105 CMR 270.000: BLOOD SCREENING OF NEWBORNS FOR TREATABLE DISEASES AND DISORDERS

Section

270.001: Purpose

The purposes of 105 CMR 270.000 are to:

(A) list those diseases and disorders with treatments that are known to be more effective if the condition is identified in the newborn/infant period and for which there is a reliable and effective blood screening test and diagnostic protocol; and

(B) ensure that every newborn is screened for markers of these diseases and disorders; and

(C) make available Optional Newborn Blood Screening for another set of diseases and disorders with treatments that are thought to be effective if the condition is identified in the newborn/infant period and for which testing shall be offered under an optional protocol to collect critical population-based data on prevalence of these diseases and disorders in the Massachusetts population, their natural history, and the efficacy of screening, diagnostic, and treatment protocols.

270.004: Definitions

Department means the Massachusetts Department of Public Health or its designated agent.

Diagnostic Evaluation means the clinical evaluation of an individual or the laboratory testing of a clinical specimen from an individual with signs or symptoms or screening indicators of a disease or disorder in order to confirm or rule out the disease or disorder in that individual.

Health Care Provider means the current primary care or other current physician for a person. Health Care Provider shall also include Advanced Practice Registered Nurses, duly licensed and registered to engage in advanced practice nursing activities by the Massachusetts Board of Registration in Nursing, and Physician Assistants, duly licensed and registered to practice as a physician assistant by the Massachusetts Board of Registration of Physician Assistants, who are currently providing care to a person.

Health Care Provider Attending a Newborn means the Director of Newborn Medicine or other treating physician designated by the hospital of birth/neonatal care unit to care for the newborn, or the physician who certifies the birth of the newborn. Health Care Provider Attending a Newborn shall also include Advanced Practice Registered Nurses, duly licensed and registered to engage in advanced practice nursing activities by the Massachusetts Board of Registration in Nursing, and Physician Assistants, duly licensed and registered to practice as a physician assistant by the Massachusetts Board of Registration of Physician Assistants, who care for the newborn.

Infant means any liveborn person who has yet to have had a first year birthday.

Mandated Newborn Blood Screening means the required statewide collection and testing of newborn/infant blood specimens from all newborns/infants in Massachusetts and related follow up activities for the benefit of the child tested (subject to religions exemption only) for diseases and disorders for which:

270.004: continued

(1) there is a significant, life-challenging risk of morbidity or mortality to those who have the disease or disorder if they are not treated in the newborn/infant period;
(2) a standard of care screening test is universally available;
(3) a standard of care diagnostic evaluation is universally available for all newborns/infants whose newborn screening results warrant such;
(4) a standard of care treatment for the screened newborn/infant is universally available;
(5) a standard of care treatment in the newborn/infant period is beneficial to the screened newborn with a confirmed diagnosis;
(6) resources for and access to treatment and counseling are available; and
(7) the positive health benefits outweigh the risks and burdens of screening and treatment.

Newborn means any liveborn infant who has not yet attained the age of 31 days from a birth occurring in the Commonwealth of Massachusetts or from a birth prior to transfer to a hospital in the Commonwealth of Massachusetts.

Newborn Blood Screening Program means the program operated either by the Department or its agent to conduct Mandated and Optional Newborn Blood Screening for the Commonwealth's newborns.

Optional Newborn Blood Screening Program means the universal offering of one or more Pilot Studies to the newborn population.

Pilot Study means a research protocol with an informed consent process approved by the Department's Institutional Review Board that includes statewide testing of newborn blood specimens and related follow up activities offered for those diseases and disorders that do not meet the criteria for Mandated Newborn Screening but are likely, based on an evaluation of additional information to be gained through the pilot study, to have the potential to meet the criteria for mandatory screening and provide a benefit to newborns. Pilot studies provide for the maintenance of specimen identifiers, allowing study results to be linked to, and reported for, specific individuals.

Residual Specimen means any dried blood spot collected in accordance with 105 CMR 270.000 or derivative specimen that remains following the completion of newborn blood screening and any necessary follow-up for the benefit of the child tested.

Screening Tests means the laboratory testing of clinical specimens from a population of individuals regardless of health status to detect markers or risk factors of a disease or a disorder.

Specimen means a blood sample collected according to Newborn Blood Screening Program guidelines on the filter paper that is provided by the Newborn Blood Screening Program.

270.005: Newborn Blood Screening Advisory Committee

(A) The Commissioner shall establish a permanent advisory committee to advise the Department on matters pertaining to the Newborn Blood Screening Program including, but not limited to:
(1) the listing of treatable diseases and disorders for which newborn blood screening should be mandated or offered as optional newborn blood screening;
(2) how to best provide newborn screening services for the residents of the Commonwealth;
(3) quality assurance and control measures utilized for the operation of the Newborn Blood Screening Program; and
(4) new and emerging research in newborn screening.

(B) Membership of the committee shall include, but not be limited to, parents and other consumers, practicing pediatricians, public health officials, neonatologists, obstetricians, clinicians and researchers specializing in newborn diseases and disorders, clinical geneticists, birth hospital representatives, Newborn Blood Screening Program professionals, medical ethicists, and other experts as needed to represent a variety of related fields such as emerging technologies and health insurance.

