Disorders Detected by Newborn Screening in Washington State

The Washington State Newborn Screening Program tests all infants born in the state for 28 rare but serious health disorders that can be treated if caught early in life. Disorders in red can be life-threatening during an infant’s first week of life.

### Amino Acid Disorders (6)
- Argininosuccinic acidemia (ASA)
- Citrullinemia type I (CIT)
- Homocystinuria (HCY)
- Maple syrup urine disease (MSUD)
- Phenylketonuria (PKU)
- Tyrosinemia type I (TYR-I)

### Fatty Acid Oxidation Disorders (5)
- Carnitine Uptake Defect (CUD)
- Long-chain L-3-OH Acyl-CoA dehydrogenase (LCHAD) deficiency
- Medium-chain Acyl-CoA dehydrogenase (MCAD) deficiency
- Trifunctional protein deficiency (TFP)
- Very-long-chain acyl-CoA dehydrogenase (VLCAD) deficiency

### Organic Acid Disorders (8)
- 3-hydroxy-3-methylglutaric aciduria (HMG)
- β-ketothiolase (BKT) deficiency
- Glutaric acidemia type I (GA-I)
- Isovaleric acidemia (IVA)
- Methylmalonic acidemias (CblA/B and MUT)
- Multiple carboxylase deficiency (MCD)
- Propionic acidemia (PA)

### Other Disorders (9)
- Biotinidase (BIOT) deficiency
- Congenital adrenal hyperplasia (CAH)
- Congenital hypothyroidism (CH)
- Cystic fibrosis (CF)
- Galactosemia (GALT)
- Severe combined immunodeficiency (SCID)
- Sickle cell diseases & Hemoglobinopathies (Hb)
**Newborn Screening DOs & DON’Ts**

**DO ...**

- **DO...** call it the newborn screen, metabolic panel, heel-stick, or dried blood spot test
- **DO...** use same newborn screening kit (pink hearing card and dried blood spot card) for each child - the barcode numbers should match
- **DO...** accurately complete all the information requested on the card
- **DO...** double check the blood specimen collected on the card matches the demographic information written (especially with multiple births, twins or triplets)
- **DO...** collect the 1st specimen in the recommended timeframe of 18-48 hours of age (must be collected before 48 hours per state law)
- **DO...** collect a 2nd specimen for all infants between 7 and 14 days of age
- **DO...** practice aseptic collection techniques by:
  - wearing gloves and using sterile lancets
  - avoiding contamination with urine, feces, or other interfering substances
  - completely filling each blood circle before moving onto the next, allowing blood to soak through the filter paper
- **DO...** air dry specimens horizontally for 3 hours before closing the flap
- **DO...** send specimens to the State Newborn Screening laboratory as soon as it is dry (initial specimen cards must be received at the State Lab within 72 hours of collection per state law)
- **DO...** call us for high priority specimens that require STAT testing (i.e. clinical symptoms or family history of newborn screening disorder)

**DON’T ...**

- **DON’T...** wait to send more than one day’s set of specimens (“batching” cards can delay testing & ultimately diagnosis & treatment of babies with life-threatening disorders)
- **DON’T...** collect blood less 18 hours after delivery [unless interfering substances will be administered (HA/TPN, antibiotics, steroids, RBC transfusion), or if the baby will not be with a medical provider in the recommended timeframe of 18-48 hours]
- **DON’T...** use EDTA tubes (purple top) to collect blood
- **DON’T...** expose specimens to heat, direct sunlight, humidity, or place in plastic bags
- **DON’T...** contaminate or scratch the filter paper
- **DON’T...** collect blood on both sides of the filter paper
- **DON’T...** check the “twin A” box if baby is a single birth (singleton)
- **DON’T...** mark or place stickers in the “do not use” section of the collection card
- **DON’T...** call it the “PKU test!” (We screen for a number of other disorders in addition to PKU)
- **DON’T...** re-apply blood to the same circle once blood on the card has already dried
Newborn Screening Collection Card Instructions

Please use your supply of existing older cards prior to use of the new cards.

