



# **Newborn Screening Section CY2023 Annual Report**

**Submitted by collaborated team of:**

- **Pat Purifoy, RN NBS Manager**
- **Jomeka Edwards, M.S.N.S. NBS Lab Manager**
- **Leslie Himstedt, BSMT (ASCP) PHL Clinical/Biological Branch Chief**
- **Wanda Simon, Sr. Epidemiologist**
- **Tsai Mei Lin, Data Processing Senior Project Leader/DPHP Health Statistics**

## Introduction

Newborn screening (NBS) for inborn conditions has been mandatory in Arkansas since Act 192 of 1967 stipulated screening of all newborns for phenylketonuria. Since that time, the number of conditions screened for has grown substantially. The program oversees follow-up on 30+ genetic disorders screened using the blood spot card in addition to two point of care tests, hearing screen and critical congenital heart disease, for a total of 35 core disorders.

The NBS Program is located within the Child and Adolescent Health Section (CAH) and the Division of Division for Health Advancement of the Arkansas Department of Health (ADH). The NBS Lab and Follow-Up programs normal working hours are Monday – Friday from 8am – 4:30pm. The NBS Follow-Up program nurses work alterative hours from 7am – 6pm on Monday – Friday. Saturdays and holidays the NBS Lab and Follow-up nurses provide coverage for emergent cases.

- How many are served by the NBS program?

In 2023, ninety-seven-point six percent (97.6%) of the approximately 34,264 babies born in Arkansas were screened for genetic disorders.

- How is the NBS program funded?

The NBS program is funded by newborn screening fees, currently billed at \$131 per specimen.

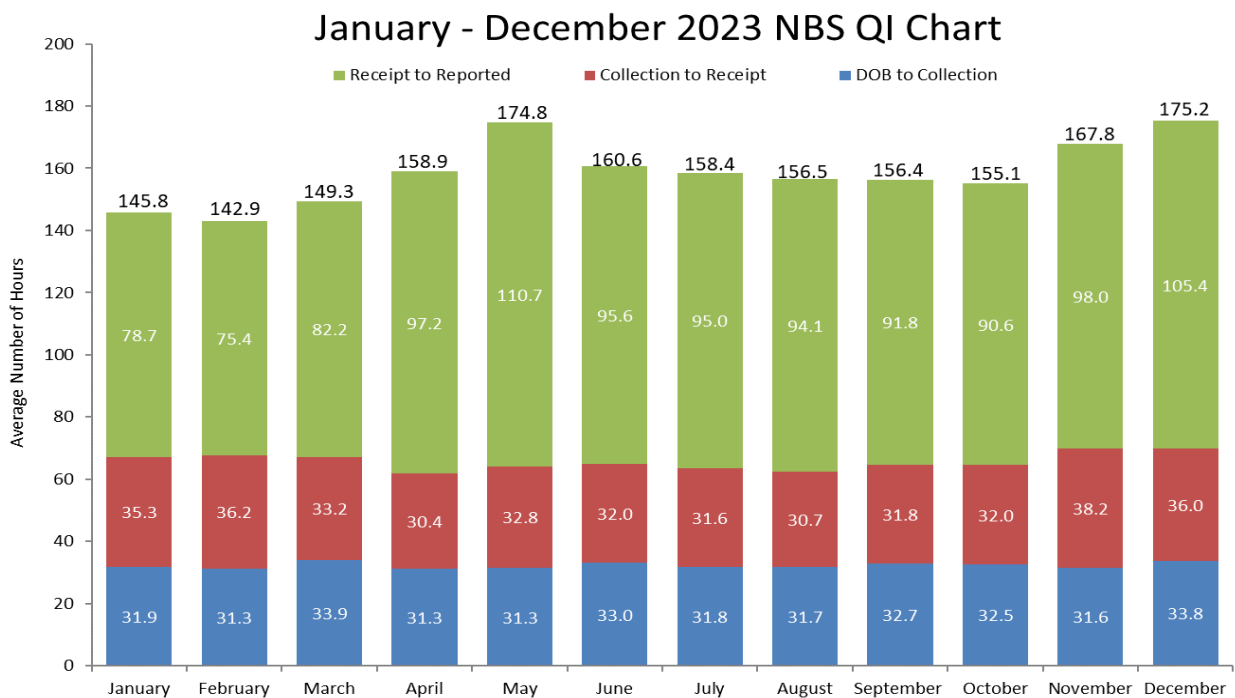
- How are services provided?

The NBS program is responsible for providing follow up actions for abnormal, borderline, inconclusive or indeterminate screening results for inborn conditions. ADH NBS program collaborates with AR Children's Hospital medical consultants to develop and implement NBS disorder protocols.

## Reports

Several reports are prepared throughout the year and shared with stakeholders.

