



Newborn Screening Section CY2021 Annual Report

Submitted by collaborated team of:

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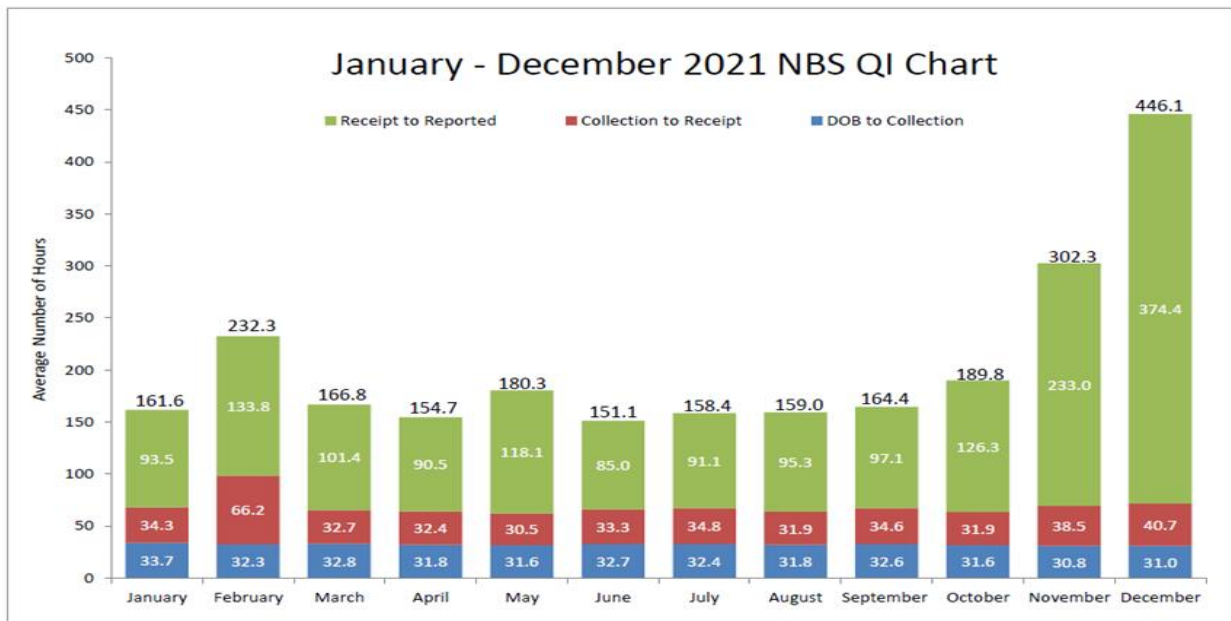
Introduction

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 34 core disorders. In 2021, ninety-seven-point seven percent (97.7%) of the approximately 35,070 babies born in Arkansas were screened for genetic disorders.

Reports

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

Newborn Screening Quality Improvement (QI) Graph: Three key data points are monitored monthly: time of birth to time of collection, time of collection to time of receipt in the lab and time of receipt to time of reporting results. Data is provided to ADH Health Statistics representative for compilation. The Public Health Lab and Newborn Screening managers work diligently and continue to monitor monthly timeliness of data at three key points. Time is measured in hours and a goal of less than 168 hours (7 days) from birth to reporting of test results was set. **The 2021 report indicated the average was 205.5 hours.**



Notes:

1. Data Source: 11November2021-QI data.xlsx & 12December2021-QI data.xlsx
2. Removed 18 records on missing DOB or Date of Collection from 11November2021-QI data.xlsx & 12December2021-QI data.xlsx
3. Removed 12 bad records from 11November2021-QI data.xlsx & 12December2021-QI data.xlsx
4. Report contains 5,714 records for November through December 2021
5. Average Hours includes holidays and weekend
6. Number at top of bar equal total hours of each month

Figure 1

As illustrated in Figure 1: The NBS QI 2021 report indicated the average number of hours that elapsed from birth to reporting of NBS results was 205.5 hours. The factors that contributed to the increase NBS QI 2021 hours was due to a statewide shutdown due to inclement weather during one week in February and lab equipment issues. The Newborn Screening contingency plan was developed after the weather got back to normal. It has since been implemented but had no effect on the February 2021 weather situation. During May, October, November, and December the lab experienced equipment issues involving MSMS (Tandem Mass Spectrometry (MSMS): Fatty, Organic, Amino Acids, X-ALD and the HPLC (Hemoglobin) disorders. PerkinElmer, the vendor for the Arkansas Public Health Lab worked with the PHL Director, Section Chief, and NBS Lab Manager to resolve these issues. Arkansas and Pennsylvania State Labs worked in partnership to complete testing of these disorders. During this time frame, the Arkansas NBS Lab and Follow-up managers developed a protocol/process with Pennsylvania state lab to receive critical results in a timely manner.

