PROPOSED RULE MAKING

CR-102 (June 2024)
(Implements RCW 34.05.320)
Do **NOT** use for expedited rule making

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STATE OF WASHINGTON
FILED

DATE: September 30, 2025
TIME: 10:09 AM

WSR 25-20-091

Agency: Washington State Board of Health

☒ **Original Notice**
☐ **Supplemental Notice to WSR**
☐ **Continuance of WSR**

☒ **Preproposal Statement of Inquiry was filed as WSR** WSR 22-05-012 and WSR 23-24-016 **; or**
☐ **Expedited Rule Making--Proposed notice was filed as WSR _____; or**
☐ **Proposal is exempt under RCW 34.05.310(4) or 34.05.330(1); or**
☐ **Proposal is exempt under RCW _____.**

Title of rule and other identifying information: (describe subject) Chapter 246-650 WAC, Newborn Screening. The Washington State Board of Health (Board) is proposing to amend the newborn screening (NBS) rules to add three conditions to the list of conditions that the Department of Health's Newborn Screening Program screens all babies for in Washington State. These conditions are Ornithine Transcarbamylase Deficiency (OTCD), Guanidinoacetate Methyltransferase (GAMT) Deficiency, and Arginase 1 Deficiency (ARG1-D).

Hearing location(s):

Date:	**Time:**	**Location:** (be specific)	**Comment:**
11/19/2025	11:00 am	Washington State Department of Health 111 Israel Road S.E. Tumwater, WA 98501 Building: Town Center Two (TC2, Rooms 166 & 167) Register to participate via Zoom here: https://us02web.zoom.us/webinar/register/WN_DlI0Jo2yQUe1KBgX_fIccw	The rules hearing will be hybrid. Individuals may attend either virtually or in-person.

Date of intended adoption: 11/19/2025 (Note: This is **NOT** the **effective** date)

Submit written comments to:
Name Molly Dinardo
Address PO Box 47990, Olympia, WA 98504-7990
Email molly.dinardo@sboh.wa.gov
Fax N/A
Other https://airtable.com/appUWFVMQBM3iV6S3/pagbZb1PlqDm5JhVf/form
Beginning (date and time) Date and time of filing
By (date and time) October 27, 2025 at 11:59 p.m.

Assistance for persons with disabilities:
Contact Molly Dinardo
Phone 360-236-4110
Fax N/A
TTY 711
Email molly.dinardo@sboh.wa.gov
Other wsboh@sboh.wa.gov
By (date) 11/05/2025

Purpose of the proposal and its anticipated effects, including any changes in existing rules: The purpose of this proposal is to amend chapter 246-650 WAC to add OCTD, GAMT Deficiency, and ARG1-D to the Washington State newborn screening panel.

The Board formed Technical Advisory Committees (TACs) in 2021 and 2023 to review these conditions. After reviewing evidence, hearing from families, and consulting experts, both TACs recommended adding the conditions to the screening panel. The Board considered these recommendations at its meetings in October 2021 and October 2023 and voted to proceed with rulemaking to amend chapter 246-650 WAC.

The Department of Health (Department) operates the newborn screening program, which, screens all babies for rare but treatable health conditions using a dried blood sample. State law (RCW 70.83.050) requires that the Board create rules for newborn screening. These rules are found in chapter 246-650 WAC. To implement screening for these conditions, the Board must amend WAC 246-650-010 and WAC 246-650-020.

In addition, adding OTCD, GAMT deficiency, and ARG1-D to the newborn screening panel requires the Department to request an increase in the state's newborn screening fee. This fee, collected for each infant in Washington, covers the costs of laboratory screening, follow-up services, and coordination with specialty care. Additionally, the Washington State Health Care Authority (HCA) covers more than 40% of births in the state. Therefore, it must also seek approval to adjust Medicaid managed care rates to cover the increased fee. After multiple requests to the Legislature, the Department and HCA secured funding during the 2025 session to implement screening, which is expected to begin in early 2026.

