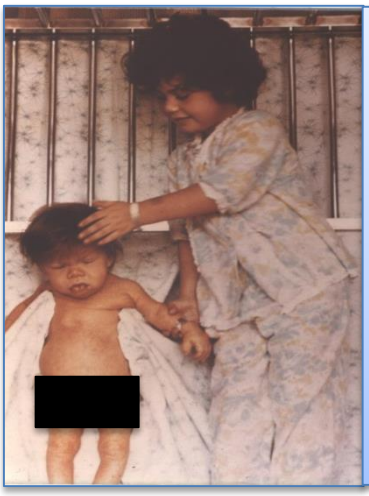
Newborn Screening
Blood Spot Test

Approximately **200** infants each year are saved through newborn blood spot screening

Why?

Without screening, these rare disorders are not likely to be detected and treated before causing severe disability or death.



Two 6 year old girls with congenital hypothyroidism.

The girl on the left was not treated until after symptoms appeared.

The girl on the right was detected through screening and treated early.

How?

The baby's heel is pricked to collect several drops of blood onto a filter paper card that is then dried and sent to the State Laboratory in Shoreline, WA for testing.

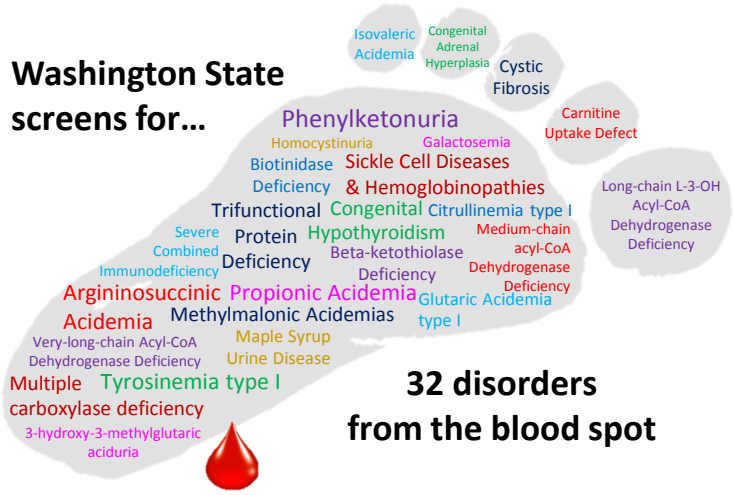
The State Newborn Screening Program works closely with the baby's health care provider to provide screening results and recommend further evaluation and/or diagnostic testing if a disorder is suspected.

When?

1st screen: shortly after birth, when the baby is 18 – 48 hours old (This screen is required)

2nd screen: at a well-child visit, when the baby is 7 – 14 days old (This screen is standard of care)

Washington State screens for…



32 disorders from the blood spot

Families Should Know:

- Babies with these disorders usually look healthy at birth.
- Serious problems can be prevented with screening and early treatment.
- Babies detected and treated early can grow up to be healthy individuals.
- All newborns are required by Washington State law to get a blood spot screen before they are two days old to ensure healthy outcomes.
- A second screen is recommended because some of the disorders may not be detected until the baby is older.

Washington State Department of Health ◆Newborn Screening Program

Phone: 206-418-5410 | Fax: 206-363-1610 | E-mail: NBS.Prog@doh.wa.gov | Website: www.doh.wa.gov/nbs

Washington State Department of Health

DOH 304-118 (NBS) Aug 2020

COMING SOON!

New Conditions!

The State Board of Health approved three additional conditions to the state screening panel. Now all babies born in Washington State will be screened for a total of 32 rare, serious conditions that are treatable when found early in life.

Glycogen Storage Disease Type II (Pompe)

Babies with Pompe have problems processing a type of sugar found in food. When this sugar builds up, it causes muscle weakness and heart problems. Treatment with enzyme replacement therapy can save the lives of infants with Pompe.

- Occurs in ~ 1 in 28,000 births
- Pompe is known as "floppy baby syndrome"
- Screening began in late 2019
- Is a lysosomal storage disorder

Mucopolysaccharidosis Type I (MPS I)

Babies with MPS I have problems processing a certain type of sugar found in food. This causes a build up in the body that leads to skeleton, organ, and brain problems. Treatment with a stem cell transplant prevents the loss of IQ points and other physical problems.

- Occurs in ~ 1 in 35,700 births
- Severe MPS I is also called "Hurler syndrome"
- Screening began in late 2019
- Is a lysosomal storage disorder

Spinal Muscular Atrophy (SMA)

Babies with SMA are unable to produce a protein critical to the function of motor neurons — nerves that control muscles. The motor neurons eventually die and cannot be regenerated leading to loss of the ability to walk, eat, or breathe. Treatments and therapies early in life can slow disease progression.

- Occurs in ~ 1 in 10,000 births
- Leading genetic cause of infant death in the United States
- Screening begins Summer 2020

Saving lives with a simple blood spot

www.doh.wa.gov/nbs
206-418-5410
NBS.Prog@doh.wa.gov

Washington State Department of Health

304-122 Jul 2019

Saving lives with a simple blood spot

Disorders Detected by Newborn Screening in Washington State

The Washington State Newborn Screening Program tests all infants born in the state for 32 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)

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)

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

Endocrine Disorders (2)

Congenital adrenal hyperplasia (CAH)
Congenital hypothyroidism (CH)

Lysosomal Storage Disorders (2)

Glycogen storage disease type II (Pompe)
Mucopolysaccharidosis type I (MPS I)

Other Disorders (6)

Biotinidase (BIOT) deficiency
Cystic fibrosis (CF)
Galactosemia (GALT)
Severe combined immunodeficiency (SCID)
Sickle cell diseases & Hemoglobinopathies (Hb)
X-linked adrenoleukodystrophy (X-ALD)
Spinal Muscular Atrophy (SMA)

Washington State Department of Health ◆ Newborn Screening Program
Phone: 206-418-5410 | Fax: 206-363-1610 | E-mail: NBS.Prog@doh.wa.gov |Website: www.doh.wa.gov/nbs

Washington State Department of Health
DOH 951-124 Aug 2020

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 biohazard 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… call it the "PKU test!" (We screen for a many other disorders in addition to PKU)

DON'T… wait to send more than one day's set of specimens ("batching" cards can delay diagnosis & treatment for 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 enclose in plastic bags

