



# Newborn Screening in Washington State

*Saving lives with a simple blood spot*

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**PUBLIC HEALTH**  
ALWAYS WORKING FOR A SAFER AND  
HEALTHIER COMMUNITY



# What is *Newborn Screening*?

An integrated system that includes:

- Universal screening for all infants
- Follow-up to assure appropriate clinical response
- Diagnosis of affected infants
- Appropriate treatment and clinical care
- Evaluation of system effectiveness

# Newborn Screening Goal

Correctly identify babies with congenital disorders and assure that they receive treatment as soon as possible.



# What does Washington screen for?

Phenylketonuria (PKU)  
Congenital Hypothyroidism  
Congenital Adrenal Hyperplasia  
Sickle Cell Diseases & Hemoglobinopathies  
Biotinidase Deficiency  
Galactosemia  
Homocystinuria  
Maple Syrup Urine Disease  
MCAD Deficiency  
Cystic Fibrosis  
Isovaleric Acidemia  
Glutaric Acidemia type I  
Methylmalonic Acidemia  
Propionic Acidemia  
Long-chain L-3-OH Acyl-CoA Dehydrogenase  
Deficiency  
Trifunctional Protein Deficiency

Very-long-chain Acyl-CoA Dehydrogenase  
Deficiency  
Citrullinemia type I  
Argininosuccinic Acidemia  
Carnitine Uptake Defect  
Holocarboxylase Synthetase Deficiency  
b-Ketothiolase Deficiency  
3-hydroxy-3-methylglutaric aciduria  
Tyrosinemia type I  
Severe Combined Immunodeficiency

# 28 Disorders

# Washington Newborn Screening Numbers

~86,000 newborns are screened each year

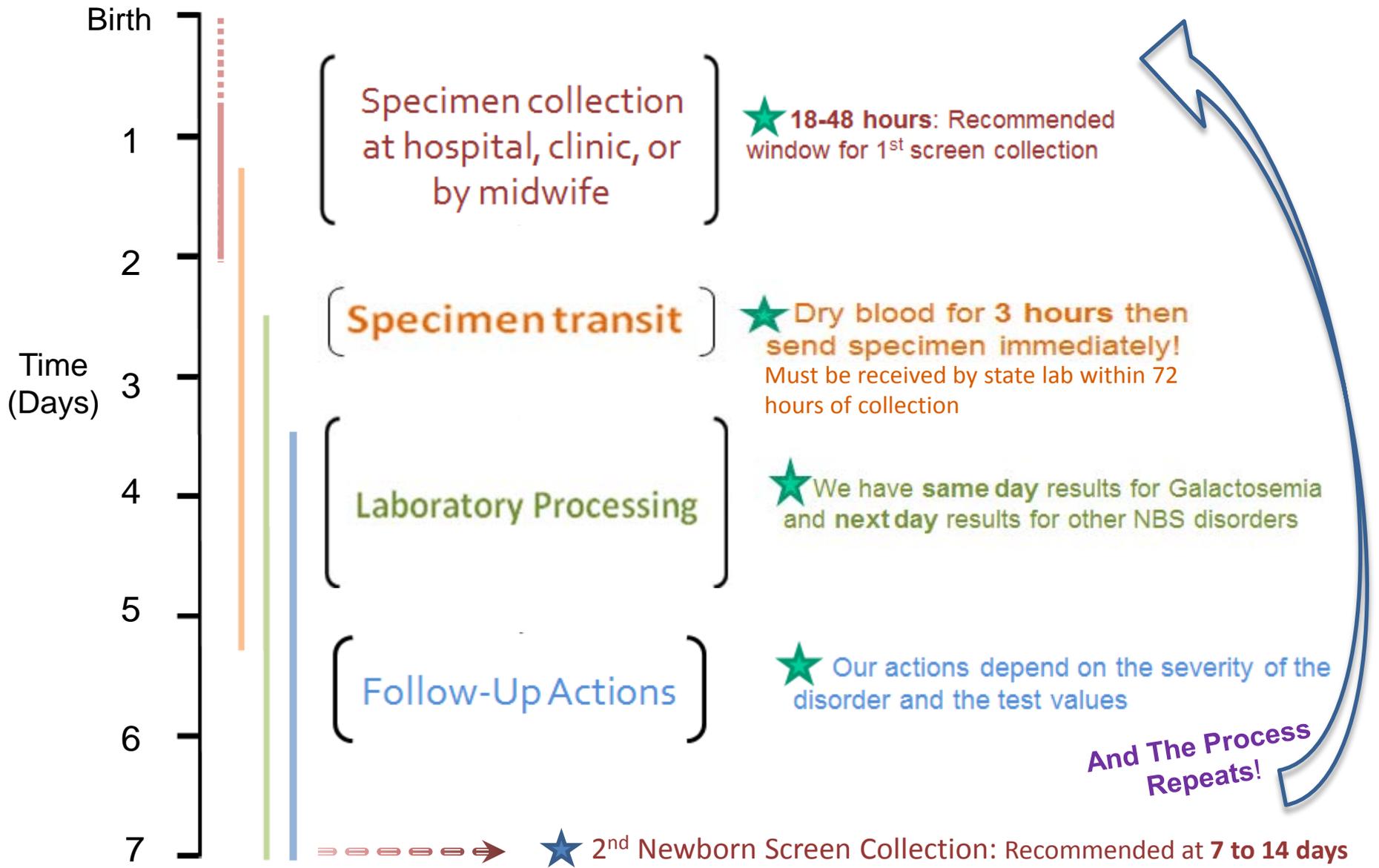
~174,000 specimens are processed

~10 million tests are performed

~2,100 false positives

Prevents death or disability for 150 - 200 babies/year  
2014 = 189 true cases in Washington

