MASSACHUSETTS DEPARTMENT OF PUBLIC HEALTH

POLICY ON THE RETENTION, STORAGE, AND USE OF NEWBORN SCREENING DATA AND RESIDUAL SPECIMENS

DECEMBER 2015

I. Introduction
II. Background
III. Definitions
IV. Rationale for Retaining and Storing Data and Residual Specimens
V. Retention Periods and Storage Conditions of Data and Residual Specimens and Data
VI. Priorities For Use of Residual Specimens
VII. Use of Residual Specimens and Data for Research
VIII. Penalties
IX. Notice

I. Introduction

This Massachusetts Department of Public Health (MDPH) Policy ("MDPH Policy") shall be implemented by the New England Newborn Screening Program (NENSP) of the University of Massachusetts Medical School (UMMS) in conjunction with the "Agreement between the MDPH and the UMMS to Provide Newborn Screening Services" ("Agreement").

This MDPH Policy applies to all specimens collected pursuant to MDPH authority on Massachusetts Newborn Screening (NBS) collection devices for purposes of providing newborn screening services, and to all data generated from collected specimens, as authorized under law and regulations. NENSP policies or standard operating procedures may address additional matters not covered by this MDPH Policy. In the event that there is a conflict between the MDPH Policy and the policies or standard operating procedures of the NENSP, the provisions of the MDPH Policy shall be controlling with respect to the retention, storage, use, and destruction of residual specimens collected on Massachusetts NBS collection devices for purposes of providing newborn screening services and data derived from such specimens. MDPH recognizes that the requirements for retention, storage, use, and destruction of specimens and data must be compatible with the operating budget of the NENSP.

II. Background

MDPH is authorized pursuant to Massachusetts General Laws chapter 111, sections 4E and 110A, to establish a newborn screening program and to promulgate regulations directing that newborns be tested for treatable genetic or biochemical disorders or infectious diseases as determined by the Commissioner of Public Health. These diseases and disorders are listed in 105 CMR 270.000: *Blood Screening of Newborns for Treatable Diseases and Disorders.*

Pursuant to the Agreement, MDPH has authorized UMMS, as an agent of MDPH, to provide newborn screening services for children born in Massachusetts or treated in Massachusetts neonatal units. The NENSP of UMMS tests Massachusetts newborns for those specific diseases and disorders set forth in 105 CMR 270.000 and is the custodian of all specimens and data that are collected or generated as part of newborn screening services.

The NENSP performs laboratory testing on dried blood spot and other specified clinical samples for newborn screening for the diseases and disorders that it is authorized to screen for pursuant to 105 CMR 270.000. To ensure optimal newborn screening services, MDPH requires the NENSP to conduct Quality Assurance, which includes use of some specimens for Laboratory Quality Control, Laboratory Validation, Proficiency Testing and Continuous Quality Improvements, as further described in IV. B. below.

III. Definitions

Approved Research means a research study that has been approved (according to procedures described in Section VII) by the NENSP and MDPH, and at least one federally approved Institutional Review Board.

Coded Specimen means a series of letters and/or numbers that are affixed to a specimen instead of direct subject identifiers. If a "key" that links the code with potentially identifiable information about the individual is disclosed with the coded specimen, such coded specimens are not de-identified.

De-Identified Information means information that alone, or in combination with other information provided or readily accessible, to an investigator, does not identify a particular individual to said investigator. In compliance with the Newborn Screening Saves Lives Act of 2014, Section 12, any dried blood spots collected after March 18, 2015, are not considered de-identified even if no other information about the individual is provided with the specimens.

Research means a systematic investigation, including research development, testing, and evaluation, designed to develop or contribute to generalizable knowledge. The term "research" includes activities designed to test a hypothesis and permit conclusions to be drawn. Research is usually described in a formal protocol that sets forth an objective and a set of procedures designed to reach that objective. Laboratory Quality Assurance activities, as described in Section IV, are generally excluded from the definition of research.

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

IV. Rationale for Retaining and Storing Data and Residual Specimens

Newborn screening data and residual specimens are retained and may only be used for:

A. Legal Accountability and Compliance with Legal Mandates

Legal accountability includes the ability to negate or confirm previously reported newborn screening results for a specimen belonging to a child for whom the clinical outcome appears to be in conflict with the previously reported result.

Compliance with legal mandates includes:

1. Confirmation of collection of an adequate specimen
2. Compliance with record retention requirements of the federal Clinical Laboratory Improvement Amendments and the Massachusetts Records Retention Laws.

B. Laboratory Quality Assurance

Laboratory quality assurance purposes, which include quality control, validation, proficiency testing and continuous quality improvement.

1. Previously characterized residual specimens (specimens that have already been tested and for which the results are known) are necessary for use as quality controls in the performance of assays; previously characterized specimens may be used as either negative or positive controls within an assay.
2. Previously characterized residual specimens are necessary to validate newly purchased reagent lots, newly purchased replicate instruments or updates to ongoing methodologies
3. Previously characterized residual specimens are necessary to validate alternatives to currently used reagents, alternative instruments or alternative methodologies for detection of a currently evaluated analyte.