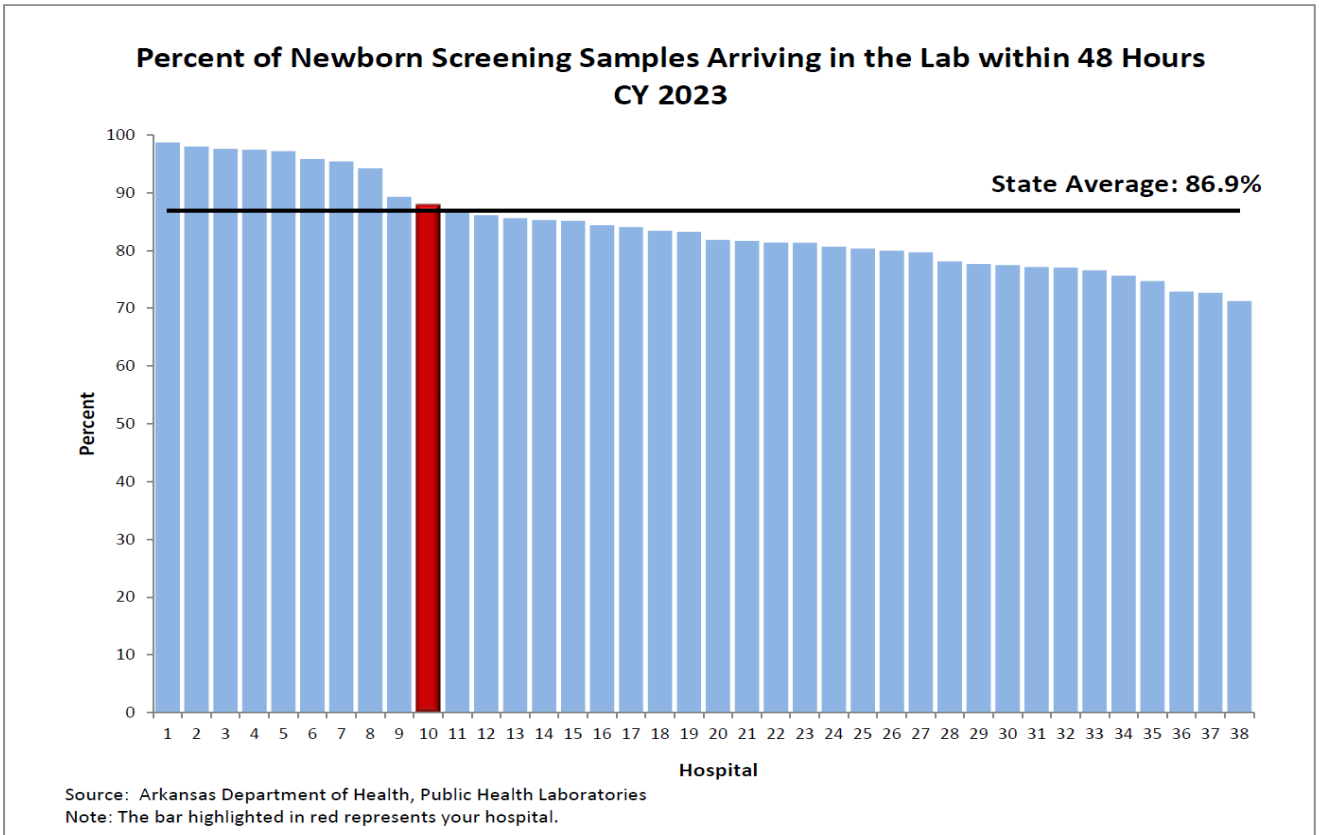
**Newborn Screening Quality Improvement (QI) Graph:** The Public Health Lab and Newborn Screening managers work diligently and continue to monitor monthly timeliness of data at three key points. These three key data points are: time of birth to time of collection, time of collection to time of receipt in the lab and time of receipt in the lab to time of reporting all results. Time is measured in hours and a goal of less than 168 hours (7 days) from birth to reporting of test results was set. Data is provided to ADH Health Statistics representative for compilation.



**Figure 1**

Figure 1 illustrates the NBS QI 2023 report, indicating the number of hours that elapsed from birth to reporting of results. The average time is calculated to be 158.5 hours, meeting the QI project goal of less than 168 hours. The program provides support to partner hospitals with virtual education opportunities and technical assistance to ensure effective collection to receipt in the lab for processing.

**Yearly 2023 Comparison Report:**



**Figure 2**

Figure 2 illustrates a yearly comparison graph, sent in addition to the quarterly report example shown in Figure 3, representing 37 birthing hospitals plus Arkansas Children’s Hospital that are distributed. This anonymized bar graph report shows placement among peers, with the specific hospital’s bar in red, along with the average submission for the state.

