Annual Report

Newborn Screening 2012

October 2013

Washington State Department of Health

Disease Control and Health Statistics
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Contents

Executive Summary

In 2012 there were 86,180 infants born in Washington (an additional 1,179 were born at two military facilities in our state that do not participate in this program). The Department of Health's Newborn Screening (NBS) Program tested these infants for 27 treatable, but potentially deadly or disabling disorders that the Washington State Board of Health has specified in chapter 246-650 Washington Administrative Code (WAC). Among these infants, 115 were affected with a severe form of one of the disorders and were quickly enrolled in appropriate preventive care systems before they suffered irreversible damage from their conditions.

In addition, 94 infants were identified with a mild form of one of the disorders that required treatment or close monitoring, and 1,244 infants were identified with abnormalities of hemoglobin that, while not directly harmful, can have important implications for future reproduction choices for the infants and their parents. In these cases the infants' health care providers were notified of the findings, their implications, and were provided a list of resources to help families understand how the findings might impact them.

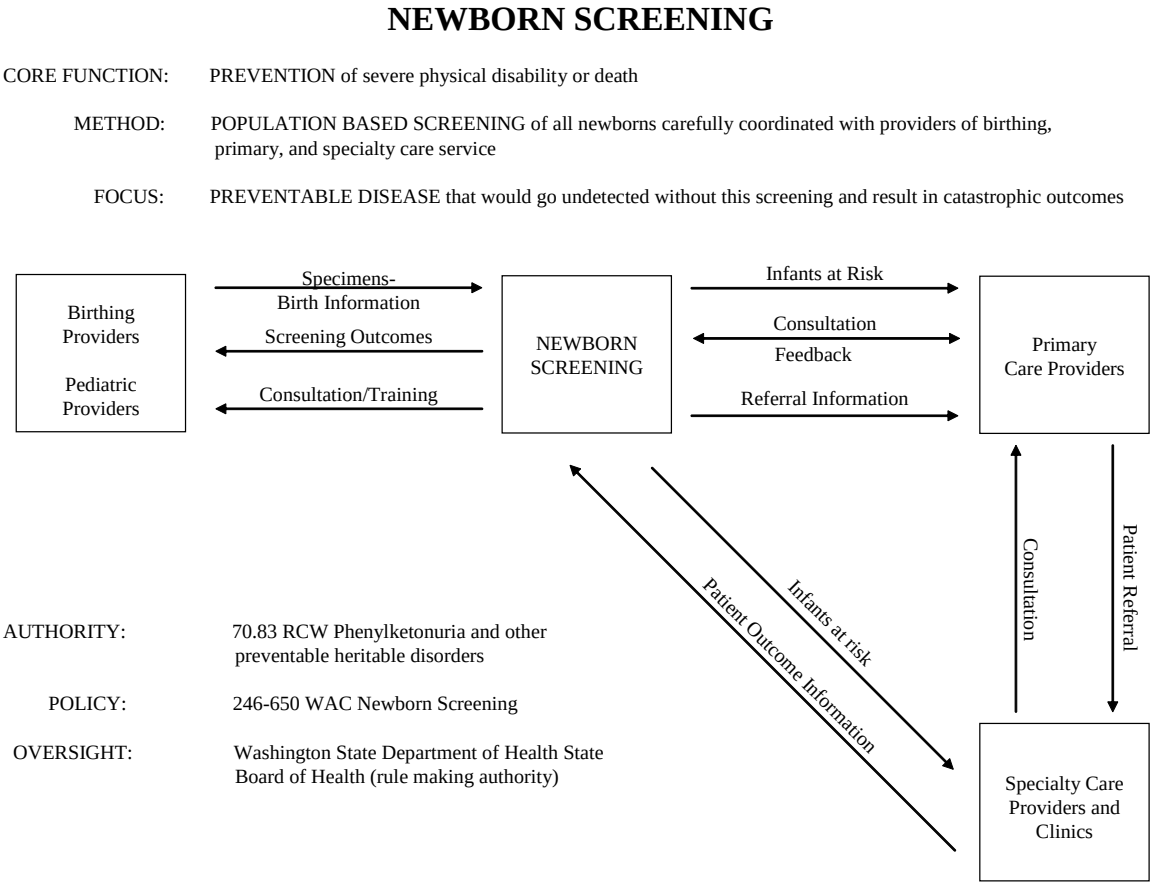
The department's cost to operate the program, including follow-up, education, and evaluation as well as the laboratory testing, is covered through a fee charged for each infant through the hospital of birth. In 2012 this charge was $60.90 for each child. This modest investment is typically covered by insurance and other third party payers. In return, the state's health care system realizes significant savings by avoiding the costs of lifetime treatment for disabling conditions.

