



ARIZONA DEPARTMENT OF HEALTH SERVICES

PREPAREDNESS

Guide to Laboratory Services: Newborn Screening

**Arizona Department of Health Services
Bureau of State Laboratory Services
250 North 17th Avenue
Phoenix, Arizona 85007
(602) 542-1188**

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General Information

Bureau Chief, Laboratory Services

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Assistant Bureau Chief

Kathryn Fitzpatrick

Hours of Operation: 8:00 am to 5:00 pm Monday through Friday. Sample receipt services available on Saturdays (9:00 am to 4:30 pm).

Annual Holiday Schedule: Laboratory Services observes all state recognized holidays.

Location: 250 North 17th Avenue, Phoenix, Arizona 85007

ASPHL Telephone Number: (602) 542-1188

WATTS Line: (800) 525-8915

ASPHL Fax Number: (602) 542-0760

Emergency Phone

(Weekends/After Hours): (480) 955-0888

Arizona State Public Health Laboratory Contact Information

Section	Supervisor	Contact Information
Chief, Newborn Screening	Trung (Joe) Huynh	(480) 261-9351 trung.huynh@azdhs.gov
Receiving / Shipping	Adrian Fichter	(602) 542-1190 labreceiving@azdhs.gov
Newborn Screening Demographics	Yvonne Carroll	(602) 364-1468 yvonne.carroll@azdhs.gov
Newborn Screening Follow-Up	Fran Altmaier	(602) 364-1814 fran.altmaier@azdhs.gov
Newborn Screening Education Quality Improvement Secure Result Viewer	Katherine Fullerton	(602) 364-0128 NBSeducation@azdhs.gov
Newborn Screening Laboratory	Okate Bilante Nam Nguyen Debbie Wright	(602) 542-1184 nbslab@azdhs.gov

Office of Newborn Screening
Telephone Number: 602-364-1409
Fax Number: 602-364-1495

Core Functions and Capabilities of State Public Health Laboratories

State public health laboratories face the broad challenge of working towards prevention, control of disease and improvement of health. To function in this capacity, the Office of Newborn Screening (ONBS) at the Arizona State Public Health Laboratory (ASPHL) provides screenings of congenital conditions for Arizona newborns. Since these rare conditions can have a devastating effect on the infant's health and development, early detection, diagnosis, and intervention can prevent disability or death. In Arizona, newborns are routinely screened for certain genetic, endocrine, and metabolic disorders using drops of blood from the newborn's heel. Newborns are also tested for hearing differences and critical congenital heart defects (CCHDs) prior to discharge from a hospital or birthing center.

The goals of the ONBS are:

1. To identify newborns with congenital disorders that can affect a child's long-term health or survival.
2. To ensure timely initial follow-up services for identified newborns, including confirmatory diagnostic testing, timely treatment and intervention, as well as long term follow up and surveillance.
3. To assist the families of the affected newborns and infants.

Newborn Screening Services

Per A.R.S. § 36-694, the Arizona State Public Health Laboratory (ASPHL) serves as the laboratory to provide testing services for the Arizona Newborn Screening (NBS) Program. Arizona screens for both core and secondary disorders currently on the Recommended Uniform Screening Panel ([RUSP](#)). All infants born in Arizona are required by law (Arizona Administrative Code R9-13-204) to have a Newborn Screening ordered. The A.A.C R9-13-205 also mandates the collection of second screen specimens for all infants born in Arizona.

