MISSOURI
NEWBORN SCREENING

2011 Annual Report

Missouri Department of Health and Senior Services

Governor Jay Nixon
Gail Vasterling, 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 dedicated to helping 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
Missouri State Public Health Laboratory

Table of Contents

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.

The goal of Missouri's newborn screening program is for every newborn to be screened for certain harmful or potentially fatal disorders that aren't otherwise apparent at birth.

Newborn screening tests are required to be collected before a newborn leaves the hospital. These newborn screening samples are promptly delivered to the Missouri State Public Health Laboratory for testing. Babies are screened in an effort 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 screen, 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 collecting the blood on a filter paper. The paper is sent to the State Newborn Screening Laboratory for screening, and results are sent back to the hospital of birth and the physician of record. If results are considered to be out of normal range, the family will be contacted for further testing of the baby's blood.

Many changes have been instituted since newborn screening became a standard practice more than 45 years ago. Missouri and other states mandate newborn screening of all infants born within their border. Affected newborns typically appear normal at birth with no sign of any disorder until a developmental disability or death occurs. Upon detection of a condition, specialists formulate a plan of medical management that allows most affected newborns to develop normally. Newborn screening is a model for public health-based population genetic screening. It is recognized nationally and internationally as an essential public health program that provides for the best outcomes for the nation's most vulnerable population. Another newborn screening is a hearing screen. 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 tell the health care staff if further screening or an audiological assessment might be necessary.

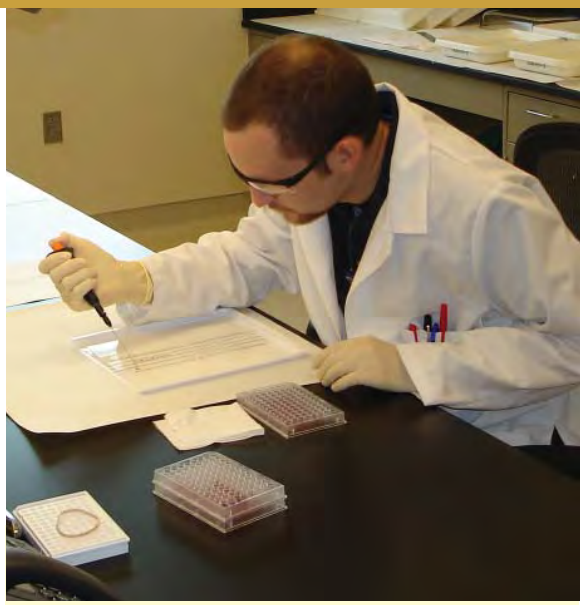
A number of accomplishments were achieved in 2011 in Missouri's Newborn Bloodspot Program. Among them were:

Newborn Screening Sample Storage Law Implementation
The Missouri Newborn Screening (NBS) Sample Storage Process was implemented on July 1, 2011. Missouri State Law (Section 191.317) requires the Missouri State Public Health Laboratory (MSPHL) to retain the leftover NBS samples for five years after the testing has been completed and then to destroy them after the five years of storage has ended. The law allows the department to release portions of the samples for the purpose of anonymous research, and allows the department to charge a reasonable fee for the use of such samples for anonymous research and for preparing and supplying samples for anonymous research proposals approved by the department. This new process is highlighted in the Screening Spotlight on page 5.

Follow-up of Newborns with Abnormal Results
Missouri successfully extended contracts with all of the genetic tertiary centers, the hemoglobinopathy centers, and cystic fibrosis centers to continue partnering with them in following-up on newborns having an abnormal newborn screen. The partnership has worked exceptionally well and has ultimately benefitted Missouri's families in achieving timely intervention in the care of their newborn. A total of 414 infants were referred to the genetic tertiary centers, cystic fibrosis centers and hemoglobinopathy centers to follow-up on infants considered to be high risk or moderate risk as a result of their newborn screening results. From these referrals, 165 infants were confirmed positive for a disorder.

Next Steps
In calendar year 2011, Missouri screened for all 29 core conditions recommended by the American College of Medical Genetics and the March of Dimes. When considering secondary conditions, a total of 66 disorders can be detected through newborn bloodspot screening.

Missouri will be closely monitoring the recommendations of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) in the coming years as recommendations for further screening are made. Newborn screening continues to evolve as advancements are made in the technology to detect disorders and as emerging treatments are discovered or known treatments are modified for treating people affected by these disorders.



In 2009, HB 716, known as the Brady Allen Cunningham Act was Truly Agreed to and Finally Passed. This bill requires the department to screen for the following five lysosomal storage diseases (LSDs) - Krabbe disease, Pompe disease, Gaucher disease, Niemann-Pick disease, and Fabry disease by July 2012. When these five screens are added to the newborn screening panel, Missouri will be among the first states to screen newborns for LSDs. The newborn screening blood spot panel will increase from 66 disorders to 71 disorders. It is estimated that there will be eight to 10 infants each year who will be confirmed positive for one of the five LSDs, but the true incidence is unknown until full population screening has been in place for a few years.