Introduction

This report is presented in accordance with Washington Administrative Code (WAC) 246-650-040 which calls for an annual report of information on newborn screening to the Board of Health. Information on newborn screening during 2012 is presented in the attached series of tables and accompanying explanations. Data relating to all births were extracted from 2012 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.

The data exclude information relating to infants born at Oak Harbor Naval Hospital and Bremerton Naval Hospital in 2012. These military hospitals have their babies screened elsewhere and did not participate in Washington's Newborn Screening Program during this time.

Newborn Screening Schematic Overview



The Newborn Screening Program also strives to assure families' involvement in this system through their primary care providers and, for affected infants, through the specialty care providers and clinics.

Description of Disorders and Abbreviations Used

Following is a brief description of the disorders identified through the Newborn Screening Program and abbreviations that are used throughout the report.

AA **Amino acid disorders;** disorders of metabolism characterized by the body's inability to correctly process amino acids or the inability to process the ammonia that is released during the break down of amino acids. The accumulation of amino acids, ammonia or other by-products may cause severe complications including intellectual disabilities, coma, seizures, and possibly death. Early detection and treatment can prevent most or all of these consequences. Disorders are:

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

BIO **Biotinidase deficiency;** deficiency of the enzyme that affects normal recycling of biotin, one of the B vitamins. If untreated, a severe deficiency of biotin can result in metabolic crisis, coma and death. Treatment with biotin can prevent all symptoms.

CAH **Congenital adrenal hyperplasia**; excessive production of androgenic hormones due to a defect in the pathway that converts cholesterol into the hormone cortisol. The most common defect, and the primary target of our screening is deficiency of the enzyme 21-hydroxylase. Deficiency of this enzyme accounts for approximately 95% of all cases of CAH. If untreated, CAH can lead to an imbalance in the body's concentration of salts which in turn can rapidly lead to shock and death. It also causes excessive masculinization and extremely premature sexual maturation. Treatment consists of providing cortisol which normalizes hormone production. Proper treatment prevents death and stops the masculinization process. Affected females may require surgical correction of masculinized genitalia.

CH **Congenital hypothyroidism**; insufficient production of the thyroid hormone thyroxine due to malformation or malfunction of the thyroid gland. If untreated, CH can result in severe neurological and developmental damage. Treatment consists of hormone replacement with synthetic thyroxine. Affected infants develop normally with proper treatment.

CF **Cystic Fibrosis;** defect in the cystic fibrosis transmembrane conductor regulator (CFTR) gene which regulates the body's salt and water secretions. This results in the build-up of thick, sticky mucus in the lungs and digestive system. Treatment consists of pancreatic enzymes, vitamin supplements, chest physiotherapy, and antibiotics. Early treatment improves physical growth and cognitive function, and possibly lung function.