4. Previously characterized residual specimens are necessary for annual staff competencies and, in the absence of alternative sources, for inter-laboratory comparison.
5. Previously characterized residual specimens from infants known to have a particular condition and from infants believed to be free of a particular condition are necessary for continuous quality improvements to the NBS program, e.g. developing new methods for detection of conditions in current newborn screening panels.
6. Residual specimens (yet-to-be-characterized) from infants known to have a particular condition and from infants believed to be free of a particular condition are necessary for continuous quality improvements to the NBS program, e.g. developing new methods and validating new methods for detection of conditions likely to be included in future newborn screening panels.

Note: When there is a question concerning whether a quality assurance protocol is purely quality assurance or may have a research component, the protocol will be submitted to the MDPH Institutional Review Board (IRB) for an opinion. If the protocol is determined to have a research component, the protocol must be reviewed and approved in accordance with the research requirements in section VII.

C. Individual or Family Clinical Benefit

When no other specimen is available, at the request of a parent/guardian or physician so authorized by either, residual specimens (when available) and/or their respective data will be made available for additional testing by a clinical laboratory for the medical benefit of the specimen subject or for the clinical benefit of that individual's family. Specimens will only be released for research studies pursuant to the procedures outlined in Section VII.

D. Individual or Family Forensic Purposes

Residual specimens (when available) and/or their respective data may be used by parents or legal guardians of a specimen subject for the identification of missing, disfigured or deceased children when the parent or the legal guardian of the specimen subject provides written authorization for release of the residual specimen or data.

E. Uses Authorized by Law

Residual specimens or data may be used and/or disclosed as required by law.

1. NENSP may provide a residual specimen or data in response to a court order. Upon receipt of a court order, the NENSP shall use its best efforts to notify the specimen subject (if not a minor) or the parent or legal guardian of the specimen subject, of a court order, to afford them an opportunity to object to the order in court.

2. Residual specimens or data may be used pursuant to the authority of the Office of the Medical Examiner under M.G.L. c. 38, § 2A. At the request of the local district attorney, any local child fatality review team established under M.G.L. c. 38, § 2A shall be provided information and records relevant to the cause of death of a child whose death is being reviewed by the local team.

F. Approved Research

Residual specimens and data have scientific value and under appropriate circumstances (see Section VII, Use of Residual Specimens and Data for Research) may be made available for research projects that seek to:

1. Further understand conditions included in newborn screening activities;
2. Further understand other health conditions of newborns when no other specimens are available; and
3. Contribute to general medical knowledge when there is no other way to conduct the proposed research.

Approved research includes only those research activities approved under the procedures outlined in Section VII.

V. Retention Periods and Storage Conditions of Residual Specimens and Data

A. Duration of Specimen Storage

Except as provided in V. B. below, residual specimens shall be retained for a minimum of 15 years and a maximum of 16 years from date of receipt by the NENSP. A residual specimen may be retained for a longer period with written consent from the parent/guardian of the specimen subject. This retention period:

1. Provides the individual's family adequate time to request access to the specimen or to request a longer storage period.
2. Ensures availability of evidence for potential lawsuits for a reasonable period of time concerning the accuracy of the testing performed. Clinical presentation of symptoms for newborn screening conditions have natural histories with initial clinical presentations occurring in infancy and early childhood and very few initial clinical presentations occurring in adulthood.
3. Maximizes the possibility that a sufficient number of specimens from individuals with rare conditions will be available for NENSP quality assurance activities.

B. Exception to Duration of Specimen Storage

Upon written request that is signed by all parents or legal guardians of a child, the NENSP will destroy residual specimens as soon as reasonably feasible, but no later than the later of two dates: the end of the legal compliance period (as specified in IV. A.) or one year after receipt of the written request.

C. Specimen Storage Conditions

1. Residual specimens shall be stored in conditions that promote the likelihood of scientific utility (generally –20 degrees C). It is recognized that the absolute value of a measure of an analyte may or may not be stable over time of storage. With appropriate controls, the interpretation of the measured value can be useful for some applications.

2. Residual specimens shall be stored with unique identifiers to enable future retrieval and identification for all purposes summarized in Section IV. All identifiers attached to the specimen will be in codified form so that patient identification will require crosslinking with separate data sources.

3. Residual specimens shall be retained in a secure environment under the direction of the NENSP. NENSP protocols and procedures shall ensure that no specimen or group of specimens shall be retrieved for any purposes other than authorized uses specified in this Policy.

D. Data Storage Conditions

Newborn screening data shall be securely stored and maintained in a manner that prevents unauthorized access and unauthorized disclosure.

VI. Priorities For Use of Residual Specimens

Newborn screening specimens are limited in quantity to that collected on the collection device. The following shall guide the priority of uses of such specimens to ensure their maximum utility.

