

Mountain States Genetics Regional Collaborative Center
 Newborn Screening Committee
 Mid-year Meeting
 April 24, 2007

State: Arizona

Newborn Screening Laboratory Contact: Joan Apt

Newborn Screening Follow-up Program Contact: Jan Kerrigan

Birthrate 2006: 101,888

Number of disorders Screened for: During 2006, the Arizona NBS panel increased from 8 disorders to 27.

Please list disorders screened for and 2006 presumed positive and confirmed cases:

We are still compiling presumed positive and should have that later this week. Listed here are disorders and confirmed cases.

Phenylketonuria (Classic)	4
Primary Congenital Hypothyroidism	45
Galactosemia (Classic)	0
Biotinidase Deficiency	3
Homocystinuria	0
Congenital Adrenal Hyperplasia (Salt Wasting)	9
Congenital Adrenal Hyperplasia (Simple Virilizing)	2
Congenital Adrenal Hyperplasia (21 OH Deficient)	1
Maple Syrup Urine Disease (MSUD)	0
Sickle Cell Disease (FS)	8
Hemoglobin SC (FSC)	5
Sickle Cell - Beta Thalassemia (FSA)	1
Other Hemoglobin Disease	3
Citrullinemia	
Tyrosinemia type I	
Argininosuccinic acidemia	
Medium chain acyl-CoA dehydrogenase deficiency	
Very long-chain acyl-CoA dehydrogenase deficiency	
Long-chain L-3-OH acyl-CoA dehydrogenase deficiency	
Trifunctional protein deficiency	
Carnitine uptake defect	
Isovaleric acidemia	
Glutaric acidemia type I	
3-OH 3-CH3 glutaric aciduria	
Multiple carboxylase deficiency	
Methylmalonic acidemia	
Methylmalonic acidemia (mutase deficiency)	
3-Methylcrotonyl-CoA carboxylase deficiency	
Propionic acidemia	

Does your state have a NBS Committee? Yes

If so provided contact information for chair and staff contact: No chair. Staff contact is Jan Kerrigan, Newborn Screening Program Manager.

How long does you state store residual specimens, where and under what conditions? 30 days unless retained for quality assurance reasons.

If you state has a written guideline (policy, rule, etc.) for the storage, retention and use of residual specimens please attach. I am attaching a policy that is not yet approved (see below). Draft is expected to be approved this month.

ARIZONA DEPARTMENT OF HEALTH SERVICES
POLICY AND PROCEDURE

SUBJECT **Newborn Screening Specimen Storage, Retention, and Access**

PURPOSE To provide guidance regarding retention, storage, access, and release of newborn screening dried blood spot specimen and related specimen or patient information.

AUTHORITY

APPLICABILITY

Arizona Department of Health Services, Newborn Screening Program and contracted Newborn Screening Laboratory.

DEFINITIONS

Abnormal means a result of an analysis performed as part of a newborn screening test that deviates from the range of values established by the Department.

Disorder means a disease or medical condition that may be identified by a laboratory analysis.

Document means to establish and maintain information in written, photographic, electronic, or other form.

Electronic means relating to technology that has electrical, digital, magnetic, wireless, optical, or electromagnetic capabilities or similar capabilities.

First specimen means the initial specimen that is collected from a newborn who is less than five days of age and sent to the screening laboratory for testing and recording of demographic information.

Guardian means an individual appointed by a court under A.R.S. Title 14, Chapter 5, Article 2.

Health care facility means a health care institution defined in A.R.S. § 36-401 where obstetrical care or newborn care is provided.

Health care provider means a physician, physician assistant, or registered nurse practitioner, or midwife.

Identification code means an account number assigned by the newborn screening laboratory.

Midwife means an individual licensed under A.R.S. Title 36, Chapter 6, Article 7 or certified under A.R.S. Title 32, Chapter 15.

Newborn means a human from birth through 28 days of age for whom a certificate of live birth is required to be filed under A.R.S. § 36-322.

Newborn screening laboratory means an entity contracted with the Department under A.R.S. § 36-694(C) to perform the newborn screening test.

Newborn screening test means multiple laboratory analyses performed on a first specimen and a second specimen to detect the presence of endocrine disorders, metabolic disorders, or hemoglobinopathies listed in A.A.C. R9-13-202.

Parent means a natural, adoptive, or custodial mother or father of a newborn.

Person means the state, a municipality, district, or other political subdivision, a cooperative, institution, corporation, company, firm, partnership, individual, or other legal entity.

Physician means an individual licensed under A.R.S. Title 32, Chapters 13, 14, 17, or 29.

