

Newborn Screening

Train the Trainer



**Oklahoma State Department of Health
Newborn Screening Program**

Phone: 1-405-271-6617

Toll Free: 1-800-766-2223

Fax: 1-405-271-4892

NewbornScreen@health.ok.gov



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Newborn Bloodspot Screening

▶ Purpose

- Newborn screening (NBS) is the practice of testing **every** newborn for harmful or potentially fatal disorders that are not otherwise apparent at birth.
- **Early detection** and **prompt treatment** can make the difference between healthy development or lifelong impairment and possible death.



Screening vs. Diagnostic

- ▶ The newborn screen is just that... a *screen*.
 - Screening results, by themselves, **cannot** determine the presence or absence of a disorder.
- ▶ Diagnostic results refer to the combination of signs, symptoms, and test results that allows the doctor to **confirm** the diagnosis of a respective disease.



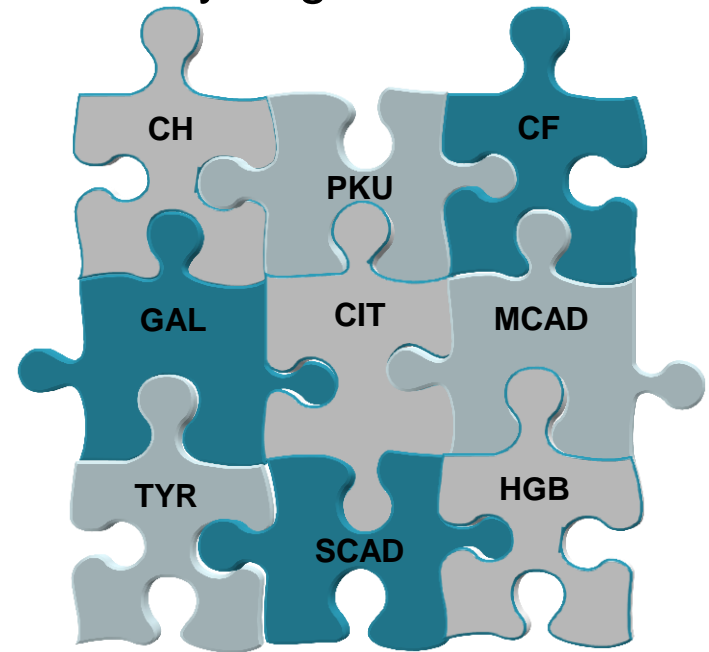
Who Decides?

▶ By law:

- The Oklahoma Board of Health decides which disorders are screened for on the panel.
- The proposed disorders must be approved by Legislation before they are added for screening.

▶ Oklahoma currently screens for more than **50** possible hidden disorders.

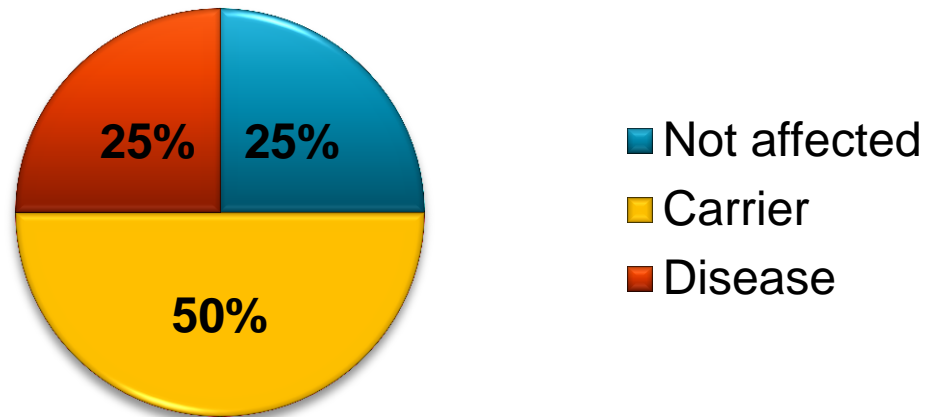
▶ Oklahoma will continue to expand the NBS panel.



Autosomal Recessive

- ▶ Most NBS disorders are autosomal recessive with the exception of:
 - Congenital Hypothyroidism (CH)
 - Some forms of Severe Combined Immunodeficiency (SCID)
- ▶ Usually no prior family history
- ▶ Risk for each pregnancy if both parents are a carrier of a disorder:

Possible Outcomes for Offspring of Parental Disease Carriers



Education for Parents

Aware

of the importance, need,
and benefits of screening

Understand

what the NBS is and that
additional follow-up may be
required

Involved

by asking baby's doctor for
NBS results



Parent Education

- ▶ NBS is collected on **every** baby born in Oklahoma.
- ▶ Importance of **correct** contact info & PCP for follow-up.
 - *No news is not good news!* Update NBS Program with changes in home address and/or PCP.
- ▶ Review hidden disorders, using NBS pamphlet as a guide.
- ▶ Specimens are kept by the OSDH lab for 42 days before being destroyed.
- ▶ Explain that most affected newborns do not exhibit signs & symptoms early on.
- ▶ Prompt identification & treatment of disorders is critical.



Parent Education

- ▶ Instruct parents to ask for screen results on first visit to PCP.
- ▶ Tell parents to bring the **Blue** or **Pink** slip to their baby's first doctor's visit.

Oklahoma State Department of Health
PUBLIC HEALTH PROTECTING YOUR BABY

OKLAHOMA
NEWBORN
METABOLIC
DISORDER
SCREENING
PROGRAM

Baby's Last Name: _____ Baby's First Name: _____

THE NEWBORN METABOLIC DISORDER SCREENING TEST

A special blood test has been done to protect your baby from hidden disease. The test screens for congenital hypothyroidism, galactosemia, phenylketonuria (PKU) and sickle cell disease. These disorders are harmful if treatment is not started within the first month of life (each disorder is explained on the back of this sheet).

WILL FURTHER TESTING BE REQUIRED?

TIME OF TESTING
 Under 24 hours of age Over 24 hours of age

ATTENTION PROVIDER
DETACH AND GIVE TO PARENT OR GUARDIAN

ASK YOUR BABY'S DOCTOR FOR THE TEST RESULTS

Please take this form with you to your baby's first doctor visit and ask for test results. If your baby's doctor does not have the test results and you have not been notified by mail, please call the Oklahoma State Department of Health when your baby is three weeks of age at (405) 271-6617 or 1-800-766-2223.

Ser. No. 711571

DETACH AND GIVE TO PARENT OR GUARDIAN
NEWBORN METABOLIC DISORDER SCREENING

Parent Education

- ▶ Review reasons why a repeat screen may be needed:
 - **Unsatisfactory Specimen**
 - **Out-of-range results**
 - Possible disorder identified
 - Hgb Trait condition
 - **Specimens collected less than 24 hours**
 - Risk for missing some disorders
 - **Premature or Sick Infants** (TPN & antibiotics could affect results)
 - **Not collected prior to a blood transfusion**



Filling out the Form

1591162 Newborn Screening Form
Oklahoma State Department of Health, P.O. Box 24106,
Oklahoma City, OK 73124-0106 (405) 271-5576 ODH #450 REV 03-2016

DO NOT WRITE IN THIS BOX

SPECIMEN INFORMATION

Refused (Check if caregiver refuses-Fax signed refusal form)

1. Collection Date: MM DD YY Collection Time: : 24 Hour Clock

2. Transfusion Date: MM DD YY Time: : 24 Hour Clock

Do not write in this box

3. Has a previous metabolic blood test been done anywhere? Yes No
Previous OSDH Lab Number

4. Check all that apply at time of screening:
 TPN Antibiotics Lactose-Free Formula (Soy)
 Meconium ileus Family History of CF

5. Test Requested:
 All Tests HGB Only GALT CFTR Phe Monitor
 Adoption (check if baby is being adopted)

HEARING SCREENING INFORMATION

1. Infant's Last Name: _____ Infant's First Name: _____

2. Sex: M F 3. Date of Birth: MM DD YY 4. Birth Time: : 24 Hour Clock 5. Gest. Age: _____

6. Birthweight in Grams: _____ 7. If Multiple Birth Indicate Birth Order: A-H 8. Infant's Medical Record or I.D. #: _____

9. Provider ID: _____ 10. Infant's Provider or Physician's Name: _____

11. Mom's Medicaid Number: _____ 12. Provider's Phone Number: () - -

1. Mom's Last Name, First Name: _____ 2. Mom's Age: _____

3. Mom's Address: _____ 4. Apt. #: _____

5. Mom's City: _____ 6. State: _____ 7. Zip: _____

8. Mom's Telephone or Contact: () - - 9. Mom's Social Security #: - -

10. Mom's Race/Ethnic: 1. White 2. Black 3. Hispanic 4. Asian 5. Indian 6. Other

Pulse Oximetry (CCHD) Screen
 Not Performed Pass Fail Echo Refused

SUBMITTING HEALTH PROVIDER ID # _____
Return to Submitter at this address: _____

Hearing Screening Results:

Right Ear	Left Ear	Screen Method
<input type="checkbox"/> Pass	<input type="checkbox"/> Pass	<input type="checkbox"/> ABR <input type="checkbox"/> Other (Specify) _____
<input type="checkbox"/> Refer	<input type="checkbox"/> Refer	<input type="checkbox"/> OAE

If not screened, reason:
 Technical problem No equipment Delayed
 Caregiver refused Baby discharged Other _____

Hearing risk status—Check all that apply:
 Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
 Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
 Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
 Infant had exchange transfusion.
 Infant has serum bilirubin level \geq 15 mg/dL.
 Infant was placed in a Level II or III nursery for more than 24 hours.

CHART COPY
DETACH AND PLACE IN MEDICAL RECORD
NEWBORN SCREENING FORM

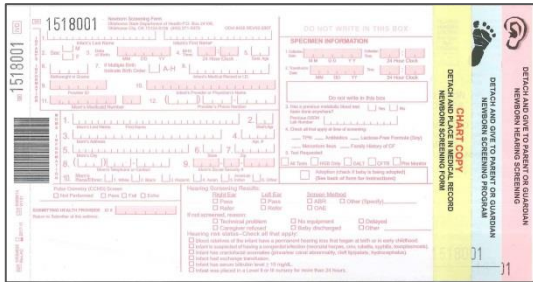
DETACH AND GIVE TO PARENT OR GUARDIAN
NEWBORN SCREENING PROGRAM

DETACH AND GIVE TO PARENT OR GUARDIAN
NEWBORN HEARING SCREENING

105178 / 316104
LOT 2019-03

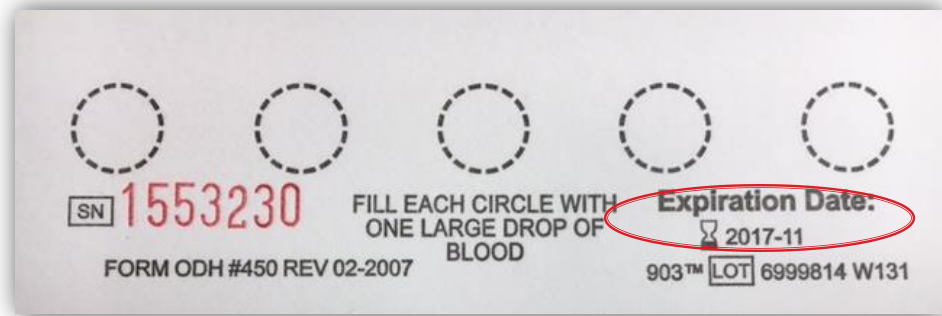
Specimen testing will be delayed if the form is incomplete!

Filling out the Form



The image shows a detailed form for specimen collection. It includes a header with the number 1518001, a barcode, and various sections for patient information, specimen information, and laboratory use. The form is titled 'CERTIFICATE OF SPECIMEN INFORMATION' and 'CHART COPY'. It contains numerous checkboxes and text boxes for data entry.

- Check expiration date
 - Ex: 2017-11 means the filter paper expires the last day of November 2017.
 - If filter paper is expired, discard the paper, check the stock of filter paper kits it came from to ensure they are not all expired, and collect on a kit that is not expired.



The image shows a close-up of a filter paper kit. It features five dashed circles for blood collection. Below the circles, the text reads: 'SN 1553230', 'FILL EACH CIRCLE WITH ONE LARGE DROP OF BLOOD', and 'Expiration Date: 2017-11'. The expiration date is circled in red. At the bottom, it says 'FORM ODH #450 REV 02-2007' and '903™ LOT 6999814 W131'.

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Infant's Information

1518001

DO NOT WRITE IN THIS BOX

CHART COPY

OFFICIAL AND NOT TO BE REPRODUCED OR QUOTED IN ANY MANNER WITHOUT THE WRITTEN PERMISSION OF THE OKLAHOMA STATE DEPARTMENT OF HEALTH

OFFICIAL AND NOT TO BE REPRODUCED OR QUOTED IN ANY MANNER WITHOUT THE WRITTEN PERMISSION OF THE OKLAHOMA STATE DEPARTMENT OF HEALTH

CHART COPY

OFFICIAL AND NOT TO BE REPRODUCED OR QUOTED IN ANY MANNER WITHOUT THE WRITTEN PERMISSION OF THE OKLAHOMA STATE DEPARTMENT OF HEALTH

- Baby's first & last name
 - May include "BG" or "Female", "BB" or "Male" for first name if unknown

1591162

INVD

SN 1591162

Newborn Screening Form
Oklahoma State Department of Health-P.O. Box 24106
Oklahoma City, OK 73124-0106 (405) 271-2070 ODH 6450 REV 03-2016

1. Infant's Last Name

2. Sex: M F

3. Date of Birth: MM DD YY

4. Birth Time: : 24 Hour Clock

5. Gest. Age

6. Birthweight in Grams

7. If Multiple Birth Indicate Birth Order: A-H

8. Infant's Medical Record or I.D.

9. Provider ID

10. Infant's Provider or Physician's Name

11. Mom's Medicaid Number

12. () - Provider's Phone Number

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Infant's Information

A small image of the newborn screening form. A red circle highlights the 'SEX/GENDER' section, which contains checkboxes for 'M' (Male) and 'F' (Female). The form also includes fields for 'Infant's Last Name', 'Infant's First Name', 'Date of Birth', 'Birth Time', 'Gest. Age', 'Birthweight in Grams', 'If Multiple Birth Indicate Birth Order', 'Provider ID', 'Infant's Provider or Physician's Name', 'Mom's Medicaid Number', and 'Provider's Phone Number'.

- Sex/Gender
 - check "M" or "F"

A larger image of the newborn screening form. A red circle highlights the 'SEX/GENDER' section, which contains checkboxes for 'M' (Male) and 'F' (Female). The form also includes fields for 'Infant's Last Name', 'Infant's First Name', 'Date of Birth', 'Birth Time', 'Gest. Age', 'Birthweight in Grams', 'If Multiple Birth Indicate Birth Order', 'Provider ID', 'Infant's Provider or Physician's Name', 'Mom's Medicaid Number', and 'Provider's Phone Number'.

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Infant's Information

Thumbnail of the Newborn Screening Form. A red circle highlights the 'Date of Birth' field in the 'Infant's Information' section. The form includes fields for infant name, sex, date of birth, birth weight, provider information, and maternal information.

➤ Date & time of birth

Close-up of the Newborn Screening Form. The form is titled 'Newborn Screening Form' and includes the Oklahoma State Department of Health contact information. The 'Infant's Information' section contains the following fields:

- 1. Infant's Last Name
- 2. Sex: M F
- 3. Date of Birth: MM DD YY (circled in red)
- 4. Birth Time: 24 Hour Clock (circled in red)
- 5. Gest. Age
- 6. Birthweight in Grams
- 7. If Multiple Birth Indicate Birth Order: A-H
- 8. Infant's Medical Record or I.D.
- 9. Provider ID
- 10. Infant's Provider or Physician's Name
- 11. Mom's Medicaid Number
- 12. Provider's Phone Number

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Infant's Information

The image shows a small version of the newborn screening form. A red circle highlights the 'Gestational Age' field, which is field number 5 on the form. The form includes various sections for patient information, medical history, and laboratory results.

- Gestational Age
 - List gestational age at birth.
 - Follow-up for abnormal SCID results are gestational age dependent.

The image shows a larger version of the newborn screening form. The form is titled 'Newborn Screening Form' and includes the Oklahoma State Department of Health contact information. The form is numbered '1591162' and '1591162'. The 'Gest. Age' field (field number 5) is circled in red. The form includes fields for Infant's Last Name, Infant's First Name, Sex, Date of Birth, Birth Time, Birthweight in Grams, Multiple Birth, Infant's Medical Record or I.D., Provider ID, Infant's Provider or Physician's Name, Mom's Medicaid Number, and Provider's Phone Number.

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Infant's Information

A small image of the newborn screening form. A red circle highlights the 'BIRTHWEIGHT IN GRAMS' field, which is currently empty. The form includes various sections for infant information, including name, sex, date of birth, and provider information.

- Birthweight (in grams)
 - Follow-up for abnormal CAH results are dependent on birth weight.

A larger image of the newborn screening form. The form is titled 'Newborn Screening Form' and includes the Oklahoma State Department of Health contact information. The form is numbered '1591162' and 'ODH #450 REV 03-2016'. The 'BIRTHWEIGHT IN GRAMS' field is circled in red. The form includes fields for infant's last name, first name, sex, date of birth, birth time, gestational age, provider ID, provider name, mom's Medicaid number, and provider's phone number.

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Infant's Information

A small image of the newborn screening form. A red circle highlights the 'Infant's Information' section, which is the focus of the slide. The form includes fields for infant's name, sex, date of birth, birth weight, and provider information.

- Birth order (if multiple birth is present)
 - Indicate "A", "B", "C", etc.. if baby is of a multiple birth (twin, triplet, etc..).
 - Do not mark anything in this space if baby is of a single birth.

A larger image of the newborn screening form. A red circle highlights the field '7. If Multiple Birth Indicate Birth Order: A-H'. The form includes fields for infant's name, sex, date of birth, birth weight, provider information, and mom's Medicaid number.

1591162 IVD

SN 1591162

Newborn Screening Form
Oklahoma State Department of Health-P.O. Box 24106,
Oklahoma City, OK 73124-0106 (405) 271-5070 ODH #450 REV 03-2016

1. Infant's Last Name

2. Sex: M F

3. Date of Birth: MM DD YY

4. Birth Time: : 24 Hour Clock

5. Gest. Age

6. Birthweight in Grams

7. If Multiple Birth Indicate Birth Order: A-H

8. Infant's Medical Record or I.D.

9. Provider ID

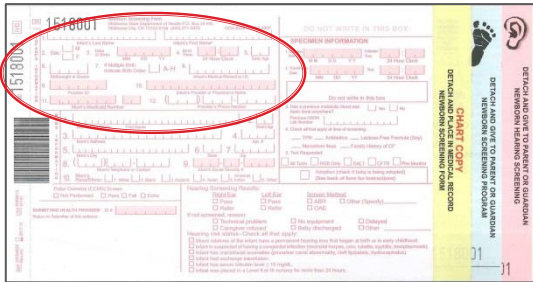
10. Infant's Provider or Physician's Name

11. Mom's Medicaid Number

12. () - Provider's Phone Number

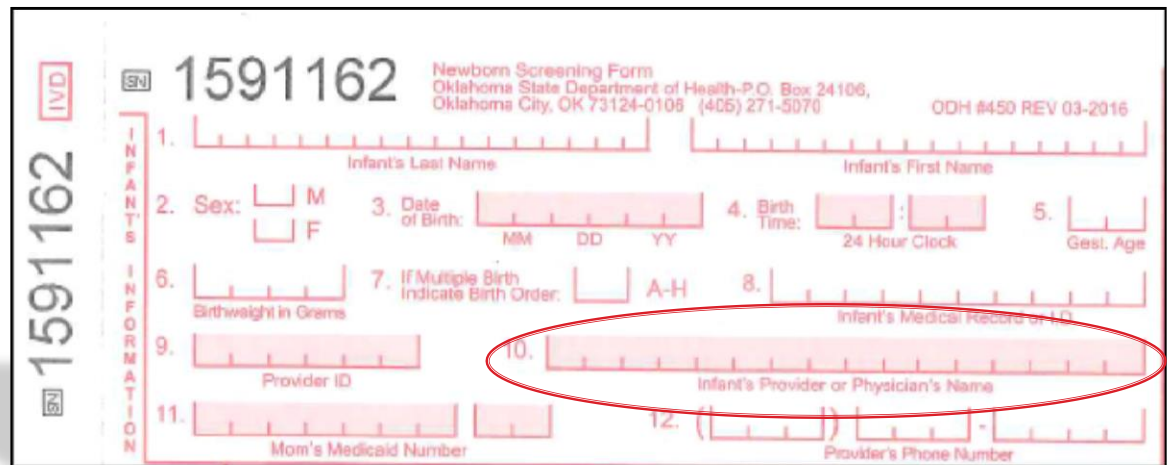
Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Infant's Information



A small image of the newborn screening form. A red circle highlights the 'Infant's Information' section, which is the focus of the slide. The form includes fields for infant name, sex, date of birth, birth weight, provider ID, and mom's Medicaid number.

