Newborn Screening

Oklahoma State Department of Health
Newborn Screening Program

Phone: (405) 426-8310
Toll Free: 1 (800) 766-2223
Fax: (405) 900-7556
NewbornScreen@health.ok.gov

OKLAHOMA
State Department of Health

1

Why Newborn Screening

Allow me to Introduce….

OKLAHOMA State Department of Health

2

Jase's Story – Galactosemia

Oklahoma State Department of Health | Newborn Screening

3

History of Newborn Screening

Then and Now

OKLAHOMA State Department of Health

4

History of Newborn Screening

- Newborn screening **originated** with Dr. Robert Guthrie who developed a test for elevated phenylalanine in dried blood spots in **1960**. (PKU Disease)
- **Before** the blood test existed, most children with PKU were not diagnosed until after they had irreversible brain damage.
- **Early** test > **Early** diagnosis > **Early** treatment > Mitigated brain damage.
- PKU was the first condition identified by NBS, so some people still refer to all NBS as the "PKU test." However, **this term is not accurate as the newborn screen now tests for 57 total disorders, not solely PKU.**

Oklahoma State Department of Health | Newborn Screening

5

Before NBS: Parents Had to Lose One to Save One

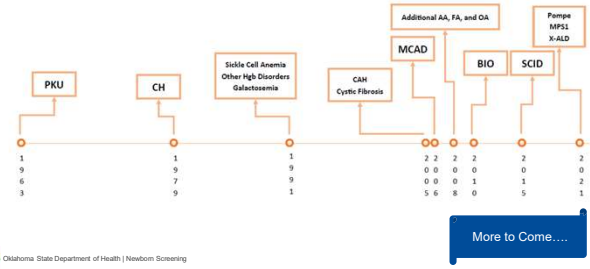
Untreated versus Treated PKU

Oklahoma State Department of Health | Newborn Screening

6

Scientific Progress Translates to More Infant Lives Positively Impacted

Newborn Screening Timeline



More to Come….

Newborn Screening Today: A Three Part Process

- Newborn screening checks a baby for certain conditions present at birth that benefit from early treatment or intervention.
  - Blood spot screening, which determines if a baby might have one of many serious conditions
  - Pulse oximetry screening, which determines if a newborn might have certain heart conditions
  - Hearing screening, which determines if a newborn might be deaf or hard of hearing

PURPOSE – Every Baby Deserves A Newborn Screen

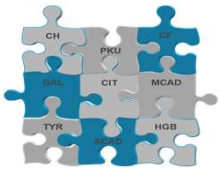
- Newborn screening (NBS) is the practice of testing **every** newborn for harmful or potentially fatal disorders that are not otherwise apparent at birth.
- **Early detection** and **prompt treatment** can make the difference between healthy development or lifelong impairment and possible death.

Screening VS Diagnostic

- **Screening results**, by themselves, cannot determine the presence or absence of a disorder. The purpose of a screen is to detect risk factors for disease in large numbers of apparently healthy individuals – in this case, newborns.
- **Diagnostic results** refers to the combination of signs, symptoms, and test results that allows the provider to confirm the diagnosis of the respective disease.

Who Decides?

- In 2022 the Oklahoma legislature passed statute stating that the Oklahoma NBS panel will match the national Recommended Uniform Screening Panel (RUSP) to the extent practicable.
- Once a condition is added to the RUSP the NBS Program (lab and follow up) will determine practicability and readiness.
- The Infant and Children's Health Advisory Committee will provide recommendations to the Commissioner of Health to add the disorder.
- The Commissioner of Health will give final approval.
- Oklahoma currently screens for 57 possible hidden disorders.
- Oklahoma will continue to expand.



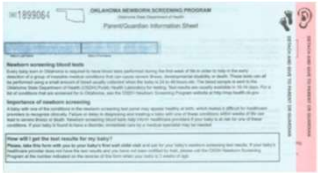
Autosomal Recessive

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

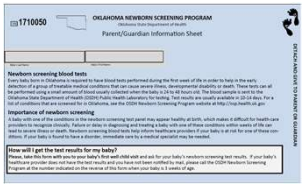
Possible Outcomes for Offspring of Parental Disease Carriers

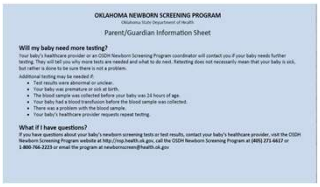
Parent Education

- Instruct parents to ask for their baby's newborn screening results:
  - Baby's Pediatrician
  - Local County Health Department
  - OSDH Newborn Screening Program
- Tell parents to hang onto the Blue or Pink slip from their baby's filter paper for reference.