Reasons supporting proposal: Population-based newborn screening is the most effective way to detect OTCD, GAMT, and ARG1-D early, when treatment can prevent severe illness, lifelong disability, or death. Without early screening, most affected infants are not diagnosed until after serious and irreversible health problems develop.

Alternatives such as waiting for symptoms or testing only infants with a known family history are not effective, as they miss most cases and delay treatment. Universal screening ensures timely diagnosis and care, giving babies the best chance for healthy development.

If this rule is not adopted, infants with these conditions will continue to face delayed diagnoses, preventable complications, and in some cases, loss of life.

Statutory authority for adoption: RCW 70.83.050

Statute being implemented: RCW 70.83

Is rule necessary because of a:

Federal Law?	☐ Yes	☒ No
Federal Court Decision?	☐ Yes	☒ No
State Court Decision?	☐ Yes	☒ No

If yes, CITATION:

Agency comments or recommendations, if any, as to statutory language, implementation, enforcement, and fiscal matters: None

Name of proponent: (person or organization) Washington State Board of Health
Type of proponent: ☐ Private. ☐ Public. ☒ Governmental.

Name of agency personnel responsible for:

	Name	Office Location	Phone
Drafting	Molly Dinardo	111 Israel Road SE Tumwater WA 98501	360-236-4110
Implementation	Megan McCrillis	1610 NE 150th Street, P.O. Box 55729, Shoreline, WA 98155	206-418-5410
Enforcement	Megan McCrillis	1610 NE 150th Street, P.O. Box 55729, Shoreline, WA 98155	206-418-5410

Is a school district fiscal impact statement required under RCW 28A.305.135? ☐ Yes ☒ No
If yes, insert statement here:

The public may obtain a copy of the school district fiscal impact statement by contacting:
Name
Address
Phone

Fax
TTY
Email
Other

Is a cost-benefit analysis required under RCW 34.05.328?

☒ Yes: A preliminary cost-benefit analysis may be obtained by contacting:

Name Molly Dinardo
Address P.O. Box 47822, Olympia, WA 98504-7822

Phone 360-236-4110
Fax N/A
TTY 711
Email molly.dinardo@sboh.wa.gov
Other

☐ No: Please explain:

Regulatory Fairness Act and Small Business Economic Impact Statement

Note: The Governor's Office for Regulatory Innovation and Assistance (ORIA) provides support in completing this part.

(1) Identification of exemptions:

This rule proposal, or portions of the proposal, **may be exempt** from requirements of the Regulatory Fairness Act (see chapter 19.85 RCW). For additional information on exemptions, consult the exemption guide published by ORIA. Please check the box for any applicable exemption(s):

☐ This rule proposal, or portions of the proposal, is exempt under RCW 19.85.061 because this rule making is being adopted solely to conform and/or comply with federal statute or regulations. Please cite the specific federal statute or regulation this rule is being adopted to conform or comply with, and describe the consequences to the state if the rule is not adopted.
Citation and description:

☐ This rule proposal, or portions of the proposal, is exempt because the agency has completed the pilot rule process defined by RCW 34.05.313 before filing the notice of this proposed rule.

☐ This rule proposal, or portions of the proposal, is exempt under the provisions of RCW 15.65.570(2) because it was adopted by a referendum.

☒ This rule proposal, or portions of the proposal, is exempt under RCW 19.85.025(3). Check all that apply:

- ☐ RCW 34.05.310 (4)(b) (Internal government operations)
- ☐ RCW 34.05.310 (4)(c) (Incorporation by reference)
- ☒ RCW 34.05.310 (4)(d) (Correct or clarify language)
- ☐ RCW 34.05.310 (4)(e) (Dictated by statute)
- ☐ RCW 34.05.310 (4)(f) (Set or adjust fees)
- ☐ RCW 34.05.310 (4)(g) ((i) Relating to agency hearings; or (ii) process requirements for applying to an agency for a license or permit)

☐ This rule proposal, or portions of the proposal, is exempt under RCW 19.85.025(4). (Does not affect small businesses).