Description of Disorders and Abbreviations Used, continued

FAO **Fatty acid oxidation disorders;** these are disorders of metabolism characterized by the inability to efficiently use fat to make energy. During times of extra energy need such as prolonged fasting or acute illness, affected infants can suffer dangerously low blood sugar and metabolic crises resulting in serious damage affecting the brain, liver, heart, eyes, muscle, and possibly death. Early detection and treatment can prevent most or all of these consequences. Disorders are:

- Carnitine uptake deficiency
- 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

GAL **Galactosemia;** deficiency in one of three enzymes that help convert galactose into glucose. Our screening targets the most common, classic form of the disorder. If untreated, an affected baby may develop jaundice, vomiting, lethargy, hepatosplenomegaly, cataracts and failure to thrive. Also, the condition can lead to liver failure, sepsis and death. A galactose-free diet with strict avoidance of lactose (milk sugar) and lactose containing foods prevents death, and assists normal growth and development.

HB **Hemoglobinopathies:**

SCD **Sickle cell disease;** a condition marked by a tendency for the blood cells to take on a sickle shape due to an abnormal structure of the hemoglobin molecule. The altered shape results in anemia due to shortened life span of the blood cells and impedes circulation, especially in capillaries. Children with sickle cell disease are highly susceptible to bacterial infections and rapid pooling of blood in their spleens. Either can rapidly lead to death. Treatment consists of regular doses of penicillin to prevent infection and training parents to recognize splenic crisis. Proper treatment dramatically reduces infections and death.

Other **Significant hemoglobinopathies;** hemoglobin abnormalities, other than sickle cell disease, that have significant clinical consequences (for example, transfusion dependent thalassemia). These conditions generally don't require immediate treatment but early identification allows families to adjust to the diagnosis, anticipate the medical needs, and begin early treatment plans as necessary.

OA **Organic acid disorders;** disorders of metabolism characterized by the accumulation of non-amino organic acids and toxic intermediates. This may lead to metabolic crisis with increases in acid and ammonia in the blood, and dangerously low blood sugar resulting in severe nerve and physical damage and possibly death. Early detection and treatment can prevent most or all of these consequences. Disorders are:

- 3-OH 3-CH3 glutaric aciduria (HMG)
- Beta-ketothiolase deficiency (BKT)
- Glutaric acidemia type I (GA 1)
- Isovaleric acidemia (IVA)
- Methylmalonic acidemia (Cbl A, B)

- Methylmalonic acidemia (mutase deficiency) (MUT)
- Multiple carboxylase deficiency (MCD)
- Propionic acidemia (PROP)

Table I: Births by County of Occurrence – Infants Detected by County of Residence

COUNTY	2012 BIRTHS	2012 INFANTS DETECTED									ALL INFANTS
		AA	BIO	CAH	CH	CF	FAO	GAL	HB	OA	
Adams	538	0	0	0	1	0	0	0	0	0	1
Asotin	1	0	0	0	0	0	0	0	0	0	0
Benton	4,315	0	0	0	2	0	0	0	0	0	2
Chelan	1,445	0	0	0	0	1	0	0	0	0	1
Clallam	602	0	0	0	1	0	0	0	0	0	1
Clark	5,229	1	0	0	4	1	0	1	1	0	8
Columbia	0	0	0	0	0	0	0	0	0	0	0
Cowlitz	1,036	0	0	0	0	1	0	0	0	0	1
Douglas	5	0	0	0	0	0	0	0	0	0	0
Ferry	2	0	0	0	0	0	0	0	0	0	0
Franklin	418	0	0	0	2	1	0	0	0	0	3
Garfield	0	0	0	0	0	0	0	0	0	0	0
Grant	1,229	0	0	1	0	1	0	0	0	0	2
Grays Harbor	581	1	0	0	1	0	0	1	0	0	3
Island[a]	198	0	0	0	1	0	0	0	0	1	2
Jefferson	102	0	0	0	0	0	0	0	0	0	0
King	29,325	4	1	4	63	2	3	1	10	1	89
Kitsap[a]	2,001	0	0	0	0	1	0	0	0	0	1
Kittitas	369	0	0	0	0	0	0	0	0	0	0
Klickitat	34	0	0	0	0	0	0	0	0	0	0
Lewis	711	0	0	0	0	1	0	0	0	0	1
Lincoln	3	0	0	0	0	0	0	0	0	0	0
Mason	283	0	0	0	0	0	0	0	0	0	0
Okanogan	496	0	0	0	0	0	0	0	0	0	0
Pacific	3	0	0	0	0	0	1	0	0	0	1
Pend Oreille	95	0	0	0	0	0	0	0	0	0	0
Pierce	11,943	0	0	3	10	4	1	6	6	1	31
San Juan	2	0	0	0	0	0	0	0	0	0	0
Skagit	1,643	0	0	1	2	0	1	0	0	0	4
Skamania	1	0	0	0	0	0	0	0	0	0	0
Snohomish	6,025	1	0	0	10	2	0	3	5	0	21
Spokane	6,879	1	0	0	10	0	0	3	1	1	16
Stevens	290	0	0	0	1	0	0	0	0	0	1
Thurston	2,974	0	0	0	2	0	0	0	0	0	2
Wahkiakum	0	0	0	0	0	0	0	0	0	0	0
Walla Walla	869	0	0	0	2	0	0	1	0	0	3
Whatcom	2,160	1	1	0	0	0	0	0	1	0	3
Whitman	498	0	0	0	1	0	0	0	0	0	1
Yakima	3,875	2	1	1	4	1	1	1	0	0	11
TOTAL[a]	**86,180**	**11**	**3**	**10**	**117**	**16**	**7**	**17**	**24**	**4**	**209**