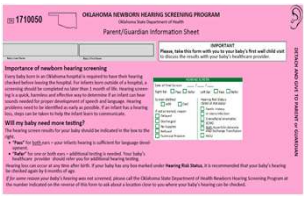


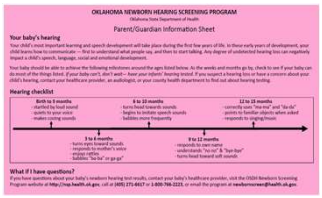
Oklahoma State Rules and Statutes – Filter Paper Education Pages Blood spot





Oklahoma State Rules and Statutes – Filter Paper Education Pages Hearing Screen





Parent Education

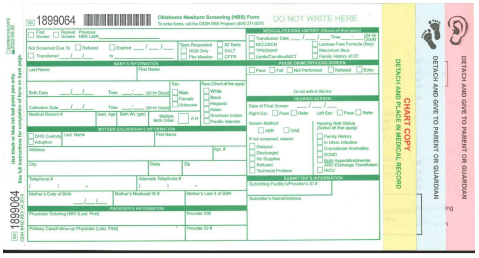
- NBS is collected on **every** baby born in Oklahoma.
- Importance of **correct** contact info & PCP for follow-up.
- *No news is not good news!* Update NBS Program with changes in home address and/or PCP.
- Review hidden disorders, using NBS pamphlet as a guide.
- Specimens are kept by the OSDH lab 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.

Indications for Repeat Screen

- The NBS testing results are out of range for one or more disorders – PCP notified by phone, parent by mail.
- The NBS sample was damaged or is otherwise unsatisfactory for testing – PCP and parent notified by mail.
- The infant received a transfusion prior to NBS collection – usually in NICU, will notify PCP when time for recollection.
- The infant's screen was collected prior to 24 hours old – if normal result, this report will be on the portal, if out of range, PCP notified by phone, parent by mail.
- The infant is premature or sick (TPN and antibiotics could affect results.

Filling Out the Demographic Form

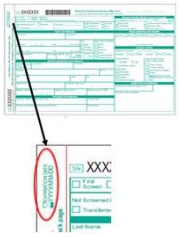
Filling out the Form



Specimen testing will be delayed if the form is incomplete!

Filling out the Form

Specimen testing will be delayed if the form is incomplete!



- **Check expiration date**
  - If the filter paper is expired, discard the paper, and check the stock of filter paper kits and discard all expired kits.
  - Collect the specimen on a kit that is not expired.

Filling out the Form: Specimen Information

Specimen testing will be delayed if the form is incomplete!

- If this is the first specimen collected for the baby, check the "First Screen" box.
- If baby has had a previous screen, check the "Repeat Screen" box.
  - List the previous OSDH Lab Number, if you have it.

If you don't mark "repeat screen" the correct linking to the original screen could be delayed.

Filling out the Form: Specimen Information

Specimen testing will be delayed if the form is incomplete!

- If baby expires before a screen can be collected:
  - Check the "Expired" box
  - Enter the date that baby passed away
  - Submit the filter paper form to the OSDH PHL

Filling out the Form: Specimen Information

Specimen testing will be delayed if the form is incomplete!

If baby is transferred prior to specimen collection:

- Check the "Transferred " box
- Enter the date that baby transferred and the facility that baby was transferred to
- It is the responsibility of the receiving facility to collect the newborn screen

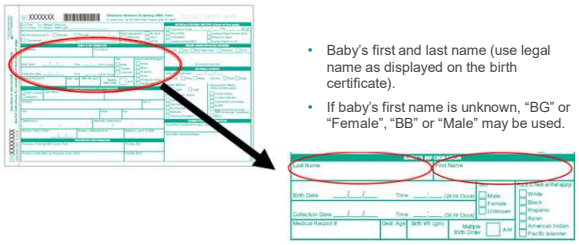
Filling out the Form: Specimen Information

Specimen testing will be delayed if the form is incomplete!