Missouri Newborn Screening Disorders

Newborn screening disorders tested and reported in Missouri are:

- Biotinidase deficiency (BIO)
- Classical galactosemia (GALT)
- Congenital adrenal hyperplasia (CAH)
- Congenital primary hypothyroidism (CH)
- Cystic fibrosis (CF)

- Amino Acid Disorders
  - Arginemia (ARG, arginase deficiency)
  - Argininosuccinate acidemia (ASA, argininosuccinase)
  - Citrullinemia type I (CIT-I, argininosuccinate synthetase)
  - Citrullinemia type II (CIT-II, citrin deficiency)
  - Defects of biopterin cofactor biosynthesis (BIOPT-BS)
  - Defects of biopterin cofactor regeneration (BIOPT-RG)
  - 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 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"
- Hemoglobin-C beta zero thalassemia disease
- Hemoglobin-C beta plus thalassemia disease
- Hemoglobin-E beta zero thalassemia disease
- Hemoglobin-E beta plus thalassemia disease
- Hemoglobin-H disease
- Homozygous beta zero thalassemia disease
- Homozygous-C disease
- Homozygous-E disorder
- Double heterozygous beta thalassemia disease

• Others
- Hearing

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.

For more details about any of the above mentioned disorders and how they are screened by the NBS Lab, visit the NBS Laboratory website at: http://health.mo.gov/lab/newborn.

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

Screening Spotlight: Missouri Newborn Screening Sample Storage and Release Policy

The Missouri Newborn Screening (NBS) Sample Storage Process was implemented on July 1, 2011. Missouri State Law (Section 191.317) requires the Missouri State Public Health Laboratory (MSPHL) to retain the NBS samples for five years after the testing has been completed and then to destroy them after the five years of storage has ended. The law allows the department to release the samples for the purpose of anonymous research, and allows the department to charge a reasonable fee for the use of such samples for anonymous research and for preparing and supplying samples for anonymous research proposals approved by the department.

This same law provides three opt-out/dissent options for the parents or legal guardian if they do NOT wish the department to release their child's leftover NBS sample for anonymous research. These options are provided to parents at the time of sample collection in an information sheet that is detached from the NBS sample collection card and provided to the mother of the newborn by the hospital staff. This information sheet is available in English, Spanish, Bosnian and Vietnamese.

The three opt-out options available to the parents after their child's NBS testing is completed are:

- Return the leftover sample to the parents
- Destroy the leftover sample in a scientifically acceptable manner
- Store the leftover sample for five years but do not release it for anonymous research

To opt-out of the sample storage and/or release, the parent must write the MSPHL and request the opt-out choice in writing, and they may do so at any time during the five year storage process. If the parent does not choose one of these options, the specimen will automatically be stored at MSPHL and may be released for approved anonymous research after the first three months storage time has transpired and until an opt-out letter is received by MSPHL. When MSPHL receives a letter from parents requesting one of the opt-out selections, their request is immediately granted. The MSPHL sends a letter back to the parents stating that their request was fulfilled along with a copy of their original opt-out letter. The MSPHL keeps both electronic and hard copy records of all opt-out cases.

There are numerous benefits to public health in retaining residual NBS samples. Residual NBS samples are the only available opportunity for a complete population study to be conducted since there is a sample

received on virtually every baby born. In addition to this, the NBS sample is sometimes the only remaining evidence available to the family from their child if their child becomes missing. The main benefits to NBS sample storage are:

- Quality assurance and improvement for the NBS laboratory.
- Research for new technologies and for detecting new disorders.
- Research for new treatments and cures for major childhood diseases.
- Population incidence research on disorders and environmental contaminant exposures.
- Parents can recall the specimens to help determine the cause of an unexplained death of their child such as sudden infant death syndrome (SIDS).
- Parents can recall the specimens to aid law enforcement in identifying their missing child.

The NBS Sample Storage and Release Subcommittee, a subcommittee of the Missouri Genetics Advisory Committee, reviews all research requests for stored NBS samples, determines priorities for the types of research proposals to be considered, and assures that only anonymous research is conducted. This committee determines if the MSPHL should proceed with sending the request through the Department of Health and Senior Services (DHSS) Institutional Review Board (IRB) for review and final approval.

If the research request is approved by the DHSS IRB, MSPHL will contact the researcher that made the request and provide an estimate of the cost for processing their research request. The MSPHL will provide approved researchers with small punches from the samples, and no records will be kept of which samples were used for any particular research request. No samples will be provided for research projects until they have been stored for at least three months to allow time for parents who wish to write MSPHL to opt-out.

The DHSS will maintain an information page on the NBS website displaying all the anonymous research projects that were granted approval for using stored NBS samples, the parent opt-out process and other related information regarding NBS sample storage and release.

For more details on Missouri Newborn Screening Sample Storage and Release Policy, please visit the NBS Laboratory website at http://health.mo.gov/lab/newborn/pdf/nbsstoragereleasepolicy.pdf

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

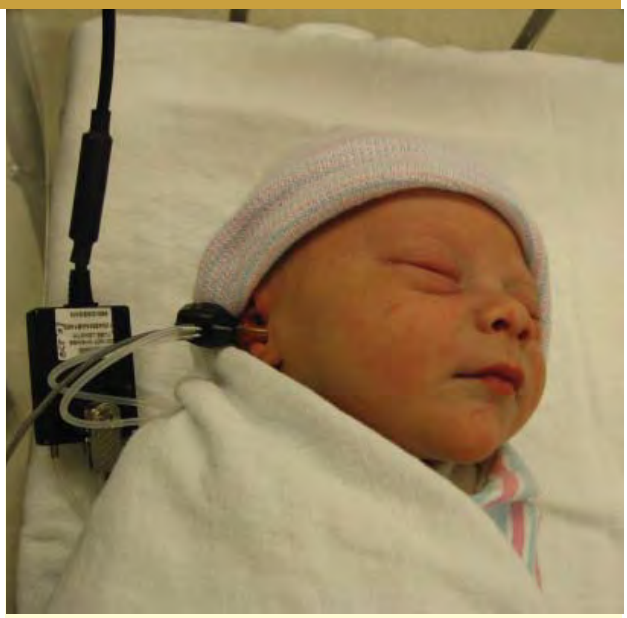
The Centers for Disease Control and Prevention (CDC) recommends that all infants be screened for hearing loss by one month of age. Infants who screen positive for possible hearing loss should receive an audiologic evaluation by 3 months of age, and infants with confirmed hearing loss should receive early intervention services by 6 months of age.

