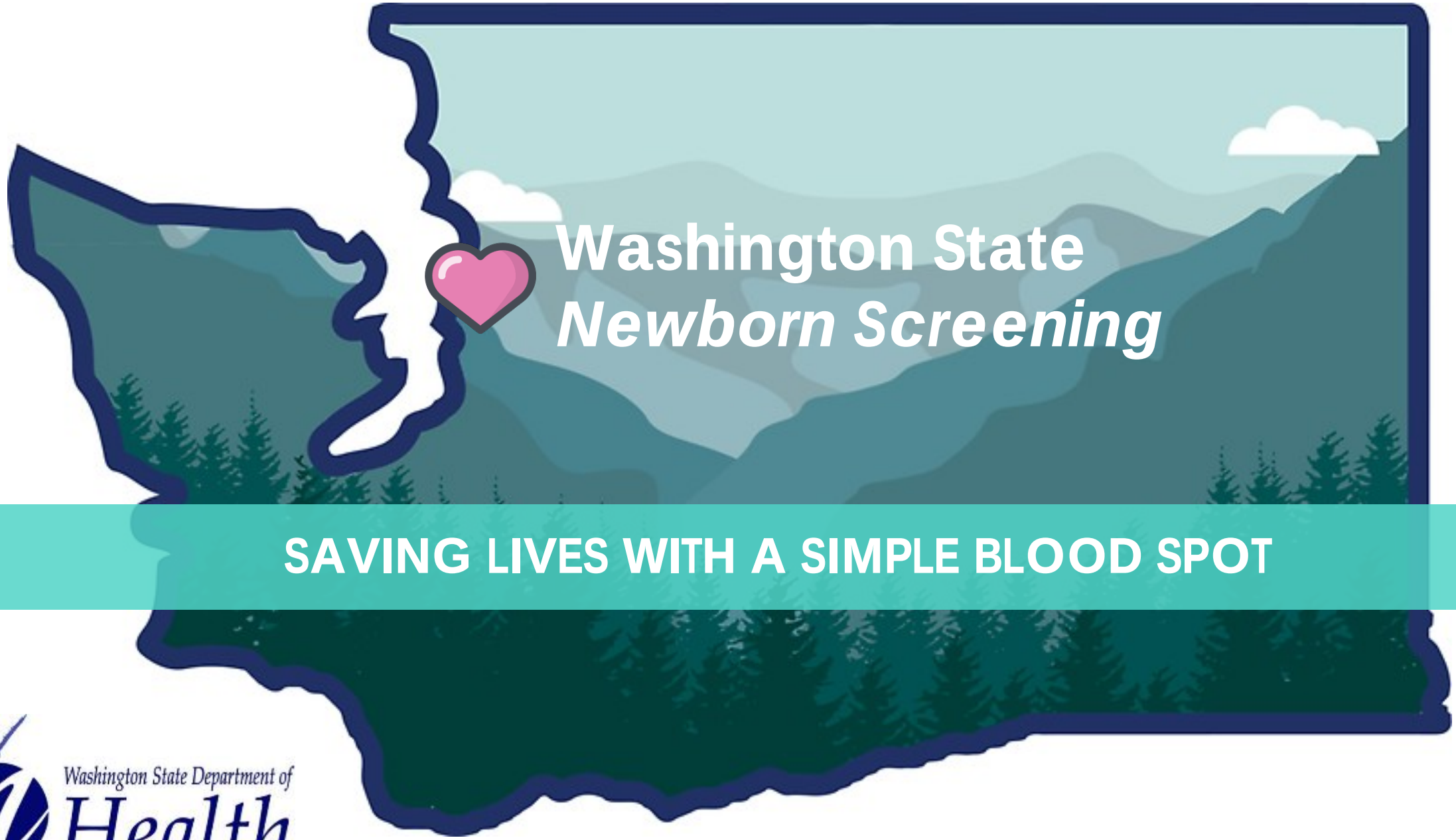


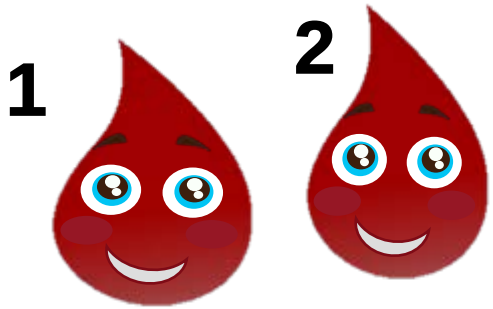
Washington State *Newborn Screening*

SAVING LIVES WITH A SIMPLE BLOOD SPOT

Washington State Department of Health

Washington State Numbers

90,000 newborns



2-screen state

173,000 specimens

17 lab staff

5 disorder FU staff

6 program staff

Washington Screens for... **29 disorders!**

Amino Acid Disorders (6)	Organic Acid Disorders (8)
Phenylketonuria	Isovaleric acidemia
Homocystinuria	Glutaric acidemia type I
Maple syrup urine disease	Methylmalonic acidemias (CblA/B and MUT)
Citrullinemia	Propionic acidemia
Argininosuccinic acidemia	Multiple carboxylase deficiency
Tyrosinemia type I	Beta-ketothiolase deficiency
	3-hydroxy-3-methylglutaric aciduria
Fatty Acid Oxidation Disorders (5)	**Other Disorders (10)**
Medium-chain acyl-CoA dehydrogenase deficiency	Congenital hypothyroidism
Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency	Galactosemia
Trifunctional protein deficiency	Congenital adrenal hyperplasia
Very long-chain acyl-CoA dehydrogenase deficiency	Biotinidase deficiency
Carnitine uptake defect	Cystic fibrosis
	Sickle Cell Diseases & Hemoglobinopathies
	Severe combined immunodeficiency
	X-linked adrenoleukodystrophy

COMING SOON

3 *Additional Screening Conditions!*

March 1, 2018:

- **X-ALD**

Sometime in 2019:
(Was supposed to be Fall 2018…)

- MPS-I
- Pompe

NBS Lab Construction

NBS Lab Construction

NBS Lab Construction

NBS Lab Construction

Equipment Procurement

Xevo TQD MS/MS

- We had one already in use (of four MS/MS units)
  - Amino acids
  - Acylcarnitines
- Started paperwork in January 2017 (need two more)
- Legislature working on budget – approved June 30th
- Working in earnest with DOH contracts office and Waters Corp.
- Contract finalized on Sept. 29th (287 pages)
- Request for additional delegation on Oct. 6th (granted October 23rd)
- Sole source justification filed on Oct. 25th (approved Dec. 5th)
- Instruments ordered on Dec. 11th (delivered Dec. 22nd)
- Installation and validation planned for Jan.
- March 1st start date

NBS Fee Increase

X-ALD

- Requested increase of $10.00 per birth
- Received authorization to increase fee by only $8.10 per birth

MPS I/Pompe

- Requested increase of $9.00 per birth
- Did not receive authorization
- Will request the increase again in the 2019-2021 biennium budget

NewSTEPs and the New Disorders Implementation Project

Three Project Goals

- Develop a Long-Term Follow-up Program for X-ALD, Pompe and MPS I
- Perform pilot testing to determine screening method for X-ALD, Pompe and MPS I
- Develop an outreach program and materials for X-ALD, Pompe and MPS I

Goal 1: Long-Term Follow-up

Learning from other programs:

Complete:

- In-person learning exchange with Amy Gaviglio and Tony Steyermark from Minnesota
- Conference call with Lisa Feuchtbaum, Jamie Matteson and Stanley Sciortino from California

In the works:

- Conference call with Nicole Brown, LTFU Supervisor in Minnesota
- In-person learning exchange with another LTFU program (or 2)

Goal 1: Long-Term Follow-up

Identify specialists who diagnose, monitor & treat X-ALD:

- Biochemical Geneticists
- Neurologists
- Endocrinologists
- Radiologists
- Transplant teams

Conduct periodic meetings with team of specialists to assess needs, strategize and identify solutions

Goal 1: Long-Term Follow-up

Still in the works...