270.005: continued

(C) On an annual basis, the Newborn Blood Screening Program shall submit to the committee a report on the quality assurance and control measures utilized for the operation of the Newborn Blood Screening Program and the committee shall provide review and guidance on these measures.

270.006: Diseases and Disorders Included in Newborn Blood Screening

(A) Mandated Newborn Blood Screening. The following diseases and disorders shall be included in mandated newborn screening:

(1) Inborn Errors of Metabolism.

(a) Amino Acid Disorders:

1. Maple Syrup Urine Disease (MSUD);
2. Homocystinuria (HCY);
3. Phenylketonuria (PKU);
4. Tyrosinemia, Type I (TYR I).

(b) Fatty Acid Oxidation Disorders:

1. Carnitine-Acylcarnitine Translocase Deficiency (CACT);
2. Carnitine Uptake Defect (CUD);
3. Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD);
4. Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD);
5. Very long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD).

(c) Organic Acidemias:

1. β-Ketothiolase Deficiency (BKT);
2. Glutaric Acidemia type I (GAI);
3. 3-Hydroxy-3-Methylglutaric Aciduria (HMG);
4. Isovaleric Acidemia (IVA);
5. Methylmalonic Acidemia: methylmalonyl CoA mutase deficiency (MUT);
6. Methylmalonic Acidemia: cobalamin A, B (Cbl A,B);
7. Methylmalonic Acidemia: cobalamin C,D (Cbl C,D);
8. Propionic Acidemia (PROP).

(d) Urea Cycle Disorders:

1. Argininemia (ARG) aka Arginase Deficiency;
2. Argininosuccinic Aciduria (ASA) aka Argininosuccinate Lyase Deficiency;
3. Carbamylphosphate Synthetase Deficiency (CPS);
4. Citrullinemia, Type I (CIT) aka Argininosuccinate Synthetase Deficiency;
5. Ornithine Transcarbamylase Deficiency (OTC).

(2) Other Miscellaneous Disorders of Involving Metabolic Pathways:

(a) Biotinidase Deficiency (BIOT);

(b) Galactosemia, Classical (GALT).

(c) Endocrinopathies:

1. Congenital Adrenal Hyperplasia (CAH);
2. Congenital hypothyroidism (CH).

(d) Infectious Diseases: Congenital Toxoplasmosis (TOXO).

(e) Hemoglobin Disorders:

1. Sickle cell anemia (Hb SS);
2. Hb S/C disease (Hb SC);
3. Hb S/ -thalassemia (Hb S/ Th).

(f) Other Genetic Disorders:

1. Cystic Fibrosis (CF);
2. Severe Combined Immunodeficiency (SCID).

(B) Pilot Studies. The Newborn Blood Screening Program shall identify and maintain a list of diseases and disorders that shall be included in the optional newborn blood screening as pilot studies. A current list of diseases and conditions available for optional screening shall be included in the New England Newborn Screening brochure provided to parents and guardians and shall be maintained on the web page for the New England Newborn Screening Program.

270.006: continued

(C) By-product Conditions. Due to the technology used for some screening tests, and/or the physiology associated with the disorders being tested for, some conditions not listed in 105 CMR 270.006(A) or offered as pilot studies pursuant to 105 CMR 270.006(B) may be identified during the screening process. These by-product conditions do not currently meet the criteria for mandated screening or pilot studies. These by-product conditions are listed in the New England Newborn Screening brochure provided to parents and guardians and are maintained on the web page for the New England Newborn Screening Program.

270.007: Collection and Submission of Newborn Blood Specimens

(A) Except as provided in 105 CMR 270.007(C), the health care provider attending a newborn shall ensure that a blood specimen is collected from the newborn between 24 and 48 hours after the birth. If a newborn child is discharged within the first 24 hours after the birth, the health care provider attending a newborn shall ensure that a blood specimen is collected prior to discharge and shall instruct the parents/guardians of the newborn to have a second specimen collected from the newborn within 48 hours of birth. The health care provider of a child after discharge from the birth hospital shall ensure that any repeat specimens requested by the Newborn Blood Screening Program are collected.

(B) The health care provider attending a newborn shall ensure that the blood specimen is collected from the newborn using the filter paper blood collection device provided by the Newborn Blood Screening Program and shall ensure that the specimen is submitted within 24 hours of collection or at next available specimen shipment for delivery to the Newborn Blood Screening Program in accordance with directions provided on the device and other supplemental information that may be provided by the Newborn Blood Screening Program. All information requested must be provided on the form associated with the device.

(C) If any parent or guardian objects to mandated newborn blood screening, the health care provider attending a newborn shall provide information about the benefits of mandated newborn screening and about the risks of refusing mandated newborn screening. A translator should be provided if necessary. If all of the infant's parents or guardians with legal custody refuse mandated newborn blood screening based on a conflict with their religious tenets and practices after being provided this information, the health care provider attending a newborn shall not conduct the newborn blood screening required by 105 CMR 270.007(A) and shall document the refusal on a form provided by the Newborn Blood Screening Program. The completed and signed form shall be sent to the Newborn Blood Screening Program and to the infant's health care provider of record.

