Arizona Department of Health Services

Fall/Winter 2015

Newborn Screening Connection

The Office of Newborn Screening Newsletter

AzEHDI Pages 4-5

Welcome to the Fall/Winter Edition

Arizona has joined 36 other states in implementing Critical Congenital Heart Defect (CCHD) screening for newborns. Babies born in our state now have access to non-invasive pulse oximetry screening, a point of care test done at about 24 hours of life, before discharge from the hospital or birth setting. Read a statement related to guidelines and recommendations from the Assistant Bureau Chief of the Office of Newborn Screening by clicking on the link or visiting the website at www.aznewborn.com

A Message from the Medical Director

Pulse Oximetry Screening Now Required

Lisa Villarroel, MD MPH

As of July 1, 2015, Arizona implemented critical congenital heart defect screening (CCHD) for all newborns across the state (Administrative Code Title 9, 13.2). **Newborns are now required to be screened with pulse oximetry**, in addition to tests for bloodspot and hearing disorders.

This is the first addition to the Arizona screening panel since 2007. New instructions for data collection and submission are posted on the website www.aznewborn.com, which consists of a minor change on the bloodspot cards. CCHD screening should not delay the submission of bloodspot cards.

Most Arizona providers have already been screening for critical congenital heart defects in newborns. The difference is that now both screening and reporting of results to public health is required. Failed reporting forms and other information can be found on the website or by emailing nbseducation@azdhs.gov.

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Arizona Department of Health Services Office of Newborn Screening · 250 N 17th Ave, 1st Fl · Phoenix, AZ 85007 · Phone: 602-364-1409 · Fax: 602-364-1495 · nbseducation@azdhs.gov

CCHD

Critical Congenital Heart Defect Screening

ACT Sheet

Provider Resources

Failed Form

American College of Medical Genetics **ACT SHEET**

Newborn Screening ACT Sheet
[Hypoxemia]
Critical Congenital Heart Disease

Differential Diagnosis: Critical congenital heart disease (CCHD); congenital heart disease (CHD); patent ductus arteriosus (PDA); persistent pulmonary hypertension of the newborn (PPHN); pulmonary and infectious diseases; metabolic, endocrine, and hemoglobin disorders.

Condition Description: Critical congenital heart disease (CCHD) is a subset of congenital heart disease (CHD), comprising about 25% of CHD cases. CCHD includes common truncus arteriosus, hypoplastic left heart syndrome, pulmonary valvular atresia, Tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great vessels, and tricuspid valvular atresia. These are ductus dependent malformations in which the children appear well but when the ductus closes the patient can deteriorate rapidly. There are other types of CCHD (such as coarctation of the aorta) that, although less likely to cause arterial desaturation, may be detected by physical examination and/or pulse oximetry. CCHD may be associated with decreased levels of oxygen in the arterial blood (hypoxemia) and may bring a significant risk of morbidity and mortality if not diagnosed soon after birth.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Inform the family of the newborn screening result.
- Evaluate the infant for cardiac and non-cardiac causes of hypoxemia.
- If non-cardiac cause for hypoxemia has been excluded, consult with a pediatric cardiologist to arrange for urgent echocardiogram.
- Transfer to appropriate referral center, if necessary.

Diagnostic Evaluation: Echocardiogram and cardiology consultation to determine cardiac anatomy and function. Noncardiac causes of hypoxemia are to be addressed by studies appropriate to such etiologies as infectious or pulmonary disease.

Clinical Considerations: CCHD comprises ductus-dependent malformations in which the newborn may appear well, but when the ductus closes the patient can deteriorate rapidly. Signs of CCHD may include visible cyanosis, tachypnea, murmur, difficulty feeding, and failure to thrive. If untreated, CCHD may lead to significant morbidity or mortality. Health care providers will want to remember that the diagnosis of CCHD, even when a specific anatomical cardiac defect is identified, necessitates that a variety of etiologies, some of them heritable or genetic, be explored. While CCHD often occurs as an isolated finding, it may occur with other anatomic features in genetic syndromes of chromosomal origin (Down syndrome, Turner syndrome and 22q11.2 deletion syndrome.) Single gene mutations, specifically affecting cardiac development or resulting in recognized syndromes (Alagille syndrome and Noonan syndrome), have been identified as causing CCHD. Copy number variation may play an as yet undefined role in CCHD.