- Identify outcomes to be measured
- Identify data to be collected
- Identify database

Considerations...

- How can we best meet the needs of families and the specialists?
- What is the appropriate amount of time to follow families?
- How to keep families engaged when kiddos appear healthy?
- How big will the program get as we continually add cases?

Goal 1: Long-Term Follow-up

Challenges

- LTFU presents a significant paradigm shift for NBS
- With so many specialists involved in monitoring & treatment…
  - Who will serve as the medical home?
  - Who will be responsible for coordinating tests & appointments?
  - What are their expectations?
  - Arranging meetings can be a big challenge!
- What is the role of the NBS program?

Goal 1: Long-Term Follow-up

Solutions & Lessons Learned

- Reach out to other programs – someone is always willing to collaborate and share
- Nurture relationships with specialists

Goal 2: Method Validation

Planned Activities:

- Validate X-ALD using negative ion mode
- Validate X-ALD using positive ion mode
- Evaluate a modification to the NY method using a 3-hour incubation period for Pompe and MPS I
- Validate X-ALD in negative ion mode and Pompe and MPS I in positive ion mode (switch ion modes during sample injection phase)
- Validate Pompe and MPS I using digital microfluidics

Goal 2: Method Validation

- Background research of available testing methods & platforms
- Cost analysis of testing methods, materials & instruments
- Analysis of workflow, space and staffing requirements.
- Review and discussion for method of choice
- Risk Assessment and Validation Plan proposal
- Plan approval prior to performing any work
- Validation study - conducting experiments, analyzing data, and summarizing reports
- Validation Report review
- SOP sign-off

Goal 2: Method Validation

Challenges, Solutions & Lessons Learned	
Infrastructure required for validation	Planning should include extra time to accommodate the unexpected
Testing materials, such as columns, may be discontinued	Allow time to choose and validate replacements
Known positive samples can be scarce and difficult to obtain	Reach out to specialists early so they can request use of DBS at regular visits
Large number of samples required to establish normal ranges	Allow time to punch and test specimens and analyze data

* Special thanks to Aranjeet Singh

Goal 2: Method Validation

Current Status:

- Validation of X-ALD using negative ion mode is complete

- Currently increasing testing capacity by validating two additional Xevo Mass Spectrometer instruments.

Key Milestone:

- Completed testing of all backlogged samples this week

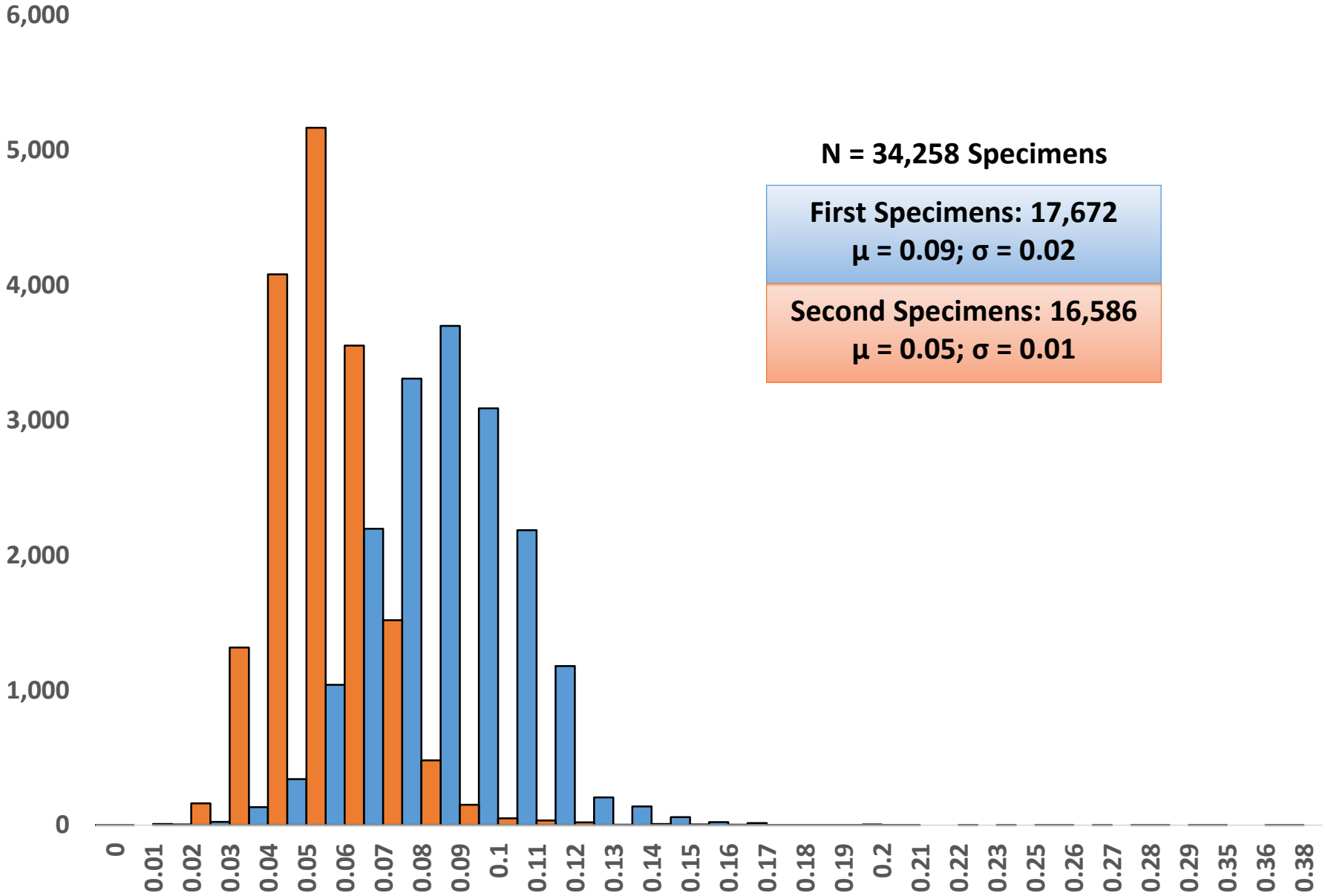
Special thanks to Bill Hoffman and the lab team

C26:0 LPC (lysophosphatidylcholine)

- Currently, specimens with C26:0 LPC ≥15.0 µmol/L are reported as elevated

- Diagnostic tests are recommended for newborns if two or more specimens have C26:0 LPC ≥15.0 µmol/L
  - Very Long Chain Fatty Acids
  - Potentially DNA

Distribution of C26:0 LPC Values: First and Second NBS Specimens



Persistently Elevated C26:0 LPC

- Ninety-one newborns had at least one elevated C26:0 LPC
- Seventy-five were resolved by a normal subsequent specimen
- Eleven are pending results of subsequent specimen
- Diagnostic tests were recommended for four newborns with persistently elevated C26:0 LPC levels

1st Specimen	2nd Specimen	3rd Specimen	VLCFA
0.35 µmol/L @ 26 hours	0.36 µmol/L @ 14 days	N/A	Consistent with X-ALD
N/A	0.16 µmol/L @ 19 days	0.28 µmol/L @ 56 days	Normal
0.27 µmol/L @ 20 hours	0.17 µmol/L @ 12 days	N/A	Normal
0.08 µmol/L @ 24 hours	0.23 µmol/L @ 14 days	0.15 µmol/L @ 52 days	Normal

Goal 3: Conduct a Statewide Educational Outreach Campaign

Planned Activities:

- Create and/or modify educational materials
- Create outreach presentation
- Schedule and conduct outreach sessions
- Identify or create measures to track impact of education materials for providers
- Conduct knowledge assessments after outreach sessions

Who are we?