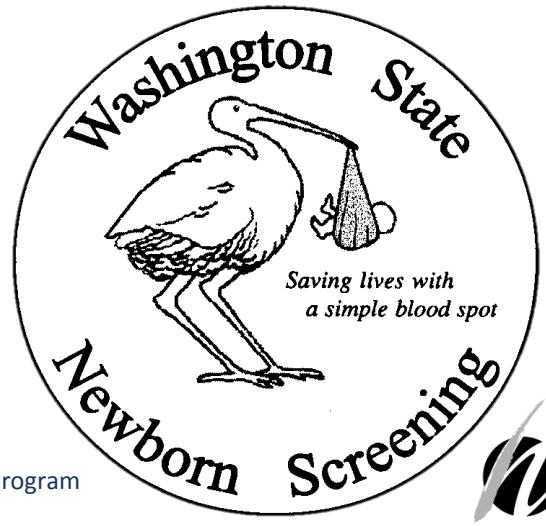
DON'T… contaminate or scratch the filter paper

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… re-apply blood to the same circle once blood on the card has already dried



Washington State Department of Health • Newborn Screening Program
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Email: NBS.Prog@doh.wa.gov • www.doh.wa.gov/nbs

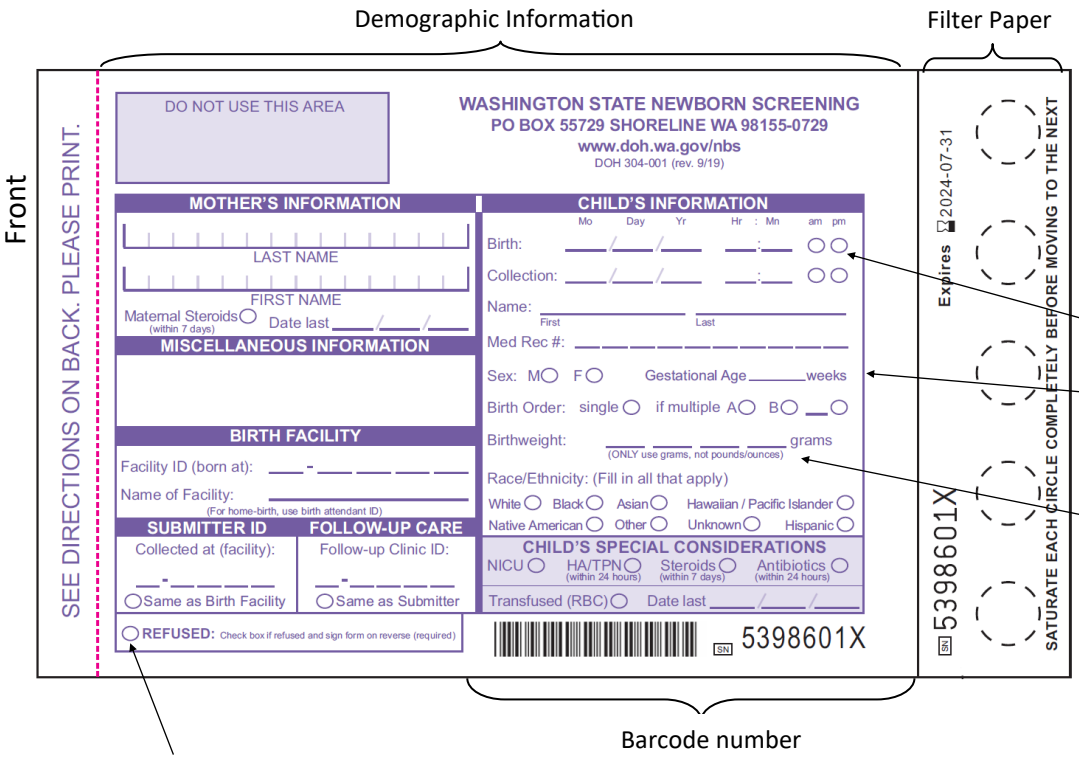
Washington State Department of Health

DOH 951-119 June 2015

Newborn Screening Collection Card Instructions

DOH 951-125 Sep 2019

Collection Cards — Fall 2019



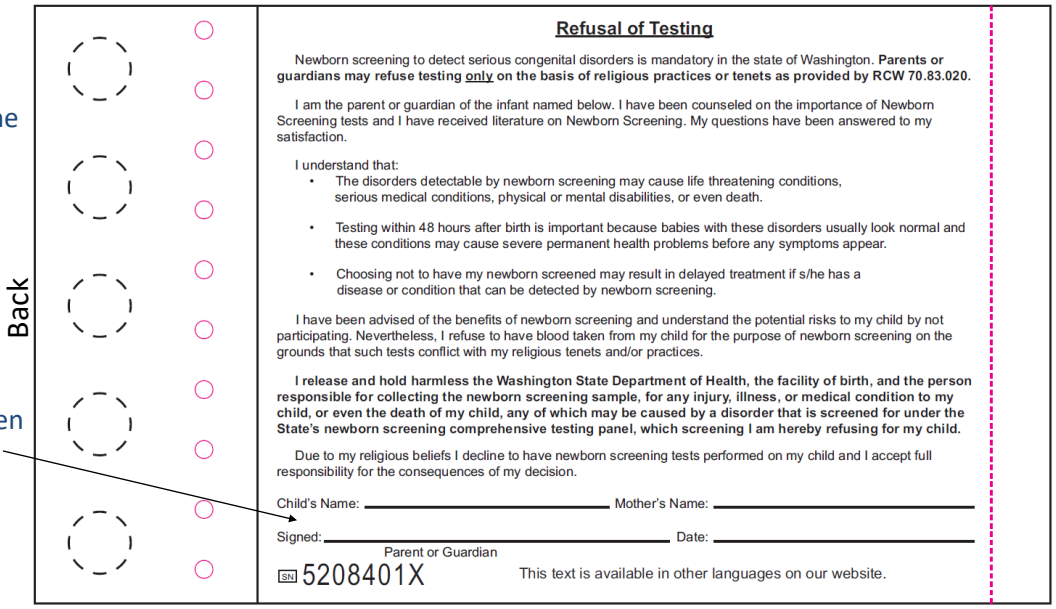
Please use your supply of existing older cards prior to use of the new cards.