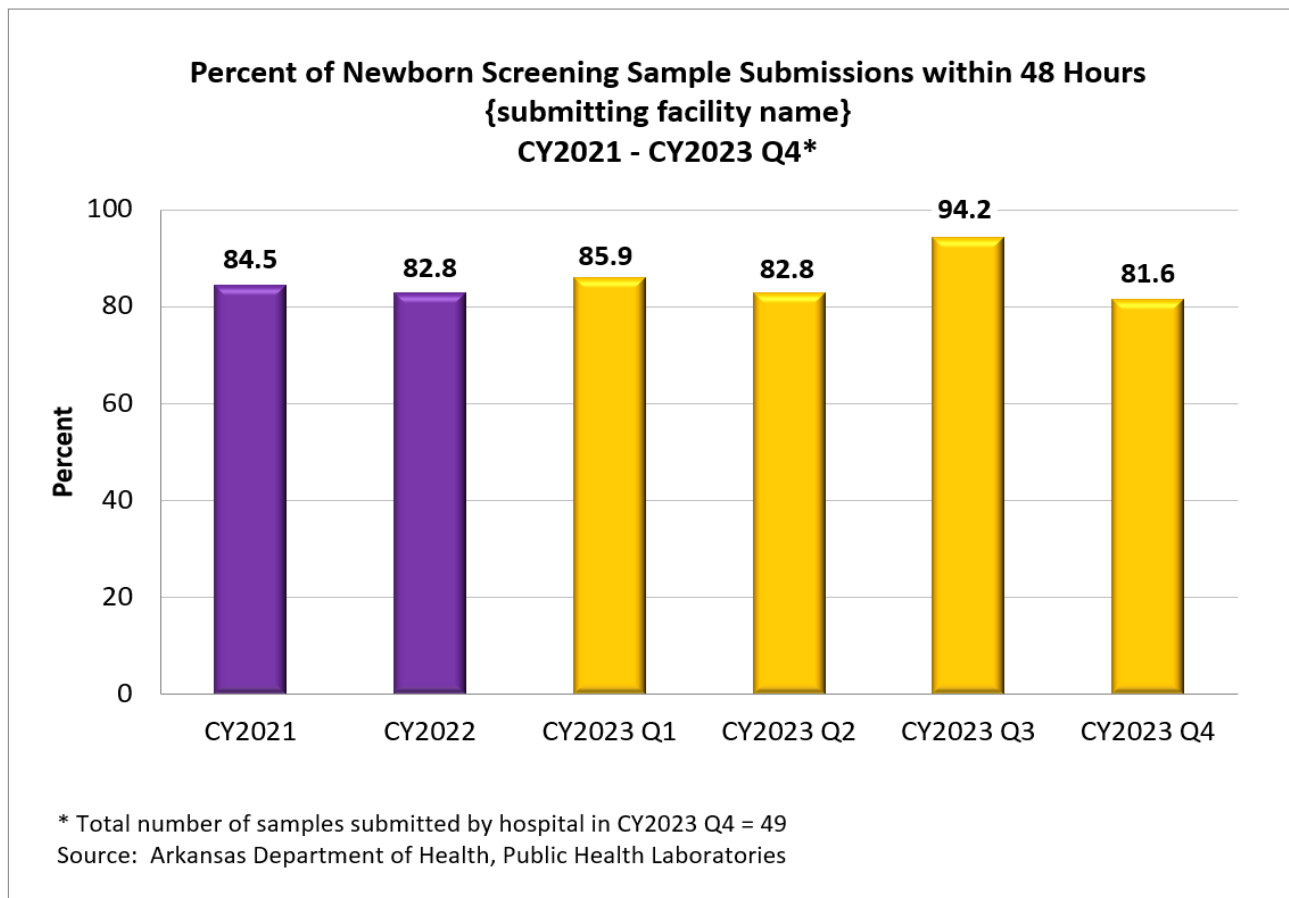


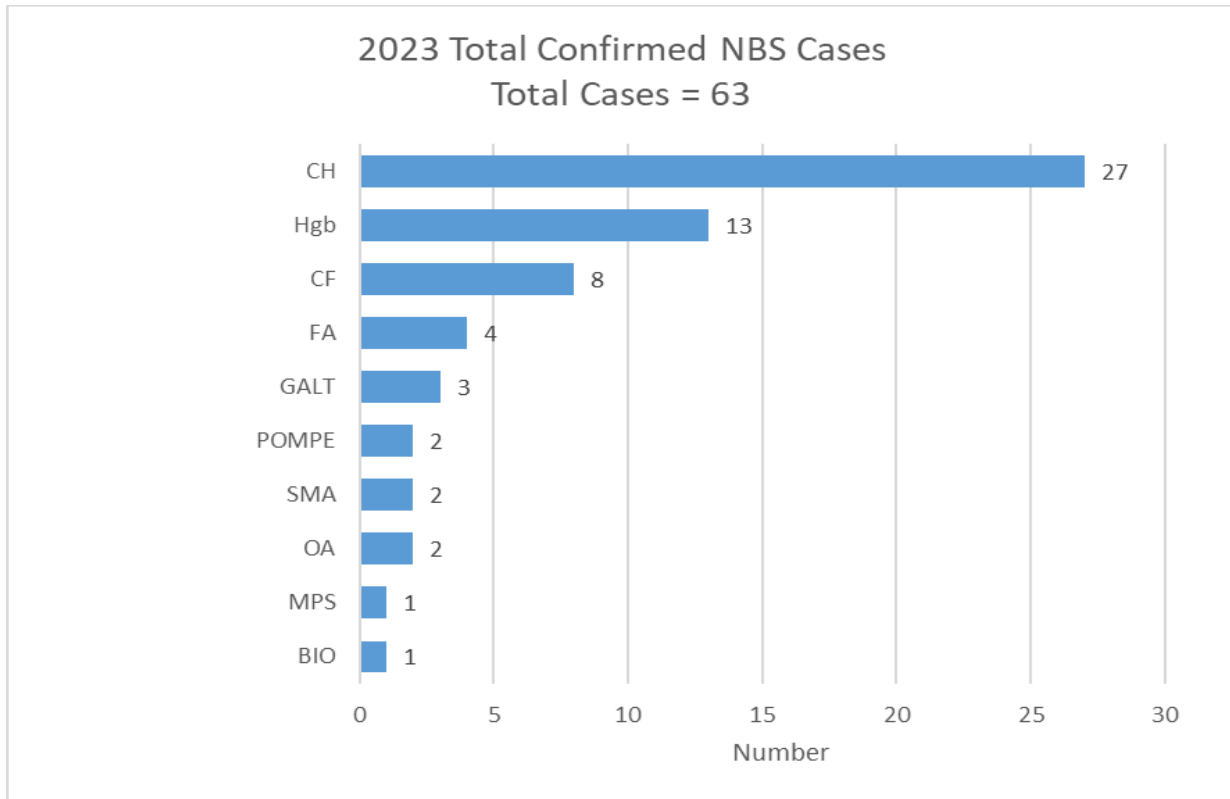
Figure 3

Figure 3 illustrates an example of the quarterly report sent to the hospital administrators, lab, and Nursery/Nursery Intensive Care (NICU) managers at each birthing hospital and Arkansas Children’s Hospital (ACH).

Each birthing hospital receives a quarterly Hospital Timeliness Report to identify the number of specimens collected and received by the NBS lab within 48 hours of collection.

Any birthing hospital that does not meet the goal of 80% of specimens reaching the lab with 48 hours is contacted to discuss potential issues related to timely specimen submission. The ADH NBS Follow-up Manager and AR State Genetics Coordinator offer to all facilities or persons educational or troubleshooting sessions via ZOOM to assist in resolving issues. **At the end of 2023, the yearly comparison report of all birthing facilities and Arkansas Children’s Hospital had an average of 86.9% which exceeds the 80% goal.**

The NBS Lab and Follow-Up managers began monitoring timeliness and NBS QI projects internally monthly in 2017. This action seems to assist birthing hospitals and submitters with early solutions before receiving the quarterly reports. The NBS lab and Follow-Up programs continue this practice currently.