Heidi Lovejoy, MSc **Gauri Gupta, MScPH**

Health Services Consultants

Office of Newborn Screening

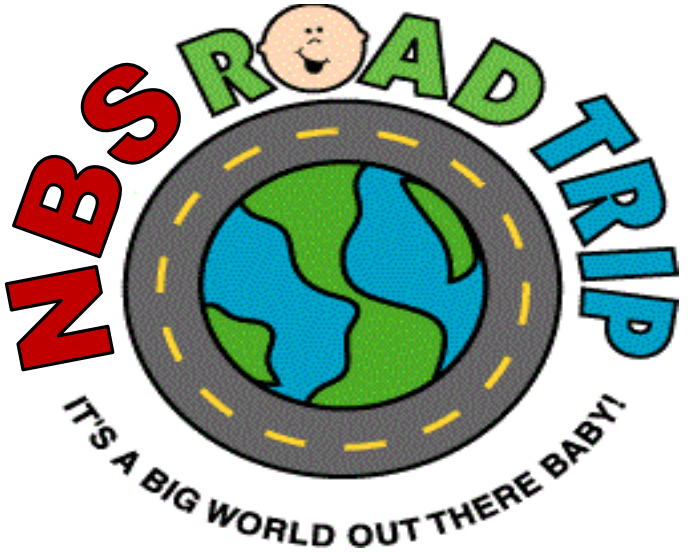
Washington State Department of Health

Statewide Education Campaign

- Inform providers about the new conditions
- Provide refresher training about NBS
  - Importance of screening, required timeframes for collection and transit, completing demographics, collection of quality specimens

- About 1,000 hospitals, clinics, labs, and midwives in our state…
  - Focus on birthing facilities (and midwives)
  - Major clinics and lab groups nearby

Grant provided funding for travel/lodging to regions of the state we don't often get to



Travel Diary

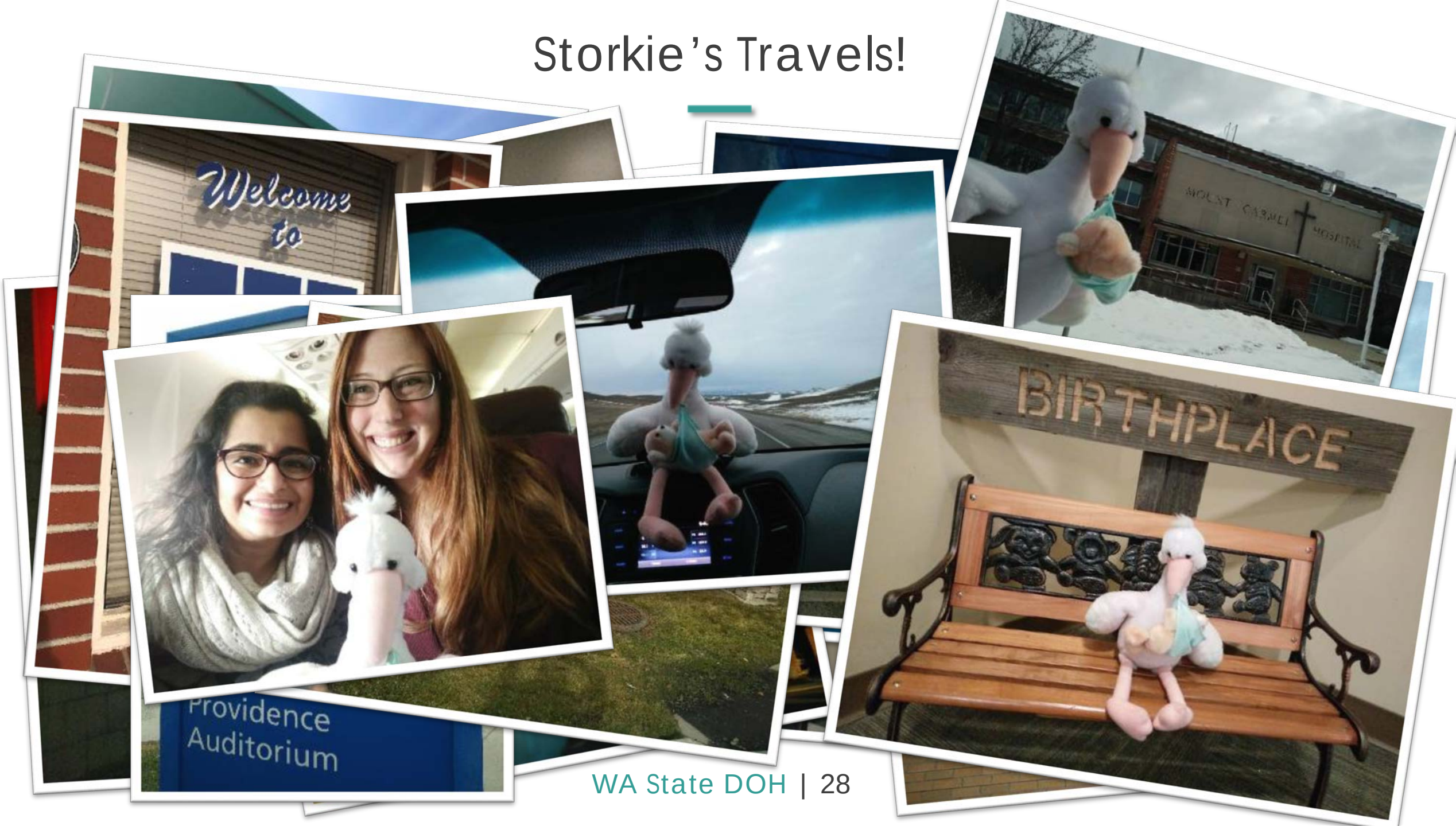
Regional visits so far

- 5 trips over 15 days
- 39 presentations
- 13 Hospitals
- 10 clinics/labs
- 2 midwife-run birth centers
- 300 health care providers

Coming soon…

- Olympic Peninsula – next week – 3 days, 6 presentations, 5 hospitals
- SW Washington
- Central Washington
- South Puget Sound
- Greater Seattle area

Storkie's Travels!



Lessons Learned

- Spokane area-- needs parent materials in Russian, Bosnian, Arabic
- Refusal section on our specimen card-- need translated into other languages
  - Translated into 8 most common WA languages. Posted on website.
- Parents in the Tri-Cities area routinely do not pick PCP
  - Hospitals do not know what to put on our card for follow-up facility

Common questions:

- More conditions = more blood?
- What about babies born before X-ALD testing began?