Key features of new cards:

- Now "bubbles" instead of "boxes. Please fill in bubbles completely
- New Field— Gestational Age! Please write in full weeks. Do not use days or decimals.
- Birth weight in grams only now Do not use kilogram, pounds/ounces
- 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. Text is available on our website in other languages for reference only.
- Complete all demographic information on the front of the card AND check the box indicating "Refused"
- Parents must sign and date specimen card to indicate refusal of testing
- Mail refusal cards to the State Laboratory right away, just like a blood specimen



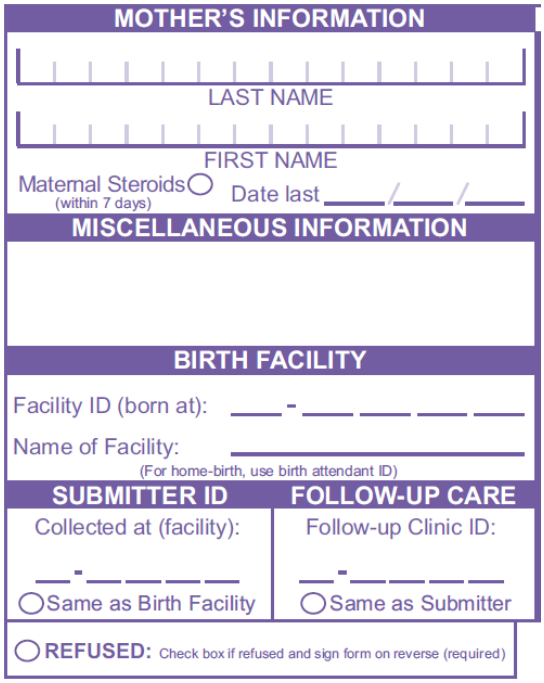
Please:

- 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

Front Left



Complete list of ID numbers available online:
www.doh.wa.gov/NBS/IDNumberDirectories

Mother's Information

- Write mother's legal first and last name (Do not include middle names)
- Fill in bubble 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 fill the bubble if same as birth facility
- Test results will be mailed to the submitter

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 fill the bubble 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

Front Right

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 fill appropriate AM/PM bubbles
    - ♦ 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)
- Fill the bubble for 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
  - — Do not use pounds/ounces, kilograms, or punctuation
- For Race/Ethnicity, fill all bubbles that apply (if known)

Child's Special Considerations

- Fill the NICU bubble if child is or will be in the Intensive Care Unit or Special Care Nursery
- Fill the HA/TPN bubble if the child received hyperalimentation/total parenteral nutrition, or IV supplementation including amino acids **in the last 24 hours**
- Fill the STERIODS bubble if the child received steroids **in the last 7 days**
- Fill the ANTIBIOTICS bubble if the child received antibiotics **in the last 24 hours**
- Fill the TRANSFUSED bubble if the child received red blood cell transfusion

Saving lives with a simple blood spot

Washington State Newborn Screening Frequently Asked Questions

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 32 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)

Washington State Department of Health
DOH 951-122 Aug 2020

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.

Washington State Department of Health

Questions? Contact Us!
Newborn Screening Program
Phone: 206-418-5410
E-mail: NBS.Prog@doh.wa.gov
Website: www.doh.wa.gov/nbs

PUBLIC HEALTH
ALWAYS WORKING FOR A SAFER AND HEALTHIER COMMUNITY

Washington State Newborn Screening Program
Key: Unsatisfactory Specimen Descriptions

Washington State Department of Health
DOH 951-143-Unsatisfactory Key
Apr 2015

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

Newborn Screening Special Considerations

Newborn screening for premature & sick infants

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

Washington State Department of Health | ***Newborn Screening Program***
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Website: www.doh.wa.gov/nbs

Washington State Department of Health
DOH 951-121 Mar 2018

Saving lives with a simple blood spot

Disorders Detected by Newborn Blood Spot Screening

Washington State Department of Health
DOH 951-145 Aug 2020

The Washington State Newborn Screening Program tests all infants born in the state for a set of rare but serious health disorders that can be treated if caught early in life. Washington State screens for 32 disorders.

Galactosemia
1 in 11,000 births

Babies with galactosemia cannot digest galactose, a sugar present in milk. When babies drink milk (including breast milk), galactose builds up in the body and can cause blindness, mental disability, or death. A lifelong diet without milk products can prevent these complications.

Congenital Hypothyroidism
1 in 950 births

Babies with congenital hypothyroidism do not produce enough thyroid hormone to grow and develop normally. Early treatment with thyroid medication can prevent developmental disability and ensure normal growth and development.

Cystic Fibrosis
1 in 5,500 births

Babies with cystic fibrosis develop poor lung function and struggle with malnutrition. This leads to serious health problems and a shortened lifespan. Early treatment can improve growth and development, and decrease the risk of infections and other complications.

Biotinidase Deficiency
1 in 86,000 births

Babies with biotinidase deficiency cannot efficiently use a vitamin called biotin. If untreated, this can cause rashes, hearing loss, seizures and developmental delay. Lifelong treatment with biotin supplements can prevent these problems.

Congenital Adrenal Hyperplasia
1 in 14,000 births

Babies with congenital adrenal hyperplasia have adrenal glands that cannot make enough of the hormones needed for healthy body function. These infants can have life-threatening episodes of dehydration and coma. Baby girls may have abnormal genitalia. Early treatment to replace the needed hormones can restore healthy body function.

Severe Combined Immunodeficiency
1 in 88,000 births

Babies with severe combined immunodeficiency are born without a working immune system. They cannot fight germs that cause disease and even the most common infections can be life-threatening. A bone-marrow transplant early in life cures the baby by giving them a working immune system to prevent and fight infections.

Sickle Cell & Hemoglobinopathies
1 in 4700 births

Babies with sickle cell disease or other hemoglobinopathies have abnormal red blood cells that are unable to carry oxygen efficiently throughout the body. These disorders can cause frequent infection, severe pain, anemia and other complications. Early treatment and proper lifelong management can prevent serious health problems. Note: Some babies have a hemoglobin trait; this is not a disease and will not affect their health.

Fatty Acid Oxidation Disorders (5)
1 in 11,000 births

Babies with fatty acid oxidation disorders cannot use fats in the body for energy. If these babies do not eat often, severe damage to the heart, liver and other organs can occur. If untreated, this will result in serious health problems and sometimes death. A special lifelong diet, eating frequently, and medications can help prevent these problems.

Organic Acid Disorders (9)
1 in 29,000 births

Babies with organic acid disorders cannot digest certain parts of proteins found in food. If untreated, harmful substances build up in their blood and urine, which can have serious effects on their health, growth, and learning and can result in death. This can be prevented by early treatment with a special lifelong diet, close monitoring, and medications.