- Infant's provider/physician
 - Planned health care provider upon discharge from birthing facility
 - Extremely important to include in case newborn screen results are abnormal and require follow-up



A larger image of the newborn screening form. A red circle highlights the 'Infant's Provider or Physician's Name' field (item 10). The form includes fields for infant name, sex, date of birth, birth weight, provider ID, mom's Medicaid number, and provider's phone number.

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Mom's Information

A screenshot of the Oklahoma State Department of Health form. A red oval highlights the 'MOM'S INFORMATION' section, which is the focus of the slide. The form includes various fields for personal and contact information, as well as checkboxes for medical history and other details.

➤ Mom's first & last name

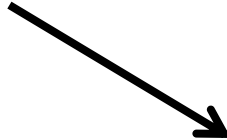
A close-up of the 'MOM'S INFORMATION' section of the form. A red oval highlights the 'MOM'S FIRST NAME' and 'MOM'S AGE' fields. The form includes various fields for personal and contact information, as well as checkboxes for medical history and other details.

Note: If baby is adopted, be sure to check the **Adoption** box on the filter paper. Also be sure to list agency/law firm information in this section. If DHS is involved, include case worker information & write “DHS Custody” on the filter paper.

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Mom's Information

- Mom's address:
 - Street, Apt # (if applicable), City, State, Zip



Note: If baby is adopted, be sure to check the **Adoption** box on the filter paper. Also be sure to list agency/law firm information in this section. If DHS is involved, include case worker information & write “DHS Custody” on the filter paper.

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Mom's Information

1518001

DO NOT WRITE IN THIS BOX

DEFENDANT CENTER FOR PREVENTION OF GOUTDAN
NEWBORN SCREENING FORM

CHART COPY
DEFENDANT CENTER FOR PREVENTION OF GOUTDAN
NEWBORN SCREENING FORM

DEFENDANT CENTER FOR PREVENTION OF GOUTDAN
NEWBORN SCREENING FORM

- Mom's telephone number
 - Extremely important to include in case newborn screen results are abnormal and require follow-up

MOM'S INFORMATION

1. Mom's Last Name, First Name

2. Mom's Age

3. Mom's Address

4. Apt. #

5. Mom's City

6. State

7. Zip

8. Mom's Telephone or Contact

9. Mom's Social Security #

10. Mom's Race/Ethnic: 1 White 2 Black 3 Hispanic 4 Asian 5 American Indian 6 Other

Note: If baby is adopted, be sure to check the **Adoption** box on the filter paper. Also be sure to list agency/law firm information in this section. If DHS is involved, include case worker information & write "DHS Custody" on the filter paper.

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Specimen Information

The image shows a full newborn screening form. A red circle highlights the 'SPECIMEN INFORMATION' section, which is the focus of the slide. The form includes fields for parent/guardian information, specimen collection details, and a refusal section.

- Check the box if parents refuse the NBS
 - Provide parents with a NBS brochure & answer any questions they might have about the screen
 - Ensure the parents fill out a Refusal Form; keep a copy for baby's record & fax a copy to the NBS Program using fax # 405-271-4892.

The image is a close-up of the 'SPECIMEN INFORMATION' section of the form. A red circle highlights the checkbox for 'Refused (Check if caregiver refuses-Fax signed refusal form)'. Below this are two rows of date and time fields:

Field	MM	DD	YY	Time	24 Hour Clock
1. Collection Date:					
2. Transfusion Date:					

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Specimen Information

1518001
SPECIMEN INFORMATION

DO NOT WRITE IN THIS BOX

DEPT. OF HEALTH
CENTRAL LABORATORY
CHILD COPY

DEPT. OF HEALTH
CENTRAL LABORATORY
CHILD COPY

- Date & time of specimen collection
 - Ideal time for well, term newborn:
24 hours + 1 minute of age

SPECIMEN INFORMATION

Refused (Check if caregiver refuses-Fax signed refusal form)

1. Collection Date: MM DD YY Collection Time: : 24 Hour Clock

2. Transfusion Date: MM DD YY Time: : 24 Hour Clock

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Specimen Information

The image shows a scan of a blood transfusion form. A red oval highlights the 'SPECIMEN INFORMATION' section, which is the focus of the subsequent slide. The form includes various fields for patient information, blood type, and transfusion details.

- Date & time of blood transfusion
 - If applicable

The image shows a close-up of the 'SPECIMEN INFORMATION' section of the form. A red oval highlights the date and time fields. The fields are labeled '1. Collection Date:' and 'Collection Time:', and '2. Transfusion Date:' and 'Time:'. Each field has a dropdown menu for the month (MM), day (DD), and year (YY), and a dropdown menu for the time in a 24-hour clock format.

Specimen testing will be delayed if the form is incomplete!



Filling out the Form: Specimen Information

The image shows the full newborn screening form. A red circle highlights the 'SPECIMEN INFORMATION' section, which includes questions about previous metabolic blood tests and the OSDH Lab Number.

- Check “Yes” if a previous newborn screen has been collected
 - List previous OSDH Lab Number, if applicable.

This is a close-up of the 'SPECIMEN INFORMATION' section of the form. A red circle highlights the following text:

3. Has a previous metabolic blood test been done anywhere? Yes No
Previous OSDH Lab Number _____

4. Check all that apply at time of screening:
 TPN Antibiotics Lactose-Free Formula (Soy)
 Meconium ileus Family History of CF

5. Test Requested:
 All Tests HGB Only GALT CFTR Phe Monitor

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Specimen Information

1518001

DO NOT WRITE IN THIS BOX

SPECIMEN INFORMATION

TPN Antibiotics Lactose-Free (Soy) Formula Meconium Ileus Family History of Cystic Fibrosis (CF)

CHART COPY

OKLAHOMA STATE DEPARTMENT OF HEALTH

- Check all that apply for baby at the time of specimen collection:
 - TPN
 - Antibiotics
 - Lactose-Free (Soy) Formula
 - Meconium Ileus
 - Family History of Cystic Fibrosis (CF)

3. Has a previous metabolic blood test been done anywhere? Yes No

Previous OSDH Lab Number _____

4. Check all that apply at time of screening:

TPN Antibiotics Lactose-Free Formula (Soy)

Meconium Ileus Family History of CF

5. Test Requested:

All Tests HGB Only GALT CFTR Phe Monitor

Specimen testing will be delayed if the form is incomplete!

Filling out the Form: Specimen Information

The image shows the Oklahoma newborn screening form. A red circle highlights the 'Test Requested' section at the bottom of the form. The form includes fields for 'Specimen Information', 'Parental Consent', and 'Test Requested'. The 'Test Requested' section has checkboxes for 'All Tests', 'HGB Only', 'GALT', 'CFTR', and 'Phe Monitor'.

- **Test Requested:** check the appropriate box(es)
 - **All Tests** – always check unless test is for HGB Only or Phe Monitor. This ensures the lab screens for all disorders on the newborn screening panel
 - **HGB Only** – check if repeat screen is follow-up for initial abnormal HGB result
 - **CFTR** – check (alongside All Tests) if baby has clinical concerns for Cystic Fibrosis, meconium ileus, and/or family history of CF
 - **Phe Monitor** – check only if baby has been diagnosed with PKU

The image shows a close-up of the 'Test Requested' section of the form. A red circle highlights the checkboxes for 'All Tests', 'HGB Only', 'GALT', 'CFTR', and 'Phe Monitor'. The text above the checkboxes reads: '5. Test Requested.'

Specimen testing will be delayed if the form is incomplete!

Filling out the Form

The image shows a specimen collection form with a red circle highlighting the 'Adoption' checkbox in the 'MOM'S INFORMATION' section. The form includes fields for patient information, specimen information, and a section for 'MOM'S INFORMATION' with checkboxes for 'Adoption', 'Surrogate', and 'Other'. The 'Adoption' checkbox is circled in red.

- Adoption (if applicable)
 - Check the Adoption box & list the agency/law firm name (& full contact information) that is handling the adoption in the “Mom’s Information” section.



Adoption (check if baby is being adopted)
(See back of form for instructions)

Specimen testing will be delayed if the form is incomplete!

Filling out the Form

The image shows a scan of a 'Pulse Oximetry (CCHD) Screen' form. The form is titled 'Pulse Oximetry (CCHD) Screen' and includes a section for 'SPECIMEN INFORMATION'. A red circle highlights the 'Pulse Oximetry (CCHD) Screen' section at the bottom of the form, which contains five checkboxes: 'Not Performed', 'Pass', 'Fail', 'Echo', and 'Refused'. An arrow points from this section to a larger, clearer view of the same section below.

- Pulse Oximetry (CCHD) Screen: check one
 - Not Performed
 - Pass
 - Fail
 - Echo
 - Refused

The image shows a close-up of the 'Pulse Oximetry (CCHD) Screen' section of the form. The section is titled 'Pulse Oximetry (CCHD) Screen' and contains five checkboxes: 'Not Performed', 'Pass', 'Fail', 'Echo', and 'Refused'. A red circle highlights the entire section.

- **Note:** If parents refuse the pulse oximetry screen, provide them with a pulse oximetry brochure and answer any questions they might have about the screen. Ensure the parents fill out a Refusal Form; keep a copy for baby's record & fax a copy to the NBS Program using fax # 405-271-4892.

Specimen testing will be delayed if the form is incomplete!

Filling out the Form

- Submitting Health Provider ID #
 - This is the ID of the provider/facility who collected the specimen.



Specimen testing will be delayed if the form is incomplete!

Filling out the Form

➤ Unsatisfactory Specimen Follow-up

- Scanned images of unsatisfactory specimens are emailed back to hospitals to be used for continuing education. Specimen collectors can place their initials and unit in the indicated area, as shown below, for identification of who collected the specimen in the event that it is unsuitable for testing. This allows for easier identification of the collecting individual so that further education on specimen collection can be provided.

SN! 1553228

FILL EACH CIRCLE WITH ONE LARGE DROP OF BLOOD

Expiration Date: 2017-11

903™ LOT 6999814 W131

RH
NICU

Collecting the Specimen



Time of Screening: Healthy Newborn

“24 hours plus one minute” of age
Or
Prior to discharge

****WHICHEVER COMES FIRST****



Time of Screening: Premature or Sick Newborns



24 hours + 1
minute of age



14 days of age



Prior to red blood
cell transfusion,
even if collected at
<24 hours of age

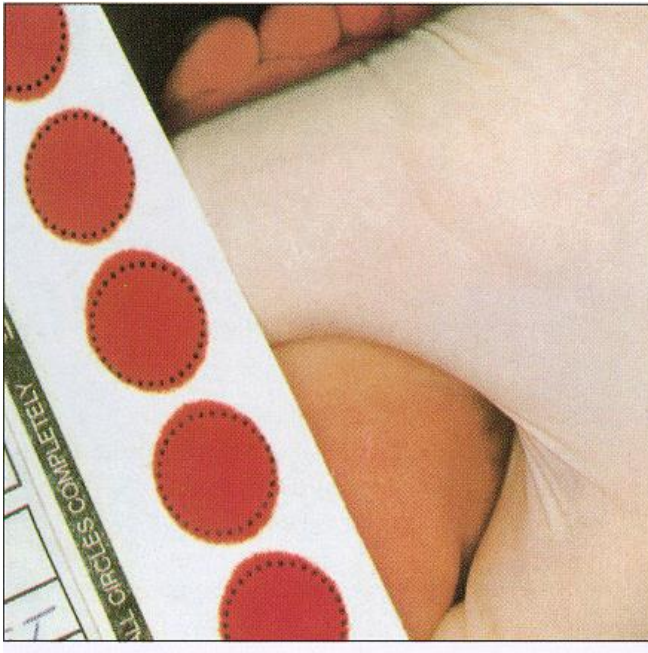
Collect no later
than 3-7 days
of age

Or immediately prior to
discharge, whichever comes first



Specimen Collection

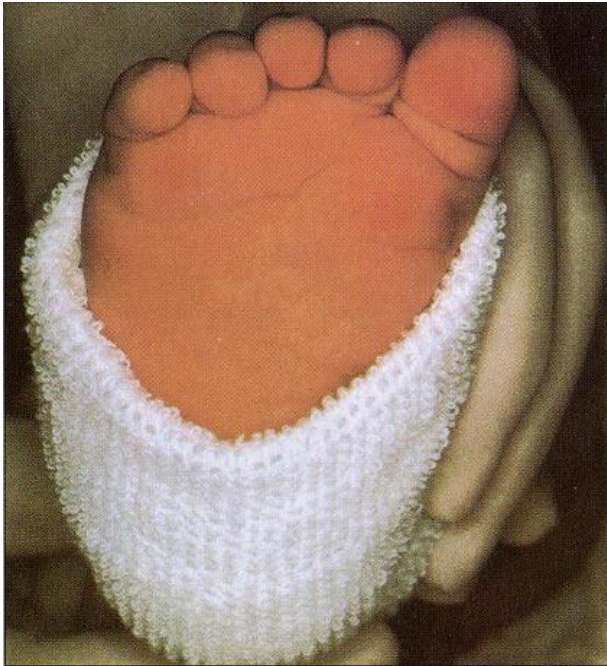
- ▶ **Heel Stick / Direct Application**
 - Preferred, recommended method



****Start with clean, dry hands
before handling the filter paper****



Direct Application



Prepare the Site

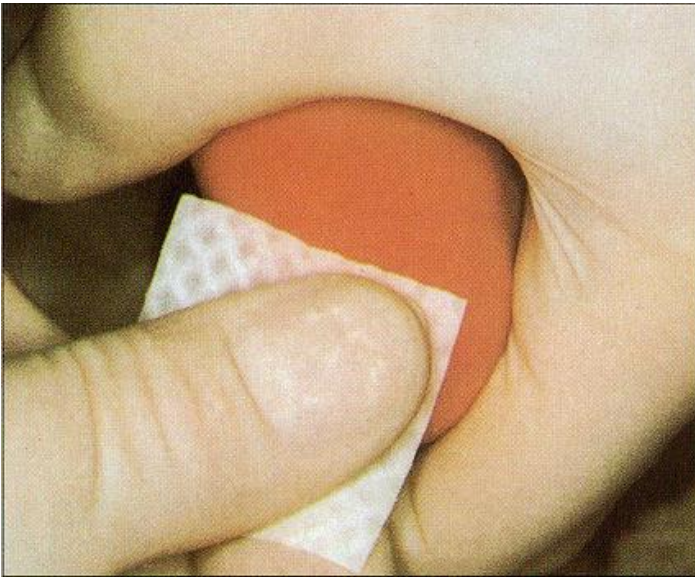
- ▶ Warm the heel with a heel warmer or a soft cloth, moistened with warm water up to 41° C for 3 to 5 minutes.
 - Warmth leads to vasodilation, which increases bloodflow and chance of collection success.

Follow your hospital protocol regarding which warming device to use



Direct Application

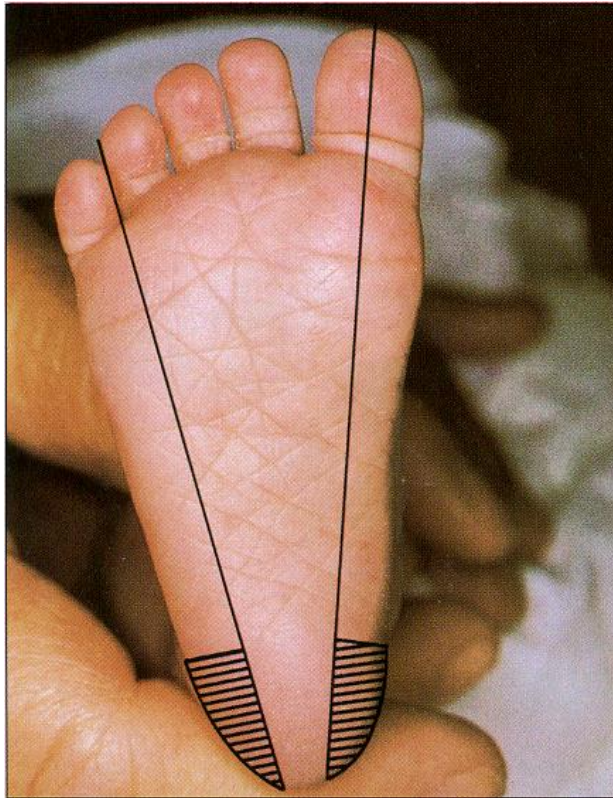
Prepare the Site



- ▶ Encourage skin-to-skin contact between newborn and parent during specimen collection.
 - Decreases stress response in newborn
 - Encourages bonding
- ▶ Position the infant's leg lower than the heart.
 - This increases venous pressure, which results in increased blood flow and a greater chance of collection success.
- ▶ Wearing gloves, wipe the infant's heel with 70% isopropyl alcohol.
- ▶ Allow the heel to air dry!
 - Residual alcohol can affect NBS results &/or lead to unsat specimens.



Direct Application



Lancet Placement

- ▶ Hatched areas are safe for puncture
 - Damage to nerves and/or the heel bone may occur for punctures outside of the hatched region.



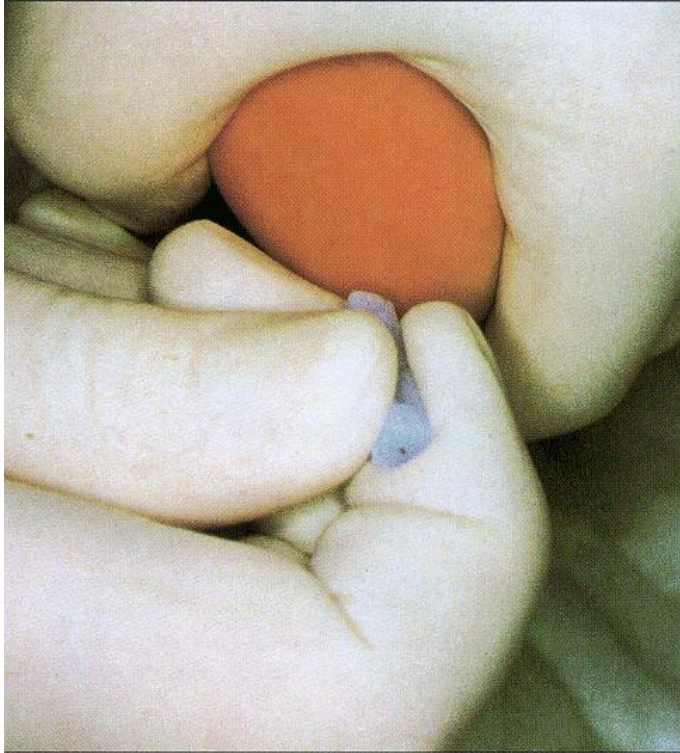
Direct Application

Perform the Puncture: Lancet Specifications

- ▶ **Full term infants (gest. age \geq 37 wks):**
 - Standard incision is 2.5 mm in length & 1 mm in depth.
 - The vascular bed of an infant's heel is about 0.35 to 0.82 mm beneath the skin; the 1 mm length of the lancet incises across this capillary-rich area for optimum blood flow. Pain fibers increase in abundance below 2.4 mm; the 1 mm incision depth also works to reduce the pain experience.
- ▶ **Premature infants (gest. age < 37 wks):**
 - Incision specifications are 1.75 mm in length & 0.85 mm in depth.
 - This is about 40% smaller than the incision for a full term infant.
- ▶ Summarized from the *Clinical and Laboratory Standards Institute (CLSI)* guidelines. Please refer to these guidelines for further information.



Direct Application

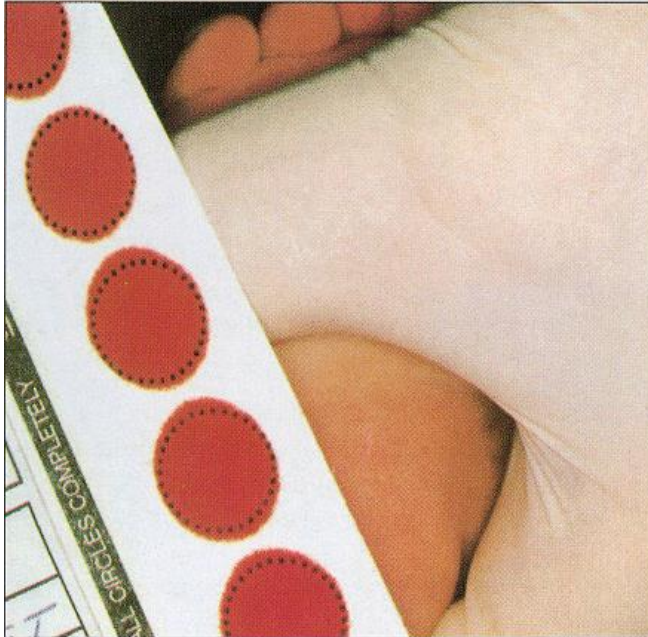


Perform the Puncture

- ▶ Using a sterile lancet, perform the puncture.
- ▶ Gently wipe off the first drop of blood with a sterile gauze or cotton ball.
- ▶ Apply gentle pressure with thumb and around heel but not near the puncture site; ease intermittently as drops of blood form.
- ▶ Avoid “milking” the puncture site.