**Figure 4**

Figure 4 illustrates the breakdown of the 2023 total number of confirmed cases ranging from the most to the least cases.

**Guide for abbreviations:**

- Congenital Hypothyroidism (CH)
- Hemoglobinopathies (Hgb)
- Cystic Fibrosis (CF)
- Fatty Acids (FA)
- Organic Acid (OA)
- Spinal Muscular Atrophy (SMA)
- Pompe
- Biotinidase Deficiency (BIO.)
- Mucopolysaccharidosis Type 1(MPS-1)

**Summary of 2023 confirmed cases:** There were **63** confirmed cases in 2023. Congenital Hypothyroidism (CH) with 27 confirmed cases was the leading category. Hemoglobinopathies (Hgb) had 13 cases, and Cystic Fibrosis (CF) had 8. Fatty Acids-4 cases and Organic Acid-2 cases for a total of Tandem Mass Spectrometry (MSMS) 6 cases. Spinal Muscular Atrophy (SMA) and Pompe had 2 cases. Biotinidase Deficiency and MPS-1 resulted in 1 case each.

Unlike previous years there were no cases of Congenital Adrenal Hyperplasia (CAH), Amino Acids (AA), Severe Combined Immunodeficiency (SCID), X-linked adrenoleukodystrophy (X-ALD) and Critical Congenital Heart Disease (CCHD).

