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

*See Appendix 5 for hemoglobinopathy results.

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

Results	Turnaround Times
High Risk Result*	1.5 days
Borderline Risk**	5 - 6 days
Normal Result **	5 - 6 days

* the result is telephoned and 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

Outcome Data - Newborn Screening Specimens and Results

- In 2007 there were 81,100 babies tested in the state newborn screening laboratory. There were 90,276 blood specimens received in the laboratory. Specimens received included:

Initial	Repeat	Unsatisfactory
81,100	7,862	2,288

- Abnormal test results from laboratory screening of these specimens, including hemoglobinopathy results from Appendix 5, were:

High Risk	Moderate Risk	Borderline Risk
349	62	2,346

- One hundred eleven (111) confirmed disorders were diagnosed from these abnormal results**.**

Appendix 4: 2007 Unsatisfactory Samples

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.	609
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.	584
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.	400
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.	331
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.	160
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.	128
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.	20
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.	17
LABORATORY ACCIDENT: Unable to test; sample damaged at laboratory.	16
FILTER AND FORM BARCODES DO NOT MATCH: Bar code on filter does not match bar code on Newborn Screening Form. Collection forms contain barcodes on demographic, hearing and filter portions. The barcodes may not be altered in any way. If incorrect baby is sampled do not remove filter and attach to a different demographic portion. If a sampling error occurs the entire form needs to be voided and sample needs to be recollected on a new form. All barcodes must match, laboratory copy, submitter copy, newborn hearing screen, and filter.	7
NO BLOOD: Filter submitted without blood.	6
OTHER UNSUITABLE	4
OLD FORM: Sample received on out-of-date form.	4
MISSING OR INCOMPLETE PATIENT INFORMATION: Missing or incomplete demographic information.	2
Total Unsatisfactory Specimens Received	2,288 (2.6%)

Appendix 5: Hemoglobinopathy Report 2007

Specimens Received:

Initial:	81,100 (88.7%)
Repeat:	7,862 (8.6%)
Unsatisfactory:	2,296 (2.5%)
Whole Blood:	214 (.2%)
Total:	**91,472**