Direct Application



Application

- ▶ Gently touch the filter paper card to the blood drop and fill each printed circle with **one** large drop of blood.
- ▶ Apply blood to **one** side only.
- ▶ Observe the saturation of each printed circle as the blood flows through the filter paper.

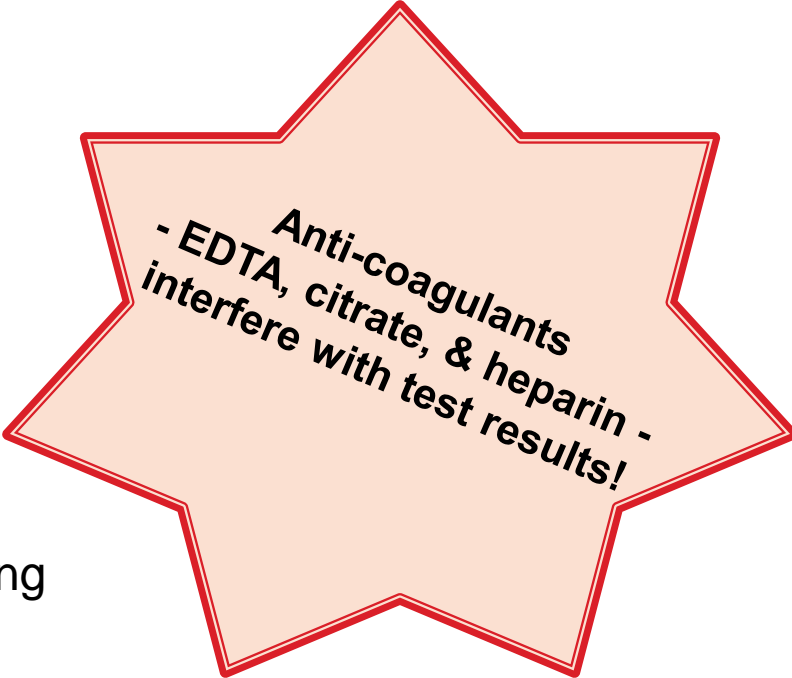


Alternative Specimen Collection

What about capillary tubes?

➤ Not preferred

- Higher risk for collection error
- If used, must be sterile/clean & plain.
 - No additives! Must be anti-coagulant free.
 - However... no anti-coagulants = risk for clotting
- Risk of scratching the filter paper.
 - Avoid touching the capillary tip to the paper.
- Use a new tube for each pre-printed circle.



Anti-coagulants
- EDTA, citrate, & heparin -
interfere with test results!



Alternative Specimen Collection

▶ What about venous samples?

- **Discouraged**
- May be appropriate under certain circumstances (e.g. NICU).
- More invasive than a heel stick.
- Do not draw blood from extremity with infusing IV fluids.
- Please refer to current CLSI guidelines for more information.



Alternative Specimen Collection

▶ What about umbilical catheters?

- **Discouraged**
- May be appropriate under certain circumstances (e.g. NICU).
- Ensure the line is cleared by withdrawing 2 – 2.5 cc (mL) of blood prior to collection a specimen for NBS.
- Please refer to current CLSI guidelines for more information.



Alternative Specimen Collection

▶ What about umbilical cord blood?

- **Discouraged**
- May be appropriate under certain circumstances (e.g. NICU).
- Risk for maternal blood contamination.
- Repeat the newborn screen using the heel stick method when indicated.

- Please refer to current CLSI guidelines for more information.



Specimen Collection: What NOT to Do

- Do NOT dab or “color in” the filter paper circles.
- Do NOT apply multiple drops of blood per circle.
- Do NOT scratch the filter paper.
- Do NOT contaminate specimens.
 - insufficient drying of alcohol, oils on hands, lotions, compressing the circles, spills, etc..
- Do NOT stack specimens.
 - risk for leaching & cross-contamination between specimens
- Do NOT submit wet specimens.
- Do NOT place specimens in direct sunlight or in front of air vents or other sources of moving air.
- Do NOT place wet specimens in plastic bags.
- Do NOT batch (hold onto) specimens.



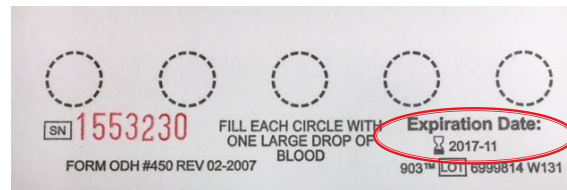
Collection Reminders

Pre-collection:

- ✓ **Check the Expiration Date of the filter paper**

Ex: 2017-11 means the filter paper expires on the last day of November 2017

If filter paper is expired, discard the paper, check the stock of filter paper kits it came from to ensure they are not all expired, and collect on a kit that is not expired.



Post-collection:

- ✓ **Air dry specimen horizontally for 3-4 hours**

Transporting wet specimens can make them unsatisfactory for testing.

- ✓ **Send specimen with Courier within 24 hours of collection**

Delayed receipt of specimens to the Public Health Laboratory can delay identification of and treatment for a disorder, which can result in lifelong disability or even death for Oklahoma newborns.

Know the courier schedule and location for your facility! Ensure all staff involved in newborn screening are also aware of the process.

- ✓ **Maintain specimen collection log & ensure screening results are received & recorded**

- ✓ **Ensure that everybody who handles the filter paper or is involved in the newborn bloodspot collection process is trained**

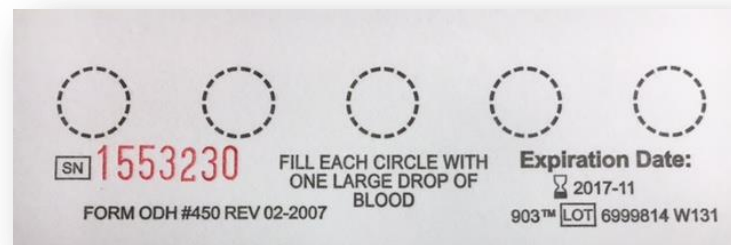
NBS Filter Paper Review

Unsatisfactory (“Unsat”) Specimen Examples



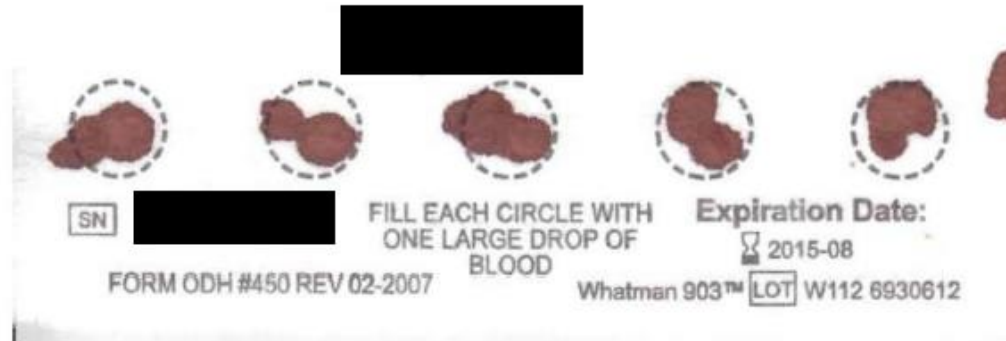
Filter Paper

- ▶ The filter paper is part of the NBS Form. It is a medical device designed to absorb a specific volume of blood within each pre-printed filter paper circle.
- ▶ If an analyte for any disorder is either too high or too low, this is an indication that additional testing is needed.
- ▶ Accurate results depend upon proper absorption of blood onto the filter paper.
 - Too much or too little blood may result in inaccurate results.

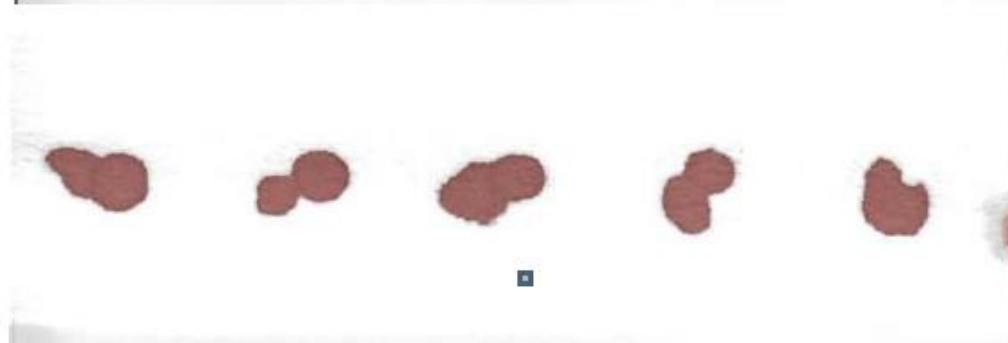


Multiple Application

Front



Back



Back

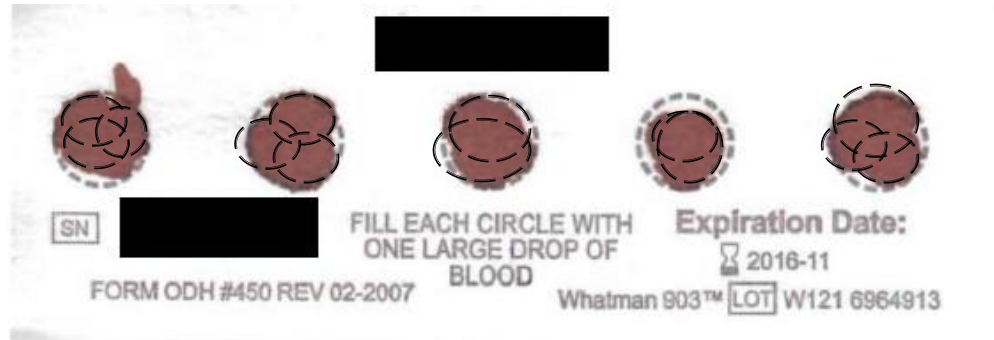
➤ Why Unsat?

- When bloodspots overlap or touch, as is the case in the sample above, it creates an uneven absorption of blood.
- Analyte levels cannot be accurately measured.
- Testing these specimens will result in inaccurate results.



Multiple Application

Front



Back



Back

It may be easier to identify multiple application of blood drops by observing the back side of the filter paper.

➤ Why Unsat?

- When bloodspots overlap or touch, as is the case in the sample above, it creates an uneven absorption of blood.
- Analyte levels cannot be accurately measured.
- Testing these specimens will result in inaccurate results.

Multiple Application – Prevention

- ▶ Apply one large drop of blood to fill each pre-printed circle
- ▶ Patience, patience, patience!
 - Wait for a full, healthy drop of blood before applying to the filter paper.
- ▶ Avoid overlapping blood spots.
- ▶ **Submitting an unsatisfactory specimen delays screening & potential identification and treatment of a disorder. If in doubt, recollect immediately!**



Clotted or Caked Blood



➤ Why Unsat?

- Clots can occur using capillary tubes or if too much blood is applied to the pre-printed circles.
- Samples with clots are not suitable for testing.



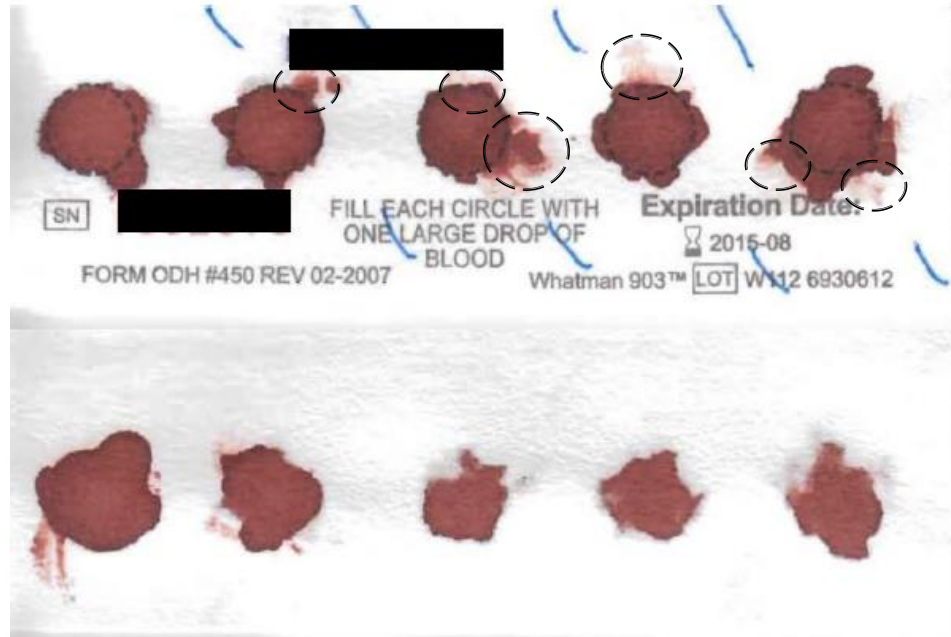
Clotted or Caked Blood - Prevention

- ▶ Avoid using capillary tubes for collection, if possible.
 - ▶ If a capillary tube is used, it must be sterile/clean and plain (anti-coagulant free) due to additives interfering with test results.
- ▶ Do not wait too long to apply the blood to the filter paper.
- ▶ It is easier to identify clots when the specimen is dry.
- ▶ **Submitting an unsatisfactory specimen delays screening & potential identification and treatment of a disorder. If in doubt, recollect immediately!**



Serum Rings

Front



Back

➤ Why Unsat?

- Notice the halos around the periphery of most of the pre-printed circles above. This can occur due to the following:
 - Insufficient drying of alcohol on the baby's heel prior to heelstick
 - Drying the specimen vertically instead of horizontally
 - Closing the flap of the filter paper on top of the circles while the specimen is still wet
 - Placing wet specimens in plastic bags
 - Milking or squeezing the puncture site

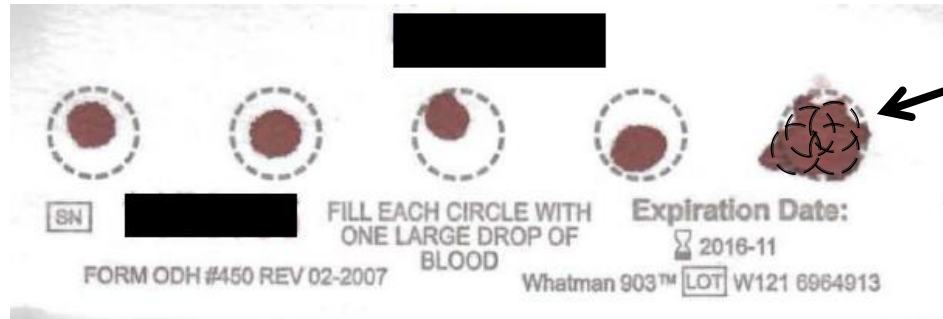
Serum Rings - Prevention

- ▶ Ensure the alcohol dries on the newborn's heel prior to puncture.
- ▶ Avoid milking the heel site. Work your way around the infant's heel, using intermittent pressure as the drop of blood forms.
- ▶ Allow specimens to dry appropriately after collection:
 - ▶ Lay horizontally for 3-4 hours.
 - ▶ Do not place wet specimens in plastic bags.
 - ▶ Do not close the flap over the pre-printed circles until the specimen is dry.
 - ▶ Avoid contamination, including alcohol and hand lotion.
- ▶ **Submitting an unsatisfactory specimen delays screening & potential identification and treatment of a disorder. If in doubt, recollect immediately!**



Inadequate Amount of Blood

Front



Note the multiple application of blood drops.

Back



➤ **Why Unsat?**

- The above filter paper circles are not sufficiently filled with blood for testing.

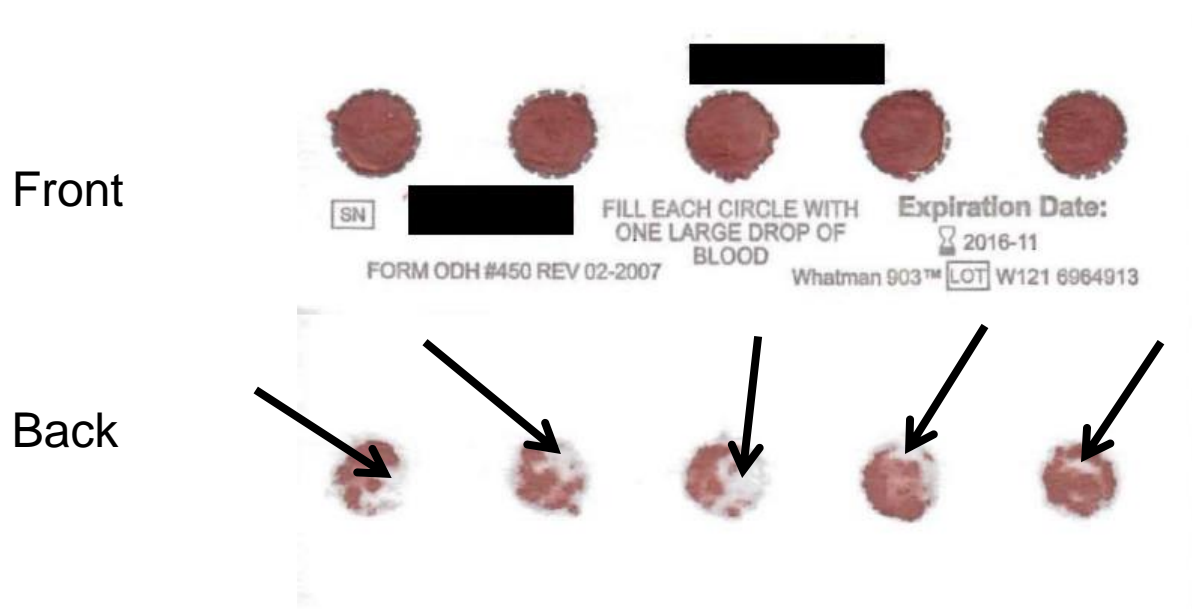


Inadequate Amount of Blood- Prevention

- ▶ Set yourself up for success! Prior to specimen collection, ensure:
 - ▶ Baby's heel has been pre-warmed via warm cloth or heel warmer for up to 3-5 minutes.
 - ▶ This leads to vasodilation, which increases bloodflow.
 - ▶ Baby's heel is lower than the heart.
 - ▶ This increases venous pressure, which enhances bloodflow.
- ▶ Patience, patience, patience!
 - ▶ Wait for a full, healthy drop of blood before applying to the filter paper.
- ▶ After each blood drop is applied, check to ensure that the blood has soaked completely through the filter paper.
- ▶ **Submitting an unsatisfactory specimen delays screening & potential identification and treatment of a disorder. If in doubt, recollect immediately!**



Under-Saturation



➤ *Why Unsat?*

- Notice how the blood has not soaked all the way through the filter paper. There simply is not enough blood in this sample for testing.



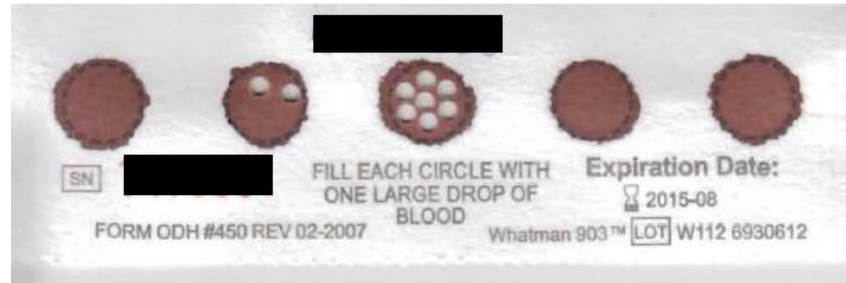
Under-Saturation - Prevention

- ▶ After each blood drop is applied, check to ensure that the blood has soaked completely through the filter paper.
- ▶ Patience, patience, patience!
 - ▶ Wait for a full, healthy drop of blood before applying to the filter paper.
- ▶ **Submitting an unsatisfactory specimen delays screening & potential identification and treatment of a disorder. If in doubt, recollect immediately!**

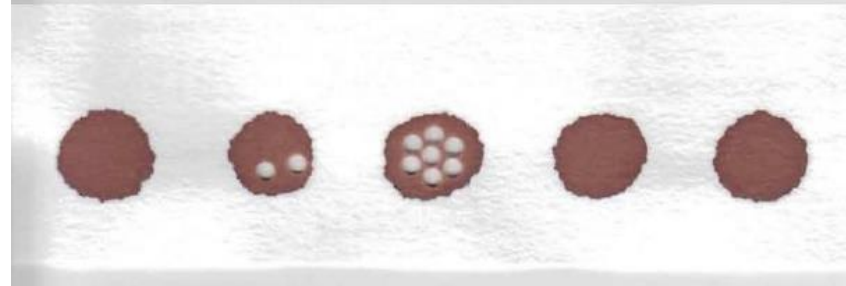


Acceptable Filter Paper

Front



Back

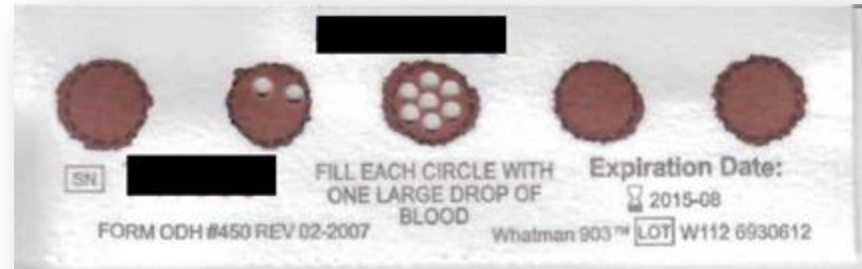


➤ **Why Acceptable?**

- Pre-printed circles are completely filled with blood
- Blood has soaked all the way through the filter paper
- Absence of clots or caked blood
- Absence of serum rings



Are All 5 Circles Needed?