Provisional 2011 calendar year data for Missouri show:

- 77,135 occurrent births (source: DHSS Vital Records)
- 76,918 occurrent births (source: Missouri Health Strategic Architectures and Information Cooperative [MOHSAIC]*)
- 98.2 percent (75,544) screened
- 98.2 percent (74,212) screened by 1 month of age
- 3.6 percent (2,718) failed their final screening
- 45.4 percent (612) of the infants that failed their final screening received audiologic evaluation by 3 months of age
- 121 infants diagnosed with a permanent hearing loss (three lived out-of-state)
- 73 enrolled in Missouri's Part C of the Individuals with Disabilities Act program, First Steps
- 53.4 percent (39) of the infants enrolled in First Steps did so by 6 months of age

*The difference of 217 births between the occurrent birth count in the program data management system, MOHSAIC, and the total occurrent births reported by Vital Records is a result of records that do not yet have a Department Client Number (DCN) assigned. Records are not released from the Vital Records system to MOHSAIC until the assignment is complete. Non-complete records are due to issues such as paternity and adoptions. This report is based upon MOHSAIC records.

The Missouri Newborn Hearing Screening Program's (MNHSP) MOHear Project continues to make great strides. Funded by a Health Resources and Services Administration (HRSA) grant beginning in 2009, the MOHear Project consists of: 1) unbiased service coordination for families of newborns diagnosed with severe to profound permanent hearing loss; and 2) linkage of newborns to services by regionally-focused efforts at resolving loss to documentation and follow-up at each step of the early hearing detection and intervention process. The effort involves collaboration



The Centers for Disease Control and Prevention recommends that all infants be screened for hearing loss by one month of age.

with the Missouri Department of Elementary and Secondary Education (DESE) and a contract with Missouri State University (MSU) for management of the MOHear Project.

More than 98 percent of newborns in Missouri were screened for hearing loss by one month of age.

Six professionals comprise the MOHear Project – one manager and five MOHears. The MOHears are professionals (either audiologist, educator of deaf, or speech pathologist) with expertise in the unique needs of infants with hearing loss. In collaborations with DESE's Part C of the Individuals with Disabilities Education Act (IDEA) program known as First Steps, System Point of Entry (SPOE) staff contact MOHears to participate in initial intake interviews with the families of infants recently diagnosed with a permanent hearing loss. In 2011, SPOES utilized MOHears on 38 occasions.

The MOHear work at reducing loss to follow-up resulted in a steady decrease in loss to follow-up percentages. The loss to follow-up rate following an infant's final failure to pass a hearing screening (known as a "refer" screening result) decreased from 58.8 percent in 2009, to 40.8 percent in 2010, to 34.8 percent in 2011. Activities the MOHears engage in to resolve and prevent loss to follow-up include the following: phone calls to families believed to be lost to follow-up, individual outpatient screenings, rescreening clinics, visits to hospitals to review procedures and conduct trainings, equipment loans, and contact with audiology clinics in order to solicit recent diagnostics.

Also in 2011, MNHSP partnered with the National Initiative for Children's Healthcare Quality (NICHQ) and HRSA to participate in a multi-state learning collaborative aimed at enhancing the performance of early hearing detection and intervention (EHDI) programs throughout the country. The improvement effort, known as "Improving Hearing Screening and Intervention System," used collaborative learning approaches with quality improvement methodologies to test and implement new ways to improve the quality and timeliness of screening, audiologic diagnosis and entry into intervention. Missouri tested and implemented strategies to improve system performance based on promising changes recommended by experts and identified by past collaborative teams. The MNHSP used NICHQ process of working from an initial theory to a "small test of change," to a wider test of change, to implementation, and finally to a change that results in improvement. Collaborating with parents, screening hospital programs, audiology clinics and early intervention (EI) programs, MNHSP developed some best practices for Missouri EHDI.

Changes in Missouri EHDI occurred as a direct result of the NICHQ learning collaborative. The MNHSP made internal modifications in process and procedure, and began inclusion of "Sherri's Letter" (a letter written by a Missouri parent of a child with hearing loss) and a Missouri Parent Road Map (a detailed list of further action to take) in follow-up letters sent by MNHSP to parents of children who do not pass the hearing screening and need to return for rescreening or testing. Small tests in the use of the "Sherri's Letter" showed that parents could more easily relate to the experiences of another parent than the information provided in the MNHSP parent letter. Furthermore, participating screening programs and audiology clinics adopted the practice of documenting that pre-appointment instructions had been given verbally and in writing to parents, thereby increasing the chance the follow-up recommendations would be followed, documented and reported to MNHSP. Finally, MNHSP began working with DESE to develop an interagency agreement for sharing identifiable early intervention

enrollment data. An interagency agreement with DESE ensures one definition of enrollment into First Steps and clarifies the type of information to be shared between the agencies.