[a] Excludes infants born in military hospitals that do not participate in the Newborn Screening Program (364 born at Oak Harbor Naval Hospital and 815 born at Bremerton Naval Hospital). Total excluded =1,179.

Table II: Births and Infants Detected by Infant's Race

INFANTS RACE[a]	2012 BIRTHS	2012 INFANTS DETECTED[a]									ALL INFANTS
		A.A.	BIO	CAH	CH	CF	FAO	GAL	HB	O.A.	
White	56,286	11	2	6	71	14	6	15	1	4	**130**
African American	6,419	0	0	1	4	1	0	0	9	0	**15**
Asian	10,844	0	0	3	29	0	0	0	7	0	**39**
Native American	2,536	0	0	0	2	0	0	1	0	0	**3**
Other[b]	10,095	0	1	0	11	1	1	1	7	0	**22**
TOTAL[c]	**86,180**	**11**	**3**	**10**	**117**	**16**	**7**	**17**	**24**	**4**	**209**
Hispanic[d]	**19,429**	2	1	1	13	1	1	1	0	0	**20**

[a] The infant's race for 2012 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] Reflects Hispanic race and multiracial (more than one race designation on the screening form).

[c] Excludes infants born in military hospitals that do not participate in the Newborn Screening Program (364 born at Oak Harbor Naval Hospital and 815 born at Bremerton Naval Hospital). Total excluded =1,179.

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

Newborn Screening Follow-Up Procedures

All specimens that are determined to be presumptive positive through the Newborn Screening Program are followed up immediately through direct telephone contact with the child's physician. This is to ensure that diagnostic testing and treatment, if indicated, is initiated as quickly as possible. Following a definitive diagnosis, a long-term, disease-specific medical management program is implemented as follows:

Phenylketonuria (PKU) - Children are seen monthly in Seattle and every other month in Spokane by the Department of Health (DOH) subsidized University of Washington Phenylketonuria Clinic. An interdisciplinary team consisting of a pediatric metabolic physician, nutritionists, social worker, and genetic counselor work closely with each family to optimize the child's dietary compliance through intensive education and support services. Periodic neuropsychological assessments are also provided for all patients to monitor cognitive and emotional development. For adults with PKU, consultative, support and nutritionist management services are provided at the University of Washington Division of Metabolism and Nutrition. Critical reproductive counseling and maternity services for women at risk of fetal damage due to Maternal PKU are also available. Other program components include a summer camp and Science Night through the private, non-profit PKU Action Group.