- Tests Requested: Check all that apply.
  - **All Tests**- always check unless test is for HGB Only. This ensures the lab screens for all disorders on the NBS panel.
  - **HGB Only**- Check if repeat screen is for follow-up of initial abnormal HGB result.
  - **GALT**- Check GALT in addition to All Tests if there is a family history of galactosemia or if baby is on lactose-free (soy) formula at time screen is collected.
  - **Phe Monitor**- Check only if baby has been diagnosed with PKU (typically metabolic specialists only)
  - **CFTR**- Check in addition to All Tests if baby has clinical concerns for CF, meconium ileus, and/or family history of CF.

Filling out the Form: Infant's Information – Initial screen

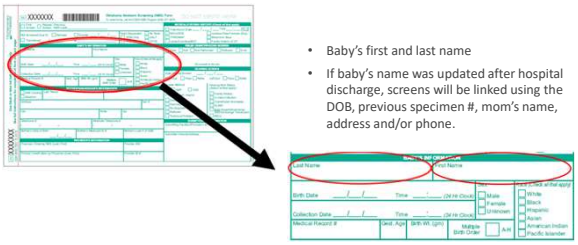
Specimen testing will be delayed if the form is incomplete!



- Baby's first and last name (use legal name as displayed on the birth certificate).
- If baby's first name is unknown, "BG" or "Female", "BB" or "Male" may be used.

Filling out the Form: Infant's Information – for repeat screen

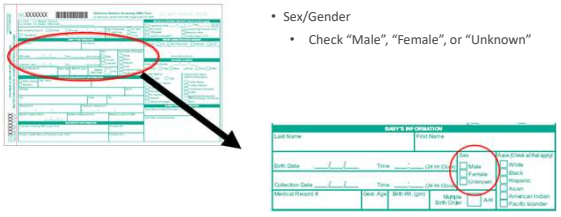
Specimen testing will be delayed if the form is incomplete!



- Baby's first and last name
- If baby's name was updated after hospital discharge, screens will be linked using the DOB, previous specimen #, mom's name, address and/or phone.

Filling out the Form: Infant's Information

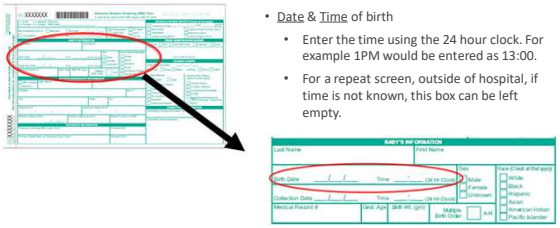
Specimen testing will be delayed if the form is incomplete!



- Sex/Gender
  - Check "Male", "Female", or "Unknown"

Filling out the Form: Infant's Information

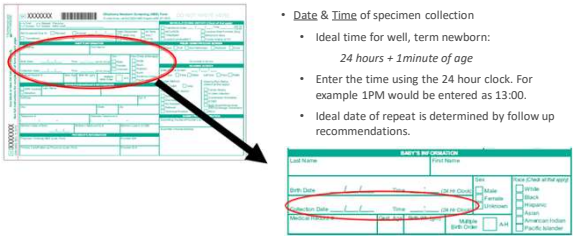
Specimen testing will be delayed if the form is incomplete!



- Date & Time of birth
  - Enter the time using the 24 hour clock. For example 1PM would be entered as 13:00.
  - For a repeat screen, outside of hospital, if time is not known, this box can be left empty.

Filling out the Form: Infant's Information

Specimen testing will be delayed if the form is incomplete!



- Date & Time of specimen collection
  - Ideal time for well, term newborn:

    24 hours + 1minute of age
  - Enter the time using the 24 hour clock. For example 1PM would be entered as 13:00.
  - Ideal date of repeat is determined by follow up recommendations.

Filling out the Form: Infant's Information

Specimen testing will be delayed if the form is incomplete!

- Medical record number
  - Baby's medical record number
  - If a multiple birth, take extreme care here

Filling out the Form: Infant's Information
Specimen testing will be delayed if the form is incomplete!

- Gestational Age
  - List gestational age at birth, may leave blank on a repeat collection.
  - Lab cut off values for abnormal severe combined immunodeficiency (SCID) are gestational age dependent.