☐ This rule proposal, or portions of the proposal, is exempt under RCW _____.

Explanation of how the above exemption(s) applies to the proposed rule: Proposed changes in WAC 246-650-010 add definitions for OTCD, GAMT and ARG1-D. These proposed changes are exempt from the SBEIS under RCW 34.05.310(4)(d) because they clarify the meaning of terms used in the rules. Defining these conditions is not setting screening standards.

(2) Scope of exemptions: *Check one.*

☐ The rule proposal: Is fully exempt. *(Skip section 3.)* Exemptions identified above apply to all portions of the rule proposal.

☒ The rule proposal: Is partially exempt. *(Complete section 3.)* The exemptions identified above apply to portions of the rule proposal, but less than the entire rule proposal. Provide details here (consider using this template from ORIA):

☐ The rule proposal: Is not exempt. *(Complete section 3.)* No exemptions were identified above.

(3) Small business economic impact statement: *Complete this section if any portion is not exempt.*

If any portion of the proposed rule is **not exempt**, does it impose more-than-minor costs (as defined by RCW 19.85.020(2)) on businesses?

☒ No Briefly summarize the agency's minor cost analysis and how the agency determined the proposed rule did not impose more-than-minor costs.

A brief description of the proposed rule including the current situation/rule, followed by the history of the issue and why the proposed rule is needed. A description of the probable compliance requirements and the kinds of professional services that a small business is likely to need in order to comply with the proposed rule.
The Washington State Board of Health (board) in collaboration with the Department of Health (department) is proposing amendments to the newborn screening requirements in WAC 246-650-010 and WAC 246-650-020. The proposed rule expands the Washington State newborn screening (NBS) panel of required tests by adding three new conditions: Ornithine Transcarbamylase Deficiency (OTCD), Guanidinoacetate Methyltransferase (GAMT) Deficiency, and Arginase 1 Deficiency (ARG1-D). To implement screening for these conditions, the board must amend WAC 246-650-010 and WAC 246-650-020. Early detection of OTCD, GAMT deficiency, and ARG1-D is critical for saving lives and improving long-term health outcomes for affected infants and their families.

The NBS program, operated by the Washington State Department of Health (department) , tests all babies for rare but treatable health conditions using a dried blood sample. Washington State law (RCW 70.83.050) requires that the board create rules for newborn screening. These rules are found in chapter 246-650 WAC. Specifically, WAC 246-650-010 and WAC 246-650-020 define and list the conditions that the department must screen all babies for after birth.

Anyone may petition the board to review a condition for possible inclusion in the NBS panel, including members of the public, department staff, and board members. To evaluate candidate conditions, the board convenes a Technical Advisory Committee (TAC), which reviews evidence against guiding principles and established criteria. The TAC provides findings and recommendations for board consideration.

In 2021, the board formed a Technical Advisory Committee (TAC) to review OTCD following a petition from a family whose child had passed away from the condition. In 2023, the board formed another TAC to review GAMT Deficiency and ARG1-D after two parent advocates submitted petitions. After reviewing evidence, hearing from families, and consulting experts, both TACs recommended adding the conditions to the screening panel. The board considered these recommendations at its meetings in October 2021 and October 2023 and voted to proceed with rulemaking to amend chapter 246-650 WAC. Through this rulemaking, the board proposes amendments to chapter 246-650 WAC to support the addition of three new conditions to Washington's newborn screening panel: OTCD, GAMT deficiency, and ARG1-D. These amendments include adding formal definitions for each of the three conditions and incorporating them into the list of newborn screening tests performed by the Department of Health.

