Data Summary

Newborn Screening 2005 – 2006

With Preliminary Data for 2007

September 2008

Washington State Department of Health
Epidemiology, Health Statistics
and Public Health Laboratories

Introduction

This following information is presented by the Washington State Department of Health (department) to the Washington State Board of Health in accordance with the newborn screening regulations:

WAC 246-650-040 Report to the board.

The department shall report to the board annually the following information concerning tests conducted pursuant to this section:

> *(1) The costs of tests as charged by the department;*
>
> *(2) The results of each category of tests, by county of birth and ethnic group, as reported on the newborn screening form and, if available, birth certificates; and*
>
> *(3) Follow-up procedures and the results of such follow-up procedures.*

Information on newborn screening during 2005 - 2006 and preliminary information on 2007 are presented in the attached series of tables and accompanying explanations. Data relating to all births were extracted from birth certificates by the department's Center for Health Statistics. These data relate to live-birth occurrences within the state. Data relating to infants detected, infants screened, and costs were extracted from data routinely maintained by the department. Information for 2007 is considered preliminary because birth certificate data is not yet available.

Additional is provided which briefly describes all the disorders that are currently tested for by the newborn screening program.

Screening Costs 2005 – 2007

The department's cost to operate the program, including laboratory testing, monitoring to assure adequate screening for all infants, follow up of all abnormal findings, education, and evaluation, is covered through a fee charged for each infant through the facility of birth. For the period covered, the charge was $60.90 for each child.

In addition to the screening fee, a separate charge of $3.50 per birth was collected during this period to support specialty clinic care for infants diagnosed through newborn screening.

An additional $3.10 fee to support specialty clinic care was authorized by the 2005 legislature and implemented in November 2005. The $3.10 fee expired on June 30, 2007. Lost revenue from this fee was replaced with state general funds by the 2007 legislature.

2005 Births and Conditions Detected by County

County	2005 Births[1]	2005 INFANTS DETECTED[2]									ALL Infants detected
		PKU	CH	CAH	Hb	GAL	BIO	MCAD	MSUD	HCY	
Adams	551	0	0	0	0	0	0	0	0	0	0
Asotin	3	0	0	0	0	0	0	0	0	0	0
Benton	3,528	0	0	0	1	0	0	0	0	0	1
Chelan	1,398	0	0	0	0	0	0	0	0	0	0
Clallam	588	0	0	0	0	0	0	1	0	0	1
Clark	5,144	2	1	0	2	0	0	0	0	0	5
Columbia	0	0	0	0	0	0	0	0	0	0	0
Cowlitz	1,191	0	1	0	0	0	0	0	0	0	1
Douglas	0	0	1	0	0	0	0	0	0	0	1
Ferry	5	0	0	0	0	0	0	0	0	0	0
Franklin	486	0	0	0	0	0	0	0	0	0	0
Garfield	0	0	0	0	0	0	0	0	0	0	0
Grant	1,090	1	1	0	0	0	0	0	0	0	2
Grays Harbor	590	0	0	0	0	0	0	0	0	0	0
Island	269	0	0	1	0	0	0	0	0	0	1
Jefferson	126	0	0	0	0	0	0	0	0	0	0
King	27,160	5	18	3	16	2	0	2	0	0	46
Kitsap	1,980	0	0	0	1	0	0	0	0	0	1
Kittitas	304	0	0	0	0	0	0	0	0	0	0
Klickitat	413	0	0	0	0	1	0	0	0	0	1
Lewis	595	0	1	0	0	0	0	0	0	0	1
Lincoln	0	0	0	0	0	0	0	0	0	0	0
Mason	344	0	2	0	0	0	0	0	0	0	2
Okanogan	531	1	0	1	0	0	0	0	0	0	2
Pacific	7	0	0	0	0	0	0	0	0	0	0
Pend Oreille	111	1	0	0	0	0	0	0	0	0	1
Pierce	8,612	1	7	0	9	3	1	1	0	0	22
San Juan	6	0	0	0	0	0	0	0	0	0	0
Skagit	1,601	1	2	0	0	0	0	0	0	0	3
Skamania	1	0	0	0	0	0	0	0	0	0	0
Snohomish	5,808	1	8	0	5	0	0	0	0	0	14
Spokane	6,452	1	7	0	0	1	3	0	0	0	12
Stevens	281	0	0	0	0	0	0	1	0	0	1
Thurston	2,798	0	1	0	0	0	0	0	0	0	1
Wahkiakum	0	0	0	0	0	0	0	0	0	0	0
Walla Walla	920	1	0	0	0	1	0	0	0	0	2
Whatcom	2,130	1	2	0	1	0	1	0	0	0	5
Whitman	422	0	0	0	0	0	0	0	0	0	0
Yakima	4,082	1	3	2	0	0	1	0	0	0	7
TOTAL	**79,257**	**17**	**55**	**7**	**35**	**8**	**6**	**5**	**0**	**0**	**133**