The MNHSP conducted its biennial parent satisfaction survey in 2011. The MNHSP mailed 500 surveys and received 105 replies (21.0 percent return rate). Survey recipients were families who had a child without risk factors for hearing loss and who referred on the initial hearing screening in the three months prior to mailing. Key programmatic findings included:

- 66 percent of the respondents reported that the birth hospital provided written information about the hearing screening prior to the hearing screening
- 91 percent of the respondents reported the hospital notified them of the results of their baby's hearing screening
- 66 percent of the respondents reported that hospital staff explained the importance of knowing whether a baby has a hearing loss early in life

Next Steps

The MOHear Project will expand its outreach by inviting diagnosing audiologists to refer to the MOHear Project by:

- Informing the family of the MOHear Project
- Offering to have MOHear call the family
- Providing MOHear flyers to families
- Recommending MOHear on the electronic diagnostic result form MOHSAIC

Each of these tactics would lead to the initiation of MOHear contact with the family.

The MNHSP will continue to engage in improvement activities using the "small tests of change" technique learned in the NICHQ Collaborative. The MNHSP will explore ways to implement some of the NICHQ-identified processes known to reduce loss to follow-up such as ensuring hospital screening programs identify on the result form the name of the physician who will undertake the pediatric care of the infant, and schedule appointments for rescreening and diagnostic evaluations.

The MNHSP will continue development of an interagency agreement with DESE to clearly define the responsibilities of DESE and DHSS regarding the coordinated provision of appropriate early intervention service for infants and toddlers suspected of or diagnosed with a permanent hearing loss. Additionally, the MNHSP and First Steps will work together to create process documents. The documents will provide specific instructions for obtaining parental release of information and sharing identifiable information with MNHSP as well as specific instructions for ensuring parents are informed of the MOHear Project and understand how to have a MOHear participate in their first meeting with the First Steps SPOE staff.

Newborn Screening Contact Information

Telephone Contacts:

Newborn Screening Laboratory main number	573-751-2662
Order newborn screening specimen forms	573-751-3334
Genetics and Healthy Childhood, follow-up information	800-877-6246

Web Addresses:

Newborn Screening Laboratory – http://health.mo.gov/lab/newborn

Newborn Screening Program – http://health.mo.gov/newbornscreening

Newborn Hearing Screening Program – http://health.mo.gov/newbornhearing

Appendix 1: Disorders Confirmed for 2011 and Projected Incidence Rates

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Amino Acid Disorders	**16**	**1/8,000***
Arginemia		
Argininosuccinate acidemia		
Citrullinemia type I	1	
Citrullinemia type II		
Defects of biopterin cofactor biosynthesis		
Defects of biopterin cofactor regeneration		
Homocystinuria		
Hypermethioninemia		
Hyperphenylalaninemia	3	
Hyperphenylalaninemia, benign	1	
Maple syrup urine disease		
Maternal PKU		
Phenylketonuria (PKU)	9	
Tyrosinemia type I		
Tyrosinemia type II	1	
Tyrosinemia type III	1	
Biotinidase Deficiency (BIOT)	**8**	**1/40,000***
Partial biotindase deficiency	3	
Profound biotindase deficiency	5	
Galactosemia (GALT)	**12**	**1/50,000****
Classical galactosemia	2	
Duarte galactosemia	10	
Congenital adrenal hyperplasia (CAH)	**7**	**1/13,000**
Congenital primary hypothyroidism (CH)	**43**	**1/3,000**
Cystic fibrosis (CF)	**18**	**1/4,000**
Fatty Acid Oxidation Disorders	**17**	**1/10,000***
Carnitine acylcarnitine translocase deficiency		
Carnitine uptake deficiency	2	
Carnitine palmitoyl transferase deficiency I		
Carnitine palmitoyl transferase deficiency II	1	
Dienoyl-CoA reductase deficiency		
Glutaric acidemia type II		
Long-chain hydroxyacyl-CoA dehydrogenase deficiency		
Medium-chain acyl-CoA dehydrogenase deficiency	7	
Medium-chain ketoacyl-CoA thiolase deficiency		
Medium/Short chain L-3 hydroxy acyl-CoA dehydrogenase deficiency	0	

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Short-chain acyl-CoA dehydrogenase deficiency	5	
Trifunctional protein deficiency		
Very-long chain acyl-CoA dehydrogenase deficiency	2	
Organic Acid Disorders	**9**	**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		
Isovaleric acidemia	1	
Malonic acidemia	1	
Methylmalonic acidemia (CBL A,B; vitamin B12 disorders)	1	
Methylmalonic acidemia (CBL, C,D)		
Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)		
Multiple carboxylase deficiency	1	
Propionic acidemia	1	
Forminioglutamic acid (FIGLU) not a disorder on the newborn screening panel but is found	2	
Secondary aciduria, undetermined metabolic disorder		
Hemoglobinopathies	**35**	**1/1,700***
Sickle cell anemia disease (Hb S/S)	16	**1/3,000** Total population **1/400** African-American population
Sickle hemoglobin-C disease (FSC)	11	
Sickle beta zero thalassemia disease		
Sickle beta plus thalassemia disease (FSA)		
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)	2	
Hemoglobin-C beta zero thalassemia disease		
Hemoglobin-C beta plus thalassemia disease	5	
Homozygous-E disorder (FE)	1	

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
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)		
Other (FSX) compound heterozygous Hb S and G-Taipei		

* Combined incidence of all disorders in this category.
**Incidence only for classical galactosemia.

