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

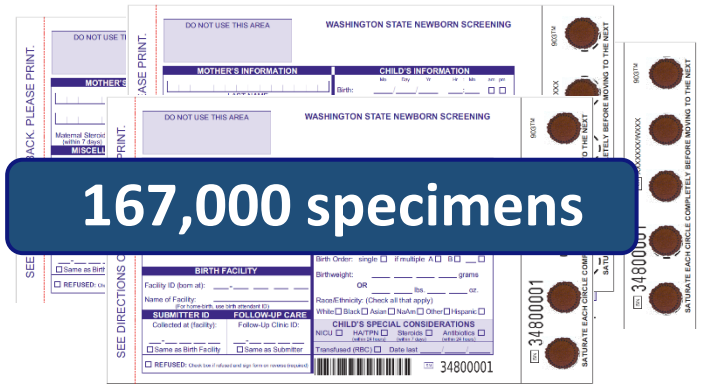
Two 6 year old girls with Congenital Hypothyroidism
Healthy 18 year old with CH, detected through Washington Newborn Screening as a baby

Washington Screens for… 32 disorders!

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

Washington State Numbers

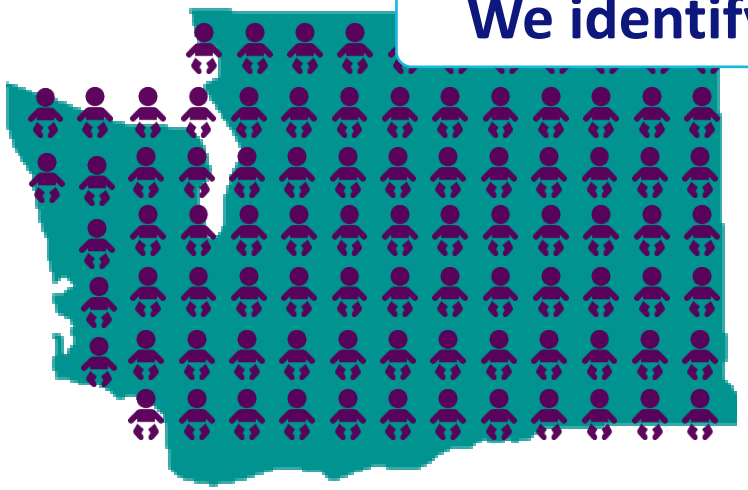
84,000 newborns



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)

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:

- Call baby's health care provider to check clinical status, recommend immediate evaluation and diagnostics for life-threatening conditions
- Ensure repeat specimen is submitted to resolve borderline results
- 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

Recommended Uniform Screening Panel (RUSP)

Federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)

- Nomination to RUSP
- Full evidence review and voting

- GAMT added in January 2023
- ARG1 is considered a Secondary Condition

~Thank you~

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

Washington State Department of Health