Amino Acid Disorders (6)
1 in 10,000 births

Babies with amino acid disorders cannot process foods containing protein. If untreated, amino acids (the building blocks of protein) and other toxic substances build up in the body and have serious effects on health, growth and learning and can result in death. A special lifelong diet and supplements can help prevent these problems. An example of an amino acid disorder is phenylketonuria (PKU).

Saving lives with a simple blood spot

Disorders Detected by Newborn Blood Spot Screening

Washington State Department of Health
DOH 951-145 Aug 2020

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X-Linked Adrenoleukodystrophy
1 in 17,000 births

Babies with X-linked adrenoleukodystropy cannot break down very long chain fatty acids. The build-up of these fats may cause fatty covering of the nerves, brain, and spinal cord (myelin) to break down. Boys can have this conditions but girls can be carriers. Possible treatments can include steroids, stem cell transplant, gene therapy, and supportive therapies and management like physical therapy.

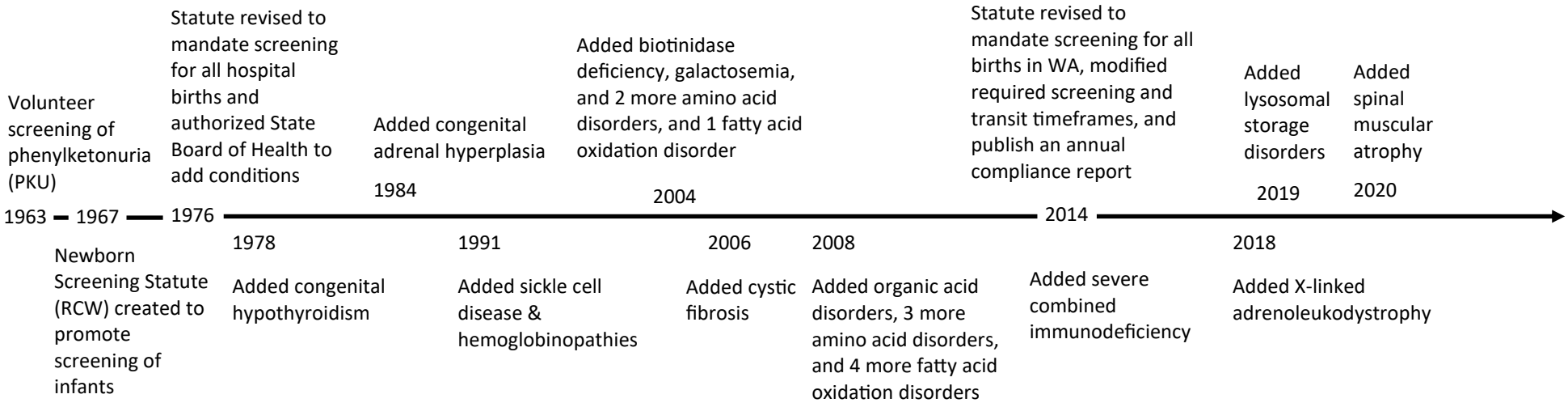
Lysosomal Storage Disorders (2)
1 in 32,000 births

Babies with lysosomal storage disorders have lysosomes that are unable to break down sugars and fats causing a toxic build-up. These infants have health problems of the brain, bone, muscle, or heart. Possible treatments can include enzyme replacement therapy, stem cell transplant, and supportive therapies and management like physical therapy.

Spinal Muscular Atrophy
1 in 15,000 births

Babies with spinal muscular atrophy lose motor nerve cells in their spinal cord. When these cells cannot send messages to the muscles, the no longer work. SMA can be mild or severe. Without treatment, SMA can cause missed motor milestones, difficulty breathing or eating, or even death.

History of Newborn Screening in Washington State



Washington State Newborn Screening

Saving lives with a simple blood spot

Washington State Department of Health

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Newborn Screening saving lives with a simple blood spot

Newborn blood spot screening is a Department of Health program that identifies babies born in Washington State with rare genetic disorders.

Infants with these disorders usually appear normal at birth and, without screening, the disorders are not likely to be detected and treated in time to prevent death or severe disability.

To make sure these disorders are identified quickly, State law requires a newborn blood spot to be collected for all babies before they are 2 days old.

Parents can legally refuse newborn screening if it conflicts with their religious tenets or practices.

Babies with these disorders can have serious health problems, but screening and early treatment can help your baby grow up healthy.

A nurse, midwife, or phlebotomist will collect several drops of blood from your baby's heel onto a filter paper card, that is then dried and sent to the State Laboratory for testing.

Newborn Screening is one of the **MOST SUCCESSFUL** public health initiatives in the USA

In Washington State, about **200** infants each year are diagnosed with one of the **32 disorders** on the screening panel.

2 screens in WA

1st shortly after birth, when your baby is 18-48 hours old

2nd at a well-child visit, when your baby is 7-14 days old

DOH 951-144 Aug 2020

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Babies with these disorders can have serious health problems, but screening and early treatment can help your baby grow up healthy.

To make sure these disorders are identified quickly, State law requires a newborn blood spot to be collected for all babies before they are 2 days old.

Parents can legally refuse newborn screening if it conflicts with their religious tenets or practices.

A nurse, midwife, or phlebotomist will collect several drops of blood from your baby's heel onto a filter paper card, that is then dried and sent to the State Laboratory for testing.

2 screens in WA

1st shortly after birth, when your baby is 18-48 hours old

2nd at a well-child visit, when your baby is 7-14 days old

DOH 951-144 Aug 2020

Washington State Newborn Screening

Saving lives with a simple blood spot

Washington State Department of Health

Public Health Laboratories
1610 NE 150th Street
Shoreline, WA 98155

Phone: (206) 418-5410
1-866-660-9050
Fax: (206) 363-1610
E-mail: NBS.Prog@doh.wa.gov

www.doh.wa.gov/nbs

Newborn Screening in Washington State

There are about **89,000** babies born in Washington State each year

Babies should be **SCREENED AROUND 24 HOURS** after birth

24

To make sure these disorders are found quickly, WA law requires newborn blood spot screening for **ALL BABIES**

Our State screens for **33** nationally **RECOMMENDED HEALTH CONDITIONS**

Z z z

Hearing Screening is typically done while the baby is asleep

Newborn Screening helps keep babies healthy through a **HEEL STICK, HEARING TEST & PULSE OXIMETRY**

Nationwide, more than **1 IN 300 NEWBORNS** have a condition detectable through newborn screening

Most babies with serious but treatable conditions caught by newborn screening **GROW UP HEALTHY** with expected development

Did you know...