Below are the core disorders tested by the ASPHL for Newborn Screening dried blood spot:

Disorder(s)	Testing Method	First Screen Panel	Second Screen Panel
Congenital Hypothyroidism	¹ DELFI [®]	✓	✓
Congenital Adrenal Hyperplasia	DELFI [®]	✓	✓
Amino Acid Disorders: Phenylketonuria (PKU) Maple syrup urine disease (MSUD) Homocystinuria (HCY) Citrullinemia type 1 (CIT I) Tyrosinemia type I (TYR I) Argininosuccinic acidemia (ASA)	FIA-MS/MS ²	✓	✓
Fatty Acid Oxidation Disorders: Medium chain acyl-CoA dehydrogenase deficiency (MCAD) Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD) Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD) Trifunctional protein deficiency (TFP) Carnitine uptake defect (CUD)	FIA-MS/MS	✓	✓
Organic Acid Disorders: Isovaleric acidemia (IVA) Glutaric acidemia type I (GA1)			

3-Hydroxy-3-Methylglutaric Aciduria (HMG) Multiple carboxylase deficiency (MCD) Methylmalonic acidemia (Cobalamin defects) (Cbl A,B) Methylmalonic acidemia (mutase deficiency) (MUT) 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC) Propionic acidemia (PROP) Beta-ketothiolase deficiency (BKT)	FIA-MS/MS	✓	✓
Hemoglobin Disorders: Hb S/Beta-thalassemia (S, β Th) Hb S/C disease (Hb S/C) Sickle cell anemia (Hb SS)	1st tier - IEF ^{3,6} 2nd tier - HPLC ^{4,6}	✓	*
Galactosemia (GALT)	Enzymatic assay	✓	*
Cystic Fibrosis (CF)	1st tier - DELFIA 2nd tier - CFTR gene analysis	✓	*
Severe Combined Immunodeficiency (SCID) ⁶	Real-time PCR	✓	*
Spinal Muscular Atrophy (SMA ^{**}) ⁶	Real-time PCR	✓	*
X-Linked Adrenoleukodystrophy (XALD)	1st tier - FIA-MS/MS 2nd tier - LC-MS/MS ^{5,6}	✓	*
Pompe Disease (Glucosidase acid-1, 4-alpha deficiency)	Enzymatic digital microfluidic	✓	*
Mucopolysaccharidosis Type I (MPS I)	Enzymatic digital microfluidic	✓	*
Mucopolysaccharidosis Type II (MPS II)⁶	Fluorometric enzymatic assay	✓	*
Biotinidase deficiency (BIOT) ⁶	Enzymatic assay	✓	*
Guanidinoacetate methyltransferase deficiency (GAMT) 6,7	FIA-MS/MS	✓	✓

¹ Dissociation-enhanced lanthanide fluorescence immunoassay

² Flow-injection analysis–tandem mass spectrometry

³ Isoelectric focusing

⁴ High-performance liquid chromatography

⁵ Liquid Chromatography with tandem mass spectrometry

⁶ These tests are not FDA-approved and they were developed and their performance characteristics determined by the Arizona State Public Health Laboratory

⁷ Schedule to be implemented by 02/01/2025

* These disorders are not normally performed on the second screen specimen after a successful first screen. However, the laboratory may add them to the second screen panel as needed.

** The real-time PCR assay used for SMA screening can detect only SMA caused by the homologous loss of SMN1 exon 7, which accounts for 95% of cases. This assay cannot detect SMA cases caused by variants outside of the SMN1 exon 7 region.

In addition to the above genetic/congenital disorders, all newborns in Arizona are required to have a hearing screen (HEAR) and Critical Congenital Heart Defects (CCHD) screen prior to discharge from the hospital. These tests are not performed by the ASPHL; rather they are “point of care” tests that are completed by the hospital or midwife.

Requesting Collection Kits and Mailing Envelopes

Per R9-13-203, submitters and/or primary care providers should only use a specimen collection kit supplied by the ASPHL to collect NBS screens. To order NBS collection kits, please fill out the [Newborn Screening Bloodspot Collection Kit Order Form](#) and submit it by either faxing or emailing the Receiving Section at:

Arizona Department of Health Services
Bureau of State Laboratory Services
ATTN: Receiving Section
250 North 17th Avenue
Phoenix, AZ 85007
Fax: (602) 364-0758
Phone: (602) 542-1190
Email: labreceiving@azdhs.gov

The form is also available at <https://www.azdhs.gov/preparedness/state-laboratory/shipping-receiving/index.php#forms-supply-order>. Please request materials before they are required as the expected turn-around time per order is **FIVE** business days.

