


Newborn Bloodspot Collection and Screening 2021

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Three types of newborn screening

- Bloodspot (NBS)
- CCHD/pulse ox*
- Hearing*



GOAL of Newborn Screening: Diagnose and treat disorders before they cause permanent harm- we screen because we can treat!

*typically performed by birthing provider





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What clinicians need to know about newborn bloodspot screening

Why it's important:

- Prevents death and/or disability
- Babies may look and act healthy at birth
- The disorders are not very common
- The disorders we screen for have treatments
- It's not just the "PKU" test!

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Newborn Screening Saves Lives

- Baby boy, uncomplicated pregnancy, normal vaginal delivery at 38 weeks, joining two healthy parents and a healthy sibling.
- Apgars were 8&9, discharged after 24 hours with mom and baby doing very well, bilirubin at discharge was low-risk at 5.8. Newborn bloodspot screen (NBS) collected prior to discharge.
- 3 days of age: appt with PCP, gaining weight well, feeding well (exclusively breastmilk) every 2-4 hours, no jaundice, no parental or provider concerns.
- 9 days of age: presented to PCP for circumcision, parents and provider noted jaundice. Child otherwise well-appearing. NBS results received that day concerning for galactosemia (GALT of 0.0; Galactose of 30 (nl <20))



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Continued...

- Due to NBS results in conjunction with the jaundice, child sent straight to emergency dept, and found to be in liver failure: Tbili at 32.9, Dbili 18.5, INR 2.12, AST 185, ALT 208, Alk Phos 786.
- Admitted to neonatal ICU
 - Immediate cessation of breast feeding
 - IV fluid with D10, then PO feedings with Isomil (soy) infant formula
 - Phototherapy x2 days for jaundice
- Discharged on admit day 4 in good health



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Neonatal Emergency

- Children who aren't quickly identified with galactosemia have a high rate of mortality (liver failure, overwhelming sepsis, bleeding)
- Rare: 1:40,000-1:60,000 births in Western countries
- Making the diagnosis early will save the baby's life: transition feeding from breast milk or lactose-containing formula (typically to soy formula) and supportive care until condition improves.
- Lactose-free diet for life.



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Newborn Bloodspot Screening includes



- Cystic Fibrosis
- Endocrine disorders
- Congenital hypothyroidism
- Fatty acid disorders
- Amino acid disorders
- Organic acid disorders
- Immune disorders
- Galactosemia
- Hemoglobin disorders
- Lysosomal storage disease
- More coming!



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Dry Blood Spot Collection

- DBS is whole blood collected on filter paper from a heel stick
- Screening infants includes *proper specimen collection, proper handling and packaging, prompt shipment to the state lab for testing*
- An issue in any of these areas can result in a unsatisfactory screening attempt! This can delay a life-saving diagnosis!



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What to tell parents?

- Educate parents about newborn screening
- Know your pediatrician or provider at time of birth
 - *we need to know where to result out to or who to contact. This is so important and often missed!*
- Ask your provider about the results
 - *don't assume no news is good news*
- Look for the card that is part 2 of the "kit" and bring it to your baby's doctor



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Parents should be sent home after birth with part 2 of the "kit" and a brochure about testing.

They should bring this to their baby's Doctor for the 2-week check-up.

OR

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What to tell parents?

- The heel stick isn't a dangerous or risky procedure, and you can also help it be as comfortable for the baby as possible:
 - No anesthetic creams can be used (might interfere with the analytic testing) BUT:
 - You can gently massage the newborn's leg before doing a heel stick
 - You can provide non-nutritive sucking (such as a pacifier)
 - You can allow the newborn to breastfeed during the heel stick
 - You can give an oral glucose: sucrose (20-30%) solution a couple minutes before the heel stick
 - Swaddle the baby in an upright position during the heel stick

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Timing of Specimen Collection

- Normal newborns should be tested at 24-48 hours old. Do it as soon as the baby has achieved 24 hours of life!
- The second screening for a normal newborn is collected between 10-14 days of age (typically at the 2 week follow up visit)
- NICU infants will get 3 screens
- Non-critical abnormal results: may require an *urgent* repeat screen, or early 2nd screen
- Unsatisfactory Specimen: requires an *urgent* repeat screen
- The goal is to have lab results to report at 5 days of life!

