Newborn Screening in Washington State

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Objectives
- Newborn Screening Overview
- Process and Law
- Completing Collection Cards
- Video Clip
- Unsuitable Specimens
- Q & A

What is Newborn Screening?
Newborn screening is a public health system that detects infants with serious but treatable conditions that may not be apparent at birth.

There are 3 types of newborn screening programs:

Washington State Department of Health

Why is Newborn Screening Important?
- It prevents death and disability for thousands of infants every year in the USA by providing early treatment
- The public benefits through savings in health care and disability support costs

Newborn Screening Goal:
Correctly identify babies with congenital disorders and assure that they receive treatment as soon as possible.

Each year in Washington State...
The NBS Program performs approximately:
- 10 million tests
- on 172,000 specimens
- for 86,000 newborns
- and we identify:
- 200 babies every year who benefit from early diagnosis and treatment
- 1,200 infants with a hemoglobin trait (not disease)
Criteria for Screening
- A good screening test exists
- Diagnostic testing and treatment are available
- Early identification benefits the newborn
- Nature of the condition justifies population-based screening
- The benefits justify the costs of screening

Washington Screens for... 28 disorders!

Amino Acid Disorders (6)
- Phenylketonuria (PKU)
- Homocystinuria
- Maple Syrup Urine Disease
- Citrullinemia type I
- Argininosuccinic Acidemia
- Tyrosinemia type I

Organic Acid Disorders (8)
- Isovaleric Acidemia
- Isovaleric Aciduria
- Propionic Acidemia
- Holocarboxylase Synthetase Deficiency
- 3-Hydroxy-3-methylglutaric aciduria
- 3-hydroxy-3-methylglutaric aciduria
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Fatty Acid Oxidation Disorders (5)
- Medium-chain Acyl-CoA Dehydrogenase Deficiency
- Long-chain 3-0H Acyl-CoA Dehydrogenase Deficiency
- Trifunctional Protein Deficiency
- Very long-chain Acyl-CoA Dehydrogenase Deficiency
- Carnitine Uptake Defect

Other Disorders (9)
- Galactosemia
- Congenital Adrenal Hyperplasia
- Cystic Fibrosis
- Cystinosis
- Sickle Cell Disease & Hemoglobinopathies
- Severe Combined Immune Deficiency

Immediately Life Threatening Conditions

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WA Newborn Screening Process

Specimen Collection: Initial Newborn Screen
- Collect specimens after 18 hours of age
- Unless interfering substances are administered
- Inform parents about the importance of newborn screening
- Most affected babies appear healthy at birth
- Fill out screening cards with complete and accurate information
- Even cards where parents refuse screening

Note: Parents can legally refuse screening if it conflicts with their religious beliefs.

WA Newborn Screening Process

Disorders

28 disorders!
**Specimen Transit**
The initial blood specimen must be received by the Newborn Screening Laboratory within 72 hours of collection (excluding Sundays and Thanksgiving) (RCW 70.83.020)

- Dry specimens for 3 hours then send immediately
- Send each specimen in its own newborn screening envelope
- To ensure timely receipt, using a courier is recommended

Newborn Screening Lab receiving hours:
- Weekdays: 7:00 am – 5:00 pm
- Saturdays: 10:00 am – 12:00 pm
- Holidays: 8:00 am – 11:00 am
- Closed: Sundays and Thanksgiving

Note: Specimens received by 10:00 am on weekdays are tested the same day.

**WA Newborn Screening Process**

1. **Specimen Preparation**
   - Specimens sorted, checked for suitability, and accessioned

2. **Testing Preparation**
   - Eight punches are taken from each specimen for testing the 28 disorders

3. **Specimen Testing**
   - A variety of testing platforms are used to screen for the 28 disorders
   - Any abnormal result is repeated in duplicate for confirmation

| Hemoglobin Gel | Electrophoresis | Flow Cytometry
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**Laboratory Processing**

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Disorder Follow-Up
Reviews abnormal screening results and decides the appropriate follow-up actions for each baby.
These include:
- Immediately reporting abnormal results to the health care provider.
- Ensuring a subsequent specimen is submitted to resolve any borderline results.
- Facilitating diagnostic testing when needed.

Disorder Follow-up also:
- Assures proper medical care is taken for babies with confirmed diagnoses.
- Tracks abnormal results and confirms diagnoses over time.

WA Newborn Screening Process

Why Collect a 2nd Newborn Screen?
- Not mandated, but standard of care (ideally collected between 7 & 14 days).
- The first screen will miss some infants with these disorders, such as milder and later onset forms that may benefit from treatment.
- May be necessary for detection of some conditions and is critical for assessment of cystic fibrosis.
- Verifies hemoglobin traits, eliminating need for diagnostic lab work.

Quality Assurance & Development

Tracking & Reporting
- Send quarterly reports to each hospital about their performance in meeting newborn screening guidelines.
- Specimen Collection and Transit Timing Compliance.
- Specimen Quality.
- Demographic Errors.

Surveillance
- Ensure every baby receives a valid newborn screen.

Education & Outreach
- Promote newborn screening in the community.
- Provide technical assistance to health care facilities.
- Create educational materials on newborn screening.