(D) The health care provider attending a newborn shall ensure that a copy of the Newborn Screening Program brochure is provided to parents or guardians to inform them about the mandated screening tests and the availability of optional screening for pilot studies as specified in 105 CMR 270.006(B).

(E) The health care provider attending a newborn shall ensure that the consent and documentation procedures, as specified by the Newborn Blood Screening Program, are followed.

270.008: Newborn Blood Screening Fees

The Newborn Blood Screening Program shall bill the hospital of birth, or the parents for out-of-hospital births, a reasonable charge for the testing of newborns for those diseases or disorders screened for pursuant to 105 CMR 270.006 and for notification and follow-up to ensure treatment of affected newborns.

270.009: Notification of Newborn Blood Screening Test Results

The Newborn Blood Screening Program shall notify the hospital of birth and the health care provider listed by the hospital on the newborn screening collection device or the health care provider determined to be the infant's health care provider of the results of the newborn blood screening tests as soon as feasible. The infant's health care provider shall notify the parent or guardian of the newborn of the newborn's screening test results.

270.009: continued

The Newborn Blood Screening Program shall provide to the health care provider, upon request, the results of all or specified newborn blood screening tests for his or her patient.

270.010: Follow-up of Newborn Blood Screening

For the purposes of quality assurance, quality improvement and ongoing evaluation of the effectiveness of the Newborn Blood Screening Program, including the determination of those disorders and diseases that should be included in the Department's Newborn Blood Screening Program, the health care provider shall report to the Newborn Blood Screening Program, upon request, the following information within 30 days of the request:

(A) Diagnostic and long term outcomes for all newborns whose newborn screening results warranted diagnostic evaluation for a newborn screening disorder or disease; and

(B) Any additional, relevant information regarding these diagnostic and long term outcomes as specified by the Newborn Blood Screening Program.

270.011: Confidentiality of Newborn Screening Test Results and Information

(A) The Newborn Blood Screening Program shall maintain the confidentiality of testing results generated by the Program and information submitted to the Program for purposes of newborn screening and follow-up of newborn testing.

(B) Other than as required for routine notification of newborn blood testing as described in 105 CMR 270.009, the Newborn Blood Screening Program shall not disclose newborn screening results or any information or patient identifiers which because of name, identifying number, mark or description can be readily associated with a particular individual or the individual's parents/guardians, or any newborn screening information that could be used in combination with other information to identify an individual, except to:

(1) that individual;
(2) the individual's parents or guardians if a minor;
(3) the executor or administrator of the individual's estate;
(4) anyone authorized in writing by that individual or the individual's legal representative;
(5) authorized Department and New England Newborn Screening Program personnel;
(6) authorized staff at other state newborn screening programs responsible for the follow up of the particular newborn;
(7) anyone authorized to receive such information pursuant to a court order; or
(8) any researcher approved by the Department to conduct a specific study pursuant to M.G.L. c. 111, § 24A, and when approved by the Department's or the University of Massachusetts Medical School's Institutional Review Board. Except as provided in 105 CMR 270.011(C), newborn screening information or data that could be used to identify an individual shall not be disclosed to a researcher without the written consent of a parent or guardian.

(C) The Newborn Screening Program may provide a Limited Data Set, as defined in the Health Insurance Portability and Accountability Act (HIPAA) Privacy Rule (45 CFR §164.514(e)), of newborn screening results or information to researchers upon approval from the Department and subject to the terms of a written data use agreement.

(D) All individually identifiable testing results and information, as defined by the Department, shall be considered confidential and shall not be available as a public record under M.G.L. c. 66.

270.012: Storage and Use of Residual Specimens and Data

(A) Residual specimens and data shall be retained, stored, used, and destroyed by the Newborn Screening Program only in accordance with the Massachusetts Department of Public Health Policy on the Retention, Storage and Use of Newborn Screening Data and Residual Specimens.

270.012: continued

(B) Except as provided in 105 CMR 270.012(C), the Newborn Blood Screening Program shall retain and use residual specimens for a minimum of 15 years and a maximum of 16 years for the purposes of:

(1) Laboratory Quality Assurance, including laboratory quality control, laboratory validation, participation in proficiency testing, and the practice of continuous quality improvements;

(2) Individual or family clinical benefit;

(3) Individual or family forensic purposes;

(4) Uses authorized by law;

(5) Research studies approved by the Department or UMMS Institutional Review Board provided that a residual specimen is not provided to a researcher without the written consent of the parent or guardian for specimens collected on or after March 16, 2015. The IRB retains discretion to require consent for use of any stored residual specimen for any particular research study.

(C) Upon written request from all parents or legal guardians of a child, the Newborn Blood Screening Program shall destroy a stored residual specimen within a reasonable period of time not to exceed one year from the receipt of the written request.

REGULATORY AUTHORITY

105 CMR 270.000: M.G.L. c. 111, §§ 3, 4E, 5, 6, 24A and 110A.