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Why Two Screens?

If you only do one screen, some disorders may be missed!
 A percentage of some disorders are found on the 2nd screen after a normal first screen

- Hypothyroidism 10%
- Adrenal Hyperplasia 20%
- Non-PKU Aminoacidopathies 10-50%
- MCAD 5%
- CPT1 78%
- Carnitine Uptake 60%
- LCHAD/VLCAD 15%

*Not all testing performed on a first screen is repeated on the second screen.



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Who is responsible for which screen?

- Before leaving birth facility (due to early discharge home, or transfer to another facility for any reason), a first screen must be obtained
- This may mean the first NBS screen is obtained prior to 24hrs of age
 - please check the box on the card if this is the case!
 - if this cannot be done for a specific medical reason, this must be documented in the chart and the accepting facility must be informed
 - the goal is to prevent a missed first screen
- Subsequent screenings will happen at the accepting facility
 - at the usual timing intervals
 - as requested by the Oregon newborn screening follow up team
- After discharge home, the primary care provider is responsible for newborn bloodspot screening
 - Must obtain, review, and act upon (as needed) all previous screens
 - Must collect any additional screens as needed



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Two Screens or Three Screens?

Table 2 — Age of infant at specimen collection

	Collection Kit	First specimen	Second specimen	Third specimen
Routine Birth	Double Kit	As soon as possible after 24 hours of age but before 48 hours of age	10-14 days	Not Collected
NICU infants transfused prior to 24 hours of age	Triple Kit	Prior to transfusion	48-72 hours after birth	~ 1 month, no sooner than 28 days
NICU infants not transfused prior to 24 hours of age	Triple Kit	As soon as possible after 24 hours of age but before 36 hours of age and prior to transfusion	10-14 days of age (11-15 days of life)	~ 1 month, no sooner than 28 days



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Avoid capillary tubes or needles when applying blood to filter paper



- Scratches or tears filter paper
- Causes uneven saturation
- Needles & cap tubes lyse the red cells, which leads to false negative or positive results!



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Heel Stick Procedure, step-by-step:

- Fill out the demographic data on the card. It must be complete- every detail counts when we are interpreting the results or need to follow up
- Use appropriate patient identification technique. Make sure you have the right filter paper kit (Part 1 or 2)
 - Note: be careful not to crush or compress filter paper while it is being stored for use, or put anything on top of the card- it will keep the blood from saturating properly
- Don't touch the filter paper- handle properly and wear gloves to avoid contamination.
 - Always use Universal Safety Precautions (as with any other specimen collection)
 - Do not touch or contaminate filter paper on the card with hands, gloves, bodily fluids, powder, formula, water, coffee, or anything else
 - DNA testing is sometimes performed as part of the NBS process, and it is important to prevent contamination with extraneous DNA from handlers!



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Heel Stick Procedure, step-by-step:

- Apply heat pack- this will improve your blood flow!
- Cleans the site on the baby's heel with alcohol swab and air dry
- Lance the site, and *wipe away the first drop of blood* (it will be contaminated)
- Allow one large drop to collect, and drop to the filter paper so that it fills up the circle completely AND is able to saturate the filter paper evenly- the front side and back side should look the same. Continue for each circle
 - Take care to avoid touching the heel directly to the filter paper! Only the blood drop itself should come into contact with the paper!



- Apply pressure to heel until bleeding stops



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Helpful Hints

- Positioning baby
- Massage the blood downwards
- Apply lancet to the heel with some pressure
- Forewarn parents: it might take more than one heel stick
- You can apply blood anywhere on the filter paper

An example:

<https://www.youtube.com/watch?v=30qbkh1jQ8>
<https://www.youtube.com/watch?v=u5S3OfWFelc>



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Helpful Hints

- Neatness does not count!
- You can use either side of the filter paper to fill, but only fill from one side
- Don't superimpose or put blood drops on top of each other ("layering")
- Don't "milk" the heel- you will get serosanguinous fluid and we need whole blood only
- If the blood flow is slow, restick!