Newborn Screening is one of the **MOST SUCCESSFUL** public health initiatives in the USA

Adapted from BabysFirstTest.org

Heel Stick

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Fax: (206) 363-1610
E-mail: NBS.Prog@doh.wa.gov
Web: www.doh.wa.gov/nbs

Pulse Oximetry

Talk to a prenatal or pediatric provider about critical congenital heart defect (CCHD) screening for newborns.

Hearing Test

Tel: (206) 418-5613
1-888-WAEHDDI (923-4334)
Fax: (206) 364-0074
E-mail: ehddi2@doh.wa.gov
Web: www.doh.wa.gov/earlyhearingloss

Washington State Department of Health

Public Health Laboratories
1610 NE 150th Street
Shoreline, WA 98155

DOH 304-120 Aug 2020

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Adapted from BabysFirstTest.org

Screening Checklist

First Trimester

WEEK

- [ ] First Trimester Ultrasound — 5 - 8
  Determines: Viable pregnancy, heartbeat, gestational age, molar or ectopic pregnancies, abnormal gestation
- [ ] Prenatal Blood Work — 8
  Determines: Blood type, Rh factor, glucose, iron and hemoglobin levels, rubella immunity, STDs, hepatitis, toxoplasmosis infection
- [ ] First Trimester Screening — 11 - 14
  Assesses: Chance of Down Syndrome and Trisomy 18

Second Trimester

- [ ] Second Trimester Screening — 15 - 20
  Assesses: Chance of Down Syndrome, Trisomy 18, and neural tube defects
- [ ] Second Trimester Ultrasound — 18 - 20
  Determines: Structural abnormalities, amniotic fluid levels, well-being
- [ ] Glucose Screening — 24 - 28
  Determines: Mother's risk of gestational diabetes

Third Trimester

- [ ] Strep B Test — 35 - 37
  Determines: Presence of group B strep infection

Newborn Screenings

- [ ] Blood Test — 24-48 hours
  Results:
- [ ] Hearing Screens — 24-48 hours
  Results:
- [ ] Pulse Oximetry Test — 24-48 hours
  Results:

My Contacts

OB/GYN

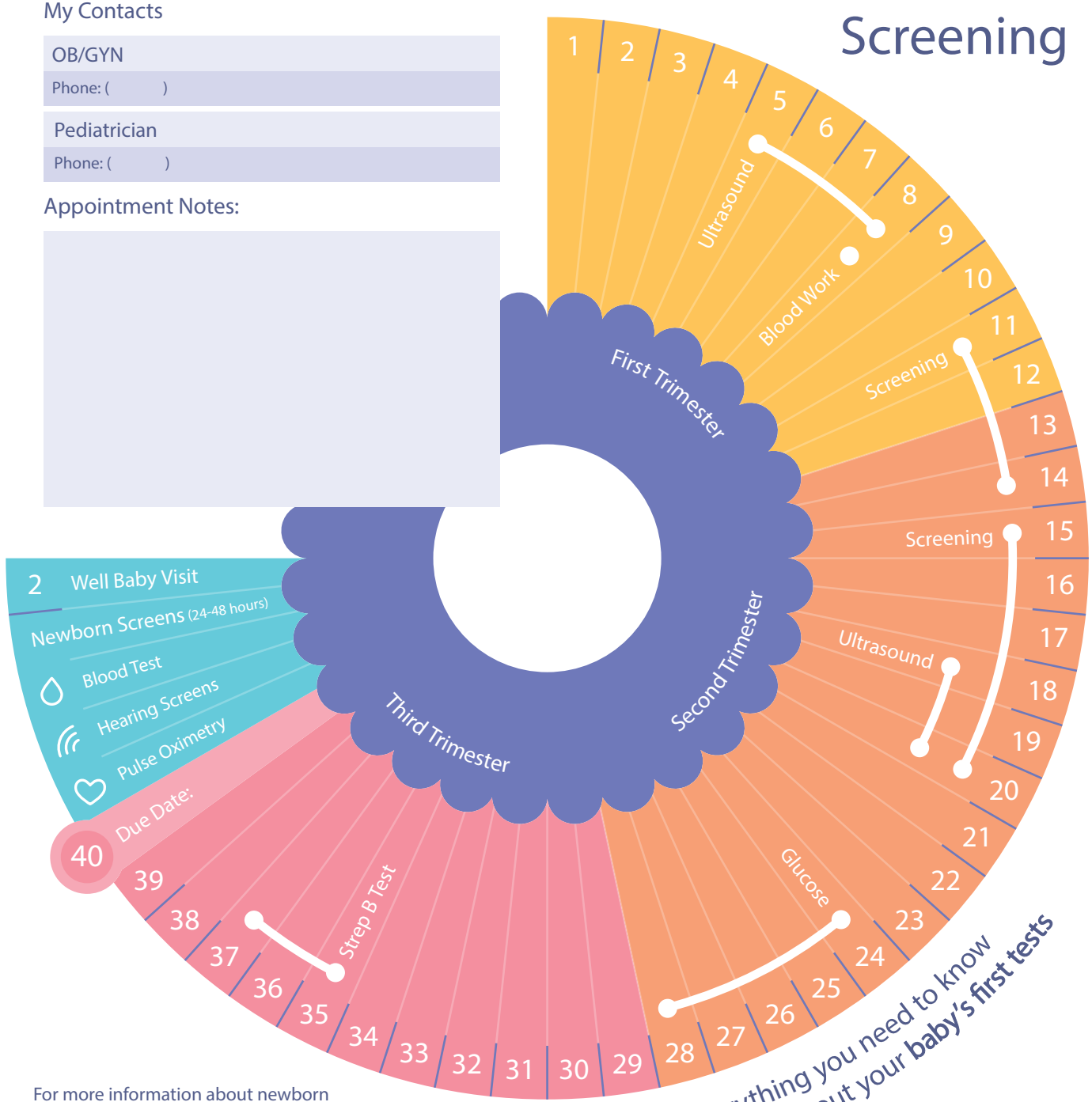
Phone: ( )

Pediatrician

Phone: ( )

Appointment Notes:

Prenatal & Newborn Screening



Everything you need to know about your **baby's first tests**

For more information about newborn screening, please visit www.babysfirsttest.org

About Prenatal & Newborn Screening

Prenatal Screenings ensure you and your baby are on track for a healthy pregnancy. They also prepare parents for potential health conditions and treatments before birth.