Reporting and Evaluation
The Newborn Screening Program will publish an annual report regarding hospitals’ compliance in meeting the 72 hour transit and 48 hour specimen collection timelines. (RCW 70.83.020)

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<td>Specimen Quality</td>
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<td>2.0% Unsatisfactory specimens</td>
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<td>13.5% Demographic errors (cards with one or more errors)</td>
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Full annual report is available online at www.doh.wa.gov/nbs.
Special Considerations for Premature & Sick Infants
Infants who are receiving transfusions, nutritional support (HA/TPN), steroids, antibiotics or are in the intensive or special care nursery

- If possible, collect the first NBS before treatment (even if before 18 hours of age)
  - Except for antibiotics (no need to collect specimen prior to treatment)
- If collected after treatment, mark the appropriate boxes in the "Special Considerations" section of collection card
  - HA/TPN: if within 24 hours prior to specimen collection
  - Steroids: if within 7 days prior to specimen collection
  - Antibiotics: if within 24 hours prior to specimen collection
  - RBC Transfusions: indicate the date of child’s last transfusion
- A third specimen is recommended at 4-6 weeks of age for these infants

Good Drops, Bad Drops: Unsuitables

- What is an "unsuitable" specimen?
  - A specimen that was improperly collected, dried, or transported

- Types of unsuitable specimens:
  - INCSAT
  - DAMAGED
  - LAYERED
  - CONTAMINATED
  - PLASTIC
  - INSUB-H/A/S/E
  - TRANSPLED
  - TOO OLD

- Why does suitability matter?
  - Effects the accuracy of test results
  - Causes non-uniform analyte concentrations
  - Falsely elevated or lowered analytes
  - Risks delaying treatment for an affected baby

Most Common Unsuitables:
Incomplete Saturation or Insufficient Quantity (INCSAT)

- Problem?
  - Too little blood

- Solution:
  - Use large drops of blood
  - Piggyback drops within 10 seconds
  - Check for complete saturation after every circle
**Most Common Unsuitables:**
Layered/Clotted, or Supersaturated (LAYERED)

- **Problem?**
  - Too much blood

- **Solution:**
  - Do not re-apply blood after blood on card has begun to clot/dry
  - Apply blood to collection card on one side only
  - Do not over-saturate filter paper by applying too much blood

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**If Having Trouble Collecting a Good Specimen:**

**Try:**
- Hydrating baby before collecting specimen
- Warming the baby's heel before collecting
- Gently massaging the leg before heel-stick (makes the blood flow better)
- Have the baby feed during collection (breast or bottle)
- Ask parent to hold baby (comfort factor)
- Keep feet down/swaddle in upright position (gravity works!)

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**Best Practices for 1st Newborn Screens**

- Collect between 18-48 hours of age
- Or before interfering substances are administered
- Specimen is suitable for testing (enough blood, good quality)
- Dried for three hours
- Has complete and clear demographic information
  - Includes parental signature if testing refused
- Mailed same day to us, and arrives at the State Lab within 72 hours of collection

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**Case Study #1**

- Phone call from UW PKU clinic asking about a new referral
- Check NBS results – elevated IRT (screen for cystic fibrosis)
- Several phone calls later: clinic had called it an abnormal “PKU test”
- Crusade to change terminology!

We test for 28 disorders now, please call it the:
- Newborn screen
- Metabolic panel
- Heel-stick
- Dried blood spot test

- **PKU Test**
Case Study #2
- Day 13 – NBS staff determines 1st specimen was not collected for baby – verifies with birth hospital and requests clinic to collect NBS
- Day 13 – 1st screen collected
- Day 22 – NBS staff reports abnormal IRT, requests immediate 2nd specimen
- Day 23 – 2nd screen collected
- Day 30 – NBS staff recommends DX testing due to abnormal IRT on 2nd screen
- Day 34 – Baby diagnosed with CF

Case Study #3
- Initial hemoglobin results were indicative of transfusion (AA)
- Results from second NBS were inconsistent with first results (FA)
- Analyzed all four blood spots – obviously blood from two individuals: one baby and one adult

Case Study #3 Continued
- Baby did not bleed well and phlebotomist supplemented with someone else’s blood to fill circles
- Letter from our Program Director to Hospital Management about this dangerous practice

Case Study #4
- Day 1 - Baby born at a local birthing hospital (< 25 miles from NBS lab) NBS specimen collected at 26h
- Day 3 - Patient was admitted at tertiary hospital because of high blood ammonia levels
- Day 4 - Received a call from a metabolic specialist inquiring about NBS results - specimen not received in NBS lab
- Specimens sent via courier to Seattle Children’s lab confirming diagnosis of Propionic Acidemia
- Patient underwent dialysis
- Day 5 - NBS specimen received, STAT testing revealed elevated C3 (propionyl carnitine)
- Day 8 - recovered from metabolic crisis

Case Study #4 Continued
- We requested that the lab supervisor conduct an investigation regarding the delay
- Staff in charge of sending NBS specimens was out of the office for 2 days
- No adequate back-up system was in place, causing batching and courier issues
- Take home message: Timely collection and submission is of utmost importance to prevent death and disability!!!