In addition, the Board is proposing minor housekeeping changes. These include updating the definition of amino acid disorders and reclassifying relevant conditions under a new definition, "urea cycle disorders" as OTCD, ARG1-D and citrullinemia type I (CIT) are specific types or urea cycle disorders. The rulemaking also removes outdated language related to prior screening requirements that sunset in 2020 and are no longer applicable.

Adding OTCD, GAMT deficiency, and ARG1-D to the newborn screening (NBS) panel requires an increase in the NBS fee. This fee is not adopted in rule, and falls under the department's fee authority established in RCW 43.70.250(2), which requires legislative approval to increase spending authority for department programs, permit fees, etc.
The department charges a one-time screening fee that covers all required newborn screens in Washington State, including repeat or follow-up screens when medically necessary. This fee, collected for each tested infant in Washington, covers the costs of laboratory screening, follow-up services, and coordination with specialty care.
The department bills the screening fee to the facility that collects the infant's initial specimen. This is typically the birth hospital but may also include a clinic, birth center, or laboratory. Facilities can usually recover the cost through insurance reimbursement or patient billing. Medical facilities may charge their own administrative fees for collecting or processing specimens, which are separate from the department's screening fee.

For specimens collected in an at home setting, the department bills the client's insurance directly when the specimen is accompanied by the insurance information. If the patient is uninsured, they must pay the screening fee out of pocket via check or money order.

To implement screening for these additional conditions, the department secured increased spending authority from the Legislature during the 2025 legislative session. Beginning January 1, 2026, the screening fee will increase by $4.02 to cover the cost of testing for OTCD, GAMT deficiency, and ARG1-D. This increase is expected to be covered by public or private insurance, or by families directly.

The proposed rule does not impose any new compliance requirements on small businesses. It applies only to the Department of Health's administration of the NBS Program and updates the list of conditions for which newborns are screened. Healthcare providers, birthing centers, midwives, and hospitals already participate in the program, and have for decades, by collecting blood samples and sending them to the state laboratory—practices that will remain unchanged. The same blood sample is used to test for these new conditions. There are no new reporting, procedural, or equipment requirements for these entities. Because the proposed rule does not alter existing procedures for facilities, no additional professional services are anticipated to be needed by small businesses to comply.

Identification and summary of which businesses are required to comply with the proposed rule using the North American Industry Classification System (NAICS).

SBEIS Table 1. Summary of Businesses Required to comply to the Proposed Rule

NAICS Code (4, 5 or 6 digit)	NAICS Business Description	Number of businesses in Washington State	Minor Cost Threshold
524114	Direct Health and Medical Insurance Carriers[1]	61	$111,059.38
621399	Offices of All Other Miscellaneous Health Practitioners[2]	5,023	$927.25

Analysis of probable costs of businesses in the industry to comply to the proposed rule and includes the cost of equipment, supplies, labor, professional services, and administrative costs. The analysis considers if compliance with the proposed rule will cause businesses in the industry to lose sales or revenue.

The following SBEIS Table 2 identifies rule sections or portions of rule sections that have been determined exempt based on the exemptions.

SBEIS Table 2. Exempted Sections

WAC Section and Title	Description of Proposed Changes	Rationale for Exemption Determination
WAC 246-650-010 – Definitions	Adding definitions for OTCD, GAMT deficiency, and ARG1-D; amending the definition of amino acid disorders; and adding a definition for urea cycle disorders.	Exempt from SBEIS under RCW 34.05.310 (4)(d) because the proposed changes clarify the meaning of terms used in the rules by add new definitions for OTCD, GAMT, and ARG1-D. Defining these conditions is not setting screening standards. Tests for screening (required newborn screening tests) are listed under WAC 246-650-020.

WAC 246-650-010 Definitions.