\*CCHD test is completed at the birthing facility via pulse oximetry and recorded in the birthing record Electronic Registration of Arkansas Vital Event (ERAVE). NBS Follow-up program monitors/reports the positive cases.

**Breakdown for number of tests performed:**

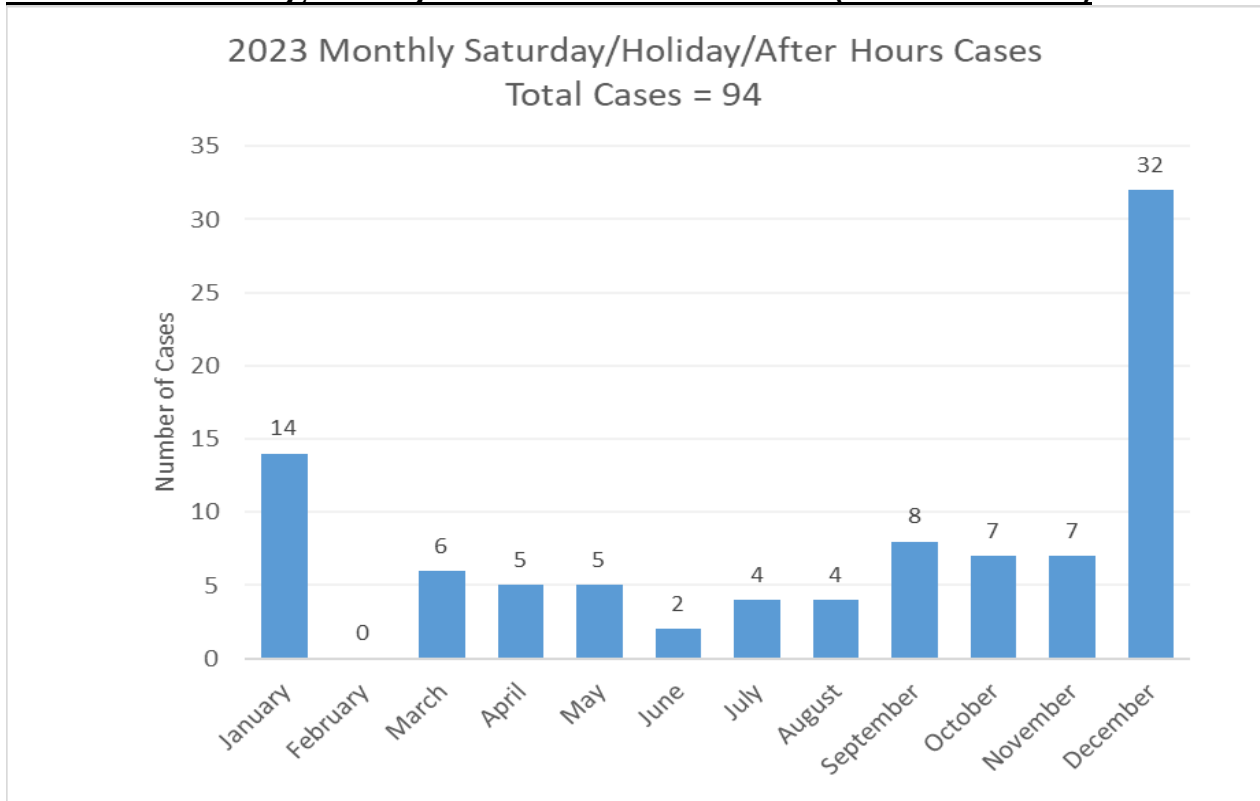
Total number of tests from Jan. 1, 2023 – Dec. 31, 2023.

The total number of tests are divided into nine (9) categories due to the addition of Mucopolysaccharidosis Type 1 (MPS-1) and Pompe to the testing panel in March 2023.

1. Total number of births by occurrence: **34,264**
2. Total number of samples received for CY2023: **33,554**
3. Total number of tests performed for CY2023: **301,986**
4. Total Number of births receiving at least one screen - with samples rejected:  
**33,554**
5. Total Number of births receiving at least one screen - without samples rejected:  
**33,450**
6. Data source year: **2023**
7. Number of presumptive positive, indeterminate, or inconclusive screens (on any screen – initial or repeat): **6002**
8. Number confirmed cases: **63**
9. Number referred for treatment: **63**

***Note: There were 866 less births in Arkansas in 2023 than in 2022. The NBS follow-up program completed 6,002 extensive case investigation until cases were resolved.***

**NBS 2023 Saturday, holidays or after hours call cases (total cases = 94)**



**Figure 5**

Figure 5 illustrates the number of 2023 critical cases reported on Saturday, holidays or after hours:

- January = 14 cases
- February = 0 cases
- March = 6 cases
- April = 5 cases
- May = 5 cases
- June = 2 cases
- July = 4 cases
- August = 4 cases
- September = 8 cases
- October = 7 cases
- November = 7 cases
- December = 32 cases

**Note:** December (32) represented the largest number of critical cases in 2023. In accordance with the 48-hour limit of closure, the Christmas holiday coverage by the lab and follow up nurses was adjusted to ensure the emergent results were followed up on in the timeframe.

