Chapter 246-650 WAC
NEWBORN SCREENING

WAC

246-650-001	Purpose.
246-650-010	Definitions.
246-650-020	Performance of screening tests.
246-650-030	Implementation of screening to detect amino acid disorders, fatty acid oxidation disorders and organic acid disorders.
246-650-040	Report to the board.
246-650-050	Privacy and security of screening specimen/information forms.
246-650-990	Screening charge.
246-650-991	Specialty clinic support fee.

WAC 246-650-001 Purpose. The purpose of this chapter is to establish board rules to detect, in newborns, congenital disorders leading to developmental impairment or physical disabilities as required by RCW 70.83.050 and to provide protections for the confidentiality of information and human biological specimens submitted pursuant to these requirements.

[Statutory Authority: Chapters 70.83, 43.20 RCW. 03-24-026, § 246-650-001, filed 11/24/03, effective 12/25/03. Statutory Authority: RCW 43.20.050. 91-02-051 (Order 124B), recodified as § 246-650-001, filed 12/27/90, effective 1/31/91. Statutory Authority: RCW 43.20.050 and 70.83.050. 87-11-040 (Order 303), § 248-103-001, filed 5/18/87.]

WAC 246-650-010 Definitions. For the purposes of this chapter:

(1) "Amino acid disorders" means disorders of metabolism characterized by the body's inability to correctly process amino acids or the inability to detoxify the ammonia released during the breakdown of amino acids. The accumulation of amino acids or their by-products may cause severe complications including mental retardation, coma, seizures, and possibly death. For the purpose of this chapter amino acid disorders include: Argininosuccinic acidemia (ASA), citrullinemia (CIT), homocystinuria (HCY), maple syrup urine disease (MSUD), phenylketonuria (PKU), and tyrosinemia type I (TYR I).

(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) "Cystic fibrosis" means a life-shortening disease 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.

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

(8) "Fatty acid oxidation disorders" means disorders of metabolism characterized by the inability to efficiently use fat to make energy. When the body needs extra energy, such as during prolonged fasting or acute illness, these disorders can lead to hypoglycemia and metabolic crises resulting in serious damage affecting the brain, liver, heart, eyes, muscle, and possibly death. For the purpose of this chapter fatty acid oxidation disorders include: 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).

(9) "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.

(10) "Hemoglobinopathy" means a hereditary blood disorder caused by genetic alteration of hemoglobin which results in characteristic clinical and laboratory abnormalities and which leads to developmental impairment or physical disabilities.

(11) "Organic acid disorders" means disorders of metabolism characterized by the accumulation of nonamino organic acids and toxic intermediates. This may lead to metabolic crisis with ketoacidosis, hyperammonemia and hypoglycemia resulting in severe neurological and physical damage and possibly death. For the purpose of this chapter organic acid disorders include: 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).

(12) "Newborn" means an infant born in a hospital in the state of Washington prior to discharge from the hospital of birth or transfer.

(13) "Newborn screening specimen/information form" means the information form provided by the department including the filter paper portion and associated dried blood spots. A specimen/information form containing patient information is "Health care information" as defined by the Uniform Health Care Information Act, RCW 70.02.010(6).

(14) "Significant screening test result" means a laboratory test result indicating a suspicion of abnormality and

requiring further diagnostic evaluation of the involved infant for the specific disorder.

[Statutory Authority: Chapter 70.83 RCW. 08-13-073, § 246-650-010, filed 6/16/08, effective 7/17/08. Statutory Authority: Chapters 70.83, 43.20 RCW. 06-04-009, § 246-650-010, filed 1/20/06, effective 2/20/06; 03-24-026, § 246-650-010, filed 11/24/03, effective 12/25/03. Statutory Authority: RCW 43.20.050. 91-02-051 (Order 124B), recodified as § 246-650-010, filed 12/27/90, effective 1/31/91. Statutory Authority: Chapters 43.20 and 70.83 RCW. 91-01-032 (Order 114B), § 248-103-010, filed 12/11/90, effective 1/11/91. Statutory Authority: RCW 43.20.050 and 70.83.050. 87-11-040 (Order 303), § 248-103-010, filed 5/18/87.]