Newborn Screening is a state public health program that tests for serious and treatable conditions. Babies who test positive for treatable conditions are able to start treatment before harmful effects occur.

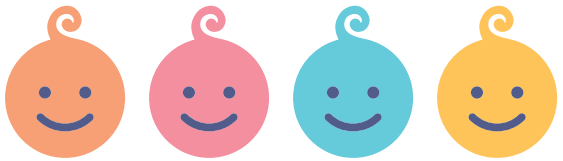
Blood Test
A small blood sample is taken from the baby's heel, placed on a newborn screening card, and sent to the state laboratory for analysis.

Hearing Screens
Determines if the ear and auditory brain stem respond to sound. No response can indicate hearing loss.

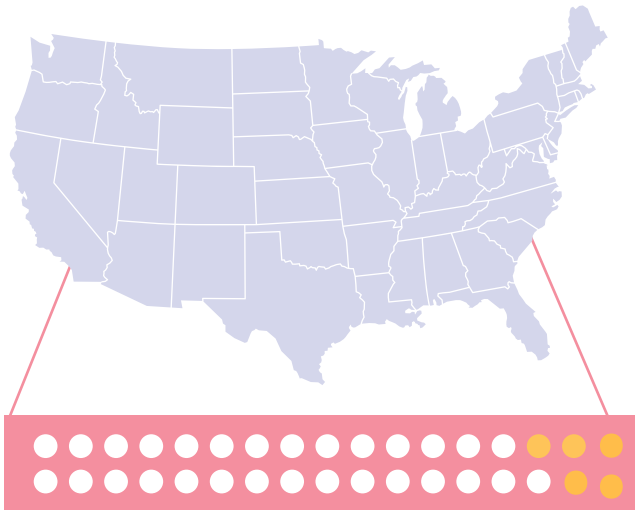
Pulse Oximetry Test
A sensor measures oxygen in the blood and can detect Critical Congenital Heart Disease (CCHD).

Why is screening so important?

Babies who appear healthy and come from healthy families can still have serious medical conditions. Newborn screening helps health professionals identify and treat conditions before they make a baby sick. Most babies identified at birth are treated early and grow up healthy.



Nearly **4 million babies** are born every year in the United States

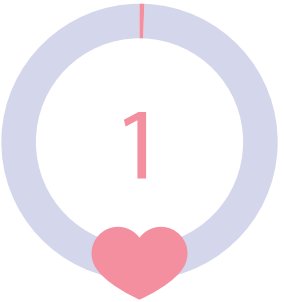


Most states screen for **29 out of 34** recommended health conditions

Each year, **12,000 babies** with serious, but treatable conditions grow up **healthy,** thanks to newborn screening

Every baby born in the United States can undergo newborn screening.

More than 1 in 300 newborns have a condition detectable through newborn screening



About 1 in 125 newborns have a Congenital Heart Defect

All newborns should be screened between 24-48 hours after birth

According to the Centers for Disease Control and Prevention

One Mother's Perspective

"Newborn screening saved my son's life. Although he appeared perfectly healthy and our family has no history of any disorders, his screening came back positive for a metabolic condition called MCADD. Thanks to the information we gained through his newborn screening, he is a perfectly healthy little boy and we know how to care for him to keep him that way.

I urge all new parents to learn about the life-saving potential of newborn screening and to thank the hospital staff who perform this very important test to ensure the health and safety of your newborn."

- A Grateful Mother in Colorado

This project is funded through cooperative agreement #U36MC16509 from the Maternal and Child Health Bureau (MCHB), Health Resources and Services Administration (HRSA).

Lista para las pruebas de detección

Primer trimeste

WEEK

- Ultrasonido del primer trimeste — 5 - 8

  Determina: si el embarazo es viable, el latido de corazón, la edad gestacional, el embarazo molar o ectópico, gestación anormal

- Análisis de sangre prenatal — 8

  Determina: el grupo sanguíneo, el factor Rh, los niveles de hierro y hemoglobina, la inmunidad, a la rubéola, las ITS, hepatitis, infección de la toxoplasmosis

- Pruebas de detección prenatales — 11 - 14

  Evalúa: la prob. del síndrome Down, trisomía 18

Segundo trimestre

- Pruebas de detección del segundo trimestre — 15 - 20

  Evalúa: la probabilidad del síndrome Down, trisomía 18, los defectos en el tubo neural

- Ultrasonido del segundo trimestre — 18 - 20

  Determina: las anormalidades estructurales, los niveles de líquido amniótico, el bienestar

- Pruebas de detección de glucosa — 24 - 28

  Determina: el riesgo materno de la diabetes gestacional

Tercer trimestre

- Prueba del estreptococo del grupo B (EGB) — 35 - 37

  Determina: la presencia de la infección del estreptococo del grupo B

Pruebas de detección en recién nacidos

- Prueba de sangre — 24-48 horas

  Resultado:

- Evaluación auditiva — 24-48 horas

  Resultado:

- Pulsioximetría — 24-48 horas

  Resultado:

