MISSOURI NEWBORN SCREENING

2014 Annual Report



Missouri Department of Health and Senior Services

Governor Jay Nixon

Acknowledgments

The Missouri State Genetic Advisory Committee and its ancillary Newborn Screening Standing Committee, Sickle Cell Standing Committee, Cystic Fibrosis Standing Committee, Newborn Hearing Screening Standing Committee, and the Lysosomal Storage Disorder Task Force 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.

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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

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Missouri Newborn Screening Disorders

Newborn screening disorders tested and reported in Missouri are as follows:

- 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)

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.

- Lysosomal Storage Disorders
  - Fabry Disease
  - Gaucher Disease
  - Hurler Syndrome
  - Krabbe Disease
  - Pompe Disease

- Organic Acid Disorders
  - 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
  - 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)
  - 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 hereditary persistence of fetal hemoglobin (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

- Other
  - Critical Congenital Heart Disease
  - Hearing

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

The Missouri Newborn Screening (NBS) 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 on any of the above-mentioned disorders and how they are screened by the NBS Laboratory, please visit the State NBS Laboratory website at: http://health.mo.gov/lab/newborn/

Screening Spotlight: Screening Newborns for Critical Congenital Heart Disease

During the 2013 legislative session, Missouri lawmakers voted to pass Senate Bill 230, otherwise known as Chloe's Law. The new law would require all babies born in Missouri to be screened for critical congenital heart disease (CCHD) beginning January 1, 2014. Chloe, for whom the law was named, was born in Missouri in 2009 and was nearly sent home from the hospital without her CCHD being detected. However, due to her mother's intuition and persistence, Chloe was diagnosed and received life saving treatment. Chloe's Law now requires all babies to be screened shortly after birth in order to help make sure babies with CCHDs can receive the same timely diagnosis and treatment.

Congenital heart defects are the most common birth defect found among babies born in the United States and they are the leading cause of birth defect-associated infant illness and death. Approximately 1% or 40,000 babies are born each year with a congenital heart defect. About one in every four babies born with a heart defect is diagnosed with critical congenital heart disease (CCHD). CCHD occurs when a baby's heart or major blood vessels do not form correctly, causing a defect. There are many different types of heart defects that range from mild to severe. Babies with "critical" heart defects need urgent treatment, which may include medicine or surgery. If left untreated, these defects can lead to death or can cause serious developmental delays.

Screening newborns for CCHD is important because while prenatal ultrasounds may detect some cases of CCHD, not all CCHDs can be detected before birth. Without screening shortly after birth, babies with CCHD are sometimes sent home without care because they appear healthy. At home, these babies can develop serious health problems within the first few days or weeks of life and often require emergency care. Babies with a missed or undiagnosed CCHD are at risk for cardiogenic shock or death. Cardiogenic shock is a condition where the heart has been damaged to the point where it is unable to supply enough blood to meet the body's needs. Those babies that do survive are at a much greater risk for permanent damage and developmental delays. If CCHD is detected early, however, infants can be treated early and lead healthier lives.

The pulse oximeter uses an infrared light sensor that is gently wrapped around the baby's right hand and one foot.

CCHD screening is a simple test that can be done at the bedside to determine the amount of oxygen in the baby's blood. Low oxygen levels can be a sign of CCHD. The test is done using a machine called a pulse oximeter. The pulse oximeter uses an infrared light sensor that is gently wrapped around the baby's right hand and one foot. Light passes through the skin and tissue and is read by the sensor to estimate the blood oxygen level. The test is painless and takes just a few minutes.

CCHD screening should be performed 24 to 48 hours after birth or before discharge from the hospital. If a baby is born at home or in a birthing center, parents should initiate discussions with the midwife or the baby's doctor to determine how and when a CCHD screen will be completed. Screening should be done while baby is warm, calm and awake. If the baby is crying, moving, fussing or cold; the screening will take longer and may need to be repeated.

A healthy baby may have a low oxygen reading. Babies with low oxygen levels may have a CCHD. Other conditions like breathing problems or infections may also cause a low blood oxygen level. If a baby has a low oxygen reading, the health care provider will check the baby carefully. An ultrasound of the heart (also called an echocardiogram or "echo") may be done to look for a CCHD. The echocardiogram may be done in a hospital or a doctor's office. It will need to be read by a children's heart doctor (pediatric cardiologist). If the echocardiogram shows a problem, the medical team will discuss the next steps with the parents.