Appendix 2: Newborn Screening Laboratory Report – Samples Received 2011

	Newborn Samples Received			
	Initial	Repeat	Poor Quality Samples	Total Infant Samples
Jan	6,054	958	142	7,154
Feb	5,766	833	143	6,742
Mar	6,517	1,135	138	7,790
Apr	5,895	1,037	94	7,026
May	6,009	980	87	7,076
Jun	6,999	1,226	114	8,339
Jul	6,350	1,112	96	7,558
Aug	7,429	1,271	129	8,829
Sep	6,569	1,186	100	7,855
Oct	6,138	1,120	110	7,368
Nov	6,219	998	157	7,374
Dec	6,029	1,104	130	7,263
Y.T.D.	**75,974 (84.07%)**	**12,960 (14.34%)**	**1,440 (1.59%)**	**90,374**

Appendix 3: Newborn Screening Laboratory Report – Abnormal Results 2011

Disorder		Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec	Y.T.D.
BIO	Confirmed	0	1	0	1	1	2	1	0	0	1	0	1	8
	High Risk	0	1	0	2	4	4	1	0	0	2	0	1	15
	Borderline Risk	6	2	2	3	2	2	4	3	1	2	1	0	28
CAH	Confirmed	1	0	1	2	0	0	0	0	2	0	1	0	7
	High Risk	2	3	1	3	1	1	1	2	4	1	3	1	23
	Borderline Risk	38	44	39	35	39	38	34	55	31	43	43	58	497
CF	Confirmed	1	1	1	4	1	0	0	2	1	2	2	3	18
	Referred	7	7	12	9	12	8	3	15	13	5	10	11	112
	Initial IRT	43	29	46	35	38	35	19	53	37	34	36	39	444
CH	Confirmed	2	4	4	6	4	5	4	4	3	1	3	3	43
	High Risk	3	6	5	6	6	7	4	4	3	1	4	3	52
	Borderline Risk	82	78	88	80	71	127	91	128	107	89	97	96	1134
GAL	Confirmed	0	2	4	0	1	2	2	1	0	0	0	0	12
	High Risk	0	2	4	0	1	3	3	2	0	1	0	0	16
	Borderline Risk	3	3	4	2	8	4	13	8	2	1	2	2	52
AA	Confirmed	1	0	1	0	1	4	1	3	2	1	0	2	16
	High Risk	1	1	1	0	1	3	1	3	2	1	0	1	15
	Moderate Risk	1	0	0	1	2	2	0	0	3	1	0	2	12
	Low Risk	81	71	84	81	70	53	61	47	51	43	42	54	738
OA	Confirmed	1	2	1	0	1	0	1	1	0	1	1	0	9
	High Risk	1	2	0	0	1	0	0	2	0	1	0	0	7
	Moderate Risk	0	0	2	3	0	0	2	2	3	3	0	3	18
	Low Risk	20	14	36	31	49	34	35	23	36	30	41	53	402
FA	Confirmed	1	1	2	1	0	3	2	2	0	0	0	5	17
	High Risk	0	1	3	1	1	1	0	0	0	0	2	3	12
	Moderate Risk	2	0	5	0	7	1	6	2	1	0	3	2	29
	Low Risk	32	28	50	31	38	56	56	51	54	41	43	57	537
Hb	Sickle Cell Disease	1	1	2	3	1	3	2	4	2	4	2	2	27
	Other Hemoglobinopathies	0	2	1	1	1	1	0	0	0	0	1	1	8
	Abnormal Traits	122	130	130	130	95	143	111	137	136	124	169	154	1581

BIO = biotinidase deficiency
CAH = congenital adrenal hyperplasia
CF = cystic fibrosis
CH = congenital hypothyroidism
GAL = galactosemia
AA = amino acid
OA = organic acid
FA = fatty acid
Hb = Hemoglobinopathies

Total Confirmed **165**

Outcome Data - Newborn Screening Samples and Results

- In 2011 there were 75,974 initial samples tested in the state newborn screening laboratory. There were a total of 90,374 blood spot samples received in the laboratory. Samples received included:

Initial	Repeat	Poor Quality Samples
75,974	12,960	1,440

- In the process of screening newborns for 66 genetic and metabolic conditions, it is the newborn screening laboratory's role to assess the risk of any abnormal screening results by evaluating the marker analytes present and the levels that were detected. This risk assessment then dictates different levels of action and follow up protocols. The three categories of risk and the number of test results falling in these categories during 2011 were:

High Risk	Moderate Risk	Low/Borderline Risk
287 (0.38%)	59 (0.07%)	3,884 (5.1%)

High Risk - Results are immediately phoned and faxed to the physician of record and to the contracted genetic referral centers for consultation and confirmatory testing. Final laboratory reports are mailed to the facility that submitted the sample and the physician of record.

Moderate Risk - Results are immediately phoned and faxed to the physician of record and to the contracted genetic referral centers for consultation and confirmatory testing. Final laboratory reports are mailed to the facility that submitted the sample and the physician of record.

Low/Borderline Risk – Final laboratory results are mailed to the physician of record and submitting facility and a repeat newborn screen is necessary.

- During 2011, 165 confirmed disorders were diagnosed from these abnormal newborn screening results.