Mis contactos

Ginecobstetra

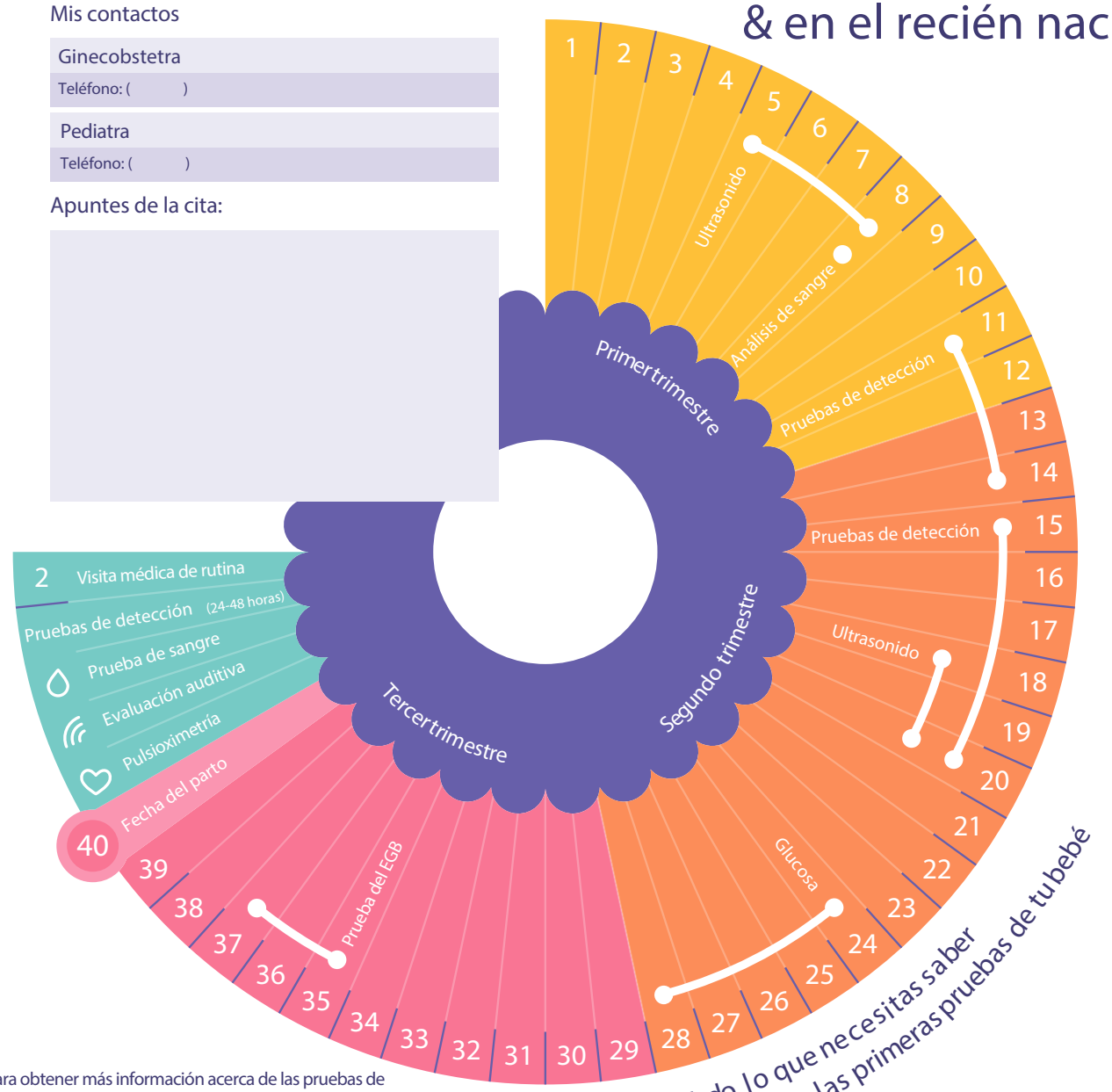
Teléfono: ( )

Pediatra

Teléfono: ( )

Apuntes de la cita:

Las pruebas de detección prenatales & en el recién nacido



Todo lo que necesitas saber sobre las primeras pruebas de tu bebé

Para obtener más información acerca de las pruebas de detección en recién nacidos, por favor visita Spanish.BabysFirstTest.org

Sobre las pruebas de detección prenatales & en el recién nacido

Las pruebas de detección prenatales aseguran de que tú y tu bebé estén en camino a un embarazo sano. También preparan a los padres antes del nacimiento para que se enteren sobre posibles condiciones de salud y tratamientos.

Las pruebas de detección en recién nacidos son un programa de salud pública estatal que busca detectar condiciones graves y tratables. Los bebés cuyas pruebas de detección dan resultados positivos a condiciones tratables, podrán comenzar con un tratamiento antes de tener efectos dañinos.

La prueba de sangre

Se extraen unas gotas de sangre del talón del bebé y se colocan en un tarjetón especial que se usa en las pruebas de detección. Este tarjetón es enviado al laboratorio estatal para un análisis.

La evaluación auditiva

Determina si el oído y el tronco cerebral responden al sonido. Si no hay respuesta, puede señalar la pérdida auditiva.

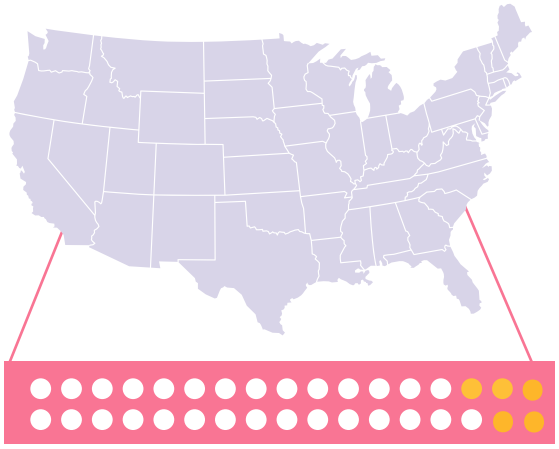
La prueba de pulsioximetría

Un sensor mide el oxígeno en la sangre y puede detectar la cardiopatía congénita crítica (CCHD).

¿Por qué son tan importantes las pruebas de detección?

Aun los bebés que tienen aspecto sano y provienen de familias sanas, pueden presentarse con condiciones médicas serias. Las pruebas de detección ayudan a que los profesionales médicos identifiquen y traten condiciones antes de que se enferme el bebé. La mayoría de los bebés identificados al nacer que reciben tratamiento temprano crecen saludables.

Nacen más de **4 millones de bebés** cada año en los Estados Unidos.



La mayoría de los estados realizan pruebas para detectar **29 de las 34** condiciones médicas recomendadas

Cada año, **12,000 bebés** con condiciones graves pero tratables crecen **sanos** gracias a las pruebas de detección.

A todo bebé que nace en los Estados Unidos le puede hacer las pruebas de detección.

Más de 1 bebé de cada 300 nace con una condición detectable mediante las pruebas de detección.

Alrededor de 1 bebé de cada 125 tiene la cardiopatía congénita.

A todos los recién nacidos se les deberían hacer las pruebas de detección entre las 24 a 48 horas después de nacer.

Según los Centros de Control y Prevención de Enfermedades (CDC)

La perspectiva de una madre

"Las pruebas de detección le salvaron la vida a mi hijo. A pesar de que tenía un aspecto perfectamente sano y que en nuestra familia no hubo un historial médico de cualquier trastorno, su prueba dio un resultado positivo para una condición metabólica llamada la MCADD. Gracias a la información que obtuvimos a través de sus pruebas de detección, es un niño completamente sano y sabemos cómo cuidarlo para que se mantenga así.