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 2021 Comparison Report:

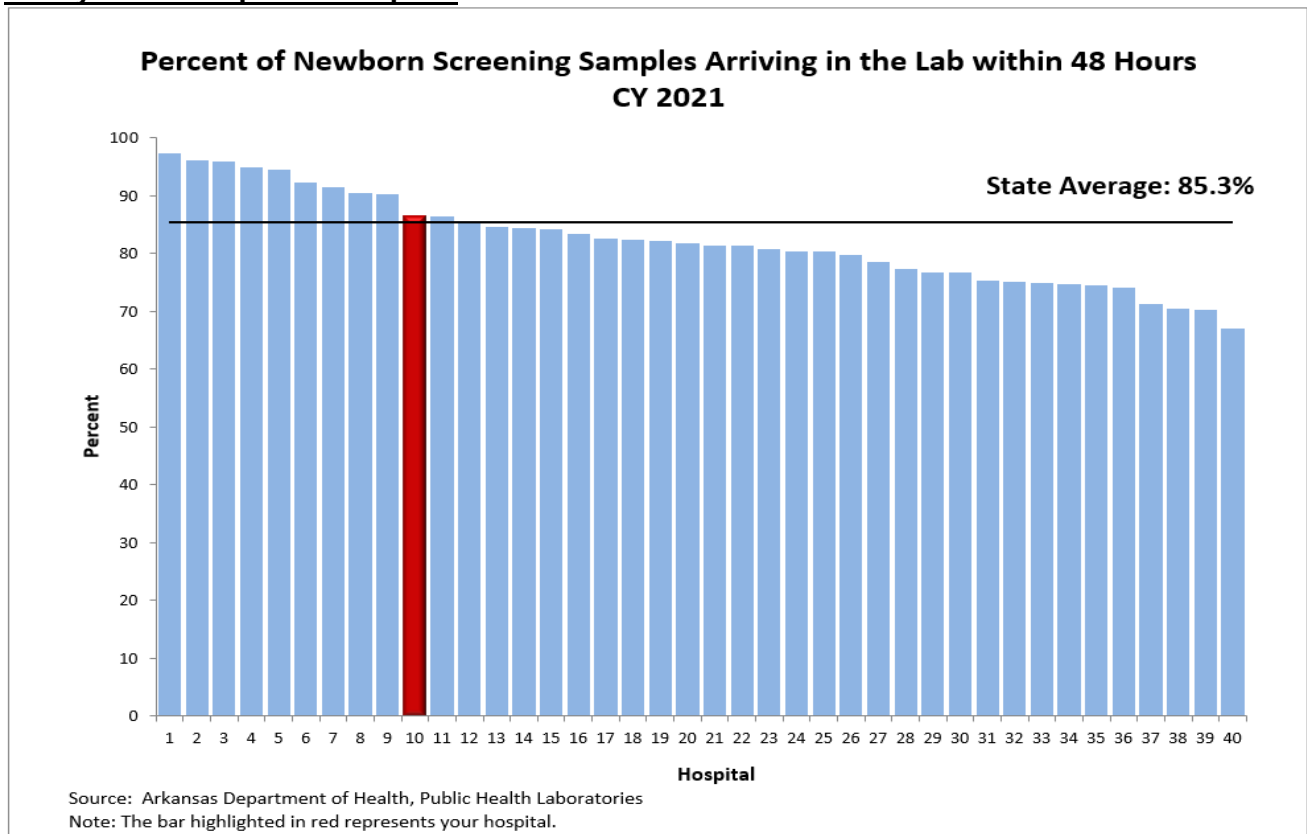


Figure 2

As illustrated in Figure 2: A yearly comparison graph in addition to the quarterly reports, representing all 40 facilities 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.