As a reminder, please:

- Complete Birth Facility field
- Identify the Follow-Up Care ID# for the baby’s outpatient clinic or facility (midwives use “M#”)
- If applicable, use check boxes to indicate same ID number(s)
- Newest cards are in royal purple ink

If parents refuse newborn screening for religious reasons:

- Have parents read the Refusal of Testing statement on the back of the screening card
- Complete all demographic information on the front of the card AND check the box indicating “Refused”
- Parents must sign and date to indicate refusal of testing
- Mail refusal cards to the State Laboratory right away, just like a blood specimen

Please:

- Do not place stickers/tracking labels over any demographic information or the “DO NOT USE THIS AREA” section
- Do not separate the filter paper from the demographic information. The barcode number for the filter paper, demographic information section, and hearing card (if present) must match for each child
- Keep record of the unique barcode number in the child’s chart and/or on a tracking log of screening specimens submitted

For people with disabilities, this document is available upon request in other formats. To submit a request, please call 1-800-525-0127 (TDD/TTY call 711).
Newborn Screening Collection Cards Instructions

**Child’s Information**
- Write the date AND time the child was born
- Write the date AND time the specimen was collected
  - Use 24-hour based time OR check appropriate AM/PM boxes
  - Tests are specific to the child’s exact age (in hours) when the specimen was collected
- Write the child’s legal name and Medical Record # (if known)
- Write the sex and birth order of the child
  - This ensures the correct child is being identified
- Write the weight of the child at birth in grams OR pounds/ounces
  - Do not use commas or other punctuation
- For Race/Ethnicity, check all boxes that apply (if known)

**Child’s Special Considerations**
- Check NICU box if child is or will be in the Intensive Care Unit or Special Care Nursery
- Check HA/TPN box if the child received hyperalimentation/total parenteral nutrition, or IV supplementation including amino acids in the last 24 hours
- Check STERIODS box if the child received steroids in the last 7 days
- Check ANTIBIOTICS box if the child received antibiotics in the last 24 hours
- Check TRANSFUSED box if the child received red blood cell transfusion
  - Indicate the date the child was last transfused with red blood cells

**Mother’s Information**
- Write mother’s legal first and last name (Do not include middle names)
- Check box if the mother received steroids within the last 7 days
- Indicate the date when steroids were last administered to the mother

**Miscellaneous Information**
- Indicate anything relevant, such as: adoption, foster care, surrogacy, CPS, family history of NBS disorders, moving/transferring out of state

**Birth Facility**
- Write the ID# for the hospital or birth center where the infant was born
- The card’s yellow flap has a list of all birth facility ID#s for your use
- If home birth, write the individual midwife ID# (“M#”)

**Submitter ID**
- Write the ID# for the facility where the specimen was collected
- If home collection, write the individual midwife ID# (“M#”)
- Or check the box if same as birth facility
- Test results will be mailed to the submitter
- Check box if parents refuse testing AND obtain signature on back of card

**Follow-Up Care**
- Write the ID# of the facility where the child will receive outpatient care*
- If child will remain in-house, write the hospital’s ID#
- Or check the box if same as submitter
- This facility will be contacted when abnormal results require follow-up
  *No longer use individual provider ID#s

**Refused**
- Check box if parents refuse testing AND obtain signature on back of card

Complete list of ID numbers available online: [www.doh.wa.gov/NBS/IDNumberDirectories](http://www.doh.wa.gov/NBS/IDNumberDirectories)
Why is newborn screening done?

Newborn screening tests infants shortly after birth for a number of rare but treatable congenital disorders. If left untreated, these disorders can lead to stunted growth, blindness, brain damage, and sometimes death. Infants with these conditions usually appear normal at birth and, without screening, the disorders are not likely to be detected and treated before causing severe disability or death.

How common are newborn screening disorders?

Each year, about 200 infants born in Washington State are diagnosed with one of the 28 disorders included in the State screening panel and need prompt medical attention to prevent disability or death.