Why?

- ▶ If a result is flagging out-of-range, the specimen will be retested and the final result will be an average of three results. Each test requires an additional punch to be taken from the pre-printed circles.
- ▶ If the results for Congenital Adrenal Hyperplasia (CAH) are out-of-range, **two entire pre-printed circles** will be removed & shipped to another laboratory for steroid profile testing.
- ▶ Disorders will continue to be added to the newborn screening panel.
- ▶ The specialist and family may request for the specimen to be sent to another laboratory for additional testing to assist in determining diagnosis.

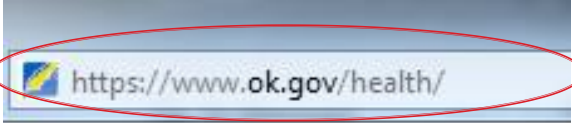



For Reference...

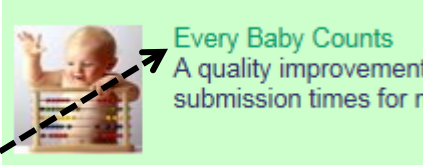
- ▶ Refer to *Clinical and Laboratory Standards Institute (CLSI)* for collection guidelines.

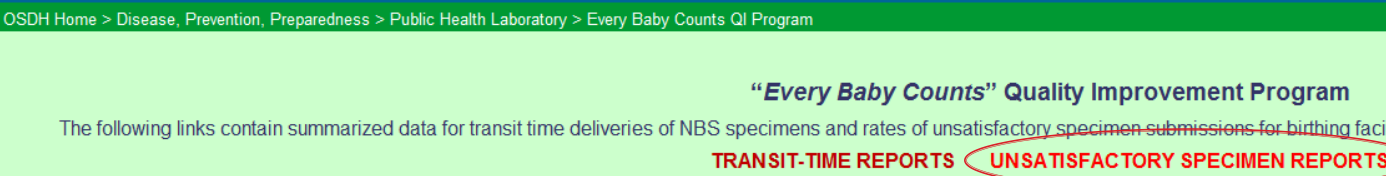


Unsat Reports – Web Access

1. 

2. 
Oklahoma State Department of Health
Contact | A-Z Health Index | Events & Meetings
Search Site
OSDH Home
Board of Health
Birth and Death Certificates
Forms, Regulations, Licensing
Disease, Prevention, Preparedness
Community & Family Health
Data and Statistics
Protective Health
Current Features
Zika Virus
Information about the Zika Virus...
West Nile Virus
Current Statistics for the West Nile Virus in Oklahoma...
Healthy Oklahoma 2020: OHIP Update
Oklahoma's strategic plan to improve health outcomes...
Every Baby Counts
A quality improvement (QI) program designed to improve submission times for newborn screening specimens...


Every Baby Counts
A quality improvement submission times for n

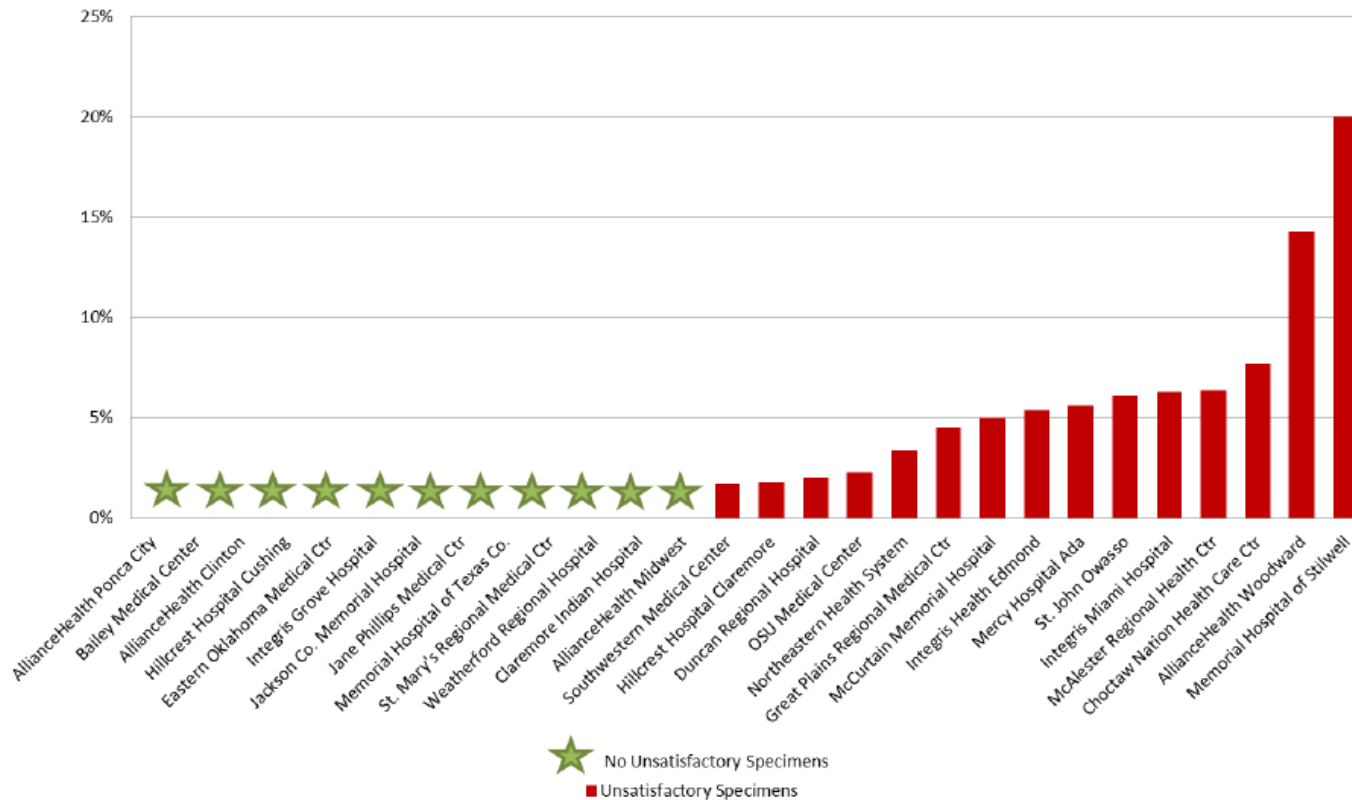
3. 
OSDH Home > Disease, Prevention, Preparedness > Public Health Laboratory > Every Baby Counts QI Program
“Every Baby Counts” Quality Improvement Program
The following links contain summarized data for transit time deliveries of NBS specimens and rates of unsatisfactory specimen submissions for birthing facilities.
TRANSIT-TIME REPORTS **UNSATISFACTORY SPECIMEN REPORTS**


UNSATISFACTORY SPECIMEN REPORTS

- ▶ Unsatisfactory reports will be emailed to representatives at each facility.

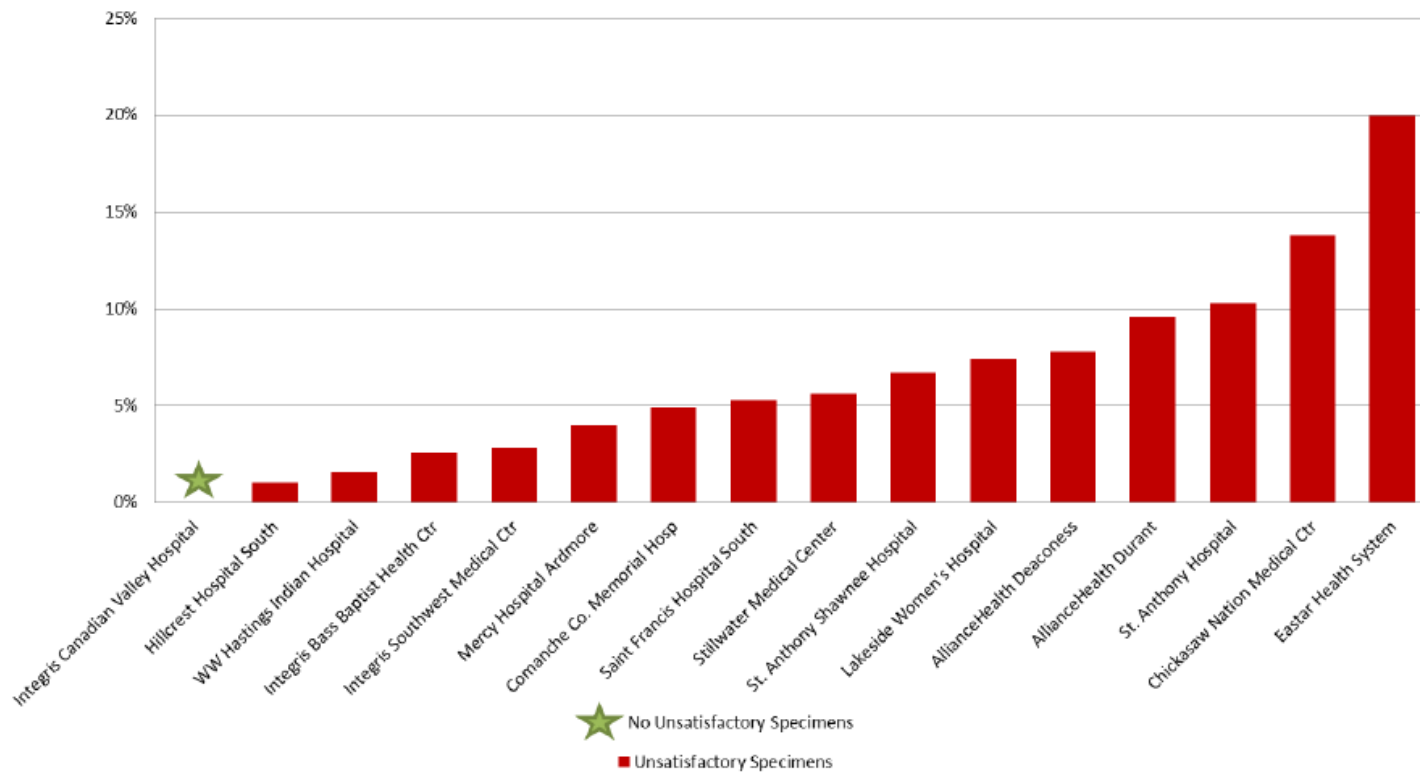
Unsat Specimen Report Example

*DEC 2016: Unsatisfactory Specimen Report
Low Volume Hospitals (Less than 731 Specimens per Year)*



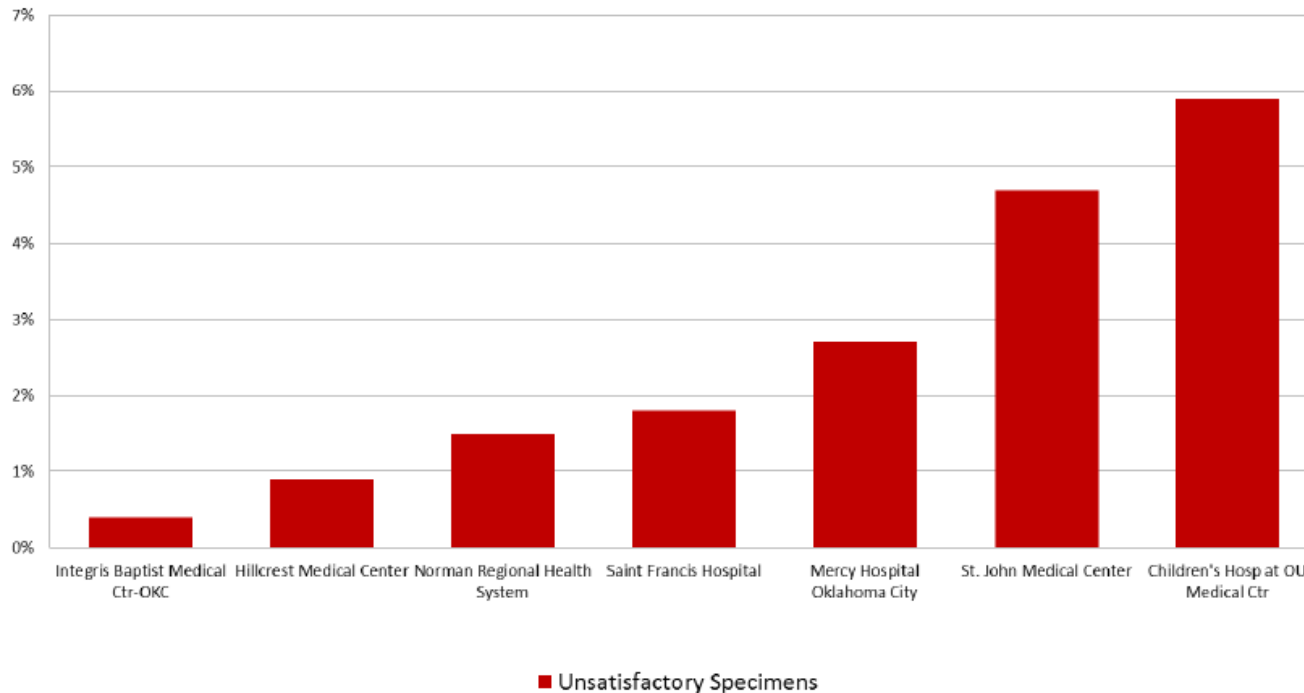
Unsat Specimen Report Example

*DEC 2016: Unsatisfactory Specimen Report
Medium Volume Hospitals (731-2499 Specimens per Year)*



Unsat Specimen Report Example

*DEC 2016: Unsatisfactory Specimen Report
High Volume Hospitals (2500 or More Specimens per Year)*



NICU & Special Considerations



Time of Screening: Premature or Sick Newborns



24 hours + 1
minute of age



14 days of age



Prior to red blood
cell transfusion,
even if collected at
<24 hours of age

Collect no later
than 3-7 days
of age



Or immediately prior to
discharge, whichever comes first



NICU Special Considerations

Factors that can interfere with screening results may include:

Infant

- Prematurity & LBW may affect TSH & 17-OHP results
- Hypoxia, CMV, septicemia, trisomies, biliary atresia may affect IRT levels
- Liver immaturity may affect amino acid results
- Carrier status may affect all NBS results

Treatment

- TPN, SNAP, & carnitine may affect amino acid, fatty acid, or organic acid results
- Steroids may affect 17-OHP results
- ECLS & blood transfusions may affect all NBS results

Maternal

- PTU therapy or radioactive iodine may affect infant TSH results
- Steroids may affect infant 17-OHP results

Collection Issues

- Contamination: oils/lotion from hands, spills, standing water, residual alcohol, heat/humidity
- Early/delayed specimen collection
- Transit time delays
- Unsatisfactory specimens

NICU Special Considerations

MELEV

(multiple elevations
of analytes)

- This result is often due to additives to feedings, such as TPN, carnitines, or MCT oil.
 - A repeat screen for an MELEV result should be collected after TPN is discontinued, at day 14 of life or prior to discharge, whichever comes first.

Transfusions

- Can affect **all** newborn screening results (especially HGB, Galactosemia, & Biotinidase Deficiency). Obtain NBS specimen prior to a blood transfusion, if possible.
 - If baby is transfused prior to initial screen, obtain two repeat screens:
 - 7 days post-transfusion &
 - 90-120 days post-transfusion



Additional Information



Hospital Responsibilities

- ▶ Ensure **all** infants are screened prior to discharge.
- ▶ Ensure specimens are received in a timely manner to the OSDH PHL for testing.
- ▶ Infants who are transferred:
 - Receiving hospital to ensure the NBS is collected.
- ▶ Submit **Satisfactory** specimens:
 - Collected properly
 - **All** requested information is documented on the filter paper
 - Submitted timely



Refusal



- ▶ Religious Tenets and Practices only
- ▶ **Check the box(es) on the filter paper form if parents refuse the NBS and/or the pulse oximetry screen.**
 - Provide parents with a NBS blood spot and/or pulse oximetry brochure(s) & answer any questions they might have about the screen(s).

A scan of a Newborn Screening (NBS) filter paper form. The form is white with a yellow vertical strip on the right side. A red circle highlights the 'Refused' checkbox in the 'SPECIMEN INFORMATION' section.A close-up of the 'SPECIMEN INFORMATION' section of the NBS filter paper form. The 'Refused' checkbox is circled in red. Below it are fields for '1. Collection Date' (MM, DD, YY) and 'Collection Time' (24 Hour Clock), and '2. Transfusion Date' (MM, DD, YY) and 'Time' (24 Hour Clock).A scan of a Newborn Screening (NBS) filter paper form. The form is white with a yellow vertical strip on the right side. A red circle highlights the 'Pulse Oximetry (COHD) Screen' section.A close-up of the 'Pulse Oximetry (COHD) Screen' section of the NBS filter paper form. The 'Refused' checkbox is circled in red. Other options include 'Not Performed', 'Pass', 'Fail', and 'Echo'.

- ▶ Ensure the parents fill out a Refusal Form. Keep a copy for baby's record & fax a copy to the NBS Program using fax # 405-271-4892.

Disorders: in Brief

The newborn screen tests for harmful or potentially fatal disorders that are not otherwise apparent at birth.



Congenital Adrenal Hyperplasia (CAH)

- Endocrine disorder of the adrenal glands resulting in a lack of the hormones cortisol and aldosterone. Cortisol protects the body during stress/illness & regulates blood glucose. This disorder is **life-threatening** & can lead to complications **within days after birth**.
- **Monitor BMPs** (for salt-wasting – hyponatremia; hyperkalemia). Low levels of sodium can lead to adrenal insufficiency & s/s resembling dehydration.
- Observe for ambiguous genitalia (females), hypospadias (males), & hyperpigmentation (males).
 - If baby has ambiguous genitalia upon birth, collect a newborn screen *immediately* & contact the Newborn Screening Program at 405-271-6617.

Signs of adrenal insufficiency:

- Lethargy
- Vomiting
- Poor feeding
- Hypovolemia
- Rapid heart rate
- Failure to Thrive
- Shock

Exposure to steroids can mask signs & symptoms of CAH

-
- ▶ **Treatment** for classic CAH requires hormonal supplementation.



Congenital Hypothyroidism (CH)

- ▶ Endocrine disorder of the thyroid gland, in which the gland fails to develop or function properly, resulting in a lack of adequate thyroid hormone production.
- ▶ A transient rise of TSH may occur physiologically in premature newborns and/or during early specimen collection (< 24 hrs of age) for premature and term newborns.

Signs of CH: *signs may not become apparent for months, after brain damage has already occurred.*

- Sluggishness
- Hypotonia
- Delayed reflexes
- Slow to feed
- Swollen tongue
- Hoarse cry
- Delayed growth
- Constipation

Untreated CH results in developmental delay & poor growth

-
- ▶ **Treatment** requires thyroid hormone supplementation



Cystic Fibrosis (CF)

- ▶ Mutations in the Cystic Fibrosis Conductance Regulator (CFTR) gene, which regulates chloride transport. Characterized by chronic pulmonary disease & gastrointestinal abnormalities.
 - ▶ Abnormal, thick mucus can obstruct airways and increase risk for recurrent pulmonary infections. May also affect the pancreas, liver, and reproductive organs.
- ▶ Check **Family History of CF** or **Meconium Ileus** on the filter paper if applicable. Mark **CFTR** under “Test Requested” if baby exhibits any other concerns for CF.
 - ▶ Doing this will result in an additional test to be performed to look for mutations for CF. Most infants with CF are born to parents who are unknowingly asymptomatic CF carriers and have NO known history of CF in the family.

Signs of CF:

- “Salty” sweat
- Meconium Ileus
- Persistent coughing/wheezing
- Thick mucus
- Loose pale, smelly stools
- Failure to thrive

-
- ▶ **Treatment** may include mucus-thinners, bronchodilators, anti-inflammatories, & antibiotics. A high-calorie diet, pancreatic enzymes, and vitamin supplements may also be indicated.