Physician assistant means an individual licensed under A.R.S. Title 32, Chapter 25.

Satisfactory specimen means a specimen collection kit, on which demographic information has been written and blood applied to the filter paper of that specimen collection kit, that meets the newborn screening test requirements.

Second specimen means a specimen that is sent to the screening laboratory for testing and recording of demographic information, after being collected: from a newborn after a first specimen; or from an individual at least five days and not older than one year of age, regardless of whether a first specimen was collected.

Specimen means capillary or venous blood, but not cord blood, applied to the filter paper of the specimen collection kit.

Specimen collection kit means a strip of filter paper for collecting a blood sample attached to a form for obtaining the information specified in A.A.C. R9-13-203(A)(3) about a newborn or infant.

Test means a laboratory analysis performed on body fluid, tissue, or excretion to determine the presence or absence of a disorder.

Unsatisfactory specimen means a specimen collection kit, on which demographic information has been written and blood applied to the filter paper of that specimen collection kit that is rejected by the newborn screening laboratory for any of the reasons specified in R9-13_____.

Verify means to obtain information through sources that include the newborn screening program, a health care provider, a health care facility, or a documented record.

POLICY:

- 1) Newborn Screening bloodspot specimens and attached information submitted to the Arizona Department of Health Services (AHDS) are the property of the ADHS.
- 2) Access to stored specimen/information forms shall be restricted to ADHS employees and those contractors or others approved by the Newborn Screening Program Manager as necessary to meet specific program needs. Access is contingent upon compliance with all applicable state laws, regulations, and policies safeguarding the privacy and confidentiality of medical information. The ADHS Director or designee shall assure that those granted access understand confidentiality requirements and

have a signed confidentiality agreement on file.

3) Specimen Retention and Storage

- a) All specimens received by the Newborn Screening Laboratory will be kept for approximately 3 months.
- b) Samples will be stored at room temperature in a plastic container. Containers will be numbered with the identifying Julian date.
- c) After 3 months, the filter paper portion of the blood spot card is separated from the demographic section of the sample. One copy of the demographic information portion of the card is archived in a medical records long-term storage facility for 22 years and then discarded. Other copies of the demographic information portion of the cards are shredded and discarded. The blood spots are autoclaved at a temperature of 121° for 60 minutes and are then discarded.
- d) Any specimens of interest or with abnormal results may be saved at the discretion of the lab, and stored at <-20°C for an indefinite amount of time.

4) Use of Specimens by the Laboratory

- a) Residual dried blood specimens remaining after the newborn screening tests have been performed may be used by the lab for methods development, comparison and validation studies, and reanalysis.

5) Release of Specimens

Dried blood spot specimen and specimen information may only be released by the ADHS when required by state or federal law or under the following conditions:

- a) A sample from a specimen and copies of associated information (patient information and testing results, if requested) may be released to:
 - i) A health care provider at the request of the parent or the newborn's legal representative after completing and signing a written request form approved by the department. The release form must be provided to the director of newborn screening before the request will be fulfilled. Newborn Screening bloodspot specimens will not be returned directly back to the newborn's parent or guardian.
 - ii) A researcher with the written, informed consent of the patient or their patient's legal representative as part of a research project that has been reviewed and approved by the ADHS human subjects review board and the Director or designee of the ADHS.
 - iii) A named person in a legally executed subpoena following review and approval of the state attorney general.
 - iv) A person to whom release is mandated by order of a court of competent jurisdiction.
 - v) The Office of the Medical Examiner (ME) with a written request from the ME and approval from the ADHS Director or designee. A portion of the sample will be cut from the card and the patients name will be written on the sample

- 6) For research using DNA derived from dried-blood spots:
- a) There must be de-identification, which can most easily be accomplished by simply snipping off a piece of the specimen and providing no other information; or
 - b) There must be parental or legal guardian written authorization on a Privacy Rule complaint form; or
 - c) There must be a waiver of the need for authorization properly granted by a Privacy Board or IRB; or
 - d) There must be a Limited Data Set containing only general geographic information and relevant dates, coupled with a data use agreement signed by the researcher
- 7) The ADHS shall notify parents of the specimen storage, retention/destruction and access requirements through the department's newborn screening informational pamphlet.