Exhibitions

Hosting info booths at maternity expos across the State

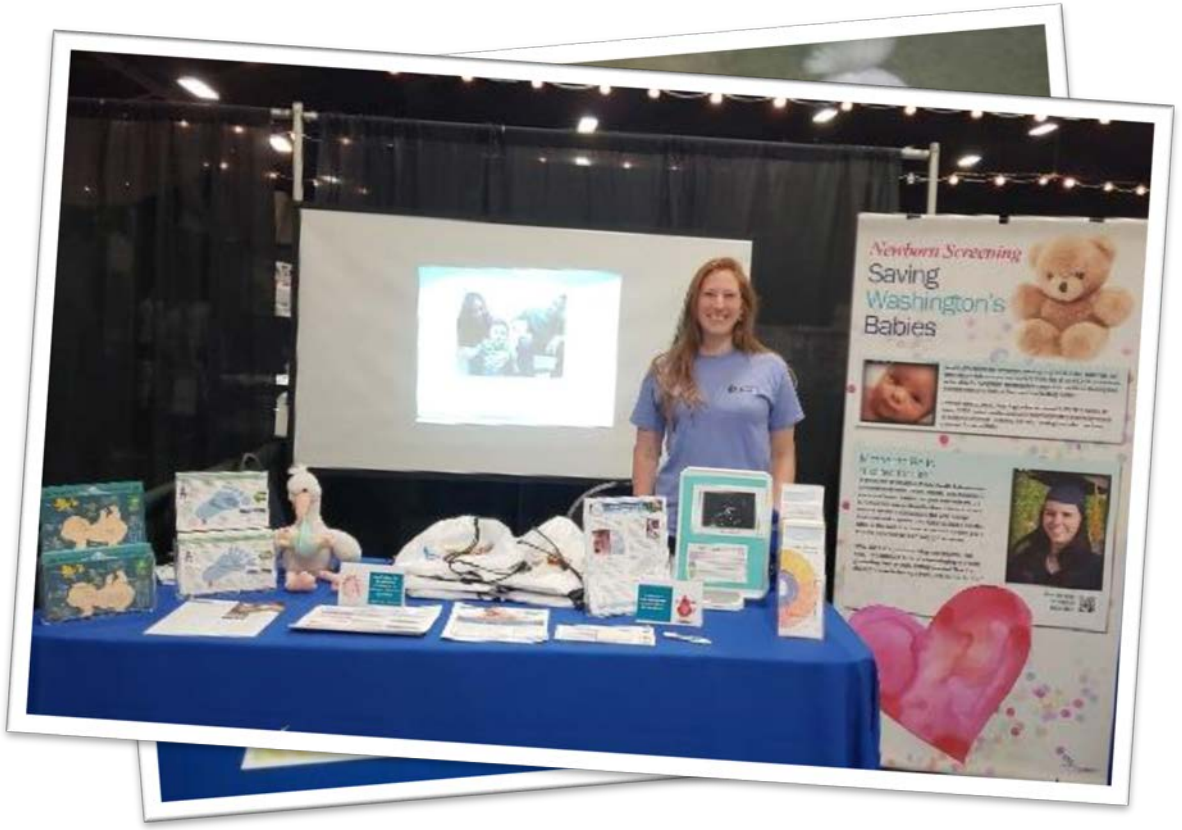
- Previously attended expos only in the Seattle area
- Interact with new/expecting families and introduce them to NBS
- Answer questions and provide swag bags with info to hundreds of new and expecting parents
- Created handout on new conditions
- Brought many of our materials in Spanish to the Tri-Cities

Spokane, March 2018

Tri-Cities, May 2018

Bellevue, August 2018

EXPOs



Tri-Cities Mom & Baby Expo

Spokane Baby Fair

Conferences

REACHE

- Conference for child birth educators, midwives, and doulas
- Displayed resources they could use for parent education
- Handout about new conditions

MAWS

- Invited by Midwives' Association of Washington State
- Advanced topics in newborn screening such as false positives, active vs. passive FU, new conditions

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 31 rare, serious conditions that are treatable when found early in life.

X-Linked Adrenoleukodystropy (X-ALD)

Babies with X-ALD cannot break down very long-chain fatty acids. When these don't break down, they build up in the body and cause serious health problems. A stem cell transplant early in life can help the baby grow up healthy.

- Occurs in ~ 1 in 34,000 births
- Affects males more severely
- Screening began March 1, 2018
- Is a peroxisomal 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 begins in 2019
- Is a lysosomal storage disorder

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 begins in 2019
- Is a lysosomal storage disorder

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 Mar 2018

MUY PRONTO

¡Nuevas enfermedades!

La State Board of Health (Junta Estatal de Salud) aprobó otras tres enfermedades en el grupo de exámenes de detección. A partir de ahora, se les deberá realizar un examen a todos los bebés nacidos en el estado de Washington para detectar un total de 31 enfermedades graves poco frecuentes que se pueden tratar si se descubren a tiempo.

Adrenoleucodistrofia ligada al cromosoma X (X-ADL)

Los bebés con X-ALD no pueden descomponer los ácidos grasos de cadena muy larga. Cuando estos ácidos no se descomponen, se acumulan en el cuerpo y causan problemas de salud graves. Un trasplante de células madre en los primeros años de vida puede ayudar a que el bebé crezca sano.

- Se presenta en ~1 de 34 000 nacimientos.
- Afecta especialmente a los varones.
- Los exámenes comenzaron el 1 de marzo de 2018.
- Es un trastorno en el almacenamiento peroxisomal.

Mucopolisacaridosis tipo I (MPS I)

Los bebés con MPS I tienen problemas para procesar un cierto tipo de azúcar que se encuentra en los alimentos. Esto produce una acumulación en el cuerpo que lleva a problemas en el esqueleto, los órganos y el cerebro. El tratamiento con un trasplante de células madre previene la pérdida de puntos del coeficiente intelectual (CI) y otros problemas físicos.

- Se presenta en ~1 de 35 700 nacimientos.
- La MPS I grave también se denomina "síndrome de Hurler".
- Los exámenes comienzan en 2019.
- Es un trastorno en el almacenamiento lisosomal.

Enfermedad de almacenamiento de glucógeno tipo II (Pompe)

Los bebés con la enfermedad de Pompe tienen problemas para procesar un tipo de azúcar que se encuentra en los alimentos. Cuando esta azúcar se acumula, causa debilidad muscular y problemas cardíacos. El tratamiento con terapia de reemplazo de enzimas puede salvar la vida de los niños con la enfermedad de Pompe.

- Se presenta en ~1 de 28 000 nacimientos.
- La enfermedad de Pompe es conocida como el "síndrome del bebé flojo".
- Los exámenes comienzan en 2019.
- Es un trastorno en el almacenamiento lisosomal.

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 Mar 2018 Spanish

Audience Response System

Purchased TurningPoint

- PowerPoint add-in that does real time polling
- Each attendee gets a ‘clicker’ to answer questions/polls during the presentation

So far so good!