[1] By county where birth occurred
[2] By county where infant resides

2006 Births and Conditions Detected by County

County	2006 Births[1]	2006 INFANTS DETECTED[2]										ALL Infants Detected
		PKU	CH	CAH	Hb	GAL	BIO	MCAD	MSUD	HCY	CF	
Adams	551	0	0	0	0	0	0	0	0	0	0	0
Asotin	0	0	0	0	0	0	0	0	0	0	0	0
Benton	3,767	0	1	1	0	0	0	0	0	0	1	3
Chelan	1,500	0	2	0	0	0	0	0	0	0	2	4
Clallam	607	0	0	0	0	0	0	0	0	0	0	0
Clark	5,602	0	3	0	0	0	0	1	0	0	1	4
Columbia	0	0	0	0	0	0	0	0	0	0	0	0
Cowlitz	1,255	0	0	0	0	0	0	0	0	0	0	0
Douglas	0	0	0	0	0	0	0	0	0	0	0	0
Ferry	4	0	0	0	0	0	0	0	0	0	0	0
Franklin	473	0	0	0	0	0	0	0	0	0	0	0
Garfield	0	0	0	0	0	0	0	0	0	0	0	0
Grant	1,182	0	0	0	0	0	0	1	0	0	0	1
Grays Harbor	584	0	0	0	0	0	0	0	0	0	0	0
Island[a]	215	0	0	0	1	0	0	0	0	0	0	1
Jefferson	150	0	0	0	0	0	0	0	0	0	0	0
King	28,930	4	16	1	9	2	2	0	0	1	4	39
Kitsap[a]	2,088	0	0	0	0	0	0	0	0	0	0	0
Kittitas	343	0	0	0	0	0	0	0	0	0	0	0
Klickitat	121	0	0	0	0	0	0	0	0	0	0	0
Lewis	623	0	0	0	0	0	0	0	0	0	0	0
Lincoln	3	0	0	0	0	0	0	0	0	0	0	0
Mason	329	0	1	0	0	0	0	0	0	0	0	1
Okanogan	580	1	1	0	0	0	0	0	0	0	0	2
Pacific	4	0	0	0	0	0	0	0	0	0	0	0
Pend Oreille	115	0	0	0	0	0	0	0	0	0	0	0
Pierce	9,148	0	6	1	1	1	0	1	0	0	3	13
San Juan	4	0	0	0	0	0	0	0	0	0	0	0
Skagit	1,636	0	1	1	0	0	0	0	0	0	0	2
Skamania	4	0	0	0	0	0	0	0	0	0	0	0
Snohomish	5,850	0	5	1	0	0	1	0	0	0	1	8
Spokane	6,894	1	2	0	0	1	0	0	0	0	0	4
Stevens	289	0	0	0	0	1	0	0	0	0	0	1
Thurston	2,862	0	2	0	1	0	0	0	0	0	0	3
Wahkiakum	0	0	0	0	0	0	0	0	0	0	0	0
Walla Walla	959	0	0	0	0	0	0	0	0	0	0	0
Whatcom	2,183	0	1	0	1	1	0	0	0	0	1	4
Whitman	387	0	1	0	0	0	0	0	0	0	0	1
Yakima	4,230	1	1	0	0	0	0	0	0	0	0	2
TOTAL	**83,472**	**7**	**43**	**5**	**12**	**6**	**3**	**3**	**0**	**1**	**13**	**93**