Each birthing facility 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 facility 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. These sessions are conducted via ZOOM which has the capability to involve responsible parties at the birthing facility along with ADH NBS Follow-up Manager and AR State Genetics Coordinator. **At the end of 2021, a yearly comparison report of all birthing facilities had an average of 85.3%.**

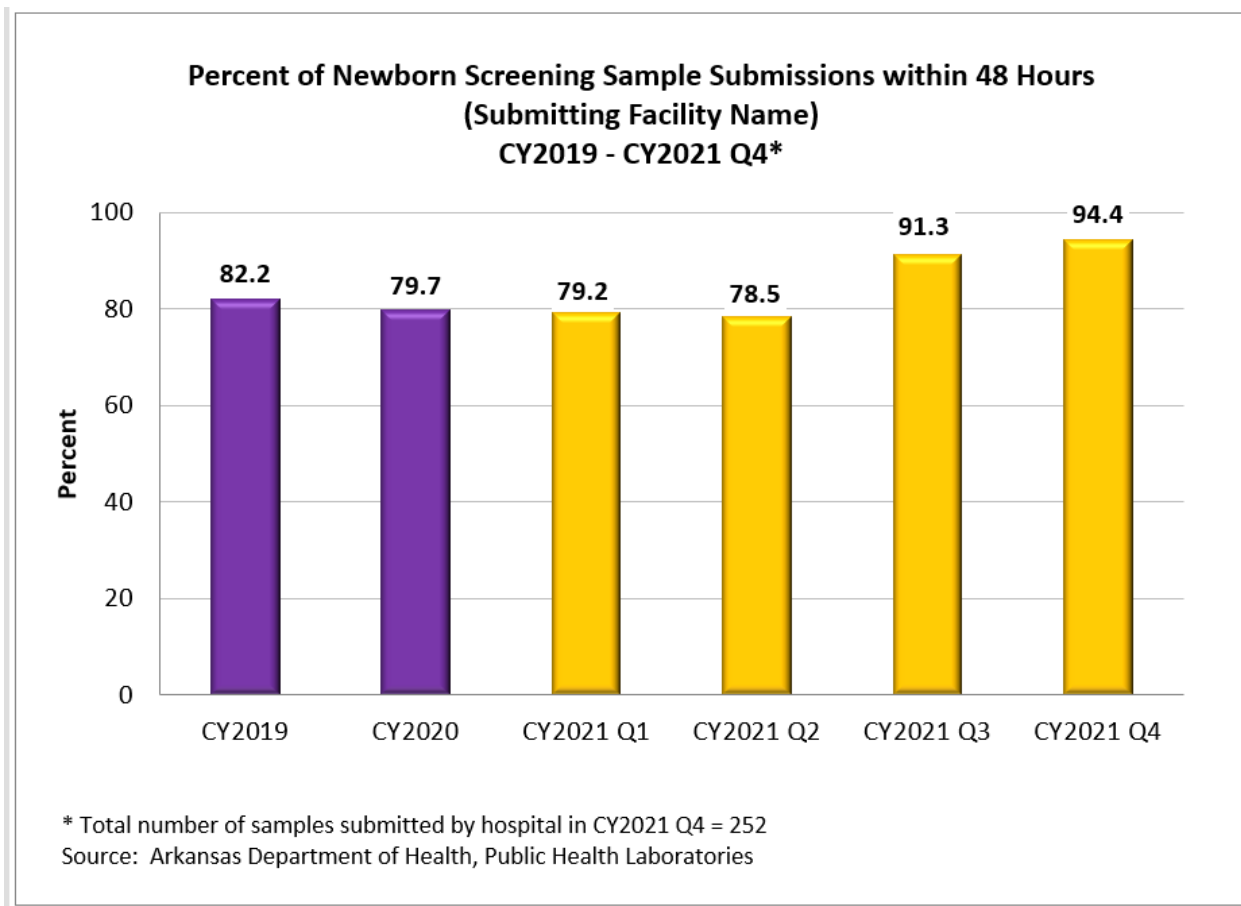


Figure 3

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 facility and Arkansas Children's Hospital (ACH).