Appendix 4: 2011 Poor Quality Samples

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.	103
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.	435
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.	36
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.	445
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.	265
OTHER	1
OLD SPECIMEN Specimen greater than 15 days old when received at State Public Health Laboratory.	42
NO BLOOD Filter submitted without blood.	4
OLD FORM Sample received on out-of-date form.	11
FILTER AND FORM BARCODES DO NOT MATCH Barcode on filter does not match barcode 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.	1
MISSING OR INCOMPLETE PATIENT INFORMATION Missing or incomplete demographic information.	10
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.	50
BLOOD ON OVERLAY COVER Overlay cover came in contact with wet blood specimen. Possible causes: Sample is poor quality status 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. It is very important that the wet filter paper does not come in contact with any surface until completely dry.	37
Total Poor Quality Samples Received	1,440 (1.59%)

Appendix 5: Hemoglobinopathy Report 2011

Specimens Received:

Initial:	75,974	(83.8%)
Repeat:	12,960	(14.3%)
Unsatisfactory:	1,440	(01.6%)
Whole Blood:	235	(00.3%)
Total:	**90,609**	

Significant Screening Results = 1,616					
Sickle Cell Disease		**Other Disease Conditions**		**Trait Conditions**	
FS	16	FC	2	FAS	1,021
FSC	11	FCA	5	FSAINC	58
		FE	1	FAC	273
				FCAINC	11
				FAE	32
				FAD	43
				FAX	136
				FACX	2
				Slightly Elevated Barts	2
				Other Trait Condition	3
Total	**27 (1.7%)**	**Total**	**8 (0.5%)**	**Total**	**1,581 (97.8%)**

Geographic Follow-up of Significant Disease

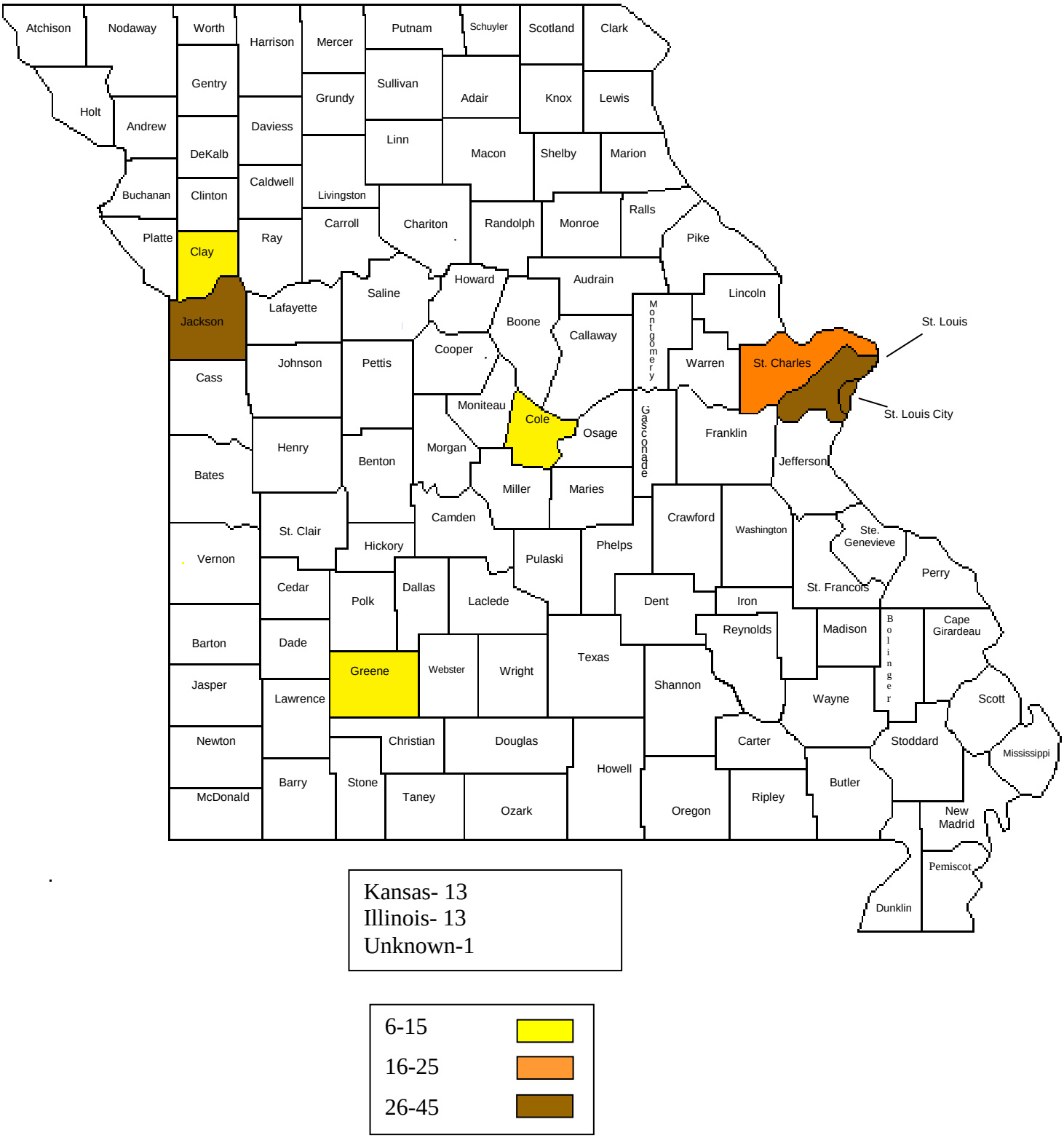
Significant Disease Conditions		
St. Louis Area	25	71%
Kansas City Area	7	20%
Remainder of Missouri	3	9%
Total	**35****	**100%**

