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) "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) "Newborn" means an infant born in any setting in the state of Washington.

(13) "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) "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) "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) "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) "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) "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) "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.

[Statutory Authority: RCW 70.83.050, 70.83.090 and RCW 70.83.020. WSR 19-20-025, § 246-650-010, filed 9/23/19, effective 10/24/19. Statutory Authority: RCW 70.83.050 and 70.83.020. WSR 18-01-024, § 246-650-010, filed 12/8/17, effective 3/1/18. Statutory Authority: Chapter 70.83 RCW. WSR 14-21-017, § 246-650-010, filed 10/2/14, effective 11/2/14. Statutory Authority: RCW 70.83.020. WSR 13-24-072, § 246-650-010, filed 11/26/13, effective 1/1/14. Statutory Authority: Chapter 70.83 RCW. WSR 08-13-073, § 246-650-010, filed 6/16/08, effective 7/17/08. Statutory Authority: Chapters 70.83, 43.20 RCW. WSR 06-04-009, § 246-650-010, filed 1/20/06, effective 2/20/06; WSR 03-24-026, § 246-650-010, filed 11/24/03, effective 12/25/03. Statutory Authority: RCW 43.20.050. WSR 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. WSR 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. WSR 87-11-040 (Order 303), § 248-103-010, filed 5/18/87.]