Description: Defines terms that apply throughout the rule chapter. The proposed rule adds definitions for OTCD, GAMT and ARG1-D. These proposed changes are exempt from the SBEIS under RCW 34.05.310(4)(d) because they clarify the meaning of terms used in the rules.Defining these conditions is not setting screening standards. Tests for screening (required newborn screening tests) are listed under WAC 246-650-020. The impacts of adding the new conditions are analyzed below.

WAC 246-650-020 Performance of Screening Tests.

Description: This section of the rule establishes the list of heritable conditions that all babies in Washington must be screened for within 48 hours after birth. It also sets the responsibilities of hospitals and birth care providers in conducting newborn screening, including parental notification, specimen collection, and timely submission. In addition, the rule details the department's newborn screening program duties upon receipt of specimens, including the specific heritable conditions to be tested.

[1] This U.S. industry comprises establishments primarily engaged in initially underwriting (i.e., assuming the risk and assigning premiums) health and medical insurance policies. Group hospitalization plans and HMO establishments that provide health and medical insurance policies without providing health care services are included in this industry.

[2] : This U.S. industry comprises establishments of independent health practitioners. We are using this code to account for midwives with their own practices, that collect the newborn screening blood spot sample, and send to the PHL.

The board is proposing to amend WAC 246-650-020(2)(b) to add three new candidate conditions, OTCD, GAMT, and ARG1-D to the list of conditions on the Washington State mandatory newborn screening panel.

Additionally, the board is proposing to repeal subsection (3) as the requirement for the attending health care provider to notify the department of the date which the test results were disclosed to the parent or guardian expired January 1, 2020.
Cost(s): Total cost of the rule: $4.02 fee increase per baby * 83,000 estimated births annually in Washington = **$333,660.** These costs will be covered by either private insurance, Medicaid, or patients will pay out of pocket.

Summary of all Cost(s):

Approximately 50% of births in Washington are covered by Medicaid; the remaining 50% are assumed to be covered by private insurance. Based on this, the cost impact is estimated as follows:

- Medicaid: $166,830
- Private Insurance: $166,830

To estimate potential cost impacts for providers serving uninsured populations (i.e., not covered by Medicaid or private insurance), we assume an 8% uninsured rate, based on 2024 U.S. Census[3] and CDC data.[4] Accordingly, the estimated cost impact on health providers collecting newborn screening specimens for uninsured patients is: $333,660 * 0.08 = $26,692

SBEIS Table 3. Summary of Section 3 probable cost(s)

WAC Section and Title	Probable Cost(s)
WAC 246-650-020 – Performance of Screening Tests, Addition of OTCD	The cost of screening would be approximately $594,241.00 per year, or $8.03 per baby. Total expenses, including the costs of screening, courier service, and false positive diagnostic testing, would be $633,179.55.
WAC 246-650-020 – Performance of Screening Tests, Addition of GAMT Deficiency	The cost of screening would be approximately $82,008.19 per year, or $0.99 per baby. Total expenses, including the costs of screening and those associated with false positives, would be $84,186.94 per year.
WAC 246-650-020 – Performance of Screening Tests, Addition of ARG1-D	The cost of screening would be approximately $82,008.19 per year, or $0.99 per baby. This includes startup lab costs, laboratory staffing, and supplies. Total expenses, including the costs of screening and those associated with false positives, would be $87,263.32 per year.

Analysis on if the proposed rule may impose more than minor costs for businesses in the industry. Includes a summary of how the costs were calculated.

No, the costs of the proposed rule (annual average per establishment of $2,735) are less than the minor cost threshold ($111,059.38) for direct health and medical insurance carriers.
No, the costs of the proposed rule (annual average per establishment up to $126.50) are less than the minor cost threshold ($927.25) for Offices of All other Miscellaneous Health Practitioners.