Galactosemia, Biotinidase deficiency, Amino Acid, Organic Acid, & Fatty Acid Oxidation Disorders- All children with these disorders are seen periodically as needed by the DOH subsidized University of Washington or Seattle Children's Biochemical Genetics Clinics in Seattle or Mary Bridge Children's Hospital in Tacoma. There are quarterly satellite clinics held in Spokane. Like PKU, an interdisciplinary team; pediatric metabolic physician, nurses, nutritionists, and genetic counselor work closely with each family to optimize the child's compliance with the specific treatment plan through intensive education and support services.

Congenital Hypothyroidism (CH) - Thyroid hormone therapy is monitored by the child's primary health care provider and/or pediatric endocrinologist. The DOH subsidized Congenital Hypothyroidism Developmental Evaluation Clinic at the University of Washington provides developmental, neuropsychological and occupational therapy assessments for affected children.

Congenital Adrenal Hyperplasia (CAH) - All children are seen for a diagnostic work-up by a pediatric endocrinologist to establish appropriate steroid hormone therapy. Long-term management is monitored by the child's primary health care provider and/or pediatric endocrinologist. Affected females with genital abnormalities related to the disorder are referred for surgical consultation.

Cystic Fibrosis (CF) – All children with cystic fibrosis are seen periodically as needed by one of the four regional CF Foundation accredited Clinics – Children's Hospital (Seattle), Mary Bridge (Tacoma), Deaconess Hospital (Spokane), or Oregon Health Sciences University (Portland). Like PKU, an interdisciplinary team consisting of a pediatric pulmonologist, nutritionist, social worker, and nurse work closely with each family to optimize the child's growth and minimize medical complications of the condition, particularly lung disease.

Sickle Cell Disease (SCD) - Affected children receive prophylactic penicillin and folic acid. Long-term management is provided by a pediatric hematologist or an interdisciplinary team consisting of a pediatric hematologist, nurse, social worker and genetic counselor at a DOH subsidized Comprehensive Sickle Cell Clinic – Children's Hospital Odessa Brown Center (Seattle) or Mary Bridge Children's Center (Tacoma). The clinic staff works closely with each family to optimize the child's health and well-being through intensive education and support services. Periodic assessments are also provided for all patients to monitor cognitive and

emotional development. Other sickle cell disease program components include a summer camp and other educational and support activities through the DOH supported Northwest Sickle Cell Collaborative.

Table III: Follow-Up Status of Infants Detected (Severe Disease)

FOLLOW-UP	2012 INFANTS DETECTED									ALL INFANTS
	A.A.	BIO	CAH	CH	CF	FAO	GAL	SCD	O.A.	
Followed by medical specialist – (i.e., pediatric endocrinologist, hematologist, or comprehensive clinic	7	1	9	53	16	4	1	7	4	102
Followed by primary care provider, with some consultation from specialist	0	0	0	6	0	0	0	4	0	10
Expired or Lost to Follow-up	1[a]	0	0	0	0	1[b]	0	1[c]	0	3
TOTAL	**8**	**1**	**9**	**59**	**16**	**5**	**1**	**12**	**4**	**115**

[a] Infant with an atypical form of an amino acid disorder – parents non-compliant with recommendations for continued monitoring

[b] Infant died at two days of age from multiple organ failure; unclear whether death was a result of condition detected.

[c] Infant moved to Louisiana shortly after diagnosis

Table IV: Age at which Treatment Began for Infants Detected (Severe Disease)

DISORDER	NUMBER OF INFANTS	AGE TREATMENT BEGAN (DAYS)	
		AVERAGE	RANGE
A.A.	6[a]	8	6 – 10
BIO	1	17	n/a
CAH	7[b]	7	2 – 12
CH	59	18	3 – 71
CF	16	29	11 – 79
FAO	4[c]	12	11 – 14
GAL	1	11	n/a
SCD	11[d]	30	9 – 45
O.A.	3[e]	35	14 – 47

[a] Excludes two infants; one treated on day one due to known older affected sib, and one infant indicated in the footnote below Table III above whose final diagnosis was not fully established

[b] Excludes two very atypical cases that took months to resolve

[c] Excludes infant who died of multiple organ failure as indicated in the footnote below Table III above

[d] Excludes baby who moved to Louisiana – we eventually found out that penicillin was initiated at 147 days of age

[e] Excludes one baby who was diagnosed prenatally

Screening Costs 2012

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 $8.40 per birth was collected during this period to support specialty clinic care for infants diagnosed through newborn screening.