- Attendees are excited to participate!
- Easy to use software
- Required Department IT exceptions and support
- Collects data and provides reports of responses

TurningPoint®

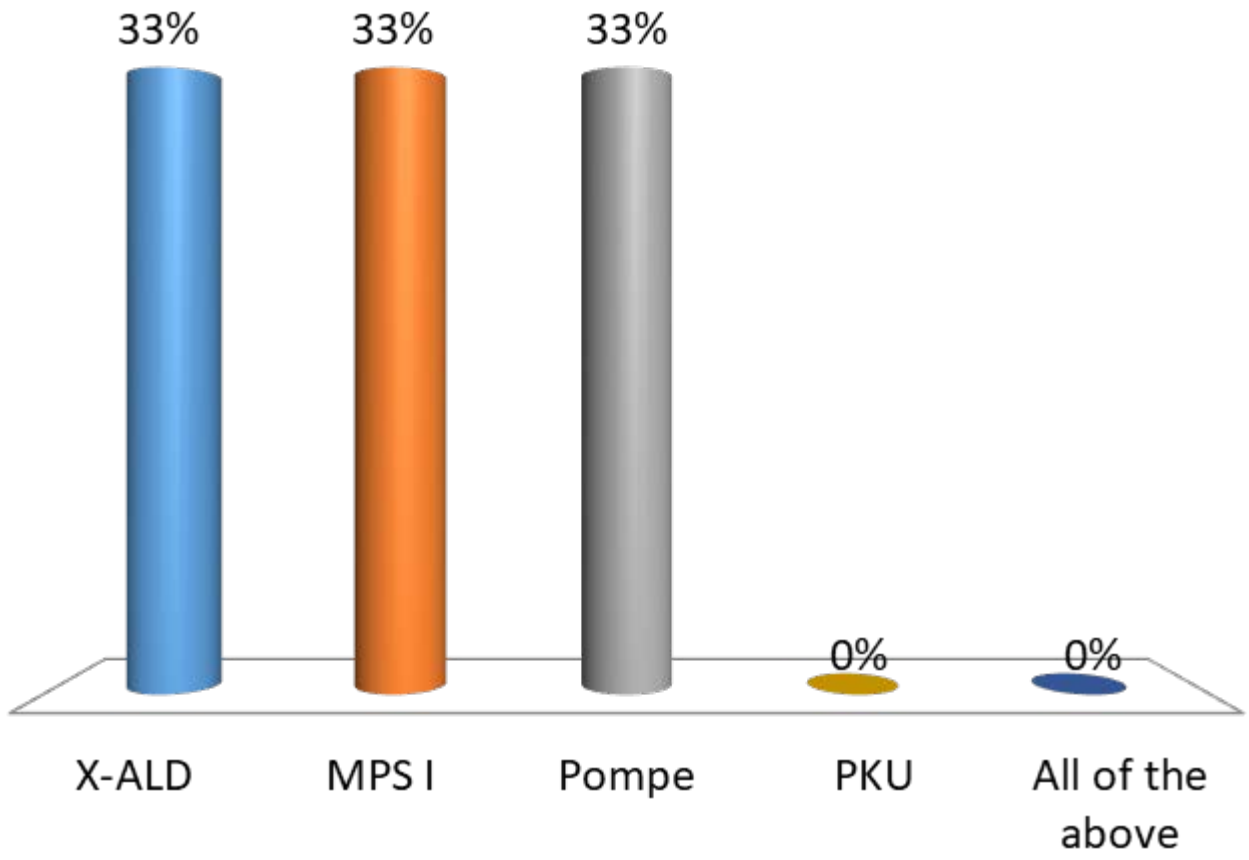
Which condition(s) primarily affects males?

A. X-ALD
B. MPS I
C. Pompe
D. PKU
E. All of the above

Which condition(s) primarily affects males?

✓ A. X-ALD
B. MPS I
C. Pompe
D. PKU
E. All of the above

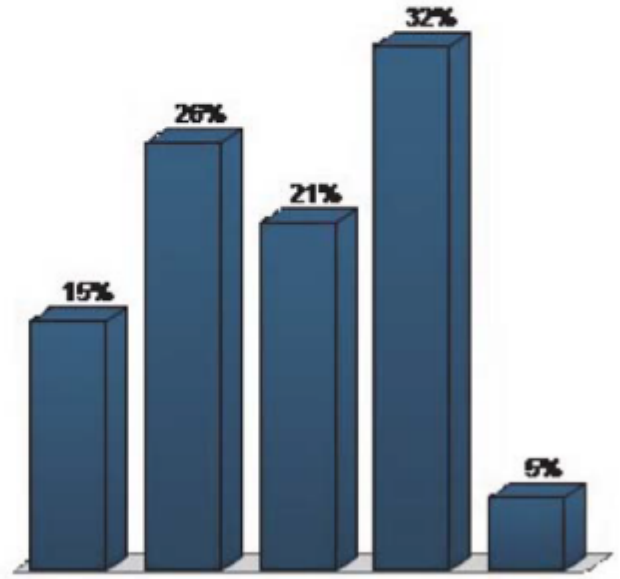
Boys are primarily affected by X-ALD because they have only one X-chromosome



Poll (start of presentation)

2. What do you want us to go over today? (Priority Ranking)

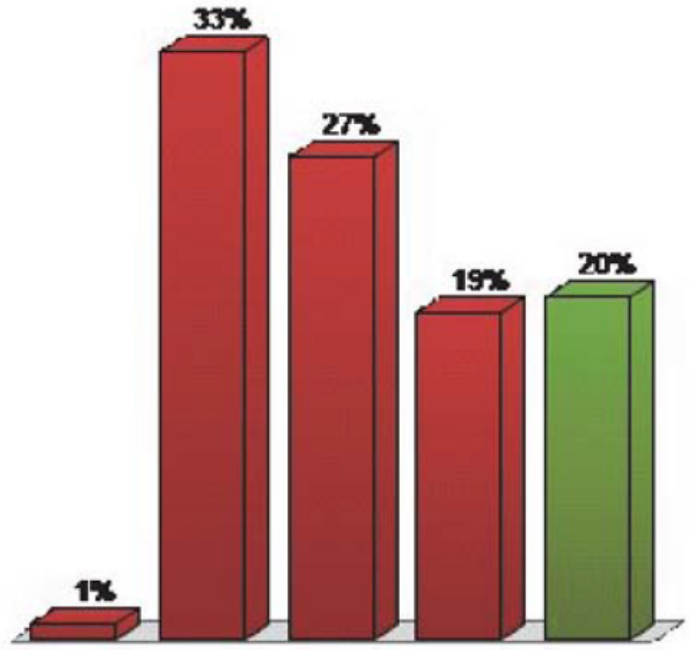
	Responses	
	Percent	**Weighted Count**
What is newborn screening	15.37%	697
Guidelines in Washington State	26.34%	1195
How to complete the collection cards	21.41%	971
How to avoid unsuitable specimens	32.36%	1468
Something else	4.52%	205
Totals	**100%**	**4536**



Pop Quiz (During presentation)

3. What is the appropriate name for blood spot test? (Multiple Choice)

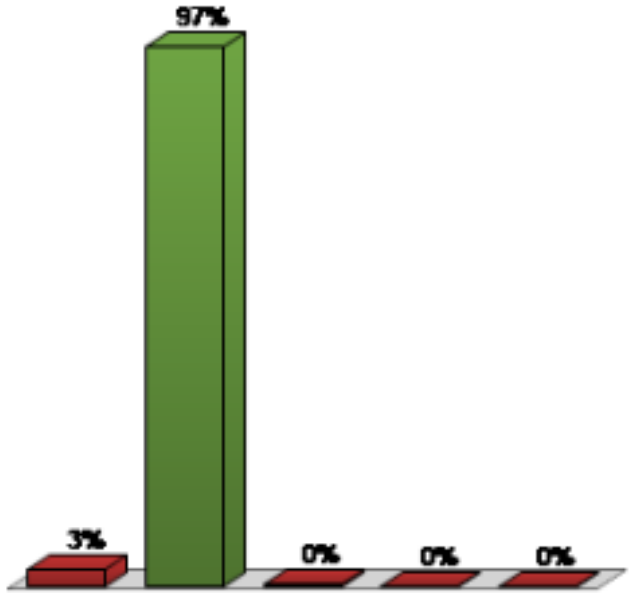
	Responses	
	Percent	**Count**
Heel-stick test	0.93%	2
Newborn screen	33.49%	72
PKU test	27.44%	59
Metabolic screen	18.6%	40
All except C *(c)*	19.53%	42
Totals	**100%**	**215**