Summary of how the costs were calculated
Health insurance carriers
The total cost of the rule is $4.02 per baby * 83,000 estimated births in Washington State (annual) = $333,660
The following assumptions and calculations were used to estimate the potential average annual costs per businesses in this sector to comply with the proposed rule:

- Medicaid covers about fifty percent of the births in Washington state. The other half are assumed to be covered by private insurance.[5] Total annual costs ($333,660) / 2 (half of births) = $166,830 annually.
- Average cost of establishment was calculated for each of the estimated 61 businesses, $166,830 / 61 businesses = $2,735

Offices of miscellaneous health providers – including midwives and other health practitioners offices
This U.S. industry comprises establishments of independent health practitioners. The department and board selected this NAICS code to account for midwives with their own practices, that collect the newborn screening blood spot sample, and send to the public health lab. This NAICS code includes not only midwives' offices but also other health practitioner offices

[3] U.S. Census Bureau Health Insurance Coverage by State: 2023 and 2024. Health Insurance Coverage by State: 2023 and 2024

[4] Centers for Disease Control and Prevention (CDC). National Center for Health Statistics. FastStats Health Insurance Coverage. FastStats - Health Insurance Coverage

[5] Uninsured births were not assumed in the calculated estimate, which likely leads to an overestimate.

that do not collect newborn blood spot specimens (e.g., dental hygienists' offices and herbalists' offices). According to the Midwives Association of Washington State there are approximately 211 licensed midwives practicing in Washington State.[6]
The total cost of the rule: $4.02 fee increase per baby * 83,000 estimated births = $333,660.
The following assumptions and calculations were used to estimate the potential average annual costs per businesses in this sector to comply with the proposed rule:

- Medicaid covers about fifty percent of the births in Washington state. The other half are assumed to be covered by private insurance. Total annual costs ($333,660) / 2 (half of births) = $166,830 annually.
- Assume that 8% of births annually are uninsured.[7] Total costs $333,660 * (0.08) = $26,692
- Average cost of establishment was calculated for each of the estimated 5,023 businesses and the estimated 211 licensed midwife businesses operating in Washington State.
    - $26,692 / 5,023 businesses = $5.32
    - $26,692 / 211 businesses = $126.50

☐ Yes Calculations show the rule proposal likely imposes more-than-minor cost to businesses and a small business economic impact statement is required. Insert the required small business economic impact statement here:

The public may obtain a copy of the small business economic impact statement or the detailed cost calculations by contacting:

Name Molly Dinardo
Address 111 Israel Road SE Tumwater WA 98501
Phone 360-236-4110
Fax N/A
TTY 711
Email molly.dinardo@sboh.wa.gov
Other

Date: 9/29/2025 **Name:** Michelle A. Davis **Title:** Executive Director	**Signature:** 

[6] Midwives Association of Washington State. 2025 Licensed Midwifery in Washington State FAQ. 2025 LM Licensed Midwifery in Washington State Fact Sheet

[7] U.S. Census Bureau Health Insurance Coverage by State: 2023 and 2024. Health Insurance Coverage by State: 2023 and 2024 & Centers for Disease Control and Prevention (CDC). National Center for Health Statistics. FastStats Health Insurance Coverage. FastStats - Health Insurance Coverage

AMENDATORY SECTION (Amending WSR 20-15-032, filed 7/7/20, effective 8/7/20)

WAC 246-650-010 Definitions. The definitions in this section apply throughout this chapter unless the context clearly requires otherwise.

(1) "Amino acid disorders" means ((~~argininosuccinic acidemia (ASA), citrullinemia type I (CIT),~~)) homocystinuria (HCY), maple syrup urine disease (MSUD), phenylketonuria (PKU), and tyrosinemia type I (TYR I), which may cause severe complications including intellectual disability, coma, seizures, and possibly death.

(2) "Board" means the Washington state board of health.

(3) "Biotinidase deficiency" means a deficiency of an enzyme (biotinidase) that facilitates the body's recycling of biotin. The result is biotin deficiency, which if undetected and untreated, may result in severe neurological damage or death.