Please only order according to the birth rate at your facility. Stockpiling of NBS collection cards can result in using expired kits to collect specimens and, as a result, affect newborn care.

Specimen Collection

It is the responsibility of the birthing institution to ensure that a first screen specimen is collected ideally between 24-36 hours but no later than 72 hours, or prior to discharge. The second screen specimen should be collected between 5 and 10 days after the birth of the newborn. Specimens should arrive in the laboratory no later than 14 days from date of collection. Specimens collected from a newborn / infant less than one year old are acceptable for testing.

The ASPHL has two types of specimen collection kits which serve the different functions listed below:

- **Linked Kit** (*Figure 1*)

The Linked Kit (pictured below) allows for automatic linkage of the first and second Newborn Screen Specimens. Names of newborn infants frequently change during the first weeks of life. The linked kits allow better identification of these newborns/infants. These paired kits have a common collection kit number, which is used to link first and second specimens collected from an individual newborn/infant. The first dried bloodspot (DBS) card (red) of the kit is used to collect a heel-stick specimen at the hospital or birthing institution prior to the baby's discharge. The second DBS card (blue) of the linked kit is sent home with the mother along with the NBS education brochure in the provided pink envelope. The second DBS can be collected at the Primary Care Physician at the first well care visit or by a commercial lab provider.

Newborn Screening		PRINT ALL INFORMATION LEGIBLY		Accession Number:	
1st		SPECIMEN		Date / Time Stamp	
EXPIRATION 2028-07-31 IVD		DO NOT WRITE IN THIS SPACE		Submitter / Physician Information AZ251846701	
Baby's Name Last: _____ First: _____ Date of Birth _____ Time of Birth _____ Birth Weight _____ Sex <input type="checkbox"/> M <input type="checkbox"/> F Date of Collection _____ Time of Collection _____ Current Weight _____ Baby's AHCCCS # _____ Gestational Age _____ MR # _____ Weeks _____ Days _____ <input type="checkbox"/> Single Birth <input type="checkbox"/> Multiple Birth (circle one) A B C D		Submitter Name/ID: _____ Ordering Physician (Hosp.): _____ Follow-up Physician Name (Last,First): _____ Phone: (____) _____ Practice Address: _____ City, State, Zip: _____		Birth Mother's Information Mom's Name _____ First: _____ Mom's Date of Birth: ____/____/____ Maiden Name: _____ (OR) Other Person with Custody: _____ Street Address: _____ City, State, Zip: _____ Phone: (____) _____ Mom's AHCCCS# _____	
Race <input type="checkbox"/> (1) White <input type="checkbox"/> (2) African Amer. <input type="checkbox"/> (3) Asian <input type="checkbox"/> (4) Amer. Indian <input type="checkbox"/> (5) Other <input type="checkbox"/> Hispanic <input type="checkbox"/> TPN <input type="checkbox"/> Never Fed		Food Source <input type="checkbox"/> (1) Breast Only <input type="checkbox"/> (2) Milk / Lactose Formula <input type="checkbox"/> (3) Soy Formula <input type="checkbox"/> (4) Breast & Lactose <input type="checkbox"/> (5) Breast & Soy		Status <input type="checkbox"/> Meconium Test <input type="checkbox"/> In NICU/Special Care Nursery <input type="checkbox"/> Transfusion (RBC ONLY) before collection <input type="checkbox"/> YES <input type="checkbox"/> NO <input type="checkbox"/> Date FIRST transfused _____ Phlebotomy / FINAL Screen Results <input type="checkbox"/> Pass (passed on attempt) <input type="checkbox"/> Not screened <input type="checkbox"/> Parental Refusal <input type="checkbox"/> Parental Consent/Depose <input type="checkbox"/> Reason (please one) <input type="checkbox"/> Invalid/NG/OTC <input type="checkbox"/> Other _____ <input type="checkbox"/> Fail	
EXP DATE 2028-07-31 PerkinElmer 226 LOT 116265 / 50010007		EXP DATE 2028-07-31 PerkinElmer 226 LOT 116265 / 50010007		EXP DATE 2028-07-31 PerkinElmer 226 LOT 116265 / 50010007	