Comprehension Check (after)

8. When should the first newborn screen be collected? (Multiple Choice)

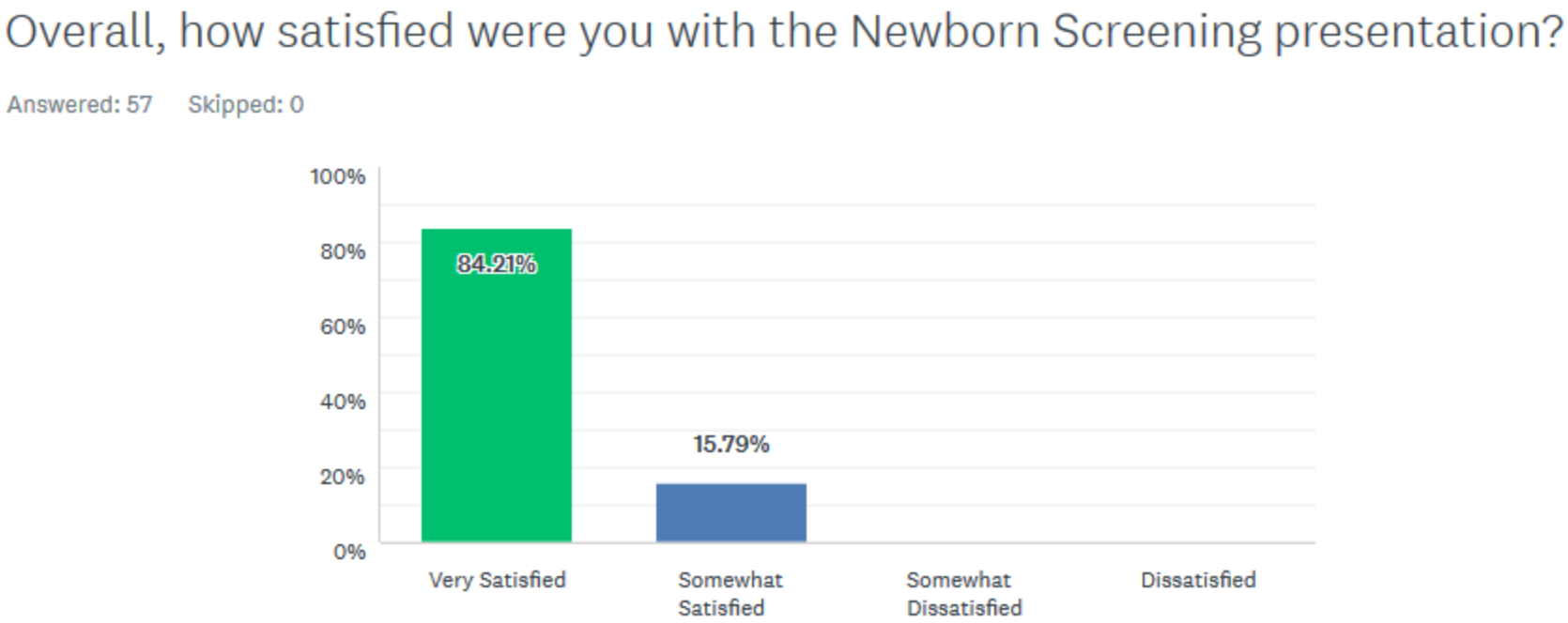
	Responses	
	Percent	**Count**
Anytime before hospital discharge	2.99%	6
Between 18-48 hours of age *(c)*	96.52%	194
After the baby feeds	0.5%	1
When the baby is asleep	0%	0
At baby's first well-child visit	0%	0
Totals	**100%**	**201**



Do people like our presentations?

We send anonymous evaluation surveys post-presentation

YES!



Survey Feedback

I have done these for 38 years. It was really helpful to have this education in person with real time question and answer.

That I learned a new technique on getting the newborn screen..

Great and useful information

5/31/2018 12:50 PM

5/31/2018 2:28 PM

the clicker interactions was a good way to keep the audience involved.

Learning more about the different disorders was helpful and being more confident to explain the importance of doing the Newborn screenings.

Thank you for the presentation! Lots of great info and well presented!

You guys were great! Thanks.

5/29/2018 7:57 AM

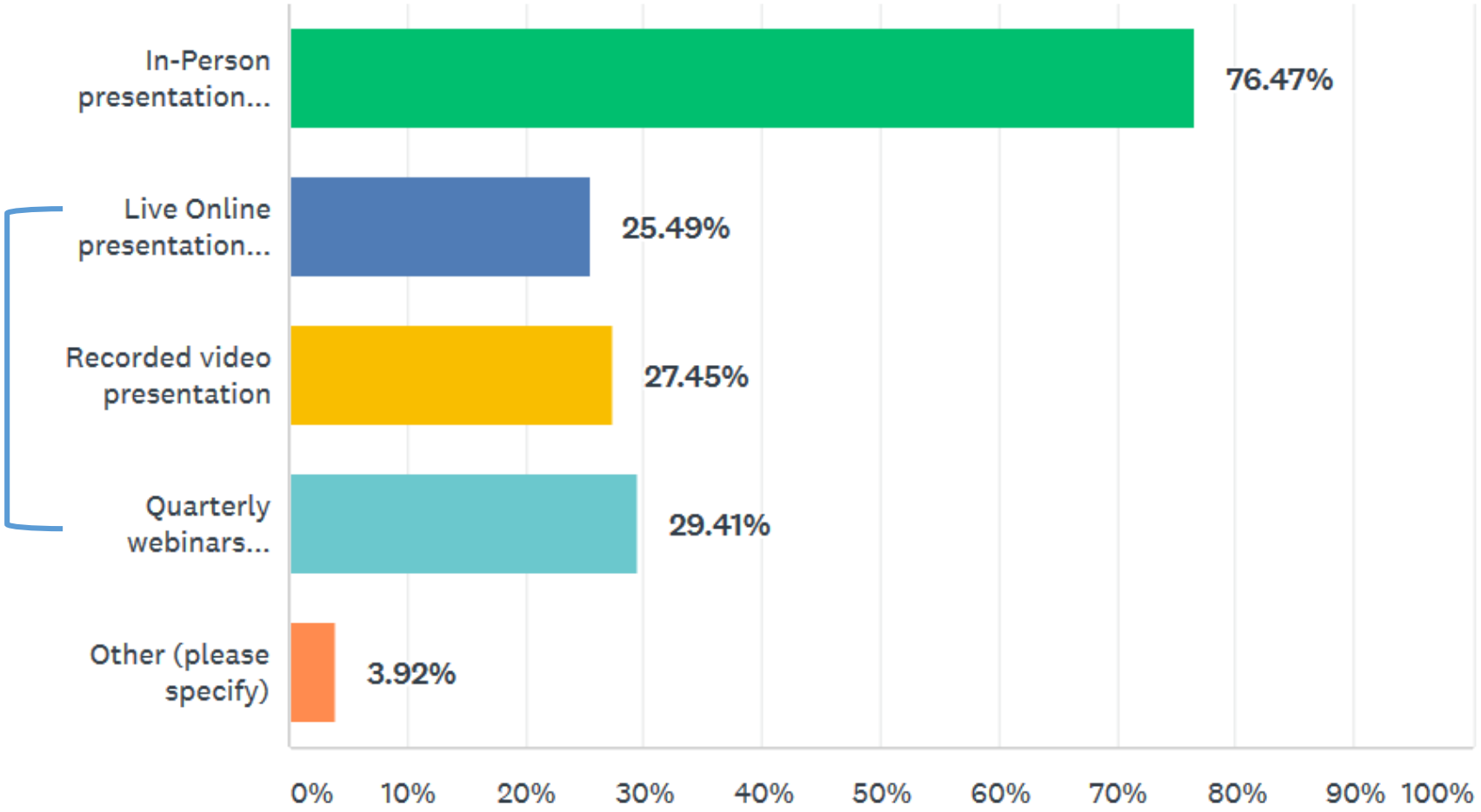
5/24/2018 10:59 PM

I loved it and was really happy to have this presented to our Lab staff. I liked the interactive part the best! Thank you

I loved the clicker interactions. It was fun to see what everyone else chose for answers and how we received information.