Newborn Screening 2021 confirmed cases

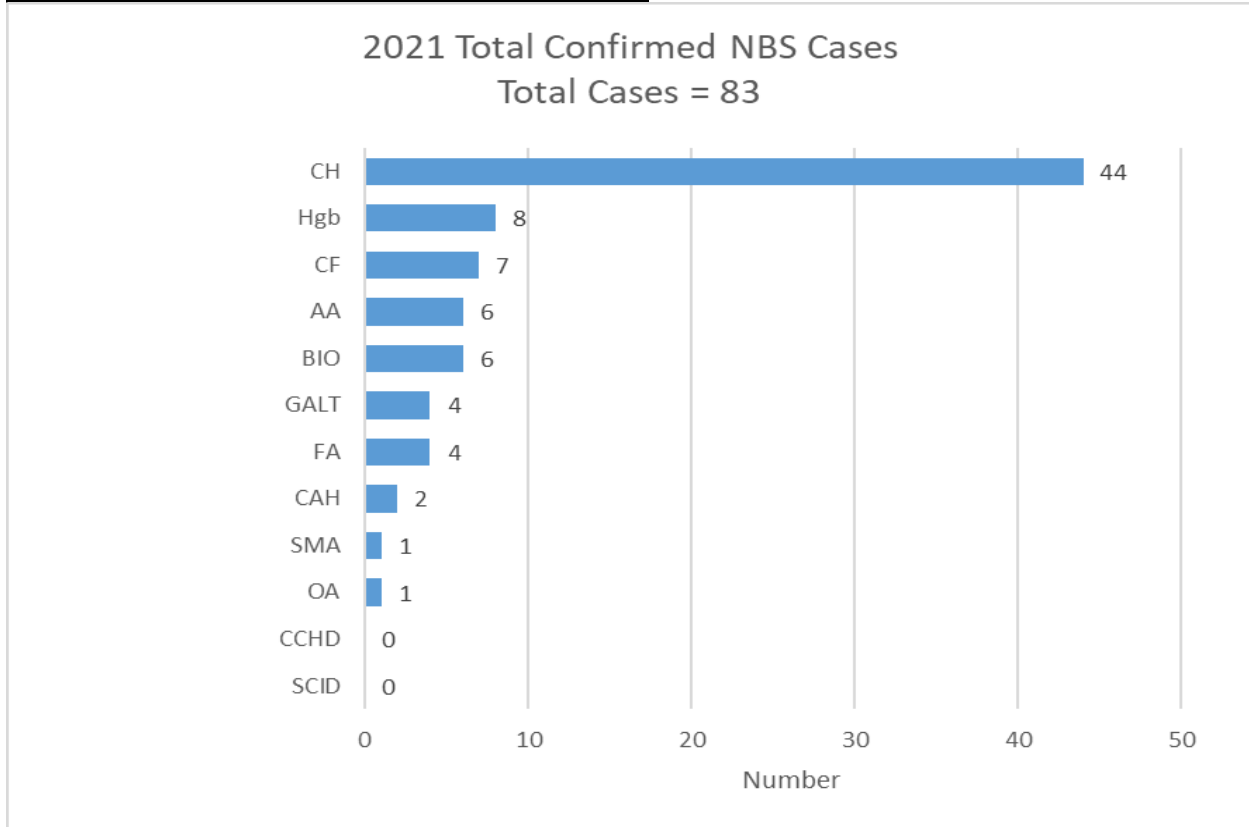


Figure 4

In Figure 4 illustrates the breakdown of the 2021 (**83**) total number of confirmed cases ranging from the most to the least cases. Congenital Hypothyroidism (44 cases) was the leading category with Severe Combined Immune Deficiency (SCID) and Spinal Muscular Atrophy (SMA) being the least at 1 case for each category.

2021 Disorders/Categories

Biotinidase Deficiency = 6

Congenital Adrenal Hyperplasia = 2

Congenital Hypothyroidism = 44

Cystic Fibrosis = 7

Galactosemia (classical) = 4

Hemoglobinopathies = 8 Sickle cell

SCID = 0

SMA = 1

Fatty Acid Oxidation = 4 (2-MCAD & 2 VLCAD)

Amino Acid = 6 (4 PKU & 2 CIT)

Organic Acid = 1(1 GA1) Disorders

Critical Congenital Heart Disease (CCHD) = 0

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

X-linked adrenoleukodystrophy (X-ALD) was added to the testing panel in November 2021. In 2021, there were no cases confirmed.