Filling out the Form: Infant's Information
Specimen testing will be delayed if the form is incomplete!

- Birthweight (in grams), leave blank if unknown (for repeat screen out of hospital)
  - Lab cut off values for abnormal congenital adrenal hyperplasia (CAH) results are dependent on birth weight.

Filling out the Form: Infant's Information
Specimen testing will be delayed if the form is incomplete!

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

Filling out the Form: Mom's Information
Specimen testing will be delayed if the form is incomplete!

- DHS Custody or Adoption

Note: If baby is adopted, be sure to check the **Adoption** box on the filter paper form and enter the agency/law firm information in this section. If DHS is involved, enter case worker information in this section and check the **DHS Custody** box.

Filling out the Form: Mom's Information
Specimen testing will be delayed if the form is incomplete!

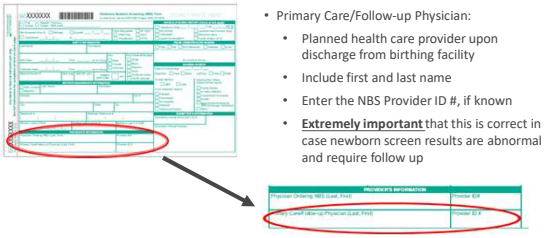
- Mom's first and last name
- Mom's mailing address:
  - Street, Apt # (if applicable), City, State, Zip
- Mom's telephone number:
  - Extremely important to include in case newborn screen results are abnormal and require follow-up.

Filling out the Form: Provider's Information
Specimen testing will be delayed if the form is incomplete!

- Physician Ordering the NBS:
  - Include first and last name
  - Enter the NBS Provider ID #, if known
  - May leave blank if ordering Physician unknown on a repeat collection, outside of hospital

Filling out the Form: Provider's Information

Specimen testing will be delayed if the form is incomplete!



- Primary Care/Follow-up Physician:
  - Planned health care provider upon discharge from birthing facility
  - Include first and last name
  - Enter the NBS Provider ID #, if known
  - **Extremely important** that this is correct in case newborn screen results are abnormal and require follow up

Filling out the Form: Medical/Feeding History

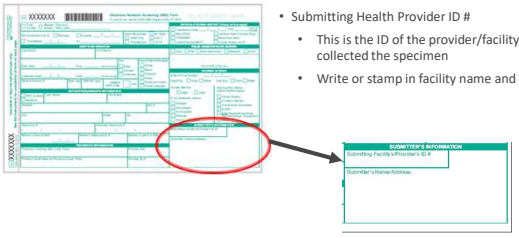
Specimen testing will be delayed if the form is incomplete!

Check all that apply for baby at the time of specimen collection

- If transfused enter the date and time of transfusion
- NICU/Special Care Nursery
- TPN/SNAP
- Lipids/Carnitine/MCT
- Lactose-Free (Soy) Formula
- Meconium Ileus
- Family History of Cystic Fibrosis (CF)

Filling out the Form: Submitter ID

Specimen testing will be delayed if the form is incomplete!



- Submitting Health Provider ID #
  - This is the ID of the provider/facility who collected the specimen
  - Write or stamp in facility name and address

Filling out the Form: Collector's Initials

Allows for thorough follow up of an unsatisfactory screen

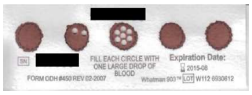
Note: Do not touch the filter paper in any other area when writing initials and unit.

- **Unsatisfactory Specimen Follow-up**
  - Specimen collectors can place their initials and unit in the area below for identification purposes, in the event of an unsatisfactory specimen. This allows for easier identification of the individual who collected the specimen so that further education and/or training can be provided.

Collecting the Specimen

How Is An Infant Screened?

- Blood spot screen-heel stick
- Performed when the infant is "24 hours plus one minute" or prior to discharge, whichever comes first.
- Small amount of blood is placed on a small card and sent to the OSDH Newborn Screen lab
- Newborn screening specimens are picked from birthing hospitals and county health department via a contract courier service and then brought to the PHL for testing.

