MISSOURI NEWBORN SCREENING

2017 Annual Report

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Missouri Department of Health and Senior Services

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.

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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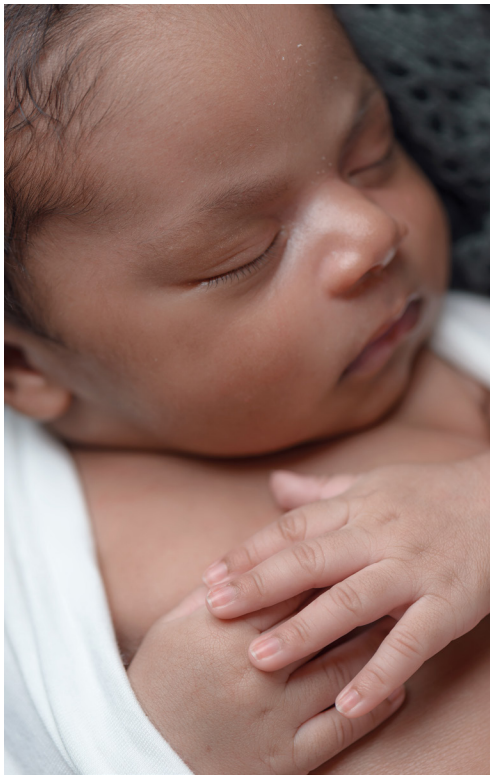
Screening Spotlight: Newborn Screening for Severe Combined Immunodeficiency

Severe combined immunodeficiency (SCID), sometimes in the past referred to as "Bubble Boy Disease," is a genetic condition characterized by the body's inability to fight off infection. The body's immune system is made up of different parts that work together to keep the body from getting sick. In SCID, certain parts of the immune system do not work properly, specifically the T-cells and B-cells. T-cells and B-cells are very important lymphocytes, or white blood cells, which help our bodies fight off infection.

It is estimated that SCID occurs in one out of every 40,000 – 100,000 live births. Babies affected by SCID may appear healthy and normal at birth, and for the first few months of life, until the immunity given to them by their mother begins to disappear. Children with a normal immune system can fight off everyday germs, but children with SCID will not be able to recover from something as simple as the common cold.

Symptoms of SCID include, but are not limited to: recurrent bouts of diarrhea, pneumonia, ear infections, thrush, and bronchitis that do not respond to usual therapies. Without early detection and treatment, children affected by SCID will die of infections before their first or second birthday. Treatment with bone marrow transplant within the first few months of life, before an infection has occurred, has a 96% likelihood of curing SCID.

In 2010, after Wisconsin and several other states had success in screening newborns for SCID, the disorder was added to the Recommended Uniform Screening Panel (RUSP). The RUSP is a list of conditions, which includes 34 core conditions and 26 secondary conditions, for which the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children recommends every baby be screened. In 2016, Missouri state legislators passed legislation, RSMo 191.332, requiring all babies born in Missouri to be screened for SCID beginning January 1, 2017.



After the passage of legislation, a SCID Task Force was formed. The Task Force consisted of several physicians specializing in immunology, hematology, and bone marrow transplants; laboratory scientists from the Missouri State Public Health Laboratory (MSPHL); staff from Missouri's Newborn Screening Follow-up Program; and SCID parent advocates. The Task Force served to provide assistance and guidance throughout the implementation process of newborn screening for SCID.

After laboratory staff were trained on use of the equipment, validation of the testing methodology was completed and follow-up protocols were created. Full population pilot testing for SCID began on the first working day of the year, January 3, 2017. The full population pilot phase continued until enough data had been collected to confirm laboratory cut-off values were accurate, thus allowing for the identification of SCID. Live reporting of results on newborn screening reports began October 9, 2017.

In Missouri's first year of screening, one child was confirmed positive for SCID and successfully treated through stem cell transplant. There were also many other children with other syndromes and lymphopenias who were identified and treated as a result of newborn screening for SCID. With collaboration between the SCID Task Force members, MSPHL staff, and Newborn Screening Follow-up staff, newborn screening for SCID continues to be successful.

For more information about Missouri's Newborn Screening Program, please visit www.health.mo.gov/newbornscreening.