(4) "Congenital adrenal hyperplasia" means a severe disorder of adrenal steroid metabolism which may result in death of an infant during the neonatal period if undetected and untreated.

(5) "Congenital hypothyroidism" means a disorder of thyroid function during the neonatal period causing impaired mental functioning if undetected and untreated.

(6) "Critical congenital heart disease" means an abnormality in the structure or function of the heart that exists at birth, causes severe, life-threatening symptoms, and requires medical intervention within the first year of life.

(7) "Cystic fibrosis" means a life-shortening disorder caused by mutations in the gene encoding the cystic fibrosis transmembrane conductance regulator (CFTR), a transmembrane protein involved in ion transport. Affected individuals suffer from chronic, progressive pulmonary disease and nutritional deficits. Early detection and enrollment in a comprehensive care system provides improved outcomes and avoids the significant nutritional and growth deficits that are evident when diagnosed later.

(8) "Department" means the Washington state department of health.

(9) "Fatty acid oxidation disorders" means carnitine uptake defect (CUD), long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHADD), medium-chain acyl-CoA dehydrogenase deficiency (MCADD), trifunctional protein deficiency (TFP), and very long-chain acyl-CoA dehydrogenase deficiency (VLCADD). These disorders can lead to hypoglycemia and metabolic crises resulting in serious damage affecting the brain, liver, heart, eyes, muscle, and possibly death.

(10) "Galactosemia" means a deficiency of enzymes that help the body convert the simple sugar galactose into glucose resulting in a buildup of galactose and galactose-1-PO_4 in the blood. If undetected and untreated, accumulated galactose-1-PO_4 may cause significant tissue and organ damage often leading to sepsis and death.

(11) "Guanidinoacetate methyltransferase (GAMT) deficiency" means an inherited condition that affects the body's ability to produce creatine. Without enough creatine, the body cannot use or store energy properly. If undetected and untreated, severe neurological problems can occur.

(12) "Hemoglobinopathies" means a group of hereditary blood disorders caused by genetic alteration of hemoglobin which results in

characteristic clinical and laboratory abnormalities and which leads to developmental impairment or physical disabilities.

((~~(12)~~)) (13) "Newborn" means an infant born in any setting in the state of Washington.

((~~(13)~~)) (14) "Newborn screening specimen/information form" means a form provided by the department for collecting a newborn's dried blood spots and information used to screen for congenital disorders under this chapter. This includes the filter paper portion and associated dried blood spots.

((~~(14)~~)) (15) "Mucopolysaccharidosis I (MPS-I)" means a multisystem disorder caused by mutations in the alpha-L-iduronidase gene in which a lysosomal enzyme is deficient, leading to accumulation of mucopolysaccharides (a type of carbohydrate) and other metabolites. This includes Hurler, Hurler-Scheie, and Scheie syndromes.

((~~(15)~~)) (16) "Organic acid disorders" means 3-OH 3-CH3 glutaric aciduria (HMG), beta-ketothiolase deficiency (BKT), glutaric acidemia type I (GA 1), isovaleric acidemia (IVA), methylmalonic acidemia (CblA,B), methylmalonic acidemia (mutase deficiency) (MUT), multiple carboxylase deficiency (MCD), and propionic acidemia (PROP). These disorders can lead to metabolic crises resulting in severe nerve damage, physical damage, and possibly death.

((~~(16)~~)) (17) "Pompe disease" means a neuromuscular disorder caused by mutations in the acid glucosidase gene which result in reduced or absent activity of the acid alpha glucosidase enzyme.

((~~(17)~~)) (18) "Significant screening test result" means a laboratory test result indicating a suspicion of abnormality and requiring diagnostic evaluation of the involved infant for a specific congenital disorder.

((~~(18)~~)) (19) "Severe combined immunodeficiency (SCID)" means a group of congenital disorders characterized by profound deficiencies in T- and B- lymphocyte function. This results in very low or absent production of the body's primary infection fighting processes that, if left untreated, results in severe recurrent, and often life-threatening infections within the first year of life.