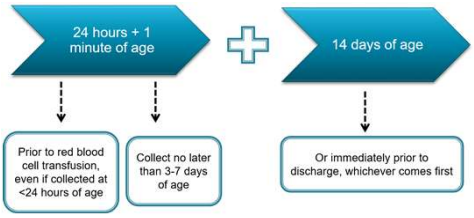


Time of Screening: Healthy Newborn

"**24 hours** plus one minute" of age
Or
Prior to discharge

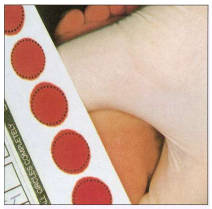
WHICHEVER COMES FIRST

Time of Screening: Premature or Sick Newborns

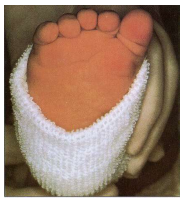


Specimen Collection

- **Heel Stick / Direct Application**
  - Preferred, recommended method
  - Start with clean, dry hands before handling the filter paper.



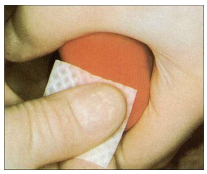
Direct Application



Prepare the Site

- Warm the heel with a heel warmer or a soft cloth, moistened with warm water up to 41 C for 3-5 min.
- Warmth leads to vasodilation, which increases blood flow and chance of collection success.
- **Follow your facility protocol regarding which warming device to use.**

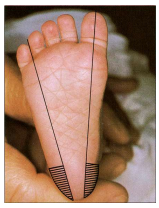
Direct Application



Prepare the Site

- If desired, parent may hold infant during 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 and/or lead to unsatisfactory specimens.

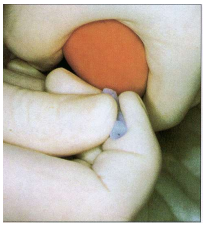
Direct Application



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.

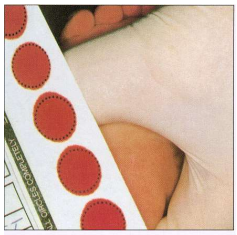
Direct Application



Perform the Puncture

- 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 and plain.
- No additives, must be anticoagulant free, however no anti-coagulants = risk of clotting.
- Risk of scratching the filter paper, avoid touching tip of tube to the paper.
- Use a new tube for EACH 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 CLSI guidelines for more information.

Only in certain circumstances

Alternative Specimen Collection

What about umbilical catheters? >> **Discouraged**

- May be appropriate under certain circumstances (e.g. NICU).
- Ensure the line is cleared with withdrawing 2-2.5 cc (ml) of blood to collecting a specimen for newborn screen.
- Please refer to 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 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 specimens in plastic bags.
- Do NOT batch (hold onto) specimens.

Collection Reminders

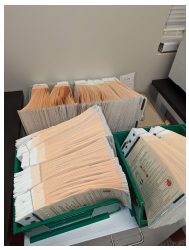
Pre-collection:

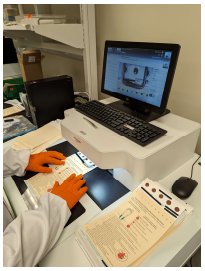
- **Check the Expiration Date of the filter paper**
  If filter paper is expired, discard the paper, check the stock of filter paper kits it came from to ensure they are not all expired, and collect on a kit that is not expired.

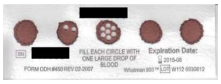
Post-collection:

- **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**

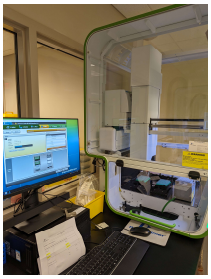
A Peek Inside the Laboratory.....



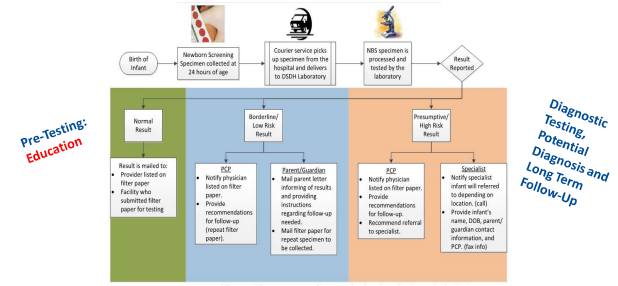




Inside the Laboratory



Blood Spot Screening is a system, not an event.



*Short-term Follow-up continues tracking case until confirmed normal or diagnosed with a disorder.