Most babies who pass the CCHD screening will not have a CCHD. It is important to know that screening cannot identify every child with a heart problem. Parents should watch for the following warning signs:

- Bluish color to the lips or skin
- Grunting
- Fast breathing
- Poor feeding
- Poor weight gain
- Sweating around the forehead – especially during feeding

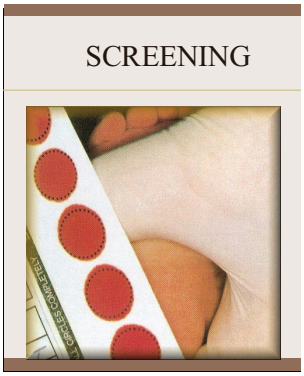
If you see any of these signs, contact your baby's health care provider right away!

For more information on CCHDs during the newborn period, please go to http://health.mo.gov/living/families/genetics/birthdefects/cchd.php

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.

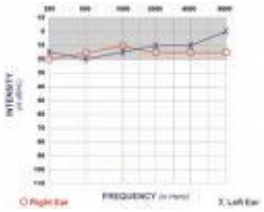
- Baby's physician consults with the specialist appropriate to the condition.

4: TREATMENT & MANAGEMENT

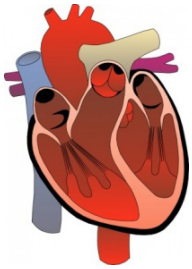
- Once diagnosis is made, treatment begins. For some life-threatening conditions, treatment may occur prior to diagnosis – on the recommendation of a specialist.

- 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

The Newborn Hearing Screening Process 7

1: SCREENING	2: FOLLOW-UP	3: EVALUATION	4: INTERVENTION
Baby is born. Hospital screens for hearing loss and checks for risk factors for late onset hearing loss prior to discharge.  Hospital submits results to the Missouri Department of Health and Senior Services (DHSS) via the Missouri Electronic Vital Records (MoEVR) system or on a paper form.  DHSS retrieves results from the Missouri Health Strategic Architectures and Information Cooperative (MOHSAIC) data system. 	Hospital reports results to parents and baby's physician.  DHSS sends letters to parents and physicians of newborns who did not pass or who missed the screening.  Parents return baby to hospital/health care provider 1-3 weeks after initial referral. 	Audiologist evaluates babies that don't pass a hearing screening by 3 months of age.  Audiologist reports evaluation results to DHSS.  Audiologist identifies risk factors and makes recommendations.  DHSS sends letter to families of children diagnosed with permanent hearing loss and refers to Missouri's Part C of the Individuals with Disabilities Education Act (IDEA) program, First Steps.	Babies diagnosed with permanent hearing loss enroll in First Steps (early intervention service) by 6 months of age.  Babies receive services from the following as appropriate: Primary Care Physician, Otolaryngologist, Geneticist, and Ophthalmologist.  Baby may be a candidate for: hearing aids, cochlear implant, sign language instruction, or speech and language services. 

The Newborn Critical Congenital Heart Disease Screening Process

1: SCREENING	2: FOLLOW-UP	3: EVALUATION	4: INTERVENTION
• Baby is born.  • Hospital or midwife screens for critical congenital heart disease (CCHD) between 24 and 48 hours after birth or prior to discharge. • Screening should be in accordance with the American Academy of Pediatrics and American Heart Association guidelines.  • Screening should be done while baby is warm, calm, and awake. 	• If screening is normal, no further action is necessary. • If baby does not pass the screening, further evaluation will be necessary and the primary care provider should be contacted as soon as possible. 	• The baby's primary care provider will perform a thorough physical examination to rule out any non-cardiac issues that may have prevented baby from passing the CCHD screen.  • An echocardiogram may be done to look for a CCHD.  • The echocardiogram should be read by a pediatric cardiologist 	• Babies diagnosed with CCHDs will typically require surgical or catheter intervention within the first year of life.  • Parents will receive treatment guidelines and education.  • Babies may receive services from the following as appropriate: primary care provider, pediatric cardiologist, geneticist, nurse, nutritionist, pharmacist, social worker, and child life specialist. 

