# SAVING LIVES WITH A SIMPLE BLOOD SPOT

## NEWBORN SCREENING

Washington State Department of Health

# What is Newborn Screening?

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

There are 3 types of newborn screening programs:

1

**Blood-spot**

2

**Blood-spot Hearing**

3

**Pulse Oximetry**

# Why is Newborn Screening Important?

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

*Two 6 year old girls with Congenital Hypothyroidism*
*Healthy 18 year old with CH, detected through Washington Newborn Screening as a baby*

# Washington Screens for... 32 disorders!

| Amino Acid Disorders (6) | Fatty Acid Oxidation Disorders (5) | Organic Acid Disorders (8) |
|---|---|---|
| Phenylketonuria | Medium-chain acyl-CoA dehydrogenase deficiency | Isovaleric acidemia |
| Homocystinuria | Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency | Glutaric acidemia type I |
| Maple syrup urine disease | Trifunction[illegible]ency | Methylmalonic acidemias (CblA/B and MUT) |
| Citrullinemia type I | Very long-chain acyl-Co[illegible]e deficiency | Propionic acidemia |
| Argininosuccinic acidemia | Carnitine [illegible]ct | Multiple carboxylase deficiency |
| Tyrosinemia type I | | Beta-ketothiolase deficiency |
| | | 3-hydroxy-3-methylglutaric aciduria |

**32 Disorders**

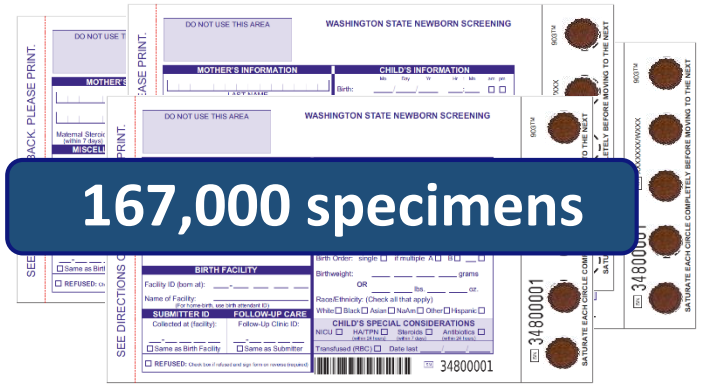
| Endocrine Disorders (2) | Lysosomal Storage Disorders (2) | Other Disorders (10) |
|---|---|---|
| Congenital hypothyroidism | Mucopolysaccharidosis type I | Galactosemia |
| Congenital adrenal hyperplasia | Glycogen storage disorder type II (Pompe) | Biotinidase deficiency |
| | | Cystic fibrosis |
| | | Sickle Cell Diseases & Hemoglobinopathies |
| | | Severe combined immunodeficiency |
| | | X-linked adrenoleukodystrophy |
| | | Spinal muscular atrophy |

# Immediately Life Threatening Conditions

| Amino Acid Disorders (6) | Fatty Acid Oxidation Disorders (5) | Organic Acid Disorders (8) |
|---|---|---|
| Phenylketonuria<br>Homocystinuria<br>Maple syrup urine disease<br>Citrullinemia type I<br>Argininosuccinic acidemia<br>Tyrosinemia type I | Medium-chain acyl-CoA dehydrogenase deficiency<br>Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency<br>Trifunctional protein deficiency<br>Very long-chain acyl-CoA dehydrogenase deficiency<br>Carnitine uptake defect | Isovaleric acidemia<br>Glutaric acidemia type I<br>Methylmalonic acidemias (CblA/B and MUT)<br>Propionic acidemia<br>Multiple carboxylase deficiency<br>Beta-ketothiolase deficiency<br>3-hydroxy-3-methylglutaric aciduria |
| **Endocrine Disorders (2)** | **Lysosomal Storage Disorders (2)** | **Other Disorders (10)** |
| Congenital hypothyroidism<br>Congenital adrenal hyperplasia | Mucopolysaccharidosis type I<br>Glycogen storage disorder type II (Pompe) | Galactosemia<br>Biotinidase deficiency<br>Cystic fibrosis<br>Sickle Cell Diseases & Hemoglobinopathies<br>Severe combined immunodeficiency<br>X-linked adrenoleukodystrophy |