### **NBS Education: on-line courses available on TRAIN:**

- AR NBS-protecting Babies Through a Simple Test: Course #1053880 (1.5 contact hrs.)
- Medical Aspects of NBS: Course #1075665 (2.0 contact hrs.)
- Introduction to CAH: Course #1076702 (2.0 contact hrs.)
- CCHD Screening in Arkansas: Course #1087269 (2.0 contact hrs.)
- ERAVE Enduring for Hospital Users: Course #1086116 (2.0 contact hrs.)-**NOTE this course is taken after the learner has trained on site with the hospital trainer).**
- ERAVE Enduring for Hospital Users (**LIVE**) **Course # 1087154** (2.0 contact hours) **NOTE this course is taken after the learner has attended a LIVE training (ZOOM or face to face) with Traci Massery, ADH ERAVE trainer.**
- Introduction to SMA (Spinal Muscular Atrophy) Enduring Course # 1091128 (2.0 contact hours).
- Introduction to X-ALD (X-Linked Adrenoleukodystrophy Disease) Enduring Course #1094925 (2.0 contact hours).
- Introduction to Galactosemia (GALT) #1076704 (1.45 contact hours)

Educational upcoming on-line courses:

- Introduction to Mucopolysaccharidosis Type 1 (MPS-1) submitted to ADH/Approved Provider Unit #AR225-7 on 12/01/23-TBA.
- Introduction to Pompe

### **Website: National Coordinating Center for the Regional Genetics Networks**

<https://nccrcg.org/act-sheet/>

<https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/ACT-Sheets-and-Algorithms.aspx>

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under Cooperative Agreement #UH9MC30770 from 6/2020- 5/2024 for \$800,000 per award year.

The ACMG ACT Sheets and their accompanying algorithms are a great resource for health care providers looking for information on genetic conditions (identified through newborn screening and beyond) to help inform clinical decision making. Developed by the American College of Medical Genetics and Genomics and the National Coordinating Center for the Regional Genetics Networks ([NCC](#)), ACMG ACT Sheets are available on the ACMG website. Given the rarity of many genetic conditions—ACT Sheets and algorithms are excellent refreshers on the conditions, diagnoses, and next steps for patients.

### **NBS ADH Pediatrician:**

Dr. Steven Schexnayder, Professor of Pediatrics, and Internal Medicine, Executive Vice Chair, Dept. of Pediatrics at UAMS College of Medicine Chief, Critical Care Medicine, at Arkansas Children's Hospital joined Arkansas Department of Health as the Medical Director of Child and Adolescent Health.

Dr. Schexnayder' s contract ended June 30, 2023. The agency is in the process of filling this vacant position by early in 2024.

**Medical Professional and Educational events presented and/or attended:**

Title:	<a href="#">Midwifery Bridge Certificate (MBC) Home Birth Services</a>
Author(s)/Organization(s):	<a href="#">Dr. Laura Hays, PhD/APRN (UAMS/ACH NBS consultant)</a>
Meeting/Conference Name:	<a href="#">2023 Midwives conference</a>
Year Presented:	<a href="#">April 13-14, 2023</a>
Media Type:	<a href="#">In-person</a>
Target Audience:	<a href="#">Certified &amp; lay midwives</a>
To obtain copies (URL or email):	<a href="https://healthy.arkansas.gov/programs-services/topics/newbornscreening">Healthy.arkansas.gov/programs-services/topics/newbornscreening</a>
Keyword	<a href="#">Midwife/home births</a>
Status	<a href="#">current</a>
Action	<a href="#">To provide NBS education &amp; open discussion with AR midwives.</a>

Title:	<a href="#">MPS-1 &amp; Pompe</a>
Author(s)/Organization(s):	<a href="#">APHL</a>
Meeting/Conference Name:	<a href="#">APHL Virtual yearly Conference focusing on MPS-1 &amp; Pompe</a>
Year Presented:	<a href="#">April 10 – 12, 2023</a>
Media Type:	<a href="#">ZOOM</a>
Target Audience:	<a href="#">NBS medical professionals (Cecilia Walker, RN &amp; Keri Lafferty, BSN/RN attended &amp; received 10 continues nursing educational credits).</a>
To obtain copies (URL or email):	
Keyword	<a href="#">Update NBS test panel disorder</a>
Status	
Action	<a href="#">To increase awareness &amp; educate about MPS-1 &amp; Pompe disorders which was added to test panel March 2023.</a>