Some parents only want one newborn screen – which one should they do?

The first one – some disorders can be life threatening within the baby’s first week of life. The first screen, collected between 18 to 48 hours of age, is critical for early detection and treatment to avoid severe disability or death (it is also the screen required by law).

Why collect a second screen?

The first screen will miss a small number of infants with the conditions, including severe, later-onset, or milder forms of the disorders. This is why a second screen is recommended between 7 to 14 days of age to catch anything not found on the first. If it is uncertain whether an infant has received newborn screening, a screen may be collected up to six months of age.

Who collects newborn screens?

It is the birth hospital or out-of-hospital birth attendant’s responsibility to ensure that the initial newborn screening specimen is collected before 48 hours of age, even if they do not collect it themselves. If an infant is transferred to another hospital, NICU, or provider, the transfer facility/provider may collect the specimen.

Most routine second screens are collected at the infant’s two-week well-child appointment at their pediatrician’s clinic, though many are also collected at a hospital, laboratory, or by a midwife.

(Over)
What if the baby is not with a medical provider during the recommended 18-48 hour collection timeframe?

We recommend the initial screen is collected before the infant is discharged home from the hospital or, for home births, before the birth attendant leaves the family’s home, even if this means collection prior to 18 hours of age. This complies with the law and is valuable for early detection of life threatening disorders.

Note: It is not necessary to wait for an infant to feed before collecting the newborn screening specimen.

How is newborn screening paid for?

Newborn screening is fully supported by fees collected for the newborn screening laboratory testing services. The Department of Health bills the facility that collected the infant’s initial screen and the facility typically then bills the fee to patient insurance. The fee is one-time and includes all screens a child receives, such as the routine second screen or any repeat screens needed.

The infant’s family has no history of these conditions – why do screening?

Since the disorders are so rare, most children who are born with these conditions come from families with no previous history of the disorders.

Can parents refuse screening?

Parents can legally refuse screening if it conflicts with their religious beliefs. Other concerns about newborn screening, such as not wanting to poke the baby, the collection timeframe isn’t convenient, not wanting to pay for testing, or privacy concerns are not valid reasons for refusing screening. We ask health care providers help explain to families the risk of refusal and the importance of newborn screening if they have these concerns so their babies can benefit from newborn screening. There are instructions on NBS collection cards on how to legally document when parents refuse testing for religious purposes.

What if a screen is improperly collected or transported?

Improper collection or handling may cause a specimen to be unsuitable. Our laboratory tests all unsuitable specimens, however, the results are considered invalid and a repeat newborn screen is required from the submitting facility or from the infant’s follow-up care clinic. If extreme values are found on an unsuitable specimen, the primary care provider will be contacted.

Questions? Contact Us!
Newborn Screening Program
Phone: 206-418-5410
E-mail: NBS.Prog@doh.wa.gov
Website: www.doh.wa.gov/nbs
### DISORDERS DETECTED BY THE WASHINGTON NEWBORN SCREEN (2014)