Blood Spot Disorders Confirmed for 2017

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE
Amino Acid Disorders	***9***
Argininosuccinic acidemia	1
Citrullinemia type I	1
Hyperphenylalaninemia, benign	1
Phenylketonuria (PKU)	6
Biotinidase Deficiency (BIOT)	***18***
Partial biotinidase deficiency	13
Profound biotinidase deficiency	5
Congenital Adrenal Hyperplasia (CAH)	***3***
Congenital adrenal hyperplasia non salt waster	2
Congenital adrenal hyperplasia salt waster	1
Congenital Primary Hypothyroidism (CH)	***34***
Cystic Fibrosis (CF)	***23***
Cystic fibrosis	19
Cystic fibrosis transmembrane conductance (CRTR) - related metabolic syndrome (CRMS)	4
Fatty Acid Oxidation Disorders	***18***
Carnitine uptake deficiency	2
Maternal carnitine uptake deficiency	1
Medium-chain acyl-CoA dehydrogenase deficiency	4
Short-chain acyl-CoA dehydrogenase deficiency	7
Very-long chain acyl-CoA dehydrogenase deficiency	4
Galactosemia (GALT)	***9***
Classical galactosemia	2
Duarte galactosemia	7
Hemoglobinopathies	***38***
Disease conditions	
Sickle cell anemia disease	24
Sickle hemoglobin-C disease	5
Sickle beta zero thalassemia disease	1
Sickle beta plus thalassemia disease	5
Hemoglobin-C beta zero thalassemia disease	1
Homozygous-E disease	1
Heterozygosity sickle cell and unidentified variant	1

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE
Lysosomal Storage Disorders (LSD)	***33***
Fabry Disease	***23***
Fabry	6
Fabry A143T	11
Genotype of unknown significance	6
Gaucher Disease	***2***
Gaucher type 1 (non-neuropathic)	1
Genotype of unknown significance	1
Hurler Syndrome	***4***
Genotype of unknown significance	4
Pompe Disease	***4***
Later onset	4
Organic Acid Disorders	***11***
3-Methylcrotonyl-CoA carboxylase deficiency	1
Glutaric acidemia, type I	1
Malonic acidemia	1
Propionic acidemia	4
Forminioglutamic acid (FIGLU) not a disorder on the newborn screening panel but is found	3
Maternal 3-Methylcrotonyl-CoA carboxylase deficiency	1
Severe Combined Immunodeficiency (SCID)	***12***
X-linked	1
Secondary conditions detected through SCID newborn screening	11

CALENDAR YEAR 2017
NEWBORN SCREENING LABORATORY REPORT
ABNORMAL RESULTS

Disorder		Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec	Y.T.D.
BIO	Confirmed Profound	0	0	1	0	0	1	2	0	0	0	1	0	**5**
	Confirmed Partial	1	0	2	1	0	0	2	4	2	0	0	1	**13**
	High Risk	3	0	4	3	0	2	5	5	2	1	1	1	**27**
	Borderline Risk	5	4	3	7	7	8	16	9	7	3	2	5	**76**
CAH	Confirmed	0	0	0	2	0	0	0	0	1	0	0	0	**3**
	High Risk	5	3	8	4	0	4	3	2	3	1	1	0	**34**
	Borderline Risk	35	24	38	23	33	33	38	41	48	37	41	46	**437**
CF	Confirmed CF	3	2	3	1	1	0	1	4	0	0	2	2	**19**
	Confirmed CRMS	0	2	0	0	0	0	1	0	0	1	0	0	**4**
	Carriers Identified by NBS	2	10	11	9	9	11	10	12	9	8	11	7	**109**
	Referred	6	14	15	11	11	13	13	20	9	10	14	9	**145**
	Initial IRT	50	52	63	56	71	42	51	68	68	61	49	59	**690**
CH	Confirmed	1	2	2	4	3	2	6	0	4	6	2	2	**34**
	High Risk	1	2	2	5	3	3	6	4	5	7	5	2	**45**
	Borderline Risk	133	103	71	58	63	52	71	87	88	89	84	81	**980**
GALT	Confirmed Classical	0	1	0	0	1	0	0	0	0	0	0	0	**2**
	Confirmed DG	1	0	2	1	0	0	1	0	1	1	0	0	**7**
	High Risk	2	2	3	2	1	2	4	1	1	2	1	1	**22**
	Borderline Risk	3	2	2	4	1	0	1	2	2	1	2	1	**21**
AA	Confirmed	0	2	0	0	2	1	0	1	2	0	0	1	**9**
	Referred	2	4	1	0	2	1	0	3	3	1	2	3	**22**
	Low Risk	68	65	78	58	69	61	70	65	53	48	47	61	**743**
OA	Confirmed	3	0	0	0	1	1	2	2	1	0	0	1	**11**
	Referred	6	0	2	2	4	1	4	4	2	2	3	5	**35**
	Low Risk	63	63	72	66	73	62	67	50	39	48	53	52	**708**
FA	Confirmed	3	0	1	2	1	3	1	0	4	0	1	2	**18**
	Referred	6	4	3	3	3	3	3	1	6	7	7	3	**49**
	Low Risk	50	52	58	51	66	51	74	81	50	56	49	42	**680**
Hb	Sickle Cell Disease	3	4	4	2	4	4	3	3	1	4	3	1	**36**
	Other Hemoglobinopathies	0	0	0	0	0	1	0	0	0	0	0	1	**2**
	Traits Identified by NBS	134	115	121	133	147	122	139	137	142	123	112	128	**1553**
LSD	Confirmed Disorder	5	2	3	2	3	3	1	1	5	4	2	2	**33**
	Confirmed Carrier	1	3	5	3	7	2	3	2	8	4	2	2	**42**
	Confirmed Pseudo Def.	2	5	10	4	4	1	3	1	6	2	6	2	**46**
	High Risk	20	17	23	17	25	16	18	18	27	18	18	16	**233**
	Borderline Risk	83	66	110	114	142	114	122	115	111	68	43	63	**1151**
SCID	Confirmed	1	1	0	0	0	0	2	3	1	3	1	0	**12**
	Referred	0	0	0	0	0	0	0	0	0	6	3	0	**9**
	Low Risk	0	0	0	0	0	0	0	0	0	13	11	8	**32**