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, for follow-up information	800-877-6246

Web Addresses:

Critical Congenital Heart Disease –
http://health.mo.gov/living/families/genetics/birthdefects/cchd.php

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

Newborn Screening Program –
http://health.mo.gov/living/families/genetics/newbornscreening/index.php

Newborn Hearing Screening Program –
http://health.mo.gov/living/families/genetics/newbornhearing/index.php

Appendix 1: Disorders Confirmed for 2014 and Projected Incidence Rates

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Amino Acid Disorders	**7**	**1/11,000***
Arginemia		
Argininosuccinate acidemia	1	
Citrullinemia type I		
Citrullinemia type II		
Defects of biopterin cofactor biosynthesis		
Defects of biopterin cofactor regeneration		
Homocystinuria		
Hypermethioninemia		
Hyperphenylalaninemia	1	
Hyperphenylalaninemia, benign		
Maple syrup urine disease		
Maternal PKU		
Phenylketonuria (PKU)	4	
Tyrosinemia type I		
Tyrosinemia type II	1	
Tyrosinemia type III		
Biotinidase deficiency (BIOT)	**9**	**1/8,600***
Partial biotinidase deficiency	6	
Profound biotindase deficiency	3	
Congenital adrenal hyperplasia (CAH)	**6**	**1/12,800**
Congenital adrenal hyperplasia non salt water	0	
Congenital adrenal hyperplasia salt water	6	
Congenital primary hypothyroidism (CH)	**38**	**1/2,000**
Cystic fibrosis (CF)	**17**	**1/4,500**
Fatty Acid Oxidation Disorders	**19**	**1/4,100***
Carnitine acylcarnitine translocase deficiency		
Carnitine uptake deficiency		
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		
Maternal carnitine uptake deficiency	1	
Medium-chain acyl-CoA dehydrogenase	8	

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
deficiency		
Medium-chain ketoacyl-CoA thiolase deficiency		
Medium/Short chain L-3 hydroxy acyl-CoA dehydrogenase deficiency		
Short-chain acyl-CoA dehydrogenase deficiency	4	
Trifunctional protein deficiency		
Very-long chain acyl-CoA dehydrogenase deficiency	5	
Galactosemia (GALT)	**14**	
Classical galactosemia	2	**1/39,000***
Duarte galactosemia	12	
Lysosomal Storage Disorders (LSD)	**31**	**1/4,500****
Fabry Disease	**22**	
Fabry		
Unknown onset		
Genotype of unknown significance		
Gaucher Disease	**0**	
Gaucher type 1 (non-neuropathic)		
Genotype of unknown significance		
Hurler Syndrome	**0**	
Hurler Syndrome - severe		
Krabbe Disease	**2**	
Genotype of unknown significance	2	
Krabbe unknown risk of onset		
Pompe Disease	**7**	
Classical Infantile Onset	1	
Non-classical infantile onset	1	
Later onset	5	
Unknown onset		
Genotype of unknown significance		
Organic Acid Disorders	**6**	**1/12,800****
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	

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Isovaleric acidemia	1	
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		
Forminioglutamic acid (FIGLU) not a disorder on the newborn screening panel but is found	1	
Hemoglobinopathies	**41**	**1/2,500****
Sickle cell anemia disease (Hb S/S)	18	**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)	5	
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	2	
Homozygous-E disorder (FE)	3	
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		

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

Appendix 2: Newborn Screening Laboratory Report
Samples Received 2014

	Newborn Samples Received			
	Initial	Repeat	Poor Quality	Total Infant Samples
Jan	6,252	1,260	163	7,675
Feb	5,755	1,106	141	7,002
Mar	5,949	1,144	135	7,228
Apr	6,402	1,224	143	7,769
May	6,136	1,170	116	7,422
Jun	6,103	1,157	93	7,353
Jul	6,769	1,317	94	8,180
Aug	6,471	1,168	65	7,704
Sep	7,072	1,188	87	8,347
Oct	6,429	1,168	105	7,702
Nov	5,363	1,001	140	6,504
Dec	6,897	1,359	160	8,416
Y.T.D.	**75,598 (82.80%)**	**14,262 (15.62%)**	**1,442 (1.58%)**	**91,302**

CALENDAR YEAR 2014
NEWBORN SCREENING LABORATORY REPORT
ABNORMAL RESULTS

Disorder		Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec	Y.T.D.
BIOT	Confirmed Profound	0	0	0	0	0	0	1	0	1	0	1	0	**3**
	Confirmed Partial	0	2	1	0	0	0	2	0	1	0	0	0	**6**
	High Risk	0	2	1	1	0	1	3	2	2	0	2	0	**14**
	Borderline Risk	0	2	3	3	6	5	7	4	2	2	1	2	**37**
CAH	Confirmed	0	0	0	1	1	0	1	1	0	0	0	2	**6**
	High Risk	5	2	1	6	4	2	3	3	3	0	1	3	**33**
	Borderline Risk	88	78	52	78	59	44	47	48	55	46	49	62	**706**
CF	Confirmed CF	3	0	0	4	3	1	0	1	1	1	2	1	**17**
	Confirmed CRMS	4	0	0	0	0	0	0	0	0	1	0	0	**5**
	Carriers Identified by NBS	16	8	10	8	12	10	17	11	14	14	6	14	**140**
	Referred	24	9	15	14	16	14	18	13	20	20	10	18	**191**
	Borderline IRT	70	84	73	62	61	61	50	60	77	72	51	74	**795**
CH	Confirmed	4	6	3	3	3	1	2	1	2	5	3	5	**38**
	High Risk	5	6	4	3	4	1	5	2	4	5	3	6	**48**
	Borderline Risk	100	86	165	122	96	77	89	38	49	69	126	159	**1176**
GALT	Confirmed Classical	0	0	0	0	0	0	1	0	0	0	0	1	**2**
	Confirmed DG	0	2	0	2	1	3	1	2	1	0	0	0	**12**
	High Risk	0	3	0	2	5	4	2	3	3	1	0	1	**24**
	Borderline Risk	4	1	3	2	5	3	7	1	6	0	2	0	**34**
AA	Confirmed	1	2	0	1	1	0	0	0	1	0	1	0	**7**
	Referred	3	5	3	3	3	0	0	2	1	0	3	0	**23**
	Low Risk	78	81	70	66	57	58	63	69	76	60	59	56	**793**
OA	Confirmed	3	0	0	0	1	0	0	1	0	0	1	0	**6**
	Referred	3	2	1	0	1	0	1	2	2	0	1	0	**13**
	Low Risk	47	42	35	45	39	39	40	40	25	43	33	37	**465**
FA	Confirmed	0	0	3	2	2	3	1	2	2	2	2	0	**19**
	Referred	5	6	8	9	8	4	3	2	7	7	5	3	**67**
	Low Risk	51	55	58	47	54	42	44	70	49	47	46	46	**609**
Hb	Sickle Cell Disease	4	1	3	2	5	1	3	1	4	5	0	5	**34**
	Other Hemoglobinopathies	0	0	0	1	1	1	1	0	1	0	1	1	**7**
	Traits Identified by NBS	136	119	125	102	112	113	130	140	140	127	120	161	**1525**
LSD	Confirmed Disorder	2	1	2	4	2	2	4	1	5	2	4	2	**31**
	Confirmed Carrier	2	2	0	2	1	2	2	2	4	1	4	3	**29**
	Confirmed Pseudo Def.	3	0	3	1	2	1	3	2	2	2	1	1	**21**
	High Risk	8	6	10	8	10	14	14	13	14	15	10	12	**134**
	Borderline Risk	0	0	0	0	0	0	0	0	0	0	0	0	**0**
SCID	Confirmed	0	0	0	0	0	0	0	0	0	0	0	0	**0**
	Referred	0	0	0	0	0	0	0	0	0	0	0	0	**0**
	Low Risk	0	0	0	0	0	0	0	0	0	0	0	0	**0**

Total Confirmed **188**

BIOT = biotinidase deficiency
CF = cystic fibrosis
GALT = galactosemia
OA = organic acid
Hb = Hemoglobinopathies
CAH = congenital adrenal hyperplasia
CH = congenital hypothyroidism
AA = amino acid
FA = fatty acid
LSD = lysosomal storage disorder
SCID = Severe Combined Immunodeficiency

Appendix 4: Outcome Data – Newborn Screening Samples and Results

- In 2014 there were 75,598 babies tested in the state newborn screening laboratory. There were 91,302 blood spot samples received in the laboratory. Samples received included:

Initial	Repeat	Poor Quality
75,598	14,262	1,442

- In the process of screening newborns for 70 genetic and metabolic conditions, it is the newborn screening laboratory's role to assess the risk of any abnormal screening by evaluating the marker analytes and the levels that were detected. This risk assessment then dictates different levels of action and follow-up protocols. The 91,302 newborn screening samples received at the state newborn screening laboratory can be separated into two risk categories. The number/percentage of test results falling into these categories during 2014 were:

High Risk / Referred	Low Risk / Borderline Risk
588 (0.64%)	4,615 (5.0%)

High Risk / Referred – 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 specimen and the physician of record.

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

- One hundred and eighty-eight (188) confirmed disorders were diagnosed from these abnormal newborn screen results during 2014.

Appendix 5: Poor Quality Samples 2014

QUANTITY NOT SUFFICIENT: Quantity of blood on filter paper not sufficient for testing. Possible causes: removing filter paper before blood has completely filled circle; not allowing an ample size blood drop to form before applying to filter paper; inadequate heel stick procedure.	206
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 sample collection.	533
SAMPLE ABRADED: Filter paper 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.	38
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.	478
DILUTED, DISCOLORED OR CONTAMINATED: Possible causes: squeezing or milking of area surrounding the puncture site; allowing filter paper to come into contact with gloved or ungloved hands, or substances such as alcohol, formula, antiseptic solutions, water, hand lotion, powder, etc., either before or after blood sample collection; exposing blood spots to direct heat; allowing blood spots to come into contact with tabletop, etc. while drying the sample.	102
OLD SAMPLE: Sample greater than 15 days old when received at State Public Health Laboratory.	48
LABORATORY ACCIDENT: Unable to test; sample damaged at laboratory.	1
OTHER:	1
NO BLOOD: Filter paper submitted without blood.	1
OLD FORM: Sample received on out-of-date form.	1

FILTER PAPER AND FORM BARCODES DO NOT MATCH: Bar code on filter paper does not match bar code on Newborn Screening Form. Collection forms contain barcodes on demographic, hearing and filter paper portions. The barcodes may not be altered in any way. If incorrect baby is sampled do not remove filter paper 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 paper.	3
MISSING, INCOMPLETE OR CONFLICTING PATIENT INFORMATION: Missing, incomplete or conflicting demographic information.	7
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.	20
BLOOD ON OVERLAY COVER: Overlay cover came in contact with wet blood sample. Possible causes: sample is poor quality status because blood soaked from back of filter paper onto the gold colored backing of the form. The filter paper circles are designed to hold a specific quantity of blood. If the wet filter paper is allowed to come into contact with the paper backing of form, blood can be drawn out of filter making the quantative tests performed by the Newborn Screening Laboratory invalid. It is very important that the wet filter paper does not come into contact with any surface until completely dry.	3
Total Poor Quality Samples Received	1442 (1.58%)

Appendix 6: Newborn Bloodspot Screening Hemoglobinopathy Report 2014

Specimens Received:

Initial:	75,598	(82.7%)
Repeat:	14,262	(15.6%)
Unsatisfactory:	1,442	(1.6%)
Whole Blood:	124	(0.1%)
Total:	**91,426**	

Significant Results = 1,566					
Sickle Cell Disease		**Other Disease Conditions**		**Trait Conditions**	
FS	18	FCA	2	FAS	993
FSA	5	FE	3	FSAINC	9
FSC	11			FAC	296
FC	2			FCAINC	10
				FAE	38
				FAD	42
				FAX	125
				FASX	3
				FACX	1
				Slightly Elevated Barts	8
				Other Trait condition	0
Total	**36**	**Total**	**5**	**Total**	**1,525**

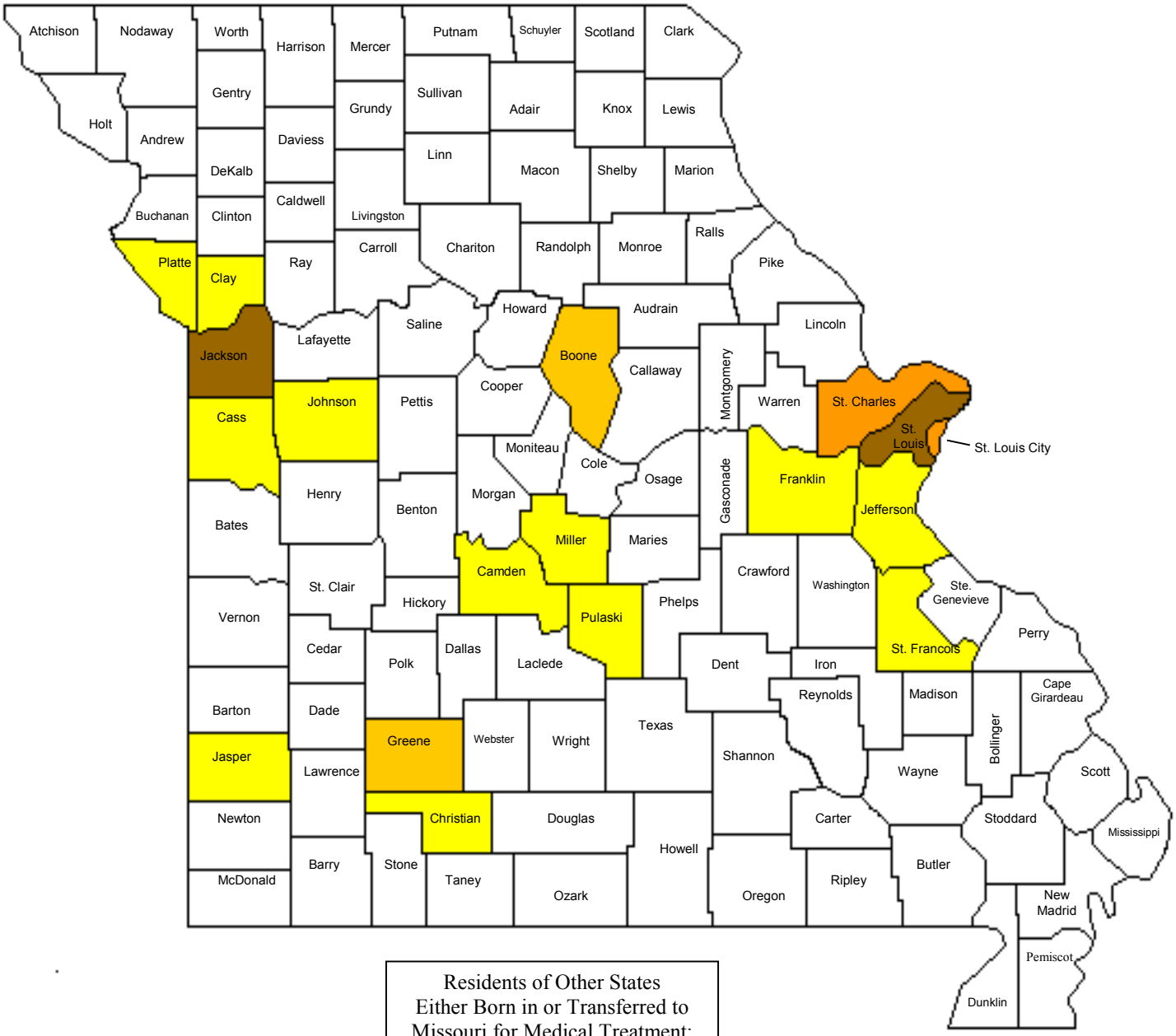
Appendix 7: Missouri Newborn Hearing Screening Data for 2014

2014 calendar year provisional data for Missouri shows:

- 76,725 occurrent births (source: Department of Health and Senior Services Vital Records)
- 76,525 occurrent births (source: Missouri Health Strategic Architectures and Information Cooperative [MOHSAIC]*)
- 97.9 percent (74,982) of newborns were screened
- 97.3 percent (73,000) of infants were screened by 1 month of age
- 1.67 percent (1,259) of infants failed the final screening
- 47.6 percent (600) of the infants who failed their final screening and received an audiologic evaluation were evaluated and diagnosed by 3 months of age
- 104 infants were diagnosed with a permanent hearing loss
- 70 infants were enrolled in Missouri's Part C of the Individual with Disabilities Education Act (IDEA) program, First Steps
- 67.3 percent (70) of the infants enrolled in First Steps did so by 6 months of age

*The difference of 200 births between the occurrent birth count in the program data management system, the Missouri Health Strategic Architectures Information Collaborative (MOHSAIC), and the total occurrent births reported by Vital Records is the result of records that do not yet have an assigned Department Client Number (DCN) and records that are sealed. Records are not released from the Vital Records system to MOHSAIC until the DCN assignment is complete. Non-complete records are due to issues such as paternity and adoptions. Sealed birth records are neither displayed nor counted in MOHSAIC. This report is based upon MOHSAIC records.

Appendix 8: Number of Newborns with Abnormal Newborn Blood Spot Screens Referred for Follow-up by County in 2014

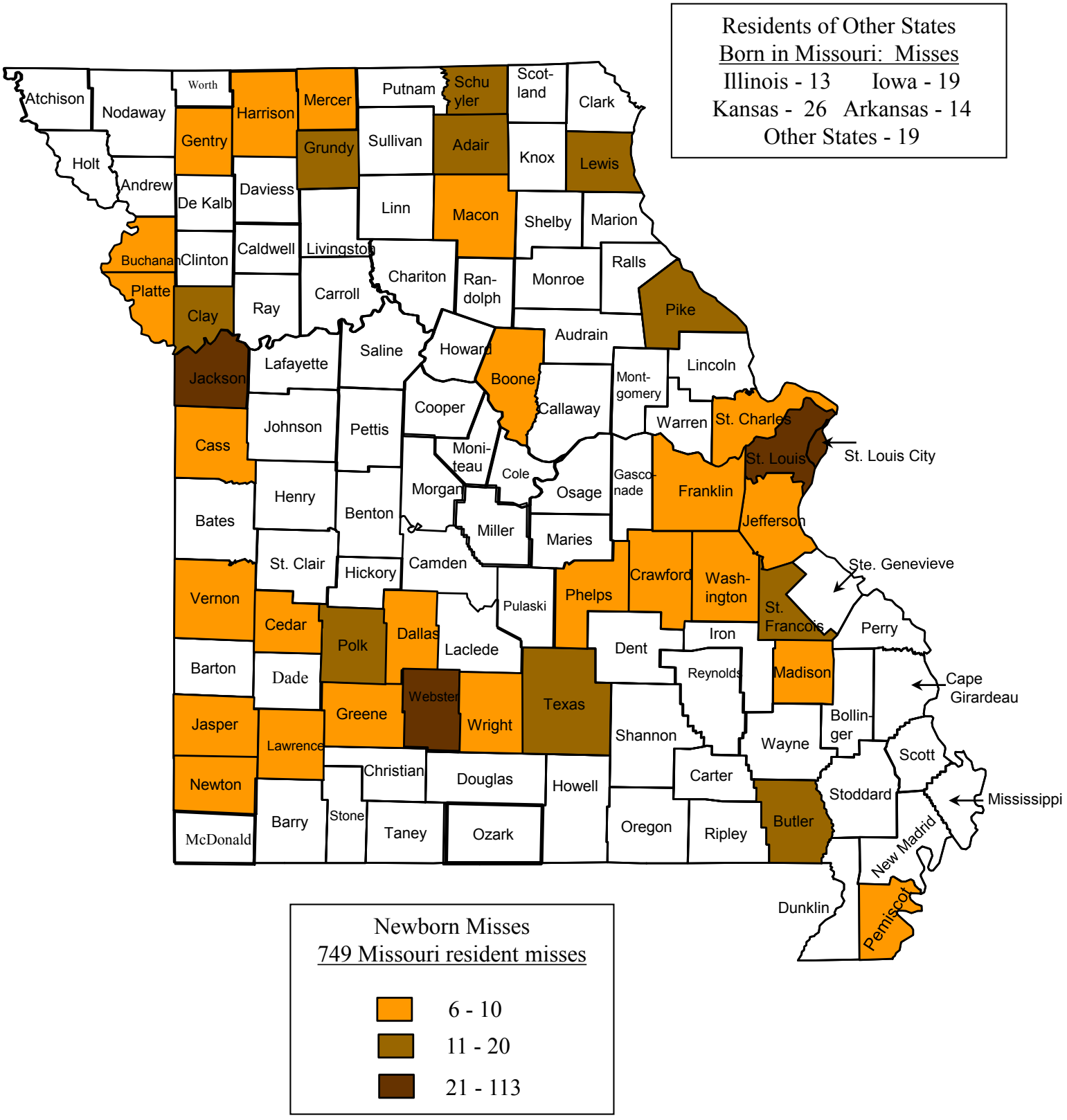


Residents of Other States Either Born in or Transferred to Missouri for Medical Treatment:

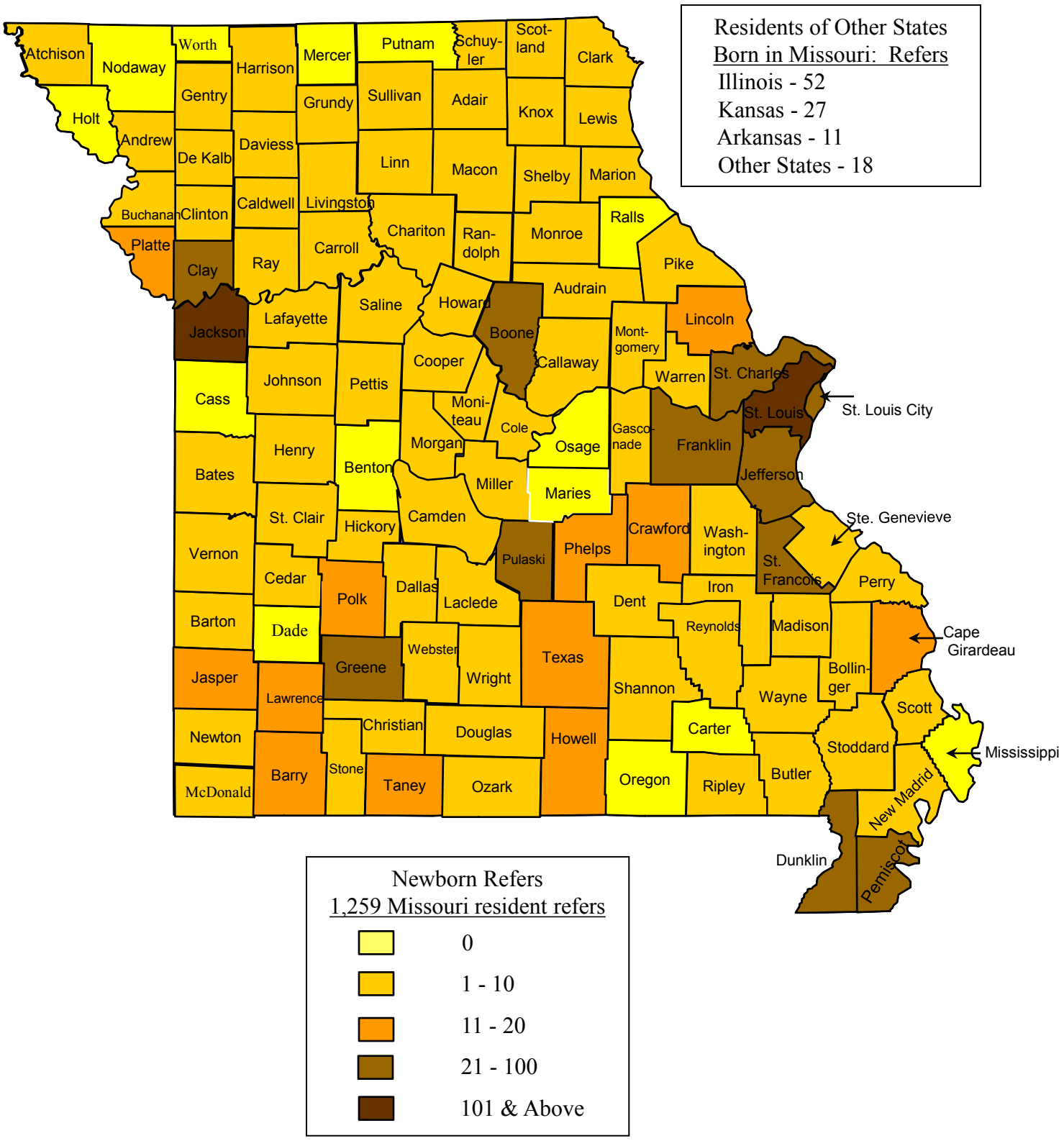
Kansas - 22 Iowa - 1
Illinois - 20 Arkansas - 1
Other States - 4

6 - 15
16 - 25
26 - 45
46 - 100

Appendix 9: Number of Newborns that Missed a Hearing Screening by County during 2014



Appendix 10: Number of Newborns Referred after a Hearing Screen by County during 2014



Appendix 11: Newborn Screening Parent Satisfaction Surveys

A satisfaction survey of parents was conducted for families of babies having abnormal newborn screening results reported in 2014. There were 120 satisfaction surveys mailed and 12 were returned for a survey return rate of 10%. Key findings:

Newborn Screening Parent Satisfaction Survey			
	Very Satisfied	Satisfied	Not Satisfied
Staff explained my baby's condition in a way I could understand	83%	17%	
Able to ask questions and discuss decisions about my baby's health care	100%		
Offered reassurance and support	92%	8%	
The treatment staff was knowledgeable	92%	8%	
My questions and concerns were addressed in a timely manner	83%	17%	
The staff provided me with useful referrals and resources	92%	8%	
Received high quality care during my appointments	83%	17%	

A satisfaction survey of parents and children receiving services provided by the hemoglobinopathy resource centers was completed in 2013. *This survey is completed every two years. A survey will be mailed in 2015.* There were 1065 surveys mailed and 340 were returned for a survey return rate of 32%. Key findings:

Hemoglobinopathy Resource Center Satisfaction Survey – Parent Response			
	Very Satisfied	Satisfied	Not Satisfied
Treated with respect	97%	1%	2%
Treatment staff was knowledgeable	88%	12%	0%
Questions/concerns addressed in a timely manner	86%	13%	1%
Staff provided useful referrals and resources	83%	15%	2%
Provided with the services needed	97%	2%	1%
Medical care/services received	76%	23%	1%
Received services or treatment without experiencing any problems	97%	0%	3%

Reasons parents responded as not satisfied with services were because of a long wait time. Parents did not indicate what a long wait time meant to them.

Appendix 12: Newborn Hearing Screening Parent Satisfaction Survey

In March 2014* a 2013 satisfaction survey was mailed to parents of children born in Missouri who failed their initial newborn hearing screening between October 2013 and December 2013. There were 578 surveys mailed and 123 were returned for a survey return rate of 21%. 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:

- 78% of the respondents reported that the birth hospital provided them with written information about the hearing screening prior to the hearing screening.
- 98% of the respondents reported that the birth hospital notified them of the screening result.
- 74% of the respondents reported that the hospital staff explained the importance of knowing whether a baby has a hearing loss early in life.

*Survey conducted every two years.

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