Figure 1 - Arizona Newborn Screening Linked Kit

- Supplemental Kit (Figure 2)**

The primary use of the Supplemental Kit (pictured below) is to collect a sample for repeat testing of a previously reported abnormal or borderline result or for a repeat testing of an unsatisfactory specimen. This kit is routinely used at commercial laboratories, at physicians' offices and by midwives.

When filling out the information on this card for a repeat testing of a previously reported abnormal or borderline result, please ensure to check the box "Recall" on the card and write the name of the disorder that was screened abnormal, borderline or unsatisfactory next to the box.

The NBS laboratory also accepts this card for the second screen in situations when the linked kit is lost or otherwise unavailable. Please ensure to check the "Second Screen" box when filling out the information on the card. The use of the Supplemental Kit for the first screen is acceptable, but not preferable.

EXPIRATION
 2028-07-31

IVD

AZ

PRINT ALL INFORMATION LEGIBLY Accession Number: _____

SUPPLEMENTAL SCREENING FORM DO NOT WRITE IN THIS SPACE

Newborn Screening

Date / Time Stamp

☐ First Screen
 ☐ Second Screen
 ☐ Recal
 (Indicate disorder & re-screening)

Baby's Name _____

Last: _____ **First:** _____

Date of Birth _____ a.m. _____ p.m.
 Birth Weight _____ Grams
 Sex ☐ M ☐ F

Date of Collection _____ a.m. _____ p.m.
 Current Weight _____ Grams

Baby's AHCCCS # _____ **Gestational Age** _____ Weeks _____ Days

MR # _____

☐ Single Birth
 ☐ Multiple Birth (circle one)
 A B C D

Race	Food Source	Status
<input type="checkbox"/> 1 White	<input type="checkbox"/> 1 Breast Only	Meconium passed in NICU/Special Care Nursery <input type="checkbox"/> Y <input type="checkbox"/> N
<input type="checkbox"/> 2 African Amer.	<input type="checkbox"/> 2 Milk / Lactose Formula	<input type="checkbox"/> Y <input type="checkbox"/> N
<input type="checkbox"/> 3 Asian	<input type="checkbox"/> 3 Soy Formula	Transferrin (RBC ONLY) before collection? <input type="checkbox"/> Y <input type="checkbox"/> N
<input type="checkbox"/> 4 Amer. Indian	<input type="checkbox"/> 4 Breast & Lactose	if YES, date FIRST transfused
<input type="checkbox"/> 5 Other	<input type="checkbox"/> 5 Breast & Soy	
<input type="checkbox"/> 6 Hispanic <input type="checkbox"/> Y <input type="checkbox"/> N	<input type="checkbox"/> 6 TPN	<input type="checkbox"/> 0 Never Fed

Pulse Oximetry FINAL Screen Results

☐ Pass (assessed on attempt) 1st _____ 2nd _____ 3rd _____
 ☐ Not screened
 ☐ Parental Refusal
 ☐ Prenatal Cardiac Diagnosis Reason (choose one)
 ☐ Maternal NICU/SCH
 ☐ Other _____