APPENDIX A: Newborn Hemoglobin Screening – Explanation and Definitions of Phenotypes Found

Hemoglobins are perhaps the most complex of the conditions detected by newborn screening. More than a dozen genes are involved in hemoglobin production and over 700 abnormalities have been described by researchers and clinicians. Also, a variety of combinations are possible for any individual. Many of the abnormalities are rare and most have no clinical implications. A primary objective of our program is to identify those infants with sickle cell disease because these infants will suffer far less illness and death if they are promptly entered into a comprehensive health care program that includes prophylactic treatment with penicillin.

PHENOTYPE	MOST LIKELY GENOTYPE/CLINICAL IMPLICATIONS
FSS	Homozygous for hemoglobin S. Results in sickle cell anemia, a severe form of sickle cell disease.
FS─	Hemoglobin S in combination with β-thalassemia[a] major. A severe form of sickle cell disease.
FSC	Hemoglobin S in combination with hemoglobin C. Results in sickle C disease, a moderate to severe form of sickle cell disease.
F-Only	β-thalassemia[a] major. A severe hemolytic anemia requiring regular blood transfusions.
FE─	Hemoglobin E in combination with β-thalassemia[a] major. A moderate to severe hemolytic anemia.
FSE	Hemoglobin S in combination with hemoglobin E. Results in sickle E disease, a moderate form of sickle cell disease.
FAA + High Bart's	High level of hemoglobin Bart's indicative of multiple α-thalassemia[b] genes. Likelihood of Hemoglobin H disease, a moderate to severe hemolytic anemia.
FCC	Homozygous for hemoglobin C. A mild to moderate hemolytic anemia.
FCA	Hemoglobin C in combination with β-thalassemia[a] minor. A mild to moderate hemolytic anemia.
FAE+CS+ High Bart's	Hemoglobin E trait with combination with two hemoglobins (Constant Spring and Bart's) indicative of multiple α-thalassemia[b] genes. Likelihood of Hemoglobin H/Constant Spring disease, a moderate to severe hemolytic anemia (might be somewhat ameliorated by presence of hemoglobin E).

[a] Decreased production of β globin chains; benign to severe anemia. Significant interaction with other β chain variants such as hemoglobin S, E, and D.
[b] Decreased production of α globin chains; benign to severe anemia depending on how many of the four α globin genes are affected.

Appendix A, continued

PHENOTYPE	MOST LIKELY GENOTYPE/CLINICAL IMPLICATIONS
FEE	Homozygous for hemoglobin E. Mild anemia.
FEE+Bart's	Homozygous hemoglobin E in combination with α-thalassemia[b]. Mild anemia.
FA+CS+Bart's	Two hemoglobins (Constant Spring and Bart's) indicative of α-thalassemia [b] genes. Mild anemia.
FAE+CS+Bart's	Hemoglobin E trait in combination with two hemoglobins (Constant Spring and Bart's) indicative of α-thalassemia [b] genes. Mild anemia.
FAS+Bart's	Hemoglobin S trait in combination with α-thalassemia [b]. No clinical implications for S trait (see FAS, below). Benign to mild anemia.
FAE+Bart's	Hemoglobin E trait in combination with α-thalassemia [b]. No clinical implications for E trait (see FAE, below). Benign to mild anemia.
FAA+Bart's	α-thalassemia [b]. Benign to mild anemia.
FA+Var+Bart's	An unidentified hemoglobin variant trait and α-thalassemia [b]. Benign to mild anemia.
FAS+Var	Hemoglobin S and unidentified variant trait. No clinical implications for S trait; clinical effects from variant trait unlikely. Family may be at risk for sickle cell disease.
FAE+Var	Hemoglobin E and unidentified variant trait. No clinical implications for E trait; clinical effects from variant trait unlikely.
FAS	Hemoglobin S trait. No clinical implications for child. Family may be at risk for sickle cell disease.
FAE	Hemoglobin E trait. No clinical implications for child. Family may be at risk for hemoglobin E/β-thalassemia[a], a significant hemoglobin disease.
FAC	Hemoglobin C trait. No clinical implications for child. Family may be at risk for homozygous C, a mild to moderate hemolytic anemia or hemoglobin SC, a moderate to severe form of sickle cell disease.
FAD	Hemoglobin D trait. No clinical implications for child. Homozygous state is benign; however, family may be at risk for hemoglobin SD, a moderate to severe form of sickle cell disease.
FA+Var	Unidentified variant trait. Clinical effects unlikely.