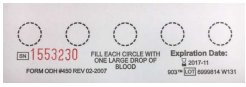
NBS Filter Paper Review

Unsatisfactory (Unsat) Specimen Examples

OKLAHOMA State Department of Health

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

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.

Clotted or Caked Blood

Front

Back

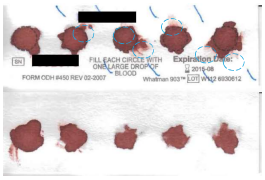
Why Unsat?

- 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.

Serum Rings

Front

Back



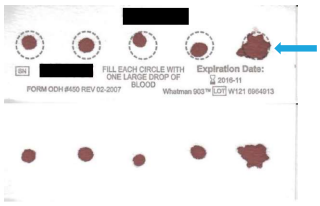
Why Unsat?

- 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

Inadequate Amount of Blood

Front

Back



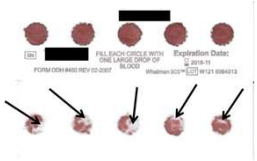
Why Unsat?

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

Under-Saturation

Front

Back



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.

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 with 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.

NICU and Special Considerations

NICU Special Considerations

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

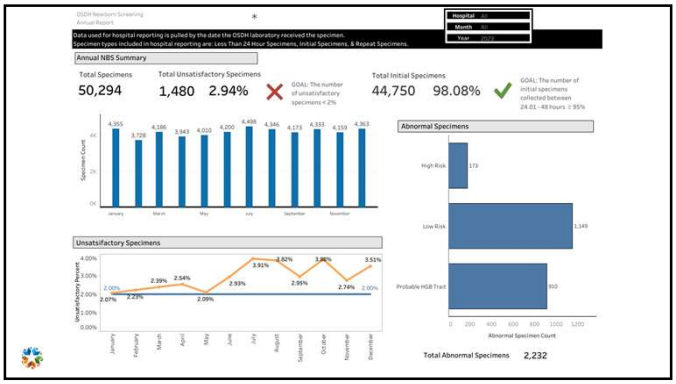
Maternal
- PTU therapy or radioactive iodine may affect infant TSH results
- Steroids may affect infant 17-OHP results

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

Additional Information

Hospital Responsibilities

- Ensure **ALL** infants are screened prior to discharge. (Keep a log book to ensure compliance.)
- Ensure specimens are received in a timely manner to the OSDH PHL for testing.
- Infants who are transferred: >> The **RECEIVING** hospital is to ensure that the NBS has been collected.
- Submit **SATISFACTORY** specimens:
  - Collected properly
  - All requested information is documented on the demographic form attached to filter paper
  - Submitted timely

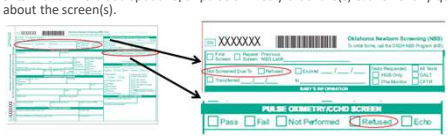


Refusal of screening

- Parents may refuse screens based on Religious Tenets and Practices.
- Refusal form must be signed and placed in the medical record with a copy mailed to the Newborn Screening Program.
- Please note that parent must indicate which of 3 portions of screen are being refused: Hearing, bloodspot, and/or CCHD.
- Demographic form with no specimen on filter paper should be sent into PHL with courier, with "X" marked in refusal box.

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).



- Ensure the parents fill out a Refusal Form. Keep a copy for baby's record & fax a copy to the NBS Program using fax # 405-900-7556.

Refusal Form


OKLAHOMA State Department of Health

Newborn Screening Program
Religious Tenets and Practices Refusal Form

Infant's Name: _____ Date of Birth: _____ Gender: M / F
Parent/Guardian's Name: _____ Medical Record #: _____
Street Address: _____ Apt/Unit #: _____
City/State/Zip: _____ Phone #: _____
Place of Birth (check one): ___Hospital ___ Birthing Facility ___ Home Birth
Hospital/Facility Name: _____ Attending Physician/Midwife: _____
Child's Dr/Planned Primary Care Provider: _____ Dr's Phone #: _____
Type of Screen Refused: ___Newborn Blood Spot ___Pulse Oximetry Screen ___Hearing Screen
(Check any that apply & complete the corresponding section(s) below)

I, (Guardian's name) _____, have been fully informed of the importance of newborn screening, and I understand that all newborns are required by law to have the newborn screening tests performed. Although the benefits of newborn screening and the dangers of not being screened have been explained to me, I elect to refuse the newborn screening test(s) checked above for my child, (Infant's name) _____, born on ___/___/___, on that such testing of my infant interferes with my religious tenets and practices. My decision was made freely, and I accept the legal responsibility for the consequences of this decision. I have discussed the newborn screening tests with _____, my child's healthcare provider, and I understand the risks to my child if the newborn screen(s) are not completed.