**Table 1:** Disorders on this page can be deadly if not detected and treated within days following birth.

<table>
<thead>
<tr>
<th>Disorder (Prevalence in WA)</th>
<th>Definition</th>
<th>Screening Test</th>
<th>Impact without Early Treatment</th>
<th>Treatment</th>
<th>Benefits of Early Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Galactosemia (1 in 40,000)</td>
<td>Inability to break down galactose, a major sugar found in milk</td>
<td>Measure activity of enzyme needed to break down galactose</td>
<td>Severe intellectual and developmental disability, liver disease, blindness, overwhelming infections and death</td>
<td>Dietary restriction of milk and other foods containing galactose</td>
<td>Prevent death, improve mental function &amp; reduce other morbidity</td>
</tr>
<tr>
<td>Congenital Adrenal Hyperplasia (CAH) (1 in 16,000)</td>
<td>Impaired production of cortisol and other adrenal hormones</td>
<td>Measure adrenal hormone: 17-hydroxyprogesterone (17-OHP) level</td>
<td>Salt loss &amp; shock may result in early sudden death, also virilization &amp; abnormal growth</td>
<td>Cortisol &amp; salt-retaining hormone replacement</td>
<td>Prevent death, reduce virilization &amp; abnormal growth</td>
</tr>
<tr>
<td>Organic Acid Disorders (1 in 25,000) (see list below)</td>
<td>Inability to process or break down organic acids, byproducts of protein and fatty acid metabolism</td>
<td>Measure acylcarnitine levels by tandem mass spectrometry</td>
<td>Severe nerve and physical damage &amp; death</td>
<td>Dietary restriction of offending amino acid(s) &amp; needs special metabolic formula</td>
<td>Prevent death, intellectual and developmental disability and other neurological damage</td>
</tr>
<tr>
<td>Fatty Acid Oxidation Disorders (1 in 13,000) (see list below)</td>
<td>Inability to process or break down fats in the body</td>
<td>Measure acylcarnitine levels by tandem mass spectrometry</td>
<td>Serious damage to brain, liver, heart, eyes, muscles &amp; death</td>
<td>High carbohydrate, low-fat diet &amp; avoidance of fasting</td>
<td>Prevent death, intellectual and developmental disability and other neurological damage</td>
</tr>
<tr>
<td>Amino Acid Disorders (1 in 10,000) (see list below)</td>
<td>Inability to break down amino acids, found in all foods containing protein</td>
<td>Measure amino acid levels by tandem mass spectrometry</td>
<td>Intellectual and developmental disability, seizures, coma &amp; death</td>
<td>Dietary restriction of offending amino acid(s) &amp; needs special metabolic formula</td>
<td>Prevent death, intellectual and developmental disability and other neurological damage</td>
</tr>
</tbody>
</table>

### Amino Acid Disorders

* Argininosuccinic acidemia (ASA)  
* Citrullinemia (CIT)  
* Homocystinuria (HCYS)  
* Maple Syrup Urine Disease (MSUD)  
* Phenylketonuria (PKU)  
* Tyrosinemia type I (TYR-I)  

### Organic Acid Disorders

* 3-OH 3-CH3 glutaric aciduria (HMG)  
* Glutaric acidemia type I (GA-I)  
* Beta-Ketothiolase deficiency (BKT)  
* Isovaleric acidemia (IVA)  
* Methylmalonic acidemia (Cbl A, B)  
* Methylmalonic acidemia (mutase deficiency) (MUT)  
* Multiple carboxylase deficiency (MCD)  
* Propionic acidemia (PROP)  

### Fatty Acid Oxidation Disorders

* Long-chain L-3-OH acyl-CoA dehydrogenase (LCHAD) deficiency  
* Medium chain acyl-CoA dehydrogenase (MCAD) deficiency  
* Trifunctional protein (TFP) deficiency  
* Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency  

* Not all amino acid, organic acid and fatty acid oxidation disorders are life-threatening within days of birth. The disorders noted by an asterisk can be deadly if not detected and treated within days of birth
## DISORDERS DETECTED BY THE WASHINGTON NEWBORN SCREEN (2014)

Table 2: Disorders on this page are not deadly within days of birth, but delay in treatment may result in later death or profound, permanent disability