**See Appendix 1

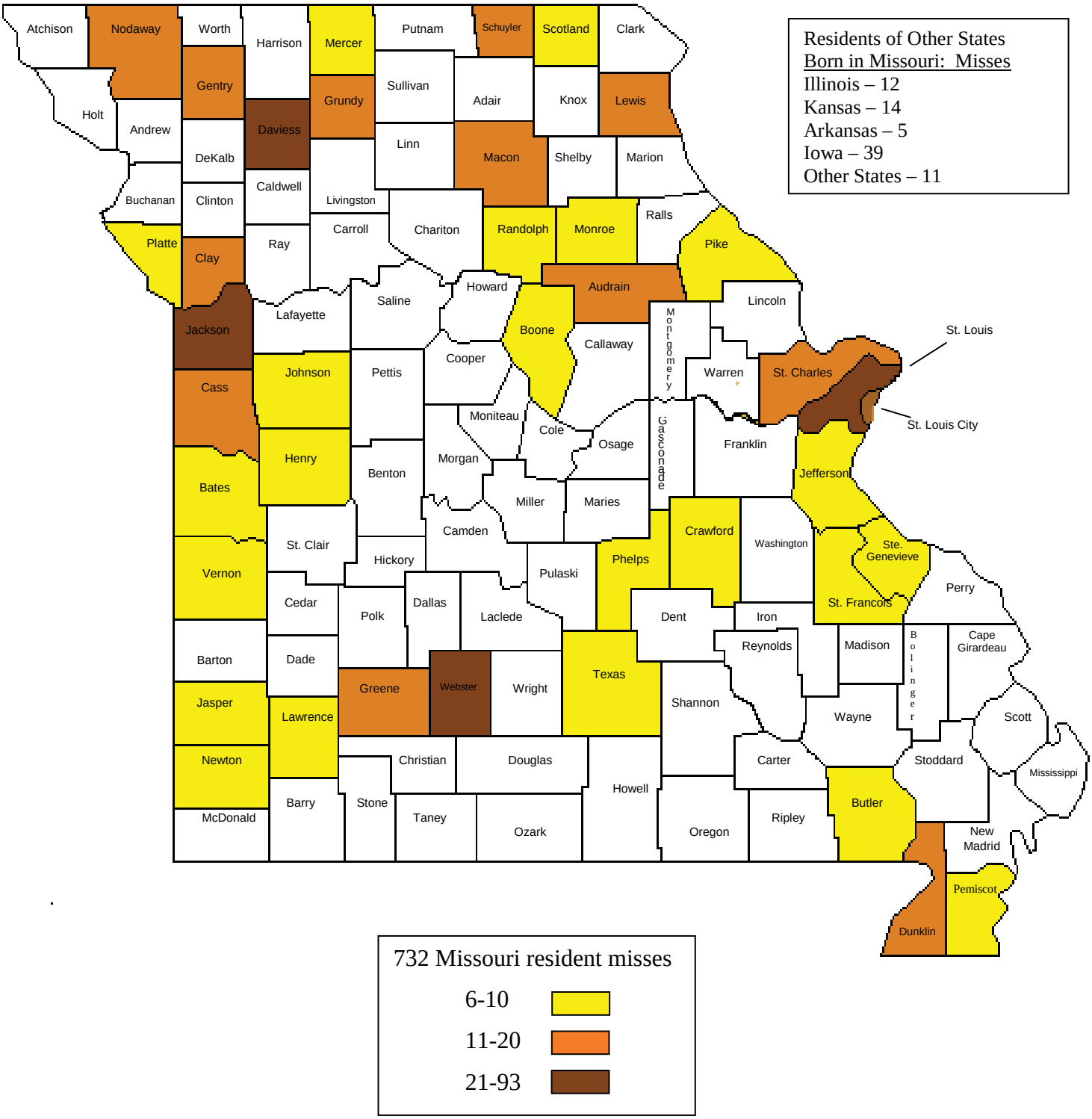
Hemoglobinopathies	**35**	
Sickle cell disease (Hb S/S)	16	**1/3,000** Total population; **1/400** African-American population
Sickle hemoglobin-C disease	10	
Sickle beta zero thalassemia disease		
Sickle beta plus thalassemia disease		
Sickle hemoglobin-D disease		
Sickle hemoglobin-E disease		
Sickle hemoglobin-O-Arab disease		
Sickle hemoglobin Lepore Boston disease		
Sickle HPFH disorder		
Sickle “Unidentified”		
Homozygous-C disease	1	
Hemoglobin-C beta zero thalassemia disease		
Hemoglobin-C beta plus thalassemia disease	2	
Homozygous-E disorder	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		

Five lost to follow-up (1 FSC, 3 FCA, 1 FC)