4/18/06

Still need:

CLIA regulations – need property established in contract (Mary Beth Joublanc)

Lab definition of room temperature (Joan Apt)

Letter to Dona Markley (Jan Kerrigan)

Change language in 3A. Currently too vague, not consistent with rest of policy. (Mary Beth Joublanc)

Add language to “return specimens to ADHS or destroy” (Mary Beth Joublanc)

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State: Colorado

Newborn Screening Laboratory Contact:

Dan Wright, Newborn Screening Unit Supervisor
303-692-3673 (NBS lab main number 303-692-3670).
daniel.wright@state.co.us

Newborn Screening Follow-up Program Contact:

Laura Taylor
Follow-up Coordinator, Newborn Metabolic Screening Program
303-692-2425; FAX: 303-753-9249
laura.taylor@state.co.us

Birthrate 2005: Occurrent births: 69,210

Number of disorders Screened for: In 2005 we screened for seven conditions: Biotinidase Deficiency, Congenital Adrenal Hyperplasia, Congenital Hypothyroidism, Cystic Fibrosis, Galactosemia, Hemoglobinopathies, and PKU. (On July 1, 2006 we began expanded screening for the complete ACMG/March of Dimes panel of recommended diseases.)

Disorders screened for and 2006 presumed positive and confirmed cases:

Below, the first number is the number of presumptive positive results -- the second is the number of confirmed cases (these counts do not include borderlines, but this is all the information I currently have available.)

Biotinidase Deficiency	4/4
CAH	23/6
Hypothyroidism	83/40
Cystic Fibrosis	64/18
Galactosemia	4/2
Hemoglobinopathies	8/6 (as noted above, this does not include traits)
PKU	38/1

Does your state have a NBS Committee? Yes

We do have a newborn screening program advisory committee that has met quarterly since 1979. The current Chair is Dr. Philip Zeitler, a pediatric endocrinologist with The Children's Hospital, Denver. Dr. Zeitler can be reached at 303-861-6128, or by email at phil.zeitler@uchsc.edu. Laura Taylor is staff person (see contact info above).

How long does you state store residual specimens, where and under what conditions?

The laboratory stores specimens for six months at room temperature. All blotters will be retained, at room temperature for 6 months at which time they will be sealed in a biohazard bag with autoclave reactive tape that, after autoclaving, will clearly indicate that the contents have been sterilized. This sterilization will occur at 250°C at 23 lbs. pressure for 1 hour (1 cycle). The blotters will then be discarded in the general garbage.

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State: MONTANA

Newborn Screening Laboratory Contact: Denise Higgins dehiggins@mt.gov

Newborn Screening Follow-up Program Contact: Sib Clack sclack@mt.gov

Birthrate 2006: preliminary number = 12,000+ (won't be finalized until mid-May 2007)

Number of disorders Screened for:

4 mandatory, 24 others optional

We have a bill before the current Legislative Session to bring MT up to the mandatory 28 plus hearing screening. Session ends Mid-April.

Please list disorders screened for and 2006 presumed positive and confirmed cases:

PKU - 1 hyperphe

GALT - 1

CH - 1

HbB - no Sickle Cell Disease

Optional test data are incomplete at this time.

Does your state have a NBS Committee?

Ad Hoc committee, not a formal standing committee.

How long does you state store residual specimens, where and under what conditions?

Six weeks to three months depending on storage space in the Laboratory freezer which is maintained at 2-8 degrees Celsius.

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State: Nevada

Newborn Screening Laboratory Contact: Brad Towle or Jo Malay, RN, MPH.
Otherwise Oregon Health Sciences Laboratory, Leanne C Rien
[Leanne.C.Rien@state.or.us] is the one we are usually in contact with.

Newborn Screening Follow-up Program Contact: Brad Towle overseer

Birthrate 2006: not available yet. 2005 was 37,083

Number of disorders Screened for: 31

Please list disorders screened for and 2006 presumed positive and confirmed cases:

See below. Confirmed cases for 2005 included 15 presumptive PKU, none confirmed, 400 presumptive congenital hypothyroidism, 19 confirmed, 78 galactosemia, none confirmed, 18 sickle cell disease with 13 confirmed, 22 Biotinidase Deficiency with 2 confirmed, 19 hemoglobinopathies with 14 confirmed, 302 CAH with 3 confirmed, 1 MCAD presumptive and confirmed, 1 Hyperphenylalaninemia presumptive and confirmed, and 7 other presumptive and confirmed.

Does your state have a NBS Committee? The Maternal and Child Health Advisory Board serves as an advisory committee. Changing of fees for NBS goes to the Board of Health.

If so provided contact information for chair and staff contact: Current Chair is
Terrance McGaw, MD.
- Chairman
75 Pringle Way Ste 801
Reno, NV 89502
775-688-5850
775-688-5809 fax

How long does you state store residual specimens, where and under what conditions? This is handled by our contractor, Oregon Health Sciences Laboratory.