((~~(19)~~)) (20) "Spinal muscular atrophy (SMA)" means a genetic disorder caused by mutations in the survival motor neuron 1 (SMN1) gene, which impairs the function of the survival motor neuron (SMN) protein. This results in the loss of motor neurons and causes progressive atrophy of skeletal muscles.

((~~(20)~~)) (21) "Urea cycle disorders" means argininosuccinic acidemia (ASA), arginase 1 deficiency (ARG1-D), citrullinemia type I (CIT), and ornithine transcarbamylase deficiency (OTCD). These disorders can lead to an elevation of ammonia in blood, which is toxic to the body and can lead to severe nervous system damage.

(22) "X-linked adrenoleukodystrophy (X-ALD)" means a peroxisomal disorder caused by mutations in the ABCD1 gene located on the X chromosome. If untreated this can lead to adrenocortical deficiency, damage to the nerve cells of the brain, paralysis of the lower limbs, mental decline, disability, or death.

AMENDATORY SECTION (Amending WSR 20-15-032, filed 7/7/20, effective 8/7/20)

WAC 246-650-020 Performance of screening tests. (1) Hospitals and other providers of birth and delivery services or neonatal care to infants shall:

(a) Inform parents or guardians, by providing a departmental information pamphlet or by other means, of:

(i) The purpose of screening newborns for congenital disorders;

(ii) Disorders of concern as listed in WAC 246-650-020(2);

(iii) The requirement for newborn screening;

(iv) The legal right of parents or guardians to refuse testing because of religious tenets or practices as specified in RCW 70.83.020; and

(v) The specimen storage, retention and access requirements specified in WAC 246-650-050.

(b) Obtain a blood specimen for laboratory testing as specified by the department from each newborn no later than forty-eight hours following birth.

(c) Use department-approved newborn screening specimen/information forms and directions for obtaining specimens.

(d) Enter all identifying and related information required on the newborn screening specimen/information form following directions of the department.

(e) In the event a parent or guardian refuses to allow newborn screening, obtain signatures from parents or guardians on the newborn screening specimen/information form.

(f) Forward the newborn screening specimen/information form with dried blood spots or signed refusal to the Washington state public health laboratory so that it will be received no later than seventy-two hours following collection of the specimen, excluding any day that the state laboratory is closed.

(2) Upon receipt of specimens, the department shall:

(a) Record the time and date of receipt;

(b) Perform appropriate screening tests for:

(i) Amino acid disorders;

(ii) Biotinidase deficiency;

(iii) Congenital hypothyroidism;

(iv) Congenital adrenal hyperplasia;

(v) Cystic fibrosis;

(vi) Fatty acid oxidation disorders;

(vii) Galactosemia;

(viii) Guanidinoacetate Methyltransferase (GAMT) deficiency;

(ix) Hemoglobinopathies;

((~~(ix)~~)) (x) Mucopolysaccharidosis type I (MPS-I);

((~~(x)~~)) (xi) Organic acid disorders;

((~~(xi)~~)) (xii) Pompe disease;

((~~(xii)~~)) (xiii) Severe combined immunodeficiency (SCID);

((~~(xiii)~~)) (xiv) Spinal muscular atrophy (SMA);

((~~(xiv)~~)) (xv) Urea cycle disorders;

(xvi) X-linked adrenoleukodystrophy (X-ALD).

(c) Report significant screening test results to the infant's attending health care provider or parent or guardian if an attending health care provider cannot be identified; and

(d) Offer diagnostic and treatment resources to health care providers attending infants with significant screening test results within limits determined by the department.

((~~(3) Once the department notifies the attending health care provider of significant screening test results, the attending health care provider shall notify the department of the date upon which the results were disclosed to the parent or guardian of the infant. This requirement expires January 1, 2020.~~))