# Washington State Numbers

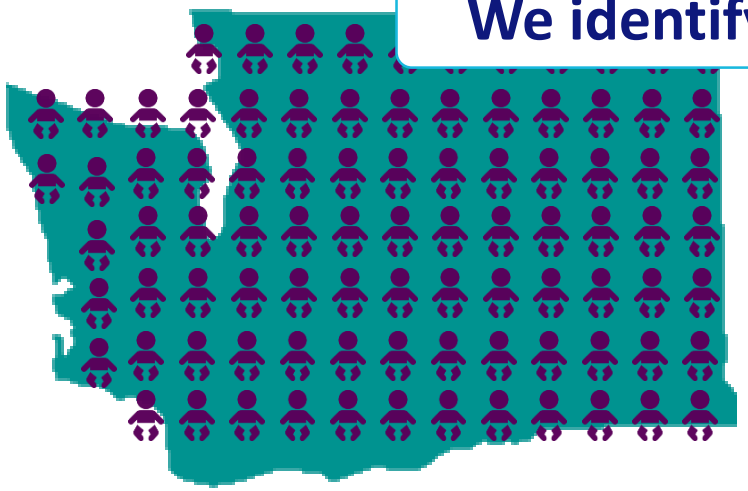
**85,000 newborns**




**167,000 specimens**

**12 million tests**

**We identify:**



**200 infants**

**every year who benefit from early diagnosis and treatment**

**1,300 infants with a hemoglobin trait (not disease)**

# WA Newborn Screening Process

**BABY IS BORN!**

**18-48**
**HOURS AFTER BIRTH**

**Collect first specimen**

**Dry specimen**
for 3 hours


**Transport specimen**
to the State Lab
ASAP!


**BEST PRACTICE**
**24**
**HOUR TRANSIT**

At the Lab, punches are taken from the dried blood spots to test for each of the disorders

Same day/next day results


Results are mailed back to who submitted the specimen

# What happens when a baby has abnormal results?

Dedicated team ensures the baby gets the care they need

- Depends on what the results are and which condition is suspected

Can include:

- Ensure repeat specimen is submitted to resolve borderline results
- Facilitate prompt diagnostic testing and treatment for non life-threatening conditions
- Call baby's health care provider to check clinical status, recommend immediate evaluation and diagnostics for life-threatening conditions
- After confirmed diagnoses, ensure baby is linked into specialty care

# Specialty Care Partners

**Consultants:**

- Seattle Children's Hospital
  - Endocrinologist
  - Pediatric Hematologist
  - Biochemical Geneticists
  - Immunologists
  - Pulmonologists
- Mary Bridge Children's
  - Biochemical Geneticists

**Community:**

- Northwest Sickle Cell Collaborative

**Specialty Care Clinics:**

- University of Washington
  - PKU Clinic
  - Congenital Hypothyroidism Developmental Evaluation Clinic
  - Neuropsych Evaluation Program
  - Biochemical Genetics Clinic
- Seattle Children's Hospital
  - Biochemical Genetics Clinic
  - Odessa Brown Sickle Cell Clinic
- Mary Bridge Sickle Cell Clinic

Mary Bridge Children's Hospital · Clinics · Foundation
1955 · CELEBRATING 60 YEARS · 2015

UW Medicine
DEPARTMENT OF MEDICINE
DEPARTMENT OF PEDIATRICS
UNIVERSITY of WASHINGTON

Seattle Children's
HOSPITAL · RESEARCH · FOUNDATION

northwest sickle cell COLLABORATIVE

ODESSA BROWN CHILDREN'S CLINIC

Cristine M. Trahms Program for Phenylketonuria
ΦKU Seattle
A Member Forever

# How Much Does Screening Cost?

- Fee for screening: $119.30 as of August 7, 2020
- This one-time fee covers all newborn screens an infant receives in WA

  (No additional charge for 2nd or 3rd screens!)

- The Department of Health bills the facility that collected the baby's initial specimen
- The facility then bills the patient's insurance

# Quality Assurance & Development

## Surveillance

- Ensure every baby in the state receives a valid newborn screen

## Education & Outreach

- Provide assistance to health care facilities
- Create educational materials
- Promote newborn screening in the community

## Tracking & Reporting

- Send quarterly reports to each facility about their performance in meeting newborn screening guidelines
  - Specimen Collection and Transit Timing Compliance
  - Specimen Quality
  - Demographic Errors

# 2019 WA State Disorder Summary

| General | |
|---|---|
| Hospital, Birth Center & Home Births | 84,831 |
| Specimens Tested | 165,626 |
| **Infants Diagnosed** | |
| Amino Acid Disorders | 6 |
| Biotinidase Deficiency | 0 |
| Congenital Adrenal Hyperlasia | 4 |
| Congenital Hypothyroidism | 99 |
| Cystic Fibrosis | 10 |
| Fatty Acid Oxidation Disorders | 7 |
| Galactosemia | 1 |
| Organic Acid Disorders | 0 |
| Severe Combined Immunodeficiency | 2 |
| Sickle Cell Disease and Other Clinically Significant Hemoglobinopathies | 25 |
| X-linked Adrenoleukodystrophy | 8 |
| Lysosomal Storage Disorders | 0 |
| **TOTAL** | **162** |

~Thank you~

Together we protect the lives of
Washington's youngest citizens.

Washington State Department of Health