In the future, would you be interested in attending or watching any of the following? (check all that apply)

Answered: 51 Skipped: 6



GoToTraining

Web based educational platform for teaching hospitals, clinics, laboratories, and midwives

- We love providing in-person presentations, but can't do this all the time
- Ability to reach audiences across the state without leaving our desk!

Still exploring functionality, but seems useful so far

- Similar to 'GoToWebinar' but with teaching tools:
  - Real time polls and quizzes
  - Distribute course materials
  - Option to record trainings = view anytime
  - Send out comprehension test afterwards
  - Detailed attendance reports, test results, and evaluations

Newborn Screening Tests & Your Baby

Newborn Screening Program
P.O. Box 55729
1610 NE 150th Street
Shoreline, WA 98155-0729
Phone: (206) 418-5410
1-866-660-9050
FAX: (206) 418-5415
Email: NBS.Prog@doh.wa.gov
Website: www.doh.wa.gov/nbs

Washington State Department of Health
DOH 304-007
Rev. 06/14

What is newborn screening?

Newborn screening is a way to identify babies who are at risk for serious disorders that are treatable, but not apparent at birth. State law requires that a blood-spot specimen be collected from every baby born in Washington within 48 hours of birth. This specimen is used to test for potentially life-threatening disorders. (Chapter 70.83 RCW and Chapter 246-650 WAC)

Why is screening important?

This screening identifies disorders that, if not detected and treated early, can result in developmental delays, severe illness or even death. A newborn baby may look perfectly healthy, but still have a serious disorder. Finding these problems early and treating them can prevent many serious complications. Fortunately, treatment is available to prevent or greatly reduce the effects of these disorders.

Newborn screening tests are one important way to provide your baby with the best possible health care. A simple blood test can give you and your baby's health care provider information about your baby's health that you may not otherwise know.

How is screening done?

All tests are done from a few drops of blood taken from your baby's heel. The blood is collected on a special absorbent paper and sent to the Newborn Screening Program at the State Public Health Laboratories in Shoreline for testing. The hospital or health care provider who submitted the specimen is notified of the results within a few days.

When should screening be done?

Generally the first screening specimen should be collected when the baby is between 18 and 48 hours of age. This allows affected infants to be treated as soon as possible. The routine second specimen should be collected between 7 and 14 days of age, but it is still beneficial for older babies. Additional testing should also be done when requested by your baby's health care provider.

Why are two specimens recommended?

Most of the disorders will be detected on the first specimen collected taken on the day of birth. The second specimen is recommended because some disorders may not be detected until the baby is slightly older.

Sometimes more than two specimens may be requested. This does not mean your baby has one of the disorders. The most common reason for requesting an additional specimen is that the previous results were inconclusive.

There is only one charge per infant for the screening. Additional specimens are tested at no additional charge. However, your health care provider may charge a fee to collect the specimen. Diagnostic testing, if needed, will involve additional costs.

What disorders are detected?

For a complete list of conditions, please see the back the of this pamphlet

What happens if a disorder is suspected?

If the newborn screening test indicates a possible problem, your baby's health care provider will be contacted immediately. Diagnostic testing will be recommended so treatment can be started without delay if your baby is affected with one of the disorders.

How can I find out the results?

If you have questions about the results from your baby's screening tests, please contact your health care provider. If your health care provider does not have the results, he or she should contact the Newborn Screening Program to obtain a copy.

As a parent, may I refuse to have newborn screening done?

The law gives parents the right to refuse the screening tests for their baby only if this testing conflicts with their religious beliefs or practices. If this is true for you, be sure to tell the hospital staff or your health care provider.

Where can I get more information about newborn screening?

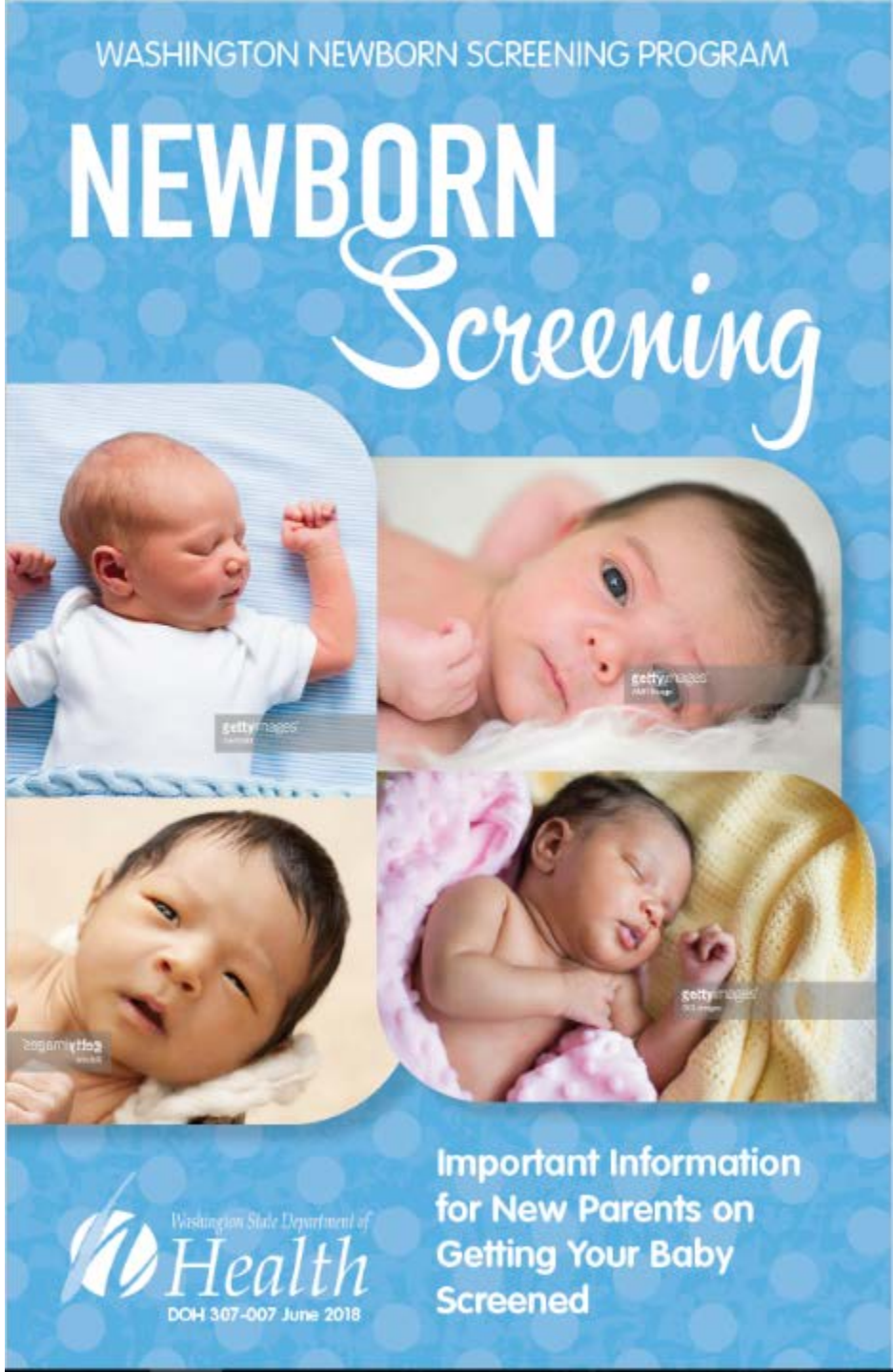
For more information, speak with your health care provider or contact the Newborn Screening Program using theinformation provided on the front of this pamphlet.

