

AMENDATORY SECTION (Amending WSR 06-04-009, filed 1/20/06, effective 2/20/06)

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.

~~((2))~~ (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.

~~((3))~~ (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.

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

((5)) (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.

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

((7)) (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), ~~carnitine palmitoyl transferase deficiency type 1-A (CPT 1),~~ 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-PO₄ in the blood. If undetected and untreated, accumulated galactose-1-PO₄ may cause significant tissue and organ damage often leading to sepsis and death.

~~((8))~~ (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.

~~((9))~~ "Homocystinuria" means deficiency of enzymes necessary to break down or recycle the amino acid homocysteine resulting in a buildup of methionine and homocysteine. If undetected and untreated may cause thromboembolism, mental and physical disabilities.

~~(10)~~ "Maple syrup urine disease" (MSUD) means deficiency of enzymes necessary to breakdown the branch chained amino acids leucine, isoleucine, and valine resulting in a buildup of these and metabolic intermediates in the blood. If undetected and untreated may result in mental and physical retardation or death.

~~(11)~~ "Medium chain acyl-coA dehydrogenase deficiency" (MCADD) means deficiency of an enzyme (medium chain acyl-coA dehydrogenase) necessary to breakdown medium chain length fatty acids. If undetected and untreated, fasting, infection or stress may trigger acute hypoglycemia leading to physical and

~~neurological damage or death.))~~ (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 Healthcare Information Act, RCW 70.02.010(6).

~~(14) ("Phenylketonuria" (PKU) means a deficiency of an enzyme necessary to convert the amino acid phenylalanine into tyrosine resulting in a buildup of phenylalanine in the blood. If undetected and untreated may cause severely impaired mental functioning.~~

~~(15))~~ "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: 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.]

AMENDATORY SECTION (Amending WSR 06-04-009, filed 1/20/06, effective 2/20/06)

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((τ))i

(ii) Congenital hypothyroidism((τ))i

(iii) Congenital adrenal hyperplasia((τ))i

(iv) Galactosemia((τ));
(v) Homocystinuria((τ));
(vi) Hemoglobinopathies((τ));
(vii) Maple syrup urine disease((τ)) (MSDU);
(viii) Medium chain acyl-coA dehydrogenase deficiency((τ and)) (MCADD);

(ix) Phenylketonuria (PKU);

~~((iii))~~ (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), ~~carnitine palmitoyl transferase deficiency type 1-A (CPT 1)~~, 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: 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.]

AMENDATORY SECTION (Amending WSR 06-04-009, filed 1/20/06, effective 2/20/06)

WAC 246-650-030 Implementation of screening to detect ~~((cystic fibrosis))~~ amino acid disorders, fatty acid oxidation disorders and organic acid disorders. The department shall implement screening to detect ~~((cystic fibrosis))~~ 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 ~~((June—2006))~~ September 2008.

[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.]