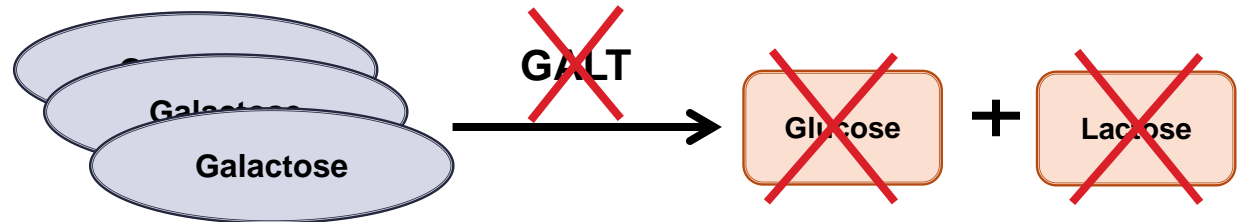


Galactosemia (GAL)

- ▶ Deficient GALT enzyme, which is required to break down the sugar galactose, which is found in all foods that contain milk. As a result, galactose accumulates in the blood & is unable to be utilized for energy. Multi-organ dysfunction can result.
- ▶ Blood transfusions can invalidate galactosemia results on the NBS. If transfused, ensure the transfusion date is recorded on the filter paper.
- ▶ Check **Lactose-Free Formula (Soy)** on the filter paper if applicable. Mark **GALT** under “Test Requested” if baby exhibits any concerns for galactosemia
- ▶ Doing this will result in an additional test to be performed to look for activity of the GALT enzyme.

Signs of Galactosemia:

- Lethargy
- Poor feeding
- Vomiting
- Seizures
- Hypoglycemia
- Failure to thrive
- Liver damage/jaundice with high direct bilirubin



Death can occur within days of birth from gram negative sepsis

- ▶ **Treatment** includes dietary management to avoid foods containing lactose and galactose.

Biotinidase (BIO) Deficiency

- Deficient activity of biotinidase enzyme, resulting in a deficiency of the biotin vitamin.
 - **Biotin:** required to break down fats, proteins, and carbohydrates.

Signs of Biotinidase Deficiency:

- Lethargy
- Hypotonia
- Vomiting/Diarrhea
- Dermatitis
- Alopecia
- Seizures
- Ketoacidosis
- Hearing loss
- Breathing problems: hyperventilation, stridor, or apnea
- Mild hyperammonemia
- Developmental delays

-
- ▶ **Treatment** includes biotin supplementation.

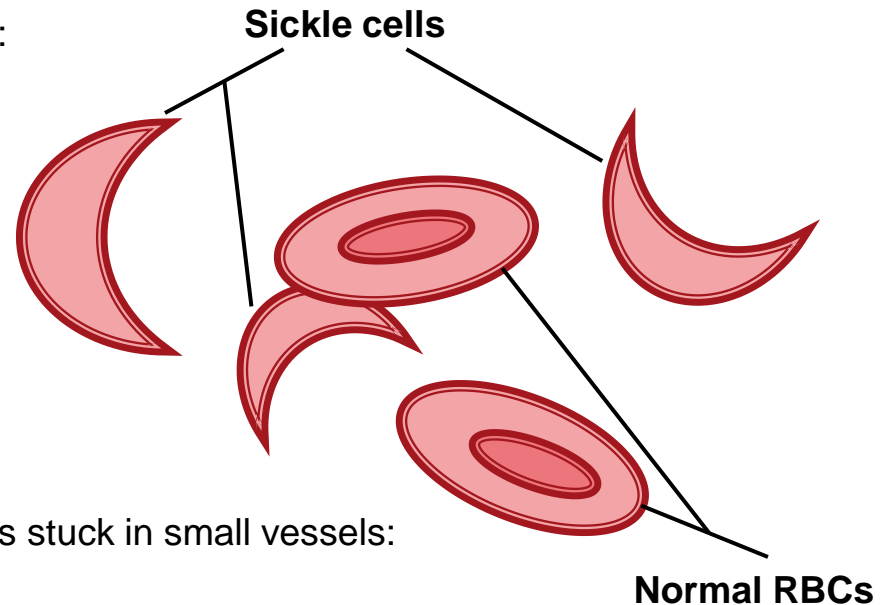


Hemoglobinopathies (HGB)

- ▶ A group of disorders that affect the number or shape of hemoglobin
 - Hemoglobin: protein in RBCs that delivers O₂ to cells throughout the body

Signs of a hemoglobinopathy:

- Anemia related to premature RBC breakdown:
 - Jaundice
 - Fatigue
 - Shortness of breath
 - Cold hands/feet
 - Pale skin
- Repeated infections:
 - Splenic damage
 - Risk for sepsis
- Intermittent pain related to stiff, inflexible RBCs stuck in small vessels:
 - O₂ deprivation to organs
 - Sickle cell crises



- ▶ **Treatment:** may include pain medication, prophylactic antibiotics, and/or blood transfusions. To avoid sickle cell crises: avoid dehydration, temperature extremes, & high altitudes.



Severe Combined Immunodeficiency (SCID)

- ▶ An immune disorder in which there is an impairment in both T & B lymphocytes
 - ▶ T lymphocytes fail to develop & B lymphocytes are either absent or compromised
- ▶ If left untreated, can lead to **life-threatening infections**
- ▶ Blood transfusions, heparin, & prematurity are factors that can affect SCID results
- ▶ Until SCID is ruled out, avoid sick contacts, pets, crowded areas (such as malls & daycares), and administering live vaccines. Mix infant formula with boiled or distilled water, not well water.

Signs of SCID:

- Frequent infections
- Infections that do not improve with antibiotic treatment
- Fungal infection (thrush) in the mouth or throat that does not go away
- Diarrhea
- Failure to thrive

-
- ▶ **Treatment:** may include immunoglobulin replacement therapy, prophylactic antibiotics, and/or bone marrow transplant.



Amino Acidopathy (AA) Disorders

- ▶ Characterized by the body's inability to metabolize certain amino acids (protein building blocks) OR the inability to detoxify ammonia through the urea cycle.
- ▶ The buildup of amino acids and/or ammonia can lead to severe medical complications, including:
 - mental retardation
 - developmental delays
 - failure to thrive
 - death
- ▶ Symptoms may not be apparent for months (PKU) to **hours/days** following birth (MSUD, CIT)



Amino Acidopathy (AA) Disorders

- ▶ Phenylketonuria (PKU)
- ▶ Citrullinemia (Cit)
- ▶ Maple Syrup Urine Disorder (MSUD)
- ▶ Homocystinuria (Hcy)
- ▶ Argininemia (Arg)
- ▶ Tyrosinemia (Tyr)

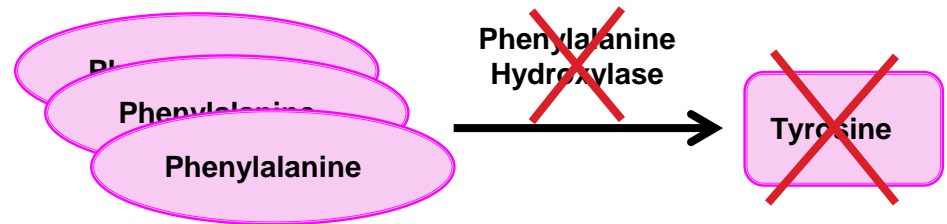


Phenylketonuria (PKU) - Amino Acidopathy

- ▶ Reduced or absent activity of *phenylalanine hydroxylase (PAH)*, an enzyme responsible for converting the amino acid phenylalanine into tyrosine. The result is a toxic buildup of phenylalanine in the blood, which can lead to irreversible brain damage. Sources of phenylalanine include protein & some artificial sweeteners.

Signs of Classic PKU: *signs may not become apparent for months, after brain damage has already occurred.*

- Irritability
- Seizures
- “Musty” or “Mouse-like” body odor
- Pale skin & hair
- Developmental delays



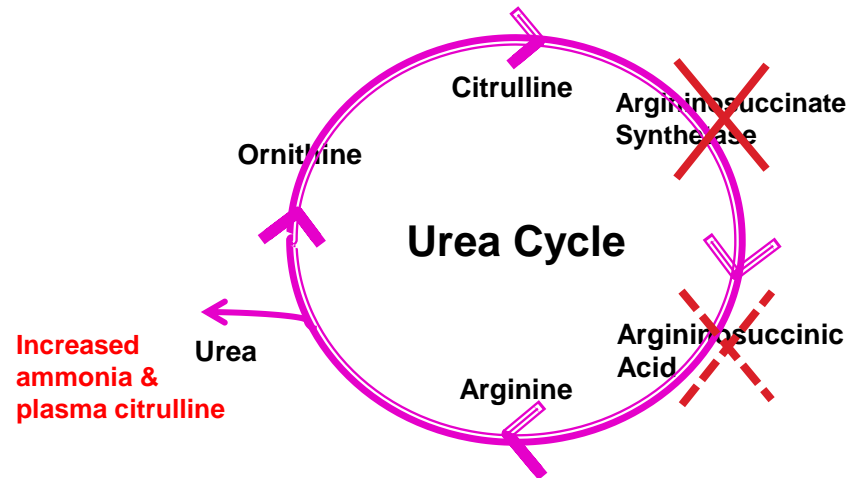
- ▶ **Treatment** requires dietary monitoring for phenylalanine-free, low protein foods & formula.

Citrullinemia (CIT) - Amino Acidopathy

- ▶ *Urea Cycle Disorder* – a **life-threatening** defect in the metabolism of waste nitrogen produced by the breakdown of protein, resulting in the toxic accumulation of ammonia during the **first few hours or days of life**.

Signs of Citrullinemia: Infants often appear normal initially but *rapidly* develop cerebral edema as well as s/s of hyperammonemia:

- Lethargy
- Poor feeding
- Hypothermia
- Seizures
- Coma
- Tachypnea
- Vomiting
- Signs of liver disease



Acute encephalopathy can occur within hours to days after birth

- ▶ **Treatment** requires dietary monitoring. Medications may be necessary to lower ammonia levels in the blood.

Fatty Acid Oxidation (FAO) Disorders

- ▶ Characterized by an enzyme defect in the fatty acid metabolic pathway, in which fats cannot be properly broken down for use in the body.
 - Stores of glucose are utilized by the body when energy is needed; however, these glucose stores are relatively small in newborns & are quickly depleted, resulting in fatty acid oxidation being turned “on” for fat breakdown for energy. But with a fatty acid disorder, certain fats are unable to be broken down and toxic byproducts accumulate. As a result, frequent feedings of glucose are crucial and special monitoring is required during periods of illness and/or stress, states in which the body requires higher energy consumption.
- ▶ Symptoms vary by disorder & are a result of the accumulation of toxic byproducts:
 - Metabolic crises:
 - Lethargy
 - Hypotonia
 - Seizures
 - Respiratory failure
 - Cardiac arrest
 - Developmental disabilities
- ▶ **Frequent feedings (every 3-4 hours) & special care during times of illness and/or stress are crucial!**
 - *Poor maternal breast milk production in the first 1-2 weeks can cause enough caloric restriction to result in sudden death of the infant within hours.*



Fatty Acid Oxidation (FAO) Disorders

- ▶ Medium-chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
- ▶ Carnitine Uptake Defect (CUD)
- ▶ Very Long-chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency
- ▶ Long-chain Acyl-CoA Dehydrogenase (LCHAD) Deficiency/Trifunctional Protein (TFP) Deficiency
- ▶ Short-chain Acyl-CoA Dehydrogenase (SCAD) Deficiency
- ▶ Carnitine Palmitoyl Transferase 1 (CPT-1) Deficiency
- ▶ Carnitine Acylcarnitine Translocatse (CACT) Deficiency/Carnitine Palmitoyl Transferase 2 (CPT-2) Deficiency

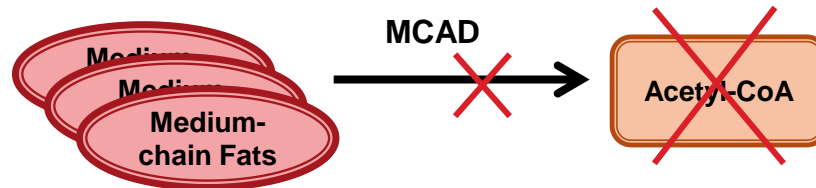


MCAD Deficiency – Fatty Acid Oxidation Disorder

- ▶ Reduced or absent activity of *MCAD* enzyme, in which medium-chain fats are unable to be broken down for energy. A **life-threatening** disorder, especially during periods of fasting when glucose stores have been depleted & also during periods of high-energy demand, such as stress & illness.
- ▶ **Ensure baby feeds routinely (every 3-4 hours) & tolerates feeds**

Signs of Metabolic Crisis:

- Irritability
- Lethargy
- Vomiting
- Seizures
- Coma
- Liver dysfunction/hepatomegaly
- Hypoglycemia with absent or 'trace' urinary ketones



- ▶ **Treatment** requires an avoidance of fasting & dietary monitoring for low-fat, high-carbohydrate foods.

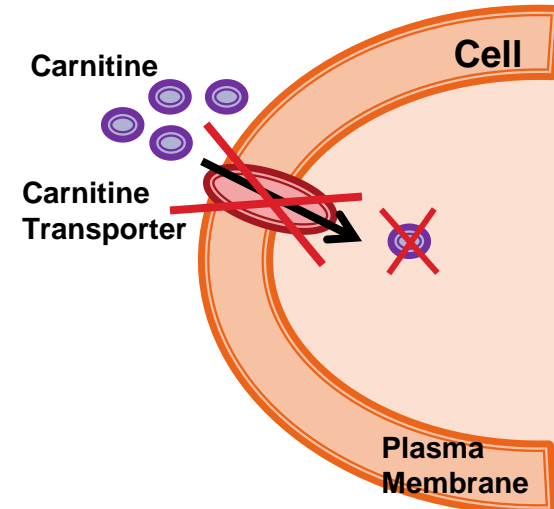


CUD – Fatty Acid Oxidation Disorder

- ▶ Defect in the carnitine transporter, which moves carnitine across the cellular plasma membrane. A **life-threatening** disorder, especially during periods of fasting when glucose stores have been depleted & also during periods of high-energy demand, such as stress & illness.
- ▶ **Ensure baby feeds routinely (every 3-4 hours) & tolerates feeds**

Signs of Metabolic Crisis:

- Irritability
- Lethargy
- Hypotonia
- Vomiting
- Cardiac decompensation
- Liver dysfunction/hepatomegaly
- Hypoglycemia



- ▶ **Treatment** requires an avoidance of fasting & dietary monitoring for low-fat, high-carbohydrate foods. L-carnitine supplementation may be indicated.

Organic Acid (OA) Disorders

- ▶ Characterized by an enzyme defect in which certain proteins are unable to be broken down, resulting in an accumulation of organic acid intermediates that become toxic.
 - Stores of glucose are utilized by the body when energy is needed; however, these glucose stores are relatively small in newborns & are quickly depleted, resulting in protein breakdown for energy. But with an organic acid disorder, certain proteins are unable to be broken down and toxic byproducts accumulate. As a result, frequent feedings of glucose are crucial and special monitoring is required during periods of illness and/or stress, states in which the body requires higher energy consumption.

- ▶ Symptoms vary by disorder & are a result of the accumulation of toxic byproducts:
 - Metabolic crises:
 - Lethargy
 - Hypotonia
 - Ketoacidosis
 - Seizures
 - Respiratory failure
 - Cardiac arrest
 - Developmental disabilities
 - Coma
 - Death

- ▶ **Frequent feedings (every 3-4 hours) & special care during times of illness and/or stress are crucial!**

Death can occur within hours to days.



Organic Acid (OA) Disorders

- ▶ Glutaryl-CoA Dehydrogenase (GA-1) Deficiency
- ▶ Propionic Acidemia (PROP)/Methylmalonic Acidemia (MMA)
- ▶ HMG/3MCC/3MBG/MCD/2M3HBA/BKT
- ▶ Malonic Acidemia (MAL)
- ▶ Isovaleric Acidemia (IVA)/2-Methylbutyrylglycinuria (2MBG)
- ▶ Isobutyryl-CoA Dehydrogenase (IBG) Deficiency

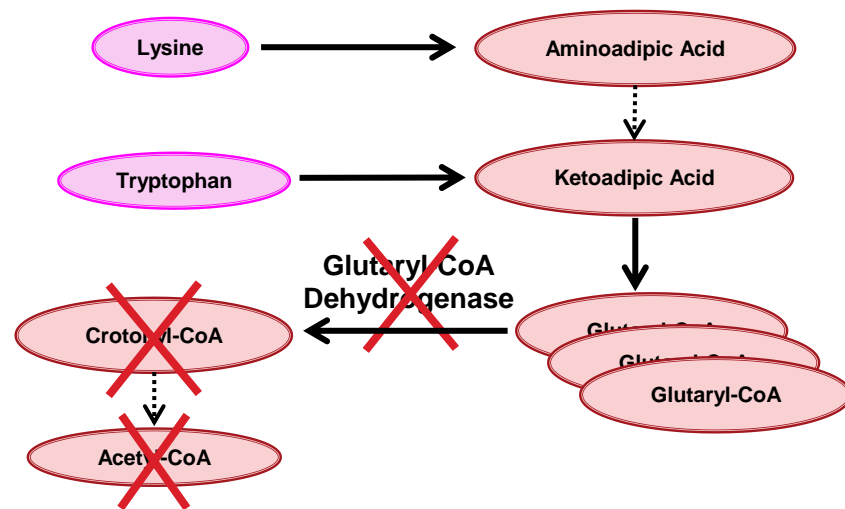


GA-1 – Organic Acid Disorder

- ▶ Reduced or absent activity of *glutaryl-CoA dehydrogenase* enzyme, in which the amino acids lysine & tryptophan are unable to be broken down. A **life-threatening** disorder, especially during periods of fasting when glucose stores have been depleted & also during periods of high-energy demand, such as stress & illness.
- ▶ **Ensure baby feeds routinely (every 3-4 hours) & tolerates feeds.**

Signs of Metabolic Crisis:

- Lethargy
- Hypotonia/muscle weakness
- Vomiting
- Seizures
- Hypoglycemia
- Macrocephaly
- Coma
- Neurological problems



- ▶ **Treatment** requires an avoidance of fasting & dietary monitoring for low-protein (lysine & tryptophan) foods & formula. L-carnitine supplementation may be indicated.

Transit Time

Prompt delivery of specimens to the Public Health Laboratory for testing can make all the difference.



Transit Time: What is it?

- “The time between the collection of a newborn screening specimen to its receipt at the OSDH Public Health Laboratory for testing.”



Transit Time

➤ Guidelines:

- Specimens should be received at the OSDH Lab within **48 hours** from the time of collection.
- Oklahoma Law: *OS 63 Sections 1-533 and 1-534*

Delays in receiving the specimen

Delays in testing the specimen

Delays in diagnosis & treatment

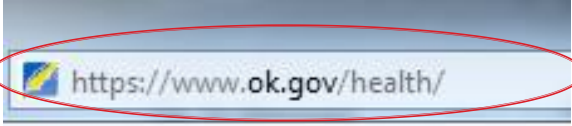



Transit Time: Tips for Improvement

- ▶ Ensure everyone involved in NBS collection/handling knows about courier pick-up time, location, and importance.
- ▶ Do not batch specimens.
- ▶ Ensure the NBS is collected at 24 hr + 1 min of age & goes out with the courier as soon as possible after it has dried (~3-4 hours of drying time).
- ▶ Set timelines and goals specific for your facility.
- ▶ Maintain a courier/transport log.
- ▶ Review transit time reports.
- ▶ Contact the PHL if the courier does not present to pick up the NBS specimens.



Transit Time Reports – Web Access

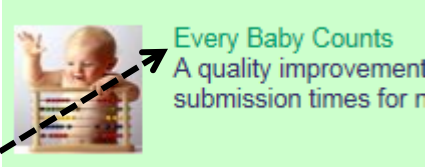
1. 

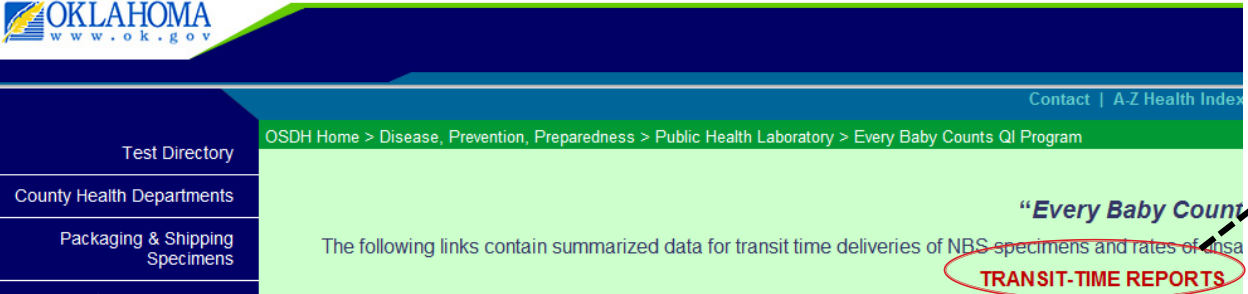
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OSDH Home

Current Features

- Zika Virus: Information about the Zika Virus...
- West Nile Virus: Current Statistics for the West Nile Virus in Oklahoma...
- Every Baby Counts: A quality improvement (QI) program designed to improve submission times for newborn screening specimens...