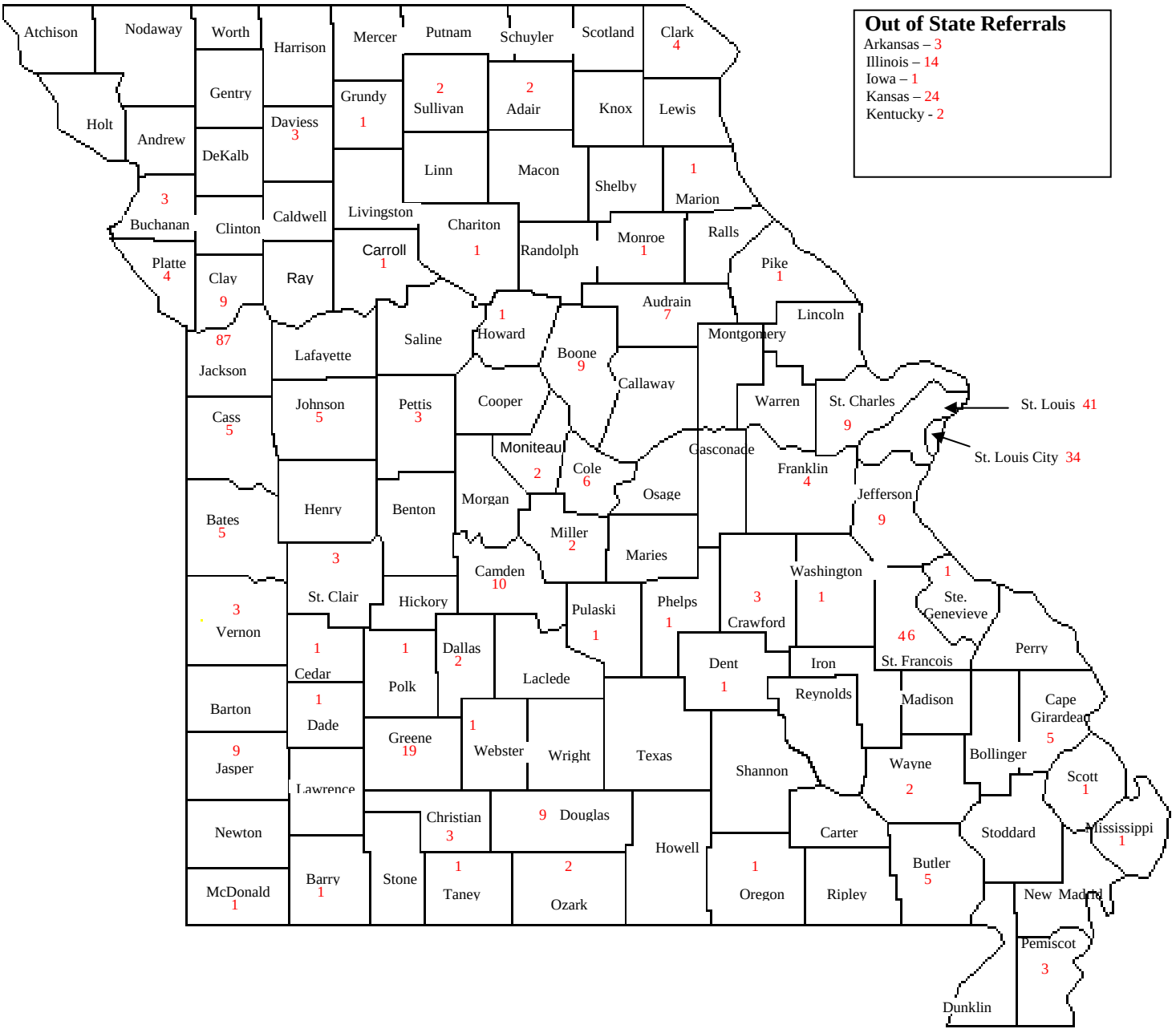
Analyses (Tests) Performed:	**IEF**	**HPLC**	**Total**
First Tests:	91,259 (85.1%)	-	91,259 (78.2%)
Retests:	1,965 (1.8%)	5,521 (58.7%)	7,486 (6.4%)
Controls/Standards:	13,673 (12.8%)	3,583 (38.1%)	17,256 (14.8%)
Proficiency Testing:	84 (.1%)	62 (.7%)	146 (.1%)
Whole Blood Tests:	253 (.2%)	246 (2.6%)	499 (.4%)
Total:	**107,234**	**9,412**	**116,646**

Significant Results = 1,690					
Sickle Cell Disease		**Other Disease Conditions**		**Trait Conditions**	
FS	12	FC	3	FAS	1,090
FSC	7	FCA	0	FAC	343
FSA	3	FE	1	FAX	116
		Highly Elevated Barts	1	FAE	35
		FCX	0	FAD	33
		F-Only	0	FASX	2
		Other (FCD)	1	FACX	1
				Slightly Elevated Barts	10
				FAG	5
				FSAINC	21
				FCAINC	6
Total	**22(1.3%)**	**Total**	**6 (.4%)**	**Total**	**1,662 (98.3%)**