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

(a) Inform parents or responsible parties, 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, and

(iv) The legal right of parents or responsible parties 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 prior to discharge from the hospital or, if not yet discharged, no later than five days of age.

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

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

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

(f) Forward the specimen/information form with dried blood spots or signed refusal to the Washington state public health laboratory no later than the day after collection or refusal signature.

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

(a) Perform appropriate screening tests for:

(i) Biotinidase deficiency;

(ii) Congenital hypothyroidism;

(iii) Congenital adrenal hyperplasia;

(iv) Galactosemia;

(v) Homocystinuria;

(vi) Hemoglobinopathies;

(vii) Maple syrup urine disease (MSDU);

(viii) Medium chain acyl-coA dehydrogenase deficiency (MCADD);

(ix) Phenylketonuria (PKU);

(x) Cystic fibrosis;

(xi) The amino acid disorders: Argininosuccinic acidemia (ASA), citrullinemia (CIT), and tyrosinemia type I (TYR 1) according to the schedule in WAC 246-650-030;

(xii) The fatty acid oxidation disorders: Carnitine uptake defect (CUD), long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHADD), trifunctional protein deficiency (TFP), and very long-chain acyl-CoA dehydrogenase deficiency (VLCADD) according to the schedule in WAC 246-650-030;

(xiii) The organic acid disorders: 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), propionic acidemia (PROP) according to the schedule in WAC 246-650-030;

(b) Report significant screening test results to the infant's attending physician or family if an attending physician cannot be identified; and

(c) Offer diagnostic and treatment resources of the department to physicians attending infants with presumptive positive screening tests within limits determined by the department.

[Statutory Authority: Chapter 70.83 RCW. 08-13-073, § 246-650-020, filed 6/16/08, effective 7/17/08. Statutory Authority: Chapters 70.83, 43.20 RCW. 06-04-009, § 246-650-020, filed 1/20/06, effective 2/20/06; 03-24-026, § 246-650-020, filed 11/24/03, effective 12/25/03. Statutory Authority: RCW 43.20.050 and 70.83.050. 92-02-019 (Order 225B), § 246-650-020, filed 12/23/91, effective 1/23/92. Statutory Authority: RCW 43.20.050. 91-02-051 (Order 124B), recodified as § 246-650-020, filed 12/27/90, effective 1/31/91. Statutory Authority: Chapters 43.20 and 70.83 RCW. 91-01-032 (Order 114B), § 248-103-020, filed 12/11/90, effective 1/11/91. Statutory Authority: RCW 43.20.050 and 70.83.050. 87-11-040 (Order 303), § 248-103-020, filed 5/18/87.]

WAC 246-650-030 Implementation of screening to detect amino acid disorders, fatty acid oxidation disorders and organic acid disorders. The department shall implement screening to detect the amino acid disorders, fatty acid oxidation disorders, and organic acid disorders listed in WAC 246-650-020 (2)(a)(xi), (xii) and (xiii) as quickly as feasible and not later than September 2008.

[Statutory Authority: Chapter 70.83 RCW. 08-13-073, § 246-650-030, filed 6/16/08, effective 7/17/08. Statutory Authority: Chapters 70.83, 43.20 RCW. 06-04-009, § 246-650-030, filed 1/20/06, effective 2/20/06; 03-24-026, § 246-650-030, filed 11/24/03, effective 12/25/03. Statutory Authority: RCW 43.20.050. 91-02-051 (Order 124B), recodified as § 246-650-030, filed 12/27/90, effective 1/31/91. Statutory Authority: Chapters 43.20 and 70.83 RCW. 91-01-032 (Order 114B), § 248-103-040, filed 12/11/90, effective 1/11/91.]

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.

[Statutory Authority: Chapters 70.83, 43.20 RCW. 03-24-026, § 246-650-040, filed 11/24/03, effective 12/25/03.]

WAC 246-650-050 Privacy and security of screening specimen/information forms. The specimen/information form submitted to the department pursuant to WAC 246-650-

020 becomes the property of the state of Washington upon receipt by the Washington state public health laboratory. The department shall protect the privacy of newborns and their families and assure that all specimen/information forms submitted for screening are protected from inappropriate use or access.

(1) Storage: The specimen/information forms shall be kept at ambient temperature in secured storage to preserve their confidentiality and prevent access by unauthorized persons.