Blood Spot Refusal
I, (Guardian's name) _____, understand the disorders the newborn metabolic screen test for are easily detected by testing a small blood sample from my baby's heel. I am aware that the signs and symptoms of these disorders sometimes do not appear for several weeks or months, and irreversible damage can occur before symptoms become apparent. I have been informed that these conditions are treatable but if left untreated may cause permanent damage to my child, including mental retardation, growth failure, and even death.

Pulse Oximetry Refusal
I, (Guardian's name) _____, understand the congenital heart defects that the pulse oximetry screen can be detected by measuring the amount of oxygen in my baby's blood. I am aware that the signs and symptoms of these defects sometimes do not appear for several weeks or months, and irreversible damage or death can occur if not identified early.

Hearing Refusal
I, (Guardian's name) _____, understand the importance of finding out if my baby can hear sounds needed to listen and talk. It has been explained to me that most babies born with hearing loss have parents who can hear and there is no history of hearing loss in their family. I understand that any degree of hearing loss has the potential to interrupt speech, language, cognitive, emotional and/or social development.

Print Parent/legal Guardian's Name _____ Signature of Parent/Legal Guardian _____ Date ___/___/___

Transit Time

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

OKLAHOMA State Department of Health

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

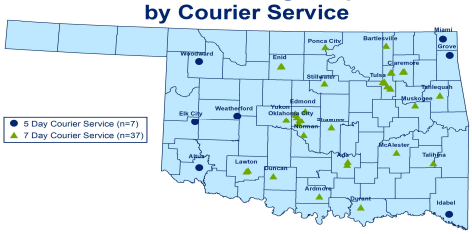
- **Guidelines:**
  - Specimens should be received at the OSDH Lab within **48 hours** from the time of collection.
  - Oklahoma Law: *OS 63 Sections 1-533 and 1-534*

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.

Oklahoma Birthing Hospitals by Courier Service



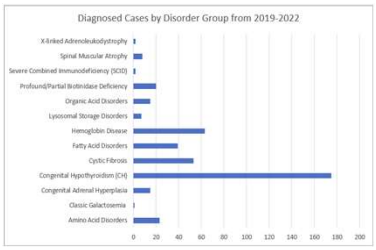
OKLAHOMA State Department of Health | Public Health Laboratory

Screening & Special Services
Oklahoma State Department of Health
Date created: 07/08/2022
Coordinate system: USA_Contiguous_Albers_Equal_Area_Conic
*64 birthing hospitals as of May 2023

Impact

Blood Spot Screening Statistics in Oklahoma

- In 2023 **50,294** dried bloodspot specimens were collected.
- 3 in 50 infants will have an abnormal screen that requires further testing.
- 1 in 450 infants will be identified with a disorder
- Specimens are kept by the OSDH lab for 42 days before being destroyed



**LSD Disorders began screening in June 2021.

Newborn Screening WINS – True Story Timeline

Case Study from 2023

- Born on a Tuesday at 0121
- NBS collected Wednesday at 0150
- Specimen arrived at PHL Wednesday at 1900
- Preliminary critical result called to NBS follow up nurse on Thursday at 1210
- Baby was found to be still in hospital, in Mother/Baby unit. Mother/Baby nurse was notified, Genetic specialist notified and confirmatory labs were ordered. Feeding precautions initiated, decision made to delay discharge another night.
- NBS critical result finalized on Friday, called to NBS follow up nurse who promptly notified Mother/Baby staff and Genetics Specialist. Emergency management protocol in place, confirmatory labs are pending. Geneticist speaks with infant's parents at bedside for initial consultation. Treatment is initiated on Friday (3 days after birth), in anticipation of confirmatory testing results.
- Confirmatory lab results are finalized 8 days after birth. Ongoing care has been established with geneticist with plans for life long management in place.
- Delay of diagnosis or lack of treatment for this fatty acid disorder would have resulted in infant mortality.