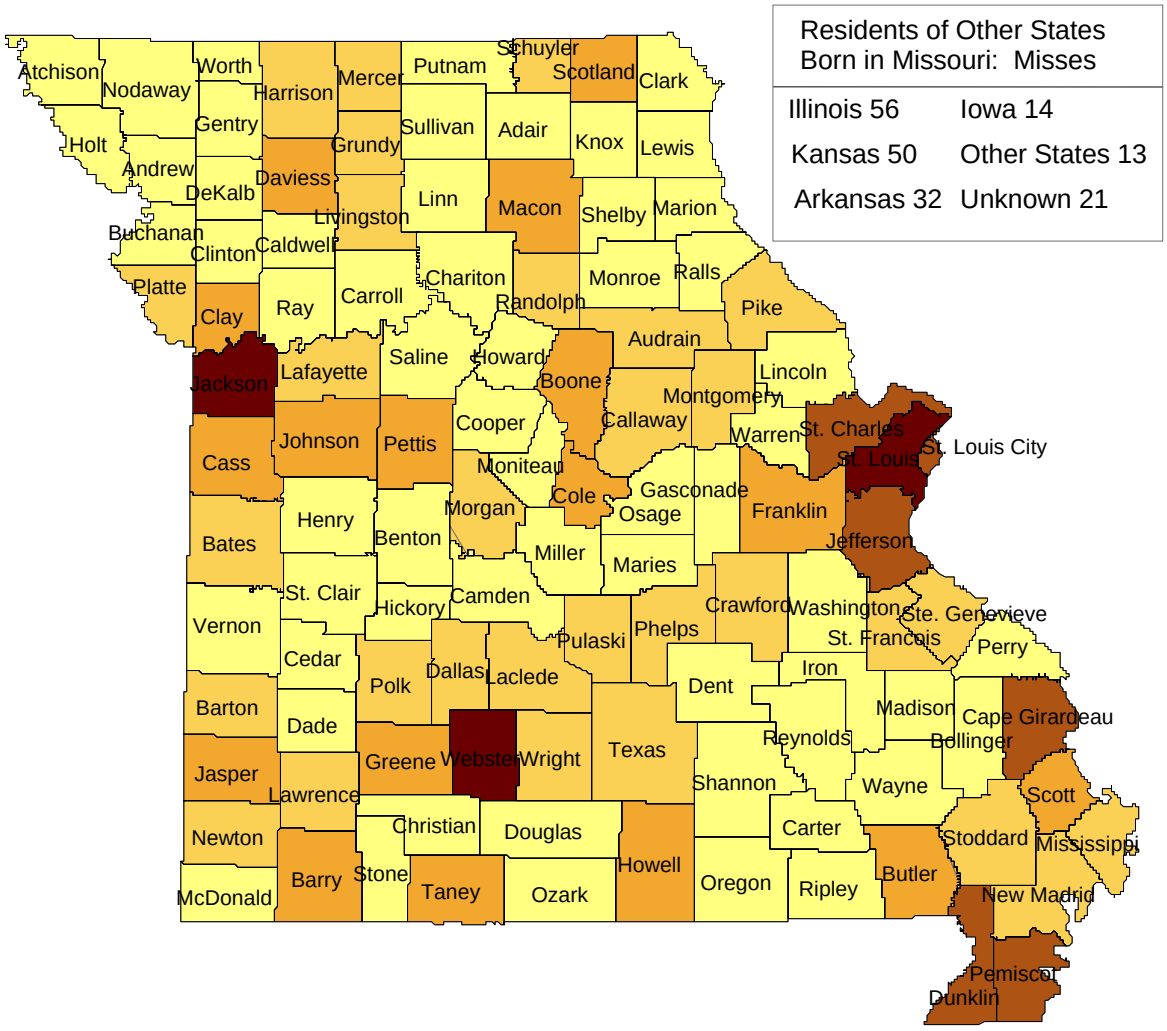
Geographic Follow-up of Significant Disease and Trait Conditions					
Significant Disease Conditions			**"S" Trait Conditions (includes repeats)**		
St. Louis Area	14	50%	St. Louis Area	647	56%
Kansas City Area	9	32%	Kansas City Area	322	28%
Remainder of MO	5	18%	Remainder of MO	186	16%
Total	**28**	**100%**	**Total**	**1155**	**100%**

