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

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	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	Trifunctional protein deficiency	Methylmalonic acidemias (CblA/B and MUT)
Citrullinemia type I	Very long-chain acyl-CoA dehydrogenase deficiency	Propionic acidemia
Argininosuccinic acidemia	Carnitine uptake defect	Multiple carboxylase deficiency
Tyrosinemia type I		Beta-ketothiolase deficiency
		3-hydroxy-3-methylglutaric aciduria

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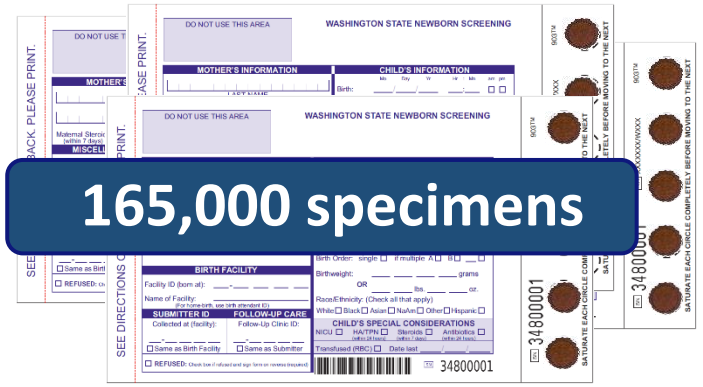
Coming Soon!

Screening for Ornithine Transcarbamylase Deficiency (OTCD)

Anticipate starting screening in Summer 2023
(pending budget approval)

Washington State Numbers

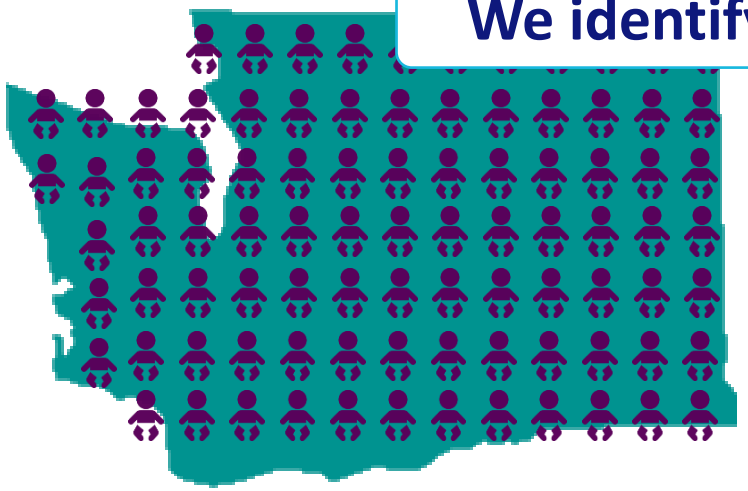
84,000 newborns



165,000 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

2020 WA State Disorder Summary

General	
Hospital, Birth Center & Home Births	84,529
Specimens Tested	159,922
Infants Diagnosed	
Amino Acid Disorders	9
Biotinidase Deficiency	2
Congenital Adrenal Hyperlasia	6
Congenital Hypothyroidism	108
Cystic Fibrosis	14
Fatty Acid Oxidation Disorders	7
Galactosemia	3
Organic Acid Disorders	0
Severe Combined Immunodeficiency	5
Sickle Cell Disease and Other Clinically Significant Hemoglobinopathies	14
X-linked Adrenoleukodystrophy	13
Lysosomal Storage Disorders	4
Spinal Muscular Atrophy	1
TOTAL	**186**

~Thank you~

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

Washington State Department of Health