Pulse Oximetry Screening

CCHD – Critical Congenital Heart Disease

Critical Congenital Heart Disease (CCHD)

- Screening began in 2014.
- Screening is done by utilizing pulse-oximetry.
- Critical congenital heart defects are conditions that are present at birth and can affect the structure of a baby's heart and the way it works.

Pulse Oximetry Screening

Purpose:

- 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.

Pulse-Oximetry Screening

- Simple and painless way to measure the amount of oxygen in the baby's blood.
- Congenital heart disease is the **most common** birth defect
- 1 in 110 infants will have a heart defect, 25% of those cases will have CCHD.
- Most affected will not have symptoms early on.
- Most will require surgery shortly after birth.

Normal Heart: Blood Flow

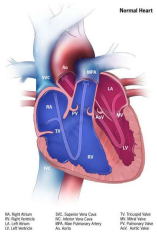


Image credit: CDC (2014)

- 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.

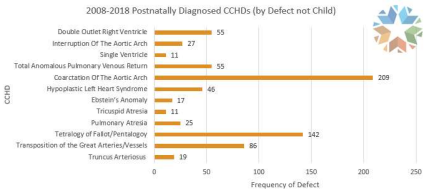
CCHD: What to Watch For

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.**

CCHDs in Oklahoma



Data provided by the Oklahoma Birth Defects Registry. Data does not reflect cases identified solely through pulse oximetry screening for CCHDs.

Pulse-Oximetry

The Screen and 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: pre-ductal measurement
- Either foot: post-ductal 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 and 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

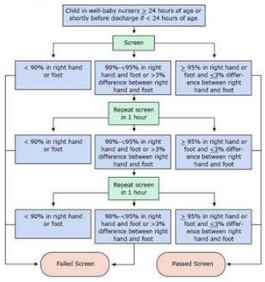
Pulse Ox Dos

- 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



Screening Results

Negative Screen (Pass):

- Oxygen saturation ≥ 95% in Right Hand and/or Left or Right Foot

AND

- Difference between the Right Hand and Left/Right Foot ≤ 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

Interpretation of Results: CCHD Screening for Newborn without Cardiovascular or Respiratory Distress

Oxygen Saturation (O2 Sats; %)

Right Hand (RH)	Either Foot (F)											
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	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.
Fail	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.

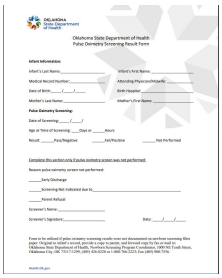
Adapted from *Critical Congenital Heart Disease Screening with Pulse Oximetry in the Neonatal Intensive Care Unit* Lakshminrusimha S., et al., e-Journal of Neonatology Research 2012;2(2:96-101.

Reporting Results for CCHD: Filter Paper

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 # 405-900-7556.

- Pulse Oximetry Screen: Check Only ONE
  - Pass
  - Fail
  - Not Performed
  - Refused
  - Echo

Reporting Results for CCHD - Pulse Oximetry Result Form



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/Midwife: _____

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 performed:

_____Early Discharge

_____Screening Not indicated due to: _____

_____Parent Refusal

Screener's Name: _____

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

Fax to be utilized if pulse oximetry screening results were not documented on newborn screening filter paper. Please fax form to the Newborn Screening Program at 405-900-7556. Keep a copy for your records. Oklahoma State Department of Health, Newborn Screening Program Coordinator, 1000 NE Tenth Street, Oklahoma City, OK 73117-299; (405) 426-8220 or 1-888-766-2223, Fax (405) 900-7556.

Health.ok.gov

Newborn Screening Contacts

- **Bloodspot, Pulse Oximetry, & Hearing Screening**

  Screening & Special Services
  123 Robert S. Kerr
  Oklahoma City, OK 73102-6406

  Phone: 1-405-426-8220
  Toll Free: 1-800-766-2223
  Fax: 1-405-900-7556
  NewbornScreen@health.ok.gov

- **Public Health Laboratory**

  Newborn Screening Section
  Public Health Laboratory Service
  4615 W. Lakeview RD
  Stillwater, OK 74075

  Phone: 1-405-564-7750
  Toll Free: 1-800-766-2223
  Fax: 1-405-900-7611
  Publichealthlab@health.ok.gov

Thank you for your time!

Questions?