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Canadian College of Medical Geneticists
Guidelines for Storage of DNA Samples

Purpose of the Present Statement:

The purpose of the present statement is to recommend guidelines for the storage of DNA samples for banking and retention practices in clinical genome diagnostics laboratories in Canada. **It is expected to serve as a resource for molecular genetics and genomics laboratories as they produce their own individual policies for storage of DNA samples (or DNA banking/retention).** The statement was produced by the CCMG Molecular Genetics Committee in 2006-7 and approved by the CCMG Board of Directors on January 23, 2008. The statement was revised by the CCMG Laboratory Practice Committee in 2020 and approved by the CCMG Board of Directors on November 23, 2020.

Scope of Present Statement:

The present statement provides guidelines for DNA storage (or banking/retention) in the context of medical genetic diagnosis in Canadian molecular genetics laboratories, genome diagnostics laboratories and clinical molecular genetic laboratories. The statement addresses both long-term banking of samples and storage of residual clinical samples remaining after diagnostic testing. This statement also applies to storage of RNA, blood or other tissues sent to molecular genetics and genomics laboratories for the expressed intent of extraction of genetic material for molecular genetic diagnostic purposes. For simplicity, the use of the term 'DNA storage (or banking/retention)' throughout the document includes the storage of the aforementioned materials.

Details about consent for research are not covered in the present statement. The process used for obtaining consent for research purposes should follow the CCMG / ACMG Joint Statement on The Process of Informed Consent for Genetic Research (https://www.ccmg-ccgm.org/documents/Policies_etc/Pos_Statements/PosStmt_EPP_CAGCInformedConsent_16Jul2008.pdf).

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Points for Consideration:

1. *Retention of long-term banked DNA samples versus storage of residual clinical DNA samples.*

Storage of DNA samples can be divided into two:

1) **Banking:** Long-term banked samples, where the purpose of retention is extended storage of genetic material for potential future use in clinical molecular diagnostic testing. Long-term banking is typically for ongoing complex analyses, for future investigations, or future testing of other family members.

2) **Retention:** Residual clinical samples, where genetic material remains after a current molecular genetic test is completed. Storage of residual clinical samples may occur for similar reasons. In addition, residual clinical samples might be used by laboratories for other purposes such as test development, validation or quality control.

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Recommendations for Laboratories:

Clinical genome diagnostics laboratories have a responsibility to define, document and disseminate their policies for DNA storage (or banking/retention), with respect to storage both of samples for long-term banking and residual clinical samples.

Referring health care providers have a responsibility to take into account laboratory banking policies, and to request appropriate long-term banking of samples when warranted. In the absence of special directives from the referring health care provider, the laboratory will be at liberty to invoke the communicated laboratory default policy with respect to disposal of residual clinical samples.

It is recommended that laboratory DNA storage (or banking/retention) policies include information regarding:

A. *Information Required for Informed Consent*

The consent process is the responsibility of the referring health care provider. The health care provider requires information about laboratory's DNA storage (or banking/retention) policies in order to adequately inform the patient. **It is required that the laboratory policies on DNA storage (or banking/retention) include information on the following:**

1. Duration of sample storage, for long-term banked samples and for residual clinical samples.
2. Procedures to be undertaken for disposal of long-term banked samples at the request of a patient.
3. Purpose for which the samples may be used other than those requested by the depositor (e.g. anonymously for test development, validation or quality control).
4. Risks, such as the possibility that the sample may be lost or otherwise compromised, and the laboratory procedures to deal with such an outcome.
5. Who has access to the sample, and if samples are to be stored, who will have access in future (e.g. patient, next of kin).

Laboratories may consider including a general statement as a reminder that in signing the requisition the clinician confirms that consent has been obtained.

B. Deposition of Samples

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1. The laboratory should accept samples only from health care professionals, and only when accompanied by proper documentation. Laboratories should develop policies outlining the minimal required information accompanying a sample and procedures undertaken in the event that inadequate information is provided. Similarly, policies should be developed outlining the minimal requirement for labeling samples and the procedures undertaken in the event that a sample is improperly labeled.

2. Special consideration may be needed for samples received from remote or atypical sources or from other laboratories.

C. Storage of Samples

1. Duration of storage: Length of storage for long-term banking should be at least one generation (25 years[4,5]). It is recommended that residual clinical DNA samples be stored for a minimum of one year after the laboratory has reported test results. However, it is recognized that laboratories may implement shorter storage times for residual DNA samples based on tests performed, clinical needs and local resources. Laboratories should establish and clearly document and disseminate their respective policies to referring health care providers.

2. Methods of storage: The relative stability of DNA and sensitivity of many assays allows a variety of options for sample storage to be considered, each with associated costs, limitations and risks and should be determined by each laboratory. There should be documented description of methods for specimen storage before and after testing.

3. Sample labelling: Coded labelling of samples is recommended. Each storage container should be labelled with two unique identifiers, and all other relevant information tied to the unique identifiers through data records. Laboratories should establish procedures undertaken in the event that a sample is improperly labeled.

D. Retrieval of Samples

1. Procedures for retrieval of samples from storage and associated handling should be developed.

2. Policies for destruction of samples at the end of the storage period should be developed.

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3. Policies concerning use of residual clinical samples for other clinical laboratory purposes (e.g. test development, validation and positive controls in the clinical laboratory) should be developed. Appropriate consideration should be given to anonymization and removal of identifiers from samples before use in these activities.

4. Use of samples for research must be conducted under institutional Research Ethics Board oversight.

5. Sharing of samples with other laboratories: Policies and consents should address who may have access to stored samples. When samples are shared with other laboratories for test validation and quality control purposes, they should be de-identified. It is recommended that whenever possible, and particularly for long-term banked samples, the laboratory responsible for shipping a sample should retain a back-up aliquot in case of sample loss, or for other future needs.

E. Information Linked to Samples

1. Policies should define how patient confidentiality will be maintained for banked samples. Laboratory facilities maintaining banked samples and banking records must be appropriately secured from unauthorized access.

2. Laboratory policies should include actions to be taken in case of loss or compromise of banked samples (e.g. a letter to the referring health care provider). Laboratory policies should also take into account actions to be taken in the case of an extended lapse of time between when the sample was submitted and the discovery of its loss or compromise. Residual clinical samples may not require this level of oversight.

3. The laboratory should have a policy to define when and how it will communicate new developments to referring health care providers.

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