Submitter / Physician Information **AZ250349701** SN

Submitter Name/ID: _____

Ordering Physician (Hosp.): _____

Follow-up Physician Name (Last, First): _____

Phone: (____) _____

Practice Address: _____

City, State, Zip: _____

Birth Mother's Information

Mom's Name _____ **First:** _____

Mom's Date of Birth: ____ / ____ / ____ **Maiden Name:** _____

(OR) Other Person with Custody: _____

Street Address: _____

City, State, Zip: _____

Phone: (____) _____

Mom's AHCCCS# _____

☐ Insurance papers included
☐ Parent Refused Bloodspot Testing

EXP DATE
 2028-07-31

LOT

Ahlstrom PerkinElmer 226

116285 / 30330006

AZ250349701 SN

Figure 2 - Arizona Newborn Screening Supplemental Kit

Sample Submission Guidelines

Submit all samples to the following location:

Arizona Department of Health Services
Bureau of State Laboratory Services
Attn: Newborn Screening
250 N. 17th Ave
Phoenix, AZ 85007
(602) 542-1188
Ordering: labreceiving@azdhs.gov

All samples and their envelopes must be identified with the appropriate labels, client, and patient information. Any samples that have been damaged and/or not properly identified will be rejected.

Newborn Screening Blood Spot Cards

- Primary and secondary Newborn Screening DBS cards being sent for disorder testing are considered non-infectious and are exempt from the hazardous material regulations. See the Requesting Collection Kits and Mailing Containers section for information on requesting screening cards from the ADHS Shipping and Receiving section.

Shipment of Specimens

Per A.A.C R9-13-203, all newborn demographic information must be filled out completely on the DBS card top sheet before specimen collection. Specimens must be **completely dry** before insertion into a mailing envelope. For all College of American Pathologists (CAP) regulated collection laboratories, **a packing list of all samples must be included with each shipment of samples** submitted to the ASPHL.

This packing list must include each of the following items:

1. Submitter name and phone number
2. Date the list is generated (date sent)
3. Newborn first and last name, and Medical Record Number
4. Newborn date of birth
5. Courier tracking number; if applicable

Additional requirements for the shipment of the DBS cards are as follows:

1. All fields on the top sheet of the bloodspot card must be filled out accurately, legibly and completely.
2. Bloodspot cards must be organized in the same order that they are listed on the packing list (alphabetical or otherwise).
3. Bloodspot cards must be rubber-banded together (no tape or staples).
4. **Do not package the dried blood spot specimens inside a sealed plastic bag.** The lack of air exchange inside the sealed plastic bag may cause heat buildup, moisture accumulation, and/or chemical leaks from the plastic, which can damage specimen integrity.

Within 24 hours of collection, the specimens should either be sent by courier (Specialized Delivery Services (SDS), FedEx, or other) or mailed to the ASPHL in Phoenix. The ASPHL should receive specimens within 3 days of collection to allow for timely detection of time critical disorders. Specimens not received within 14 days of collection will be rejected as “Unsatisfactory: Specimen too old upon receipt.”

If you need to set up a courier service or a FedEx account to send samples to the ASPHL, please send your request via email to nbseducation@azdhs.gov and CC: labreceiving@azdhs.gov

See the Sample Submission Guidelines section for submission address and information.

Specimen Storage and Retention Policy

After the newborn screening process is complete, specimens are stored at room temperature for approximately 3 months (90 days). The patient information is detached and shredded and the sample portion is autoclaved and discarded. The laboratory may keep the de-identified sample portion for quality assurance testing or validating new laboratory methods for newborn screening. The samples are not used for research.

Reporting and Interpretation of Results

Normal laboratory results are reported as “Normal Findings” or “Screen Negative”.

All unsatisfactory samples (i.e. rejected samples) are reported with a specific code (please see Specimen Rejection Policy below). A sample can be unsatisfactory for all of the tests in the panel or only for specific tests. The ONBS Follow-up staff will contact the submitter or the primary care provider to ask for another NBS sample for repeat testing.