Additional Information:

CDC Website
AHA/AAP Article

Genetics Home Reference

Referral (local, state, regional and national):

ACMG American College of Medical Genetics and Genomics

American College of Medical Genetics
https://www.acmg.net/

Arizona Department of Health Services, Office of Newborn Screening
Failed Pulse Oximetry (CCHD) Report

Complete within 30 days of a failed screen and return via fax to ADHS Office of Newborn Screening at: **602-364-1495**

Newborn Demographics

Last Name: _____ First Name: _____
Date of Birth (mm/dd/yyyy): _____ Time of Birth (military time): _____
Mother's Name: _____ Mother's DOB: _____
Birth Hospital / Facility/ or Site: _____

Pulse Oximetry Test Results

Screening Facility: _____ Infant's Medical Record Number (MRN): _____

Screening Results/Values:

First attempt
Date and Time of screen: _____ R Hand _____% R / L Foot _____%

Second attempt
Date and Time of screen: _____ R Hand _____% R / L Foot _____%

Third attempt
Date and Time of screen: _____ R Hand _____% R / L Foot _____%

Referral and Diagnosis

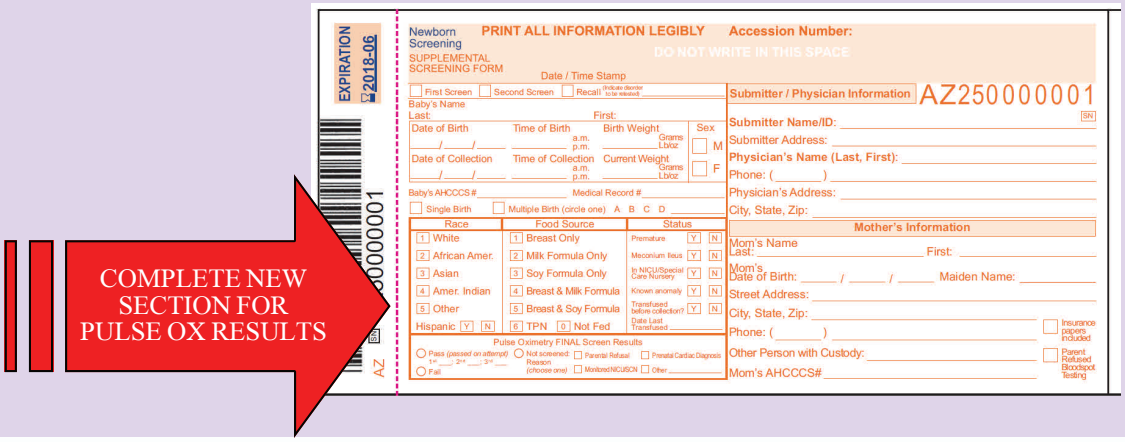
1. Was the infant referred and/or transferred to another facility? Yes No If yes, where?
2. Did the infant have a known or suspected Congenital Heart Defect prior to screening? (circle one) Yes No Unknown
   If yes, provide diagnosis and/or any pertinent information: _____
3. Does the infant have other congenital diagnoses? (circle one) Yes No Unknown
   If yes, provide diagnosis and/or any pertinent information: _____
4. Did the infant receive an echo and/or additional testing following the Pulse Ox Screen: (circle one) Yes No Unknown
   If yes, provide diagnosis and/or any pertinent information: _____
5. Final cardiac and/or additional diagnoses, if known. Or, other explanation of failed screen: _____

Name, Title, and Phone # of Person Completing Form: _____

July 2015

http://azdhs.gov/lab/aznewborn/providers/

The Redesigned Collection Kit



Parent Resources

Pulse Oximetry Screening for Critical Congenital Heart Defects

Understanding Critical Congenital Heart Defects

Congenital heart defects account for 24 % of infant deaths due to birth defects. In the United States, about 4,800 (or 11.6% per 1000) babies born every year have one of seven ***critical*** congenital heart defects (CCHDs). These seven CCHDs are:

- Hypoplastic left heart syndrome
- Pulmonary atresia (with intact septum)
- Tetralogy of Fallot
- Total anomalous pulmonary venous return
- Transportation of the great arteries
- Tricuspid atresia
- Truncus arteriosus

1 in 110

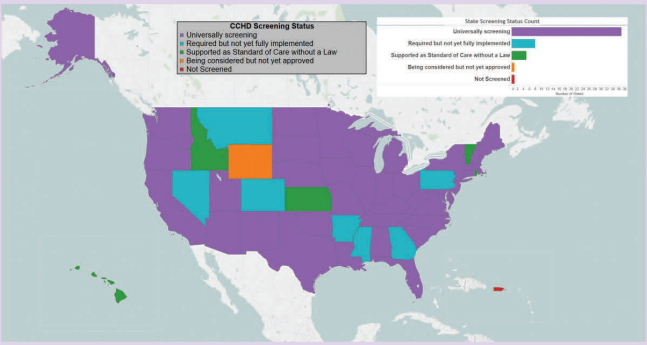
Babies with one of these CCHDs are at significant risk for death or disability if their CCHD is not diagnosed and treated soon after birth.