[1] By county where birth occurred
[2] By county where infant resides

2005 Births and Infants Detected by Infant's Race/ Hispanic Ethnicity

INFANTS RACE	2005 BIRTHS[a]	2005 INFANTS DETECTED[b]									ALL INFANTS
		PKU	CH	CAH	Hb	GAL	BIO	MCAD	MSUD	HCY	
White	52,349	15	39	5	1	8	6	5	0	0	79
African American	4,875	0	1	0	11	0	0	0	0	0	12
Asian/Pacific Islander	8,245	0	7	1	18	0	0	0	0	0	26
Native American	2,350	1	2	0	0	0	0	0	0	0	3
Unknown/ Other	11,438	1	6	1	5	0	0	0	0	0	13
TOTAL	**79,257[c]**	**17**	**55**	**7**	**35**	**8**	**6**	**5**	**0**	**0**	**133**
Hispanic[d]	**17,707**	**1**	**7**	**1**	**1**	**1**	**1**	**0**	**0**	**0**	**12**

[a] From birth certificate data determined by an algorithm of mother and father's race developed by the National Center for Health Statistics.

[b] From information provided on the newborn screening test form; includes multiracial (more than one race designation on the screening form) or unknown (no designation made).

[c] Excludes infants born in military hospitals that do not participate in the Washington State Newborn Screening Program (1,972 born at Madigan Army Medical Center, 426 born at Oak Harbor Naval Hospital and 709 born at Bremerton Naval Hospital). Total excluded = 3,107.

[d] Hispanics can be of any race; they are included in the figures above.

2006 Births and Infants Detected by Infant's Race/ Hispanic Ethnicity

INFANTS RACE[a]	2006 BIRTHS	2006 INFANTS DETECTED[a]										ALL INFANTS
		PKU	CH	CAH	Hb	GAL	BIO	MCAD	MSUD	HCY	CF	
White	65,906	7	25	4	0	6	3	2	0	0	12	59
African American	5,527	0	1	0	8	0	0	0	0	0	0	9
Asian/Pacific Islander	8,897	0	5	0	2	0	0	0	0	1	0	8
Native American	2,542	0	2	0	0	0	0	1	0	0	0	3
Unknown/ Other [c]	600	0	10	1	2	0	0	0	0	0	1	14
TOTAL	**83,472[b]**	**7**	**43**	**5**	**12**	**6**	**3**	**3**	**0**	**1**	**13**	**93**
Hispanic[d]	**11,169**	**0**	**7**	**0**	**0**	**0**	**1**	**0**	**0**	**0**	**0**	**8**

[a] The infant's race for 2006 is from birth certificate data and was determined by an algorithm of mother and father's race developed by the National Center for Health Statistics. The race of infants detected is from information provided on the newborn screening test form.

[b] Excludes infants born in military hospitals that do not participate in the Washington State Newborn Screening Program (2,193 born at Madigan Army Medical Center, 404 born at Oak Harbor Naval Hospital and 730 born at Bremerton Naval Hospital). Total excluded = 3,327.

[c] Includes multiracial (more than one race designation on the screening form) or unknown (no designation made).

[d] Hispanics can be of any race; they are included in the figures above.