Newborn Screening Disorders – as of 9/12/03

Endocrine

Congenital Adrenal Hyperplasia
Congenital hypothyroidism

Hemoglobin

Sickle cell disease and other hemoglobinopathies

Metabolic Disorders

Biotinidase deficiency

Galactosemia

Amino Acid Disorders

- Arginase Deficiency
- Argininosuccinate lyase deficiency (ASA)
- Citrullinemia
- Homocystinuria
- Hyperphenylalanemia, including phenylketonuria
- Tyrosinemia

Organic Acid Disorders

- Beta-ketothiolase deficiency
- Glutaric academia, Type I
- Isobutyryl CoA dehydrogenase deficiency
- Isovaleric academia
- Malonic aciduria
- Maple syrup urine disease
- Methylmalonic acidemias (8 types)
- Propionic academia
- 3-Hydroxy-3-methylglutaryl (HMG) CoA lyase deficiency
- 2-Methyl-3-hydroxybutyryl CoA dehydrogenase deficiency
- 2-Methylbutyryl CoA dehydrogenase deficiency
- 3-Methylcrotonyl CoA carboxylase deficiency
- 3-Methylglutaconyl CoA hydratase deficiency
- Multiple carboxylase deficiency

Fatty Acid Oxidation Disorders

- Carnitine uptake/transport defects
- Multiple acyl-CoA dehydrogenase deficiency (MADD)
- Short chain acyl-CoA dehydrogenase deficiency (SCAD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
- Long chain 3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)

Mountain States Genetics Regional Collaborative Center
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State: New Mexico

Newborn Screening Laboratory Contact: Oregon State Public Health 503-229-5466

Newborn Screening Follow-up Program Contact: Carla Ortiz RN BSN 505-476-8858

Birthrate 2006: 28,800

Number of disorders Screened for: 6

Please list disorders screened for and 2006 presumed positive and confirmed cases:
Congenital Hypothyroidism, Congenital Adrenal Hyperplasia, Phenylketonuria,
Galactosemia, Sickle Cell Conditions, Biotinidase Deficiency.

January 2007 we have increased to 26 disorders.

Number of presumed positive: PKU-10, CH-181,-Galt-4, SC-0, Biotinidase-20, CAH-308

Number of Confirmed cases: Variant hyperphenlalaniemia-1, CH-14, Transient hypothyroidism-2, Galactocemia Variant-6

Does your state have a NBS Committee? Yes

If so provided contact information for chair and staff contact: Chair is Carla Ortiz 505-476-8858

How long does you state store residual specimens, where and under what conditions? Until age 21 Room Temp.

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State: Texas

Newborn Screening Laboratory Contact: Susan Tanksley, Ph.D., Biochemistry and Genetics Branch Manager & Lynette Borgfeld, Newborn Screening Group Manager

Newborn Screening Follow-up Program Contact: Margaret Drummond-Borg, M.D. & David Martinez

Birthrate 2006: 405,822

Number of disorders Screened for: 27

Please list disorders screened for and 2006 presumed positive and confirmed cases:

AMINO ACID DISORDERS: Argininosuccinic Acidemia (ASA), Citrullinemia (CIT), Homocystinuria (HCY), Maple Syrup Urine Disease (MSUD), Phenylketonuria (PKU), Tyrosinemia Type I (TYRI). FATTY ACID DISORDERS: Medium Chain Acyl-CoA Dehydrogenase Def. (MCAD), Very Long Chain Acyl-CoA Dehydrogenase Def. (VLCAD), Long Chain Hydroxyacyl-CoA Dehydrogenase (LCHAD), Trifunctional Protein Def. (TFP), Carnitine Uptake Def. (CUD), Carnitine Palmitoyl Transferase Def.1 (CPT1). ORGANIC ACID DISORDERS: Glutaric Acidemia I (GA-I), 3-OH 3-Methyl Glutaric Aciduria (HMG), Isovaleric Acidemia (IVA), Multiple Carboxylase Def. (MCD), 3 -Methyl Crotonyl-CoA Carboxylase Def. (3-MCC), Methylmalonic Acidemia (MMA), Propionic Acidemia (PA), Beta-Ketothiolase Def. (BKT). GALACTOSEMIA. BIOTINIDASE DEFICIENCY. CONGENITAL HYPOTHYROIDISM (CH), CONGENITAL ADRENAL HYPERPLASIA (CAH), HEMOGLOBINOPATHIES, including Hb, S/S, Hb S/C, Hb S-Beta thalassemia

Number of presumed positive: CAH – 3900, GALT – 631, PKU – 256, Thyroid – 7668, Hemoglobinopathies - 22202 (with traits), Hemoglobinopathies (clinically significant) - 427

Does your state have a NBS Committee? We have three ad hoc consultant groups.