Title:	<b>APHL Training</b>
Author(s)/Organization(s):	APHL/NewSTEPS
Meeting/Conference Name:	APHL training for NBS Follow-Up Nurse
Year Presented:	May 16 -19, 2023
Media Type:	In-person Silver Springs, Maryland
Target Audience:	Pat Purifoy, RN NBS Manager nominated Jennifer Simmons, BSN/RN to attend. APHL ONLY selected 25 to attend, 65 applied & Jennifer was one that was selected. (APHL funded travel, per diem & accommodations).
To obtain copies (URL or email):	
Keyword	Newborn screening follow-up
Status	
Action	Compare all states protocol/process, & review of educational tools

Title:	<b>CF 10-year project (2013-2023)</b>
Author(s)/Organization(s):	NewSTEPS AR Cystic Fibrosis (Ruby Moore, RN CF Lead Nurse provided AR data)
Meeting/Conference Name:	Updated/Review CF 10-year project
Year Presented:	06/29/23
Media Type:	ZOOM
Target Audience:	All states test for CF
To obtain copies (URL or email):	
Keyword	CF data
Status	
Action	Compared, discussed all states data of CF in the past 10 years

Title:	<b>SMA consultant process with AR Children's &amp; ADH/NBS</b>
Author(s)/Organization(s):	Arkansas Children's Hospital (AC)- Dr. Kapil Arya & SMA team/ ADH NBS Lead Nurse-Ruby Moore, RN & Team
Meeting/Conference Name:	Review of SMA consultant process with AR Children's & ADH/NBS
Year Presented:	07/20/23
Media Type:	In-person
Target Audience:	AC/SMA & ADH/NBS Team members
To obtain copies (URL or email):	
Keyword	SMA
Status	
Action	Meet new ACH/SMA nurse Coord. -Crystal Salser, BSN/RN, identified ACH/SMA & ADH/NBS job responsibilities.

Title:	<b>ACE Summit 2023</b>
Author(s)/Organization(s):	Dr. Chad Rogers, pediatrician- Chairperson of ACE Summit
Meeting/Conference Name:	ACE Summit 2023
Year Presented:	08/03/23 8am-4:30pm
Media Type:	In-person @ Pulaski Tech campus in NLR (Pat Purifoy, RN presented AR NBS data for NBS).
Target Audience:	Medical professional specialized in infants, children, caregivers, parents, stakeholders, religions, mental health or any pediatric practice
To obtain copies (URL or email):	
Keyword	Childrens health & family dynamics
Status	
Action	Holistic Presentation for all aspects of a child's development. Dr. Alan Mease (retired-ADH Child Health Pediatrician received the ACE Summit Outstanding Award).

Title:	<b>2023 APHL/ISNS Newborn Screening Symposium</b>
Author(s)/Organization(s):	APHL/NewSTEPs
Meeting/Conference Name:	October 15 – 19, 2023 Sacramento, CA
Year Presented:	2023
Media Type:	Conference provided <b>LIVE or Virtual</b> . NBS Lab Manager: Jomeka Edwards, MSNS & Asst. Manager Keacha Payne, BSM attended LIVE conference. Ruby Moore, RN, Keri Lafferty, BSN/RN & Courtney Waggle, RN attended Virtual conference. The NBS nurses received a total of 19 nursing continues educational credits towards licensure.
Target Audience:	Medical professionals with expertise and focus in NBS (physicians, genetic counselors, nurses)
To obtain copies (URL or email):	
Keyword	NBS, genetic testing, lab, follow-up process
Status	
Action	Empowered to confidently perform follow-up activities and will be able to describe the newborn screening system, communicate NBS results to providers and families, explain the numerous diseases on NBS panels, and participate in data analytics and new disease implementation activities. Information will be shared with staff members and management on lab and follow-up topics pertinent to the processes.

Title:	<b>UAMS Head Start/Early Head Start Health Service &amp; Education Advisory Committee</b>
Author(s)/Organization(s):	UAMS Early Head Start
Meeting/Conference Name:	The following NBS nurses were asked to service on the Advisory Board: Tanja Brown, LPN II-Asst. NBS Nurse Coord. /Outreach Pat Purifoy, RN NBS Manager
Year Presented:	Jan 2023 & July 2023
Media Type:	Quarterly meeting
Target Audience:	Program professionals involved in Head start programs (Medical, Dental, Nutrition, Disabilities, Mental health & Education)
To obtain copies (URL or email):	
Keyword	Head Start

Status	Medical, Dental, Nutrition, Disabilities, Mental health & Education
Action	Provide resources for families

**Newborn Screening Workgroup (NBS WG):**

The NBS WG schedules quarterly (Virtual/ZOOM) meetings on the 4<sup>th</sup> Tuesday of the first month of the quarter. The meeting dates were: 01/24/23, 04/25/23, 07/25/23 and 10/24/23.