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

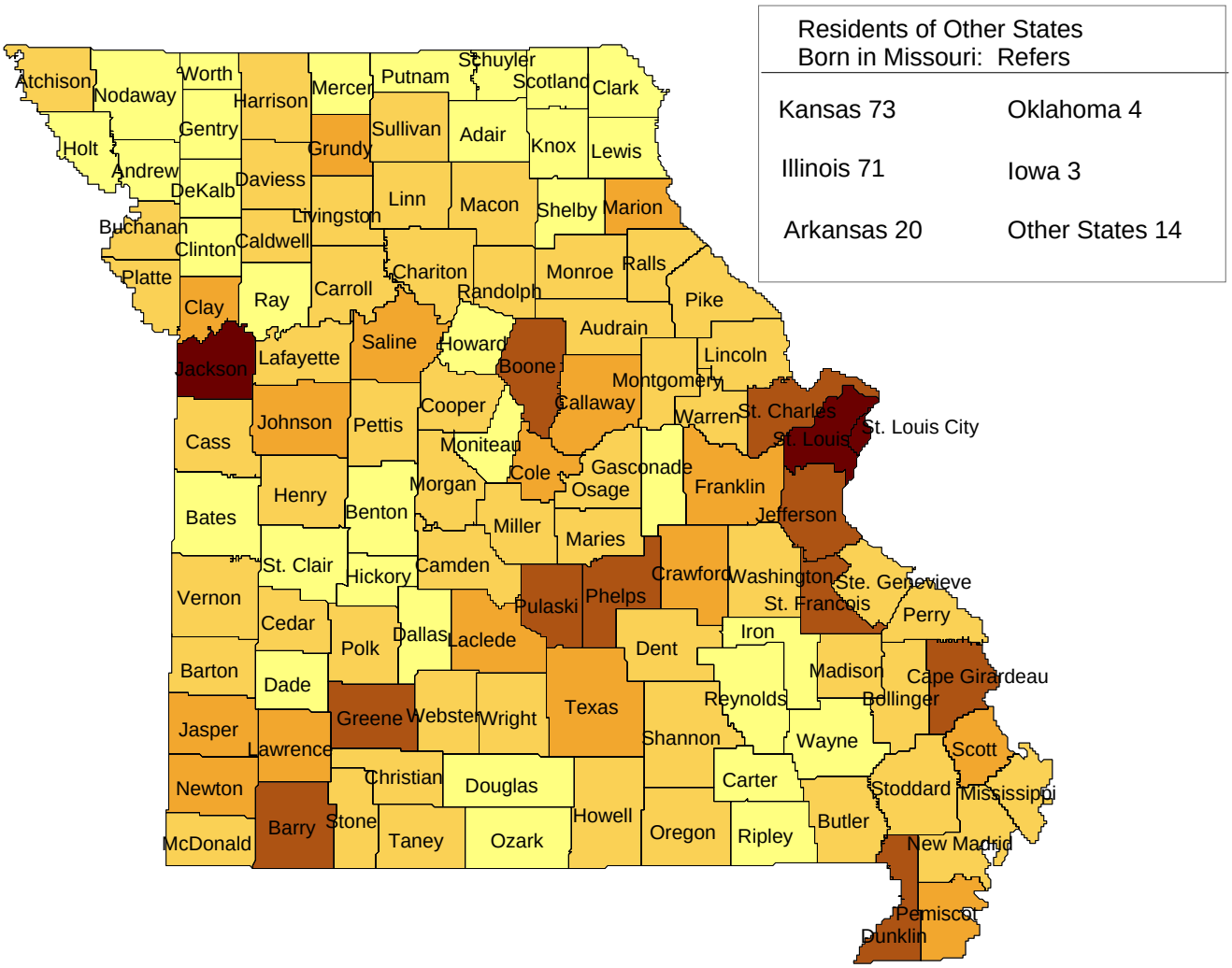
Appendix 6: 2007 Referrals from Missouri Newborn Bloodspot Screening Program



Appendix 7: 2007 Misses from Missouri Newborn Hearing Screening



Appendix 8: 2007 Refers from Missouri Newborn Hearing Screening



Appendix 9: 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 infants and children receiving services provided by the hemoglobinopathy resource centers was completed in 2007. Key findings:

Hemoglobinopathy Resource Center Satisfaction Survey - Parent Response			
	Very Satisfied	Satisfied	Not Satisfied
Treated with respect	88%	12%	0%
Treatment staff was knowledgeable	86%	14%	0%
Questions/concerns addressed in a timely manner	83%	17%	0%
Staff provided useful referrals and resources	81%	19%	0%
Provided with the services needed	83%	17%	0%
Medical care/services received	78%	22%	0%
Received services or treatment without experiencing any problems	99%	0%	1%

Newborn Hearing Screening Survey

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