(2) Retention/destruction: The specimen/information form shall be retained until the child is twenty-one years old in accordance with the requirements for hospitals specified in RCW 70.41.190. After this time the form will be destroyed.

EXCEPTION FOR PARENTAL REQUEST: Upon request of a parent or guardian (or a patient who is over the age of eighteen years), the department will destroy the specimen/information form only after all required screening tests have been performed and if the patient's screening/clinical status related to these tests is not in question.

(3) Access: Access to stored specimen/information forms shall be restricted to department employees and those contractors or others approved by the department as necessary to meet specific program needs. Access is contingent upon compliance with all applicable federal and state laws, regulations, and policies safeguarding the privacy and confidentiality of medical information. The department shall assure that those granted access understand the confidentiality requirements and have a signed confidentiality agreement on file.

(4) Release: Dried blood spot samples and specimen information may only be released when required by state or federal law or under the following conditions:

(a) A sample from a specimen and copies of associated information (patient information and testing results, if requested) may be released to:

(i) A health care provider at the request of the patient or their legal representative after completing and signing a written request form approved by the department. The release form must be provided to the director of newborn screening before the request will be fulfilled.

(ii) A researcher with the written, informed consent of the patient or their patient's legal representative as part of a research project that has been reviewed and approved by the DOH/DSHS human subjects research review board and the secretary or designee of the department of health.

(iii) A named person in a legally executed subpoena following review and approval of the state attorney general.

(iv) A person to whom release is mandated by order of a court of competent jurisdiction.

(b) Anonymous samples may be released if the department determines that the intended use has significant potential health benefit and that each of the following criteria have been met:

(i) The investigation design is adequate to assure anonymity will be preserved.

(ii) All newborn screening tests have been completed and the status of the infant is resolved.

(iii) At least one fully adequate spot will remain after the anonymous sample has been taken.

(iv) Sufficient resources (personnel) are available for extracting the samples.

(v) The DOH/DSHS human subjects research review board has reviewed and approved the investigation. This requirement may be waived by the department for a very small (i.e., less than 100 sample) pilot study where the intent is to evaluate a testing tool, as opposed to an evaluation where the intent is to measure some characteristic of a population.

(5) Notification: The department shall notify parents of the specimen storage, retention/destruction and access requirements through the department's newborn screening informational pamphlet.

[Statutory Authority: Chapters 70.83, 43.20 RCW. 03-24-026, § 246-650-050, filed 11/24/03, effective 12/25/03.]

WAC 246-650-990 Screening charge. The department has authority under RCW 43.20B.020 to require a reasonable charge from parents or responsible parties for the costs of newborn screening. The charge is to be collected through the facility where the specimen was obtained.

[Statutory Authority: RCW 70.83.040. 99-20-036, § 246-650-990, filed 9/29/99, effective 10/30/99. Statutory Authority: RCW 43.20B.020. 92-02-018 (Order 224), § 246-650-990, filed 12/23/91, effective 1/23/92. Statutory Authority: RCW 43.20.050. 91-02-051 (Order 124B), recodified as § 246-650-990, filed 12/27/90, effective 1/31/91. Statutory Authority: RCW 43.20.050 and 70.83.050. 87-11-040 (Order 303), § 248-103-030, filed 5/18/87.]

WAC 246-650-991 Specialty clinic support fee. (1) The department has the authority under RCW 70.83.040 to collect a fee for each infant screened to fund specialty clinics that provide treatment services for hemoglobin diseases, phenylketonuria, congenital adrenal hyperplasia, congenital hypothyroidism and other disorders defined by the state board of health under RCW 70.83.020.

(2) The specialty clinic support fee is $3.50. It is to be collected in conjunction with the screening charge from the parents or other responsible party through the facility where the screening specimen is obtained.

(3) However, effective through June 30, 2007, the department will collect an additional $3.10 to fund specialty clinics that provide treatment services for other disorders defined by the board under RCW 70.83.020.

[Statutory Authority: RCW 70.83.040. 05-20-108, § 246-650-991, filed 10/5/05, effective 11/5/05; 99-20-036, § 246-650-991, filed 9/29/99, effective 10/30/99.]