Chairperson

- Barbara Holcombe, BSN/RN, ADH/ACH NBS Liaison Nurse retired September 2023. In October 2023, Dominique DuBois, RN, CPM, APRN moved into the Chairperson from Co-Chairperson.

Co-Chairperson

- Dominique DuBois, RN, CPM, APRN served in this position until October 2023. In the 2024 NBS WG January meeting, a candidate for this position will be nominated and voted on.

Purpose:

- Provides an opportunity for the AR Dept. of Health/NBS lab, Follow-up program and AR Children’s Hospital NBS staff and medical consultants to discuss, update, educate and share information that affects outcomes of the newborn screening process.

**AR Genetic Health Committee (AGHC):**

The AGHC meet quarterly (Virtual/Zoom) on the 2<sup>nd</sup> Thursday of the first month of the quarter. The meeting dates were: 01/12/23, 04/13/23, 07/13/23 and 10/26/23- cancelled.

Chairperson

- Dr. Laura Hays, PhD/APRN.

Co-Chairperson

- Ruby Moore, RN/AR State Genetic and ADH/NBS Nurse Coordinator.

Purpose:

- Implementation and monitoring of the Arkansas State Genetics Plan, five-year (2022 – 2026) plan of genetics services in Arkansas.

Summary of topics discussed during AGHC 2023 meetings:

- AR Public Health Continuity of Operations Plan (COOP) - recommendations and guidance to achieve uninterrupted provision of state services during operational challenges.
- Providing educational opportunities for presentations pertaining to newborn screening program.

- ACT 490 - the adding of new disorders from Recommended Uniform Screening Panel (RUSP) which will impact Arkansas NBS program.
- Development of AGHC Website – this website link would provide helpful information to stakeholders concerning newborn screening with several components involved such as Arkansas Department of Health (ADH) activities, University of AR for Medical Sciences (UAMS) activities, National level items, capability to translate website into Spanish, links to other websites and the ability to continuously update website.

**Newborn Screening Program 2023 update including Plans for 2024:**

In April of 2023, ACT 490/HB1102 was voted into law by the State of Arkansas Legislative body.

**Act 490 (Summary) HB1102**

An act to create the universal newborn screening act; to ensure that newborns are screened for conditions recommended by the United States Department of Health and Human Services; and for other purposes:

All newborn infants shall be tested for core medical conditions as listed in the recommended uniform screening panel recommended by the United States Secretary of Health and Human Services, in order to provide appropriate newborn screening guidelines to protect the health and welfare of newborns.

- For each core medical condition added to the list of recommended uniform screening panel by the United States Secretary of Health and Human Services, the Department of Health or its designee, in accordance with §20-15-304, shall begin testing for the newly added core medical conditions upon approval of the appropriation of funding by the General Assembly or funding under §20-15-304, if required, or within thirty-six (36) months upon introduction to the recommended uniform screening panel recommended by the United States Secretary of Health and Human Services.

In July 2023, Arkansas State Board of Health: Rules Pertaining To Testing Of Newborn Infants was approved and began the process to obtain agency signature to implement in early 2024.

**Note:** The below statement is in compliant to ACT 490.

**Laboratory Analysis**

1. The Department shall be responsible for provision of forms and instructions for the blood specimen collection; processing and recording of the specimen received; analysis of specimen; determination of abnormal results; and reporting of lab results within a time period which would allow preventive medical intervention. Testing for core medical conditions newly added to the RUSP shall begin within thirty-six (36) months upon introduction to the RUSP.

Website to view updated document is located at:

[https://www.healthy.arkansas.gov/images/uploads/rules/Newborn\\_Screening\\_Final\\_Rule.pdf](https://www.healthy.arkansas.gov/images/uploads/rules/Newborn_Screening_Final_Rule.pdf)

The Newborn Screening Laboratory will start the process of identifying needs and activities to incorporate the two most recent core disorders added to the RUSP for dried blood spot screening. Those two disorders are Mucopolysaccharidosis Type II (MPS-2) and Guanidinoacetate methyltransferase deficiency (GAMT).

In addition, the NBS lab has plans to implement a statewide data entry project that will allow submitters to send patient demographics and receive test results electronically through the StarLIMS information system. StarLIMS is a Web-based-off-the-shelf Laboratory Information Management System which is a comprehensive solution that offers the functionality to support operational excellence. StarLIMS lets you seamlessly collect, share, and analyze lab data to facilitate improved overall patient care.

The Arkansas NBS program prides itself in being consistent to meet the established mission and vision.

- Mission: To protect and improve the health and well-being of all infants, children, and youth in Arkansas.
- Vision: Infants, children and youth in Arkansas are healthy and reach full developmental and academic *potential*.

The Arkansas NBS program is constantly working to better serve Arkansas families and health professionals.

## Arkansas Department of Health Newborn Screening Program

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