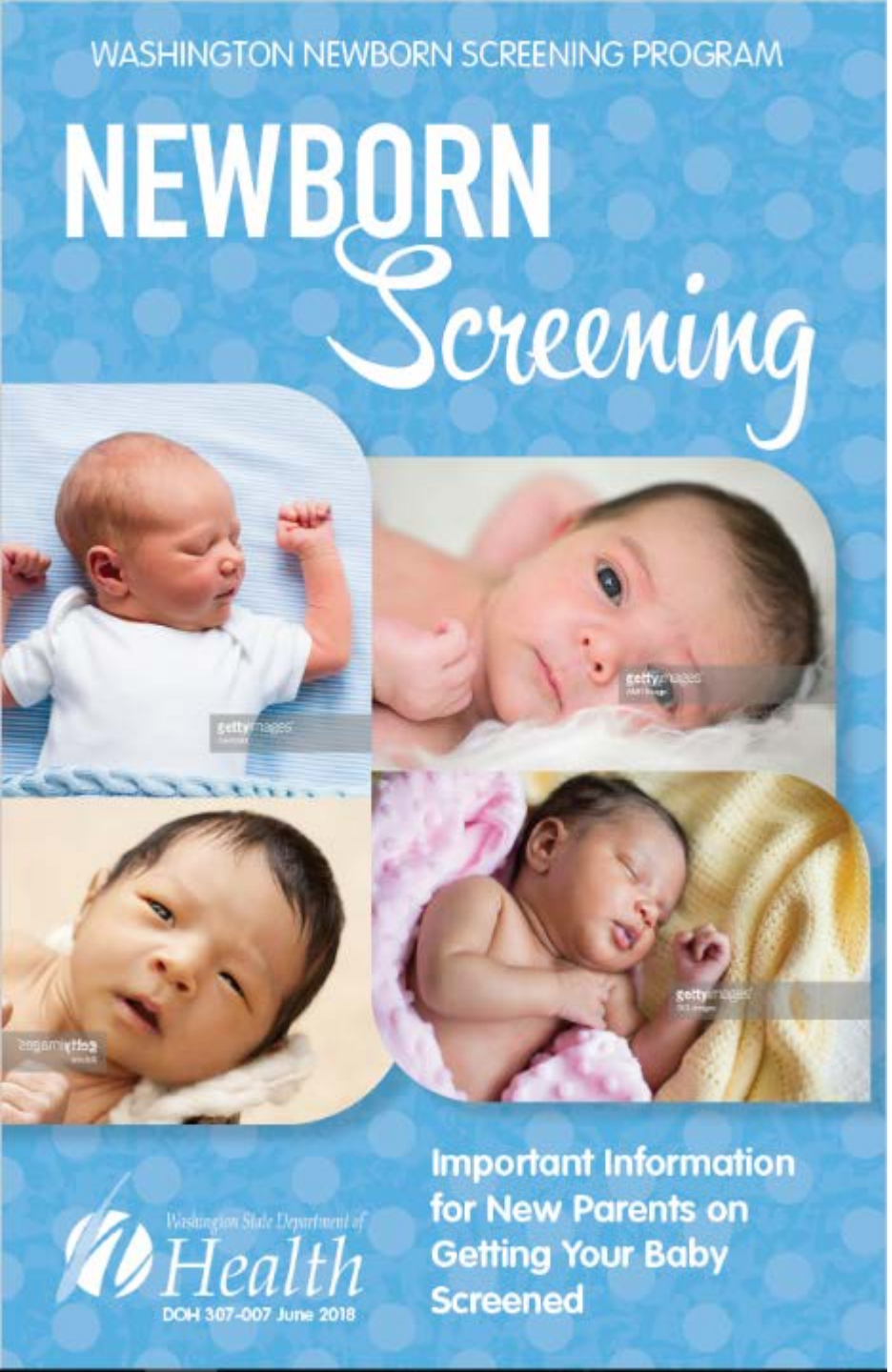
- Dated design
- Not visually appealing
- Limited use of photos/fonts/colors
- Not in ‘plain talk’
- Unsure if parents understand
- People just throw them away…
- We’re ready for something new!

Out with the old,

in with the new!

(DRAFT)





New Booklet

Working with our DOH Health Promotion & Communication team (never worked with them before)

- Two health educators, graphic designer, project manager

  - New modern design
    - Visually appealing!
  - Consulted with Baby's First Test on content
  - Revised language for 'plain talk' = understandable by broader audience
  - Will translate into 15 languages
  - Audience testing (in progress)
  - Longer booklet style allows for more information and nice layout
    - Combines several of our standalone handouts into one, easier to distribute

Sneak Peak!

X-Linked Adrenoleukodystrophy (X-ALD)

1 IN 34,000 BIRTHS

Babies with X-ALD cannot break down certain fatty acids. When these build up, it can cause the fatty covering of the nerves, brain, and spinal cord to break down. X-ALD primarily affects males, but females may also show symptoms and pass X-ALD onto their children.

TREATMENT

Possible treatments can include steroids, stem cell transplant, and/or gene therapy. Supportive therapies and management, such as physical therapy, can help the baby grow up healthy.

Lysosomal Storage Disorders (2)

1 IN 32,000 BIRTHS

There are two types of lysosomal storage disorders. Babies with lysosomal storage disorders are unable to break down the sugars and fats in food. This can lead to a build-up of toxins in the body that can cause problems with the baby's the brain, bones, muscles, or heart.

TREATMENT

Possible treatments include enzyme replacement therapy and stem cell transplant. Supportive therapies and management such as physical therapy can help the baby grow up healthy.

Sickle Cell & Hemoglobinopathies (HGB)

1 IN 4,700 BIRTHS

Babies with sickle cell disease or other hemoglobinopathies have abnormal red blood cells. The blood cells are unable to carry oxygen efficiently throughout the body. These disorders can cause frequent infections, severe pain, anemia, and other complications.

TREATMENT

Early treatment and proper lifelong management can prevent serious health problems.

Newborn Screening Checklist

- ☐ Make sure the newborn screening was completed after birth.
- ☐ Check with hospital staff or your midwife that they have the right phone number and address to reach you.
- ☐ Choose a health care provider for your baby.
- ☐ Take your baby in for a well-child visit when they are one to two weeks old to get their second newborn screen.
- ☐ If you don't receive screening results, call your health care provider to see if they have them.
- ☐ Follow any instructions for more tests, appointments, or follow up care.

Notes & Questions to Ask

Thank You

Lani Culley
lani.culley@doh.wa.gov

Gauri Gupta
gauri.gupta@doh.wa.gov

Heidi Lovejoy
heidi.lovejoy@doh.wa.gov

Washington State Department of Health
Newborn Screening Program
(206) 418-5410
NBS.Prog@doh.wa.gov
www.doh.wa.gov/nbs