A. Upon written notice of an actual or pending legal action pertaining to a particular specimen (e.g., related to a clinical presentation or medical finding that appears to be inconsistent with an “In Range” NBS result or an absence of a clinical presentation or medical finding that appears to be inconsistent with an “Out of Range” NBS result), such court action for access to the particular residual specimens shall have the highest priority.
B. The next highest priority of use of any residual specimen after VI. A. above is for NBS laboratory quality assurance (See Section IVB) and NENSP staff shall have unlimited access to stored specimens for the purposes of quality assurance.
C. Any residual specimen that has not been depleted by activities described in VI. A. or B. above may be made available for activities related to individual or family

clinical benefit and individual or family forensic purposes. (See Sections IV.C. and D.)

D. Any residual specimen that has not been depleted by activities described in VI. A., B., or C. above may be made available for activities related to approved research. (See Section IV.F.)
E. Any residual specimen that has not been depleted by activities described in VI. A., B., C., or D. above may be made available for activities related to uses authorized by law (See Section IV.E.).
F. Approved research has the lowest priority; consequently no research project may be approved unless the NENSP determines that the research study would not deplete a residual specimen and the research cannot practicably be carried out without the use of the residual specimen. Within the subsection IV.F. Approved Research, there is a hierarchy of priorities for use of residual specimens. Highest priority shall be for research projects that seek to further the understanding of conditions included in newborn screening activities; next priority shall be for research projects that seek to further the understanding of other health conditions of newborns; lowest priority shall be for research projects that seek to contribute to general medical knowledge. Residual specimens may not be used for research when other sources of available information can be utilized to achieve the same results.

VII. Use of Residual Specimens and Data for Research

Residual specimens and data may be provided to approved researchers, at the discretion of the NENSP and MDPH, under the conditions specified below. NENSP and MDPH recognize the importance of promoting public health research while protecting the rights of research subjects to the greatest extent possible. In order to respect the privacy rights of specimen and data subjects and their families, NENSP and MDPH conduct rigorous reviews of proposed research protocols. Approved researchers must adhere to strict requirements to protect the confidentiality of NBS specimens and data, and safeguard such specimens and data from unauthorized access or use.

A. Research Involving Residual Specimens

Use of residual specimens collected on or after March 18, 2015, for an approved research project always requires the written consent of the parent/guardian prior to release of the specimen to the approved researchers, regardless of whether or not potentially identifiable information is provided with the specimen. For all other specimens, the IRB shall determine whether or not consent is required for use of residual specimens.

Research using residual specimens shall only be allowed as follows:

For use of any residual specimen collected prior to March 18, 2015, that would be provided to the investigator as a de-identified specimen, the specimen may only be provided if:

1. The NENSP has reviewed and approved the research protocol and consent forms; and
2. Such research study is approved by the Commissioner of Public Health pursuant to M.G.L. c.111, § 24A; and
3. The NENSP determines that use of a residual specimen for the approved research study will not deplete all residual specimens for an individual; and
4. The research is approved by a federally authorized Institutional Review Board and, when MDPH or NENSP staff are engaged in the research study, the study is also approved by the MDPH IRB and/or University of Massachusetts Medical School IRB; and
5. The NENSP has confirmed that the parents or guardians have not requested in writing that their child's newborn screening residual specimen not be used for research.

For use of any residual specimen with identifiers linked to the newborn or for use of any residual specimen collected on or after March 18, 2015 (with or without identifiers), the specimen may only be provided if:

1. Conditions 1-4 above have been met; and
2. The parent/guardian provides written informed consent for the use the residual specimen for research.

B. Research Involving Newborn Screening Data

Researchers who are requesting access only to newborn screening data may follow the process in VII. C. below to request access to such data for research. NENSP shall not provide to any researchers the newborn screening data for any child whose parents or guardians, or the data subject if not a minor, have requested in writing that their child's newborn screening information not be used for research.

C. Process for Consideration of Research Proposals

Researchers interested in using newborn screening specimens or data for research should contact NENSP to discuss the feasibility of the project. NENSP, in consultation with MDPH, will determine whether the project may be feasible and warrants submission for review, and, if so, will instruct the researcher to submit an application for public health research to MDPH through IRBNet. MDPH, in consultation with NENSP, will carefully review the proposed study to determine whether it meets conditions for approval, including, but not limited to, adherence to MDPH policy, scientific rigor, adequate privacy and security protections, and priority of resources.

D. Costs

UMMS may charge researchers for costs related to specimen access, including, but not limited to: Management of assurance of consent, retrieval of the residual specimen(s), distribution of the specimen(s), monitoring of specimen use, return of any unused residual specimen to storage (or destruction), and documentation of such.

In addition, UMMS may charge researchers for costs related to data access, including but not limited to: Management of assurance of consent, development and implementation of data queries, development and submission of reports.

VIII. Penalties

A. MDPH and NENSP are responsible for ensuring that this Policy and all applicable human subject research and applicable state and federal privacy protection requirements are followed with regard to access and use of residual specimens and data.

B. Violations will be referred to the MDPH Office of General Counsel and the appropriate oversight agencies including the Office of Civil Rights, Office of Human Research Protections, and the Massachusetts Attorney General's Office for investigation and sanction as warranted.

****

REVISED and EFFECTIVE: December 1, 2015