Download the flyer by visiting:

http://www.cdc.gov/ncbddd/heartdefects/cchd-facts.html

Centers for Disease Control and Prevention
CDC 24/7: Saving Lives. Protecting People™

CCHD Screening Map
July 1, 2015



https://www.newsteps.org/cchd

NewSTEPs
A Program of The Association of Public Health Laboratories®

Mended Little Hearts of Phoenix
A program of The Mended Hearts, Inc.

http://www.mendedlittleheartsofphx.org/
A support program for parents and caregivers of children with Congenital Heart Defects

march of dimes

www.marchofdimes.org/baby/congenital-heart-defects.aspx

http://www.babysfirsttest.org/newborn-screening/conditions/critical-congenital-heart-disease-cchd#sources

baby's first test

AzEHDI

Early Hearing Detection and Intervention

EHDI Coordinator Corner

Lylis Olsen, MPH, C-AAA

New Laws Means New Rules

The statute changes for hearing, under Newborn Screening, were only a few words that included hearing along with the other conditions that receive mandatory newborn screening. Since all hospitals were already screening, the impact on them was minimal. Many midwives were already screening as well so there are a small number of births each year that must now be screened that weren't previously. Rule changes always follow statute (law) and serve to explain how the new statute is to be implemented. Some of the requirements in the rules include the provision of educational materials to the parent before the hearing screen, a second screen if an atypical result was obtained and referral for diagnostic evaluation for those infants who did not pass a two stage screen or after the first screen for those who were in the Neonatal Intensive Care Unit for more than five days. Also new is the requirement to provide the date of the appointment and who the provider will be for any subsequent testing if known. The audiologist is required to submit all of their results to the Department of Health including the narrative report. The change in the rules will help the Department to provide the follow up needed to ensure that all newborns and infants who are Deaf or Hard of Hearing are identified early. The rules draft as submitted in May 2015 can be found here.

http://azdhs.gov/lab/aznewborn/azehdi/index.php?pg=resources.

Mark Your Calendars! Upcoming Event

Save the Date for NAD

The National Association for the Deaf (NAD) is a civil rights organization of, by and for deaf and hard of hearing individuals in the United States. Their Biennial conference will be held in **Phoenix July 5-9, 2016.** The meeting includes activities, workshops and a council meeting. You can find out more information under *Events* on their website

EHDI Chapter Champion

Bradley Golner, M.D.

EHDI and Arizona PCPs: How do Arizona's primary care physicians feel about infant and childhood hearing loss? EHDI programs across the country and here in Arizona are present to help educate and guide both physicians and parents involved with infants and children with hearing loss.

In 2012, a survey developed by NCHAM was given to Arizona physicians regarding newborn hearing screening. One of the findings that stood out was that 50% of PCPs do not feel their training was adequate to meet the needs of infants with permanent hearing loss. In addition, less than 50% of PCPs felt confident talking to parents about bilateral moderate-profound hearing loss consequences. There are many other components of hearing loss included in the survey but these results are just a small portion that reminds us that we at EHDI still have a lot of work to accomplish.

Arizona Department of Health Services (ADHS) is an excellent resource for parents and providers (http://azdhs.gov/lab/aznewborn/azehdi/index.php?pg=providers) and it is our continuous goal to not only educate our PCPs, but also to identify all newborns with congenital or late-onset hearing loss.

Case Management Coordinator

Fran Altmaier, BSW

Welcome to our newest hearing follow up team member, **Blanca Ramirez.** Blanca recently joined Gidget and Amie on the hearing team and will be providing follow up to the babies that spend > 5 days in the NICU. She brings a wealth of experience in early childhood development and is bilingual too! Blanca has a BSW and a M. Ed., in Special Education of Infants & Young Children.

Also new to our team for the Fall '15 and Spring '16 semesters, we are fortunate to have 2 BSW interns from Arizona State University's, College of Public Service & Community Solutions, School of Social Work. Jodi Goguen and Natasha Saragosa will be helping the hearing and bloodspot teams with follow up and the education team with outreach and family resource development.

HiTrack Tip

Melanie , AuD, CCC-A, Consulting Audiologist

When to add physicians to demographics

The primary care physician for the baby (not the hospital attending) is required to be entered into the demographics page on babies who have referred (inpatient or outpatient), were missed, refused or whose test results were inconclusive. This is important because the testing information is sent on to the physician and they will help remind the parents to return to your facility or proceed on to diagnostic audiology to complete the testing for the child.

You can select the name of the physician from the drop down menu on the demographics page. If the PCP is unknown, you may choose that option as long as it is accompanied by a note in the "Notes" section. If the PCP is known but not on the list, you can ask the State to add that individual by sending a request to hearing@azdhs.gov.

Your AzEHDI Team

Fran Altmaier	Case Management Coordinator	fran.altmaier@azdhs.gov	602-364-1814
Gidget Carle	Hearing Team Lead	Gidget.carle@azdhs.gov	602-364-1405
Kostas Petritis	Data Team Manager	Kostas.Petritis@azdhs.gov	602-364-1475
Sondi Aponte	QI and Education Manager	Sondi.aponte@azdhs.gov	602-364-1642
Lylis Olsen	AzEHDI State Coordinator	EHDI@earfoundationaz.com	602-753-5273
Melanie Moore	Consulting audiologist	audiology@earfoundationaz.com	602-753-5274
NCHAM Help Desk	Technical Support for HiTrack	ncham.helpdesk@usu.edu	435-797-3584
Brad Golner, MD	AAP Chapter Champion	hearing@azdhs.gov	602-364-1409
Najwa Ghattas	Exec. Director, AZ Hands & Voices	najwa1020@hotmail.com	602-828-1020

Meet Melanie Moore, Au.D., CCC-A, FAAA

Melanie holds a B.S. in Speech and Hearing Sciences and an M.S. in Audiology from the University of Arizona in 2000 and 2002, respectively. She completed her Clinical Fellowship Year at East Valley Hearing Center and continued on at that location for just under ten years. During that time, she earned her Transitional Doctorate of Audiology (Au.D.) from the Arizona School of Health Sciences (ASHS) in 2008.

Dr. Moore is a long-standing member of the American Speech-Language-Hearing Association (ASHA) as well as a Fellow of the American Academy of Audiology (AAA) and is currently serving her second term as the Audiology Representative for the Arizona Speech-Language-Hearing Association (ArSHA) while managing an informal audiology list serve for providers in Arizona.

Melanie also enjoys volunteering her time as a supervisor for the Heathy Hearing program within the Arizona branch of the Special Olympics. She also looks forward to bringing her passion for audiology and patient care to her new role with the Office of Newborn Screening as a consulting audiologist.

Process Improvement

When Fast isn't Fast enough; How to Expedite a Newborn Screen

Newborn screening turn-around times are intended to be fast and efficient to quickly detect babies that require further attention. Sometimes though, *fast isn't fast enough*. Maybe the baby was born with physical characteristics that raise suspicion, or perhaps they arrive symptomatic in an ER or physician office, or possibly they have had an abnormal prior newborn screen with a "critical" value. Regardless of the reason, it is important to know that properly marking the card and envelope can lead to even faster results.

First, it is important to mark the collection card and the envelope "**STAT**". Then, indicate which disorder is suspected (if known). Next, call the Office of Newborn Screening at **602-364-1409** to alert the lab and follow up teams that the sample is on the way. In extremely urgent situations, the Office of Newborn Screening can assist with getting the expedited sample to the lab.

Stay Connected

A Must Read

THIRTY MILLION WORDS
BUILDING A CHILD'S BRAIN
TUNE IN · TALK MORE · TAKE TURNS
DANA SUSKIND, MD

To learn more about what's happening at the AZ Department of Health, visit Director Cara Christ, MD on her weekly blog
www.feeds.feedburner.com/ADHSdirectorsblog
-or-
Read it here:
www.directorsblog.health.azdhs.gov

Join our constant contact list and receive updates and news delivered right to your inbox!

Simply do the following to add yourself to the list:

- Visit this page: http://visitor.constantcontact.com/email.jsp?m=1101362445484
- Click the box next to the topics you would like to receive information on from ADHS
- Be sure to choose *Office of Newborn Screening* and *AZEHDI: Early Hearing Detection and Intervention*
- Fill out the contact info in the column to the right so we can include you in specified correspondence as well
- Forward and share with a friend!

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