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Proper Handling Post-Collection

- Take care not to touch or smear your blood spots
- Allow to air-dry horizontally for 3-4 hours at room temp
- Keep away from direct sunlight
- No not heat, stack or allow the blood spots to touch other surfaces during the drying process



- No need to dry longer than 3-4 hrs
- No not hang filter paper in a dependent position
- Do not cover blood spots with end paper until dry
- Do not store or ship in plastic bags



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And then ship to the OSPHL

- Place the protective cover over the DBS
- When stacking multiple cards, reverse ends so blood spots don't touch
- Make sure all demographic data is complete and card is fully filled out. Inspect to be sure the DBS are adequate
- Prepare a packing list of the specimens
- Put the specimens into their sealable paper envelopes or your large mailing envelope (when sending multiple specimens)
- Ship the same day by courier, express mail or a postal service
- Don't accumulate specimens
- Consider weather and holidays



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Weekends and Holidays

- Keep your specimens in a cool, dry, and at room temperature until able to send out. AVOID sunlight, heat, humidity, hot mailboxes, or similar conditions.
- **Send by overnight or express mail on the following business day.**
- **Even with holidays, the goal is always to have the specimen received by the OSPHL within 1-2 days of the specimen collection.**



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A Good Specimen Collection

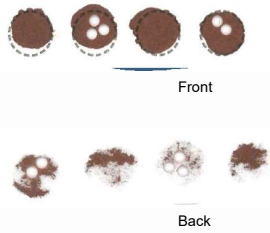
- The circles are completely filled
- Blood is evenly saturated
- Looks the same on both sides of filter paper
- No contamination evident
- No heat/humidity exposure
- Was laid flat to dry



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Uneven Saturation

- When blood applied with needle or capillary tube
- Touching filter paper with gloves or hands
- Other contaminate or damage to paper
- Hanging them to dry
- Poor blood flow!



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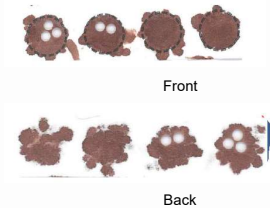
Uneven Saturation



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Layering

- Usually due to collecting multiple small spots to fill in the circle
- Happens when blood flow is poor



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Quantity Not Sufficient

- When not enough blood is applied within the circles
- When the drops of blood are too small to saturate the filter paper
- Inadequate blood flow

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Scratched or abraded

- Applying blood with a capillary tube or a needle
- Oversaturation can abrade the paper
- Not dried properly

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Odd color, or wet

- Specimen not dried sufficiently
- Sometimes if baby is very anemic pre-transfusion

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Clotted, layered, or super-saturated

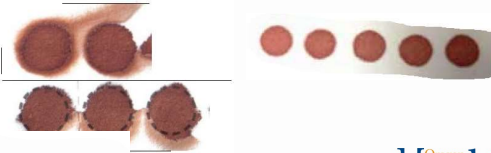
- Applying excess blood, usually from a device
- Blood applied from both sides of the filter paper
- Blood applied on top of dried or semi-dried or already-saturated blood- usually when a restick should have been done due to insufficient blood flow.



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Serum rings and contamination

- Not letting alcohol dry, not wiping away the first drop
- Contamination, with IV fluid, water, another substance
- Milking heel- sero sanguinous fluid
- Improper drying- exposed to heat or humidity, sunlight
- Cap tubes- can separate blood components



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I'm happy to help!

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Education



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Resource from: Babysfirsttest.org

Screening Checklist

First Trimester 11-14 weeks

- Bloodwork to measure nuchal translucency, free beta hCG, and PAPP-A
- Ultrasound to measure nuchal translucency, crown-rump length, and fetal heart rate
- Amniocentesis or chorionic villus sampling (CVS) to check for chromosomal changes
- Noninvasive prenatal testing (NIPT) to check for chromosomal changes
- Amniocentesis or CVS to check for genetic changes

Second Trimester 15-20 weeks

- Bloodwork to measure alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), and unconjugated estriol (uE3)
- Ultrasound to check for structural changes
- Amniocentesis or CVS to check for genetic changes

Third Trimester 21-30 weeks

- Bloodwork to check for gestational diabetes

Newborn Screenings 24-48 hours

- Bloodwork to check for metabolic and genetic changes

My Details

Maternal:

Fetal:

Appointment Note:

Prenatal & Newborn Screening

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Everything you need to know about your baby's health.



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