

SAVING LIVES AND PREVENTING DISABILITY



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:







Pulse Oximetry

Blood-spot

Hearing

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



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Healthy 18 year old with CH, detected through Washington Newborn Screening as a baby

Mandated Screening 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 (CbIA/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

Immediately Life-Threatening Conditions

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Screening for Ornithine Transcarbamylase Deficiency (OTCD)

Anticipate starting screening in Summer 2023 (pending budget approval)

WA Newborn Screening Process



What happens when a baby has abnormal results?

Dedicated team ensures the baby gets the care they need

• Depends on the results 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 lifethreatening conditions
- After confirmed diagnoses, ensure baby is linked into specialty care



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



Washington State Numbers





200 infants with blood spot conditions **170 infants** with early hearing loss



EHDDI Program Overview

Early Hearing Detection Diagnosis Intervention



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EHDDI Program Goals

National 1-3-6 Goals for all state EHDDI Programs



All infants receive a hearing screen before they are **one** month old.

Infants who do not pass two hearing screens have a diagnostic evaluation before they are **three** months old.



Infants who are deaf or hard of hearing start early intervention (EI) services before they are **six** months old.

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What's The Rush?



The first months of an infant's life are a critical time for developing language. Delays in identification can lead to developmental delays.

Research shows that children who are deaf or hard of hearing have better outcomes when they receive early intervention prior to 6 months of age.



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Why Screen all infants?

Hearing loss is a common condition present at birth

1-3 per 1000 births

It's invisible



EHDDI Program Follow-up

- Monitor that EHDDI 1-3-6 goals are met by collecting and reviewing data:
 - Hearing screening results
 - Reported on hearing screen cards by hospitals, midwives, and audiologists
 - Diagnostic hearing evaluation results
 - Reported online and via fax by audiologists
 - Early intervention enrollment data
 - Obtained through an electronic data exchange with the Early Support for Infants and Toddlers (ESIT) program

Risk Factors for Late-Onset Hearing Loss

- Neonatal Intensive Care (NICU) stay of >five days
- Stigmata or other findings associated with a syndrome known to include hearing loss
- Family history of permanent hearing loss
- Craniofacial anomalies
- In-utero infections with cytomegalovirus, herpes, toxoplasmosis, rubella, or syphilis

D	EPARTMENT OF HEALTH
Early Hearing-los	s Detection, Diagnosis, and Intervention Program
1610 N.E. Phone 206 418 5613 Toll	150 th Street · Shoreline, Washington 98155 Free 1 888 WAEHDDI (1 888 923 4334) Fey 206 364 0074
Filone 200-418-3013 10ii	Fiee 1-000-WAEHDDI (1-000-925-4554) Fax 200-504-0074
Action Needed: Passed H	learing Screen but has Risk Factor for Hearing Loss
TO: WASHINGTON STATE PEDIATRICS	
REVINKE SKYWALKEP	
DOB: May 12, 1977	EHDDI #:
Birth Facility: NEWBORN SCREENING	
Newborn Hearing Screen Results	PASSED <u>BUT</u> AT RISK
Risk Factor(s) for Late-onset/Progressive Lo	DSS IN UTERO INFECTION
Follow-up Needed	DIAGNOSTIC AUDIOLOGY EVALUATION
Due Date	3 MONTHS OF AGE FOR CYTOMEGALOVIRIUS (CMV) 9 MONTHS OF AGE FOR OTHER IN LITERO INFECTIONS
For a list of audiology clinics for infants, plea	ase visit <u>www.doh.wa.gov/infantaudiology</u> .
Children with cytomegalovirus infection, syn disorders, trauma, or culture-positive postna nore frequent evaluations. For more informa www.doh.wa.gov/hearingriskfactors.	dromes associated with progressive hearing loss, neurodegenerative tal infections associated with sensorineural hearing loss may need earlier and ation about risk factors for late-onset or progressive hearing loss, please visit
PLEASE FAX THE FOLLOWING INFORM	ATION REGARDING YOUR ACTIONS TO (206) 364-0074.
] This patient was referred to	on (name of audiology clinic) (date)
] We shared the hearing screen results a and they understand the recommendat	and recommendations with the patient's parent or legal guardian ions.
] This patient does not have any of the ri	sk factors indicated.
] This patient's parent or legal guardian of	declined further testing.

EHDDI Program Follow-up

- Recommend follow-up through primary care providers (PCPs) when an infant needs additional testing or services.
- Work with audiologists, Family Resources Coordinators (FRCs), and PCPs to ensure audiology and early intervention referrals are placed and received.
- Provide families with resources when a child is referred for diagnostic testing and identified as deaf or hard of hearing.
 - ~170 infants are identified each year in Washington



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