All “not-within-normal” results are reported with the quantitative or qualitative value, if applicable, and the result interpretation (i.e. Abnormal, Borderline, Elevated, etc.). Result interpretations will include a comment on the laboratory report to provide guidance to the provider. The ONBS Follow-up staff will provide immediate case management services for any newborn identified with critical presumptive positive for any condition on the ONBS testing panel.

Within 24 hours of completion of laboratory testing and demographic data entry, laboratory reports (mailers) are generated and available on the Secure Remote Viewer (SRV) portal. In certain instances, these may be mailed to the submitter and the follow-up physician or ordering provider. For any sample whose submitter and follow-up physician are missing on the DBS card, the physical laboratory report will be destroyed.

Laboratory reports are available on the [Secure Results Viewer Portal](#) (SRV). To request access for an SRV account, please contact nbseducation@azdhs.gov.

Specimen Rejection Policy

Specimens are rejected as unsatisfactory to test for the following reasons:

Unsat code	Unsat code description and instruction
UCC	The sample quality is unsatisfactory due to clotted or caked blood. Please submit another Newborn Screening sample no later than one week (if not already collected).
UCE	The sample was collected on an expired specimen card. Please submit another Newborn Screening sample no later than one week (if not already collected). In addition, please review your inventory and discard all expired collection cards.
UII	Unsatisfactory specimen due to improper specimen information. Please submit another Newborn Screening sample no later than one week (if not already collected).
UIO	The Newborn Screening Program cannot test babies over one year of age. Please do not collect additional samples. If clinically indicated, please consult with a medical specialist.
UIS	There was insufficient specimen collected for this sample to be tested. Please submit another Newborn Screening no later than one week (if not already collected).
ULA	Due to unforeseen circumstances the laboratory was not able to test the Newborn Screening sample. Please submit another sample no later than one week (if not already collected). We apologize for any inconvenience.
UMA	The sample quality is unsatisfactory due to multiple specimen applications. Please submit another Newborn Screening sample no later than one week (if not already collected). For assistance with proper collection techniques, please email us at NBSEducation@azdhs.gov .
UNI	The sample was not tested due to missing critical information to verify the identity of the patient. Please review A.A.C. R9-13-204(B) for required patient information. Please submit another Newborn Screening sample no later than one week (if not already collected).
UNO	There was no sample collected for this patient. Please submit another Newborn Screening sample no later than one week (if not already collected). For assistance with proper collection techniques, please email us at NBSEducation@azdhs.gov .
UNS	The sample quality is unsatisfactory due to incomplete saturation of the blood through the filter paper. Please submit another Newborn Screening sample no later than one week (if not already collected). For assistance with proper collection techniques, please email us at NBSEducation@azdhs.gov .
UPR	The Newborn Screening was not conducted due to parent(s) or guardian(s) refusal. Please provide education on newborn screening per A.A.C. R9-13-

	203(G)(H). Babies can be tested up to one year of age. If clinically indicated, please consult with medical specialists.
USC	The specimen was contaminated upon receipt. Please submit another Newborn Screening sample no later than one week (if not already collected).
USD	The specimen was detached from the collection kit upon receipt. The laboratory was unable to verify the patient's identity. Please submit another Newborn Screening sample no later than one week (if not already collected).
UST	The sample was not tested due to serum separation. Please submit another Newborn Screening sample no later than one week (if not already collected). For assistance with proper collection techniques, please email us at NBSEducation@azdhs.gov .
UTO	This specimen was received more than 14 days after collection and was not tested. Please submit another Newborn Screening no later than one week (if not already collected). For assistance with transit time, please email us at NBSEducation@azdhs.gov . If clinically indicated, please consult with a medical specialist.
UTS	The sample was not tested due to torn or scratched circles. Please submit another Newborn Screening sample no later than one week (if not already collected). For assistance with proper collection techniques, please email us at NBSEducation@azdhs.gov .
NOT	The newborn has received two successful Newborn Screenings. Additional samples are no longer needed and will not be tested.