# Washington Newborn Screening Process



# Laboratory Processing

## 1. Specimen Preparation

- Specimen sorted, checked, and numbered

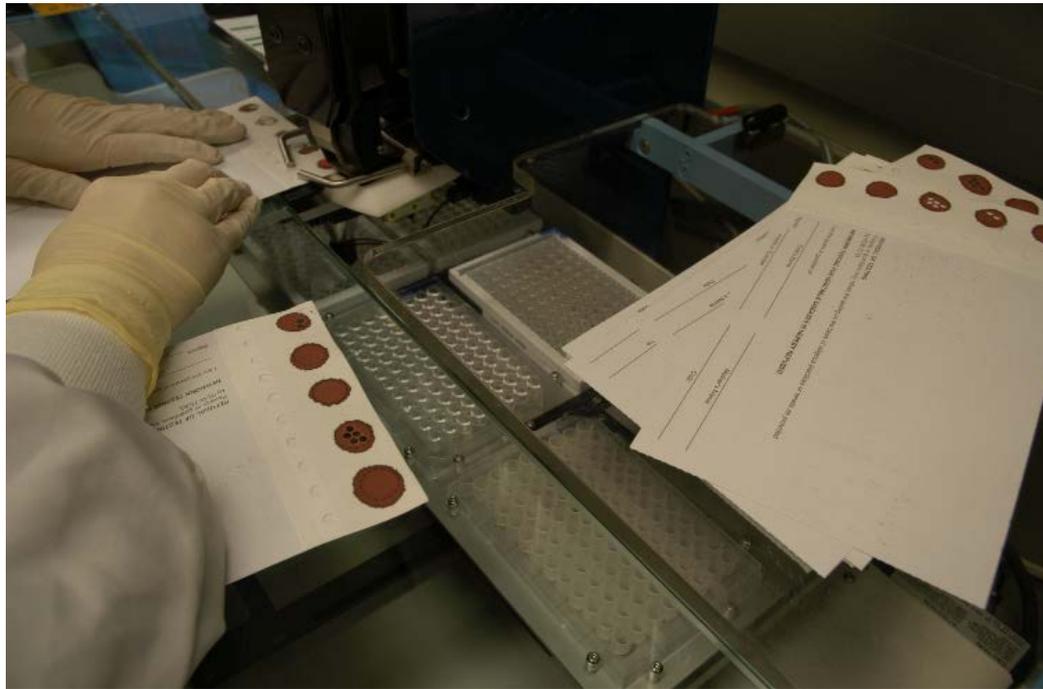


*Specimens sorted and checked*

# Laboratory Processing

## 2. Setting Up Tests

- Samples “punched” to test for different disorders



*Specimens are “punched”*

# Laboratory Processing

## 3. Testing and Reporting Results

- Each test has specific cutoff ranges and follow-up responses
- Critical abnormal results reported to follow-up immediately
- Redundant system – email reports and action manager



*Tandem Mass Spectrometer*

*SCID PCR  
Instrument*



*Fluoroimmunoassay Instrument*



*Galactosemia Instrument*



*Hemoglobin Gel Electrophoresis*

# Laboratory Processing

## 4. Quality Control

- Test results checked for accuracy and precision
- Use standards and controls and track performance over time
- Additional tests ordered if needed

# Disorder Follow-Up

- Reviews abnormal screening results and decides appropriate follow-up actions
- Reports results to health care provider and facilitates diagnostic testing
- Assures proper medical care is taken for babies with confirmed diagnoses
- Ensures subsequent specimens are submitted to resolve borderline results
- Tracks abnormal results and confirmed diagnoses over time



# Newborn Screening System



# Quality Assurance & Development

- Surveillance – Ensures that...
  - Every baby has a valid screen
- Compliance – Reports...
  - Collection compliance
  - Transit compliance
- Education – Provides...
  - Technical assistance to health care facilities
  - Educational materials on newborn screening

# 2014 Washington State Disorder Summary

Hospital, Birth Center & Home Births	87,398
Specimens Tested	173,908

Infants	Diagnosis
6	Amino Acid disorders
1	Biotinidase Deficiency
5	Congenital Adrenal Hyperplasia
113	Congenital Hypothyroidism
14	Cystic Fibrosis
10	Fatty Acid Oxidation disorders
10	Galactosemia
3	Organic Acid disorders
1	Severe Combined Immunodeficiency
26	Sickle Cell Disease and Other Clinically Significant Hemoglobinopathies
<b>189</b>	<b>Total Dried Blood Spot Disorders</b>

# Our Newborn Screening Team