2005 Follow-Up Status of Infants Detected – Severe Disease[1]

FOLLOW-UP	2005 INFANTS DETECTED									ALL INFANTS
	PKU	CH	CAH	SCD	GAL	BIO	MCAD	MSUD	HCY	
Followed by medical specialist – (i.e., pediatric endocrinologist, hematologist, or comprehensive clinic,	9	52	7	4	2	0	5	0	0	79
Followed by primary care provider, with some consultation from specialist	0	0	0	0	0	0	0	0	0	0
Lost to follow-up	0	0	0	0	0	0	0	0	0	0
TOTAL	**9**	**52**	**7**	**4**	**2**	**0**	**5**	**0**	**0**	**79**

2006 Follow-Up Status of Infants Detected – Severe Disease[1]

FOLLOW-UP	2006 INFANTS DETECTED										ALL INFANTS
	PKU	CH	CAH	SCD	GAL	BIO	MCAD	MSUD	HCY	CF	
Followed by medical specialist – (i.e., pediatric endocrinologist, hematologist, or comprehensive clinic)	3	29	5	7	1	2	2	0	1	12	62
Followed by primary care provider, with some consultation from specialist	0	3	0	0	0	0	0	0	0	0	3
Lost to Follow-up	0	0	0	0	0	0	0	0	0	0	0
TOTAL	**3**	**32**	**5**	**7**	**1**	**2**	**2**	**0**	**1**	**12**	**65**

[1] All affected infants require follow-up, however, those with milder forms do not require immediate treatment.

2005 Age at which Treatment Began for Infants Detected – Severe Disease

Disorder	Number of Infants	Age Treatment began (days)	
		AVERAGE	RANGE
PKU	7	8	6 – 11
CH	52	13[a]	1 – 52
CAH	7	7	1 – 14
SCD	4	23	18 – 30
GAL	2	8	7 – 8
BIO	0	n/a	–
MCAD	5	13	8 – 22[b]
MSUD	0	n/a	–
HCY	0	n/a	–

[a] Includes seven infants who weren't detected until their second or third screen.

[b] Excludes one infant whose diagnosis was delayed until 73 days of age because his disorder was being masked by intravenous feedings he was receiving for his extended hospital stay for surgery.

2006 Age at which Treatment Began for Infants Detected – Severe Disease

Disorder	Number of Infants	Age Treatment began (days)	
		AVERAGE	RANGE
PKU	3	6	3 – 9
CH	32	15	4 – 43[a]
CAH	5	7	0 – 14[b]
SCD	7	40	22 – 68
GAL	1	6	n/a
BIO	2	6	3 – 9
MCAD	2	13	10 – 15
MSUD	0	n/a	n/a
HCY	1	31[c]	n/a
CF	12	20	0 - 61

[a] Includes six infants who weren't detected until their second or third screen.

[b] Includes one infant whose treatment was delayed until 14 days due to initially inconclusive diagnostic testing results.

[c] Infant wasn't detected until her second screen.

Preliminary Data for 2007 – Infants Detected

In 2007, about 85,000 infants were tested by the Washington State Newborn Screening Program. This excludes approximately 3,000 infants born at the three Washington military hospitals.

DISORDER	INFANTS DETECTED
Phenylketonuria (PKU)	7
Congenital Hypothyroidism (CH)	49
Congenital Adrenal Hyperplasia (CAH)	5
Cystic Fibrosis	14
Hemoglobinopathies[a]	23
Galactosemia	7
Biotinidase Deficiency	1
Medium chain acyl-CoA dehydrogenase deficiency (MCAD)	6
Maple Syrup Urine Disease (MSUD)	0
Homocystinuria	0
TOTAL	**112**

[a] Over 1,000 additional hemoglobin traits were detected; trait requires no treatment but the information is made available to the family through the child's health care provider.