**Every Baby Counts**
A quality improvement submission times for n

3. 

OSDH Home > Disease, Prevention, Preparedness > Public Health Laboratory > Every Baby Counts QI Program

“Every Baby Counts”

The following links contain summarized data for transit time deliveries of NBS specimens and rates of ~~ansa~~

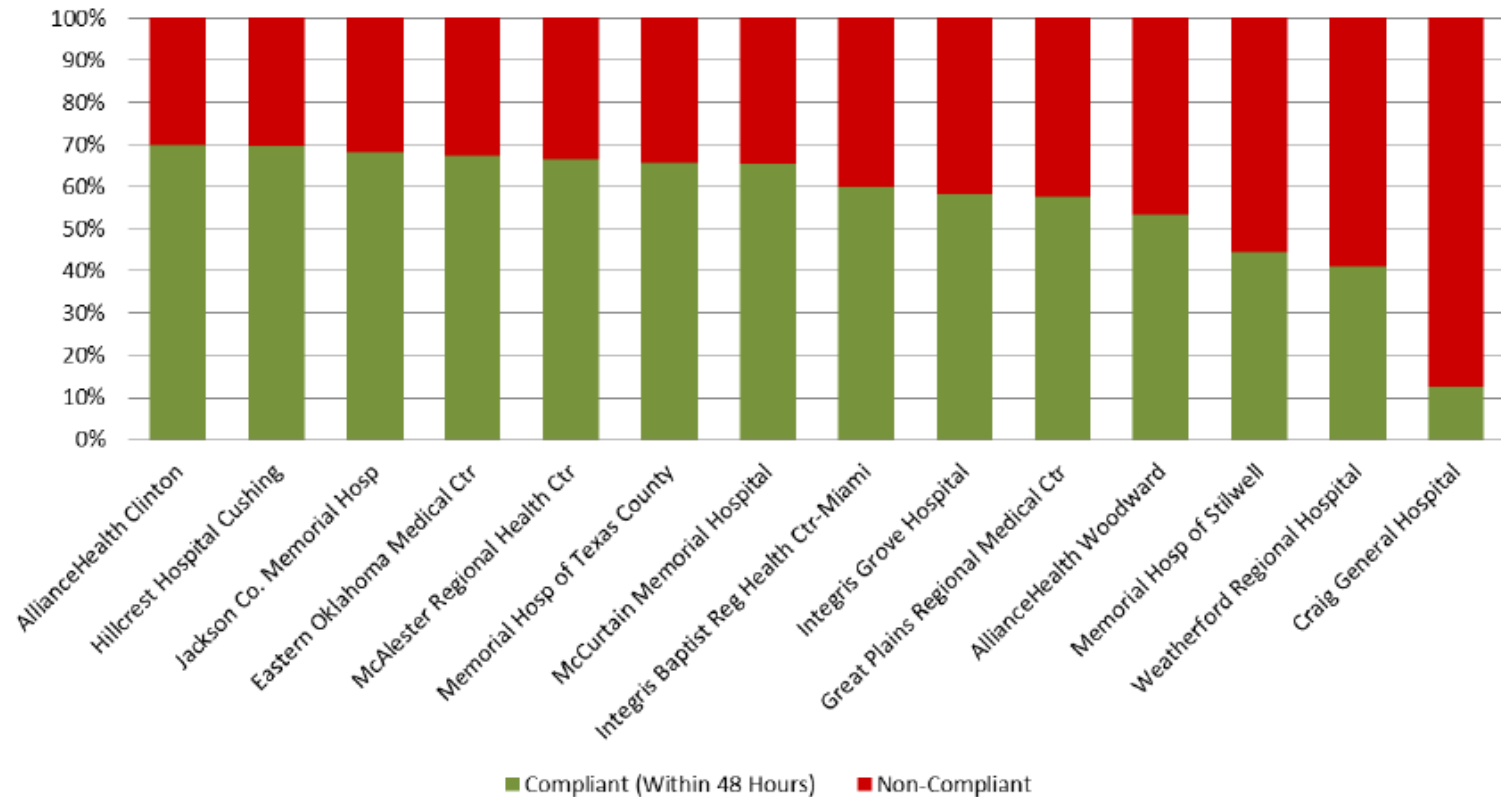
TRANSIT-TIME REPORTS

TRANSIT-TIME REPORTS

- ▶ Transit Time reports will be emailed to representatives at each facility.

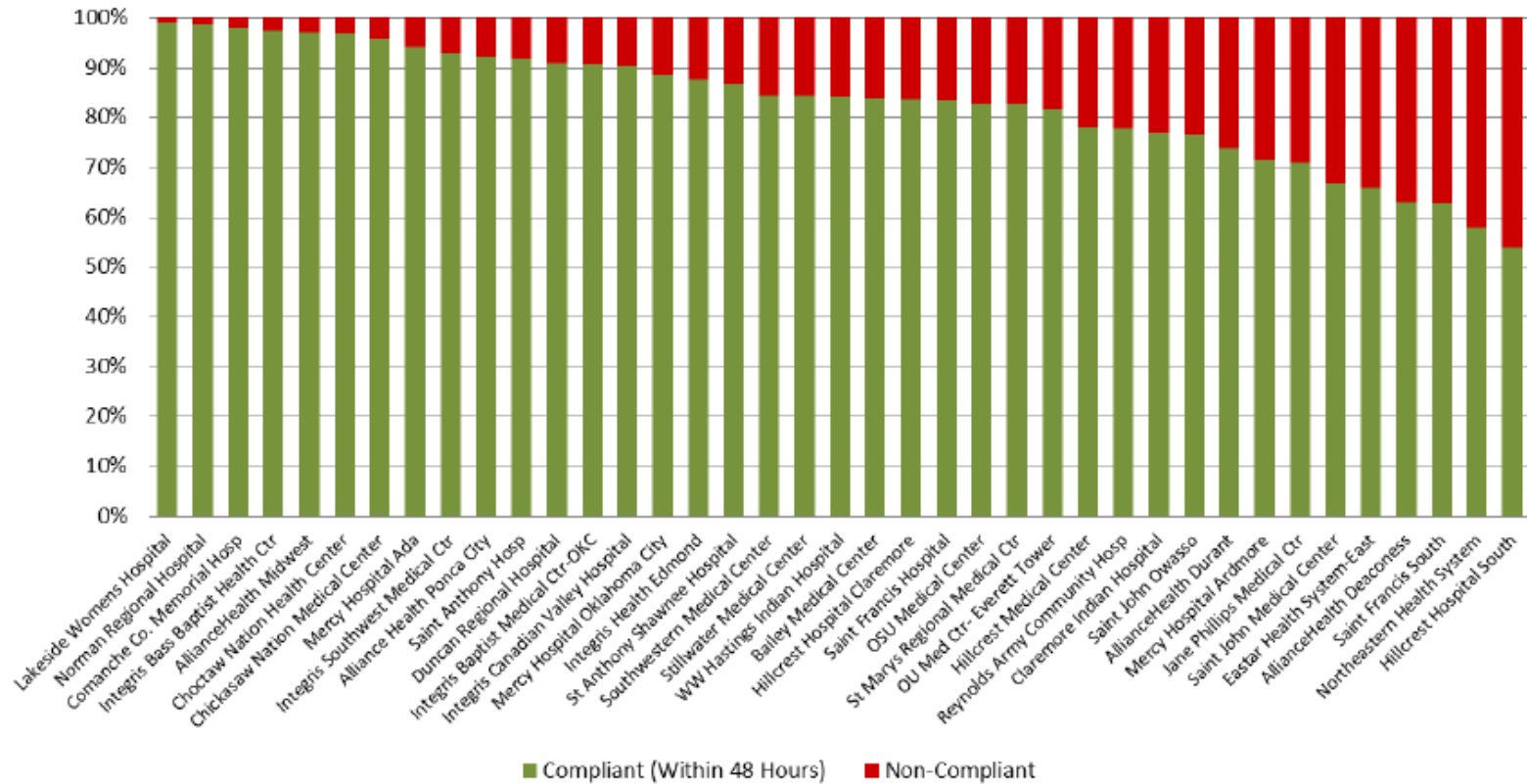
Transit Time Report Example

2016 2nd Quarter: % Compliance Ranking for Hospitals with 5-Day Courier Service



Transit Time Report Example

2016 2nd Quarter: % Compliance Ranking for Hospitals with 7-Day Courier Service



Newborn Hearing Screening



Newborn Hearing Screening

▶ Purpose:

- to screen **all** newborns prior to discharge using physiologic measure **and** risk factor checklist to identify babies who are more likely to have a hearing loss



Rationale

Waiting until a child is developmentally ready for behavioral testing diminishes language acquisition outcomes of children with a hearing loss

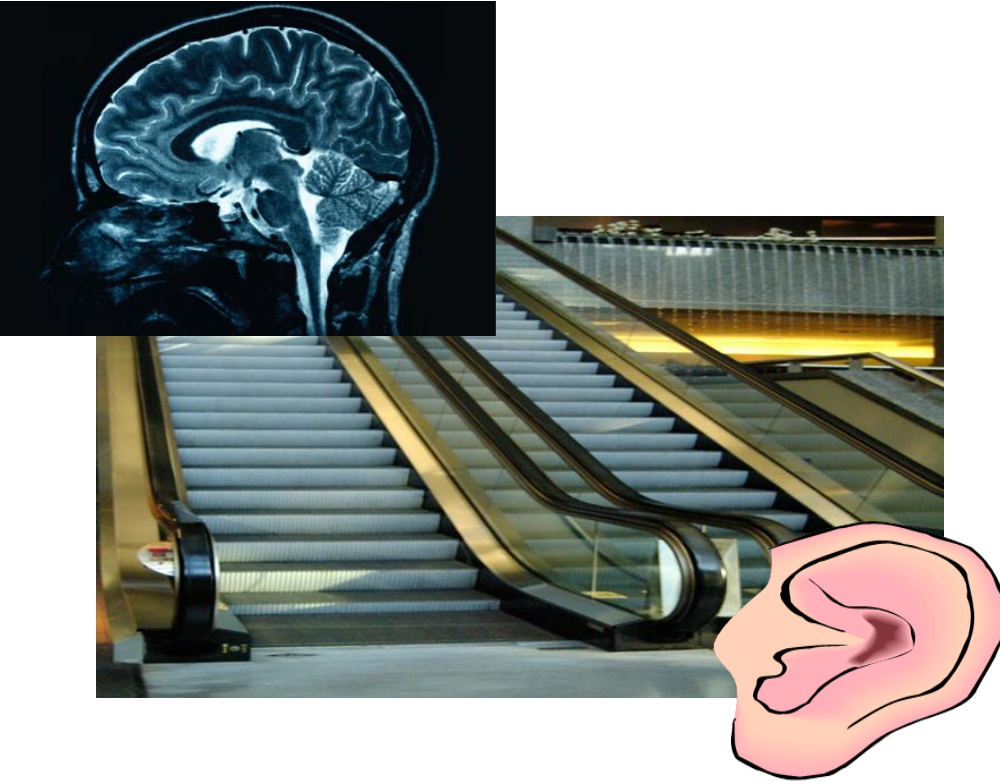


Implications

- Hearing loss is one of the **most common** birth defects
- In the United States, approximately 12,000 babies a year are born with hearing loss
 - **Approximately 180 babies per year in Oklahoma**
- An Additional 4,000-6000 who pass newborn hearing screening will acquire late onset hearing loss before age 3.



Why Universal Newborn Hearing Screening?



The Ear

- ▶ It's all about communication; the earlier the better!
- ▶ Children who are identified early and are receiving appropriate intervention of the family's choosing by 6-months of age have same or similar outcomes as children with normal hearing.
- ▶ The ear is the conveyor belt to the brain!



Hearing Risk Factors

- ▶ Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
- ▶ Infant was placed in a Level II or III nursery for more than 24 hours.
- ▶ Infant received an exchange transfusion.
- ▶ Infant had serum bilirubin level ≥ 15 mg/dL.
- ▶ Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
- ▶ Infant has craniofacial anomalies (such as pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).

The image shows a 'CHART COPY ALL RECORDS' form for newborn hearing screening. The form is divided into several sections: 'PATIENT INFORMATION', 'SPECIMEN INFORMATION', 'HEARING SCREENING RESULTS', and 'HEARING RISK STATUS'. The 'HEARING RISK STATUS' section is circled in red and contains a list of risk factors for hearing loss, which are detailed in the adjacent box.



Hearing Screening Results:

Right Ear	Left Ear	Screen Method
<input type="checkbox"/> Pass	<input type="checkbox"/> Pass	<input type="checkbox"/> ABR <input type="checkbox"/> Other (Specify) _____
<input type="checkbox"/> Refer	<input type="checkbox"/> Refer	<input type="checkbox"/> OAE

If not screened, reason:

<input type="checkbox"/> Technical problem	<input type="checkbox"/> No equipment	<input type="checkbox"/> Delayed
<input type="checkbox"/> Caregiver refused	<input type="checkbox"/> Baby discharged	<input type="checkbox"/> Other _____

Hearing risk status—Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
- Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
- Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
- Infant had exchange transfusion.
- Infant has serum bilirubin level ≥ 15 mg/dL.
- Infant was placed in a Level II or III nursery for more than 24 hours.

Conducting the Screen

- » Preparation of the baby and how to troubleshoot common problems



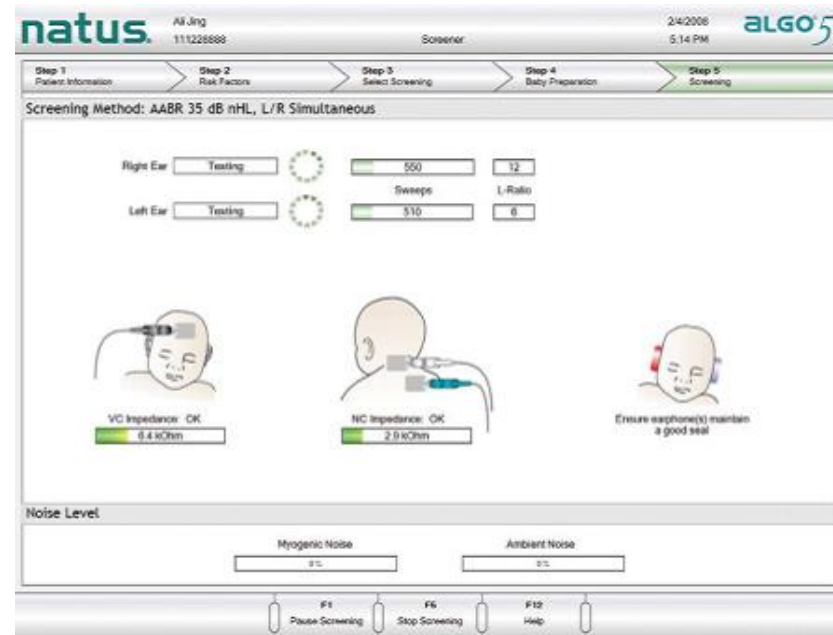
Performing the Screen: Points to Remember

- ▶ At least 5-6 hours after delivery
- ▶ Not immediately following a bath
 - Baby needs to be completely dry with no lotion applied to skin
- ▶ Quiet environment, away from other machines
- ▶ Quiet, still baby; swaddled if possible
- ▶ 2 screens before discharge (if needed):
 - Not immediately following each other
 - No more than 3 screens (only used if baby is fussy during screen and has to start again)
- ▶ Additional information can be found in the Natus user manual



Preparing to Screen

- ▶ Clean and prepare the skin (NuPrep, Alcohol)
- ▶ Do not use gel; may use small dab of water
- ▶ Proper Electrode placement:
 - Nape/neck = **WHITE** collar
 - High forehead (vertex) = **BLACK** top hat
 - Shoulder/cheek (common) = **GREEN** common
- ▶ Connect leads with the snap/alligator clips
- ▶ Place earphone over each ear
 - (Right on **Red**, “Left over” on **Blue**)



Potential Problems During Screening

- ▶ High impedances
 - Skin preparation
 - Equipment check

- ▶ Myogenic (Muscle) Noise
 - Feed baby
 - Swaddled
 - Neck rolls
 - Pacifier



Troubleshooting

▶ **Electrical Noise**

- Electrical outlets
- Change room location
- Check placement of cords
- Note: Monitors/cell phone can cause interference
- If the AABR is taking more than 12-15 minutes to complete, stop screening and troubleshoot.

▶ **Also check the following:**

- Check for sound coming through earphones.
- Check placement of earphones and make sure they fit snugly.
- Check electrode placement and ensure they are connected.
- Check to make sure cables are working properly by running an equipment check (using the clear box/docking station) – if not, get new cables as soon as possible.



Natus Information

- ▶ **Natus video and manual accompanying screening unit**
- ▶ **Technical Support**

Natus Technical Support

(650) 802-0400

(888) 496-2887 (Toll free)

technical_service@natus.com

Hours: 24/7



Reporting Results

- »» How to send correct information in compliance with Oklahoma law



HEARING SCREENING INSTRUCTIONS

Hearing screening is to be completed with results recorded and forwarded to the Oklahoma State Department of Health at the same time as the blood specimen. Follow the instructions below:

- 1 Screen the infant's hearing using the available technology.
- 2 Record results in the **Hearing Screening Results** area on the front page of the form. Place a check mark in the appropriate Pass or Refer box for the right ear and the left ear.
- 3 Indicate the method used to screen hearing (ABR, OAE, Other). If "Other" is checked, specify the technology used.
- 4 **If hearing cannot be screened**, check the appropriate box for the reason; **if screening will be delayed**, follow instructions below.
- 5 Complete the **Hearing risk status** indicator section by placing a check mark in the box of any item that applies to this infant. The first question about familial hearing loss is to be asked of the birth mother. Information for the other indicators should be available in the infant's chart.
- 6 Detach and give the Newborn Hearing Screening parent form (pink sheet) to the infant's parent or guardian at discharge.

DO NOT DELAY SENDING THE BLOOD SPECIMEN. ALL BLOOD SPECIMENS MUST BE SENT WITHIN 24 HOURS OF COLLECTION.

For infants whose hearing screening cannot be completed by the time the blood specimen must be sent (including those transferred within the facility) and it is anticipated hearing will be screened prior to discharge, do the following:

- 1 On the original form in the **If not screened, reason:** area, mark the "Delayed" box.
- 2 Complete the Hearing risk status section. For infants placed in "special care" nursery, be sure to mark the **Infant was placed in a Level II or III nursery for more than 24 hours** box. **Be certain there are no marks in the Screen Method box.**
- 3 Detach and retain the parent's copy of the hearing screening form (pink sheet). It will be used to record hearing screening results.
- 4 Be sure that the infant's last and first names are legible on the detached document.
- 5 Mail blood specimen.
- 6 Perform the hearing screening prior to discharge.
- 7 Record the results as indicated above in the appropriate boxes on the pink parent copy.
- 8 Mark any appropriate boxes in the Hearing risk status area if this has not already been completed.
- 9 Photocopy the front of the completed form (pink sheet). **Be certain that infant's name and the form's serial number are legible on photocopy.**
- 10 Mail the photocopy to OSDH, Public Health Laboratory Service, PO Box 24106, Oklahoma City, OK 73124-0106.
- 11 Give the completed pink sheet to the infant's parent or guardian.





Hearing Results Section

Oklahoma State Department of Health

Newborn Hearing Screening

OKLAHOMA
NEWBORN
HEARING
SCREENING
PROGRAM

Baby's Last Name **Baby's First Name**

THE NEWBORN HEARING SCREENING TEST
 Newborn hearing screening checks to see if your baby's hearing is okay. Good hearing is important for speech/language development. Hearing problems need to be identified as early as possible. If your baby has a hearing loss, steps can be taken to help your baby develop communication.

CAN YOUR NEWBORN HEAR?
 Your baby's nurse or doctor can tell you the hearing screening results. The screening results also are shown in the box below where it says Hearing Screening Results. Look for check marks in the "Pass" boxes. If there is a mark in each "Pass" box, your baby's hearing was okay. If your baby gets a "Refer" for one or both ears, more testing is needed. Your baby's doctor may refer you to an audiologist for additional testing. An audiologist is a hearing specialist. *If for some reason your baby's hearing was not screened, please call 1-800-766-2223 or 405-271-6617 to ask about a location close to you where hearing can be checked.*

IF YOUR BABY PASSES THE SCREENING, WILL HEARING NEED TO BE TESTED AGAIN?
 Perhaps. There are some conditions that cause hearing loss later in life. One is a family history of deafness. Others include various illnesses or conditions at birth. If there is a check mark in any of the boxes under "Hearing risk status" it is recommended that hearing be checked again by six months of age.