X-ALD is a genetic disease that affects the nervous system and the adrenal glands (small glands located on top of each kidney). People with this disease often have progressive loss of the fatty covering (myelin) that surrounds the nerves in the brain and spinal cord.

Breakdown for number of tests performed:

Total number of tests from Jan. 1, 2021 – Dec. 31, 2021, the total number of tests are nine (9) categories due to the addition of Spinal Muscular Atrophy (SMA) to the testing panel.

1. Total number of births by occurrence: **35,070**
2. Total number of samples received for CY2021: **38,905**
3. Total number of tests performed for CY2021: **380,287**
4. Total Number of births receiving at least one screen - with samples rejected: **34,358**
5. Total Number of births receiving at least one screen - without samples rejected: **34,270**
6. Data source year: **2021**
7. Number of presumptive positive, indeterminate, or inconclusive screens (on any screen – initial or repeat): **8,931**
8. Number confirmed cases: **83**
9. Number referred for treatment: **83**

Note: 8,931 newborn screens involved extensive follow-up by the NBS nurses until cases were resolved.

NBS 2021 Saturday call cases:

October 2014 the ADH Lab began Saturday hours to prevent delays in testing follow-up exceeding 48 hours from collection. In February 2021, NBS Follow-Up implementing after hours call coverage. A total of **75** critical cases were identified during the Saturday call in 2021.

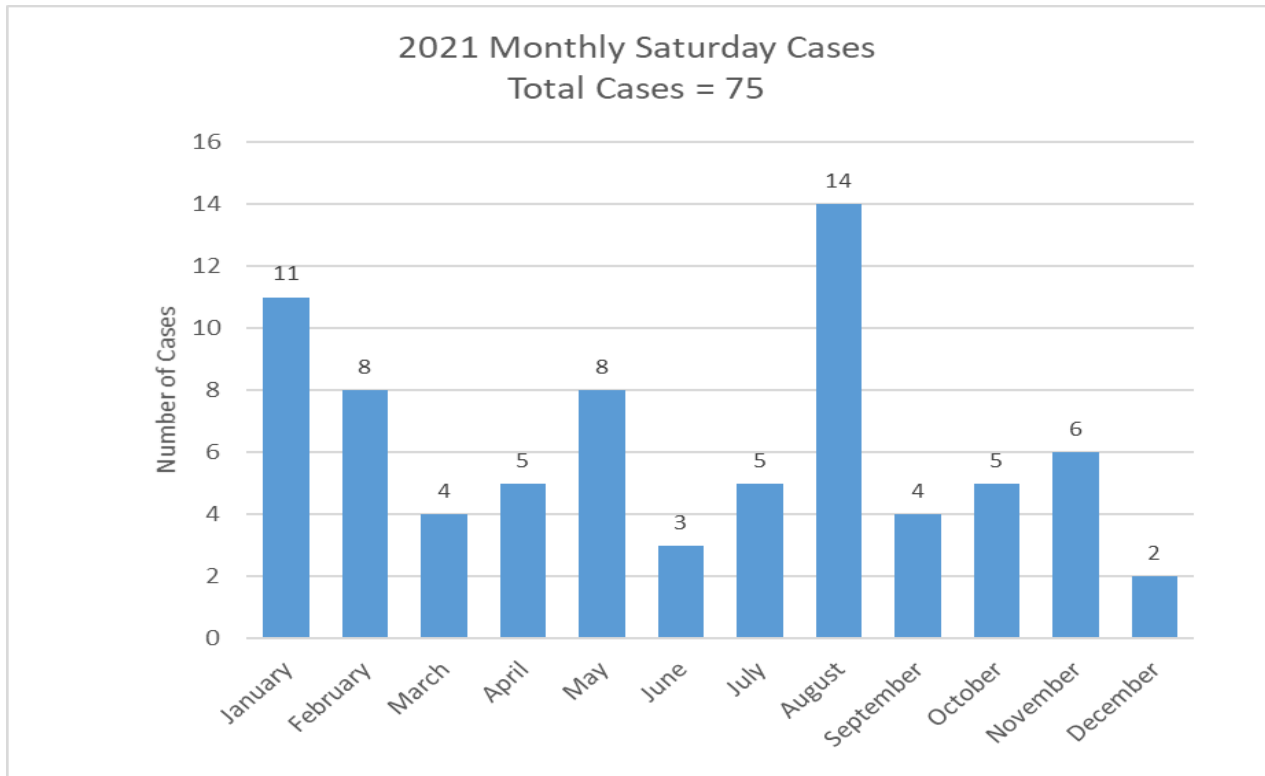


Figure 5

Figure 5 illustrates the number of critical cases reported on Saturday in 2021: August 2021 presented the largest number of critical cases (14) identified on Saturdays.