NEWBORN SCREENING INFORMATION MATRIX

Disorders Currently Screened in Washington – September 2008

Disorder & (Prevalence in WA)	Definition	Screening Test	Impact Without early Treatment	Treatment	Benefits of Early Treatment
Biotinidase deficiency (1 in 60,000)	Deficiency of biotin, part of the Vitamin B complex	Enzyme assay: Measure Biotinidase activity	Seizures, damage to immune system, mental retardation, hearing loss	Oral biotin supplementation	Prevent all adverse consequences
Congenital adrenal hyperplasia (CAH) (1 in 16,000)	Impaired production of cortisol and other adrenal hormones	Measure Adrenal hormone: 17-hydroxyprogesterone (17-OHP)level	Salt loss & shock may result in early sudden death, virilization & abnormal growth	Cortisol & salt-retaining hormone replacement	Prevent death, reduce virilization & abnormal growth
Congenital hypothyroidism (1 in 1,600)	Inadequate production of thyroid hormone	Measure thyroid stimulating hormone (TSH) level	Mental retardation, growth failure	Thyroid hormone replacement	Normal growth and mental development
Cystic fibrosis (1 in 3000 expected)	Defect in the cystic fibrosis transmembrane conductance regulator (CFTR) gene	Measure immunoreactive trypsinogen (IRT) level	Thick, sticky mucus builds up in the lungs and digestive system	Pancreatic enzymes, vitamin supplements, chest psysiotherapy, antibiotics	Improve physical growth, cognitive function & possibly lung function
Galactosemia (1 in 40,000)	Inability to break down galactose, a major sugar found in milk	Enzyme assay: measure galactose-1-phosphate uridyl transferase (GALT) activity	Galactose accumulates in vital organs, leading to severe mental retardation, liver disease, blindness, overwhelming infections and death	Dietary restriction of galactose	Prevent death, improve mental function & reduce other morbidity
Sickle cell disease (1 in 10,000)	Production of abnormal hemoglobin	Separate and visualize hemoglobin proteins by electrophoresis	Severe infections and possible death	Antibiotic prophylaxis to help prevent infections & parental education to recognize health crises	Prevent death, reduce infections and other morbidity

NEWBORN SCREENING INFORMATION MATRIX

Disorders Currently Screened in Washington – September 2008

Disorder & (Prevalence in WA)	Definition	Screening Test	Impact Without early Treatment	Treatment	Benefits of Early Treatment
Amino acid disorders (1 in 10,000)	Inability to break down amino acids, found in all foods containing protein	Measure amino acid levels by MS/MS	Mental retardation, seizures, coma & death	Dietary restriction of offending amino acid(s) using a special metabolic formula	Prevent mental retardation and other neurological damage
Fatty acid disorders (1 in 13,000)	Inability to process or break down fats in the body due to missing or dysfunctional enzymes	Measure acylcarnitine levels by MS/MS	Serious damage to brain, liver, heart, eyes and muscles & death	High carbohydrate, low-fat diet & avoidance of fasting	Prevent mental retardation and other neurological damage
Organic acid disorders (1 in 25,000)	Inability to process or break down organic acids, byproducts of protein and fatty acid metabolism	Measure acylcarnitine levels by MS/MS	Severe nerve and physical damage & death	Dietary restriction of offending amino acid(s) and use of a special metabolic formula	Prevent mental retardation and other neurological damage

NEWBORN SCREENING INFORMATION MATRIX

Disorders Currently Screened in Washington – September 2008

Amino acid disorders:

- Argininosuccinic acidemia (ASA)
- Citrullinemia (CIT)
- Homocystinuria (HCYS)
- Maple Syrup Urine Disease (MSUD)
- Phenylketonuria (PKU)
- Tyrosinemia type I (TYR-I)

Organic acid disorders:

- 3-OH 3-CH3 glutaric aciduria (HMG)
- Beta-Ketothiolase deficiency (BKT)
- Glutaric acidemia type I (GA-I)
- Isovaleric acidemia (IVA)
- Methylmalonic acidemia (Cbl A, B)
- Methylmalonic acidemia *(mutase deficiency)* (MUT)
- Multiple carboxylase deficiency (MCD)
- Propionic acidemia (PROP)

Fatty acid oxidation disorders:

- Carnitine uptake defect (CUD)
- Long-chain L-3-OH acyl-CoA dehydrogenase (LCHAD) deficiency
- Medium chain acyl-CoA dehydrogenase (MCAD) deficiency
- Trifunctional protein (TFP) deficiency
- Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency