Iowa Department of Public Health

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Iowa Department of Public Health Policy on the Purpose, Storage, and Use of Specimens Residual to Those Collected for Newborn Metabolic Screening Services

I Overview

The Iowa Department of Public Health (IDPH) is authorized pursuant to Iowa Code 136A and Iowa Administrative Code (IAC) 641-4.3 to establish a newborn metabolic screening program and directs that all newborns and infants born in the state of Iowa be tested for specific hereditary and congenital disorders as determined by the Director of Public Health and approved by the State Board of Health. These disorders are listed in IAC, 641-4.3(1). Comprehensive newborn screening services including laboratory, follow-up, consultative, and educational services are provided through the Iowa Neonatal Metabolic Screening Program (INMSP).

INMSP is a program under the Center for Congenital and Inherited Disorders (CCID) pursuant to Iowa Code chapter 136A and Iowa Administrative code 614-4.3. The CCID provides administrative oversight to the INMSP for the Iowa Department of Public Health. The University Hygienic Laboratory (UHL) is the designated central screening laboratory pursuant to IC chapter 136A and IAC 641-4.3. UHL tests Iowa newborns for those specific disorders set forth in 64.1-4.3 (1) and is the custodian of the residual specimens collected for newborn metabolic screening services in Iowa. The University of Iowa (UI) Department of Pediatrics, Divisions of Medical Genetics, Endocrinology and Diabetes, and Hematology are the designated consultants for the INMSP pursuant to IAC 641-4.3 and provide consultative, follow-up, and education activities. IDPH and the University of Iowa entered into a 28E agreement in March 1999 to ensure the provision of comprehensive newborn screening services for hereditary and congenital disorders in the state of Iowa through the INMSP.

This Policy shall be implemented by the INMSP, in conjunction with and upon revision of the UHL policy for storage, retention, use and disposition of specimens that are residual to testing performed by the INMSP. The new policy will address the definition of residual newborn metabolic screening specimens, stewardship, storage conditions, notice of storage of specimens to parents, validation of specimen integrity for specified analytes at defined time periods, access to residual specimens for specific diagnostic/clinical, quality assurance/ improvement and research purposes and standard operating procedures for inclusion to, monitoring of, and retrieval from the specimen repository. It is understood that the UHL policy may address additional technical and implementation matters not covered by this policy. In the event that there is a conflict between the policies, the provision of the IDPH Policy shall be controlling with respect to the use of residual specimens collected for newborn screening services in Iowa.

II. Basis for Retaining Residual Newborn Metabolic Screening Specimens

A residual newborn metabolic screening specimen is defined as a metabolic screening specimen using the INMSP collection form that is collected at the birth hospital, at the hospital of transfer, at the health care provider's office, an outpatient laboratory, or at home as a result of IAC 641-4.3(1) a **and** is leftover after the completed newborn screening services of the Iowa Neonatal Metabolic Screening Program. The INMSP collection form consists of dried blood spots on filter paper and attached baby and birthing center information.

Specimens are retained for several reasons:

1. Legal Accountability
    a. To confirm the existence of a specimen and its adequate collection
    b. Reconfirm newborn screening analytical results
    c. Allow for retesting of a specimen when a child has been subsequently diagnosed with a screenable disorder

2. Laboratory Quality Assurance/Improvement
    a. Necessary for laboratory to perform continuous quality assurance and improvement of testing methodologies

3. New Method Evaluations and Comparisons
    a. Necessary for laboratory to compare testing methodologies
    b. Necessary for laboratory to develop and validate new testing methodologies

4. Diagnostic/clinical purposes
    a. Availability of a specimen for diagnostic purposes in the event of an unexplained infant death or SIDS death. Definitive diagnosis is beneficial for counseling families and providing risk assessment for future pregnancies.
    b. Availability of a specimen when parent requests additional testing.

5. Epidemiological research to benefit the public health
    a. Permit the conduction of population studies on anonymized specimens to determine the incidence and prevalence of biochemical markers and/or genetic polymorphisms for disorders and diseases. An anonymized specimen is defined as one which cannot be traced back to or linked with the particular infant from whom the specimen was obtained.

6. Basic health-related research
    a. Permit the conduction of individualized studies on anonymized and identifiable specimens to advance medical knowledge on birth defects, disorders and disease.

Specimens shall be retained for a minimum of five years. The specimens shall be retained for one year at –70° C and then archived for four additional years at room temperature.

III. Continuous Quality Improvement

Continuous quality improvement (CQI) includes those activities designed to ensure test accuracy and to determine the feasibility of modified, additional or enhanced newborn screening tests. Activities also include validation of the integrity of specimens for specified analytes at defined time periods in order to justify retention period. INMSP investigators shall have unlimited access to specimens for the purposes of CQI. The INMSP may use linked specimens in pilot studies approved by the Birth Defects Advisory Committee for the purpose of incorporating new tests or evaluating new test methodologies. When there is any concern that the activity may have a research component, the project will be submitted to the University of Iowa Institutional Review Board for an opinion on how to proceed, including whether informed consent is required.

IV. Research

Research on anonymized or identifiable specimens shall be allowed in instances where such research would further 1) newborn screening activities; 2) the health of a newborn or child, for whom no other specimens are available or readily attainable; or 3) general medical knowledge for existing public health surveillance activities. Research on anonymized specimens shall also be allowed for population studies to benefit public health medicine.

A. Requests for use of anonymized residual newborn metabolic screening specimens
Researchers/investigators shall submit a proposal to the Center for Congenital and Inherited Disorders within the Iowa Department of Public Health. Documentation of human subjects review committee approval shall accompany the proposal.

The proposal shall:

1. Discuss project objectives, methodology, and rationale of how it will benefit the public health.

2. Identify the person or persons who will perform the study, their qualifications and organizational affiliation.

3. List the number of residual specimens needed for the study and anticipated duration of the study.

4. Discuss how long the residual specimens will be kept and how the specimens will be discarded at the conclusion of the study.
5. Describe how the results will be disseminated.

6. Indicate that residual specimens will be used only for the purpose stated in the proposal.

Review process:

1. The State Coordinator for Genetic Services will distribute the proposal to the Center for Congenital and Inherited Disorders' advisory committee members and schedule a presentation of the proposal for the next planned committee meeting.

2. The advisory committee will provide their written recommendations to the Iowa Department of Public Health director within two weeks of the presentation.

3. The IDPH director will review the recommendations and the proposal. Research projects approved by the IDPH director will be presented to the Iowa Board of Health.

4. The Iowa Board of Health shall give final approval of proposals. The State Coordinator for Genetic Services and/or the researcher will present the proposal at the next scheduled Iowa Board of Health meeting.

5. The State Coordinator for Genetics Services, the advisory committee chairperson and the IDPH director shall provide a written reply within six months of proposal submission.

B. Requests for use of identifiable residual newborn metabolic screening specimens submit a proposal to the Center for Congenital and Inherited Disorders within the Iowa Department of Public Health. Documentation of human subjects review committee approval of the research project and the parental consent form shall accompany the proposal.

The proposal shall:

1. Discuss project objectives, methodology, and rationale of how it will benefit public and individual health.

2. Identify the person or persons who will perform the study, their qualifications and organizational affiliation.

3. List the number of residual specimens needed for the study, the protocol for selecting and contacting subjects, and anticipated duration of the study.

4. Provide the protocol for selecting and contacting subjects.

5. Discuss how long the residual specimens will be kept, how the confidentiality of the specimens will be maintained, and how the specimens will be returned at the conclusion of the study.

6. Describe how the results will be disseminated.

7. Indicate that residual specimens will be used only for the purpose stated in the proposal.

8. Include a copy of the materials to be provided to the parent/guardian of the subject(s) of the residual specimens (i.e. educational materials, informed consent form).

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V. Individual Diagnostic/Clinical Purposes

When no other specimen is available, specimens will be made available for additional testing by a diagnostic laboratory at the request of a parent/guardian or health care provider so authorized either for the medical benefit of the specimen subject, or for the benefit of that individual's family. Specimens will only be released for research studies pursuant to the procedures outlined in Section IV. Specimens will also be made available for additional testing to an authorized officer from the State of Iowa Office of the Medical Examiner for diagnostic purposes in the cases of unexplained infant and child deaths, SIDS cases and child abuse cases upon receipt of parental/guardian consent.

VI. Confidentiality

INMSP shall maintain the confidentiality of all newborn metabolic screening records in accordance with state and federal laws and regulations. The UHL newborn screening laboratory shall not disclose personally identifiable records and/or residual specimens without informed parental consent. The INMSP personnel including CCID, UHL, and UI pediatric consultants shall not release any confidential newborn metabolic screening records to investigators for the purposes of determining research subjects.

VII. Access to Residual Specimens

Only UHL newborn screening laboratory personnel shall have access to residual specimens. Personally identifiable records and/or residual specimens shall only be released to researchers upon documentation of project approval, list of residual specimens needed, and informed parental consents. Identifiable records and/or residual specimens shall only be released to the medical examiner's office or to a diagnostic laboratory upon documentation of informed parental consent. The UHL shall only provide to researchers, the medical examiner's office, or diagnostic, a portion of the residual newborn screening specimen(s). The UHL shall assess a fee for retrieval of residual specimens and the fee structure will be outlined in the UHL retention policy.

VIII. Notice

Notice shall be given to parents at the time they receive the pamphlet entitled " Parent's Guide to Newborn Metabolic Screening" that (1) residual specimens, if available, will be stored and may be used without identifying information for research, and (2) permission will be sought for any proposed research, which would require identification of specimens.