QUESTIONS ABOUT HEARING OR WHERE TO HAVE YOUR BABY'S HEARING CHECKED?
 Please call the Newborn Screening Program for answers. The toll-free number is 800-766-2223. The Oklahoma City metropolitan area number is 271-6617. The phone is answered Monday through Friday from 8:00 AM until 5:00 PM. E-mail: newbornscreen@health.ok.gov

Hearing Screening Results:

Right Ear	Left Ear	Screen Method	
<input type="checkbox"/> Pass	<input type="checkbox"/> Pass	<input type="checkbox"/> ABR	<input type="checkbox"/> Other (Specify) _____
<input type="checkbox"/> Refer	<input type="checkbox"/> Refer	<input type="checkbox"/> OAE	

If not screened, reason:

<input type="checkbox"/> Technical problem	<input type="checkbox"/> No equipment	<input type="checkbox"/> Delayed
<input type="checkbox"/> Caregiver refused	<input type="checkbox"/> Baby discharged	<input type="checkbox"/> Other _____

Hearing risk status—Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
- Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
- Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
- Infant had exchange transfusion.
- Infant has serum bilirubin level \geq 15 mg/dL.
- Infant was placed in a Level II or III nursery for more than 24 hours.

ATTENTION PROVIDER

DETACH AND GIVE TO PARENT OR GUARDIAN

DETACH AND GIVE TO PARENT OR GUARDIAN
NEWBORN HEARING SCREENING

1504928

Instruct parents to ask for screen results on first visit to PCP.
 Tell parents to bring the **Blue** or **Pink** slip to their baby's first doctor's visit.

Filter Paper: Marking Practice

Hearing Screening Results:

Right Ear

Pass

Refer

Left Ear

Pass

Refer

Screen Method

ABR

OAE

Other (Specify) _____

If not screened, reason:

Technical problem

Caregiver refused

No equipment

Baby discharged

Delayed

Other _____

Hearing risk status – Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
- Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
- Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
- Infant had exchange transfusion.
- Infant has serum bilirubin level ≥ 15 mg/dL.
- Infant was placed in a Level II or III nursery for more than 24 hours.



Example 1: Pass Bilaterally, No Risk

Hearing Screening Results:

Right Ear
 Pass
 Refer

Left Ear
 Pass
 Refer

Screen Method
 ABR Other (Specify)_____
 OAE

If not screened, reason:

Technical problem No equipment Delayed
 Caregiver refused Baby discharged Other_____

Hearing risk status – Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
- Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
- Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
- Infant had exchange transfusion.
- Infant has serum bilirubin level ≥ 15 mg/dL.
- Infant was placed in a Level II or III nursery for more than 24 hours.

Example 2: Pass/Refer; No Risk

Hearing Screening Results:

Right Ear
 Pass
 Refer

Left Ear
 Pass
 Refer

Screen Method
 ABR Other (Specify)_____
 OAE

If not screened, reason:

Technical problem No equipment Delayed
 Caregiver refused Baby discharged Other_____

Hearing risk status – Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
- Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
- Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
- Infant had exchange transfusion.
- Infant has serum bilirubin level ≥ 15 mg/dL.
- Infant was placed in a Level II or III nursery for more than 24 hours.

Example 3: Not Screened, NICU

Hearing Screening Results:

Right Ear
 Pass
 Refer

Left Ear
 Pass
 Refer

Screen Method
 ABR Other (Specify)_____
 OAE

If not screened, reason:

Technical problem No equipment Delayed
 Caregiver refused Baby discharged Other_____

Hearing risk status – Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
- Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
- Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
- Infant had exchange transfusion.
- Infant has serum bilirubin level ≥ 15 mg/dL.
- Infant was placed in a Level II or III nursery for more than 24 hours.

Example 4: Equipment Malfunction, Risk

Hearing Screening Results:

Right Ear
 Pass
 Refer

Left Ear
 Pass
 Refer

Screen Method
 ABR Other (Specify)_____
 OAE

If not screened, reason:

Technical problem No equipment Delayed
 Caregiver refused Baby discharged Other_____

Hearing risk status – Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
- Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
- Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
- Infant had exchange transfusion.
- Infant has serum bilirubin level ≥ 15 mg/dL.
- Infant was placed in a Level II or III nursery for more than 24 hours.

Example 5: Bilateral Refer, Multiple Risk Factors

Hearing Screening Results:

Right Ear

Pass

Refer

Left Ear

Pass

Refer

Screen Method

ABR Other (Specify)_____

OAE

If not screened, reason:

- Technical problem No equipment Delayed
 Caregiver refused Baby discharged Other_____

Hearing risk status - Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
 Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
 Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
 Infant had exchange transfusion.
 Infant has serum bilirubin level ≥ 15 mg/dL.
 Infant was placed in a Level II or III nursery for more than 24 hours.

Example 6: One Ear Screened

Hearing Screening Results:

Right Ear

Pass

Refer

Left Ear

Pass

Refer

Screen Method

ABR Other (Specify)_____

OAE

If not screened, reason:

- Technical problem No equipment Delayed
 Caregiver refused Baby discharged Other_awake__

Hearing risk status - Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
 Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
 Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
 Infant had exchange transfusion.
 Infant has serum bilirubin level ≥ 15 mg/dL.
 Infant was placed in a Level II or III nursery for more than 24 hours.

Making Corrections

- ▶ Use single line through incorrect mark
- ▶ Print "error"
- ▶ Initial the change

Right Ear

~~X Pass~~ error SS

Refer

Left Ear

Pass

Refer

Screen Method

ABR

OAE

Filter Paper Already Submitted?

- ▶ **Never delay submitting the filter paper**
 - Send the specimen promptly to the OSDH Public Health Laboratory via the courier system (i.e., **within 24 hours of collection**) for testing.
 - Some babies can pass away within the first seven days of life if not treated for some of the rare disorders we identify.
 - See instructions on insert of the filter paper (page where the circles are located).



How to Report Hearing Results after the Filter Paper has been Submitted

▶ **Never delay the bloodspot filter paper**

- Call the Newborn Hearing Screening Program with updated results: 405-271-9444 ext. 56741
- Email updated results to our office: NewbornScreen@health.ok.gov
- Fax copy of updated results on copy of filter paper to our office: 405-271-4892
- Fax a copy of the updated results label to our office: 405-271-4892



Giving Results to Parents

- » Communicating effectively, clearly, and helpfully





Relaying Results to Parents

Oklahoma State Department of Health

Newborn Hearing Screening

OKLAHOMA
NEWBORN
HEARING
SCREENING
PROGRAM

THE NEWBORN HEARING SCREENING TEST
Newborn hearing screening checks to see if your baby's hearing is okay. Good hearing is important for speech/language development. Hearing problems need to be identified as early as possible. If your baby has a hearing loss, steps can be taken to help your baby develop communication.

CAN YOUR NEWBORN HEAR?
Your baby's nurse or doctor can tell you the hearing screening results. The screening results also are shown in the box below where it says Hearing Screening Results. Look for check marks in the "Pass" boxes. If there is a mark in each "Pass" box, your baby's hearing was okay. If your baby gets a "Refer" for one or both ears, more testing is needed. Your baby's doctor may refer you to an audiologist for additional testing. An audiologist is a hearing specialist. *If for some reason your baby's hearing was not screened, please call 1-800-766-2223 or 405-271-6617 to ask about a location close to you where hearing can be checked.*

IF YOUR BABY PASSES THE SCREENING, WILL HEARING NEED TO BE TESTED AGAIN?
Perhaps. There are some conditions that cause hearing loss later in life. One is a family history of deafness. Others include various illnesses or conditions at birth. If there is a check mark in any of the boxes under "Hearing risk status" it is recommended that hearing be checked again by six months of age.

QUESTIONS ABOUT HEARING OR WHERE TO HAVE YOUR BABY'S HEARING CHECKED?
Please call the Newborn Screening Program for answers. The toll-free number is 800-766-2223. The Oklahoma City metropolitan area number is 271-6617. The phone is answered Monday through Friday from 8:00 AM until 5:00 PM. E-mail: newbornscreen@health.ok.gov

Hearing Screening Results:

Right Ear	Left Ear	Screen Method	
<input type="checkbox"/> Pass	<input type="checkbox"/> Pass	<input type="checkbox"/> ABR	<input type="checkbox"/> Other (Specify) _____
<input type="checkbox"/> Refer	<input type="checkbox"/> Refer	<input type="checkbox"/> OAE	

If not screened, reason:

<input type="checkbox"/> Technical problem	<input type="checkbox"/> No equipment	<input type="checkbox"/> Delayed
<input type="checkbox"/> Caregiver refused	<input type="checkbox"/> Baby discharged	<input type="checkbox"/> Other _____

Hearing risk status—Check all that apply:

- Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
- Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
- Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
- Infant had exchange transfusion.
- Infant has serum bilirubin level \geq 15 mg/dL.
- Infant was placed in a Level II or III nursery for more than 24 hours.

**ATTENTION
PROVIDER**

**DETACH AND
GIVE TO
PARENT
OR GUARDIAN**

DETACH AND GIVE TO PARENT OR GUARDIAN
NEWBORN HEARING SCREENING

1504928

Instruct parents to ask for screen results on first visit to PCP.
Tell parents to bring the **Blue** or **Pink** slip to their baby's first doctor's visit.

Sharing Information: Special Considerations

- ▶ Try to be mindful of how much the parents are understanding and of their possible emotional reaction.
- ▶ Emphasize how important it is that the parents schedule a follow-up appointment for their baby, if needed.
- ▶ Ensure the family knows what to do next.



Result: Pass

PASS RESULT (95% of your babies)

- ✓ Your baby passed the hearing screening today.
- ✓ Your baby's hearing is critical for normal speech and language development.
- ✓ It is important that you speak to your baby's doctor who can help you in knowing if your baby should have further tests with a pediatric audiologist.
- ✓ Your PCP can also help you to monitor for normal speech and language development.
- ✓ At any time if you are concerned with your child's hearing or speech, ask your PCP -"When in doubt, check it out."

Do NOT say:

- ✓ Your baby passed with flying colors.

Always remember to report any hearing screening results to the Newborn Hearing Screening Program (NHSP).

Result: Refer

REFER RESULT

- ✓ Your baby did not pass the hearing screening today.
- ✓ That means that your baby needs to have some more testing done so we can make sure they are hearing well enough for on-time speech and language development.
 - Refer to a county health department, audiology program, AND/OR refer to their baby's primary care provider.
- ✓ This does not mean that your baby cannot hear.

Do NOT say:

- ✓ Your baby did not pass the hearing screening today but it's probably because they've got fluid in their ears.
- ✓ Your baby did not pass the hearing screening on the (left/right) ear today, but all the babies have been referring on that ear so it's probably just the machine.

Always remember to report any hearing screening results to the Newborn Hearing Screening Program (NHSP).



Result: Pass at Risk

PASS AT RISK RESULT

- ✓ Your baby passed the hearing screening today, but because of risk factor(s), national recommendations are that your baby's hearing be checked again at 6 months of age.
 - Delayed onset hearing loss can occur after birth and are associated with risk factors. Explain which risk factors are present.
- ✓ Your baby's hearing is critical for normal speech and language development.
- ✓ When your baby is 5 months of age, you and the PCP on record will get a letter reminding you to get your baby's hearing screened again. You can choose if you would like to go to your local county health department or to a private practice provider.
- ✓ Your PCP can also help you to monitor for normal speech and language development.
- ✓ At any time if you are concerned with your child's hearing or speech, ask your PCP --"When in doubt, check it out."

Always remember to report any hearing screening results to the Newborn Hearing Screening Program (NHSP).

Pulse Oximetry Screening



Pulse Oximetry Screening

▶ Purpose:

- to screen **all** newborns between 24-48 hours of life with pulse oximetry to detect select defects related to critical congenital heart disease.

Rationale

- ▶ Some newborns may appear healthy at first *despite* having a CCHD. Early detection and prompt treatment can prevent lifelong disability and early death.

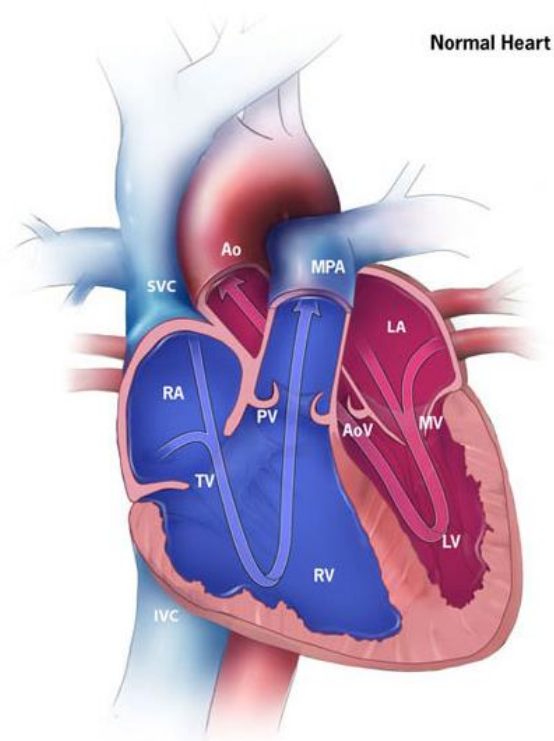


Implications

- Congenital heart disease is the **most common** birth defect
- 1 in 110 infants will have a heart defect
 - **25%** of those cases will have a CCHD
- Most affected are asymptomatic early on
- Most will require surgery shortly after birth



Normal Heart: Blood Flow



Normal Heart

RA. Right Atrium
RV. Right Ventricle
LA. Left Atrium
LV. Left Ventricle

SVC. Superior Vena Cava
IVC. Inferior Vena Cava
MPA. Main Pulmonary Artery
Ao. Aorta

TV. Tricuspid Valve
MV. Mitral Valve
PV. Pulmonary Valve
AoV. Aortic Valve

Image credit: CDC (2014)

Blood from body tissues goes to the right side of the heart and enters the lungs, where the blood becomes oxygenated. The blood is then delivered to the left side of the heart, which is responsible for pumping the oxygenated blood out to the body in order to provide oxygenation to the body tissues. After being utilized, the deoxygenated blood is returned to the right side of the heart, and the cycle continues. Valves within the heart help to prevent backflow of blood during this process.

Fetal openings between the atria, ventricles, and blood vessels begin to close shortly after birth.

Fetal-Neonatal Circulation

- The first *breath of life* leads to important changes in neonatal circulation:
 - Makes way for use of neonatal lungs (The lungs were not utilized in utero, as the placenta provided oxygenation to the fetus; after birth, however, an enormous amount of pressure is necessary in order for the newborn to close the diversions used to bypass the lungs in utero and instead allow for use of the lungs.)
 - Increased pressure change in the left side of heart compared to the right (The left side becomes the body's "pump") resulting in:
 - Closure of the Ductus Arteriosus (fetal opening between aorta and pulmonary artery)
 - Closure of the Foramen Ovale (fetal opening between the right and left atria)

❖ *Failure of closure of fetal openings can result in complications*



CCHD: Screening Targets & Symptomatology



CCHD Targets

Most likely detected by pulse oximetry screening

- ▶ Hypoplastic Left Heart Syndrome (HLHS)
 - ▶ Pulmonary Atresia
 - ▶ Tetralogy of Fallot
 - ▶ Total Anomalous Pulmonary Venous Return
 - ▶ Transposition of the Great Arteries
 - ▶ Tricuspid Atresia
 - ▶ Truncus Arteriosus
- *These heart defects lead to low levels of oxygen in the blood.*



CCHD Targets

Potentially detected by pulse oximetry screening

- ▶ Double Outlet Right Ventricle (DORV)
 - ▶ Ebstein's Anomaly
 - ▶ Coarctation of the Aortic Arch
 - ▶ Interruption of the Aortic Arch
 - ▶ Single Ventricle
- *Also potentially detected by pulse oximetry screening: other hypoxic cardiac or non-cardiac conditions.*



CCHD: What to Watch For

▶ **Signs:**

- Cyanosis
- Tachypnea
- Increased work of breathing
- Swelling
- Tires easily during feeds
- Sweating
- Poor weight gain

➤ **If at any time, the newborn should become symptomatic, the family should *immediately* take the baby to the closest emergency room for evaluation.**



CCHD: Heart Defects Review



CCHD: A Closer Look

▶ Hypoplastic Left Heart Syndrome (HLHS):

Underdevelopment of the left side of the heart

- ▶ Underdeveloped aorta
 - ▶ Underdeveloped left ventricle
 - ▶ Large Patent Ductus Arteriosus (PDA) supplying the blood flow to the body
 - ▶ Atrial Septal Defect (ASD) allowing blood return from lungs to the single ventricle
-
- ▶ The left ventricle is the heart's “powerhouse” that pumps the blood from the heart to the **entire** body. Imagine how difficult it would be to keep the body's tissues oxygenated when the body's powerhouse is underdeveloped!

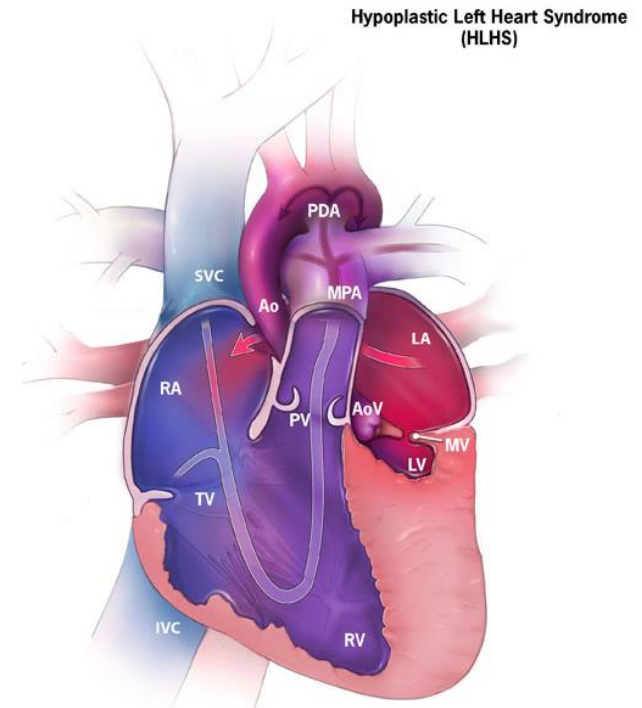


Image credit: CDC (2014)

CCHD: A Closer Look

- ▶ **Pulmonary Atresia:**
 - Defect of the pulmonary valve in which the valve failed to form and no blood can pass through to get from the heart to the lungs
 - Presence of a patent ductus arteriosus (PDA) allows for shunting of blood

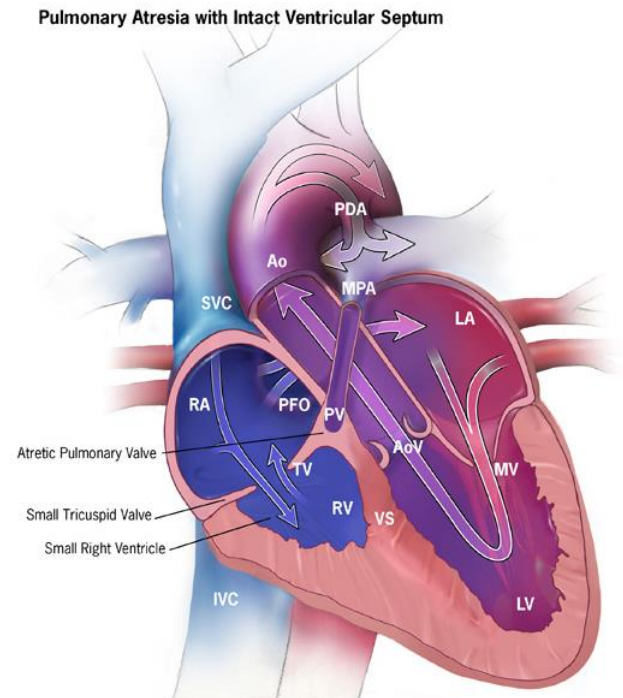


Image credit: CDC (2014)

CCHD: A Closer Look

▶ Tetralogy of Fallot:

Combination of four heart defects

- ▶ Pulmonary stenosis
- ▶ Right ventricular hypertrophy
- ▶ Overriding aorta (the aorta arises from both ventricles as opposed to solely the left ventricle)
- ▶ Ventricular septal defect (VSD), resulting in right-to-left shunting of blood