- January = 11 cases
- February = 8 cases
- March = 4 cases
- April = 5 cases
- May = 8 cases
- June = 3 cases
- July = 5 cases
- August = 14 cases
- September = 4 cases
- October = 5 cases
- November = 6 cases
- December = 2 cases

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 hrs.)
- Introduction to X-ALD (X-Linked Adrenoleukodystrophy Disease) Enduring Course #1094925 (2.0 contact hours. Course launched in TRAIN 03/15/21)

Educational upcoming on-line courses:

- Introduction to Galactosemia (GALT)

NBS ADH Pediatrician:

In March 2021; 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.

Medical Professional and Educational events attended:

- April 14 and 15, 2021

St. Jude Hospital in Memphis, TN provided a virtual conference on Sickle Cell Disease. Three NBS Nurse Coordinators attended the event. Each nurse earned a total of 6.25 professional nursing contact hours.

- August 25-26, 2021

The APHL NewSTEPS New Disorders and Short-Term Follow-up Virtual Meeting provided training and ideas to support NBS programs in improving efficiency, increasing knowledge, and expanding capacity.

- September 23, 2021

The ACH NBS Team: Dr. Brad Schaefer, NBS Medical Director, Barbara Holcombe, BSN/RN, NBS Liaison Nurse and Jo Ann Bolick, APRN NBS Program Director provided a three-hour NBS Training Session virtual learning course on NBS. All ADH NBS nurses attended the event.

- September 29, 2021

The American National Cystic Fibrosis (CF) Organization had scheduled a one-day LIVE conference in San Antonio, TX but due to COVID-19 the arrangements to change the conference to virtual meeting was unsuccessful. The meeting was cancelled.

- October 5 -14, 2021 (Tuesday, Wednesday, and Thursday of both weeks)

The Association of Public Health Laboratories (APHL) NBS 2021 National Symposium was a virtual educational session. The sessions began at 11am on those days. Four NBS Follow-Up nurses were able to attend the virtual sessions and was awarded professional nursing contact hours.

- October 25, 2021

The NewSTEPS (national newborn screening resource center) had a virtual meeting: “Updated NewSTEPS Repository Data Cystic Fibrous (CF) Cases. This meeting involved NBS follow-up nurses from several states discussing current screening status of new and prospective of CF, review tiered testing, evolving therapies, strategies, identify tools to build the workforce and providing educational tools for parents of children with CF.

Educational Material:

<https://www.youtube.com/channel/UC1qysK3DUhkP8iJIQrVdRuA>

In 2021, Heartland Genetic Collaborative’s Spanish Newborn Screening Video was developed for educational purposes. The goal is to provide parents, medical professionals, and all stakeholders with outreach material in Spanish about the Arkansas Newborn Screening Program. This material is available on the ADH/NBS website, or by request and will be utilized in promotion setting like conferences, seminars, meetings, and health fairs. <https://www.healthy.arkansas.gov/programs-services/topics/newborn-screening>

AR Genetic Health Committee (AGHC):

Due to COVID-19, Dr. Brad Schaefer (NBS Medical Director) scheduled ZOOM meetings. The goal of this committee was to update the Arkansas State Genetics Plan (2021-2025). The Arkansas State Genetics Plan is a five-year plan for implementation and monitoring of genetics services in Arkansas. In October 2021, the AGHC five-year strategic plan dates were changed to 2022-2026. The draft was provided to all committee members for review and editing. The updated plan will be presented at the 2022 AGHC January meeting. The final draft will be reviewed and approved to implement.

Newborn Screening Plans for 2022:

In November 2021, X-ALD disorder was added to the blood testing panel giving us a total of 30+ disorders. Additionally, Critical Congenital Heart Disease (CCHD) and hearing screening are completed at the birthing facility. The plans to increase the blood testing panel within 2021 – 2022 include adding the following disorders: Glycogen Storage Disease Type II (Pompe), and Mucopolysaccharidosis Type 1 (MPS-1).

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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