[a] Decreased production of β globin chains; benign to severe anemia. Significant interaction with other β chain variants such as hemoglobin S, E, and D.
[b] Decreased production of α globin chains; benign to severe anemia depending on how many of the four α globin genes are affected.

APPENDIX B: Newborn Hemoglobin Screening – Infants Detected by Phenotype and Race/Ethnicity

January through December 2012; Number of Infants = 86,180

PHENOTYPE	TOTAL	WHITE	BLACK	ASIAN	NAT. AM.	OTHER[a]	HISPANIC[b]
FSS	4	0	4	0	0	0	0
FS-	1	0	0	0	0	1	0
FSC	3	0	3	0	0	0	0
F-Only	1	0	0	0	0	1	0
FE-	2	0	0	2	0	0	0
FSE	4	0	0	0	0	4	0
FAA+High Bart's	6	1	0	3	0	2	0
FCC	1	0	1	0	0	0	0
FCA	1	0	1	0	0	0	0
FAE+CS+ High Bart's	1	0	0	1	0	0	0
FEE	11	0	0	7	0	4	0
FEE+Barts	1	0	0	0	0	1	0
FA+CS+Bart's	19	0	1	10	0	8	2
FAE+CS+Bart's	4	0	0	4	0	0	0
FAS+Bart's	1	0	1	0	0	0	0
FAE+Bart's	8	0	3	4	0	1	1
FAA+ Bart's	174	7	21	73	0	73	15
FA+Var+Bart's	1	0	0	1	0	0	0
FAS+Var	1	0	0	0	0	1	0
FAE+Var	2	0	0	0	0	2	0
FAS	440	44	163	5	5	223	103
FAE	261	9	7	127	0	118	18
FAC	116	11	43	0	0	62	18
FAD	26	9	0	4	1	12	6
FA+Var	179	72	7	13	3	84	63
TOTAL	**1268**	**153**	255	**254**	**9**	**176**	**226**

[a] Includes multi-racial (more than one race designation on the screening form) or unknown (no designation made).
[b] Hispanics can be of any race; they are included in figures to the left.