"Les ruego a todos los nuevos padres que aprendan sobre el potencial de las pruebas de detección para salvar vidas, y que agradezcan al personal del hospital que realizan estas importantes pruebas que protegen la salud y la seguridad de su recién nacido."

- Una madre agradecida de Colorado

Este proyecto es financiado por el acuerdo de cooperación #U36MC16509 del Maternal and Child Health Bureau (MCHB), Health Resources and Services Administration (HRSA)

DISORDERS DETECTED BY THE WASHINGTON NEWBORN SCREEN (2020)

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

Disorder (Prevalence in WA)	Definition	Screening Test	Impact without Early Treatment	Treatment	Benefits of Early Treatment
Galactosemia (1 in 11,000)	Inability to break down galactose, a major sugar found in milk	Measure activity of enzyme needed to break down galactose; DNA test if indicated	Severe intellectual and developmental disability, liver disease, blindness, overwhelming infections and death	Dietary restriction of milk and other foods containing galactose	Prevent death, improve intellectual function, and reduce other morbidity
Congenital Adrenal Hyperplasia (CAH) (1 in 14,000)	Impaired production of cortisol and other adrenal hormones	Measure adrenal hormone: 17-hydroxyprogesterone (17-OHP) level	Salt loss and shock may result in early sudden death; virilization and abnormal growth	Cortisol and salt-retaining hormone replacement	Prevent death, reduce virilization and abnormal growth
Organic Acid Disorders (1 in 29,000) (see list below)	Inability to process or break down organic acids, byproducts of protein and fatty acid metabolism	Measure acylcarnitine levels by tandem mass spectrometry	Severe nerve and physical damage and death	Dietary restriction of offending amino acid(s) and use of special metabolic formula	Prevent death, intellectual and developmental disability and other neurological damage
Fatty Acid Oxidation Disorders (1 in 11,000) (see list below)	Inability to process or break down fats in the body	Measure acylcarnitine levels by tandem mass spectrometry	Serious damage to brain, liver, heart, eyes and muscles, and death	High carbohydrate, low-fat diet and avoidance of fasting	Prevent death, intellectual and developmental disability and other neurological damage
Amino Acid Disorders (1 in 10,000) (see list below)	Inability to break down amino acids, found in all foods containing protein	Measure amino acid levels by tandem mass spectrometry	Intellectual and developmental disability, seizures, coma, and death	Dietary restriction of offending amino acid(s) and use of special metabolic formula	Prevent death, intellectual and developmental disability and other neurological damage

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

Carnitine uptake defect (CUD)
*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

Questions? Please contact:
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Phone: 206-418-5410
Website: www.doh.wa.gov/nbs
E-mail: NBS.Prog@doh.wa.gov

Washington State Department of Health
DOH 951-123
Aug 2020

DISORDERS DETECTED BY THE WASHINGTON NEWBORN SCREEN (2020)

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***

Disorder (Prevalence in WA)	Definition	Screening Test	Impact without Early Treatment	Treatment	Benefits of Early Treatment
Sickle Cell Diseases and Hemoglobinopathies (1 in 4,700)	Production of abnormal hemoglobin	Separate and visualize hemoglobin proteins by isoelectric focusing, with confirmation by high performance liquid chromatography and DNA analysis, if indicated	Severe infections and possible death	Antibiotic prophylaxis to help prevent infections and parental education to recognize health crises	Prevent death, reduce infections and other morbidity
Congenital Hypothyroidism (1 in 950)	Inadequate production of thyroid hormone	Measure thyroid stimulating hormone (TSH) level	Intellectual and developmental disability, growth failure	Thyroid hormone replacement	Normal growth and intellectual development
Biotinidase Deficiency (1 in 86,000)	Deficiency of biotin, part of the Vitamin B complex	Measure activity of enzyme needed to recycle biotin	Seizures, damage to immune system, intellectual and developmental disability, hearing loss	Oral biotin supplementation	Prevent all adverse consequences
Cystic Fibrosis (CF) (1 in 5,500)	Defect in the cystic fibrosis transmembrane conductance regulator (CFTR) gene	Measure immunoreactive trypsinogen (IRT) level; DNA test if indicated	Significant nutritional deficits due to thick, sticky mucus in the digestive system. Severe lung infections due to mucus	Pancreatic enzymes, vitamin supplements, chest physiotherapy, antibiotics	Improve physical growth, cognitive function and possibly lung function
Severe Combined Immunodeficiency (SCID) (1 in 88,000)	Complete lack of immune system	DNA test: measure number of T-cell excision circles (TRECs) by real-time PCR	Severe life-threatening infections that complicate treatment and possible death	Stem-cell transplant or gene therapy, depending on the genotype	Prevent death and cure the condition
X-linked Adrenoleukodystropy (X-ALD) (1 in 17,000)	Peroxisomal disorder caused by mutations in ABCD1 gene leading to accumulation of very long chain fatty acids (VLCFA) in tissues and organs	Measure very long chain fatty acid levels by tandem mass spectrometry	Severe debilitating sensorimotor, behavioral and cognitive functions that can lead to death 2-4 years after onset of symptoms for those affected with childhood cerebral ALD	Cortisol (hormone replacement), Stem cell transplant (HSCT)	Treatment with HSCT may prevent death and disability

DOH 951-123
Aug 2020

Glycogen Storage Disorder (Pompe disease) (1 in 28,000)	Inability to break down glycogen, (a complex sugar)	Measure activity of enzyme needed to break down glycogen	Muscle weakness, possible cardiac and respiratory failure, and possible death	Enzyme replacement therapy	Prevent death, reduce need for mechanical ventilation and other morbidity
Mucopolysaccharidosis type I (MPS-I) (1 in 36,000)	Inability to break down glycosaminoglycans (large sugar molecules)	Measure activity of enzyme needed to break down glycosaminoglycans	Progressive cognitive decline	Hematopoietic stem-cell transplantation and/or Enzyme replacement therapy	Slow or halt cognitive decline, reduce other morbidity
Spinal Muscular Atrophy (SMA) (1 in 15,000)	Genetic disorder that results in lack of survival motor neuron (SMN) protein, causing progressive death of nerve cells in the spinal cord	DNA test: detect the presence/absence of exon 7 of SMN1 by real-time PCR	Muscle weakness, possible difficulty walking, swallowing, breathing, or even death.	One-time gene therapy or regularly administered intrathecal or oral medications	Prevent death, slow or halt disease progression

DOH 951-123
Aug 2020