<table>
<thead>
<tr>
<th>Disorder (Prevalence in WA)</th>
<th>Definition</th>
<th>Screening Test</th>
<th>Impact without Early Treatment</th>
<th>Treatment</th>
<th>Benefits of Early Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sickle Cell Diseases and Hemoglobinopathies (1 in 10,000)</td>
<td>Production of abnormal hemoglobin</td>
<td>Separate and visualize hemoglobin proteins by isoelectric focusing, with confirmation by high performance liquid chromatography and DNA analysis</td>
<td>Severe infections and possible death within 2-3 months following birth</td>
<td>Antibiotic prophylaxis to help prevent infections &amp; parental education to recognize health crises</td>
<td>Prevent death, reduce infections and other morbidity</td>
</tr>
<tr>
<td>Congenital Hypothyroidism (1 in 1,600)</td>
<td>Inadequate production of thyroid hormone</td>
<td>Measure thyroid stimulating hormone (TSH) level</td>
<td>Intellectual and developmental disability, growth failure</td>
<td>Thyroid hormone replacement</td>
<td>Normal growth and mental development</td>
</tr>
<tr>
<td>Biotinidase Deficiency (1 in 60,000)</td>
<td>Deficiency of biotin, part of the Vitamin B complex</td>
<td>Measure activity of enzyme needed to recycle biotin</td>
<td>Seizures, damage to immune system, intellectual and developmental disability, hearing loss</td>
<td>Oral biotin supplementation</td>
<td>Prevent all adverse consequences</td>
</tr>
<tr>
<td>Cystic Fibrosis (1 in 5,000)</td>
<td>Defect in the cystic fibrosis transmembrane conductance regulator (CFTR) gene</td>
<td>Measure immunoreactive trypsinogen (IRT) level</td>
<td>Significant nutritional deficits due to thick, sticky mucus in the digestive system. Severe lung infections due to mucus</td>
<td>Pancreatic enzymes, vitamin supplements, chest physiotherapy, antibiotics</td>
<td>Improve physical growth, cognitive function &amp; possibly lung function</td>
</tr>
<tr>
<td>Severe Combined Immunodeficiency (1 in 45,000)</td>
<td>Complete lack of immune system in the baby</td>
<td>DNA test: measure number of T-cell excision circles (TRECs) by real-time PCR</td>
<td>Severe life-threatening infections that complicate treatment and possible death</td>
<td>Stem-cell transplant or gene therapy, depending on the genotype of the baby</td>
<td>Prevent death and cure the condition</td>
</tr>
</tbody>
</table>
Some specimens are considered unsatisfactory due to the quality of specimen collection or handling. These specimens are tested for extreme values but another specimen must be obtained to complete screening. The need to obtain a repeat specimen could delay diagnosis and treatment of an affected infant.