If so provided contact information for chair and staff contact: These are coordinated through the NBS Case Management Program. It is not a formal committee & therefore does not have a chair.

How long does you state store residual specimens, where and under what conditions? Indefinitely since July 2002. See policy for specific information.

Mountain States Genetics Regional Collaborative Center
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State: Utah

Newborn Screening Laboratory Contact: Norm Brown, Section Chief

Newborn Screening Follow-up Program Contact: Fay Keune, Program Manager

Birthrate 2006: preliminary occurant births - 54,521

Number of disorders Screened for: 36

Disorders screened for and 2006 presumed positive and confirmed cases:

(Data is preliminary)

<u>Category</u>	<u>Presumed positive</u>	<u>Confirmed</u>
Amino Acids:	260	10
Acylcarnitines	725	17
Biotinidase	9	1
Congenital Hypothyroidism	361	63
Congenital Adrenal Hyperplasia	213	7
Galactosemia	64	56
Hemoglobinemias	310	281

Does your state have a NBS Committee? Yes

Chair and staff contact:

Jeff Botkin MD

jeffrey.botkin@hsc.utah.edu

(801) 581-7170

How long does you state store residual specimens, where and under what conditions?

We are currently involved in a grant project with the Utah Birth Defects Network. For this project all specimens will be held for 2 years in a -20 degree freezer, in low gas permeable bags with desiccant.

Our routine storage is 90 days in a refrigerator (2-8 degrees) in cardboard boxes.

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State: Wyoming

Newborn Screening Laboratory Contact: Dr. Jim Bebee (303) 692-3488
Dan Wright (303) 692-3673

Newborn Screening Follow-up Program Contact: Paul Ramirez (307) 777-5413
Sheli Gonzales (307) 777-7943

Birthrate 2006: 6,925

Number of disorders Screened for: 29 (including hearing)

Please list disorders screened for and 2006 presumed positive and confirmed cases:

Jan. 1, 2006 through June 30, 2006 we screened for the first eight conditions listed below.
As of July 1, 2006 our screening added the remaining conditions listed.

Congenital Adrenal Hyperplasia (CAH)
Congenital Hypothyroidism (CH)
Biotinidase Deficiency (BIOT)
Cystic Fibrosis (CF)
Hearing (HEAR)
Hemoglobinopathies (Hgb)
Galactosemia (GALT)
Phenylketonuria (PKU)
Argininosuccinic Acidemia (ASA)
Citrullinemia (CIT)
Homocystinuria (HCY)
Maple Syrup Urine Disease (MSUD)
Tyrosinemia (TYR 1)
Carnitine Uptake Defect (CUD)
Long-chain L-3-OH acyl-CoA Dehydrogenase Deficiency (LCHAD)
Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
Trifunctional Protein Deficiency (TCP)
Very long-chain acyl-CoA Dehydrogenase Deficiency (VLCAD)
3-Hydroxy 3-Methyl Glutaric Acid (HMG)
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)
B-Ketothiolase Deficiency (BKT)
Glutaric Acidemia Type 1 (GA1)
Isovaleric Acidemia (IVA)
Methylmalonic Acidemia – Mutase Deficiency (MUT)
Methylmalonic Acidemia (Cbl, A, B)
Multiple Carboxylase Deficiency (MCD)
Propionic Acidemia (PROP)

The conditions listed below are the 2006 presumed positive and confirmed cases:

Conditions:	Presumed:	Confirmed:
Congenital Adrenal Hyperplasia (CAH)	23	1
Congenital Hypothyroidism (CH)	37	1
Biotinidase Deficiency (BIOT)	6	0
Cystic Fibrosis (CF)	5	1
Hemoglobinopathies (Hgb)	1	0
Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	1	1

Does your state have a NBS Committee? No

How long does you state store residual specimens, where and under what conditions?

6 mos @ room temperature

Blotter Disposal

All blotters will be retained, at room temperature for 6 months at which time they will be sealed in a biohazard bag with autoclave reactive tape that, after autoclaving, will clearly indicate that the contents have been sterilized. This sterilization will occur at 250°C at 23 lbs. pressure for 1 hour (1 cycle). The blotters will then be discarded in the general garbage.