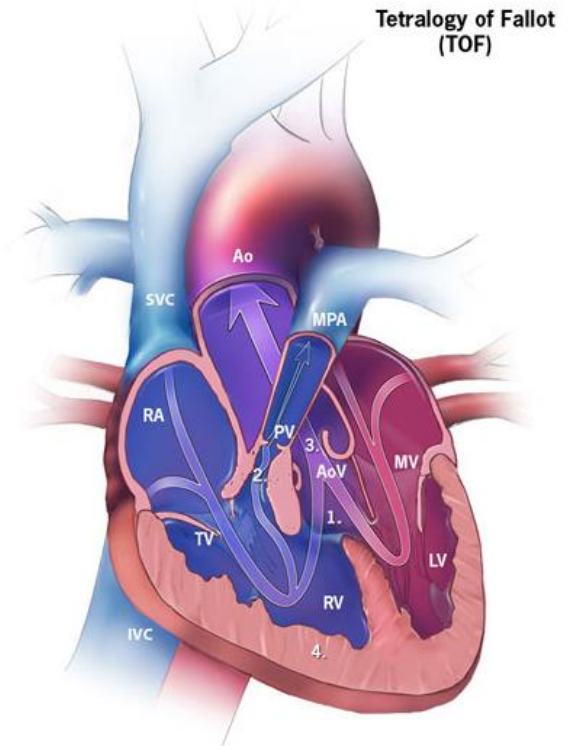


Image credit: CDC (2014)

CCHD: A Closer Look

- ▶ **Total Anomalous Pulmonary Venous Return:**
- Defect in which the pulmonary veins, which carry oxygenated blood from the lungs back to the heart, do not connect to the heart's left atrium like normal but instead go to the heart via abnormal routes

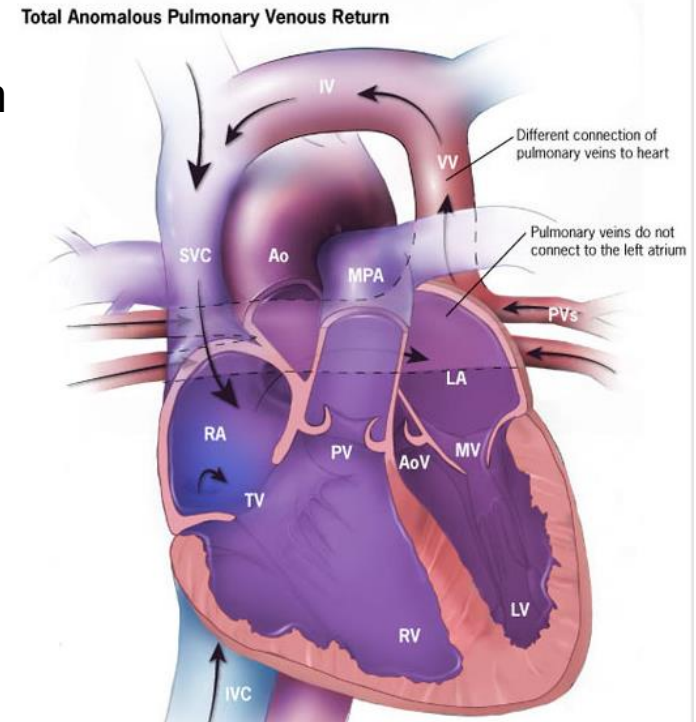


Image credit: CDC (2014)

CCHD: A Closer Look

- ▶ **Transposition of the Great Arteries/Vessels:**
 - Defect in which the aorta and the pulmonary artery are switched (“transposed”), resulting in deoxygenated blood being pumped to the body and bypassing the lungs while oxygenated blood travels from the lungs to the heart and back to the lungs

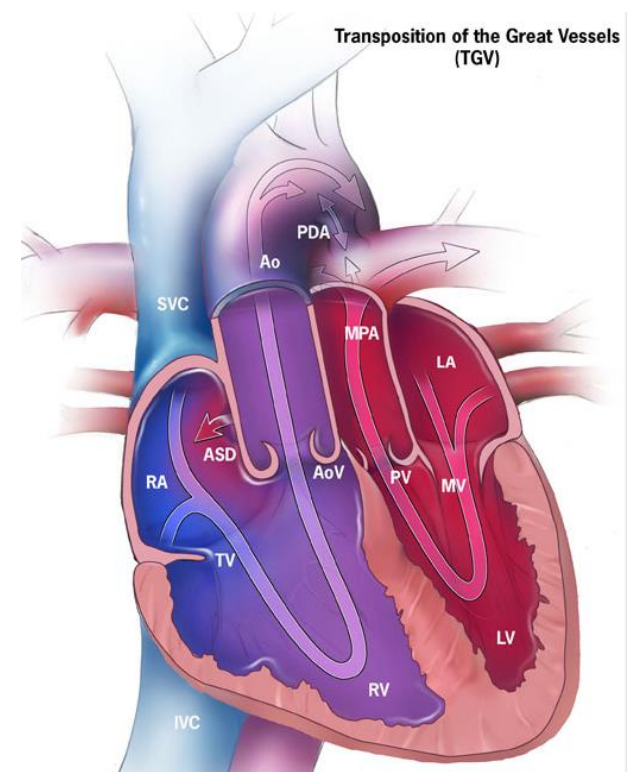


Image credit: CDC (2014)



CCHD: A Closer Look

▶ **Tricuspid Atresia:**

- ▶ Defect of the tricuspid valve in which the valve failed to form and no blood can pass through
- ▶ Presence of an atrial septal defect (ASD) or a ventricular septal defect (VSD)
- ▶ Presence of a patent ductus arteriosus (PDA)

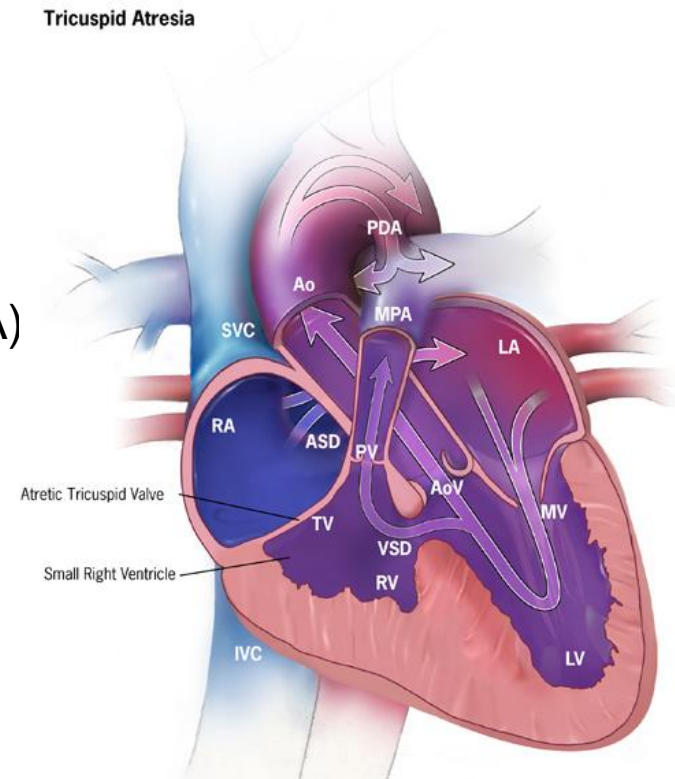


Image credit: CDC (2014)

CCHD: A Closer Look

▶ **Truncus Arteriosus:**

- ▶ Defect in which the aorta and pulmonary artery failed to separate during development, resulting in the mixture of oxygenated and deoxygenated blood
- ▶ Single common truncal valve instead of separate aortic valve and pulmonary valve
- ▶ Presence of a ventricular septal defect (VSD)

Truncus arteriosus

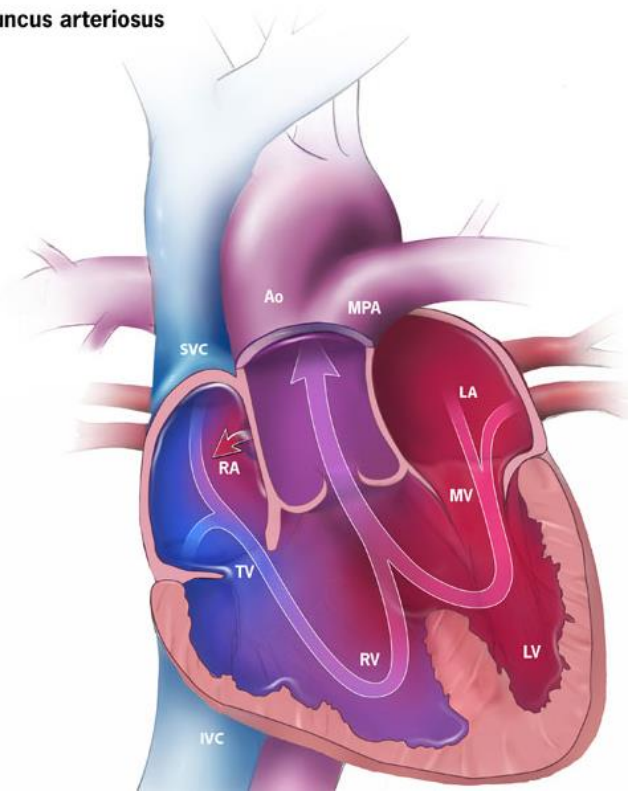


Image credit: CDC (2014)

Pulse Oximetry: the Screen & the Oximeter



Pulse Oximetry: Context

Who is screened?

- **All newborns:**
 - Must be calm & well; not crying
 - Warm extremities (temperature affects readings)
 - Skin clean & dry (dried blood affects readings)
 - Using room air; not on supplemental oxygen

When is screening performed?

- **Healthy Newborn:** Between 24-48 hours of life
- **Sick Newborn:** Between 24-48 hours of life
 - May delay if on supplemental oxygen
- **Before 24 hours:** higher risk for false positives (fetal-neonatal circulation transition not fully established)
- **After 48 hours:** delayed identification & treatment of affected newborns



The Pulse Oximeter

What is it?

- ▶ **Screening tool:** measures the percent of oxygen saturation of hemoglobin in the blood; and pulse rate
 - Simple
 - Painless
 - Non-invasive
 - Quick



The Pulse Oximeter

Oximeter Probe: 2 main parts

- light emitter
- Photodetector

Where is the probe placed?

- Right hand: preductal measurement
- Either foot: postductal measurement



Points to Consider

- Pulse oximeter must be FDA approved (AAP, 2015)
- Regular calibration of the oximeter is required
- Pulse oximetry readings are averages
- Skin color and jaundice **do not** affect pulse oximetry readings
- ***Continuous pulse oximetry monitoring does not replace the pulse oximetry screen.***



Screening How-To, Protocol, & Guidelines



How is the Screen Performed?

1. **Select site:** right hand; either foot
2. Place photodetector on outer aspect of hand/foot (under 4th-5th finger/toe)
3. Wrap sensor tape around extremity
4. Ensure light emitter is **directly opposite** the photodetector
5. If using a reusable sensor, secure the sensor using wrap recommended by vendor; **do not tape** or use hand to secure sensor to site



Photo credit: Masimo 2011



Guidance for Screeners

Pulse Ox Dos

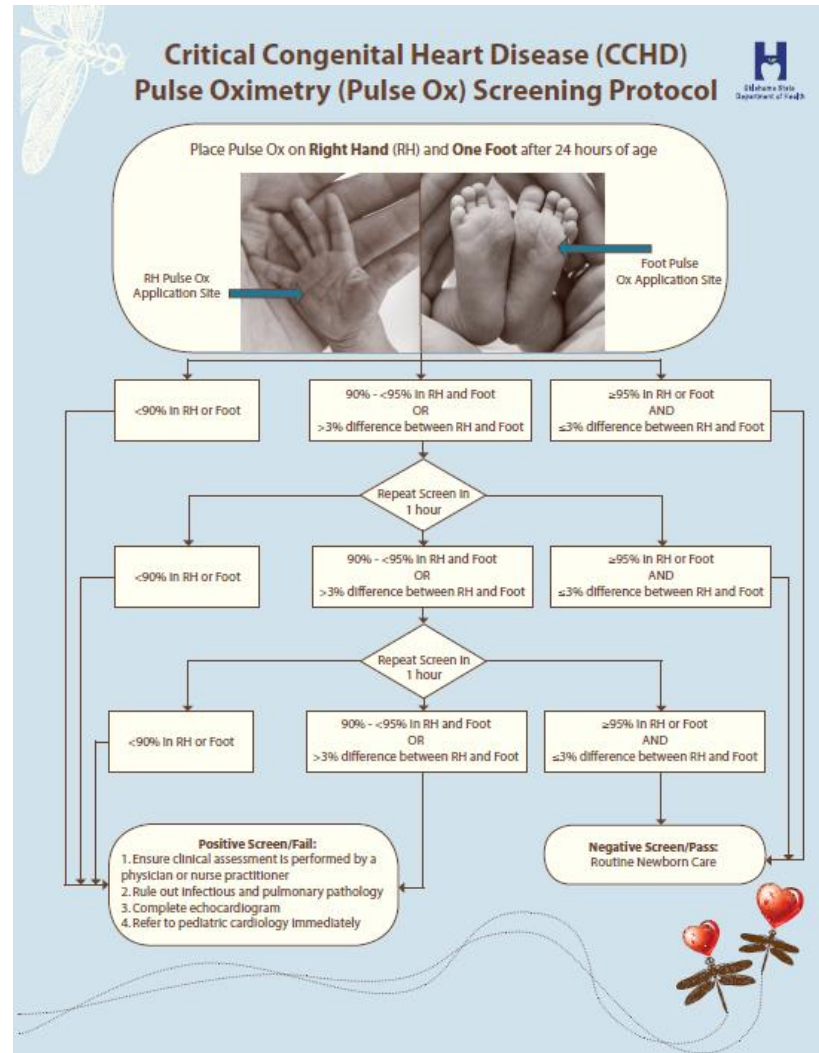
- ▶ If disposable, use a new, clean sensor; if reusable, clean between use
- ▶ Clean according to manufacturer recommendations
- ▶ Ensure newborn is calm and warm, not crying; encourage family involvement
- ▶ Ensure newborn skin is clean and dry
- ▶ Ensure no gaps between sensor and newborn's skin
- ▶ Light emitter and photodetector should be **directly opposite** of one another
- ▶ Use alongside physical examination
- ▶ Ensure pulse: no pulse, no oximetry!

Pulse Ox Don'ts

- ▶ Do not use an adult probe
- ▶ Do not tape pulse oximeter in place (use disposable wrap as indicated)
- ▶ Do not use your own hand to hold sensor in place
- ▶ Do not obtain reading from same extremity with blood pressure cuff
- ▶ Bilirubin lamps & surgical lights can affect accuracy of reading; cover pulse oximetry sensor with a blanket if such instruments are in use
- ▶ Do not use in isolation



Pulse Oximetry Screening Protocol



Interpretation of Results & Follow-Up



Screening Results

▶ **Negative Screen (Pass):**

- Oxygen saturation $\geq 95\%$ in Right Hand and/or Left or Right Foot
AND
- Difference between the Right Hand and Left/Right Foot $\leq 3\%$

▶ **Positive Screen (Refer/Fail):**

- Oxygen saturation $< 90\%$ in Right Hand or Left/Right Foot during **any** screen
 - Oxygen saturation 90 - 94% for **all** 3 screens (1 hour between each screen)
 - Difference between the Right Hand and Left/Right Foot $> 3\%$ for **all** 3 screens (1 hour between each screen)
- **If at *any* time, the newborn should become symptomatic, the family should *immediately* take the baby to the closest emergency room for evaluation.**



Interpretation of Results

Negative = Pass

- ▶ Results are in-range
- ▶ Blood oxygen level WNL
- ▶ CCHD still possible (if symptomatic, a cardiac evaluation is warranted)
- ▶ Monitor baby's status:
 - ✓ Heart rate – too fast/slow?
 - ✓ Energy – overly sleepy/fussy/lethargic?
 - ✓ Appearance – pale/blue skin?
 - ✓ Respiration – too fast/slow?
 - ✓ Temperature – cold to touch?
 - ✓ Feeding – difficulties?

Positive = Fail/Refer

- ▶ Results are out-of-range
- ▶ Blood oxygen level is low
- ▶ High risk; not diagnostic
- ▶ Confirmatory procedures & referral for treatment are warranted



Pulse Oximetry Screening for Critical Congenital Heart Defects (CCHDs) in Newborns without Cardiovascular or Respiratory Distress

Interpretation of Pulse Oximetry Results

Oxygen Saturation (%)

Right Hand (RH)	Either Foot (F)											
	100	99	98	97	96	95	94	93	92	91	90	89 or lower
100	100	99	98	97	96	95	94	93	92	91	90	89 or lower
99	100	99	98	97	96	95	94	93	92	91	90	89 or lower
98	100	99	98	97	96	95	94	93	92	91	90	89 or lower
97	100	99	98	97	96	95	94	93	92	91	90	89 or lower
96	100	99	98	97	96	95	94	93	92	91	90	89 or lower
95	100	99	98	97	96	95	94	93	92	91	90	89 or lower
94	100	99	98	97	96	95	94	93	92	91	90	89 or lower
93	100	99	98	97	96	95	94	93	92	91	90	89 or lower
92	100	99	98	97	96	95	94	93	92	91	90	89 or lower
91	100	99	98	97	96	95	94	93	92	91	90	89 or lower
90	100	99	98	97	96	95	94	93	92	91	90	89 or lower
89 or lower	100	99	98	97	96	95	94	93	92	91	90	89 or lower

Pass/Negative 95 % or higher in right hand (RH) or either foot (F) AND difference of 3% or less between RH and F.

Rescreen 90-94% in RH and F OR difference of 4% or more between RH and F. Screen up to 3 times, 1 hr btwn each screen.

Fail/Positive 89% or lower in RH or F (at any time)
OR
3rd screen: 90-94% in RH and F OR difference of 4% or more between RH and F.

Reference:

Michigan Department of Community Health. Critical Congenital Heart Disease Newborn Screening Program. (2013). Pulse ox screening visual aid. Retrieved from http://www.michigan.gov/documents/mdch/PO_Screen_Graphic_422859_7.pdf

Follow-up

- **Newborn Referral Indicated:** *What to do next?*
 - Contact the infant's provider immediately.
 - Document results on the Newborn Screening Filter Paper or on the Pulse Oximetry Screening Report Form
 - Proceed with follow-up according to protocol
 - ECHO indicated



Reporting Results for CCHD

▶ Filter Paper

▶ Pulse Oximetry Result Form

1591162 INVD

SN 1591162 Newborn Screening Form
Oklahoma State Department of Health
Oklahoma City, OK 73124-0106 (405)

INFANT'S INFORMATION

1. Infant's Last Name: _____

2. Sex: M F 3. Date of Birth: _____ 4. _____
MM DD YY

6. Birthweight in Grams: _____ 7. If Multiple Birth Indicate Birth Order: A-H

9. Provider ID: _____ 10. Infant: _____

11. Mom's Medicaid Number: _____ 12. ()

MOM'S INFORMATION

1. Mom's Last Name, First Name: _____

3. Mom's Address: _____

5. Mom's City: _____

8. () - _____
Mom's Telephone or Contact

10. Mom's Race/Ethnicity: 1. White 2. Black 3. Hispat

Pulse Oximetry (CCHD) Screen

Not Performed Pass Fail Echo Refused

105178 / 1316104

SUBMITTING HEALTH PROVIDER TO: _____
Return to Submitter at this address:

H

Oklahoma State Department of Health
Pulse Oximetry Screening Result Form

Infant Information:

Infant's Last Name: _____ Infant's First Name: _____

Medical Record Number: _____ Attending Physician: _____

Date of Birth: ____/____/____ Birth Hospital: _____

Mother's Last Name: _____ Mother's First Name: _____

Pulse Oximetry Screening:

Date of Screening: ____/____/____

Age at Time of Screening: _____ Days or _____ Hours

Result: Pass/Negative Fail/Positive Not Performed

Complete this section only if pulse oximetry screen was not performed:

Reason pulse oximetry screen not perform:

Early Discharge

Screening Not Indicated due to _____

Parent Refusal

Screeener's Name: _____

Screeener's Signature: _____ Date: ____/____/____

Form to be utilized if pulse oximetry screening results were not documented on newborn screening filter paper. Original to infant's record, provide a copy to parent, and forward copy by fax or mail to: Oklahoma State Department of Health, Newborn Screening Program Coordinator, 1000 NE Tenth Street, Oklahoma City, OK 73117-1299, (405) 271-6617 or 1-800-766-2223; Fax (405) 271-4892.

Newborn Screening Contacts

▶ Bloodspot, Pulse Oximetry, & Hearing Screening

Screening & Special Services
1000 NE 10th Street
Oklahoma City, OK 73117-1299

Phone: 1-405-271-6617
Toll Free: 1-800-766-2223
Fax: 1-405-271-4892

▶ Public Health Laboratory

Newborn Screening Section
Public Health Laboratory Service
1000 NE 10th Street
Oklahoma City, OK 73117-1299

Phone: 1-405-271-5070
Toll Free: 1-800-766-2223
Fax: 1-405-271-4850