### Unsatisfactory Specimen Errors

<table>
<thead>
<tr>
<th>Error</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Layered or Supersaturated</td>
<td>Blood was layered, clotted or supersaturated. Caused by:</td>
</tr>
<tr>
<td></td>
<td>• Repeated application of blood to the same filter paper circle</td>
</tr>
<tr>
<td></td>
<td>• Blood applied to both sides of the filter paper</td>
</tr>
<tr>
<td></td>
<td>• Blood clotting in a capillary tube</td>
</tr>
<tr>
<td></td>
<td>• Application of too much blood</td>
</tr>
<tr>
<td>Incompletely Saturated</td>
<td>Blood did not completely soak through the filter paper or not enough blood on the filter paper. Caused by:</td>
</tr>
<tr>
<td></td>
<td>• Filter paper circles not fully saturated or not completely filled</td>
</tr>
<tr>
<td></td>
<td>• Application of small blood spots</td>
</tr>
<tr>
<td></td>
<td>• Blood applied to both sides of the filter paper</td>
</tr>
<tr>
<td>Contaminated</td>
<td>Blood was diluted, discolored, contaminated or exhibited serum rings. Caused by:</td>
</tr>
<tr>
<td></td>
<td>• Alcohol not completely drying before skin puncture</td>
</tr>
<tr>
<td></td>
<td>• Puncture site squeezed or 'milked' to expel blood</td>
</tr>
<tr>
<td></td>
<td>• Improper drying of specimen</td>
</tr>
<tr>
<td></td>
<td>• Exposure to high temperatures</td>
</tr>
<tr>
<td></td>
<td>• Filter paper contact with gloved or ungloved hands, or by substances such as alcohol, feeding or antiseptic solutions, hand lotion or powder</td>
</tr>
<tr>
<td>Specimen Too Old</td>
<td>Specimen was delayed in transit and is too old due to deterioration of the dried blood spots. Caused by:</td>
</tr>
<tr>
<td></td>
<td>• Specimens received more than 14 days after collection are too old for hemoglobin and galactosemia testing</td>
</tr>
<tr>
<td></td>
<td>• Specimens received more than 30 days after collection are too old for all tests</td>
</tr>
<tr>
<td>Abraded</td>
<td>Specimen surface was scratched, dented, or abraded. Caused by:</td>
</tr>
<tr>
<td></td>
<td>• Improper application of blood with capillary tube or other device</td>
</tr>
<tr>
<td>Partial Unsuitable</td>
<td>Validation of the preliminary screening results was not possible due to the unsuitability of the residual blood. Caused by:</td>
</tr>
<tr>
<td></td>
<td>• Partial abrasion, contamination, damage, or oversaturation of residual blood</td>
</tr>
<tr>
<td></td>
<td>• Insufficient quantity of blood</td>
</tr>
<tr>
<td>Ambiguous Degradation</td>
<td>Hemoglobin screening results indicate degradation or chemical modification of hemoglobins present causing assay interference.</td>
</tr>
<tr>
<td>Damaged Specimen</td>
<td>Specimen was damaged during transport and may be ripped or contaminated by rain and/or other substances.</td>
</tr>
<tr>
<td>Old Collection Card</td>
<td>Specimen was submitted on a collection card past its expiration date. Cards expire three years after their manufacture date.</td>
</tr>
<tr>
<td>Received in Plastic</td>
<td>Specimen was received in a sealed plastic bag and may be damaged by heat exposure and moisture accumulation.</td>
</tr>
</tbody>
</table>
Special attention is required when infants are premature, sick, or are administered substances that interfere with newborn screening tests. Please follow these Special Considerations to assure that all infants receive appropriate newborn screening.

Whenever possible, collect the infant’s newborn screening specimen prior to administering any of the following interfering substances:

- HA/TPN
- Steroids
- RBC Blood Transfusion
- Dopamine

When an infant has received interfering substances prior to specimen collection, check the appropriate box in the Special Considerations section of the newborn screening collection card if the substance was received in the timeframe below:

- NICU: When an infant is or will be in the NICU or Special Care Nursery
- HA/TPN: If the child received HA/TPN within 24 hours prior to specimen collection
- Steroids: If the child received steroids within 7 days prior to specimen collection*
- Antibiotics: If the child received antibiotics within 24 hours prior to specimen collection
- RBC Blood Transfusions: Indicate the date of child’s last transfusion
- Dopamine: Write this substance in the “Miscellaneous Information” section

Timing of Newborn Screening Specimens

1st Screen: Collect the first screen before interfering substances are administered, even if prior to 18 hours of age. If no therapies are administered, the first specimen should be collected during the regular recommended timeframe of 18 to 48 hours of age. The first specimen is mandatory and must be collected no later than 48 hours of age and received by the Newborn Screening Laboratory within 72 hours of collection.

2nd Screen: A second screen is recommended for all infants (including healthy infants) between 7-14 days of age.

3rd Screen: A third specimen is recommended for all premature, sick (requiring three or more weeks of hospitalization), low birth weight infants (under 1500g), or infants that have received interfering substances.

- For RBC transfusions, a subsequent screen should be collected at least 4 weeks after the last transfusion
- For infants that receive steroids, a screen should be collected at least 7 days after discontinuing therapy
- Otherwise, the third specimen should be collected at 4-6 weeks of age or just prior to hospital discharge, whichever is sooner.

Additional recommendations:

- If the mother received steroids within 7 days prior to newborn screening specimen collection, please indicate this in the space provided in the “Mother’s Information” section of the collection cards*
  * Please take care to correctly indicate whether the mother or child has received steroids
- Do not collect specimens using tubes containing EDTA (purple tops) as it will interfere with screening tests
- Please note that it is no longer recommended to collect specimens prior to administering antibiotics