APPENDIX C: Infants Detected by Newborn Screening 2008 - 2011

Disorder	2008*	2009	2010	2011
Amino Acid Disorders:				
• Phenylketonuria (PKU)	6	6	7	6
• Maple Syrup Urine Disease (MSUD)	2	0	0	1
• Citrullinemia (CIT)	0	0	0	1
• Tyrosinemia type 1 (TYR-1)	1	0	0	1
• Homocystinuria (HCY)	0	0	0	1
Biotinidase Deficiency (BIO)	0	0	0	2
Congenital Adrenal Hyperplasia (CAH)	8	4	3	11
Congenital Hypothyroidism (CH)	84	73	77	104
Cystic Fibrosis (CF)	16	14	23	17
Fatty Acid Oxidation Disorders:				
• Medium chain acyl-CoA dehydrogenase (MCAD) deficiency	5	4	7	5
• Very Long Chain acyl-CoA dehydrogenase (VLCAD) deficiency	0	2	3	1
• Carnitine uptake deficiency (CUD)	0	0	1	0
Galactosemia (GALT)	2	1	3	11
Hemoglobinopathies (Hb)				
• Sickle Cell Disease	9	7	9	7
• Hemoglobin E-beta thalassemia	1	1	1	3
• Hemoglobin H disease	6	5	6	4
• Other moderate to severe hemoglobinopathies	3	4	1	1
• Mild hemoglobin conditions & traits (excluded in summaries below)	1,415	1,158	1,199	1,130
Organic Acid Disorders:	0			
• Glutaric acidemia type 1 (GA-1)		3	0	0
• Methylmalonic acidemia (MUT)		0	1	2
• Propionic acidemia (PROP)		0	0	2
• Beta-ketothiolase deficiency (BKT)		1	1	0
Non-panel Disorders:	0			
• 3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency		1	0	1
• Glutaric acidemia type II (GA-II)		0	1	0
• Methylmalonic acidemia Cbl C		0	1	3
• 2-methylbutyryl-CoA dehydrogenase (2-MBD) deficiency		0	0	3
• 3-methylglutaconic aciduria (3-MGA)		0	0	1
Total Infants Detected** (excludes mild hemoglobin conditions & traits)	143	126	145	188
Total Infants Screened	86,058	84,871	83,086	84,918
Overall frequency (excludes mild hemoglobin conditions and traits)	1 in 601	1 in 674	1 in 573	1 in 452

*expanded MS/MS screening began mid-year

APPENDIX D: History of Conditions Added to Washington's Newborn Screening Panel

In 1963 phenylketonuria (PKU) screening was offered through the state's Public Health Laboratory as a voluntary service. In 1967 the state legislature passed a statute that directed the Department of Health to "...promote screening tests of all newborn infants..." for PKU. Despite these efforts, however, many infants were not being screened and the quality of screening was highly variable between sites. As a result, some affected infants were not detected in time and suffered the irreversible mental and physical damage caused by PKU. This led the legislature to adopt revisions to the statute in 1976 to require screening of all infants unless the parents refused on religious grounds. The legislation also gave authority to the Board of Health to determine which other disorders should be included in the screening and to adopt rules to achieve the intent of the law. The table below is a timeline of additional disorders added to the panel:

YEAR	DISORDERS ADDED
1963	PKU - test available - voluntary
1967	- statute adopted, promotes screening
1976	- statute revised, mandates screening & BOH given authority to add conditions;
	adopt rules to carry out intent of statute
1978	Congenital hypothyroidism (CH)
1984	Congenital adrenal hyperplasia (CAH)
1991	Hemoglobinopathies (Hb)
2004	Biotinidase deficiency (BIO)
	Galactosemia (GALT)
	Homocystinuria (HCY)
	Maple syrup urine disease (MSUD)
	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
2006	Cystic fibrosis (CF)
2008	Amino acid (AA) disorders:
	Argininosuccinic acidemia (ASA)
	Citrullinemia (CIT)
	Tyrosinemia type 1 (TYR-1)
	Fatty acid oxidation (FAO) disorders:
	Carnitine uptake deficiency (CUD)
	Long-chain L-3-hydroxy acyl-CoA dehydrogenase (LCHAD) deficiency
	Trifunctional protein (TFP) deficiency
	Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency
	Organic acid disorders (OA)
	3-hydroxy-3-methylglutaric aciduria (Hydroxymethylglutaric aciduria - HMG)
	Beta-ketothiolase (BKT) deficiency
	Glutaric acidemia type 1 (GA-1)
	Isovaleric acidemia (IVA)
	Methylmalonic acidemia - mutase (MUT) deficiency
	Methylmalonic acidemia – cobalamin A, B (Cbl A,B) deficiency
	Multiple carboxylase deficiency (MCD)
	Propionic acidemia (PROP)