BIO = biotinidase deficiency
CF = cystic fibrosis
GAL = 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

Total Confirmed **208**

Outcome Data - Newborn Screening Samples and Results

- In 2017 there were 90,489 samples tested in the state newborn screening laboratory. Samples received included:

Initial	Repeat	Poor Quality
72,440	16,082	1,967

- In the process of screening newborns for over 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 90,489 newborn screening samples received at the state newborn screening laboratory can be separated into the following categories: normal, low risk, borderline risk, high risk, and no results. The number/percentage of test results falling into the high risk, borderline risk, and low risk categories during 2017 were:

High Risk / Referred	Borderline Risk / Low Risk
659 (0.73%)	*5,518 (6.1%)*

High Risk / Referred - Results are immediately phoned and faxed to the physician of record and to the contracted genetic, cystic fibrosis, or hemoglobinopathy referral centers for consultation and confirmatory testing. Final laboratory reports are mailed to the physician of record.

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

- **208** confirmed disorders were diagnosed from these abnormal newborn screen results during 2017.

Poor Quality Samples for 2017

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.	303
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.	583
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.	37
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; or clotted swirl marks from improper capillary application.	854
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.	110
OLD SAMPLE: Sample greater than 15 days old when received at State Public Health Laboratory.	4
DAMAGED SPECIMEN: Specimen damaged in transit.	0
NO BLOOD: Filter paper submitted without blood.	0
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.	2

MISSING, INCOMPLETE OR CONFLICTING PATIENT INFORMATION: Missing, incomplete or conflicting demographic information.	12
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.	34
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 quantitative 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.	10
OLD FORM: Sample received on out-of-date form.	17
WET SPECIMEN: Specimen submitted before drying thoroughly. Allow blood spots to thoroughly dry for at least 3 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 card.	1
TOTAL POOR QUALITY SAMPLES RECEIVED	1,967 (2.17%)

Newborn Hearing Screening Data for 2017

2017 calendar year provisional data for Missouri shows:

- *73,868 occurrent births (source: Department of Health and Senior Services Vital Records)*
- *73,839 occurrent births (source: Missouri Health Strategic Architectures and Information Cooperative [MOHSAIC]*)*
- *98.1 percent (72,452) of newborns were screened*
- *98 percent (71,000) of infants were screened by 1 month of age*
- *1.5 percent (1,100) of infants failed the final screening*
- *93.9 percent (692) of the infants who failed their final screening and received an audiologic evaluation were evaluated and diagnosed by 3 months of age*
- *113 infants were diagnosed with a permanent hearing loss*

*The difference of 29 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 Department of Health and Senior Services Bureau of 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 BVR system to MOHSAIC until the DCN assignment is complete. Incomplete and sealed 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.

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