Appendix 6: 2011 Refers from Missouri Newborn Bloodspot Screening Program

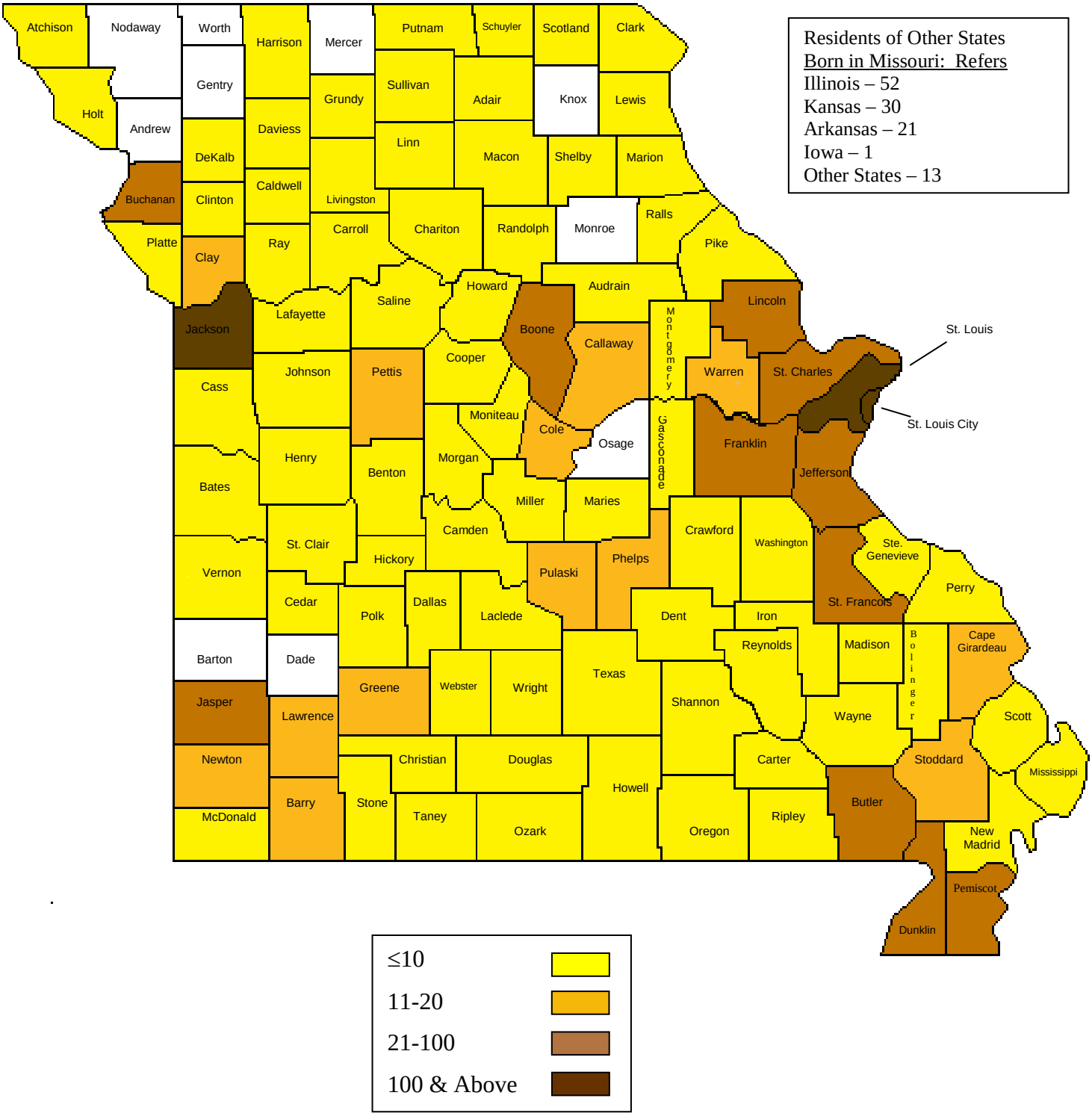


Appendix 7: Newborn Hearing Screening
2011 Misses* from Missouri



*Misses are those babies with no record of a hearing screen result.

Appendix 8: Newborn Hearing Screening
2011 Refers from Missouri



Appendix 9: Newborn Screening Satisfaction Surveys

A satisfaction survey of parents was conducted for families of babies having abnormal newborn screening results reported in 2011. Key findings:

Newborn Screening Parent Satisfaction Survey			
	Very Satisfied	Satisfied	Not Satisfied
I was treated with respect.	100%		
My questions and concerns were addressed in a timely manner.	100%		
The staff provided me with useful referrals and resources.	83%	17%	
I was provided with the services I needed.	100%		
Overall satisfied with quality of care.	100%		

A satisfaction survey of parents of infants and children receiving services provided by the hemoglobinopathy resource centers was completed in 2011. Key findings:

Hemoglobinopathy Resource Center Satisfaction Survey - Parent Response			
	Very Satisfied	Satisfied	Not Satisfied*
Treated with respect.	95%	5%	0%
Treatment staff was knowledgeable.	91%	9%	0%
Questions/concerns addressed in a timely manner.	83%	16%	1%
Staff provided useful referrals and resources.	77%	20%	3%
Provided with the services needed.	89%	9%	2%
Medical care/services received.	87%	11%	2%
Received services or treatment without experiencing any problems.	95%	0%	5%

*The reasons parents responded as “not satisfied” with services were because of long wait time.

Appendix 10: Newborn Hearing Screening Survey

A satisfaction survey of parents of children born in Missouri who failed their initial newborn hearing screening between October 2011 and December 2011 was completed in March 2012. The survey examined factors influencing the follow-up time between a failed newborn hearing screening and a repeat screening or an audiologic evaluation.

Key Findings

- 66 percent of the respondents reported that the birth hospital provided them with written information about the hearing screening prior to the hearing screening (an increase of 1 percent from the 2009 survey).
- 91 percent of the respondents reported that the birth hospital notified them of the screening result (an increase of 17 percent from the 2009 survey).
- 66 percent of the respondents reported that the hospital staff explained the importance of knowing whether a baby has a hearing loss early in life. (This question will be asked again in the 2013 survey.)

Missouri Department of Health and Senior Services
P.O. Box 570
Jefferson City, MO 65102
www.health.mo.gov